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# Pediatrics

TENTH EDITION

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- ▶ NEW clinical vignette-style questions
- ▶ Targets what you really need to know
- ▶ Student-tested and reviewed

*Robert J. Yetman • Mark D. Hormann*



# Pediatrics

PreTest® Self-Assessment and Review

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# Pediatrics

PreTest® Self-Assessment and Review  
Tenth Edition

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# Contents

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*Introduction* . . . . . ix

## **General Pediatrics**

*Questions* . . . . . I  
*Answers, Explanations, and References* . . . . . 35

## **The Newborn Infant**

*Questions* . . . . . 61  
*Answers, Explanations, and References* . . . . . 89

## **The Cardiovascular System**

*Questions* . . . . . 113  
*Answers, Explanations, and References* . . . . . 121

## **The Respiratory System**

*Questions* . . . . . 131  
*Answers, Explanations, and References* . . . . . 143

## **The Gastrointestinal Tract**

*Questions* . . . . . 155  
*Answers, Explanations, and References* . . . . . 171

## **The Urinary Tract**

*Questions* . . . . . 185  
*Answers, Explanations, and References* . . . . . 197

## **The Neuromuscular System**

*Questions* . . . . . 207  
*Answers, Explanations, and References* . . . . . 220

**Infectious Diseases and Immunology**

*Questions* . . . . . **233**  
*Answers, Explanations, and References* . . . . . **250**

**Hematologic and Neoplastic Diseases**

*Questions* . . . . . **269**  
*Answers, Explanations, and References* . . . . . **278**

**Endocrine, Metabolic, and Genetic Disorders**

*Questions* . . . . . **289**  
*Answers, Explanations, and References* . . . . . **306**

**The Adolescent**

*Questions* . . . . . **327**  
*Answers, Explanations, and References* . . . . . **335**

*Bibliography* . . . . . **343**  
*Index* . . . . . **345**

# Introduction

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*Pediatrics: PreTest® Self-Assessment and Review*, Tenth Edition, provides comprehensive self-assessment and review within the field of pediatrics. The 500 questions contained in the book have been designed to be similar in format and degree of difficulty to the questions contained in Step 2 of the United States Medical Licensing Examination (USMLE).

Each question has the correct answer, an explanation, and a specific reference to a textbook. A bibliography that lists the sources used in the book follows the last chapter.

Perhaps the most effective way to use this book is to allow yourself one minute to answer each question in a given chapter in order to approximate the time limits imposed by the USMLE Step 2. As you proceed, indicate your answer to each question.

When you have finished answering the questions in a chapter, you should then spend as much time as you need verifying your answers and reading the explanations. Although you should pay special attention to the explanations for the questions you answered incorrectly, you should read every explanation. The editor has designed the explanations to reinforce and supplement the information tested by the questions. If, after reading the explanations, you want more information, you should consult and study the references indicated.

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# Pediatrics

PreTest® Self-Assessment and Review

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# General Pediatrics

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## Questions

**DIRECTIONS:** Each item below contains a question or incomplete statement followed by suggested responses. Select the **one best** response to each question.

**1.** Two weeks after a viral syndrome, a 9-year-old girl presents to your clinic with a complaint of several days of drooping of her mouth. In addition to the drooping of the left side of her mouth, you note that she is unable to completely shut her left eye. Her smile is asymmetric, but her examination is otherwise normal. This girl likely has

- a. Guillain-Barré syndrome
- b. Botulism
- c. Cerebral vascular accident
- d. Brainstem tumor
- e. Bell palsy

**2.** An infant can move his head from side to side while following a moving object, can lift his head from a prone position 45° off the examining table, smiles when encouraged, and makes cooing sounds. He cannot maintain a seated position. The most likely age of the infant is

- a. 1 month
- b. 3 months
- c. 6 months
- d. 9 months
- e. 12 months

**3.** A child is brought to your clinic for a routine examine. She can dress with help, can ride a tricycle, knows her own age, and can speak in short sentences. She had difficulty in copying a square. The age of this child is most likely

- a. 1 year
- b. 2 years
- c. 3 years
- d. 4 years
- e. 5 years

## 2 Pediatrics

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4. A 4-year-old girl is noticed by her grandmother to have a limp and a somewhat swollen left knee. The parents report that the patient occasionally complains of pain in that knee. An ophthalmologic examination reveals findings as depicted in the photograph. The condition most likely to be associated with these findings is



- a. Juvenile rheumatoid arthritis
- b. Slipped capital femoral epiphysis
- c. Henoch-Schönlein purpura
- d. Legg-Calvé-Perthes disease
- e. Osgood-Schlatter disease

5. The previously healthy 4-year-old child pictured presents to the emergency room with a 2-day history of a brightly erythematous rash and temperature of 40°C (104°F). The exquisitely tender, generalized rash is worse in the flexural and perioral areas. The child is admitted and over the next day develops crusting and fissuring around the eyes, mouth, and nose. The desquamation of skin shown occurs with gentle traction. This child most likely has

**A**

(Courtesy Adelaide Hebert, M.D.)

**B**

(Courtesy Adelaide Hebert, M.D.)

- Epidermolysis bullosa
- Staphylococcal scalded skin syndrome
- Erythema multiforme
- Drug eruption
- Scarlet fever

- 6.** You are counseling an adolescent patient about the long-term management of her asthma while she is away at college. She brought an article she found on the Internet that promotes an herbal supplement purported to be a “safe, natural” treatment for asthma. You recall a recent study in the medical literature showing inhaled steroids to be statistically superior to the advertised herbal supplement at preventing asthma exacerbations at the  $p < 0.05$  level. You explain to her that this means that
- The inhaled steroids are 5% better than herbal treatment
  - A critical threshold for medical significance has been reached
  - Patients will not benefit from the herbal treatment 5% of the time
  - The odds are less than 1 in 20 that the differences observed were only a chance variation
  - It would be unethical to use herbal treatment

### Items 7–8

- 7.** A previously healthy 8-year-old boy has a 3-week history of low-grade fever of unknown source, fatigue, weight loss, myalgia, and headaches. On repeated examinations during this time, he is found to have developed a heart murmur, petechiae, and mild splenomegaly. The most likely diagnosis is
- Rheumatic fever
  - Kawasaki disease
  - Scarlet fever
  - Endocarditis
  - Tuberculosis
- 8.** After you make the diagnosis in the previous case, you explain the findings to the family and instruct the family to
- Restrict the child from all strenuous activities
  - Give the child a no-salt-added diet
  - Ensure that the patient receives antibiotic prophylaxis for dental procedures
  - Test all family members in the home
  - Avoid allowing the child to get upset

9. A 5-year-old boy who was previously healthy has a 1-day history of low-grade fever, colicky abdominal pain, and a skin rash. He is alert but irritable; temperature is 38.6°C (101.5°F). A diffuse, erythematous, maculopapular, and petechial rash is present on his buttocks and lower extremities, as shown in the following figure. There is no localized abdominal tenderness or rebound; bowel sounds are active. Laboratory data demonstrate

Urinalysis: 30 red blood cells per high-powered field,  
2+ protein  
Stool: guaiac positive  
Platelet count: 135,000/ $\mu$ L

These findings are most consistent with



(Courtesy Adelaide Hebert, M.D.)

- a. Anaphylactoid purpura
- b. Meningococemia
- c. Child abuse
- d. Leukemia
- e. Hemophilia B

**10.** A 4-month-old baby boy has just arrived in the emergency room. He is cold and stiff. History from the parents is that the seemingly healthy infant had been placed in his crib for the night, and when they next saw him, in the morning, he was dead. Physical examination is uninformative. Routine whole-body x-rays are shown. The most likely diagnosis is



*(Courtesy Susan John, M.D.)*

- a. Scurvy
- b. Congenital syphilis
- c. Sudden infant death syndrome (SIDS)
- d. Osteogenesis imperfecta
- e. Battery

**11.** A mother brings an 18-month-old to the emergency center with the concern that the child may have ingested a substance. Which of the following is a contraindication to the use of ipecac in this child

- a. Age less than 5 years
- b. Breast-feeding
- c. Ingestion of alkali
- d. Ingestion of iron
- e. Concurrent administration of intravenous glucose

**12.** A mother calls you on the telephone and says that her 4-year-old son bit the hand of her 2-year-old son 2 days previously. The area around the laceration has become red, indurated, and swollen, and he has a temperature of 39.4°C (103°F). Your response should be to

- a. Arrange for a plastic surgery consultation to be scheduled in 3 days
- b. Admit the child to the hospital immediately for surgical debridement and antibiotic treatment
- c. Prescribe penicillin over the telephone and have the mother apply warm soaks for 15 min qid
- d. Suggest purchase of bacitracin ointment to apply to the lesion tid
- e. See the patient in the emergency room to suture the laceration

**Items 13–14**

**13.** The adolescent shown presents with a 14-day history of multiple oval lesions over her back. The rash began with a single lesion over the lower abdomen (A); the other lesions developed over the next days (B). These lesions are slightly pruritic. The likely diagnosis is



**A**

*(Courtesy Adelaide Hebert, M.D.)*



**B** (Courtesy Adelaide Hebert, M.D.)

- a. Contact dermatitis
- b. Pityriasis rosea
- c. Seborrheic dermatitis
- d. Lichen planus
- e. Psoriasis

**14.** The most appropriate initial therapy for the patient in the previous question is

- a. Phototherapy
- b. High-dose topical steroid therapy
- c. Systemic antifungal agents
- d. Coal-tar shampoos
- e. Observation and topical emollients

### **Items 15–16**

**15.** A very concerned mother brings a 2-year-old child to your office because of multiple episodes of a brief, shrill cry followed by a prolonged expiration and apnea. You have been following this child in your practice since birth and know that the child is a product of a normal pregnancy and delivery, has been growing and developing normally, and has no acute medical problems. The mother relates that the first episode in question occurred immediately after the mother refused to give the child some juice. The child became cyanotic and unconscious and had generalized clonic jerks. A few moments later the child awakened and had no residual effects. A second episode of identical nature occurred at the grocery store when the father of the child refused to purchase a toy for the child. Your physical examination reveals a totally delightful and normal child. The most likely diagnosis in this case is

- a. Seizure disorder
- b. Drug ingestion
- c. Hyperactivity with attention deficit
- d. Pervasive development disorder
- e. Breath-holding spell

**16.** The most appropriate course of action in the previous case would be to

- a. Obtain an EEG and neurologic consultation prior to starting anticonvulsants
- b. Begin anticonvulsants while awaiting the results of an EEG, a neurologic consultation, and a urine drug screen
- c. Initiate a trial of methylphenidate (Ritalin)
- d. Instruct the family to splash cold water on the child's face and begin mouth-to-mouth resuscitation should another episode occur
- e. Reassure the family of the likely benign nature of the problem and offer counseling for appropriate behavior modification

**17.** The 3-day-old infant pictured has a facial rash. The most likely diagnosis is



*(Courtesy Adelaide Hebert, M.D.)*

- a. Herpes
- b. Neonatal acne
- c. Milia
- d. Seborrheic dermatitis
- e. Eczema

**18.** You are called to the emergency room to see one of your patients. The father of this 3-year-old was spraying the yard with an unknown insecticide. In the emergency room, the child is noted to have bradycardia, muscle fasciculations, meiosis, wheezing, and profound drooling. The most likely agent included in this pesticide is

- a. Organophosphate
- b. Chlorophenothane (DDT)
- c. Sodium cyanide
- d. Warfarin
- e. Paraquat

## 12 Pediatrics

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**19.** A 2-year-old child (A) presents with a four-day history of a rash limited to the feet and ankles. The papular rash is both pruritic and erythematous. The 3-month-old sibling of this patient (B) has similar lesions also involving the head and neck. Appropriate treatment for this condition includes



**A**

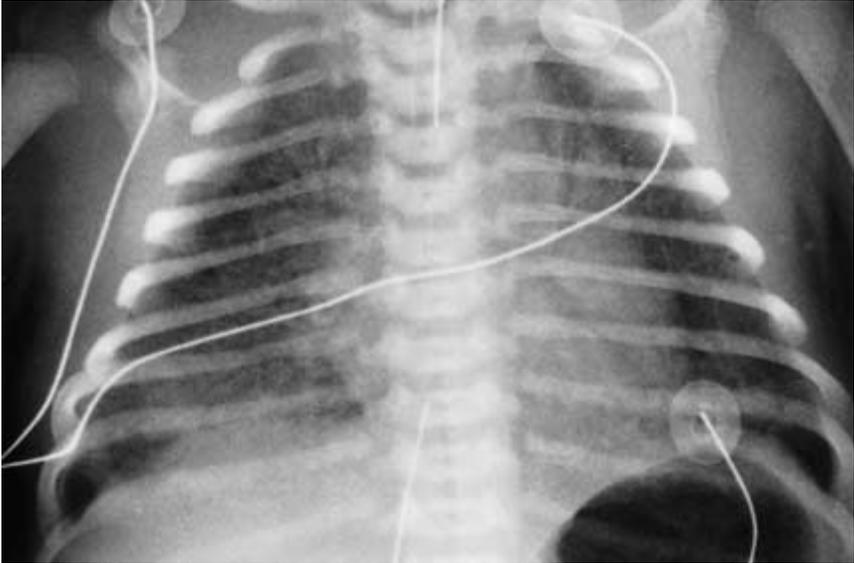


**B**

*(Courtesy Adelaide Hebert, M.D.)*

- a. Coal-tar soap
- b. Permethrin
- c. Hydrocortisone cream
- d. Emollients
- e. Topical antifungal cream

**20.** An 8-h-old infant develops increased respiratory distress, hypothermia, and hypotension. A CBC demonstrates a WBC of  $2500/\mu\text{L}$  with 80% bands. Which of the following diagnoses is most likely?



*(Courtesy Susan John, M.D.)*

- a. Gonococcal eye infection
- b. Diaphragmatic hernia
- c. Group B streptococcal pneumonia
- d. Transient tachypnea of the newborn
- e. Chlamydial pneumonia

**21.** A 16-year-old basketball player complains of pain in his knees. A physical examination reveals, in addition to tenderness, a swollen and prominent tibial tuberosity. Radiographs of the area are unremarkable. The most likely diagnosis is

- a. Osgood-Schlatter disease
- b. Popliteal cyst
- c. Slipped capital femoral epiphysis
- d. Legg-Calvé-Perthes disease
- e. Gonococcal arthritis

**22.** You are performing a well-child examination on a new patient, the 1-year-old child shown in the picture. Your next action should be to



*(Courtesy Kathryn Musgrove, M.D.)*

- a. Patch the eye with the greater refractive error
- b. Patch the eye that deviates
- c. Defer patching or ophthalmologic exam until the child is older and better able to cooperate
- d. Reassure the mother that he will outgrow it
- e. Refer immediately to ophthalmology, as central vision may fail to develop if the diagnosis and treatment are delayed

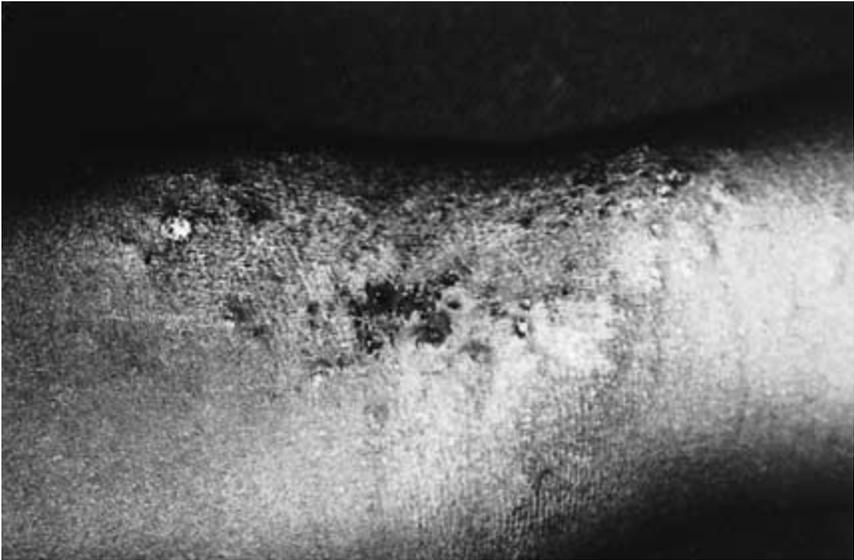
**23.** You are seeing a 2-month-old infant, brought by her father for a well-child examination. He has concerns about maintaining a safe environment for his child. In providing age-appropriate anticipatory guidance, you tell him that

- a. He should set his water heater to 71°C (160°F) to ensure the sterility of dishes and clothes, thereby decreasing the risk of infections
- b. His 2-month-old should not be given solid food at this time
- c. Pillows in the crib should be soft to provide a comfortable sleeping environment
- d. Infants should be placed on their stomachs to sleep, as this decreases the risk of choking if they spit up
- e. An infant should never be left unattended in the bathtub until she learns to sit on her own, and then only for brief periods of time

**24.** An infant who sits with only minimal support, attempts to attain a toy beyond reach, and rolls over from the supine to the prone position, but does not have a pincer grasp, is at a developmental level of

- a. 2 months
- b. 4 months
- c. 6 months
- d. 9 months
- e. 1 year

**25.** A 5-year-old boy presents with the severe rash pictured as follows. The rash is pruritic, and it is especially intense in the flexural areas. The mother reports that the symptoms began in infancy (when it also involved the face) and that her 6-month-old child has similar symptoms. The most likely diagnosis of this condition is



*(Courtesy Adelaide Hebert, M.D.)*

- a. Seborrheic dermatitis
- b. Superficial candidiasis
- c. Psoriasis
- d. Eczema
- e. Contact dermatitis

**26.** Appropriate treatment for the condition described in the previous question includes



*(Courtesy Adelaide Hebert, M.D.)*

- a. Coal-tar soaps and shampoo
- b. Topical antifungal cream
- c. Ultraviolet light therapy
- d. Moisturizers and topical steroids
- e. Topical antibiotics

**27.** An 18-month-old infant has an intensely pruritic scalp, especially in the occipital region, with 0.5-mm lesions noted at the base of hair shafts, as shown in the picture. The mother of this infant has been using the same hairbrush for herself and for her other two children (ages 2 months and 36 months). Which of the following therapies should be avoided?



*(Courtesy Adelaide Hebert, M.D.)*

- a. Treatment of all household contacts with 1% lindane (Kwell)
- b. Use of 1:1 vinegar-water rinse for hair for nit removal
- c. Washing of all clothing and bedding in very hot water
- d. Replacement of all commonly used brushes
- e. Advice to the mother that treatment will again be necessary in 7 to 10 days

**28.** You are called by a general practitioner to consult on a patient admitted to the hospital 4 days ago. The patient is a 7-month-old white boy with poor weight gain for the past 3 months, who has not gained weight in the hospital despite seemingly adequate nutrition. His guardian is his maternal aunt, as his mother is in jail for unknown reasons. You take a detailed diet history from the guardian, and the amounts of formula and baby food intake seem appropriate for age. Physical examination reveals an active, alert infant with a strong suck reflex who appears wasted. You note generalized lymphadenopathy with hepatomegaly. In addition, you find a severe case of oral candidiasis that apparently has been resistant to treatment. Which of the following is the most appropriate next step in the evaluation or treatment of this child?

- a. Increase caloric intake because this is probably a case of underfeeding
- b. Order HIV PCR testing because this is likely the presentation of congenitally acquired HIV
- c. Draw blood cultures because this could be sepsis
- d. Perform a sweat chloride test because this is probably cystic fibrosis
- e. Send stool for fecal fat because this is probably a malabsorption syndrome

**29.** A 2-year-old boy has been vomiting intermittently for 3 weeks and has been irritable, listless, and anorectic. His use of language has regressed to speaking single words. In your evaluation of this patient, the *least* likely diagnosis to consider is

- a. Subdural hematoma
- b. Brain tumor
- c. Tuberculous meningitis
- d. Food allergy
- e. Lead poisoning

**30.** Among the following, the *least* likely risk factor for hearing impairment is

- a. A maternal history of use of phenytoin during pregnancy
- b. A family history of hearing impairment
- c. Craniofacial abnormalities
- d. Birth weight less than 1500 g
- e. Neonatal hyperbilirubinemia

**Items 31–32**

**31.** An 8-month-old infant has a 2-day history of diarrhea and poor fluid intake. You diagnose a 10 to 15% dehydration. Which of the following fluids is appropriate to begin immediate resuscitation?

- a. D<sub>5</sub> ¼ normal saline
- b. D<sub>5</sub> ½ normal saline
- c. Normal saline
- d. Whole blood
- e. D<sub>10</sub>W

**32.** In this patient, you decide to give 20 mL/kg of the chosen fluid. A reasonable period of time over which to infuse this fluid is

- a. 2.5 min
- b. 60 min
- c. 4 h
- d. 8 h
- e. 24 h

**33.** You are seeing an established patient, a four-year-old girl brought in by her mother for vaginal itching and irritation. She is toilet trained and has not complained of frequency or urgency, nor has she noted any blood in her urine. Her mother noted she has been afebrile and has not complained of abdominal pain. Mom denies the risk of inappropriate contact; the girl also denies anyone “touching her there.” Your physical examination of the perineum is significant for the lack of foul odor or discharge. You do note some erythema of the vulvar area, but the hymenal ring is intact, without evidence of trauma. The appropriate course of action should be to

- a. Refer to pediatric gynecology for removal under anesthesia of a suspected foreign body in the vagina
- b. Reassure the mother, and counsel her to stop giving the girl bubble baths, have the girl wear only cotton underwear, and improve hygiene
- c. Refer to social services for suspected physical or sexual abuse
- d. Swab for gonorrhea and plate on chocolate agar
- e. Treat with an antifungal cream for suspected yeast infection

**34.** A 20-month-old child is brought to the emergency department because of fever and irritability and refusal to move his right lower extremity. Physical examination reveals a swollen and tender right knee that resists passive motion. The most important test to confirm the impression of septic arthritis is

- a. Examination of joint fluid
- b. X-ray of the knee
- c. Erythrocyte sedimentation rate (ESR)
- d. Complete blood count (CBC) and differential
- e. Blood culture

**35.** A 16-year-old high school boy sustained an abrasion of the knee after a fall while Rollerblading in the school yard. School records reveal that his last DPT booster was at age 6. In this situation, which of the following is appropriate?

- a. Tetanus toxoid
- b. Adult tetanus and diphtheria toxoid (Td)
- c. DPT booster
- d. Tetanus toxoid and tetanus immune globulin
- e. No immunization

**36.** A five-year-old boy is brought into the emergency room immediately after an unfortunate altercation with a neighbor's immunized Chihuahua that occurred while the child was attempting to dress the dog as a superhero. The fully immunized child has a small, irregular, superficial laceration on his right forearm that has stopped bleeding. His neuromuscular examination is completely normal, and his perfusion is intact. Management should include

- a. Antimicrobial prophylaxis
- b. Tetanus booster immunization and tetanus toxoid in the wound
- c. Copious irrigation
- d. Primary rabies vaccination for the child
- e. Destruction of the dog and examination of brain tissue for rabies

**37.** Aunt Mary is helping her family move to a new apartment. During the confusion, 3-year-old Jimmy is noted to be stumbling about, his face flushed and his speech slurred. The contents of Aunt Mary's purse are strewn about on the floor. In the emergency room, Jimmy is found to have a rapid heart-beat, blood pressure of 42/20, and dilated pupils. ECG shows prolonged QRS and QT intervals. Jimmy suddenly starts to convulse. His condition is most likely to be the result of poisoning with

- a. Barbiturates
- b. Tricyclic antidepressants
- c. Diazepam
- d. Organophosphates
- e. Arsenic

**38.** As a city public health officer, you have been charged with the task of screening high-risk children for lead poisoning. The best screen for this purpose is

- a. Careful physical examination of each infant and child
- b. Erythrocyte protoporphyrin levels (EP, FEP, or ZPP)
- c. CBC and blood smear
- d. Blood lead level
- e. Environmental history

**39.** Universal immunization of infants with a three-dose series of intramuscular, genetically engineered hepatitis B surface antigen vaccine is recommended. Implementation of this recommendation should decrease the incidence of which of the following?

- a. Neonatal hyperbilirubinemia
- b. Alcoholic liver disease
- c. Dubin-Johnson syndrome
- d. Hepatocellular carcinoma
- e. Hydrops of the gallbladder

**40.** New parents ask you how to reduce the chance of their baby suffering from sudden infant death syndrome (SIDS). You tell them to place the child in which of the following for sleep?

- a. Supine position
- b. Prone position
- c. Seated position
- d. Trendelenburg position
- e. A hammock

**41.** A mentally retarded 14-year-old boy has a long face, large ears, micropenis, and large testes. Chromosome analysis is likely to demonstrate which of the following?

- a. Trisomy 21
- b. Trisomy 18
- c. Trisomy 13
- d. Fragile X syndrome
- e. William syndrome

**42.** A 5-month-old child presents to the emergency room with generalized tonic clonic seizure activity of about 30-min duration that stops upon the administration of lorazepam. The most helpful information to gather from the mother would be

- a. Whether the child has had congestion without fever for the past 3 days
- b. Whether the child is developmentally normal, as are his siblings
- c. Whether the mother has been diluting the infant's formula to make it last longer
- d. The number of pets at home
- e. Whether the mother works as a secretary in an energy trading firm

**DIRECTIONS:** Each group of questions below consists of lettered options followed by numbered items. For each numbered item, select the appropriate lettered option(s). Each lettered option may be used once, more than once, or not at all. **Choose exactly the number of options indicated following each item.**

### Items 43–48

Many rashes and skin lesions can be found first in the newborn period. For each of the descriptions listed below, select the most likely diagnosis.

- a. Sebaceous nevi
- b. Salmon patch
- c. Neonatal acne
- d. Pustular melanosis
- e. Erythema toxicum
- f. Seborrheic dermatitis
- g. Milia

**43.** A 1-week-old child's mother complains that the child has a transient rash that has splotchy areas of erythema with a central clear pustule. Your microscopic examination of the liquid in the pustule reveals eosinophils. **(SELECT 1 DIAGNOSIS)**

**44.** An adolescent boy complains of a splotchy red rash on the nape of his neck, discovered when he had his head shaved for football season. The rash seems to get more prominent with exercise or emotion. His mother notes that he has had the rash since infancy, but that it became invisible as hair grew. He had a similar rash on his eyelids that resolved in the newborn period. **(SELECT 1 DIAGNOSIS)**

**45.** A nurse calls you to evaluate an African American baby that seems to have a bacterial skin infection, with many scattered pustules full of a milky fluid. Upon examining a pustule, it easily wipes away, revealing a hyperpigmented spot. **(SELECT 1 DIAGNOSIS)**

**46.** The obstetrical resident on call asks you to evaluate an area of a newborn's scalp that seems to have no hair and is scaly and yellowish. **(SELECT 1 DIAGNOSIS)**

**47.** A newborn's mother complains that her infant seems to have very small white dots all over his nose. The dots do not wipe off with bathing, but they are also not erythematous. **(SELECT 1 DIAGNOSIS)**

**48.** A newborn's father complains that his son has dandruff, with many waxy flakes of skin on the scalp. When he scrapes the lesions, hair often comes off with the flakes of skin. In addition, the baby has flaking of the eyebrows. **(SELECT 1 DIAGNOSIS)**

**Items 49–53**

For each otherwise normal child presented, choose the sleep disturbance most consistent with the history.

- a. Night terrors
- b. Nightmares
- c. Learned behavior
- d. Obstructive sleep apnea
- e. Somniloquy

**49.** A 3-year-old boy awakens every night around 2:00 A.M. screaming incoherently. His parents note that he is agitated, seems awake but unresponsive, and goes back to sleep within a few minutes. He has no memory of the episodes in the morning.

**50.** A 15-month-old toddler continues to wake up crying every night. Her parents give her a nighttime bottle, rock her, and sing to her to help her go back to sleep. Her parents are exhausted and ask you if she is having bad dreams.

**51.** Parents hear over their baby monitor that their 5-year-old girl regularly calls out during the night. When the parents check on her, she is sleeping comfortably and is in no apparent distress.

**52.** A 4-year-old boy occasionally wakes in the middle of the night crying. When his parents check on him, he seems visibly frightened and tells his parents that dogs were chasing him.

**53.** A 5-year-old child refuses to sleep in his bed, claiming there are monsters in his closet and that he has bad dreams. The parents allow him to sleep with them in their bed to avoid the otherwise inevitable screaming fit. The parents note that the child sleeps soundly, waking only at sunrise.

**Items 54–58**

For each of the injuries listed below, select the age at which it is most likely to occur.

- a. 6 months
- b. 1 year
- c. 2 years
- d. 6 years
- e. 10 years

**54.** Asphyxiation and choking (**SELECT 1 AGE**)

**55.** Drowning in a swimming pool (**SELECT 1 AGE**)

**56.** Pedestrian injury (**SELECT 1 AGE**)

**57.** Baby-walker injuries (**SELECT 1 AGE**)

**58.** Accidental poisoning (**SELECT 1 AGE**)

**Items 59–62**

For each disorder below, select the dietary deficiency that is likely to be responsible.

- a. Caloric deficiency
- b. Thiamine deficiency
- c. Niacin deficiency
- d. Vitamin D deficiency
- e. Vitamin C deficiency
- f. Vitamin B<sub>12</sub> deficiency
- g. Vitamin B<sub>6</sub> deficiency
- h. Biotin deficiency
- i. Riboflavin deficiency

**59.** Progressive weight loss, constipation, muscular atrophy, loss of skin turgor, hypothermia, and edema (**SELECT 1 DEFICIENCY**)

**60.** Dermatitis, diarrhea, and dementia (**SELECT 1 DEFICIENCY**)

**61.** Congestive heart failure, peripheral neuritis, and psychic disturbances (**SELECT 1 DEFICIENCY**)

**62.** Enlargement of costochondral junction, craniotabes, and scoliosis (**SELECT 1 DEFICIENCY**)

**Items 63–66**

Identify the syndrome based on the clinical presentation.

- a. Waardenburg syndrome
- b. Prader-Willi syndrome
- c. Marfan syndrome
- d. Treacher Collins syndrome
- e. Down syndrome

**63.** A 2-year-old boy has speech delay but otherwise normal intelligence and development. He has a broad nasal bridge, white forelock, heterochromic irises, and a ventriculoseptal defect diagnosed by echocardiogram. His mother wears a hearing aid and also has a white forelock.

**64.** A 17-year-old boy is well over 6 feet tall and has long slender limbs, hypermobile joints, myopia, and a midsystolic click on physical examination.

**65.** A 6-year-old boy weighs 150 lb and has small hands and feet, almond-shaped eyes, and hypogonadism.

**66.** A 4-year-old has conductive hearing loss, micrognathia, down-slanting palpebral fissures, abnormally shaped ears, and normal intelligence.

**Items 67–70**

For each case listed below, match the most likely diagnosis.

- a. Legg-Calvé-Perthes disease
- b. Slipped capital femoral epiphysis
- c. Osteomyelitis
- d. Septic arthritis of the hip
- e. Transient synovitis

**67.** An afebrile, obese 14-year-old boy has developed pain at the right knee and a limp.

**68.** A 6-year-old boy has developed a limp and has limited mobility of the hip, but denies pain and fever.

**69.** A 2-year-old refuses to walk, has fever, has significant pain with external rotation of the right leg, and has an elevated white blood cell count.

**70.** A 3-year-old refuses to walk, is afebrile, had an upper respiratory tract infection a week ago, has right hip pain with movement, and has a normal white blood cell count.

**Items 71–75**

For the most likely toxic substance involved in the cases below, match the appropriate treatment.

- a. Atropine and pralidoxime (2-PAM)
- b. *N*-acetylcysteine (Mucomyst)
- c. Meso-2,3-dimercaptosuccinic acid (DMSA succimer)
- d. Naloxone (Narcan)
- e. Sodium bicarbonate

**71.** Over the last several weeks, a 2-year-old girl has exhibited developmental regression, abnormal sleep patterns, anorexia, irritability, and decreased activity. These symptoms have progressed to acute encephalopathy with vomiting, ataxia, and variable consciousness. The family recently moved, and they are in the process of restoring the interior of their home. (**SELECT 1 TREATMENT**)

**72.** After a fight with her boyfriend, a 16-year-old girl took “some pills.” At presentation she is alert and complains of emesis, diaphoresis, and malaise. Her initial liver function tests, obtained about 12 h postingestion, are elevated. Repeat levels at 24 h show markedly elevated AST and ALT, along with abnormal coagulation studies and an elevated bilirubin. **(SELECT 1 TREATMENT)**

**73.** You are called to the delivery room. A newborn infant seems lethargic and has poor tone with only marginal respiratory effort, but his heart rate is above 100 beats per min. The mother had an uncomplicated pregnancy, and delivery was uncomplicated and vaginal 10 min after spontaneous rupture of membranes. The mother received only pain medications while in labor. **(SELECT 1 TREATMENT)**

**74.** A 4-year-old girl comes into the emergency room after eating a bottle full of small, chewable pills she found while at her grandfather’s house. She has an increased respiratory rate, elevated temperature, and vomiting, and is disoriented. She is intermittently complaining of “a bell is ringing” in her ears. She has a metabolic acidosis on an arterial blood gas. **(SELECT 1 TREATMENT)**

**75.** After helping his father in the yard, a 14-year-old boy complains of weakness and feels like his muscles are twitching. He begins to drool, and then collapses in a generalized tonic clonic seizure. Upon the arrival of EMS, his heart rate is found to be 40 beats per min and his pupils are pinpoint. **(SELECT 1 TREATMENT)**

### Items 76–78

For the most likely toxic substance involved in the cases below, match the appropriate treatment.

- a. Deferoxamine mesylate
- b. Diphenhydramine (Benadryl)
- c. Acetazolamide and sodium bicarbonate
- d. Ethanol
- e. Dimercaprol (BAL)

**76.** A 14-year-old male presents after taking a “happy pill” that his friend gave him. He is alert and oriented, but complains of a muscle spasm in his neck, making his head lean on his right shoulder. You also notice he is arching his back in an unusual manner. **(SELECT 1 TREATMENT)**

**77.** A 2-year-old boy found a bottle of his mother’s prenatal vitamins and consumed the majority of them. He now has hematemesis and abdominal pain. He is febrile, and labs reveal a leukocytosis and hyperglycemia. **(SELECT 1 TREATMENT)**

**78.** A 17-year-old is brought into the emergency department by his friends at about 10:00 A.M. They were at a party the night before and drank some “homemade” alcohol. He is disoriented and confused, and has an anion-gap acidosis. He begins to have seizures as he is placed on a stretcher. **(SELECT 1 TREATMENT)**

### **Items 79–83**

Excess vitamin intake has been shown to have deleterious effects. Match the vitamin with the toxic effect.

- a. Kidney stones
- b. Sensory neuropathy
- c. Edema
- d. Hyperostosis
- e. Calcification of the heart
- f. Decreased cerebrospinal fluid pressure
- g. Erythema of skin

**79.** Vitamin A **(SELECT 1 EFFECT)**

**80.** Nicotinic acid **(SELECT 1 EFFECT)**

**81.** Vitamin C **(SELECT 1 EFFECT)**

**82.** Vitamin D **(SELECT 1 EFFECT)**

**83.** Pyridoxine **(SELECT 1 EFFECT)**

**Items 84–88**

The normal development of the fetus can be adversely affected by exposure to a number of environmental factors, including infectious agents, physical agents, chemical agents, and maternal metabolic and genetic agents. Match each maternal history of teratogen exposure with the most likely clinical presentation.

- a. Small palpebral fissures, ptosis, midfacial hypoplasia, smooth philtrum
- b. Hypoplasia of distal phalanges, small nails
- c. Bilateral microtia or anotia
- d. Spina bifida
- e. Heart block
- f. Sacral agenesis
- g. Aniridia
- h. Hemangiomas

**84.** A multiparous mother has a 1-h glucose tolerance test value of >190 mg/dL and a 3-h glucose tolerance test of >150 mg/dL. **(SELECT 1 PRESENTATION)**

**85.** A pregnant mother is taking phenytoin for a long-standing generalized seizure disorder. **(SELECT 1 PRESENTATION)**

**86.** A 15-year-old mother is taking valproate for absence seizures. **(SELECT 1 PRESENTATION)**

**87.** A primiparous mother reports drinking two beers a day during the pregnancy. **(SELECT 1 PRESENTATION)**

**88.** A 23-year-old pregnant woman has a malar rash that worsens with sun exposure, oral ulcerations, and joint pain. Her obstetrician has told her she has proteinuria and anemia, but several other labs are pending. **(SELECT 1 PRESENTATION)**

**Items 89–92**

Match each common skin condition with the most appropriate therapy.

- a. Mild cleansing cream, topical moisturizers, and topical steroids
- b. Topical or oral antibacterial agents
- c. Oral antihistamines alone
- d. Brief application of liquid nitrogen or cantharidin 0.9%, or extrusion of lesion's central plug with curette
- e. Permethrin 5% cream (Elimite)
- f. Topical acyclovir
- g. Topical antifungal agents

**89.** In the late summer, a 5-year-old boy presents with several sores on his arms and legs. The lesions have a honey-colored crust, and there is minimal surrounding erythema. The lesions seem to have spread by the child scratching. **(SELECT 1 TREATMENT)**

**90.** A 13-year-old girl complains of intense itching of her skin for 2 weeks. You find papules and pustules with a few vesicles distributed over the torso and extremities, but the lesions seem to spare the face. She has areas of lichenification from repeated scratching. The outbreak seems worse on the hands, and you identify raised, linear lesions in the webbing of her hands. **(SELECT 1 TREATMENT)**

**91.** A mother complains of “moles” on her child's face for 6 months. You find four flesh-colored, papular lesions, the largest being about a centimeter in diameter. The lesions have a central umbilication. The mother reports no scratching of the lesions. **(SELECT 1 TREATMENT)**

**92.** A 10-year-old boy presents with a dry, scaly rash in his antecubital and popliteal fossas, much of it with areas of lichenification. His father notes that this rash always seems to get worse in the winter months. **(SELECT 1 TREATMENT)**

**Items 93–95**

Heart failure is possible at any age. Match each clinical condition with the most likely diagnosis.

- a. Glomerulonephritis
- b. Severe anemia
- c. Heart block
- d. Ventricular septal defect
- e. Arteriovenous malformation
- f. Coarctation of the aorta

**93.** A 6-month-old child has a loud systolic murmur at the left lower sternal border. **(SELECT 1 DIAGNOSIS)**

**94.** A 14-year-old child has headache, hypertension, edema, and a change in urine output and color. **(SELECT 1 DIAGNOSIS)**

**95.** A 3-day-old infant was born to a mother with active systemic lupus erythematosus (SLE). **(SELECT 1 DIAGNOSIS)**

**Items 96–102**

The Committee on Nutrition of the American Academy of Pediatrics has concluded that children on a normal diet do not need vitamin supplements. There are, however, some clinical situations in which special needs do occur. Match each situation with the appropriate supplement.

- a. All fat-soluble vitamins
- b. Pyridoxine
- c. Vitamin A
- d. Vitamin D
- e. Vitamin K
- f. Folate

**96.** Isoniazid therapy in a pregnant teenager **(SELECT 1 SUPPLEMENT)**

**97.** Administration of phenytoin **(SELECT 1 SUPPLEMENT)**

**98.** Measles in developing countries **(SELECT 1 SUPPLEMENT)**

**99.** Liver disease (**SELECT 1 SUPPLEMENT**)

**100.** Breast-fed infant in Alaska (**SELECT 1 SUPPLEMENT**)

**101.** Sickle cell disease (**SELECT 1 SUPPLEMENT**)

**102.** One-day-old newborn (**SELECT 1 SUPPLEMENT**)

**Items 103–107**

Match each clinical condition with the most appropriate diagnostic laboratory test.

- a. Erythrocyte sedimentation rate
- b. Serum immunoglobulin levels
- c. Nitroblue tetrazolium (NBT) test
- d. CH<sub>50</sub> assay
- e. CBC demonstrating Howell-Jolly bodies
- f. Platelet count
- g. Intradermal skin test using *Candida albicans*

**103.** A 1-year-old boy has been admitted three times in the past with abscess formation requiring incision and drainage. He is now admitted for surgical drainage of a hepatic abscess identified on ultrasound. (**SELECT 1 TEST**)

**104.** A 5-month-old infant is admitted with severe varicella infection. The lesions cover the infant's entire body, and the infant is beginning to show symptoms of respiratory distress. Past medical history is significant for a history of atopic dermatitis. The family also notes frequent epistaxis; the last episode required nasal packing in the emergency department. (**SELECT 1 TEST**)

**105.** A 3-year-old has had repeated episodes of sinusitis and otitis media. He was recently admitted for osteomyelitis of his femur with *Staphylococcus aureus*. The family notes that while his first four or five months of life were normal, he has been persistently ill with multiple infections in the ensuing months. The mother notes that her brother had similar problems with infections and died at the age of 3 years from a "lung infection." Physical examination is significant for the absence of lymph nodes and tonsillar tissue. (**SELECT 1 TEST**)

**106.** A general practitioner refers to you for evaluation a 3-year-old boy with frequent infections. You note the child to have a loud systolic murmur, posteriorly rotated ears that are small and low-set, down-slanting and widely spaced eyes, a small jaw, and an upturned nose. At birth the child spent 2 weeks in the nursery for “low calcium” and seizures, and still receives calcium supplementation, but the mother does not know why. You would like to order a rapid diagnostic test for this child. **(SELECT 1 TEST)**

**107.** A 2-year-old girl has had two episodes of *Neisseria meningitidis* septicemia and is now admitted for *Streptococcus pneumoniae* septicemia. **(SELECT 1 TEST)**

# General Pediatrics

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## Answers

**1. The answer is e.** (*Behrman, 16/e, p 1893. McMillan, 3/e, p 1963. Rudolph, 21/e, p 2366.*) Bell palsy is an acute, unilateral facial nerve palsy that begins about 2 weeks after a viral infection. The exact pathophysiology is unknown, but it is thought to be immune or allergic. On the affected side, the upper and lower face are typically paretic, the mouth droops, and the patient cannot close the eye. Treatment consists of maintaining moisture to the affected eye (especially at night) to prevent keratitis. Complete, spontaneous resolution occurs in about 85% of cases, 10% of cases have mild residual disease, and about 5% of cases do not resolve.

**2. The answer is b.** (*Behrman, 16/e, p 35. McMillan, 3/e, pp 756–761. Rudolph, 21/e, p 14.*) By 8 weeks of age, infants who are developing normally should be able to smile and coo when smiled at or talked to. By 3 months of age, infants should be able to follow a moving toy not only from side to side but also in the vertical plane. When placed on his or her abdomen, a normal 3-month-old infant can raise his or her face 45° to 90° from the horizontal. Not until 6 to 8 months of age should an infant be able to maintain a seated position.

**3. The answer is c.** (*Behrman, 16/e, p 38. McMillan, 3/e, pp 756–761. Rudolph, 21/e, p 440.*) By 3 years of age, children become quite skilled in many areas. Most can say many words and speak in sentences. They can help dress and undress themselves, with the exception of shoelaces and sometimes buttons. They can ride a tricycle and can alternate feet when climbing stairs, although they still place both feet on each step when going down stairs. They can identify at least one color by name, know their age and gender, but have not progressed beyond copying a circle and a crude cross. Only at 4 to 5 years of age can a child copy a square.

**4. The answer is a.** (*Behrman, 16/e, pp 704–709. McMillan, 3/e, pp 2156–2160. Rudolph, 21/e, pp 836–839.*) Pauciarticular rheumatoid arthritis asymmetrically involves large joints, especially the knee, and often has no other symptoms. The major morbidity of pauciarticular rheumatoid arthri-

tis is blindness. About 20% of girls who have the monoarthritic or pauci-articular form of juvenile rheumatoid arthritis have iridocyclitis as their only significant systemic manifestation. Because this eye disorder can require treatment with local or systemic steroids and develop without signs or symptoms, it is recommended that all children with this form of arthritis have frequent slit-lamp eye examinations.

**5. The answer is b.** (*Behrman, 16/e, p 2031. McMillan, 3/e, pp 378–379, 418–420. Rudolph, 21/e, pp 1225–1226.*) Also known as Ritter disease, staphylococcal scalded skin disease is seen most commonly in children less than 5 years of age. The rash is preceded by fever, irritability, erythema, and extraordinary tenderness of the skin. Circumoral erythema; crusting of the eyes, mouth, and nose; and blisters on the skin can develop. Intraoral mucosal surfaces are not affected. Peeling of the epidermis in response to mild shearing forces (Nikolsky sign) leaves the patient susceptible to problems similar to those of a burn injury, including infection and fluid and electrolyte imbalance. Cultures of the bullae are negative, but the source site is often positive. Treatment includes antibiotics (to cover resistant *Staphylococcus aureus*) and localized skin care. Recovery without scarring can be expected.

**6. The answer is d.** (*McMillan, 3/e, pp 38–39.*) The probability given is an estimation of the odds that the observed differences could have occurred by chance alone. The interpretation of these results depends on an assessment of factors such as study design, the size of the sample, the type of controls used, the severity of the disease, the side effects, and the importance of the treatment. The tendency for negative results to remain unpublished should also be kept in mind.

**7–8. The answers are 7-d, 8-c.** (*Behrman, 16/e, pp 1424–1428. McMillan, 3/e, pp 1405–1417. Rudolph, 21/e, pp 909–913.*) The presentation of infective endocarditis can be quite variable, ranging from prolonged fever with few other symptoms to an acute and severe course with early toxicity. A high index of suspicion is necessary to make the diagnosis quickly. Identification of the causative organism (frequently *Streptococcus* sp. or *Staphylococcus* sp.) through multiple blood cultures is imperative for appropriate treatment. Echocardiography may identify valvular vegetations and can be predictive of impending embolic events. Treatment usually consists of 4 to

6 weeks of appropriate antimicrobial therapy. Bed rest should be instituted only for heart failure. Antimicrobial prophylaxis prior to and after dental cleaning is indicated.

**9. The answer is a.** (*Behrman, 16/e, pp 728–729. McMillan, 3/e, pp 1490–1491, 2176–2179. Rudolph, 21/e, pp 842–844.*) The clinical presentation described supports the diagnosis of anaphylactoid purpura, a generalized, acute vasculitis of unknown cause involving small blood vessels. In this condition, the skin lesion, which is classic in character and distribution, is often accompanied by arthritis, usually of the large joints, and by gastrointestinal symptoms. Colicky abdominal pain, vomiting, and melena are common. Renal involvement occurs in a significant number of patients and is potentially the most serious manifestation of the disease. Although most children with this complication recover, some will develop chronic nephritis. Laboratory studies are not diagnostic. Serum complement and IgA levels can be normal or elevated. Coagulation studies and platelets are normal.

**10. The answer is e.** (*Behrman, 16/e, pp 110–114. McMillan, 3/e, pp 507–524. Rudolph, 21/e, pp 463–469.*) The x-ray showing a fracture (or an x-ray showing multiple fractures in various stages of healing) indicates trauma. This information should be reported to the medical examiner and appropriate social agencies, including the police, so that an investigation can be started and other children in the home or under the care of the same babysitter can be protected. Although an autopsy (and death-scene investigation) should be done in every such case, there sometimes develops a tendency for medical examiners to diagnose SIDS without an autopsy, particularly if the parents object to one, unless further information is provided by the emergency room staff, as in this case.

**11. The answer is c.** (*Behrman 16/e, pp 1126–1127. McMillan, 3/e, pp 618–619. Rudolph, 21/e, p 1395.*) The induction of emesis with syrup of ipecac is a very effective method of removing swallowed poisons. It has been used safely in children as young as 6 months of age. Ipecac should not be given to a patient who has ingested a corrosive poison, such as strong acid or alkali, because of the possibility of gastric perforation and further necrosis of the esophagus. When vomiting imposes a risk of aspiration, as in the case of an obtunded or comatose patient or in the case of

impending seizures precipitated by the ingested drug, syrup of ipecac is contraindicated. Hydrocarbons such as mineral oil have a low viscosity and surface tension; the latter property accounts for the spreading tendency of the product and the potential for aspiration. Very minute amounts of aspirated hydrocarbon can cause a necrotizing pneumonia. Syrup of ipecac should be used only when the primary toxicity of the hydrocarbon is systemic, as, for example, with carbon tetrachloride or benzene. Administration of activated charcoal alone without prior gut emptying is being used for certain ingestions. Proper instruction of parents regarding toxic ingestions should be provided during well-child visits. Efforts should be aimed at prevention, with age-appropriate anticipatory guidance regarding environmental hazards.

**12. The answer is b.** (*Behrman, 16/e, pp 790–792. McMillan, 3/e, pp 587–589.*) Human bites can pose a significant problem. They can become infected with oropharyngeal bacteria, including *Staphylococcus aureus*, viridans streptococci, *Bacteroides spp.*, and anaerobes. A patient with an infected human bite of the hand requires hospitalization for appropriate drainage procedures, Gram stain and culture of the exudate, vigorous cleaning, debridement, and appropriate antibiotics. The wound should be left open and allowed to heal by secondary intention.

**13–14. The answers are 13-b, 14-e.** (*Behrman, 16/e, pp 2001–2007. McMillan, 3/e, pp 689, 720. Rudolph, 21/e, p 1181.*) Pityriasis rosea is a benign condition that usually presents with a herald patch, a single round or oval lesion appearing anywhere on the body. Usually about 5 to 10 days after the appearance of the herald patch, a more diffuse rash involving the upper extremities and trunk appears. These lesions are oval or round, slightly raised, and pink to brown in color. The lesion is covered in a fine scale with some central clearing possible. The rash can appear in the Christmas tree pattern on the back, identified by the aligning of the long axis of the lesions with the cutaneous cleavage lines. The rash lasts 2 to 12 weeks and can be pruritic. This rash is commonly mistaken for tinea corporis, and the consideration of secondary syphilis is important. Treatment is usually unnecessary but can consist of topical emollients and oral antihistamines as needed. More uncommonly, topical steroids can be helpful if the itching is severe.

Lichen planus is rare in children. It is intensely pruritic, and additional lesions can be induced with scratching. The lesion is commonly found on the flexor surfaces of the wrists, forearms, inner thighs, and occasionally on the oral mucosa.

Seborrheic dermatitis can begin anytime during life; it frequently presents as cradle cap in the newborn period. This rash is commonly greasy, scaly, and erythematous and, in smaller children, involves the face, neck, axilla, and diaper area. In older children, the rash can be localized to the scalp and intertriginous areas. Pruritus can be marked.

Contact dermatitis is characterized by redness, weeping, and oozing of the affected skin. The pattern of distribution can be helpful in identification of the offending agent. The rash can be pruritic; removal of the causative agent and use of topical emollients or steroids is curative.

Psoriasis consists of red papules that coalesce to form plaques with sharp edges. A thick, silvery scale develops on the surface and leaves a drop of blood upon its removal (Auspitz sign). Additional lesions develop upon scratching older lesions. Commonly affected sites include scalp, knees, elbows, umbilicus, and genitalia.

**15–16. The answers are 15-e, 16-e.** (*Behrman, 16/e, pp 1829–1830. Rudolph, 21/e, p 2274.*) The child in this question most likely has breath-holding spells. Two forms exist. Cyanotic spells consist of the symptoms outlined and are predictable upon upsetting or scolding the child. They are rare before 6 months of age, peak at about 2 years of age, and resolve by about 5 years of age. Avoidance of reinforcing this behavior is the treatment of choice. Pallid breath-holding spells are less common and are usually caused by a painful experience (such as a fall). With these events, the child will stop breathing, lose consciousness, become pale and hypotonic, and may have a brief tonic episode. These, too, resolve spontaneously. Again, avoidance of reinforcing behavior is indicated.

**17. The answer is b.** (*Behrman, 16/e, pp 2044–2050. McMillan, 3/e, pp 684, 730. Rudolph, 21/e, pp 1169, 1210.*) The pictured infant has a classic case of neonatal acne, which peaks at 2 to 4 weeks of age. The condition results from maternal hormone transmission. It resolves in a few weeks to months, and occasionally is severe enough to require treatment with agents such as 2.5% benzoyl peroxide.

**18. The answer is a.** (Behrman, 16/e, pp 2152–2153, 2162. McMillan, 3/e, p 621. Rudolph, 21/e, pp 373–374.) When the clinical signs of constricted pupils, bradycardia, and muscle fasciculations are associated with the sudden onset of neurologic symptoms, progressive respiratory distress, diaphoresis, diarrhea, and overabundant salivation, a diagnosis of organophosphate poisoning should be suspected. Intake of organophosphate agents can occur by ingestion, inhalation, or absorption through skin or mucosa. Organophosphates inhibit carboxylic esterase enzymes, including acetylcholinesterase and pseudocholinesterase; toxicity depends primarily on the inactivation or inhibition of acetylcholinesterase.

Treatment consists of gastric lavage, if the poison has been ingested, or decontamination of the skin, if exposure has been through contact. Maintenance of adequate ventilation and fluid and electrolyte balance also is indicated. All symptomatic children should receive atropine and, if severely affected, cholinesterase-reactivating oximes as well. Cholinesterase-reactivating oximes quickly restore consciousness by inhibiting muscarine- and nicotine-like synaptic actions of acetylcholine. Cholinesterase-reactivating oximes include pralidoxime chloride or obidoxime.

**19. The answer is b.** (Behrman, 16/e, pp 2044–2046. McMillan, 3/e, pp 716–717. Rudolph, 21/e, pp 1153–1154, 1233–1234.) Scabies is caused by the mite *Sarcoptes scabiei* var. *hominis*. Most older children and adults present with intensely pruritic and threadlike burrows in the interdigital areas, groin, elbows, and ankles; the palms, soles, face, and head are spared. Infants, however, usually present with bullae and pustules, and the areas spared in adults are often involved in infants. The clinical manifestations closely resemble those of atopic dermatitis. Because of the potential neurotoxic effect to infants of gamma benzene hexachloride through percutaneous absorption, an excellent alternative—5% permethrin cream (Elimite)—is available and is more often recommended.

**20. The answer is c.** (Behrman, 16/e, pp 810–816. McMillan, 3/e, pp 421–427. Rudolph, 21/e, pp 999–1001.) The rapid onset of the symptoms, the low WBC count with left shift, and the depicted chest x-ray findings are typical of a patient with group B streptococcus pneumonia. Appropriate management would include rapid recognition of symptoms, respiratory support, and rapid institution of appropriate antibiotics. Despite these

measures, mortality from this infection is not uncommon. The other infectious causes listed do not present so early, and the noninfectious causes listed do not cause elevations in the band count.

**21. The answer is a.** (*Behrman, 16/e, pp 2075–2076, 2080–2082, 2106. McMillan, 3/e, pp 749, 2109–2110. Rudolph, 21/e, pp 2432, 2437–2438.*) This history is typical of Osgood-Schlatter disease. Microfractures in the area of the insertion of the patellar tendon into the tibial tubercle are common in athletic adolescents. Swelling, tenderness, and an increase in size of the tibial tuberosity are found. Radiographs can be necessary to rule out other conditions. Treatment consists of rest.

Legg-Calvé-Perthes disease is avascular necrosis of the femoral head. This condition usually produces mild or intermittent pain in the anterior thigh but can also present as a painless limp.

Gonococcal arthritis, although common in this age range, is uncommon in this anatomic site. More significant systemic signs and symptoms, including chills, fever, migratory polyarthralgias, and rash, are commonly seen.

Slipped capital femoral epiphysis is usually seen in a younger, more obese child (mean age about 10 years) or in a thinner, older child who has just undergone a rapid growth spurt. Pain upon movement of the hip is diagnostic.

Popliteal cysts are found on the posterior aspect of the knee.

**22. The answer is e.** (*Behrman, 16/e, pp 1904–1907. McMillan, 3/e, pp 662, 672–673. Rudolph, 21/e, pp 2402–2404, 2353.*) To prevent monocular blindness and to ensure the development of normal binocular vision, early recognition and treatment of strabismus are essential. Infants can be screened for strabismus by observing the location of a light reflection in the pupils when the patient fixes on a light source. Normally, it should be in the center or just nasal of the center in each pupil. Persistence of a transient or fixed deviation of an eye beyond 4 months of age requires referral to an ophthalmologist. Another method of testing for strabismus is the “cover” or “cover-uncover” test, using the principle that children with strabismus will use the “good” eye for fixation. By covering the good eye, the other eye will deviate. The aim of treatment is to prevent loss of central vision from foveal suppression of a confusing image in the deviating eye. This is accomplished by surgery, eyeglasses, or patching of the normal eye. The prognosis for

normal vision if diagnosis is delayed beyond 6 years of age is guarded. Routine vision and strabismus screening is essential at age 3 to 4 years.

**23. The answer is b.** (*Behrman, 16/e, pp 231, 1279–1280. McMillan, 3/e, pp 500, 597–598, Rudolph, 21/e, pp 37–39, 1935–1936.*) Childhood fatalities from asphyxiation are concentrated among children younger than 4. The most common cause of asphyxiation is choking on foods of the size, shape, and consistency to occlude the upper airway completely. Foods that are particularly dangerous include round, hard candies, hot dogs, whole grapes, and pieces of raw apple or carrot. Children are also at risk of aspirating small objects such as round, pliable toys, uninflated balloons, makeshift pacifiers, bottle nipples, and plastic-lined disposable diapers. Water temperature should be set to 49°C (120°F); this will minimize the risk of accidental scald burns. While attractive, pillows are not allowed in an infant's crib, because infants with poor head control may asphyxiate in them. The current recommendation of the American Academy of Pediatrics is to place infants to sleep on their back. This recommendation has the apparent result of decreasing the risk of SIDS. Infants and young children should never be left alone in the bathtub, regardless of water depth; a child may drown in as little as 1 to 2 in. of water.

**24. The answer is c.** (*Behrman, 16/e, pp 35–37. McMillan, 3/e, pp 756–761. Rudolph, 21/e, p 14.*) At 6 to 6½ months of age, infants will be able to sit alone, leaning forward to support themselves with arms extended, in the so-called tripod position. They can reach for an object by changing the orientation of the torso. They can purposefully roll from a prone to a supine as well as from a supine to a prone position. By 12 months, they can grasp a pellet between thumb and forefinger without ulnar support. Motor development occurs in a cephalocaudal and central-to-peripheral direction. Therefore, truncal control precedes arm control, which precedes finger dexterity.

**25–26. The answers are 25-d, 26-d.** (*Behrman, 16/e, pp 680–684. McMillan, 3/e, pp 704–706. Rudolph, 21/e, pp 1177–1179.*) Eczema is a chronic dermatitis that occurs in a population with a strong personal or family history of atopy. The skin presents initially as an erythematous, papulovesicular, weeping eruption, which progresses over time to a scaly,

lichenified dermatitis. From 3 months to 2 years of age, the rash is prominent on the cheeks, wrists, scalp, postauricular areas, and arms and legs. In the young child 2 to 12 years of age, mainly the extensor surfaces of arms, legs, and neck are involved. Pruritus is a predominant feature, and scratching leads to excoriation, secondary infection, and lichenification of the skin. The rash has a chronic and relapsing course, and treatment is determined by the major clinical features. Cutaneous irritants (bathing in hot water, scrubbing vigorously with soap, wearing wool or synthetic clothing) should be avoided, and maximal skin hydration with emollients is essential. Topical moisturizers and steroids are the mainstays of therapy for atopic dermatitis. The use of antihistamines can provide additional relief from pruritus.

**27. The answer is a.** (*Behrman, 16/e, pp 2046–2047. McMillan, 3/e, p 716. Rudolph, 21/e, pp 1155–1156.*) The photo and history are consistent with head lice (*Pediculus capitis*). All the treatments outlined for this patient are appropriate for the treatment of lice except for use of 1% lindane in small infants. The treatment of choice for these small children is permethrin 1% cream rinse (Nix).

**28. The answer is b.** (*Behrman, 16/e, pp 1025–1028. McMillan, 3/e, p 870. Rudolph, 21/e, pp 1046–1048.*) This child is presenting with failure to thrive (FTT), and the differential diagnosis of this problem is extensive. While any of the answers provided may have a place in an FTT evaluation, the best single recommendation in this case would be to evaluate for HIV. With a mother in jail for unknown reasons (possibly for prostitution, drugs, or other high-risk activities), this child has increased risk for congenital HIV. In addition, the presenting symptoms are most consistent with HIV.

**29. The answer is d.** (*Behrman, 16/e, pp 653–654, 890–891, 1859, 2107, 2156–2159. McMillan, 3/e, pp 615–616, 632, 1033, 1511–1512, 1689–1692. Rudolph, 21/e, pp 368–371, 818–819, 953, 2209–2210, 2246–2247.*) Unfortunately, the seriousness of the illness in a child such as this will often be unrecognized, and the patient will be diagnosed as having an incipient nondisease and treated symptomatically. Although it is true that many patients with nonspecific complaints have trivial diseases that resolve spontaneously, the patient presented here has a 3-week history with poten-

tially grave implications. Diagnoses other than food allergy should be considered foremost.

**30. The answer is a.** (*Behrman, 16/e, pp 1940–1947. McMillan, 3/e, pp 770–771, 2255. Rudolph, 21/e, pp 21–22.*) The recognition of hearing impairment in a neonate is difficult. The delay in diagnosis that often occurs can seriously jeopardize normal speech development and learning. To aid in early diagnosis, the behavioral or electrophysiologic responses (or both) of infants who are at higher-than-average risk of hearing impairment are monitored closely during the early months of life. Neonatal factors that confer high risk include craniofacial defects (including abnormal ear structure), neonatal asphyxia, bacterial meningitis, congenital infections, significant hyperbilirubinemia, use of ototoxic medications for more than 5 days, mechanical ventilation for more than 10 days, stigmata of syndromes causing hearing loss, birth weight less than 1500 g, and family history of deafness. A pattern of malformation that can include hypoplasia of digits, nails, and midface; growth retardation; cardiac defects; and mental deficiency is associated with the maternal use of phenytoin during pregnancy. Hearing loss is not a manifestation of the fetal hydantoin syndrome. The American Academy of Pediatrics currently recommends universal screening for hearing loss for all newborn infants.

**31–32. The answers are 31-c, 32-b.** (*Behrman, 16/e, pp 215–217. McMillan, 3/e, pp 67–72. Rudolph, 20/e, pp 1643–1647.*) Rapid expansion of the vascular space is needed. Appropriate intravenous fluids include Ringer's lactate and normal saline. Albumin, plasma, and blood offer no significant advantages over the cheaper and more available Ringer's lactate or normal saline.

If this patient is in shock, it is vital to restore blood volume quickly, thereby improving tissue perfusion and shifting anaerobic toward aerobic metabolism. The restoration of vascular volume would also improve functioning of the intestines so that diarrhea will abate and would improve circulation to the kidney so that renal function would be restored. A volume of 20 mL/kg is about one-quarter the blood volume of the patient; therefore, it should be given at a rate that will not produce pulmonary edema. An emergency phase lasting 1 to 2 h or less has worked well in practice. The dextrose fluids listed are various forms of maintenance fluids.

**33. The answer is b.** (Behrman, 16/e, pp 1660–1663.) A nonspecific vulvovaginitis is common in this age group, usually caused by chemical irritants such as bubble baths or by poor hygiene. Mothers should be counseled to use only cotton underwear for young children, stop bubble baths (or at least splash fresh, clean water in the vaginal area at the end of a bath), and reemphasize wiping front to back after bowel movements. Vaginal foreign bodies in young girls are usually either toilet paper or stool, and are accompanied by a foul odor and discharge that is sometimes bloody. Removal of vaginal foreign bodies is frequently under general anesthesia in the operating room. While child sexual abuse is always a possibility, there is no verbal or physical evidence that this is a problem in this child. Gonorrhea would go along with abuse and is usually accompanied by a mild discharge in the prepubertal child.

**34. The answer is a.** (Behrman, 16/e, pp 777–780. McMillan, 3/e, pp 416–418. Rudolph, 21/e, pp 907–908.) Examination of the joint fluid is the key to diagnosis. The joint tap will reveal cloudy fluid containing a predominance of polymorphonuclear leukocytes. Organisms are readily seen on Gram stain examination; cultures of joint fluid and blood are usually positive. X-ray reveals a widened joint space. Finding pus in the joint indicates the need for immediate surgical drainage and prompt institution of IV antibiotic therapy to avoid serious damage to the joint and permanent loss of function. The most common organism found to cause septic arthritis is *Staphylococcus aureus*. Since immunization against *Haemophilus influenzae* type B has become an established practice, invasive disease such as septic arthritis caused by this organism is rarely seen. In sexually active adolescents, *Neisseria gonorrhoeae* is a common cause of septic arthritis.

**35. The answer is b.** (Behrman, 16/e, pp 878–880. McMillan, 3/e, pp 486–487, 940. Rudolph, 21/e, pp 1007–1008.) The patient should receive a booster immunization with adult Td, which should be given every 10 years to maintain immunity against both diphtheria and tetanus. He does not need passive immunization with tetanus immune globulin because the wound is superficial and there appears to be little or no risk of tetanus. As of 1993, pertussis immunization is discontinued at age 7 years.

**36. The answer is c.** (Behrman, 16/e, pp 790–792. McMillan, 3/e, p 587.) Mammalian bites should be promptly and thoroughly scrubbed with soap

and water and debrided. The decision to suture depends on the location, age, and nature of the wound. Antibiotic prophylaxis should be considered in cat, human, or monkey bites. Only 4% of dog bites become infected (and do not necessarily need antibiotic prophylaxis), compared with 35% of cat bites and 50% of monkey bites (which require antibiotics in most cases). Cat bites are usually deep punctures. Human bites almost invariably become infected. The etiologies of these infections are polymicrobial. *Pasteurella multocida* is a common organism in infected cat and dog bites. Infected human bites tend to have positive cultures for *Streptococcus viridans*, *Staphylococcus aureus*, and *Bacteroides* spp. Treatment with oral amoxicillin-clavulanate or erythromycin is recommended. Antibiotic prophylaxis is recommended for any bite sustained by an infant, a diabetic, or an immunocompromised patient because of the higher risk of infection in these persons. Since the child is fully immunized, tetanus boosters are not required. Similarly, as the dog was provoked and was fully immunized, rabies should not be a concern.

**37. The answer is b.** (Behrman, 16/e, p 91. McMillan, 3/e, p 622. Rudolph, 21/e, pp 364–365.) Poisoning with tricyclic antidepressants is a leading reason for admissions to pediatric intensive care units and a leading cause of fatal drug overdose in adolescents. These preparations produce a variety of pharmacologic effects, including inhibition of muscarinic/cholinergic receptors, blockade of norepinephrine and serotonin uptake, and depression of sodium channels responsible for cardiac cell membrane depolarization. The toxic-to-therapeutic ratio is low. Young children often ingest poisons and drugs during times of household disruption. Visitors' handbags are a great temptation for the inquisitive toddler.

**38. The answer is d.** (Behrman, 16/e, pp 2156–2159. McMillan, 3/e, pp 632–633. Rudolph, 21/e, pp 23–24.) Because of evidence that impairment of cognitive function can occur at blood lead levels previously thought to be safe, the toxic concentration of lead in whole blood was revised downward in 1991 from 25  $\mu\text{g}/\text{dL}$  to 10  $\mu\text{g}/\text{dL}$ . The blood erythrocyte protoporphyrin concentration is not elevated in low-level poisoning; therefore, testing for erythrocyte protoporphyrin is no longer a valid screen. The definitive screen is the blood lead level. Venipuncture avoids the risk of environmental contamination with lead that is more likely with finger sticks. Most lead poisoning is clinically inapparent. A careful history will help to identify

sources of lead in the environment. Neither the history nor the anemia that accompanies severe lead poisoning, however, is an appropriate means of screening for lead poisoning. Some authorities state that screening of the environment and removal of sources of lead is preferable to screening of children.

**39. The answer is d.** (*Behrman, 16/e, pp 771–773, 1083–1084. McMillan, 3/e, pp 447–448, 1724–1725. Rudolph, 21/e, pp 1497–1498.*) In the United States, the hepatitis B virus is transmitted mainly by exposure to blood and blood products, through sexual contact, and from mothers to newborns. Close family contact has also been implicated as a factor in transmission. About one-third of cases have no identifiable risk factor. Although the infection usually presents as viral hepatitis, its full impact may not be realized until years later. Chronic active hepatitis is a complication characterized by unresolving inflammation, which can eventually lead to necrosis and fibrosis of the liver and possible progression to cirrhosis and liver failure. Hepatocellular carcinoma is another long-term complication of hepatitis B infection. Acute fulminant hepatitis causes massive destruction of hepatic parenchyma, but is fortunately a rare complication of infection with the virus. Hydrops of the gallbladder is not related to hepatitis B infection. It is usually idiopathic but can be a complication of Kawasaki disease, among other diseases.

**40. The answer is a.** (*Behrman, 16/e, p 2142. McMillan, 3/e, pp 597, 602. Rudolph, 21/e, p 1936.*) An increasing number of studies in infants are pointing to the prone (face down) sleeping position as a risk factor for sudden infant death syndrome (SIDS). A higher risk of SIDS in the prone position has been noted when the infant sleeps on a soft, porous mattress or in an overheated room, is swaddled, or has recently been ill. The American Academy of Pediatrics recommends that healthy infants be positioned on their side or back when being put down for sleep. (The prone position is still recommended for infants with certain medical conditions.) The rate of SIDS has declined in areas where the change from prone to supine (face up) sleeping positions has been effected.

**41. The answer is d.** (*Behrman, 16/e, p 331. McMillan, 3/e, p 2232. Rudolph, 21/e, p 720.*) The physical features associated with the fragile X syndrome

become more obvious after puberty. They include a long face, large ears, prominent jaw, macroorchidism, hypotonia, repetitive speech, gaze avoidance, and hand flapping. Even in the absence of physical findings, boys of all ages with developmental delay, autism, and abnormal temperament of unknown cause probably should be tested for the fragile X syndrome.

The genetics of the fragile X syndrome are unique. Most males who carry the fragile X mutation are mentally impaired and show the clinical phenotype; however, 20% of males who inherit the genetic mutation are normal in intelligence and physical appearance. They are also cytogenetically normal in that the fragile site on their X chromosome is not seen by the karyotyping method. These normal transmitting males (NTMs) transmit the fragile X mental retardation (FMR-1) gene to all of their daughters and often have severely affected grandchildren. It is thought that a premutation carried by these NTMs must go through oogenesis in their daughters to become a full mutation. Daughters of NTMs are usually normal but are obligate carriers of the FMR-1 gene. Daughters who inherit the gene from the mother are mentally retarded about one-third of the time. Because both males and females can be affected, the fragile X syndrome is best described as a dominant X-linked disorder with reduced penetrance in females. Cytogenetic testing for the fragile site on the X chromosome is an unsatisfactory method of identifying persons with this mutation for the following reasons: (1) not all cells in the affected males show the fragile site, (2) NTMs have no fragile X site demonstrable by this method, and (3) a significant percentage of carrier females are cytogenetically normal. In 1991, the FMR-1 gene was cloned. Direct DNA analysis is now available for diagnosis of phenotypically affected persons as well as suspected carriers of fragile X; it can also be used for prenatal testing.

**42. The answer is c.** (*McMillan, 3/e, p 77, Rudolph, 21/e, pp 1650–1651.*) The differential diagnosis of seizure activity is extensive, and a detailed history is essential. While knowing the occupation of the mother and the developmental status of the children is important, the most helpful historical detail for acute management of this patient is the dilution of the formula. Over time, inappropriate dilution can cause hyponatremia and water intoxication, leading to seizure activity. Slow correction of the sodium is curative.

**43–48. The answers are 43-e, 44-b, 45-d, 46-a, 47-g, 48-f.** (*Behrman, 16/e, pp 1971–1972, 1983, 1996–1997. McMillan, 3/e, pp 373, 375–377, 380,*

683–684, 706, 723, 730. *Rudolph, 21/e, pp 86–87, 1169, 1175–1176, 1210.*) Erythema toxicum is a benign, self-limited condition of unknown etiology. It is found in about 50% of term newborns. Lesions are yellow-white and 1 to 2 mm in size, with a surrounding edge of erythema. This rash waxes and wanes over the first days to weeks of life. Examination of the fluid from these lesions demonstrates eosinophils. No therapy is indicated.

Salmon patches are flat vascular lesions that occur in the listed regions and appear more prominent during crying. The lesions on the face fade over the first weeks of life. Lesions found over the nuchal and occipital areas often persist. No therapy is indicated.

Pustular melanosis is another benign, self-limited disease of unknown etiology of the newborn period. It is more common in blacks than in whites. These lesions are usually found at birth and consist of 1- to 2-mm pustules that result in a hyperpigmented lesion upon rupture of the pustule. The pustular stage of these lesions occurs during the first few days of life, with the hyperpigmented stage lasting for weeks to months. No therapy is indicated.

Sebaceous nevi (nevus of Jadassohn) are small, sharply edged lesions that occur most commonly on the head and neck of infants. These lesions are yellow-orange in color and are slightly elevated. They usually are hairless. Malignant degeneration is possible, most commonly after adolescence.

Milia are fine, yellowish white, 1- to 2-mm lesions scattered over the face and gingivae of the neonate. They are cysts that contain keratinized material. Commonly, these lesions resolve spontaneously without therapy.

Seborrheic dermatitis can begin anytime during life and frequently presents as cradle cap in the newborn period. This rash is commonly greasy, scaly, and erythematous and in smaller children involves the face, neck, axilla, and diaper area. In older children, the rash can be localized to the scalp and intertriginous areas. Pruritus can be marked.

**49–53. The answers are 49-a, 50-c, 51-e, 52-b, 53-c.** (*Behrman, 16/e, p 16. McMillan, 3/e, pp 822–827. Rudolph, 21/e, pp 417–420.*) Sleep disturbances are fairly common in childhood. Many children resist going to bed, and parents frequently give in just to get the child to sleep by allowing the child to sleep in the parents' bed or allowing them to stay up late. Unfortunately children learn remarkably well how to get what they want, and the parents' concessions only make the problem worse. Learned behavior is the root of many sleep disturbances in young children.

Other types of sleep disturbance in children fall into the category of sleep disruptions, such as nightmares and night terrors. A nightmare is a scary or disturbing dream that usually awakens the child and causes agitation about the content of the dream. Nightmares occur during rapid eye movement (REM) sleep. Many children and adults have an occasional nightmare; recurrent or frequent nightmares, however, may be indicative of an ongoing stress in the child's life.

Night terrors (*pavor nocturnus*) are non-REM phenomena seen less commonly than nightmares, occurring in 1 to 6% of all children. The child will be described as apparently awake but unresponsive. The child will seem to awake abruptly and have evidence of autonomic arousal such as tachycardia, sweating, and tachypnea, and appear frightened and agitated. Attempts at calming the child are usually not effective, and the child will eventually go back to sleep. Although usually a problem in early childhood, night terrors can sometimes continue through adolescence.

Somnambulism, or sleepwalking, occurs in 15% of children and is described as recurrent episodes of rising from bed and walking around. The child is typically hard to arouse and will have amnesia after the event. This usually happens in the first third of the sleep cycle, during stage 4 non-REM sleep. Somniloquy, or sleepwalking, can occur at any sleep stage and is seen in all ages.

**54–58. The answers are 54-b, 55-c, 56-d, 57-a, 58-c.** (*Behrman, 16/e, pp 231–237. McMillan, 3/e, pp 497, 499–502. Rudolph, pp 37–39.*) The majority of deaths from asphyxia or choking are due to food items (hot dogs, candy, grapes); occasionally they are also caused by nonfood items such as pacifiers, balloons, and small toys.

Drowning risks are age-dependent. Most drownings are accidental. Infants drown in bathtubs; unsupervised toddlers and young children fall into swimming pools, ponds, and streams. Alcohol and drugs are factors in many teenage drownings. Swimming pool drownings of children generally occur between ages 1 and 3 years. It is essential to counsel parents never to allow young children to swim or bathe unsupervised. Pools should be fenced, and meticulous attention must be paid to locking the gate.

Pedestrian injuries are the leading cause of death due to trauma in 5- to 9-year-old children and are most likely to occur with boys. Parents

should be advised to dress children who play outdoors in brightly colored, reflective clothing and to teach them the fundamentals of pedestrian safety.

Baby walkers are popular with many parents who believe erroneously that they are helpful to a baby who is learning to walk. They increase the mobility of an infant, which increases the infant's risks to the level of a more mobile toddler.

The Poison Prevention Packaging Act of 1970 achieved considerable success in reducing the number of accidental poisonings in children by mandating the manufacture of child-resistant containers, reducing the quantity of drug in a container to safer limits, and ordering the printing of warnings on container labels. Despite these safeguards, children, especially those less than 5 years of age, are still at risk of accidental poisonings. These accidents are more likely to occur when children are left unattended or unsupervised and when there is a disruption in the usual family routines. Anticipatory guidance regarding accidental ingestion is an essential part of well-child medical care.

**59–62. The answers are 59-a, 60-c, 61-b, 62-d.** (*Behrman, 16/e, pp 169, 179–181, 184–187, 2054. McMillan, 3/e, 470–475. Rudolph, pp 1323, 1336, 2156–2160.*) Marasmus (infantile atrophy) is due to inadequate caloric intake that can be linked with such factors as insufficient food resources, poor feeding techniques, metabolic disorders, and congenital anomalies. Patients with marasmus have progressive weight loss, constipation, muscular atrophy, loss of skin turgor, hypothermia, and, possibly, edema. In advanced disease, affected infants are lethargic and can have starvation diarrhea, with small, mucus-containing stools. These children appear emaciated and cachectic.

Pellagra, which literally means “rough skin,” is due to a deficiency of niacin (nicotinic acid). Niacin is an essential component of two enzymes—nicotinamide adenine dinucleotide (NAD) and nicotinamide adenine dinucleotide phosphate (NADP)—that are needed for electron transfer and glycolysis. Pellagra is most prevalent in areas that rely on corn as a basic foodstuff (corn contains little tryptophan, which can be converted into niacin). The classic (3-D) triad of clinical symptoms of pellagra consists of dermatitis, diarrhea, and dementia, although classic symptoms are not well developed in infants.

Beriberi results from a deficiency of thiamine (vitamin B<sub>1</sub>), which is essential for the synthesis of acetylcholine and for the operation of certain enzyme systems in carbohydrate metabolism. Thiamine is present in fair amounts in breast milk, cow's milk, cereals, fruits, vegetables, and eggs; meat and legumes are good sources. Thiamine is destroyed by heat and cooking in water, and polishing of grains reduces their thiamine content by removing the coverings that contain most of the vitamin. Clinical disturbances stemming from thiamine deficiency are congestive heart failure, peripheral neuritis, and psychic disturbances.

Rickets is a disorder of growing bone characterized by defective mineralization of matrix. Rickets that responds to administration of physiologic doses of vitamin D is termed vitamin D-deficient rickets. Deficiency of vitamin D can lead to osseous changes, such as enlargement of the costochondral junctions ("rachitic rosary") and craniotabes, within a few months; advanced rickets can cause scoliosis, pelvic and leg deformities, "pigeon breast," rachitic dwarfism, and other disorders. It is most commonly seen in industrialized nations in black children, children undergoing rapid growth (low-birth-weight infants and adolescents), and children with malabsorption syndromes.

**63–66. The answers are 63-a, 64-c, 65-b, 66-d.** (*Behrman, 16/e, pp 1113, 1746–1747, 1986, 2131–2132. McMillan, 3/e, pp 1893–1895, 2241, 2246, 2249, 2251. Rudolph, 21/e, pp 737, 754–755, 762–764, 1185, 2303.*) Waardenburg syndrome is the most common of several syndromes that are characterized by both deafness and pigmentary changes. Features of this syndrome, which is inherited as an autosomal dominant disorder, include a distinctive white forelock, heterochromia irides, unilateral or bilateral congenital deafness, and lateral displacement of the inner canthi.

Marfan syndrome is characterized by tall stature, long slender limbs, hypermobile joints, lens subluxation and myopia, and mitral valve prolapse. It is autosomal dominant with variation in expression. Aortic dilatation and dissection are potential complications.

Prader-Willi syndrome is characterized by infantile hypotonia and poor feeding, sometimes requiring gastrostomy tube feeding. As these children get older, however, they begin to overeat and develop obesity. They are prone to fits of rage, especially when denied food. The parents may report having to lock the refrigerator to keep the child from eating. Children with this syndrome have mild to moderate mental deficiency.

Treacher Collins syndrome patients are normal in intelligence. The primary problem is facial abnormalities, with malar hypoplasia, micrognathia, abnormally shaped ears, and hearing loss. The syndrome is inherited in an autosomal dominant manner.

**67–70. The answers are 67-b, 68-a, 69-d, 70-e.** (*Behrman, 16/e, pp 2080–2082, 2092. McMillan, 3/e, 2105–2107. Rudolph, 21/e, pp 907–908, 2436–2439.*) Legg-Calvé-Perthes disease is avascular necrosis or idiopathic osteonecrosis of the femoral head; the cause of this disorder is unknown. Boys between the ages of 2 and 12 years are most frequently affected (incidence in boys is greater by four- to fivefold; mean of 6 to 7 years old). Presenting symptoms include a limp and pain in the anterior thigh, groin, or knee, although classic symptoms include a painless limp.

Slipped capital femoral epiphysis is a disease of unknown etiology and occurs typically in adolescents; the disorder is most common among obese boys with delayed skeletal maturation or in thin, tall adolescents having recently enjoyed a growth spurt. Onset of this disorder is frequently gradual; pain referred to the knee in 20% of cases can mask the hip pathology.

Transient synovitis is a disorder of unknown etiology, affecting children usually from 2 to 6 years of age. These children usually present with a painful limp. This is a diagnosis of exclusion; septic hip and osteomyelitis must be ruled out. While the white count and sedimentation rate may be normal, they also may be slightly elevated. Early aspiration of the joint space may assist in diagnosis. Transient synovitis is a self-limited disorder.

Septic arthritis requires urgent intervention to preserve joint mobility. Joint aspiration is diagnostic and can be helpful in treatment. Opening the joint space may be required in a septic hip to assist in draining purulent material. These children need treatment for 4 to 6 weeks.

Osteomyelitis usually presents with focal bone tenderness and fever. Early evaluation is best done through nuclear medicine studies, as plain film bony changes usually take a week or so before becoming evident.

**71–75. The answers are 71-c, 72-b, 73-d, 74-e, 75-a.** (*Behrman, 16/e, pp 2160–2167. McMillan, 3/e, pp 620–621, 623–627, 629–635. Rudolph, 21/e, pp 360, 368–371, 373–376, 1657.*) The most important aspect of the management of lead poisoning is the identification and withdrawal of the source of the lead. Patients with symptomatic lead poisoning or high lead levels in the blood (over 70  $\mu\text{g/dL}$ ) should be

treated with both dimercaprol and calcium EDTA. With milder poisoning, intravenous or intramuscular calcium EDTA or, more likely, oral dimercaptosuccinic acid can be used.

*N*-acetylcysteine (NAC) is an effective treatment for acetaminophen poisoning and acts by removing hepatotoxic metabolites. It should be given within 16 h of ingestion; after 36 h it is probably ineffective.

Morphine and other narcotics produce their major toxic effect by suppression of ventilation. Ventilatory support can be necessary initially, but naloxone is a specific antidote and can be very rapidly effective. The effect of naloxone can wear off more quickly than the effects of the drug for which it was given, so careful observation and repeated doses may be necessary.

Salicylate poisoning is not treated with any of these drugs. It produces metabolic acidosis and respiratory alkalosis (although this latter feature is often missed in young children), hyperglycemia and hypoglycemia, dehydration, and lethargy. Excretion of salicylates in the urine can be markedly enhanced by the administration of acetazolamide and intravenous sodium bicarbonate. Hemodialysis can also be used.

Organophosphate insecticides are absorbed from all sites and act by inhibiting cholinesterases, thereby leading to the accumulation of high levels of acetylcholine. This affects the parasympathetic nervous system, muscle, and the central nervous system. Treatment of a patient contaminated with organophosphate insecticide will include thorough washing of the pesticide from the skin, inducing emesis or performing gastric lavage, supporting ventilation, and administering atropine followed by pralidoxime (2-PAM).

**76–78. The answers are 76-b, 77-a, 78-d.** (*Behrman, 16/e, pp 2160–2169. McMillan, 3/e, pp 620–621. Rudolph, 21/e, pp 347, 367–368, 371–372.*) Phenothiazine toxicity can cause extrapyramidal symptoms such as oculogyric crisis, tremors, torticollis, opisthotonus, and dysphagia. These dystonic symptoms respond surprisingly quickly to the intravenous or intramuscular administration of diphenhydramine (Benadryl).

Iron in the form of salts such as ferrous sulfate or gluconate used to treat iron deficiency anemia can be highly toxic to infants; as few as three tablets can cause severe symptoms and as few as 10 tablets can be lethal to young children. Symptoms occur in two phases: gastrointestinal symptoms

such as bloody vomiting or diarrhea and abdominal pain, followed by a latent period of up to 12 h or more and terminating with cardiovascular collapse. Deferoxamine given intravenously or intramuscularly forms a complex with the iron and is excreted in the urine, to which it imparts the color of vin rosé (red wine).

Methanol, also known as methyl alcohol or wood alcohol, is present in a number of household products and is a frequent contaminant of bootleg alcohol. Toxicity causes a profound metabolic acidosis. Treatment includes emptying the stomach by inducing emesis or by gastric lavage, the intravenous infusion of ethanol to saturate the enzyme systems that convert methanol to toxins, and, in severe poisoning, the use of hemodialysis to remove the methanol.

**79–83. The answers are 79-d, 80-g, 81-a, 82-e, 83-b.** (*Behrman, 16/e, pp 146–147, 178, 181–184, 187–188. McMillan, 3/e, pp 83–84, 474–475. Rudolph, 21/e, pp 1322, 1323.*) So far, there is very little or no well-documented clinical evidence to justify the use of large doses of vitamins beyond the levels found in a regular, balanced diet. Vitamin excess can be dangerous. Vitamin A in large doses slows growth and causes hyperostosis (excess bone growth), hepatomegaly, increased CSF pressure, and drying of skin. Nicotinic acid, a vasodilator, causes skin flushing and pruritus; long-term use can cause tachycardia, liver damage, hyperglycemia, and hyperuricemia. Excessive doses of vitamin C, in addition to causing kidney stones, diarrhea, and cramps, increase the normal requirement for the vitamin when large doses are discontinued. Prolonged excessive intake of vitamin D can cause nausea, diarrhea, weight loss, polyuria, and soft-tissue calcification of heart, kidney, blood vessels, bronchi, and stomach. Excessive intake of pyridoxine (B<sub>6</sub>) can result in sensory neuropathy with altered sensation of touch, pain, and fever.

**84–88. The answers are 84-f, 85-b, 86-d, 87-a, 88-e.** (*Behrman, 16/e, pp 468–469, 473, 716. McMillan, 3/e, pp 2165, 2227, 2228, Rudolph, 21/e, pp 774–779.*) The fetal environment is affected by factors that alter maternal metabolism—among them, diseases such as juvenile-onset insulin-dependent diabetes mellitus and lupus erythematosus. Infants of diabetic mothers have a two to three times greater risk than the general population of having anomalies. These anomalies include sacral agenesis (which

is pathognomonic for maternal diabetes mellitus), cardiac defects (e.g., transposition of the great arteries and ventricular septal defects), renal defects, and anencephaly.

The use of the anticonvulsants phenytoin and valproate during pregnancy to control seizures has been associated with findings of fetal malformation. Prenatal exposure to phenytoin can result in the characteristic features of the fetal hydantoin syndrome: pre- and postnatal growth retardation, hypoplasia of distal phalanges and nails, and alterations in central nervous system performance. The incidence of neural tube defects in the fetus can be increased in mothers receiving valproate during the first trimester of pregnancy. Although the risk (1 to 2%, as estimated by the Centers for Disease Control and Prevention) is similar to that for non-epileptic women who have had children with neural tube defects, caution is advised in the use of this drug during pregnancy.

Heavy alcohol ingestion is associated with a syndrome (fetal alcohol syndrome) consisting of pre- and postnatal growth retardation, developmental delay, and a characteristic facies that includes microcephaly, microphthalmia, and a flattened philtrum area. Skeletal, joint, and cardiac abnormalities have also been associated with this syndrome. It has been stated that infants of mothers who drink heavily (>3 oz of absolute alcohol daily) have a 30 to 50% risk of abnormalities of growth and performance. No safe amount of alcohol consumption during pregnancy is known; therefore, pregnant women and those who might become pregnant should be advised to abstain from alcohol.

The clinical findings associated with neonatal lupus erythematosus include skin lesions and cardiac abnormalities, among them AV block (which is a permanent condition), complete transposition of the great arteries, and valvular and septal defects. Most mothers are symptomatic prior to or during pregnancy, but occasionally the diagnosis of lupus in the mother is made when her newborn is found to have heart block. Most babies with lupus erythematosus will have anti-Ro antibodies, which are considered markers for the disease. Antinuclear antibodies, which are also present in most infants, also disappear by 6 months of age.

**89–92. The answers are 89-b, 90-c, 91-d, 92-a.** (*Behrman, 16/e, pp 680–684, 2028–2029, 2042, 2044–2046. McMillan, 3/e, pp 688–689, 702, 704–706, 716–717, 2059–2063. Rudolph, 21/e, pp 1056, 1153–1154, 1177–1179, 1217.*) Impetigo is a common skin condition of children that

begins on the face or extremities after the skin has been traumatized. The two organisms that cause most of the infections are *Staphylococcus aureus* and  $\beta$ -hemolytic streptococci. In most cases, a tiny vesicle or pustule forms, which rapidly degenerates into a honey-colored, crusty plaque. Usually no pain or systemic symptoms are associated with these lesions. Complications are rare, but acute poststreptococcal glomerulonephritis is possible. Treatment is by topical or systemic antibiotics.

Scabies is caused by mites (*Sarcoptes scabiei* var. *hominis*), which burrow into the skin and release toxins that result in extremely pruritic lesions. These lesions range from a threadlike burrow in older patients to bullae and pustules in infants. Common sites of infection in older patients are the interdigital spaces, wrist, axillary folds, ankles, buttocks, and groin. An infant's most common locations are feet, palms, axilla, and scalp. The lesions are rarely found above the neck in older children. The diagnosis can be made either clinically or by microscopic identification of the mites. Therapy consists of treatment with either 1% lindane (not in smaller children) or permethrin 5% cream (less toxicity for smaller children).

Molluscum contagiosum is caused by a poxvirus. Lesions are skin-colored, dome-shaped, and about 1 to 5 mm in size. They can be distinguished by the central umbilication found on most lesions. Areas of predilection include the face, eyelids, neck, axillae, and thighs. Diagnosis can usually be made on clinical grounds. Therapy includes observation (the disease can be self-limited), application of liquid nitrogen to the lesion, extrusion of the central plug with a needle or curette, or a topical application of cantharidin 0.9% to each lesion.

Atopic dermatitis, an immediate hypersensitivity reaction to common environmental irritants, has a prevalence of 2 to 3% in children. Inflammatory patches and weeping, crusted plaques on the neck, face, groin, and extensor surfaces characterize the infantile form. In older children, dermatitis of the flexural areas is common. Soaps and hot water are common irritants. Therapy is based on avoidance of irritants, adequate hydration of the skin, and use of topical steroids, as well as treatment of infected lesions.

**93–95. The answers are 93-d, 94-a, 95-c.** (Behrman, 16/e, pp 716, 1369–1371, 1575–1576, 1581–1582. McMillan, 3/e, pp 381, 1354–1357, 1581–1598, 2165. Rudolph, 21/e, pp 1677–1691, 1788–1791.) The ventricular septal defect is the most common congenital cardiac malformation. Although small lesions result in insignificant left-to-right shunts, the

murmur associated with them can be significantly louder due to turbulent blood flow. Larger lesions result in significant left-to-right shunting of blood and can result in dyspnea, poor growth, and heart failure, usually during early infancy.

The classic presentation of acute poststreptococcal glomerulonephritis consists of the sudden onset of change in urine color (bloody), edema, hypertension, and renal insufficiency. This disease follows an infection of the throat or the skin with a nephrogenic strain of group A  $\beta$ -hemolytic streptococci. The oliguria can progress to heart failure if fluid overload occurs.

Neonatal lupus can be responsible for a variety of problems in the newborn. Mothers with antibodies to Ro/SSA and some with antibodies to La/SSB can deliver infants with rashes, thrombocytopenia, and congenital heart block among the more common problems. Whereas the other symptoms usually resolve during the first months of life, the congenital heart block can be irreversible and can result in heart failure, need for early pacemakers, and an increased incidence of early death.

**96–102. The answers are 96-b, 97-f, 98-c, 99-a, 100-d, 101-f, 102-e.** (*Behrman, 16/e, pp 151, 178, 459, 1163–1164, 1467–1468. McMillan, 3/e, 367, 477, 1035–1036, 1487–1488, 1753–1754. Rudolph, 21/e, pp 31, 105, 956–957, 1055, 1513, 1529.*) Isoniazid therapy can cause peripheral neuritis as a result of competitive inhibition of pyridoxine metabolism when the dose is high and the patient is poorly nourished or is an alcoholic. It is seldom seen in childhood. To be safe, vitamin B<sub>6</sub> supplements should be given to adolescents. Children maintained on anticonvulsants can develop low folate levels that, rarely, can be associated with megaloblastic anemia. Folic acid supplementation may be indicated.

The World Health Organization (WHO) recommends that, in communities where vitamin A deficiency is prevalent, vitamin A be given to all children with measles. Compliance with this recommendation has resulted in a definite reduction in measles-related morbidity and mortality. In the United States, vitamin A supplements should be considered for use in measles patients with immunodeficiency, impaired intestinal absorption, and malnutrition. Recent immigrants from areas with a high mortality from measles and who show ophthalmologic evidence of vitamin A deficiency (blindness, Bitot's spots, or xerophthalmia) should also be included in that group.

Fat malabsorption occurs (1) in the absence of pancreatic enzymes, as in cystic fibrosis; (2) as a result of failure of micellar solubilization by bile salts, as in chronic liver disease; and (3) with a problematic mucosal uptake, as in celiac sprue. For people with these conditions, attention must be paid to the provision of fat-soluble vitamins A, D, E, and K.

Both human and cow's milk are low in vitamin D content. Cow's milk and infant formulas are fortified with this vitamin. Breast-fed infants require supplementation with vitamin D when adequate exposure to sunlight cannot be ensured, such as in urban areas where pollutants obscure ultraviolet light, in northern areas with long periods of darkness, and in patients with darkly pigmented skin.

Patients with hemolytic anemia, such as sickle cell disease, have an ongoing compensatory erythropoiesis. To supply the increased need of rapidly dividing red blood cell precursors for folate, supplementation is necessary.

In newborns, a lack of free vitamin K in the mother and the absence of bacterial intestinal flora that synthesizes vitamin K result in a transient deficiency in vitamin K-dependent factors (II, VI, IX, X). Milk is a poor source of vitamin K. Vitamin K administered shortly after birth prevents hemorrhagic disease of the newborn.

**103–107. The answers are 103-c, 104-f, 105-b, 106-g, 107-e.** (Behrman, 16/e, p 617. McMillan, 3/e, pp 2091–2092. Rudolph, 21/e, pp 793–802.) The bulk of immunodeficiencies can be ruled out with little cost. Wiskott-Aldrich syndrome is unlikely if the platelet count is normal. Asplenia results in Howell-Jolly bodies, so a complete blood count can rule out this disease. B-cell defects are likely to result in low immunoglobulin A, G, and M levels. An intradermal skin test using *Candida albicans* will result in no response in the patient with T-cell deficiencies. The nitroblue tetrazolium (NBT) or other respiratory burst assay will help identify phagocytic-cell defects such as chronic granulomatous disease. Should any of these tests prove to be positive, more extensive, invasive, and expensive testing can be undertaken.

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# The Newborn Infant

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## Questions

**DIRECTIONS:** Each item below contains a question or incomplete statement followed by suggested responses. Select the **one best** response to each question.

**108.** The infant pictured as follows was small for gestational age. He also was found to have a ventricular septal defect on cardiac evaluation. This infant appeared to have features consistent with



- a. Perinatal phenytoin exposure
- b. Trisomy 21
- c. Alport syndrome
- d. Fetal alcohol syndrome
- e. Infant of diabetic mother

**109.** A newborn is noted to be quite jaundiced at 3 days of age. Which of the following factors is associated with an increased risk of neurologic damage in a jaundiced newborn?

- a. Metabolic alkalosis
- b. Increased attachment of bilirubin to binding sites caused by drugs such as sulfisoxazole
- c. Hyperalbuminemia
- d. Neonatal sepsis
- e. Maternal ingestion of phenobarbital during pregnancy

**110.** A full-term newborn infant is having episodes of cyanosis and apnea, which are worse when he is attempting to feed, but he seems better when he is crying. The most important next step to quickly establish the diagnosis is

- a. Echocardiogram
- b. Ventilation perfusion scan
- c. Passage of catheter into nose
- d. Hemoglobin electrophoresis
- e. Bronchoscopic evaluation of palate and larynx

**111.** The mother of a 7-day-old infant has developed chickenpox. Which of the following is the most appropriate measure?

- a. Isolate the infant from the mother
- b. Hospitalize the infant in the isolation ward
- c. Administer acyclovir to the infant
- d. Administer varicella-zoster immunoglobulin to the infant
- e. Advise the mother to continue regular well-baby care for the infant

**112.** A 2-week-old infant has had no immunizations, sleeps 18 h a day, weighs 3.5 kg, and takes 60 mL of standard infant formula four times a day, but no solid food and no iron or vitamin supplements. Of most concern is

- a. Immunization status
- b. Caloric intake
- c. Iron levels
- d. Levels of vitamins A, C, and D
- e. Circadian rhythm

**113.** A mother delivers a neonate with meconium staining and Apgar scores of 3 at 1 and 5 min of life. She had no prenatal care and the delivery was by emergency cesarean section for severe fetal bradycardia. Which of the following sequelae could be expected to develop in this intubated neonate with respiratory distress?

- a. Sustained rise in pulmonary arterial pressure
- b. Hyperactive bowel sounds
- c. Microcephaly with micrognathia
- d. Cataracts
- e. Thrombocytosis

**114.** A 2-year-old boy is being followed for congenital cytomegalovirus (CMV) infection. He is deaf and developmentally delayed. The child's mother informs you that she has just become pregnant and is concerned that the new baby will be infected. Which of the following is true?

- a. The mother has antibodies to CMV that are passed to the fetus
- b. The mother's infection cannot become reactivated
- c. The likelihood that the new baby will become clinically ill is approximately 80%
- d. Termination of pregnancy is advised
- e. The new infant should be isolated from the older child

**115.** A full-term infant is born after a normal pregnancy; delivery, however, is complicated by marginal placental separation. At 12 h of age, the child, although appearing to be in good health, passes a bloody meconium stool. For determining the cause of the bleeding, which of the following diagnostic procedures should be performed first?

- a. A barium enema
- b. An Apt test
- c. Gastric lavage with normal saline
- d. An upper gastrointestinal series
- e. A platelet count, prothrombin time, and partial thromboplastin time

**116.** As you are about to step out of a newly delivered mother's room, she mentions that she wants to breast-feed her healthy infant, but that her obstetrician was concerned about one of the medicines she was taking. Which of the woman's medicines, listed below, is clearly contraindicated in breast-feeding?

- a. Ibuprofen as needed for pain or fever
- b. Labetolol for her chronic hypertension
- c. Lithium for her bipolar disorder
- d. Carbamazepine for her seizure disorder
- e. Acyclovir for her HSV outbreak

**117.** The nurse from the level II neonatal intensive care nursery calls you to evaluate a baby. The infant, born at 32 weeks' gestation, is now 1 week old and had been doing well on increasing nasogastric feedings. This afternoon, however, the nurse noted that the infant has vomitted the last two feedings and seems less active. Your examination reveals a tense and distended abdomen with decreased bowel sounds. As you are evaluating the child, he has a grossly bloody stool. Your management of this infant should include

- a. Surgical consultation for an emergent exploratory laparotomy
- b. Continued feeding of the infant, as gastroenteritis is usually self-limited
- c. Stool culture to identify the etiology of the bloody diarrhea and an infectious diseases consultation
- d. Stopping feeds, beginning intravenous fluids, ordering serial abdominal films, and initiating systemic antibiotics
- e. Upper GI series and barium enema to evaluate for obstruction

**118.** A recovering premature infant who weighs 950 g (2 lb, 1 oz) is fed breast milk to provide 120 cal/(kg·d). Over ensuing weeks, the baby is most apt to develop

- a. Hypernatremia
- b. Hypocalcemia
- c. Blood in the stool
- d. Hyperphosphatemia
- e. Vitamin D toxicity

**119.** An infant weighing 1400 g (3 lb) is born at 32 weeks' gestation in a delivery room that has an ambient temperature of 24°C (75°F). If left in an open crib for a few minutes, this child is likely to demonstrate

- a. Ruddy complexion
- b. Shivering
- c. Hypertension
- d. Increased respiratory rate
- e. Metabolic alkalosis

**120.** A primiparous woman whose blood type is O positive gives birth at term to an infant who has A-positive blood and a hematocrit of 55%. A serum bilirubin level obtained at 36 h of age is 12 mg/dL. Which of the following laboratory findings would be characteristic of ABO hemolytic disease?

- a. A normal reticulocyte count
- b. A positive direct Coombs test
- c. Crescent-shaped red blood cells in the blood smear
- d. Elevated hemoglobin
- e. Petechiae

**121.** Two infants are born at 36 weeks' gestation. Infant A weighs 2600 g (5 lb, 12 oz) and infant B weighs 1600 g (3 lb, 8 oz). Infant B is more likely to have which of the following problems?

- a. Congenital malformations
- b. Low hematocrit
- c. Hyperglycemia
- d. Surfactant deficiency
- e. Rapid catch-up growth retardation

**122.** A 3-day-old infant born at 32 weeks' gestation and weighing 1700 g (3 lb, 12 oz) has three episodes of apnea, each lasting 20 to 25 s and occurring after a feeding. During these episodes, the heart rate drops from 140 to 100 beats per min, and the child remains motionless; between episodes, however, the child displays normal activity. Blood sugar is 50 mg/dL and serum calcium is normal. The child's apneic periods most likely are

- a. Due to an immature respiratory center
- b. A part of periodic breathing
- c. Secondary to hypoglycemia
- d. Manifestations of seizures
- e. Evidence of underlying pulmonary disease

### Items 123–124

**123.** You are seeing a 2-week-old boy at a routine visit. The mother complains that he has been constipated, jaundiced, sluggish, and excessively sleepy. The physical examination is normal except for mild jaundice and a distended abdomen in a sleepy infant. The most appropriate course to pursue initially is assessment of

- a. The mother's serum for autoantibodies to thyroid gland
- b. Levels of thyrotropin-releasing hormone (TRH), thyroid-stimulating hormone (TSH), tri-iodothyronine ( $T_3$ ), reverse  $T_3$ , levothyroxine ( $T_4$ ), and thyroglobulin in the mother
- c. The results of the neonatal metabolic screen
- d. Levels of thyrotropin-releasing hormone (TRH), thyroid-stimulating hormone (TSH), tri-iodothyronine ( $T_3$ ), reverse  $T_3$ , levothyroxine ( $T_4$ ), and thyroglobulin in the infant
- e. The effects on growth and symptoms with increasing feeds by 20% per day

**124.** The most appropriate next step is to

- a. Repeat all the baby's abnormal laboratory results (if any)
- b. Obtain x-rays of the baby's skull, wrists, and knees
- c. Begin on the baby oral sodium-L-thyroxine, 10 to 15 mg/(kg·d)
- d. Evaluate the neonate in 2 weeks for the results of symptomatic treatment
- e. Obtain a pediatric endocrinology consultation within 2 weeks

### Items 125–126

**125.** A 1-day-old infant who was born by a difficult forceps delivery is alert and active. She does not move her left arm, however, which she keeps internally rotated by her side with the forearm extended and pronated; she also does not move it during a Moro reflex. The rest of her physical examination is normal. This clinical picture most likely indicates

- a. Fracture of the left clavicle
- b. Fracture of the left humerus
- c. Left-sided Erb-Duchenne paralysis
- d. Left-sided Klumpke paralysis
- e. Spinal injury with left hemiparesis

**126.** The infant in the preceding question immediately develops tachypnea with cyanosis. She improves somewhat on oxygen but has predominantly thoracic breathing movements, and the chest x-ray, which appears to have been taken inadvertently at expiration, seems normal. The procedure most likely to provide a specific etiologic diagnosis is

- a. Venous blood gas
- b. CT scan of the head
- c. Ultrasound or fluoroscopy of the chest
- d. Bronchoalveolar lavage
- e. Blood culture

**127.** A 19-year-old primiparous woman develops toxemia in her last trimester of pregnancy and during the course of her labor is treated with magnesium sulfate. At 38 weeks' gestation, she delivers a 2100-g infant with Apgar scores of 1 at 1 min and at 5 at 5 min. Laboratory studies at 18 h of age reveal a hematocrit of 79%, platelet count of 100,000/ $\mu$ L, glucose 38 mg/dL, magnesium 2.5 meq/L, and calcium 8.7 mg/dL. Soon after, this the infant has a generalized convulsion. The most likely cause of the infant's seizure is

- a. Polycythemia
- b. Hypoglycemia
- c. Hypocalcemia
- d. Hypermagnesemia
- e. Thrombocytopenia

**128.** An infant who appears to be of normal size is noted to be lethargic and somewhat limp after birth. The mother is 28 years old, and this is her fourth delivery. The pregnancy was uncomplicated, with normal fetal monitoring prior to delivery. Labor was rapid, with local anesthesia and intravenous meperidine administered for maternal pain control. Which of the following therapeutic maneuvers is likely to improve this infant's condition most rapidly?

- a. Intravenous infusion of 10% dextrose in water
- b. Administration of naloxone (Narcan)
- c. Administration of vitamin K
- d. Measurement of electrolytes and magnesium levels
- e. Neurologic consult

**129.** At 43 weeks' gestation, a long, thin infant is delivered. The infant is apneic, limp, pale, and covered with "pea soup" amniotic fluid. The first step in the resuscitation of this infant at delivery should be

- a. Suction of the trachea under direct vision
- b. Artificial ventilation with bag and mask
- c. Artificial ventilation with endotracheal tube
- d. Administration of 100% oxygen by mask
- e. Catheterization of the umbilical vein

**130.** The newborn pictured was born at home and has puffy, tense eyelids; red conjunctivae; a copious amount of purulent ocular discharge; and chemosis 2 days after birth. The most likely diagnosis is



*(Courtesy Kathryn Musgrove, M.D.)*

- a. Dacryocystitis
- b. Chemical conjunctivitis
- c. Pneumococcal ophthalmia
- d. Gonococcal ophthalmia
- e. Chlamydial conjunctivitis

**131.** After an uneventful labor and delivery, an infant is born at 32 weeks' gestation weighing 1500 g (3 lb, 5 oz). Respiratory difficulty develops immediately after birth and increases in intensity thereafter. The child's mother (now gravida 3, para 2102) previously lost an infant because of hyaline membrane disease. At 6 h of age, the child's respiratory rate is 60 breaths per min. Examination reveals grunting, intercostal retraction, nasal flaring, and marked cyanosis in room air. Physiologic abnormalities compatible with these data include

- a. Decreased lung compliance, reduced lung volume, left-to-right shunt of blood
- b. Decreased lung compliance, reduced lung volume, right-to-left shunt of blood
- c. Decreased lung compliance, increased lung volume, left-to-right shunt of blood
- d. Normal lung compliance, reduced lung volume, left-to-right shunt of blood
- e. Normal lung compliance, increased lung volume, right-to-left shunt of blood

**132.** A term infant is born to a known HIV-positive mother. She has been taking antiretroviral medications for the weeks prior to the delivery of her infant. Routine management of the healthy infant should include

- a. Admission to the neonatal intensive care unit for close cardiovascular monitoring
- b. HIV ELISA on the infant to determine if congenital infection has occurred
- c. A course of zidovudine for the infant
- d. Chest radiographs to evaluate for congenital *Pneumocystis carinii*
- e. Administration of IVIG to the baby to decrease the risk of perinatal HIV infection

**133.** Initial examination of a full-term infant weighing less than 2500 g (5 lb, 8 oz) shows edema over the dorsum of her hands and feet. Which of the following findings would support a diagnosis of Turner syndrome?

- a. A liver palpable to 2 cm below the costal margin
- b. Tremulous movements and ankle clonus
- c. Redundant skin folds at the nape of the neck
- d. A transient, longitudinal division of the body into a red half and a pale half
- e. Softness of the parietal bones at the vertex

**134.** A 1-week-old black infant presents to you for the first time with a large, fairly well-defined, purple lesion over the buttocks bilaterally, as shown in the photograph. The lesion is not palpable, and it is not warm or tender. The mother denies trauma and reports that the lesion has been present since birth. This otherwise well-appearing infant is growing and developing normally and appears normal upon physical examination. The most likely diagnosis in this infant is



*(Courtesy Adelaide Hebert, M.D.)*

- a. Child abuse
- b. Mongolian spot
- c. Subcutaneous fat necrosis
- d. Vitamin K deficiency
- e. Hemophilia

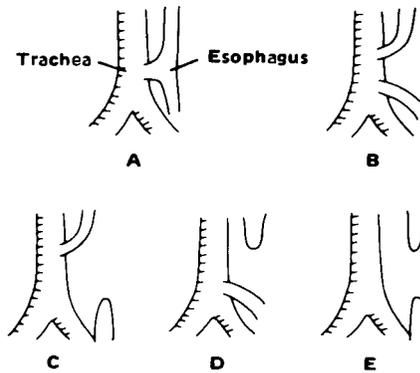
**135.** A newborn infant develops respiratory distress immediately after birth. His abdomen is scaphoid. No breath sounds are heard on the left side of his chest, but they are audible on the right. Immediate intubation is successful with little or no improvement in clinical status. Emergency chest x-ray is shown (A) along with an x-ray 2 h later (B). The most likely explanation for this infant's condition is

**A****B**

*(Courtesy Susan John, M.D.)*

- Pneumonia
- Cyanotic heart disease
- Diaphragmatic hernia
- Choanal atresia
- Pneumothorax

**136.** Shortly after birth, an infant develops abdominal distention and begins to drool. When she is given her first feeding, it runs out the side of her mouth, and she coughs and chokes. Physical examination reveals tachypnea, intercostal retractions, and bilateral pulmonary rales. The esophageal anomaly that most commonly causes these signs and symptoms is illustrated by



- A
- B
- C
- D
- E

**137.** You are speaking to a couple who are expecting their first baby in about 2 weeks. They are concerned about the safety of childhood immunizations and also about “unnecessary” medications given to newborns in the hospital. They ask about the purpose of the routine administration of intramuscular vitamin K. You explain to them about hemorrhagic disease of the newborn, stating that the untreated baby can manifest

- A deficiency of factor V
- A prolonged prothrombin time and a risk of serious hemorrhage in the days following delivery
- Development of hemorrhagic manifestations within 24 h of delivery
- More severe symptoms in a male than a female child
- Hemorrhagic symptoms at a much higher rate if the infant is fed cow’s milk rather than breast milk

**138.** You are advised by the obstetrician that the mother of a baby he has delivered is a carrier of hepatitis B surface antigen (HBsAg-positive). The most appropriate action in managing this infant would be to

- a. Screen the infant for HBsAg
- b. Isolate the infant for enteric transmission
- c. Screen the mother for hepatitis B “e” antigen (HBeAg)
- d. Administer hepatitis B immune globulin and hepatitis B vaccine to the infant
- e. Do nothing because transplacentally acquired antibody will prevent infection in the infant

**139.** You are called to a delivery of a term infant, about to be born via cesarean section to a mother with multiple medical problems including a 1-month history of a seizure disorder, for which she takes phenytoin; rheumatic heart disease, for which she must take penicillin daily for life; hypertension, for which she takes propranolol; acid reflux, for which she takes aluminum hydroxide; and a deep venous thrombosis in her left calf diagnosed two days ago, for which she was started on a heparin infusion. The obstetrician is concerned about the possible effects of the mother’s multiple medications on the newborn infant. You correctly note that the one medication most likely to cause harm in this newborn infant at delivery is

- a. Propranolol
- b. Penicillin
- c. Aluminum hydroxide
- d. Phenytoin
- e. Heparin

**140.** At the time of delivery, a woman is noted to have a large volume of amniotic fluid. At 6 h of age, her baby begins regurgitating small amounts of mucus and bile-stained fluid. Physical examination of the infant is normal, and an abdominal x-ray is obtained (shown in the following figure). The most likely diagnosis of this infant's disorder is



*(Courtesy Susan John, M.D.)*

- a. Gastric duplication
- b. Pyloric stenosis
- c. Esophageal atresia
- d. Duodenal atresia
- e. Midgut volvulus

**Items 141–142**

**141.** You are called to the normal newborn nursery to see a baby who was noted to be mildly jaundiced and has a total serum bilirubin concentration of 12 mg/dL at 48 h of age. The baby is a 3500-g boy who was born at term to a 27-year-old O-positive, Coombs-test-negative primigravida 2 h after membranes ruptured. There were no prenatal complications, and the mother had regular prenatal care. Breast-feeding has been well tolerated, and the baby's vitals have been normal. The most appropriate additional diagnostic studies to evaluate the cause of this infant's jaundice are

- a. Direct serum bilirubin, complete blood count, liver ultrasound, liver transaminases
- b. Direct serum bilirubin, complete blood count, red blood cell glucose-6-phosphate dehydrogenase levels (G6PD), baby blood type
- c. Complete blood count, direct serum bilirubin, baby blood type and Coombs, peripheral smear
- d. Blood cultures, liver transaminases, red blood cell osmotic fragility, direct serum bilirubin
- e. Abdominal radiograph, urine bilirubin, complete blood count, direct serum bilirubin

**142.** Since you are a new intern, you ordered all of the diagnostic studies you could think of instead of just the ones your senior resident told you were most appropriate. The nurse calls to inform you that the infant's studies are back. Both the mother and baby have O-positive blood. The baby's direct serum bilirubin is 0.2 mg/dL, with a repeat total serum bilirubin of 11.8 mg/dL. Urine bilirubin is positive. The mother's white count is 13,000/ $\mu$ L with a differential of 50% polymorphonuclear cells, 45% lymphocytes, and 5% monocytes. The hemoglobin is 17 g/dL, and the platelet count is 278,000/ $\mu$ L. Reticulocyte count is 1.5%. The peripheral smear does not show fragments or abnormal cell shapes. Blood cultures are pending in the laboratory. Liver enzymes and liver ultrasound are normal. G6PD levels and osmotic fragility testing are normal. The most likely diagnosis in this infant is

- a. Rh or ABO hemolytic disease
- b. Physiologic jaundice
- c. Sepsis
- d. Congenital spherocytic anemia
- e. Biliary atresia

**143.** You see the baby shown in the picture for the first time in the nursery as a newborn. You consult plastic and reconstructive surgery as well as the hospital's speech therapist. Understandably, the parents have many questions. Which of the statements listed below is appropriate anticipatory guidance for this family?



- a. Parenteral alimentation is recommended to prevent aspiration
- b. Surgical closure of the palatal defect should be done before 3 months of age
- c. Good anatomic closure will preclude the development of speech defects
- d. Recurrent otitis media and hearing loss are likely complications
- e. The chance that a sibling also would be affected is 1 in 1000

**144.** A 26-year-old gravida 3 woman has a history of gestational diabetes and a delivery of two previous infants at term that were greater than 4000 grams, each of whom had severe hypoglycemia. Which of the following maneuvers is least likely to reduce the chance of the next child's having hypoglycemia?

- a. Careful control of the maternal blood glucose levels during pregnancy
- b. Maternal intravenous loading with 10% glucose beginning 2 to 4 h prior to the expected time of delivery
- c. Careful glucose monitoring of the infant
- d. Early feedings of the infant
- e. Maintenance of the infant in a neutral thermal environment

**I45.** A term, 4200-g female infant is delivered via cesarean section because of cephalopelvic disproportion. The amniotic fluid was clear, and the infant cried almost immediately after birth. Within the first 15 min of life, however, the infant's respiratory rate increased to 80 breaths per min, and she began to have intermittent grunting respirations. The infant was transferred to the level 2 nursery and was noted to have an oxygen saturation of 94%. The chest radiograph showed fluid in the fissure, overaeration, and prominent pulmonary vascular markings. The most likely diagnosis in this infant is

- a. Diaphragmatic hernia
- b. Meconium aspiration
- c. Pneumonia
- d. Idiopathic respiratory distress syndrome
- e. Transient tachypnea of the newborn

**146.** The infant in the following pictures (A) and (B) presented with hepatosplenomegaly, anemia, persistent rhinitis, and a maculopapular rash. The most likely diagnosis for this child is



**A** (Courtesy Lawrence Fineberg, M.D.)



**B** (Courtesy Adelaide Hebert, M.D.)

- a. Toxoplasmosis
- b. Glycogen storage disease
- c. Congenital hypothyroidism
- d. Congenital syphilis
- e. Cytomegalovirus disease

**147.** A well-appearing, 3200-g black infant is noted to have fifth finger polydactyly. The extra digit has no skeletal duplications and is attached to the rest of the hand by a threadlike soft tissue pedicle (see photograph). Appropriate treatment for this condition includes



*(Courtesy Adelaide Hebert, M.D.)*

- a. Chromosomal analysis
- b. Excision of extra digit
- c. Skeletal survey for other skeletal abnormalities
- d. Echocardiogram
- e. Renal ultrasound

**148.** A routine prenatal ultrasound reveals a male fetus with meningomyelocele. The 24-year-old primigravid mother is told the infant will require surgery shortly after birth. You counsel her about the etiology of this defect and the risk of further pregnancies being similarly affected, stating that

- a. The hereditary pattern for this condition is autosomal recessive
- b. The prenatal diagnosis can be made by the detection of very low levels of  $\alpha$ -fetoprotein in the amniotic fluid
- c. Subsequent pregnancies are not at increased risk compared to the general population
- d. Supplementation of maternal diet with folate leads to a decrease in incidence of this condition
- e. Neither environmental nor social factors have been shown to influence the incidence

**149.** An infant born at 35 weeks' gestation to a mother with no prenatal care is noted to be jittery and irritable, and is having difficulty feeding. You note coarse tremors on examination. The nurses report a high-pitched cry and note several episodes of diarrhea and emesis. You suspect the infant is withdrawing from

- a. Alcohol
- b. Marijuana
- c. Heroin
- d. Cocaine
- e. Tobacco

**150.** A previously healthy full-term infant has several episodes of duskiess and apnea during the second day of life. Diagnostic considerations should include which of the following?

- a. Hemolytic anemia
- b. Congenital heart disease
- c. Idiopathic apnea
- d. Harlequin syndrome
- e. Hyperglycemia

**151.** The signs and symptoms of meningitis in an infant can be different than those in an adult. Which of the signs and symptoms of meningitis listed below is more helpful in an adult patient than in a 4-month-old?

- a. Lethargy
- b. Jaundice
- c. Vomiting
- d. Brudzinski's sign
- e. Hypothermia

**152.** A woman gives birth to twins at 38 weeks' gestation. The first twin weighs 2800 g (6 lb, 3 oz) and has a hematocrit of 70%; the second twin weighs 2100 g (4 lb, 10 oz) and has a hematocrit of 40%. Which of the following statements is correct?

- a. The second twin is at risk for developing respiratory distress, cyanosis, and congestive heart failure
- b. The first twin is more likely to have hyperbilirubinemia and convulsions
- c. The second twin is at risk for renal vein thrombosis
- d. The second twin probably has hydramnios of the amniotic sac
- e. The second twin is likely to be pale, tachycardic, and hypotensive

**153.** Parents bring a 5-day-old infant to your office. The mother is O negative and was Coombs positive at delivery. The term child weighed 3055 g at birth and had a baseline hemoglobin (16 gm/dL) and a total serum bilirubin (3 mg/dL). He passed a black tarlike stool within the first 24 h of life. He was discharged at 30 h of life with a stable axillary temperature of 36.5°C (97.7°F). Today the infant's weight is 3000 g, his axillary temperature is 35°C (95°F), and he is jaundiced to the chest. Parents report frequent yellow, seedy stool. You redraw labs and find his hemoglobin is now 14 gm/dL, and his total serum bilirubin is 13 mg/dL. The change in which of the following parameters is of most concern?

- a. Hemoglobin
- b. Temperature
- c. Body weight
- d. Bilirubin
- e. Stool

**154.** A couple expecting their first infant in a few weeks scheduled an appointment in your pediatric clinic to get a head start on child care issues (a prenatal visit). You ask about the pregnancy course, and the mother notes that she was recently told she had oligohydramnios. At delivery of this infant, you plan to evaluate for which of the following conditions?

- a. Anencephaly
- b. Trisomy 18
- c. Renal agenesis
- d. Duodenal atresia
- e. Tracheoesophageal fistula

**155.** A newborn infant becomes markedly jaundiced on the second day of life, and a faint petechial eruption first noted at birth is now a generalized purpuric rash. Hematologic studies for hemolytic diseases are negative. Acute management should include which of the following steps?

- a. Liver ultrasound
- b. Isolation of the infant from pregnant hospital personnel
- c. Urine drug screen on the infant
- d. Discharge with an early follow-up visit in 2 days to recheck bilirubin
- e. Thyroid hormone assay

**DIRECTIONS:** Each group of questions below consists of lettered options followed by a set of numbered items. For each numbered item, select the appropriate lettered option(s). Each lettered option may be used once, more than once, or not at all. **Choose exactly the number of options indicated following each item.**

**Items 156–159**

The response options for items 156–159 are the same. Select one answer for each item in the set.

- a. Needle aspiration of the chest
- b. Extracorporeal membrane oxygenation
- c. Nebulized epinephrine
- d. Nebulized steroids
- e. Systemic steroids
- f. Chest physiotherapy
- g. Emergent intubation
- h. Chest radiograph, two views
- i. Respiratory isolation (droplet precautions)

**156.** A 1-month-old infant presents to the clinic in the month of December with a several-day history of cough and rhinorrhea, and has now developed difficulty breathing. On examination, the child has marked intercostal and subcostal retractions. There are both crackles and wheezes diffusely on auscultation, and the infant is mildly hypoxic. The child's radiograph displays a diffuse interstitial infiltrate with some air trapping. The child has been placed on oxygen. A rapid RSV antigen test is positive. What is the next step in initial management? (**SELECT 1 ANSWER**)

**157.** An infant born an hour ago to a mother with severe pregnancy-induced hypertension was floppy at birth and required bag-mask ventilation for several minutes. The infant is now in distress and seemingly agitated, with tachypnea, tachycardia, and hypoxia. Perfusion is poor. Diminished breath sounds are noted on the left side of her chest, and her heart sounds are displaced to the right. What is the best initial step in management? (**SELECT 1 ANSWER**)

**158.** A 3-week-old infant has a 1-week history of a mild cough with thick nasal secretions. In the last 12 h she has developed spasmodic coughing fits during which she becomes cyanotic. When not coughing, she appears tired. She has lost weight since the 2-week visit. The family has not taken her temperature, but she has been feeding less and has not voided in about 16 h. You decide to admit her for observation and rehydration. What is the most appropriate management consideration for this patient? (**SELECT 1 ANSWER**)

**159.** A 2-day-old infant with meconium aspiration syndrome is worsening. The delivered  $\text{FiO}_2$  is 100%, and yet his arterial  $\text{PaO}_2$  is 40 mmHg on the most recent arterial blood-gas analysis. You have increased his ventilator pressures without success. What is the next step in this patient's management? (**SELECT 1 ANSWER**)

### **Items 160–163**

The response options for items 160–163 are the same. Select one answer for each item in the set.

- a. Bronchopulmonary dysplasia
- b. Respiratory distress syndrome (hyaline membrane disease)
- c. Pulmonary interstitial emphysema
- d. Bronchiolitis
- e. Primary pulmonary hypertension
- f. Pneumothorax
- g. Asthma
- h. Meconium aspiration
- i. Transient tachypnea of the newborn
- j. Bacterial pneumonia

**160.** A term infant delivered via scheduled cesarean section develops, at 15 min of age, tachypnea, grunting, flaring, and retractions. A chest radiograph reveals well-aerated lungs with fluid in the fissure on the right, prominent pulmonary vascular markings, and flat diaphragms. The child is mildly hypoxic on room air with 89% oxygen saturation. Over the next 6 h she improves and no longer requires oxygen. What is the diagnosis? **(SELECT 1 ANSWER)**

**161.** A postterm infant is born at home after a prolonged and difficult labor. The maternal grandmother brings the infant to the hospital at 1 h of life because of fast breathing. Grandmother notes that the child spit up some dark brown particulate fluid shortly after birth. Physical examination reveals an infant in marked respiratory distress. Other findings include both an umbilical cord and flaking skin with a yellow-green hue. Chest radiograph reveals patchy infiltrates bilaterally. What is the diagnosis? **(SELECT 1 ANSWER)**

**162.** An infant of uncertain dates is born via emergent cesarean section after the mother was critically injured in a motor vehicle accident. Birth weight was 1075 g. The infant has poor respiratory effort and you begin bag-mask ventilation but find it extremely difficult to cause chest wall movement. A chest radiograph reveals diffuse whiteout of both lungs, with an occasional air bronchogram. What is the diagnosis? **(SELECT 1 ANSWER)**

**163.** The same infant is now 7 weeks old. She has been extubated for 2 weeks and still requires oxygen to maintain her saturation above 93%. Her chest radiograph now reveals patchy, fluffy infiltrates with areas of lucency. She requires daily diuretic treatment. What is the diagnosis? **(SELECT 1 ANSWER)**

**Items 164–167**

Blood samples of a 3-day-old full-term infant are sent for screening to identify diseases that would have serious, permanent consequences without prompt and appropriate treatment. Match the disease with the treatment.

- a. Special diet
- b. Hormone therapy
- c. Vitamin therapy
- d. Antibiotic prophylaxis
- e. Sunlight

**164.** Galactosemia (**SELECT 1 TREATMENT**)

**165.** Phenylketonuria (**SELECT 1 TREATMENT**)

**166.** Biotinidase deficiency (**SELECT 1 TREATMENT**)

**167.** Hypothyroidism (**SELECT 1 TREATMENT**)

**Items 168–171**

For each of the following descriptions of a patient with a congenital anomaly, select the major abnormality with which it is most likely to be associated.

- a. Deafness
- b. Seizures
- c. Wilms tumor
- d. Congestive heart failure
- e. Optic glioma

**168.** You are asked to see a child in consultation. The family noticed that the child's left arm and leg seemed bigger than those on the right. In addition, the child has aniridia. You examine the family members present, and none of them have aniridia or hemihypertrophy, nor do they know of anyone else in the family with these conditions. You suggest further evaluation of the child for which condition? (**SELECT 1 ABNORMALITY**)

**169.** An infant has fusion of the eyebrows, heterochromic irises, a broad nasal root with lateral displacement of the medial canthi, and a white forelock. Testing should be performed on this child for what associated condition? (**SELECT 1 ABNORMALITY**)

**170.** You are called to examine a newborn with a rash. Upon your arrival to the nursery, you find that the rash is actually a large, flat vascular malformation over the infant's left face and scalp. The mother notes that her other child was born with a capillary hemangioma on his arm and asks if this is the same thing. You explain that this vascular malformation is different, and that you will want to monitor him for which of the listed conditions? (**SELECT 1 ABNORMALITY**)

**171.** A new mother asks you about several birthmarks on her child. She points out several hypopigmented oval macules over the child's trunk and extremities. She notes that these have been present since birth. She is concerned because she had a brother with the same spots who she thinks had "growths" in his brain. You counsel that you are concerned that the infant may develop which of the listed conditions? (**SELECT 1 ABNORMALITY**)

### **Items 172–174**

For each description of head injury that follows, select the major abnormality with which it is most likely to be associated.

- a. Intraventricular hemorrhage
- b. Caput succedaneum
- c. Subdural hemorrhage
- d. Subarachnoid hemorrhage
- e. Cephalohematoma

**172.** A 1-day-old healthy infant with a superficial swelling over the right parietotemporal region that does not cross the suture lines. **(SELECT 1 ABNORMALITY)**

**173.** A 6-month-old comatose infant with multiple broken bones in various stages of healing, bulging anterior fontanelle, and retinal hemorrhages. **(SELECT 1 ABNORMALITY)**

**174.** Previous premature infant born at 27 weeks' gestation and now 6 months of age presenting with macrocephaly and hydrocephalus on ultrasonogram. **(SELECT 1 ABNORMALITY)**

# The Newborn Infant

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## Answers

**108. The answer is d.** (*Behrman, 16/e, pp 328, 441, 531, 1580. McMillan, 3/e, pp 356–358, 1599–1600, 2228, 2230. Rudolph, 21/e, pp 124–126, 527, 732–734, 775, 776, 1699–1700.*) Fetal alcohol syndrome is a preventable cause of birth defects. Prenatal exposure to ethanol is the cause. Findings include microcephaly, small palpebral fissures, short nose, smooth philtrum, thin upper lip, ptosis, microphthalmia, cleft lip and palate, and central nervous system abnormalities including mental retardation (average IQ = 67).

Common findings with trisomy 21 include protruding tongue, Brushfield's spots, redundant neck skin, mental retardation, brachycephaly, upslanting palpebral fissures, epicanthal folds, flat face, small ears, cardiac abnormalities (especially ventricular septal defect or endocardial cushioning defect), palmar creases, and clinodactyly of the fifth digit.

Dilantin exposure causes midface hypoplasia, low nasal bridge, ocular hypertelorism, and accentuated cupid's bow of the upper lip. Other features include cleft lip and palate, growth retardation, mental deficiency, distal phalangeal hyperplasia, cardiovascular anomalies, and skeletal defects.

Alport syndrome is the most common of the hereditary nephritis conditions, frequently leading to end-stage renal disease. In 85% of patients with Alport syndrome, an X-linked dominant form of inheritance is found; about 15% are autosomal recessive. All cause hematuria and progressive nephritis. Other findings include deafness and ocular defects.

Infants of diabetic mothers have an increased chance of congenital heart disease, caudal regression syndrome, and a small left colon; are large for gestation age; and have a number of biochemical abnormalities such as hypoglycemia or hypocalcemia.

**109. The answer is d.** (*Behrman, 16/e, pp 513–517. McMillan, 3/e, 197–206. Rudolph, 21/e, pp 164–169.*) Significant unconjugated serum bilirubin levels in full-term newborn infants can lead to diffusion of bilirubin into brain tissue and to neurologic damage. Sulfisoxazole and other drugs compete with bilirubin for binding sites on albumin; therefore, the presence of these drugs can cause dislocation, not increased affinity, of

bilirubin to tissues. Metabolic acidosis also reduces binding of bilirubin, and neonatal sepsis interrupts the blood-brain barrier, thus allowing diffusion of bilirubin into the brain. Administration of phenobarbital has been used to induce glucuronyl transferase in newborn infants and can reduce, rather than exacerbate, neonatal jaundice. Other factors that reduce the amount of unconjugated bilirubin bound to albumin (and therefore cause an increase in free unconjugated bilirubin) include hypoalbuminemia and certain compounds (e.g., nonesterified fatty acids, which are elevated during cold stress) that compete with bilirubin for albumin binding sites.

**110. The answer is c.** (*Behrman, 16/e, pp 495, 1258–1259. Rudolph, 21/e, p 1260.*) It is important to make the diagnosis of choanal atresia quickly because it responds to treatment but can be lethal if unrecognized and untreated. Most neonates are obligate nose breathers because they cannot breathe adequately through their mouths. Infants with choanal atresia have increased breathing difficulty during feeding and sleeping and improved respirations when crying. A variety of temporizing measures to maintain an open airway have been used, including oropharyngeal airways, positioning, tongue fixation, and endotracheal intubation, but surgical correction with placement of nasal tubes is most effective. The diagnosis can be made by failure to pass a catheter through the nose to the pharynx.

**111. The answer is e.** (*Behrman, 16/e, pp 974, 977, 1088. McMillan, 3/e, pp 436–437. Rudolph, 21/e, pp 1043, 1224.*) Varicella-zoster immunoglobulin (VZIG) should be administered to the infant immediately after delivery if the mother had the onset of varicella within 5 days prior to delivery, and immediately upon diagnosis if her chickenpox started within 2 days after delivery. If untreated, about half of these infants will develop serious varicella as early as 1 day of age. If a normal full-term newborn is exposed to chickenpox 2 or more days postnatally, VZIG and isolation are not necessary because these babies appear to be at no greater risk for complications than older children. Acyclovir is not approved for use in early infancy.

**112. The answer is b.** (*Behrman, 16/e, p 164. McMillan, 3/e, p 470. Rudolph, 21/e, p 31.*) A normal 2-week-old infant has a basal caloric expenditure of about 65 cal/(kg·d). Adding calories for activity and growth brings the caloric requirement to about 110 cal/kg. A 3.5-kg infant, therefore, requires about 385 cal in the diet per day. Standard infant formulas

have a caloric density of 0.67 cal/mL, so it would take about 575 mL of infant formula per day to supply adequate calories for this infant. For the infant in question, 60 mL four times a day (240 mL per day) is less than half of the amount that the infant needs and should be of concern. The other factors in the question are normal for the infant's age.

**113. The answer is a.** (*Behrman, 16/e, pp 493–495. McMillan, 3/e, pp 179–180, 235–237. Rudolph, 21/e, pp 98–99.*) The low Apgar scores, meconium staining, and ensuing respiratory distress suggest that asphyxia has occurred. During a period of asphyxia, the resulting hypoxemia, acidosis, and poor perfusion can damage a neonate's brain, heart, kidney, liver, and lungs. The resulting clinical abnormalities include cerebral edema, irritability, seizures, cardiomegaly, heart failure, renal failure, poor liver function, disseminated intravascular coagulopathy, and respiratory distress syndrome. There can be excessively high pulmonary arterial pressure at the same time systemic blood pressure begins to fall, resulting in a persistent right-to-left shunt across a patent ductus arteriosus or foramen ovale.

**114. The answer is a.** (*Behrman, 16/e, pp 981–983. McMillan, 3/e, pp 429–433. Rudolph, 21/e, pp 1031–1035.*) Cytomegalic inclusion disease is the most common congenital viral infection. In the United States, 20 to 90% of women of childbearing age have serologic evidence of a past infection with CMV. Symptomatic congenital disease usually occurs when a mother has a primary CMV infection in the first trimester of pregnancy. Many of these babies die, and those who survive are severely affected. In the event of reactivation of CMV infection during pregnancy, maternal IgG, passed transplacentally, protects the infant from serious infection. Although most infants infected during this secondary maternal infection are asymptomatic, about 10% of them eventually manifest hearing and neurologic problems. Some recommend keeping a child with congenital CMV infection away from susceptible pregnant (or about to become pregnant) women because CMV excretion can persist for months to years; at the very least, good hand washing practices should be instituted. If infected shortly after birth, the younger sibling will probably be asymptomatic since he or she has maternal IgG in the circulation. CMV is primarily an occult infection. Of toddlers in day-care centers, 20 to 80% acquire CMV and shed it in saliva and urine for years. The “gold,” or criterion, standard for diagnosis is a urine viral culture for CMV.

**115. The answer is b.** (*Behrman, 16/e, pp 526–527, 1101. McMillan, 3/e, p 360. Rudolph, 21/e, p 1372.*) Hematemesis and melena are not uncommon in the neonatal period, especially if gross placental bleeding has occurred at the time of delivery. The diagnostic procedure that should be done first is the Apt test, which differentiates fetal from adult hemoglobin in a bloody specimen. If the blood in an affected infant's gastric contents or stool is maternal in origin, further workup of the infant is obviated.

**116. The answer is c.** (*Behrman, 16/e, p 460. McMillan, 3/e, p 477.*) Most medications are secreted to some extent in breast milk. Some lipid-soluble medications may be concentrated in breast milk. Although the list of contraindicated medications is short, caution should always be exercised when giving a medication to a breast-feeding woman. Medications that are clearly contraindicated include lithium, cyclosporin, antineoplastic agents, illicit drugs including cocaine and heroin, ergotamines, and bromocriptine (which suppresses lactation). Although some suggest that oral contraceptives may have a negative impact on milk production, the association has not been proven conclusively. In general, antibiotics are safe, with only a few exceptions. While sedatives and narcotic pain medications are probably safe, the infant must be observed carefully for sedation. All of the medications listed in the question are considered safe, except for lithium.

**117. The answer is d.** (*Behrman, 16/e, pp 512–513. McMillan, 3/e, pp 325–332. Rudolph, 21/e, pp 140–143.*) The infant presented in the question has necrotizing enterocolitis (NEC), a potentially life-threatening disease of the neonate. It is more common in premature infants, but has been described in term infants as well. Although several organisms have been isolated from NEC patients, no clear cause for this condition has been identified. Patients present with feeding intolerance and a distended abdomen. About a quarter have grossly bloody stool. Pneumatosis intestinalis is found on plain radiograph of the abdomen and is diagnostic for NEC in this age group. Management depends initially on perforation; if there is no evidence of free peritoneal air, the infant should be put on bowel rest with nasogastric decompression, and systemic antibiotics are initiated. Electrolytes and vital signs should be monitored closely, and serial abdominal films should be performed to evaluate for perforation. If free air is identified on plain radiographs or if the infant clinically worsens with medical management, surgical consultation is required. An exploratory laparotomy

is usually performed, and any necrotic gut is removed. Occasionally, removal of necrotic gut will result in an infant without adequate intestinal surface area to absorb nutrition, a condition known as *short bowel syndrome*.

**118. The answer is b.** (*Behrman, 16/e, pp 480–482. McMillan, 3/e, pp 146–148.*) It is usually impossible with any combination of parenteral and enteral nutrition to match what the infant would have accumulated in utero. The average, healthy, low-birth-weight infant of this size requires a daily intake of calcium of about 200 mg/kg. Breast milk has much less calcium (and phosphorus) than do commercial formulas. The breast milk can be supplemented with calcium, or it can be mixed with commercial formulas designed for the premature infant. Breast milk promotes gut maturation and prevents intestinal atrophy induced by lack of enteral feeding. Breast milk, however, is likely to have insufficient calcium and phosphorus for catch-up growth.

**119. The answer is d.** (*McMillan, 3/e, p 218. Rudolph, 21/e, pp 84–85, 123.*) A room temperature of 24°C (approximately 75°F) provides a cold environment for newborn infants. Aside from the fact that these infants emerge from a warm, 37.6°C (99.5°F) intrauterine environment, at birth, infants (and especially preterm infants) are wet, have a relatively large surface area for their weight, and have little subcutaneous fat. Within minutes of delivery, the infants are likely to become pale or blue and their body temperatures will drop. In order to bring body temperature back to normal, they must increase their metabolic rate; ventilation, in turn, must increase proportionally to ensure an adequate oxygen supply. Because a preterm infant is likely to have respiratory problems and be unable to oxygenate adequately, lactate can accumulate and lead to a metabolic acidosis. Infants rarely shiver in response to a need to increase heat production.

**120. The answer is b.** (*Behrman, 16/e, pp 521–525. McMillan, 3/e, p 361. Rudolph, 21/e, pp 177, 1531–1532.*) If a mother is O positive and her baby is A positive, the baby has a chance of developing hemolytic disease. Hemolytic disease and jaundice caused by a major blood-group incompatibility are usually less severe than with Rh incompatibility. Although the hematocrit of affected infants usually is normal, elevation of the reticulocyte count and the presence of nucleated red blood cells and microspherocytes in the blood smear provide evidence of hemolysis. In comparison

with hemolytic disease caused by Rh incompatibility, where it is usually strongly positive, major blood-group incompatibility is often associated with a direct Coombs test that is frequently weakly positive. Petechiae are usually associated with decreased number of platelets. Crescent-shaped (sickled) red blood cells are found with sickle cell disease.

**121. The answer is a.** (*Behrman, 16/e, pp 475–477. McMillan, 3/e, pp 206–209. Rudolph, 21/e, pp 122, 2022–2023.*) Small-for-dates infants are subject to a different set of complications than preterm infants whose size is appropriate for gestational age. The small-for-dates infants have a higher incidence of major congenital anomalies and are at increased risk for future growth retardation, especially if length and head circumference as well as weight are small for gestational age. Also more common are neonatal asphyxia and the meconium aspiration syndrome, which can lead to pneumothorax, pneumomediastinum, or pulmonary hemorrhage. These, rather than hyaline membrane disease, are the major pulmonary problems in these infants. Because neonatal symptomatic hypoglycemia is more commonly found in small-for-dates infants, careful blood glucose monitoring and early feeding are appropriate precautions. Normal or elevated hematocrit is also more common in these infants.

**122. The answer is a.** (*Behrman, 16/e, pp 497–498. McMillan, 3/e, pp 265–266. Rudolph, 21/e, p 1934.*) Apneic episodes are characterized by an absence of respirations for more than 20 s and may be accompanied by bradycardia and cyanosis. A large number of conditions can cause apnea. Periods of apnea are generally thought to be secondary to an incompletely developed respiratory center, particularly when they are seen, as is common, associated with prematurity. Although seizures, hypoglycemia, and pulmonary disease accompanied by hypoxia can lead to apnea, these causes are less likely in the infant described, given that no unusual movements occur during the apneic spells, that the blood sugar level is more than 40 mg/dL, and that the child appears well between episodes. Periodic breathing, a common pattern of respiration in low-birth-weight babies, is characterized by recurrent breathing pauses of 3 to 10 s.

**123–124. The answers are 123-c, 124-c.** (*Behrman, 16/e, pp 1698–1703. McMillan, 3/e, pp 1806–1809. Rudolph, 21/e, pp 2065–2070.*) The main thrust of both questions is directed at the need for speed in the diagnosis and treat-

ment of congenital hypothyroidism because the earlier treatment is started with thyroid hormone, the better the prognosis for intellectual function. Time should not be spent in exhaustive investigation. Regardless of the reason for the hypothyroidism, treatment with replacement thyroid hormone is indicated. If it turns out that the initial diagnosis was erroneous, little harm will be done by treating an infant with a physiologic dose of thyroid hormone for a few days. Waiting for laboratory tests or x-rays to be performed, interpreted, and probably repeated is inappropriate if this will delay treatment. Eventually, they should be done, along with an evaluation of the mother's immune status, her health history, and a complete family history looking for one of the many known, although relatively rare, causes of congenital hypothyroidism.

Thyroid dysgenesis is found in 90% of the cases. Neonatal screening for hypothyroidism has allowed for the much earlier diagnosis of hypothyroidism, resulting in improvement of prognosis, so that frank cretinism is now quite rare. Most industrialized countries test for phenylketonuria and hypothyroidism; there is variability in testing for other metabolic and genetic diseases.

**125–126. The answers are 125-c, 126-c.** (*Behrman, 16/e, p 491. McMillan, 3/e, pp 164, 2122. Rudolph, 21/e, p 2447.*) In a difficult delivery in which traction is applied to the head and neck, several injuries, including all those listed in the question, may occur. Erb-Duchenne paralysis affects the fifth and sixth cervical nerves; the affected arm cannot be abducted or externally rotated at the shoulder, and the forearm cannot be supinated. Injury to the seventh and eighth cervical and first thoracic nerves (Klumpke paralysis) results in palsy of the hand and also can produce Horner's syndrome. Fractures in the upper limb are not associated with a characteristic posture, and passive movement usually elicits pain. Spinal injury causes complete paralysis below the level of injury.

When paralysis of an upper extremity from injury to the brachial plexus is found in a neonate, injury to the phrenic nerve should also be suspected because the nerve roots are close together and can be injured concurrently. The paralyzed diaphragm can be noted to remain elevated on a chest x-ray taken during deep inspiration when it will contrast with the opposite normal diaphragm in its lower normal position; on expiration, this asymmetry cannot be seen. On inspiration, not only is breathing impaired since the paralyzed diaphragm does not contract, but the negative

pressure generated by the intact diaphragm pulls the mediastinum toward the normal side, impairing ventilation further. The diagnosis can easily be made by fluoroscopy, where these characteristic movements on inspiration and expiration can be seen. Rarely, both diaphragms can be paralyzed, producing much more severe ventilatory impairment. Fortunately, these injuries frequently improve spontaneously.

**127. The answer is a.** (*Behrman, 16/e, pp 525–526. McMillan, 3/e, pp 240–245, 362–363. Rudolph, 21/e, pp 192–193.*) An infant of 2100 g at 38 weeks would be considered small for gestational age (SGA), a not uncommon consequence of maternal toxemia. Pregnancy-induced hypertension can produce a decrease in uteroplacental blood flow and areas of placental infarction. This can result in fetal nutritional deprivation and intermittent fetal hypoxemia, with a decrease in glycogen storage and a relative erythrocytosis, respectively. Hence, neonatal hypoglycemia and polycythemia are common clinical findings in these infants. A blood glucose level of 30 mg/dL in a full-term infant, however, is probably normal during the first postnatal day, and an infant is very unlikely to have a convulsion as a result of a level of 38 mg. Serum calcium levels usually decline during the first 2 to 3 postnatal days, but will only be considered abnormally low in a term infant when they fall below 7.5 to 8 mg/dL. Neonatal hypermagnesemia is common in an infant whose mother has received  $MgSO_4$  therapy, but is usually asymptomatic or produces decreased muscle tone or floppiness. A persistent venous hematocrit of greater than 65% in a neonate is regarded as polycythemia and will be accompanied by an increase in blood viscosity. Manifestations of the “hyperviscosity syndrome” include tremulousness or jitteriness that can progress to seizure activity because of sludging of blood in the cerebral microcirculation or frank thrombus formation, renal vein thrombosis, necrotizing enterocolitis, and tachypnea. Therapy by partial exchange transfusion with albumin is probably more likely to be useful if performed prophylactically before significant symptoms have developed.

**128. The answer is b.** (*Behrman, 16/e, p 495. McMillan, 3/e, pp 173–178. Rudolph, 21/e, pp 238–243.*) In the description provided, the most likely cause of the neonatal depression is maternal analgesic narcotic drug administration. While controlling the pain of the delivery in the mother, use of narcotics can result in depression of the newborn via crossing of the placenta. Appropriate first steps in the management of this infant (after

managing the ABCs of airway, breathing, and circulation) is the administration of naloxone, 0.1 mg/kg, IM, IV, or intratracheal. The other possibilities are unlikely, given the clinical information provided.

**129. The answer is a.** (*Behrman, 16/e, pp 505–506. McMillan, 3/e, pp 173–178. Rudolph, 21/e, pp 238–243.*) Infants who are postmature (more than 42 weeks' gestation) and show evidence of chronic placental insufficiency (low birth weight for gestational age and wasted appearance) have a higher-than-average chance of being asphyxiated, and passage of meconium into the amniotic fluid thus places these infants at risk for meconium aspiration. To prevent or minimize this risk, these infants should have immediate nasopharyngeal suction as their heads are delivered. Immediately after delivery and before initiation of respiration, their tracheas should be carefully and thoroughly suctioned through an endotracheal tube under direct vision with a laryngoscope. Afterward, appropriate resuscitative measures should be undertaken to establish adequate ventilation and circulation. Artificial ventilation performed before tracheal suction could force meconium into smaller airways.

**130. The answer is d.** (*Behrman, 16/e, pp 1911–1914. McMillan, 3/e, pp 668–669. Rudolph, 21/e, p 2370.*) The time of onset of symptoms is somewhat helpful in the diagnosis of ophthalmia neonatorum. Chemical conjunctivitis is a self-limited condition that presents within 6 to 12 h of birth as a consequence of silver nitrate or erythromycin prophylaxis. Silver nitrate is no longer made for ocular prophylaxis in the United States. Gonococcal conjunctivitis has its onset within 2 to 5 days after birth and is the most serious of the bacterial infections. Prompt and aggressive topical treatment and systemic antibiotics are indicated to prevent serious complications such as corneal ulceration, perforation, and resulting blindness. Parents should be treated to avoid the risk to the child of reinfection. Silver nitrate is believed by some to be ineffective prophylaxis against chlamydial conjunctivitis, which occurs 5 to 14 days after birth. To avoid the risk of chlamydial pneumonia, treatment with systemic antibiotics is indicated for the infant as well as both parents in cases of chlamydial conjunctivitis.

**131. The answer is b.** (*Behrman, 16/e, pp 498–505. McMillan, 3/e, pp 254–258. Rudolph, 21/e, pp 127–134.*) For the child described in the question, prematurity and the clinical picture presented make the diagnosis of

hyaline membrane disease (infant respiratory distress syndrome) likely. In this disease, lung compliance is reduced; lung volume also is reduced, and a significant right-to-left shunt of blood can occur. Some of the shunt can result from a patent ductus arteriosus or foramen ovale, and some can be due to shunting in the lung. Minute ventilation is higher than normal, and affected infants must work harder in order to sustain adequate breathing.

**132. The answer is c.** (*Behrman, 16/e, pp 1028, 1031. McMillan, 3/e, pp 872, 874. Rudolph, 21/e, pp 1046, 1052.*) The transmission of HIV from mother to infant has decreased in recent years, due in large part to perinatal administration of antiretroviral medications to the mother and a course of zidovudine to the exposed infant. Studies suggest that a better than 50% decrease in transmission can be seen with appropriate medications as outlined. An ELISA is an antibody test and will be positive in the infant born to an HIV-infected mother due to maternal antibodies that are passed through the placenta; it is not a useful test in the newborn infant to determine neonatal infection. IVIG has not been shown to have a role in decreasing perinatal transmission. Healthy asymptomatic term infants born to HIV-infected mothers do not need special monitoring nor do they need routine radiographs.

**133. The answer is c.** (*Behrman, 16/e, pp 1753–1755. McMillan, 3/e, pp 1775, 2231. Rudolph, 21/e, pp 2087–2089.*) Turner syndrome is a genetic disorder; the 45,XO karyotype is the most common. At birth, affected infants have low weights, short stature, edema over the dorsum of the hands and feet, and loose skin folds at the nape of the neck. Some other findings with this syndrome include sexual infantilism, streak gonads, typical faces, shield chest, low hairline, coarctation of the aorta, hypertension, bicuspid aortic valve, high palate, and horseshoe kidney. Coarse, tremulous movements accompanied by ankle clonus; vascular instability as evidenced, for example, by a harlequin color change (a transient, longitudinal division of a body into red and pale halves); softness of parietal bones at the vertex (craniotabes); and a liver that is palpable down to 2 cm below the costal margin are all findings often demonstrated by normal infants and are of no diagnostic significance in the clinical situation presented.

**134. The answer is b.** (*Behrman, 16/e, pp 455, 1970. McMillan, 3/e, pp 372, 682. Rudolph, 21/e, p 1192.*) Mongolian spots are bluish-gray lesions located

over the buttocks, lower back, and occasionally, the extensor surfaces of the extremities. They are common in blacks, Asians, and Latin Americans. They tend to disappear by 1 to 2 years of age, although those on the extremities may not fully resolve. Child abuse is unlikely to present with bruises alone; children frequently present with more extensive injuries. Subcutaneous fat necrosis is usually found as a sharply demarcated, hard lesion on the cheeks, buttocks, and limbs. The lesion usually is red. Hemophilia and vitamin K deficiency rarely present with subcutaneous lesions as described and are more likely to present as a bleeding episode.

**135. The answer is c.** (*Behrman, 16/e, pp 1231–1233. McMillan, 3/e, pp 177, 217–218, 222. Rudolph, 21/e, pp 189–191.*) Diaphragmatic hernia occurs with the transmittal of abdominal contents across a congenital or traumatic defect in the diaphragm. In the newborn, this condition results in profound respiratory distress with significant mortality. Prenatal diagnosis is common and, when found, necessitates that the birth take place at a tertiary level center. In the neonate, respiratory failure in the first hours of life, a scaphoid abdomen, and presence of bowel sounds in the chest are common findings. Intensive respiratory support including mechanical ventilation and extracorporeal membrane oxygenation (ECMO) has increased survival. Mortality can be as high as 50% despite aggressive treatment.

**136. The answer is d.** (*Behrman, 16/e, pp 1122–1123. McMillan, 3/e, pp 309–310. Rudolph, 21/e, pp 1385–1387.*) Abdominal distention, choking, drooling, and coughing associated with feedings are symptoms of esophageal anomalies. The anomaly illustrated by **D** is the most common; that of **A** can be diagnosed after repeated episodes of pneumonia. The anomalies in **E** and **C** are associated with all the same symptoms except abdominal distention, which cannot develop because air cannot enter the gastrointestinal tract. **B** and **C** are the least common; in these, the upper esophageal segment is connected directly to the trachea, and massive entry of fluid into the lungs occurs.

VATER syndrome, a complex of cardiovascular malformations, skeletal malformations, and renal abnormalities, has tracheoesophageal fistula as a common finding.

**137. The answer is b.** (*Behrman, 16/e, p 526. McMillan, 3/e, pp 367, 1487–1488. Rudolph, 21/e, pp 1574–1575.*) Failure to give vitamin K pro-

phylactically to newborn infants is associated with a decline in the levels of vitamin K-dependent coagulation factors. In less than 1% of infants (but especially those fed human breast milk), the levels reached are low enough to produce classic hemorrhagic manifestations on the second to seventh day of life. These manifestations include melena, hematuria, and bleeding from the circumcision; intracranial hemorrhage and hypovolemic shock are serious complications. Diagnosis of this condition is indicated by a prolonged prothrombin time, which reflects inadequate concentrations of factors II, VII, IX, and X.

**138. The answer is d.** (*Behrman, 16/e, pp 771–773, 775–776. McMillan, 3/e, pp 447–449, 1724–1725. Rudolph, 21/e, pp 49–50.*) The infant of a mother who is a carrier of hepatitis B surface antigen has a significant risk of acquiring infection. This usually occurs at the time of delivery, but infection can also be acquired during pregnancy and postnatally. A small percentage of infected neonates develop acute icteric hepatitis, but the majority remain asymptomatic. Of these infected asymptomatic infants, 80% or more will develop chronic antigenemia, the long-term consequences of which are chronic liver disease and, possibly, hepatocellular carcinoma. Combined passive-active immunoprophylaxis in the form of immune globulin and hepatitis B vaccine affords protection not only from immediate perinatal infection but also from infection that may be acquired as a result of continued exposure in the household of a chronic carrier.

Immunization in this infant is indicated regardless of the presence of hepatitis B “e” antigen (HBeAg) in the mother. Although the presence of HBeAg, especially in the absence of antibody to HBeAg, is associated with high rates of transmission to neonates, any woman positive for hepatitis B surface antigen (HbsAg) is potentially infectious. It is not necessary to isolate infants born to carriers of HBsAg, and screening of neonates for HBsAg is not indicated. Testing for HBsAg and anti-HBsAg at least 1 month after the third dose of hepatitis B vaccine will determine the efficacy of these measures.

**139. The answer is a.** (*Behrman, 16/e, pp 467–469. McMillan, 3/e, pp 137–139.*) The effect of a drug on the fetus is determined by the nature of the drug and by the timing and degree of exposure. Heparin does not cross the placental barrier and does not appear to directly affect the fetus once

pregnancy is well established. Phenytoin may cause birth defects when given during the first trimester. Penicillin and aluminum hydroxide have not been found to affect the fetus. Propranolol, which may cause growth retardation when given throughout pregnancy, diminishes the ability of an asphyxiated infant to increase heart rate and cardiac output. It has also been associated with hypoglycemia and apnea.

**140. The answer is d.** (*Behrman, 16/e, pp 1133–1134. McMillan, 3/e, p 312. Rudolph, 21/e, pp 1403–1404.*) The findings of polyhydramnios signal high intestinal obstruction, signs of which include abdominal distention and early and repeated regurgitation. Distention usually is not present, as vomiting keeps the intestine decompressed. The bile-stained vomitus of the infant places the obstruction distal to the ampulla of Vater, eliminating esophageal atresia and pyloric stenosis from consideration. The “double bubble” sign on the x-ray is characteristic of duodenal atresia, which is compatible with the history. Midgut volvulus, which may obstruct the bowel in the area of the duodenojejunal junction, most often produces signs after an affected infant is 3 or 4 days old with acute onset of bilious vomitus. Gastric duplication does not usually produce intestinal obstruction; a cystic mass may be palpated on abdominal examination. Patients with duodenal atresia should be examined closely for evidence of other conditions such as Down syndrome or heart disease.

**141–142. The answers are 141-c, 142-b.** (*Behrman, 16/e, pp 513–517. McMillan, 3/e, pp 197–206. Rudolph, 21/e, pp 164–169.*) The development of jaundice in a healthy full-term baby may be considered the result of a normal physiologic process if the time of onset and duration of the jaundice and the pattern of serially determined serum concentrations of bilirubin are in conformity with currently accepted safe criteria. Physiologic jaundice becomes apparent on the second or third day of life, peaks to levels no higher than about 12 mg/dL on the fourth or fifth day, and disappears by the end of the week. The rate of rise is less than 5 mg/dL per 24 h and levels of conjugated bilirubin do not exceed about 1 mg/dL. Concern about neonatal jaundice relates to the risk of the neurotoxic effects of unconjugated bilirubin. The precise level and duration of exposure necessary to produce toxic effects are not known, but bilirubin encephalopathy, or kernicterus, is rare in term infants whose bilirubin level is kept below 18 to 20 mg/dL. Certain

risk factors affecting premature or sick newborns increase their susceptibility to kernicterus at much lower levels of bilirubin. The diagnosis of physiologic jaundice is made by excluding other causes of hyperbilirubinemia by means of history, physical examination, and laboratory determinations. Jaundice appearing in the first 24 h is usually a feature of hemolytic states and is accompanied by an indirect hyperbilirubinemia, reticulocytosis, and evidence of red-cell destruction on smear. In the absence of blood group or Rh incompatibility, congenital hemolytic states (e.g., spherocytic anemia) or G6PD deficiency should be considered. With infection, hemolytic and hepatotoxic factors are reflected in the increased levels of both direct and indirect bilirubin.

Studies should include maternal and infant Rh types and blood groups and Coombs tests to detect blood group or Rh incompatibility and sensitization. Measurements of total and direct bilirubin concentrations help to determine the level of production of bilirubin and the presence of conjugated hyperbilirubinemia. Hematocrit and reticulocyte count provide information as to the degree of hemolysis and anemia, and a complete blood count screens for the possibility of sepsis and the need for cultures. Examination of the blood smear is useful in differentiating common hemolytic disorders. Except for determinations of total and direct bilirubin, tests of liver function are not particularly helpful in establishing the cause of early-onset jaundice. Transient elevations of transaminases (AST and ALT) related to the trauma of delivery and to hypoxia have been noted. Biliary atresia and neonatal hepatitis can be accompanied by elevated levels of transaminase but characteristically present as chronic cholestatic jaundice with mixed hyperbilirubinemia after the first week of life.

**143. The answer is d.** (*Behrman, 16/e, pp 1111–1113. McMillan, 3/e, pp 391–394, 2253, 2256. Rudolph, 21/e, pp 748–753.*) The infant pictured has bilateral cleft lip and palate. This defect occurs in about 4% of the siblings of affected infants; its incidence in the general population is 1 in 1000. Evaluation for other structural and chromosomal abnormalities is indicated. Although affected infants are likely to have feeding problems initially, these problems usually can be overcome by feeding in a propped-up position and using special nipples. Complications include recurrent otitis media and hearing loss as well as speech defects, which may be present despite good anatomic closure. Repair of a cleft lip usually is performed

within the first 2 to 3 months of life; the palate is repaired later, usually between the ages of 6 months and 5 years.

**144. The answer is b.** (*Behrman, 16/e, pp 441, 532–533. McMillan, 3/e, pp 346–347, 356–358. Rudolph, 21/e, pp 124–126, 155–159.*) Glucose loading of the mother will result in fetal hyperglycemia, which causes insulin release and reactive hypoglycemia. Careful medical support of the antepartum woman diminishes the hypertrophy of the fetal islet cells. Careful monitoring of the infant with early feeding or intravenous infusion of glucose can prevent hypoglycemia. A neutral thermal environment diminishes glucose consumption and, therefore, helps with glucose homeostasis.

**145. The answer is e.** (*Behrman, 16/e, p 505. McMillan, 3/e, p 259. Rudolph, 21/e, pp 179–181.*) Transient tachypnea of the newborn is usually seen after a normal vaginal or, especially, after a cesarean delivery. These patients have tachypnea, retractions, grunting, and sometimes, cyanosis. The chest examination is usually normal; the chest radiograph demonstrates prominent pulmonary vascular markings with fluid in the fissures and hyperexpansion (flat diaphragms). Therapy is supportive, with maintenance of normal oxygen saturation. Resolution usually occurs in the first 3 days of life.

**146. The answer is d.** (*Behrman, 16/e, pp 903–907. McMillan, 3/e, pp 438–442. Rudolph, 21/e, pp 1004–1005.*) The clinical presentation of congenital syphilis is varied. Many newborns appear normal at birth and continue to be asymptomatic for the first few weeks or months of life. Most untreated infants will develop a skin lesion, the usual one being an infiltrative, maculopapular peeling rash that is most prominent on the face, palms, and soles. Involvement of the nasal mucous membranes causes rhinitis with a resultant serous and occasionally purulent, blood-tinged discharge (snuffles). This, as well as scrapings from the skin lesions, contains abundant viable treponemes. Hepatosplenomegaly and lymphadenopathy are common, and early jaundice is a manifestation of syphilitic hepatitis. Among the later manifestations, or stigmata, of congenital syphilis is interstitial keratitis, which is an acute inflammation of the cornea that begins in early childhood (most commonly between 6 and 14 years of age). Interstitial keratitis represents the response of the tissue to earlier sensitization. Findings

include marked photophobia, lacrimation, corneal haziness, and eventual scarring.

**147. The answer is b.** (*Behrman, 16/e, p 2093. McMillan, 3/e, p 2123. Rudolph, 21/e, p 2449.*) Fifth-finger polydactyly is 10 times more common in black than in white children. This finding in otherwise healthy black children should raise no special concern. In a white child, careful examination of the cardiac system is warranted.

**148. The answer is d.** (*Behrman, 16/e, pp 1803–1806. McMillan, 3/e, pp 156, 223–224. Rudolph, 21/e, pp 2186–2187.*) Diseases that are due to defects in a single gene are designated as autosomal or X-linked depending on whether the affected gene is located on an autosome or an X chromosome. Genetically determined diseases that are multifactorial in origin (i.e., neural tube defects) do not conform to the Mendelian pattern of inheritance but exhibit a variable outcome that reflects the interaction between a particular genotype and an environment. The relatives of persons with diseases of multifactorial origin have an increased risk of having similar abnormalities. The recurrence risk for most single primary defects of multifactorial inheritance (e.g., neural tube defects) is increased with each child affected. This increased risk forms the basis for assuming that genetic factors play a role in the occurrence of these abnormalities. Other factors, such as race, sex, and ethnic and racial background, influence the frequency with which an abnormality of multifactorial inheritance occurs in relatives. The prenatal diagnosis of neural tube defects (anencephaly and meningomyelocele) can be made by the detection of elevated levels of  $\alpha$ -fetoprotein in the amniotic fluid. To reduce the risk of neural tube defects, it is now recommended that all women capable of becoming pregnant take 4 mg of folic acid daily.

**149. The answer is c.** (*Behrman, 16/e, p 530. Rudolph, 21/e, p 2196.*) Infants born to narcotic addicts are more likely than other children to exhibit a variety of problems, including perinatal complications, prematurity, and low birth weight. The onset of withdrawal commonly occurs during an infant's first 2 days of life and is characterized by hyperirritability and coarse tremors, along with vomiting, diarrhea, fever, high-pitched cry, and hyperventilation; seizures and respiratory depression are less common. The production of surfactant can be accelerated in the infant of a heroin-addicted mother.

**150. The answer is b.** (*Behrman, 16/e, pp 486, 497–498. McMillan, 3/e, pp 265–266. Rudolph, 21/e, pp 1934–1936.*) Idiopathic apnea is common in premature infants but is not expected in the full-term newborn. When apnea occurs in the term infant, there is almost always an identifiable cause. Sepsis, gastroesophageal reflux, congenital heart disease, seizures, hypoglycemia, and airway obstruction can cause apnea in term newborns. Harlequin syndrome is a transient change in the skin color of the otherwise asymptomatic newborn (usually preterm) in which the dependent side of the entire body turns red while the upper side remains pale.

**151. The answer is d.** (*Behrman, 16/e, pp 544–546, 548, 753, 811–814. McMillan, 3/e, pp 406–407, 858. Rudolph, 21/e, pp 106–107.*) Neonatal sepsis, a clinical syndrome of systemic illness accompanied by bacteremia, often results in spread of infection to the meninges and other distant sites. The diagnosis of serious infection, including meningitis, in a neonate is difficult because the signs and symptoms are subtle and nonspecific. They include lethargy; feeding problems including abdominal distention, vomiting, and diarrhea; temperature instability; respiratory distress or apnea; and jaundice. Nuchal rigidity and Kernig and Brudzinski signs are frequently not present in the neonate with meningitis.

**152. The answer is b.** (*Behrman, 16/e, pp 475–477. McMillan, 3/e, pp 362–363. Rudolph, 21/e, pp 63, 68, 118–119.*) Twin-to-twin transfusions occur in about 15% of monochorionic twins and commonly cause intrauterine death. This disorder should be suspected when the hematocrits of twins differ by more than 15 mg/dL. The donor twin is likely to have oligohydramnios, anemia, and hypovolemia with evidence of shock if the hematocrit is significantly reduced; the recipient twin is likely to have hydramnios and plethora and to be larger than the donor twin. As the central venous hematocrit rises above 65%, infants can develop hyperviscosity, respiratory distress, hyperbilirubinemia, hypocalcemia, renal vein thrombosis, congestive heart failure, and convulsions.

**153. The answer is b.** (*Behrman, 16/e, pp 32, 458, 516, 1499. McMillan, 3/e, pp 186–187, 192–193, 197, 2224. Rudolph, 21/e, p 207.*) There is loss of body weight of 1.5 to 2% per day for the first 5 days of life for a normal newborn infant as excessive fluid is excreted. This would tend to produce an increase in hematocrit, but, to the contrary, the hematocrit falls as an

adaptation to an environment of higher oxygen. Infants usually have several meconium stools during the first day or two of life, changing to soft yellow stools at 1 to 2 days of life. As the hematocrit falls, there is a corresponding increase in serum bilirubin that peaks around 3 to 5 days of life. Temperature should not change; temperature instability in a term infant is frequently a sign of serious infection.

**154. The answer is c.** (*Behrman, 16/e, p 463. McMillan, 3/e, pp 158, 309, 312, 336. Rudolph, 21/e, pp 191, 1639.*) It is generally presumed that duodenal atresia and tracheoesophageal fistula lead to hydramnios (polyhydramnios) by interference with reabsorption of swallowed amniotic fluid. Hydramnios is also associated with approximately 80% of infants who have trisomy 18. Approximately 50% of women with anencephalic fetuses have polyhydramnios. Oligohydramnios occurs in association with congenital abnormalities of the fetal kidneys, such as renal agenesis, that inhibit formation of fetal urine.

**155. The answer is b.** (*Behrman, 16/e, pp 544–546, 548–549, 981–983, 1698–1703. McMillan, 3/e, pp 197–206, 354–355, 404–416, 429–433, 438–444, 1806–1808. Rudolph, 21/e, pp 106, 1004–1005, 1032–1033, 2065–2070.*) Although hypothyroid neonates may develop hyperbilirubinemia, the patient described most likely has a congenital or acquired infection requiring immediate diagnosis and treatment. Among the important causes of neonatal sepsis are prenatal infections, including congenital syphilis, toxoplasmosis, cytomegalic inclusion disease, and rubella. Useful diagnostic studies, in addition to cultures for bacteria, include specific serologic tests for pathogens, viral cultures, lumbar puncture, and x-rays of the chest and long bones. Longitudinal striations in the metaphyses are characteristic of congenital rubella, whereas osteochondritis or periostitis usually indicates congenital syphilis. Congenital syphilis, cytomegalovirus, and rubella can be highly contagious. Urine can contain rubella virus for more than 6 months and is, therefore, a special hazard to nonimmune pregnant women.

**156–159. The answers are 156-c, 157-a, 158-i, 159-b.** (*Behrman, 16/e, pp 507–508, 838–842, 1250, 1285–1287. McMillan, 3/e, pp 260, 262, 264–265, 990–993, 1214–1216, 1267. Rudolph, 21/e, pp 148–151, 319–320, 973–975, 1984–1985.*) Acute bronchiolitis is usually a viral illness with a

peak incidence at about six months of life. More than half of the cases are caused by respiratory syncytial virus (RSV). Although some children do not respond to any therapy, inhaled epinephrine seems to be more useful than inhaled albuterol. Steroids do not seem to be helpful in children with normal pulmonary function. Bronchiolitis is usually self-limited, but in children with underlying lung disease (such as bronchopulmonary dysplasia), the infection can be severe and sometimes fatal.

A tension pneumothorax occurs when the pressure of free air in the pleural space exceeds that of atmospheric pressure, thereby causing compression of the opposite lung with mediastinal shift. Infants with this condition develop cardiovascular compromise and require emergent needle aspiration of the pneumothorax. When time allows, a chest tube should be placed.

Infants under two months of age are not yet immunized against pertussis, and as such are at increased risk of contracting the disease. Pertussis usually has three distinct stages, starting with the *catarrhal* stage, followed by the *paroxysmal* stage (the infant described is in this stage), and concluding with the *convalescent* stage. The paroxysms can be tiring, and many infants experience posttussive emesis. Pertussis is one of the most communicable diseases known and is spread by droplets aerosolized by coughing. Although children are immunized against pertussis, immunity is now thought to wane in the adult years. Hospitalized patients suspected of having pertussis infection should be isolated with droplet precautions.

Infants in severe respiratory failure from any number of illnesses—such as meconium aspiration, severe pneumonia, persistent fetal circulation, and diaphragmatic hernia—may fail conventional ventilation. Extracorporeal membrane oxygenation (ECMO) is an alternative to conventional ventilation. Essentially the ECMO takes deoxygenated blood, oxygenates it, and sends it back into the infant's circulation. With reversible disease, ECMO can be extremely effective.

**160–163. The answers are 160-i, 161-h, 162-b, 163-a.** (*Behrman, 16/e, pp 498–506. McMillan, 3/e, pp 254–260, 262, 266–269. Rudolph, 21/e, pp 127–134, 179–185, 194–196, 1964–1966.*) Infants born via cesarean section have a higher risk of transient tachypnea of the newborn, a self-limited condition thought to result at least in part from persistent fetal lung fluid. These infants display evidence of respiratory distress shortly after delivery, occasionally requiring oxygen. The radiograph findings are often described

as a “starburst” pattern with diffuse streaky marks that seem to create a star around the mediastinal structures. Over the course of a day or so, the symptoms resolve spontaneously. Differential diagnosis considerations must include pneumonia.

Infants who are under perinatal stress will occasionally pass meconium in utero. The aspiration of this meconium into the lungs can cause a chemical pneumonitis and subsequent respiratory distress. These infants are also at risk for developing primary pulmonary hypertension (persistent fetal circulation); some become quite ill and require extracorporeal membrane oxygenation (ECMO). Meconium stains skin and umbilical cords. Premature infants are usually not at risk for meconium aspiration.

Hyaline membrane disease (respiratory distress syndrome, primary surfactant deficiency) is primarily seen in infants of less than 34 weeks’ gestation. Pulmonary surfactant starts to be secreted around 28 weeks’ gestation; this onset time is remarkably variable, however. Surfactant decreases surface tension in the alveoli and permits inflation of lung tissue at lower pressures. Infants born with immature lung development frequently cannot generate enough negative pressure to inflate lung tissue. Historically these infants required aggressive ventilator support that ultimately resulted in damage to the lung tissue. Since the 1980s, however, exogenous surfactant replacement therapy has been available and is now used routinely in many parts of the world with good results.

Bronchopulmonary dysplasia (BPD) is a chronic lung disease diagnosed in premature infants who have persistent oxygen requirements, clinical findings of respiratory difficulties, and abnormal chest radiographs. These children have airway hyperresponsiveness, and many have pulmonary edema that requires daily diuretic medication. Although the morbidity and mortality rates of children with BPD have improved over the last two decades, these children still have an increased risk of developmental problems and asthma.

**164–167. The answers are 164-a, 165-a, 166-c, 167-b.** (*Behrman, 16/e, pp 344–347, 357, 413, 1698–1703. McMillan, 3/e, pp 354–355, 1829–1830, 1845, 1859. Rudolph, 21/e, pp 609–611, 626, 640–641, 1486, 2065–2070.*) In galactosemia, an enzyme deficiency (galactose-1-phosphate uridylyl transferase) results in a block in the metabolic pathway of galactose and leads to the accumulation of galactose-1-phosphate in the tissues. Infants with this condition develop serious damage to liver, brain, and eyes after being fed

milk containing lactose (a disaccharide compound of glucose and galactose). Clinical manifestations include lethargy, vomiting and diarrhea, hypotonia, hepatomegaly and jaundice, failure to thrive, and cataracts. The course of the disease in untreated patients is variable; death from liver failure and inanition can occur; most untreated patients develop physical and mental retardation. Treatment consists of prompt elimination of lactose-containing milk from the diet in infancy and, as a more varied diet is introduced, exclusion of foods that contain casein, dry milk solids, whey, or curds.

Phenylketonuria, a genetically determined disorder with an autosomal recessive pattern of inheritance, is caused by the absence of an enzyme that metabolizes phenylalanine to tyrosine. The resultant accumulation of phenylalanine and its metabolites in the blood leads to severe mental retardation in untreated patients. Treatment consists of a diet that maintains phenylalanine at levels low enough to prevent brain damage but adequate to support normal physical and mental development. Careful supervision of the low-phenylalanine diet and monitoring of blood levels are necessary. Special formulas are available for the infant; older children have difficulty following the diet. It is not clear when and if the diet can be discontinued.

Biotinidase is the enzyme responsible for breakdown of biocytin (the lysyl precursor of biotin) to free biotin. Deficiency of the enzyme, which is inherited as an autosomal recessive trait, results in malfunctioning of the biotin-dependent mitochondrial enzymes and in organic acidemia. Clinical problems related to the deficiency appear several months or years after birth and include dermatitis, alopecia, ataxia, hypotonia, seizures, developmental delay, deafness, immunodeficiency, and metabolic acidosis. The treatment is lifelong administration of free biotin.

The treatment of congenital hypothyroidism with oral levothyroxine sodium should begin as early as possible to prevent psychomotor retardation. Periodic measurement of  $T_3$ ,  $T_4$ , and TSH is necessary to assess the response to therapy and the need for adjustment of the dose of thyroxine. Careful evaluation of somatic growth by plotting sequential measurements and monitoring bone age is essential.

**168–171. The answers are 168-c, 169-a, 170-b, 171-b.** (Behrman, 16/e, pp 350, 1554–1556, 1837–1838, 1901–1903, 1976, 1986. McMillan, 3/e, pp 714–715, 727–728, 1515–1517, 2023–2025, 2251, 2255. Rudolph, 21/e, pp 770–772, 1185, 1614–1616, 2347–2348.) Aniridia is found in 1 to 2% of children with Wilms tumor. Genitourinary anomalies are found in 4 to 5%, and

hemihypertrophy is associated with this tumor in 2 to 3% of patients. Wilms tumor is the most common primary renal malignancy in childhood. Presentation is usually an abdominal mass, sometimes with hypertension, hematuria, abdominal pain, and fever. Prognosis is generally good.

Waardenburg syndrome is inherited as an autosomal dominant trait with variable penetrance. It includes, in decreasing order of frequency, the following anomalies: lateral displacement of the medial canthi, broad nasal bridge, medial hyperplasia of the eyebrows, partial albinism commonly expressed by a white forelock or heterochromia (or both), and deafness in 20% of cases.

A flat capillary vascular malformation in the distribution of the trigeminal nerve is the basic lesion in the Sturge-Weber syndrome. The malformation also involves the meninges and results in atrophy to the underlying cerebral cortex. The damage is manifested clinically by grand mal seizures, mental deficiency, and hemiparesis or hemianopsia on the contralateral side. The cause is unknown.

Infants who have tuberous sclerosis are often born with hypopigmented oval or irregularly shaped skin macules (ash leaf). Cerebral sclerotic tubers also present from birth and become visible radiographically by the third to fourth year of life. Myoclonic seizures, present in infancy, can convert to grand mal seizures later in childhood. Adenoma sebaceum appears at 4 to 7 years of age. The disease, which also affects the eyes, kidneys, heart, bones, and lungs, is inherited as an autosomal dominant trait with variable expression; new mutations are very common.

**172–174. The answers are 172-e, 173-c, 174-a.** (*Behrman, 16/e, pp 110–114, 488–491, 2107. McMillan, 3/e, pp 163, 230–235, 519–520, 610–611, 614–615. Rudolph, 21/e, pp 144, 186–187, 465, 2193–2195, 2242.*) Subdural hematomas are commonly seen as part of the shaken baby syndrome. This lesion occurs when the bridging cortical veins that drain the cerebral cortex have been ruptured, leading to a collection of blood between the dura and the cerebral mantle. Repeated trauma can lead to additional collections of blood. In many children, additional findings of abuse such as broken bones, bruises, and retinal hemorrhages are found.

Caput succedaneum is soft-tissue swelling of the scalp involving the presenting delivery portion of the head. This lesion is sometimes ecchymotic and can extend across the suture lines. The edema resolves in the first few days of life.

Cephalhematomas do not cross the suture line since they are subperiosteal hemorrhages. No discoloration of the scalp is seen, and the swelling usually progresses over the first few hours of life. Occasionally, skull fractures are present as well. Most cephalhematomas resolve within the first few weeks or months of life without residual findings.

Intraventricular hemorrhage (IVH) is commonly seen in very small, preterm infants. The incidence of IVH increases with smaller-size infants and in those with perinatal complications. It occurs in the gelatinous subependymal germinal matrix of the brain and can lead to progressive posthemorrhagic hydrocephalus. Hydrocephalus in these children can present with enlarging head circumference, apnea and bradycardia, lethargy, bulging fontanelle, widely split sutures, or no signs at all. Therapy can include ventricular-peritoneal shunting.

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# The Cardiovascular System

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## Questions

**DIRECTIONS:** Each item below contains a question or incomplete statement followed by suggested responses. Select the **one best** response to each question.

**175.** A 9-month-old infant accidentally ingests an unknown quantity of digitalis. The most important noncardiac manifestation of toxicity in this infant is

- a. Fever
- b. Dizziness
- c. Vomiting
- d. Visual disturbances
- e. Urticaria

### Items 176–177

**176.** A 10-year-old boy, the star goalie for the Salt Lake City Little League soccer team, had a sore throat about two weeks ago but did not tell anyone because he was afraid he would miss the play-offs. Since several children have been diagnosed with rheumatic fever in the area, his mother is worried that he may be at risk as well. You tell her that several criteria must be met to make the diagnosis but that the most common finding is

- a. Carditis
- b. Arthralgia
- c. Erythema marginatum
- d. Chorea
- e. Subcutaneous nodules

**177.** In the same patient, of the following manifestations of acute rheumatic fever, which is not relieved by salicylate or steroid therapy?

- a. Carditis
- b. Abdominal pain
- c. Arthritis
- d. Chorea
- e. Fever

**178.** The parents of a 2-month-old boy are concerned about his risk of coronary artery disease because of the recent death of his 40-year-old maternal uncle from a myocardial infarction. In managing this situation, you would do which of the following?

- a. Screen the parents for total cholesterol
- b. Counsel the parents regarding appropriate dietary practices for a 2-month-old infant and test him for total cholesterol at 6 months of age
- c. Reduce the infant's dietary fat to less than 30% of his calories by giving him skim milk
- d. Initiate lipid lowering agents
- e. Recommend yearly ECGs for the patient

**179.** For the past year, a 12-year-old boy has had recurrent episodes of swelling of his hands and feet, which has been getting worse recently. These episodes occur following exercise and emotional stress, last for two to three days, and resolve spontaneously. The last episode was accompanied by abdominal pain, vomiting, and diarrhea. The results of routine laboratory workup are normal. An older sister and a maternal uncle have had similar episodes but were not told a diagnosis. The most compatible diagnosis is

- a. Systemic lupus erythematosus
- b. Focal glomerulosclerosis
- c. Congenital nephrotic syndrome
- d. Hereditary angioedema
- e. Henoch-Schönlein purpura

**180.** A 15-year-old girl with short stature, neck webbing, and sexual infantilism is found to have coarctation of the aorta. A chromosomal analysis would demonstrate

- a. Mutation at chromosome 15q21.1
- b. Trisomy 21
- c. XO karyotype
- d. Defect at chromosome 4p16
- e. Normal chromosome analysis

**181.** A newborn is diagnosed with congenital heart disease. You counsel the family that the incidence of heart disease in future children is

- a. 1%
- b. 2 to 6%
- c. 8 to 10%
- d. 15 to 20%
- e. 25 to 30%

**182.** During a regular checkup on an 8-year-old child, you note a loud first heart sound with a fixed and widely split second heart sound at the upper left sternal border that does not change with respirations. The patient is otherwise active and healthy. The mostly likely heart lesion to explain these findings is

- a. Atrial septal defect
- b. Ventricular septal defect
- c. Isolated tricuspid regurgitation
- d. Tetralogy of Fallot
- e. Mitral valve prolapse

### Items 183–184

A 2-year-old boy is brought into the emergency room with a complaint of fever for 6 days and development of a limp. On examination, he is found to have an erythematous macular exanthem over his body, ocular conjunctivitis, dry and cracked lips, a red throat, and cervical lymphadenopathy. There is a grade II/VI vibratory systolic ejection murmur at the lower left sternal border. A white blood cell count and differential show predominant neutrophils with increased platelets on smear.

**183.** The most likely diagnosis is

- a. Scarlet fever
- b. Rheumatic fever
- c. Kawasaki disease
- d. Juvenile rheumatoid arthritis
- e. Infectious mononucleosis

**184.** The next steps in management of the child in the preceding question would be

- a. Intramuscular or oral penicillin, outpatient follow-up
- b. Pooled immunoglobulin (IVIG) and high-dose aspirin, inpatient admission
- c. Nonsteroidal anti-inflammatory medication, outpatient follow-up
- d. Blood cultures, intravenous vancomycin, inpatient admission
- e. Viral titers, contact sports precautions, outpatient follow-up

**185.** An ill-appearing 2-week-old girl is brought to the emergency room. She is pale and dyspneic with a respiratory rate of 80 breaths per min. Heart rate is 195 beats per min, heart sounds are distant, and a gallop is heard. There is cardiomegaly by x-ray. An echocardiogram demonstrates poor ventricular function, dilated ventricles, and dilation of the left atrium. An electrocardiogram shows ventricular depolarization complexes that have low voltage. The diagnosis suggested by this clinical picture is

- a. Myocarditis
- b. Endocardial fibroelastosis
- c. Pericarditis
- d. Aberrant left coronary artery arising from pulmonary artery
- e. Glycogen storage disease of the heart

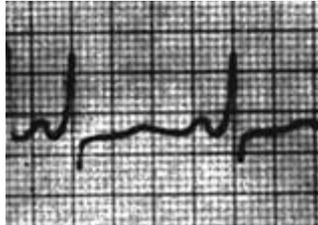
**186.** A newborn infant has mild cyanosis, diaphoresis, poor peripheral pulses, hepatomegaly, and cardiomegaly. Respiratory rate is 60 breaths per min, and heart rate is 250 beats per min. The child most likely has congestive heart failure caused by

- a. A large atrial septal defect and valvular pulmonic stenosis
- b. A ventricular septal defect and transposition of the great vessels
- c. Total anomalous pulmonary venous return
- d. Hypoplastic left heart syndrome
- e. Paroxysmal atrial tachycardia

**187.** A 2-year-old child with minimal cyanosis has an  $S_3$  and  $S_4$  (a quadruple rhythm), a systolic murmur in the pulmonic area, and a mid-diastolic murmur along the lower left sternal border. An electrocardiogram shows right atrial hypertrophy and a ventricular block pattern in the right chest leads. The child most likely has

- a. Tricuspid regurgitation and pulmonic stenosis
- b. Pulmonic stenosis and a ventricular septal defect (tetralogy of Fallot)
- c. An atrioventricular canal
- d. Ebstein's anomaly
- e. Wolff-Parkinson-White syndrome

**188.** A 4-year-old girl is brought to the pediatrician's office. Her father reports that she suddenly became pale and stopped running while he had been playfully chasing her. After 30 min, she was no longer pale and wanted to resume the game. She has never had a previous episode or ever been cyanotic. Her physical examination was normal, as were her chest x-ray and echocardiogram. An electrocardiogram showed the pattern seen below, which indicates



- a. Paroxysmal ventricular tachycardia
- b. Paroxysmal supraventricular tachycardia
- c. Wolff-Parkinson-White syndrome
- d. Stokes-Adams pattern
- e. Excessive stress during play

**189.** A child has a history of spiking fevers, which have been as high as  $40^{\circ}\text{C}$  ( $104^{\circ}\text{F}$ ). She has spindle-shaped swelling of finger joints and complains of upper sternal pain. The most likely diagnosis is

- a. Rheumatic fever
- b. Juvenile rheumatoid arthritis
- c. Toxic synovitis
- d. Septic arthritis
- e. Osteoarthritis

**190.** A cyanotic newborn is suspected of having congenital heart disease. The ECG shows left axis deviation and left ventricular hypertrophy (LVH). The most likely diagnosis is

- a. Transposition of the great arteries
- b. Truncus arteriosus
- c. Tricuspid atresia
- d. Tetralogy of Fallot
- e. Persistent fetal circulation

**191.** A 3-day-old infant with a single second heart sound has had progressively deepening cyanosis since birth but no respiratory distress. Chest radiography demonstrates no cardiomegaly and normal pulmonary vasculature. An electrocardiogram shows an axis of  $120^\circ$  and right ventricular prominence. The congenital cardiac malformation most likely responsible for the cyanosis is

- a. Tetralogy of Fallot
- b. Transposition of the great vessels
- c. Tricuspid atresia
- d. Pulmonary atresia with intact ventricular septum
- e. Total anomalous pulmonary venous return below the diaphragm

**192.** During a physical examination for participation in a sport, a 16-year-old girl is noted to have a late apical systolic murmur, which is preceded by a click. The rest of the cardiac examination is normal. She states that her mother also has some type of heart “murmur” but knows nothing else about it. The most likely diagnosis is

- a. Atrial septal defect
- b. Aortic stenosis
- c. Tricuspid regurgitation
- d. Mitral valve prolapse
- e. Ventricular septal defect

**193.** Appropriate management for the patient in the preceding question includes

- a. Cardiac catheterization
- b. Penicillin prophylaxis for dental procedures
- c. Avoidance of strenuous activity
- d. Beta blockers (propranolol)
- e. Digitalis

**DIRECTIONS:** Each group of questions below consists of lettered options followed by a set of numbered items. For each numbered item, select the appropriate lettered option(s). Each lettered option may be used once, more than once, or not at all. **Choose exactly the number of options indicated following each item.**

**Items 194–196**

For each case presented, select the cardiovascular defect with which the exam findings are most likely to be associated.

- a. Coarctation of the aorta
- b. Septal defect
- c. Supravalvular aortic stenosis
- d. Aortic aneurysm
- e. Patent ductus arteriosus

**194.** An infant comes to your office for a 2-week checkup. You find that the baby has a nonopposable thumb that seems to be triphalangeal. You note that the mother has abnormal thumbs as well. **(SELECT 1 DEFECT)**

**195.** A 15-year-old adolescent girl comes to your office for a sports physical. Her weight is in the twenty-fifth percentile and her height is above the ninety-fifth percentile. Physical examination reveals a tall, thin girl wearing rather thick glasses. She has a high-arched palate, dental crowding, and a mild scoliosis. Her fingers seem remarkably long and flexible. She has Tanner stage IV development. **(SELECT 1 DEFECT)**

**196.** A happy and friendly 10-year-old girl has prominent, thick lips; a long philtrum; short stature; and mild cognitive impairments. Her mother mentions a 2-week hospital stay after birth for electrolyte problems and poor feeding. **(SELECT 1 DEFECT)**

**Items 197–200**

For each of the cases presented below, choose the most likely cardiac diagnosis based on the patient's presentation.

- a. Congenital heart block
- b. Hypertrophic cardiomyopathy
- c. Prolonged QT syndrome
- d. Congestive heart failure
- e. Cor pulmonale

**197.** A 15-year-old adolescent male presents to the office for a sports physical. In his screening questionnaire, he notes that he occasionally gets short of breath and dizzy during exercise, with occasional chest pain. He lost consciousness once last summer during football practice, but attributed it to the heat. His grandfather died suddenly at age 35 of unknown etiology, but otherwise the family is healthy. **(SELECT 1 DIAGNOSIS)**

**198.** A 15-year-old adolescent female comes to be evaluated for syncopal episodes. Her only other medical problem is congenital deafness. She notes through a signing interpreter that syncopal episodes happen during stressful or emotional situations, and started only within the last year or so. Her mother and father are from the same small farming town and are second cousins. **(SELECT 1 DIAGNOSIS)**

**199.** You are called to the nursery to evaluate a newborn. The mother has a history of systemic lupus erythematosus and gestational diabetes. The nurses are concerned because the baby has developed petechiae and bruising after his bath. Vital signs have been stable, with a heart rate in the 60s and a respiratory rate in the 40s. You note a large liver, scattered petechiae, and an erythematous rash on the cheeks and the bridge of the nose. **(SELECT 1 DIAGNOSIS)**

**200.** An infant previously diagnosed with a large muscular ventricular septal defect comes to the office with complaints from the mother of fatigue and poor feeding over the past month. You note the child has not gained weight since the previous visit two months ago. The child is apathetic, tachypneic, and has wheezes and crackles on lung auscultation. **(SELECT 1 DIAGNOSIS)**

# The Cardiovascular System

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## Answers

**175. The answer is c.** (*Behrman, 16/e, p 2254. Rudolph, 21/e, pp 365–366.*) An important noncardiac manifestation of digitalis toxicity in infants is vomiting. Affected infants also exhibit electrocardiographic changes, including sinus arrhythmia (bradycardia) and a wandering pacemaker, paroxysmal tachycardia, and a heart rate of less than 100 beats per min. The commonly used digitalis preparation in infants is digoxin. Digoxin blood levels of 2 ng/dL or less are usually therapeutic in adults; in contrast, therapeutic digoxin blood levels in infants range from 1 to 5 ng/dL, but the benefit of the higher levels in infants is doubtful.

**176–177. The answers are 176-b, 177-d.** (*Behrman, 16/e, pp 806–810. McMillan, 3/e, pp 1417–1422. Rudolph, 21/e, pp 1901–1904.*) It is often difficult to establish a diagnosis of rheumatic fever because there is no single clinical manifestation or laboratory test that is confirmatory. The importance of doing so relates to the need to treat the acute problems promptly and effectively, as well as to the importance of instituting long-term antibiotic prophylaxis to prevent recurrences. To assist in diagnosis, the American Heart Association identified a set of major and minor standards relating to the manifestations of the disease, called the Jones criteria (modified), and recommends that these criteria be applied in the diagnosis of every patient with possible rheumatic fever. The major criteria are carditis, arthritis, erythema marginatum, chorea, and subcutaneous nodules. The minor criteria are arthralgia (joint pain with no objective findings), fever or history of rheumatic fever, increased erythrocyte sedimentation rate (ESR), positive C-reactive protein, increased WBC and anemia, and prolonged PR and QT intervals on ECG. To make the diagnosis of rheumatic fever, the following criteria should be met: two major manifestations, or one major and two minors plus strong evidence of a preceding group A  $\beta$ -hemolytic streptococcal infection (culture, rapid antigen-antibody rise or elevation) or scarlet fever.

Administration of salicylates and steroids can relieve the inflammatory manifestations of acute rheumatic fever. Steroids are used to treat affected children who have carditis with an enlarged heart. Neither salicylates nor corticosteroids have a therapeutic effect on chorea, but barbiturates and chlorpromazine may be helpful.

**178. The answer is a.** (*Behrman, 16/e, pp 389–390. McMillan, 3/e, pp 1864–1865. Rudolph, 21/e, pp 696–698.*) Identification of those with a genetic predisposition to hypercholesterolemia and of the factors that increase the risk of the condition is recommended so that dietary and other measures to reduce serum lipids can be introduced if indicated. Children with a first- or second-degree relative with early onset of coronary heart disease should be evaluated early in life but after 2 years of age. Other known risk factors include obesity, diabetes, hypertension, and smoking. No change in current infant dietary practice is recommended for children under 2 years of age. The high total fat content of the infant diet is considered to be biologically sound, given the need for lipid for a developing nervous system and the infant's limited capacity for an intake of high volume during this period of rapid growth. It is generally agreed that a dietary fat intake of 40% of calories is excessive over the age of 2 years. There is, however, concern about the potential loss of minerals such as iron, zinc, and calcium when dietary fat is reduced below 30% of calories in children 2 to 18 years of age. Principal sources of fat in the American diet are meat and milk, which are also sources of minerals such as iron and calcium. Simple modifications in the current U.S. diet of children of these ages (trimming excess fat from meat and drinking 1% fat milk) would effect a reduction of fat intake by 5% of calories without the risk of lowering mineral intake. It would be helpful in this situation to determine whether the uncle had hypercholesterolemia. Yearly ECGs and lipid-lowering agents are not indicated in this situation.

**179. The answer is d.** (*Behrman, 16/e, pp 632–633, 684–685. McMillan, 3/e, pp 2049–2053. Rudolph, 21/e, p 1196.*) This condition often worsens in adolescents. Although hereditary angioedema is relatively rare as a cause of edema, the recurrent episodes in late childhood, the normal laboratory results, and the family history make the other choices less likely. Hereditary angioedema, transmitted as an autosomal dominant trait, is due to inadequate function (due to either deficient quantity or quality) of an inhibitor of the first step in the complement cascade, which results in the excessive

production of a vasoactive kinin. In addition to otherwise asymptomatic subcutaneous edema, edema can occur in the gastrointestinal tract and produce the symptoms mentioned in the question. Laryngeal edema with airway obstruction can also occur.

**180. The answer is c.** (*Behrman, 16/e, pp 1753–1755. McMillan, 3/e, pp 2230–2231. Rudolph, 21/e, pp 476, 1781, 2087–2089.*) Short stature, neck webbing, sexual infantilism, and a shieldlike chest with widely spaced nipples are signs of Turner syndrome, which is usually associated with an XO karyotype. Aortic coarctation occurs in about 15 to 20% of those with this disorder. Down syndrome is most commonly associated with endocardial cushioning defects or ventricular septal defect. Marfan syndrome (mutation 15q21.1) is associated with dilatation of the aorta and mitral and aortic regurgitation. Ellis–van Creveld syndrome (defect is 4p16) is associated with atrial septal defects.

**181. The answer is b.** (*Behrman, 16/e, p 1363. McMillan, 3/e, p 279. Rudolph, 21/e, p 1781.*) The incidence of congenital heart disease in the population is about 1%. The risk of congenital heart disease in a family with one child born with heart disease varies depending on the type of lesion the first child had, but, overall, the rate averages from about 2 to 6% if the heart disease is not associated with a diagnosable syndrome. The risk of congenital heart disease for an infant is increased if there is a history of congenital heart disease in the mother. If there are two children with congenital heart disease, the risk escalates to 20 to 30%.

**182. The answer is a.** (*Behrman, 16/e, pp 1365–1366, 1369–1371, 1383–1390. McMillan, 3/e, pp 287, 1329–1332, 1346–1350, 1354–1357, 1378–1380. Rudolph, 21/e, pp 1788–1794, 1797–1798, 1820–1823.*) Most commonly, children with an atrial septal defect (ASD) are asymptomatic, with the lesion found during a routine examination. In older children, exercise intolerance can be noted if the lesion is of significant size. On examination, the pulses are normal, a right ventricular systolic lift at the left sternal border is palpable, and a fixed splitting of the second heart sound is audible. For lesser degrees of ASD, surgical treatment is more controversial. Ventricular septal defects commonly present as a harsh or blowing holosystolic murmur best heard along the left lower sternum, often with radiation throughout the precordium. Tricuspid regurgitation is a mid-diastolic rum-

ble at the lower left sternal border. Often, a history of birth asphyxia or findings of other cardiac lesions are present. Tetralogy of Fallot is a very common form of congenital heart disease. The four abnormalities include right ventricular outflow obstruction, ventricular septal defect, dextroposition of the aorta, and right ventricular hypertrophy. The cyanosis presents in infants and in young children. Mitral valve prolapse occurs with the billowing into the atria of one or both mitral valve leaflets at the end of systole. It is a congenital abnormality that frequently manifests only during adolescence or later. It is more common in girls than in boys and seems to be inherited in an autosomal dominant fashion. On clinical examination, an apical murmur is noted late in systole, which can be preceded by a midsystolic click. The diagnosis is confirmed with an echocardiogram that shows prolapse of the mitral leaflets during mid- to late systole. Antibiotic prophylaxis is recommended for dental work (especially if a murmur is present), as the incidence of endocarditis can be higher in these patients.

**183–184. The answers are 183-c, 184-b.** (*Behrman, 16/e, pp 725–727. McMillan, 3/e, pp 924–932. Rudolph, 21/e, pp 844–845.*) Many conditions can be associated with prolonged fever, a limp caused by arthralgia, exanthem, adenopathy, and pharyngitis. Conjunctivitis, however, is suggestive of Kawasaki disease. The fissured lips, although common in Kawasaki disease, could occur after a long period of fever from any cause if the child became dehydrated. The predominance of neutrophils and high sedimentation rate are common to all. An increase in platelets within this constellation of symptoms, however, is found only in Kawasaki disease. Kawasaki disease presents a picture of prolonged fever, rash, epidermal peeling on the hands and feet (especially around the fingertips), conjunctivitis, lymphadenopathy, fissured lips, oropharyngeal mucosal erythema, and arthralgia or arthritis. The diagnosis is still possible in the absence of one or two of these physical findings. Coronary artery aneurysms can develop, as can aneurysms in other areas.

Initial treatment is typically IVIG and high-dose aspirin. The child will usually defervesce shortly after the infusion. Aspirin is typically kept at a higher dose until the platelet count begins to decrease, and then is continued at a lower dose for several weeks. While bacterial infection is in the differential diagnosis for this patient's presentation and blood cultures are usually part of the evaluation, intravenous vancomycin should be reserved

for a culture-proven susceptible organism resistant to other antibiotics, or as empiric therapy in a critically ill patient.

**185. The answer is a.** (*Behrman, 16/e, pp 1434–1435. McMillan, 3/e, pp 1389–1397. Rudolph, 21/e, pp 1860–1861.*) The findings of pallor, dyspnea, tachypnea, tachycardia, and cardiomegaly are common in congestive heart failure regardless of the cause. The most common causes of myocarditis include adenovirus and coxsackievirus B, although many other viruses can cause this condition. The constellation of findings in the question point to myocarditis as the etiology of this patient's condition. The lack of echocardiographic findings other than ventricular and left atrial dilatation and poor ventricular function is inconsistent with both glycogen storage disease of the heart, in which there is muscle thickening, and pericarditis, since there is no pericardial effusion. It is also not consistent with an aberrant origin of the left coronary artery, although the origin of the coronary arteries can be more easily missed. On electrocardiogram, the voltages of the ventricular complexes seen with aberrant origin of the left coronary artery are not diminished, and a pattern of myocardial infarction can be seen. Voltages from the left ventricle are usually high in endocardial fibroelastosis, and both right and left ventricular forces are high in glycogen storage disease of the heart.

**186. The answer is e.** (*Behrman, 16/e, pp 1418–1421. McMillan, 3/e, pp 290–291, 1431–1432. Rudolph, 21/e, pp 1854–1856.*) Congestive heart failure from any cause can result in mild cyanosis, even in the absence of a right-to-left shunt, and in poor peripheral pulses when cardiac output is low. Congestive heart failure from many causes can be associated with a rapid pulse rate (up to 200 beats per min). A pulse rate greater than 250 beats per min, however, should suggest the presence of a tachyarrhythmia. Common causes for supraventricular tachycardia includes Wolff-Parkinson-White (WPW) syndrome, congenital heart disease, and sympathomimetic drugs. In this patient, evaluation for WPW and cardiac abnormalities must be accomplished after the congestive heart failure from the increased heart rate is under control.

**187. The answer is d.** (*Behrman, 16/e, pp 1384, 1393–1395. Rudolph, 21/e, pp 1817–1818.*) A quadruple rhythm associated with the murmur of tricus-

pid regurgitation and a mid-diastolic murmur at the lower left sternum suggests the diagnosis of Ebstein anomaly (downward displacement of the tricuspid valve). The presence of right atrial hypertrophy and right ventricular conduction defects confirms the diagnosis. Both tricuspid regurgitation with pulmonic stenosis and tetralogy of Fallot give electrocardiographic evidence of right ventricular enlargement. The Wolff-Parkinson-White syndrome, which frequently accompanies Ebstein malformation, is not associated with murmurs or cyanosis as an isolated entity but also includes supraventricular tachycardia.

**188. The answer is c.** (*Behrman, 16/e, pp 1418–1420. McMillan, 3/e, pp 1431–1432. Rudolph, 21/e, pp 1452–1453.*) The child described in the question, who has no cyanosis or murmur, no cardiac or pulmonary vascular abnormalities by chest x-ray, and no evidence of structural anomalies by echocardiogram, is unlikely to have an underlying gross anatomic defect. The electrocardiographic pattern in the figure shows the configuration of preexcitation, the pattern seen in the Wolff-Parkinson-White syndrome (WPW). These patients have an aberrant atrioventricular conduction pathway, which causes the early ventricular depolarization appearing on the electrocardiogram as a shortened PR interval. The initial slow ventricular depolarization wave is referred to as the *delta wave*. Seventy percent of patients with WPW have single or repeated episodes of paroxysmal supraventricular tachycardia, which can cause the symptoms described in the question. The preexcitation electrocardiographic pattern and WPW can occur in Ebstein malformation, but this is unlikely in the absence of cyanosis and with a normal echocardiogram. If ventricular tachycardia were present, the symptoms would likely be more profound. Active play and exposure to over-the-counter medications containing sympathomimetics in a healthy 4-year-old child can cause symptoms such as those described in the question in children with WPW by precipitating paroxysmal supraventricular tachycardia.

**189. The answer is b.** (*Behrman, 16/e, pp 704–709. McMillan, 3/e, pp 2156–2160. Rudolph, 21/e, pp 836–839.*) Juvenile rheumatoid arthritis (JRA) frequently causes spindle-shaped swelling of finger joints and can involve unusual joints such as the sternoclavicular joint. Presentation of JRA occurs as either polyarthritis (5 or more joints, systemic symptoms not so severe or persistent), pauciarticular (4 or fewer joints, lower extremity joints,

extra-articular disease unusual), or systemic disease (severe constitutional disease, systemic symptoms prior to arthritis, rheumatoid rash, high-spiking fevers, variable joints). This disorder can be associated with spiking high fevers, which are not a feature of rheumatic fever, toxic synovitis, or osteoarthritis. Although septic arthritis can affect any joint, it would not be likely to affect finger joints by causing spindle-shaped swellings. Toxic synovitis usually involves larger joints such as the hip, and osteoarthritis is not a disease of childhood.

**190. The answer is c.** (*Behrman, 16/e, pp 1391–1392. McMillan, 3/e, pp 1327–1329. Rudolph, 21/e, pp 1816–1817.*) Tricuspid atresia has a hypoplastic right ventricle and, therefore, the ECG shows left axis deviation and left ventricular hypertrophy. Almost all other forms of cyanotic congenital heart disease are associated with elevated pressures in the right ventricle. In those, therefore, the ECG will show right axis deviation and right ventricular hypertrophy.

**191. The answer is b.** (*Behrman, 16/e, pp 1395–1398. McMillan, 3/e, pp 1322–1324. Rudolph, 21/e, pp 1823–1826.*) Transposition of the great vessels with an intact ventricular septum presents with early cyanosis, a normal-sized heart (classic “egg on a string” radiographic pattern in one-third of cases), normal or slightly increased pulmonary vascular markings, and an electrocardiogram showing right axis deviation and right ventricular hypertrophy. In tetralogy of Fallot, cyanosis is often not seen in the first few days of life. Tricuspid atresia, a cause of early cyanosis, causes diminished pulmonary arterial blood flow; the pulmonary fields on x-ray demonstrate a diminution of pulmonary vascularity. There is a left axis and left ventricular hypertrophy shown by electrocardiogram. Total anomalous pulmonary venous return below the diaphragm is associated with obstruction to pulmonary venous return and a classic radiographic finding of marked, fluffy-appearing venous congestion (“snowman”). In pulmonic atresia with intact ventricular septum, cyanosis appears early, the lung markings are normal to diminished, and the heart is large.

**192–193. The answers are 192-d, 193-b.** (*Behrman, 16/e, pp 1383–1384. McMillan, 3/e, pp 1378–1380. Rudolph, 21/e, pp 1797–1798.*) Mitral valve prolapse occurs with the billowing into the atria of one or both mitral valve leaflets at the end of systole. It is a congenital abnormality that frequently

manifests only during adolescence or later. It is more common in girls than in boys and seems to be inherited in an autosomal dominant fashion. On clinical examination, an apical murmur is noted late in systole, which can be preceded by a click. The diagnosis is confirmed with an echocardiogram that shows prolapse of the mitral leaflets during mid- to late systole. The ECG and chest x-ray are usually normal. Beta blockers and digitalis are unlikely to be required, but penicillin prophylaxis for dental procedures for patients with mitral valve prolapse, especially if a murmur is present, is indicated.

**194–196. The answers are 194-b, 195-d, 196-c.** (*Behrman, 16/e, pp 328, 331, 1365–1366, 1377, 1753–1755, 2131–2132. McMillan, 3/e, pp 1347, 2230–2231, 2241, 2251. Rudolph, 21/e, pp 732–734, 738, 740, 762–764, 1781, 1805, 2087–2089.*) Holt-Oram syndrome is autosomal dominant, and includes an atrial or, less commonly, ventricular septal defect with skeletal abnormalities primarily of the forearm and hand. All affected have carpal bone anomalies.

Major findings in Marfan syndrome include ectopia lentis (subluxation of the ocular lens), progressive dilatation of the aortic root and ascending aorta, aortic aneurysm, excessive height for age, and disproportionately long and thin extremities, with hypermobility and ligamentous laxity.

William syndrome is most characteristically associated with supravalvular aortic stenosis, but children with the syndrome may also have peripheral pulmonic stenosis, pulmonary valve stenosis, atrial septal defect, and ventricular septal defect. These children frequently have hypercalcemia in infancy, mental retardation, and characteristic facies. The defect is on chromosome 7.

Common features of Turner syndrome include female phenotype, short stature, sexual infantilism, streak gonads, broad chest, low hairline, webbed neck, congenital lymphedema of the hands and feet, coarctation of the aorta, and a variety of other anomalies.

Down syndrome has many diagnostic features including microcephaly, centrally placed hair whorl, small ears, redundant skin on the nape of the neck, upslanting palpebral fissures, epicanthal folds, flat nasal bridge, Brushfield spots, protruding tongue, short and broad hands, single palmar creases, widely spaced first and second toe, and hypotonia. Cardiac lesions are found in 30 to 50% of children with Down syndrome, including endocardial cushion defect (30%), ventricular septal defect (30%), and tetralogy of Fallot (about 30%).

**197–200. The answers are 197-b, 198-c, 199-a, 200-d.** (Behrman, 16/e, pp 716, 1433–1434, 1440–1441, 1830. McMillan, 3/e, pp 381, 1354–1356, 1401–1403, 2165, Rudolph, 21/e, pp 847, 1858, 1860–1862, 1869–1877.) Hypertrophic cardiomyopathy (HCM), also called *ideopathic hypertrophic subaortic stenosis*, is often inherited in autosomal dominant fashion with variable penetrance. Many children are completely asymptomatic, while others may have complaints of weakness, fatigue, chest pain, and syncope. A midsystolic ejection murmur and a left ventricular lift sometimes can be appreciated on physical examination. The diagnosis is confirmed by echocardiography. HCM can be a cause of arrhythmia and death in young athletes. Prolonged QT syndrome occurs in 1:10,000 to 1:15,000 children, usually first causing syncopal episodes in late childhood or adolescence. During the syncope episodes arrhythmias may be noted, including ventricular fibrillation. These episodes may result in death. QT intervals are elongated on electrocardiography. An autosomal recessive form associated with deafness (Jervell-Lange-Nielsen syndrome) and an autosomal dominant form (Romano-Ward syndrome) have been described.

Neonatal lupus is a rare manifestation of transferred maternal IgG autoantibodies. Infants can have thrombocytopenia, neutropenia, rash, liver dysfunction, and a congenital heart block. Most manifestations are self-resolved; however, the congenital heart block is permanent and frequently requires pacing.

Congestive heart failure can result from a number of causes, including congenital heart disease, Kawasaki disease, metabolic cardiomyopathies, arrhythmias, and viral myocarditis. The case presented is typical of an infant with heart failure. Such infants are weak, diaphoretic, have poor weight gain, and may be tachypneic with retractions. Lung findings can include crackles, wheezes, or both. Prompt evaluation by a pediatric cardiologist is imperative.

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# The Respiratory System

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## Questions

**DIRECTIONS:** Each item below contains a question or incomplete statement followed by suggested responses. Select the **one best** response to each question.

### Items 201–202

**201.** A previously well 1-year-old infant has had a runny nose and has been sneezing and coughing for 2 days. Two other members of the family had similar symptoms. Four hours ago, his cough became much worse. On physical examination, he is in moderate respiratory distress with nasal flaring, hyperexpansion of the chest, and easily audible wheezing without rales. The most likely diagnosis is

- a. Bronchiolitis
- b. Viral croup
- c. Asthma
- d. Epiglottitis
- e. Diphtheria

**202.** The most likely agent responsible for the infant's condition in the preceding question is

- a. *Staphylococcus aureus*
- b. *Haemophilus influenzae*
- c. *Corynebacterium diphtheriae*
- d. Respiratory syncytial virus
- e. Echovirus

**203.** A 10-month-old infant has poor weight gain, a persistent cough, and a history of several bouts of pneumonitis. The mother describes the child as having very large, foul-smelling stools for months. Which of the following diagnostic maneuvers is likely to result in the correct diagnosis of this child?

- a. CT of the chest
- b. Serum immunoglobulins
- c. TB skin test
- d. Inspiratory and expiratory chest x-ray
- e. Sweat chloride test

**204.** A 13-year-old girl with a history of 2 days of cough and fever has the chest x-ray shown here. The most appropriate treatment is



- a. N-acetylcysteine
- b. Prolonged course of ampicillin combined with a  $\beta$ -lactamase inhibitor
- c. Lobectomy
- d. Postural drainage
- e. Thoracentesis and chest tube

**205.** A 2-year-old girl is playing in the garage, only partially supervised by her father, who is mowing the grass. He finds her in the garage, gagging and vomiting. She smells of gasoline. In a few minutes she stops vomiting, but later that day she develops cough, tachypnea, and subcostal retractions. She is brought to your emergency center. The first step in her management is to

- a. Administer charcoal
- b. Begin nasogastric lavage
- c. Administer ipecac
- d. Perform pulse oximetry and arterial blood gas
- e. Administer gasoline binding agent intravenously

**206.** A 3-year-old girl is admitted with the x-ray pictured. The child lives with her parents and a 6-week-old brother. Her grandfather stayed with the family for 2 months before his return to the West Indies 1 month ago. The grandfather had a 3-month history of weight loss, fever, and hemoptysis. Appropriate management of this problem includes



- a. Bronchoscopy and culture of washings for all family members
- b. Placement of a Mantoux test on the 6-week-old sibling
- c. Isolating the 3-year-old patient for 1 month
- d. Treating the 3-year-old patient with isoniazid (INH) and rifampin
- e. HIV testing for all family members

**207.** A tine test administered at a routine visit of a 2½-year-old boy is positive. The child is asymptomatic and thriving; there is no known exposure to tuberculosis. Your initial course of action would be to

- a. Admit the child and perform a workup for tuberculosis
- b. Start the child on isoniazid (INH)
- c. Repeat the tine test
- d. Screen all contacts with a tuberculin test and chest x-ray
- e. Administer a Mantoux (PPD) test

**208.** A previously healthy, active 18-month-old child presents with unilateral nasal obstruction and foul-smelling discharge. The child's examination is otherwise unremarkable. The most likely diagnosis is

- a. Foreign body
- b. Nasal polyps
- c. Frontal sinusitis
- d. Deviated septum
- e. Choanal atresia

**209.** A 7-year-old child is brought by her mother for a school physical. His growth parameters show his height to be fiftieth percentile and his weight to be significantly higher than ninety-fifth percentile. His mother complains that he always seems sleepy during the day and that he has started complaining of headaches. His second-grade teacher has commented that he has difficulty staying awake in class. His mother complains that he wakes up the whole house with his snoring at night. The next step in evaluating and managing this condition should be to

- a. Try steroids to decrease tonsillar and adenoid hypertrophy
- b. Refer to an otolaryngologist for tonsillectomy and adenoidectomy
- c. Arrange for continuous positive airway pressure (CPAP) at home
- d. Arrange for home oxygen therapy for use at night
- e. Arrange for polysomnography

**210.** You admitted to the hospital the previous evening a 4-year-old boy who presented with cough, fever, and mild hypoxia. At the time of his admission, he had evidence of a right upper lobe consolidation on his chest radiograph. A blood culture has become positive in less than 24 h for *Staphylococcus aureus*. Approximately 20 h into his hospitalization, the nurse calls you because the child has acutely worsened over the last few minutes, with markedly increased work of breathing and increasing oxygen requirement. As you move swiftly to the child's hospital room, you call ahead and order

- a. A second chest radiograph to evaluate for pneumatocele formation
- b. A large-bore needle and chest tube kit for aspiration of a probable tension pneumothorax
- c. A change in antibiotics to include gentamicin
- d. A sedative to treat the child's attack of severe anxiety
- e. A thoracentesis kit to drain his probable pleural effusion

**211.** A fully immunized 2-year-old presents to the emergency room with several days of low-grade fever, barking cough, and noisy breathing. Over the last few hours he has developed a fever to 40°C (104°F) and looks toxic. He has inspiratory and expiratory stridor. The family has not noticed drooling, and he seems to be drinking without pain. Direct laryngoscopy reveals a normal epiglottis. The management of this disease process includes

- a. Intubation and intravenous antibiotics
- b. Inhaled epinephrine and oral steroids
- c. Inhaled steroids
- d. Observation only
- e. Oral antibiotics and outpatient follow-up

**212.** A 6-year-old boy is brought to the emergency room with a 3-h history of fever to 39.5°C (103.1°F) and sore throat. The child appears alert but anxious and toxic. He has mild inspiratory stridor and is drooling. You should immediately

- a. Examine the throat and obtain a culture
- b. Obtain an arterial blood gas and start an IV line
- c. Order a chest x-ray and lateral view of the neck
- d. Prepare to establish an airway
- e. Admit the child and place him in a mist tent

**213.** A 3-year-old boy has a young puppy and also has a history of pica. He has a recent onset of wheezing, hepatomegaly, and marked eosinophilia (60% eosinophils). The test most likely to produce a specific diagnosis is

- a. Tuberculin skin test
- b. Histoplasmin test
- c. ELISA for *Toxocara*
- d. Silver stain of gastric aspirate
- e. Stool examination for ova and parasites

**214.** A 10-year-old has had a “cold” for 14 days. In the 2 days prior to the visit to your office, she has developed a fever to 39°C (102.2°F), purulent nasal discharge, facial pain, and a daytime cough. Examination of the nose after topical decongestants shows pus in the middle meatus. The most likely diagnosis is

- a. Brain abscess
- b. Maxillary sinusitis
- c. Streptococcal throat infection
- d. Sphenoid sinusitis
- e. Middle-ear infection

**215.** You are awakened in the night by your 2-year-old son, who has developed noisy breathing on inspiration, marked retractions of the chest wall, flaring of the nostrils, and a barking cough. He has had a mild upper respiratory infection (URI) for 2 days. The most likely diagnosis is

- a. Asthma
- b. Epiglottitis
- c. Bronchiolitis
- d. Viral croup
- e. Foreign body in the right mainstem bronchus

**216.** In patients on theophylline therapy for asthma, certain other drugs change theophylline clearance and raise or lower theophylline levels. Which of the following medications may be safely administered with theophylline?

- a. Erythromycin
- b. Phenytoin
- c. Cimetidine
- d. Rifampin
- e. Penicillin

**217.** A 13-year-old develops fever, malaise, sore throat, and a dry hacking cough over several days. He does not appear to be particularly sick, but his chest examination is significant for diffuse rales and rhonchi. The chest radiograph is shown. The most likely pathogen is



(Courtesy Susan John, M.D.)

- Staphylococcus aureus*
- Mycobacterium tuberculosis*
- Haemophilus influenzae*
- Streptococcus pneumoniae*
- Mycoplasma pneumoniae*

**218.** As a doctor in a clinic, you have just given a 10-year-old boy an injection of pollen extract as prescribed by his allergist. You are about to move on to the next patient when the boy starts to complain about a funny feeling in his chest, and his face becomes red and swollen. He then develops severe respiratory distress with wheezing, and as he starts to fall, you catch him and place him on a bed. The *least* important measure to be instituted immediately would be

- Endotracheal intubation
- Placement of a tourniquet above the injection site
- Administration of oxygen
- Subcutaneous injection of 1:1000 epinephrine 0.2 mL
- Administration of corticosteroids

**219.** A previously healthy 18-month-old has been in a separate room from his family. The family notices the sudden onset of coughing, which resolves over a few minutes. Subsequently, the patient appears to be normal except for increased amounts of drooling and refusal to take foods orally. The most likely explanation for this toddler's condition is

- a. Severe gastroesophageal reflux
- b. Foreign body in the airway
- c. Croup
- d. Epiglottitis
- e. Foreign body in the esophagus

### **Items 220–223**

**220.** You receive a telephone call from the mother of a 4-year-old child with sickle cell anemia. She tells you that the child is breathing fast, coughing, and has a temperature of 40°C (104°F). The most conservative, prudent approach is to

- a. Prescribe aspirin and ask her to call back if the fever does not respond
- b. Make an office appointment for the next available opening
- c. Make an office appointment for the next day
- d. Refer the child to the laboratory for an immediate hematocrit, white blood cell count, and differential
- e. Admit the child to the hospital as an emergency

**221.** Although the patient is in respiratory distress, a lack of cyanosis indicates

- a. Insignificant hypoxia
- b. That the patient has pulmonary thrombi rather than pneumonia
- c. Very little since it is not a useful indication of hypoxia in a patient with anemia
- d. Shift of the oxyhemoglobin curve to the right due to increased levels of BPG (previously known as DPG)
- e. An adequate hemoglobin level for the given activity level

**222.** The laboratory workup of the patient reveals the following: hemoglobin 5 g/dL; hematocrit 16%; white blood cell count 30,000/ $\mu$ L; and arterial blood, while breathing room air, of pH 7.1, PO<sub>2</sub> 35 mmHg, and PaCO<sub>2</sub> 28 mmHg. These values indicate

- a. Acidemia, metabolic acidosis, respiratory alkalosis, and hypoxia
- b. Alkalemia, respiratory acidosis, metabolic alkalosis, and hypoxia
- c. Acidosis with compensatory hypoventilation
- d. Long-term metabolic compensation for respiratory alkalosis
- e. Primary respiratory alkalosis

**223.** While waiting for further studies of the patient, it is appropriate to administer

- a. Sedation
- b. Bicarbonate by IV push
- c. Urea
- d. 100% oxygen
- e. 40% oxygen with 5% carbon dioxide

**224.** A 13-year-old boy has a 3-day history of low-grade fever, upper respiratory symptoms, and a sore throat. A few hours before his presentation to the emergency room, he has an abrupt onset of high fever, difficulty swallowing, and poor handling of his secretions. He indicates that he has a marked worsening in the severity of his sore throat. His pharynx has a fluctuant bulge in the posterior wall. Appropriate initial therapy for this patient would be

- a. Narcotic analgesics
- b. Trial of oral penicillin V
- c. Surgical consultation for incision and drainage under general anesthesia
- d. Rapid streptococcal screen
- e. Monospot test

**225.** A 6-week-old child develops increased respiratory rate and a non-productive cough. Physical examination is significant for rales and rhonchi. The past medical history for the child is positive for an eye discharge at 3 weeks of age, which was treated with a topical antibiotic drug. The most likely organism causing this child's condition is

- a. *Neisseria gonorrhoeae*
- b. *Staphylococcus aureus*
- c. Group B streptococcus
- d. *Chlamydia trachomatis*
- e. Herpesvirus

**226.** Appropriate therapy for the patient in the previous question includes

- a. Ceftriaxone
- b. Acyclovir
- c. Penicillin G
- d. Nafcillin
- e. Erythromycin

**227.** A previously healthy 2-year-old black child has developed a chronic cough over the previous 6 weeks. He has been seen in different emergency rooms on two occasions during this period and placed on antibiotics for pneumonia. Upon auscultation, you hear normal breath sounds on the left. On the right side, you hear decreased air movement during inspiration but none upon expiration. The routine chest radiograph shows no infiltrate, but the heart is shifted slightly to the left. The appropriate next step in making the diagnosis in this patient is to

- a. Measure the patient's sweat chloride
- b. Obtain inspiratory and expiratory chest radiographs
- c. Prescribe broad-spectrum oral antibiotics
- d. Initiate a trial of inhaled  $\beta$  agonists
- e. Prescribe appropriate doses of oral prednisone

**DIRECTIONS:** Each group of questions below consists of lettered options followed by a set of numbered items. For each numbered item, select the appropriate lettered option(s). Each lettered option may be used once, more than once, or not at all. **Choose exactly the number of options indicated following each item.**

### Items 228–230

Match each management procedure below with the appropriate set of arterial blood gas results of patients spontaneously breathing room air.

	pH	PCO <sub>2</sub> (mmHg)	PO <sub>2</sub> (mmHg)	Base Excess (meq/L)
a.	7.20	28	95	16
b.	7.20	70	41	2
c.	7.64	18	94	1
d.	7.34	32	39	8
e.	none of the above			

**228.** Have patient rebreathe in a paper bag (**SELECT 1 RESULT SET**)

**229.** Administer FiO<sub>2</sub> 0.4 (**SELECT 1 RESULT SET**)

**230.** Perform thoracentesis to remove air under pressure (**SELECT 1 RESULT SET**)

### Items 231–234

Match each management procedure below with the appropriate arterial blood gas results of patients spontaneously breathing room air.

	pH	PCO <sub>2</sub> (mmHg)	PO <sub>2</sub> (mmHg)	Base Excess (meq/L)
a.	6.92	101	19	15
b.	7.36	60	50	7
c.	7.50	46	76	11
d.	7.41	60	90	10
e.	none of the above			

**231.** Place the patient on a ventilator with an  $\text{FIO}_2$  of 1.0 (**SELECT 1 RESULT SET**)

**232.** Discontinue diuretics, discontinue base, and increase KCl in IV fluids (**SELECT 1 RESULT SET**)

**233.** Perform tonsillectomy (**SELECT 1 RESULT SET**)

**234.** Repeat the test because of obvious laboratory error (**SELECT 1 RESULT SET**)

# The Respiratory System

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## Answers

**201–202. The answers are 201-a, 202-d.** (*Behrman, 16/e, pp 1285–1287. McMillan, 3/e, pp 1214–1216. Rudolph, 21/e, pp 1984–1985.*) Of the choices given, bronchiolitis is the most likely, although asthma, pertussis, and bronchopneumonia can present similarly. The family history of upper respiratory infections, the previous upper respiratory illness in the patient, and signs of intrathoracic airway obstruction make the diagnosis of bronchiolitis more likely. Viral croup, epiglottitis, and diphtheria are not reasonable choices because there are no signs of extrathoracic airway obstruction.

The most likely cause of the illness is infection by respiratory syncytial virus, which causes outbreaks of bronchiolitis of varying severity, usually in the winter and spring. Other viruses, such as parainfluenza and the adenoviruses, have also been implicated in producing bronchiolitis. Treatment is usually supportive in this usually self-limited condition.

**203. The answer is e.** (*Behrman, 16/e, pp 1315–1327. McMillan, 3/e, pp 1242–1254. Rudolph, 21/e, pp 1967–1979.*) Cystic fibrosis (CF) is a multisystem disease caused by an abnormally functioning cystic fibrosis transmembrane regulator protein. Abnormal secretions are produced as a result of decreased permeability of ionized chloride in the secretory epithelium of a number of organs. Progressive lung failure is caused by accumulation of viscid secretions that obstruct the airway and lead to infection, bronchiectasis, and inflammatory changes. Survival has improved during the past few decades as a result of prompt recognition of CF and aggressive treatment; the median age at death has increased from less than 10 years to more than 30 years. Therapeutic approaches have included inhalation therapy, chest physical therapy, aggressive antibiotic administration, bronchodilators, oxygen, and nutritional support. Heart-lung transplants have prolonged life and improved quality of life for some terminal patients. Several new approaches to the treatment of CF have been proposed, namely, the use of amiloride, purified human plasma alpha<sub>1</sub>-antitrypsin, recombinant DNAase, and gene therapy. The rationale for these therapeutic modalities is that they focus directly on ameliorating or correcting the basic deficit: amiloride by inhibiting sodium, and with it water reabsorption, and thereby improving airway

hydration; alpha<sub>1</sub>-antitrypsin by counteracting the effects on the lungs of neutrophil elastase, a proteolytic enzyme released by neutrophils; DNAase by reacting with DNA released by dead leukocytes to reduce sputum viscosity; and gene therapy by altering genetic material. Lung cancer does not appear to be associated with cystic fibrosis.

Unlike many other tests, there is almost no overlap in chloride values in sweat between patients with cystic fibrosis and normal control participants. A chloride concentration of greater than 60 meq/L is diagnostic, values less than 40 are normal, and values between 40 and 60 are intermediate. Conditions other than cystic fibrosis can manifest an elevated sweat chloride including adrenal insufficiency, ectodermal dysplasia, nephrogenic diabetes insipidus, hypothyroidism, and malnutrition.

**204. The answer is b.** (*Behrman, 16/e, pp 1309–1310. McMillan, 3/e, pp 1227–1230. Rudolph, 21/e, p 1982.*) The x-ray reveals a lung abscess involving the right upper lobe characterized by the round density, the air-fluid level, and the opaque rim. Lung abscesses are usually caused by anaerobic bacteria such as bacteroides, fusobacteria, and anaerobic streptococci, and on occasion by *Staphylococcus aureus* and *Klebsiella*. The organisms were previously sensitive to penicillin, but some anaerobic organisms (especially bacteroides) are now resistant due to  $\beta$ -lactam production. Lung abscesses frequently respond surprisingly well to treatment with antibiotics alone.

**205. The answer is d.** (*Behrman, 16/e, pp 1289–1290, McMillan, 3/e, pp 621–622. Rudolph, 21/e, pp 366–367.*) Hydrocarbons with low viscosity and high volatility are the most likely agents to cause respiratory symptoms. Gasoline, kerosene, and furniture polish (which contain hydrocarbons) are common agents responsible for hydrocarbon aspiration. Hydrocarbon aspiration can produce dyspnea, cyanosis, and respiratory failure. Treatment is symptomatic, sometimes requiring intubation and mechanical ventilation. Induction of emesis is contraindicated, as this may cause further aspiration. Placement of a nasogastric tube is used only in high-volume ingestions or when the hydrocarbon is mixed with another toxin. Charcoal is not useful, and no intravenous binding agent is available.

**206. The answer is d.** (*Behrman, 16/e, pp 885–897. McMillan, 3/e, pp 1026–1039. Rudolph, 21/e, pp 949–959.*) The key to controlling tuberculosis in children and eradicating the disease is early detection and appropriate

treatment of adult cases; the child, once infected, is at lifelong risk for the development of the disease and for infecting others unless given isoniazid prophylaxis. The usual source of the disease is an infected adult. Household contacts of a person with newly diagnosed active disease have a considerable risk of developing active tuberculosis, and the risk is greatest for infants and children. Therefore, when tuberculosis is diagnosed in a child, the immediate family and close contacts should be tested with tuberculin skin tests and chest radiographs and treated appropriately when indicated. Bronchoscopy would be indicated only in unusual circumstances. Three to eight weeks is required after exposure before hypersensitivity to tuberculin develops. This means that the tuberculin test must be repeated in exposed persons if there is a negative reaction at the time that contact with the source of infection is broken. TB skin tests are usually negative in infants of this age, even when active disease is ongoing. A logical preventive measure is the administration of isoniazid to the baby for 3 months when a Mantoux (purified protein derivative, PPD) can then be placed. Transmission of tuberculosis occurs when bacilli-laden, small-sized droplets are dispersed into the air by the cough or sneeze of an infected adult. Small children with primary pulmonary tuberculosis are not considered infectious to others, and they are not capable of coughing up and producing sputum. Sputum, when produced, is promptly swallowed, and for this reason specimens for microbial confirmation can be obtained by means of gastric lavage from smaller children.

**207. The answer is e.** (*Behrman, 16/e, p 887. McMillan, 3/e, pp 1028–1029. Rudolph, 21/e, pp 955–956.*) The tuberculin skin test is based on the detection of delayed hypersensitivity to the antigen of *Mycobacterium tuberculosis*. Two to ten weeks after infection, the intradermal injection of antigen will result in a positive response indicated by induration. The tine test, a multipuncture skin test used widely for mass screening because of its ease of administration, uses a plastic unit with four stainless steel blades treated with a crude filtrate of culture medium containing old tuberculin (OT). The tine test has a number of problems, such as a lack of standardization and variable results. Its use is discouraged, especially if TB is suspected. Retesting positive or doubtful tine reactions with the Mantoux test is indicated. The Mantoux intracutaneous tuberculin test uses a protein precipitate derived from OT (purified protein derivative, PPD) and is the preferred skin test. It is a more reliable test because of the efforts used in its

preparation to standardize and preserve potency and because it delivers a defined amount of the antigen. The most appropriate next step in managing an asymptomatic, thriving child with a positive tine test is to administer a Mantoux test.

**208. The answer is a.** (*Behrman, 16/e, pp 1259–1260. McMillan, 3/e, p 639. Rudolph, 21/e, p 1265.*) Small children frequently introduce any number of small objects into their noses, ranging from food to small toys. Initially, only local irritation occurs. Later, as prolonged obstruction is seen, symptoms increase to include worsening of pain, and a purulent, malodorous, bloody discharge can be seen. Unilateral nasal discharge in the presence of obstruction suggests the need to examine the patient for a nasal foreign body.

**209. The answer is e.** (*Behrman, 16/e, pp 1268–1271. McMillan, 3/e, pp 1256–1258. Rudolph, 21/e, pp 1937–1941.*) A nasopharyngeal airway, the use of nasal CPAP, and tonsillectomy and adenoidectomy can be effective treatments for obstructive sleep apnea (OSA). However, the diagnosis of OSA should first be made by polysomnography (sleep study) to exclude other causes of snoring. Administering oxygen may decrease the respiratory drive in severe OSA, and thus should be used only with caution. Irradiation should not be used for fear of development of malignancy, particularly of the thyroid.

**210. The answer is b.** (*Behrman, 16/e, pp 764, 795, 1331–1332. McMillan, 3/e, pp 1010, 1229–1230. Rudolph, 21/e, pp 149–150, 1981.*) Tension pneumothorax, a recognized complication of staphylococcal pneumonia, can be quickly lethal if not recognized and treated. This makes a high index of suspicion and prompt diagnosis mandatory. The other complications can occur also but do not require as prompt a response. Immediate action to relieve the tension is mandatory. This can be done by inserting a needle or catheter into the second or third intercostal spaces in the midclavicular line, with the patient supine. A three-way stopcock on the syringe is an added refinement.

**211. The answer is a.** (*Behrman, 16/e, p 1279, McMillan, 3/e, p 1309, Rudolph, 21/e, p 1944.*) Bacterial tracheitis is an uncommon but severe and life-threatening sequella of a viral laryngotracheobronchitis. The typical story is that presented in the case, with several days of viral upper respira-

tory symptoms, followed by an acute elevation of temperature and an increase in respiratory distress. Children may also present acutely and without the initial viral symptoms. The differential must include epiglottitis; the lack of drooling and dysphagia help make this a case of tracheitis. Management includes establishing an airway with endotracheal intubation and IV antibiotics. Special attention is focused on preservation of the airway, as even intubated children with tracheitis can have secretions thick and copious enough to occlude the airway.

**212. The answer is d.** (Behrman, 16/e, pp 1275–1276. McMillan, 3/e, pp 572, 1093, 1310. Rudolph, 21/e, p 1276.) In the past, this disease was most commonly caused by invasive *Haemophilus influenzae* type B. Due to the widespread use of the Hib vaccine, this condition is now more commonly caused by group A  $\beta$ -hemolytic streptococcus, *Moraxella catarrhalis*, or *Streptococcus pneumoniae*. Epiglottitis is a life-threatening form of infection-produced upper airway obstruction. The course of the illness is brief and prodromal symptoms are lacking. There is a sudden onset of sore throat, high fever, and prostration that is out of proportion to the duration of the illness. Drooling and difficulty in swallowing, a muffled voice, and preference for a characteristic sitting posture with the neck hyperextended may be noted. Unless preparations are made for immediate intubation by skilled personnel, any attempt to visualize the epiglottis should be avoided. Morbidity and mortality are usually related to a delay in establishing an airway early in the disease. Therefore, radiography of the neck, which may delay definitive treatment, is unwise.

**213. The answer is c.** (Behrman, 16/e, p 1068, McMillan, 3/e, pp 1199–1202. Rudolph, 21/e, pp 1110–1111.) The presentation described is characteristic of visceral larva migrans from infestation with a common parasite of dogs, *Toxocara canis*. Dirt-eating children ingest the infectious ova. The larvae penetrate the intestine and migrate to visceral sites, such as the liver, lung, and brain, but do not return to the intestine, so the stools do not contain the ova or parasites. The diagnosis can be made by a specific enzyme-linked immunosorbent assay (ELISA) for *Toxocara*.

**214. The answer is b.** (Behrman, 16/e, p 1264. McMillan, 3/e, pp 1278–1284. Rudolph, 21/e, pp 1263–1264.) Maxillary and ethmoid sinuses are large enough to harbor infections from infancy. Frontal sinuses are

rarely large enough to harbor infections until the sixth to tenth year of life. Sphenoid sinuses do not become large until about the third to fifth year of life. In general, a cold lasting longer than 10 to 14 days with fever and facial pain is indicative of sinusitis. Examination of the nose can reveal pus draining from the middle meatus in maxillary, frontal, or anterior ethmoid sinusitis. Pus in the superior meatus indicates sphenoid or posterior ethmoid sinuses. Diagnosis is on clinical grounds and can be difficult. Positive findings on plain sinus films in a symptomatic child are supportive of sinusitis. CT scans are more sensitive, but are usually reserved for the more complicated cases. The treatment is usually oral antibiotics for 10 to 14 days. Decongestants and antihistamines have not been shown to be helpful or necessary.

**215. The answer is d.** (*Behrman, 16/e, pp 1275–1278. McMillan, 3/e, pp 1307–1311. Rudolph, 21/e, pp 1275–1276.*) The signs of illness described are those involving the airway above the point at which the trachea enters the neck and leaves the thorax, as in croup syndrome. Intrathoracic airway diseases, such as asthma or bronchiolitis, produce breathing difficulty on expiration, with expiratory wheezing, prolonged expiration, and signs of air trapping due to the increased narrowing during expiration as the airways are exposed to the same intrathoracic pressure changes as the alveoli. The extrathoracic airway, to the contrary, tends to collapse on inspiration, producing the characteristic findings this patient demonstrates.

Agents causing croup include parainfluenza types I and III, influenza A and B, RSV, and occasionally, other viruses. Treatment is usually supportive, but racemic epinephrine and corticosteroids reduce the length of time in the emergency room and hospitalizations.

**216. The answer is e.** (*Behrman, 16/e, p 656. McMillan, 3/e, pp 575, 2044. Rudolph, 21/e, pp 1956, 1959.*) Therapeutic serum levels of theophylline are 10 to 20  $\mu\text{g}/\text{mL}$ . Below these levels, a therapeutic effect may not be obtained, and above them, toxicity supervenes, with nausea, vomiting, cardiac arrhythmias, convulsions, and death. The narrow therapeutic window, its possible relationship to increased morbidity when combined with some  $\beta$  agonists, and low efficacy, have reduced this drug's popularity, but it is still listed as an alternative in the most recent asthma guidelines from the National Asthma Education and Prevention group of the NIH. Thus, it is important to know the effects of prescribing additional drugs for patients

taking theophylline and of giving theophylline to patients taking these drugs. Erythromycin and cimetidine decrease the clearance of theophylline and increase its serum levels. Phenytoin and rifampin have the opposite effect. Penicillin has no effect on theophylline levels.

**217. The answer is e.** (Behrman, 16/e, pp 762–765, 795, 885–897, 914–915. McMillan, 3/e, pp 893–894, 1026–1039, 1227–1230. Rudolph, 21/e, pp 949–959, 965–967, 1980–1982.) The radiograph shown has reticulonodular opacities in the right upper lobe and prominence of the right hilum (lymphadenopathy) consistent with *Mycoplasma pneumoniae*. Infections with *M. pneumoniae* are common in older children and young adults. Although the infection typically produces lower-lobe bronchopneumonia, its effects are characteristically nonspecific, and it can produce lobar pneumonia as well. It can produce upper respiratory infection, pharyngitis, otitis media and externa, bronchiolitis, hemolytic anemia, and Guillain-Barré syndrome. Treatment of choice is a macrolide antibiotic. *Staphylococcus aureus* is seen in infants less than 6 months of age; *Haemophilus influenzae* is rarely seen with the widespread use of *H. influenzae* B vaccine. At this age, a child with *Mycobacterium tuberculosis* presents with hilar adenopathy and localized broncholobar pneumonia in the upper lobe most typically, and *Streptococcus pneumoniae* causes the sudden onset of high fever and a lobar pneumonia, often with pleural effusion. A TB skin test for this patient would be a good idea.

**218. The answer is b.** (Behrman, 16/e, pp 686–688. McMillan, 3/e, p 2070. Rudolph, 21/e, pp 821–822.) Treatment is similar to that of an insect sting in an allergic child. If respiratory distress is severe, intubation is necessary. Additional treatment can include bagging with oxygen and administration of epinephrine, plasma expanders, diphenhydramine, and cimetidine as indicated by the clinical course of the patient. Additional treatment such as administration of corticosteroids should be started early, but the effect will be delayed. Tourniquet therapy is not indicated and is potentially harmful.

**219. The answer is c.** (Behrman, 16/e, pp 1127–1128. Rudolph, 21/e, pp 1397–1398.) Many types of objects produce esophageal obstruction in young children, including small toys, coins, and food. Most are usually lodged below the cricopharyngeal muscle at the level of the aortic arch. Initially, the foreign

body may cause a cough, drooling, and choking. Later, pain, avoidance of food (liquids are tolerated better), and shortness of breath can develop. Diagnosis is by history (as outlined in the question) and by radiographs (especially if the object is radiopaque). The usual treatment is removal of the object via esophagoscopy.

**220–223. The answers are 220-e, 221-c, 222-a, 223-d.** (*Behrman, 16/e, pp 204–210, 743–744, 799–801, 1248–1250, 1479–1483. McMillan, 3/e, pp 1450–1451. Rudolph, 21/e, pp 1531–1534.*) Fever, cough, and tachypnea in a patient with sickle cell anemia can be manifestations of pneumonia, pulmonary thromboemboli, or sepsis. Aside from being relatively common in patients with sickle cell anemia, these diseases can be rapidly progressive and quickly fatal. It is, therefore, important for the patient to be evaluated and treated on an emergency basis. The treatment requires hospitalization because it will almost certainly include systemic antibiotics, intravenous fluids, oxygen, and, perhaps, blood transfusion.

In order to see cyanosis, there must be about 5 g of unoxygenated hemoglobin in the skin capillaries. In anemia this may not be possible, as the total hemoglobin level can be beneath that. In addition, dark skin pigmentation and poor lighting contribute to making cyanosis an unreliable negative sign. With anemia and pulmonary disease, it should be assumed that the patient has impaired oxygenation.

The low pH in the arterial blood can be called acidemia. In this context, it is likely that the hydrogen ions come from lactic acid produced by anaerobic metabolism in tissues with inadequate oxygen delivery. Inadequate oxygenation is caused by the low  $PO_2$ , the low oxygen-carrying capacity of the blood (Hb 5 g/dL), and circulatory inadequacy due to the sickling itself and to the vascular disease it produces. The low  $PCO_2$  reflects the hyperventilation, which is secondary to the respiratory difficulty, and to the anemia, and is also respiratory compensation for the metabolic acidosis.

Administration of 100% oxygen will rapidly raise alveolar oxygen concentration and, in the absence of substantial right-to-left shunting of blood, will fully saturate the arterial hemoglobin. It will also dissolve 0.003 mL of oxygen per mmHg of oxygen partial pressure in each deciliter of blood. This will serve to decrease the tissue hypoxia and increase the concentration of mixed venous oxygen, which may decrease the amount of sickling. The other choices are all undesirable or contraindicated.

**224. The answer is c.** (Behrman, 16/e, p 1266. McMillan, 3/e, pp 572–573, 1299–1301. Rudolph, 21/e, p 1944.) Suppurative infection of the chain of lymph nodes between the posterior pharyngeal wall and the prevertebral fascia leads to retropharyngeal abscesses. The most common causative organisms are *Staphylococcus aureus*, group A  $\beta$ -hemolytic streptococci, and oral anaerobes. Presenting signs and symptoms include a history of pharyngitis, abrupt onset of fever with severe sore throat, refusal of food, drooling, and muffled or noisy breathing. A bulge in the posterior pharyngeal wall is diagnostic, as are radiographs of the lateral neck that reveal the retropharyngeal mass. Palpation (with adequate provision for emergency control of the airway in case of rupture) reveals a fluctuant mass. Treatment should include incision and drainage if fluctuance is present.

**225–226. The answers are 225-d, 226-e.** (Behrman, 16/e, p 920. McMillan, 3/e, pp 451–453, 896–897. Rudolph, 21/e, pp 930, 2372.) Chlamydiae, sexually transmitted among adults, are spread to infants during birth from genitally infected mothers. The sites of infection in infants are the conjunctivae and the lungs, where chlamydiae cause inclusion conjunctivitis and afebrile pneumonia, respectively, in infants between 2 and 12 weeks of age. Diagnosis is confirmed by culture of secretions and by antibody titers. In adolescents, chlamydial infections may be a cause of cervicitis, salpingitis, endometritis, and epididymitis and appear to be an important cause of tubal infertility. The most common treatment for this condition includes macrolide antibiotics orally, which clears both the nasopharyngeal secretions when a conjunctivitis is present and prevents the pneumonia that can occur later. Topical treatment for chlamydia conjunctivitis is not effective in clearing the nasopharynx.

**227. The answer is b.** (Behrman, 16/e, pp 1279–1282. McMillan, 3/e, pp 570–571, 640. Rudolph, 21/e, pp 1277–1278.) Recurrent pneumonias in an otherwise healthy child should indicate the potential for anatomic blockage of an airway. In the patient in this question, the findings on clinical examination suggest a foreign body in the airway. Inspiratory and expiratory films can be helpful. Routine inspiratory films are likely to appear normal or near normal (as outlined in the question). Expiratory films will identify air trapping behind the foreign body. It is uncommon for the foreign body to be visible on the plain radiograph; a high index of suspicion is necessary to make the diag-

nosis. Suspected foreign bodies in the airway are potentially diagnosed with fluoroscopy, but rigid bronchoscopy is not only diagnostic but also the treatment of choice for removal of the foreign body.

**228–230. The answers are 228-c, 229-d, 230-b.** (*Behrman, 16/e, pp 204–210, 1248–1250. Rudolph, 21/e, pp 278–284, 1656–1657.*) The laboratory results of row c indicate a striking respiratory alkalosis. This could be secondary to voluntary hyperventilation or inappropriate respirator settings for a patient on a ventilator and is also typical of acute hyperventilation syndrome secondary to anxiety. Such a patient can complain of dyspnea, chest pain, tingling, and dizziness and can even have generalized convulsions secondary to low ionized calcium levels. Rebreathing into a paper bag can be both therapeutic and diagnostic.

The blood gases of row d are the only ones given that are relatively normal except for a low oxygen partial pressure. The mild respiratory alkalosis and metabolic acidosis can be a consequence of the hypoxia. These results could be obtained in a patient with moderately severe pneumonia, bronchiolitis, or asthma, secondary to ventilation-perfusion inequality, with some areas of the lung underventilated with respect to perfusion. This cause of hypoxia can be easily corrected by giving the patient relatively small increases in oxygen concentration to breathe. These results would also be typical of findings in patients with right-to-left shunting of blood, as in tetralogy of Fallot, in which case, giving oxygen would not help.

The results in row a show fairly severe metabolic acidosis with respiratory compensation and without hypoxia. These would be typical for someone in early shock and would most commonly be seen in children with diarrhea. Treatment of this type of acidosis is hydration.

The blood gases of row b demonstrate an uncompensated respiratory acidosis with hypoxia but with no metabolic acidosis. This is compatible with acute hypoventilation, which could be produced by a tension pneumothorax, for example. This can be treated easily by placing a needle or catheter in the pleural space and evacuating the air.

**231–234. The answers are 231-a, 232-c, 233-b, 234-d.** (*Behrman, 16/e, pp 204–210, 1248–1250, 1267–1268. Rudolph, 21/e, pp 278–284, 1656–1657.*) The data on row a indicate severe acidemia and severe hypoxia with a marked respiratory acidosis and metabolic acidosis. These are manifestations of severe ventilatory failure, probably accompanied by circulatory failure or

cardiac arrest. This mandates the most aggressive therapy, including assisted ventilation with administration of high oxygen levels. Other measures to restore circulation and improve the acidemia are also indicated.

The results of row c show metabolic alkalosis. The high  $\text{PCO}_2$  and low  $\text{PO}_2$  result from compensatory hypoventilation. This can all be secondary to excessive body potassium losses from diuretics.

The results in row b indicate respiratory acidosis with metabolic compensation, indicating chronic upper airway obstruction. A common cause of chronic hypoventilation in children is hypertrophied tonsils and adenoids, which may be an indication for tonsillectomy or adenoidectomy (or both).

The blood gases of row d are impossible in a patient breathing room air. The  $\text{PCO}_2$  cannot go up without the  $\text{PO}_2$  dropping roughly proportionately. An increase in  $\text{PCO}_2$  of 20 mmHg from 40 to 60 mmHg should, therefore, produce a fall in  $\text{PO}_2$  from 90 to 70 mmHg. The test should be repeated after the blood gas equipment has been checked and recalibrated.

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# The Gastrointestinal Tract

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## Questions

**DIRECTIONS:** Each item below contains a question or incomplete statement followed by suggested responses. Select the **one best** response to each question.

**235.** A 4-year-old boy presents with a history of constipation since the age of 6 months. His stools, produced every 3 to 4 days, are described as large and hard. Physical examination is normal; rectal examination reveals a large ampulla, poor sphincter tone, and stool in the rectal vault. The plain film of his abdomen is shown. The next step in the management of this child would be



*(Courtesy Susan John, M.D.)*

- a. Lower GI barium study
- b. Parental reassurance and dietary counseling
- c. Serum electrolyte measurement
- d. Upper GI barium study
- e. Initiation of synthroid

**236.** A 10-year-old boy has been having bellyaches for about 2 years. They occur at night as well as during the day. Occasionally, he vomits after the onset of pain. Occult blood has been found in his stool. His father also gets frequent stomachaches. The most likely diagnosis is

- a. Peptic ulcer
- b. Appendicitis
- c. Meckel diverticulum
- d. Intussusception
- e. Pinworm infestation

**237.** A 5-year-old child is noted to be iron deficient. Upon questioning, his family reports several episodes of grossly bloody (maroon) stools. Which of the following is a likely explanation for his anemia?

- a. Increased demands for iron due to accelerated growth
- b. Meckel diverticulum
- c. Infestation by hookworm
- d. Peptic ulcer
- e. Recurrent epistaxis

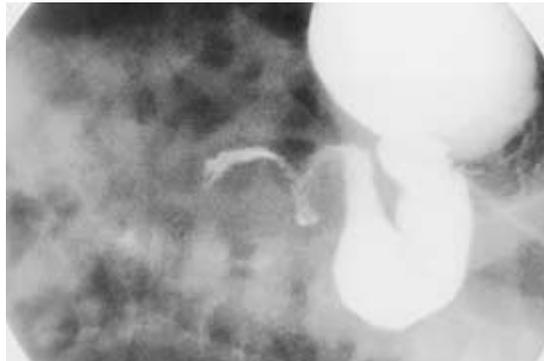
**238.** A 6-month-old infant has large, foul-smelling stools and is not gaining weight. Sweat chloride level was 68 mmol/L. Appropriate nutritional recommendations include

- a. Supplementation with water-soluble vitamins
- b. Pancreatic enzyme replacement
- c. High-fat diet
- d. Low-protein diet
- e. Low-carbohydrate diet

**239.** A 1-week-old previously healthy infant presents to the emergency room with the acute onset of bilious vomiting. The abdominal plain film in the emergency department (**A**) and the barium enema done after admission (**B**) are shown. The most likely diagnosis for this patient is



**A** (Courtesy Susan John, M.D.)



**B** (Courtesy Susan John, M.D.)

- a. Jejunal atresia
- b. Hypertrophic pyloric stenosis
- c. Malrotation of the intestines with volvulus
- d. Appendicitis
- e. Intussusception

**240.** A 14-month-old infant receives chronic total parenteral nutrition for short gut syndrome. Which of the following is the most common complication?

- a. Sepsis
- b. Liver disease
- c. Pulmonary emboli
- d. Air emboli
- e. Irreversible atrophy of the mucosa of the small intestine

**241.** A 4-week-old boy presents with a 10-day history of vomiting that has increased in frequency and forcefulness. The vomitus is not bile stained. The child feeds avidly and looks well, but he has been losing weight. An ultrasound of the abdomen is shown. The most likely diagnosis is



- a. Pyloric stenosis
- b. Small intestinal obstruction
- c. Gastroenteritis
- d. Intussusception
- e. Brain tumor

**242.** A 3-year-old child presents to your office for an evaluation of constipation. The mother notes that since birth and despite frequent use of stool softeners, the child has only about one stool per week. He does not have fecal soiling or diarrhea. He was born at term and without pregnancy complications. The child stayed an extra day in the hospital at birth because he did not stool for 48 h, but has not been in the hospital since. Initial evaluation of this child should include

- a. A child psychiatry evaluation for stool retention and parenting assistance
- b. A barium enema and rectal manometry
- c. Plain films of the abdomen
- d. Dietary log and observation
- e. Beginning oral antispasmodic medication

**243.** A 14-year-old boy has sickle cell disease. He presents to the emergency room with the complaints of increased jaundice, abdominal pain, nausea, vomiting, and fever. His examination is remarkable for jaundice, pain of the right upper quadrant with guarding, and a clear chest. Chest radiographs appear normal. The test most likely to reveal the cause of this pain is

- a. Serum chemistries
- b. Complete blood count with platelets and differential
- c. Ultrasound of the right upper quadrant
- d. Upper GI series
- e. Hepatitis panel

**244.** An 8-year-old is accidentally hit in the abdomen by a baseball bat. After several minutes of discomfort, he seems to be fine. Over the ensuing 24 h, however, he develops a fever, abdominal pain radiating to the back, and persistent vomiting. On examination, the child appears quite uncomfortable. The abdomen is tender, with decreased bowel sounds throughout, but especially painful, with guarding in the midepigastic region. The test likely to confirm your suspicions is

- a. Serum amylase
- b. Complete blood count with differential and platelets
- c. Serum total and direct bilirubin levels
- d. Abdominal radiograph
- e. Electrolyte panel

**245.** A 10-month-old boy, recently arrived from Guyana, has a 5-h history of crying, with intermittent drawing up of his knees to his chest. On the way to the emergency room he passes a loose, bloody stool. He has had no vomiting and has refused his bottle since the crying began. Physical examination is noteworthy for an irritable infant whose abdomen is very difficult to examine because of constant crying. His temperature is 38.8°C (101.8°F). The rectal ampulla is empty, but there is some gross blood on the examining finger. The most helpful study in the immediate management of this patient would be

- a. Stool culture
- b. Examination of the stool for ova and parasites
- c. An air-contrast enema
- d. Examination of the blood smear
- e. Coagulation studies

**246.** A 12-month-old girl has been spitting up her meals since 1 month of age. Her growth is at the ninety-fifth percentile, and she is otherwise asymptomatic and without findings on physical examination. The most likely diagnosis is

- a. Pyloric stenosis
- b. Partial duodenal atresia
- c. Hypothyroidism
- d. Gastroesophageal reflux disease
- e. Tracheoesophageal fistula

**247.** A 14-year-old girl has a 9-month history of diarrhea, abdominal pain (usually periumbilical and postprandial), fever, and weight loss. She has had several episodes of blood in her stools. Which of the following is the most likely diagnosis in this child?

- a. Chronic appendicitis
- b. Chronic pancreatitis
- c. Crohn disease
- d. Bulimia
- e. Gallstones

**248.** A 6-month-old infant has eaten a diet with the following content and intake for the past 5 months: protein 4% of calories, fat 50% of calories, carbohydrates 46% of calories, calories 105 per kilogram of body weight per day. The patient's disturbance is

- a. Rickets
- b. Marasmus
- c. Obesity
- d. Tetany
- e. Kwashiorkor

**249.** A 5-month-old child regularly regurgitates a large portion of her feeds. A pH probe study showed significant periods of low esophageal pH. The child has normal growth, and no other significant past medical history. The best management at this point is

- a. Barium swallow and upper GI series
- b. Oral reflux medications
- c. Esophageal manometry
- d. Close observation only
- e. Surgical correction with fundoplication

**250.** A 2-year-old presents to the emergency center with several days of rectal bleeding. The mother first noticed reddish-colored stools 2 days prior to arrival and has since changed several diapers with just blood. The child is afebrile, alert, and playful, and is eating well without emesis. He is slightly tachycardic, and his abdominal examination is normal. The best diagnostic study at this time is

- a. Exploratory laparotomy
- b. Barium enema
- c. Ultrasound of the abdomen
- d. Radionucleotide scan
- e. Stool culture

**251.** A 15-year-old otherwise healthy boy presents with a complaint of intermittent abdominal distention, crampy abdominal pain, and excessive flatulence. He first started noticing these symptoms when he moved into his father's house, and his stepmother insisted on milk at dinner every night. He has normal growth, has not lost weight, and has no travel history. The most appropriate diagnostic study to diagnose his condition would be

- a. Barium swallow and upper GI
- b. Hydrogen excretion in breath after oral administration of lactose
- c. Esophageal manometry
- d. Stool pH
- e. Serum lactose levels

**252.** A 6-week-old infant is admitted to the hospital with jaundice. Her outpatient blood work demonstrated a total bilirubin of 12 mg/dL with a direct portion of 3.5 mg/dL. Which of the following disorders is likely to be responsible?

- a. ABO incompatibility
- b. Choledochal cyst
- c. Rh incompatibility
- d. Gilbert disease
- e. Crigler-Najjar syndrome

**253.** An 11-year-old child has been diagnosed with hepatitis C infection. Which of the following may be considered in the treatment of chronic hepatitis C infection?

- a. Active vaccination
- b. Interferon
- c. Gamma globulin
- d. Zidovudine
- e. Low-protein diet

**254.** An awake, alert infant with a 2-day history of diarrhea presents with a depressed fontanelle, tachycardia, sunken eyes, and the loss of skin elasticity. The appropriate percentage of dehydration is

- a. Less than 1%
- b. 1 to 5%
- c. 5 to 9%
- d. 10 to 15%
- e. More than 20%

**255.** A 9-month-old is brought to the emergency center by ambulance. The child had been having emesis and diarrhea with decreased urine output for several days, and the parents noted that she was hard to wake up this morning. Her weight is 9 kg, down from 11 kg the week prior at her 9-month checkup. You note her heart rate and blood pressure to be normal. She is lethargic, and her skin is noted to be “doughy.” After confirming that her respiratory status is stable, you send electrolytes, which you expect to be abnormal. You start an IV. The best solution for an initial IV bolus would be

- a.  $\frac{1}{4}$  normal saline (38.5 meq sodium/L)
- b. D10 water (100 g glucose/L)
- c. Normal saline (154 meq sodium/L)
- d. 3% saline (513 meq sodium/L)
- e. Fresh-frozen plasma

**256.** In the preceding case, management for this child should include

- a. Slow rehydration over 48 h
- b. Rapid volume expansion with  $\frac{1}{4}$  NS
- c. Packed red blood cells
- d. CT scan to evaluate for CNS tumor causing SIADH
- e. Urinary electrolytes

**257.** A mother of a 6-month-old infant is concerned that her baby may be teething. You explain to her that the first teeth to erupt in most children are the

- a. Mandibular central incisors
- b. Maxillary lateral incisors
- c. Maxillary first molars
- d. Mandibular cuspids (canines)
- e. First premolars (bicuspid)

**258.** The 4-year-old child pictured is noted to have the tooth decay shown. This characteristic pattern of tooth decay is due to



- a. Excessive use of fluoride
- b. Tetracycline
- c. Use of bottled water that lacks fluoride
- d. Caries from nursing with a baby bottle
- e. Consumption of too much candy

**259.** An 8-year-old boy is brought to your office with the complaint of abdominal pain. The pain is worse during the week and seems to be less prominent during the weekends and during the summer. The patient's growth and development are normal. The physical examination is unremarkable. Laboratory screening, including stool for guaiac, complete blood count, urinalysis, and chemistry panel, yields normal results. The next step in the care of this patient should be to

- a. Perform an upper GI series
- b. Perform CT of the abdomen
- c. Administer a trial of H<sub>2</sub> blockers
- d. Observe the patient and reassure the patient and family
- e. Recommend a lactose-free diet

**260.** A 55-day-old infant born 27 weeks prematurely is shown. The swelling is not tender, firm, hot, or red, and it does not transilluminate. It seems to resolve with pressure, but returns when the infant begins to perform the Valsalva maneuver. The most appropriate course of action at this point is to



- Obtain a surgical consultation
- Perform a needle aspiration
- Order a barium enema
- Order a KUB (plain radiographs of kidney, ureter, and bladder)
- Observe the patient and reassure the patient and family

**261.** A 12-year-old girl was hit in the face by a baseball 15 min earlier and has had her mandibular incisors knocked out. Which of the following represents an accurate plan of action?

- The teeth should be rinsed in hot water
- Foreign matter adhering to the teeth should be immediately scrubbed off
- The teeth may be transported in tea, juice, or cola
- Avulsed teeth can be transported in the mouth of the older, cooperative pediatric patient if they cannot be reinserted at the scene
- A dental appointment should be made within 72 h

**262.** An 18-month-old infant is found with the contents of a bottle of drain cleaner in his mouth. Which of the following treatment options is appropriate?

- a. Emesis is the immediate emergency treatment
- b. Endoscopic examination is indicated within the first 12 to 24 h
- c. Decontamination by activated charcoal is effective
- d. Neutralization by drinking a solution of the opposite pH is effective
- e. Having the patient drink copious amounts of milk or water is effective

**263.** A 16-year-old male, despondent over a recent breakup, takes an unknown quantity of an unknown material he found at home to commit suicide. He is brought to the emergency center by his parents within 30 min of the ingestion. For which of the following household materials and medications should he be given activated charcoal as part of his emergency center treatment?

- a. Drain cleaner
- b. Ethylene glycol
- c. Bleach
- d. Phenobarbital
- e. Lithium

**DIRECTIONS:** Each group of questions below consists of lettered options followed by a set of numbered items. For each numbered item, select the appropriate lettered option(s). Each lettered option may be used once, more than once, or not at all. **Choose exactly the number of options indicated following each item.**

### Items 264–268

For each of the patient descriptions below, choose the best initial diagnostic step in the evaluation of the patient's apparent gastrointestinal hemorrhage.

- a. Abdominal series
- b. Fiberoptic endoscopy
- c. Apt test
- d. Routine stool culture
- e. Barium enema
- f. No immediate intervention
- g. Meckel scan
- h. Stool culture on sorbitol-MacConkey medium

**264.** A newborn infant, the product of an emergency cesarean section, is 24 h old and has a grossly bloody stool. She looks well otherwise. **(SELECT 1 DIAGNOSIS)**

**265.** A 7-day-old premature infant born at 26 weeks' gestation now has a grossly bloody stool, abdominal distention, and increasing oxygen requirements. **(SELECT 1 DIAGNOSIS)**

**266.** A 2-year-old has crampy abdominal pain and grossly bloody diarrheal stool, but no fever. His abdominal examination reveals no masses. A family member, who ate at the same local hamburger shop the night prior, has the same symptoms. **(SELECT 1 DIAGNOSIS)**

**267.** A 7-year-old has been vomiting for 2 days and has had diarrhea for 1 day. He now notes that he has small streaks of blood in his emesis. The rest of his family has had similar symptoms. **(SELECT 1 DIAGNOSIS)**

**268.** A 10-year-old has complained for 1 month of intermittent epigastric pain that awakens him from sleep. He notes that eating food sometimes helps. He reports black stools during the prior week, and also admits that he has occasionally vomited frank blood. **(SELECT 1 DIAGNOSIS)**

**Items 269–274**

For each presented child, choose the appropriate vitamin or trace element replacement therapy or therapies to treat the described condition.

- a. Vitamin A
- b. Vitamin B<sub>12</sub>
- c. Vitamin C
- d. Vitamin D
- e. Vitamin E
- f. Iron
- g. Vitamin K
- h. Folate
- i. Niacin

**269.** A 5-year-old caucasian child has a history of poor growth, fatty stools, nasal polyps, chronic cough, and frequent pulmonary infections. **(SELECT 4 VITAMINS OR TRACE ELEMENTS)**

**270.** A 9-month-old infant, who has been fed cow's milk for 4 months, is tachycardic and pale. **(CHOOSE 1 VITAMIN OR TRACE ELEMENT)**

**271.** A 3-day-old infant born at home is brought to the emergency center with bloody stools, hematemesis, and purpura. His circumcision is oozing blood. **(CHOOSE 1 VITAMIN OR TRACE ELEMENT)**

**272.** A 17-month-old toddler has been irritable over the last month. She now refuses to walk and seems to have tenderness in both of her legs. She has had a low-grade fever, and she has petichiae on her skin and mucous membranes. She has a small cut that has not healed well. Radiographs of the legs reveal generalized bony atrophy with epiphyseal separation. **(CHOOSE 1 VITAMIN OR TRACE ELEMENT)**

**273.** A 4-year-old whose diet consists mostly of cheese puffs and cola begins to have problems walking at night, complaining that he cannot see well. In addition, his skin has become dry and scaly, and he has complained of headache for a month. **(CHOOSE 1 VITAMIN OR TRACE ELEMENT)**

**274.** An exclusively breast-fed 2-year-old is brought to the emergency center with pain in his right leg after a fall. Physical examination reveals a small child with a 3-cm anterior fontanelle, a flattened occiput, a prominent forehead, significant dental caries, bumpy ribs, and bowed extremities. Radiographs reveal a greenstick fracture at the site of pain, along with fraying at the distal ends of the femur. **(CHOOSE 1 VITAMIN OR TRACE ELEMENT)**

**Items 275–278**

For each description below, select the disorder with which it is most likely to be associated.

- a. Behçet disease
- b. Colic
- c. Short bowel syndrome
- d. Gluten-sensitive enteropathy
- e. Cystic fibrosis
- f. Wolman disease
- g. Reye syndrome

**275.** A 6-year-old child with vomiting, seizures, coma, and liver failure 6 days after a varicella infection **(SELECT 1 DISORDER)**

**276.** Foul-smelling, bulky stools in a 3-month-old child with a history of failure to thrive **(SELECT 1 DISORDER)**

**277.** A 1-month-old otherwise healthy child with daily periods of increased crying, fussiness, and passage of flatus during the evening hours **(SELECT 1 DISORDER)**

**278.** An irritable 8-month-old child with diarrhea and poor weight gain each time cereal is introduced into his diet **(SELECT 1 DISORDER)**

# The Gastrointestinal Tract

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## Answers

**235. The answer is b.** (*Behrman, 16/e, pp 1105, 1138–1141. McMillan, 3/e, pp 313–315, 1637–1639, 2281–2282. Rudolph, 21/e, pp 1368–1370.*) The radiograph demonstrates a stool-filled megacolon. Hirschsprung disease is usually suspected in the chronically constipated child despite the fact that 98% of such children have functional constipation. Finding a dilated, stool-filled anal canal with poor tone on the physical examination of a well-grown child supports the diagnosis of functional constipation. The difficulty in treating functional constipation once it has been established emphasizes the need for prompt identification and treatment of problems with defecation and for counseling of parents regarding proper toileting behavior. The extensive workup of this patient would likely be negative and expensive, and is not indicated.

**236. The answer is a.** (*Behrman, 16/e, pp 1147–1150. McMillan, 3/e, pp 1195, 1642–1655, 1702–1704. Rudolph, 21/e, pp 1105–1106, 1405–1408, 1433–1434, 1450–1451.*) The presence of nocturnal abdominal pain and gastrointestinal bleeding and a positive family history support a diagnosis of peptic ulcer disease. Pain is the most common symptom. Symptoms often persist for several years before diagnosis. The increased incidence of peptic ulcer disease in families (25 to 50%) and concordance in monozygotic twins suggest a genetic basis for the disease. Antibiotic treatment for *Helicobacter pylori* in patients not responding to conventional therapy can cure this disease in some patients. Appendicitis and intussusception are acute events. Pinworms produce perianal pruritus but do not commonly cause abdominal pain or other serious problems. Meckel diverticulum causes painless rectal bleeding, usually during early childhood.

**237. The answer is b.** (*Behrman, 16/e, pp 1065–1066, 1137–1138, 1147–1150, 1260, 1469–1471. McMillan, 3/e, pp 1447–1448, 1642–1652. Rudolph, 21/e, pp 1108–1109, 1261, 1405–1406, 1433–1434.*) Iron deficiency is most common between 6 months and 3 years of age. During this period,

the need for iron is increased because of accelerated growth, and this need is often not met when the diet consists predominantly of cow's milk, a uniquely iron-poor food. Growth plateaus during the preschool and preadolescent years and the mixed diets of children in this period are more likely to provide them with adequate iron. Chronic loss of blood from the intestinal tract or from nosebleeds can quickly deplete the iron stores of children. In adolescence, there are increased requirements for iron because of rapid growth. The adolescent female has the additional risk of becoming iron-deficient from the loss of menstrual blood.

Grossly positive blood in the stool of this patient requires further evaluation. The Meckel diverticulum results from an embryologic remnant of the vitelline duct. Gastric tissue in the area ulcerates, causing intermittent bleeding. Blood can be microscopic or massive and grossly positive. The bleeding is often painless and is intermittent. The diverticulum can serve as a lead point for intussusception with resultant symptoms of obstruction. The diagnosis is with a Meckel scan (Tc99m pertechnetate nuclear scan), which demonstrates a lesion in the right lower quadrant.

Hookworms are caused by two species of roundworm, *Necator americanus* and *Ancylostoma duodenale*. Symptoms include abdominal pain, weakness, dizziness, eosinophilia, pica, guaiac positive stools, and anemia. Gross bleeding and melena are rare.

Peptic ulcer disease often presents with epigastric pain, discomfort, vomiting, anorexia, early satiety, and weight loss. Bleeding is usually noted as hematemesis or melena. Bright red blood from the rectum is unusual without severe disease.

Epistaxis causes anemia of chronic blood loss, but the blood in the stool is usually only guaiac positive or is melanotic. Bright red blood from the rectum from nosebleeds is distinctly abnormal.

**238. The answer is b.** (Behrman, 16/e, pp 1315–1327. McMillan, 3/e, pp 1242–1254. Rudolph, 21/e, pp 1967–1979.) The goal in treatment of patients with cystic fibrosis is to supply enough calories to maintain normal growth. Extra calories are needed to replace those lost as a result of gastrointestinal losses, to offset the catabolic and anorectic effects of respiratory infection, and for catch-up growth. Almost 90% of patients have poor digestion of fat and protein as a result of the deficiency of pancreatic function, which may be partially corrected by oral pancreatic enzyme replacement; protein and fat losses are reduced, and patients are usually able to tolerate a reasonably

normal diet. The high caloric density of fat is useful in meeting the patient's caloric needs. Supplementation with fat-soluble vitamins (A, D, E, and K) is necessary. With severe disease, feedings by nasogastric tube, percutaneous enterostomy, or intravenous nutrition can be necessary.

**239. The answer is c.** (*Behrman, 16/e, pp 1135–1136. McMillan, 3/e, pp 311–312. Rudolph, 21/e, pp 1400–1402.*) The plain films demonstrate dilated stomach and proximal loops of bowel. The cross-table upper GI demonstrates a “curly Q” twist of barium as it passes through the malrotated portion of bowel. Malrotation results when incomplete rotation of the intestines occurs during embryologic development. The most common type of malrotation is failure of the cecum to move to its correct location in the right lower quadrant. Most patients present in the first weeks of life with bilious vomiting indicative of bowel obstruction and/or intermittent abdominal pain. Acute presentation, similar to that in the question, is caused by a volvulus of the intestines. The diagnosis is confirmed by radiographs; barium contrast studies (upper GI and/or enema) demonstrate malposition of the cecum in the vast majority of cases. Treatment is surgical.

**240. The answer is a.** (*Behrman, 16/e, p 1165. McMillan, 3/e, pp 332–335. Rudolph, 21/e, pp 1347–1348, 1427–1429.*) Parenteral nutrition, particularly when a central line is used, has a relatively high risk of complications, so it should not be used without good indication. The most common complication, and one of the most serious, is sepsis (1 to 5% of patients). In addition to the complications mentioned, thrombosis of central veins, perforations of veins with infusion and bleeding into the pleural space and other sites, pneumothorax, brachial plexus injury, and skin sloughing can also occur. Small intestinal mucosal atrophy is reversed with oral feedings. Awareness of these complications and the practice of careful techniques may minimize the risks of this sometimes life-saving nutritional procedure.

**241. The answer is a.** (*Behrman, 16/e, pp 1130–1131. McMillan, 3/e, p 311. Rudolph, 21/e, p 1402.*) A history of nonbilious vomiting of 10 days' duration in a child who does not look ill points to infantile hypertrophic pyloric stenosis as the most likely diagnosis. The ultrasound demonstrates the thickened pylorus. The incidence of this condition in infants is between 1:200 and 1:750, with males affected more often than females. Although there is no specific pattern of inheritance, a familial incidence has been

observed in about 15% of patients. Information about predisposing ancestry is conflicting, as is data concerning the assertion of a first-born predilection. Metabolic alkalosis with low serum potassium and chloride levels is frequently seen in pyloric stenosis as a result of loss of gastric contents from vomiting.

**242. The answer is b.** (*Behrman, 16/e, pp 1139–1141. McMillan, 3/e, pp 313–315. Rudolph, 21/e, pp 1461–1463.*) The diagnosis of Hirschsprung's disease should be suspected in a child with intractable chronic constipation without fecal soiling (approximately 3% have fecal soiling). In contrast, overflow diarrhea caused by leakage of the unformed fecal stream around a rectal impaction is common in functional constipation. A neonatal history of delayed passage of meconium is often obtained, and the infant can continue to be constipated and to have bouts of abdominal distention and vomiting. The infant is also at risk of developing enterocolitis, a life-threatening consequence of the partial obstruction. Radiologic study by barium enema and rectal manometry are accurate diagnostic tools. Identification of an aganglionic segment of bowel by punch or suction biopsy can establish the diagnosis. Histochemical tissue examination showing increased amounts of acetylcholinesterase and an absence of ganglia cells is confirmatory. Rectal manometric studies have shown that in aganglionic megacolon, the usual relaxation of the internal rectal sphincter in response to balloon inflation does not occur. Surgery is indicated as soon as the diagnosis is made.

**243. The answer is c.** (*Behrman, 16/e, pp 1213, 1479–1483. McMillan, 3/e, pp 1450–1451, 1736–1737. Rudolph, 21/e, pp 1507–1508.*) Cholecystitis and cholelithiasis are unusual diseases in children and are usually associated with predisposing disorders such as hemolytic anemia. Pain of the right upper quadrant, nausea, vomiting, fever, and jaundice are symptoms of acute cholecystitis. The diagnosis is confirmed with an ultrasound of the gallbladder.

**244. The answer is a.** (*Behrman, 16/e, pp 1191–1193. McMillan, 3/e, pp 1711–1712. Rudolph, 21/e, pp 1466–1467.*) The causes of pancreatitis in children are varied, with about one-fourth of cases without predisposing etiology and about one-third of cases as a feature of another systemic disease. Traumatic cases are usually due to blunt trauma to the abdomen.

Acute pancreatitis is difficult to diagnose; a high index of suspicion is necessary. Common clinical features include severe pain with nausea and vomiting. Tenderness, guarding or rebound pain, abdominal distention, and a paralytic ileus are signs and symptoms often seen. No diagnostic test is completely accurate. An elevated total serum amylase with the correct clinical history and signs and symptoms is the best diagnostic tool. Plain films of the abdomen exclude other diagnoses; ultrasonography of the pancreas can reveal enlargement of the pancreas, gallstones, cysts, and pseudocysts. Supportive care is indicated until the condition resolves.

**245. The answer is c.** (*Behrman, 16/e, pp 1142–1143. McMillan, 3/e, pp 1652–1655. Rudolph, 21/e, pp 1407–1408.*) The usual presentation of intussusception is that of an infant between 4 and 10 months of age who has a sudden onset of intermittent colicky abdominal pain. The child can appear normal when the pain abates, but as it recurs with increasing frequency, the child can begin to vomit and become progressively more obtunded. The passage of stool containing blood and mucus, and resembling currant jelly, is often observed. Early examination of the abdomen can be unremarkable, but as the problem persists, a sausage-shaped mass in the right upper quadrant is frequently palpated. A contrast enema examination under fluoroscopic control can be therapeutic as well as diagnostic when the hydrostatic effects of the column of air serve to reduce the intussusception. Early diagnosis prevents bowel ischemia. The cause of most intussusceptions is unknown, but a Meckel's diverticulum or polyp can serve as a lead point.

**246. The answer is d.** (*Behrman, 16/e, pp 1125–1126. McMillan, 3/e, pp 320–321. Rudolph, 21/e, pp 1389–1393.*) Gastroesophageal reflux is a common pediatric complaint, often seen in the first 1 to 2 months of life and resolving by 1 to 2 years of age. About 7% of children have reflux severe enough to come to medical attention, and only 2% of that group requires investigation. For most children, conservative treatment (small feeds, thickened formula, avoiding high-fat meals, etc.) suffices. A smaller number need pharmacologic therapy, and an even smaller number require surgery.

**247. The answer is c.** (*Behrman, 16/e, pp 1154–1157. McMillan, 3/e, pp 1674–1681. Rudolph, 21/e, pp 1441–1444.*) The presentation of Crohn depends on the location and extent of lesions. Onset of the gastrointestinal or extraintestinal symptoms is insidious. The common presentation is as

described. Crohn disease (granulomatous colitis) characteristically is associated with transmural, granulomatous intestinal lesions that are discontinuous and can appear in both the small and large intestines. Although Crohn disease can first appear as a rectal fissure or fistula, the rectum is often spared. Arthritis/arthralgia occurs in a minority (11%) of affected children. Other extraintestinal symptoms include erythema nodosum or pyoderma gangrenosum, liver disease, renal calculi, uveitis, anemia, specific nutrient deficiency, and growth failure. In relation to the general population, the risk of colonic carcinoma in affected persons is increased but not nearly to the degree associated with ulcerative colitis.

**248. The answer is c.** (*Behrman, 16/e, pp 169–171. McMillan, 3/e, p 1764. Rudolph, 21/e, pp 1336–1337.*) The syndrome of kwashiorkor is caused by a diet that is deficient in protein, leading to low serum albumin, which causes decreased plasma volume and increased interstitial fluid or edema. The term *marasmus* refers to a combined inadequacy of protein and energy in which the deficiency is dominated by the lack of food in general. The minimum requirement for protein is 8% of the total daily calories when the protein provided is from high-quality animal sources. These patients have a high death rate from intercurrent infections.

**249. The answer is d.** (*Behrman, 16/e, pp 1125–1126. McMillan, 3/e, pp 320–321. Rudolph, 21/e, pp 1390–1393.*) Barium swallow and upper GI series are helpful in detecting anatomic abnormalities such as antral web, pyloric stenosis, malrotation, and annular pancreas. To confirm the presence of gastroesophageal reflux (GER), the best test is the esophageal pH probe, which measures the frequency and duration of falls in pH, thereby indicating acid reflux. Esophageal manometry is used to identify poor tone of the lower esophageal sphincter. In this child, who is growing well and developing properly and has no other medical problems such as reflux-induced apnea or bradycardia or aspiration pneumonia, pharmacologic treatment is not necessary, as most infants will outgrow this common problem of esophageal reflux. Surgical correction is reserved for infants with poor weight gain or reflux-associated apnea and bradycardia who have failed pharmacologic therapy.

**250. The answer is d.** (*Behrman, 16/e, pp 1137–1138. McMillan, 3/e, pp 1644–1645. Rudolph, 21/e, pp 1405–1406.*) The child described has a typical

presentation for Meckel diverticulum. Children with this condition usually present with painless rectal bleeding in the first 2 years of life but can have symptoms throughout the first decade. The embryonic duct connecting the yolk sac to the intestine can fail to regress completely and persist as a diverticulum attached to the ileum. The lining of this Meckel diverticulum usually contains acid-secreting gastric mucosa that can produce ulcerations of the diverticulum itself or the adjacent ileum. Bleeding, perforation, or diverticulitis can occur. More seriously, the diverticulum can lead to volvulus of itself and of the small intestine, and it can also undergo eversion and intussusception. Diagnosis can be made by technetium scan, and treatment is by surgical excision. Barium studies do not readily reveal the diverticulum, nor do plain films.

**251. The answer is b.** (*Behrman, 16/e, pp 1168, 1175–1176. McMillan, 3/e, pp 1698–1699. Rudolph, 21/e, p 1424.*) Lactase is a disaccharidase localized in the brush border of the intestinal villous cells. It hydrolyzes lactose to its constituent monosaccharides, glucose and galactose. Intestinal lactase levels are usually normal at birth in all populations; however, lactase deficiency is a common, genetically predetermined condition. Sucrose, also a disaccharide, is a nonreducing sugar composed of glucose and fructose that is hydrolyzed by the brush border enzyme sucrase. Lactase activity is not readily increased by the oral administration of substrate or the inclusion of lactose in the diet. The clinical symptoms of lactose malabsorption are due to the presence of osmotically active, undigested lactose, which may act to increase intestinal fluid volume, alter transit time, and produce the symptoms of abdominal cramps, distention, and, occasionally, watery diarrhea. Bacterial metabolism of the nonabsorbed carbohydrates in the colon to carbon dioxide and hydrogen may contribute to the clinical symptoms. Acquired lactase deficiency is often associated with conditions of the gastrointestinal tract that cause intestinal mucosal injury (e.g., sprue and regional enteritis).

Diagnostic techniques for lactose intolerance include removal of the offending sugar, with a reproduction of symptoms upon reintroduction. Although the ingestion of even small amounts of lactose can be diagnostic if gastrointestinal symptoms occur, the measurement of breath hydrogen is more specific, as it is not affected by glucose metabolism or gastric emptying. Similarly, an acidic stool pH in the presence of reducing substances would be diagnostic. Direct measurement of enzyme levels combined with

histologic evaluation helps to differentiate an acquired (secondary versus primary) lactase deficiency in which the intestinal histology is normal.

**252. The answer is b.** (*Behrman, 16/e, pp 1198–1201. McMillan, 3/e, pp 205–206, 315–319. Rudolph, 21/e, p 1482.*) Obstructive jaundice (i.e., direct-reacting bilirubin greater than 20% of the total) requires investigation in all infants. Cystic fibrosis and alpha<sub>1</sub>-antitrypsin deficiency should be considered in the diagnostic evaluation of any child with obstructive jaundice. Other diseases to be excluded are galactosemia, tyrosinemia, and urinary tract or other infections, including toxoplasmosis, cytomegalovirus, rubella, syphilis, and herpesvirus. Ultrasound examination to rule out choledochal cyst may be included with a <sup>99</sup>Tc hepatic iminodiacetic acid (HIDA) scan to assess the patency of the biliary tree. Liver biopsy can assist in the diagnosis by providing a histologic diagnosis (e.g., hepatitis), tissue for enzyme activity (i.e., inborn error of metabolism), or tissue for microscopic determination of storage diseases. ABO and Rh incompatibility could cause direct hyperbilirubinemia if there were brisk hemolysis at birth, which would then lead to inspissated bile syndrome. All of the other causes listed typically lead to indirect hyperbilirubinemia.

**253. The answer is b.** (*Behrman, 16/e, pp 773–774. McMillan, 3/e, pp 449, 1726. Rudolph, 21/e, p 1499.*) Compared with treatment of chronic hepatitis in adults, little is known about optimal treatment of these life-threatening conditions in children. For chronic hepatitis B and C infection, interferon  $\alpha$  and ribavirin have shown promise. The long-term response to these agents remains unknown; ongoing studies may clarify the role of these agents in the treatment of this condition.

**254. The answer is c.** (*Behrman, 16/e, p 213. McMillan, 3/e, p 69. Rudolph, 21/e, p 1645.*) A moribund state is characteristic of a loss of greater than 10% of body weight from dehydration. The other findings are characteristic of a loss of body weight of 5 to 9% when there is no hypernatremia. Additional findings at this level of dehydration can be restlessness, absent or reduced tears, weak radial pulses, and, possibly, orthostatic hypotension.

**255–256. The answers are 255-c, 256-a.** (*Behrman, 16/e, pp 212–215. McMillan, 3/e, pp 69–72. Rudolph, 21/e, p 1326.*) The description is that of a child with hypernatremia (in this case, the child's sodium was 170 meq/dL);

the “doughy” skin is often seen in hypernatremia. The extracellular fluid and circulating blood volumes tend to be preserved with hypernatremic dehydration at the expense of the intracellular volume. Therefore, hypotension may not be observed, and neither are the other signs of circulatory inadequacy that are typical of isotonic or hypotonic dehydration. Signs suggesting involvement of the central nervous system (such as irritability or lethargy) are characteristic of hypertonic dehydration. Slow correction of this hypernatremia (over 24 to 48 h) prevents significant fluid shifts and increased intracranial pressure. The story in the case is not consistent with SIADH, and packed cells are not indicated with the information given. Initial bolus therapy should be with isotonic fluid such as normal saline or lactated Ringer’s solution. Blood products such as fresh-frozen plasma are not indicated, and hypotonic solutions such as D10W and 1/4 NS could cause rapid fluid shifts, resulting in cerebral edema and death. Hypertonic (3%) saline is used in the event of seizures caused by rapid rehydration, along with other measures typically used to reduce cerebral edema.

**257. The answer is a.** (*Behrman, 16/e, pp 37, 1109. McMillan, 3/e, pp 642–643. Rudolph, 21/e, p 1285.*) In general, mandibular teeth erupt before maxillary teeth; teeth tend to erupt in girls before boys. The first teeth to erupt usually are the mandibular central incisors at 5 to 7 months, followed by the maxillary central incisors at 6 to 8 months. Lateral incisors (mandibular then maxillary) erupt next at 7 to 11 months, followed by the first molars (10 to 16 months), the cuspids (16 to 20 months), and the second molars (20 to 30 months).

**258. The answer is d.** (*Behrman, 16/e, pp 1114–1116. McMillan, 3/e, pp 651–652, 1287–1288. Rudolph, 21/e, p 1289.*) The pattern of dental disease found with the use of bottles that contain high concentrations of sugars, which promotes dental disease, includes extensive maxillary decay (especially frontal) and posterior maxillary and mandibular decay, but essentially normal mandibular frontal teeth. Prevention of this disease is possible through counseling families to avoid the use of fruit juices in bottles or sweetened pacifiers. Genetic predisposition seems to play a role as well.

**259. The answer is d.** (*Behrman, 16/e, pp 1176–1178. McMillan, 3/e, pp 1685–1686. Rudolph, 21/e, pp 1357–1363.*) Recurrent abdominal pain is a common complaint occurring in at least 10% of school-aged children. In

children older than 2 years, less than 10% of cases have an identifiable organic cause. Management of these children is difficult and frustrating for the physician and the family. Excessive testing and treatments are contraindicated. A thorough history and physical examination, including growth parameters, are frequently helpful in separating organic from nonorganic causes of abdominal pain. Any signs or symptoms of organic causes should be pursued. If nothing in the history or physical examination is found, as is likely in the case described, reassurance of the children and family members is indicated. Close follow-up for new or changing symptoms as well as further reassurance to the family is important.

**260. The answer is a.** (*Behrman, 16/e, pp 1185–1188. McMillan, 3/e, pp 1640–1642. Rudolph, 21/e, p 1742.*) Inguinal hernias are commonly seen in premature infants (up to 16 to 25%). Incarceration is common; elective repair is often considered prior to hospital discharge. The diagnosis is so common that rarely are diagnostic tests indicated.

**261. The answer is d.** (*Behrman, 16/e, pp 1117–1118. McMillan, 3/e, pp 655–656. Rudolph, 21/e, pp 1303–1304.*) The earlier that permanent teeth are replanted, the greater the rate of success, going down from 90% in the first 30 min to 5% after 2 h. The rate of success is a function of the integrity of the periodontal ligament. The teeth can be rinsed in cold water, but not brushed, to avoid damage to the root and periodontal ligament. Milk is a good transport medium if the child is uncooperative or for some other reason the teeth cannot be reinserted at the scene. Teeth may also be transported in the mouth of the older, cooperative patient or the parent. The immediate application of acrylic splints is needed to keep the teeth in place, so immediate dental attention is required. The need for full tetanus immunization should be evaluated. A completely avulsed primary tooth is generally not replaced, to avoid damage to the underlying permanent tooth.

**262. The answer is b.** (*Behrman, 16/e, pp 1126–1127. McMillan, 3/e, pp 618–619. Rudolph, 21/e, pp 1395–1396.*) Endoscopic examination of the esophagus and stomach is a diagnostic method of determining the extent of the mucosal injury. Vomiting is to be avoided since it would expose the mucosal surfaces to the caustic agent a second time. The child can be given small amounts of milk or water, but large amounts, which might cause vomiting, are unwise. Neutralization of the caustic can result in an exother-

mic reaction and produce a thermal burn. The use of steroids after endoscopy in second-degree chemical burns of the esophagus has been effective in diminishing the inflammatory response in some patients. Optimal treatment is still controversial and requires expert consultation or review of the most current literature. Charcoal, however, does not absorb the alkaline agent in drain cleaner.

**263. The answer is d.** (*Behrman, 16/e, pp 2161–2162, 2167. McMillan, 3/e, pp 618–619. Rudolph, 21/e, p 359.*) The absorption of certain toxins from the gastrointestinal tract is diminished by the use of activated charcoal administered during the first few hours after the ingestion in the minimum dose of 1 g/kg of body weight or 5 to 10 times the quantity of the ingested material. Activated charcoal exerts its effect by adsorbing particles of toxin on its surface. Compounds not adsorbed include alcohols, acids, ferrous sulfate, strong bases (such as drain cleaners and oven cleaners), cyanide, lithium, and potassium. For drugs with an enterohepatic circulation (e.g., phenobarbital and tricyclic antidepressants), or those with prolonged absorption (e.g., sustained-release theophylline), the use of multiple-dose activated charcoal can be effective in decreasing the half-life and increasing the total body clearance of the toxic substance.

**264–268. The answers are 264-c, 265-a, 266-h, 267-f, 268-b.** (*Behrman, 16/e, pp 512–513, 527, 851, 1127, 1149–1150. McMillan, 3/e, pp 325–332, 360, 967–968, 1644, 1646–1652. Rudolph, 21/e, pp 140–143, 987, 1372, 1432–1434.*) Swallowed maternal blood can be differentiated from neonatal hemorrhage by the Apt test. The Apt test, initially described by Leonard Apt and William Downey in 1953, distinguishes fetal hemoglobin from adult hemoglobin based on the specimen's reaction to alkali (fetal hemoglobin is unchanged, whereas adult hemoglobin changes to hematin). Infants may swallow blood during delivery or from a cracked nipple during breast-feeding.

Necrotizing enterocolitis is a life-threatening condition seen mostly in premature infants. Although the precise etiology is unknown, contributing factors include gastrointestinal tract ischemia, impaired host immunity, the presence of bacterial or viral pathogens, and the presence of breast milk or formula in the gut. Findings include bloody stools, abdominal distension, hypoxia, acidosis, and emesis. The initial diagnostic test of choice is plain film radiographs. The characteristic finding in necrotizing enterocolitis

(NEC) is pneumatosis intestinalis; free air in the peritoneum may also be seen. Perforation is a surgical emergency.

Enterohemorrhagic *Escherichia coli* are pathogens found in poorly cooked beef, and some have been responsible for outbreaks of bloody diarrhea that were well-publicized in the media. These organisms secrete shiga toxin. Routine stool cultures do not isolate this particular pathogen; the lab must use sorbitol-MacConkey agar to isolate the bacteria. Enzyme assays for shiga toxin are becoming available as well.

Forceful emesis can result in small tears in the esophagus, termed Mallory-Weiss syndrome. This is usually a benign condition, only occasionally resulting in significant blood loss. In a patient who is otherwise stable, diagnostic procedures are not indicated.

Peptic ulcer disease can result in hematemesis and melena, along with the typically epigastric abdominal pain. Children can have both chronic and acute blood loss associated with an ulceration. Fiberoptic endoscopy is the diagnostic method of choice. An upper gastrointestinal series can sometimes reveal an ulcer as well.

**269–274. The answers are 269-a,d,e,g, 270-f, 271-g, 272-c, 273-a, 274-d.** (Behrman, 16/e, pp 176–178, 182–187, 526–527, 1325, 1469–1471. McMillin, 3/e, pp 471, 473–474, 1246, 1253, 1447–1448, 1487. Rudolph, 21/e, pp 200–202, 1322–1333, 1525–1528, 2156.) Children with cystic fibrosis have pancreatic insufficiency, and, thus, have deficiencies in the fat-soluble vitamins A, D, E, and K. A commercial preparation combines these vitamins for use in cystic fibrosis and other malabsorptive diseases.

Cow's milk contains an insufficient quantity of iron to sustain normal red blood cell production. Therefore, children whose primary caloric source is cow's milk are likely to develop iron-deficiency anemia, characterized by microcytosis and hypochromia on the peripheral smear.

Hemorrhagic disease of the newborn is now rare, as the vast majority of newborns receive a vitamin K injection shortly after birth. Classic disease presents within the first week of life and is characterized by hematemesis, hematuria, umbilical stump and circumcision oozing, and purpura.

Vitamin C deficiency impairs wound healing. In its severe form, also termed *scurvy*, children can have diffuse tenderness, which is worse in the legs; evidence of hemorrhage; irritability; low-grade fever; swelling; tachypnea; and poor appetite. Diagnosis is based on clinical picture and radiographic findings; there are no definitive laboratory studies.

Vitamin A deficiency manifests first in visual changes, including night blindness. Deficiency can also cause drying of the conjunctivas and sclera. Skin is frequently dry. Poor growth and impaired cognition are also seen.

Vitamin D deficiency leads to rickets, a failure of bone mineralization, and the clinical picture described in the question. Vitamin D deficiency is usually not a problem in developed parts of the world. In addition to nutritional rickets, there are congenital forms of rickets as well. Despite vitamin D supplementation, these children can have permanent bony disfigurement.

**275–278. The answers are 275-g, 276-e, 277-b, 278-d.** (*Behrman, 16/e, pp 167, 404–405, 1157–1159, 1165–1167, 1214–1216, 1315–1327, 1743. McMillan, 3/e, pp 1242–1254, 1694–1697, 1750, 1957–1958, 2055, 2174–2175. Rudolph, 21/e, pp 414–417, 919, 1490, 1967–1979.*) Reye syndrome is becoming increasingly rare. Its hallmarks are acute encephalopathy and fatty degeneration of the liver. The association of this disease with aspirin use in children having recently suffered a bout with influenza or varicella has resulted in a dramatic decrease in aspirin use in children. No tests are diagnostic of this condition, although elevated liver function studies and serum ammonia levels are helpful. Diagnosis of this condition is dependent on a high index of suspicion. Treatment is supportive.

Cystic fibrosis is a multisystem disease primarily affecting the lungs and the intestines. Respiratory symptoms upon presentation include cough with frequent pneumonias. Intestinal symptoms include large, bulky, foul-smelling stools in the failure-to-thrive infant. It is most common in white children and far less common in Hispanics and blacks. The diagnosis is made with the sweat test and chromosomal analysis.

Colic is a frustrating condition for the child, the parents, and the physician. It commonly occurs before the age of 3 months and is of unknown etiology. The child has the sudden onset of crying for several hours, the abdomen is tense and distended, the legs are drawn up to the abdomen, and the child frequently passes gas. Many theories have been forwarded as to the cause, including swallowed air, milk intolerance, and maternal dietary changes. After a careful history and physical examination reveals no other likely pathology, reassurance to the parents is indicated. A variety of measures such as feeding the child in the upright position (avoiding air swallowing), frequent burping, gentle rocking, assistance in passing stools or flatus, and other measures are commonly tried with a variable response.

A permanent sensitivity to dietary gluten (commonly found in wheat, rye, oats, and barley) results in small-bowel mucosal damage in gluten's presence. Affected children usually present between 1 to 2 years of age with diarrhea, but can also present with failure to thrive (frequently with food avoidance) or vomiting. Clinically, children can have abdominal distention and pallor. Laboratory tests can reveal anemia and hypoproteinemia. Small-bowel biopsy is the gold standard for diagnosis. A lifelong, gluten-free diet is curative.

Behçet syndrome is a vasculitis of small- or medium-sized arteries and veins. Classically, this causes a nondestructive arthritis in multiple joints, especially large joints such as knees. Associated symptoms include fever, erythema nodosum, aphthous stomatitis, uveitis, and central nervous system abnormalities ranging from trivial complaints to pseudotumor cerebri.

Wolman syndrome is a lysosomal acid lipase deficiency. It presents in infancy with hepatosplenomegaly, diarrhea and vomiting, failure to thrive, icterus, and malabsorption. A KUB (plain radiograph of the kidney, ureter, and bladder) shows calcification of the adrenal gland. There is no treatment known for this condition.

Short bowel syndrome is caused by perinatal events including volvulus, gastroschisis, small bowel atresia, necrotizing enterocolitis, meconium ileus, Crohn disease, and trauma. Nutrient malabsorption can occur. High-quality oral feeds, potentially with parenteral nutrition, can be required.

# The Urinary Tract

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## Questions

**DIRECTIONS:** Each item below contains a question or incomplete statement followed by suggested responses. Select the **one best** response to each question.

**279.** A 7-year-old child is noted to have 2+ protein on urinalysis. A 24-h collection of urine reveals a protein excretion of 2.5 g/24 h. A thorough history might reveal ingestion of which of the following medications?

- a. Tetracycline
- b. Streptomycin
- c. Trimethadione
- d. Diazepam
- e. Chlorambucil

**280.** The delivery of a newborn boy is remarkable for oligohydramnios. The pictured infant is also noted to have undescended testes and clubfeet, and to be in respiratory distress. The most likely diagnosis to explain these findings is



*(Courtesy Michael L. Ritchey, M.D.)*

- a. Surfactant deficiency
- b. Turner syndrome
- c. Prune belly syndrome
- d. Hermaphroditism
- e. Congenital adrenal hyperplasia

**281.** A 7-year-old boy suffers multiple injuries as a result of blunt abdominal trauma. Which of the following statements concerning the proper assessment and treatment of the injuries is true?

- a. Most renal injuries require operative intervention
- b. Major vascular injuries require rapid surgical intervention
- c. Rupture of a full bladder is highly unlikely
- d. The renal capsule surrounding the kidney precludes the need for surgical exploration in most cases of traumatic hematocele
- e. Visible blood in the urine following trauma nearly always indicates significant renal damage

**282.** A 1-year-old child presents with failure to thrive, frequent large voids of dilute urine, excessive thirst, and three episodes of dehydration not associated with vomiting or diarrhea. Over the years, other family members reportedly have had similar histories. The likely diagnosis is

- a. Water intoxication
- b. Diabetes mellitus
- c. Diabetes insipidus
- d. Child abuse
- e. Nephrotic syndrome

**283.** An 18-year-old adolescent develops anorexia, polydipsia and polyuria, vomiting, and unexplained fevers. Laboratory evaluation demonstrates glucosuria but normal blood sugar concentration, abnormally high urine pH in the presence of moderate serum hyperchloremic metabolic acidosis, and mild albuminuria with normal serum protein and albumin. The ingested agent most likely to result in these symptoms is

- a. Penicillamine
- b. Lithium
- c. Outdated tetracycline
- d. Nonsteroidal anti-inflammatory drugs (NSAIDs)
- e. Sulfonamide antibiotics

**284.** A 9-year-old girl presents with an elevated creatinine. To further her evaluation, an inulin clearance test is ordered to measure her glomerular filtration rate. Inulin is chosen for this test because it is an exogenous substance that is

- a. Physiologically active
- b. Capable of binding with plasma proteins
- c. Freely filterable at the glomerulus
- d. Secreted by the renal tubule
- e. Reabsorbed by the renal tubule

**285.** After her first urinary tract infection, a 1-year-old has a voiding cystourethrogram with findings as shown. Which of the following is an appropriate treatment option?



*(Courtesy Susan John, M.D.)*

- a. Low-dose daily antibiotics
- b. Immediate surgical reimplantation of the ureters
- c. Weekly urinalyses and culture
- d. Diet low in protein
- e. Early toilet training

**Items 286–287**

**286.** Physical examination of a baby boy shortly after birth reveals a large bladder and palpable kidneys. A voiding cystourethrogram performed on the baby demonstrates an area of obstruction and proximal dilatation of the urethra, ureters, and kidney. He appears to be otherwise normal. Which of the following is the likely diagnosis?

- a. Ureteropelvic junction obstruction
- b. Posterior urethral valve
- c. Prune belly syndrome
- d. Duplication of the collecting system
- e. Horseshoe kidney

**287.** Despite vigorous medical and surgical efforts, this child has an 80% reduction of glomerular filtration rate and elevated levels of blood urea nitrogen (BUN) and creatinine. He is in chronic renal failure. He may eventually exhibit which of the following?

- a. Polycythemia
- b. Rickets
- c. Frequent episodes of hypoglycemia
- d. Early onset of puberty
- e. Metabolic alkalosis

**288.** Funduscopic examination of a 13-year-old girl shows general and focal arteriolar narrowing. A hemorrhage is observed in the left retina, and sclerosis is present. Her blood pressure is 180/110 mmHg. This girl would be likely to exhibit which of the following symptoms or signs?

- a. Multiple cranial nerve palsy
- b. Headache
- c. Hyporeflexia
- d. Increased urine output
- e. Right ventricular hypertrophy

**289.** A 2-year-old boy with undescended testes is referred to a urologist. Surgical correction will probably eliminate the risk of

- a. Testicular malignancy
- b. Decreased sperm count
- c. Torsion of testes
- d. Urinary tract infection
- e. Epididymitis

**290.** A 6-week-old child is being evaluated for a fever of unknown etiology. As part of the laboratory evaluation, a urine specimen was obtained that grew *Escherichia coli* with a colony count of 2000/ $\mu$ L. These findings would be definite evidence of a urinary tract infection if the sampled urine

- a. Has a specific gravity of 1.008
- b. Is from a bag attached to the perineum of an uncircumcised boy
- c. Is from an ileal-loop bag
- d. Is from a suprapubic tap
- e. Is the first morning sample

**291.** A 6-year-old black boy has brown urine and healing impetigo lesions. He presents with hypertension, dyspnea, periorbital edema, and hepatomegaly. The most likely cause of his problem is

- a. IgA nephropathy
- b. Poststreptococcal glomerulonephritis
- c. Idiopathic hypercalciuria
- d. Pyelonephritis
- e. Sexually transmitted disease

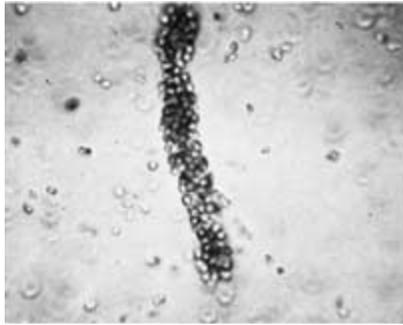
**292.** A 2-year-old boy develops bloody diarrhea shortly after eating in a fast-food restaurant. A few days later, he develops pallor and lethargy; his face looks swollen and his mother reports that he has been urinating very little. Laboratory evaluation reveals low hematocrit and platelet count and positive blood and protein in the urine. Which of the following diagnoses is likely to explain these symptoms?

- a. Henoch-Schönlein purpura
- b. IgA nephropathy
- c. Intussusception
- d. Meckel diverticulum
- e. Hemolytic-uremic syndrome

**293.** A 6-year-old girl is brought to the emergency room because her urine is red. She has been healthy and asymptomatic otherwise. Which of the following conditions would result in a positive dipstick for heme?

- a. Ingestion of blackberries
- b. Ingestion of beets
- c. Phenolphthalein catharsis
- d. Presence of myoglobin
- e. Ingestion of Kool-Aid

**294.** The photomicrograph is of a urine specimen from a 15-year-old girl. She has had intermittent fever, malaise, and weight loss over the previous several months. Recently she has developed swollen hands, wrists, and ankles, the pain of which seems out of proportion to the clinical findings. She also complains of cold extremities with some ulcerations of her distal digits. The laboratory test most likely to assist in the diagnosis of this condition is



- a. Antibodies to nDNA and Sm nuclear antigens
- b. Throat culture for group A  $\beta$ -hemolytic streptococcus
- c. Simultaneously acquired urine and serum bicarbonate levels
- d. A urine culture
- e. Erythrocyte sedimentation rate

**295.** A 5-year-old boy comes to the emergency department at midnight with a complaint of severe scrotal pain since 7 P.M. There is no history of trauma. Your first step is to

- a. Order a surgical consult immediately
- b. Order a radioisotope scan as an emergency
- c. Order a urinalysis and Gram stain for bacteria
- d. Arrange for an ultrasound examination
- e. Order a Doppler examination

**296.** The mother of a 2-year-old girl reports that her daughter complains of burning when she urinates and that she has foul-smelling discharge from her vagina. She has noticed that there is some slight staining on the front of her underwear, but she reports no fever, nausea, vomiting, or other constitutional signs. The child does not attend day care, and she has demonstrated no change in behavior. The physical examination is normal with an intact hymen, but the child's vulva is reddened and with a malodorous scent noted. Her urinalysis and culture are normal. The most likely explanation for this child's condition is

- a. Sexual abuse
- b. Pinworms in the vagina
- c. Chemical vulvovaginitis
- d. Pediculosis pubis
- e. Giardiasis

**297.** A 7-year-old boy has crampy abdominal pain and a rash mainly on the back of his legs and buttocks as well as on the extensor surfaces of his forearms. Laboratory analysis reveals proteinuria and microhematuria. He is most likely to be affected by

- a. Systemic lupus erythematosus
- b. Anaphylactoid purpura
- c. Poststreptococcal glomerulonephritis
- d. Takayasu arteritis
- e. Dermatomyositis

**298.** A 13-year-old boy's scrotum is shown. He complains of several months of swelling but no pain just above his left testicle. He is sexually active but states that he uses condoms. On physical examination, the area in question feels like a "bag of worms." Which of the following is appropriate for this condition?



*(Courtesy Michael L. Ritchey, M.D.)*

- a. Doppler flow study of testes
- b. Radionuclide scan of testes
- c. Urinalysis and culture
- d. Ceftriaxone intramuscularly and doxycycline orally
- e. Reassurance and education only at this time

**299.** A 3-day-old infant's scrotum is shown. Palpation reveals a tense, fluid-filled area surrounding the right testicle. The scrotum transilluminates well, and the amount of fluid does not vary with mild pressure. The appropriate approach to this condition is to



*(Courtesy Michael L. Ritchey, M.D.)*

- a. Request a surgical consultation
- b. Begin furosemide orally
- c. Administer prophylactic antibiotics
- d. Observe only
- e. Perform a chromosome determination

**300.** A 2-year-old patient has microscopic and occasionally gross hematuria. His father has hearing loss and end-stage renal disease. The most likely cause of this child's hematuria is

- a. Alport syndrome
- b. Berger's nephropathy (IgA nephropathy)
- c. Idiopathic hypercalciuria
- d. Membranous glomerulopathy
- e. Goodpasture syndrome

**301.** The parents of a 7-year-old boy complain that they have been unable to potty train their son. Although he is dry during the day, he rarely goes an entire night without wetting the bed. A thorough physical examination and a screening urinalysis are normal. You counsel the family that

- a. The condition is three times more common in girls than boys
- b. Most patients with this condition have a psychiatric illness as the cause
- c. Spontaneous cure rates are high regardless of therapy
- d. Family history of this condition is uncommon
- e. Short courses of desmopressin acetate (DDAVP) lead to permanent cure in 25% of cases

**302.** A 21-year-old woman presents to the emergency room in active labor. She has had no prenatal care, but her last menstrual period was approximately 9 months prior. Her membranes are artificially ruptured, yielding no amniotic fluid. She delivers an 1800-g term infant who develops significant respiratory distress immediately at birth. The first chest radiograph on this infant demonstrates hypoplastic lungs. Which of the following is an appropriate next step for this infant?

- a. Cardiac catheterization
- b. Renal ultrasound
- c. MRI of the brain
- d. Liver and spleen scan
- e. Upper GI

**DIRECTIONS:** Each group of questions below consists of lettered options followed by a set of numbered items. For each numbered item, select the appropriate lettered option(s). Each lettered option may be used once, more than once, or not at all. **Choose exactly the number of options indicated following each item.**

**Items 303–306**

For each condition listed below, match the category to which it belongs.

- a. Nephrotic syndrome
- b. Renal tubular acidosis
- c. Bartter syndrome
- d. Acute glomerulonephritis
- e. IgA nephropathy (Berger nephropathy)
- f. Idiopathic hypercalciuria

**303.** A 4-year-old with early-morning periorbital swelling presents with a serum protein of 1.5 g/dL (**SELECT 1 DISORDER**)

**304.** A 14-year-old with dark urine and a blood pressure of 140/90 mmHg (**SELECT 1 DISORDER**)

**305.** Elevated levels of cholesterol and triglycerides found in a 6-year-old boy who has had unexpected weight gain and scrotal edema (**SELECT 1 DISORDER**)

**306.** An 8-month-old girl whose height and weight are less than the fifth percentile and who has a serum potassium of 2.7 meq/L. (**SELECT 1 DISORDER**)

# The Urinary Tract

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## Answers

**279. The answer is c.** (*Behrman, 16/e, pp 1592–1596. McMillan, 3/e, pp 590–591. Rudolph, 21/e, pp 1691–1696.*) The patient described appears to have nephrotic syndrome (protein excretion of greater than 2 g/24 h). Drug-related nephrotic syndrome has been described in connection with the use of trimethadione, penicillamine, captopril, probenecid, ethosuximide, methimazole, lithium, procainamide, chlorpropamide, phenytoin, paramethadione, tolbutamide, some of the nonsteroidal anti-inflammatory drugs (NSAIDs), and certain heavy metals (e.g., gold, mercury-containing medications).

**280. The answer is c.** (*Behrman, 16/e, pp 1635–1637. McMillan, 3/e, pp 341–342, 1558–1559. Rudolph, 21/e, pp 1737–1738.*) Prune belly syndrome, a malformation that occurs mostly in males, is characterized by a lax, wrinkled abdominal wall, a dilated urinary tract, and intra-abdominal testes. There are additional urinary tract abnormalities including significant renal dysfunction or dysplasia. Oligohydramnios and commonly associated pulmonary complications, such as pulmonary hypoplasia and pneumothorax, are seen frequently. Congenital hip dislocation, clubfeet, and intestinal malrotation with possible secondary volvulus can occur. There does not appear to be a genetic predisposition to prune belly syndrome.

**281. The answer is b.** (*Behrman, 16/e, pp 1654–1655. McMillan, 3/e, p 2214.*) Most injuries to the kidney are mild and do not require surgery but, rather, close observation. Identification of those with the serious injury requiring surgery can be difficult. The CT scan or intravenous pyelogram are helpful to identify serious injury (i.e., those with major vascular injury requiring rapid surgical intervention or those with traumatic hydroceles). Because it is an abdominal organ in children, the bladder, especially when full, is often ruptured by blunt trauma and lower abdominal wounds. Although small bladder tears can be treated conservatively by catheter drainage, surgical exploration is likely to be needed. Extensive urethral injuries can require surgical drainage of periurethral hematoma, primary surgical repair, or even urinary diversion procedures. Most ureteral injuries

require prompt surgical intervention, although such injuries are rare because of the protected position of the ureter. A retrograde cystourethrogram and intravenous urography can be helpful, especially with pelvic fracture or suspected renal trauma.

**282. The answer is c.** (*Behrman, 16/e, pp 1600–1601. McMillan, 3/e, pp 1621–1622, 1789. Rudolph, 21/e, p 1714.*) Nephrogenic diabetes insipidus is a hereditary congenital disorder in which the urine is hypotonic and produced in large volumes because the kidneys fail to respond to antidiuretic hormone. Most North American patients thus involved are descendants of Ulster Scots who came to Nova Scotia in 1761 on the ship Hopewell. Males are primarily affected, apparently through an X-linked recessive modal, though there can be a variable expression in heterozygous females. The defect is unknown, but the disorder is felt to result from primary unresponsiveness of the distal tubule and collecting duct to vasopressin. Although the condition is present at birth, the diagnosis is often not made until several months later, when excessive thirst, frequent voiding of large volumes of dilute urine, dehydration, and failure to thrive become obvious. Maintenance of adequate fluid intake and diet and use of saluretic drugs are the bases of therapy for this incurable disease. Water intoxication would not present with episodes of dehydration; diabetes mellitus rarely presents with a protracted course in such a young child (it is usually a more acute illness and often with vomiting). Child abuse would be unlikely, especially with a family history as noted. Nephrotic syndrome would be expected to present with other signs such as edema, proteinuria, and other symptoms.

**283. The answer is c.** (*Behrman, 16/e, pp 1603–1604. McMillan, 3/e, pp 1613–1615, 2202. Rudolph, 21/e, pp 1674–1677, 1691–1696, 1708–1710.*) The nonspecific findings of anorexia, polydipsia and polyuria, vomiting, and unexplained fevers, along with the more specific laboratory abnormalities of glucosuria but normal blood sugar, abnormally high urine pH in the face of mild or moderate serum hyperchloremic metabolic acidosis, and mild albuminuria in the presence of normal serum protein and albumin suggest Fanconi syndrome (panproximal tubular dysfunction). A number of agents can cause FS including gentamicin (or other aminoglycosides), outdated tetracycline, cephalothin, cidofovir, valproic acid, streptozocin, 6-mercaptopurine, azathioprine, cisplatin, ifosfamide, heavy metals (e.g.,

lead, mercury, cadmium, uranium, platinum), paraquat, maleic acid, and glue sniffing (toluene). The other agents listed are also potentially nephrotoxic. The mechanisms of actions of these agents is through acute tubular necrosis, alteration of renal blood flow, intratubular obstruction, or allergic reactions within the kidney itself. Many of these toxic effects are reduced or eliminated with removal of the offending agent. Nephrotic syndrome is associated with penicillamine; lithium can cause renal tubular acidosis and nephrogenic diabetes insipidus; NSAIDs can cause nephrotic syndrome or interstitial nephritis; and sulfonamide agents can be associated with vasculitis and nephritis (although newer agents tend to have reduced incidence of this).

**284. The answer is c.** (*Behrman, 16/e, pp 1573–1575. McMillan, 3/e, p 73. Rudolph, 21/e, p 1638.*) Inulin (a fructose polymer) is ideal for a clearance test because it is an exogenously administered substance that is freely filtered across the capillary wall of the glomerulus, is not metabolized, is not bound by plasma proteins, and is not secreted or reabsorbed by the renal tubules. Substances that do not possess these properties will result in a falsely elevated or decreased estimates of glomerular function. In practice, creatinine is often used to estimate glomerular filtration rates, but the inulin test will be used when more precise measures are required.

**285. The answer is a.** (*Behrman, 16/e, pp 1625–1629. McMillan, 3/e, pp 1548–1549. Rudolph, 21/e, pp 1671–1673.*) The radiograph shows reflux into the ureters and into the kidney with dilation of the renal pelvis. In children, primary reflux of urine into the ureter and kidney is more common. The higher the reflux into the renal system, especially if the renal pelvis is dilated, the more likely it is for renal damage to occur. Low-grade lesions (grade 1 and grade 2) are conservatively managed with close observation, daily low-dose antibiotics, and urinalyses and cultures every 3 to 4 months. Higher-grade lesions (grade 4 and grade 5) ultimately may require surgical reimplantation of a ureter if the findings persist. Those lesions in between these two extremes are treatment dilemmas.

**286. The answer is b.** (*Behrman, 16/e, pp 1636–1637. McMillan, 3/e, pp 1549–1551. Rudolph, 21/e, p 1737.*) The constellation of findings described points to posterior urethral valves. The clinical picture may range from that described in the question to severe renal obstruction with renal failure and

pulmonary hypoplasia. Urinary tract infections are common complications in older children, and sepsis can occasionally be the presenting sign in afflicted newborns. Despite early recognition and correction of the obstruction, the prognosis for normal renal function is guarded. Prenatal diagnosis is often accomplished secondary to ultrasound diagnosis.

**287. The answer is b.** (*Behrman, 16/e, pp 1609–1611. McMillan, 3/e, pp 1568–1572. Rudolph, 21/e, pp 1718–1728.*) Renal osteodystrophy, which occurs in chronic renal failure, is a designation that encompasses osteitis fibrosa cystica, caused by secondary hyperparathyroidism, and rickets, or osteomalacia, caused by deficiency of 1,25(OH)<sub>2</sub>D<sub>3</sub>; they can occur together. Growth retardation, rather than precocious puberty, occurs in chronic renal failure due to malnutrition (poor appetite, low somatomedin C level), renal osteodystrophy, excessive urinary loss of water and sodium, metabolic acidosis, and anemia. Bone age is lower than chronologic age. Normocytic, normochromic anemia is seen in advanced chronic renal failure, mainly as a result of a relative deficiency of erythropoietin; its other causes include shortened life span of red blood cells in uremic patients, gastrointestinal blood loss caused by abnormal platelet function, iatrogenic blood loss during hemodialysis and from frequent blood sampling, folic acid deficiency, dialysis-related aluminum toxicity, and poor appetite. Glucose intolerance results from peripheral resistance to endogenous insulin. Elevated glucagon and growth hormone levels may be contributing factors. Neurodevelopmental problems occur, especially when uremia develops during early infancy. Brain growth is subnormal and gross motor and developmental delays occur. In untreated children, progressive encephalopathy ensues.

**288. The answer is b.** (*Behrman, 16/e, pp 1450–1455. McMillan, 3/e, pp 1433–1434. Rudolph, 21/e, pp 1877–1878.*) Hypertension is usually asymptomatic, but with marked hypertension, children can develop headache, dizziness, visual disturbances, irritability, and nocturnal waking. Hypertensive encephalopathy can be preceded or accompanied by vomiting, hyperreflexia, ataxia, and focal or generalized seizures. Facial nerve palsy can be the sole manifestation of severe hypertension. Isolated left ventricular hypertrophy, decreased urine output, and abdominal bruits may be possible. When marked fundal changes are present or when there are signs of vascular compromise, emergency treatment of the accompanying hyper-

tension is warranted. Such hypertensive persons require immediate hospitalization for diagnosis and therapy.

**289. The answer is c.** (*Behrman, 16/e, pp 1650–1651. McMillan, 3/e, pp 1555–1556. Rudolph, 21/e, pp 1740–1742.*) At 1 year of age, 0.7% of boys born at term still have cryptorchidism. In adults with cryptorchidism, the risk of testicular malignancy is much higher than in unaffected men. Orchiopexy does not eliminate this risk, but repositioning the testes makes them accessible for periodic examinations. Whether the testes are brought into the scrotum or not, the sperm count can be reduced. The failure of the testes to develop, and their subsequent atrophy, can be detected by 6 months of age. Torsion of the testis is a potential risk because of the excessive mobility of the undescended testis. Orchiopexy helps to eliminate this problem.

**290. The answer is d.** (*Behrman, 16/e, pp 1622–1623. McMillan, 3/e, pp 420–421. Rudolph, 21/e, p 1668.*) The mechanism by which urine is collected is paramount in determining the interpretation of cultures. No bacteria should grow in a properly obtained urine sample from a suprapubic tap unless infection is present; any bacterial growth at all from a properly obtained suprapubic tap is significant. For children who undergo bladder catheterization, colony counts of  $10^3$  to  $10^4$  are considered significant. Ileal-loop bags are usually contaminated and are not useful. Bag specimens in smaller children (obtained by placing an adhesive, sealed, sterile collection system onto the perineum) can be helpful if the culture is negative. Contamination is common, especially in girls and in uncircumcised boys; positive cultures in this latter group of children, especially if they contain more than one organism, are suspect. In properly obtained midstream clean-catch urines, colony counts greater than  $10^5$  in the asymptomatic child and colony counts of  $10^4$  in the symptomatic child may be significant. Clean-catch urines in an uncircumcised male who cannot retract the foreskin, “bagged” specimens in girls, and “bagged” specimens in uncircumcised boys are suspect.

**291. The answer is b.** (*Behrman, 16/e, pp 1579–1582, 1587. McMillan, 3/e, pp 1583–1586. Rudolph, 21/e, pp 998, 1677–1682.*) Acute poststreptococcal glomerulonephritis follows a skin or throat infection with certain nephritogenic strains of group A,  $\beta$ -hemolytic streptococci. Hematuria often colors

the urine dark, and decreased urinary output can result in circulatory congestion from volume overload with pulmonary edema, periorbital edema, tachycardia, and hepatomegaly. This can be avoided by fluid restriction. Acute hypertension is common and can be associated with headache, vomiting, and encephalopathy with seizures. Pyelonephritis and sexually transmitted diseases can cause bloody urine, but rarely the other symptoms of impetigo as described in the question.

IgA nephropathy is rare in black children and rarely presents with hypertension. Idiopathic hypercalciuria can present with blood and renal stones, but the other findings are distinctly unusual.

**292. The answer is e.** (*Behrman, 16/e, pp 1586–1587. McMillan, 3/e, pp 2204–2206. Rudolph, 21/e, pp 1696–1698.*) Hemolytic-uremic syndrome is characterized by an acute microangiopathic hemolytic anemia, thrombocytopenia from increased platelet utilization, and renal insufficiency from vascular endothelial injury and local fibrin deposition. Ischemic changes result in renal cortical necrosis and damage to other organs such as colon, liver, heart, brain, and adrenal. Laboratory findings associated with hemolytic-uremic syndrome include low hemoglobin level, decreased platelet count, hypoalbuminemia, and evidence of hemolysis on peripheral smear (burr cells, helmet cells, schistocytes). Urinalysis reveals hematuria and proteinuria. A marked reduction of renal function leads to oliguria and rising levels of blood urea nitrogen (BUN) and creatinine. Gastrointestinal bleeding and obstruction, ascites, and central nervous system findings such as somnolence, convulsions, and coma can occur. In the past decade, infection by the verotoxin-producing *Escherichia coli* 0157:H7 has been implicated as a cause of hemolytic-uremic syndrome. This organism is epizootic in cattle. Outbreaks associated with undercooked contaminated hamburgers have been reported in several states. Roast beef, cow's milk, and fresh apple cider have been implicated as well. The Coombs test is not positive in this type of hemolytic anemia.

**293. The answer is d.** (*Behrman, 16/e, p 1577. Rudolph, 21/e, p 1659.*) A number of pH-dependent substances can impart a red color to urine. Use of phenolphthalein, a cathartic agent, or phenindione, an anticoagulant, can cause red urine; ingestion of blackberries or beets also can lead to red coloration ("beeturia"). None of these choices, however, would cause the heme test to be positive. Myoglobin, on the other hand, does test positive

for heme. It is the only choice that can test positive for heme. Hematuria should be confirmed by dipstick testing as well as by microscopic examination of urinary sediment.

**294. The answer is a.** (*Behrman, 16/e, pp 1581–1584, 1597–1599. McMillan, 3/e, pp 1583–1585, 1607–1613, 2162–2165. Rudolph, 21/e, pp 847–850, 1687–1688.*) The figure accompanying the question depicts a red blood cell cast characteristically found in the urine of patients with glomerular disease. The fever, malaise, and weight loss as well as the arthritis involving mainly small joints are common findings of systemic lupus erythematosus (SLE). Raynaud phenomenon resulting in digital ulceration and gangrene in a few patients may also be seen. Not described in this patient is the oft-seen malar rash in a butterfly distribution across the bridge of the nose and the cheeks. Simultaneous urine and serum electrolytes for bicarbonate determination hint at renal tubular acidosis that often presents with failure to thrive and unexplained acidosis, sometimes with repeated episodes of dehydration and anorexia. Poststreptococcal glomerulonephritis can present with red cell casts, but the symptoms usually develop 8 to 14 days after the acute throat infection. Urinary tract infections, especially of the upper system, can result in casts, but they would be expected to consist of white rather than red cells. Elevation of the erythrocyte sedimentation rate may be found in SLE, but this is a nonspecific finding noted in many other chronic and acute inflammatory conditions; its diagnostic usefulness in this situation is limited.

**295. The answer is a.** (*Behrman, 16/e, pp 1651–1652, McMillan, 3/e, pp 1556–1557. Rudolph, 21/e, p 1740–1741.*) The majority of all cases of acute scrotal pain and swelling in boys under 6 years of age are caused by testicular torsion. If surgical exploration occurs within 4 to 6 h, the testes can be saved 90% of the time. Too often, delay caused by scheduling of various imaging modalities and laboratory tests results in an unsalvageable gonad.

**296. The answer is c.** (*Behrman, 16/e, pp 1660–1663, McMillan, 3/e, pp 560–561.*) The symptoms listed are those of vulvovaginitis, with nonspecific (or chemical) vulvovaginitis accounting for 70% of all pediatric vulvovaginitis cases. The discharge in nonspecific vulvovaginitis is usually brown or green and with a fetid odor. The burning with urination occurs because of contact between raw skin and urine. Further history in this case might reveal use of tight-fitting clothing (including rubber pants), prolonged bubble baths

with contamination of the vagina with soap products, use of perfumed lotions in the vaginal area, or improper toilet habits (wiping of fecal material toward rather than away from vagina). Attention to these causative conditions usually results in resolution of the symptoms. The finding of a normal hymen points away from sexual abuse. Pinworms can infest the vagina, but symptoms usually include significant itching of the rectum and vagina. Pediculosis pubis requires pubic hair and, thus, is usually not seen before adolescence. Giardiasis can result in vaginal discharge, but associated symptoms usually include diarrhea and malabsorption syndrome as well. In a sexually active adolescent (or in a sexually abused younger child) a variety of infectious agents such as candida, *Chlamydia trachomatis*, *Trichomonas vaginalis*, *Gardnerella vaginalis*, and *Neisseria gonorrhoeae* would be higher on the differential.

**297. The answer is b.** (Behrman, 16/e, pp 728–729. McMillan, 3/e, pp 2176–2179. Rudolph, 21/e, pp 842–844, 1688–1689.) The rash of anaphylactoid purpura most often involves extensor surfaces of the extremities; the face, soles, palms, and trunk are less often affected. Other significant symptoms include edema, arthralgia or arthritis, colicky abdominal pain with GI bleeding, acute scrotal pain, and renal abnormalities ranging from microscopic hematuria to renal failure. Both systemic lupus erythematosus and dermatomyositis are often accompanied by typical facial rashes (butterfly and heliotrope, respectively). People who have Takayasu arteritis usually do not present with a rash. The scarlatiniform rash characteristic of streptococcal infections generally does not coincide with the development of poststreptococcal nephritis; impetiginous lesions, however, may still be present.

**298. The answer is e.** (Behrman, 16/e, p 1653. McMillan, 3/e, p 1558. Rudolph, 21/e, p 1742.) Varicocele, a common condition seen after 10 years of age, occurs in about 15% of adult males. It results from the dilatation of the pampiniform venous plexus (usually on the left side) due to valvular incompetence of the spermatic vein. Reduced sperm counts are possible with this condition; surgery may ultimately be indicated for infertility problems. Typically, these lesions are not painful but can become tender with strenuous exercise. Its typical “bag of worms” appearance on palpation makes its diagnosis apparent in most cases. For a 13-year-old boy, reassurance and education seem appropriate.

**299. The answer is d.** (*Behrman, 16/e, p 1653. McMillan, 3/e, pp 1557–1558, 1640–1641. Rudolph, 21/e, p 1742.*) The description is that of a hydrocele, an accumulation of fluid in the tunica vaginalis. Small hydroceles usually resolve spontaneously in the first year of life. Larger ones or those that have a variable fluid level with time will likely need surgical repair.

**300. The answer is a.** (*Behrman, 16/e, p 1580. McMillan, 3/e, pp 1599–1601. Rudolph, 21/e, pp 1699–1700.*) The most common type of hereditary nephritis is Alport syndrome. Clinically, patients present with asymptomatic microscopic hematuria, but gross hematuria is also possible. Hearing loss, eventually leading to deafness, is associated with Alport syndrome in 30 to 75% of cases. End-stage renal disease is common by the second or third decades of life. This syndrome is thought to be an X-linked dominant disorder, which explains the more severe course in males.

**301. The answer is c.** (*Behrman, 16/e, pp 72–73. McMillan, 3/e, pp 1559–1600. Rudolph, 21/e, pp 1742–1743.*) Nocturnal enuresis is involuntary voiding at night at an age when control of micturition is expected. It is common in children under the age of 5 years (approximately 20%), resolving in approximately 15% of these cases per year thereafter. A careful history and physical examination will usually identify any potential organic causes. In most cases, no etiology is found. A family history is common. Minimal laboratory testing beyond screening urinalysis is indicated. The condition is more common in boys than in girls. Therapy is aimed at reassurance to the parents that the condition is self-limited, avoidance of punitive measures, and consideration of purchasing a bed-wetting alarm. Spontaneous cure rates are high regardless of therapy. Short courses of DDAVP lead to control in 60 to 75% of cases while the patient takes the medication, but frequently the episodes return when the medication is stopped.

**302. The answer is b.** (*Behrman, 16/e, p 461. McMillan, 3/e, p 1336. Rudolph, 21/e, p 1639.*) Oligohydramnios can cause a number of serious problems in the infant, including constraint deformities (such as clubfoot) and pulmonary hypoplasia. These infants have usually experienced intrauterine growth retardation and frequently have an associated serious renal abnormality. Ultrasound of the kidneys is important to rule out renal involvement as a cause of the oligohydramnios.

**303–306. The answers are 303-a, 304-d, 305-a, 306-c.** (*Behrman, 16/e, pp 1577–1578, 1592–1596, 1601–1602. McMillan, 3/e, pp 1590–1599, 1624–1625. Rudolph, 21/e, pp 1677–1696, 1710–1713.*) Elevated levels of cholesterol and triglycerides are common in nephrotic syndrome because of generalized protein synthesis in the liver (including lipoproteins) and because of a decrease in lipid metabolism due to reduced plasma lipoprotein lipase levels. In the nephrotic syndrome, albumin is lost in the urine and, despite increased hepatic synthesis, serum levels drop. The upper limit of protein excretion in healthy children is 0.15 g/24 h. In nephrotic syndrome, proteinuria can exceed 2.0 g/24 h. When the level drops low enough, the oncotic pressure of the plasma becomes too low to balance the hydrostatic pressure. Plasma volume, therefore, decreases as edema occurs. Endocrine and renal mechanisms then partially compensate by retaining water and salt. Careful monitoring and restriction of water and salt intake are not usually required.

With acute glomerulonephritis, on the other hand, oliguria frequently occurs as a direct consequence of the disease process itself, and, on occasion, it can be profound with virtual anuria for several days. During this period of time, it is vital to monitor and restrict fluid intake lest massive edema, hypervolemia, and even pulmonary edema and death occur.

Hypertension commonly accompanies glomerulonephritis, but only occasionally accompanies nephrotic syndrome. Diuretics are sometimes used in both nephrotic syndrome and glomerulonephritis with temporary effect, but are not curative. A combination of albumin infusions followed by a diuretic also has been used to temporarily decrease the edema in patients with nephrotic syndrome. Because both illnesses are usually self-limited, temporary measures are important.

Bartter syndrome (also known as juxtaglomerular hyperplasia) causes hypokalemia, alkalosis, hyperaldosteronism, normotension, and hyperreninemia. It may be inherited as an autosomal recessive trait. Clinical presentations occurring frequently between 6 to 12 months of age include failure to thrive with constipation, weakness, vomiting, polyuria, and polydipsia. Treatment is aimed at returning the potassium level to normal.

# The Neuromuscular System

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## Questions

**DIRECTIONS:** Each item below contains a question or incomplete statement followed by suggested responses. Select the **one best** response to each question.

**307.** You receive a call from your next-door neighbor about her grandchild who lives in another state. She does not know much detail, but tells you that the child is 12 months old and is “delayed.” You explain to her that, without examining the child and talking with the child’s mother, it is difficult to know for sure what is going on, but among the developmental abnormalities and deviations of early childhood, the one most amenable to early intervention is

- a. Autism
- b. Mental retardation of unknown cause
- c. Psychomotor retardation
- d. Developmental language disorder
- e. Trisomy 21

**308.** A 4-year-old child falls from the back of a three-wheeled vehicle, hitting his head. He experiences no loss of consciousness. In the emergency room, he is alert and oriented without focal findings on examination. He has blood behind his left tympanic membrane. CT scan of the skull is likely to show

- a. Subdural hematoma
- b. Epidural hematoma
- c. Intraventricular hemorrhage
- d. Basilar skull fracture
- e. Hydrocephalus

**309.** The examination of a newborn's back reveals a quarter-size "lump" of soft tissue overlying the lower spine. Evaluation with ultrasound of this lesion may demonstrate

- a. Ebstein pearl
- b. Mongolian spot
- c. Cephalohematoma
- d. Omphalocele
- e. Occult spina bifida

**310.** A 6-month-old child was noted to be normal at birth, but over the ensuing months you have been somewhat concerned about his slowish weight gain and his mild delay in achieving developmental milestones. The family calls you urgently at 7:00 A.M. noting that their child seems unable to move the right side of his body. Which of the following conditions might explain this child's condition?

- a. Phenylketonuria
- b. Homocystinuria
- c. Cystathioninuria
- d. Maple syrup urine disease
- e. Histidinemia

**311.** An 11-month-old girl presents to your emergency department with irritability, fever, and stiff neck. When performing the lumbar puncture in this child, you should

- a. Quickly push the needle in all the way until a pop is felt
- b. Push the needle in until resistance is felt and then withdraw the stylet
- c. Use a needle without a stylet
- d. Twirl the needle repeatedly
- e. Advance the needle by small increments and remove the stylet after each advance to see if cerebrospinal fluid (CSF) comes out

**312.** A 4-year-old child is observed to hold his eyelids open with his fingers and to close one eye periodically, especially in the evening. He has some trouble swallowing his food. He usually appears sad, although he laughs often enough. He can throw a ball, and he runs well. Which of the following is likely to result in the diagnosis?

- a. Muscle biopsy
- b. Creatine phosphokinase (CPK)
- c. Effect of a test dose of endrophonium
- d. Chest x-ray
- e. Antinuclear antibodies (ANAs)

**313.** A 3-year-old child is unconscious, with symmetrically small pupils. These findings occur in which of the following?

- a. Pontine hemorrhage
- b. Phenylephrine overdose
- c. Trauma with resultant third nerve palsy
- d. Atropine-induced coma
- e. Cyclopentolate exposure

**314.** A previously healthy 7-year-old child suddenly complains of a headache and falls to the floor. When examined in the emergency room, he is lethargic and has a left central facial weakness and left hemiparesis with conjugate ocular deviation to the right. The most likely diagnosis is

- a. Hemiplegic migraine
- b. Supratentorial tumor
- c. Todd's paralysis
- d. Acute subdural hematoma
- e. Acute infantile hemiplegia

**315.** A 5-month-old child was normal at birth, but the family reports that the child does not seem to look at them any longer. They also report the child seems to "startle" more easily than he had before. Testing of his white blood cells identifies the absence of hexosaminidase A activity, confirming the diagnosis of

- a. Niemann-Pick disease
- b. Infantile Gaucher's disease
- c. Tay-Sachs disease
- d. Krabbe's disease
- e. Fabry's disease

**316.** The family of a 4-year-old boy has recently moved into your area. The child was recently brought to the emergency department for an evaluation of abdominal pain. Although appendicitis was ruled out in the ED and the child's abdominal pain has resolved, the ED physician requested that the family follow up in your office to evaluate an incidental finding of an elevated creatine kinase. The family notes that he was a late walker (began walking independently about 18 months of age), that he is more clumsy than their daughter was at the same age, and that he seems to be somewhat sluggish when he runs, climbs stairs, rises from the ground after he sits, and rides his tricycle. A thorough history and physical examination are likely to reveal which of the following?

- a. Hirsutism
- b. Past seizure activity
- c. Severe proximal muscle weakness
- d. Cataracts
- e. Enlarged gonads

**317.** A 9-year-old girl is brought to her pediatrician with the complaint of severe, intermittent headaches for the last several months. The physical examination, including a careful neurologic examination, is normal. Which of the following characteristics support the diagnosis of childhood migraine in this patient?

- a. Strong family history of migraine
- b. Frequently isolated to the occipital region
- c. Frequently associated with attention deficit hyperactive disorder
- d. Duration of headache more than 24 h
- e. Persistence of headache after sleep

**318.** Examination of the cerebrospinal fluid of an 8-year-old, mildly febrile child with nuchal rigidity and intermittent stupor shows the following: white blood cells 100/ $\mu$ L (all lymphocytes), negative Gram stain, protein 150 mg/dL, and glucose 15 mg/dL. The most likely diagnosis is

- a. Tuberos sclerosis
- b. Tuberculous meningitis
- c. Stroke
- d. Acute bacterial meningitis
- e. Pseudotumor cerebri

**319.** A 6-year-old child has a somewhat unsteady but nonspecific gait and is irritable. Physical examination reveals a very mild left facial weakness, brisk stretch reflexes in all four extremities, bilateral extensor plantar responses (Babinski reflex), and mild hypertonicity of the left upper and lower extremities; there is no muscular weakness. The most likely diagnosis is

- a. Pontine glioma
- b. Cerebellar astrocytoma
- c. Tumor of the right cerebral hemisphere
- d. Subacute sclerosing panencephalitis
- e. Subacute necrotizing leukoencephalopathy

**320.** A 2-year-old boy was just admitted to the hospital because of a convulsion 6 h ago, followed by coma. The emergency room doctor ordered a CT scan with contrast, which showed enhancement of the basal cisterns by the contrast material. No other history is available. The most likely diagnosis is

- a. Battered child syndrome
- b. Malformation of the vein of Galen
- c. Meningococcal meningitis
- d. Tuberculous meningitis
- e. Eastern equine encephalitis

**321.** A 6-year-old child is hospitalized for observation because of a short period of unconsciousness after a fall from a playground swing. He has developed unilateral pupillary dilatation, focal seizures, recurrence of depressed consciousness, and hemiplegia. Appropriate management would be

- a. Spinal tap
- b. CT scan
- c. Rapid fluid hydration
- d. Naloxone
- e. Gastric decontamination with charcoal

**322.** A 6-year-old boy is brought to the clinic with the complaint of poor growth. His growth curves demonstrate his linear development to be well below the fifth percentile. Upon close questioning, his family reports that he has had headaches in the last 2 or 3 weeks and occasional vomiting. Your physical examination reveals mild papilledema. Which of the following might also be expected to be seen in this patient?

- a. Sixth nerve palsy
- b. Unilateral cerebellar ataxia
- c. Unilateral pupillary dilatation
- d. Unilateral anosmia
- e. Bitemporal hemianopsia

**323.** A 6-year-old boy presents with the sudden onset of ataxia. Which of the following is the likely cause?

- a. Drug intoxication
- b. Agenesis of the corpus callosum
- c. Ataxia telangiectasia
- d. Muscular dystrophy
- e. Friedreich ataxia

**324.** A 9-year-old child has developed headaches that are more frequent in the morning and are followed by vomiting. Over the previous few months, his family has noted a change in his behavior (generally more irritable than usual) and his school performance has begun to drop. Imaging of this child is most likely to reveal a lesion that is

- a. Subtentorial
- b. Supratentorial
- c. Intraventricular
- d. In the spinal canal
- e. In the peripheral nervous system

**325.** A young infant is noted to have facial diplegia and difficulty sucking and swallowing. The child has been colicky, and the maternal grandmother has been treating the child with a mixture of weak tea, rice water, and honey. Which of the following disorders is likely the culprit in this child?

- a. Infantile spinal muscular atrophy
- b. Myasthenia gravis
- c. Congenital myotonic dystrophy
- d. Duchenne muscular dystrophy
- e. Botulism

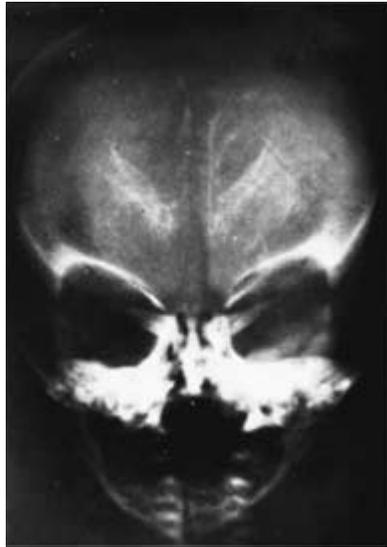
**326.** At birth, an infant is noted to have an abnormal neurologic examination. Over the next few weeks he develops severe progressive central nervous system degeneration, an enlarged liver and spleen, macroglossia, coarse facial features, and a cherry-red spot in the eye. The laboratory finding likely to explain this child's problem is

- a. Reduced serum hexosaminidase A activity
- b. Deficient activity of acid beta-galactosidase
- c. A defective gene on the X chromosome
- d. Complete lack of acid beta-galactosidase activity
- e. Deficient activity of galactosyl-3-sulfate-ceramide sulfatase (cerebroside sulfatase)

**327.** An 18-month-old child is brought to the emergency center after the family reports she had a seizure. They note that the child was in her normal good state of health at bedtime, but when they went to investigate noises they heard coming from her room they found her having "a seizure." They picked her up to put her on the floor and noticed that she was very warm. Her tonic-clonic activity resolved, and by the time they have reached the emergency room she appears to be back to her normal state of good health. Which of the following statements is supportive of this child's having had a simple febrile seizure?

- a. There is usually a mild pleocytosis in the cerebrospinal fluid (CSF)
- b. Such seizures usually occur in association with infections outside the central nervous system
- c. Such seizures often last more than 15 min
- d. Affected children usually are between 2 months and 10 years of age
- e. Focal activity is typical

**328.** The developmentally delayed 6-month-old child in the picture had intrauterine growth retardation (including microcephaly), hepatosplenomegaly, prolonged neonatal jaundice, and purpura at birth. The calcific densities in the skull x-ray shown are likely the result of



- a. Congenital cytomegalovirus infection
- b. Congenital toxoplasmosis
- c. Congenital syphilis
- d. Tuberculous meningitis
- e. Craniopharyngioma

**329.** The infant pictured develops infantile spasms. The disorder most likely to be affecting this infant is



- a. Neurofibromatosis
- b. Tuberous sclerosis
- c. Incontinentia pigmenti
- d. Pityriasis rosea
- e. Psoriasis

**330.** A 4-year-old patient in coma with severe brain injury has developed diabetes insipidus, with urine output of 100 mL/h. The diabetes insipidus can be managed by which of the following?

- a. Replacing excessive urine output with a solution of 5% glucose and normal saline
- b. Fluid restriction
- c. Oral (or nasogastric) hypoglycemic agents
- d. Giving insulin
- e. Giving DDAVP (synthetic vasopressin) intranasally

**331.** A 3-year-old boy's parents complain that their child has difficulty walking. The child rolled, sat, and first stood at essentially normal ages and first walked at 13 months of age. Over the past several months, the family has noticed an increased inward curvature of the lower spine as he walks and that his gait has become more "waddling" in nature. On examination, you confirm these findings and also notice that he has enlargement of his calves. This child most likely has

- a. Occult spina bifida
- b. Muscular dystrophy
- c. Brain tumor
- d. Guillain-Barré syndrome
- e. Botulism

**332.** About 12 days after a mild upper respiratory infection, a 12-year-old boy complains of weakness in his lower extremities. Over several days, the weakness progresses to include his trunk. On physical examination, he has the weakness described and no lower extremity deep tendon reflexes, muscle atrophy, or pain. Spinal fluid studies are notable for elevated protein only. The most likely diagnosis in this patient is

- a. Bell palsy
- b. Muscular dystrophy
- c. Guillain-Barré syndrome
- d. Charcot-Marie-Tooth disease
- e. Werdnig-Hoffmann disease

### **Items 333–334**

**333.** Your 6-year-old son awakens at 1:00 A.M. screaming. You note that he is hyperventilating, is tachycardic, and has dilated pupils. He cannot be consoled, does not respond, and is unaware of his environment. After a few minutes, he returns to normal sleep. He recalls nothing the following morning. The most likely diagnosis is

- a. Seizure disorder
- b. Night terrors
- c. Drug ingestion
- d. Psychiatric disorder
- e. Migraine headache

**334.** The most appropriate approach to this type of patient in the office setting is to

- a. Obtain an EEG
- b. Refer the patient to a child psychiatrist
- c. Begin beta-blocker therapy
- d. Reassure the patient and parents
- e. Obtain a serum and urine drug screen

**DIRECTIONS:** Each group of questions below consists of lettered options followed by a set of numbered items. For each numbered item, select the appropriate lettered option(s). Each lettered option may be used once, more than once, or not at all. **Choose exactly the number of options indicated following each item.**

### Items 335–337

Certain arthropods elaborate venom that, when introduced into the human body, can be clinically important. For each arthropod, select the symptom or sign its venom is most likely to produce.

- a. A 9-year-old otherwise healthy child who has ataxia and areflexia and then has progressive ascending flaccid paralysis
- b. An 11-year-old with dull, crampy pain around the site of a lesion on the legs that over the course of about an hour develops into painful spasms of the abdominal muscles
- c. A 12-year-old with a chronic, necrotizing ulceration on his ankle
- d. An 8-year-old who appears somewhat drunk reports he has a painful lesion on his arm, over the ensuing few hours he becomes irritable, agitated, and complains of blurred vision
- e. An 18-year-old with a creeping eruption of the skin of his legs after a spring break trip to Mexico

**335.** Scorpion (**SELECT 1 FINDING**)

**336.** Black widow spider (**SELECT 1 FINDING**)

**337.** Tick (**SELECT 1 FINDING**)

### Items 338–340

For each description, select the most likely diagnosis.

- a. Transient tic disorder of childhood
- b. Tourette syndrome
- c. Sydenham chorea
- d. Tardive dyskinesia
- e. Cerebral palsy

**338.** Eye blinking or throat-clearing noises in an otherwise healthy 8-year-old boy (**SELECT 1 DIAGNOSIS**)

**339.** A 6-year-old boy with eye twitching and ecolalia (**SELECT 1 DIAGNOSIS**)

**340.** A 2-year-old infant who was born prematurely and is unable to walk or talk (**SELECT 1 DIAGNOSIS**)

**Items 341–343**

For each diagnosis listed, select the most common clinical sign or symptom.

- a. A 6-month-old with blindness on the same side as a large facial lesion
- b. An infant with infantile spasms, a hypsarrhythmic EEG pattern, and ash-leaf depigmentation on her back
- c. An 18-year-old with a history of fractures and optic gliomas, who now has developed a malignant schwannoma
- d. A 2-year-old with multiple episodes of skin infection and failure to thrive
- e. A 14-year-old girl with a history of precocious puberty who now develops a large goiter

**341.** Neurofibromatosis type 1 (**SELECT 1 FINDING**)

**342.** McCune-Albright syndrome (**SELECT 1 FINDING**)

**343.** Tuberous sclerosis (**SELECT 1 FINDING**)

# The Neuromuscular System

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## Answers

**307. The answer is d.** (*Behrman, 16/e, pp 94–100. McMillan, 3/e, pp 776–782. Rudolph, 21/e, pp 17–19.*) Although early intervention is currently advocated for all of these conditions as soon as they are identified, studies of outcome are unable to validate that development is significantly improved in any but developmental language disorder. In support of such intervention is the fact that parental involvement is strengthened and parental needs supported. Developmental language disorder, if uncomplicated by other developmental impairments such as autism or mental retardation, is highly responsive to speech therapy, an intervention that can have long-term positive results.

**308. The answer is d.** (*Behrman, 16/e, p 1961. McMillan, 3/e, p 611. Rudolph, 21/e, pp 2242–2244.*) The history, signs, and symptoms as outlined in the question are characteristic of a basilar skull fracture. Those patients with rupture of the tympanic membrane allowing otorrhea and those with rhinorrhea after the injury are at increased risk of complications of infection. For these children, a semi-upright position and observation for 72 h for evidence of increased intracranial pressure or infection without use of prophylactic antibiotics is appropriate. Cerebrospinal fluid (CSF) drainage frequently stops within 72 h. Drainage beyond 72 h can require surgical closure; the risk of complications such as infection increases after this time.

**309. The answer is e.** (*Behrman, 16/e, pp 1803–1806. McMillan, 3/e, pp 223–224. Rudolph, 21/e, p 2185.*) Mongolian spot is not related to any abnormality. Virtually any abnormality (except Mongolian spots) over the lower spine points to the possibility of occult spinal dysraphism. This designation includes a number of spinal cord and vertebral anomalies that frequently produce severe loss of neurologic function, particularly in the region of the back, the lower extremities, and the urinary system. Examples of these abnormalities are subcutaneous lipomeningomyelocele, diastematomyelia,

hamartoma, lipoma, tight filum terminale, tethered cord, dermal and epidermal cysts, dermal sinuses, neurenteric canals, and angiomas. Occasionally, the loss of neurologic function from such anomalies is mild and, as a result, easily overlooked. Prompt evaluation of these lesions via CT, MRI, or ultrasound is indicated.

**310. The answer is b.** (*Behrman, 16/e, pp 344–347, 350–357, 373. McMillan, 3/e, pp 1829–1830, 1832–1835, 1841–1842. Rudolph, 21/e, pp 609–611, 613–617, 2236–2237.*) Homocystinuria can cause thromboembolic phenomena in the pulmonary and systemic arteries and particularly in the cerebral vasculature; vascular occlusive disease is, in turn, one of the many causes of acute infantile hemiplegia. None of the other disorders listed in the question is associated with acute hemiplegia. Phenylketonuria causes retardation and, on occasion, seizures; maple syrup urine disease, an abnormality of the metabolism of leucine, leads to seizures and rapid deterioration of the central nervous system in newborn infants; histidinemia is associated with speech impairments and other minor neurologic difficulties; and cystathioninuria is most likely a benign amino-aciduria with no effect on the central nervous system.

**311. The answer is e.** (*Behrman, 16/e, pp 1800–1801. McMillan, 3/e, pp 2267–2268.*) In the infant, in contrast to the adult, it is often not possible to sense the sudden release of resistance that may occur as the needle penetrates the dura and enters the subarachnoid space. It is, therefore, very common for the physician not to realize that the tip of the needle is in the proper spot for fluid withdrawal and to mistakenly continue to advance the needle, which may penetrate a blood vessel, producing a traumatic tap and confusing the diagnosis. The stylet should be removed repeatedly as the needle is inserted to increase the chance that the fluid will be clear. Although not using a stylet at all can increase the chance of avoiding a traumatic tap, it produces the risk of inserting a core of epidermis into the subarachnoid space and producing an epidermoid tumor. The other wrong answers decrease the chances of success.

**312. The answer is c.** (*Behrman, 16/e, pp 1885–1887. McMillan, 3/e, pp 1967–1969. Rudolph, 21/e, pp 2285–2287.*) Myasthenia gravis is an autoimmune disorder in which circulating acetylcholine receptor-binding antibodies are found in approximately 90% of cases, less commonly, when the

patient demonstrates isolated eye disease. Other nonspecific evidence of autoimmune disease includes elevated ANAs, presence of abnormal immune complexes, and an association with the hypothyroidism related to lymphocytic thyroiditis, also an autoimmune disorder. The earliest signs of myasthenia gravis are ptosis and weakness of the extraocular muscles, followed by dysphagia and facial muscle weakness. The distinguishing hallmark of this disease is rapid fatiguing of the involved muscles. The conduction velocity of the motor nerve is normal in this condition. When the involved muscle is repetitively stimulated for diagnostic purposes, the EMG shows a decremental response, which can be reversed by the administration of cholinesterase inhibitors. In older children this test is accomplished with edrophonium chloride, but it should be avoided in young infants, as cardiac arrhythmias may result (neostigmine would be used instead). Cholinesterase inhibitors are the primary therapeutic agents, and intramuscular neostigmine methylsulfate, oral neostigmine bromide, and oral pyridostigmine are available. Other therapeutic modalities for myasthenia gravis include immunosuppression, plasmapheresis, thymectomy (an enlarged thymus is frequently seen on chest x-ray), and treatment of hypothyroidism. CPK should be normal.

**313. The answer is a.** (*McMillan, 3/e, p 1953. Rudolph, 21/e, pp 2386–2387.*)

Pinpoint pupils are found in coma, narcotics use, pilocarpine treatment, Horner syndrome, pontine hemorrhage, posterior synechia formation, pesticide or nerve gas exposure, and tertiary syphilis. Dilated pupils are seen with drugs such as epinephrine, phenylephrine, topical antihistamines/vasoconstrictor combinations and atropine-like agents. In addition, trauma, third nerve paralysis, or closed-angle glaucoma can also cause pinpoint pupils.

**314. The answer is e.** (*Behrman, 16/e, pp 1854–1855. McMillan, 3/e, pp 614–615, 1929, 1931–1933. Rudolph, 21/e, pp 2246–2248, 2257–2258, 2261, 2274–2275.*) The abrupt onset of a hemisindrome, especially with the eyes looking away from the paralyzed side, strongly indicates a diagnosis of acute infantile hemiplegia. Most frequently, this represents a thromboembolic occlusion of the middle cerebral artery or one of its major branches. Hemiplegic migraine commonly occurs in children with a history of migraine headaches. Todd's paralysis follows after a focal or Jacksonian seizure and generally does not last more than 24 to 48 h. The clinical onset of supratentorial brain tumor is subacute, with repeated headaches and

gradually developing weakness. A history of trauma usually precedes the signs of an acute subdural hematoma. Clinical signs of other diseases can appear fairly rapidly, but not often with the abruptness of occlusive vascular disease.

**315. The answer is c.** (*Behrman, 16/e, pp 400, 1850. McMillan, 3/e, pp 1875–1878. Rudolph, 21/e, pp 2324–2325.*) Children who have Tay-Sachs disease are characterized by progressive developmental deterioration; physical signs include macular cherry-red spots and exquisite sensitivity to noise. Diagnosis of this disorder can be confirmed biochemically by the absence of hexosaminidase A activity in white blood cells. Tay-Sachs disease is inherited as an autosomal recessive trait; frequently, affected children are of Eastern European Jewish ancestry. The other disorders listed in the question are associated with enzyme deficiencies as follows: Niemann-Pick disease, sphingomyelinase; infantile Gaucher disease, beta-glucosidase; Krabbe disease (globoid cell leukodystrophy), galactocerebroside beta-galactosidase; and Fabry disease, alpha-galactosidase.

**316. The answer is d.** (*Behrman, 16/e, pp 1878–1879. McMillan, 3/e, pp 1973–1974. Rudolph, 21/e, pp 2293–2294, 2406.*) The child in the question appears to have myotonic muscular dystrophy. An elevated creatine kinase (especially in the preclinical phase) is often found and psychomotor retardation can be the presenting complaint (but may be identified only in retrospect). Ptosis, baldness, hypogonadism, facial immobility with distal muscle wasting (in older children), and neonatal respiratory distress (in the newborn period) are major features of this disorder. Cataracts are commonly seen, presenting either congenitally or at any point during childhood. The diagnosis is confirmed by identifying typical findings on muscle biopsy. Seizures are not a feature of myotonic dystrophy. Enlarged gonads are associated with fragile X syndrome, and hirsutism is found (among other things) in children with congenital adrenal hyperplasia.

**317. The answer is a.** (*Behrman, 16/e, pp 1832–1834. McMillan, 3/e, pp 679–680, 1931–1932, 2027–2029. Rudolph, 21/e, pp 2274–2276.*) In contrast to adults, children with migraine most often have “common” migraine: bifrontal headache without an aura or diffuse throbbing headache of only a few hours’ duration. As with adults, the headaches can be terminated with vomiting or sleep. Family history is frequently positive. Association with

attention deficit hyperactive disorder is not common, but a relationship with seizure disorder can be seen.

**318. The answer is b.** (*Behrman, 16/e, p 759. McMillan, 3/e, p 859. Rudolph, 21/e, pp 900–904, 953.*) Included among those things that can cause the clinical picture are viral meningitis, tuberculous meningitis, meningeal leukemia, and medulloblastoma, all of which can cause pleocytosis as well as elevated protein and lowered glucose concentrations in cerebrospinal fluid (CSF). Of the four diseases (and the likely finding of this patient), tuberculous meningitis is associated with the lowest glucose levels in CSF. The cellular response to viral meningitis eventually will be predominantly lymphocytic. Cells found in the CSF of a child who has meningeal leukemia most commonly are lymphocytes or lymphoblasts. Children who have a medulloblastoma generally present with the signs and symptoms caused by a mass in the posterior cranial fossa; their pleocytotic cerebrospinal fluid contains unusual-appearing cells of the monocytic variety. The decrease in the glucose concentration of CSF associated with these disorders has been attributed to a disturbance of glucose transport as a result of meningeal irritation. In pseudotumor cerebri, the constituents of CSF are generally normal except for a low protein content in some instances. Acute bacterial disease typically causes polymorphonuclear cells and positive Gram stains. Neither tuberous sclerosis nor stroke typically causes these findings on CFS examination.

**319. The answer is a.** (*Behrman, 16/e, pp 1858–1862. McMillan, 3/e, pp 1511–1515. Rudolph, 21/e, pp 2219–2221.*) A child who has a subacute disorder of the central nervous system that produces cranial nerve abnormalities (especially of cranial nerve VII and the lower bulbar nerves), long-tract signs, unsteady gait secondary to spasticity, and some behavioral changes is most likely to have a pontine glioma. Tumors of the cerebellar hemispheres can, in later stages, produce upper motor neuron signs, but the gait disturbance would be ataxia. Dysmetria and nystagmus also would be present. Supratentorial tumors are quite uncommon in 6-year-old children; headache and vomiting would be likely presenting symptoms and papilledema a finding on physical examination.

**320. The answer is d.** (*Behrman, 16/e, pp 890–891. McMillan, 3/e, p 1033. Rudolph, 21/e, p 953.*) The CT scan with contrast can be an excellent clue for

diagnosing tuberculous meningitis. Exudate in the basal cisterns that shows enhancement by contrast material is typical; tuberculomas, lucenecies, edema, and infarction can be apparent; and hydrocephalus can develop. Confirmation with culture is mandatory. The x-ray of the chest will be likely to show signs of pulmonary tuberculosis. A high index of suspicion is necessary to diagnose tuberculous meningitis early.

**321. The answer is b.** (*Behrman, 16/e, pp 272–273. McMillan, 3/e, pp 614–615. Rudolph, 21/e, pp 2246–2248.*) Compression of cranial nerve III and distortion of the brainstem, resulting in unilateral pupillary dilatation, hemiplegia, focal seizures, and depressed consciousness, suggest a progressively enlarging mass, most likely a subdural hematoma. Such a hematoma displaces the temporal lobe into the tentorial notch and presses on the ipsilateral cranial nerve III. Brainstem compression by this additional tissue mass leads to progressive deterioration in consciousness. Rising blood pressure and falling pulse rate are characteristic of increasing intracranial pressure. The most urgent test to diagnose this condition is a CT scan.

**322. The answer is c.** (*Behrman, 16/e, p 1861. Rudolph, 21/e, pp 2226–2227.*) The findings of poor growth and papilledema could be explained by a craniopharyngioma. This tumor is one of the most common supratentorial tumors in children, often causing growth failure through disruption of pituitary excretions such as growth hormone. Upward growth of a craniopharyngioma results in compression of the optic chiasm. Particularly affected are the fibers derived from the nasal portions of both retinas (in other words, from those parts of the eyes receiving stimulation from the temporal visual field). Early in the growth of a craniopharyngioma, a unilateral superior quadrantanopsic defect can develop, and an irregularly growing tumor can impinge upon the optic chiasm and cause homonymous hemianopia.

**323. The answer is a.** (*Behrman, 16/e, pp 1839–1840. Rudolph, 21/e, pp 2314–2315.*) Cerebellar ataxia in childhood can occur in association with infection, metabolic abnormalities, ingestion of toxins, hydrocephalus, cerebellar lesions, multiple sclerosis, labyrinthitis, polyradiculopathy, and neuroblastoma. Although the other diagnoses (except agenesis of the corpus collasum) can cause ataxia, it is more of a chronic nature than acute. Ingestion (intentional or accidental) of barbiturates, phenytoin, alcohol, and other

drugs also must be considered. Agenesis of the corpus callosum is usually diagnosed by imaging studies; however, it does not cause acute ataxia.

**324. The answer is a.** (*Behrman, 16/e, pp 1858–1862. McMillan, 3/e, pp 1511–1513. Rudolph, 21/e, pp 2207–2210.*) Between 50 and 60% of tumors of the nervous system in children 4 to 11 years old are infratentorial (posterior fossa) and include cerebellar and brainstem tumors, often either medulloblastoma or cerebellar astrocytoma. In adults and infants, most intracranial tumors originate above the tentorium; only 25 to 30% of brain tumors in adults are subtentorial.

**325. The answer is e.** (*Behrman, 16/e, pp 875–878. McMillan, 3/e, p 941. Rudolph, 21/e, pp 2287–2288.*) Spinal muscular atrophy in a neonate is associated with hypotonia and feeding difficulties; a muscle biopsy can confirm this diagnosis. Neonatal myasthenia gravis and botulism, though uncommon, must be considered in a newborn infant who has the symptoms described in the question. The symptoms presented also could represent myotonic dystrophy; this diagnosis is confirmed by examination of both parents for percussion and grip myotonia and by electromyographic depiction of myotonic discharges. Duchenne (pseudohypertrophic) muscular dystrophy clinically appears in children who are about 2 or 3 years of age. Botulism has clearly been associated with the ingestion of raw honey. In this child, although the other possibilities listed in the question may be the etiology, it seems most likely that botulism is the culprit.

**326. The answer is b.** (*Behrman, 16/e, p 1928. McMillan, 3/e, pp 1875–1878, 1993–1995, 2004–2005. Rudolph, 21/e, pp 668, 2323–2324.*) The cherry-red spot represents the center of a normal retinal macula that is surrounded by ganglion cells in which there is an abnormal accumulation of lipid. This alters the surrounding retinal color so that it is yellowish or grayish white and is seen more often in such disorders as GM<sub>1</sub> generalized gangliosidosis type 1, Sandhoff disease, and Niemann-Pick disease type A, in which there is lipid material deposited in the ganglion cells. Generalized gangliosidosis type 1 (Type 1 GM<sub>1</sub> gangliosidosis) presents as noted in the question, with symptoms often present at birth; these infants have a complete lack of acid beta-galactosidase activity. Other findings with generalized gangliosidosis type 1 (and not listed in the question) include gingival hyperplasia, hernias, joint stiffness, dorsal kyphosis, and edema of the extremities. Hexosaminidase A

deficiency (GM<sub>2</sub> gangliosidosis, type 1 or Tay-Sachs disease) presents as psychomotor retardation and hypotonia beginning at about 6 to 12 months of age; the children are usually normal at birth. A pronounced startle reflex and severe hyperacusis, seizures, loss of vision (with cherry-red macular spots), and macrocephaly are seen. Reduced activity of alpha-galactosidase (Fabry's disease) presents in older children as acroparesthesia (numbness or tingling in one or more extremities), intermittent painful crisis of the extremities or the abdomen, frequently low-grade fevers, and sometimes cataracts. Patients with Rett syndrome (the etiology of which has been traced to a defective gene called MeCP2 on the X chromosome) present as normal children at birth, but then have a rapid decline in motor and cognitive functions beginning between 6 and 18 months of age. Affected girls demonstrate loss in the use of their hands and loss in their ability to communicate and socialize. Metachromatic leukodystrophy (deficient activity of galactosyl-3-sulfate-ceramide sulfatase) has its onset between 1 and 2 years of age, notable for progressive ataxia, weakness, and peripheral neuropathy. In this disorder, gray macular lesions can be seen that look somewhat similar to cherry-red spots.

**327. The answer is b.** (*Behrman, 16/e, pp 1818–1819. McMillan, 3/e, pp 1949–1952. Rudolph, 21/e, pp 2270–2271.*) Febrile seizures usually occur in children between the ages of 9 months and 5 years and generally in association with upper respiratory illness, roseola, shigellosis, or gastroenteritis. The generalized seizures are mostly brief (2 to 5 min), and the cerebrospinal fluid is normal. Infants who have seizures that are prolonged (longer than 15 min), focal, or lateralized, or who had neurologic problems before the febrile seizure, are at a higher risk of developing an afebrile seizure disorder during the next 5 to 7 years.

**328. The answer is a.** (*Behrman, 16/e, pp 981–984, 1054–1062. McMillan, 3/e, pp 429–433, 444–447, 1033. Rudolph, 21/e, pp 1031–1035, 1146–1148.*) Periventricular calcifications are a characteristic finding in infants who have congenital cytomegalovirus (CMV) infection. The encephalitic process especially affects the subependymal tissue around the lateral ventricles and thus results in the periventricular deposition of calcium. Calcified tuberculomas, if visible radiographically, are not particularly periventricular; congenital tuberculosis presenting such as the patient described would be extraordinarily unusual. Granulomatous encephalitis caused by congenital toxoplasmosis is

associated with scattered and soft-appearing intracranial calcification, and suprasellar calcifications are typical of craniopharyngiomas. Congenital syphilis does not produce intracranial calcifications. The unscientific but sometimes useful way of keeping intracranial calcifications caused by CMV straight from those caused by toxoplasmosis is to remember that “CMV” has a “V” in it, as does “periventricular”; “toxoplasmosis” has an “x” in it, and the lesions associated with it are scattered throughout the “cortex,” which also has an “x” in it.

**329. The answer is b.** (*Behrman, 16/e, p 1837. McMillan, 3/e, pp 2023–2024. Rudolph, 21/e, pp 770–772.*) In infants, achromic skin patches, especially in association with infantile spasms, are characteristic of tuberous sclerosis. Other dermal abnormalities (adenoma sebaceum and subungual fibromata) associated with this disorder appear later in childhood. Although children who have neurofibromatosis may have a few achromic patches, the identifying dermal lesions are café au lait spots. Incontinentia pigmenti also is associated with seizures; the skin lesions typical of this disorder begin as bullous eruptions that later become hyperpigmented lesions. Pityriasis rosea and psoriasis are not associated with infantile spasms.

**330. The answer is e.** (*Behrman, 16/e, pp 195–196, 1681–1683. McMillan, 3/e, pp 1783, 1792, 1955. Rudolph, 21/e, pp 2244–2245.*) Diabetes insipidus is a common complication of severe head trauma and is due to a deficiency in secretion of antidiuretic hormone. It must be distinguished from the polyuria that can occur several days after head injury, as the high antidiuretic hormone levels associated with the head injury resolve. This form of diabetes insipidus can be treated by replacement of urinary losses intravenously or enterally with solutions low in sodium as long as it is possible to keep up with the losses. Vasopressin given intramuscularly or as an intranasal preparation makes care easier and safer as long as water balance is carefully monitored.

**331. The answer is b.** (*Behrman, 16/e, pp 1873–1877. McMillan, 3/e, pp 1972–1976. Rudolph, 21/e, pp 2289–2293.*) The most common form of muscular dystrophy is Duchenne muscular dystrophy. It is inherited as an X-linked recessive trait. Male infants are rarely diagnosed at birth or early infancy since they often reach gross milestones at the expected age. Soon after beginning to walk, however, the features of this disease become more

evident. While these children walk at the appropriate age, the hip girdle weakness is seen by age 2. Increased lordosis when standing is evidence of gluteal weakness. Gower sign (use of the hands to “climb up” the legs in order to assume the upright position) is seen by 3 to 5 years of age, as is the hip waddle gait. Ambulation ability remains through about 7 to 12 years, after which use of a wheelchair is common. Associated features include mental impairment and cardiomyopathy. Death due to respiratory failure, heart failure, pneumonia, or aspiration is common by 18 years of age.

**332. The answer is c.** (*Behrman, 16/e, pp 1892–1893. McMillan, 3/e, pp 1959–1965, 1972–1976. Rudolph, 21/e, pp 2281–2283.*) The paralysis of Guillain-Barré occurs about 10 days after a nonspecific viral illness. Weakness is gradual over days or weeks, beginning in the lower extremities and progressing toward the trunk. Later, the upper limbs and the bulbar muscles can become involved. Involvement of the respiratory muscles is life-threatening. The syndrome seems to be caused by a demyelination in the motor and, occasionally, the sensory nerves. Measurement of spinal fluid protein is helpful in the diagnosis; protein levels are increased to more than twice normal, while glucose and cell counts are normal. Hospitalization for observation is indicated. Treatment can consist of intravenous immunoglobulin, steroids, or plasmapheresis. Recovery is not always complete. Bell palsy usually follows a mild upper respiratory infection, resulting in the rapid development of weakness of the entire side of the face. Muscular dystrophy encompasses a number of entities that include weakness over months. Charcot-Marie-Tooth disease has a clinical onset including peroneal and intrinsic foot muscle atrophy, later extending to the intrinsic hand muscles and proximal legs. Werdnig-Hoffmann disease is an anterior horn disorder that presents either in utero (in about one-third of cases) or by the first 6 months of life with hypotonia, weakness, and delayed developmental motor milestones.

**333–334. The answers are 333-b, 334-d.** (*Behrman, 16/e, pp 16, 1829. McMillan, 3/e, p 826. Rudolph, 21/e, pp 34, 418, 2273.*) Night terrors are most common in boys between the ages of 5 and 7 years. The child awakens suddenly, appears frightened and unaware of his surroundings, and has the clinical signs outlined in the question. He cannot be consoled by the parents. After a few minutes, sleep returns, and the patient cannot recall the event in the morning. Sleepwalking is common in these children. Explor-

ing the family dynamics for emotional disorders may be helpful; usually pharmacologic therapy is not required, and family reassurance is indicated.

**335–337. The answers are 335-d, 336-b, 337-a.** (*Behrman, 16/e, pp 1887, 2175–2177. McMillan, 3/e, pp 1209–1210, 1970–1971. Rudolph, 21/e, pp 393–395, 1152–1153, 1235, 2288.*) Tick paralysis can be caused by such ticks as the eastern dog or the Rocky Mountain wood tick, which inject toxin in their saliva while they remain attached. It can be difficult to diagnose because the only sign of illness can be the paralysis. The paralysis begins as ataxia and areflexia about 1 to 2 days after tick attachment, later progressing to progressive ascending paralysis. Ticks should be aggressively sought, since removal usually brings cure and averts possible death. The scorpion sting is usually easier to diagnose, since scorpions are easier to see and the sting induces a severe reaction with pain and swelling and can also induce a severe systemic reaction with shock, excessive salivation, and convulsions. In addition to supportive therapy, including the use of tourniquets and phenobarbital (but not morphine), specific antivenins are available.

Black widow spiders are also capable of making their presence known not only by their distinctive markings, but also by their ability to attract one's attention to the site of their bite by a marked, painful local reaction followed by a systemic reaction, with nausea, vomiting, sweating, hypertension, and cramping abdominal pain. Pain can be treated with meperidine (Demerol) or intravenous calcium gluconate, and a specific antivenin should be used.

The patient in the question with chronic skin ulceration hints at brown recluse spider bites, while the 18-year-old may have contracted cutaneous larval migrans, perhaps from walking barefoot on a beach where dogs also frequent.

**338–340. The answers are 338-a, 339-b, 340-e.** (*Behrman, 16/e, pp 1842–1845. McMillan, 3/e, pp 1910–1917, 2014–2015. Rudolph, 21/e, pp 462, 2197–2202.*) Tics are commonly seen in a pediatric practice. All have in common the nonrhythmic, spasmodic, involuntary, stereotypical behaviors that involve any muscle group. Transient tic disorder is the most common and is seen more often in boys; a family history is often noted. In this condition, the patient has eye blinking, facial movements, or throat clearing lasting for weeks to about a year. No medications are needed. Chronic

motor tics persist throughout life and can incorporate motor movements involving up to three muscle groups.

Gilles de la Tourette syndrome is a lifelong condition that is characterized by motor and vocal tics, obsessive-compulsive behavior, and a high incidence of attention deficit disorder with hyperactivity (ADDH). Therapy for the ADDH is helpful, as is medication to control the motor or vocal tics. Multiple psychosocial problems exist with these children; a multidisciplinary approach is helpful.

Cerebral palsy is a static condition of movement and posture disorders frequently associated with epilepsy and abnormalities of vision, speech, and intellect. A defect in the developing brain is felt to be the cause. No significant change in the incidence of cerebral palsy has been noted in the past few decades despite drastically improved obstetric and neonatal care. No treatment of cerebral palsy is available; a multidisciplinary approach to manage the many medical problems associated with the condition is helpful.

**341–343. The answers are 341-c, 342-e, 343-b.** (*Behrman, 16/e, pp 1692–1693, 1835–1837. McMillan, 3/e, p 2026. Rudolph, 21/e, pp 769–772, 2344–2347.*) Neurofibromatosis type 1 (NF1) is a progressive neurocutaneous syndrome that results from a defect in neural crest differentiation and migration during the early stages of development. It has an autosomal dominant pattern of inheritance; the gene locus has been identified on chromosome 17. Any organ or system can be affected, neurologic complications are frequent, and patients are at high risk of developing malignant neoplasms of various types. The presence of any two of the following findings confirms the diagnosis of NF1.

1. Five or more café au lait spots over 5 mm in diameter in prepubertal patients; six or more over 15 mm in diameter in postpubertal patients
2. Axillary freckling
3. Two or more Lisch nodules (hamartomas of the iris)
4. Two or more neurofibromas, typically involving the skin and appearing during adolescence or pregnancy; or one plexiform neuroma involving a nerve track present at birth
5. Bony lesions leading to pathologic fracture and kyphoscoliosis
6. Optic glioma
7. NF1 in a first-degree relative

McCune-Albright syndrome and NF1 share many of the same clinical findings: skin, bones, and endocrine glands can be involved in both. The characteristic features of McCune-Albright syndrome include large, irregular, usually unilateral café au lait spots and fibrous dysplasia of bones in association with precocious puberty in girls. Although the last finding has been the major recognized endocrinopathy in the syndrome, disorders of pituitary, thyroid, and adrenal glands have been noted with greater frequency in recent years. The endocrinopathies are ascribed to autonomous hyperfunctioning of the involved gland with goiter.

Tuberous sclerosis, an autosomal dominant condition, can result in severe mental retardation and seizures. Infantile spasms, a hypsarrhythmic EEG pattern, hypopigmented lesions (ash-leaf spots), cardiac tumors, sebaceous adenomas, a shagreen patch (a roughened, raised lesion over the sacrum), and calcifications on the CT scan are all features of this condition. No specific treatment is available.

# Infectious Diseases and Immunology

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## Questions

**DIRECTIONS:** Each item below contains a question or incomplete statement followed by suggested responses. Select the **one best** response to each question.

**344.** Your 18-year-old college freshman calls to state that he has had fever, muscular pain (especially in the neck), headache, and malaise. He describes the area from the back of his mandible toward the mastoid space as being full and tender and that his earlobe on the affected side appears to be sticking upward and outward. Drinking sour liquids causes much pain in the affected area. You quickly retrieve his immunization card and suddenly realize that he has failed to get vaccinated for

- a. Mumps
- b. Varicella
- c. Rubella
- d. Measles
- e. Herpangina

**345.** A 2-month-old child of an HIV-positive mother is followed in your pediatric practice. Which of the following therapies should be considered for this child?

- a. Monthly evaluation for Kaposi's sarcoma
- b. Prophylaxis against *Pneumocystis carinii* pneumonia (PCP)
- c. Vitamin C supplementation
- d. Oral polio virus
- e. Bone marrow transplantation

**346.** Influenza virus vaccine is usually administered annually because of antigen shift and limited duration of protection. Which of the following groups should receive the vaccine?

- a. Infant with Still's murmur
- b. Government employees
- c. Infants under 6 months of age
- d. Patients on long-term aspirin therapy
- e. Young patients with seizure disorder

**347.** An 18-month-old child presents to the emergency center having had a brief, generalized tonic clonic seizure. He is now postictal and has a temperature of 40°C (104°F). During the lumbar puncture (which proves to be normal), he has a large, watery stool that has both blood and mucus in it. The most likely diagnosis in this patient is

- a. *Salmonella*
- b. Enterovirus
- c. Rotavirus
- d. *Campylobacter*
- e. *Shigella*

**348.** A 4-year-old child with fever, peeling skin, abdominal pain, and a red throat has been admitted to the hospital because of overlap between presenting signs and symptoms of scarlet fever and Kawasaki disease. Which of the following statements comparing these two diseases is true?

- a. Neither has cardiac complications
- b. Serologic tests are helpful in diagnosing both
- c. Kawasaki disease has mucocutaneous and lymph node involvement, but scarlet fever does not
- d. Pharyngeal culture aids in the diagnosis of scarlet fever but not Kawasaki disease
- e. A specific therapy is recommended for scarlet fever, but supportive care only is recommended for Kawasaki disease

**349.** A patient with hair loss is shown below. The lesion does not fluoresce with a Wood's lamp and has not responded well to a variety of topical agents. The lesion is boggy, is spreading, and has tiny pinpoint black dots throughout. The most likely diagnosis is



- a. Traction alopecia
- b. Infection with *Trichophyton tonsurans*
- c. Alopecia areata
- d. Biotinidase deficiency
- e. Hypothyroidism

**350.** An 8-year-old sickle cell patient arrives at the emergency room in respiratory distress. Over the previous several days, the child has become progressively tired and pale. The child's hemoglobin concentration in the emergency room is 3.1 mg/dL. Which of the following viruses commonly causes such a clinical picture?

- a. Roseola
- b. Parvovirus B-19
- c. Coxsackie A-16
- d. Echovirus 11
- e. Cytomegalovirus

**Items 351–352**

**351.** A 17-year-old male complains of acute, painful, scrotal swelling. Ultrasound of the testes is normal, but the urinalysis reveals pyuria. The most likely diagnosis in this patient is

- a. Incarcerated inguinal hernia
- b. Torsion of the testes
- c. Varicocele
- d. Epididymitis
- e. Urinary tract infection

**352.** Appropriate therapy for this patient is

- a. Surgical consultation
- b. Culture, intramuscular ceftriaxone, and a course of oral doxycycline
- c. Voiding cystourethrogram
- d. Oral sulfonamides for 3 days
- e. Warm compresses and counseling

**353.** A 15-month-old boy is brought to the emergency room because of fever and a rash. Six hours earlier he was fine, except for tugging on his ears; another physician diagnosed otitis media and prescribed amoxicillin. During the interim period, the child has developed an erythematous rash on his face, trunk, and extremities. Some of the lesions, which are of variable size, do not blanch on pressure. The child is now very irritable, and he does not interact well with the examiner. Temperature is 39.5°C (103.1°F). He continues to have injected, immobile tympanic membranes, but you are concerned about his change in mental status. The most appropriate next step in the management of this infant is to

- a. Begin administration of intravenous ampicillin
- b. Begin diphenhydramine
- c. Discontinue administration of ampicillin and begin trimethoprim with sulfamethoxazole
- d. Perform bilateral myringotomies
- e. Perform a lumbar puncture

**354.** The 3-year-old sister of a newborn baby develops a cough diagnosed as pertussis by nasopharyngeal culture. The mother gives a history of having been immunized as a child. A correct statement regarding this clinical situation is

- a. The mother has no risk of acquiring the disease because she was immunized
- b. Hyperimmune globulin is effective in protecting the infant
- c. The risk to the infant depends on the immune status of the mother
- d. Erythromycin should be administered to the infant
- e. The 3-year-old sister should be immediately immunized with an additional dose of pertussis vaccine

**355.** A 14-year-old boy is seen in the emergency room because of a 3-week history of fever between 38.3°C and 38.9°C (101°F and 102°F), lethargy, and a 6-lb weight loss. Physical examination reveals marked cervical and inguinal adenopathy, enlarged tonsils with exudate, small hemorrhages on the soft palate, a WBC differential that has 50% lymphocytes (10% atypical), and a palpable spleen 2 cm below the left costal margin. Which of the following conditions is the likely diagnosis?

- a. HIV disease
- b. Varicella
- c. Kawasaki disease
- d. Streptococcal throat infection
- e. Infectious mononucleosis

**356.** A 2-year-old child is seen in the emergency center with the complaint of fever and a limp. As you begin your evaluation of this child, you remember that acute osteomyelitis

- a. Is most commonly caused by *Streptococcus pyogenes*
- b. Often arises following development of deep cellulitis
- c. Results in tenderness in the region of infection that is diffuse, not localized
- d. Causes bony changes visible radiographically within 48 h of the beginning of symptoms
- e. Requires antibiotic therapy usually for 10 to 14 days

**357.** A 16-day-old infant presents with fever, irritability, poor feeding, and a bulging fontanelle. Spinal fluid demonstrates gram-positive cocci. The most likely diagnosis is

- a. *Listeria monocytogenes*
- b. group A streptococcus
- c. group B streptococcus
- d. *Streptococcus pneumoniae*
- e. *Staphylococcus aureus*

**358.** A 4-day-old infant is brought to your office and is noted to be sucking on a honey-filled pacifier. You caution the mother against its use because its contents can cause a syndrome notable for

- a. Diffuse hypertonia and increased deep tendon reflexes
- b. Residual neurologic damage
- c. Oculomotor palsies
- d. Hyperresponsive gag reflex
- e. Diarrhea, hyperphagia, tachycardia, and irritability

**359.** A 14-month-old infant suddenly develops a fever of 40.2°C (104.4°F). Physical examination shows an alert, active infant who drinks milk eagerly. No physical abnormalities are noted. The white blood cell count is 22,000/ $\mu$ L with 78% polymorphonuclear leukocytes, 18% of which are band forms. The most likely diagnosis is

- a. Pneumococcal bacteremia
- b. Roseola
- c. Streptococcosis
- d. Typhoid fever
- e. Diphtheria

**360.** A 21-year-old woman is seronegative for rubella. You caution her against exposure to rubella early in pregnancy because it can result in an infant with

- a. Thrombocytosis
- b. Wide-set eyes
- c. Glaucoma
- d. Tetralogy of Fallot
- e. Duodenal atresia

**361.** The 3-year-old boy seen in the following photograph is brought to the emergency room with a swollen eye. He was well until 3 days ago, when he developed symptoms of an upper respiratory infection, which gradually worsened. On the night before admission, he was noted to have a temperature of  $39.6^{\circ}\text{C}$  ( $103.3^{\circ}\text{F}$ ). The child presents with proptosis, limitation of eye movement, chemosis, and decreased visual acuity. Your diagnosis for this patient is



*(Courtesy Binita Shah, M.D.)*

- a. Retinoblastoma
- b. Periorbital cellulitis
- c. Glaucoma
- d. Orbital cellulitis
- e. Myasthenia gravis

**362.** The pictured child presents with a 3-day history of malaise, fever to 41.1°C (106°F), cough, coryza, and conjunctivitis. He then develops the erythematous, maculopapular rash pictured. He is noted to have white pin-point lesions on a bright red buccal mucosa in the area opposite his lower molars. The most likely diagnosis is



*(Courtesy Adelaide Hebert, M.D.)*

- a. Parvovirus
- b. Rubella
- c. Herpes
- d. Rubeola
- e. Varicella

**363.** A 3-year-old child awakens at night with a fever of 39.6°C (103.3°F), a severe sore throat, and a barking cough. Physical examination of the child, who is drooling, shows a very red throat and inspiratory stridor. Optimal management would include

- a. Immediate hospitalization for possible intubation
- b. Immediate inhalation therapy with racemic epinephrine
- c. Treatment with oral ampicillin, 50 mg (kg·d)
- d. Suctioning of the pharynx and hourly examinations of the hypopharynx
- e. A throat culture, expectorant, and mist therapy

**364.** A 14-year-old girl awakens with a mild sore throat, low-grade fever, and a diffuse maculopapular rash. During the next 24 h, she develops tender swelling of her wrists and redness of her eyes. In addition, her physician notes mild tenderness and marked swelling of her posterior cervical and occipital lymph nodes. Four days after the onset of her illness, the rash has vanished. The most likely diagnosis of this girl's condition is

- a. Rubella
- b. Rubeola
- c. Roseola
- d. Erythema infectiosum
- e. Erythema multiforme

**365.** A 4-year-old child presents in the clinic with an illness notable for swelling in front of and in back of the ear on the affected side and altered taste sensation. Correct statements about this condition include which of the following?

- a. Arthritis is a common presenting complaint in children
- b. The disease could have been prevented by prior immunization with killed whole cell vaccine
- c. Involvement of the CNS may occur 10 days after the resolution of the parotitis
- d. Orchitis can occur and is almost exclusively in prepubertal males
- e. Subendocardial fibroelastosis is a common complication in a child of this age

**366.** A 2-year-old participant in a day-care center is diagnosed with hepatitis A. As the physician consultant to the day-care center, you develop a fact sheet for other families, explaining to them that

- a. Hepatitis A has a long incubation period (50 to 80 days)
- b. Hepatitis A presents after a prolonged viremic stage
- c. The presentation of hepatitis A is characterized by the sudden onset of fever, nausea, and vomiting
- d. Hepatitis A is spread by parenteral route
- e. An infection with hepatitis A results in elevations of liver function studies and serum bilirubin levels for 6 months or more

**367.** A 6-year-old comes to the clinic with the rapid onset of fever, muscle pain, and rash. The rash that developed is maculopapular, beginning on the flexor surfaces of the wrist and evolving to a hemorrhagic-type appearance. The history is positive for his going camping in the deep woods with the Boy Scouts 9 days prior to presentation. Which of the following is likely?

- a. Lyme disease
- b. Tularemia
- c. Measles
- d. Toxic shock syndrome
- e. Rocky Mountain spotted fever

**368.** A routine placement of a Mantoux (PPD) test on a 1-year-old child living in a homeless shelter results in a 12-mm indurated area 48 h later, indicating which of the following?

- a. The child was exposed to tuberculosis 2 to 5 days prior to the placement of the PPD
- b. The child is contagious
- c. The size of the induration is not large enough to require antimicrobial therapy
- d. The test is likely to revert to negative after 6 months
- e. The child may have an infection with atypical mycobacteria

**369.** The rash and mucous membrane lesions shown in the photograph develop in an infant 5 days into the course of an upper respiratory infection with otitis media, being treated with amoxicillin. The child's condition is likely



- a. Urticaria
- b. Rubella
- c. Stevens-Johnson syndrome
- d. Kawasaki disease
- e. Scarlet fever

**370.** An 8-year-old immigrant from rural Central America presents with complaints of weakness and fever. A CBC reveals eosinophilia. Which of the following parasites is most likely to be responsible?

- a. *Cryptosporidium parvum*
- b. *Sporothrix schenckii*
- c. *Giardia lamblia*
- d. *Enterobius vermicularis*
- e. *Trichinella spiralis*

**371.** A 10-year-old boy from the Connecticut coast is seen because of discomfort in his right knee. He had a large, annular, erythematous lesion on his back that disappeared 4 weeks prior to the present visit. His mother recalls pulling a small tick off his back. The correct statement about this child's likely illness includes which of the following?

- a. The tick was probably a *Dermacentor andersoni*
- b. The disease is caused by a rickettsial agent that is transmitted by the bite of a tick
- c. In addition to skin and joint involvement, CNS and cardiac abnormalities may be present
- d. Therapy with antibiotics has little effect on the resolution of symptoms
- e. The pathognomonic skin lesion is required for diagnosis

**372.** The patient pictured presents with a 6-h history of fever to 38.9°C (102°F). Her mother reports that the patient appeared to be feeling poorly, that she had been eating less than normal, and that she vomited once. About 2 h prior to arrival at the emergency room, the mother states that she noted a few purple spots scattered about the body on the patient, especially on the buttocks and legs. On the 30-min ride to the emergency room, the rash spread rapidly as shown, and the patient is now obtunded. Of the following, the most likely diagnosis is



(Courtesy Binita Shah, M.D.)

- a. Stevens-Johnson syndrome
- b. Toxic shock syndrome caused by *Staphylococcus aureus*
- c. Measles
- d. Rocky Mountain spotted fever
- e. Meningococcemia

**373.** In this 6-month-old girl, which of the following conditions would be a contraindication to a diagnostic lumbar puncture?

- a. Uncorrected bleeding diathesis
- b. Bulging fontanelle
- c. Lumbar puncture 2 days before
- d. Marked uncooperativeness on the part of the patient
- e. Significantly elevated WBC count consistent with bacteremia

**374.** Your office is called by the mother of one of your regular patients. She reports that her daughter has a 3-day history of subjective fever, hoarseness, and a bad barking cough. You arrange for her to be seen in your office that morning. Upon seeing this child, you would expect to find

- a. A temperature greater than 38.9°C (102°F)
- b. Expiratory stridor
- c. Infection with parainfluenza virus
- d. Hyperinflation on chest x-ray
- e. A child aged between 6 and 8 years

**375.** Two weeks ago, a 5-year-old boy developed diarrhea, which has persisted to the present time, despite dietary management. His stools have been watery, pale, and frothy. He has been afebrile. Microscopic examination of his stools is likely to show which of the following?

- a. *Salmonella sonnei*
- b. *Enterobius vermicularis*
- c. *Sporothrix schenckii*
- d. *Toxoplasma gondii*
- e. *Cryptosporidium*

**DIRECTIONS:** Each group of questions below consists of lettered options followed by a set of numbered items. For each numbered item, select the appropriate lettered option(s). Each lettered option may be used once, more than once, or not at all. **Choose exactly the number of options indicated following each item.**

**Items 376–379**

For each set of immunologic abnormalities listed in the table below, **SELECT 1 DIAGNOSIS.**

- a. A 5-year-old boy who, after 3 months of age, developed recurrent otitis media, pneumonia, diarrhea, and sinusitis, often with simultaneous infections at two or more disparate sites
- b. A distinctive-appearing 8-month-old boy with an interrupted aortic arch, hypocalcemia, and cleft palate
- c. A 1-year-old boy with severe eczema, recurrent middle-ear infections, lymphopenia, and thrombocytopenia
- d. A 9-year-old with an eczema-like rash and recurrent severe staphylococcal infections.
- e. A 4-month-old who has had failure to thrive, chronic diarrhea, a variety of rashes, and recurrent serious bacterial, fungal, and viral infections

	<b>Serum IgG</b>	<b>Serum IgA</b>	<b>Serum IgM</b>	<b>T-Cell Function</b>	<b>Parathyroid Function</b>
<b>376.</b>	Normal	Normal	Normal	Decreased	Decreased
<b>377.</b>	Low	Low	Low	Normal	Normal
<b>378.</b>	Low	Low	Low	Decreased	Normal
<b>379.</b>	Normal	High	Low	Decreased	Normal

**Items 380–385**

Match the disease with the associated organism.

- a. *Rochalimaea henselae*
- b. *Pseudomonas*
- c. Rubivirus
- d. Human herpesvirus 6
- e. *Escherichia coli*
- f. *Helicobacter pylori*
- g. Group B streptococci
- h. *Listeria monocytogenes*
- i. Epstein-Barr virus
- j. *Toxocara cati*
- k. *Campylobacter jejuni*

**380.** A 4-year-old with frequent pneumonias, chronic diarrhea, and poor growth (**SELECT 1 ORGANISM**)

**381.** Foot puncture wound through a tennis shoe of an adolescent exploring a construction site (**SELECT 1 ORGANISM**)

**382.** Warm, red, tender axillary lymph nodes in a 5-year-old girl; she has a few red papules on her hand at the site of a feline scratch (**SELECT 1 ORGANISM**)

**383.** The appearance of an evanescent, erythematous, maculopapular rash following the rapid defervescence of several days of high fever in a 9-month-old boy (**SELECT 1 ORGANISM**)

**384.** A weepy, red lesion on the arm of an 18-month-old who came into contact with a hot curling iron (**SELECT 1 ORGANISM**)

**385.** Several-week history of epigastric and periumbilical pain that worsens with fasting and is relieved by eating, in a 9-year-old with guaiac-positive stools (**SELECT 1 ORGANISM**)

**Items 386–388**

Match the diagnostic test with the associated disease state.

- a. A 15-year-old sexually active girl with a small, painful, red blister on her labia
- b. An otherwise healthy 9-year-old boy with pinpoint black dots in the center of an itchy 3-cm boggy patch of hair loss on his scalp
- c. A 2-month-old with fever and foul-smelling urine
- d. A toddler with severe perianal itching, especially at night
- e. Recurring fever and anemia in an immigrant from central Africa

**386.** Wood's lamp (**SELECT 1 DISEASE**)

**387.** Tzanck smear (**SELECT 1 DISEASE**)

**388.** Scotch tape preparation (**SELECT 1 DISEASE**)

**Items 389–392**

Match each clinical state with the most likely form of pneumonia.

- a. Mycoplasmal pneumonia
- b. Pneumococcal pneumonia
- c. Chlamydial pneumonia
- d. Viral pneumonia
- e. Tuberculous pneumonia
- f. Fungal pneumonia
- g. Staphylococcal pneumonia

**389.** A 6-week-old infant with tachypnea and history of eye discharge at 2 weeks of age (**SELECT 1 DIAGNOSIS**)

**390.** A 14-year-old girl with low-grade fever, cough of 3 weeks' duration, and interstitial pneumonia (**SELECT 1 DIAGNOSIS**)

**391.** A 2-month-old boy with a 3-day history of upper respiratory infection who suddenly develops high fever, cough, and respiratory distress; within 48 h, the patient has developed a pneumatocele and a left-sided pneumothorax (**SELECT 1 DIAGNOSIS**)

**392.** An 8-year-old girl with fever, tachypnea, and interstitial infiltrate and hilar adenopathy on chest x-ray; her social history is positive for an uncle living in the home who has a cough and was recently released from prison (**SELECT 1 DIAGNOSIS**)

# Infectious Diseases and Immunology

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## Answers

**344. The answer is a.** (*Behrman, 16/e, pp 817–820, 946–955, 961–962, 973–977. McMillan 3/e, pp 704, 1127–1130, 1134–1142. Rudolph, 21/e, pp 1042–1045, 1053–1058, 1075–1079, 1223.*) In addition to the findings described, mumps typically swells to the opposite side in a day or so after symptoms appear on the first side. Other findings include redness and swelling at the opening of Stensen's duct, edema and swelling in the pharynx, and displacement of the uvula on the affected side. A rash would not be expected. Measles presents in a child with a several-day history of malaise, fever, cough, coryza, and conjunctivitis followed by the typical, widespread, erythematous, maculopapular rash. Koplik spots, white pinpoint lesions on a bright red buccal mucosa often in the area opposite his lower molars, appear transiently and are pathognomonic. Symptoms of rubella, usually a mild disease, include a diffuse maculopapular rash that lasts for 3 days, marked enlargement of the posterior cervical and occipital lymph nodes, low-grade fever, mild sore throat, and, occasionally, conjunctivitis, arthralgia, or arthritis. Signs and symptoms of varicella include a prodrome of fever, anorexia, headache, and mild abdominal pain, followed 24 to 48 h later by the typical clear, fluid-filled vesicles (dewdrop on a rose petal). The rash of varicella typically starts on the scalp, face, or trunk. The lesions are pruritic and appear in crops over the next several days, with old lesions crusting over as new lesions develop. Herpangina causes sudden (usually high) fever, headache, backache, and, frequently, vomiting. Oral lesions are vesicles or ulcers usually found on the anterior tonsillar pillars, but can occur nearly anywhere in the mouth. They are caused by an enterovirus for which vaccination is not available.

**345. The answer is b.** (*Behrman, 16/e, pp 1022–1032. McMillan 3/e, pp 867–893. Rudolph, 21/e, pp 1045–1053.*) The drug of choice for treatment of *Pneumocystis carinii* pneumonia (PCP) is trimethoprim-sulfamethoxazole. In certain situations, such as in an infant born to an HIV-positive mother, it is used as prophylaxis against PCP infection. While awaiting final deter-

mination of this infant's HIV status, which can take several months, prophylaxis with trimethoprim-sulfamethoxazole is usually instituted.

Severe anemia can be associated with AIDS and can require blood transfusion, especially if there is evidence of respiratory compromise. Donor blood should be cytomegalovirus (CMV) negative to avoid CMV infection, a serious and possibly fatal disease in the immunocompromised patient. Bone marrow transplants in AIDS patients have been unsuccessful thus far because of the persistence of virus in macrophages throughout the body.

HIV-positive and AIDS patients should receive primary and booster immunization with diphtheria-pertussis-tetanus (DPT) vaccine, measles-mumps-rubella (MMR) vaccine, hepatitis B vaccine, and *Haemophilus influenzae* type B conjugated vaccine (HibCV). Inactivated polio vaccine (IPV) is recommended in place of oral polio vaccine (OPV) because of the theoretical risk of paralytic polio. Although vaccination with MMR is precluded in most other immunodeficient patients, those with AIDS should be immunized against measles, mumps, and rubella. The morbidity and mortality associated with these diseases in AIDS patients significantly override the risk of vaccine complications. If these children are exposed to measles, they should receive a protective dose of measles immunoglobulin regardless of their immunization history. Pneumovax at age 2 and influenza vaccine are recommended annually.

Eventually, all patients infected with HIV will lose weight and fail to grow. The factors responsible include reduced caloric intake because of poor appetite, intestinal malabsorption, and increased resting energy expenditure associated with chronic infection. Maintaining adequate nutrition in AIDS patients is very difficult. Vitamin C supplementation, by itself, has no special benefits.

Kaposi sarcoma is rare in children.

**346. The answer is d.** (*Behrman, 16/e, p 989. McMillan 3/e, pp 1083, 1089–1091. Rudolph, 21/e, pp 1069–1073.*) Patients targeted to receive influenza vaccine are those with chronic cardiac, pulmonary, hematologic, and neurologic problems in which respiratory compromise is part of the syndrome, as well as children with symptomatic HIV infection. Sound immunization practice regarding influenza stresses the importance of immunizing not only the high-risk patient but also all close contacts and medical personnel. Health professionals are at increased risk of exposure to influenza, and, in the event of an epidemic, their absence from work

because of illness would seriously compromise patient care. Reye syndrome, which has occurred less frequently in recent years, has been associated with influenza (and varicella) in patients receiving aspirin, and influenza vaccination may prevent this. Influenza vaccine is not approved for use in infants under 6 months old. No information is available about reactivity, immunogenicity, and adverse reactions in this age group. New recommendations from the American Academy of Pediatrics (and others) are beginning to suggest immunization of children 6 months to 2 years of age, although this practice has not yet become widespread.

**347. The answer is e.** (*Behrman, 16/e, pp 848–850. McMillan 3/e, pp 1006–1009. Rudolph, 21/e, pp 985–987.*) Clinical manifestations of shigellosis range from watery stools for several days to severe infection, with high fever, abdominal pain, and generalized seizures. In general, about 50% of these children have emesis, greater than two-thirds have fever, 10 to 35% have seizures, and 40% have blood in their stool. Often, the seizure precedes diarrhea and is the complaint that brings the family to the physician. Fever usually lasts about 72 h, and the diarrhea resolves within 1 week. Presumptive diagnosis can be made on the clinical history; confirmation is through stool culture. Supportive care, including adequate fluid and electrolyte support, is the mainstay of therapy. Antibiotic treatment is problematic; resistance to trimethoprim-sulfamethoxazole is common, necessitating therapy with third-generation cephalosporins in many cases.

**348. The answer is d.** (*Behrman, 16/e, pp 725–727, 803–804. McMillan, 3/e, pp 924–932, 1012–1017. Rudolph, 21/e, pp 844–845, 993–999.*) Kawasaki disease is an acute febrile illness of unknown etiology and shares many of its clinical manifestations with scarlet fever. Scarletiform rash, desquamation, erythema of the mucous membranes that produces an injected pharynx and strawberry tongue, and cervical lymphadenopathy are prominent findings in both. The most serious complication of Kawasaki disease and scarlet fever is cardiac involvement. Erythrogenic toxin-producing group A  $\beta$ -hemolytic streptococcus is the agent responsible for scarlet fever. Isolation of the organism from the nasopharynx and a rise in antistreptolysin titers will confirm the diagnosis. Serologic tests for a variety of infectious agents, both viral and bacterial, have been negative in Kawasaki disease. Rheumatic heart disease is a serious sequela of streptococcal pharyngitis, which can be prevented by appropriate treatment with

penicillin. Coronary artery aneurysm and thrombosis are the most serious complications of Kawasaki disease. The current approach to treatment, which includes aspirin and IV gamma globulin administered within a week of the onset of fever, appears to lower the prevalence of coronary artery dilatation and aneurysm and shorten the acute phase of the illness.

**349. The answer is b.** (*Behrman, 16/e, pp 2037–2038. McMillan, 3/e, pp 1153–1155. Rudolph, 21/e, pp 1226–1230.*) *Trichophyton tonsurans* is a major cause of tinea capitis. It produces an infection within the hair follicle that is unresponsive to topical treatment alone and requires long-term therapy with griseofulvin for eradication. Fluorescence is absent on examination by Wood's lamp. Diagnosis is made by microscopic examination of KOH preparation of infected hairs and by culture on appropriate media. A severe form of tinea capitis known as *kerion* is shown in the photograph. Keep in mind that a diagnosis of tinea capitis and not seborrhea should be considered in any child between the ages of 6 months and puberty who presents with scaldiness and hair loss, even if mild. Seborrhea rarely occurs in that age group.

**350. The answer is b.** (*Behrman, 16/e, pp 964–966, 1497–1499. McMillan, 3/e, pp 1098–1100. Rudolph, 20/e, pp 1058–1059, 1531–1534.*) Fifth disease (erythema infectiosum), long recognized as a benign mild exanthem of school-aged children, is now known to be caused by human parvovirus B-19. In the compromised patient, the parvovirus can cause serious anemia by infecting red-cell precursors and causing them to lyse. Patients with hemolytic anemia, such as sickle cell anemia, thus develop a transient aplastic crisis. A poorly functioning bone marrow (for a week or more) in a patient with a reduced red-cell life span (about 30 days) can result in profound anemia. Other problems can result in patients infected with parvovirus B-19. In patients with immunodeficiency, the B-19 infection can be persistent and lead to life-threatening chronic anemia. Infection in a pregnant woman can result in severe anemia in the infected fetus, with secondary hydrops fetalis and death. Roseola is now thought to be caused most often by the human herpesvirus 6. Coxsackie A-16 virus causes hand-foot-and-mouth disease. Echo-11 virus frequently causes viral meningitis, and cytomegalovirus causes a congenital infection.

**351–352. The answers are 351-d, 352-b.** (*Behrman, 16/e, pp 919–920, 1652–1653. McMillan, 3/e, p 1558. Rudolph, 21/e, p 261–266, 929–931,*

967–970.) Acute scrotal swelling and pain in a sexually active adolescent should raise the suspicion of epididymitis. Causative organisms are usually gonococci or chlamydiae, but the offending organism is frequently not recovered. Supportive evidence includes pyuria without organisms. Treatment is bed rest, antibiotics, and counseling. The most important differential diagnosis is torsion of the testes.

**353. The answer is e.** (Behrman, 16/e, pp 751–757. McMillan, 3/e, pp 413–416, 855–883. Rudolph, 21/e, pp 970–972, 977–979.) Unsuspected bacteremia due to *Haemophilus influenzae* type B (now rare), *Neisseria meningitidis*, or *Streptococcus pneumoniae* should be considered before prescribing treatment for otitis media in a young, febrile, toxic-appearing infant. Blood culture should be performed before antibiotic therapy is initiated, and examination of the cerebrospinal fluid is indicated if meningitis is suspected. The classic signs of meningitis are found with increasing reliability in children over the age of 6 months. Nevertheless, a febrile, irritable, inconsolable infant with an altered state of alertness deserves a lumbar puncture even in the absence of meningeal signs. A petechial rash, characteristically associated with meningococcal infection, has been known to occur with other bacterial infections as well. Organisms may be identified on smear of these lesions.

A fever accompanied by inability to flex rather than rotate the neck immediately suggests meningitis. An indolent clinical course does not rule out bacterial meningitis. A lumbar puncture is of prime diagnostic importance in determining the presence of bacterial meningitis, which requires immediate antibiotic therapy. A delay in treatment can lead to complications such as cerebrovascular thrombosis, obstructive hydrocephalus, cerebritis with seizures or acute increased intracranial pressure, coma, or death. In the described patient, lumbar puncture is warranted because of the change in his clinical status.

**354. The answer is d.** (Behrman, 16/e, pp 838–842. McMillan, 3/e, pp 990–993. Rudolph, 21/e, pp 973–975.) Newborn infants exposed to pertussis are at considerable risk of being infected regardless of the immune status of the mother. In contrast to other childhood infectious diseases, pertussis is not prevented by transplacentally acquired antibody. Hyperimmune globulin is ineffective and not recommended. Natural immunity conferred by

infection is lifelong. Because immunity acquired by immunization declines with age, however, many adults who were immunized in infancy are susceptible to pertussis. Erythromycin achieves high concentrations in respiratory secretions and is effective in eliminating organisms from the respiratory tract of patients. In exposed, susceptible persons, erythromycin may be effective in preventing or lessening severity of the disease if administered during the preparoxysmal stage. Immunization against pertussis is unnecessary if the patient has had culture-proven pertussis.

**355. The answer is e.** (*Behrman, 16/e, pp 977–981. McMillan, 3/e, pp 1107–1110. Rudolph, 21/e, pp 1035–1039.*) To prove a diagnosis of infectious mononucleosis, a triad of findings should be present. First, physical findings can include diffuse adenopathy, tonsillar enlargement, an enlarged spleen, small hemorrhages on the soft palate, and periorbital swelling. Second, the hematologic changes should reveal a predominance of lymphocytes with at least 10% of these cells being atypical. Third, the characteristic antibody response should be present. Traditionally, heterophil antibodies can be detected when confirming a diagnosis of infectious mononucleosis. These antibodies may not be present, however, particularly in young children. Alternatively, specific antibodies against viral antigens on the Epstein-Barr virus can be measured. Antibodies to viral capsid antigen (VCA) and to anti-D early antigen are elevated prior to the appearance of Epstein-Barr nuclear antigen (EBNA) and are, therefore, markers for acute infection. IgG VCA and EBNA persist for life, whereas anti-D disappears after 6 months.

**356. The answer is b.** (*Behrman, 16/e, pp 776–780. McMillan, 3/e, pp 1009–1012, 2127–2128. Rudolph, 21/e, pp 835, 904–907.*) Acute osteomyelitis tends to begin abruptly, with fever and marked, localized bone tenderness that usually occurs at the metaphysis. Redness and swelling frequently follow. Although usually the result of hematogenous bacterial spread, particularly of *Staphylococcus aureus*, acute osteomyelitis can follow an episode of deep cellulitis and should be suspected whenever deep cellulitis occurs. Diagnosis often must be based on clinical grounds because bone changes may not be visible on x-ray for up to 12 days after onset of the disease. Bone scans with radionuclides, however, can be useful in the early diagnosis of osteomyelitis and in its differentiation from cellulitis and septic arthritis. Caution must be exercised, however, when interpreting a normal bone scan

in a patient suspected of having osteomyelitis; falsely normal bone scans do occur in patients with active bone infection. Antibiotic treatment must be initiated immediately to avoid further extension of infection into bone, where adequate drug levels are difficult to achieve. Treatment is usually continued for at least 3 weeks.

**357. The answer is c.** (*Behrman, 16/e, pp 810–813. McMillan, 3/e, pp 404–413, 421–427. Rudolph, 21/e, pp 900–904.*) Many organisms can cause meningitis in the neonate, including *Escherichia coli*, *Listeria monocytogenes*, *Haemophilus influenzae*, gram-negative rods, group B streptococci, and coagulase-positive and coagulase-negative staphylococci. Clinical manifestations of meningitis in neonates include lethargy, bulging fontanelle, seizures, and nuchal rigidity. The diagnosis is made with examination and culture of the cerebrospinal fluid. Treatment is begun while awaiting the results of the spinal fluid analysis. Appropriate antibiotic coverage must include activity against gram-positive and gram-negative organisms (ampicillin and gentamicin or cefotaxime).

**358. The answer is c.** (*Behrman, 16/e, pp 875–878. McMillan, 3/e, pp 940–942, 1969–1970. Rudolph, 21/e, pp 917–919.*) Infant botulism is a neuromuscular disease caused by the toxin of *Clostridium botulinum*. The disease is distinct from classic botulism in that spores are ingested and the toxin is synthesized by the organism while it resides in the infant's intestine. The toxin is then absorbed and produces weakness and paralysis (including those of the eyes) because of impaired release of acetylcholine at the neuromuscular synapse. Presenting signs and symptoms include poor feeding, lack of stooling, lethargy, weak cry, decreased spontaneous movement, dysphagia, poor head control, and loss of suck and gag reflexes. Death can occur if the respiratory muscles are involved or if the infant cannot control their secretion. Recent evidence suggests a broad clinical spectrum of infant botulism; some infants may never require hospitalization and demonstrate only minimal feeding difficulties. Infants who survive show a complete resolution of symptoms.

**359. The answer is a.** (*Behrman, 16/e, pp 743, 803. McMillan, 3/e, pp 850–855, 961–964, 995–998, 1002–1006, 1124–1126. Rudolph, 21/e, pp 933–936, 977–979, 981–985, 993–999, 1039–1040.*) In an infant who appears otherwise normal, the sudden onset of high fever, together with a

marked elevation and shift to the left of the white blood cell count, suggests pneumococcal bacteremia. Viral infections such as roseola seldom cause such profound shifts in the blood leukocyte count. Streptococcosis refers to prolonged, low-grade, insidious nasopharyngitis that sometimes occurs in infants infected with group A  $\beta$ -hemolytic streptococci. Neither typhoid fever nor diphtheria produces markedly high white blood cell counts; both are characterized by headache, malaise, and other systemic signs. Other bacteria that should be considered in a child with this presentation include *Haemophilus influenzae* type B and meningococcus.

**360. The answer is c.** (*Behrman, 16/e, pp 951–953. McMillan, 3/e, pp 442–444, 1135–1136. Rudolph, 21/e, pp 1075–1079, 2312–2313.*) When German measles (rubella) occurs during the first 2 months of pregnancy, it has a severe effect on the fetus, including cardiac defects, cataracts, and glaucoma. The most common cardiac defects are patent ductus arteriosus, which can be accompanied by peripheral pulmonary artery stenosis, and atrial and ventricular septal defects. A myriad of other complications vary in incidence with the timing of the infection during pregnancy, including thrombocytopenia, hepatosplenomegaly, hepatitis, hemolytic anemia, microcephaly, and a higher risk of developing insulin-dependent diabetes mellitus.

**361. The answer is d.** (*Behrman, 16/e, pp 836, 1934–1935. McMillan, 3/e, p 670. Rudolph, 21/e, pp 1218–1219, 2409.*) The presence of proptosis, pain, edema of the conjunctiva, ophthalmoplegia, and a decrease or loss of visual acuity distinguishes orbital cellulitis from periorbital or preseptal cellulitis. In the latter condition, the globe has a full range of motion, and vision is unimpaired. The most common cause of orbital cellulitis in children is infection in the adjacent paranasal sinuses. Computed tomography (CT) scan of the orbit is indicated to determine the extent of orbital involvement and the need for surgical intervention for drainage of infected sinuses, orbital abscesses, or subperiosteal abscesses.

**362. The answer is d.** (*Behrman, 16/e, pp 946–950. McMillan, 3/e, pp 1137–1141. Rudolph, 21/e, pp 1053–1056.*) The picture and clinical history presented are most common for the diagnosis of measles (rubeola). This is an uncommon disease in areas where immunization rates are high, but sporadic outbreaks do occur. The rash typically lasts 6 days. Complications are common, including pneumonia, laryngitis, myocarditis, and encephalitis.

**363. The answer is a.** (*Behrman, 16/e, pp 1275–1278. McMillan, 3/e, pp 572, 971, 997, 1307–1310. Rudolph, 21/e, pp 1276, 1943.*) Children who have acute epiglottitis, a life-threatening infection of the hypopharynx caused by *Haemophilus influenzae*, typically present with high fever, extremely sore throat, and a croupy cough. It is now a rare disease due to the widespread use of Hib vaccine. An increased incidence of epiglottitis is now seen due to *Streptococcus pneumoniae* and in immunocompromised children. Physical examination characteristically shows a red throat and a red, swollen epiglottis that can be obscured by exudate or so distorted that its identity is misinterpreted. It is important that caution be exercised while attempting to visualize the epiglottis. Abrupt glottic spasm is a well-recognized, potentially fatal complication in these patients. Affected children often are unable to swallow saliva, and because the swollen epiglottis can unpredictably and suddenly cause total and fatal airway obstruction, immediate hospitalization is mandatory, even in the absence of severe respiratory distress. If a diagnosis of acute epiglottitis is uncertain, a lateral x-ray of the neck will differentiate epiglottic from subglottic swelling, the latter being associated with a less serious disease, viral croup.

**364. The answer is a.** (*Behrman, 16/e, pp 946–953, 964–966, 984–986, 1989–1990. McMillan, 3/e, pp 721–722, 1098–1100, 1124–1125, 1134–1140. Rudolph, 21/e, pp 1039–1040, 1053–1056, 1058–1059, 1075–1079, 1195–1197.*) Symptoms of rubella, usually a mild disease, include a diffuse maculopapular rash that lasts for 3 days, marked enlargement of the posterior cervical and occipital lymph nodes, low-grade fever, mild sore throat, and, occasionally, conjunctivitis, arthralgia, or arthritis. Persons with rubeola develop a severe cough, coryza, photophobia, conjunctivitis, and a high fever that reaches its peak at the height of the generalized macular rash, which typically lasts for 5 days. Koplik spots on the buccal mucosa are diagnostic. Roseola is a viral exanthem of infants, in which the high fever abruptly abates as a rash appears. Erythema infectiosum (fifth disease) begins with bright erythema on the cheeks (“slapped cheek” sign), followed by a red maculopapular rash on the trunk and extremities, which fades centrally at first. Erythema multiforme is a poorly understood syndrome consisting of skin lesions and involvement of mucous membranes. A number of infectious agents and drugs have been associated with this syndrome.

**365. The answer is c.** (*Behrman, 16/e, pp 954–955. McMillan, 3/e, pp 1141–1142. Rudolph, 21/e, pp 1056–1058.*) The child in this question likely

has mumps. Although mumps is usually thought of as a parotitis, it is a generalized infection and, as such, can have widespread effects and a variety of clinical presentations. Many infections with the mumps virus are unrecognized because of the substantial rate of subclinical attacks. Meningitis, pancreatitis, and renal involvement can occur as part of the disease. The meningitis that occurs with mumps can occur at the same time as the parotitis, or following the parotitis by about 10 days. Orchitis, seen most frequently in postpubertal males, has been reported in young children as well. All of these problems can be prevented by prior immunization with live attenuated virus vaccine. Many patients with mumps have some white cells in their spinal fluid. Arthritis is rare in children. Subendocardial fibroelastosis in a newborn was once thought to be associated with prenatal infection with mumps, but this does not appear to be true.

**366. The answer is c.** (*Behrman, 16/e, pp 769–771. McMillan, 3/e, pp 447–448, 1723–1725. Rudolph, 21/e, pp 1495–1500.*) Hepatitis A (infectious hepatitis) is characterized by a relatively short incubation period (15 to 50 days) following transmission of the virus, which is primarily by the fecal-oral route. Its onset is abrupt, with sudden fever, nausea, vomiting, anorexia, and tenderness of the liver, soon followed by jaundice. Subclinical infections are common. Elevated serum levels of bilirubin and aspartate aminotransferase (serum glutamic-oxaloacetic transaminase, SGOT) are transient, usually not persisting for more than 3 weeks. Viremia is brief and the period of maximum infectivity of stools usually occurs during the 2 weeks prior to the onset of jaundice. Hepatitis B (serum hepatitis), usually transmitted parenterally via blood or blood products, can also be transmitted nonparenterally via body fluids such as saliva or semen. Following a long incubation period (40 to 180 days), there is a gradual onset of low fever, anorexia, and jaundice, often preceded or accompanied by extrahepatic manifestations such as macular rashes, arthralgias, or urticaria, which may mimic serum sickness. Levels of SGOT and serum bilirubin can be elevated for months, the latter sometimes rising to levels greater than 20 mg/dL when associated with the fulminant hepatitis more often seen with hepatitis B infection. Viremia usually persists throughout the clinical course of hepatitis B infections and can progress to a chronic carrier state in 10% of affected persons, most of whom are asymptomatic; these can be identified by the persistence of the viral surface antigen HBsAg in their blood. A third type of hepatitis, hepatitis C (formerly known as non-A, non-B hepatitis), is important because it accounts for 80 to 90% of post-

transfusion hepatitis in the United States. In addition, it is associated with a high rate of chronicity (25 to 50%).

**367. The answer is e.** (Behrman, 16/e, pp 797–798, 865–867, 910–914, 922–925. McMillan, 3/e, pp 717–718, 898–902, 951–953, 1012, 1017–1021, 1137–1140, 2160–2162. Rudolph, 21/e, pp 946–948, 990–993, 1008–1009, 1013–1015, 1053–1056, 1225–1226.) The incubation period for Rocky Mountain spotted fever (RMSF) has a range of 1 to 14 days. A brief prodromal period consisting of headache and malaise is typically followed by the abrupt onset of fever and chills. A maculopapular rash starts on the second to fourth day of illness on the flexor surfaces of the wrists and ankles before moving in a central direction. Typically, the palms and soles are involved. The rash can become hemorrhagic within 1 or 2 days. Hyponatremia and thrombocytopenia may be seen.

Tularemia has a variable presentation including abrupt onset of fever, chills, malaise, weakness, and headache, and also a variety of skin rashes. Children often have fever, pharyngitis, hepatosplenomegaly, and nonspecific constitutional symptoms.

In the differential diagnosis of RMSF are a number of other diseases. A morbilliform eruption can precede a petechial rash caused by *Neisseria meningitidis*. Viral infections, particularly by the enteroviruses, can cause a severe illness that resembles RMSF. Atypical measles is seen primarily in persons who received the killed measles vaccine before 1968. After exposure to wild-type measles, such a person can develop a prodrome consisting of fever, cough, headache, and myalgia. This is usually followed by the development of pneumonia and an urticarial rash beginning on the extremities. Toxic shock syndrome (TSS) is a disease characterized by sudden onset of fever, diarrhea, shock, inflammation of mucous membranes, and a diffuse macular rash resulting in desquamation of the hands and feet. Lyme disease is seen with an early period of localized disease including erythema migrans, possibly with flulike symptoms, followed by a distinctive period of erythema migrans, arthralgia, arthritis, meningitis, neuritis, and carditis.

**368. The answer is e.** (Behrman, 16/e, pp 885–897. McMillan, 3/e, pp 1026–1039. Rudolph, 21/e, pp 949–959.) Allergic response to tuberculous and nontuberculous mycobacteria is the basis for the intracutaneous Mantoux test; the test becomes positive within 2 to 10 weeks after infection.

Cross-reactions to atypical mycobacteria sometimes occur, especially in the southeastern United States, with reactions up to 15 mm occurring (but usually less than 10 mm). The Mantoux test can become negative during advanced stages of tuberculosis or briefly after immunization with live virus vaccines (such as those for measles, mumps, and rubella) and during administration of corticosteroids or immunosuppressive drugs; otherwise, it is positive for life. Reaction to nontuberculous mycobacteria, however, can wane over time.

Determining what size of reaction is positive can be difficult, as overlap between normal and diseased children occurs. In a child with no known risk factors a reaction over 15 mm is probably positive. For a child in a high-risk group (homeless, prisoners, foreign-born from high-prevalence areas, IV or other drug abusers, and children living with adults in any of the previous groups) a reaction less than 15 mm may be considered positive. In some children (those with suspicious chest radiographs or clinical symptoms of tuberculosis, HIV positive children, and children known to be exposed to an adult with contagious disease), induration as small as 5 mm is often considered positive. A positive skin test in a child likely warrants antimicrobial therapy. Children, who rarely develop cavitory disease, are not contagious.

**369. The answer is c.** (Behrman, 16/e, p 1990. McMillan, 3/e, pp 721–722. Rudolph, 21/e, pp 1195–1197.) The combination of erythema multiforme and vesicular, ulcerated lesions of the mucous membranes of the eyes, mouth, anus, and urethra defines the Stevens-Johnson syndrome (erythema multiforme major). Fever is common, and even pulmonary involvement occasionally is noted; the mortality rate can approach 10%. Common complications include corneal ulceration; dehydration due to severe stomatitis, and, subsequently, poor fluid intake; and urinary retention caused by dysuria. Among the known causes of the Stevens-Johnson syndrome are allergy to various drugs (including phenytoin, barbiturates, sulfonamides, and penicillin) and infection with a variety of organisms including *Mycoplasma pneumoniae* or herpes type 1.

Erythema multiforme is sometimes confused with urticaria early in the course because both can have a target-like lesion. The former has a target center that contains a papule or vesicle, which progresses to blister and necrose. In contrast, urticaria can have a bluish center, but the lesions are transient (often lasting less than 24 h) and are pruritic. Kawasaki disease is

an acute febrile illness of unknown etiology, sharing many of its clinical manifestations with scarlet fever. Scarletiform rash, desquamation, erythema of the mucous membranes that produces an injected pharynx and strawberry tongue (but not sloughing, as in the case presentation), and cervical lymphadenopathy are prominent findings in both. Persons with rubella develop a severe cough, coryza, photophobia, conjunctivitis, and a high fever that reaches its peak at the height of the generalized macular rash, which typically lasts for 5 days. The oral changes include Koplik spots (transient white pinpoint lesions on a bright red buccal mucosa often in the area opposite the lower molars) but not sloughing.

**370. The answer is e.** (Behrman, 16/e, pp 944–945, 1036–1040, 1067–1068, 1072–1073. McMillan, 3/e, pp 1164–1166, 1174–1177, 1193–1195, 1198–1203. Rudolph, 21/e, pp 1095–1097, 1105–1106, 1111–1112, 1126–1128, 1131–1132.) One to seven days following the ingestion of pork or other improperly cooked meat infected with *Trichinella spiralis*, symptoms develop, including abdominal pain, nausea, vomiting, and malaise. During the second week, muscle invasion occurs, which causes edema of eyelids, myalgia, weakness, fever, and eosinophilia. The muscle organisms can become encysted and remain viable for years. Other common parasites travel through body tissue to create disease as well. *Ascaris lumbricoides* larvae travel through the intestinal wall and end up, by way of the liver, in the lungs, where they commonly produce pneumonia and peripheral eosinophilia (Löffler syndrome); worms mature in the small intestine, where they sometimes cause obstruction. The larvae of *Toxocara canis* migrate from the intestine to all parts of the body, where granulomatous reactions can occur (visceral larva migrans). Hookworms (*Necator americanus*) can cause intestinal blood loss from mucosal laceration; cutaneous larva migrans occurs when hookworm larvae fail to enter cutaneous blood vessels after penetrating the skin. *Enterobius vermicularis* (pinworm) causes a localized infestation resulting in perianal pruritis and does not have a tissue phase that would cause eosinophilia. Sporotrichosis is a chronic fungal infection that typically is limited to cutaneous and subcutaneous tissues. *Giardia* and *Cryptosporidium* rarely penetrate the gut wall, instead causing gas, diarrhea, and bloating as common findings, but not fever and eosinophilia.

**371. The answer is c.** (Behrman, 16/e, pp 910–914. McMillan, 3/e, pp 951–953, 2160–2162. Rudolph, 21/e, pp 946–948.) Lyme disease, caused by

the spirochete *Borrelia burgdorferi* and transmitted mostly by ticks of the ioxodes family, is characterized by a unique skin lesion, recurrent attacks of arthritis, and occasional involvement of the heart and central nervous system (CNS). Illness usually appears in late summer or early fall, 2 to 30 days after a bite by an infecting tick. Erythema chronicum migrans begins as a red macule, usually on the trunk at the site of tick attachment that enlarges in a circular fashion with central clearing. Nonspecific systemic signs include headache, fever, and malaise. Joint involvement generally occurs days to years after onset of the rash. Cardiac disease consists primarily of disturbances of rhythm. Involvement of the CNS is evidenced by headache and stiff neck. The diagnosis should be suspected when any of the signs and symptoms occur, because the disease can present in an atypical manner. The characteristic lesion of erythema chronicum migrans as well as the history of tick bite have frequently not been noted by the patient. It is not until late joint, heart, or neurologic manifestations occur, and Lyme disease is suspected, that serologic evidence confirms the etiology. Serologic evidence is sought when the patient has spent time in summer months in endemic areas or there is a risk of tick bite. Treatment with penicillin or tetracycline results in a faster resolution of symptoms and prevention of later complications, especially if given early in the course of the disease.

**372–373. The answers are 372-e, 373-a.** (Behrman, 16/e, pp 826–828, 1800–1801. McMillan, 3/e, pp 901, 982, 2267–2268. Rudolph, 21/e, pp 970–972, 1013–1017.) Rocky Mountain spotted fever and meningococemia can present in a similar fashion. Meningococemia can be complicated by a variety of disorders, including meningitis, purulent pericarditis, endocarditis, pneumonia, otitis media, and arthritis. (Arthritis associated with meningococemia may be mediated by an immune mechanism rather than bacterial invasion of the joint.) The potent endotoxin of the causative organism, *Neisseria meningitidis*, can induce shock, disseminated intravascular coagulation with associated hemorrhaging, and acute adrenal failure caused by localized intraadrenal bleeding; these reactions can be collectively referred to as the Waterhouse-Friderichsen syndrome. Vaccines against *N. meningitidis* groups A, C, Y, and W135 are now available. Prophylaxis with sulfadiazine for sensitive organisms or with rifampin for those in close contact with affected persons is recommended. In contrast, Rocky Mountain spotted fever is often a more indolent infection, with a rash developing days after the onset of the fever and other symptoms.

This course is not invariable, however, and antibiotic therapy for the patient for whom Rocky Mountain spotted fever and meningococcemia cannot be differentiated should not be delayed.

The importance and urgency of the lumbar puncture in cases of suspected meningitis outweigh the usual niceties in the performance of procedures. Infants and children require adequate restraints, preferably local anesthesia, and sometimes sedation. Contraindications are few and include increased intracranial pressure in the patient without an open fontanelle that can result in herniation, severe cardiorespiratory distress, skin infection at the puncture site, and severe thrombocytopenia.

**374. The answer is c.** (Behrman, 16/e, pp 1277–1278. McMillan, 3/e, pp 1307–1310. Rudolph, 21/e, pp 1275–1277.) The description is that of croup. This infection involves the larynx and trachea; it usually is caused by parainfluenza or respiratory syncytial viruses. The usual age range for presentation is 6 months to 6 years. Symptoms include low-grade fever, barking cough, and hoarse, inspiratory stridor without wheezing. The pharynx can be normal or slightly red, and the lungs usually clear. In children in severe respiratory distress, prolonged dyspnea can progress to physical exhaustion and fatal respiratory failure. Because agitation can be a sign of hypoxia, sedation should not be ordered. Hyperinflation on chest x-ray is seen in asthma, not croup. One condition in the differential in this child is epiglottitis, a now rare disease owing to widespread use of the vaccine. Its presentation is more abrupt, with higher fever in rather toxic-appearing patients.

**375. The answer is e.** (Behrman, 16/e, pp 842–848, 944–945, 1039–1040, 1054–1062, 1067–1068. McMillan, 3/e, pp 1002–1006, 1164–1166, 1174–1176, 1184–1193, 1995. Rudolph, 21/e, pp 981–985, 1095–1097, 1105–1106, 1126–1129, 1146–1148.) *Cryptosporidium* has become an important cause of diarrhea in immunocompromised patients, particularly those with AIDS. It can also affect patients who are immunocompetent, and, with increased experience in the laboratory detection of the organism, it has been recognized as an agent responsible for epidemics of diarrhea in day-care centers. Persistent, nonsuppurative diarrhea can be caused by such organisms as amebas, whipworms (trichuriasis), *Cryptosporidium*, or *Giardia lamblia*. *Salmonella sonnei* can be grown in culture, but microscopy is not helpful other than finding fecal leukocytes. *Enterobius vermicularis* is

pinworms, which causes rectal itching and not diarrhea. Sporotrichosis is a fungal infection of the cutaneous and subcutaneous tissue that typically does not cause diarrhea. Acquired *Toxoplasma gondii* can infest any body tissue. Infection can result in fever, myalgia, lymphadenopathy, maculopapular rash, hepatomegaly, pneumonia, encephalitis, chorioretinitis, or myocarditis. This intracellular parasite does not ordinarily cause diarrhea and is not found in stools. Congenital toxoplasmosis can occur if a mother first acquires the parasite during pregnancy. Her infected newborn infant can demonstrate jaundice, hepatosplenomegaly, hydrocephalus or microcephaly, intracranial calcification, or chorioretinitis.

**376–379. The answers are 376-b, 377-a, 378-e, 379-c.** (Behrman, 16/e, pp 331, 595–597, 599–600, 602–604. McMillan, 3/e, pp 867, 2071–2072, 2092–2101. Rudolph, 21/e, pp 793–802, 1189.) Many primary immunologic deficiencies can be classified as defects of T-lymphocyte function (containment of fungi, protozoa, acid-fast bacteria, and certain viruses) and B-lymphocyte function (synthesis and secretion of immunoglobulins). Among the T-cell diseases is DiGeorge anomaly, in which defective embryologic development of the third and fourth pharyngeal pouches results in hypoplasia of both thymus and parathyroid glands. Associated findings with DiGeorge anomaly include CATCH: C for cardiac, A for abnormal faces, T for thymic hypoplasia, C for cleft palate, and H for hypocalcemia.

Primary B-cell diseases include panhypogammaglobulinemia (Bruton disease), an X-linked deficiency of all three major classes of immunoglobulins, as well as other selective deficiencies of the immunoglobulins or their subgroups. This condition usually presents after 3 months of age (after maternal antibodies wane) with recurrent and often simultaneous bouts of otitis media, pneumonia, diarrhea, and sinusitis.

Combined T- and B-cell diseases include the X-linked recessive Wiskott-Aldrich syndrome of mild T-cell dysfunction, diminished serum IgM, marked elevation of IgA and IgE, eczema, recurrent middle-ear infections, lymphopenia, and thrombocytopenia. Patients with the catastrophic combined T- and B-cell disease, known as severe combined immunodeficiency disease (Swiss-type lymphopenic agammaglobulinemia or SCID), have deficient T- and B-cells. Consequently, they are both marked lymphopenia and agammaglobulinemia, as well as hypoplasia of the thymus. Chronic diarrhea;

rashes; recurrent, serious bacterial, fungal, or viral infections; wasting; and early death are characteristic. Other T- and B-cell deficiencies include ataxia-telangiectasia and chronic mucocutaneous candidiasis.

Job-Buckley syndrome is a disorder of phagocytic chemotaxis associated with hypergammaglobulin E, eczema-like rash, and recurrent severe staphylococcal infections.

**380–385. The answers are 380-b, 381-b, 382-a, 383-d, 384-b, 385-f.** (Behrman, 16/e, pp 862–864, 872–873, 984–986, 1147–1150, 1315–1317. McMillan, 3/e, pp 956–961, 998–1101, 1124–1125. Rudolph, 21/e, pp 923–928, 945–946, 987–990, 999–1001, 1035–1040, 1110–1111, 1430–1431.) The evidence is mounting that *Rochalimaea henselae* is the major etiologic agent for cat-scratch disease. History of a scratch or a bite from a kitten is often positive, and fleas are often a factor in transmission. Diagnosis is usually by history and presenting signs and symptoms as described in the question. Another form of the disease, Parinaud oculoglandular syndrome, occurs when the primary site of inoculation occurs in or near the conjunctivae, resulting in a moderately severe conjunctivitis and preauricular lymphadenopathy.

Infection with *Helicobacter pylori* is associated with antral gastritis and primary duodenal ulcer disease, with the symptoms described in the question. Culture for *H. pylori* requires endoscopy and gastric biopsy. The biopsied tissue can be tested for urease activity and examined histologically after special staining. Alternatively, noninvasive tests involving detection of metabolic products of *H. pylori* (urease activity) or antibodies against *H. pylori* can be demonstrated.

Roseola (exanthema subitum) is a common acute illness of young children, characterized by several days of high fever followed by a rapid defervescence and the appearance of an evanescent, erythematous, maculopapular rash. Human herpesvirus 6 (HHV-6) has been identified as its main cause.

*Pseudomonas* is a ubiquitous organism. Most infections with the organism are opportunistic, involving several organs. Skin infections related to burns, trauma (such as in a puncture wound through a tennis shoe), and use of swimming pools are not uncommon. Injuries through a tennis shoe are prone to pseudomonas infections due to the warm, moist nature of the shoe's environment. In contrast, a wound through a bare foot would be associated with cutaneous flora such as *Staphylococcus*. Cystic fibrosis

(which can present as failure to thrive, recurrent pneumonias, and chronic diarrhea) is characterized by a predilection for colonization of airways with *Staphylococcus aureus* and *Pseudomonas aeruginosa*. *Pseudomonas cepacia* infection is not as common, but it is more problematic in patients with cystic fibrosis.

**386–388. The answers are 386-b, 387-a, 388-d.** (Behrman, 16/e, pp 1067–1068, 1967. McMillan, 3/e, pp 918, 1153–1155, 1195. Rudolph, 21/e, pp 1105–1106, 1168, 1173, 1226–1230.) Several simple tests are available to assist in the diagnosis of dermatologic conditions. Among these are the Wood's lamp, which uses a wavelength of 365 nm to identify fungal infections of the skin. Blue-green fluorescence is detected at the base of hair shafts infected with certain strains of fungus responsible for tinea capitis. A negative Wood's lamp result does not rule out fungal infection because not all strains become apparent. The black pinpoint dots described are hair follicles; the boggy appearance is a result of the host immune response and usually does not indicate secondary bacterial infection.

For a Tzanck smear, the base of a freshly ruptured blister is scraped, stained with Giemsa stain, and examined under the microscope. Some viral infections such as herpes (which is what the girl in this question likely has) and varicella will demonstrate balloon cells and multinucleated giant cells.

Pinworms (*Enterobius vermicularis*) lay their eggs at night on the perianal region, resulting in a very pruritic rash (usually a toddler). These eggs can be gathered by pressing cellophane tape to the perianal region at night or early in the morning. This tape is then examined under the microscope to detect the characteristic eggs.

The central African immigrant probably merits at least malaria preparation of his blood, and the infant with foul-smelling urine and fever should have blood and urine cultures, and perhaps a lumbar puncture if the clinical picture warrants.

**389–392. The answers are 389-c, 390-a, 391-g, 392-e.** (Behrman, 16/e, pp 761–765, 885–897, 914–921. McMillan, 3/e, pp 451–452, 893–894, 897, 1009–1012, 1026–1039, 1217, 1227–1230. Rudolph, 21/e, pp 929–931, 949–959, 965–967, 1980–1986.) Approximately 10 to 20% of infants born to mothers with *Chlamydia trachomatis* infection develop pneumonia. The presentation of this pneumonia usually occurs between 1 and 3 months of age, with cough, tachypnea, and lack of fever. Examination reveals rales

but not wheezing. Laboratory data suggestive of *C. trachomatis* infection include an increase in eosinophils in the peripheral blood. The chest radiograph shows hyperinflation with interstitial infiltrates.

*Mycoplasma pneumoniae* is a common cause of pneumonia in the school-aged child or the young adult. Usual presentation includes the gradual onset of headache, malaise, fever, and lower respiratory symptoms. Typically, the cough (often nonproductive) worsens for the first 2 weeks of the illness, then slowly resolves over the ensuing 3 to 4 weeks. Early in the disease, the physical examination is remarkable for a paucity of signs; the patient usually has a few fine rales. Later, the dyspnea and fever become worse. Radiographic findings include an interstitial or bronchial pattern, especially in the lower lobes, and commonly only on one side. The diagnosis is usually made on clinical grounds.

Staphylococcal pneumonia is caused by *Staphylococcus aureus*. It is a rapidly progressive and life-threatening form of pneumonia most commonly seen in children less than 1 year of age. Commonly, the child has an upper respiratory infection for several days, with the abrupt onset of fever and respiratory distress. Pleural effusion, empyema, and pyopneumothorax are common complications. Laboratory evidence of this disease can include a markedly elevated WBC count with left shift. Radiographic findings include nonspecific bronchopneumonia early in the disease, which later becomes more dense and homogeneous and involves an entire lobe or hemithorax.

A high index of suspicion is helpful to diagnose tuberculosis. Tuberculosis should be considered in any patient with pneumonia who belongs to a high-risk group, including poor and indigent persons, present and former residents of correctional institutions, homeless persons, IV drug abusers, health care workers caring for high-risk patients, and children exposed to high-risk adults. Clinical and radiographic findings are variable in children, but include nonspecific cough and dyspnea. Radiographic findings often include hilar adenopathy, focal hyperinflation, and atelectasis.

# Hematologic and Neoplastic Diseases

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## Questions

**DIRECTIONS:** Each item below contains a question or incomplete statement followed by suggested responses. Select the **one best** response to each question.

### Items 393–394

**393.** Two weeks after a viral syndrome, a 2-year-old child develops bruising and generalized petechiae, more prominent over the legs. No hepatosplenomegaly or lymph node enlargement is noted. The examination is otherwise unremarkable. Laboratory testing shows the patient to have a normal hemoglobin, hematocrit, and white blood count and differential. The platelet count is 15,000/ $\mu$ L. The most likely diagnosis is

- a. von Willebrand disease
- b. Acute leukemia
- c. Idiopathic (immune) thrombocytopenic purpura
- d. Aplastic anemia
- e. Thrombotic thrombocytopenic purpura

**394.** Appropriate treatment of this child includes

- a. Intravenous gamma globulin
- b. Platelet transfusion
- c. Aspirin therapy
- d. Factor VIII infusion
- e. Prednisone, vincristine, and asparaginase induction followed by methotrexate and 6-mercaptopurine

**395.** A newborn infant is born with petechiae scattered across his body. His platelet count is noted to be 22,000/ $\mu\text{L}$  with a hemoglobin of 12 mg/dL. Which of the following is most likely to explain this infant's condition?

- a. Congenital cytomegalovirus infection
- b. Uncomplicated prematurity
- c. Chlamydial conjunctivitis
- d. Maternal ingestion of aspirin
- e. Nasolacrimal duct stenosis

**396.** On a routine-screening complete blood count, a 1-year-old is noted to have a microcytic anemia. A follow-up hemoglobin electrophoresis demonstrates an increased concentration of hemoglobin  $A_2$ . The child is most likely to have

- a. Iron deficiency
- b.  $\beta$ -thalassemia trait
- c. Sickle cell anemia
- d. Chronic systemic illness
- e. Lead poisoning

**397.** A 3-year-old child presents with a petechial rash but is otherwise well and without physical findings. Platelet count is 20,000/ $\mu\text{L}$ ; hemoglobin and WBC count are normal. The most likely diagnosis is

- a. Immune thrombocytopenic purpura (ITP)
- b. Henoch-Schönlein purpura
- c. Disseminated intravascular coagulopathy (DIC)
- d. Acute lymphoblastic leukemia
- e. Systemic lupus erythematosus (SLE)

**398.** A 4-year-old previously well boy develops pallor, dark urine, and jaundice. There has been no apparent exposure to a jaundiced person or to any toxins. He is taking trimethoprim-sulfamethoxazole for otitis media. You consider the possibility of a hemolytic crisis caused by glucose-6-phosphate dehydrogenase (G6PD) deficiency. In which of the following ethnic groups is the incidence *lowest*?

- a. African American
- b. Greek
- c. Chinese
- d. Middle Eastern
- e. Scandinavian

**399.** On a routine newborn screen, a 2-week-old child in your clinic is noted to have sickle cell disease. As you discuss treatment options with this family, you recommend

- a. Monthly injections of vitamin B<sub>12</sub>
- b. Tetracycline prophylaxis
- c. Meningococcal vaccine at 2, 4, and 6 months of age
- d. Education of parents regarding abdominal palpation and temperature taking
- e. Infusion of immunoglobulin every 6 weeks

**400.** A 2950-g black baby boy is born at home at term. On arrival at the hospital, he appears pale, but the physical examination is otherwise normal. Laboratory studies reveal the following: mother's blood type A, Rh-positive; baby's blood type O, Rh-positive; hematocrit 38%; reticulocyte count 5%. Which of the following is the most likely cause of the anemia?

- a. Fetomaternal transfusion
- b. ABO incompatibility
- c. Physiologic anemia of the newborn
- d. Sickle cell anemia
- e. Iron-deficiency anemia

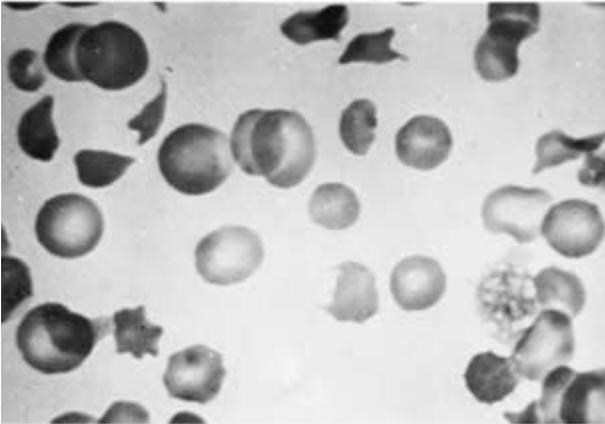
**401.** The family of a child just diagnosed with acute lymphoblastic leukemia asks for information about the child's prognosis. Included as a poor prognostic sign is which of the following?

- a. Presence of a mediastinal mass
- b. Hyperdiploidy with more than 50 chromosomes
- c. White blood cell count at diagnosis of less than 100,000/ $\mu$ L
- d. Age between 1 and 10 years
- e. Early pre-B-cell variety of the disease

**402.** While bathing her 2-year-old son, a mother feels a mass in his abdomen. A thorough medical evaluation of the child reveals aniridia, hypospadias, horseshoe kidney, and hemihypertrophy. The most likely diagnosis for this child is

- a. Neuroblastoma
- b. Wilms tumor
- c. Hepatoblastoma
- d. Rhabdomyosarcoma
- e. Testicular cancer

**403.** A 2-year-old child in shock has multiple nonblanching purple lesions of various sizes scattered about on the trunk and extremities; petechiae are noted, and oozing from the puncture site has been observed. The child's peripheral blood smear is presented. Clotting studies are likely to show which of the following?



- a. Increased levels of factor V and VIII
- b. A decreased prothrombin level
- c. An increased fibrinogen level
- d. The presence of fibrin split products
- e. Normal partial thromboplastin time (PTT)

**404.** During a routine-screening CBC, a 1-year-old is noted to have eosinophilia. Which of the following most commonly causes increased eosinophilia in the peripheral blood smear?

- a. Bacterial infections
- b. Chronic allergic rhinitis
- c. Fungal infections
- d. Helminth infestation
- e. Tuberculosis

**405.** A preterm black male infant was found to be jaundiced 12 h after birth. At 36 h of age, his serum bilirubin was 18 mg/dL, hemoglobin concentration was 12.5 g/dL, and reticulocyte count 9%. Many nucleated red cells and some spherocytes were seen in the peripheral blood smear. The differential diagnosis should include which of the following?

- a. Pyruvate kinase deficiency
- b. Hereditary spherocytosis
- c. Sickle cell anemia
- d. Rh incompatibility
- e. Polycythemia

### Items 406–407

**406.** On a routine well-child examination, a 1-year-old boy is noted to be pale. He is in the seventy-fifth percentile for weight and the twenty-fifth percentile for length. Results of physical examination are otherwise normal. His hematocrit is 24%. Of the following questions, which is most likely to be helpful in making a diagnosis?

- a. What is the child's usual daily diet?
- b. Did the child receive phototherapy for neonatal jaundice?
- c. Has anyone in the family received a blood transfusion?
- d. Is the child on any medications?
- e. What is the pattern and appearance of his bowel movements?

**407.** Having performed a complete history and physical examination on the patient, you proceed with a diagnostic workup. Initial laboratory results are as follows: hemoglobin 8 g/dL; hematocrit 24%; leukocyte count 11,000/ $\mu$ L with 38% neutrophils, 7% bands, 55% lymphocytes; hypochromia on smear; free erythrocyte protoporphyrin (FEP) 110  $\mu$ g/dL; lead level 7  $\mu$ g/dL whole blood; platelet count adequate; reticulocyte count 0.5%; sickle cell preparation negative; stool guaiac negative; and mean corpuscular volume (MCV) 65 fl. You would most appropriately recommend

- a. Blood transfusion
- b. Oral ferrous sulfate
- c. Intramuscular iron dextran
- d. An iron-fortified cereal
- e. Calcium EDTA

**408.** A 10-year-old boy is admitted to the hospital because of bleeding. Pertinent laboratory findings include a platelet count of 50,000/ $\mu\text{L}$ , prothrombin time (PT) of 15 s (control 11.5 s), activated partial thromboplastin time (aPTT) of 51 s (control 36 s), thrombin time (TT) of 13.7 s (control 10.5 s), and factor VIII level of 14% (normal 38 to 178%). The most likely cause of his bleeding is

- a. Immune thrombocytopenic purpura (ITP)
- b. Vitamin K deficiency
- c. Disseminated intravascular coagulation (DIC)
- d. Hemophilia A
- e. Hemophilia B

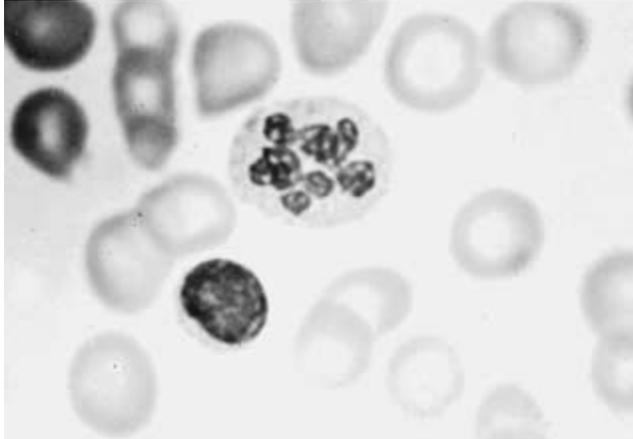
**409.** An 8-year-old child being treated with a combination of chemotherapy agents develops very red, inflamed sores in the mouth and esophagus. He has difficulty eating and drinking food and liquids. Which of the following antineoplastic agents is the most likely etiology?

- a. Vincristine
- b. Prednisone
- c. Methotrexate
- d. 6-mercaptopurine
- e. Doxorubicin (Adriamycin)

**410.** An otherwise healthy 17-year-old complains of swollen glands in his neck and groin for the last 6 months and an increasing cough over the previous 2 weeks. He also reports some fevers, especially at night, and possibly some weight loss. On examination, you notice that he has nontender cervical, supraclavicular, axillary, and inguinal nodes, no hepatosplenomegaly, and otherwise looks to be fairly healthy. Which of the following would be the appropriate *next* step?

- a. Biopsy of a node
- b. Complete blood count and differential
- c. Trial of antituberculous drugs
- d. Chest radiograph
- e. Cat-scratch titers

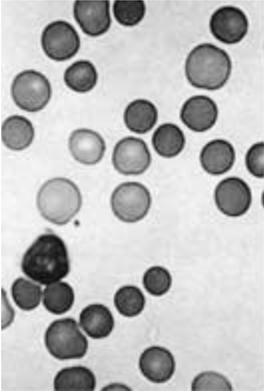
**411.** An otherwise healthy child has on his 1-year-old routine CBC the polymorphonuclear neutrophil shown in the following illustration. This child likely has



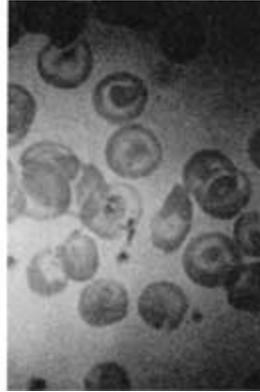
- a. Malignancy
- b. Iron deficiency
- c. Folic acid deficiency
- d. Döhle inclusion bodies
- e. The Pelger-Huët nuclear anomaly

**DIRECTIONS:** The group of questions below consists of lettered options (in figure) followed by a set of numbered items. For each numbered item, select the appropriate lettered option(s). Each lettered option may be used once, more than once, or not at all. **Choose exactly the number of options indicated following each item.**

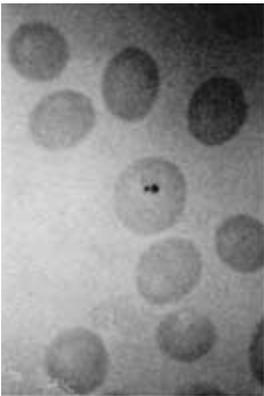
**Items 412–415**



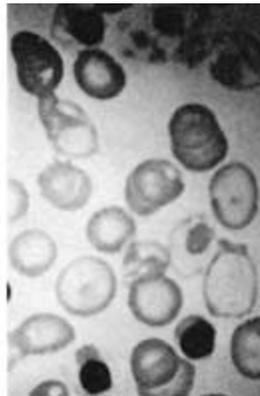
**a**



**b**



**c**



**d**

For each disorder listed below, select the peripheral blood smear with which it is most likely to be associated.

- 412.** An 8-year-old patient with sickle cell anemia (**SELECT 1 SMEAR**)
- 413.** A 7-month-old boy with severe anemia requiring transfusions, heart failure, hepatosplenomegaly, and weakness (**SELECT 1 SMEAR**)
- 414.** A 3-day-old newborn with anemia and pathologic hyperbilirubinemia requiring phototherapy (**SELECT 1 SMEAR**)
- 415.** A completely asymptomatic, healthy 1-year-old whose routine CBC reveals an abnormality. (**SELECT 1 SMEAR**)

# Hematologic and Neoplastic Diseases

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## Answers

**393–394. The answers are 393-c, 394-a.** (*Behrman, 16/e, pp 1520–1522. McMillan, 3/e, pp 367, 1477–1479. Rudolph, 21/e, pp 1556–1557.*) In children, idiopathic or immune thrombocytopenic purpura (ITP) is the most common form of thrombocytopenic purpura. In most cases a preceding viral infection can be noted. No diagnostic test exists for this disease; exclusion of the other diseases listed in the question is necessary. In this disease, the platelet count is frequently less than 20,000/ $\mu\text{L}$ , but other laboratory tests yield essentially normal results, including the bone marrow aspiration (if done). For ITP, platelets are sequestered and destroyed at the spleen by the reticuloendothelial system (RES) that binds self-immunoglobulins attached to the platelet. Exogenous IV gamma globulin can work to saturate the RES binding sites for platelet-bound self-immunoglobulin. Thus, there is less platelet uptake and destruction by the spleen.

Aplastic anemia is unlikely if the other cell lines are normal. Von Willebrand disease might be expected to present with bleeding and not just bruising. It is unlikely that acute leukemia would present with thrombocytopenia only. Thrombotic thrombocytopenic purpura is rare in children. Treatment for ITP consists of observation and/or gamma globulin and steroids. Splenectomy is reserved for the most severe and chronic forms.

**395. The answer is a.** (*Behrman, 16/e, pp 526–527, 981–983, 1523. McMillan, 3/e, pp 367, 1475–1476, 1479–1480. Rudolph, 21/e, pp 1031–1035, 1558, 2372–2373.*) Thrombocytopenia and hemolytic anemia, as seen in this patient, are common manifestations of the TORCH (toxoplasmosis, other, rubella, cytomegalovirus, and herpes simplex) infections as well as congenital syphilis. Both increased platelet destruction and impaired production of platelets may be the mechanisms involved. Some mothers who have had ITP and who have high levels of antiplatelet antibody in the maternal plasma can give birth to thrombocytopenic infants because of transplacental crossing of antiplatelet IgG antibody. The syndrome of congenital amegakaryocytic thrombocytopenia and bilateral absence of the radii (thrombocytopenia-

absent radius, or TAR, syndrome) is a well-known entity. Maternal ingestion of aspirin can lead to bleeding in the newborn, not as a result of thrombocytopenia but as a consequence of transient impairment of the infant's platelet aggregation. Neither chlamydial conjunctivitis nor nasolacrimal duct stenosis is usually associated with thrombocytopenia.

**396. The answer is b.** (*Behrman, 16/e, p 1460. McMillan, 3/e, pp 1447–1448, 1451–1453. Rudolph, 21/e, pp 842–844, 1536–1538.*) The concentration of hemoglobin A<sub>2</sub> is increased in  $\beta$ -thalassemia trait. In severe iron deficiency, hemoglobin A<sub>2</sub> may be decreased. In mild-to-moderate iron deficiency, the level of hemoglobin A<sub>2</sub> is normal. The level is also normal in sickle cell anemia, chronic systemic illness, and lead poisoning.

**397. The answer is a.** (*Behrman, 16/e, pp 1520–1522. McMillan, 3/e, pp 1477–1479, 1488, 1490–1491, 1493–1507, 2162–2165, 2176–2177. Rudolph, 21/e, pp 298–301, 847–851, 1556–1557, 1575, 1594–1600, 1688–1689.*) The mean age of presentation of ITP is 6 years. Patients look well except for petechial rash. Patients with acute lymphoblastic leukemia frequently have symptoms of pallor and fever in addition to bleeding. Nearly 50% of them have hepatomegaly and splenomegaly. CBC reveals anemia, leukocytosis or leukopenia, and thrombocytopenia. Disseminated intravascular coagulopathy (DIC) is secondary to a severe underlying disease, such as fulminant bacterial sepsis with hypotension or profound hypoxia. Patients invariably appear ill and have leukocytosis, thrombocytopenia, and abnormal coagulation studies (e.g., prolonged PT and PTT, decreased fibrinogen concentration, and elevated fibrin split products). Patients with Henoch-Schönlein purpura have symptoms of skin rash and abdominal or joint pain. The rash is usually urticarial and purpuric and present over the buttocks or lower extremities. The platelet count is normal or elevated. Systemic lupus erythematosus (SLE) is very rare in a 3-year-old child. Findings include fever, joint pain, and skin rash. CBC can reveal anemia, leukopenia, and thrombocytopenia.

**398. The answer is c.** (*Behrman, 16/e, pp 1489–1491. McMillan, 3/e, pp 1455–1456, 2086. Rudolph, 21/e, pp 1542–1543.*) Synthesis of the red cell enzyme glucose-6-phosphate dehydrogenase (G6PD) is determined by genes on the X chromosome, and the pattern of inheritance is X-linked recessive. The enzyme found in most populations is termed G6PDB1. There are over

380 deficient variants of the enzyme affecting over 100 million people worldwide, among them G6PDA1, a mutant enzyme affecting about 13% of African American males and 2% of African American females; G6PDB or Mediterranean, a deficient mutant occurring among Middle Eastern, African, and Asian groups; and G6PD Canton occurring in 5% of Chinese.

In people with G6PDB1, enzyme activity is reduced to 50% during the 120-day life span of the erythrocyte. In persons affected with all G6PD deficiency variants, enzyme activity is always 10% of normal or less. Deficiency of G6PD compromises the generation of reduced glutathione, and upon exposure to oxidant agents such as sulfa drugs, antimalarials, nitrofurans, naphthalene mothballs, or infection, a hemolytic episode usually occurs. The degree of hemolysis depends on the nature of the oxidant and severity of the enzyme deficiency. In African Americans, the older, more G6PD-deficient cells are destroyed, but since young cells have sufficient enzyme to prevent further red cell destruction even if the inciting factor is still present, the hemolytic crisis is usually self-limited. Blood transfusion may be unnecessary.

In African Americans, premature testing for the enzyme immediately after a hemolytic episode can lead to a false negative result since the newly produced red cells in the circulation have a higher G6PD enzyme activity. The older red cells containing Heinz bodies (insoluble precipitates resulting from oxidation), the "bite cells" (red cells after the removal of the Heinz bodies), and cell fragments are removed from the circulation within 3 to 4 days. In the severe Mediterranean type, young as well as old red cells are enzyme-deficient. Recovery is signaled by the appearance of reticulocytes and a rise in hemoglobin.

**399. The answer is d.** (*Behrman, 16/e, pp 1479–1483. McMillan, 3/e, pp 1450–1451. Rudolph, 21/e, pp 1531–1534.*) Mass neonatal screening for hemoglobinopathy has made it possible to institute comprehensive care in very early infancy. In the United States, the incidence of SS, SC, and S  $\beta$ -thalassemia combined is 1:400. Acute splenic sequestration is a serious complication of sickle cell disease, usually occurring in patients under 5 years of age. Sudden massive pooling of blood in the splenic vasculature leads to hypovolemia and circulatory collapse. Early detection of splenomegaly by parents who have been taught the technique of abdominal palpation is an important aspect of patient management. The ability

of the spleen to opsonize and phagocytize encapsulated organisms, particularly *Streptococcus pneumoniae* and *Haemophilus influenzae* B, is impaired in children with sickle cell disease as early as 6 months of age, and, as a result, they are at an increased risk of infection with these organisms. A temperature of 38.5°C to 39°C (101.3°F to 102.2°F) calls for emergency evaluation even if the child does not look very sick. Without treatment, death from overwhelming sepsis, commonly due to pneumococcus, can occur within hours. Prophylactic oral penicillin early in infancy has been used to prevent pneumococcal infection. All children receive the 7-valent pneumococcal vaccine beginning at 2 months of age, and patients with sickle cell disease receive the 23-valent pneumococcal vaccine beginning at 2 years of age. Invasive *H. influenzae* B infections have been responsible for significant morbidity and mortality in patients with sickle cell disease. Special efforts should be made to immunize these children, as well as other immunocompromised children, with Hib vaccine as soon as they enter the health care system, even if they are over 5 years of age. Folic acid requirement is increased in sickle cell disease (as in all hemolytic anemias) and, therefore, supplementation is prudent. Vitamin B<sub>12</sub> supplementation is not necessary, and tetracycline is contraindicated because of damage to teeth; meningococcal vaccine is not indicated in children less than 2 years of age, and immunoglobulins have no place in the therapy for this condition.

**400. The answer is a.** (*Behrman, 16/e, pp 519–521. McMillan, 3/e, pp 359–362. Rudolph, 21/e, pp 174–177, 197–199, 1525–1528, 1531–1534.*) The absence of a major blood-group incompatibility and the finding of a normal reticulocyte count argue strongly in favor of a recent fetomaternal transfusion, probably at the time of delivery. A Betke-Kleihauer stain for fetal hemoglobin-containing red cells in the mother's blood would confirm the diagnosis. After birth, erythropoiesis ceases, and the progressive decline in hemoglobin values, reaching a nadir at 6 to 8 weeks of age, has been termed physiologic anemia of infancy. Iron-deficiency anemia is common in the term infant between 9 and 24 months of age when the iron stores derived from circulating hemoglobin have been exhausted and an exogenous dietary source of iron has not been provided. The manifestations of sickle cell disease do not appear until 4 to 6 months of life, coincident with the replacement of fetal hemoglobin with sickle hemoglobin.

**401. The answer is a.** (*Behrman, 16/e, pp 1543–1548. McMillan, 3/e, pp 1493–1507. Rudolph, 21/e, pp 1594–1600.*) Age less than 12 months or more than 10 to 15 years, the presence at diagnosis of central nervous system leukemia or a white blood cell count of 100,000/ $\mu$ L or higher, and the appearance of a mediastinal mass indicate a poor prognosis for children who have acute lymphocytic leukemia. The first two prognostic factors listed above are the most reliable. Hyperdiploidy with more than 50 chromosomes is a good prognostic factor for treatment with antimetabolic-based chemotherapy. Most of the children with these poor prognostic signs have the thymic (T cell) variety of the disease. In addition to the conventionally employed regimen of prednisone and vincristine, other chemotherapeutic agents should be administered. A smaller fraction of these patients with poor prognostic features can be expected to achieve long-term disease-free survival, and, once they relapse, many fail to go into remission despite aggressive chemotherapy. Bone marrow transplantation offers hope in some cases.

**402. The answer is b.** (*Behrman, 16/e, pp 1554–1556. McMillan, 3/e, pp 1515–1517. Rudolph, 21/e, pp 1614–1620.*) An abdominal mass is palpated in 85% of patients with Wilms tumor; abdominal pain is present in 40%, hypertension in about 60%, and hematuria in 12 to 24%. Because of the association of hemihypertrophy and aniridia with Wilms tumor, children with these findings should be followed with periodic physical examinations and abdominal sonograms, especially during their first 5 years. Wilms tumor and aniridia are associated with abnormalities in chromosome 11. Neuroblastoma should also be considered in the differential diagnosis of abdominal mass, especially if fever, irritability, bone pain, limp, and diarrhea are present.

**403. The answer is d.** (*Behrman, 16/e, pp 1519–1520. McMillan, 3/e, p 1488. Rudolph, 21/e, p 1575.*) The clinical history and blood-smear findings (fragmented cells and few platelets) presented in the question are typical of disseminated intravascular coagulation. The disorder, which can be triggered by endotoxin shock, results ultimately in the initiation of the intrinsic clotting mechanism and the generation of thrombin (prolonged PT and PTT, decreased fibrinogen concentration, and an increase in fibrin split products). Fibrin deposited in the microcirculatory system can lead to tissue

ischemia and necrosis, further capillary damage, release of thromboplastic substances, and increased thrombin generation. Simultaneous activation of the fibrinolytic system produces increased amounts of fibrin split products, which inhibit thrombin activity. Of utmost importance in the treatment of children who have disseminated intravascular coagulation is the management of the condition that precipitated the disorder.

**404. The answer is d.** (*Behrman, 16/e, pp 614–615, 651–652, 666. McMillan, 3/e, pp 1464–1465. Rudolph, 21/e, p 1555.*) Some common causes of eosinophilia in the peripheral blood smear include asthma, recurrent urticaria, infantile eczema, serum sickness, angioneurotic edema, helminth infections, collagen vascular disease, and some neoplasms. Allergic rhinitis can cause eosinophilia in nasal secretions, but usually does not cause dramatic peripheral eosinophilia.

**405. The answer is b.** (*Behrman, 16/e, pp 513–517, 1475–1477, 1479–1483, 1488. McMillan, 3/e, pp 1450–1451, 1453–1455. Rudolph, 21/e, pp 174–177, 197–199, 1531–1534, 1539–1540, 1543–1544.*) Spherocytosis can be seen in hyperthermia, hereditary spherocytosis, G6PD deficiency, or ABO incompatibility. Hyperbilirubinemia has been associated with black preterm infants with G6PD deficiency. The blood smear of the affected infant usually reveals nucleated red cells, spherocytes, poikilocytes, “blister” cells, and fragmented cells. Neonatal hyperbilirubinemia occurs in about 50% of patients with hereditary spherocytosis. Spherocytosis occurs in ABO incompatibility but not in Rh incompatibility. The hemolytic manifestations of ABO incompatibility and hereditary spherocytosis are very similar. The blood types of the mother and of the infant should be determined along with the results of a direct Coombs test on the infant and the presence or absence of a family history of hemolytic disease (spherocytosis). Sick cell disease would not be expected to cause problems in newborns due to protection by fetal hemoglobin.

**406. The answer is a.** (*Behrman, 16/e, pp 1469–1471. McMillan, 3/e, pp 1447–1448. Rudolph, 21/e, pp 1525–1529.*) Iron-deficiency anemia is the most common nutritional deficiency in children between 9 and 15 months of age. Low availability of dietary iron, impaired absorption of iron related to frequent infections, high requirements for iron for growth, and, occa-

sionally, blood losses, favor the development of iron deficiency in infants. A history regarding anemia in the family, blood loss, and gestational age and weight can help to establish the cause of an anemia. The strong likelihood is that anemia in a 1-year-old child is nutritional in origin, and its cause will be suggested by a detailed nutritional history.

**407. The answer is b.** (*Behrman, 16/e, pp 1469–1471. McMillan, 3/e, pp 1447–1448. Rudolph, 21/e, pp 1525–1529.*) Response to a therapeutic trial of iron is an appropriate and cost-effective method of diagnosing iron-deficiency anemia. A prompt reticulocytosis and rise in hemoglobin and hematocrit follow the administration of an oral preparation of ferrous sulfate. Intramuscular iron dextran should be reserved for situations in which compliance cannot be achieved. This is because this treatment is expensive, painful, and no more effective than oral iron. Dietary modifications, such as limiting the intake of cow's milk and including iron-fortified cereals along with a mixed diet, are appropriate long-term measures, but they will not make enough iron available to replenish iron stores. The gradual onset of iron-deficiency anemia enables a child to adapt to surprisingly low hemoglobin concentrations. Transfusion is rarely indicated unless a child becomes symptomatic or is further compromised by a superimposed infection. When the iron available for production of hemoglobin is limited, free protoporphyrins accumulate in the blood. Levels of erythrocyte protoporphyrin (EP) are also elevated in lead poisoning. Iron-deficiency anemia can be differentiated from lead intoxication by measuring blood lead, which should be less than 10  $\mu\text{g/dL}$ .

**408. The answer is c.** (*Behrman, 16/e, pp 1519–1520. McMillan, 3/e, p 1448. Rudolph, 21/e, p 1575.*) The prolongation of PT, aPTT, and TT excludes the diagnosis of ITP. PT tests principally for factors I, II, V, VII, and X and is not prolonged in hemophilia A (factor VIII deficiency) or hemophilia B (factor IX deficiency). In vitamin K deficiency, there is a decrease in the production of factors II, VII, IX, and X, and PT and aPTT are prolonged. The thrombin time, which tests for conversion of fibrinogen to fibrin, however, should be normal and the platelet count should also be normal. In DIC, there is consumption of fibrinogen; factors II, V, and VIII; and platelets. Therefore, there is prolongation of PT, aPTT, and TT and a decrease in factor VIII level and platelet count. In addition, the titer of fibrin split production is usually increased.

**409. The answer is c.** (*Behrman, 16/e, p 1538. Rudolph, 21/e, p 1298.*) The description is that of a child with mucositis. The toxicity of methotrexate is dependent on the dose, schedule, and route of administration. The major toxicities include gastrointestinal mucositis, bone marrow suppression, skin erythema, and hepatic dysfunction. The main toxicities of vincristine include peripheral neuropathy, constipation, jaw pain, and inappropriate antidiuretic hormone secretion. The major side effects of prednisone include Cushingoid facies, truncal obesity, salt and water retention, hypertension, increased susceptibility to infection, gastric irritation, and osteoporosis. 6-Mercaptopurine can cause nausea, vomiting, marrow suppression, and hepatic dysfunction. Doxorubicin (Adriamycin) can lead to alopecia, nausea, vomiting, stomatitis, tissue necrosis (if drug extravasates), and bone marrow suppression. The dose-limiting factor is cardiotoxicity, and the risk of cardiotoxicity increases at cumulative doses of doxorubicin above 550 mg/m<sup>2</sup>. In the latter two cases (6-mercaptopurine and doxorubicin), mucositis is possible but is seen less commonly than with methotrexate.

**410. The answer is d.** (*Behrman, 16/e, pp 1548–1550. McMillan, 3/e, pp 1464–1465, 1507–1509. Rudolph, 21/e, pp 1607–1610.*) The patient in this question, especially with the pulmonary findings, may have Hodgkin disease. Although a biopsy of the node may prove to be necessary at some point, the first step would involve a radiograph, which may show mediastinal mass suspicious for Hodgkin disease. Depending on the results of the radiograph, a biopsy of a node may be indicated, especially if the question of Hodgkin (or other malignancy) remained high. A tuberculosis skin test may be indicated, but a trial of antituberculosis drugs without a chest radiograph, a positive PPD, or other diagnostic evaluation indicating tuberculosis would be unwise. In Hodgkin disease, the CBC is not diagnostic but may show anemia, neutropenia, or thrombocytopenia; they are useful in the staging process after the diagnosis has been confirmed by biopsy. The presenting complaint with multiple groups of swollen nodes is unlikely to be related to cat-scratch disease.

In underdeveloped countries, the peak incidence of Hodgkin disease is in children under 10 years of age; however, in developed countries, the peak incidence occurs in late adolescence and young adulthood. Systemic symptoms of Hodgkin disease include fever, night sweats, malaise, weight loss, and pruritus. In the Ann Arbor staging system, however, only fever, night

sweats, and weight loss are considered significant systemic symptoms that have prognostic importance. In most instances, the initial mode of spread of Hodgkin disease is predictable involvement of contiguous lymphoid tissue. The objective of surgical staging is to determine whether there is occult intraabdominal disease in patients who clinically have only apparent supra-diaphragmatic involvement. The information provided by staging laparotomy is important if radiation therapy is the only modality of treatment contemplated. In patients who have obvious intraabdominal disease by noninvasive studies (such as CT scan or lymphangiogram) or obvious metastases outside the lymphatic system (e.g., bone marrow), combination chemotherapy with or without radiation therapy is generally recommended, and staging laparotomy is then not required.

**411. The answer is c.** (*Behrman, 16/e, pp 1467–1468. McMillan, 3/e, pp 1448–1449. Rudolph, 21/e, pp 1529–1530.*) The finding of hypersegmented neutrophils in the peripheral blood is one of the most useful laboratory aids in making an early diagnosis of folate deficiency. Serum folate levels become low in weeks with an inadequate dietary source. The Pelger-Huët anomaly is an inherited disorder in which neutrophils have no more than two lobes. Neutrophils in severe bacterial infections have toxic granulation, Döhle inclusion bodies, and cytoplasmic vacuoles.

**412–415. The answers are 412-c, 413-d, 414-a, 415-b.** (*Behrman, 16/e, pp 1475–1477, 1479–1487, 1526–1527. McMillan, 3/e, pp 1451–1454, 1466, 1468, 2091. Rudolph, 21/e, pp 1531–1534, 1536–1540, 1561.*) Howell-Jolly bodies (slide C) are small, spherical nuclear remnants seen in the reticulocytes and, rarely, erythrocytes of persons who have no spleen (due to congenital asplenia or splenectomy) or who have a poorly functioning spleen (e.g., in hyposplenism associated with sickle cell disease). Ultrafiltration of blood is a unique function of the spleen that cannot be assumed by other reticuloendothelial organs.

A target cell is an erythrocyte with a membrane that is too large for its hemoglobin content; a thin rim of hemoglobin at the cell's periphery and a small disk in the center give the cell a target-like appearance. Target cells, which are more resistant to osmotic fragility than are other erythrocytes, are seen in children who have  $\beta$ -thalassemia, hemoglobin C disease, or liver disease (e.g., obstructive jaundice or cirrhosis). Thalassemia major (slide D) presents in the second 6 months of life with severe anemia requir-

ing transfusion, heart failure, hepatosplenomegaly, and weakness. Later, the typical facial deformities (maxillary hyperplasia and malocclusion) can be seen in a patient inadequately transfused. The diagnosis can be made on peripheral blood smear by the presence of poorly hemoglobinized normoblasts in addition to target cells in the peripheral blood.

Uniformly small microspherocyte (less than  $6\ \mu$  in diameter) are typical of hereditary spherocytosis (slide A). Because of a decreased surface-to-volume ratio, these osmotically fragile red blood cells have an increased density of hemoglobin. Although spherical red blood cells also can appear in other hemolytic states (e.g., immune hemolytic anemia, microangiopathy, ABO incompatibility, and hypersplenism), their cellular volume is only irregularly augmented. Patients with hereditary spherocytosis can present in the newborn period with anemia, hyperbilirubinemia, and reticulocytosis. They may remain asymptomatic until adulthood, when they develop symptoms. After infancy, hepatosplenomegaly and gallstones are common.

Although hemoglobin C disease (slide B) is frequently a mild disorder, target cells constitute a far greater percentage of total red blood cells than in thalassemia major. In the heterozygous state, no anemia or disease is noted, but target cells are seen. In the homozygous state, a moderately severe hemolytic anemia, reticulocytosis, and splenomegaly are seen, along with a smear containing a large number of target cells.



# Endocrine, Metabolic, and Genetic Disorders

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## Questions

**DIRECTIONS:** Each item below contains a question or incomplete statement followed by suggested responses. Select the **one best** response to each question.

**416.** A 6-month-old infant has been exclusively fed a commercially available infant formula. Upon introduction of fruit juices, however, the child develops jaundice, hepatomegaly, vomiting, lethargy, irritability, and seizures. Tests for urine reducing substances are positive. Which of the following is likely to explain this child's condition?

- a. Tyrosinosis
- b. Galactosemia
- c. Fructosemia
- d.  $\alpha_1$  antitrypsin
- e. Glucose-6-phosphatase deficiency

**417.** A 12-year-old healthy Jewish girl is found on routine blood count to have a mild anemia, leukopenia, and thrombocytopenia. Physical examination reveals an enlarged spleen. An x-ray of the femur is described as appearing to be an Erlenmeyer flask. Bone marrow examination shows abnormal cells. The diagnosis can be confirmed by measurement of activity of which of the following?

- a. Sphingomyelinase activity
- b. Hexosamidase A
- c. Sulfatase A
- d. Glucocerebrosidase
- e. Ceramide trihexosidase

**418.** An infant is born to a woman who has received very little prenatal care. The mother is anxious, complains of heat intolerance and fatigue, and reports that she has not gained much weight despite having an increased appetite. On examination the mom is tachycardic, has a tremor, and has fullness in her neck and in her eyes. The infant is most likely at risk for development of

- a. Constipation
- b. Heart failure
- c. Macrocephaly
- d. Third-degree heart block
- e. Thrombocytosis

**419.** An otherwise healthy 7-year-old girl is brought to your office by her father because she has some acne, breast development, and fine pubic hair. The most likely etiology for her condition is

- a. A feminizing ovarian tumor
- b. A gonadotropin-producing tumor
- c. A lesion of the central nervous system
- d. Exogenous estrogens
- e. Early onset of “normal” puberty (constitutional)

### **Items 420–421**

**420.** The parents of a 14-year-old boy are concerned about his short stature and lack of sexual development. By history, you learn that his birth weight and length were 3 kg and 50 cm, respectively, and that he had a normal growth pattern, although he was always shorter than children his age. The physical examination is normal. His upper-to-lower segment ratio is 0.98. A small amount of fine axillary and pubic hair is present. There is no scrotal pigmentation; his testes measure 4.0 cm<sup>3</sup> and his penis is 6 cm in length. In this situation you should

- a. Measure pituitary gonadotropin
- b. Obtain a CT scan of the pituitary area
- c. Biopsy his testes
- d. Measure serum testosterone levels
- e. Reassure the parents that the boy is normal

**421.** Which of the following is the most likely diagnosis for the patient in the previous question?

- a. Hypopituitarism
- b. Klinefelter syndrome
- c. Hypothyroidism
- d. Constitutionally short stature with delayed puberty
- e. Male Turner syndrome

**Items 422–423**

**422.** A 13-year-old asymptomatic girl is shown. She states that the findings demonstrated began more than a year ago. The most likely diagnosis is



*(Courtesy Adelaide Hebert, M.D.)*

- a. Iodine deficiency
- b. Congenital hypothyroidism
- c. Graves disease
- d. Exogenous ingestion of synthroid
- e. Lymphocytic (Hashimoto) thyroiditis

**423.** Treatment for the patient in the previous question includes

- a. Iodine
- b. Synthroid if she becomes symptomatic
- c. PTU (propylthiouracil)
- d. Psychiatry consult
- e. Surgical removal of thyroid

**Items 424–425**

**424.** A 10-year-old obese boy has central fat distribution, arrested growth, hypertension, plethora, purple striae, and osteoporosis. Which of the following disorders is most likely to be responsible for the clinical picture that this boy presents?

- a. Bilateral adrenal hyperplasia
- b. Adrenal adenoma
- c. Adrenal carcinoma
- d. Craniopharyngioma
- e. Ectopic adrenocorticotropin-producing tumor

**425.** Appropriate initial management of this young man is

- a. Measurement of evening cortisol levels
- b. MRI of the adrenals
- c. Bilateral inferior petrosal blood sampling
- d. MRI of the brain and pituitary
- e. Adrenal scintigraphy with radiocholesterol

**426.** A 3-month-old child has been diagnosed by sweat chloride testing to have cystic fibrosis. The appropriate next step is

- a. Have all siblings assessed for cystic fibrosis with sweat chloride testing
- b. Determine parental carrier state with DNA analysis for carrier state
- c. Determine siblings' carrier state with DNA analysis
- d. Determine the couple's carrier state by sweat chloride testing
- e. Determine siblings' carrier state with sweat chloride testing

**427.** The 2-week-old female pictured is noted to have a thin membrane adhering together the upper portion of the labia minora. The most appropriate course of action for this condition is to



*(Courtesy Michael L. Ritchey, M.D.)*

- a. Apply estrogen cream daily
- b. Refer for surgical repair
- c. Apply traction to the opposing labia until the membrane breaks
- d. Evaluate the patient for congenital adrenal hyperplasia
- e. Do nothing, as the lesions are of no consequence

**428.** An otherwise healthy 7-year-old child is brought to you to be evaluated because he is the shortest child in his class. Careful measurements of his upper and lower body segments demonstrate normal body proportions for his age. Which of the following disorders of growth is likely?

- a. Achondroplasia
- b. Morquio disease
- c. Hypothyroidism
- d. Growth hormone deficiency
- e. Marfan syndrome

**429.** A 9-year-old child has aplastic anemia unresponsive to conventional therapy. A bone marrow transplant has been recommended. As the family begins their quest for the best possible human leukocyte antigen (HLA) match, which of the following statements is true?

- a. There is a 1 in 2 chance that two siblings are HLA-matched
- b. Each parent is diploidentical to the child
- c. Identical twins are not necessarily HLA-matched
- d. Unrelated donors can occasionally be matched
- e. It is always better to use a family member as a donor

**430.** A 13-year-old boy is below the third percentile for height (fiftieth percentile for age 9). Which of the following would give him the best prognosis for normal adult height?

- a. A bone age of 9 years
- b. A bone age of 13 years
- c. A bone age of 15 years
- d. Being at the fiftieth percentile for weight
- e. Being at the third percentile for weight

**431.** A 12-year-old girl has a mass in her neck. Physical examination reveals a thyroid nodule, but the rest of the gland is not palpable. A technetium scan reveals a “cold” nodule. The child appears to be euthyroid. Which of the following diagnoses is the *least* likely?

- a. Simple adenoma
- b. Follicular carcinoma
- c. Papillary carcinoma
- d. Cyst
- e. Dysgenetic thyroid gland

432. The child pictured has



- a. Achondrogenesis
- b. Achondroplasia
- c. Metatrophic dwarfism
- d. Thanatophoric dwarfism
- e. Chondroectodermal dysplasia

**433.** The 16-month-old male infant pictured was recently brought from a developing country to the United States. The family history reveals that his father had an eye and a leg removed. The most likely diagnosis is



*(Courtesy Kathryn H. Musgrove, M.D.)*

- a. Coloboma of the choroid
- b. Retinal detachment
- c. Nematode endophthalmitis
- d. Retinoblastoma
- e. Persistent hyperplastic primary vitreous

**434.** Bone age will be advanced in short stature caused by which of the following?

- a. Environmental deprivation syndrome
- b. Hypopituitarism
- c. Hypothyroidism
- d. Congenital adrenal hyperplasia
- e. Chronic administration of glucocorticoids in high doses

**435.** A 4-year-old child has mental retardation, shortness of stature, brachydactyly (especially of the fourth and fifth digits), and obesity with round facies and short neck. The child is followed by an ophthalmologist for subcapsular cataracts, and has previously been noted to have cutaneous, subcutaneous, and perivascular calcifications of the basal ganglia. This patient is likely to have which of the following features?

- a. Hypercalcemia
- b. Hypophosphatemia
- c. Elevated concentrations of parathyroid hormone
- d. Advanced height age
- e. Decreased bone density, particularly in the skull

**436.** A 15-year-old boy has been immobilized in a double hip spica for 6 weeks after having fractured his femur in a skiing accident. He has become depressed and listless during the past few days and has complained of nausea and constipation. He is found to have microscopic hematuria and a blood pressure of 150/100 mmHg. You should

- a. Request a psychiatric evaluation
- b. Check blood pressure every 2 h for 2 days
- c. Collect urine for measurement of the calcium-creatinine ratio
- d. Order a renal sonogram and intravenous pyelogram (IVP)
- e. Measure 24-h urinary protein

**437.** An adolescent with type I diabetes has a dramatically elevated glycosylated hemoglobin (hemoglobin A<sub>1C</sub>) level, indicating poor control of his diabetes over at least the preceding

- a. 8 h
- b. 1 week
- c. 1 month
- d. 2 months
- e. 6 months

**Items 438–439**

**438.** A 7-day-old boy is admitted to a hospital for evaluation of vomiting and dehydration. Physical examination is otherwise normal except for minimal hyperpigmentation of the nipples. Serum sodium and potassium concentrations are 120 meq/L and 9 meq/L, respectively; serum glucose is 120 mg/dL. The most likely diagnosis is

- a. Pyloric stenosis
- b. Congenital adrenal hyperplasia
- c. Secondary hypothyroidism
- d. Panhypopituitarism
- e. Hyperaldosteronism

**439.** The diagnosis can be confirmed in this patient by

- a. Obtaining a barium swallow
- b. Measurement of 17-hydroxyprogesterone
- c. Measurement of T3, T4, and TSH
- d. Somatomedin C measurement
- e. Measurement of serum renin levels

**440.** An infant is brought to a hospital because her wet diapers turn black when they are exposed to air. Physical examination is normal. Urine is positive both for reducing substance and when tested with ferric chloride. This disorder is caused by a deficiency of

- a. Homogentisic acid oxidase
- b. Phenylalanine hydroxylase
- c. l-histidine ammonia-lyase
- d. Ketoacid decarboxylase
- e. Isovaleryl-CoA dehydrogenase

**441.** An 18-year-old girl has hepatosplenomegaly, an intention tremor, dysarthria, dystonia, and deterioration in her school performance. She also developed abnormal urine with excess glucose, protein, and uric acid. She has a several-year history of elevated liver enzymes of unknown etiology. Which of the following best explains her condition?

- a. Indian childhood cirrhosis
- b.  $\alpha_1$ -antitrypsin deficiency
- c. Menkes syndrome
- d. Dubin-Johnson syndrome
- e. Wilson disease

**442.** A 2-week-old girl is noted to have bilateral inguinal hernias, but otherwise appears to be normal. Which of the following conditions is most likely to be present?

- a. Congenital adrenal hyperplasia (adrenogenital syndrome)
- b. Cushing syndrome
- c. Androgen-producing ovarian tumor
- d. Testicular feminization
- e. Administration of exogenous androgens

**443.** A very upset mother brings her 8-month-old child to the emergency room because he will not move his leg. She reports that she was carrying him to the car about  $\frac{1}{2}$  hour prior, slipped on some ice, and fell on top of him. The mother, an 18-year-old African American woman, has been exclusively breast-feeding her child. She has only just recently started him on cereals, and has not supplemented his diet with vitamins. Which of the following laboratory findings would be expected?



*(Courtesy Susan John, M.D.)*

- a. Hypocalcemia
- b. Hypophosphaturia
- c. Reduced serum alkaline phosphatase
- d. Hypocalciuria
- e. Hyperphosphatemia

**444.** An abnormally tall 11-year-old with normal mentation is most likely to have which of the following syndromes?

- a. Cerebral gigantism (Sotos syndrome)
- b. Homocystinuria
- c. XXY (Klinefelter syndrome)
- d. Marfan syndrome
- e. XYY

**445.** A small-for-gestation-age infant is born at 30 weeks' gestation. At 1 h of age, his serum glucose is noted to be 20 mg/dL (normally greater than 40 mg/dL). The most likely explanation for hypoglycemia in this infant is

- a. Inadequate stores of nutrients
- b. Adrenal immaturity
- c. Pituitary immaturity
- d. Insulin excess
- e. Glucagon deficiency

**446.** A 1-day-old infant develops tetany and convulsions. He was born at 34 weeks' gestation with Apgar scores of 2 and 4 (at 1 and 5 min, respectively) to a woman whose pregnancy was complicated by diabetes mellitus and pregnancy-induced hypertension. Which of the following serum chemistry values is likely to be the explanation for his condition?

- a. Serum bicarbonate level of 22 meq/dL
- b. Serum calcium of 6.2 mg/dL
- c. Serum glucose of 45 mg/dL
- d. Serum magnesium level of 5.0 mg/dL
- e. Intracranial hemorrhage

**DIRECTIONS:** Each group of questions below consists of lettered options followed by a set of numbered items. For each numbered item, select the appropriate lettered option(s). Each lettered option may be used once, more than once, or not at all. **Choose exactly the number of options indicated following each item.**

**Items 447–451**

For each of the disorders listed below, select the serum concentration of calcium (Ca) and phosphate ( $\text{PO}_4$ ) with which it is most likely to be associated.

- a. Low  $\text{PO}_4$ , normal Ca
- b. Low  $\text{PO}_4$ , high Ca
- c. Normal  $\text{PO}_4$ , low Ca
- d. Normal  $\text{PO}_4$ , normal Ca
- e. High  $\text{PO}_4$ , low Ca

**447.** Three boys in a family with hypodense calcification of their bones and a history of fractures; their condition has been unresponsive to dietary changes or vitamin supplementation (**SELECT 1 FINDING**)

**448.** A short, 4-year-old mentally retarded child with brachydactyly of the fourth and fifth digits; obesity with round facies; short neck; subcapsular cataracts; and cutaneous, subcutaneous, and perivascular calcifications of the basal ganglia (**SELECT 1 FINDING**)

**449.** A 4-year-old child with blue sclera and a history of multiple fractures with minimal trauma (**SELECT 1 FINDING**)

**450.** A 6-year-old child who complains of numbness and tingling of the hands, and later develops tonic clinic seizures (**SELECT 1 FINDING**)

**451.** An otherwise healthy 18-year-old girl with medullary thyroid carcinoma (**SELECT 1 FINDING**)

**Items 452–457**

All the syndromes listed below are associated with obesity in children. For each of the other clinical findings that follow, select the syndrome with which it is most likely to be associated.

- a. Prader-Willi syndrome
- b. Laurence-Moon-Biedl syndrome
- c. Cushing syndrome
- d. Fröhlich syndrome
- e. Pseudohypoparathyroidism

**452.** A 3-year-old mentally retarded girl followed by ophthalmology for cataracts (**SELECT 1 SYNDROME**)

**453.** A 6-day-old infant with severe hypotonia and poor feeding since birth (**SELECT 1 SYNDROME**)

**454.** An obese 5-year-old boy with mental retardation and polydactyly (**SELECT 1 SYNDROME**)

**455.** A 5-day-old girl with brachydactyly, round facies, and short neck (**SELECT 1 SYNDROME**)

**456.** A short 11-year-old girl in special education classes who has basal ganglia calcifications (**SELECT 1 SYNDROME**)

**457.** A 14-year-old boy with hypogonadism and night blindness with retinitis pigmentosa (**SELECT 1 SYNDROME**)

**Items 458–463**

For each of the following disorders, select the serum concentrations (meq/L) of sodium ( $\text{Na}^+$ ) and potassium ( $\text{K}^+$ ) with which it is most likely to be associated in a dehydrated patient.

- a.  $\text{Na}^+$  118,  $\text{K}^+$  7.5
- b.  $\text{Na}^+$  125,  $\text{K}^+$  3.0
- c.  $\text{Na}^+$  134,  $\text{K}^+$  6.0
- d.  $\text{Na}^+$  144,  $\text{K}^+$  2.9
- e.  $\text{Na}^+$  155,  $\text{K}^+$  5.5

- 458.** A 4-month-old boy with salt-losing 21-hydroxylase deficiency (adrenogenital syndrome) (**SELECT 1 FINDING**)
- 459.** An 11-year-old boy with central diabetes insipidus secondary to an automobile accident (**SELECT 1 FINDING**)
- 460.** A 2-year-old girl with nephrogenic diabetes insipidus (**SELECT 1 FINDING**)
- 461.** A 4-year-old boy with hyperaldosteronism (**SELECT 1 FINDING**)
- 462.** An 8-year-old boy with Addison disease (in crisis) (**SELECT 1 FINDING**)
- 463.** A 1-year-old girl with glucose-6-phosphatase deficiency (von Gierke disease) (**SELECT 1 FINDING**)

**Items 464–468**

For each diagnosis that follows, select the mode of inheritance with which it is usually associated.

- a. Autosomal dominant
- b. Autosomal recessive
- c. X-linked dominant
- d. X-linked recessive
- e. None of the above

- 464.** A 1-year-old African American boy with poor calcification of his bones, unresponsive to vitamin supplementation (**SELECT 1 MODE**)
- 465.** An 8-year-old white girl with a history of renal failure as an infant, who now has hepatic fibrosis and is developing portal hypertension (**SELECT 1 MODE**)
- 466.** An 11-year-old African American girl with a butterfly rash on her face and protein in her urine (**SELECT 1 MODE**)

**467.** An 18-month-old white child, normal until about 18 months of age, has now developed Fanconi syndrome, renal failure, failure to thrive, and photophobia due to corneal crystals found on ophthalmologic examination (**SELECT 1 MODE**)

**468.** A 4-year-old Hispanic boy who appears normal but whose father has recently been noted to have hypertension, chronic renal failure, and large cysts within his kidneys (**SELECT 1 MODE**)

**Items 469–470**

New discoveries made possible by advances in molecular genetics have broadened our understanding of nontraditional inheritance. Match the disease with the appropriate genetic mechanism listed below.

- a. Mitochondrial inheritance
- b. Mosaicism
- c. Genomic imprinting
- d. Sex chromosome imbalance

**469.** A 3-year-old boy with myoclonus, ataxia, weakness, and seizures who has cytochrome oxidase negative ragged red fibers noted on muscle biopsy (**SELECT 1 MECHANISM**)

**470.** A mentally retarded 4-year-old boy, who was noted to be hypotonic at birth and had failure to thrive in infancy, now has a tremendous appetite, obesity, hypogonadism, and small hands and feet (**SELECT 1 MECHANISM**)

**Items 471–472**

Select the first sign of pubertal changes in boys and girls.

- a. Enlargement of the testes
- b. Development of pubic hair
- c. Acne
- d. Development of breast buds
- e. Penile enlargement
- f. Ovulation
- g. Thickening of the vaginal mucosa
- h. Growth spurt

**471.** The first sign of pubertal development in a 13-year-old boy (**SELECT 1 SIGN**)

**472.** The first sign of pubertal development in an 11-year-old girl (**SELECT 1 SIGN**)

**Items 473–474**

Match each patient with the appropriate findings.

- a. Tracheoesophageal fistula
- b. Endocardial cushion defect
- c. Short stature
- d. Lactose intolerance
- e. Increased incidence in younger mothers
- f. History of duodenal atresia at birth
- g. Heterochromia
- h. Panhypopituitarism
- i. Streak gonads
- j. Renal failure
- k. Increased incidence of cystic fibrosis
- l. Coarctation of the aorta

**473.** A child with Down syndrome (**SELECT 3 FINDINGS**)

**474.** A child with Turner syndrome (**SELECT 3 FINDINGS**)

# Endocrine, Metabolic, and Genetic Disorders

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## Answers

**416. The answer is c.** (Behrman, 16/e, pp 344–348, 407, 413–414. McMillan, 3/e, pp 1747–1750, 1830–1831, 1859. Rudolph, 21/e, pp 612, 1487–1488, 1490–1491, 1542–1543, 1996–1997.) Galactosemia, fructosemia, tyrosinosis, and glucose-6-phosphatase deficiency represent diseases in which a congenital deficiency of enzyme causes an interruption of a normal metabolic pathway and an accumulation of metabolic precursors that damage vital organs. Galactosemia (found in milk) and fructosemia (found in fruit juices) produce urinary reducing substances. The mode of inheritance of galactosemia, fructosemia, and most forms of glucose-6-phosphatase deficiency is autosomal recessive. In galactosemia and fructosemia, errors in carbohydrate metabolism cause the accumulation of toxic metabolites when specific dietary sugars are introduced (lactose in galactosemia; fructose and sucrose in fructosemia). Exclusion of the offending carbohydrate from the diet will prevent liver damage. In tyrosinemia type I, or tyrosinosis, the accumulation of tyrosine and its metabolites is associated with severe involvement of liver, kidney, and central nervous system (CNS). Manifestations of acute liver failure can appear in infancy. A chronic form of the disorder presents as progressive cirrhosis and leads to liver failure or hepatoma. Dietary management does not prevent liver disease. Glucose-6-phosphatase deficiency often presents at 3 to 4 months of age with failure to thrive, hypoglycemia, hepatomegaly, and acidosis.  $\alpha_1$  antitrypsin deficiency causes liver disease through accumulation of an abnormal protein, caused by a single amino acid substitution on chromosome 14. It has a variable presentation, but cholestasis; bleeding into the CNS, GI tract, or at the umbilical stump; and elevation of transaminase concentrations are common in infancy. In childhood, a picture of chronic hepatitis with cirrhosis and portal hypertension is seen.

**417. The answer is d.** (Behrman, 16/e, pp 400–404. McMillan, 3/e, pp 1876–1877. Rudolph, 21/e, pp 2326–2328.) Gaucher disease is characterized by  $\beta$ -glucocerebrosidase deficiency, which causes an abnormal accumula-

tion of glucocerebroside in the reticuloendothelial system. Bone marrow aspirate shows the typical Gaucher cells engorged with glucocerebroside. Replacement of marrow leads to anemia, leukopenia, and thrombocytopenia. The liver and spleen can also be involved. Serum acid phosphatase is elevated. X-ray demonstrates an Erlenmeyer flask appearance of the long bones. The diagnosis of Gaucher disease is confirmed by the absence of glucocerebrosidase activity in leukocytes, in cultured skin fibroblasts, and in liver cells. Prenatal diagnosis by enzyme analysis is now possible. In the most common form of Gaucher disease, adult type I, there is no involvement of the central nervous system. Therefore, MRI of the brain is not indicated. Sphingomyelinase deficiency causes type A Niemann-Pick disease, hexosaminidase A deficiency causes Sandhoff disease, sulfatase A deficiency causes juvenile metachromatic leukodystrophy, and serum trihexosidase causes Fabry disease.

**418. The answer is b.** (*Behrman, 16/e, pp 1711–1712, McMillan, 3/e, pp 355, 1809. Rudolph, 21/e, p 2076.*) The infant is likely at risk for neonatal thyrotoxicosis. Neonatal thyrotoxicosis usually disappears within 2 to 4 months as the concentration of thyroid-stimulating immunoglobulins (7S gamma globulin) diminishes. Unlike TSI, thyroid-stimulating hormone (TSH) does not cross the placenta. All forms of thyrotoxicosis are more common in females, with the exception of neonatal thyrotoxicosis, which has an equal sex distribution. Symptoms include tachycardia and tachypnea, irritability and hyperactivity, low birth weight with microcephaly, severe vomiting and diarrhea, thrombocytopenia, jaundice, hepatosplenomegaly, and heart failure. In severely affected infants, the disease could be fatal if not treated vigorously and promptly. Third-degree heart block is not a feature of this disease, but is sometimes seen in infants born to mothers with systemic lupus erythematosus.

**419. The answer is c.** (*Behrman, 16/e, pp 1687–1690. McMillan, 3/e, pp 1772–1774. Rudolph, 21/e, pp 2097–2098.*) The patient in the question appears to have “true sexual precocity,” implying that the gonads have matured in response to the secretion of pituitary gonadotropins and have begun secreting sex steroids, causing the development of secondary sexual characteristics. Thus, ovarian tumors and exogenous estrogens, which suppress the function of the pituitary gland, do not cause true precocious puberty. In girls, the most common form of true precocious puberty is

idiopathic and is thought to be caused by early maturation of an otherwise normal hypothalamic-pituitary-gonadal feedback system. In boys, true precocious puberty is relatively rare and is more likely to be caused by lesions of the central nervous system. Gonadotropin-producing tumors, which are very rare, can cause true precocious puberty in both sexes.

**420. The answer is e.** (*Behrman, 16/e, p 1679. McMillan, 3/e, pp 1775–1780. Rudolph, 21/e, p 2103.*) A record of the sequential pattern of growth in height is very helpful in the differential diagnosis of a child with short stature. A child with constitutionally short stature and delayed puberty will have a consistent rate of growth below but parallel to the average for his or her age, whereas patients with organic disease do not follow a given percentile but progressively deviate from their prior growth percentile. A knowledge of the patterns of growth and sexual maturation of family members is helpful because such patterns are often familial. Puberty is said to be delayed in males if physical changes are not apparent by 14 years of age. Identification of the earliest signs of sexual maturation by means of careful physical examination avoids unnecessary workup. In this case, measurement of pituitary gonadotropins is unnecessary because the child already shows evidence of pubertal development (a testicular length of more than 2.5 cm, volume 3.0 cm<sup>3</sup>). The single most useful laboratory test is the determination of bone age. In those of constitutionally short stature with delayed pubertal maturation, the bone age is equal to the height age, both of which are behind chronologic age. In familial short stature, bone age is greater than height age and equal to chronologic age. In a child at any age, the administration of human chorionic gonadotropin (hCG) will stimulate interstitial cells of testes to produce testosterone, thereby serving as a method of assessing testicular function. The finding of testicular enlargement is evidence of pituitary secretion of gonadotropins and of testicular responsiveness and obviates the need for administration of hCG. Elevated serum gonadotropins are found in children 12 years of age or older who have primary hypogonadism (Klinefelter syndrome, bilateral gonadal failure from trauma or infection). Because the secretion of gonadotropins is not constant but occurs in spurts, children with constitutional delay of puberty may have normal or low levels of gonadotropins.

**421. The answer is d.** (*Behrman, 16/e, pp 1675–1680, 1703–1704, 1747–1748. McMillan, 3/e, pp 1775, 1779, 1808–1809, 2231, 2243. Rudolph, 21/e, pp 2021, 2089–2090, 2103–2105.*) In Klinefelter syndrome, the testes

are smaller than normal for age and feel firm and fibrotic. Physical examination often reveals a eunuchoid body habitus and reduced upper-to-lower body segment ratio secondary to a long lower segment. Diagnosis is established by means of buccal smear and karyotyping. Levels of luteinizing hormone are elevated after 12 years of age. Hypothyroidism and hypopituitarism are associated with a deviation from the previously established growth pattern in addition to delayed bone age and other abnormal findings on physical examination. In the so-called male Turner syndrome, many of the features of classic Turner syndrome, which occurs in females, are present. In addition, the testes are hypoplastic and are often undescended. A consistent growth pattern and normal upper-to-lower body segment ratio support the diagnosis of constitutionally short stature. Bone age consistent with height age rather than chronologic age is also a supportive finding. Reassurance that normal sexual development will occur and that a normal adult height (usually a midparental height) will be attained is frequently the only therapy indicated.

**422–423. The answers are 422-e, 423-b.** (*Behrman, 16/e, pp 1704–1705. McMillan, 3/e, pp 1808–1809. Rudolph, 21/e, pp 2070–2071.*) The photograph shows thyromegaly. Lymphocytic thyroiditis is a typical organ-specific autoimmune disease characterized by lymphocytic infiltration of the thyroid gland, with or without goiter. It is the most common cause of juvenile hypothyroidism, peaking in adolescence, and affecting as many as 1% of schoolchildren. The condition is 4 to 7 times more prevalent in girls than in boys and may persist for many years without symptoms. Patients are initially euthyroid, but, with the eventual atrophy of the gland, they become hypothyroid. Spontaneous remission can occur in one-third of the affected adolescents. Hashimoto thyroiditis is not related to endemic goiter caused by iodine deficiency.

Autoimmune thyroiditis is associated with many other autoimmune disorders; its association with Addison disease and insulin-dependent diabetes mellitus is called type II polyglandular autoimmune disease (Schmidt syndrome). Family clusters of autoimmune thyroiditis are common; nearly 50% of the patients have siblings with antithyroid antibodies.

In Hashimoto thyroiditis, thyroid function tests are often normal, although an elevated TSH level may be seen in a euthyroid child. With progressive thyroid failure,  $T_3$  and  $T_4$  levels drop and the TSH level rises. Most patients have titers of thyroid antimicrosomal antibodies; elevated antithyroglobulin titers occur infrequently. Blocking TSH antibodies are thought

to be related to development of hypothyroidism. Congenital rubella infection can cause late-onset thyroiditis and hypothyroidism on an autoimmune basis. Antirubella antibodies cannot be used for diagnostic purposes in this 13-year-old child because she has probably been immunized with the attenuated vaccine or may have had rubella in childhood.

**424–425. The answers are 424-a, 425-a.** (*Behrman, 16/e, pp 1737–1739. McMillan, 3/e, pp 1819–1820. Rudolph, 21/e, pp 2045–2049.*) Although the administration of exogenous adrenocorticotropic hormone or of glucocorticoids is the most common cause of Cushing syndrome, it can also be caused by bilateral adrenal hyperplasia. In the latter case, the concentration of adrenocorticotropic hormone can be normal or high. The basic abnormality, however, is thought to be in the hypothalamic-pituitary axis, not the adrenal gland, because a distinct pituitary adenoma is found in some patients. Furthermore, many patients who have undergone bilateral adrenalectomy develop Nelson syndrome (invasive pituitary adenoma) despite receiving adequate cortisol replacement. If the patient were an infant, however, the most likely answer would have been an adrenal carcinoma.

The initial management of this child would be to measure evening cortisol levels which, in a normal child, are expected to be less than 50% of the 8:00 A.M. value. Additionally, 24-h excretion of 17-hydroxycorticosteroids is almost always increased. In difficult-to-diagnose cases, a dexamethasone suppression test can be required. After the diagnosis has been established, further testing is indicated to determine if the condition is ACTH-dependent or ACTH-independent. At that point, corticotropin-releasing hormone testing and imaging will be required.

**426. The answer is c.** (*Behrman, 16/e, pp 1315–1327. McMillan, 3/e, pp 1242–1254. Rudolph, 21/e, pp 1967–1979.*) Cystic fibrosis (CF) has an autosomal recessive pattern of inheritance; both parents are usually heterozygous, and the affected child may be a homozygote (two copies of the same mutation) or a compound heterozygote (one copy of each of two different mutations). The  $\Delta F508$  mutation, present in about 70% of North American whites, is the most common. In Europe, the incidence of this mutation ranges from 40 to 80% in different population groups. There are many other mutations that vary in specific ethnic groups.

Identification of a CF mutation in both chromosomes, whether homozygous or compound heterozygous, would be diagnostic in 72%. DNA testing

can help predict manifestations of the disease, especially pancreatic involvement. Pancreatic insufficiency is present in 99% of patients who are homozygous for  $\Delta F508$ , in 72% who are compound homozygous for  $\Delta F508$ , and in only 32% who do not carry the  $\Delta F508$  mutation. DNA analysis for carrier testing is now recommended for relatives of CF patients and for reproductive partners of carriers. Its role in mass population screening is controversial. The sweat test is diagnostic in virtually all cases of classic CF and is equally useful in all ethnic groups, but it is not useful in detecting heterozygotes. DNA testing is of value for postmortem investigations as well as in sick premature infants and other patients when sweat collection is unsuccessful.

**427. The answer is a.** (*Behrman, 16/e, pp 1660–1661.*) Labial adhesions are a common condition and are diagnosed when a central line of adherence is noted from the area inferior to the clitoris to the fourchette. It is an asymptomatic condition seen in girls less than 6 years of age. Labial adhesions can be responsible for vulvovaginitis and increased urinary tract infections in girls due to pooling of urine in the vagina. Treatment with topical estrogen cream daily results in resolution of this problem. Mechanical separation is not advisable under normal circumstances.

**428. The answer is d.** (*Behrman, 16/e, pp 421–422, 1675–1678, 1698–1704, 2121–2123, 2131–2132. McMillan, 3/e, pp 1782–1783, 1790–1791, 1808–1809, 1873, 1893–1895, 2142. Rudolph, 21/e, pp 660, 759–760, 762–764, 2017–2021.*) Alteration of body proportion results from selective regional rates of growth at different stages during the developmental period. At birth, the head is large for the body size, the limbs are short, and the upper-to-lower segment ratio (crown to pubis/pubis to heel) of 1.7 is high. As the growth of the limbs exceeds that of the trunk from infancy to adolescence, there is a change in body proportions reflected in the upper-to-lower segment ratios: 1.3 at 3 years, 1.1 at 6 years, and 1.0 at 10 years of age. In achondroplasia, there is a disproportion between the limbs and the trunk; that is, the limbs are relatively short. The head in this condition is also disproportionately large. Achondroplasia is the most common genetic skeletal dysplasia. This disorder has an autosomal dominant mode of inheritance. Marfan syndrome is a serious disease of connective tissue that is inherited in the autosomal dominant mode. The predominant findings in this condition are bilateral subluxation of the lens, dilatation of the aortic root, and disproportionately long limbs in comparison with the

trunk. The decreased upper-to-lower segment ratio in Marfan syndrome reflects this relative increase in the length of the legs as compared with the trunk. Morquio syndrome is one of the mucopolysaccharidoses. Abnormal amounts of keratan sulfate accumulate as a result of an enzyme deficiency, and widespread storage of this material in the body results in problems in morphogenesis and function. Skeletal malformations are similar to those seen in osteochondrodysplasias, namely, short trunk with short stature, marked slowing of growth, severe scoliosis, pectus carinatum, and short neck. Thyroid hormone is necessary for physical growth and development and, along with sex hormones, has an essential role in development of bone and linear growth. Thyroid deficiency results in delayed puberty in most cases and in stunting of growth with persistence of immature body proportions; signs and symptoms of hypothyroidism are also frequently seen. In growth hormone deficiency, the upper-to-lower segment ratio is normal, often without other signs or symptoms.

**429. The answer is d.** (*Behrman, 16/e, pp 634–644. McMillan, 3/e, pp 1505–1507. Rudolph, 21/e, pp 1589–1593.*) In bone marrow transplantation, human leukocyte antigen (HLA) compatibility is important in preventing graft rejection and graft-versus-host disease (GVHD). The marrow donor can be the patient (autologous transplant); an identical twin in which, by definition, HLA is identical (syngeneic transplant); or a histocompatible donor, usually a sibling (allogeneic transplant). The HLA complex, a series of genetic loci on the short arm of chromosome 6, is usually inherited as a group or haplotype in a codominant Mendelian fashion, one from each parent. Statistically, then, 25% of siblings will inherit the same haplotype from each parent and, therefore, be HLA-matched and histocompatible. If the donor and recipient share only one haplotype, they are termed *haploidentical*, a situation that pertains to a parent-child pair, for example. Identification of potential donors has been facilitated by the International Bone Marrow Transplant Registry, a computerized list of the names and HLA types of tens of thousands of persons. In a highly HLA-disparate family, it can be preferable to look for a closely matched unrelated donor.

**430. The answer is a.** (*Behrman, 16/e, pp 56, 59–60. McMillan, 3/e, pp 1776–1780, 1790–1791. Rudolph, 21/e, p 2015–2017, 2094.*) The determination of bone age by the radiographic examination of ossification centers provides a measure of a child's level of growth that is independent of his or

her chronologic age. Height age is the age that corresponds to the fiftieth percentile for a child's height. When bone age and height age are equally retarded several years behind chronologic age, a child is described as having constitutional short stature. Such a child is usually shorter than peers in adolescence because of the delayed growth spurt, but the prognosis for normal adult height is excellent because there is still the potential for growth. Detailed questioning will usually identify other family members with a history of delayed growth and sexual maturation but with ultimately normal stature. Children with genetic or familial short stature grow at an adequate rate, but remain small throughout life; their ultimate height is consistent with predictions based on parental heights. Bone age is within the limits of normal for chronologic age, and puberty occurs at the normal time. In all cases, a thorough history and physical examination are necessary to identify any other cause of growth delay.

**431. The answer is e.** (*Behrman, 16/e, pp 1698, 1712–1713. McMillan, 3/e, pp 1806, 1811–1812. Rudolph, 21/e, p 2077.*) A dysgenetic thyroid gland can appear as a neck mass; as a rule, however, it is functional and, thus, does not appear as a cold nodule on thyroid scan. A cold thyroid nodule can be a benign or malignant lesion. With the exception of anaplastic carcinomas, most thyroid malignancies are slow-growing. The incidence of thyroid malignancy in children appears to be decreasing, possibly as a result of a decrease in the exposure of children to x-rays.

**432. The answer is b.** (*Behrman, 16/e, pp 2121–2123. McMillan, 3/e, pp 2141–2147. Rudolph, 21/e, pp 759–760.*) Achondroplasia, occurring with an incidence of approximately 1 in every 26,000 live births, is the most common genetic form of skeletal dysplasia. Affected persons bear a striking resemblance to one another and are identified by their extremely short extremities; prominent foreheads; short, stubby fingers; and marked lumbar lordosis. Although they go through normal puberty, affected females must have children by cesarean section because of the pelvic deformity.

**433. The answer is d.** (*Behrman, 16/e, pp 1561–1562. McMillan, 3/e, pp 676–677, 1525–1527. Rudolph, 21/e, pp 2395–2396.*) The child pictured is an asymmetric “red” reflex (one eye is lighter than the other). Although all the listed options can produce the symptoms described, the family history supports the diagnosis of retinoblastoma, the most common intraocular

tumor in children. Early detection can result in a survival rate of over 75%. The pattern of inheritance of retinoblastoma is complicated: the hereditary form of the disease can be transmitted by means of autosomal dominant inheritance from an affected parent, or from an unaffected parent carrying the gene, or from a new germinal mutation. Familial occurrences are usually bilateral. A second primary tumor develops in 15 to 90% of survivors of bilateral retinoblastoma, the most common of which is osteosarcoma increasing in incidence with time. Retinoblastoma is associated with a mutation or deletion of the long arm of chromosome 13. In addition to specialized ophthalmologic care, management of retinoblastoma includes molecular genetic investigation of the family to identify those who have inherited the tumor-predisposing retinoblastoma gene.

**434. The answer is d.** (*Behrman, 16/e, pp 59–60, 1675–1680, 1698–1704, 1729–1736, 1738. McMillan, 3/e, pp 1776–1780. Rudolph, 21/e, pp 7–9, 466, 2017–2021, 2038–2042, 2045.*) The appearance and union of the various centers of ossification follow a fairly definite pattern and time schedule from birth through adolescence. This process provides, through x-ray studies, a valuable criterion for estimating normal and abnormal growth. The skeletal maturity of any person is known as the *bone age*. Bone maturation is particularly influenced by the androgenic and estrogenic hormones. In congenital adrenal hyperplasia, a deficiency of enzyme (21-hydroxylase in 80% of cases) causes an interruption in the pathway for production of cortisol; the end result is hypersecretion of androgenic precursors and clinical manifestations of virilism and protein anabolism. In both males and females, muscles are well developed and there is rapid growth in stature, with marked acceleration of osseous maturation. The result is early closure of epiphyses and failure to achieve full growth. Thyroid hormone appears to act as a primary stimulant to skeletal maturation. Deficiency of thyroxine results in marked retardation of bone age. Failure to thrive as a consequence of profound environmental deprivation is characterized by very low levels of circulating pituitary hormones and is associated with a delay in skeletal maturation. Removal of the child from the abnormal environment results in dramatic catch-up growth and a rapid return of hormone levels to normal. Glucocorticoid excess, either endogenous or exogenous, is usually associated with a decrease in the rate of growth and a delay in the onset of puberty, the exception being a case of glucocorticoid excess resulting in significant virilization and rapid closure of growth plates. The effects

of glucocorticoid excess appear to be mediated at end-organ sites as part of the catabolic effects of these steroids.

**435. The answer is c.** (*Behrman, 16/e, pp 1718–1719. McMillan, 3/e, pp 1620–1621. Rudolph, 21/e, p 2151.*) The patient with the features listed likely has pseudohypoparathyroidism (Albright hereditary osteodystrophy). Such patients have chemical findings of hypoparathyroidism (low calcium, high phosphorus), but parathyroid hormone levels are high, indicating resistance to the action of this hormone. Parathyroid hormone infusion does not produce a phosphaturic response. Phenotypically, these patients demonstrate shortness of stature with delayed bone age, mental retardation, increased bone density throughout the body (especially evident in the skull), brachydactyly (especially of the fourth and fifth digits), obesity with round facies and short neck, subcapsular cataracts, cutaneous and subcutaneous calcifications, and perivascular calcifications of the basal ganglia.

**436. The answer is c.** (*Behrman, 16/e, pp 200–201, 1580–1581. McMillan, 3/e, pp 83–84, 1770–1772. Rudolph, 21/e, pp 2152–2156.*) Hypercalcemia can develop in children who are immobilized following the fracture of a weight-bearing bone. Serious complications of immobilization hypercalcemia, and the hypercalciuria that occurs as a result, include nephropathy, nephrocalcinosis, hypertensive encephalopathy, and convulsions. The early symptoms of hypercalcemia—namely, constipation, anorexia, occasional vomiting, polyuria, and lethargy—are nonspecific and may be ascribed to the effects of the injury and hospitalization. Therefore, careful monitoring of these patients with serial measurements of the serum ionized calcium and the urinary calcium-creatinine ratio is critical during their immobilization. A ratio of greater than 0.2 establishes a diagnosis of hypercalciuria. Although complete mobilization is curative, additional measures such as vigorous intravenous hydration with a balanced salt solution, dietary restrictions of dairy products, and administration of diuretics can be instituted. For patients who are at risk for symptomatic hypercalcemia, short-term therapy with calcitonin is highly effective in reducing the concentration of serum calcium by inhibiting bone resorption.

**437. The answer is d.** (*Behrman, 16/e, p 1782. McMillan, 3/e, p 1797. Rudolph, 21/e, p 2111.*) Glucose is nonenzymatically attached to hemoglo-

bin to form glycosylated hemoglobin. The major component of this reaction proceeds very slowly and is irreversible until the hemoglobin is destroyed. The concentration of glycosylated hemoglobin thus reflects glucose concentration over the half-life of the red cell, or about 2 to 3 months. The adolescent in the question may have had poor control of his diabetes for longer than 2 to 3 months, but a glycosylated hemoglobin is unable to determine this.

**438–439. The answers are 438-b, 439-b.** (*Behrman, 16/e, pp 1729–1736. McMillan, 3/e, pp 311, 1806–1809, 1816–1820. Rudolph, 21/e, pp 1402–1403, 2017–2021, 2038–2042, 2067–2071.*) Salt-losing congenital adrenal hyperplasia (adrenogenital syndrome; 21-hydroxylase deficiency) usually manifests during the first 5 to 15 days of life as anorexia, vomiting, diarrhea, and dehydration. Hypoglycemia can also occur. Affected infants can have increased pigmentation, and female infants show evidence of virilization, that is, ambiguous external genitalia. Hyponatremia, hyperkalemia, and urinary sodium wasting are the usual laboratory findings. Death can occur if the diagnosis is missed and appropriate treatment is not instituted. Although adrenal aplasia, an extremely rare disorder, presents a similar clinical picture, it has an earlier onset than adrenal hyperplasia, and virilization does not occur. In classic 21-hydroxylase deficiency, serum levels of 17-hydroxyprogesterone are markedly elevated beyond 3 days of life (in the first 3 days of life they can normally be high). Blood cortisol levels are usually low in salt-losing forms of the disease.

Pyloric stenosis seems unlikely in this infant in that the vomiting with this disease usually begins after the third week of life. Hypothyroidism would present as a lethargic, poor-feeding infant with delayed reflexes, persistent jaundice, and hypotonia. Hyperaldosteronism would be expected to cause decreased potassium, not increased levels. Panhypopituitarism usually presents with apnea, cyanosis, or severe hypoglycemia.

**440. The answer is a.** (*Behrman, 16/e, p 349. McMillan, 3/e, pp 1825, 1829–1830, 1832, 1841–1843. Rudolph, 21/e, pp 609–613, 616, 623–624.*) The infant described in the question has alkaptonuria, an autosomal recessive disorder caused by a deficiency of homogentisic acid oxidase. The diagnosis is made in infants when their urine turns dark brown or black on exposure to air due to the oxidation of homogentisic acid. Affected children are asymptomatic. In adults, ochronosis—the deposition of a bluish

pigment in cartilage and fibrous tissue—develops; symptoms of arthritis may appear later. No specific treatment is available for patients who have alkaptonuria, although supplemental ascorbic acid may delay the onset of the disorder and reduce clinical symptoms. The other deficiencies listed in the question are found in phenylketonuria, histidinemia, maple syrup urine disease, and isovaleric acidemia, respectively.

**441. The answer is e.** (*Behrman, 16/e, pp 1209–1210, 1852. McMillan, 3/e, pp 1721–1722. Rudolph, 21/e, pp 1491–1492.*) Wilson disease is an autosomal recessive disorder characterized by liver disease (usually seen in childhood), neurologic and behavioral disturbances (seen by adolescence), renal tubular dysfunction (Fanconi syndrome), and eye findings (Kayser-Fleischer rings). Its multisystem manifestations are caused by the deposition of copper in various tissues (resulting in low serum levels), and therapy is aimed at the prevention of accumulation of copper. Defective metabolism of the copper-binding protein ceruloplasmin (usually reduced) has been demonstrated by some. Indian childhood cirrhosis is a nontreatable, fatal condition found in rural India, affecting children aged 1 to 3 years with hepatomegaly, fever, anorexia, and jaundice. It rapidly progresses to cirrhosis and liver failure. Serum immunoglobulin levels and hepatic copper concentrations are elevated.  $\alpha_1$  antitrypsin deficiency causes liver disease through accumulation of an abnormal protein, caused by a single amino acid substitution on chromosome 14. It has a variable presentation, but, in infancy, cholestasis; bleeding into the CNS, GI tract, or at the umbilical stump; and elevation of transaminase concentrations are common. In childhood, a picture of chronic hepatitis with cirrhosis and portal hypertension is seen, but the neurologic and behavior changes typical of Wilson disease are not seen. Menkes syndrome presents in the first months of life to include hypothermia, hypotonia, and myoclonic seizures. These children have chubby, rosy cheeks and kinky, colorless, and friable hair. Severe mental retardation is always seen. Low serum copper and ceruloplasmin levels are found, with a copper absorption/transport problem being the cause. Dubin-Johnson syndrome is inherited as an autosomal recessive condition with patients being unable to excrete conjugated bilirubin. Such patients present during adolescence or early adulthood, sometimes earlier. Morbidity due to this condition is unusual.

**442. The answer is d.** (*Behrman, 16/e, pp 1562–1563, 1666, 1729–1739, 1758–1759, 1761–1765. McMillan, 3/e, pp 351–352, 1533–1535, 1774,*

1816–1820. *Rudolph, 21/e, pp 2038–2042, 2045–2049, 2085–2086.*) Patients with testicular feminization are genotypic males with normal testes. Complete resistance to androgens causes failure of masculinization of the external genitalia, which remain female. Palpation of tissue below the level of the inguinal ligament in a phenotypical female is suspicious for testicular tissue (more than 50% of patients with testicular feminization have inguinal hernias). The presence (or absence) of a uterus can be confirmed with a rectal examination or with an ultrasound. Chromosomes may be required. At puberty, these persons develop normal female breasts and body habitus despite the presence of testes and high concentrations of testosterone. Because of resistance to androgens, these patients have scant secondary sexual hair and, thus, do not develop hirsutism. The other disorders listed represent syndromes that can present with electrolyte abnormalities and an excess of androgens (leading to secondary sex characteristics and other findings such as hirsutism).

**443. The answer is d.** (*Behrman, 16/e, pp 184–187. McMillan, 3/e, pp 83, 471, 474, 1619, 1900. Rudolph, 21/e, pp 2156–2160.*) The x-ray demonstrates a fracture of the tibia, and also a significant decreased bone mineralization. The child in the question (exclusively breast-fed, no vitamin D supplementation, northern climate with limited sun exposure, African American mother) is at risk for simple (nutritional) rickets. Nutritional rickets is caused by a dietary deficiency of vitamin D and lack of exposure to sunlight. Intestinal absorption of calcium and phosphorus is diminished in vitamin D deficiency. Transient hypocalcemia stimulates the secretion of parathyroid hormone and the mobilization of calcium and phosphorus from bone; enhanced parathyroid hormone activity leads to phosphaturia and diminished excretion of calcium. In children with nutritional rickets, the concentration of serum calcium usually is normal and the phosphate level low. Increased serum alkaline phosphatase is a common finding. The excretion of calcium in the urine is increased only after therapy with vitamin D has been instituted.

**444. The answer is d.** (*Behrman, 16/e, pp 350–352, 1686–1687, 1747–1749, 2131–2132. McMillan, 3/e, pp 1833–1835, 2231, 2241, 2248. Rudolph, 21/e, pp 614, 762–764, 2024, 2090–2091.*) Marfan syndrome is a genetic disorder transmitted as an autosomal dominant trait with variable expression. People with this disorder usually have tall stature, arachnodactyly, sublux-

ation of the lens, dilatation of the aorta, and dissecting aneurysm. Mental retardation is not a part of this syndrome. Vascular complications are the usual cause of death. Patients with any of the other syndromes listed have tall stature and varying degrees of mental retardation among their clinical findings.

**445. The answer is a.** (*Behrman, 16/e, pp 533–534. McMillan, 3/e, p 208. Rudolph, 21/e, pp 82, 2108–2109.*) Glycogen and fat stores are diminished in premature infants and those who are small for gestational age. Energy stores are inadequate to meet the energy demands after the maternal supply of glucose is interrupted at birth, and hypoglycemia ensues. Deficiency of cortisol or growth hormone is a rare cause of neonatal hypoglycemia. Insulin excess, common in infants of diabetic mothers, is unusual in other infants. Hypoglycemia associated with a deficiency of glucagon has been reported but is very rare.

**446. The answer is b.** (*Behrman, 16/e, pp 224–227. McMillan, 3/e, pp 1768–1769. Rudolph, 21/e, pp 161–162, 2268–2269.*) Hypocalcemia of newborn infants can be divided into two groups: early (during the first approximately 72 h of life) and late (after approximately 72 h). The most common type of early neonatal hypocalcemia is the so-called idiopathic hypocalcemia. Other causes early on include maternal illness (diabetes, toxemia, and hyperparathyroidism), neonatal respiratory distress (perinatal asphyxia) or sepsis, low birth weight due to prematurity, or hypomagnesemia. Transient or permanent hypoparathyroidism and high phosphate intake are the most common factors associated with late hypocalcemia. The bicarbonate and glucose levels in the question are normal, while the elevated magnesium level may cause sedation and apnea but not tetany and seizures. Intracranial hemorrhages are less common in an infant of this gestational age, and usually do not present with tetany.

**447–451. The answers are 447-a, 448-c, 449-d, 450-b, 451-d.** (*Behrman, 16/e, pp 1713–1714, 1718–1722, 2128–2130, 2136–2137. McMillan, 3/e, pp 1618–1621, 1770–1772, 1812, 1896–1897. Rudolph, 21/e, pp 163–164, 761–762, 2057–2059, 2151, 2156–2160.*) Vitamin D-resistant rickets is caused by a genetic abnormality in the renal tubular reabsorption of phosphate with resultant hyperphosphaturia and hypophosphatemia and also in the conversion of 25-hydroxyvitamin D to 1,25-dihydroxyvitamin D. The

intestinal absorption of phosphate is also abnormal, and calcium absorption from the gut can be secondarily affected. Calcium concentration is usually normal. The disorder is usually transmitted as an X-linked dominant trait.

Patients with pseudohypoparathyroidism have the same chemical abnormality (low Ca, high  $\text{PO}_4$ ) as those with hypoparathyroidism. They are distinguished from the latter group by the phenotypic features in the question (Albright hereditary osteodystrophy) and high serum concentration of parathyroid hormone. The basic abnormality in these patients is the unresponsiveness of the renal tubules to parathyroid hormone. They are classified into two groups depending on the site of the defect. Type I patients have failure to generate cyclic AMP and do not have an increase in urinary concentration of cyclic AMP or phosphate in response to parathyroid hormone. Type II patients have a defect in the renal tubules that causes failure to respond to high concentrations of cyclic AMP. These patients, if given parathyroid hormone, have increased urinary excretion of cyclic AMP but not of phosphate.

Osteogenesis imperfecta (blue sclera, easily broken bones with minimal trauma) is transmitted as an autosomal recessive (severe form) or, more commonly, autosomal dominant (milder form) disorder. The basic defect is an abnormality in the production and composition of the matrix of bone. Serum calcium and phosphate concentrations are normal.

Hyperparathyroidism is rare in children outside the newborn period, usually presenting with neuromuscular instability such as seizures (numbness and tingling may precede the seizures). In response to high concentrations of parathyroid hormone, there is increased bone resorption. In the kidney, there is increased excretion of phosphate and enhanced formation of 1,25-dihydroxyvitamin D. Increased formation of 1,25-dihydroxyvitamin D, in turn, enhances the absorption of calcium and, secondarily, of phosphorus from the gut. The net effect is hypercalcemia and hypophosphatemia. Children with hypoparathyroidism should be followed carefully for the development of other endocrinopathies such as hypoadrenalism, hypogonadism, hypothyroidism, or diabetes mellitus; inheritance of these multiple endocrinopathies can be autosomal recessive.

Medullary carcinoma of thyroid arises from the C cells of the thyroid. These tumors secrete excessive amounts of calcitonin and, accordingly, the concentration of this hormone in the blood is increased. Despite elevated levels of calcitonin, the serum concentrations of calcium and phosphorus

are usually normal unless the patient has associated hyperparathyroidism (multiple endocrine adenomatosis, type II; see above).

**452–457. The answers are 452-e, 453-a, 454-b, 455-e, 456-e, 457-b.** (Behrman, 16/e, pp 1718–1719, 1737–1739, 1746–1747, 1922. McMillan, 3/e, pp 1620–1621, 1819–1820, 2246. Rudolph, 21/e, pp 1708, 2045–2049, 2152, 2303.) The Prader-Willi syndrome is a disorder consisting of hypotonia, hypogonadism, hyperphagia after the newborn period, hypomentia, and obesity. A deletion of a portion of chromosome 15 has been found in approximately 70% of patients. Children affected by this syndrome exhibit little movement in utero and are hypotonic during the neonatal period. Feeding difficulties and failure to thrive can be the presenting complaints in the first year; later, obesity becomes the most common presenting complaint. The enormous food intake of affected children is thought to be due to a defect in the satiety center in the hypothalamus. Stringent caloric restriction is the only known treatment.

Laurence-Moon-Biedl syndrome is transmitted as an autosomal recessive trait. Obesity, mental retardation, hypogonadism, polydactyly, and retinitis pigmentosa with night blindness are the principal findings in affected children. There is no known effective treatment.

The initial complaint in Cushing syndrome may be obesity. Accumulation of fat in the face, neck, and trunk causes the characteristic “buffalo hump” and “moon” facies. Characteristic features include growth failure, muscle wasting, thinning of the skin, plethora, and hypertension. The bone age of affected patients is retarded, and osteoporosis can be present. The disorder results from an excess of glucocorticoids that may be caused by a primary adrenal abnormality (adenoma or carcinoma) or secondary hypercortisolism, which may be due to excess adrenocorticotropin. Exogenous glucocorticoids administered in supraphysiologic doses for a prolonged period of time will produce a similar picture in normal subjects.

Pseudohypoparathyroidism is a collective term for a variety of diseases. Affected patients have biochemical findings (low serum calcium and high serum phosphorus levels) similar to those associated with hypoparathyroidism, but they also have high levels of endogenous parathyroid hormone; in addition, exogenous parathyroid hormone fails to increase their phosphate excretion or raise their serum calcium level. The defects in these patients appear to be at the hormone receptor site or in the adenylate

cyclase-cyclic AMP system. The symptoms of pseudohypoparathyroidism are due to hypocalcemia. Affected children are short, round-faced, and mildly retarded. Metacarpals and metatarsals are shortened, and subcutaneous and basal ganglia calcifications as well as cataracts can be present. The current treatment consists of large doses of vitamin D and reduction of the phosphate load.

Fröhlich syndrome has none of the characteristics listed in the question.

**458–463. The answers are 458-a, 459-e, 460-e, 461-d, 462-a, 463-b.**

(Behrman, 16/e, pp 195–196, 408–409, 1600–1601, 1726, 1729–1736, 1739–1740. McMillan, 3/e, pp 1621–1624, 1792, 1813, 1816–1819, 1855–1856. Rudolph, 21/e, pp 634–636, 1651, 1714, 2026–2028, 2038–2044, 2245.)

In the salt-losing variety of 21-hydroxylase deficiency, the synthesis of both mineralocorticoids (e.g., aldosterone) and cortisol is impaired. Aldosterone deficiency impairs the exchange of potassium for sodium in the distal renal tubule. Affected patients have hyponatremia and hyperkalemia. Dehydration, hypotension, and shock may be present.

In the absence of vasopressin, renal collecting tubules are impermeable to water, resulting in the excretion of hypotonic urine. Patients with diabetes insipidus present with polyuria and polydipsia. Net loss of water leads to dehydration and hemoconcentration and, therefore, to relatively high serum concentrations of sodium and potassium. Patients with nephrogenic diabetes insipidus have similar laboratory findings. This genetic disorder is unresponsive to antidiuretic hormone (ADH). These patients are unable to concentrate their urine and present in the neonatal period with hypernatremic dehydration.

In hyperaldosteronism, renal tubular sodium–potassium exchange is enhanced. Hypokalemia, hypernatremia, hyperchloremia, and alkalosis are the usual findings. Primary hyperaldosteronism (Conn syndrome) is very rare in children.

Addison disease is associated with a combined deficiency of glucocorticoids and mineralocorticoids. Resorption of sodium and excretion of potassium and hydrogen ions are impaired at the level of the distal renal tubules. Sodium loss results in loss of water and depletion of blood volume. Persons with compensated Addison disease can have relatively normal physical and laboratory findings; Addisonian crisis, however, characteristically produces hyponatremia, hyperkalemia, and shock. The pathophysiol-

ogy of the serum electrolyte abnormalities in this disorder is the same as in the salt-losing variety of adrenogenital syndrome.

Patients with a deficiency of glucose-6-phosphatase (von Gierke disease) are, as a rule, hyperlipidemic. Increased triglyceride concentration in the serum decreases the volume of the aqueous compartment. Because electrolytes are present only in the aqueous compartment of the serum but are expressed in milliequivalents per liter of serum as a whole, the concentrations of sodium and potassium can be factitiously low in these patients.

**464–468. The answers are 464-c, 465-b, 466-e, 467-b, 468-a.** (Behrman, 16/e, pp 713–716, 1588–1589, 1599–1600, 2134. McMillan, 3/e, pp 1545–1546, 1615–1616, 1618–1620, 1882–1883, 2162–2165. Rudolph, 21/e, pp 847–851, 1704–1707, 1709–1710, 2156–2160.) All the disorders listed in the question are familial, even systemic lupus erythematosus (butterfly rash and protein in urine), which appears to result from a combination of environmental and genetic causes. Lupus has been postulated to result from sun, drugs, or infection in genetically predisposed persons.

Hypophosphatemic rickets (vitamin D-resistant rickets) is usually inherited as an X-linked dominant trait. Affected males, therefore, have a more severe form of this disease than affected females presenting with poor calcification of their bones that is unresponsive to vitamin supplementation. Variants of the classic form also seem to be transmitted on occasion as an autosomal recessive and even as an autosomal dominant trait.

Polycystic kidney and liver disease is an autosomal recessive disorder that is associated with the development of hepatic fibrosis as children get older. Renal failure can occur early in infancy but is of variable severity. It can lead to the need for dialysis and renal transplant. Liver disease, the main source of later problems, can lead to portal hypertension.

Adult-type polycystic kidney disease, inherited in an autosomal dominant fashion, is often seen in successive generations of the same family. It presents as hypertension and chronic renal failure in an adult. The ultrasound demonstrates large cysts (sometimes filling the abdomen) originating from the kidney. If adult-type polycystic kidney disease is discovered, the family requires investigation.

Cystinosis is an autosomal recessive disease more commonly found in white children of European ancestry in which affected patients can develop renal failure in childhood or adolescence, or a more benign form in which renal failure does not occur, but crystalline deposits in the cornea

can occur. In the children with early-onset disease, symptoms begin after 6 to 18 months of age with evidence of Fanconi syndrome, progressive renal failure, failure to thrive (with frequent episodes of dehydration, electrolyte disturbance, and rickets), and characteristic corneal crystals. Now that some patients who have cystine storage disease are receiving renal allografts, the pathologic effects of cystine storage in tissues other than the kidney may become clinically important.

**469–470. The answers are 469-a, 470-c.** (*Behrman, 16/e, pp 331, 1746–1747, 1884–1885. McMillan, 3/e, pp 1979–1981, 2234, 2246. Rudolph, 21/e, pp 649–650, 717–720, 2303.*) The mitochondrial genome originates only from the ovum and is, therefore, transmitted by the mother to her offspring of both sexes. Mitochondrial disease involves mainly brain and muscle. Ragged red fibers seen on muscle biopsy are present in several inherited enzyme defects. Examples of mitochondrial inheritance include myoclonic epilepsy and ragged red fibers (MERRF); mitochondrial myopathy, encephalopathy, lactic acidosis, and strokelike episodes (MELAS); and Leber hereditary optic neuropathy (LHON), a condition not associated with myopathy. Prader-Willi syndrome, which is characterized by hypotonia, obesity, hypogonadism, mental retardation, and characteristic hands, feet, and facies, is caused by a chromosomal deletion of 15q11-13 when the chromosome is of paternal origin. In some syndromes, such as Angelman syndrome, chromosomal deletion of maternal origin results in a syndrome characterized by a specific facies, happy disposition, mental retardation, bizarre movements, and seizures. More recently, cases of Prader-Willi syndrome have been found wherein there is no DNA deletion but both copies of chromosome 15 have been inherited from the mother; similarly, some cases of Angelman syndrome have been shown to have two copies of paternally derived chromosome 15. This suggests that it is the lack of part of paternal chromosome 15 that causes Prader-Willi syndrome and the lack of part of maternal chromosome 15 that causes Angelman syndrome.

**471–472. The answers are 471-a, 472-d.** (*Behrman, 16/e, pp 52–57. McMillan, 3/e, pp 527–530. Rudolph, 21/e, pp 2093–2095.*) The first sign of pubertal changes in girls is the development of breast buds (as early as 8 years). In boys, testicular enlargement occurs first, often as early as 9.5 years. Pubertal changes then follow a rather set sequence commonly known

as Tanner staging. Normal ranges for the various levels of development of secondary sexual characteristics are available.

**473–474. The answers are 473-b, c, f; 474-c, i, l.** (*Behrman, 16/e, pp 328, 330, 1753–1755. McMillan, 3/e, pp 2230–2231. Rudolph, 21/e, pp 732–734, 741.*) Common features of Turner syndrome include female phenotype, short stature, sexual infantilism, streak gonads, broad chest, low hairline, webbed neck, congenital lymphedema of the hands and feet, coarctation of the aorta, and a variety of other anomalies.

Down syndrome has many diagnostic features, including short stature, microcephaly, centrally placed hair whorl, small ears, redundant skin on the nape of the neck, upslanting palpebral fissures, epicanthal folds, flat nasal bridge, Brushfield spots, protruding tongue, short and broad hands, simian creases, widely spaced first and second toe, and hypotonia. Cardiac lesions are found in 30 to 50% of children with Down syndrome, including endocardial cushion defect (30%), ventricular septal defect (30%), and tetralogy of Fallot (about 30%). At birth, duodenal atresia is a common finding. It causes bilious vomiting and a characteristic KUB finding of a double bubble (dilatation of the stomach and the proximal duodenum).

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# The Adolescent

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## Questions

**DIRECTIONS:** Each item below contains a question or incomplete statement followed by suggested responses. Select the **one best** response to each question.

### Items 475–476

**475.** During a routine health maintenance visit with a 14-year-old adolescent, which of the following areas of mental development should raise concern?

- a. Development of close, enduring friendships with peers of the same gender
- b. Rejection of parental standards and beliefs, such as those regarding religion or sexual conduct
- c. Disregard for physical well-being
- d. Concern for weight and body configuration
- e. Frequent bickering and quarreling with siblings

**476.** In evaluating this adolescent, which of the following should be considered acceptable?

- a. Involvement in extracurricular activities or a job to the detriment of school-work
- b. Absence of aims or plans for the near or far future
- c. Changing and fleeting romantic attachments (lasting 3 months or less) or multiple simultaneous attachments
- d. Preoccupation with physical well-being, concern for physical symptoms
- e. Constant bickering and quarreling with friends

**477.** In counseling a 15-year-old boy to avoid anabolic steroids, which are being used by some of his teammates, you indicate potential toxic effects to include which of the following?

- a. Hypoglycemia
- b. Increased testicular size
- c. Increased high-density lipoproteins
- d. Toxic hepatitis
- e. Delayed closure of epiphyses

**478.** A 15-year-old girl is brought to the pediatric emergency room by the lunchroom teacher, who observed her sitting alone and crying. On questioning, the teacher learned that the girl had taken five tablets after having had an argument with her mother about a boyfriend of whom the mother disapproved. Toxicology studies are negative, and physical examination is normal. The most appropriate course of action would be to

- a. Hospitalize the teenager on the adolescent ward
- b. Get a psychiatry consultation
- c. Get a social service consultation
- d. Arrange a family conference that includes the boyfriend
- e. Prescribe an antidepressant and arrange for a prompt clinic appointment

**479.** After watching a recent afternoon television show about eating disorders, a mother brings to your office her 15-year-old girl with the concern that “she may have one.” You try to determine whether this girl has anorexia nervosa or bulimia. Which of the following statements is correct?

- a. Rarely do anorexia and bulimia occur in the same patient simultaneously
- b. Anorexia nervosa most commonly occurs in girls less than 10 years old
- c. No signs of bulimia can be found, on careful physical examination
- d. When used appropriately, imipramine is a useful adjunct in the treatment of bulimia
- e. Girls with anorexia nervosa are usually too weak to engage in active sports

**480.** A 17-year-old sexually active girl comes to your office complaining of acne that is unresponsive to the usual treatment regimen. Physical examination reveals severe nodulocystic acne of her face and upper chest and back. You consider prescribing isotretinoin (Accutane), but you are concerned about side effects. Reviewing the literature, you find which of the following to be true about isotretinoin?

- a. Its efficacy can be profound and permanent
- b. It is not known to be a teratogen
- c. Most patients experience excessive tearing and salivation
- d. Severe arthritis necessitating cessation of the drug occurs in about 15% of patients
- e. Significant decrease in serum triglyceride levels are noted in 25% of patients

**481.** A 15-year-old presents with the complaint of a rash, as pictured. Which of the following statements is correct concerning the management of this common condition?



*(Courtesy Adelaide Hebert, M.D.)*

- a. Fried foods must be avoided
- b. Frequent scrubbing of the affected areas is key
- c. Topical antibiotics are of no value
- d. Topical benzoyl peroxide is the mainstay of treatment
- e. This rash is solely a disease of the adolescent

**482.** After a recent suicide by one of his students, the local high school principal asks you to talk with teachers and students about adolescent suicide. Which of the statements is appropriate to include in the discussion?

- a. Girls tend to use more lethal means
- b. The number of attempted suicides is much higher among boys
- c. Those who are successful have a history of a prior attempt or prior serious suicidal ideation
- d. Inquiry by pediatricians, high school teachers, parents, or friends about suicidal thoughts precipitates the act
- e. The number of suicides in adolescents 10 to 19 years of age has decreased significantly since the 1950s

**483.** After a recent news segment about scoliosis, the mother of several of your female patients calls to ask about the condition. You explain to her that idiopathic scoliosis would be most commonly found in her daughters in which age range?

- a. Birth to 3 years
- b. 4 to 10 years
- c. 11 to 18 years
- d. 19 to 25 years
- e. >25 years

**484.** A 16-year-old girl is in your office for a well-adolescent visit, accompanied by her mother. The mother asks about drug and alcohol abuse. You explain that the warning signs of abuse include which of the following?

- a. Excessive concern for weight and body configuration
- b. Improved school performance
- c. Recent changes from age-appropriate, acceptable friends to younger associates
- d. Deterioration in personal habits, hygiene, dress, grooming, speech patterns, and fluency of expression
- e. Improvement in relationships with adults, siblings, and authority figures

**485.** A 16-year-old girl has had unprotected sex for the last several months and now finds herself pregnant. Her pregnancy is at higher risk for

- a. Preeclampsia and eclampsia
- b. Postdate delivery
- c. Male gender
- d. Large-for-gestation-age infants
- e. Multiple birth

**486.** A 15-year-old boy asks how to bulk up. Appropriate advice to increase muscle mass includes

- a. Taking extra vitamins
- b. Doubling protein intake
- c. Using hormones
- d. Increasing muscle work
- e. Taking ergogenic medication

**487.** A 15-year-old sexually active girl is seen in an adolescent clinic for her yearly checkup. Appropriate screening tests for sexually transmitted diseases should be part of that visit. Which of the following sexually transmitted diseases can be diagnosed only when the patient is symptomatic?

- a. Endocervical culture for *Neisseria gonorrhoeae*
- b. Endocervical culture or antigen test for *Chlamydia trachomatis*
- c. Papanicolaou (Pap) smear
- d. Test for herpes simplex type 2
- e. Serologic test for syphilis

**488.** An 18-year-old male college student is seen in the student health clinic for urinary frequency, dysuria, and urethral discharge. Which of the following is likely to explain his condition?

- a. Herpes simplex
- b. *Escherichia coli* urinary tract infection
- c. Chlamydial urethritis
- d. Syphilis
- e. HIV infection

**489.** A 19-year-old male college student returns from spring break in Fort Lauderdale, Florida, with complaints of acute pain and swelling of the scrotum. Physical examination reveals an exquisitely tender, swollen right testis that is rather hard to examine. The cremasteric reflex is absent, but there is no swelling in the inguinal area. The rest of his genitourinary examination appears to be normal. A urine dip is negative for red and white blood cells. Which of the following is the appropriate next step in this man?

- a. Administration of antibiotics after culture of urethra for chlamydia and gonorrhea
- b. Reassurance
- c. Intravenous fluid administration, pain medications, and straining of all voids
- d. Ultrasound of the scrotum
- e. Laproscopic exploration of both inguinal regions

**Items 490–491**

**490.** A 16-year-old girl presents with lower abdominal pain and fever. On physical examination, a tender adnexal mass is felt. Further questioning in private reveals the following: she has a new sexual partner; her periods are irregular; she has a vaginal discharge. The most likely diagnosis is

- a. Appendiceal abscess
- b. Tuboovarian abscess
- c. Ovarian cyst
- d. Renal cyst
- e. Ectopic pregnancy

**491.** You hospitalize the patient for closely supervised treatment. In addition to the acute complications, there are possible long-term sequelae. These include which of the following?

- a. Ectopic pregnancy
- b. Increased incidence of twin gestation
- c. Chronic fatigue syndrome
- d. Ovarian cancer
- e. Endometriosis

**DIRECTIONS:** Each group of questions below consists of lettered options followed by a set of numbered items. For each numbered item, select the appropriate lettered option(s). Each lettered option may be used once, more than once, or not at all. **Choose exactly the number of options indicated following each item.**

**Items 492–495**

Listed below are diseases in sexually active adolescents in which genital ulcers may occur. Match each with the appropriate diagnostic test.

- a. Dark-field microscopic examination
- b. Special chocolate agar culture
- c. Bordet-Gengou medium culture
- d. Tzanck preparation for multinucleated giant cells
- e. *Chlamydia* culture
- f. Loeffler medium culture

**492.** A 16-year-old girl with a positive RPR of 1:64 and an ulcer on her labia (**SELECT 1 TEST**)

**493.** A 19-year-old male with recurrent episodes of painful, erythematous, small vesicles and ulcers on his glans penis (**SELECT 1 TEST**)

**494.** A 15-year-old male with a painless papule on his genitals that resolved but who has now developed a unilateral draining inguinal lymphadenitis (**SELECT 1 TEST**)

**495.** A 19-year-old woman has a few small papules on her labia and perineum. The papules become pustular, eroded, and ulcerated over the next few days; at the same time, the patient develops painful, tender inguinal lymphadenopathy (**SELECT 1 TEST**)

**Items 496–500**

Listed below are common injuries of the adolescent. Match each with the adolescent and his/her activity with which it is most commonly associated.

- a. A 15-year-old competitive swimmer
- b. A 17-year-old high school quarterback
- c. A 17-year-old starting center for a high school basketball team
- d. A 14-year-old long-distance runner
- e. An 11-year-old ballerina
- f. A 16-year-old high school wrestling champion
- g. A 14-year-old snow skier
- h. An 18-year-old college hockey star

**496.** Patellar tendinitis and Osgood-Schlatter disease (**SELECT 1 ACTIVITY**)

**497.** Injuries almost exclusively related to the shoulder, including rotator cuff tendinitis (**SELECT 1 ACTIVITY**)

**498.** Delayed menarche and eating disorder (**SELECT 1 ACTIVITY**)

**499.** Hyperextension of the thumb and sprains of the anterior cruciate ligament (**SELECT 1 ACTIVITY**)

**500.** Shoulder subluxation, knee injuries, and dermatologic problems such as herpes simplex, impetigo, and staphylococcal furunculosis or folliculitis (**SELECT 1 ACTIVITY**)

# The Adolescent

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## Answers

**475–476. The answers are 475-c, 476-c.** (*Behrman, 16/e, pp 52–57. McMillan, 3/e, pp 531–536. Rudolph, 21/e, pp 224–226.*) Adolescence is a time of major physical, cognitive, and emotional changes. The tasks of the adolescent are directed toward determining their ultimate adult self. They must become independent of their parents and, in so doing, take responsibility for their own welfare and start preparing for their future work or career. They must define themselves sexually and move toward lasting attachments. The range of normality is broad and the variations numerous. Evaluation of the adolescent requires weighing the normal and abnormal tendencies to determine where the balance lies. To neglect schoolwork and have no vision or plan for the future suggests either immaturity or depression. A certain degree of concern for appearance is a healthy adolescent phenomenon. On the other hand, excessive concern for physical well-being and physical symptoms suggests an abnormal level of anxiety or depression. Constant quarreling with friends indicates a lack of flexibility and accommodation. Close friends help in the separation from parents and the achievement of independence by providing mutual support and self-justification. Bickering with siblings, on the other hand, is a holdover from childhood and, if not excessive, may be considered normal. Brief superficial romantic attractions fueled more by fantasy than reality start in early to midadolescence. These are rehearsals for the more serious attachments to come.

**477. The answer is d.** (*Behrman, 16/e, pp 571–572. McMillan, 3/e, p 1774.*) As his physician, you point out the untoward medical consequences, which include decrease in testicular size, oligospermia, aggressive behavior, mood swings, liver damage, and decreased levels of high-density lipoproteins, as well as possible premature closure of epiphyses. In addition, you point out that the use of anabolic steroids may give the athlete an unfair advantage, and, therefore, using them is tantamount to cheating.

**478. The answer is a.** (*Behrman, 16/e, pp 559–560. McMillan, 3/e, pp 810–812. Rudolph, 21/e, pp 501–503.*) The adolescent who has attempted

suicide should be hospitalized so that a complete medical, psychological, and social evaluation can be performed and an appropriate treatment plan developed. Hospitalization also emphasizes the seriousness of the adolescent's action to her and to her family and the importance of cooperation in carrying out the recommendations for ongoing future therapy. The treatment plan may include continued counseling or supportive therapy with a pediatrician, outpatient psychotherapy with a psychiatrist or other mental health worker, or family therapy.

**479. The answer is d.** (*Behrman, 16/e, pp 562–564. McMillan, 3/e, pp 651, 813–820. Rudolph, 21/e, pp 231–233.*) Eating disorders have become increasingly prevalent in recent years. Symptoms of finicky appetite, progressively restricted food intake, distress at looking fat, and compulsively pursuing thinness can appear after puberty. Parents may not appreciate the magnitude of weight loss until it reaches 10% or more of body weight because those girls will not undress in their parents' presence and because the facial contours are the last parts to be affected. Bulimia usually appears in midadolescence rather than early adolescence and is characterized by sessions of gorging, often in secret and often involving a single favored snack food such as ice cream, cake, or candy, although it may also be manifested as immoderate eating at mealtimes. This gorging is followed by secret bouts of self-induced vomiting. Some bulimics also use laxatives and purgatives. Physical consequences of bulimia include esophageal varices and hemorrhage; dental decay, especially of anterior teeth (because of exposure of enamel to gastric HCl); and a swollen, reddened, irritated uvula (also from chronic HCl exposure). Physical consequences of anorexia include profound weight loss (25 to 30% or more of body weight), dehydration, facial and arm hirsutism, loss of hair of the head, bradycardia, cardiac conduction problems, hypocardia, hypothermia, impaired renal function, multiple malnutrition effects (including avitaminoses), a primary or secondary amenorrhea, and osteoporosis. There is significant mortality in treatment-resistant cases. The psychological component of these disorders is not a unitary one. Some anorexic patients have an underlying obsessive-compulsive or narcissistic personality disorder, some are borderline psychotic, and some are depressed. Bulimic patients have a significant underlying depression. Patients with eating disorders have exceedingly ambivalent feelings toward parents, especially mothers, and in turn evoke great ambivalence on the part of parents. Therapy includes behavior mod-

ification to deal with eating behavior per se, family therapy, and individual or group therapy. Imipramine, when used appropriately, is a useful adjunct treatment for this condition. In cases of life-threatening degrees of weight loss or vomiting, hospitalization to limit freedom, restore physiologic equilibrium, and provide a controlled eating environment can be indicated.

**480. The answer is a.** (*Behrman, 16/e, pp 2047–2049. McMillan, 3/e, pp 729–731, 2229. Rudolph, 21/e, pp 1208–1210.*) Isotretinoin (13-*cis*-retinoic acid; Accutane) has proved to be very effective in the treatment of refractory nodulocystic acne. The effects of treatment appear to be long-lasting. Precautions regarding its use, however, are essential. Because of its teratogenic effects (isotretinoin syndrome), the drug is contraindicated during pregnancy and within 1 month of becoming pregnant. Dry skin, eyes, and mucous membranes are the most frequent complications of therapy. Other associated problems include musculoskeletal pain and hyperostosis, inflammatory bowel disease, pseudotumor cerebri, and corneal opacities. Patients on isotretinoin therapy can often develop abnormal liver function tests, elevated triglyceride and cholesterol levels, and lowered levels of high-density lipoproteins.

**481. The answer is d.** (*Behrman, 16/e, pp 2047–2049. McMillan, 3/e, pp 729–731. Rudolph, 21/e, pp 1208–1210.*) Acne is a skin disorder that affects virtually all adolescents and is seen less commonly in older patients. There is a wide spectrum of clinical findings ranging from a few papules and comedones to a disfiguring nodulocystic disease of the face and trunk. The goals of therapy are to prevent scarring and disfigurement and to avoid loss of self-esteem. The chief benefit of benzoyl peroxide is derived from its antibacterial activity, but it also functions as an exfoliant and comedolytic. A combination of benzoyl peroxide and retinoic acid is particularly effective in sloughing the epithelium, and statics, such as oral tetracycline, and the use of topical antibiotics can be necessary to control the inflammatory component of acne. Studies have failed to demonstrate adverse effects of any particular foods on disease activity. Vigorous facial or body scrubbing can traumatize the skin and aggravate the problem.

**482. The answer is c.** (*Behrman, 16/e, pp 599–560. McMillan, 3/e, pp 810–812. Rudolph, 21/e, pp 501–503.*) Suicide among teenagers has increased steadily since 1950 and is now the third leading cause of adolescent death,

following accidents and homicides. Suicide attempts occur more often in girls, but in all age groups males outnumber females in completed suicides because boys tend to use more lethal means, such as firearms, hanging, jumping, and inhalation of carbon monoxide. Most suicide attempters and completers (where history can be established) have a history of a prior attempt or prior serious suicidal ideation. Therefore, direct questioning of the adolescent about feelings of sadness, hopelessness, concerns about death, and thoughts of committing suicide is important. There are no data to indicate that such inquiry precipitates suicidal behavior.

**483. The answer is c.** (*Behrman, 16/e, pp 2083–2084. McMillan, 3/e, pp 2117–2121. Rudolph, 21/e, pp 2000–2001.*) The most common form of scoliosis is idiopathic scoliosis. Three age ranges of idiopathic scoliosis exist: infantile (which presents at birth to 3 years of age), juvenile (presenting at 4 to 10 years of age), and adolescent (the most common form, accounting for 80% of cases and presenting from 11 years and older). To diagnose this condition, the back is viewed from behind, with the patient in the standing position. The waist, shoulders, and pelvis should be symmetric. The spine is examined for symmetry or deformity. The patient is then asked to bring the palms together in the front and bend at the waist. Viewing the patient from behind will allow for identification of any humps, valleys, or other deformities of the spine. Identified abnormalities can be confirmed radiographically. Premenarchal girls with a curvature of the spine of more than 20° on radiographs need close follow-up every 4 to 6 months because the risk of progression is high.

**484. The answer is d.** (*Behrman, 16/e, pp 564–567. McMillan, 3/e, pp 550–555. Rudolph, 21/e, pp 226–230.*) When an adolescent shows evidence of declining school performance or truancy, a change for the worse in personal habits and grooming, exaggerated mood swings, change in friends to an older and unacceptable group, and frequent hostile reactions in relationships with others, the possibility that he or she is abusing drugs or alcohol, or both, should be strongly considered. Dependence on drugs is a progressive disorder; therefore, prompt identification and intervention are required if serious complications are to be avoided. Routine health assessment of adolescents should include inquiry about the use of cigarettes, alcohol, and other drugs, and about school performance and family and peer relationships.

**485. The answer is a.** (Behrman, 16/e, pp 580–582. McMillan, 3/e, p 543.) The main obstetric complications of teenage pregnancy are preeclampsia and eclampsia, which are thought to be due to inadequate prenatal care and nutrition. The rate of prematurity is high, and this in turn is thought to account for the increased incidence of mental retardation among children of teenage mothers. The incidences of repeat pregnancy and infant mortality are high. A combination of medical, social, psychological, and educational resources must be made available to permit optimum health and development of the teenage mother and her child. Multiple births and male gender do not occur with any greater frequency in teenagers than in older women.

**486. The answer is d.** (Behrman, 16/e, p 2110. McMillan, 3/e, p 736. Rudolph, 21/e, pp 2130–2131.) Increased muscle work (along with increased calories) is the only appropriate way to increase muscle mass. Measurements of skin-fold thickness, performed serially, are a useful way to detect changes in the amount of body fat, so that obesity can be avoided. Protein loading or using drugs, hormones, and vitamins will not be helpful and may be harmful.

**487. The answer is d.** (Behrman, 16/e, pp 583–586. McMillan, 3/e, pp 555–559.) The sexually active teenager can be screened for all the conditions listed except asymptomatic genital herpes, for which there is no satisfactory screen. A Pap smear can reveal cellular changes that represent precursors of cervical neoplastic disease. Squamous intraepithelial lesions, formerly known as cervical intraepithelial neoplasia (CIN), affect about 5 to 10% of sexually active adolescents. Gonorrhea can be asymptomatic; syphilis is on the rise; chlamydial infection is present in 10 to 25% of all sexually active adolescents. Screening tests for these diseases are readily available. A wet mount of vaginal secretions can demonstrate trichomonal, candidal, and bacterial vaginosis.

**488. The answer is c.** (Behrman, 16/e, pp 583–586. McMillan, 3/e, p 562. Rudolph, 21/e, pp 260–270.) Urethritis in an adolescent male is almost always a sexually transmitted disease (STD), either gonococcal or nongonococcal urethritis (NGU). *Chlamydia trachomatis* is usually the causative agent in NGU. Less frequently, NGU can be caused by *Ureaplasma urealyticum*, *Trichomonas vaginalis*, and yeast. Herpes simplex can cause an NGU, but it is

considerably less likely than *C. trachomatis*. Gonococcal culture and Gram stain are easily available; chlamydial culture may not be. Direct monoclonal antibody tests as well as enzyme immunoassay and molecular probe tests are alternative methods for *Chlamydia* identification, although they are less sensitive and less specific than chlamydial culture. Ceftriaxone and doxycycline (among other antibiotics) would be reasonable choices as treatment of urethritis since both gonococcal and nongonococcal infections are covered by these antibiotics. Azithromycin, which has the advantage of a one-dose regimen, may be used instead of doxycycline. Serologic testing for syphilis should always be done; in none of its normal presentations is urethral discharge common. Testing for HIV should be offered and safe sexual practices encouraged; HIV does not cause urethral discharge. Urinary tract infection is not associated with a urethral discharge.

**489. The answer is d.** (Behrman, 16/e, pp 583–586, 1652–1653. McMillan, 3/e, pp 1555–1558, 1628–1629. Rudolph, 21/e, pp 260–270.) The patient in the question may have a torsion of his testis that requires immediate attention. Another possibility would be epididymitis, especially if there is a possible antecedent history of sexual activity or urinary tract infection. Prehn's sign, although not totally reliable, is elicited by gently lifting the scrotum toward the symphysis. Relief of the pain points to epididymitis; its worsening, to torsion. Doppler ultrasound (or surgical consultation) is a logical first step in this man's evaluation, demonstrating absence of flow in torsion and increased flow in epididymitis. Alternatively, a radionuclide scan will show diminished uptake in torsion and increased uptake in epididymitis. Treatment for torsion is surgical exploration and detorsion and scrotal orchiopexy. Causative organisms for epididymitis include *Neisseria gonorrhoeae*, *Chlamydia trachomatis*, and other bacteria. Treatment with appropriate antibiotics and rest is indicated. However, treating this patient with antibiotics without first excluding testicular torsion is ill-advised; loss of the testis can be expected after 4 to 6 h of absent blood flow if the testis has torsioned. Strangulated hernia is associated with evidence of intestinal obstruction.

**490–491. The answers are 490-b, 491-a.** (Behrman, 16/e, pp 583–586, 829–832, 919–920. McMillan, 3/e, pp 561–562. Rudolph, 21/e, pp 260–270.) Each year, a diagnosis of pelvic inflammatory disease (PID) is made in over 1 million women. Sexually active teenagers are at great risk of acquiring

PID because of their high-risk behavior, exposure to multiple partners, and failure to use contraceptives. The strong likelihood of PID in the patient presented should not preclude consideration of serious conditions requiring surgical intervention, such as appendiceal abscess, ectopic pregnancy, and ovarian cyst. Renal cyst does not present in the manner described. An episode of PID raises the risk of ectopic pregnancy, and about 20% of women become infertile following one episode of PID. Other sequelae include dyspareunia, pyosalpinx, tuboovarian abscess, and pelvic adhesions. Endometriosis is not related to PID.

**492–495. The answers are 492-a, 493-d, 494-e, 495-b.** (Behrman, 16/e, pp 584–585, 838, 903–906, 920, 966–972. McMillan, 3/e, pp 558–560, 1024–1025, 1120–1121. Rudolph, 21/e, pp 928–929, 1002–1005, 1168, 1173.) In the first patient, syphilis is likely. A scraping from a genital ulcer should be examined under dark-field microscopy for *Treponema pallidum*. A serologic test for syphilis is indicated for every sexually active patient recently infected with a sexually transmitted disease. In the event of a negative result, consideration should be given to repeating this test at a later date because serologic tests performed too early in the disease may not be reliable.

Herpes should be suspected in a patient with recurrent shallow, painful ulcers of the genitourinary tract. A herpes simplex ulcer can be either a primary or a secondary infection (as in the case presented). Examination of a Tzanck preparation of scraping from the ulcer reveals multinucleated giant cells and intranuclear inclusions. Herpes simplex virus is readily cultured.

Chancroid caused by *Haemophilus ducreyi* is difficult to culture. Special chocolate agar medium is only 65% sensitive. Inguinal adenopathy that suppurates and causes chronic draining of sinuses is commonly seen in chancroid and can be confused with lymphogranuloma venereum (LGV). Typically, the inguinal adenopathy of chancroid occurs at the same time as the genital ulcer, while the adenopathy in LGV occurs after the ulcer has healed. LGV is caused by serovars of *Chlamydia trachomatis*, which can be cultured. Serial serologic testing for chlamydial antibodies can be of diagnostic value retrospectively.

Genital ulcers probably play a role in the transmission of HIV infection. Moreover, manifestations of syphilis, herpes simplex, and chancroid can be more severe in HIV patients and may not respond as well to standard treatments.

**496–500. The answers are 496-c, 497-a, 498-e, 499-g, 500-f.** (*Behrman, 16/e, pp 2110–2112. McMillan, 3/e, pp 736–737, 742, 745–746, 748. Rudolph, 21/e, pp 2103, 2432, 2447, 2449–2451.*) A wide variety of injuries occur in adolescents who participate in sports. Some injuries, however, are more common in certain sports than in others.

For swimmers, shoulder injuries are the most common type of problem seen. Rotator cuff tendinitis of the biceps and/or the supraspinatus muscles presents as shoulder pain and tenderness.

In football, head and neck injuries are not uncommon, but, fortunately, serious injuries are rare. Knee injuries such as anterior cruciate, posterior cruciate, and collateral ligament tears do occur. In addition, turf toe, injury to the first metatarsophalangeal joint, is seen when play is on artificial turf.

Basketball and volleyball tend to produce lower extremity problems, including those of the knee, with such injuries as Osgood-Schlatter disease and sprains to the ligaments of the knee. Ankle injuries, too, are quite common in these sports.

Injuries due to running are frequently muscle strains in the hamstrings, adductors, soleus, and gastrocnemius muscles. Runner's knee, anterior knee pain due to patellofemoral stress, is also seen.

Ballet can be associated with delayed menarche and eating disorders (more commonly in the female dancers). In addition, a variety of mostly lower extremity problems can be seen, ranging from bunions to serious overuse knee and ankle problems. Injuries in wrestlers are frequently seen in the upper extremities, especially shoulder subluxation, and in the knees, usually prepatellar bursitis from traumatic impact to the floor. Additionally, a variety of skin conditions are common, ranging from contact dermatitis and superficial fungal infections to herpes simplex (*herpes gladiatorum*), impetigo, and staphylococcus furunculosis or folliculitis.

Availability of better equipment has resulted in a decrease in the number of serious skiing injuries. Thumb injuries during a fall, skier's thumb (abduction and hyperextension of the thumb, causing a sprain of the ulnar collateral ligament), remains the most common injury seen.

Hockey injuries range from mild contusions to significant lacerations. No particular type of injury (with the exception of, perhaps, loss of teeth) is characteristic of this sport.

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# Index

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## A

- Abdominal pain, recurrent, 165, 179–180
- Abscesses  
  lung, 132, 144  
  retropharyngeal, 139, 151
- Acetylcholinesterase, 40
- Achondroplasia, 294, 311, 313
- Acidemia, 139, 150
- Acne  
  in adolescents, 329, 337  
  in neonates, 11, 39
- Acroparesthesia, 227
- Activated charcoal, 38, 167, 181
- Addison disease, 302, 303, 309, 322–323
- Adenoma sebaceum, 110
- Adolescence  
  cognitive and emotional changes during, 327, 335  
  drug and alcohol use during, 330, 338  
  pregnancy during, 330, 339  
  sports-related injuries of, 334, 342  
  and STDs, 331–333, 339–341  
  suicide attempts during, 328, 329, 335–338
- Adrenal hyperplasia  
  bilateral, 292, 310  
  congenital, 223, 298, 302, 303, 314, 316, 322
- AIDS (see HIV infection)
- Air-contrast enemas, 161, 175
- Albright hereditary osteodystrophy (see Pseudohypoparathyroidism)
- Alcohol  
  maternal use of, 30, 56  
  poisoning from, 29, 55
- Alkaptonuria, 298, 316–317
- $\alpha_1$  antitrypsin deficiency, 306
- Alport syndrome, 89, 194, 205
- Aluminum hydroxide, maternal use of, 101
- Anabolic steroids, 327, 335
- Anaphylactoid purpura, 5, 37, 192, 204
- Ancylostoma duodenale*, 172
- Anemia  
  aplastic, 278  
  hemolytic, 278, 281  
  iron-deficiency, 169, 171–172, 182, 273, 279, 281, 283–284  
  sickle cell, 33, 59, 253, 279  
  from thalassemia major, 276, 277, 286–287
- Angelman syndrome, 324
- Angioedema, hereditary, 114, 122–123
- Animal bites, 20, 45–46
- Aniridia, 109, 271, 282
- Anorexia nervosa, 336
- Aplastic anemia, 278
- Apnea, 65, 94, 101  
  idiopathic, 80, 105  
  obstructive sleep, 134, 146
- Appendicitis, 171
- Apt test, 63, 92, 168, 181
- Arthralgia, 113, 121
- Arthritis, 259  
  gonococcal, 41  
  juvenile rheumatoid, 2, 35–36, 117, 126–127  
  from meningococemia, 263  
  septic, 27, 45, 53, 127
- Ascaris lumbricoides*, 262
- Asphyxiation, 25, 42, 50, 63, 91
- Aspirin  
  maternal use of, 279  
  and Reye syndrome, 183, 252
- Atresia  
  choanal, 90  
  duodenal, 74, 101, 106  
  pulmonic, 127  
  tricuspid, 118, 127
- Atrial septal defects (ASDs), 115, 123, 257
- Attention deficit disorder with hyperactivity (ADHD), 223–224, 231
- Auspitz sign, 39

**B**

Baby walkers, and injuries, 25, 50, 51  
Bacteremia, pneumococcal, 238, 256–257  
Bacterial tracheitis, treatment for, 135, 146–147  
*Bacteroides*, 38, 46, 144  
Barium enemas, 160, 174  
Bartter syndrome, 196, 206  
Behçet syndrome, 184  
Bell palsy, 1, 35  
Benzoyl peroxide, 329, 337  
Beriberi, 52  
 $\beta$ -glucocerebrosidase deficiency, 306–307  
 $\beta$ -thalassemia trait, 270, 279, 280  
Bilirubin, 89–90, 101–102  
Biotinidase, deficiency of, 86, 108, 109  
Bites  
  arthropod, 218, 230  
  human, 7, 38, 46  
  mammalian, 20, 45–46  
Bladder, injuries to, 197  
Blood gases, 141, 142, 152, 153  
Bone age, 294, 296, 312–314  
Bone marrow transplants, and HLA compatibility, 294, 312  
*Borrelia burgdorferi*, 263  
Botulism, infant, 212, 226, 238, 256  
Brainstem, compression of, 225  
Breast milk  
  calcium and phosphorus content of, 93  
  medication secretion through, 64, 92  
  vitamin D content of, 32, 33, 59  
Breath-holding spells, 10, 39  
Breath hydrogen test, 163, 177–178  
Bronchiolitis, 131, 143  
  acute, 106–107  
Bronchopulmonary dysplasia (BPD), 84, 107, 108  
Bruton disease, 265  
Bulimia, 336

**C**

Café au lait spots, 231  
Caloric deficiency, 25, 26, 51  
Caloric intake, in newborns, 62, 90–91  
*Candida albicans*, intradermal testing with, 33, 34, 59  
Cantharidin, 31, 56, 57  
Caput succedaneum, 87, 88, 110  
Caries, from baby-bottle nursing, 165, 179  
Cataracts, with myotonic muscular dystrophy, 210, 223  
Cat-scratch disease, 266, 285  
Cellulitis, orbital, 239, 257  
Cephalohematomas, 87, 88, 110, 111  
Cerebellar ataxia, 212, 225–226  
Cerebral palsy, 218, 219, 230, 231  
Cerebrospinal fluid (CSF)  
  lowered glucose levels in, 210, 224  
  measuring protein levels of, 229  
Chancroid, testing for, 333, 341  
Charcot-Marie-Tooth disease, 229  
Chickenpox (*see* Varicella)  
Child abuse, 99, 198  
  sexual, 45, 204  
  suspected after autopsy, 6, 37  
*Chlamydia trachomatis*, 339–341  
  congenital, 140, 151, 267  
  pneumonia from, 97, 249, 267–268  
Choanal atresia, 90  
Choking, 25, 50  
Cholinesterase-reactivating oximes, 40  
Cleft lip, 76, 102–103  
*Clostridium botulinum*, 238, 256  
Colic, 170, 183  
Colitis, granulomatous (*see* Crohn disease)  
Congenital cytomegalovirus (CMV)  
  infection, 63, 91, 214, 227–228, 251, 270, 278  
Congestive heart failure, 116, 120, 125, 129  
Conjunctivitis, 97, 124, 151  
Constipation, functional, 155, 171, 174  
Coombs test, 65, 93, 94, 102, 202, 283

Corpus callosum, agenesis of, 226  
 Coxsackie A-16 virus, 253  
 Cranial nerve III, compression of, 225  
 Craniopharyngiomas, 212, 225  
 Creatinine, 199  
 Crohn disease, 161, 175–176, 184  
 Croup, 246, 264  
 Cryptorchidism, 189, 201  
*Cryptosporidium*, 246, 264  
 Cushing syndrome, 310, 321  
 Cutaneous larval migrans, 218, 230  
 Cystathioninuria, 221  
 Cystic fibrosis (CF), 170, 178, 182, 183  
   DNA testing for, 292, 310–311  
   pancreatic enzyme replacement for, 156, 172–173  
   and predilection for bacterial disease, 266–267  
   sweat chloride test for, 132, 143, 144  
 Cystinosis, 303, 304, 323–324  
 Cysts, popliteal, 41  
 Cytomegalovirus, 253

## D

Deafness, 86, 87, 109, 110  
   from Alport syndrome, 205  
 Deferoxamine mesylate, for treating iron toxicity, 28, 29, 54–55  
 Dehydration, 163, 178, 179, 198  
 Dermatitis  
   atopic, 31, 56, 57  
   contact, 39, 342  
   seborrheic, 23, 24, 39, 48, 49  
 Dermatomyositis, 204  
 Desmopressin acetate (DDAVP), 205, 215, 228  
 Developmental language disorder, 207, 220  
 Development, physical  
   during early childhood, 1, 35  
   during infancy, 1, 35  
 Diabetes insipidus  
   from head injuries, 215, 228  
   nephrogenic, 187, 198, 199, 302, 303, 322

Diabetes mellitus, 55, 76, 89, 103, 198, 257, 309  
 Diaphragmatic hernia, 71, 99  
 Diarrhea, *Cryptosporidium*-caused, 246, 264  
 DiGeorge anomaly, 265  
 Dilantin, 89  
 Diphenhydramine, for treating phenothiazine toxicity, 28, 29, 54  
 Diphtheria, 45, 257  
 Disseminated intravascular coagulopathy (DIC), 274, 279  
   and fibrin split products, 272, 282–284  
 Diuretics, 206  
 Down syndrome, 123, 128, 305, 325  
 Doxorubicin, side effects of, 285  
 Droplet precautions, with pertussis, 83, 84, 106, 107  
 Drowning, 25, 50  
 Dubin-Johnson syndrome, 317  
 Duodenal atresia, 74, 101, 106

## E

Eating disorders, 328, 336–337, 342  
 Ebstein anomaly, 117, 125–126  
 Echo-11 virus, 253  
 Eczema, 15–16, 42–43  
 Edrophonium chloride, 209, 221, 222  
 Ellis–van Creveld syndrome, 123  
 Encephalitis, granulomatous, 227–228  
 Endocarditis, infective, 4, 36–37  
*Enterobius vermicularis* (see Pinworms)  
 Enzyme-linked immunosorbent assay (ELISA) test  
   for HIV infection, 69, 98  
   for *Toxocara canis*, 135, 147  
 Eosinophilia, 272, 283  
 Epididymitis, 236, 253, 254, 340  
 Epiglottitis, 135, 147, 241, 258, 264  
 Epinephrine, nebulized, 83, 84, 106, 107  
 Epistaxis, 172  
 Epstein-Barr virus, 255  
 Erb-Duchenne paralysis, 66, 95

- Erythema  
  chronicum migrans, 263  
  infectiosum, 253  
  multiforme, 258, 261  
  toxicum, 23, 48, 49
- Erythromycin, for treating pertussis,  
  237, 254, 255
- Escherichia coli*, with hemolytic-uremic  
  syndrome, 202
- Esophagus  
  anomalies of, in newborns, 72, 99  
  foreign bodies in, 138, 149–150
- Ethanol, for treating methanol poison-  
  ing, 28, 29, 54, 55
- Exanthema subitum (*see* Roseola)
- Extracorporeal membrane oxygenation  
  (ECMO), 83, 84, 99, 106–108
- F**
- Fabry disease, 223, 227, 307
- Factor VIII deficiency, 284
- Failure to thrive (FTT), 18, 43
- Fanconi syndrome (FS), 187, 198–199,  
  317, 324
- Ferrous sulfate, for treating iron-  
  deficiency anemia, 273, 284
- Fetal alcohol syndrome, 30, 55, 56, 61,  
  89
- Fetomaternal transfusion, 271, 281
- Fibrin split products, and DIC, 272,  
  282–284
- Fifth disease, 253
- Fluoroscopy, 67, 96, 97
- Folic acid  
  deficiency of, 275, 286  
  supplementation with, 32, 58, 80,  
  104, 281
- Foreign bodies, 151–152  
  in esophagus, 138, 149–150  
  in nose, 134, 146  
  in vagina, 45
- Formulas, infant  
  calcium and phosphorus content of, 93  
  caloric density of, 90–91  
  dilution of, 48
- Fragile X syndrome, 22, 47–48, 223
- Fröhlich syndrome, 322
- Fructosemia, 306
- G**
- Galactosemia, 86, 108–109, 289, 306
- Gallbladder, hydrops of, 47
- Gamma benzene hexachloride, 40
- Gangliosidosis type 1, generalized, 213,  
  226–227
- Gastroesophageal reflux (GER) disease,  
  161, 175, 176
- Gaucher disease, 223, 289, 306–307
- German measles (*see* Rubella)
- Giardia lamblia*, 264
- Giardiasis, 204
- Gilles de la Tourette syndrome  
  (*see* Tourette syndrome)
- Glomerulonephritis, 32, 57, 58  
  acute, 196, 206  
  poststreptococcal, 190, 201–204
- Glucose-6-phosphatase deficiency, 302,  
  303, 306, 322, 323
- Glucose-6-phosphate dehydrogenase  
  (G6PD) synthesis  
  ethnic differences of, 270, 279–280  
  and hyperbilirubinemia, 283
- Glucose intolerance, 200
- Gluten-sensitive enteropathy, 170, 183,  
  184
- Glycosylated hemoglobin, 297,  
  315–316
- Gonorrhea, 45, 339, 340
- Gower sign, 229
- Graft-versus-host disease (GVHD), 312
- Grand mal seizures, 85, 109, 110
- Growth hormone deficiencies, 293, 311,  
  312
- Guillain-Barré syndrome, 216, 229
- H**
- Haemophilus ducreyi*, 341
- Haemophilus influenzae* type B, 251, 254,  
  257  
  immunization against, 45, 147, 149,  
  258  
  and sickle cell disease, 281

- Hand-foot-and-mouth disease, 253  
 Harlequin syndrome, 105  
 Headaches  
   with hypertension, 189, 200, 202  
   migraine, 210, 222–224  
 Head lice, 17, 43  
 Hearing impairment  
   from Alport syndrome, 205  
   neonatal, 18, 44  
 Heart block, congenital, 120, 129  
 Heart disease  
   congenital, 115, 123–124, 129  
   rheumatic, 252  
*Helicobacter pylori*, 171, 248, 266  
 Hematemesis, 92  
 Hematocrit, 81, 105–106  
 Hematuria, 190, 201–203  
 Hemoglobin A<sub>2</sub>, levels of, 279  
 Hemoglobin C disease, 276, 277, 286, 287  
 Hemolytic anemia, 278, 281  
 Hemolytic disease, 65, 93–94  
 Hemolytic-uremic syndrome, 190, 202  
 Hemophilia, 99  
   type A, 284  
 Henoch-Schönlein purpura, 279  
 Heparin, maternal use of, 100–101  
 Hepatitis  
   syphilitic, 103  
   type A, 242, 259  
   type B, 21, 47, 73, 100, 178, 259  
   type C, 163, 178, 259–260  
 Hepatocellular carcinoma, 21, 47  
 Hernia  
   inguinal, 166, 180  
   strangulated, 340  
 Herpes simplex, 339–340, 342  
   testing for, 333, 341  
 Hirschsprung disease, 171, 174  
 Histidinemia, 221  
 HIV infection, 341  
   congenital, 43, 69, 98, 233, 250–251  
   testing for, 18, 43, 69, 98, 340  
 Hodgkin disease, 274, 285–286  
 Holt-Oram syndrome, 128  
 Homocystinuria, 208, 221  
 Hookworms, 172, 262  
 Horner syndrome, 95  
 Howell-Jolly bodies, 33, 34, 59, 276, 286  
 Human bites, 7, 38, 46  
 Human herpesvirus 6 (HHV-6), 248, 266  
 Human leukocyte antigen (HLA), compatibility for bone transplants, 294, 312  
 Hyaline membrane disease, 69, 85, 97–98, 107, 108  
 Hydramnios, 106  
 Hydrocele, 194, 205  
 Hydrocephalus, 111  
 Hyperaldosteronism, 316  
   primary, 302, 303, 322  
 Hyperbilirubinemia, 102, 106, 178  
   with G6PD deficiency, 283  
 Hypercalcemia, 297, 315  
 Hypercalciuria, idiopathic, 202  
 Hypercholesterolemia, 114, 122  
 Hyperglycemia, 103  
 Hypermagnesemia, neonatal, 96  
 Hypernatremia, 164, 178–179  
 Hyperostosis, 29, 55  
 Hyperparathyroidism, 200, 301, 319, 320  
 Hypertension, 189, 200–201  
   with glomerulonephritis, 202, 206  
 Hypertrophic cardiomyopathy (HCM), 120, 129  
 Hypocalcemia, neonatal, 64, 93, 300, 319  
 Hypoglycemia, 96, 101, 103, 319  
 Hypopituitarism, 309  
 Hypoplasia  
   of distant phalanges, 30, 55, 56  
   pulmonary, 205  
 Hypothyroidism, 309, 312, 316  
   congenital, 66, 86, 94–95, 108, 109  
 Hypoxia, 138, 141, 142, 150, 152–153
- I**
- Idiopathic (immune) thrombocytopenic purpura (ITP), 269, 270, 278, 279, 284

- IgA nephropathy, 202  
ileal-loop bags, 190, 201  
Immunization  
  *Haemophilus influenzae* type B, 45,  
    147, 149, 258  
  hepatitis B, 100  
  for HIV-positive patients, 251  
  influenza, 234, 251–252  
  pertussis, 45, 237, 254–255  
  rubella, 310  
  tetanus, 20, 45, 46, 180  
  varicella, 62, 90  
Immunodeficiencies, B- and T-cell, 247,  
  265–266  
Impetigo, 31, 56–57, 342  
Incontinentia pigmenti, 228  
Infantile atrophy, 51  
Infantile hemiplegia, acute, 209, 222  
Infant respiratory distress syndrome (*see*  
  Hyaline membrane disease)  
Influenza, immunization against, 234,  
  251–252  
Inheritance  
  of endocrine and metabolic diseases,  
    303, 304, 323–324  
  of genetic diseases, 48, 104, 304, 324  
Insulin, for clearance tests, 187, 199  
Interferon  $\alpha$ , for treating childhood  
  hepatitis, 163, 178  
International Bone Marrow Transplant  
  Registry, 312  
Intraventricular hemorrhage (IVH), 87,  
  88, 110, 111  
Intussusception, 171, 175  
Iridocyclitis, 2, 35–36  
Iron  
  deficiency of, 169, 171–172, 182,  
    273, 279, 281, 283–284  
  toxicity from, 29, 54–55  
Isoniazid (INH), 133, 144, 145  
Isotretinoin, side effects of, 328, 337
- J**  
Jaundice, 75, 101–102  
  obstructive, 163, 178
- Jervell-Lange-Nielsen syndrome, 129  
Job-Buckley syndrome, 266  
Joint fluid, examination of, 20, 45  
Jones criteria, 121  
Juvenile rheumatoid arthritis (JVA), 2,  
  35–36, 117, 126–127  
Juxtaglomerular hyperplasia (*see* Bartter  
  syndrome)
- K**  
Kaposi sarcoma, 251  
Kawasaki disease, 47, 116, 124–125,  
  129, 234, 252–253, 261–262  
Kayser-Fleischer rings, 317  
Kerion, 235, 253  
Kidneys, injuries to, 197  
*Klebsiella*, 144  
Klinefelter syndrome, 291, 308–309  
Koplik spots, 250, 258, 262  
Krabbe disease, 223  
Kwashiorkor, 162, 176
- L**  
Labial adhesions, 293, 311  
Lactose  
  and galactosemia, 108–109  
  intolerance of, 163, 177–178  
Laurence-Moon-Biedl syndrome, 321  
Lead poisoning, 21, 27, 46–47, 53–54,  
  279  
  versus iron-deficiency anemia, 284  
Learned behavior, with sleep distur-  
  bances, 24, 25, 49  
Leber hereditary optic neuropathy  
  (LHON), 324  
Legg-Calvé-Perthes disease, 27, 41, 53  
Leukodystrophy, metachromatic, 227  
Lichen planus, 39  
Lisch nodules, 231  
Lithium  
  and renal disease, 199  
  secretion through breast milk, 64, 92  
Löfller syndrome, 262  
Low-birth-weight infants, complications  
  in, 94

- Lumbar puncture, 208, 221, 236, 254  
for meningitis, 246, 264
- Lung abscesses, antibiotic therapy for, 132, 144
- Lupus erythematosus, 55  
cardiac abnormalities with, 30, 32, 56–58  
systemic, 191, 203, 204, 279, 307, 323
- Lyme disease, 244, 260, 262–263
- Lymphocytic leukemia, acute, 271, 282
- Lymphogranuloma venereum (LGV), 341
- M**
- Mallory-Weiss syndrome, 182
- Malrotation, intestinal, 157, 173, 197
- Mantoux (PPD) test, 133, 145–146, 242, 260–261
- Marasmus, 51, 176
- Marfan syndrome, 26, 52, 123, 128, 300, 311–312, 318–319
- McCune-Albright syndrome, 219, 231, 232
- Measles (*see* Rubeola)
- Meckel diverticulum, 156, 171–173, 175–177
- Meconium, 85, 107, 108, 174
- Mediastinal mass, with acute lymphocytic leukemia, 271, 282
- Medulloblastomas, 224
- Melena, 92
- Meningitis, 81, 105, 254, 263  
and lumbar puncture, 246, 264  
neonatal, 238, 256  
viral, 253
- Meningococemia, 245, 263–264
- Menkes' syndrome, 317
- Metabolic acidosis, 90, 141, 142, 152–153
- Metabolic alkalosis, 141, 142, 152, 153  
from pyloric stenosis, 174
- Metachromatic leukodystrophy, juvenile, 307
- Methotrexate, toxicities of, 274, 285
- Microspherocytes, 276, 286, 287
- Migraines  
“common,” 210, 223–224  
hemiplegic, 222
- Milia, 23, 24, 48, 49
- Mitral valve prolapse, 118, 124, 127–128
- Molluscum contagiosum, 31, 57
- Mongolian spots, 70, 98–99, 220
- Mononucleosis, infectious, 237, 255
- Moraxella catarrhalis*, 147
- Morquio syndrome, 312
- Mucositis, 285
- Mumps, 233, 241, 250, 258–259
- Muscular atrophy, spinal, 226
- Muscular dystrophy  
Duchenne, 216, 226, 228–229  
myotonic, 210, 223
- Myasthenia gravis, 209, 221–222, 226
- Mycobacterium tuberculosis*, 145, 149
- Mycoplasma pneumoniae*, 137, 149, 249, 267, 268
- Myocarditis, 116, 125, 129
- Myoglobin, in urine, 190, 202–203
- N**
- N-acetylcysteine (NAC), 27, 28, 53, 54
- Naloxone, 27, 28, 53, 54, 67, 96–97
- Narcotics  
poisoning from, 28, 54  
use during childbirth, 67, 96–97  
use during pregnancy, 80, 104
- Nasopharyngeal suction, 68, 97
- Necator americanus* (*see* Hookworms)
- Necrotizing enterocolitis (NEC), 64, 92–93, 181–182
- Neisseria gonorrhoeae*, 340
- Neisseria meningitidis*, 254, 260, 263
- Nelson syndrome, 310
- Nephrotic syndrome, 185, 196–199, 206
- Neural tube defects, 104
- Neuroblastomas, 282
- Neurofibromatosis, 228  
type 1 (NF1), 219, 231–232
- Niacin  
deficiency of, 25, 26, 51  
toxicity from, 29, 55

- Niemann-Pick disease, 223  
  type A, 226, 307  
Nightmares, 24, 49, 50  
Night terrors, 24, 49, 50, 216, 217,  
  229–230  
Nikolsky sign, 36  
Nitroblue tetrazolium (NBT) test, 33, 59  
Nocturnal enuresis, 195, 205  
Nongonococcal urethritis (NGU),  
  339–340  
Nonsteroidal anti-inflammatory drugs  
  (NSAIDs), 197, 199  
Nose  
  bleeding from, 172  
  foreign bodies in, 134, 146
- O**  
Obstructive sleep apnea (OSA), 134, 146  
Occult spinal dysraphism, 208, 220  
Ochronosis, 316–317  
Oligohydramnios, 82, 106, 195, 205  
Oliguria, with acute glomerulonephritis,  
  206  
Ophthalmia neonatorum, 68, 97  
Orchiopexy, 201  
Orchitis, 259  
Osgood-Schlatter disease, 13, 41  
Osteitis fibrosa cystica, 200  
Osteoarthritis, 127  
Osteogenesis imperfecta, 301, 319, 320  
Osteomyelitis, 27, 53  
  acute, 237, 255–256  
Otitis media, 76, 102, 236, 254  
Oxygen therapy, 139, 150
- P**  
Pampiniform venous plexus, 204  
Pancreatic enzyme replacement, for CF,  
  156, 172–173  
Pancreatitis, 160, 174–175  
Panhypogammaglobulinemia (*see* Bruton  
  disease)  
Panhypopituitarism, 316  
Parinaud oculoglandular syndrome, 266  
Parovirus B-19, 235, 253  
Paroxysmal supraventricular tachycardia,  
  117  
*Pasteurella multocida*, with animal bites,  
  46  
Patent ductus arteriosus, 257  
Pauciarticular rheumatoid arthritis, 2,  
  35–36  
Pedestrian injuries, 25, 50–51  
Pediculosis pubis, 204  
Pelger-Huët anomaly, 286  
Pellagra, 51  
Pelvic inflammatory disease (PID), 332,  
  340–341  
Penicillamine, 199  
Penicillin, 144  
  maternal use of, 101  
  with mitral valve prolapse, 118, 127,  
  128  
  with theophylline, 136, 149  
Peptic ulcers, 156, 171, 172, 182  
Permethrin  
  for treating head lice, 43  
  for treating scabies, 12, 31, 40, 56, 57  
Pertussis, 107  
  immunization against, 45, 237,  
  254–255  
  treatment with erythromycin, 237,  
  254, 255  
Pharyngitis, streptococcal, 252–253  
Phenindione, and urine discoloration,  
  202  
Phenolphthalein, and urine discoloration,  
  202  
Phenylketonuria, 86, 108, 109, 221  
Phenytoin  
  birth defects from, 30, 44, 55, 56, 101  
  and low folate levels, 32, 58  
  with theophylline, 149  
Pinworms, 171, 262, 264–265  
  cellophane-tape collection of, 249,  
  267  
  vaginal, 204  
Pityriasis rosea, 8–10, 38, 228  
Platelet count, testing for, 33, 59  
Pleocytosis, 224

- Pneumonia  
 from *Chlamydia trachomatis*, 97, 249, 267–268  
 group B streptococcus, 13, 40–41  
 from *Mycoplasma pneumoniae*, 137, 149, 249, 267, 268  
 necrotizing, 38  
 from *Pneumocystis carinii*, 233, 250–251  
 recurrent, 140, 151–152  
 from *Staphylococcus aureus*, 249, 267, 268  
 from *Streptococcus pneumoniae*, 147, 149, 254, 258  
 tuberculous, 249, 267, 268
- Poisoning, 25, 51  
 from analgesics, 28, 54  
 from alkali, 7, 37, 167, 180–181  
 from arthropod bites and stings, 218, 230  
 from digitalis, 113, 121  
 from hydrocarbons, 38, 132, 144  
 from lead, 21, 27, 46–47, 53–54  
 from methanol, 29, 55  
 from organophosphates, 11, 28, 40, 54  
 from phenothiazine, 29, 54  
 from tricyclic antidepressants, 21, 46  
 from vitamins, 29, 54–55
- Poison Prevention Packaging Act of 1970, 51
- Polycystic diseases, of kidneys and liver, 303, 304, 323
- Polycythemia, 67, 96
- Polydactyly, fifth-finger, 79, 104
- Polydipsia, 198
- Pontine glioma, 211, 224
- Posterior urethral valves, 189, 199–200
- Postmature infants, complications in, 68, 97
- Poststreptococcal glomerulonephritis, 190, 201–204
- Prader-Willi syndrome, 26, 52, 302, 321, 324
- Pralidoxime (2-PAM), 27, 28, 53, 54
- Prednisone, side effects of, 285
- Pregnancy  
 teenage, 330, 339  
 use of alcohol during, 30, 56  
 use of medications during, 64, 73, 92, 100–101, 279
- Prehn sign, 340
- Preterm infants, complications in, 65, 93, 94, 105, 180, 300, 319
- Prolonged QT syndrome, 120, 129
- Propranolol, maternal use of, 73, 100, 101
- Proteinuria, 206
- Prune belly syndrome, 186, 197
- Pseudohypoparathyroidism, 297, 301, 302, 315, 319–322
- Pseudomonas*, 248, 266–267
- Pseudotumor cerebri, 224
- Psoriasis, 39, 228
- Puberty  
 and bone age, 313, 314  
 delayed, 290, 308, 312  
 first signs of, 304, 305, 324–325  
 true precocious, 290, 307–308
- Pulmonary hypoplasia, 205
- Pulmonic atresia, 127
- Pupils, 209, 222
- Pustular melanosis, 23, 48, 49
- Pyelonephritis, 202
- Pyloric stenosis, 159, 173–174, 316
- Pyridoxine  
 deficiency of, 32, 58  
 toxicity from, 29, 55
- R**
- Raynaud phenomenon, 203
- Reflux, urine, 188, 199
- Renal failure, chronic, 200
- Renal osteodystrophy, 200
- Renal tubular acidosis, 199
- Respiratory acidosis, 141, 142, 152–153
- Respiratory alkalosis, 153  
 paper-bag technique for, 141, 152
- Respiratory syncytial virus (RSV), 107, 131, 143
- Retinoblastomas, 296, 313–314

- Rett syndrome, 227  
Reye syndrome, 170, 183, 252  
Rheumatic fever, 113, 114, 121  
Rickets, 200  
    Vitamin D–deficient, 52, 170, 183, 299, 318  
    Vitamin D–resistant, 301, 303, 319–320, 323  
Ringer lactate, IV fluid replacement with, 44, 179  
Ritter disease, 3, 36  
*Rochalimaea henselae*, 248, 266  
Rocky Mountain spotted fever (RMSF), 242, 260, 263–264  
Romano-Ward syndrome, 129  
Roseola, 253, 257, 258, 266  
Rubella, 241, 250, 258  
    birth defects from, 238, 257  
    immunization against, 310  
Rubeola, 240, 250, 257, 258, 262  
    atypical, 260
- S**  
Sacral agenesis, 30, 55–56  
Salicylates  
    poisoning from, 28, 54  
    for treating rheumatic fever, 114, 121, 122  
Saline, IV fluid replacement with, 19, 44  
    for treating hypernatremia, 164, 178, 179  
*Salmonella sonnei*, 264  
Salmon patches, 23, 48, 49  
Sandhoff disease, 226, 307  
Scabies, 12, 31, 40, 57  
Schmidt syndrome (*see* Type II polyglan-  
    dular autoimmune disease)  
Scoliosis, idiopathic, 330, 338  
Scorpion stings, 218, 230  
Scurvy, 182  
Sebaceous nevi, 23, 48, 49  
Seizures, 223  
    differential diagnosis of, 48  
    febrile, 213, 227  
    grand mal, 85, 109, 110  
Sepsis, neonatal, 62, 89–90, 105, 106, 200  
    from short gut syndrome, 158, 173  
Serum amylase, 160, 174, 175  
Serum immunoglobulin levels, testing  
    for, 33, 59  
Sexual abuse, 45, 204  
Sexually transmitted diseases (STDs), 202, 331–333, 339–341  
Shigellosis, 234, 252  
Short bowel syndrome, 184  
Short gut syndrome, 158, 173  
Sickle cell disease, 94, 160, 283  
    anemia from, 33, 59, 253, 279  
    respiratory complications of, 138, 139, 150  
    splenic complications of, 271, 277, 280–281, 286  
Sinusitis, 136, 147–148  
6-mercaptopurine, side effects of, 285  
Skull fractures, basilar, 207, 220  
Sleep disturbances, 24, 25, 49–50  
Sleepwalking (*see* Somnambulism)  
Slipped capital femoral epiphysis, 27, 41, 53  
Small-for-dates infants, complications in, 65, 94  
Small-for-gestational-age (SGA) infants, complications in, 67, 96, 300, 319  
Somnambulism, 50, 229  
Somniloquy, 24, 49, 50  
Sorbitol-MacConkey media, 168, 181, 182  
Spherocytosis, hereditary, 273, 276, 277, 283, 286, 287  
Spider bites, 218, 230  
Sporotrichosis, 262, 265  
Staphylococcal scalded skin disease (*see* Ritter disease)  
*Staphylococcus aureus*, 149, 151, 267  
    as cause of pneumonia, 249, 267, 268  
    with human bites, 38, 46  
    with impetigo, 57  
    with lung abscesses, 144  
    with osteomyelitis, 255  
    with septic arthritis, 45

- Steroids  
 anabolic, 327, 335  
 for treating esophageal inflammation, 181  
 for treating rheumatic fever, 114, 121, 122
- Stevens-Johnson syndrome, 243, 261
- Strabismus, 14, 41–42
- Streptococcosis, 257
- Streptococcus pneumoniae*, 147, 149, 254, 258, 281
- Streptococcus viridans*, 38, 46
- Sturge-Weber syndrome, 85, 110
- Subdural hematomas, 110, 211, 225
- Subendocardial fibroelastosis, 259
- Sucrose, 177
- Sudden infant death syndrome (SIDS), 37  
 prevention of, 21, 42, 47
- Suicide attempts, 328, 329, 335–338
- Sulfamethoxazole, combined with trimethoprim  
 for PCP prophylaxis, 250–251  
 and treatment of shigellosis, 252
- Sulfisoxazole, 89–90
- Supraventricular aortic stenosis, 119, 128
- Sweat chloride test, for CF, 132, 143, 144
- Syphilis, 339, 340  
 congenital, 78, 103–104, 228  
 testing for, 333, 341
- Syrup of ipecac, contraindications to, 7, 37–38
- Systemic lupus erythematosus (SLE), 191, 203, 204, 279, 307, 323
- T**
- Takayasu arteritis, 204
- Target cells, 276, 277, 286, 287
- Tay-Sachs disease, 209, 223, 227
- Teeth  
 avulsion of, 166, 180  
 decay of (caries), 165, 179  
 primary, eruption of, 164, 179
- Tension pneumothorax, 83, 84, 106, 107, 134, 146
- Testicular feminization, 299, 317, 318
- Testicular torsion, 191, 203, 254, 340
- Tetanus, immunization against, 20, 45, 46, 180
- Tetracycline, 281
- Tetralogy of Fallot, 124, 126–128, 152
- Thalassemia major, 276, 277, 286–287
- Theophylline, therapeutic versus toxic levels of, 136, 148–149
- Thiamine, deficiency of, 25, 26, 51, 52
- Thoracentesis, 141, 152
- Thrombocytopenia, 278–279
- Thrombotic thrombocytopenic purpura, 278
- Thyroid gland  
 dysgenesis of, 95, 294, 313  
 medullary carcinoma of, 301, 319–321
- Thyroiditis, 291, 292, 309–310
- Thyrotoxicosis, neonatal, 290, 307
- Tick bites, paralysis from, 230
- Tinea capitis, forms of, 253
- Todd's paralysis, 222
- Tourette syndrome, 218, 219, 230, 231
- Tourniquet therapy, 137, 149
- Toxic shock syndrome (TSS), 260
- Toxic synovitis, 127
- Toxocara canis*, 135, 147, 262
- Toxoplasma gondii*, acquired, 265
- Toxoplasmosis, congenital, 227–228, 265
- Tracheoesophageal fistula, 106
- Transient synovitis, 27, 53
- Transient tachypnea, 76, 85, 103, 107–108
- Transient tic disorder, 218, 230–231
- Transposition of the great vessels, 118, 127
- Treacher Collins syndrome, 26, 52, 53
- Treponema pallidum*, 341
- Trichinella spiralis*, 244, 262
- Trichophyton tonsurans*, 235, 253
- Tricuspid  
 atresia, 118, 127  
 regurgitation, 123–124, 126
- Trimethadione, 185, 197  
 combined with sulfamethoxazole, 250–252
- Trisomy 21, 89

- Tuberculosis (TB), 133, 144–145, 249, 268  
skin test for, 145  
Tuberculous meningitis, 210, 211, 224–225  
Tuberous sclerosis, 87, 110, 215, 219, 224, 228, 231, 232  
Tularemia, 260  
Tumors, brain, 224, 225  
infratentorial, 212, 226  
supratentorial, 222–223  
Tunica vaginalis, 205  
Turner syndrome, 69, 98, 123, 128, 305, 309, 325  
21-hydroxylase deficiency, 223, 298, 302, 303, 314, 316, 322  
Twins, monochorionic, 81, 105–106  
Type I GM<sub>1</sub> (*see* Gangliosidosis type I, generalized)  
Type II polyglandular autoimmune disease, 309  
Typhoid fever, 257  
Tyrosinosis, 306  
Tzanck smear, 249, 267, 341
- U**  
Ultrasound, renal, 195, 205  
Undescended testes (*see* Cryptorchidism)  
Ureters, injuries to, 197–198  
Urinary tract  
infections of, 200, 203, 340  
injuries to, 186, 197–198  
Urine  
discoloration of, 190, 201–203  
reflux of, 188, 199  
specimens, contamination of, 190, 201
- V**  
Vagina  
discharge from, 192, 203–204  
foreign bodies in, 45  
labial adhesions, 293, 311  
pinworms in, 204  
Valproate, birth defects from, 30, 55, 56  
Varicella, 62, 90, 250  
Varicocele, 193, 204  
VATER syndrome, 99  
Ventricular septal defects, 23, 32, 57–58, 123, 128, 257  
Vincristine, toxicities of, 285  
Viral croup, 136, 148  
Vitamin A  
deficiency of, 32, 58, 169, 170, 182, 183  
toxicity from, 29, 55  
Vitamin B<sub>1</sub> (*see* Thiamine, deficiency of)  
Vitamin B<sub>6</sub> (*see* Pyridoxine)  
Vitamin B<sub>12</sub>, 281  
Vitamin C  
deficiency of, 169, 182  
disease-prevention supplementation of, 251  
toxicity from, 29, 55  
Vitamin D  
deficiency of, 25, 26, 32, 33, 51, 52, 58, 59, 169, 170, 182, 183, 299, 318  
toxicity from, 29, 55  
Vitamin E, deficiency of, 169, 182  
Vitamin K, deficiency of, 32, 33, 58, 59, 72, 99–100, 169, 182, 284  
Von Willebrand disease, 278  
Vulvovaginitis, 19, 45, 192, 203–204
- W**  
Waardenburg syndrome, 26, 52, 87, 110  
Waterhouse-Friderichsen syndrome, 263  
Werdnig-Hoffmann disease, 229  
Williams syndrome, 128  
Wilms tumor, 86, 87, 109–110, 271, 282  
Wilson disease, 298, 317  
Wiskott-Aldrich syndrome, 59, 265  
Wolff-Parkinson-White (WPW) syndrome, 125, 126  
Wolman syndrome, 184  
Wood's lamp, 249, 267
- X**  
XO karyotype, 115, 123