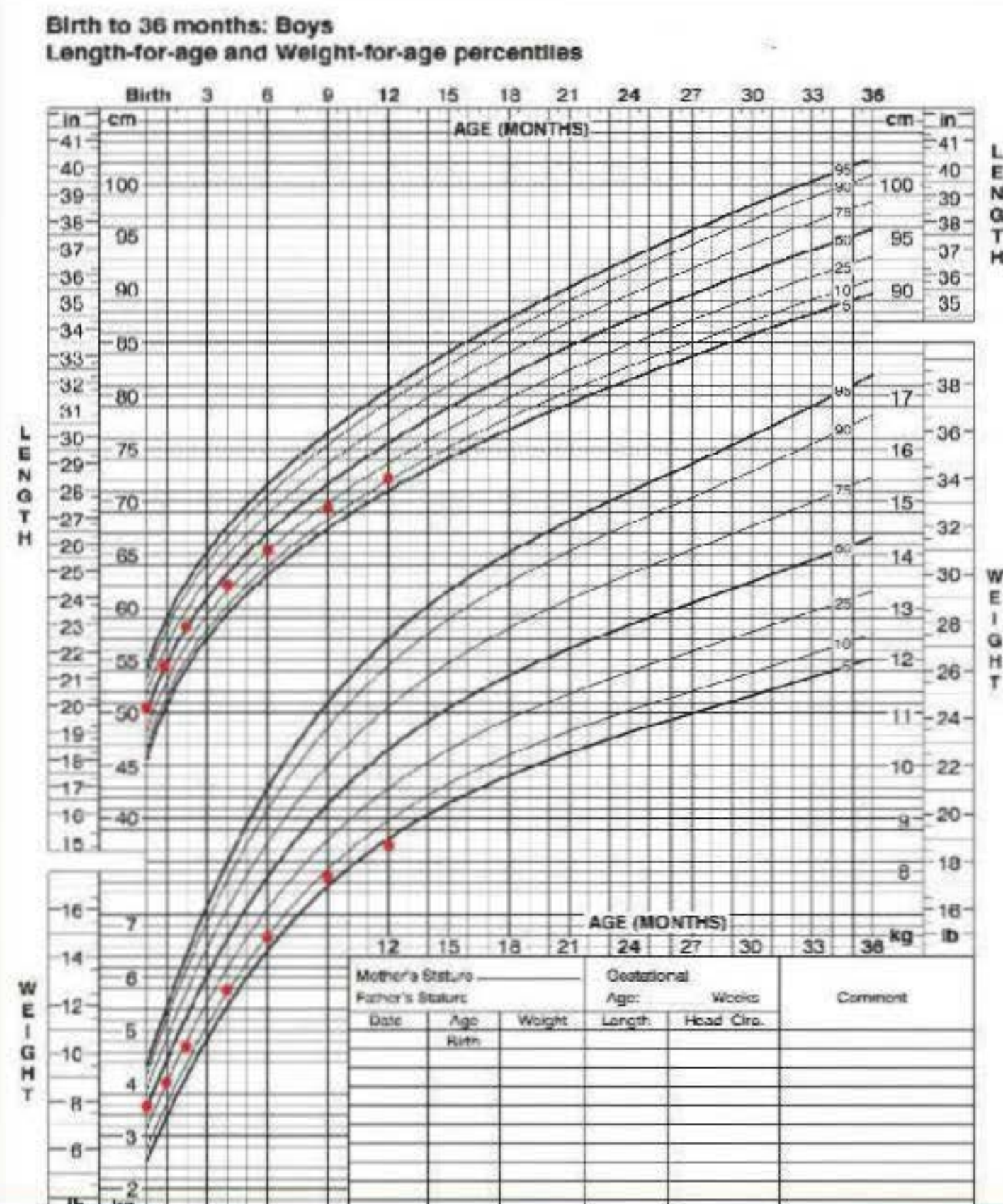
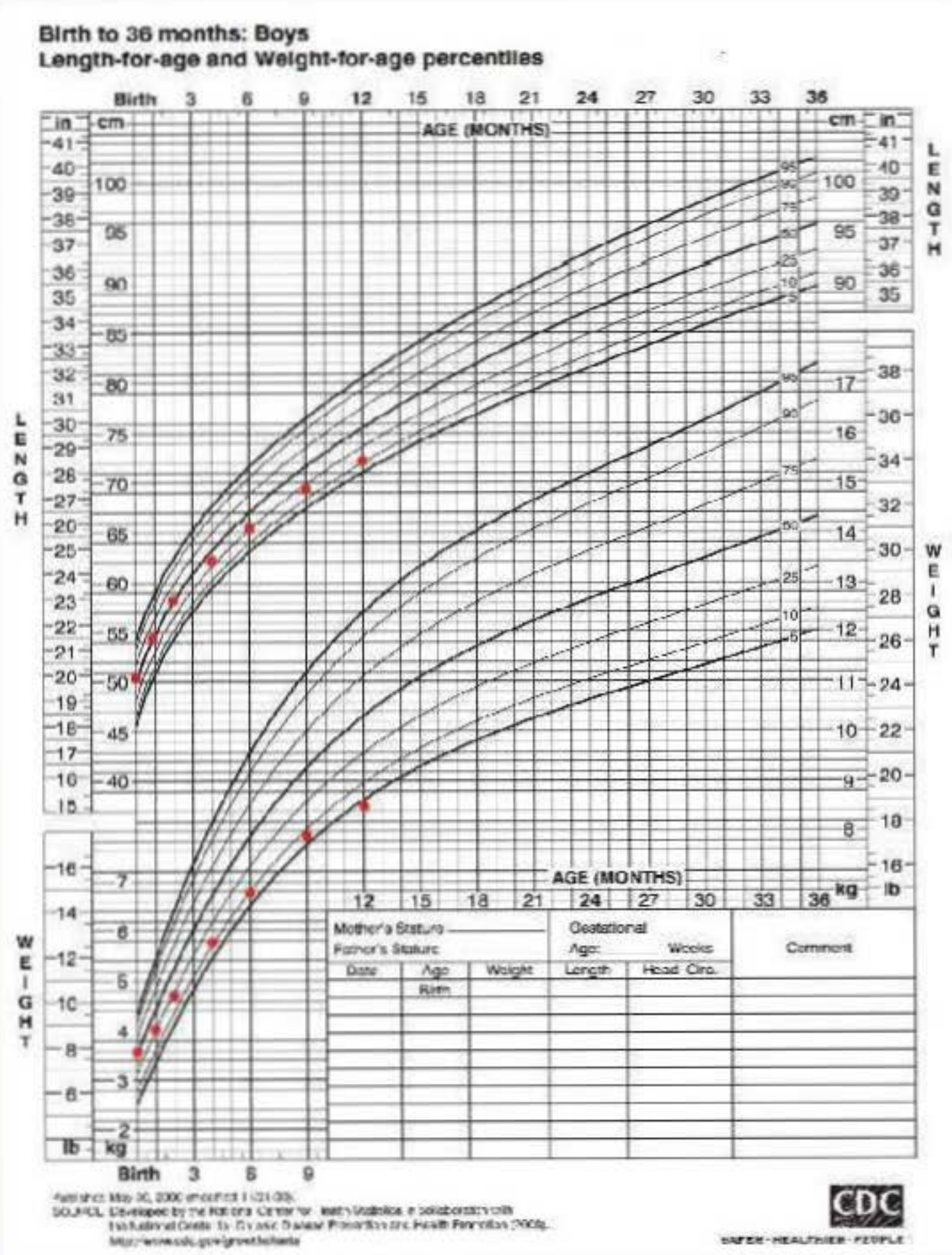


A 1-year-old boy is brought to the clinic by his mother due to poor weight gain. She is concerned that the patient is smaller than children of the same age at his day care. Since birth, the mother has trialed various infant formulas due to recurrent "greasy" and loose stools. The patient recently started drinking cow's milk; he drinks about 20 oz of whole milk and eats table food such as meats, fruits, and grains 3-4 times each day. He also has had recurrent nosebleeds over the past month. The patient's medical history is notable for 3 prior hospital admissions for bronchiolitis, and his mother states that his cough persisted for a month after his last discharge. He and his mother live with her boyfriend, who smokes cigarettes. The patient's growth chart is shown below.





notable for 3 prior hospital admissions for bronchiolitis, and his mother states that his cough persisted for a month after his last discharge. He and his mother live with her boyfriend, who smokes cigarettes. The patient's growth chart is shown below.



Physical examination shows a thin child with dried blood on his nasal turbinates. Bilateral wheezes are heard on chest auscultation. Scattered ecchymoses are noted on his extremities. What is the most likely cause of this patient's failure to thrive?











Physical examination shows a thin child with dried blood on his nasal turbinates. Bilateral wheezes are heard on chest auscultation. Scattered ecchymoses are noted on his extremities. What is the most likely cause of this patient's failure to thrive?

- ☐ A. Autoimmunity to gluten [6%]
- ☐ B. Child abuse [8%]
- ☐ C. Constitutional growth delay [1%]
- ☐ D. Inadequate caloric intake for age [1%]
- ☐ E. Lactose intolerance [3%]
- ☒ F. **Pancreatic insufficiency** [81%]

Proceed to Next Item

Explanation:

User Id: [REDACTED]

Clinical features of cystic fibrosis	
Respiratory	<ul style="list-style-type: none"> <li>• Obstructive lung disease → bronchiectasis</li> <li>• Recurrent pneumonia</li> <li>• Chronic rhinosinusitis</li> </ul>
Gastrointestinal	<ul style="list-style-type: none"> <li>• Obstruction (10%-20%)                             <ul style="list-style-type: none"> <li>◦ Meconium ileus</li> <li>◦ Distal intestinal obstruction syndrome</li> </ul> </li> <li>• Pancreatic disease                             <ul style="list-style-type: none"> <li>◦ Exocrine pancreatic insufficiency</li> <li>◦ CF-related diabetes (~25%)</li> </ul> </li> <li>• Biliary cirrhosis</li> </ul>
Reproductive	<ul style="list-style-type: none"> <li>• Infertility (&gt;95% men, ~20% women)</li> </ul>
Musculoskeletal	<ul style="list-style-type: none"> <li>• Osteopenia → fractures</li> <li>• Kyphoscoliosis</li> <li>• Digital clubbing</li> </ul>

©UWorld



## Musculoskeletal

- Osteopenia
- Kyphoscoliosis
- Digital clubbing

©UWorld

**Cystic fibrosis (CF)**, the most common autosomal recessive disorder in those of white ethnicity, is characterized by defective chloride transport resulting in viscous secretions in the lungs, sinuses, and pancreas. If not identified by newborn screening or meconium ileus at birth, growth failure and recurrent sinopulmonary infections typically raise concern for CF, as in this patient.

The most common gastrointestinal manifestation of CF is **pancreatic insufficiency**. Pancreatic duct obstruction and distension due to viscous mucus and subsequent inflammation develop in utero, eventually leading to fibrosis. Pancreatic insufficiency is present from birth in most patients with CF and results in inability to absorb fats and fat-soluble vitamins (A, D, E, and K), leading to **steatorrhea** (greasy, foul-smelling stools), **failure to thrive**, and **vitamin deficiencies**. Vitamin K deficiency causes prolonged prothrombin time with easy mucosal bleeding (eg, epistaxis) and bruising.

The growth chart in infants with CF shows normal birth measurements with subsequent deceleration in weight (decline of  $\geq 2$  major percentiles) followed by deceleration in length. In addition to malabsorption, the recurrent sinopulmonary infections impair growth due to poor appetite and increased metabolic needs during illness.

**(Choice A)** Celiac disease, an autoimmune intolerance to gluten, can result in failure to thrive due to intestinal inflammation, malabsorption, and malnutrition. However, celiac disease is not typically associated with respiratory symptoms.

**(Choice B)** Although child abuse can present with growth failure (eg, neglect) and bruising, this diagnosis is less likely given the patient's chronic cough and steatorrhea. In addition, bruises on the extremities in active infants are less concerning for abuse than those that occur in central areas (eg, back, buttocks).

**(Choice C)** Constitutional growth delay presents with a decelerated velocity of height growth in infancy. These patients experience normalization of height growth rate after age 2-3 but have short stature and delayed puberty until catch-up growth after puberty. These children are otherwise asymptomatic.

**(Choice D)** This patient's intake is adequate for a healthy 1-year-old, but he will likely need caloric supplementation due to increased metabolic demand from recurrent illnesses.

**(Choice E)** Lactose intolerance is characterized by abdominal cramping, bloating, and



bruising.

The growth chart in infants with CF shows normal birth measurements with subsequent deceleration in weight (decline of  $\geq 2$  major percentiles) followed by deceleration in length. In addition to malabsorption, the recurrent sinopulmonary infections impair growth due to poor appetite and increased metabolic needs during illness.

**(Choice A)** Celiac disease, an autoimmune intolerance to gluten, can result in failure to thrive due to intestinal inflammation, malabsorption, and malnutrition. However, celiac disease is not typically associated with respiratory symptoms.

**(Choice B)** Although child abuse can present with growth failure (eg, neglect) and bruising, this diagnosis is less likely given the patient's chronic cough and steatorrhea. In addition, bruises on the extremities in active infants are less concerning for abuse than those that occur in central areas (eg, back, buttocks).

**(Choice C)** Constitutional growth delay presents with a decelerated velocity of height growth in infancy. These patients experience normalization of height growth rate after age 2-3 but have short stature and delayed puberty until catch-up growth after puberty. These children are otherwise asymptomatic.

**(Choice D)** This patient's intake is adequate for a healthy 1-year-old, but he will likely need caloric supplementation due to increased metabolic demand from recurrent illnesses.

**(Choice E)** Lactose intolerance is characterized by abdominal cramping, bloating, and diarrhea after ingestion of lactose-containing products. It is uncommon in children age  $<6$  and is not associated with respiratory problems.

**Educational objective:**

Growth failure and recurrent respiratory infections in infants should raise concern for cystic fibrosis. Pancreatic enzyme deficiency causes steatorrhea and fat-soluble vitamin malabsorption, resulting in poor weight gain.

**References:**

1. Evolution of pancreatic function during the first year in infants with cystic fibrosis.
2. Clinical presentation of cystic fibrosis at the time of diagnosis: a multicenter study in a region without newborn screening.
3. Growth failure in children with cystic fibrosis.