

A 3-year-old boy is brought to the office by his parents for the evaluation of dry eyes and photophobia. He has some difficulty in adapting to darkness. He is a very poor eater, and his diet consists mainly of canned foods, and very rarely, fresh vegetables or milk. Examination reveals dry, scaly skin, follicular hyperkeratosis in the extensor surfaces of the extremities, and dry, silver-gray plaques on the bulbar conjunctiva. What is the most likely diagnosis of this patient?

- ☐ A. Vitamin A deficiency
- ☐ B. Thiamine deficiency
- ☐ C. Ariboflavinosis
- ☐ D. Scurvy
- ☐ E. Hypervitaminosis A

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- ☒ A. Vitamin A deficiency [92%]
- ☐ B. Thiamine deficiency [1%]
- ☐ C. Ariboflavinosis [1%]
- ☐ D. Scurvy [3%]
- ☐ E. Hypervitaminosis A [2%]

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Explanation:

User Id: [REDACTED]

This child's presentation is classic for Vitamin A deficiency. The condition usually manifests in the second or third year of life as impaired adaptation to darkness (which may progress to night blindness), photophobia, dry scaly skin, dry conjunctiva (xerosis conjunctiva), dry cornea (xerosis cornea) and a wrinkled, cloudy cornea (keratomalacia). Bitot spots (dry, silver-gray plaques on the bulbar conjunctiva) and follicular hyperkeratosis of the shoulders, buttocks, and extensor surfaces are less common findings.

(Choice B) Thiamine deficiency is associated with infantile and adult beriberi, as well as Wernicke-Korsakoff syndrome in alcoholics. Manifestations of infantile beriberi appear between the ages of two and three months and include a fulminant cardiac syndrome with cardiomegaly, tachycardia, cyanosis, dyspnea, and vomiting. Adult beriberi is categorized as dry or wet. Dry beriberi describes a symmetrical peripheral neuropathy accompanied by sensory and motor impairments, especially of the distal extremities. Wet beriberi includes this neuropathy in addition to cardiac involvement (eg, cardiomegaly, cardiomyopathy, congestive heart failure, peripheral edema, tachycardia).

(Choice C) Riboflavin is an integral component of coenzymes that participate in multiple cellular metabolic pathways (eg, energy producing respiration). This vitamin is present in many foods, including meats, fish, eggs, dairy products, green vegetables, yeast, and enriched foods. Pure riboflavin deficiency is therefore unusual in industrialized nations, but the condition has been documented in regions of the world with severe food shortages. In the United States, patients who present with riboflavin deficiencies are

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(Choice D) Ascorbic acid deficiency is characterized by impaired collagen synthesis and damaged connective tissue. The more common symptoms include ecchymoses, petechiae, bleeding gums, hyperkeratosis, Sjogren's syndrome, arthralgias, and impaired wound healing. Systemic manifestations include weakness, malaise, joint swelling, arthralgias, edema, coiled hair, depression, neuropathy, and vasomotor instability. This condition may also present with dry skin and eyes due to hyperkeratosis and keratoconjunctivitis sicca; however, the other features in this case are diagnostic for Vitamin A deficiency.

(Choice E) Hypervitaminosis A is usually due to the ingestion of excessive doses of vitamin A for several weeks or months. Symptoms include anorexia, pruritus, lack of weight gain, increased irritability, limitation of motion, tender swelling of the bones, alopecia, seborrheic cutaneous lesions, fissuring of the corners of the mouth, increased intracranial pressure, and hepatomegaly.

Educational Objective:

Suspect vitamin A deficiency in a 2 or 3-year-old child with impaired adaptation to darkness, photophobia, dry scaly skin, xerosis conjunctiva, xerosis cornea, keratomalacia, Bitot spots and follicular hyperkeratosis of the shoulders, buttocks, and extensor surfaces.