Denouement and Discussion

Nutritional Rickets

Nutritional rickets has a prevalence rate between 5% and 10% in developing countries.[1,2] By the mid 1900s, nutritional rickets was virtually eliminated in the United States. However, a recent reemergence has been witnessed by health care providers in the United States and other developed countries.[3-6] Because rickets is not a reportable disease, the exact incidence in US children is unknown.

Rickets occurs when calcium and/or phosphorus deficiency results in inadequate bone mineralization. Both 1,25-dihydroxyvitamin D and parathyroid hormone are essential to calcium and phosphorus homeostasis. The 1,25-dihydroxyvitamin D increases calcium and phosphorus absorption from the gastrointestinal tract. Parathyroid hormone increases the serum calcium level by bone resorption and decreases the serum phosphorus level by increased urinary excretion. Together, these mechanisms maintain serum calcium and phosphorus concentrations at the expense of bone mineralization.[7] Rickets occurs in patients with disorders in calcium, phosphorus, and vitamin D metabolism.

Nutritional rickets occurs when 1,25-dihydroxyvitamin D, calcium, or both are deficient. Because 25-hydroxyvitamin D is the direct precursor to 1,25-dihydroxyvitamin D, 25-hydroxyvitamin D deficiency is a direct cause of rickets. Deficiency of 25-hydroxyvitamin D is the most common cause of nutritional rickets and is associated with infants and toddlers who have dark skin, limited sunlight exposure, prolonged or exclusive breastfeeding, diets with minimal dairy intake, and improper vitamin D supplementation.[3-5]

CLINICAL FEATURES

Most of the clinical manifestations of rickets occur secondary to either impaired mineralization of the bony matrix or hypocalcemia. Skeletal findings include the following:

- Craniotabes (softening of the skull and occipital flattening)
- Delayed fontanelle closure
- Frontal bossing
- Rachitic rosary (enlargement at the costochondral junction)
- Harrison sulcus (groove in the lower ribs where the diaphragm attaches)
- Scoliosis and/or kyphosis
- Genu varum or genu valgum (in ambulatory patients)
- Bowing of the radius and ulna (in nonambulatory patients)
- Saber shin deformity (anterior bowing of the tibia)
- Thickened wrists and ankles
- Bone pain and tenderness
- Hypotonia

Extraskeletal features include the following:

- Late teeth eruption and poor enamelization of teeth
- Seizures and/or tetany (related to hypocalcemia)
- Failure to thrive or short stature
- Developmental delay
- Secondary immunodeficiency

Laboratory evaluation reveals a significantly elevated alkaline phosphatase level in all types of rickets. The calcium (total and ionized) and phosphorus levels are frequently decreased whereas the parathyroid hormone level is frequently increased in hypocalcemic rickets. The 25-hydroxyvitamin D level is decreased in vitamin D deficiency and hypocalcemic rickets.

Radiographic findings are noted in the growth plates of long bones, especially the distal ulnar, radius, and femur and the proximal tibia. In these locations, metaphyseal widening, cupping, fraying, and flaring are common. Bowing of the upper and lower extremities, depending on the weight-bearing status of the patient, is another frequent radiographic finding. Cortical thinning and osteopenia of the long bone diaphyses can be seen.

DIFFERENTIAL DIAGNOSIS

The etiology of rickets includes decreased intake of calcium, vitamin D, and phosphorus, poor absorption of minerals from hepatic and pancreatic insufficiency, renal insufficiency, interference with absorption from medications (antacids and antiepileptics), increased renal excretion of phosphorus and calcium, and alkaline phosphatase deficiency. Other rare causes of metabolic bone disease must be considered in the differential diagnosis of rickets. Additionally, Blount disease, neurofibromatosis type I, fibular hemimelia, and congenital lower extremity anomalies can result in genu varum and other bowing deformities.

TREATMENT

To prevent vitamin D deficiency, it is recommended that all infants who do not ingest a minimum of 500 mL of vitamin D–containing formula or milk receive vitamin D supplementation. Breast milk contains minimal vitamin D and is not considered a vitamin D–containing milk. Therefore, exclusively breastfed infants require supplementation before 2 months of age. Additionally, children and adolescents who do not drink a minimum of 500 mL of vitamin D–containing milk, do not have adequate sunlight exposure, or do not receive a multivitamin with at least 200 IU of vitamin D require supplementation. The prophylactic dose of vitamin D is 200 IU daily.[8] To treat nutritional rickets, high doses of oral vitamin D (2000-10 000 IU of ergocalciferol daily) and calcium (300-1000 mg of elemental calcium daily) are suggested for 3 months. Consultation with an endocrinologist or other health care provider with an interest in bone disorders may be warranted.

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Because a positive correlation between maternal and child vitamin D status has been demonstrated,[9] especially with breastfed infants,[10] a diagnosis of rickets strongly suggests maternal vitamin D deficiency. If a child at high risk for vitamin D deficiency is diagnosed with rickets, routine management should include analysis of maternal vitamin D status and appropriate therapy for the mother.[11] A daily maternal intake of approximately 2000 to 4000 IU of vitamin D is recommended since this supplementation appears to safely provide sufficient vitamin D to enter the breast milk and increase the infant's circulating 25-hydroxyvitamin D level.[12] In our case, the mother was receiving no vitamin D supplementation despite limited sun exposure, darkly pigmented skin, and inadequate diet and had a decreased 25-hydroxyvitamin D level (27 nmol/L [optimal range, 62-200 nmol/L]).

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Announcement

Submissions. The Editors welcome contributions to Picture of the Month. Submissions should describe common problems presenting uncommonly, rather than total zebras. Cases should be of interest to practicing pediatricians, highlighting problems that they are likely to at least occasionally encounter in the office or hospital setting. High-quality clinical images (in either 35-mm slide or electronic format) along with parent or patient permission to use these images must accompany the submission. The entire discussion should comprise no more than 750 words. Articles and photographs accepted for publication will bear the contributor's name. There is no charge for reproduction and printing of color illustrations. For details regarding electronic submission, please see: http://archpedi.ama-assn.org.

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