

The bony manifestation of Rickets. A case report.

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Abstract

This paper describes a case of the bony manifestation of Rickets. Although vitamin D deficiency has become less common, Rickets still is seen among the Eritrean pediatric population. Rickets is an entity in which mineralization is decreased at the level of the growth plates, resulting in growth retardation and delayed skeletal development. The patient was referred to our Out patient Department for deformities of the legs and growth disturbance, with his height far below the normal range. Bowing of long bones of the legs and cupping of the wrist metaphyseal region were the radiographic changes most evident. Although the primary management of rickets is conservative, the orthopedist's role can be crucial. The orthopedist's responsibilities include establishing a diagnosis and treating osseous complications, such as fractures, slipped capital femoral epiphysis, and bowing of the extremities. Furthermore, the orchestration of care between the pediatrician and orthopedist is highlighted here.

Key words: Bony deformity, rickets, vitamin D deficiency, renal osteodystrophy

Introduction

Rickets is the bony manifestation of altered vitamin D, calcium, and phosphorous metabolism in a child; osteomalacia is the adult form. The earliest reports describing the syndrome of rickets appeared in the English literature around 1650, suggesting that the disease is an ancient one¹. There are multiple causes of rickets and osteomalacia, and the various metabolic defects in rickets, osteomalacia, and renal osteodystrophy². Because vitamin D deficiency has become less common in this country, rickets and osteomalacia are not often considered as differential diagnoses in patients who have extremity pain or deformity^{3,4}. However, the orthopedist should remain familiar with the roentgenographic and laboratory findings that accompany these diseases. By definition, rickets occurs only in children whose growth plates have not closed. Children with rickets are described as apathetic and irritable, often with a short attention span and are seemingly indifferent. They are content to sit for long periods of time in a "Buddha" or sitting posture, due to the bowing deformity of the lower extremities.^{2,5}

In patients with advanced rickets permanent skeletal deformities may occur. The relevant clinical findings are skull-craneotabes, rachitic rosary (the enlarged ends of the ribs), scoliosis, bowing of the long bones (caused by loading), greenstick fractures, thickening of the wrist and ankle, genu varum, genu valgum, and laxity of the ligaments.^{2,6} Plain radiographic findings include physical cupping and widening, transverse radiolucent (Looser's) lines, fraying of the metaphysis, flattening of skull, enlargement of the costal cartilages (rachitic rosary); and dorsal kyphosis.^{2,7,8} Additionally, there is a form of genetic rickets, called X-linked hypophosphatemic rickets, in which some children,

often girls, may be only moderately affected. However, girls with this disorder can have rickets symptoms that are just as severe as those in boys^{5,9}. The goal of this article is to demonstrate that although there are many osseous diseases that can appear in early ages, rickets remains a formidable yet treatable disease that should be considered in the differential within this country.

Case report:

A 2 year-old boy was referred to our Out Patient Department, because his parents had noticed knock-knees, and growth disturbance. The parents were of normal stature and build, and there was no consanguinity in the family history. The patient had no features of mal-absorption or chronic renal nor hepatic disease for the past one year. On the examination the patient had a bilateral genu varum, widened wrists and short stature, however his face, skull, spinal column, diaphragmatic insertion, dentition and costochondral junctions were normal. Hearing, vision, mental and motor development were normal. The results of biochemical investigations are summarized in Table 1: alkaline phosphatase, phosphorus and creatinine were elevated; hemoglobin and total iron were low. Calcium and Serum-Bun results were at the upper limits of borderline and vitamin D_{25-OH}, vitamin D_{1, 25-OH} and PTH were not measured due to laboratory limitations. X-ray showed widening and cupping and slight fraying of the metaphysical regions, knock-knees with the patient in standing position and the cupping of the metaphysis are more calcified than the metaphysis (osteopenia). This findings were suggestive of bony manifestations of rickets.

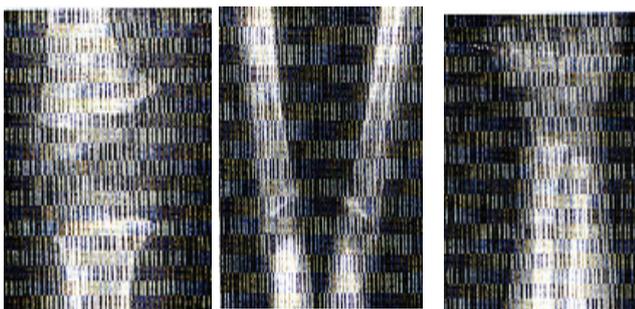
Table 1: Laboratory finding and reference ranges.

Parameter	Patient finding	Reference range
WBC	4.2x10 ³	4.5 x 10.5
Hgb	9.3 g/dl	11 - 18
Alkaline phosphatase	418 u/l	117 - 398
Bun - Serum	16 mg/dl	5 - 18
Creatinine - Serum	0.8 mg/dl	0.3 - 0.7
Calcium	10.2 mg/dl	8.8 - 11.8
Total Iron	30 mg/dl	45 - 160
Phosphorus	7.5 mmol/l	3.6 - 5.0



Deformity of the legs (genus varum)

Radiograph in a 4-year-old girl with rickets depicts bowing of the legs caused by loading and cupping of the metaphyseal area.



Cupping, slight fraying and osteopenia of the metaphyseal region are demonstrated here.

Discussion

The orthopedist's role in the diagnosis and treatment of the many disorders associated with rickets and renal osteodystrophy has shifted considerably with the understanding of the basic science of these entities. Pediatric orthopedic surgeons and pediatricians continue to be the principal diagnosticians dealing with children who have findings suggestive of rachitic disease¹⁰. The various forms of rickets should continue to be important elements of the differential diagnosis in children who present for evaluation with a bowed extremity, repeated fractures, abnormalities of the

spine, gait disturbances, diminished height, and failure to thrive. Radiography, radionucleotide imaging, and measurements of BUN, creatinine, calcium, phosphorus, alkaline phosphatase, 25-hydroxyvitamin D, 1,25-dihydroxyvitamin D, PTH, and a variety of urinary measurements, also including calcium, are invaluable aids in establishing the diagnosis of rickets and in elucidating the etiology of the disorder¹¹. If the pediatric orthopedist is making the diagnosis, communication with the primary pediatrician is then appropriate, after which further consultation may be obtained with specialists in pediatric nephrology or gastroenterology. 2.10 The term renal osteodystrophy is used to indicate a spectrum of bone disorders caused by damage to the glomerulus of the kidney. Characterized by hyperphosphatemia, elevated blood urea, uric acid, creatinine, low or normal calcium, reabsorption of the distal clavicle, brown tumors, slipped capital femoral epiphysis, ligament laxity, weakness, soft tissue calcification, low 1.25 dihydroxy Vitamin D, and high PTH^{12,13}. According to the biochemical results of this case, the patient's phosphorus is elevated which only occurs with renal osteodystrophy. In all other causes of rickets, the phosphorous is normal to low.

Management of the underlying metabolic disturbance is always the necessary first step, because it alone may be curative. Furthermore, the general health status of the individual depends on it, and orthopedic intervention without it will prove disappointing. Coordination of care between the pediatric orthopedic surgeon and the physician making the metabolic adjustments is important to ensure that metabolic response is appropriate. After the issue of nutrition is addressed, the issue of bracing arises. When treating patients with rickets, osteomalacia, and renal dystrophy, the orthopedist must always be concerned about the effect treatment may have on impaired calcium homeostasis.^{10,14,15}

The use of bracing to change the morphology of the skeleton usually generates some controversy. For physiologic alignment changes, there is little need to brace, and what in the past had been interpreted as a response to brace treatment is generally regarded as the progress of normal development. For metabolic problems, the pediatric orthopedist can observe in the face of metabolic correction, bracing to counteract the deformity should be considered. Although uncommon in rickets, refractory cases may require surgery.^{10,16}

When patients receive adequate treatment, no mortality is associated with rickets per se, but can be associated with other concomitant diseases such as pneumonia, which can arise from lack of thoracic excursion due to poor bony thoracic formation. Before surgery, management of the metabolic defect with vitamin D, phosphorus, and calcium or other appropriate measures should be carried out for several months. If the disease is not controlled metabolically, the deformity probably will recur after corrective osteotomy. However, large-dose vitamin D treatment should be discontinued for at least 3 weeks before surgery because otherwise hypercalcemia is likely to

occur with immobilization. Treatment of the metabolic defect supplemented by corrective splinting or bracing was indicated here for correcting the deformity.^{10,17}

The requisites for surgery are a reasonable life expectancy, motivated patient and parents, demonstrated improvement of bone lesions on medical management, deformities that can be corrected with one or two orthopedic procedures, and the likelihood that the surgery will significantly reduce the patient's disability. The deformities that require surgical correction most often are genu varum and genu valgum. In genu varum usually the femur, tibia, and fibula are all deformed, an osteotomy of the tibia and the fibula usually is required. Sometimes osteotomy of the femur also is necessary. Osteotomies can be done bilaterally at one operation. In genu valgum most of the bowing usually is in the femur, and a severe deformity in older children and in adults can be corrected by supracondylar osteotomy of the femur.^{10,16,17}

In conclusion rickets still occurs in Eritrea and may present in an unusual manner. When the orthopedist is confronted with a bony deformity in a young patient, it is crucial to rule out rickets as a potential cause, as is it a very treatable disease. Furthermore, inability to properly diagnose rickets can result in incorrect management and treatment of other bony disorders, which may even prove dangerous or futile.

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