Case Report

Rickets, a reemerging disease in developed countries

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Abstract

Rickets is a skeletal disease of the growing child owing to defective mineralization of the bone tissue. It is a rare entity in developed countries, however, increases due to immigration. Most cases are due to vitamin D deficiency and they may be diagnosed by an exhaustive anamnesis, typical biochemical changes and radiological signs. Its suspicion and early diagnosis are essential for an early start of treatment.

We report a case of rickets in a young child with vitamin D deficiency, to remark the importance of this problem, which is currently reemerging in our environment. The child presented legs deformity and growth retardation. All the examinations performed, including blood tests, radiological studies and clinical history oriented to rickets. Adequate treatment with vitamin D and calcium was initiated just after the diagnosis and the child improved remarkably, both growth and skeletal deformities.

Keywords: rickets, vitamin D, vitamin D deficiency

Introduction

Rickets is a skeletal disease of the growing child. It is the result of defective mineralization of the bone tissue and it is associated with decreased serum calcium or phosphate levels. Rickets is a common disease in children in developing countries [1], however, it is exceptional in developed countries where there has been a reappearance of nutritional rickets in recent years due to immigration [2]. Early diagnosis plays a pivotal role to minimize morbidity by treating children before the first year of life, so it is essential to know the population groups at greatest risk of rickets to maintain a high level of suspicion in them [3]. Most cases are due to vitamin D deficiency and they may be diagnosed by history, characteristic biochemical tests (such as decreased vitamin D levels and increased serum parathyroid hormone and alkaline phosphatase) and classical radiological changes (early metaphyseal widening, deformities of long bones) [4]. Its management is based on restoring vitamin D and calcium levels. The response to vitamin D treatment in nutritional rickets is extraordinary, confirming the diagnosis [2].

Case

A two-and-a-half-year-old Arabian girl was attended in her Health Center because her family observed that she walked with bowed legs.

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As a personal history, she was admitted to the Neonatal Unit due to neonatal hyperbilirubinemia that needed phototherapy treatment. She presented normal developmental milestones, except delayed walking at two years old. She was the only daughter of non-consanguineous parents without a family history of interest (not skeletal deformities, delayed dentition or renal diseases). To initiate the study, a nutritional survey was developed showing that the patient was breastfed until 12 months because she rejected the initiation of complementary feeding. After that, the intake had been very scarce. She had three meals per day consisting of milk, biscuits, vegetables, potatoes and very sporadic intake of animal protein (fish or chicken, once a week). Parents referred that the patient was a very active child that spent the most time at home, with limited sunlight exposure.

Physical examination showed weight and height below 3rd percentile and head circumference in the 25th percentile. Her legs were bowed in varus and she presented swelling of both wrists and ankles. During the examination, she stood up and walked fast despite legs deformity.

A blood test was performed showing normal calcium levels (9 mg/dL; normal 8-11 mg/dL), low phosphorus (2 mg/dL; normal 3.4-6.2 mg/dL) and raised alkaline phosphatase (944 IU/L; normal 0-280 IU/L). There were no alterations in liver and renal function. Intact parathyroid hormone (PTH) was 350 pg/mL (normal: 15-65 pg/mL) and 25-hydroxyvitamin-D level was decreased (<5 ng/mL; normal >20 ng/mL). She also presented iron deficiency anemia with lower hemoglobin (9.2 g/dL; normal 10.6-14.3 g/dL), mean corpuscular volume (60 fL, normal 73-89 fL) and ferritin (7 ng/mL; normal 11-140 ng/mL).

To complete the study, an x-ray was performed observing cupping and fraying of the metaphyseal regions of the radius, femur and tibia (Figure 1).

![Figure 1. X-ray of lower limbs where we can observe cupping and fraying of the metaphyseal regions of femur and tibia, such as the lateral widening of the metaphysis with growth plate expansion and bowing deformities of long bones.](image)

Regarding all the previous examinations, she was diagnosed with vitamin D deficiency rickets and initiated oral vitamin D as well as oral iron. Vitamin D was also administered intramuscularly in a single dose of 150,000 IU. Furthermore, parents were advised to increase foods rich in vitamin D in the diet as well as sunlight exposure. In a blood test performed two months after the starting treatment, alkaline phosphatase dropped to 436 IU/L, with normalization of phosphorus and vitamin D levels. Six months after treatment, alkaline phosphatase levels were in the normal range and remained normal in successive controls, with oral vitamin D supplementation at a dose of 600 IU per day. At this time, an X-ray showed almost complete resolution of the previous findings. Growth, height and weight improved, reaching the 10th percentile in a year and the deformity of the legs was slowly corrected during the follow-up.

**Discussion**

In recent decades, there has been an increase in the prevalence of rickets in developed countries due to immigration [5]. It is more prevalent in dark-skinned immigrants displaced to countries with less daily hours of sunlight exposure like our case [6]. Other risk groups are infants fed exclusively with breastfeeding, not supplemented with vitamin D, mothers with vitamin D deficiency, low-dairy diet and high in phytates, poor sunlight exposure, prematurity, low birth weight and liver or kidney disease [4]. Infants under one-year-old with exclusive breastfeeding and school-age children are the most frequently affected subjects [2].

Normal calcium levels, reduced phosphate and 25-hydroxyvitamin-D along with raised parathyroid hormone levels oriented the diagnosis of vitamin D deficiency rickets in our patient [7]. Differential diagnosis must include other forms of rickets, such as hypophosphatemic
rickets (excluded by elevated PTH levels) and vitamin D dependent rickets due to 1α-hydroxylase deficiency (unlikely because of the exceptional response to vitamin D treatment) [8]. Traditionally, it has been considered that there are two main causes of rickets: calciopenic and phosphopenic [8]. Besides these two types, the third group of diseases, related to renal tubular disorders, also causes rickets. The classical biochemical presentation of a child with calciopenic rickets is low or normal calcium levels with increased serum PTH leading to decrease renal tubular reabsorption of phosphate and low serum phosphate levels, with decreased 25-hydroxyvitamin-D [9]. However, in phosphopenic rickets, we could find normal serum calcium, low phosphate and normal levels of PTH and 25-hydroxyvitamin-D. Alkaline phosphatase levels are very increased in calciopenic rickets due to high PTH causing an increased bone turnover, but there are not so raised in phosphopenic forms [10]. Skeletal characteristics of rickets are most evident at rapidly growing ends of long bones, so the most appropriate sites for performing an X-ray study, when it is suspected, are distal radius and ulna and distal femur and proximal tibia at the knee [9,11]. Classic radiographic findings include widening of the distal physis, cupping, fraying and lateral widening of the metaphysis with the expansion of the growth plate and angular deformities of long bones. These bony manifestations of rickets may be confused with other pathologies of children such as child abuse or osteogenesis imperfecta [3,12]. Bones in rickets may also show evidence of osteopenia in calciopenic forms due to bone demineralization because of high PTH levels [4]. Nutritional rickets associated with vitamin D deficiency is managed by correcting deficiencies in both vitamin D and calcium levels [9]. Several vitamin D preparations, dosages, dosing schedules and administration routes are available. Although daily therapy provides a more stable 25-hydroxyvitamin-D level during the treatment period than stoss therapy, non-adherence to the recommendations is a risk that may result in treatment failure [13,14]. For this reason, in our patient, we administered a single dose of intramuscular vitamin D. Table 1 summarizes vitamin D dosage according to the child age [15].

Table 1. Vitamin D dosage according to the child age

<table>
<thead>
<tr>
<th>Age</th>
<th>Daily dose for 3 months (IU)</th>
<th>Single dose (IU)</th>
<th>Maintenance daily dose (IU)</th>
</tr>
</thead>
<tbody>
<tr>
<td>&lt;3 months</td>
<td>2000</td>
<td>Not available</td>
<td>400</td>
</tr>
<tr>
<td>3-12 months</td>
<td>2000</td>
<td>50000</td>
<td>400</td>
</tr>
<tr>
<td>1-12 years</td>
<td>3000-6000</td>
<td>150000</td>
<td>600</td>
</tr>
<tr>
<td>&gt;12 years</td>
<td>6000</td>
<td>300000</td>
<td>600</td>
</tr>
</tbody>
</table>

IU: International units

After treatment initiation, all patients will require monitoring of serum calcium, phosphorus, alkaline phosphatase and 25-hydroxyvitamin-D levels [6]. In this child, alkaline phosphatase level dropped after the beginning of treatment and normalized after six months. Vitamin D and calcium status in children with nutritional rickets should be maintained after treatment by ensuring sufficient vitamin D and calcium intake and by increasing sunlight exposure [4]. In our patient, in addition to giving advice the family to increase sunlight exposure and perform a diet rich in calcium and vitamin D, we maintained daily oral vitamin D supplements at a dose of 600 IU.

In conclusion, vitamin D deficiency rickets is a rare entity in developed countries, however, it increases due to immigration. Early diagnosis plays a pivotal role to minimize morbidity. The nutritional survey, physical examination, blood test including calcium, phosphorus, alkaline phosphatase, PTH and 25-hydroxyvitamin-D as well as x-ray studies may be performed to establish the diagnosis. The treatment includes vitamin D preparations, calcium intake and sunlight exposure.

Conflict of interest
The authors declare that there is no conflict of interest.

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References


