PORTUGUESE JOURNAL OF PEDIATRICS

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CASE REPORT

Missed doses, missed milestones: a case report of nutritional rickets in a high-risk infant

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Abstract

Introduction: Nutritional hypocalcemic rickets, though uncommon in developed countries, remains a relevant concern in at-risk populations. **Case-report**: We report the case of a 13-month-old female migrant infant who presented with growth failure, delayed psychomotor development, and classic signs of rickets, including craniotabes, a rachitic rosary, limb deformities, and bone pain. Contributing factors included the absence of vitamin D supplementation, an inadequate diet, and cow's milk protein allergy. Biochemical evaluation revealed low serum levels of vitamin D and phosphorus, as well as elevated parathyroid hormone and markedly increased alkaline phosphatase, supporting the diagnosis. Radiographic findings showed osteopenia and skeletal abnormalities. After six months of treatment with high-dose vitamin D and calcium supplementation, there was significant improvement in growth, motor development, and radiological. **Discussion:** This case underscores the importance of ensuring universal access to healthcare, adherence to supplementation guidelines, and appropriate nutritional education to prevent rickets and its long-term complications.

Keywords: Rickets. Failure to thrive. Nutritional counseling. Migrants.

Falha na suplementação e atraso do desenvolvimento: um relato de caso de raquitismo nutricional em lactente de alto risco

Introdução: O raquitismo hipocalcémico nutricional, embora raro em países desenvolvidos, mantém-se uma preocupação em populações de risco. **Relato de caso:** Apresentamos o caso de uma criança migrante, de 13 meses, com má progressão ponderal, atraso do desenvolvimento psicomotor e sinais clínicos de raquitismo, incluindo craniotabes, rosário raquítico, deformidades dos membros e dor óssea. A não suplementação com vitamina D, padrão alimentar insuficiente e a alergia às proteínas do leite de vaca foram os principais fatores de risco identificados. A avaliação laboratorial e imagiológica confirmou o diagnóstico, com evidência de osteopenia e deformidades esqueléticas. O tratamento com doses elevadas de vitamina D e suplementação de cálcio levou a uma melhoria significativa no crescimento, desenvolvimento psicomotor e nos achados radiológicos ao fim de seis meses. **Discussão:** O caso realça a importância de garantir o acesso universal aos cuidados de saúde, da adesão à suplementação de vitamina D e de educação nutricional apropriada para prevenir o raquitismo e suas complicações a longo prazo.

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Received: 19-08-2025
Accepted: 19-02-2026
https://pjp.spp.pt

Available online: 21-04-2026
Port J Pediatr. (Ahead of print)
DOI: 10.24875/PJP.25000065

Keypoints

What is known

- Although nutritional hypocalcemic rickets is a rare condition in developed countries, it remains a potentially serious metabolic bone disorder.
- Exclusive breastfeeding without vitamin D supplementation, inadequate diets, and dark skin pigmentation are known risk factors for vitamin D deficiency.
- Timely recognition and evidence-based treatment of rickets are essential to preventing complications and supporting healthy growth and neurodevelopment.

What is added

- Migrant families frequently exhibit difficulty navigating healthcare systems, which leaves migrant children particularly susceptible to inadequate surveillance.
- This case highlights the importance of vitamin D supplementation adherence during breastfeeding and culturally sensitive nutritional education.
- IgE-mediated cow's milk protein allergy is identified as an aggravating factor that worsens calcium deficiency and dietary restrictions.

Introduction

Nutritional hypocalcemic rickets is a metabolic bone disorder involving impaired mineralization of the bone matrix and growth cartilage. This results in skeletal deformities and growth failure. Its primary causes include vitamin D and calcium deficiencies, which are both crucial for bone metabolism and mineral homeostasis.[1-3]

Historically, rickets was extensively described in the 17th century and was linked to malnutrition and lack of sunlight exposure in urban environments.[4] The discovery of vitamin D and its critical role in calcium metabolism led to the implementation of widespread supplementation and food fortification programs, significantly reducing its prevalence in developed regions.[2] However, despite its rarity in high-income countries, cases still arise, particularly among vulnerable populations, such as those with darker skin (melanodermic), limited sun exposure, or inadequate dietary intake.[3,5] Factors such as socioeconomic barriers, limited access to healthcare, and inadequate follow-up can also contribute to delayed diagnosis and management.[1,2,6,7]

The absence of vitamin D supplementation during the first year of life significantly increases the risk of developing rickets. In individuals with dark skin pigmentation, this risk is compounded by the reduced efficiency of cutaneous vitamin D synthesis due to increased melanin production.[2,3] Furthermore, the combined deficiency of vitamin D and calcium exacerbates bone demineralization, leading to secondary hypocalcemia and compensatory hyperparathyroidism, which are key mechanisms in the pathogenesis of rickets.[2,6]

When prevention through vitamin D supplementation fails, early recognition and appropriate management of rickets are critical to avoiding complications such as growth failure, permanent skeletal deformities, and developmental delays. Evidence-based interventions, including vitamin D and calcium supplementation, effectively reverse biochemical abnormalities and restore skeletal integrity.[6,8]

Case report

A 13-month-old Black girl was referred to the general pediatrics outpatient clinic due to poor weight gain and delayed psychomotor development (Fig. 1). She was the second child of non-consanguineous parents from Angola, and had no relevant past medical history. According to WHO growth charts, she was born at 39 weeks in England with a birth weight of 3,150 g (P15-50) and a length of 47 cm (P15) and had no neonatal complications. She received routine health surveillance in the UK until 6 weeks of age, during which her weight remained at the 15th percentile and her length ranged between the 3rd and 15th percentiles. Thereafter, follow-up was discontinued, partly due to COVID-19 restrictions. The family moved from England to Portugal when she was seven months old. She was exclusively breastfed until 7 months, when complementary feeding was introduced. The mother reported lip swelling and facial erythema after ingesting cow's milk products, leading to the avoidance of dairy and delaying the introduction of most complementary foods. In Portugal, follow-up was only initiated at 13 months of age, starting with an initial primary care visit, after which a pediatric referral was made and the child was evaluated in a pediatric consultation two weeks later. At that time, her diet consisted mainly of breast milk, fruit, and eggs. She had never received vitamin D supplementation.

Physical examination revealed that she was malnourished, with a weight of 5,764 g (< 3rd percentile; z-score: −3.98), a length of 64 cm (< 3rd percentile; z-score: −4.27), and a head circumference of 45.7 cm (50-85th percentile; z-score: 0.38), according to WHO growth charts. She had limited spontaneous movement and axial hypotonia, and appeared distressed during passive mobilization, which is suggestive of bone pain (Fig. 2A). She also had a markedly enlarged anterior

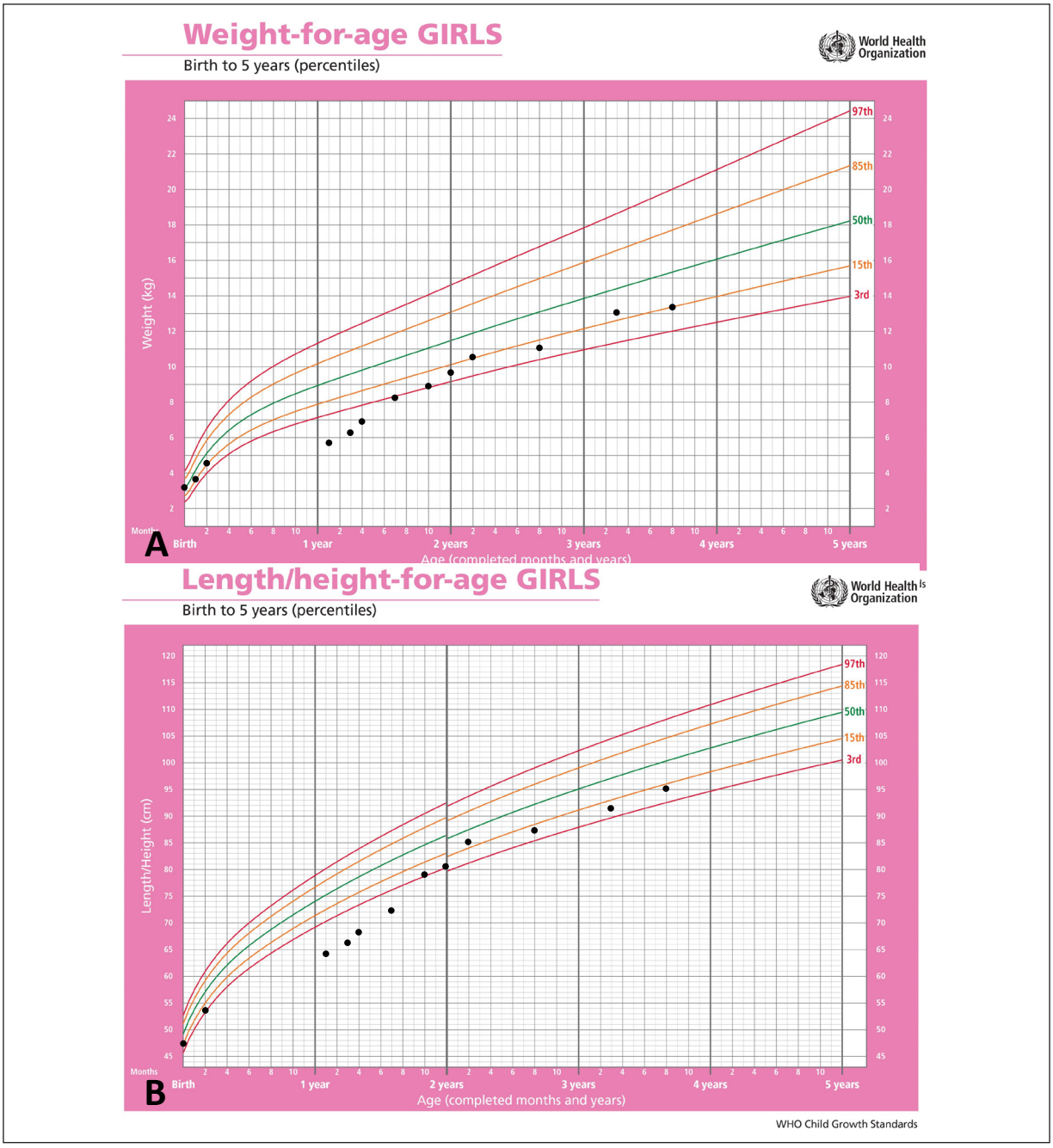

Figure 1. WHO growth charts for girls aged 0-5 years. Weight-for-age (**A**) and length-for-age (**B**) growth charts for girls aged 0– 5years, based on WHO standards. The black crosses represent the patient's anthropometric measurements plotted over time. At presentation, both weight and length were below the 3rd percentile, indicating severe growth failure. Following nutritional and medical intervention, catch-up growth is observed.

fontanelle measuring 12 × 10 cm and craniotabes. She presented swelling of the costochondral junctions (rachitic rosary), widening of the wrists, bilateral femoral varus, and absence of teeth (Fig. 3A). Her skin was melanodermic with marked xerosis and areas of eczema. A neurodevelopmental assessment revealed head control but inability to sit unsupported or bear weight. In the prone position, she was unable to lift her head. No additional neurodevelopmental abnormalities were observed. Her older sister was also evaluated during the initial consultations and showed appropriate weight and length growth (z-scores 0–2) and normal psychomotor development, with no relevant findings on physical examination.

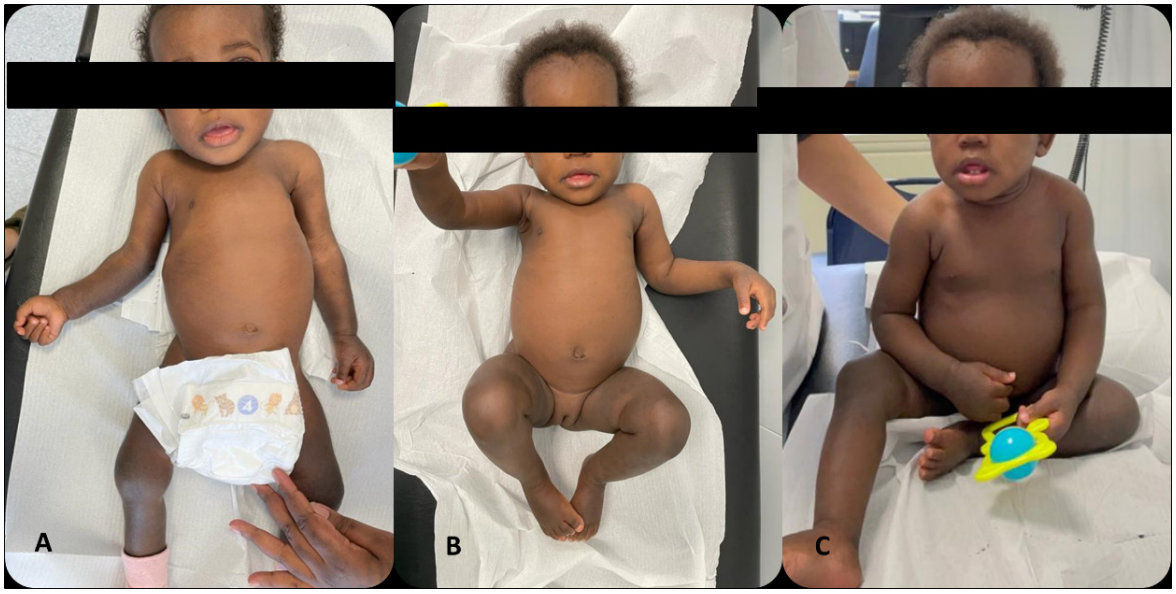

Figure 2. Clinical evolution before and after treatment. **A**: the first observation shows malnourish appearance, axial hypotonia and rachitic rosary. **B**: observation after 3 months of treatment with enhanced nutrition status and muscle tone. **C**: after 6 months of treatment with good nutrition status and adequate muscle tone.

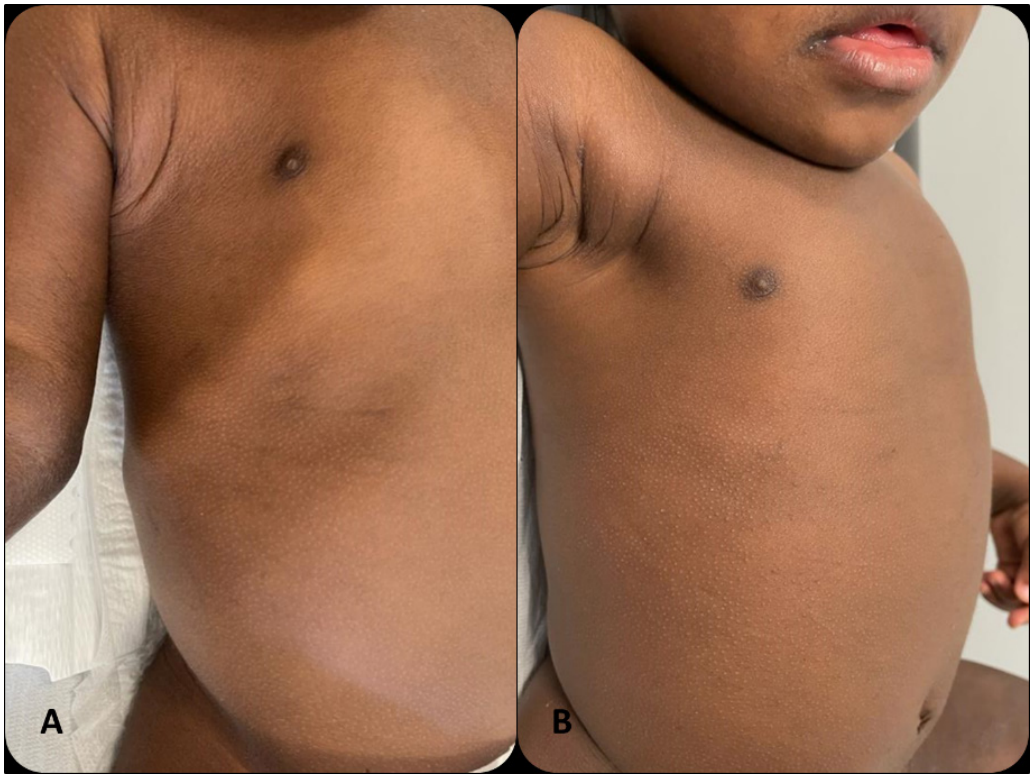

Figure 3. Chest wall changes before and after treatment. **A**: the first observation shows swelling of the costochondral joints (rachitic rosary). **B**: chest of the child after 3 months of treatment.

Table 1. Relevant laboratory findings at initial evaluation

Analyte	Result	Reference values
Hemoglobin	10.9 g/dL	10.5-14.0 g/dL
Ferritin	129 ng/mL	10-60 ng/mL
Folate	3.9 ng/mL	1.8-9.0 ng/mL
Vitamin B12	326 pg/mL	200 to 800 pg/mL*
25-OH-Vitamin D	13.9 ng/mL	30 to 60 ng/mL*
Calcium	9.3 mg/dL	8.8-10.8 mg/dL
Albumin	3.51 g/dL	3.4-4.2 g/dL
Phosphorus	2 mg/dL	3.8-6.5 mg/dL
Alkaline phosphatase	1466 U/L	145-420 U/L
Parathyroid hormone	147 pg/mL	10-65 pg/mL*
Urinary Calcium:Creatinine ratio	< 0.03	< 0.4 mg/mg

Reference ranges according to Nelson Textbook of Pediatrics, 20th edition (Elsevier, 2016), except for parameters marked with an asterisk (*), for which reference values were obtained from UpToDate.

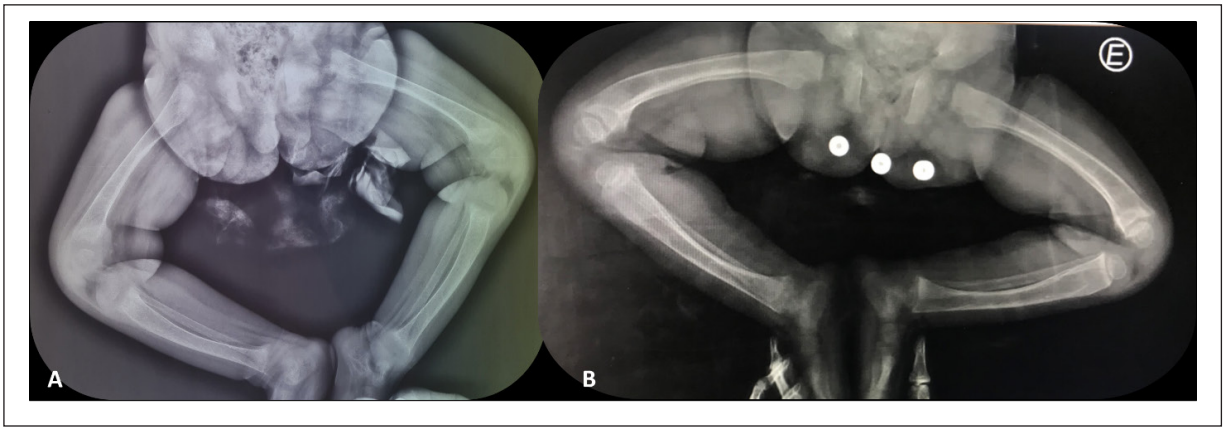

Figure 4. Radiographic changes before and after treatment. **A:** pre-treatment radiograph of the lower limbs and hip, showing loss of definition and epiphyseal widening, with no visible ossification centers, diaphyseal deformity, osteopenia, and thin cortex. **B:** radiograph of the lower limbs after 3 months of treatment, showing improvement in osteopenia, with visible ossification centers, while diaphyseal deformity persists.

Extensive laboratory investigations revealed abnormalities in phosphocalcic metabolism, which are consistent with a diagnosis of nutritional hypocalcemic rickets. These abnormalities include low levels of vitamin D and phosphorus, normal levels of calcium and high levels of alkaline phosphatase and parathyroid hormone (Table 1). Immunological findings indicated an IgE-mediated cow's milk protein allergy, with specific IgE levels corresponding to class V for beta-lactoglobulin and class III for casein. There were no anemia or other nutritional deficiencies, including ferropenia. Renal, thyroid, hepatic, and coagulation profiles were normal (Table 1).

Imaging revealed thin cranial bones, an enlarged anterior fontanelle, and defective tooth mineralization. Long bone X-rays showed osteopenic diaphyses with thin cortices, loss of definition, and epiphyseal widening without visible ossification centers, as well as diaphyseal deformities in the lower limbs (Fig 4A).

Treatment began with a daily dose of 2000 IU of vitamin D for 4 months, followed by a maintenance daily dose of 600 IU, which has been continued. Calcium carbonate was prescribed at 250 mg three times daily (equivalent to 100 mg of elemental calcium per dose, totaling 50 mg/kg/day) for 4 months. It was discontinued after the laboratory resolved there were

abnormalities and a near-complete radiological improvement, following a discussion with a pediatric radiologist. She also began complementary feeding while avoiding cow's milk proteins. After 3 months, there was significant clinical and radiological improvement (Fig 2B, 3B, 4B). At the 6-month follow-up (at 19 months of age), she had regained age-appropriate muscle tone, was walking independently, and had improved nutritional status (Fig. 2C). Catch-up growth was observed with sustained tracking between the 3rd and 15th percentiles for both weight (from 23 months of age) and length (from 32 months onward). This was accompanied by the resolution of hypotonia and motor delay. She was referred to Allergology, Dermatology, Nutrition and Social services consultations for further assessment and management. Social services were crucial in supporting the family and helping them navigate the national health system.

Discussion

Normal bone growth and mineralization require adequate calcium and phosphate availability. Rickets results from impaired mineralization at the growth plate and can be classified according to the predominant mineral deficiency into calcipenic and phosphopenic forms. Calcipenic rickets, the most common type, results from inadequate calcium availability and is most frequently caused by vitamin D deficiency or impaired vitamin D metabolism. Serum calcium levels may be low or normal, while secondary hyperparathyroidism with hypophosphatemia is a typical biochemical feature.

Despite the low overall incidence of nutritional rickets in most Western countries following the implementation of universal vitamin D supplementation, a disproportionately higher burden persists among specific risk groups with incidence rates estimated to be up to 100-fold higher.[2] Infants with darker skin pigmentation, prolonged breastfeeding without vitamin D supplementation, and those born to vitamin D–deficient mothers are particularly affected. Consequently, there is a markedly increased incidence among migrant and refugee populations from non-Western countries.[2]

This case describes a 13-month-old girl who presented with failure to thrive and delayed psychomotor development, alongside clinical and laboratory findings that were consistent with nutritional hypocalcemic rickets. Several risk factors were present, including an absence of vitamin D supplementation in the first year of life, an inadequate diet, and migrant status.[7,9,10] These factors represent a classic scenario of insufficient calcium and vitamin D intake, which is pivotal in the pathogenesis of rickets in infancy.[1-3,5] The co-occurrence of cow's milk protein allergy, confirmed by elevated specific IgE levels, contributed to dietary restrictions and likely worsened the calcium deficiency, as described by Che et al.[6,8,11] Migrant families often struggle to navigate healthcare systems, which can result in in suboptimal medical surveillance and delayed identification of growth failure and nutritional deficiencies in their children.[12]

The patient's laboratory findings — low vitamin D and phosphorus levels, elevated PTH, and markedly high alkaline phosphatase — corroborate the diagnosis of nutritional hypocalcemic rickets.[1,2] This biochemical profile indicates functional hypocalcemia, leading to compensatory hyperparathyroidism, a hallmark of cases involving simultaneous calcium and vitamin D deficiencies.[2,7] Clinically, she exhibited typical rickets features, including an enlarged fontanel, softening of the skull bones (craniotabes), a rachitic rosary, lower limb deformities, and bone pain. The patient also presented with growth failure and delayed psychomotor development. The treatment regimen together with nutritional counseling, was effective in promoting catch-up growth in length and weight, improving age-appropriate muscle tone, supporting the achievement of motor milestones, and leading to radiological resolution of bone abnormalities. She was referred to Immunoallergology for the cow´s milk protein allergy.

This case highlights the importance of early diagnosis and timely intervention in nutritional hypocalcemic rickets, as well as the need to ensure access to child health surveillance to all children. Despite its rarity in developed countries, rickets remains a concern, especially among vulnerable groups, particularly with dark skin or inadequate dietary intake. In this case, the absence of vitamin D supplementation, dietary restrictions due to cow's milk protein allergy, and inadequate complementary feeding, along with social vulnerability contributed to the development of rickets.

The successful treatment with vitamin D and calcium supplementation emphasizes the need for early recognition and adherence to evidence-based guidelines.[7] Regular follow-ups and proper nutritional counseling are essential to preventing complications such as growth delays and skeletal deformities.

Author contributions

C. Cezanne: conception and design of the report; acquisition of data either from the patient or literature; analysis or interpretation of data either from patients, research studies, and literature; drafting the article; critical review of the article for important intellectual content; final approval of the version

to be published; agreement to be accountable for the accuracy or integrity of the work. J. Bastos: conception and design of the report; acquisition of data either from the patient or literature; analysis or interpretation of data either from patients, research studies, and literature; drafting the article; critical review of the article for important intellectual content; final approval of the version to be published; agreement to be accountable for the accuracy or integrity of the work. N. Santos: critical review of the article for important intellectual content; final approval of the version to be published; agreement to be accountable for the accuracy or integrity of the work S. Fraga: critical review of the article for important intellectual content; final approval of the version to be published; agreement to be accountable for the accuracy or integrity of the work. M. Braga: critical review of the article for important intellectual content; final approval of the version to be published; agreement to be accountable for the accuracy or integrity of the work.

Funding

There were no external funding sources for the realization of this paper.

Conflicts of interest

None.

Ethical considerations

Protection of human subjects and animals. The authors declare that no experiments on humans or animals were performed for this research.

Confidentiality, informed consent, and ethical approval. The authors have followed their institution's confidentiality protocols, obtained informed consent from all patients, and secured approval from the Ethics Committee. SAGER guidelines have been followed as applicable to the nature of the study.

Declaration on the use of artificial intelligence (AI). The authors declare that no generative artificial intelligence was used in the writing or creation of the content of this manuscript.

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