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Vitamin D Deficiency Rickets in a Child: Diagnosis, Treatment, and Clinical Outcomes

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Abstract **Case Report**

This article presents the clinical case of J.S.M., a 26-month-old child residing in a rural community in northeastern Brazil, diagnosed with severe nutritional rickets due to vitamin D deficiency. The child was referred to the primary health unit due to delays in motor development, deformities in the lower limbs, and symptoms of irritability and bone pain. The clinical history revealed a lack of vitamin D supplementation, a diet deficient in essential nutrients, and limited sun exposure. Physical examination and laboratory tests confirmed hypocalcemia, hypophosphatemia, and critically low vitamin D levels, as well as characteristic bone deformities of rickets. Initial treatment included vitamin D and calcium supplementation, with improvements in laboratory parameters after 4 months of follow-up. However, persistent bone deformities indicated the need for consideration of surgical correction. The case highlights the importance of early detection, appropriate treatment, and the need for public health policies focused on preventing rickets in vulnerable populations.

Keywords: Nutritional Rickets, Vitamin D Deficiency, Bone Deformities.

INTRODUCTION

Rickets is a pediatric condition characterized by inadequate or abnormal mineralization of the growth cartilage in the epiphyseal plates, resulting in weak bones and skeletal deformities. Although it is a preventable condition, rickets still persists in various regions around the world, including Brazil, particularly among vulnerable populations. The primary cause of rickets is vitamin D deficiency, which is essential for regulating calcium and phosphorus metabolism, both crucial for normal bone development in children (Ikegawa & Hasegawa, 2023).

In Brazil, socioeconomic, cultural, and geographic factors contribute to the occurrence of rickets, especially in rural areas and low-income communities where nutrition is inadequate and sun exposure is limited (Mahmoud Mohamed, Gharib Sabaq, & Nabawy-Elasser, 2023). Vitamin D can be obtained through sunlight exposure and dietary intake; however, in areas with restricted access to vitamin D-rich foods and where cultural practices limit sun exposure, children are at higher risk of developing this condition (Singhania *et al*., 2023).

The case presented in this report illustrates the clinical and social challenges faced by a 2-year-old child residing in a rural community in the extreme south of Bahia state in northeastern Brazil, where the combination of environmental and nutritional factors led to the development of severe rickets. The relevance of this case lies not only in the identification and management of the condition but also in the need for public health strategies aimed at preventing rickets in vulnerable populations (Aftab *et al*., 2023). Recent literature reviews emphasize the importance of vitamin D supplementation in at-risk areas and strengthening health programs to improve access to nutritional education and medical care—critical elements for eradicating rickets in endemic regions (Aiello *et al*., 2023; Ikegawa & Hasegawa, 2023).

CASE PRESENTATION

J.S.M., a 26-month-old male patient, was brought to the basic health unit by his mother due to complaints of motor development delay and deformities in the lower limbs. The mother reported that J.S.M. had not yet begun walking and, in recent months, had developed bowed legs (genu varum) and difficulty sitting and crawling. The child also showed progressive irritability and complained of pain upon palpation of the lower limbs.

The patient was born full-term via normal delivery, with a birth weight of 3.2 kg, without any neonatal complications. Neuropsychomotor development was adequate until the early months, but there was a delay in autonomous walking, which prompted the search for medical evaluation. The family history did not reveal any hereditary metabolic diseases or cases of rickets, and both parents are healthy. J.S.M. lives with his family in a rural community in the extreme south of Bahia state in northeastern Brazil, an area characterized by low income and restricted access to diverse foods and health services. The patient's diet is predominantly based on carbohydrates, such as rice, beans, and cassava flour, with limited intake of dairy products and other sources of calcium and vitamin D. The mother reported that, due to intense heat, the patient is rarely exposed to the sun, and when he is, he wears clothing that covers most of his body. No vitamin D supplementation was administered during the neonatal and early childhood periods.

On physical examination, J.S.M. appeared to be in borderline nutritional status, with a weight of 9.5 kg (below the 3rd percentile) and height of 76 cm (below the 3rd percentile). The child was irritable during the examination. The skull showed partially open anterior fontanelles and slight frontal bulging. The thorax presented with pectus carinatum, with visible rachitic rosary, characterized by bony protrusions along the costochondral joints. The lower limbs exhibited significant bowing (genu varum) and widening of the ankles, suggesting typical "rachitic wrist" deformity. The wrists were also widened, and there was tenderness to bone palpation. The abdomen showed slight distension, with no evidence of hepatomegaly or splenomegaly. Neurologically, reflexes were preserved, but muscle tone was decreased, and the patient could not stand without support.

Laboratory tests revealed hypocalcemia, with serum calcium of 8.0 mg/dL (normal: 8.5 - 10.5 mg/dL), hypophosphatemia, with serum phosphorus of 3.0 mg/dL (normal: 4.5 - 5.5 mg/dL), and a significant increase in alkaline phosphatase, with levels of 700 U/L (normal: 150 - 400 U/L). Serum 25-hydroxyvitamin D levels were severely low at 12 ng/mL (sufficiency: >30 ng/mL), and parathyroid hormone (PTH) was elevated at 110 pg/mL (normal: 10 - 65 pg/mL). X-rays of the lower limbs revealed pronounced bowing of the tibias and femurs, with widening and irregularity of the metaphyseal plates, along with generalized osteopenia.

Based on the clinical, laboratory, and radiological findings, a diagnosis of severe nutritional rickets secondary to vitamin D deficiency was established.

Treatment and Management

J.S.M.'s treatment was aimed at correcting vitamin D deficiency and addressing the bone deformities associated with nutritional rickets. Initially, oral vitamin D supplementation was started at 400 IU/day, along with calcium supplementation of 500 mg/day of calcium carbonate, to restore serum vitamin D and calcium levels and promote bone mineralization. Additionally, dietary guidance was provided to increase the intake of vitamin D and calcium-rich foods, and recommendations were made to enhance safe sun exposure due to the importance of cutaneous vitamin D synthesis.

J.S.M. was re-evaluated after 4 months of follow-up. New laboratory tests showed significant improvement in biochemical parameters. Serum calcium levels normalized, increasing to 9.2 mg/dL (normal: 8.5 - 10.5 mg/dL). Serum phosphorus also improved, reaching 4.2 mg/dL (normal: 4.5 - 5.5 mg/dL). Alkaline phosphatase decreased to 450 U/L (normal: 150 - 400 U/L), indicating a positive response to treatment. Serum 25-hydroxyvitamin D levels increased to 25 ng/mL (sufficiency: >30 ng/mL), still below the ideal range but showing a trend towards improvement. Parathyroid hormone (PTH) was reduced to 70 pg/mL (normal: 10 - 65 pg/mL), reflecting therapeutic response and partial correction of hypocalcemia.

Physical examination showed a reduction in the degree of leg bowing and in the child's irritability, although the delay in walking persisted. The residual bone deformity, combined with the delay in motor development, suggested the need to consider surgical correction to address persistent deformities and optimize functional development. It was recommended that J.S.M. be evaluated by a pediatric orthopedic specialist for possible corrective surgical intervention, which may include osteotomy procedures to correct genu varum and other skeletal deformities associated with rickets. Ongoing follow-up will be essential to monitor treatment progress, adjust supplementation doses as needed, and assess the need for additional interventions.

DISCUSSION

The case of J.S.M. illustrates the severity of nutritional rickets due to vitamin D deficiency and highlights the importance of early detection and appropriate intervention. Existing literature reinforces that rickets is a largely preventable condition, but its prevalence remains significant in socioeconomically disadvantaged contexts. Timely diagnosis and treatment

are crucial to prevent long-term complications, such as permanent bone deformities and motor development delays.

Vitamin D deficiency, the primary cause of rickets, is commonly associated with inadequate diets and low sun exposure, factors evident in J.S.M.'s case. Studies show that vitamin D deficiency is particularly prevalent in populations with limited access to vitamin D-rich foods and inadequate medical care. In tropical and subtropical regions, such as northeastern Brazil, vitamin D insufficiency is often exacerbated by cultural practices that limit sun exposure and diets poor in essential nutrients.

J.S.M.'s treatment, which included vitamin D and calcium supplementation, aligns with recommendations for managing nutritional rickets. The positive response observed in laboratory tests and the improvement in clinical symptoms corroborate the effectiveness of this therapeutic approach. According to the literature, vitamin D supplementation is fundamental for correcting calcium and phosphorus deficits, promoting bone mineralization, and recovering from rickets-associated deformities.

However, the fact that J.S.M. still presents significant bone deformities despite clinical treatment underscores the need for a multifaceted approach to rickets management. Studies demonstrate that in cases of severe or prolonged rickets, surgical intervention may be necessary to correct residual bone deformities and optimize functional development. Considering surgical correction for J.S.M. exemplifies the practical application of theoretical knowledge on the treatment of rachitic deformities.

Comparisons with similar cases in the literature show that surgical correction is often indicated when bone deformities do not adequately respond to conservative treatment. Literature suggests that orthopedic surgery can be effective in correcting severe deformities and improving motor functions, particularly in children with severe rickets who experience significant mobility and quality of life impairment.

J.S.M.'s case also highlights the importance of education and access to healthcare in preventing rickets. The lack of vitamin D supplementation and inadequate diet are critical factors that can be addressed through public health policies and programs. Literature emphasizes the need for educational and community health interventions to promote adequate nutrient intake and sun exposure as effective preventive measures against rickets.

Additionally, ongoing follow-up and monitoring of patients with rickets are essential to ensure treatment efficacy and adjust interventions as needed. J.S.M.'s experience reinforces the importance of long-term follow-up to assess treatment response and implement additional interventions, such as surgical correction, when appropriate.

The theoretical implication of this case lies in the practical application of concepts related to the pathophysiology of rickets and the effectiveness of treatment strategies. The partial success of J.S.M.'s clinical treatment demonstrates the need for an integrated approach that combines nutritional therapy, sun exposure, and, when necessary, surgical intervention for effective rickets management.

Finally, J.S.M.'s case underscores the challenges and complexities of managing rickets in socioeconomically disadvantaged contexts. It serves as an example for the medical community and health policy makers about the need for comprehensive strategies to prevent and treat rickets, improve access to healthcare, and ensure equity in pediatric care. The case reinforces the need for public health policies aimed at rickets prevention and the promotion of children's bone health in vulnerable populations.

CONCLUSION

For clinical practice, this case underscores the need for continuous monitoring and long-term follow-up to adjust interventions based on treatment response. Early recognition of rickets signs and the implementation of effective treatment strategies are essential to prevent complications and improve patient prognosis.

Future research should focus on evaluating preventive and educational interventions targeting at-risk populations. Additional studies may explore the effectiveness of different supplementation regimens and nutritional education strategies to optimize rickets treatment and prevention. Research should also assess the effectiveness of surgical approaches for managing persistent bone deformities in severe cases of rickets.

In conclusion, J.S.M.'s case serves as a reminder of the importance of an integrated approach to rickets management, including prevention, clinical treatment, and surgical intervention when necessary. Enhancing preventive care and implementing public health policies are crucial for reducing rickets incidence and promoting bone health in children, especially in resource-limited areas.

Declaration of Patient Consent

The authors certify that they have obtained all appropriate consent forms from the child's legal guardians. In the form, the guardians granted permission for the publication of the child's images and other clinical information in the journal. The guardians understand that the child's name and personal information will not be disclosed, and efforts will be made to preserve the child's identity, although anonymity cannot be guaranteed.

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