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ETIOLOGY, PATHOGENESIS, CLINICAL SYMPTOMS, AND LABORATORY DIAGNOSTICS OF RICKETS IN CHILDREN

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Abstract

Rickets is a metabolic bone disease that primarily affects children and is characterized by defective mineralization of the growing bone. The condition is predominantly caused by deficiencies in vitamin D, calcium, or phosphate, but may also result from genetic mutations or underlying medical conditions. This article reviews the etiology and pathogenesis of rickets in children, its clinical presentation, and current laboratory diagnostic methods. Understanding the mechanisms of rickets is crucial for early diagnosis and effective management of this preventable condition [2,4].

Keywords: rickets, children, vitamin D deficiency, calcium deficiency, phosphate deficiency, bone deformities.

Introduction

Rickets is a disease that leads to softening and weakening of the bones in children, commonly resulting from deficiencies in vitamin D, calcium, or phosphate. It is a preventable condition that can have serious long-term health implications, including

skeletal deformities, fractures, and growth retardation. Although the global prevalence of rickets has decreased due to improved nutrition and the widespread use of vitamin D supplements, the disease continues to affect certain populations, particularly in regions with limited sunlight exposure, malnutrition, or specific genetic disorders [3,5].

Etiology of Rickets

1. Vitamin D deficiency. The most common cause of rickets is a deficiency in vitamin D, which plays a pivotal role in calcium and phosphate metabolism. Vitamin D deficiency impairs the absorption of calcium and phosphate from the intestines, leading to a lack of these minerals necessary for bone mineralization.

Risk factors include:

- Limited sunlight exposure. Vitamin D is synthesized in the skin upon exposure to ultraviolet (UV) radiation. Insufficient sunlight, common in regions with long winters or in individuals with limited outdoor activity, increases the risk of deficiency;
- Breastfeeding without supplementation. Although breast milk contains some vitamin D, it is insufficient to meet the needs of infants, especially those who are exclusively breastfed without supplementation;
- Skin pigmentation. Darker skin has higher melanin levels, which reduces the skin's ability to produce vitamin D from sunlight [1,5].

2. Calcium deficiency. Inadequate dietary intake of calcium can also contribute to rickets. Calcium is essential for bone mineralization, and a deficiency leads to decreased bone density and structural integrity. Common causes include poor dietary habits or malabsorption disorders, which can impair calcium absorption.

3. Phosphate deficiency. Hypophosphatemic rickets is a rare form of rickets caused by phosphate deficiency. It is usually due to genetic mutations affecting phosphate metabolism, such as X-linked hypophosphatemic rickets (XLH), a disorder that impairs the renal reabsorption of phosphate.

4. Genetic and metabolic disorders. Some forms of rickets are genetic, such as: X-linked hypophosphatemic rickets (XLH): A genetic disorder caused by mutations in the PHEX gene, leading to phosphate wasting and insufficient mineralization of

bone. Autosomal recessive hypophosphatemic rickets: Caused by mutations in genes that affect phosphate metabolism. In addition to genetic causes, renal tubular disorders, such as renal phosphate wasting, can lead to rickets.

Pathogenesis of Rickets

The pathogenesis of rickets involves disturbances in bone mineralization due to deficiencies in calcium, phosphate, and vitamin D. These deficiencies impair the ability of the bone matrix to mineralize properly, leading to the accumulation of unmineralized osteoid tissue, softening of the bones, and deformation. The pathophysiological mechanisms of rickets are as follows:

Defective mineralization. Vitamin D deficiency leads to decreased intestinal absorption of calcium and phosphate, resulting in low serum calcium and phosphate levels. In response to low calcium, the parathyroid glands secrete parathyroid hormone (PTH), which increases bone resorption to release calcium. However, without sufficient phosphate, bone mineralization remains impaired;

- Calcium deficiency further exacerbates bone mineralization defects, as calcium is an essential component of hydroxyapatite crystals in the bone matrix;
- Phosphate deficiency causes a reduction in the mineralization capacity of bone, leading to poorly mineralized bone tissue.

Bone deformities. The softening of bones due to defective mineralization results in skeletal deformities such as bowing of the legs (genu varum), rachitic rosary (enlarged costochondral junctions), and craniotabes (soft spots on the skull). As the bones soften, the growing bones of the child may also bend and deform under the influence of weight-bearing forces, leading to growth retardation and skeletal asymmetry [3].

Clinical symptoms of rickets. The clinical presentation of rickets in children varies depending on the severity and duration of the deficiency. Common symptoms include bone deformities:

- Genu varum (bow legs): Bowing of the legs due to the weakening of the long bones in the lower limbs;
- Genu valgum (knock knees): In severe cases, the knees may touch while the feet remain apart;

- Rachitic rosary: Enlargement of the costochondral joints, which gives the appearance of beads along the ribcage.
- Frontal bossing: Prominent forehead caused by the failure of normal skull mineralization.

Children with rickets may exhibit growth retardation, leading to short stature and delayed milestones. Bone pain and tenderness can cause reluctance to walk or crawl. Weakness of the skeletal muscles due to impaired calcium and phosphate metabolism may lead to difficulty with movement, walking, or crawling. Children may present with pain in the bones, especially in the lower limbs, pelvis, and ribs. Tenderness can be elicited upon palpation of the affected bones.

Irritability and poor appetite are often seen in children with vitamin D deficiency. In severe cases, tetany (muscle spasms) may occur due to low calcium levels in the blood.

Laboratory Diagnostics of Rickets.

1. Serum calcium and phosphate levels. In vitamin D deficiency rickets, serum calcium levels are often normal or slightly low, while phosphate levels are typically low. The ratio of calcium to phosphate is altered, impairing bone mineralization.
2. Serum alkaline phosphatase (ALP). Elevated serum alkaline phosphatase levels are a hallmark of rickets. ALP is released from the bone during the process of bone formation and is elevated when there is active bone turnover.
3. Serum vitamin D (25-hydroxyvitamin D). Low levels of 25-hydroxyvitamin D (the inactive form of vitamin D) are indicative of vitamin D deficiency. This test is critical for diagnosing rickets due to vitamin D deficiency.
4. Parathyroid hormone (PTH). Elevated PTH levels are commonly seen in rickets due to secondary hyperparathyroidism, which is a compensatory response to low calcium levels. The elevated PTH levels further contribute to bone resorption.
5. Genetic testing. Genetic testing can be used to diagnose X-linked hypophosphatemic rickets or other genetic forms of rickets, particularly in cases of hypophosphatemic rickets.

X-ray imaging. Radiographs of affected bones may reveal characteristic changes, including bone softening, fractures, and skeletal deformities like bowing of the legs or rachitic rosary.

Conclusion. Rickets in children is a preventable bone disorder, primarily caused by vitamin D, calcium, or phosphate deficiency. Understanding the etiology and pathogenesis of rickets is essential for early diagnosis and intervention. Clinical symptoms often include bone deformities, growth delay, and muscle weakness, while laboratory diagnostics focus on serum levels of calcium, phosphate, vitamin D, and markers of bone turnover. With timely diagnosis and appropriate treatment, including vitamin D and calcium supplementation, rickets can be effectively managed, preventing long-term skeletal complications.

References

1. Aldana Sierra M. C., Christian C. W. Vitamin D, rickets and child abuse: controversies and evidence //Pediatric radiology. – 2021. – Т. 51. – №. 6. – С. 1014-1022. Cannalire G. et al. Alkaline phosphatase in clinical practice in childhood: Focus on rickets //Frontiers in Endocrinology. – 2023. – Т. 14. – С. 1111445.
2. Gentile C., Chiarelli F. Rickets in children: an update //Biomedicines. – 2021. – Т. 9. – №. 7. – С. 738.
3. Miller W. L., Imel E. A. Rickets, vitamin D, and Ca/P metabolism //Hormone Research in Paediatrics. – 2022. – Т. 95. – №. 6. – С. 579-592.
4. Umarova S. S., Mukhamadiyeva L. A., Nabiyeva F. S. The pathogenesis of rheumatic fever //Journal of new century innovations. – 2023. – Т. 29. – №. 4. – С. 164-169.
5. Тюленева Н. В. Анализ клинико-диагностических критериев рахита у детей //печатается по решению редакционно-издательского совета фгбоу во «Хакасский государственный университет им. НФ Катанова». – 2023. – с. 264.