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Pathogenetic And Clinical Aspects of Rickets Associated with Connective Tissue Dysplasia in Children

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Abstract: The article presents a review of current data on the pathogenetic and clinical aspects of rickets in children with connective tissue dysplasia. Particular attention is paid to disorders of mineral metabolism, hormonal regulation, and structural insufficiency of connective tissue, which contribute to a more severe course of the disease. The main clinical manifestations, diagnostic criteria, and principles of comprehensive treatment and prevention are described. The importance of early detection of combined pathology and a multidisciplinary approach to patient management is emphasized.

Keywords: Rickets, connective tissue dysplasia, children, vitamin D, mineral metabolism, parathyroid hormone.

Introduction: Rickets is one of the most common pediatric disorders associated with impaired bone mineralization. Deficiency of vitamin D, calcium, and phosphorus leads to bone softening, skeletal deformities, growth retardation, and an increased risk of fractures. The condition is most frequently observed in children aged between three months and five years, a period characterized by rapid bone growth, during which mineral deficiencies have the greatest impact on skeletal development. Connective tissue dysplasia represents a group of genetically determined disorders affecting the structure and function of connective tissue. Increased joint hypermobility, muscle weakness, and abnormalities in bone and cartilage structure contribute to reduced mechanical strength of bones and increased fragility. The coexistence of rickets and connective tissue dysplasia constitutes a clinically significant problem, as both conditions exert a synergistic effect on bone metabolism. Children with this combined pathology often present with pronounced skeletal deformities, muscle weakness, delayed motor development, and a higher risk of fractures. Recent studies emphasize the importance of early identification of children at risk for the combined development of rickets and connective tissue dysplasia. Timely initiation of preventive and therapeutic interventions enables correction of mineral metabolism and strengthening of the skeletal system. The aim of this review article is to systematize current data on the pathogenesis, clinical manifestations, diagnosis, and комплексное лечение (comprehensive treatment) of rickets associated with connective tissue dysplasia in children.

METHODS

This study is based on a comprehensive literature review and analysis of modern scientific data related to the pathogenesis, clinical features, diagnosis, and treatment of rickets associated with connective tissue dysplasia in children.

The analysis included:

- evaluation of current clinical and experimental studies
- comparison of diagnostic approaches
- assessment of laboratory and instrumental findings
- synthesis of data on therapeutic and

preventive strategies

The methodological approach involved systematic analysis, comparison, and generalization of published data to provide an integrated understanding of the combined pathology.

RESULTS

Pathogenesis

The pathogenesis of rickets associated with connective tissue dysplasia is complex and includes disturbances in mineral metabolism, structural bone changes, and hormonal imbalance.

Disturbance of mineral metabolism

Vitamin D deficiency reduces intestinal absorption of calcium and phosphorus, leading to hypocalcemia and hypophosphatemia. In response, parathyroid hormone (PTH) secretion increases, stimulating bone resorption and renal phosphate excretion. Mineral deficiency results in osteomalacia, bone softening, and impaired formation of growth plates.

Features of connective tissue dysplasia

In connective tissue dysplasia, the structure of collagen, elastin, and other components is altered. This leads to decreased mechanical strength of bones, increased flexibility, and fragility. In combination with rickets, these changes intensify bone deformities and increase the risk of injury.

Hormonal mechanisms

Hormone	Effect
Parathyroid hormone (PTH)	Increases bone resorption and renal phosphate excretion to maintain calcium homeostasis
Calcitonin	Reduces osteoclast activity and prevents bone destruction
Vitamin D (calcitriol)	Enhances calcium and phosphorus absorption and promotes bone mineralization

Interaction of factors

The combination of vitamin D, calcium, and phosphorus deficiency with structural connective tissue abnormalities creates a vicious cycle in which bone weakness, impaired mineralization, and hormonal imbalance reinforce each other. This results in pronounced skeletal deformities and delayed bone recovery.

Scheme 1. Pathogenesis of combined rickets and connective tissue dysplasia

Vitamin D / calcium / phosphorus deficiency

↓

Hypocalcemia and hypophosphatemia

↓

Increased parathyroid hormone secretion

↓

Bone resorption + phosphate excretion

↓

Bone weakness

↓

Impaired mineralization

↓

Rickets and skeletal deformities

Clinical manifestations

Children with combined rickets and connective tissue dysplasia exhibit more severe clinical features compared to isolated forms.

- Skeletal deformities: kyphoscoliosis, postural disorders, X-shaped or O-shaped deformities of the lower limbs, chest deformities, and rib beading
- Muscle weakness and delayed motor development: hypotonia, delayed walking, impaired coordination, increased fatigue
- Other manifestations: bone pain, frequent fractures, flat feet, joint instability

Table 1. Main clinical signs

Sign	Frequency (%)	Comment

Kyphoscoliosis	65	Depends on age
O-shaped leg deformities	40	More common in severe CTD
Muscle weakness	55	Reduces physical activity
Frequent fractures	30	Occur with minimal trauma
Flat feet	25	Requires orthopedic correction

Diagnostics

Diagnosis requires a комплексный (comprehensive) approach including laboratory, instrumental, and orthopedic assessment.

Laboratory findings

- Calcium — decreased
- Phosphorus — decreased
- Alkaline phosphatase — increased
- Parathyroid hormone — increased
- Vitamin D — decreased
- Magnesium — may be reduced

Table 2. Laboratory indicators

Parameter	Normal range	Changes
Calcium	2.2–2.7 mmol/L	Decreased
Phosphorus	1.3–2.3 mmol/L	Decreased
Alkaline phosphatase	150–420 U/L	Increased
PTH	15–65 pg/mL	Increased
Vitamin D	30–50 ng/mL	Decreased

Instrumental methods

- X-ray — detects bone deformities and growth plate changes
- Densitometry (DXA) — evaluates bone mineral density
- Ultrasound — assesses bone quality
- Orthopedic assessment — evaluates deformities and joint mobility

DISCUSSION

The findings confirm that the combination of rickets and connective tissue dysplasia significantly aggravates bone pathology. The interaction between metabolic disturbances and structural defects leads to increased severity of skeletal deformities and higher fracture risk. The role of vitamin D deficiency and secondary hyperparathyroidism is central in disease progression. At the same time, connective tissue abnormalities contribute to decreased bone strength and impaired adaptation to mechanical нагрузка (mechanical load). Modern diagnostic approaches emphasize the importance of early detection using laboratory markers and imaging techniques. Multimodal assessment improves diagnostic accuracy and allows timely therapeutic intervention.

CONCLUSION

The combination of rickets and connective tissue dysplasia in children represents a significant clinical problem in pediatrics. The pathogenesis involves vitamin D, calcium, and phosphorus deficiency, secondary hyperparathyroidism, and structural abnormalities of connective tissue. Clinically, the condition manifests as pronounced skeletal deformities, muscle weakness, and increased fracture risk. Comprehensive diagnosis, early detection, and individualized therapy significantly reduce complications and improve quality of life. Effective management includes correction of mineral metabolism, supplementation with vitamin D, calcium and phosphorus, physiotherapy, and orthopedic support.

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