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PATHOLOGICAL CHANGES IN DENTAL HARD TISSUES DURING TOOTH DEVELOPMENT: GENETIC AND MOLECULAR ASPECTS

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Abstract

Pathological changes in dental hard tissues formed during tooth development represent a significant problem in modern dentistry. Disturbances in amelogenesis and dentinogenesis may result from genetic, molecular, and systemic factors that affect mineralization processes. This narrative review summarizes current data on the genetic mechanisms underlying pathological changes in enamel and dentin, discusses major hereditary dental disorders, and highlights the prospects of molecular genetic diagnostics in clinical dental practice.

Keywords: Dental hard tissues, amelogenesis, dentinogenesis, genetic factors, dentistry.

Introduction

The formation of dental hard tissues is a complex, multistage process that depends on precise interactions between cellular, molecular, and genetic mechanisms. Disruptions at any stage of tooth development may lead to pathological changes in enamel and dentin, subsequently affecting the functional integrity of the dentition and the patient's quality of life.

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In recent decades, increasing attention has been paid to the role of genetic factors in the development of dental hard tissue defects. Hereditary forms of enamel hypoplasia, amelogenesis imperfecta, and dentinogenesis imperfecta demonstrate a direct relationship between specific gene mutations and clinical dental manifestations. Understanding these mechanisms is essential for early diagnosis, prevention, and the development of personalized treatment strategies.

The aim of this review is to analyze current scientific evidence regarding genetic and molecular aspects of pathological changes in dental hard tissues during tooth development.

Physiology of Dental Hard Tissue Formation

Enamel Development

Enamel is the hardest tissue in the human body and is formed through the process of amelogenesis, which is carried out by specialized cells known as ameloblasts. This process includes the secretory stage, the maturation stage, and mineralization. Any disturbance in ameloblast activity can result in structural defects of enamel.

Dentin Formation

Dentin is formed by odontoblasts and is characterized by a lower degree of mineralization compared to enamel. Although dentinogenesis continues throughout life, the primary phase occurring during tooth development is crucial for the long-term durability of dental hard tissues.

Genetic Factors in Enamel Pathology

To date, several genes have been identified whose mutations are associated with impaired enamel formation. Among the most extensively studied are **AMELX, ENAM, MMP20, and KLK4**, which encode proteins involved in matrix organization and enamel mineralization.

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Amelogenesis imperfecta is one of the most common hereditary enamel disorders and may present in hypoplastic, hypocalcified, or hypomaturation forms. Clinical manifestations include enamel discoloration, increased wear, and a higher susceptibility to dental caries.

Genetic Disorders of Dentinogenesis

Dentin pathologies, particularly dentinogenesis imperfecta, are commonly associated with mutations in the **DSPP** gene, which encodes dentin sialophosphoprotein. Structural abnormalities in dentin result in increased tooth fragility, altered tooth morphology, and premature tooth loss.

Importantly, dentin defects may coexist with systemic conditions, including inherited connective tissue disorders, highlighting the interdisciplinary nature of this problem.

Molecular Mechanisms of Pathogenesis

At the molecular level, pathological changes in dental hard tissues are caused by disruptions in the synthesis and degradation of the organic matrix, as well as imbalances in mineralization processes. Altered expression of genes involved in calcium and phosphate transport leads to defective crystal formation in enamel and dentin.

Recent studies indicate that epigenetic factors, including DNA methylation and microRNA regulation, also play a significant role in the development of these conditions.

Clinical Significance and Diagnostics

Understanding the genetic mechanisms underlying dental hard tissue disorders enables the implementation of early diagnostic approaches, including molecular genetic testing. This is particularly important in pediatric dentistry, where early detection can reduce the severity of clinical manifestations.

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Conclusion

Genetic and molecular factors play a crucial role in the development of pathological changes in dental hard tissues during tooth formation. Advances in molecular biology and genetics open new perspectives for early diagnosis and personalized treatment of these conditions. Further research in this field will contribute to improved understanding of pathogenesis and enhanced dental care strategies.

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