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Enamel Hypoplasia in Children with Down Syndrome: Developmental Pathways and Clinical Implications for Oral Health

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Abstract

Enamel hypoplasia is a developmental defect characterized by a quantitative reduction of enamel thickness resulting from disturbances during amelogenesis. Children with Down syndrome present a markedly higher prevalence of dental anomalies, including enamel hypoplasia, compared to the general population, potentially leading to functional, esthetic, and psychosocial complications. This review synthesizes current evidence regarding the association between enamel hypoplasia and Down syndrome in children, focusing on etiological mechanisms, prevalence, and clinical implications. A narrative literature review was conducted using PubMed, Scopus, ScienceDirect, and Google Scholar databases with relevant keywords related to Down syndrome and enamel developmental defects. The reviewed studies consistently report a higher occurrence of enamel hypoplasia in children with Down syndrome, with prevalence rates ranging from approximately 30% to over 45%. The condition is associated with genetic predisposition, impaired embryonic blood supply, and systemic developmental disturbances affecting neurological and odontogenic processes. Disruption of odontoblast activity may interfere with ameloblast differentiation, leading to reduced enamel matrix secretion. Clinically, enamel hypoplasia increases susceptibility to dental caries, tooth sensitivity, enamel wear, and compromised oral health–related quality of life. Current evidence indicates a significant association between Down syndrome and enamel hypoplasia; however, further comprehensive and methodologically robust studies are required to clarify causal pathways and optimize preventive and therapeutic strategies. Early diagnosis and tailored dental management are essential to improve oral health outcomes in this vulnerable population.

Keywords: Down syndrome; Enamel hypoplasia; Children; Dental anomalies; Developmental defects

1. Introduction

Dental anomalies may result in significant functional problems affecting both jaws; therefore, a comprehensive evaluation of the factors involved in their development is essential. Specific genetic factors have been reported to contribute to the development of dental anomalies in each jaw. In addition, disturbances occurring during tooth development may lead to variations in tooth number (agenesis or supernumerary teeth), size, shape, and tooth bud position, thereby affecting both permanent and deciduous dentitions of the maxilla and mandible. Dental anomalies can be classified according to alterations in tooth number, size, shape, and structure. This classification system also categorizes anomalies based on the stage of dental development at which each anomaly is presumed to originate [1, 2].

The incidence of dental anomalies has been reported to be approximately five times higher in individuals with Down syndrome than in the general population. Among these anomalies, enamel hypoplasia is one of the most frequently observed conditions and is defined as a defect of the tooth crown surface resulting from disturbances in enamel matrix secretion, defective calcification, or impaired maturation [3]. The prevalence of enamel hypoplasia in primary teeth among one-year-old Japanese children has been reported to reach 38.5% [4]. Similarly, a study conducted in Syria reported prevalence rates of 34% in healthy children and 45% in children with Down syndrome [5]. Enamel hypoplasia

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in individuals with Down syndrome is characterized by a reduced amount of enamel matrix and decreased enamel opacity. This condition has been associated with abnormal blood supply to the embryonic jaw, which may inhibit tissue growth and cause degeneration of odontoblasts involved in dentin formation. Since dentin formation is required to induce pre-ameloblast differentiation into ameloblasts, such disturbances may ultimately result in reduced enamel matrix secretion [1, 6].

In addition to enamel hypoplasia, several other dental anomalies have been reported in individuals with Down syndrome, including anomalies of tooth number such as hypodontia and supernumerary teeth; anomalies of tooth size such as microdontia of permanent teeth; anomalies of tooth shape including short roots, taurodontia, talon cusp, fusion, and gemination; as well as anomalies of tooth structure such as enamel hypocalcification. Furthermore, tooth eruption in individuals with Down syndrome may occur asymmetrically and can be delayed by approximately two to three years compared to healthy individuals. Previous studies have identified an association between intellectual disability and enamel defects, attributed to the close relationship between enamel development and brain development. Systemic disruptions that interfere with neurological development may also affect tooth germ formation. Because damaged enamel cannot regenerate, enamel defects may provide valuable information regarding the timing and nature of insults that potentially affect other ectoderm-derived structures, including the brain [1, 7].

This study was conducted to identify correlations between enamel hypoplasia and developmental characteristics in children with Down syndrome.

2. Material and methods

This study was conducted as a narrative literature review focusing on enamel hypoplasia in children with Down syndrome and its implications for oral and dental health. The review aimed to synthesize existing evidence regarding prevalence, etiology, pathogenesis, and clinical impact without performing quantitative data pooling.

A comprehensive literature search was performed using the PubMed, Scopus, ScienceDirect, and Google Scholar databases. Relevant articles published in English were identified using combinations of the following keywords: Down syndrome, enamel hypoplasia, children, dental anomalies, and developmental defects. The search strategy was designed to capture studies addressing both clinical and developmental aspects of enamel hypoplasia in pediatric populations with Down syndrome.

Eligible articles included original research studies and review articles that specifically discussed enamel hypoplasia in children with Down syndrome, including its prevalence, etiological factors, pathogenic mechanisms, and effects on oral health. Articles that were not available in full text, published in languages other than English, or not directly related to the topic were excluded from the review.

All selected articles were assessed descriptively, and relevant findings were extracted and synthesized narratively. The evidence was organized thematically to provide a coherent overview of current knowledge regarding the association between enamel hypoplasia and developmental characteristics in children with Down syndrome.

3. Results and discussion

3.1. Types and Epidemiology of Down Syndrome

Down Syndrome (DS) is the most common chromosomal abnormality that was first discovered in humans. Three distinct types of Down syndrome have been described. Trisomy 21, which constitutes the majority of cases, occurs when there is an extra copy of chromosome 21. Mosaicism, accounting for 2–5% of cases, involves a mixture of cells containing either 46 or 47 chromosomes, with the characteristics exhibited by the affected individual depending on the number of cells involved and their chromosomal composition. The translocation type, which occurs when a segment of chromosome 21 detaches and attaches itself to another chromosome, resulting in a total of 47 chromosomes genetically. The incidence of trisomy 21 correlates strongly with increasing maternal age, with a low probability among younger mothers and a rapidly increasing risk after the age of 35 years. The condition is observed in 1 in 1,550 live births among women under 20 years old, compared with an incidence of 1 in 25 live births among women over the age of 45 [8, 9, 10].

Down syndrome occurs worldwide, with an incidence of 1 per 374 live births and an estimated affected population of approximately 8 million individuals. In 2002, the prevalence among children and adolescents aged 0–19 years was

reported as 10.3 per 10,000 live births. According to the World Health Organization (WHO), the estimated incidence ranges from 1 in 1,000 to 1 in 1,100 births, with approximately 3,000 to 5,000 babies born with DS each year. Data from the Indonesian National Basic Health Research (Riskesdas) 2018 showed that 0.41% of children were born with congenital abnormalities [5, 11, 12].

3.2. Etiology and Risk Factors of Down Syndrome

Down syndrome occurs in all racial groups, and the growth of affected children often shows developmental abnormalities, slowness, and functional barriers. Advanced maternal age has been identified as the main etiological factor due to the formation of gametes during the intrauterine phase and disruption of meiosis I, which causes oocytes to age along with the woman, as they mature only after puberty. This aging process may damage chromosomal fibers and deteriorate the centromere, resulting in an inability to separate chromosomes during anaphase I of meiosis. Another contributing factor is impaired chromosomal segregation during gametogenesis associated with advanced paternal age, particularly above 55 years. Additional risk factors include alcohol consumption, cigarette smoking, exposure to chemical substances, oral contraceptive use, genetic inheritance, a history of abortion, and environmental agents such as radiation, all of which may contribute to genetic errors [13].

3.3. Systemic and Orofacial Manifestations in Down Syndrome

Patients with Down syndrome present with a wide range of signs and symptoms, including intellectual and developmental disabilities, neurological features, congenital heart defects, gastrointestinal abnormalities, characteristic facial features, altered immune function, and other systemic abnormalities. Furthermore, children with DS exhibit several orofacial features that may aid in diagnosis, including a highly vaulted and narrow palate, large and thick lips, and a fissured tongue that develops progressively. Individuals with Down syndrome also experience multiple oral health problems, such as a reduced oral cavity size, macroglossia, and dental anomalies including microdontia, conical-shaped teeth, hypodontia, taurodontia, and enamel defects such as enamel hypoplasia and enamel hypocalcification. Moreover, higher prevalence rates of periodontal disease, dental caries, missing teeth, prolonged retention of primary teeth, and malocclusion have been reported, often exacerbated by functional limitations [14, 15].

3.4. Association Between Down Syndrome and Enamel Defects

Several studies have reported that severe illness or prolonged febrile episodes may lead to enamel hypoplasia and hypocalcification, although follow-up studies addressing this association remain limited. Other studies have identified an association between intellectual disability and enamel defects, attributed to the close relationship between enamel development and brain development. Systemic disruptions that interfere with neurological development may concurrently affect tooth germ formation [11, 15]. Among individuals with Down syndrome, nearly 50% exhibit three or more dental anomalies, and approximately 20% are affected by enamel hypoplasia. Enamel defects have been reported to occur more frequently on buccal tooth surfaces than on other surfaces, indicating that disturbances in growth and development may be reflected in dental structures [10, 11].

3.5. Syndrome Clinical Characteristics of Enamel Hypoplasia

A study conducted in Massachusetts reported that enamel hypoplasia is a common structural anomaly in individuals with Down syndrome, with a prevalence of 32%. Enamel hypoplasia is defined as a quantitative reduction in enamel resulting from disturbances during the matrix formation stage. The condition has a high prevalence among children from developing countries, children with chronic or acute malnutrition, and children with very low birth weight. Clinically, enamel hypoplasia may manifest as pitting, grooving, or partial to complete absence of enamel. In contrast, hypomineralization occurs when enamel maturation is disturbed and presents as opaque or chalky areas on normally contoured enamel surfaces. Disturbances in either matrix formation or calcification may occur depending on the stage of tooth formation at the time of injury. Hypoplastic defects may appear as pits or grooves of varying depth and distribution or as extensive areas of enamel loss, with the remaining enamel appearing translucent or opaque [1, 9].

3.6. Etiological Factors and Developmental Timing of Enamel Hypoplasia

Enamel hypoplasia or hypomineralization may result from hereditary and environmental factors. Systemic factors include nutritional deficiencies, exanthematous diseases such as measles and chickenpox, congenital syphilis, hypocalcemia, birth injury or premature birth, fluoride ingestion, and idiopathic causes, while local factors include infection or trauma affecting deciduous teeth. In individuals with Down syndrome, enamel hypoplasia has been associated with abnormal blood supply to the embryonic jaw, which inhibits tissue growth and causes degeneration of odontoblasts involved in dentin formation. Since dentin formation is required to induce pre-ameloblasts to differentiate into ameloblasts, such disturbances may ultimately result in reduced enamel matrix secretion. Previous studies have

demonstrated a relationship between the frequency of enamel hypoplasia and the chronological timing of tooth calcification. The maxillary incisors are the first teeth to begin calcification during the third to fourth prenatal months, with amelogenesis of primary incisors nearly complete at birth, whereas canines and molars continue to develop postnatally. Consequently, disturbances occurring around the 12th prenatal week may affect anterior teeth, while those occurring around the 16th week may result in hypoplastic lesions in posterior teeth [11].

3.7. Impact of Enamel Hypoplasia on Oral Health and Quality of Life

Enamel hypoplasia may negatively affect quality of life due to both functional and esthetic consequences. The condition may cause tooth discoloration, either extrinsic or intrinsic, and may impair speech, particularly in children with enamel hypoplasia and dental caries. Surface irregularities associated with hypoplastic enamel increase susceptibility to thermal sensitivity, mechanical wear, acid erosion, plaque retention, and bacterial colonization, thereby increasing the risk of tooth decay. As a result, enamel hypoplasia is considered a predisposing factor for early childhood caries and erosion. In primary dentition, incomplete enamel calcification in pits and fissures provides a favorable environment for the adhesion and colonization of cariogenic bacteria, leading to more rapid development of early childhood caries on altered tooth surfaces [11, 16, 17].

4. Conclusion

Children with Down syndrome exhibit a higher prevalence of oral and dental anomalies compared with individuals without the condition. Previous studies have indicated an association between enamel hypoplasia and Down syndrome, which may be attributed to systemic disturbances that interfere with neurological development and subsequently affect tooth germ formation. Therefore, a proactive approach with early diagnosis is essential to ensure that dental care can be appropriately planned and tailored to the specific needs of these children. Nevertheless, further comprehensive studies are required to establish a more definitive correlation between enamel hypoplasia and specific developmental characteristics in children with Down syndrome.

Compliance with ethical standards

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Disclosure of conflict of interest

The authors declare that there is no conflict of interest.

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