



HEREDITARY DISEASES OF THE HARD TISSUES OF THE TEETH

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Abstract

This article analyzes the etiology, pathogenesis, clinical manifestations, and diagnostic and treatment methods of Amelogenesis Imperfecta, Dentinogenesis Imperfecta, and Odontogenesis Imperfecta. Amelogenesis Imperfecta is a hereditary developmental defect of tooth enamel, characterized by disturbances in the quantity and quality of enamel. Dentinogenesis Imperfecta is a hereditary disorder in the formation of dentin tissue, accompanied by discoloration, brittleness, and rapid tooth decay.

The article highlights the clinical differences, radiological signs, and differential diagnosis of these pathologies. It also examines the importance of restorative and orthopedic treatment methods in modern dentistry, including composite materials, crowns, and prosthetics. The research findings are significant for early detection and the development of effective treatment strategies.

Keywords: Amelogenesis Imperfecta, Dentinogenesis Imperfecta, Odontogenesis Imperfecta, Osteogenesis Imperfecta.

Introduction

Congenital defects of the hard tissues of the teeth transmitted from generation to generation include hereditary Amelogenesis Imperfecta, hereditary Dentinogenesis Imperfecta, and hereditary Odontogenesis Imperfecta.

Dentinogenesis Imperfecta

Dentinogenesis Imperfecta is an autosomal dominant condition affecting both primary and permanent teeth. Affected teeth range in color from gray to yellowish-brown and have wide crowns with a narrowing at the cervical region, giving them a “tulip” shape. Radiologically, the pulp chambers and root canals



are absent. Enamel fractures easily, exposing the dentin. The condition arises from a mutation on chromosome 4, encoding dentin sialoproteinphosphoprotein (DSPP), which constitutes approximately 50% of the non-collagenous component of the dentin matrix. How the mutated protein leads to near-complete obliteration of the pulp is not fully understood. Some patients with Osteogenesis Imperfecta show a clinically indistinguishable dental condition. The dental defect associated with Osteogenesis Imperfecta has been classified as Type I Dentinogenesis Imperfecta (Shields classification). Subsequent research confirmed that the two conditions differ clinically. On radiographs, anterior tooth roots appear short, thin, or thickened, while posterior teeth show a single short root.

Dentinogenesis Imperfecta – Type I

Type I is caused by a mutation in the DSPP gene (locus 4q21.3), encoding dentin phosphoprotein and dentin sialoprotein. It differs from Osteogenesis Imperfecta in that it affects only the teeth, with no bone fractures. It occurs in approximately 1 in 6,000–8,000 children. Witkop and Rao (1971) preferred the term “opalescent dentin” for this isolated condition. Teeth appear opalescent, ranging from blue-gray to yellowish-brown. Enamel may detach from dentin under occlusal stress. Sauk et al. (1976) reported increased glycosaminoglycan content in EDTA-soluble dentin and decreased content in EDTA-insoluble residues compared to controls. A deficiency of dentin sialoproteinphosphoprotein has been proposed as a contributing factor. Zhang et al. (2001) studied a Chinese family with Shields Type II Dentinogenesis Imperfecta across three generations and observed discoloration, severe clinical changes in the teeth, and obliteration of pulp chambers.

Dentinogenesis Imperfecta – Type II

First identified in Brandywine, southern Maryland. Pulp damage results from exposure of the crowns of primary and permanent teeth. Dentin appears yellow and smooth. Radiography of primary teeth in the first two years shows very large pulp chambers and canals. The widening of pulp chambers in primary teeth is not



seen in DGI-1. Witkop (1975) stated that the two conditions are identical, but later research showed that DGI-1 and DGI-2 result from allelic or two closely linked gene mutations.

Clinical Types of Dentinogenesis Imperfecta

Type I – Primary and permanent teeth appear amber-colored and semi-translucent. Enamel detaches easily. Primary teeth are more severely affected.

Type II (Stainton-Capdepon Syndrome) – Both primary and permanent teeth are equally affected. Crowns discolor and wear rapidly. Enamel fractures at the margins, which become sharp and may injure the oral mucosa. Exposed dentin results from enamel loss.

Type III – Presents similarly to Types I and II but in various forms.

According to the SHIELD3 classification, Dentinogenesis Imperfecta is divided into three types based on clinical, radiological, histological, and genetic characteristics:

SHIELD3 Type I

Osteogenesis Imperfecta with blue sclera, bone fragility, joint sounds, and reduced hearing. Characterized by a defect in the 1α chain of type I collagen.

SHIELD3 Type II

Defect in the gene on chromosome 4 responsible for dentin phosphoprotein synthesis. Dentin structure similar to Type I but without osteogenic effects.

SHIELD3 Type III (Brandywine Isolate)

Found in isolated communities, first identified in southern Maryland. Primarily affects primary teeth. Teeth are shell-shaped, initially white then turning amber-yellow, and are brittle. Radiologically, roots are short, cone-shaped, and canals are narrowed.



Figure 1. *Generalized opalescent tooth discoloration.*

Hereditary Amelogenesis Imperfecta (AI)

This is a very rare disease — a developmental defect of the tooth enamel tissue distinguished by several clinical forms, varied presentations, and different modes of hereditary transmission. The cause lies in hereditary developmental failures in the embryonic ectodermal layer. AI may appear as an isolated defect or as part of combined syndromes and chromosomal disorders. In this enamel defect, enamel changes color, becomes thinner, the crown dimensions decrease, and pits, oval notches, and grooves appear on the crown surface. These signs prompt aesthetic complaints from affected children. AI is caused by disturbances in the enamel matrix: the hypoplastic type involves disturbed enamel matrix formation; the hypomaturational type involves impaired normal maturation of the matrix; the hypermineralization type results from disturbed mineral saturation of the matrix.



Six Clinical Forms of Hypoplastic AI

1. Autosomal-dominant pit-groove AI: Pits and grooves of moderate size on the vestibular and buccal surfaces. Grooves run longitudinally from the cutting edge toward the cervical region. Enamel is thinned and stained by food pigments, which can be removed mechanically. Occurs in primary and permanent teeth.
1. Autosomal-dominant hypoplastic smooth AI: Enamel is uniformly thinned across all surfaces without altering crown shape, but reducing dimensions. Color changes from light yellow to light brown. Enamel is hard, smooth, translucent, and shiny. Fine cracks and tremas may be present; hypersensitivity may occur. Occurs in primary and permanent teeth.
2. Autosomal-dominant hypoplastic granular AI: Pits and grooves irregularly distributed on the crown surface. Intervening enamel is granular, of normal hardness, firmly attached to dentin. Hypersensitivity observed; incisal edges become blunt. Molar cusps appear sharp-spindle shaped. Over time dentin is exposed and discolors. Occurs in primary and permanent teeth.
3. Autosomal-dominant localized hypoplastic AI: Large oval pits and grooves individually placed without symmetric arrangement on vestibular and buccal surfaces. Depth of discoloration depends on defect depth and child's age. Occurs in primary and permanent teeth.
4. X-linked dominant hypoplastic smooth AI (in boys): Enamel slightly thinned in all areas, smooth, shiny; anterior teeth light yellow, posterior teeth yellowish-brown. Tremas between teeth; hypersensitivity and enamel wear. Affects primary and permanent teeth.
5. X-linked dominant hypoplastic groove AI (in girls): Longitudinal ridges and grooves on vestibular and buccal enamel surfaces. Enamel hardness intact; dentin may be slightly thinned where grooves occur. Affects primary and permanent teeth.

Other Forms of AI

Autosomal-recessive hypoplastic uneven AI: Newly erupted teeth show thin enamel that cracks, reducing crown dimensions. Enamel is best preserved on



contact and palatal surfaces. Anterior teeth are light yellow, posterior teeth dark brown. Small cracks on molar cusps; tremas and hypersensitivity in both primary and permanent teeth.

Autosomal-dominant hypomaturation pigmented AI: Enamel initially smooth and of normal thickness but brittle, loses shine, and discolors to yellow or brown (or chalky-opaque). Hypersensitivity develops in primary and permanent teeth.

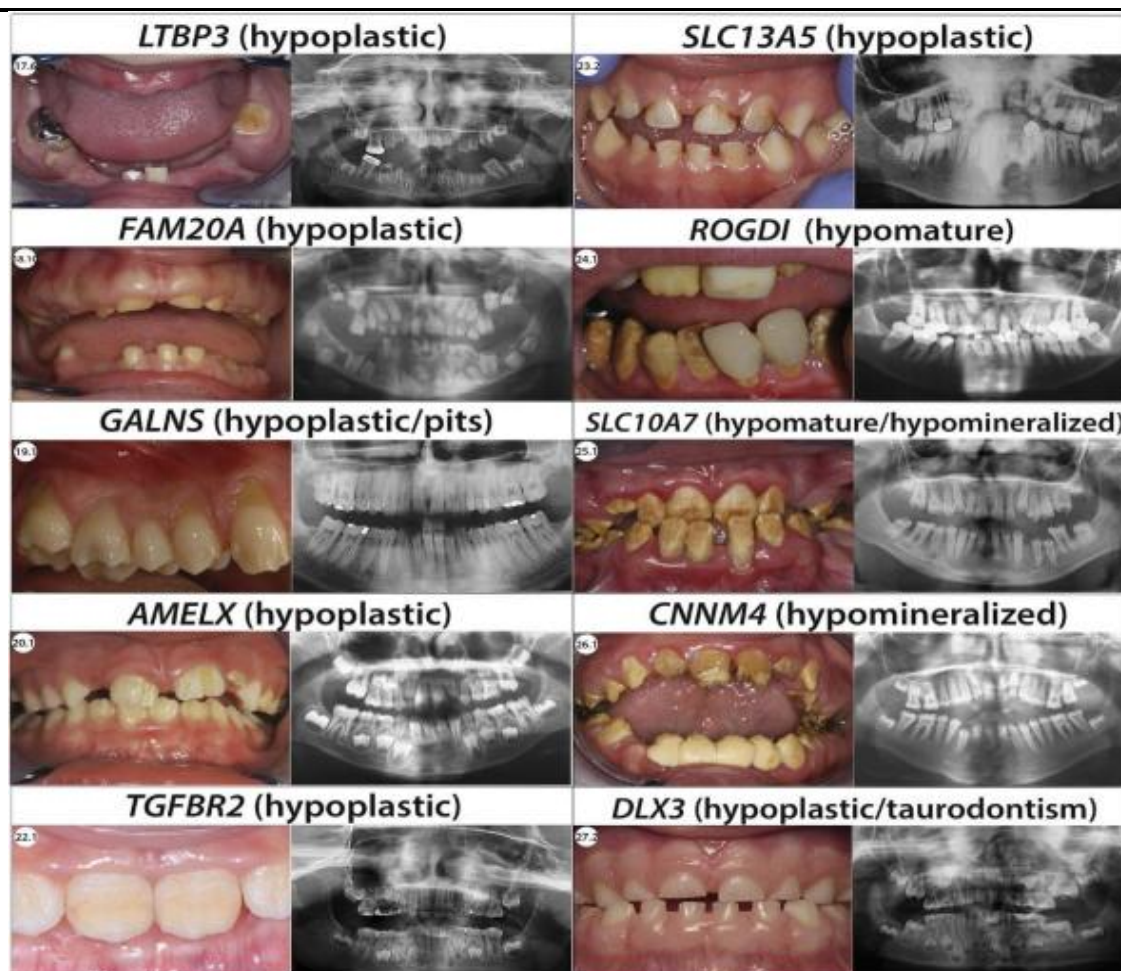
Autosomal-dominant hypomaturation “snow-cap” AI: Flat or clearly demarcated chalky-white opaque spots on vestibular or buccal surfaces. Crown enamel is smooth, hard, and of normal thickness, with no tremas or hypersensitivity. Most common on vestibular surfaces of upper anterior permanent teeth.

Autosomal-recessive hypomaturation pigmented AI: Smooth enamel of normal thickness with no tremas; color ranges from pale-opaque to pale-brown mainly on the vestibular surface. Enamel thins where spots are present. Occurs in primary and permanent teeth.

X-linked recessive hypomaturation groove AI (females only): Stripe-shaped enamel changes of varying lengths on the vestibular surfaces of homonymous teeth. Enamel color is opaque, chalky white, light yellow, or pale brown with slightly reduced hardness. Cracks and enamel fracturing may occur over time on occlusal surfaces. Affects primary and permanent teeth.

Autosomal-dominant hypomineralized AI (complete): Crown enamel initially smooth and of normal thickness, color from chalky-white to light brown with no tremas. Reduced enamel density is detectable with a probe. Over time enamel darkens, cracks, detaches, and severe pathological wear occurs. Occurs in primary and permanent teeth.

Autosomal-recessive hypomineralized AI (complete): Shortly after eruption, enamel is smooth and of normal thickness but markedly reduced in density; separates easily from dentin with an excavator. No tremas. Enamel quickly darkens; severe pathological wear and hypersensitivity observed. Enamel is preserved only at the cervical region.



Odontogenesis Imperfecta (Stainton-Capdepont Syndrome)

This disease arises from a disturbance in the activity of embryonic ectodermal and mesodermal layer tissues, leading to impaired development of both enamel and dentin. It occurs equally in boys and girls. Teeth erupt on schedule; even though tooth dimensions are not altered, enamel color is changed — usually gray, sometimes iridescent brown. Shortly after eruption, enamel cracks and dentin becomes exposed, rapidly wearing down until the contours of the tooth cavity become visible through the remaining tissue. The degree of wear depends on the child's age: the older the child, the faster the wear. Primary teeth wear more extensively and rapidly. Due to crown wear, bite disturbances and temporomandibular joint changes develop. On radiographs, the pulp chamber and root canals appear obliterated. Roots are short, thin, or thickened, with hypercementosis and focal bone osteoporosis in the apical region. Children



complain of cosmetic defects, tooth wear, gum bleeding, temporomandibular joint pain, and increased temperature sensitivity.

Osteogenesis Imperfecta

This is an autosomal dominant, very rare hereditary condition occurring equally in boys and girls. Frequent fractures of the long bones, clavicle, and ribs are typical. In young children, cranial suture closure is delayed; they lag in growth with hearing loss, blue sclera, and disturbances in tooth structure. Tooth dimensions and shape are normal, but color varies from gray and blue-gray to pale yellow or brown. Pathological wear of primary and permanent teeth is one of the constant clinical features. Radiographs show thinning of the cortical plate of the jaw bones and osteoporosis at root apices. As children age, obliteration of the pulp chamber and root canals is noted.

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