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DIAGNOSIS

Etiology of developmental enamel defects in the primary dentition

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Abstract Many systemic and local conditions can adversely affect the cells forming the enamel of the primary dentition, resulting in abnormalities that are permanently recorded on the tooth surface as changes in translucency (opacities) or reduced enamel quantity (hypoplasia). These conditions include genetic defects, such as amelogenesis imperfecta, congenital abnormalities of the liver and kidneys, premature birth, nutritional deficiencies, infections and local trauma. Developmental defects of enamel have significant clinical implications, such as reduced esthetics, tooth sensitivity and impaired masticatory function. Importantly, developmental enamel defects are now recognized as a major risk factor for early childhood caries.

Keywords Primary teeth · Dental enamel · Enamel hypoplasia

Quick reference/description

Abnormalities that originate during the formation of enamel are commonly referred to as developmental defects of enamel (DDE). During the development of enamel which occurs from approximately the 13th week of gestation to around 3 years of age, the primary dentition can be affected by many systemic and local environment insults that can affect the quality and quantity of enamel. As enamel does not remodel, the defects present on the surface provide permanent records of the changes that occur during the formation of the teeth.

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In the primary dentition, the prevalence of DDE ranges from approximately 20–30% in developed countries to over 40% in developing countries. Further, DDE is found in high prevalence in some groups of children, such as indigenous populations and the medically compromised.

Signs and symptoms

Opacities

Developmental defects of enamel associated with deranged enamel mineralization may be expressed as qualitative changes seen as alterations in the translucency or opacity of the enamel. Opacities may be diffuse or demarcated, and colored white, yellow or brown.

Enamel hypoplasia

Developmental defects of enamel arising from disruptions of matrix formation are known as enamel hypoplasia and may be expressed as pits, grooves and thin or missing enamel.

The presentation and severity of DDE is dependent on the stage of enamel development at the time and the extent and duration of the adverse condition (Table 1).

Clinical examination

Histories

Detailed family, medical and dental histories of children with DDE should be obtained. A positive family history of enamel defects may suggest a hereditary cause for the DDE, while the clinical appearance of the enamel and timing of medical conditions may help identify possible etiological factors. The dental history will help elucidate symptoms, complications and past treatment of the DDE.

Table 1 Developmental defects of enamel

Cause/ presentation	Developmental enamel defects	
	Quantitative enamel deficiencies	Qualitative enamel deficiencies
Cause	Disruptions of matrix formation	Altered enamel mineralization
Presentation	Expressed as:	Expressed as:
	Pits	Changes in the translucency or opacity of the enamel that may be diffuse or demarcated, and colored white, yellow, or brown
	Grooves	
	Thin or missing enamel	

Oral examination

A thorough examination of the teeth should be performed after cleaning and drying. Hypomineralization defects are diagnosed if there are diffuse or demarcated changes in translucency or opacity of the enamel (white, yellow, or brown in color). Pits, grooves, thin or missing enamel are suggestive of hypoplastic defects.

Intra-oral (bitewings and periapical films) as well as extra-oral (orthopantomogram) radiographs are useful for diagnosis of caries, DDE and associated developmental conditions.

Etiology

The complex cellular and biochemical events that occur during amelogenesis can be adversely affected by genetic and acquired systemic and local environmental conditions (Table 2). Genetic and systemic conditions tend to cause generalized DDE, whereas local factors are usually associated with DDE that are limited to a few teeth.

Table 2 Conditions associated with developmental defects of enamel in primary teeth

Generalized defects/ localized factors	Conditions
Generalized defects	Inherited conditions
	Amelogenesis imperfecta
	Generalized inherited disorder or syndrome with enamel defect
	Acquired
	Maternal conditions during pregnancy (e.g. cytomegalovirus infection, vitamin D deficiency)
	Birth trauma
	Preterm birth
	Isolated cleft lip and palate
	Nutritional deficiencies
	Renal and liver conditions (e.g. renal failure, biliary atresia)
	Celiac disease
	Endocrine, e.g., hypocalcemia, hypoglycemia
	Infections from bacteria, viruses and fungi
	Chemicals and toxins
	Other systemic conditions
Localized factors	Trauma (e.g. laryngoscopy)
	Radiation (e.g. cancer treatment)
	Local infection

Generalized defects

Hereditary conditions

Amelogenesis imperfecta

Amelogenesis imperfecta is a group of genetic conditions that affect mainly dental enamel, with a prevalence rate of between 1:7000 and 1:14,000. In amelogenesis imperfecta, both the primary and permanent dentitions are characterized by enamel defects. The inheritance may be autosomal dominant, recessive or X-linked. The defects in amelogenesis imperfecta may be generally classified into three types: hypoplastic hypocalcification or hypomaturation [1]. The hypoplastic types are characterized by features of enamel deficiency, such as pits, grooves or thin enamel, as shown in Fig. 1. By contrast, the hypocalcification and hypomaturation types are characterized by soft, opaque and stained enamel. In many cases, the severity of amelogenesis imperfecta may vary within and between the primary and permanent dentitions of an affected child. Further, dental variation among affected members within a family has also been reported [2].

Generalized inherited disorder or syndrome with DDE

In inherited dermatological syndromes involving the skin, hair and nails, such as ectodermal dysplasia, epidermolysis bullosa and tuberous sclerosis, both primary and permanent dentitions usually show generalized DDE. In addition, hereditary conditions with defects in vitamin D metabolism are often characterized by the presence of DDE. For example, children with vitamin D-dependent rickets often present with generalized enamel hypoplasia resulting from the chronic hypocalcemia present in this condition.

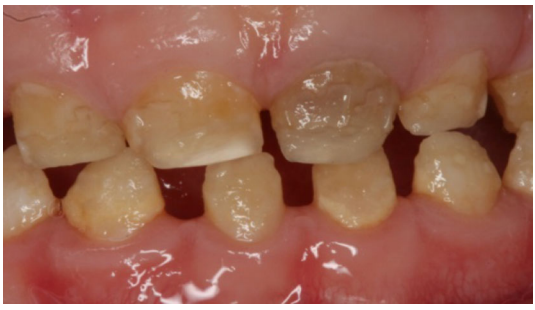

Fig. 1 A child with amelogenesis imperfecta showing generalized areas of missing enamel in all primary teeth

Acquired systemic conditions

Adverse perinatal conditions

The cells forming enamel (ameloblasts) can be adversely affected by perinatal conditions, such as hypoglycemia, hypocalcemia and neonatal tetany. Also, children with toxic levels of bilirubin (hyperbilirubinemia) resulting from congenital bile duct and liver diseases have DDE in the primary dentition. The teeth affected by hyperbilirubinemia often show a brownish green discoloration.

Preterm birth

Children who are born preterm with low birthweight have a high risk for DDE compared to children with a normal birthweight, most likely due to derangements of tooth formation and mineralization associated with organ immaturity. For example, deficient gastrointestinal absorption and inadequate supplies of calcium and phosphorus have been reported to cause severe DDE in the primary teeth of preterm children.

Isolated cleft lip and palate

Children with isolated cleft lip and palate are at risk to develop DDE from the local tissue disruption caused by the clefting anomaly on enamel formation.

Malnutrition and nutritional deficiencies

Malnutrition and nutritional deficiencies, particularly intake deficiency and gut malabsorption of vitamins A, C and D, as well as calcium and phosphorous may cause DDE in the primary dentition. Further, extended breastfeeding without solid supplementation is also thought to cause suboptimal nutrition that increases a child's risk for DDE.

Liver and renal conditions

Children with abnormalities of the liver and kidneys are at increased risk of DDE as the liver and kidneys are involved in the activation of vitamin D and the calcification of bone and teeth. Further, in congenital liver disease, high levels of circulating bilirubin can cause DDE due to their toxic effects on the ameloblasts.

Celiac disease and gastrointestinal malabsorption

The teeth of children with celiac disease are at high risk for developmental defects, most likely due to the reduced gut absorption of calcium and vitamin D.

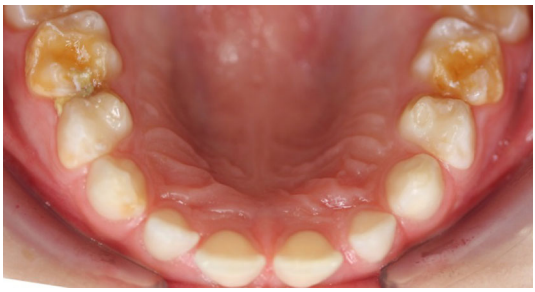

Fig. 2 Maxillary primary second molars presenting with enamel hypoplasia associated with a severe viral infection at approximately 15 months of age

Infections

Systemic infections during pregnancy and in infancy are associated with DDE in the primary dentition [3] (Fig. 2). Infective microorganisms may damage ameloblasts directly or indirectly by altering cellular function indirectly through their metabolic products. Further, the high temperatures experienced by children during infections may have adverse effects on the formation of enamel.

Chemicals and toxins

Ameloblasts are also highly sensitive to many chemicals and drugs, and exposure to agents such antibiotics (e.g. tetracycline and amoxicillin) and cytotoxic drugs may result in DDE. Further, intake of lead from environmental exposures, accidental ingestion or pica ingestion have also been shown to cause DDE in the primary dentition.

Localized factors

Trauma

Local oral trauma in a newborn may occur from laryngoscopy and endotracheal intubation that are used to treat respiratory distress. In a preterm child, a laryngoscope placed on the anterior maxillary alveolus during intubation may result in inadvertent pressure being exerted on the developing primary maxillary incisor teeth. The pressure can cause localized enamel hypoplasia or hypomineralization which is usually seen on the left-sided incisor teeth.

Radiation

Developing teeth exposed to radiation for treatment of tumors or leukemia are at risk for a range of developmental defects, including DDE. Damage to the ameloblasts presumably results from the direct effects of the radiation used for treatment.

Local infections

Local infections can cause DDE in the primary dentition [4]. As with radiation, disruption of ameloblastic function most likely occurs from the direct local infection and inflammation of the tissues.

Differential diagnosis

A positive family history of enamel defects and the presence of DDE in both dentitions in a child are suggestive of amelogenesis imperfecta. However, it is generally difficult to determine the etiology of acquired DDE from visual inspection as diverse etiological agents can produce the same presentation on a tooth surface. In some cases, the etiology of DDE may be identified from the patient's medical history and the presence of diagnostic features, such as specific intrinsic staining (e.g. greenish-brown discoloration of excessive bilirubin in chronic liver disease. Also, although rarely seen today, tetracycline ingestion during pregnancy and early infancy can cause DDE associated with brown discoloration in the primary teeth. Tetracycline stains characteristically show yellow fluorescence when exposed to ultraviolet light.

The location of a defect on the tooth surface may provide clues regarding the timing of the defect with respect to the chronology of dental development. For example, defects present on the incisal third of a primary incisor crown are suggestive of insults sustained during the first 12–20 weeks of pregnancy, and defects on the cusps of primary first molars may be associated with perinatal abnormalities. However, determination of the timing of the DDE may be problematic due to the wide variation in mineralization times of the primary teeth. Further, it is often difficult to accurately identify the specific etiologic agents responsible for the DDE.

Pitfalls and complications

Increased susceptibility to caries is an important complication of DDE in the primary dentition. DDE is now regarded as a major risk factor for early childhood caries which is one of the most significant conditions in pediatric dentistry [5].

DDE also greatly increases the risk for tooth wear and erosion due to thin and/or poorly mineralized enamel. Further, severe DDE in the anterior teeth can affect aesthetics, and tooth sensitivity from enamel hypoplasia may have detrimental effects on eating and tooth cleaning.

Further reading

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