Enamel hypoplasia associated with congenital hypoparathyroidism

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Abstract

Enamel hypoplasia is a defect in the enamel due to disturbance of ameloblastic function during amelogenesis. The etiology of such a disturbance may be either genetic or environmental in nature. This case report of undetected congenital idiopathic hypoparathyroidism emphasizes the importance of investigating the underlying cause of these enamel defects.

Introduction

Enamel hypoplasia is a defect in enamel that arises from a disturbance of ameloblastic function during amelogenesis and is characterized by a reduction in enamel thickness. Clinical manifestations may occasionally appear as pitting, furrowing and absence of enamel. Amelogenesis consists of two distinct phases: matrix formation and mineralization. The formation of the organic matrix begins when the first layer of dentin is formed and progresses in a rhythmic pattern of deposition until the entire thickness of the enamel is formed. Mineralization occurs in two stages with the first stage occurring as the matrix is laid down, resulting in partial mineralization (approximately 30%). The second stage, maturation, follows, and continues until the entire matrix is mineralized. Maturation begins before the matrix reaches full thickness, occurring on the inner matrix first while the outer matrix is being deposited. Maturation is an integration of two processes; 1) mineralization from the depth of the matrix to the surface, and 2) mineralization from the incisal to the cervical surface.

Disturbances in amelogenesis occurring during matrix formation will result in hypoplasia, whereas disturbances during mineralization will result in hypocalcification which is manifested clinically as opaque, chalky areas of enamel exhibiting normal contour. The etiological factors involved in enamel hypoplasia can be either genetic, or environmental. An environmental etiology can be either systemic or local. Generalized hypoplasia that is not of genetic origin must be evaluated for a systemic origin.

The systemic disturbances that can affect amelogenesis are nutritional deficiencies, febrile episodes, chemical intoxications, and endocrinopathies. The most frequent chronic disturbances associated with generalized hypoplasia of enamel are vitamin D refractory rickets and hypoparathyroidism. A case of severe enamel hypoplasia secondary to chronic undiagnosed hypoparathyroidism will be presented.

Report

The following is a case review of a 14-year-old white female who presented to the Department of Pedodontics at the University of Iowa for dental evaluation and treatment.

Medical History

The patient was a para 4, gravida 4, who was a product of a normal pregnancy and delivery. The mother reported that shortly after birth the child was very irritable and jumpy to tactile stimulation, including jerking of the head and flying of the arms. At 1½ weeks of age the mother noticed episodes of shaking and twitching of all extremities. After five or six days of these convulsive episodes the child was referred to Children's Memorial Hospital in Omaha, Nebraska, for evaluation. Clinical examination revealed normal development and the following symptoms: 1) a positive Moro reflex, 2) a positive Babinski sign bilaterally, and 3) hyperactive deep tendon reflexes. Laboratory examination revealed hypocalcemia and hyperphosphatemia. The child responded favorably to phenobarbital and calcium. She was discharged from the hospital with a diagnosis of tetany of the newborn and placed on daily Dilantin and calcium therapy with no further convulsive episodes. At age 1, the
medications were terminated, resulting in a recurrence of the convulsive episodes. The Dilantin and calcium therapy was reinstituted, resulting in the termination of the seizures. At age two, the medications were again discontinued with no future seizure activity.

Dental History

The mother reported that the primary teeth were somewhat delayed in eruption with the first tooth appearing at about 10 months of age. The mother stated that the primary teeth appeared “funny” but not to any degree that prompted her to seek consultation. At approximately three years of age, the child traumatized the maxillary incisors in a fall. They subsequently became excessively mobile and required extraction by the family dentist. The permanent incisors erupted at approximately age 10. No further dental care was rendered until age 14, when teasing at school made her self-conscious concerning the appearance of her teeth. At that time a dental consultation was sought from the family dentist in order to improve the esthetics of her dentition. It was the family dentist who recommended that a thorough physical evaluation be performed prior to dental care.

Physical Evaluation

The patient was evaluated by the Pediatric Department at the University of Iowa Hospitals and Clinics and appeared to be normal physically with the exception of the hypoplastic teeth and apparent slight mental retardation. Laboratory results, however, revealed hypocalcemia and hyperphosphatemia. The patient was then referred to pediatric endocrinology for a diagnostic evaluation. Further findings included brittle nailbeds with vertical fracture lines, calcifications of the lenses, calcification of the basal ganglia, a positive Chvostek sign, and a positive response to parathyroid hormone. A diagnosis of congenital idiopathic hypoparathyroidism was made. The patient was placed on daily calcium supplementation and dihydrotachysterol.

Dental Evaluation

The patient was referred to the Pedodontic Department at the University of Iowa for evaluation and treatment of the hypoplastic dentition. An investigation into the family history revealed no evidence of hypoparathyroidism or any dental disorders of a similar nature in any of the siblings or relatives going back three generations. The dental findings were: moderate to severe hypoplasia of the permanent dentition, very mild hypoplasia of the remaining primary dentition, and dental caries and shortened roots of the maxillary incisors which exhibited a significant degree of mobility (Figures 1-6B). The child exhibited dental delay as evidenced by the dental age which was calculated to be approximately 11 years, 6 months by the method described by Demirjian, et al., utilizing periapical radiographs, and was confirmed by the method described by Marshal, utilizing panoramic radiographs (Figure 5).

Dental Treatment

The treatment consisted of: 1) restoring the maxillary primary molars with amalgam, 2) adapting stainless steel crowns for the first permanent molars, 3) restoring the hypoplastic defects on the premolars and mandibular incisors to normal contour using an acid-
etch resin technique, and 4) placement of custom laminate veneers on the maxillary incisors for esthetics. These laminate restorations were placed in order to improve the appearance of the teeth, with full knowledge of the guarded prognosis of the maxillary incisors due to the presence of mobility and short roots (Figure 7).

The child’s future dental treatment may require placement of stainless steel crowns on the second permanent molars upon eruption, and, ultimately, permanent full coverage with cast crowns on all permanent molars when the permanent dentition is stable and the prosthetic needs are known.

Discussion

Enamel hypoplasia has been associated with the hypocalcemia that accompanies hypoparathyroidism, provided that it coincides with amelogenesis. The various hypocalcemic disorders that need to be addressed in a differential diagnosis will briefly be discussed.

Hypoparathyroidism

The parathyroid glands produce parathyroid hormone which acts to regulate the balance of calcium and phosphorus in the body. Decreased levels of parathyroid hormone result in an imbalance of the calcium/phosphorus ratio, leading to hypocalcemia and hyperphosphatemia. The characteristic symptoms of hypoparathyroidism are a direct result of the hypocalcemia. They include; hyperexcitation of the neuromuscular membranes, resulting in convulsions without loss of consciousness, tetany, and hyperreflexia. A positive Chvostek sign, which is the contraction of the facial muscles elicited by tapping over the facial nerve in front of the ear, is frequently present.

In addition to the signs and symptoms secondary to hypocalcemia, other clinical findings result from a lack of parathyroid hormone. These findings are; calcification of the lenses, cataracts, and calcifications of the basal ganglia. Several ectodermal signs may also be present and include varying degrees of alopecia, dry and scaly skin, and brittle fingernails and toenails that exhibit vertical fracture lines. Dental findings have been reported to include delayed eruption, shortening of roots and enamel hypoplasia, which occurs only when hypocalcemia is present during amelogenesis. Finally, physical and mental retardation may result if treatment is delayed for a prolonged period of time.

The signs and symptoms of hypoparathyroidism are similar to several clinical disease entities which can be distinguished by their history and onset. The syndromes of hypoparathyroidism that may well be
included in a differential diagnosis will be briefly discussed with respect to their distinguishing features.

**Infantile Hypoparathyroidism States**

*Transient idiopathic neonatal hypoparathyroidism* (tetany of the newborn) appears usually during the first few weeks of life. The symptoms are those related to hypocalcemia. Onset may be related to high levels of phosphorus in the diet (i.e. cow’s milk). The calcium/phosphorus imbalance is transitory and is quickly corrected with calcium and vitamin D supplementation.\(^\text{16,18}\) This entity was ruled out by virtue of the chronic nature of the patient’s condition.

*Sex-linked neonatal and infantile hypoparathyroidism* has an early onset at birth, with symptoms arising during the first week of life.\(^\text{16,21}\) The signs and symptoms are those found with hypoparathyroidism (previously discussed). This entity was rejected by the lack of genetic evidence.

**Hypoparathyroidism of Later Onset**

*Post-surgical hypoparathyroidism* develops as a result of damage to, or removal of, the parathyroid glands during thyroidectomy. Symptoms appear as a result of hypocalcemia.\(^\text{16,21}\) The absence of surgery rules out this possibility.

*Pseudohypoparathyroidism* is characterized by symptoms related to hypocalcemia. The parathyroid glands are normal or hyperplastic with normal function. The defect is primarily a failure of the end organs (kidney, skeleton) to respond to parathyroid hormone. Additional findings are: delayed growth, short and stocky build, a round face, generalized demineralization of bones, mental retardation, and a failure to respond to increased levels of parathyroid hormone.\(^\text{16,21}\) The positive response to parathyroid hormone precluded this possibility.

Pseudo-pseudohypoparathyroidism displays the usual anatomic stigmata of hypoparathyroidism, but with normal levels of serum calcium and phosphorus. This entity appears to be a sex-linked dominant trait and is thought to be a variant of pseudohypoparathyroidism. The lack of a genetic pattern and the laboratory data ruled out this entity.\(^\text{16,21}\)

*Idiopathic hypoparathyroidism of later onset* develops overt symptomology between 5 and 15 years of age. Symptoms may be manifested in infancy and the disease could most likely be detected early with proper laboratory investigations.\(^\text{16,21}\)

*Anticonvulsant drug therapy* is another factor which could influence the findings in this case due to the effect of anticonvulsant therapy on calcium metabolism.\(^\text{18}\) Patients receiving anticonvulsant therapy, especially diphenylhydantoin, exhibit a deficiency in 25-OHD\(_3\) resulting in a decreased level of serum-calcium.\(^\text{18,21}\) Severe hypocalcemia is seen in these patients with borderline intakes of vitamin D and little or no exposure to sunshine.\(^\text{31}\) It appears that significant alterations in calcium metabolism are the result of long-term anticonvulsant therapy. In fact, reports indicate that long-term anticonvulsant therapy can cause a state of drug-induced hypoparathyroidism.\(^\text{32}\) The possibility exists that a child with hypoparathyroidism may be incorrectly diagnosed as epileptic and placed on anticonvulsant therapy.\(^\text{18}\) This could superimpose a drug-induced state of hypocalcemia upon that already present from the hypoparathyroid disorder. In this case, the anticonvulsant therapy lasted for only two years and calcium supplementation was concurrent with the anticonvulsant therapy. The hypoplasia exhibited by this patient is inconsistent with the time frame of the anticonvulsant therapy.

The patterns of hypoplasia exhibited in this case are consistent with the timing and duration of the hypocalcemia which may have been mild during the first year of life when calcium supplementation was provided. However, the hypocalcemia may have been more pronounced as the child’s growth and calcium needs surpassed the level of supplementation from age one to two. The calcium requirements change rapidly during the first two years beginning with 360 mg from birth to six months of age, increasing to 540 mg until age one, and increasing again at age one to 800 mg, which remains constant until adolescence.\(^\text{34}\)

This may account for the early hypoplasia that occurred on the first permanent molar cusps and the permanent central incisors (which most likely were delayed in their development). One must assume that the child was sufficiently hypocalcemic from age 2 to 14, when no calcium supplementation was provided to cause enamel hypoplasia, but not to a degree to cause overt symptoms of hypoparathyroidism. The authors conclude by emphasizing the need for thorough investigation of cases that exhibit generalized hypoplasia of the primary or permanent dentitions, since underlying disease states may be present. The hypoplastic enamel may be the most overt clinical sign of the underlying disease.

Figure 7. Esthetic improvement with custom laminate veneers.
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References