12-year-old Male with Amelogenesis Imperfecta: a Case Report

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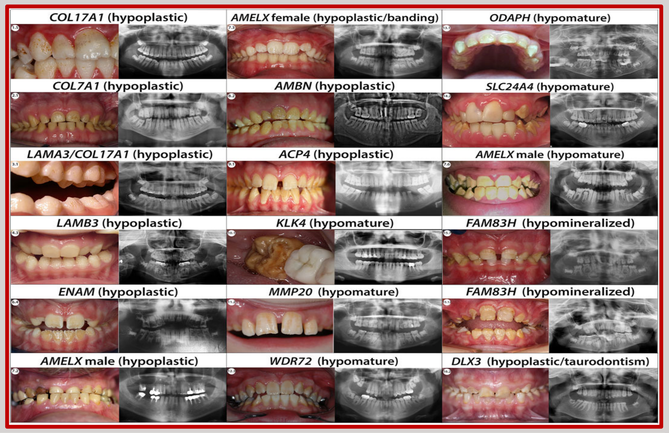
BACKGROUND

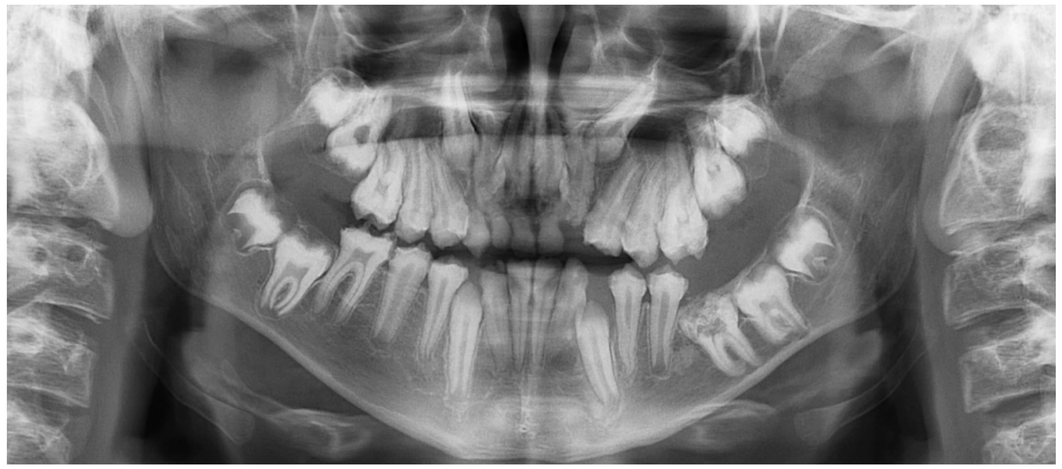
Amelogenesis Imperfecta (AI) is a complex hereditary disorder involving various conditions causing developmental alterations in enamel structure in the absence of a systemic disorder[1]. AI has an estimated frequency between 1:718 and 1:14,000 and may be inherited as autosomal dominant, autosomal recessive, or X-linked. With at least 14 different hereditary subtypes identified, AI exhibits a wide variety of clinical manifestations. In general, AI diffusely affects both deciduous and permanent dentitions[2]. AI may result from disruption and/or dysregulation of any one of the steps of enamel formation. The resulting defects can be divided into 3 categories: hypoplastic, hypocalcified, and hypomaturation[2]. There are several subtypes within each classification based on inheritance.



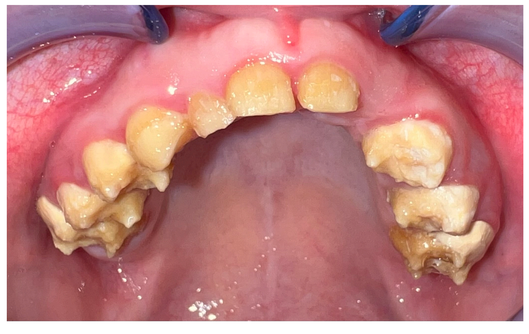
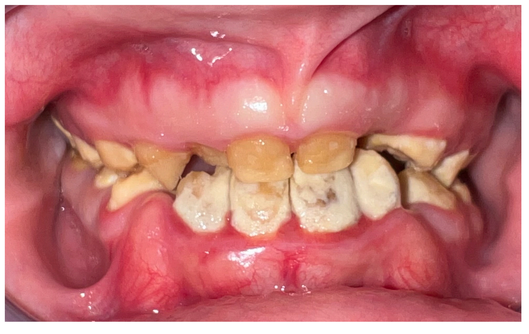
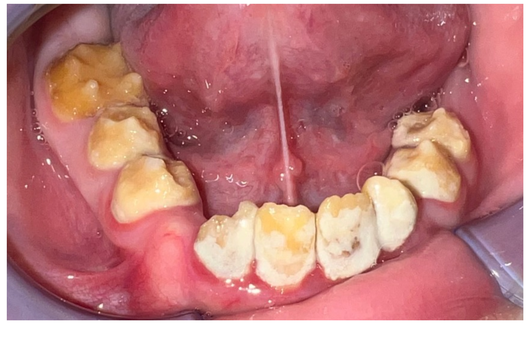
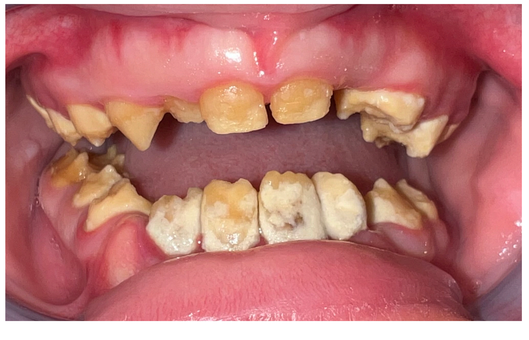
CLINICAL MANIFESTATION

Identification of AI can prove to be difficult due to the variability in appearance of the different types of AI. Some manifestations may appear normal clinically, while others present much more obvious and disfiguring[3]. Possible manifestations of AI include delayed, accelerated, and/or ectopic tooth eruption, permanent tooth impaction, anterior open bite, agenesis of permanent 2nd molars, and enlarged dental follicles[2]. Hypoplastic AI is associated with inadequate deposition of enamel matrix. Any enamel matrix present is appropriately mineralized with pits scattered across the surface of teeth. In contrast, with Hypocalcified AI, no significant enamel matrix mineralization occurs. Teeth are appropriately shaped on eruption, but with irregular, soft enamel that tends to chip from the underlying dentin. Hypomaturation AI is characterized by a defect in the maturation of enamel crystal structure. Affected teeth are also normal in shape on eruption, but with mottled, opaque, yellow-brown discoloration and soft enamel[1].

PANORAMIC RADIOGRAPH



CLINICAL PHOTOGRAPHS

CLINICAL PRESENTATION

A 12-year-old male presented to the Riley Hospital for Children Outpatient Dental Clinic for a comprehensive examination by referral due to the complexity of the patient's clinical presentation. Mom reports that the Amelogenesis Imperfecta diagnosis was confirmed through genetic testing and denies any other medical conditions or diagnoses. The patient reports frequent sensitivity to cool or warm liquids, chewing, and any tactile sensation to the teeth. Dental presentation: Mixed dentition (retained primary teeth #C, D, E, F and several unerupted permanent teeth) with all teeth exhibiting characteristics of hypocalcified/hypoplastic AI with notable severe loss of enamel resulting in misshapen and brown-yellow appearance. The dentition varies from rough/soft and depressible upon tactile examination. The patient initially presented with generalized heavy calculus throughout dentition. Radiographic exam revealed pathologically unerupted, impacted and/or misshapen teeth #6-11, 19.

TREATMENT/MANAGEMENT

It was determined that comprehensive dental treatment under general anesthesia in an operating room setting was the best modality to provide care for this patient. Issues to address include esthetics, dental sensitivity, loss of vertical dimension of occlusion, delayed eruption, tooth impaction, and retained primary teeth. The patient's specific severity and subtype of AI indicates full coverage treatment of all restorable dentition[2]. All retained primary teeth will be extracted along with the surgical extraction of malformed, impacted/unerupted tooth #19. Delay of full coverage treatment may result in loss of sufficient crown length, and full dentures may often become the only satisfactory treatment approach[3].

Type	Pattern	Specific Features	Inheritance
IA	Hypoplastic	Generalized pitted	Autosomal dominant
IB	Hypoplastic	Localized pitted	Autosomal dominant
IC	Hypoplastic	Localized pitted	Autosomal recessive
ID	Hypoplastic	Diffuse smooth	Autosomal dominant
IE	Hypoplastic	Diffuse smooth	X-linked dominant
IF	Hypoplastic	Diffuse rough	Autosomal dominant
IG	Hypoplastic	Enamel agenesis	Autosomal recessive
IIA	Hypomaturation	Diffuse pigmented	Autosomal recessive
IIB	Hypomaturation	Diffuse	X-linked recessive
IIC	Hypomaturation	Snow-capped	X-linked
IID	Hypomaturation	Snow-capped	Autosomal dominant?
IIIA	Hypocalcified	Diffuse	Autosomal dominant
IIIB	Hypocalcified	Diffuse	Autosomal recessive
IVA	Hypomaturation-hypoplastic	Taurodontism present	Autosomal dominant
IVB	Hypoplastic-hypomaturation	Taurodontism present	Autosomal dominant

REFERENCES

1) Neville B, Damn DD, Allen C, Bouquot J: Oral and Maxillofacial Pathology, ed 3, China, 2009, Elsevier, pp 99-106.
2) American Academy of Pediatric Dentistry Reference Manual 2011-2012. *Pediatr Dent*. 2011;33(6 Reference Manual): 226-331.
3) Roma M, Hegde P, Durga Nandhini M, Hegde S. Management guidelines for amelogenesis imperfecta: a case report and review of the literature. *J Med Case Rep*. 2021;15(1):67.