Amelogenesis imperfecta (AI) is a term for a clinically and genetically heterogeneous group of conditions that affect the dental enamel, occasionally in conjunction with other dental, oral and extra-oral tissues. It is a hereditary disorder with clinical impact on both deciduous and permanent teeth and first described in 1890.

In primary dentition the development of enamel starts at seventh week of gestation involving formation and secretions of an organic matrix containing the proteins amelogenin and enamelin. This is pursued by mineralisation of the matrix at nine weeks and subsequent maturation of this enamel with change in enamel protein predominantly from amelogenin to enamelin.

Amelogenesis imperfecta, the hereditary defect of enamel, occurs as a result of disturbance beginning at the organic matrix stage and continuing through calcification. The development of enamel is a multistep process, so the problem in any one of the steps may give rise to this defect. In general, the development of enamel can be divided into three major stages – elaboration of the organic matrix, mineralisation of the matrix, maturation of the enamel.

Witkop and Sauk listed the varieties of AI, divided according to whether the abnormality lay in a reduced amount of enamel (hypoplasia), deficient calcification (hypocalcification), or imperfect maturation of the enamel (hypomaturation), and also recognized the combined defects. Table-1

### Type I: Hypoplastic
- IA: Hypoplastic, pitted autosomal dominant
- IB: Hypoplastic, local autosomal dominant
- IC: Hypoplastic, pitted autosomal recessive
- ID: Hypoplastic, smooth autosomal dominant
- IE: Hypoplastic, smooth X-linked dominant
- IF: Hypoplastic, rough autosomal dominant
- IG: Enamel agenesis, autosomal recessive

### Type II: Hypomaturation
- IIA: Hypomaturation, pigmented autosomal recessive
- IB: Hypomaturation
- IIC: Snow capped teeth, X-linked
- IID: Autosomal dominant

### Type III: Hypocalcification

This clinical report describes the sequenced full mouth of a 5yr old patient with hypoplastic amelogenesis imperfecta along with a complete review that we diagnosed on the basis of clinical and radiographic features.
Clinical features of patients with AI depend on the type which is involved. AI has been classified on the basis of clinical, radiographic, and histologic appearance of the enamel defect and the mode of inheritance of the trait.4-7 AI has been categorized as hypoplastic (autosomal dominant/autosomal recessive/x-linked dominant) or hypomaturation (autosomal dominant/autosomal recessive) according to the degree of and site of enamel alteration. The radiodensity of the enamel is well mineralized but its amount is reduced and clinically, the enamel will be realized on the surface of the fine enamel, hypocalcified (autosomal dominant/autosomal recessive) shows pigmented, softened, and easily detachable enamel, hypomaturation types (autosomal recessive/x-linked recessive/autosomal dominant) shows pitted, grooved, and unbroken enamel, and hypoplastic-hypomaturation type affects the teeth exhibit mottled, opaque white-brown or yellow discoloured enamel which is softer than the normal.6,10,11

A variety of symptoms can be presented with AI. The most substantial findings comprise extensive loss of tooth tissue, tooth sensitivity, excessive attrition leading to short clinical crowns, and spaces in the anterior region of the dentition, normal or tight proximal contacts in the posterior region, and general enamel caries resistance.10,11

Literatures have investigated that mutations in five genes have been associated with amelogenesis imperfecta. Each gene can be mutated in a variety of ways, often creating diverse and distinct phenotypic patterns.2 Mutations in the amelogenin gene (AMELX) cause X-linked amelogenesis imperfecta, while mutations in the enamelin gene (ENAM) cause autosomal-inherited forms of amelogenesis imperfecta.2 Some reports involve that kalikrein-4 (KLK4) and MMP-20 cause the mutation of this gene which has been associated with the autosomal recessive, pigmented hypomutation variant of amelogenesis imperfecta, and DLX3 genes is in a group of genes that code for a number of proteins that are critical for craniofacial, tooth, hair, brain and neural development, mutation of this gene is associated with the hypoplastic-hypomaturation variants of amelogenesis imperfecta with taurodontism.2

AI may be inherited in an X-linked manner or as an autosomal dominant or recessive trait. However, there are cases where the diagnosis of AI remains tentative in apparently sporadic cases of enamel defects. Ultimately, it is anticipated that molecular genetic tools will allow more precise diagnosis.8

Treatment planning for patients with amelogenesis imperfecta is related to the age, socioeconomic factors, the type, and severity of the disorder and intraoral structures. An interdisciplinary approach may be required to evaluate, diagnose, and resolve the esthetic problem.12,13 The unbeaten management of AI during childhood requires the cooperation, preventive counselling, emotional support and motivation of the patient and parents.

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