

# Amelogenesis Imperfecta: A Case Report and Review of Literature

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## Abstract

Amelogenesis imperfecta (AI) is a hereditary disorder that causes developmental alterations in the structure of enamel. It represents a group of heterogenous conditions. AI has several names such as hereditary enamel dysplasia, hereditary brown enamel, hereditary brown opalescent teeth. In this disorder, the enamel is hypoplastic, hypomineralized or both. It may show autosomal dominant, autosomal recessive, sex-linked or sporadic pattern. It is necessary to diagnose this disorder and provide durable, functional and esthetic management of these patients to improve the quality of their lives. We present a case of AI affecting the dentition of an 18-year-old girl.

**Key words:** Amelogenesis imperfecta, Enamel, Hereditary, Management, Treatment

## INTRODUCTION

Amelogenesis imperfecta (AI) is a group of inherited defects of enamel that show clinical and genetic heterogeneity. It affects all or only some of the teeth in the primary or permanent dentition.<sup>1</sup>

In its mildest form, AI causes discoloration while in its most severe presentation; the enamel is hypo mineralized causing it to be abraded from the teeth shortly after their emergence into the oral cavity. Enamel findings are highly variable ranging from deficient enamel formation to defects in mineral and protein content.<sup>2</sup> Apart of enamel defects, AI has been also associated with abnormalities in dental eruption, congenitally missing teeth, anterior open bite, pulpal calcifications, root and crown

resorption, hypercementosis, root malformations, and taurodontism.<sup>3</sup> AI patients may experience compromised chewing function due to tooth sensitivity and the short clinical crowns caused by attrition and/or incomplete eruption.<sup>4</sup> No racial predilection of the AI has been reported. In the affected teeth, the dentin and root form are normal.<sup>5</sup>

Treatment aims to relieve pain or tooth sensitivity, to preserve as much tooth structure as possible while preventing further tooth loss, to maintain mastication and to improve the appearance because this has great psychological impact on patient's confidence.<sup>6</sup> This report presents the diagnosis and treatment planning of an 18-year-old girl whose chief concern was unaesthetic appearance of her teeth.

## CASE REPORT

An 18-year-old female patient reported to the Department of Oral Pathology, Shree Bankey Bihari Dental College and Research Centre, Ghaziabad with the chief complaint of yellow stains on all the teeth and chipping off of the teeth while biting hard food. Patient had these discolored

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teeth since childhood, and her milk teeth were similarly discolored too. Patient reported that she felt extreme dissatisfaction with the unesthetic appearance of her teeth. There was no family history, and medical history was non-contributory. Extra-oral examination did not reveal any relevant findings. On intra-oral examination, there was generalized yellowish discoloration of all the teeth along with diffuse pitting, more prominent on labial and buccal aspect and generalized attrition. The surfaces of the teeth were rough and irregular in shape and much smaller than normal with considerable tooth sensitivity (Figures 1-5). The emergence pattern and timing of teeth were normal. Examination of periodontium revealed a chronic generalized gingivitis with calculus deposition. On palpation, by probing resistance was felt and tooth material is soft in consistency with mild flaking of residual enamel. Based on history and clinical examination a provisional diagnosis of AI - hypo plastic type was made.

Patient was advised orthopantomogram that showed generalized thinning of enamel on all tooth surfaces and enamel was even absent in certain areas (Figure 6). Based on history, clinical findings, radiological report and histopathological findings the case was diagnosed as hypoplastic AI. Patient was advised oral prophylaxis and composite restoration or full-coverage crowns.



Figure 1: Rough pitted surface on labial surface of all the teeth



Figure 2: Left buccal segment



Figure 3: Right buccal segment



Figure 4: Maxillary occlusal view



Figure 5: Mandibular occlusal view



Figure 6: Orthopantomogram showing thin layer of enamel over tooth surfaces

## DISCUSSION

AI is a developmental, often inherited disorder that affects enamel. It occurs in the absence of systemic features and comprises of diverse phenotypic entities.<sup>7</sup>

AI can be subdivided at the clinical level into various forms depending on the type of defect and stage at which enamel formation is disturbed, into hypoplastic, hypo mineralized or hypo maturation type.<sup>8</sup>

In cases, with an X-linked form, the disorder results from a mutation in the amelogenin gene, AMELX. In cases of dominant forms of AI, the enamelin gene, ENAM, is implicated in the pathogenesis.<sup>9</sup>

In the hypo maturation type, the affected teeth exhibit mottled, opaque white-brown yellow, discolored enamel, which is softer than normal. In radiographs, thickness of enamel is normal, but density is the same as that of the dentin.<sup>4</sup>

The hypo calcified type shows pigmented and easily detachable enamel. Radiographically, enamel thickness is normal, but its density is even less than that of the dentin.<sup>4</sup>

In the hypoplastic type, the enamel is well-mineralized but its amount is less. Clinically grooves and pits are present on the surface of the fine enamel. The rough pattern of the hypoplastic type, exhibits thin, hard, and rough surfaced enamel. The tooth is tapered towards the incisal/occlusal face and has open contact points. Radiographs exhibit a thin peripheral outline of radiodense enamel and low or absent cusps. Clinical and radiographic appearances of the teeth of our case were similar to rough pattern hypoplastic type AI.<sup>5</sup>

AI can have a profoundly negative functional and emotional impact on individuals that may include pain and difficulty in eating as well as social avoidance, distress and low self-esteem. Dental care can be challenging and protracted.<sup>10</sup> The primary goal for the treatment is to address each concern as it presents with an overall comprehensive plan that outlines anticipated future treatment needs. Clinician treating children and adolescents with AI must understand and solve the clinical and emotional demands of these disorders with sensitivity. It is important to establish good rapport with the child and the family. Timely intervention is critical to spare the patient from psychosocial consequences of these disfiguring conditions. A comprehensive and prompt approach is reassuring to the patient and family and may help decrease their anxiety.<sup>1</sup>

The treatment options vary depending on several factors such as age of the patient, severity of the disorder, socioeconomic status and most importantly patient's age and cooperation. Numerous treatment modalities based on prognosis have been described for the rehabilitation of AI such as a combination of extractions, adhesive restorations, stainless steel crowns, overdentures, porcelain crowns and veneers after crown lengthening.<sup>11</sup>

Oral hygiene can be difficult for these patients due to the sensitivity while brushing. The use of warm water for tooth brushing will go some way towards relieving symptoms while rinsing.

Regular use of fluoride mouthwashes can help to reduce sensitivity and prevent caries.<sup>9</sup>

Root canal treatment as well as esthetic crown replacement for decayed teeth should be done to achieve the Jackson's triad of esthetic harmony, structural balance and functional efficiency.<sup>12</sup>

## CONCLUSION

AI affects the psychology of the patient negatively due to the aesthetic concerns. It should be promptly identified and treated, and existing dentition should be protected so that teeth can be conserved as much as possible. Oral rehabilitation can certainly provide a good prognosis provided there are less clinical complications. Patient should be counseled and motivated and must be taught how to maintain good oral hygiene, which goes a long way in maintaining the dentition.

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