

# Amelogenesis Imperfecta: Report and Review of a Rare Case

<sup>1</sup>Suryakanti Nayak, <sup>2</sup>Aprna Gupta, <sup>3</sup>Lipsa Bhuyan, <sup>4</sup>Saratkumar Nayak

## ABSTRACT

**Context:** Amelogenesis imperfecta (AI) is a hereditary ectodermal disorder, which is characterized by developmental alterations in the structure of enamel in the absence of any other systemic manifestations. It is transmitted through either an autosomal dominant, autosomal recessive, or X-linked mode of inheritance.

**Aims:** This study aims to report a case of AI and presents a review of the literature, to highlight the current knowledge about this condition.

**Settings and design:** A detailed case history was taken before clinical examination of general, extraoral, and intraoral features. Intraoral periapical and panoramic radiographs were taken.

**Conclusion:** Based on the patient's family history, and changes in the enamel structure as revealed by clinical examination and radiographs, the patient was diagnosed with amelogenesis AI.

**Keywords:** Amelogenesis imperfecta, Multiple impacted teeth, Pulp calcification, Teeth exfoliation.

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## INTRODUCTION

Amelogenesis imperfecta (AI) encompasses conditions characterized by quantitative or qualitative developmental abnormalities affecting the structure of tooth enamel, without any identifiable known systemic manifestations. Crown malformation and abnormal enamel density are typical manifestations.<sup>1</sup>

Both the primary and permanent dentitions may be involved. It is a purely ectodermal defect, which does

not affect mesodermal/ectomesenchymal structures; hence, dentin and root development will be normal. Interestingly, the affected teeth are more resistant to dental caries. No racial predilections of AI have been reported.<sup>2-5</sup> Amelogenesis imperfecta has been classified based on clinical, radiographic, and histologic appearance of the enamel defect and the mode of inheritance of the trait. Amelogenesis imperfecta has been categorized as hypoplastic (autosomal dominant/autosomal recessive and X-linked dominant), hypocalcified [autosomal dominant (AD)/autosomal recessive (AR)], hypomaturation (AR/X-linked recessive/AD), and hypoplastic-hypomaturation types.<sup>6</sup>

Hypoplastic AI represents 60 to 73% of all cases, hypomaturation AI represents 20 to 40%, and hypocalcification AI represents 7%.<sup>7</sup>

## CASE REPORT

A 20-year-old male reported to the Department of Oral Pathology and Microbiology, SCB Dental College and Hospital, Cuttack, Odisha with the complaint of pain and pus discharge from posterior teeth of both sides of lower jaw for 20 days. The patient reported frequent extractions from upper and lower arch following complaints of pain and pus discharge. The patient's father had got all his teeth extracted due to the same problem, but mother was not reported to be affected. The patient has one sister who also reported to have no problems. Medical history did not reveal anything relevant. The patient was of normally built; hair, skin, and nails appeared normal. There was no systemic problem.

Extraoral examination showed apparent mandibular prognathism and sparse facial hair. Intraoral examination revealed a narrow, high-arched palate, and reverse overjet with deep bite. Tongue was normal in size and shape. Gingiva was normal in color and consistency. Many teeth were missing in the right and left lower posterior region and upper arch as well. Most teeth were small in size, with open contact areas and a yellow brown hue. Severe attrition of teeth was present (Figs 1 and 2).

Right side intraoral periapical (IOPA) radiograph showed a tooth with short root, periapical radiolucency, and exposed pulp horn due to attrition (Fig. 3). Left side IOPA showed a tooth with attrition, short root, and

<sup>1</sup>Senior Resident, <sup>2,3</sup>Senior Lecturer, <sup>4</sup>Postgraduate Student

<sup>1,4</sup>Department of Oral and Maxillofacial Pathology, SCB Dental College and Hospital, Cuttack, Odisha, India

<sup>2</sup>Department of Oral and Maxillofacial Pathology, Hi-Tech Dental College and Hospital, Bhubaneswar, Odisha, India

<sup>3</sup>Department of Oral and Maxillofacial Pathology, Kalinga Institute of Dental Sciences, Bhubaneswar, Odisha, India

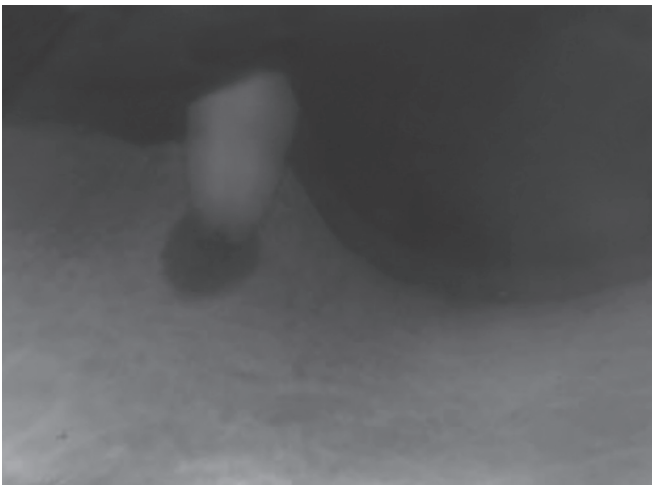
**Corresponding Author:** Suryakanti Nayak, Senior Resident Department of Oral and Maxillofacial Pathology, SCB Dental College and Hospital, Cuttack, Odisha, India, e-mail: suryakantinayak9@gmail.com



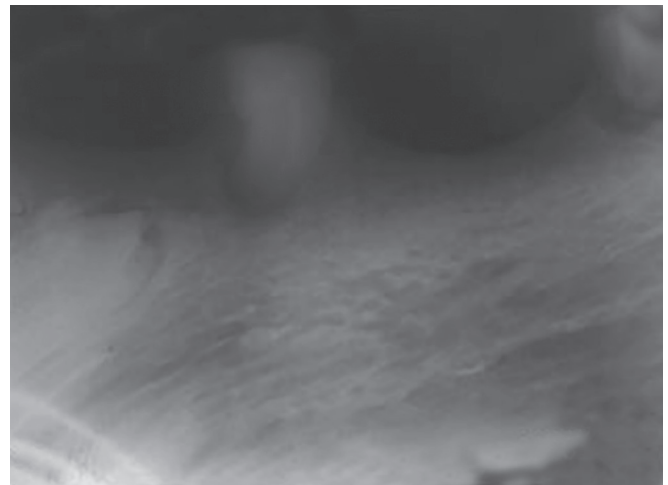
**Fig. 1:** Many missing teeth, small in size with open contacts and yellow brown hue



**Fig. 2:** Severe teeth attrition present



**Fig. 3:** Right side IOPA showing tooth with short root, periapical radiolucency and exposed pulp horn due to attrition



**Fig. 4:** Left side IOPA showed attrited tooth with short root and periapical radiolucency

periapical radiolucency (Fig. 4). Panoramic radiograph showed many teeth with stunted roots and malformed crown, multiple impacted permanent teeth, and congenitally missing teeth. Few teeth with pulp calcifications were also present. No peripheral enamel overlying the dentin could be appreciated in the radiographs, as the normal enamel–dentin contrast was absent. Multiple periapical radiolucencies were also seen, in relation to many attrited teeth (Fig. 5).

Based on the above clinical and radiological features, a diagnosis of AI was arrived at.

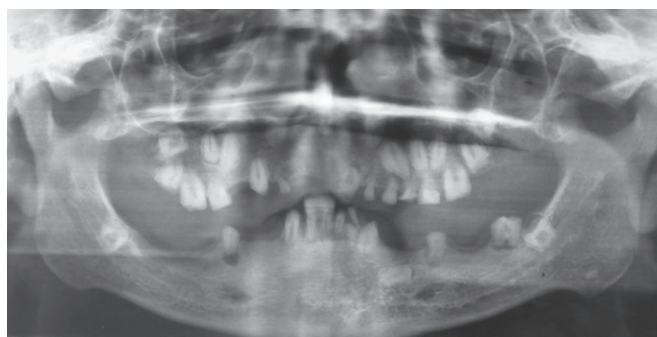
**DISCUSSION**

Amelogenesis imperfecta is a developmental, often-inherited disorder, affecting enamel. It occurs in the absence of systemic features and comprises diverse phenotypic entities.<sup>8</sup> At the clinical level, AI can be subdivided, depending on the type of defect and stage at which enamel formation is disturbed, into hypoplastic, hypomineralized, or hypomaturational type (Table 1).<sup>6</sup>

The prevalence of the condition has varied among studies conducted in different populations and has

**Table 1:** Classification of amelogenesis imperfecta (Witkop 1988<sup>6</sup>)

<i>Type I Hypoplastic</i>	<i>Type II Hypomaturational</i>	<i>Type III Hypocalcification</i>	<i>Type IV Hypomaturational–hypoplastic with taurodontism</i>
IA – Hypoplastic, pitted autosomal dominant	IIA – Hypomaturational, pigmented autosomal recessive	IIIA – Autosomal dominant	IVA – Hypomaturational–hypoplastic with taurodontism, autosomal dominant
IB – Hypoplastic, local autosomal dominant	IIB – Hypomaturational	IIIB – Autosomal recessive	IVB – Hypoplastic–hypomaturational with taurodontism, autosomal dominant
IC – Hypoplastic, local autosomal recessive	IIC – Snow-capped teeth, X-linked		
ID – Hypoplastic, smooth autosomal dominant			
IE – Hypoplastic, smooth X-linked dominant			
IF – Hypoplastic, rough autosomal dominant			
IG – Enamel agenesis, autosomal recessive	IID – Autosomal dominant?		



**Fig. 5:** Panoramic radiograph showing multiple impacted permanent teeth and congenitally missing teeth. No peripheral enamel overlying the dentin/E-D contrast absent

ranged from 1:718 to 1:14,000.<sup>7</sup> The distribution of AI types is known to vary among different populations. In a study in Sweden, 63% of the cases were inherited as AD. In contrast, in a study in the Middle East, the most common prevalent type of AI was found to be AR.<sup>9,10</sup>

Amelogenesis imperfecta may be associated with some other developmental abnormalities of dental and skeletal tissue, such as attrition, root resorption, taurodontism, delayed eruption of primary and permanent teeth, tooth impaction, dense invaginatus, pulp calcifications, severe malocclusion, and/or agenesis of teeth.<sup>5</sup> In cases with an X-linked mode of inheritance, the disorder results from a mutation in the amelogenin gene, AMELX. In cases with a dominant form of inheritance, the enamel gene, ENAM is responsible for the condition.<sup>11</sup>

In the hypomaturation type, the affected teeth exhibit mottled, opaque, whitish-brown or yellow discolored enamel, which are softer than normal. Radiographically, enamel shows normal thickness but the radiodensity will be less and comparable to dentin. The hypocalcified type shows colored and easily detachable enamel. Radiographically, enamel thickness is normal, but its density and thickness are even less than that of the dentin.<sup>12</sup>

In the hypoplastic type, the enamel is not only well mineralized but also less in quantity. Normal grooves and pits are present on the surface of enamel. The rough pattern of the hypoplastic type exhibits thin, hard, and rough-surfaced enamel. Teeth show tapering toward incisal and occlusal surfaces and often exhibit open contact points. Radiographs reveal radiodense enamel with a thin peripheral outline and cusps, which may be shortened or totally absent.<sup>13</sup>

Clinical and radiographic appearances of the teeth in our case were similar to hypoplastic type of AI. Amelogenesis imperfecta can have a negative functional and emotional impact on patients that may include difficulty in eating as well as distress, social avoidance, and low self-esteem.<sup>14</sup> Treatment planning for AI patients

depends on many factors – the age and socioeconomic status of the patient, the type and severity of the disorder, and the intraoral condition at the time the treatment is planned. Preventive aspects in the primary and mixed dentition include dietary advice, fluoride supplements, and oral hygiene instructions. Topical fluoride application can be done in the permanent dentition.<sup>15</sup>

## CONCLUSION

Amelogenesis imperfecta represents a group of developmental conditions, genomic in origin, which affects the structure and clinical appearance of enamel of all or nearly all the teeth. Amelogenesis imperfecta is a serious problem that can result in reduced oral health-related quality of life. Amelogenesis imperfecta affects the psychology of the patient negatively due to esthetic concerns. It should be promptly identified and treated and the existing dentition should be protected so that teeth can be conserved as much as possible. From this point of view, people with AI need extensive treatment. While planning for the treatment, the age, socioeconomic status of the patient, and type and severity of the disorder should be taken into consideration. Multidisciplinary approach is important for the success of the treatment if accompanied by other dental anomalies. Patients should be counseled and motivated to maintain good oral hygiene.

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