CASE REPORT AND A SHORT REVIEW OF AMELOGENESIS IMPERFECTA

ABSTRACT

Amelogenesis imperfecta (AI) is a group of genetic developmental disorders that cause abnormalities in the enamel structure and appearance of teeth. This condition poses a significant challenge as it can considerably affect an individual's oral health and lead to various physiological difficulties, thereby significantly reducing their quality of life. We present a case of a 17-year-old with amelogenesis imperfecta which was diagnosed based on clinical and radiographic features.

KEYWORDS: Amelogenesis imperfecta, amelogenesis imperfecta hypomaturation type, Amelogenesis imperfecta local hypoplastic type, dental enamel

INTRODUCTION:

Amelogenesis imperfecta (AI) is a group of genetic conditions that affect the formation and appearance of dental enamel. These conditions can cause changes in other tissues in the mouth and beyond. The term "amelogenesis imperfecta" is also used to describe enamel characteristics in related disorders.[1] Mutations in the genes responsible for amelogenesis can produce a range of diverse phenotypes with varying characteristics. Its occurrence is variable, affecting approximately 1 in every 700 to 14,000 individuals. [2-4]

CASE:

A 17-year-old male patient reported to the Department of Oral Medicine and Radiology, Kamineni Institute of Dental Sciences, Narketpally with a complaint of yellowish discolouration of all teeth since childhood. There was no relevant medical history. This is the patient’s dental visit. No history of any pernicious habits. The history did not reveal any eruption disturbances. A similar history of yellowish-brown discolouration of teeth was present in the patient’s elder sister since her childhood.

On examination, yellowish brown pigmentation of all the teeth is seen. The size of the teeth is normal. No loss of tooth-to-tooth contact. Soft tissue examination there was no abnormality detected. There is no tenderness on percussion. No chalky texture or chipping on scraping the teeth.

So, based on the history, clinical findings and morphology, a provisional diagnosis of amelogenesis imperfecta was given. A differential diagnosis of dental fluorosis, environmental enamel hypoplasia, and dentinogenesis imperfecta was given.

As a part of the radiographic examination, an orthopantamogram was taken (Figure 3). The patient's OPG showed all unerupted third molars and a normal teeth eruption pattern. However, there was prominent enamel loss in the posterior teeth.

The patient was advised pedigree plotting and full mouth rehabilitation.

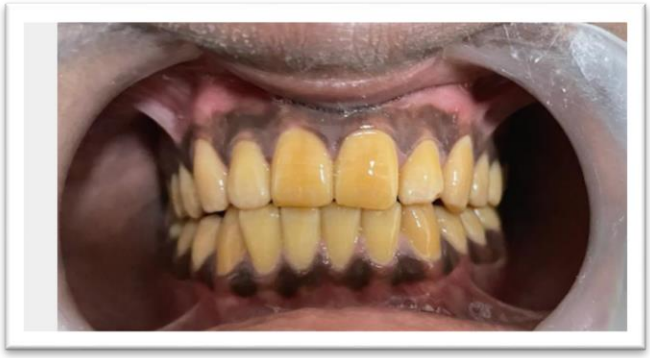

Figure1

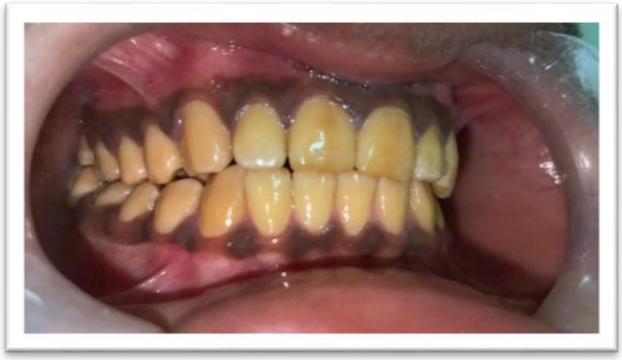

Figure 2

Figure 3

DISCUSSION:

AI is caused by mutations or altered expression in five genes: AMEL (amelogenin), ENAM (enamelin), MMP20 (matrix metalloproteinase-20), KLK4 (kallikrein-4) and FAM83H.Various classifications have been given dating back from the 1940s. Witkop's [2] classification is given below. (Figure 1)

Characteristics of hypoplastic AI

- Enamel of reduced thickness due to a defect in the formation of the normal matrix
- Pitting and grooves
- Hard and translucent enamel
- Radiographically, the enamel contrasts normally with dentine.

Characteristics of hypocalcified AI

- Defect in enamel calcification
- Enamel of normal thickness
- Weak in structure
- Appears opaque or chalky
- Teeth become stained and rapidly wear down
- Radiographically, enamel is less radio-opaque than dentine.

Characteristics of hypomaturation AI

- Enamel of normal thickness but mottled in appearance

- Slightly softer than normal and vulnerable to tooth wear, but not as severe as the hypocalcified type

- Radiographically, similar radiodensity as dentine.

Characteristics of hypomaturation-hypoplasia with taurodontism

- Mixed hypomaturation and hypoplasia appearance

- Taurodontism: body and pulp chamber enlarged, and the floor of the pulp chamber and furcation is moved apically down the root.

Individuals who have been diagnosed with AI and taurodontism are often found to suffer from Trichodentoosseous (TDO) syndrome. TDO is a condition that is inherited dominantly and is characterised by several symptoms, including splitting of the superficial layers of the nails, kinky or tightly curled hair, bone sclerosis of the long bones and skull base, zones of provisional calcification in the long bones, taurodontism, and enamel hypoplasia that occurs with hypomaturation/hypo calcification defects.[6]

To make an accurate diagnosis of enamel hypoplasia, a thorough patient history and clinical examination are essential to rule out any underlying systemic diseases. It is also important to identify the mode of inheritance through a family pedigree chart and to properly interpret radiographs. Early and accurate diagnosis enables effective genetic counselling, and precautionary measures can be taken to prevent further dental complications for the patient and any potentially affected family members.

Management is focused on prevention, restoration, and esthetics. Full mouth rehabilitation is a common treatment, along with dental hygiene maintenance supported by the dentist.

CONCLUSION

Amelogenesis imperfecta is a group of genomic conditions that affect the dental enamel. Early diagnosis with proper precautionary measures is required to prevent further complications of the disease, along with saving the potential family members who would be affected.

FIGURES AND REFERENCES

Figure 1:

Type I	Hypoplastic
Type IA	Hypoplastic, pitted autosomal dominant
Type IB	Hypoplastic, local autosomal dominant
Type IC	Hypoplastic, local autosomal recessive
Type ID	Hypoplastic, smooth autosomal dominant
Type IE	Hypoplastic, smooth X-linked dominant
Type IF	Hypoplastic, rough autosomal dominant
Type IG	Enamel agenesis, autosomal recessive
Type II	Hypomaturation
Type IIA	Hypomaturation, pigmented autosomal recessive
Type IIB	Hypomaturation, X-linked recessive
Type IIC	Hypomaturation, snow-capped teeth, X-linked
Type IID	Hypomaturation, , snow-capped teeth, autosomal dominant?
Type IIIA	Autosomal dominant
Type IIIB	Autosomal recessive
Type IV	Hypomaturation-hypoplastic with taurodontism
Type IV A	Hypomaturation-hypoplastic with taurodontism, autosomal dominant
Type IVB	Hypoplastic-hypomaturation with taurodontism, autosomal dominant

REFERENCES

1. Gadhia, K., McDonald, S., Arkutu, N. et al. Amelogenesis imperfecta: an introduction. Br Dent J 2012; 212, 377–379.
2. Witkop CJ Jr. Amelogenesis imperfecta, dentinogenesis imperfecta and dentin dysplasia revisited: Problems in classification. J Oral Pathol 1988;17:547-53.
3. Bäckman B, Holm AK. Amelogenesis imperfecta: Prevalence and incidence in a northern Swedish county. Community Dent Oral Epidemiol 1986;14:43-7.
4. Witkop CJ Jr, Sauk JJ Jr. Heritable defects of enamel. In: Stewart RE, Prescott GH, editors. Oral Facial Genetics. St. Louis: Mosby Co.; 1976. p. 151-226.
5. Wright J T, Hart T C, Hart P S et al. Human and mouse enamel phenotypes resulting from mutation or altered expression of AMEL, ENAM, MMP20 and KLK4. Cells Tissue Organs 2009; 189: 224–229.

6. Seow WK. Trichodentoosseous (TDO) syndrome: Case report and literature review. Pediatr Dent 1993;15:355-61.