Research Paper Medical Science

Amelogenesis Imperfecta- a Case Report with Genetic Influence

*** Basavaraj KF ** A U Madihalli**

*** Lecturer, Department of Oral Pathology &Oral Biology , Faculty of dentistry , Al-jabal Al-gharbi University, Gharian, Libya.**

**** Lecturer, Department of Maxillofacial Prosthosis , Faculty of dentistry , Al-jabal Al-gharbi University, Gharian, Libya.**

ABSTRACT

Amelogenesis imperfecta (AI) is a hereditary disorder expressing a group of conditions that cause developmental alterations in the structure of enamel. AI is a serious problem that reduces oral health-related quality of life and causes some physiological problems. The treatment of patients with AI may upgrade the quality of life and reinforce their self-esteem. The AI trait can be transmitted by either autosomal dominant, autosomal recessive, or X-linked modes of inheritance.

It is a diverse collection of inherited diseases that exhibit quantitative or qualitative tooth enamel defects in the absence of systemic manifestations. Also known by varied names such as Hereditary enamel dysplasia, Hereditary brown enamel, Hereditary brown opalescent teeth, this defect is entirely ectodermal, since mesodermal components of the teeth are basically normal.

It is necessary to diagnose the case and provide durable functional and esthetic management of these patients in order to improve the quality of their lives. We present a case of amelogenesis imperfecta affecting the dentition of a 13 year old Female.

Keywords : Amelogenesis Imperfecta (AI), Hereditary enamel dysplasia, Hereditary

INTRODUCTION :

Tooth enamel is the most highly mineralized structure in the human body, with 85% of its volume occupied by hydroxyapatite crystals1,2. The physical properties and physiological function of enamel are directly related to the composition, orientation, disposition, and morphology of the mineral components within the tissue3. Classifications of AI are primarily based on phenotype and mode of inheritance. The most commonly used classification was proposed in 1988 by Witkop, and revised by Nusier in 2004. Based on enamel appearance and hypothesized developmental defects, AI is classified as 4 patterns: hypoplastic, hypomaturation, hypocalcified, and hypomaturation-hypoplastic3. The disease is genetically and clinically heterogeneous, and autosomal dominant, autosomal recessive and X-linked inheritance were established in an extensive study of Swedish families with AI. However, there is no apparent correlation between the phenotype and the mode of inheritance5.

The aim of this paper is to present a case of amelogenesis imperfecta affecting a 13 year old female, with an insight on the genetic aspects of this disease.

CASE REPORT:

An 13-year-old girl [fig-1] reported with the chief complaint of discolored teeth since childhood. This little girl's parents did not seek any treatment previously, thinking that since the condition was not resulting in any other systemic manifestations and since she had inherited the condition from her father, there was little they could really do about it and accepted it as part of her appearance. It was only now, when they realized that the girl avoided hard food substances, they got her to a dentist. Apart from this, her past medical history was noncontributory. The history did not reveal any eruption disturbances. From a functional point of view, she had been avoiding hard food substances; at the same time, remaining caries free, except for a sole proximal carious lesion in 36, involving the enamel and dentin. On intraoral examination, it was found that she had a normal complement of teeth. The thickness of enamel was reduced on the teeth and was completely chipped off from some teeth exposing the dentin. The surfaces of the teeth were rough. The teeth, in general, exhibited a yellowish brown discoloration, with diffuse pitting present on the exposed tooth surfaces, more prominent on the labial and buccal aspects. The emergence pattern and timing of teeth seemed to be within the normal range. No open bite was present. Examination of the periodontium revealed the presence of chronic, generalized, marginal, and papillary gingivitis, with calculus deposition and unsatisfactory oral hygiene [Figures2]. The panoramic view showed all 32 teeth which were fully erupted. There was no retention of teeth, none of the teeth were missing. No pulp calcifications or external resorption was seen [Figure 3]. A diagnosis of amelogenesis imperfecta (hypoplastic type) was made.

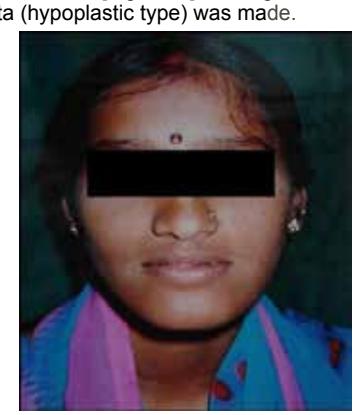

Fig:1

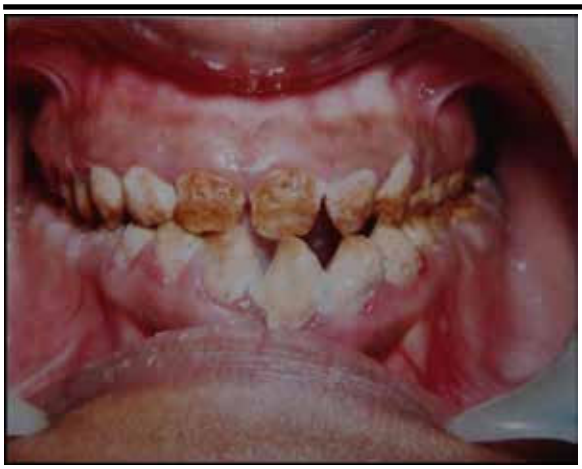

Fig:2

DISCUSSION:

Amelogenesis imperfecta (AI) encompasses a complicated group of conditions that demonstrate developmental alterations in the structure of the enamel in the absence of a systemic disorder. The prevalence of this condition has been expected to range from 1 in 718 to 1 in 14,000, depending on the population studies. Hypoplastic AI represents 60-73% of all cases, hypomaturation AI represents 20-40%, and hypocalcification AI represents 7%4.

The distribution of AI types is known to vary among different populations. In a study in Sweden, 63% of the cases were inherited as autosomal-dominant. In contrast, in a study in the Middle East, the most common prevalent type of AI was found to be autosomal-recessive6,7. 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 defects5.

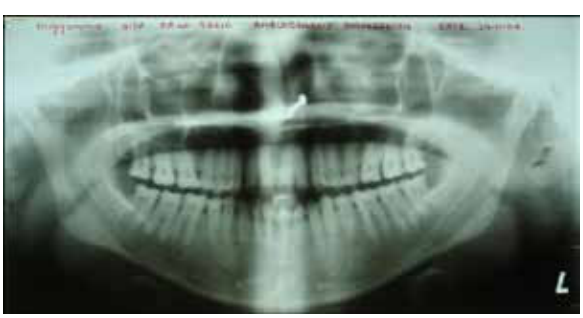

Fig:3

Early developing enamel matrix is rich in protein, but it successively loses its protein, and finally becomes highly mineralized. The matrix proteins are a heterogeneous group which are generally separated into amelogenins (major component) and enamelins. Enamelins have been suggested to function as nucleation sites for the hydroxyapatite crystals, while amelogenins are thought to regulate the rate, size and pattern of hydroxyapatite crystal growth. Two separate genes have been isolated for human amelogenin, one on the X chromosome, and one on the Y chromosome8. The histology of autosomal dominant hypomaturation-hypoplasia type of AI with taurodontism, definitively described by Winter *et al*., comprised of areas of severe hypomineralization with a pore volume of between 1 and 25%. They described a normal prismatic structure to the enamel, but with considerable post-calcification organic content and occasional bands of globular defects. The dentin was also reported as being defective, with a decreased number of tubules, an increased amount of intertubular dentin, dilatations, and cellular inclusions. All these findings were more marked in the radicular dentin. The pulp was normal, but enlarged in size9. Recently a gene encoding a second amleoblast specific protein, Enamelin has been mapped to the AIH2 critical region within 15kb of AMBN10. The enamelin gene, ENAM, has been mapped with different techniques to the same region on chromosome 4q as AIH2 and AMBN, suggesting that this region could contain a cluster of genes encoding enamel proteins. A splice site mutation in ENAM was recently found to be associated with autosomal dominant AI, where the patients presented with smooth hypoplastic enamel11.

Treatment for amelogenesis imperfecta includes replacement of the missing teeth and protecting the existing dentition with crowns and bridges. Though AI is unsightly and painful at times the oral rehabilitation has good prognosis, esthetically and functionally, if the situation is not complicated with too many anodontia or impacted teeth. Recently, new materials called second-generation laboratory composites, poliglasses or ceromers have been developed. They have different filler components that improve wear resistance, physical properties and their use has expanded into posterior intracoronal, full crown and even fixed partial denture restorations.

CONCLUSION

Amelogenesis imperfecta (AI) represents a group of developmental conditions, genomic in origin, which affect the structure and clinical appearance of enamel of all or nearly all the teeth in a more or less equal manner and which may be associated with morphologic or biochemical changes elsewhere in the body. Thus the dentist has to diagnose the condition as early as possible to offer early intervention and balance the decision for early intervention and long-term survival of the restorations. Dental practitioners should consider the social implications for these patients and intervene to relieve their suffering. Thus, this article is an attempt to improve the clinician's knowledge about the clinical diagnosis as well as intervention required for such a condition.

REFERENCES

[1] Foreman, D.E., Daniel, D.E., 1986. Permeation of compacted clay with organic chemicals. Journal of Geotechnical Engineering, ASCE 112(7), pp. 669-681. | [2] Gidigasu, M.D.,1976. Laterite Soil Engineering Pedogenesis and Engineering Principles. Elsevier Scientific Pub. Amsterdam.1976. | [3] Gnanapragasam, N., B.G Lewis, R.J Finno, 1995. Microstructural changes in sand-bentonite soils when exposed to aniline. Journal of Geotechnical Engineering, ASCE 121(7), pp.119-125. | [4] Khan, A.K., Pise, P.J.,1994. Effect of liquid wastes on the physico chemical properties of lateritic soils. Proceedings of Indian Geotechnical Conference, December 1994, Warangal, pp.189-194. | [5] Khan, A.K., Rao, C.U., Bandyopadhyay, M. Characteristics of leachates from solid wastes, Indian Journal of Environmental Health, 1994 36(4), pp.248-257. | [6] Mesri, G., Olson, R.E., 1970. Shear strength of montmorillonite. Geotechnique 20(3), pp 261-270. | [7] Nayak, S., B. M. Sunil, S. Shrihari. Shear strength characteristics and chemical characteristics of Leachate contaminated lateritic soils. A journal engineering geology 106,2009. pp 20 – 25. | [8] Ravishankar, R., Madhuri, B., Mugeraya, 2004. Characterization of the MSW leachate at Mangalore dumpyard. NITK Research Bulletin 14 (1), pp 20-25. |