AUTOSOMAL RECESSIVE ROUGH HYPOPLASTIC TYPE-1 AMELOGENESIS IMPERFECTA

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ABSTRACT: Amelogenesis Imperfecta (AI) is a severe disorder affecting patient's quality of life with relation to their oral health and may have serious psychological impact on them. From this point of view, people with AI need extensive treatment. While planning the treatment, several factors must be taken into account like patient's age, their affordability, pattern and the nature of AI. Moreover, although rare, some dental anomalies may accompany the AI cases.

This case report presents restorative management of an Autosomal Recessive Rough Hypoplastic AI. The patient was looking for a cost effective solution of her aesthetic problem and replacement of her missing teeth. The management of her case involved direct and indirect restorations. Contemporary treatment modalities comprising of adhesive restorative techniques, removable partial dentures and stainless steel metal crowns were used in this case report. Patient reported her comfort with the restorative treatment on her follow up visits.

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INTRODUCTION

There are many developmental defects which have genetic source. Some of them affect almost all the teeth in a more or less equal manner by altering the configuration and form of enamel. Amelogenesis imperfecta (AI) represents one such group, which may be related to structural or organic changes occurring somewhere else in the body. According to the populations studied, its incidence fluctuates from 1:700 to 1:14,000. The presentation of the defected enamel may be hypoplastic, hypomineralised or both resulting in sensitive, discolored dentition which is susceptible to breakdown. AI may occur alone or may be linked with other conditions. It can be of autosomal dominant, autosomal recessive, sex-linked or sporadic type. Amelogenesis imperfecta (non-syndromic form) has been found to be caused due to mutations in certain genes such as AMELX, MMP20, KLK-4 and ENAM.

These genes provide instructions for the synthesis of certain proteins that are responsible for normal tooth development including the normal enamel synthesis, which is a hard, calcium-rich protective outer layer of the teeth. Alteration in any of these genes changes the structure of these proteins interfering with their function and therefore prevents the genes from making any protein at all. As a result, the tooth enamel produced is unusually soft, thin and discolored and damages very easily.

Amelogenesis imperfecta can also be acquired in an autosomal recessive pattern resulting usually from mutations in the ENAM or MMP20 gene. In this type, mutation occurs in two copies of the gene in each cell. AI is mostly inherited in an autosomal dominant pattern which results due to an alteration in the ENAM gene. In this type, mutation in even one copy of the gene in each cell is enough to cause the disorder. Few other types of AI may result due to mutation in certain new genes or in individuals with no previous family history of this condition.

AI causes a lot of clinical problems including teeth that are very sensitive due to defective enamel. There is a loss of occlusal vertical dimension and dysfunction with compromised aesthetics. A timely treatment of these defects results in a positive psychological impact on the

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patient because of improved esthetics and function. Aberrations are seen in tooth number, crown morphology, pulp-dentine tissue, and in the eruption process. Gingival conditions and oral hygiene of such patients is usually poor, with calculus being found frequently. Patients may suffer from malocclusions.

Amelogenesis imperfecta may also present in the hypoplastic form, in which the enamel may be pitted, rough, or with irregular vertical ridges of normal and defective enamel. There is an inadequate depth of the enamel seen in this type of AI, resulting in a lack of interproximal contacts. Current case report presents restorative management of a case of type 1 AI.

CLINICAL REPORT

A 17 year old female patient was referred to the Prosthodontics department, Fatima Jinnah Dental Hospital, Karachi, for the replacement of missing anterior teeth. The girl was worried about the appearance of her teeth and wanted an immediate replacement. The girl was later referred to the Operative department because of the general appearance of her remaining permanent anterior teeth which lacked the normal form and thickness of enamel.

The clinical examination showed hypoplastic 13, 21, 22, 23, 31, 32, 33, 41, 42, 43 and all four permanent first molars. Affected teeth were thin, small teeth with rough and pitted enamel surface. Crown size was smaller than the normal with lack of proximal contact and anterior open bite affecting the upper and lower anterior teeth along with first permanent molars. Second molars and premolars however, were spared. The teeth no. 11 and 12 were missing and exhibited a significant loss of horizontal and vertical bone volume (Fig no 1 to Fig no 5). According to the patient the missing teeth never erupted. The oral hygiene of the patient was poor with hyperemic and edematous gingiva. Medical history and extra oral examination was non-contributory. A working diagnosis of Autosomal Recessive Rough Hypoplastic Type-1 Amelogenesis Imperfecta was made. Radiograph examination revealed periapical infection in tooth no 31.

An ideal treatment plan that included orthodontic repositioning of teeth to more favourable location
followed by ceramic veneer on anterior teeth and lithium disilicate crowns on first permanent molar, placement of implants and guided bone regeneration to replace congenitally missing right central and lateral incisors was turned down by patient owing to financial constraints and lack of time. Our alternate and cost effective treatment plan accepted by the patient included improvement in oral hygiene, direct composite restorations, full coverage metal crowns for the affected teeth and a removable partial denture for missing teeth.

Oral hygiene instructions, scaling, and root planning were done to improve her periodontal status. Two weeks later, the gingival edema and hyperemic appearance of gingiva resolved and bleeding on probing returned to normal.

All permanent first molars were prepared to receive full metal crowns, impression was performed with polyvinyl siloxane putty and light body (3M ESPE) in a single step technique using stock trays. The preps were temporized with self cure acrylic crowns. 5 days later full metal crowns were cemented on all the first molars using a type 1 GIC cement (GC-Gold label) (Fig no 6 & 7). Composite veneers were done on all the affected anterior upper and lower teeth using Ceram X mono composite (Dentsply) shade M1 without any preparation.

No attempt was made to close the proximal gaps between the teeth as there was not much tooth structure to support such restoration. The idea was to strengthen the remaining tooth structure, improve esthetics and to protect the teeth from further damage.

Upper and lower jaw impressions were taken with alginate for the fabrication of a partial denture for missing teeth 11 and 12. A wax trial was done to check for shape and color of the teeth. The acrylic partial denture was inserted and appropriate occlusal adjustment was done (Fig 8,9,10 & 11).
The patient was given instructions on cleaning of interproximal areas and partial denture. The patient was reexamined after 1 week. The patient was recalled for follow-up visits at 3 and at 6 months. No issues were noticed during this period related to her esthetics or function.

DISCUSSION

The presented clinical situation was rare and required use of treatment to protect and strengthen the affected teeth as well as improve esthetics and restore function. The patient was looking for a cost effective solution of her esthetic problem and replacement of missing teeth. She was not aware of the compromised structure of her first permanent molars and hypoplastic anterior teeth and therefore acceptance of treatment for those teeth required her understanding and cooperation. The approach to this situation was made by discussing the treatment options along with material available for veneering her anterior teeth for giving her esthetic results, with the patient and parents.

In cases of hypoplastic AI, the enamel is properly mineralized, but its amount is deficient. The fine enamel surface is thin, hard, and rough due to presence of ridges and grooves. The teeth have crowns tapering downwards with deficient contact points. A thin border of radiopaque enamel is seen on radiographs with very low cuspal height or entirely absent cusps of the teeth. Clinically and radiographically, our case was harmonious with rough pattern hypoplastic type AI.

Many other dental and skeletal developmental defects may be accompanied with AI, such as taurodontism, root resorption, attrition, dens in dente, pulp stones, tooth impaction, anterior open bite and agenesis of teeth. The eruption of the teeth may be delayed in such cases and sometimes teeth may not be formed entirely. In our case, missing teeth, disturbed eruption, anterior open bite were present.

The gingiva of our patient was hyperemic and edematous. Until now, 40 papers have been published reporting the gingival conditions of patients with AI, and 28 of them had edematous and hyperemic gingivae. This condition is perhaps contributed by mouth breathing and reluctance to brush because of sensitive teeth. Along with poor oral hygiene, these factors adversely affect the prognosis of the prosthetic treatment. The oral health of our patient was maintained after conventional periodontal therapy.

The management of amelogenesis imperfecta usually involves direct and/or indirect restorations. Contemporary treatment modalities comprising of adhesive restorative techniques, overdentures, fixed partial dentures, full porcelain crowns, porcelain fused-to metal crowns and inlay/onlay restorations have been reported in literature.

Full porcelain restorations are becoming increasingly popular, because of their improved esthetics, excellent biocompatibility and improved physical properties.
The recent advances in dentin bonding have revolutionized the field of esthetic dentistry. The practitioners can now predictably restore the function and esthetics to quite a satisfactory level\cite{10,17,18}. However, the main disadvantages of laminate veneers have been identified as marginal adaptation and bonding problems (16).

In the present case, full-metal restorations on posterior first molars were preferred to redouble the mechanical durability, recover strength and protect the residual dentin. The porcelain laminate veneers were our first option to improve the esthetic outcome, but were refused by our patient due to financial constraints. Patient reported her comfort with the restorative treatment on her followup visits.

**CONCLUSION**

In conclusion, AI is a severe disorder affecting the patient's quality of life with relation to their oral health and may have serious psychological impact on them. From this point of view, people with AI need extensive treatment. While planning the treatment, several factors must be taken into account like patient's age, their affordability, pattern and the nature of AI. Moreover, although rare, some dental anomalies may accompany the AI cases. In these cases, multidisciplinary approach is important for treatment success. In the present case, the patient tolerated the use of partial denture and restorations well at routine follow up visits.

**REFERENCES**