Amelogenesis imperfecta: Diagnosis and resolution of a case with hypoplasia and hypocalcification of enamel, dental agenesis, and skeletal open bite

Rosa Pulgar Encinas, DDS, MD, PhD[1]/Ignacio García-Espona, DDS, MD, PhD[2]/
Jose Manuel Navajas Rodriguez de Mondelo, DDS, MD, PhD[3]

A case of amelogenesis imperfecta with hypoplasia, hypocalcification of the enamel, congenital absence of teeth 12 and 22, delayed eruption of tooth 23, edge-to-edge incisal relationship, open bite, and bilateral posterior cross bite at the level of the first and second premolars is presented. Lateral skull teleradiography indicated a Class III skeletal pattern of maxillary origin associated with a dolichofacial pattern with multiple indicators of facial hyperdivergence. The patient presented a major esthetic abnormality of the face and required orthodontic treatment prior to a prosthetic solution with full-coverage metal-ceramic crowns in both the maxilla and the mandible. The diagnosis of cases such as this one and the therapeutic implications from an orthodontic and prosthetic standpoint are reported. *(Quintessence Int 2001;32:183–189)*

Key words: amelogenesis imperfecta, dental abnormalities, dental agenesis, enamel hypoplasia, hypodontia, open bite, oral rehabilitation

Amelogenesis imperfecta (AI) is one of a group of abnormalities known in dental pathology as *hereditary dysplasia*. The term is currently reserved for congenital enamel defects that primarily affect only enamel formation and are not accompanied by morphologic or metabolic disorders in any body system other than tooth form or eruption.[1] Amelogenesis imperfecta has been associated with inclusions and abnormalities in dental eruption, congenitally missing teeth, anterior open bite, pulpal calcifications, dentin dysplasias, root and crown resorption, hypercementosis, root malformations, and taurodontism.[2,3]

Amelogenesis imperfecta was first reported in 1890 but was not considered a clinical entity distinct from dentinogenesis imperfecta until 1938.[4] Its prevalence varies widely between studies, from 1 in 14,000[1] to 1 in 4,000,[5] depending on the diagnostic criteria used and the population group studied.

[1]Assistant Professor of Restorative Dentistry, Stomatology Department, University of Granada, Granada, Spain.

[2]Associate Professor of Orthodontics, Stomatology Department, University of Granada, Granada, Spain.

[3]Professor of Restorative Dentistry, Stomatology Department, University of Granada, Granada, Spain.

Reprint requests: Dr Rosa Pulgar Encinas, Avenida. Constitución nº 41 3ºB, 18014-Granada, Spain. E-mail: rpulgar@platon.ugr.es

There are numerous classification systems, and the most widely accepted is that proposed by Witkop and Sauk in 1976,[6] which considers the inheritance pattern of the disorder as well as its specific clinical characteristics. However, this system was recently challenged by Aldred and Crawford,[7] who proposed a new classification that not only evaluates the phenotype but also the molecular disorder, the biochemical composition of the enamel, and the mode of inheritance of the defect.[7]

Despite recent advances in the identification of the molecular defects responsible for the appearance of AI, there is still scant information on the phenotype and genotype of this group of disorders. In some reported cases, the hereditary pattern is autosomal dominant; in others, it is recessive. There are also reports linking AI to chromosome X. The only molecular defect thus far identified with the appearance of AI is related to the gene that codifies amelogenin, the most abundant protein of the original matrix.[8] It has been suggested that alterations in the process of degradation and resorption of this matrix contribute to the characteristic hypomineralization of the affected enamel.[9]

A case that illustrates the difficult diagnosis of some of the clinical subtypes of AI and presents multiple concomitant abnormalities that largely determined the therapeutic program followed is reported.

Figs 1 to 3 Views of a 15-year-old female patient diagnosed with amelogenesis imperfecta at initial presentation.

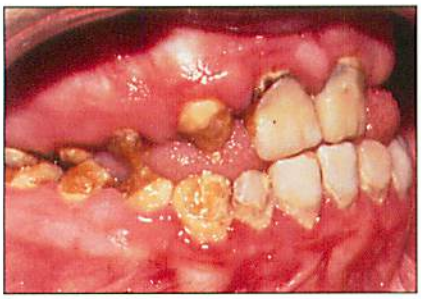


Fig 1 Right lateral view.

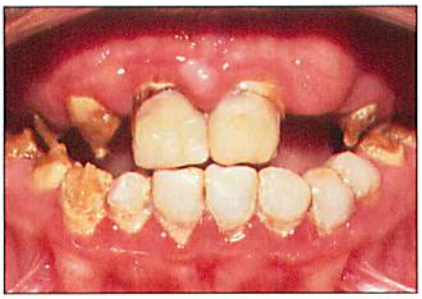


Fig 2 Frontal view.

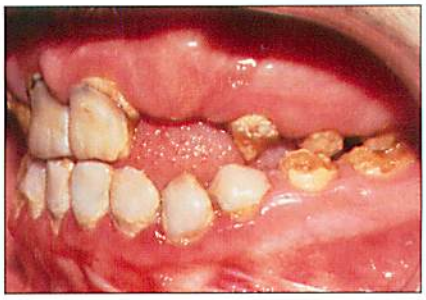


Fig 3 Left lateral view.

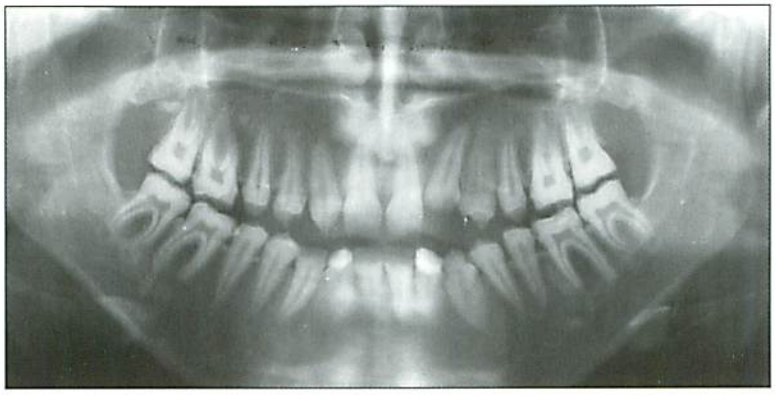


Fig 4 Panoramic radiograph.

CLINICAL REPORT

In 1995, a 15-year-old patient with AI was seeking treatment to improve the esthetic appearance of her mouth. She had previously been treated with resin composite restorations on the buccal surface of teeth 11[8], 21[9], 31[24], 34[21], 35[20], 41[25], and 42[26]. Her medical records showed a history of an intense hypersensitivity. Although the patient had no history of extractions, teeth 12[7], 22[10], and 23[11] were absent. Palpation revealed the presence of tooth 23. The erupted teeth, including premolars and molars, were very small, of abnormal morphology, brownish in color, and very rough and friable on the surface. The superficial dental layer became detached with only light pressure. There were no proximal contact points between the teeth, and a Class III occlusion with lateral open bite was recorded. Gingivitis with hyperplasia was noted at the level of the maxillary interincisal papilla (Figs 1 to 3).

The panoramic radiograph (Fig 4) confirmed that teeth 12 and 22 were congenitally missing and that tooth 23 was unerupted. It showed no intrapulpal calcifications. The teeth were deformed, with reduced crown size and a very fine film of enamel only slightly more radiopaque than the dentin in some areas of certain teeth.

After bacterial plaque was controlled, the patient was referred to the orthodontist for evaluation of the malocclusion. The orthodontist indicated improvement of the vertical disorder in order to prepare for subsequent prosthetic treatment, since lateral open bite was combined with major reduction of the posterior vertical skeletal dimension. The patient was classified as having Class I molar relationship with an edge-to-edge incisal relationship and bilateral posterior cross bite at the level of the first and second premolars. The mandibular arch presented a negative discrepancy and oval morphology, whereas the maxillary arch showed a positive discrepancy and triangular morphology, largely conditioned by loss of space of the congenitally missing maxillary lateral incisors and delayed eruption of the left maxillary canine. The patient had an atypical swallowing pattern that seemed closely related to the open bite of the lateral sectors.

Lateral teleradiography of the skull indicated a Class III skeletal pattern of maxillary origin associated with a dolichofacial pattern with multiple indicators of facial hyperdivergency (Fig 5).

The orthodontic treatment plan included reopening the spaces of the maxillary lateral incisors, correcting the posterior cross bite, repositioning the unerupted maxillary left canine, correcting the bilateral open bite,

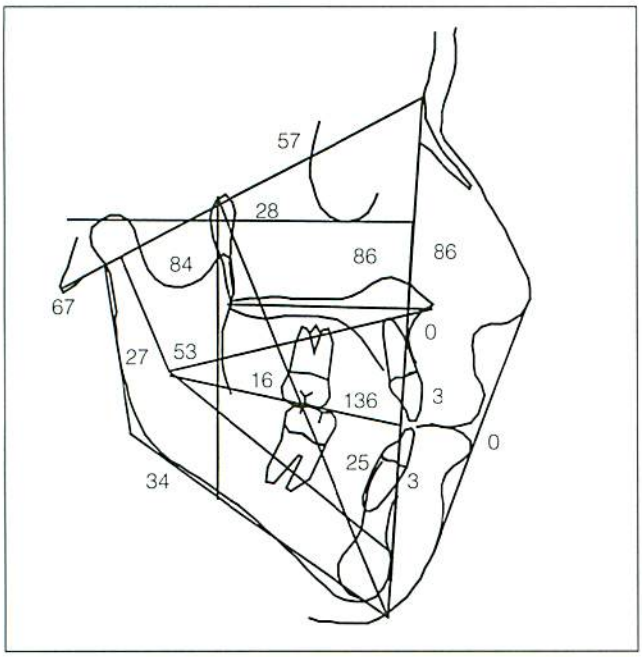
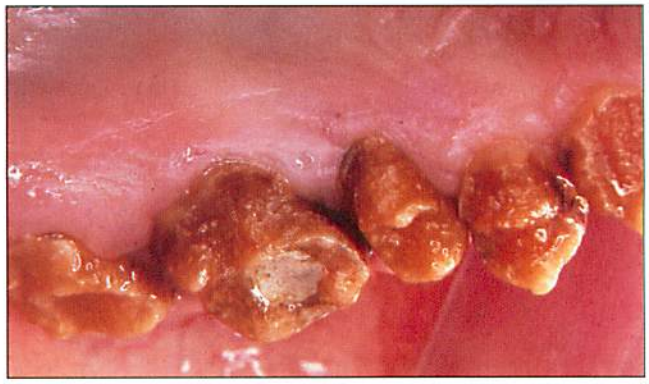


Fig 5 *(left)* Pretreatment cephalometric tracing.

Fig 6 *(below)* Maxillary teeth after endodontic treatment on the first molar.



and normalizing the vertical/sagittal incisal relationship. Finally, a moderate posterior open bite would be created as a preparation for a prosthetic restoration of the posterior sectors without consequent anterior opening of the bite. We used a preadjusted bracket appliance (0.018-inch slot), and the expansion was performed with a removable Quad-Helix. The brackets were bonded with autopolymerizable adhesive (Rely A Bond, Reliance Orthodontic Products), while Prime & Bond 2.0 (Dentsply/De Trey) was used as a photo-curable bonding agent for enamel and dentin. The bonding of the bands was with glass-ionomer cement (Ketac Cem, ESPE).

Prior to the orthodontic treatment, endodontia of the first 4 molars was carried out because of the elevated sensitivity and tendency to avoid molar contact (Fig 6). An oral surgeon performed the surgical exposure of the maxillary left canine to bond a bracket and create orthodontic traction. During the fenestration and after the incision in the mucosa, the surgeon noted a rough dental surface of earth-colored appearance that detached with only a touch with the scalpel. The collaboration of the periodontist was required during the treatment for gingival control.

After the orthodontic treatment (Figs 7 to 9), 2 thermopolymerizable acrylic retainers including the 2 missing maxillary lateral incisors were applied for 4 months, and the patient was again referred to the periodontist for evaluation. Coronal lengthening techniques were considered for improving the height of the clinical crown for the posterior sector, but were ruled out because the periodontist believed a simple gingivectomy was inadequate and that bone surgery techniques were required. The patient refused this option due to fear of surgery and psychologic exhaustion from the prolonged duration of the treatment.

Diagnostic casts were obtained, duplicated, and mounted on a semiadjustable articulator (ARH, Dentatus). The prosthetic treatment consisted of individual metal ceramic crowns in both arches excluding teeth 11 to 13 and 21 to 23 for the congenital absence of the maxillary lateral incisors and excluding the second molars because of the practical nonexistence of clinical crowns. The existing resin composite restorations were removed, and the anomalous layer of dental tissue was cut from the remaining surfaces. This superficial dental layer was softer to cut than normal enamel, although in gingival areas, it was possible to find tissue with the appearance and feel of normal enamel. The dentin presented a healthy appearance (Fig 10). The margins were situated juxtagingivally in the esthetically compromised areas and supragingivally in the areas where there was no compromise. The crowns were made from Bioethic (CM Dental) with a high gold content to minimize the gingival response, among other reasons. Following the normal clinical sequence, the marginal fitting (Fig 11), esthetic appearance, and occlusal fit were established. The restorations were cemented with a glass-ionomer cement (Ketac Cem) (Figs 12 to 14). At this time, 2 new thermo-modeled acrylic retainers were made by the orthodontist to complete the retention phase.

Figs 7 to 9 Views of patient after orthodontic treatment.

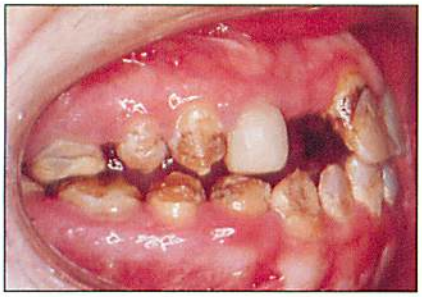


Fig 7 Right lateral view.

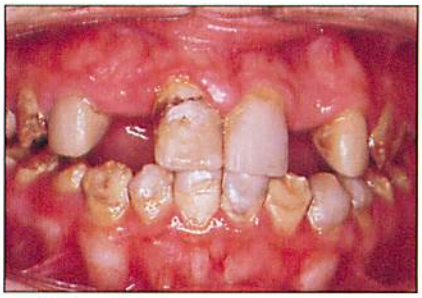


Fig 8 Frontal view.

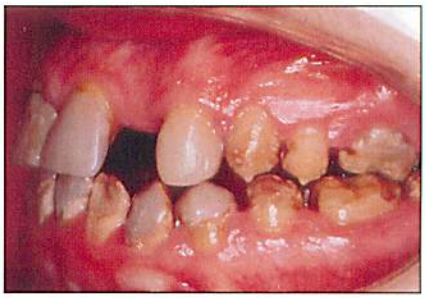


Fig 9 Left lateral view.

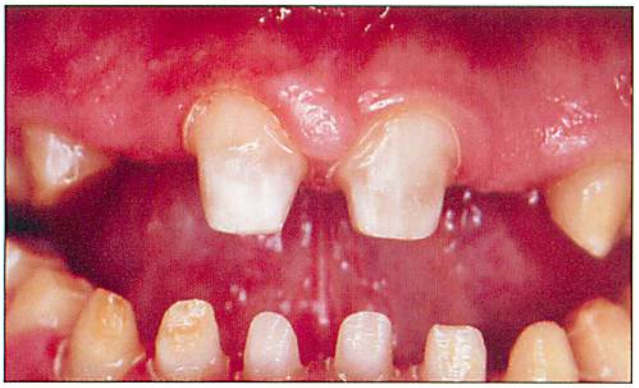


Fig 10 Normal dentin after tooth preparation.

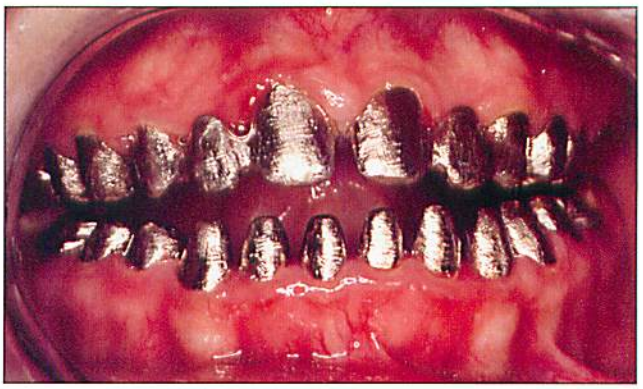


Fig 11 Marginal fitting.

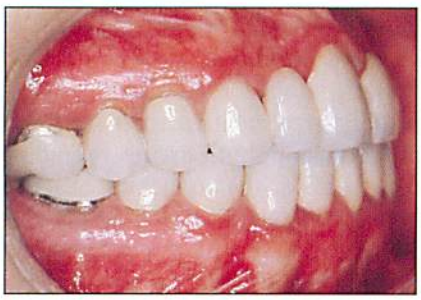


Fig 12 Right lateral view of completed restorations.

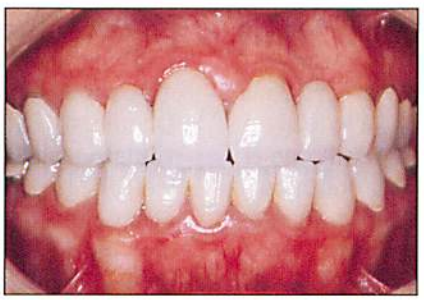


Fig 13 Frontal view after prosthetic treatment.

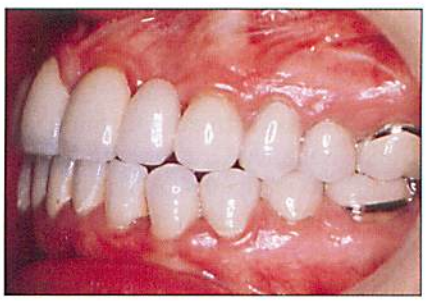


Fig 14 Left lateral view.

DISCUSSION

The Witkop and Sauk[6] system, similar to other classifications of amelogenesis imperfecta,[10] is based on the inheritance pattern of the disorder and on clinical manifestations. Our patient comes from a village, and her parents are first cousins. She is the youngest of 6 children. None of her siblings presented with dentition disorders although 2 of them are daltonic. A first cousin of her father lost her dentition at an early age due to the crumbling of her teeth, and she had a daughter with the same problem. These data suggest an autosomal recessive inheritance pattern of which expression was favored by the consanguinity between the parents[11] (Fig 15).

The clinical characteristics of the case made it difficult to include the patient in any of the subtypes described by Witkop and Sauk.[6] She had presented a major abnormality of dental morphology and size since dental eruption, due to a quantitative alteration of the enamel layer. This, together with the rough, yellow-brown dental surface, the absence of contact points, the unerupted teeth, and the open bite, suggested one of the subtypes of the hypoplastic forms with autosomal recessive inheritance. However, there was also a qualitative disorder of the enamel with a tendency to

crumbling at examination, also evident at the surgical exposure of submucosal tooth 23 and during the tooth preparation. The relative persistence of this layer in the lateral sectors may be partly accounted for by the open bite and malocclusion of the teeth. The external features of this enamel layer indicated a mineralization defect. Radiologically, this layer could hardly be distinguished from the dentin, which is a characteristic of hypocalcification. There are reports that cervical enamel is more mineralized and harder with hypomineralization, as in our case.[10]

For all of the stated reasons, we think that this case is one of the autosomal recessive forms of the hypoplastic type. The presence of hypocalcification in the enamel is characteristic of the IC subgroup in which the residual enamel is hypomineralized and soft at examination, as opposed to the normal hardness of the enamel in the other hypoplastic forms.[1] However, we stress that it is sometimes difficult to establish the diagnosis and classify a case as a particular subtype. There are reports of similar cases that were included in the IG subtype, ie, AI with enamel agenesis.[2] Peters et al[2] interpreted enamel agenesis as the absence of radiologic contrast between the dentin and an enamel layer (defined by them as agranular), although no data was given on the characteristics of this anomalous layer at the examination.

We first evaluated the patient when she was 15 years old. The morphology was very altered, but the extent to which this resulted from contact with the oral medium is not known. However, the surgical exposure of tooth 23 revealed the original fragility of this external layer, which suggested that the patient presented a combined disorder of hypoplastic type with hypocalcification. As mentioned above, there is scant information on the genotype or phenotype of this type of anomaly or on the expressivity of the defect. It is suspected that genes in various chromosomes are implicated.[3] It is most likely that the phenotypic expression is so varied that it is difficult to ascribe a case to one of the subtypes established according to a rigid clinical classification.

Unlike other anomalies, there is no correlation between the histologic findings and the phenotype of the case. Backman and Holm[12] reached this conclusion after analyzing 17 deciduous teeth of children with AI of different clinical subtypes. Polarized light microscopy, scanning elecron microscopy, and secondary ion mass spectrometry was used in the analysis. Regardless of the clinical appearance or inheritance pattern, the main findings in these 17 cases were the presence of hypomineralized enamel and what were termed "wavy bands." These observations support the idea that the diversity of clinical appearance is no more than a reflection of the variation in the expressivity of the anomaly.

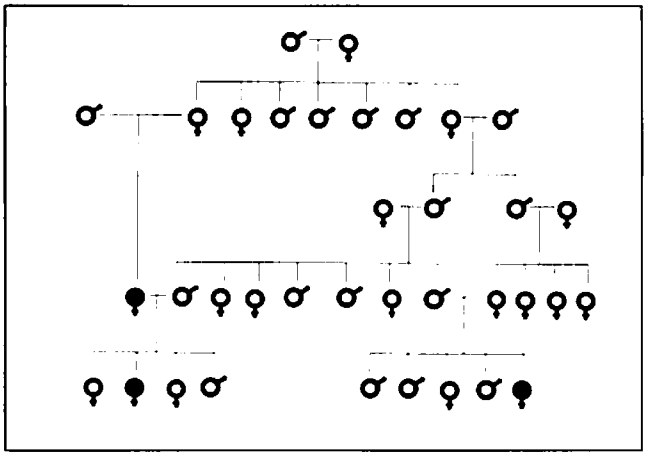


Fig 15 Pedigree of the patient with amelogenesis imperfecta.

The association of gingivitis with enamel defects may be accounted for by the persistence of bacterial plaque on the anomalous adamantine surface and by a deficient oral hygiene that is partly a consequence of the hypersensitivity presented by these patients.[13]

The first therapeutic consideration was whether the correction should be at the dental level alone or extended to the skeletal level. A combined orthodontic-surgical approach was ruled out from the beginning at the express wish of the patient, who was informed of both the skeletal/facial benefits and the special difficulties of orthognathic surgery in cases of AI.[14] This approach would have corrected the vertical and sagittal skeletal alteration, but an orthodontic camouflage approach was adopted instead.

At the start of orthodontic treatment, we had to choose between opening or closing the spaces of the congenitally missing maxillary lateral incisors. It was decided to open them, despite the greater complexity of the procedure, because closing them in the setting of a Class III malocclusion pattern would have required 2 dental extractions in the mandibular arch and a dentoalveolar distal retraction, which would have compromised the facial esthetics. There have been reports on the association between one hereditary entity and another hereditary entity, such as AI and dental agenesis.[2,15] Both are in turn associated with a delay in eruption of the permanent maxillary left canine that, as a result, requires fenestration and corresponding orthodontic traction. It is not known whether dental abnormalities associated with AI also result from the expression of a genetic amelogenesis abnormality or whether they are only associated effects. It has been proposed that eruption problems in autosomal recessive hypoplastic forms (as in this report) are related to abnormalities in the molecular control of the eruption process.[3]

The partial collapse of the posterior skeletal vertical dimension presented by our patient led us to create a moderate posterolateral open bite to allow for the insertion of crowns on the posterior teeth at a future date. This especially complicated the orthodontic treatment, since the edge-to-edge incisal relationship, in the setting of an anterior open bite skeletal pattern, had to be simultaneously normalized.

There are many reports in orthodontic literature on the association between AI and open bite, with occurrences ranging from 24%[6] to 60%.[12,16] Witkop and Sauk[6] regarded the open bite to be a result from defects in the eruption mechanism secondary to alterations in the organic matrix of the enamel. Thus, the hypersensitivity to heat and cold would be compensated by a lingual interposition to mechanically prevent the alveolar growth. Rowley et al[16] concluded that the association was due less to local factors and more to a genetic anomaly of craniofacial development, which would include skeletal open bite features.[17] Our case presented both lingual interposition and skeletal open bite, although the former appeared to be more a consequence of the vertical dysgnathia than a protective mechanism against dental hypersensitivity. Nevertheless, there are also published cases of AI with a reduction in the vertical dimension due to wear of the anomalous dental structure.[18,19]

The prosthetic treatment in this report yielded the question of whether or not to use full-coverage crowns. The external layer of the teeth was removed to reach layers of healthy tissue that would ensure a good adhesion. In the reports that used full-coverage crowns combined with inlays/onlays or partial crowns, patients were those with milder disease who had an enamel layer that provided better adhesion.[20]

The orthodontist and prosthodontist selected a bonding agent for both the enamel and the dentin because of the irregular dental surfaces. The severe porosity and deformity of the right maxillary canine caused several failures in the adhesion to the bracket, and a temporary resin crown finally had to be placed over it. However, despite the severity of our case, there were very few adhesion complications.

Various professionals must intervene to address these multiple and severe esthetic and functional problems.[20-22] A close working relationship between them is essential. It is also important for the patient to be well-informed at all times about the progress of the treatment.

At the end of the treatment, our patient experienced a considerable improvement in her facial appearance, because the opening of the spaces of the agenesic maxillary lateral incisors enhanced maxillary labial protrusion and reduced the esthetic impact of the Class III bone relationship. Despite the need to place crowns and fixed partial dentures in the posterior sectors, the vertical dimension did not cause significant facial variations.

At follow-up sessions, after treatment was completed, the patient reported her great satisfaction with the outcome, and her family described her character as having become more extroverted. This case illustrates that interdisciplinary approaches are required to take into account all of the factors involved and thus to achieve a satisfactory resolution.

ACKNOWLEDGMENTS

The authors are grateful to Dr Elena Sánchez and Dr Blas Noguerol for their contribution to the surgical and periodontal treatment of the patient, and to Richard Davies for his assistance with the English translation of the manuscript.

REFERENCES

1. Witkop CJ. Amelogenesis imperfecta, dentinogenesis imperfecta and dysplasia revisited: problems in classification. J Oral Pathol 1989;17:547-553.
2. Peters E, Cohen M, Altini M. Rough hypoplastic amelogenesis imperfecta with follicular hyperplasia. Oral Surg Oral Med Oral Pathol 1992;74:87-92.
3. Collins MA, Mauriello SM, Tyndall DA, Wright JT. Dental anomalies associated with amelogenesis imperfecta. A radiographic assessment. Oral Surg Oral Med Oral Pathol 1999;88:358-364.
4. Finn SB. Hereditary opalescent dentition. I. An analysis of the literature on hereditary anomalies of tooth colour. J Am Dent Assoc 1938;25:1240-1249.
5. Sundell S, Valentin J. Hereditary aspects and classification of hereditary amelogenesis imperfecta. Community Dent Oral Epidemiol 1986;14:211-219.
6. Witkop CI, Sauk JJ. Heritable defects of enamel. In: Stewart RE, Prescott GH (eds). Oral facial genetics. St. Louis: Mosby, 1976:156-193.
7. Aldred MJ, Crawford PJM. Amelogenesis imperfecta: Towards a new classification. Oral Dis 1995;1:2-5.
8. Lau EC, Mohandras TK, Shapiro LJ, Slavkin HC, Snead ML. Human and mouse amelogenin gene loci are on the sex chromosomes. Genomics 1989;27:121-130.
9. Takagi Y, Fujita H, Katano H, Shimokawa H, Kuroda T. Immunochemical and biochemical characteristics of enamel proteins in hypocalcified amelogenesis imperfecta. Oral Surg Oral Med Oral Pathol Oral Radiol Endod 1998;85: 424-430.
10. Schulze C. Anomalies in the development of teeth and jaws [in Spanish]. In: Gorlin RJ, Goldman HM (eds). Patología Oral. Barcelona, Salvat Editores S.A. 1983:132-158.
11. Nakata M, Wei SHY. Genetic basis of oral diseases. In: Wei SHY (ed). Pediatric Dentistry: Total patient care. Philadelphia: Lea & Febiger, 1988:352-384.
12. Backman B, Holm AK. Amelogenesis imperfecta: Prevalence and incidence in a northern Swedish country. Community Dent Oral Epidemiol 1986;14:43-47.

13. Bascones A, Alandez J, Yela M, Sicilia A, Sanz M. Amelogenesis imperfecta and gingival hyperplasia due to plaque [in Spanish]. Av Odontoestom 1987;3:105–117.
14. Hoppenreijs TJM, Voorsmit R, Freihofer HP. Openbite deformity in amelogénesis imperfecta. Part 1: An analysis of contributory factors and implications for treatment. J Craniomaxillofac Surg 1998;26:260–266.
15. Forner Navarro L, Almenar García A, Bonet Marco J, Llena-Puy MC. A study of the clinical, histopathologic and ultrastructural aspects of enamel agenesis: Report of case. ASDC J Dent Child 1999;May-Jun:208–212.
16. Rowley R, Hill FJ, Winter GB. An investigation of the association between anterior open-bite and amelogenesis imperfecta. Am J Orthod 1982;81:229–235.
17. Backmann B, Adolfsson U. Craniofacial structure related to inheritance pattern in amelogenesis imperfecta. Am J Orthod Dentofacial Orthop 1994;105:575–582.
18. García M, Martín MC, Llamas JM, Compagni R. Treatment of a case of multiple agenesis and amelogenesis imperfecta [in Spanish]. Arch Odontoestom 1992;8:418–423.
19. Rosenblum SH. Restorative and orthodontic treatment of an adolescent patient with Amelogenesis Imperfecta. Pediatr Dent 1999;21:289–292.
20. Nel JC, Pretorius JA, Weber A. Restoring function and esthetics in a patient with amelogenesis imperfecta. Int J Periodont Rest Dent 1997;17:479–483.
21. Greenfield R, Iacono V, Zove S, Baer P. Periodontal and prosthodontic treatment of amelogenesis imperfecta. J Prosthet Dent 1992;68:572–574.
22. Bouvier D, Duprez JP, Pirel C, Vincent B. Amelogenesis imperfecta-a prosthetic rehabilitation: A clinical report. J Prosthet Dent 1999;82:130–131.