CASE REPORT



Full-mouth rehabilitation choices depending on amelogenesis imperfecta's type: A familial case report

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Abstract

Amelogenesis imperfecta, a rare disease, represents inherited, congenital defects that primarily affect enamel with esthetic and functional impairment affecting everyday life. We present oral rehabilitation of a mother and her son, respectively, suffering from an hypoplastic and an hypocalcified form of AI.

K E Y W O R D S

amelogenesis imperfecta, full-mouth rehabilitation, prosthetics, rare disease

1 | INTRODUCTION

Amelogenesis imperfecta (AI) is a rare disease with a prevalence of 1 in 14,000 people.¹ AI represents a group of genetic defects in enamel's development. It affects structure of enamel with visible repercussions on all—or almost all—the teeth (deciduous and/or permanent teeth).² AI may be isolated or syndromic associated with morphologic or biochemical changes elsewhere in the body like nephrocalcinosis in enamel renal syndrome or gingival hyperplasia in gingival fibromatosis syndrome.³

Management of rare oral diseases, in France, but as in many other countries, has clearly improved over the last 15 years. It seems to us that before, and even now for many practitioners, these "dilapidated mouths" could frighten them because of the extent of the treatments to be done. Diagnostic errancy (rare diseases not well known by many health professionals) and therapeutic errancy (what to do in such situations) have left many patients without adequate oral rehabilitation. It is clear today in the literature that the earlier the treatment of amelogenesis imperfecta on temporary teeth (composites, preformed caps), the more the esthetic and functional prejudice will be reduced, and thus, children and adolescents will have an improved quality of life related to their oral health.

Patients with AI have a quality of life impacted by esthetic impairments (hypoplastic enamel with pitted and ridged appearance, hypocalcified with discolored enamel or hypomatured with whitish surface) and functional disorders (decreased vertical dimension, fragile enamel, dentin hypersensitivity, and open bite). These patients often hide their teeth, they do not smile and can be reclusive and withdrawn. Many cases have been observed of patients requesting the extraction of all their teeth and the fitting of dentures.¹

In this article, we report an original case of two patients, a mother (48 years old) and her son (24 years old), both affected by AI. They never received any prosthetic

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rehabilitation. Both patients suffered from a decrease of occlusal vertical dimension (OVD). Deviating from the three-step technique,⁴⁻⁶ developed by Dr Francesca Vailati for patients affected by severe dental erosion, we adapted it to our patients with chronic facial dysmorphism. The objective is to restore the loss of dental tissue in the most conservative way possible using indirect bonded restorations.

The complexity of these clinical situations is due to the different stages of tissue loss and the establishment of a correct vertical dimension of occlusion. Reproducing the same steps for both patients during the full-mouth rehabilitation facilitate communication between the patient, the practitioner, and the laboratory. In the complex and lengthy care of these patients, we have had to judge, as far as possible, their motivation. It is not possible, given the scope of the treatment to come in these pathologies, because here all the teeth are affected, to start a treatment without being able to finish it. The therapeutic approach is very patient-dependent and each patient, depending on age, previous care course, and motivation, should receive a perfectly adapted treatment.

2 | CLINICAL REPORT

The first part of the full-mouth rehabilitation is an esthetic evaluation (evaluating incisal edges, teeth axes, teeth shapes and sizes, occlusal plane, ideal OVD) to establish an ideal occlusal plane. To do so, full-mouth wax-ups are made by the laboratory technician and mock-ups—reproducing the wax-ups—are transferred in mouth by the dental surgeon. In the second part, the patient's posterior quadrants are restored at an increased vertical dimension (established by the dental surgeon using esthetic settings within the physiological limits of phonation and swallowing). Finally, the third part reestablishes the anterior guidance.

2.1 | The son

2.1.1 | Patient

The son is a 24-year-old man in good health affected by an isolated form of amelogenesis imperfecta. He was diagnosed with AI at the age of 11 by his orthodontist based on clinical points and without any molecular diagnosis made. He was treated by his orthodontist for a skeletal class II occlusion with removable appliance. His enamel damages never worried him until he had the feeling that it was crumbling more and more with sensitivities. Indeed, the patient first came to the University Hospital because of severe pain with the slightest thermic stimuli.

2.1.2 | First consult

The son suffered from both qualitative and quantitative impairments of enamel with an hypocalcified form of AI. He had a sagging of the labial tissues with pursed lips because of a loss of OVD, his smile was unsightly (Figure 1A,B). He had occlusal surfaces and incisal edges' wear on all teeth associated with brown stains, the enamel was soft and cleaved quickly, dentin was exposed in many spots (Figure 1C-E). We noted good oral hygiene with no plaque, no decay, and a previous restorative treatment on the upper left first molar #26 (amalgam). In intercuspal position, the patient presented an increased overjet and overbite with interposition of the lower lip-the imprint of the maxillary incisors was visible on his lower lip (Figure 1F-H). Observing the patient's opening pattern, we noted a jaw deviation to the right with repositioning along with a clicking joint noise on the left that might indicates a disc displacement with reduction on the left. An orthopantomogram was made showing that the enamel was less opaque than dentin (Figure 2).

We established an esthetic plan in order to have esthetic landmarks essentials for increasing the OVD: arbitrary occlusal plane from the upper edge of the tragus to an anterior point about 10 mm below the infraorbital point, located 8° below the Frankfurt plane. We took as reference the horizontal inter-pupillary line and the vertical reference line was the median to the inter-pupillary line, we observed that the interincisal midpoint was aligned with the vertical line. It was determined when the patient, in an upright, orthostatic position, looked at the horizon. For the patient to test the new OVD and also to reduce his left temporomandibular joint's disc dislocation, we decided to make a thermoformed plane splint. It was therefore made to obtain the following parameters: bilateral even contact with the opposing teeth on closure and in a comfortable mandibular posture (anterior position and mandibular asymmetry), opening pattern normalized without deviation and no more clicking noise on the left, presence of an occlusion space in a posture resting position, absence of contact between the dental arches during phonetic activity, pleasant appearance of the lower level of the face in intercuspal position with closed lips but not pursed.

Then, for 2 months, we saw the patient at regular intervals to modify the splint in order to slightly realign the mandibular in a posterior centric position. After 2 months, the patient adapted well to the new OVD and the disc dislocation was reduced. We estimated an increase of OVD of 10 mm in anterior region. After maxillary and mandibular alginate impression, we transferred the maxillary bone position with the splint onto the articulator using a face bow then recorded the patient's mandibular position with the splint using occlusion silicone (Futar^{*} - Kettenbach).



FIGURE 1 First consult. (A) Son's extra-oral view with pursed lips and (B) unsightly smile. (C) Son's intra-oral view with front view showing incisal edges' wear, (D) maxillary view, and (E) mandibular view with occlusal surfaces wear, exposed dentin and brown stains. (F) Smile view with interposition of the lower lip and imprint of the maxillary incisors visible on the lower lip, occlusion view with increased overbite (G), and overjet (H)



FIGURE 2 Son's orthopantomogram. First consult. Enamel is less opaque than dentin. Left condyle is in anteriorly position

2.1.3 | Full-mouth rehabilitation technique

First part of the treatment was common for both patients and consisted in assessing the new OVD to estimate the occlusal plane, evaluate the maximal intercuspal position and validate the esthetics. Without any teeth preparation, we proceeded full-mouth mock-ups restorations corresponding to the wax-ups made for the son with splints (Figure 3A,B). We used injectable resin composite technique using unheated flow composite (Tetric EvoFlow^{*} - Ivoclar Vivadent) to add material to unprepared teeth to achieve the new OVD. Several splints were made by the prosthetist to have dental contacts to perform mock-ups at the chosen OVD. The patient was seen after 2 weeks to assess the comfort with the new OVD and the wear facets of the mock-ups corresponding to the areas of greatest constraints during the function.

For this patient, full-mouth single crowns cemented on living tooth seemed to be the best long-term solution.

Second part consisted in the rehabilitation of the posterior teeth in the defined occlusal scheme at the increased OVD. Due to the son's enamel fragility and dentinal sensitivities, we performed intra-sulcular tooth preparations, under local anesthesia (Figure 4). We removed all soft enamel and tooth preparations were made with 2 mm thickness. Resin shells had been prepared beforehand by the dental technician, and we rebased them in the mouth with chemo-polymerizable resin (Unifast^{*}). An occlusal posterior equilibration was performed guided by the incisal guidance that

3 of 9

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FIGURE 3 Intra-oral view of the mock-ups (A) with difference between tooth with and without mock-ups (B)



FIGURE 4 Intra-oral view of the crown's preparations. Son's intra-sulcular peripheral preparations

remained with the mock-ups. Despite local anesthesia, the session was difficult for the patient who had to remain open mouth for a long time and felt pulp sensitivities. To avoid pain between two oral care sessions, temporary crowns were bonded with temporary cement without eugenol (Temp Bond NE[®]) and we prescribed an analgesic (Paracetamol). After gingival healing of 2 weeks, the patient came back with intense pain. At first, we thought the pain was linked to dentin hypersensitivity, but after thorough examination, the clicking noise on the left TMJ had reappeared and the pain was reproduced on masticatory muscles' palpation. The duration between sessions was increased to let the patient's muscles rest, we prescribed physiotherapy with muscle relaxant (Miorel[®] 8 mg per day during 5 days) and anti-inflammatory medicines (Ketoprofen 200 mg per day during 5 days). During the break, another issue appeared: the temporary crowns showed percolations with the natural wear of the resin. Furthermore, dentinal pain reappeared requiring rehabilitation sessions of the temporary restorations.

After 7 months of physiotherapy, the patient was freed from his myofascial pain and disc dislocation was reduced. We were able to take the impression of the posterior quadrants with silicone Aquasil[®] Dentsply (1-step 2-viscosity technique). The crowns were sent back to the laboratory technician to be glazed. A lengthy appointment was set to cement the posterior zirconia crowns and make occlusion adjustment. We used GC Fuji One^{*} capsules for crowns seating. Occlusion setting was made in maximal intercuspal position with intra- and interarch stability.

After the completion of a stable posterior occlusal support, the third part consisted of the rehabilitation of the anterior quadrants, with esthetics and functional needs, to establish a proper anterior guidance. Preparations were made at subgingival limits for better esthetic integration and above all to avoid any dentinal exposure that could induce pain. Then, following the same steps as the posterior quadrants, a temporary stage with resin crowns and impression session, we tried on the biscuit bake zirconia try-in crowns. The maxillary incisor block was composed of long teeth to obtain better anterior guidance. We set a large session in order to cement the six maxillary and six mandibular anterior crowns still using GC Fuji One capsules (Figure 5A-E). Occlusion adjustment was performed statically and dynamically (Figure 5F-H). The incisal guidance allowed posterior disocclusion in translational movement and in lateral excursion the patient had a functional canine guidance.

A follow-up appointment was given 2 weeks after the crowns seating, and a mandibular rigid bruxism splint was given to the patient. From the first consult to the 2 weeks' follow-up appointment, the treatment lasted 18 months. At 6 months follow-up, the son was pain-free and satisfied, he did not notice any temporomandibular joint (TMJ) pain.

2.2 | The mother

2.2.1 | Patient

The mother is a 48-year-old woman in good health with IA isolated form of amelogenesis imperfecta. She was diagnosed tardily when her son was diagnosed without any molecular diagnosis made either. Her request was mainly cosmetic due to the staining and the porous appearance of her teeth. She had an orthodontic treatment with bonding orthodontic bands in 2016 to compensate for an Angle class II division two malocclusion but after the brackets' removal, teeth appearance was worse than before.

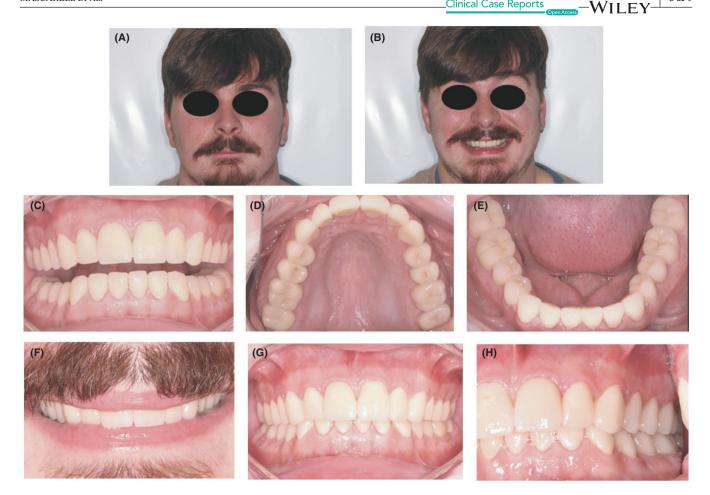


FIGURE 5 Son's extra-oral view with full-mouth rehabilitation. Day of crowns' seating. (A) Reduced pursed lips and (B) harmonious smile. Son's intra-oral view with front view (C), maxillary view (D), and mandibular view (E) with single zirconia crowns. Son's smile (F) and occlusion view with correct overbite (G) and lateral occlusion view (H)



FIGURE 6 Mother's intra-oral view. First consult. (A)Increased overbite and prominence of the maxillary bone, (B) wear of the occlusal and vestibular surfaces, irregular incisal edges, endo-alveoli of the maxillary posterior quadrants, and diastemas from 13 to 23 and (C) 43 to 33

2.2.2 First consult

The mother had a less severe form of AI with an hypoplastic enamel: wear of the occlusal and vestibular surfaces of the posterior maxillary quadrants, irregular incisal edges, quantitative defect with low thickness or even absence of enamel, presence of wells and streaks with areas of hard and translucent normal enamel. She had a decreased OVD that might be explained by night bruxism and daytime tightening. She had sagging labial tissue, diastemas

from 13 to 23 and 43 to 33 and endo-alveoli of the maxillary posterior quadrants and previous restorative treatments with amalgam on maxillary molars (Figure 6A-C). She had a gingival smile and prominent alveolar bone with impression of an extrusion of the maxillary alveolar bone. In intercuspal position, she showed an increased overlap leading to a locked occlusion. On radiographs, enamel contrasted normally from dentin.

Following the same method used with her son, after an esthetic evaluation, we established a new ideal occlusion WILEY_Clinical Case Reports

vertical dimension assessed with a splint set in centric relation. The mother adapted without problem to the new OVD. We established and increase of 6 mm in anterior quadrants (mandibular interincisal point). The laboratory technician was able to perform maxillary and mandibular anterior quadrants wax-ups and maxillary posterior wax-ups with the obtained space. To get an ideal overjet and compensate for the class II malocclusion, we asked the dental technician to realize anterior maxillary teeth in slight linguoversion.

2.2.3 | Full-mouth rehabilitation technique

We followed the same first step as for her son to assess the new OVD, estimate the occlusal plane and validate the esthetics. We proceeded maxillary and anterior mandibular quadrants pre-prep diagnostics mock-ups (Figure 7A,B). We also used injectable resin composite technique (Tetric EvoFlow - Ivoclar Vivadent). Several maxillary splints were made by the prosthetist to have dental contacts to perform mock-ups at the chosen OVD: first splint rehabilitating from the first premolar #4 to the first molar #6 -with stable supports on the second molar #7 and anterior block #3, #2, #1 without wax-ups; second splint rehabilitating the anterior block from #3 to contralateral #3 and #7 -with stable supports on #4, #5, #6 with mock-ups and mucous support. For the mandibular arch, only one splint was made rehabilitating from #33 to #43 with stable supports on posterior quadrants. A precise adjustment of the occlusion in maximal intercuspal position was carried out.

Single vitroceramic crowns bonded on the maxillary teeth and from canine to canine in the mandibular arch and direct composite restorations on the premolars and molars on the mandibular arch were the treatment chosen.

Second part consisted of the rehabilitation of the posterior teeth in the defined occlusal scheme at the increased OVD. In this case, we performed minimal invasive peripheral preparations - vertical peripheral preparations with less than 1mm thickness and occlusal preparations of 1.5 mm thickness (Figure 8). The patient's endo-alveoli of the maxillary posterior quadrants allowed us to perform very thin preparations in vestibular to have crowns with slight over-contour. The temporary stage was made with resin elements at the defined OVD. After an impression session, we tried on the biscuit bake Emax[®] (Ivoclar Vivadent) try-in crowns. A long session was planned for the bonding protocol using unheated bonding composite (Variolink Esthetic Ivoclar Vivadent) indicated for permanent cementation of glass-ceramic. Then, we lightcured 20 s per quarter surface of crown. After bonding of the four posterior quadrants, we made direct composite restoration on mandibular premolars and molars with less loss of tissue. Then, we made the occlusion adjustments in maximal intercuspal position in order to have a stable distribution occlusal contacts throughout the posterior quadrants: vestibular cusps of the premolars and mesiovestibular mandibular molars maintained contacts with mesial marginal ridges of the opposing maxillary teeth, vestibular cusps of the mandibular molars had their contacts in the opposing pits on the ridges of the maxillary molars, lingual cusps of the maxillary premolars were positioned in the triangular fossa of the mandibular premolars and mesiolingual cusps of the maxillary molars were positioned in the central fossa of the mandibular molars, the contacts on the vestibular cusp angle of the maxillary molars were accentuated due to linguoversion of mandibular teeth.

With the mother, the third part was important due to her esthetic requirements. Same steps were respected with minimal invasive peripheral preparations. The patient was eager to have small square teeth. After trying the biscuit bake try-in crowns, these last were glazed by the dental technician. The maxillary and mandibular incisor-canine block Emax^{*} (Ivoclar Vivadent) crowns were bonded (Variolink^{*} esthetic protocol). Occlusion adjustments were made to obtain an anterior guidance and canine function in lateral movements. The overbite was still accentuated explained by the extrusion of the maxillary bone, leading to an accentuated anterior guidance.

A follow-up appointment was given 2 weeks after a mandibular rigid bruxism splint was given. For the mother the whole treatment lasted 6 months with a

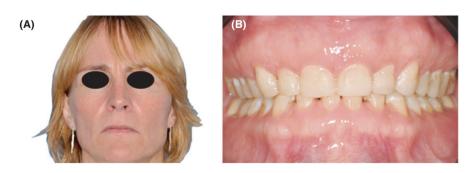


FIGURE 7 (A) Mother's extra-oral view with mock-ups (B) Intra-oral view of the mock-ups

Clinical Case Reports

painless adaptation to the increase of OVD, the patient even reported that since the prosthetic's rehabilitation she no longer grinded her teeth at night—as evidenced by her intact splint after 6 months of wearing.

At 1-year follow-up, the mother had no complain, she was completely satisfied with the rehabilitation and we noted the relaxation of her facial muscles, sign of soft tissues' adaptation to the new OVD; Figure 9A–E).

3 | DISCUSSION

Both patients noted an improvement in their everyday life with better self-esteem and more functional teeth.



FIGURE 8 Intra-oral view of the crown's preparations. Minimal invasive peripheral preparations

In order to solve the esthetic and functional issues (in particular increase of OVD), we worked in three steps. For the first step, we used an esthetic plan to assess the increase of OVD which can be opposed to the determination of the OVD by cephalometric measurements.^{7,8} Orthlieb's way of determining an increase of OVD is based on the skeletal facial typology, cephalometric references of the ratio between the maxilla and the mandible and maintenance of anterior functional contacts. For him, the increase in OVD should be at least 5 mm at the incisal level to have an esthetic impact, so esthetics' factor is not a major indicator for increasing the OVD. On the other hand, Fabbri's way of determining an increase of OVD is based especially on esthetic settings. For him, the OVD is determined regarding the horizontal plan (inter-pupillary line) and the lower lip to have the best harmony of the face, while maintaining a free space of occlusion at rest and when speaking. Due to the significant esthetic impairments of AI in these patients, we chose to use Fabbri's technique mainly based on esthetics to determine the increase in OVD. Whatever the technique used to determine an increase in OVD, only the clinical result after the test period (mock-ups, wearing a splint) can be used to validate or not the relevance of the established OVD.

An interesting point to note concerning the reconstruction's position of these two patients is that for the son we chose a more anterior mandibular position to reduce the disc dislocation, while for the mother the chosen reconstruction position was the centric relation. This difference is explained by the adaptation of the reconstruction position

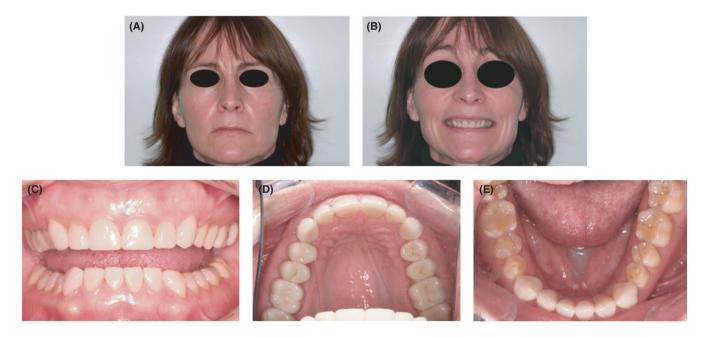


FIGURE 9 (A, B) Mother's extra-oral view with full-mouth rehabilitation. 12 months follow-up. (A) Resting position, relaxation of facial muscles compared to mock-ups (B) Smiling. (C–E) Mother's intra-oral view with front view (C), maxillary view (D), and mandibular view (E) with Emax^{*} crowns

to the needs of each patient; indeed, the reconstruction position is not an absolute value and can be adapted to the patient while respecting esthetic and functional standards.⁹

During second and third steps, one of the main stakes of this treatment was the management of the temporary stage. Indeed, the son felt inter-session pain. Despite the completion of an efficient splint therapy at the beginning of the treatment, the patient again had a disc displacement explained by the fact of having remained for a long time with his mouth open. Added to this, intraoperative pain (and personal family stress that day) led to the onset of facial myalgia. The patient had temporomandibular joint pain with myofascial pain and a disc displacement with reduction on the left. He was in emotional distress coming to the sessions for fear of aggravating his pain. The intraoperative pain as well as the establishment of an incisal guidance probably led to the contraction of the masticatory muscles and the introduction of a more locked occlusion than before that resulted in neuromuscular stress.⁹ Following the guidelines for TMJ disorders, the decision was made to pause the temporary stage with provisional crowns so that the patient could rest, prescribe the indicated medicines and do physiotherapy—at the new OVD established.¹⁰ During the break, we often saw the patient in order to maintain occlusal stability, which is an important tolerance criterion in the case of a sudden significant increase in OVD.¹¹

The different forms of AI type in the two patients led to a choice of different material type for rehabilitation, the choice of cementing or bonding crowns was also based on the type of AI. Indeed, hypocalcified (or hypomineralized) enamel initially develops normal thickness, consistency rough and soft, discolored enamel is orange-yellow at eruption and consists of poorly calcified matrix which is rapidly lost leaving dentin cores, on radiograph enamel is less opaque than dentin, teeth are sensitive to temperature changes, the crowns have a moth-eaten appearance and teeth often accumulate heavy deposits of calculus.² For the son, his dentin was exposed in many spots and no reactive dentin had formed yet leading to dentin hypersensitivity. Hypoplastic enamel does not develop normal thickness, it has pitted and ridged appearance.² Several studies have shown that composite restoration and adhesive ceramic restoration may be a reliable approach even in the presence of a compromised enamel layer.^{12,13} For the mother, enamel was well mineralized, but thickness was reduced. Plus, the mother already had an orthodontic treatment with bonding orthodontic bands, and she reported no detachment during treatment. Taking the information into account, bonding restorations appeared to be adequate for the mother. A recent study, exploring adhesive cementing on dentine and affected enamel, showed that preservation of residual enamel to enhance bonding, sandblasting the tooth surfaces to increase roughness, choosing a universal adhesive, and

reinforcing enamel and dentine by means of infiltrant resins could be clinical principles used for a perennial bonding.¹⁴

The invasiveness of the treatment was a key line of thought in our therapeutic decision. Indirect restorations are preferred over direct restorations for patients with AI.¹⁵ Following the therapeutic gradient, other possible treatments for the mother were as follows: abstention but does not respond to the patient's request; vestibular facets in the anterior quadrant but does not restore good function (accentuation of anterior guidance or canine function) and does not restore soft tissue esthetics (loss of OVD); single ceramic crowns on the anterior maxillo-mandibular quadrants and overlays on the posterior maxilla quadrants but does not restore a good surface condition of the posterior maxillary buccal surfaces; single ceramic crowns on the anterior maxillo-mandibular quadrants and posterior maxillary crowns. Single vitroceramic crowns bonded on the maxillary teeth and from canine to canine in the mandibular arch was selected because it was a long-lasting solution to restore function (increase in OVD and canine function), esthetics, and the surface condition of the teeth (no porosity, easier oral hygiene). For the son, the choice was different because he had an hypocalcified enamel-not mineralized enough-with exposed dentin that cannot be trusted for bonding.^{2,13} The perennial choice of cementing considering his AI's form was crowns seating. The use of a glass-ionomer cement was indicated because of its biocompatibility with pulp and its long-term fluoride release.¹⁶

The therapeutic choices we made allowed to restore function, increase the OVD, improve esthetics, induce a positive psychological impact for the patients with the disappearance of the pain and a better self-esteem. Recently, a retrospective clinical study observing the 6-year survival rates of three-step technique concluded that this technique is a reliable treatment option in the medium term and that protective nightguards are recommended.¹⁷ Thus, follow-ups are essential for the long-term maintenance of the rehabilitation carried out.

4 | CONCLUSION

Amelogenesis imperfecta is a rare disease with esthetics, functional, and psychosocial impairments.¹⁸ As usual in rare diseases, care course may be difficult for the patient with diagnostic and therapeutic wandering. The later the diagnosis and care, the more additional disorders have established—such as decrease in OVD—complexifying the treatment plan. Thus, these patients require our full attention in order to achieve the more precocious care to restore esthetic and function. In this type of rehabilitation, the key points of a good treatment will be importance of early diagnosis with evaluation from all domains—that is, function, activity, participation, personal, and environmental. These points are essential before starting this treatment which must have a step-by-step approach. Depending on the severity of the damage, we will be able to establish whether the rehabilitation will be bonded or cemented. These treatments were performed in a university hospital where the fees are low. In other circumstances, in private practice for example, these two treatments are quite expensive and not all patients can access them. In these circumstances, when patients cannot afford the cost of prosthetic reconstructions, a direct technique approach can be considered, even if these composite techniques are less durable over time. Furthermore, psychological support is essential with these patients with chronic facial dysmorphism and low self-esteem.^{1,18}

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CONFLICT OF INTEREST

The authors declare that they have no known competing financial interests or personal relationships that could have appeared to influence the work reported in this paper.

AUTHOR CONTRIBUTIONS

SM involved in investigation, writing—original draft, writing—review & editing, and visualization. HC and VM involved in writing—review & editing and supervision. LF involved in conceptualization, investigation, writing—review & editing, and supervision.

CONSENT

Written informed consent was obtained from the patients to publish this report in accordance with the journal's patient consent policy.

DATA AVAILABILITY STATEMENT

The data that support the findings of this study are available from the corresponding author upon reasonable request.

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