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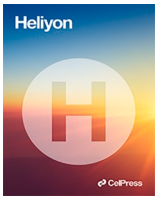

Prosthodontic rehabilitation of two siblings with hypoplastic (type 1) amelogenesis imperfecta: A case report

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ABSTRACT

Amelogenesis imperfecta is a rare genetic disorder that interferes with normal enamel formation. Of the 4 main types of amelogenesis imperfecta, hypoplastic (type 1) is the most prevalent, characterized by a quantitative alteration in enamel. The pitting or reduced thickness of the enamel results in generalized hypersensitivity, increased susceptibility to caries and infection, attrition, and a loss in vertical dimension of occlusion. Prosthodontic management of these patients can be challenging not only functionally and restoratively, but also from an emotional and psychosocial standpoint. This clinical report describes the prosthodontic management and rehabilitation of two young adult siblings with hypoplastic (type 1) amelogenesis imperfecta.

1. Introduction

Amelogenesis imperfecta (AI) is a rare hereditary disorder that interferes with normal enamel formation of both the primary and permanent dentition [1]. Although AI can be inherited as an X-linked, autosomal dominant or autosomal recessive trait, it can also occur through sporadic inheritance with no familial history [1,2]. AI has been classified into 4 main types based on phenotype, which are hypoplastic (type 1), hypomaturation (type 2), hypocalcified (type 3), and hypomaturation-hypoplastic with taurodontism [1–3]. These 4 main categories can then be even further subdivided 15 different subtypes [3]. Patients with AI often experience generalized hypersensitivity, compromised esthetics, loss in vertical dimension of occlusion, malocclusion, increased caries and infection risk, difficulty maintaining oral hygiene, and negative psychosocial impacts such as social avoidance, poor self-image, and anxiety [2,4,5].

Among the different types of AI, hypoplastic (type 1) is the most commonly occurring type [6]. Hypoplastic (type 1) AI results in a quantitative (sometimes qualitative) alteration of the enamel that can be localized or generalized pitting, localized or generalized decrease of enamel thickness, rough or smooth enamel, grooving, and in extreme cases enamel agenesis [3.7]. Because of the quantitative loss of enamel seen with hypoplastic AI, diastemas, attrition, and smaller clinical crown sizes are often observed [7]. Proper diagnosis of the specific type of AI should also be considered when determining overall treatment for patients with AI. Hypoplastic enamel, although quantitatively altered, is still of sufficient quality. Other types of AI, such as hypocalcified (type 3) AI, have a significant alteration in the quality of the enamel [1], therefore it may need a more aggressive removal of defective enamel during treatment.

Prosthodontic intervention of patients with AI is often required at an early age and should begin as early as possible [8] in order to avoid further deterioration of the dentition and more invasive procedures such as extractions and implants [9], and improve their psychosocial well-being [10]. However, orthodontic intervention [11,12] in preparation for prosthetic reconstruction should be highly

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considered in order to optimize arch form, interdental spacing, and subsequent prosthetic outcomes [13,14]. In patients with AI, a perfect occlusion may not always be achieved, but rather the focus should be on the final prosthodontic rehabilitation and how orthodontic movement can better position the teeth to allow for a successful reconstruction [13]. Clear aligner therapy for orthodontics in patients with AI [15] can also be beneficial as the aligners form an intimate fit over the teeth, sealing off areas of exposed dentin or pitting which may be causing hypersensitivity.

This report describes the fixed prosthodontic rehabilitation of 2 siblings with hypoplastic (type 1) AI. Treatment objectives were prosthodontic intervention to eliminate pain/sensitivity, restore stable function, and improve esthetics.

2. Case report

A 22-year old female (sibling A) first presented to the private-practice prosthodontic clinic (Fig. 1A). The patient's younger brother, a 20-year old male (sibling B), then also presented to the clinic after her (Fig. 1B). Siblings A's main concerns were pain and sensitivity, dissatisfaction with the appearance of her teeth (Fig. 1C), and the long-term health of her teeth. Informed consent was given from the patient, and a patient questionnaire revealed functional, physical, psychological, and social impairment from her teeth such as difficult chewing, pain and sensitivity, avoidance of smiling, embarrassment, and self-consciousness. A familial history revealed that no other family members to their knowledge had teeth like the siblings. Siblings B's concerns were similar (Fig. 1D), but in addition he was dissatisfied with the inflammation in his gums. Informed consent was given from the patient, and his patient questionnaire also revealed significant functional, physical, psychological, and social impairment due to the condition of his teeth. No contraindications to prosthodontic rehabilitation in either medical history were found.

2.1. Dental history and clinical examination

A comprehensive dental history revealed that both siblings had previously completed orthodontic treatment and crown lengthening of the maxillary anterior teeth (Fig. 2A–D). For both siblings, the little enamel that remained exhibited pitting, but was hard and did not easily flake off with hand instrumentation (Fig. 3A and B). Clinical and radiographic examination revealed a generalized decrease in enamel thickness and subsequent insufficient quantity of enamel (Fig. 4A–F). Both patients were diagnosed with hypoplastic (type 1) AI.

For sibling A, facial composite resin restorations were present on the maxillary premolars and incisors, and on the mandibular canines and first premolars. Small carious lesions were noted on the occlusal and interproximal of some of the molars. Attrition, collapsed occlusal vertical dimension, Angle Class II dental relationship, short clinical crowns in the posterior, and crossbite at the left second molars was noted. Due to a generalized decrease in enamel thickness, the teeth had an overall smaller crown size and shortened clinical crown height, especially in the posterior. Despite the smaller crown size, orthodontic therapy closed any spacing between the teeth and brought all of the teeth together.

For sibling B, full coverage restorations were present on the maxillary premolars and incisors, and facial composite resin restorations were present on the mandibular canines. Small carious lesions were noted on the occlusal and interproximal of some of the molars and gingival inflammation was noted along the existing full coverage restorations in the maxilla. Attrition, collapsed occlusal vertical dimension, Angle class II dental relationship, and 100 % overbite was also noted. Smaller crown sizes and shortened clinical

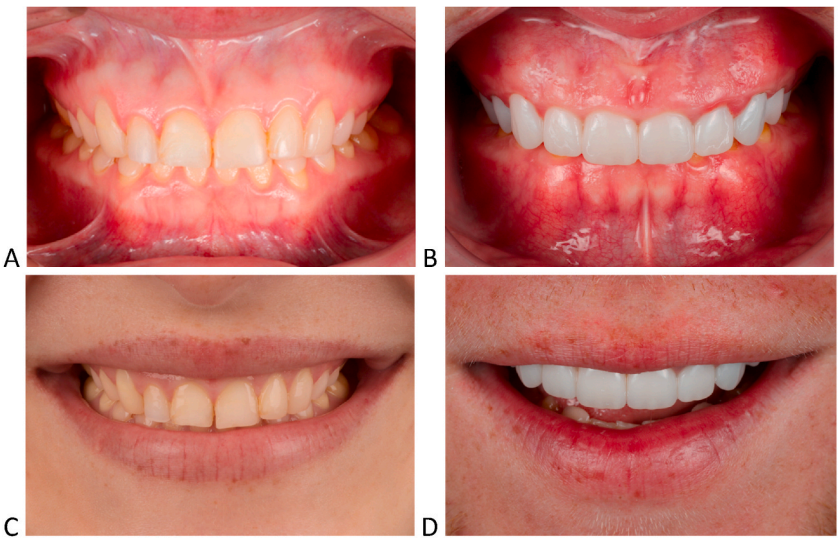

Fig. 1. Preoperative presentation. A, Intraoral presentation, sibling A. B, Intraoral presentation, sibling B. C, Frontal smile, sibling A. D, Frontal smile, sibling B.

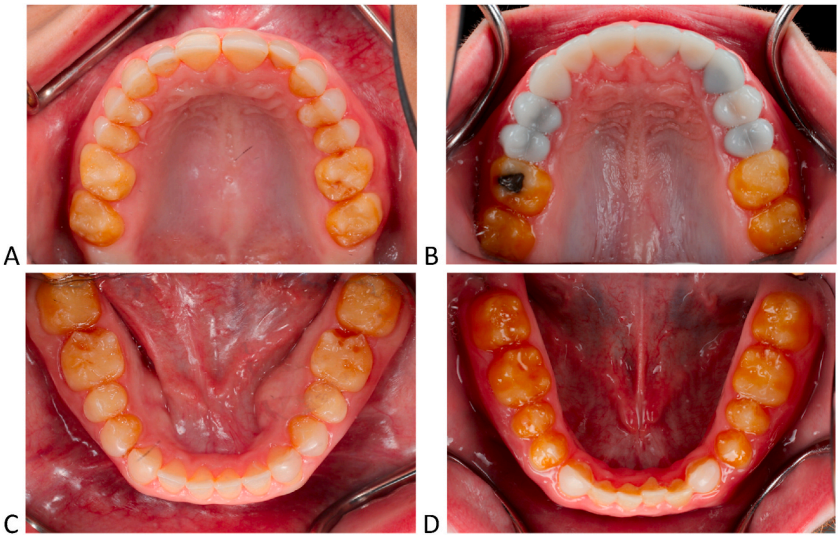

Fig. 2. A, Maxillary occlusal view, sibling A. B, Maxillary occlusal view, sibling B. C, Mandibular occlusal view, sibling A. D, Mandibular occlusal view, sibling B.

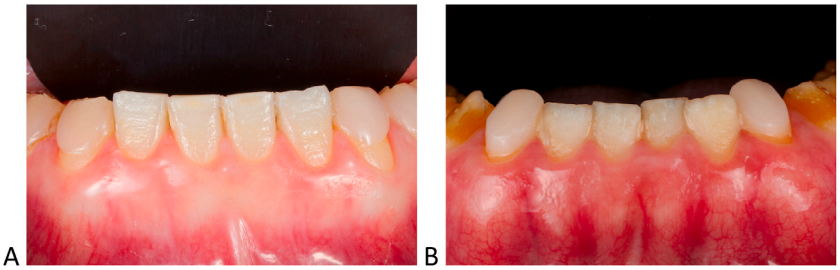

Fig. 3. Defective hypoplastic enamel pathognomonic of type 1 amelogenesis imperfecta. A, Sibling A. B, Sibling B.

crown heights were also noted, with long contacts and relatively flat interproximal surfaces on the existing full coverage restorations. Although the teeth were smaller in size, orthodontic therapy closed any spacing and brought the teeth together in the arch. Minor orthodontic relapse and crowding of the lower mandibular anterior teeth was also noted.

2.2. Treatment

After digital intraoral (TRIOS; 3Shape A/S) and extraoral facial scan were made, treatment plan options were presented and discussed. Both patients elected to proceed with full mouth reconstruction which included full coverage restorations on all of the teeth and crown lengthening and gingivectomies as prescribed. The treatment objectives were to provide a stable and functional occlusion, eliminate pain and sensitivity, improve esthetics, prevent further deterioration of the teeth, and achieve sociopsychological improvements.

For both patients, preliminary intraoral and extraoral facial scans were made and then brought into computer-aided design and computer-aided manufacture (CAD-CAM) software program (Zirkonzahn Modellier; Zirkonzahn USA Inc) to fabricate a digital waxup prior to tooth preparation (Fig. 5A and B). The digital proposal was then used to fabricate milled maxillary and mandibular polymethyl methacrylate (PMMA) shells. The teeth were prepared for complete coverage restorations along with crown lengthening and gingivectomy as prescribed (Fig. 6A–D). Carious lesions and the rough pitted enamel was removed. The PMMA shells were relined chairside at initial tooth preparation and worn for 3 months to allow for healing of the gingival tissue.

The interim restorations were then removed and definitive intraoral scanning was completed. Milled trial restorations (PRE-MIOtemp; primotech USA) were then made for a final trial period of 2 months to confirm occlusal function, esthetics, and phonetics. Minor esthetic changes were requested and incorporated into these trial restorations. During the trial period, the interim restorations exhibited minor wear while functional and esthetic needs were met. Upon patient approval, the definitive zirconia restorations (NexxZr T; Sagemax Bioceramics, Inc) were then fabricated based off the approved design. The definitive restorations were then luted with a dual-polymerizing resin cement (PANAVIA SA Cement Universal; Kurray Noritake Dental Inc) (Fig. 7A–D). A nocturnal occlusal device (Temp Premium; Zirkonzahn USA Inc) was also delivered (Fig. 8A and B).

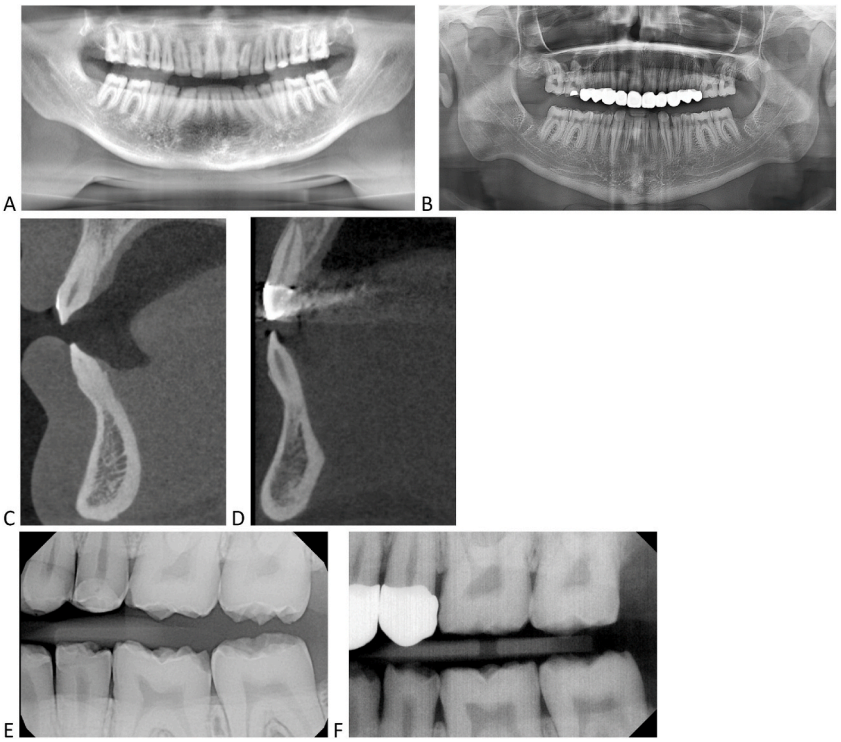

Fig. 4. A, Panoramic radiograph, sibling A. B, Panoramic radiograph, sibling B. C. CBCT cross section of incisors, sibling A. D, CBCT cross section of incisors, sibling B. E, Bitewing radiograph, sibling A. F, Bitewing radiograph, sibling B.

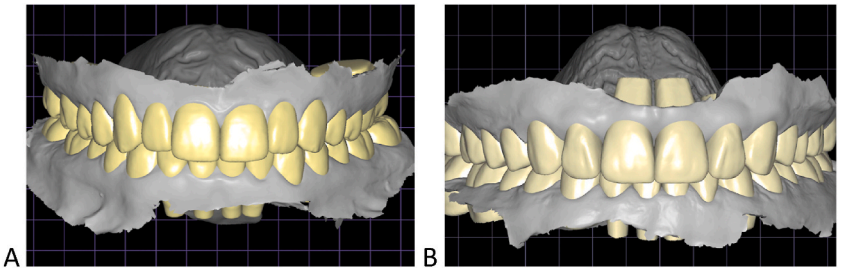

Fig. 5. A, Digital proposal of definitive restorative plan, sibling A. B, Digital proposal of definitive restorative plan, sibling B.

2.3. Results

Both siblings were pleased with the final outcome of their treatment. Esthetics and function were both restored and occlusal stability was achieved at a proper vertical dimension of occlusion. This improved not only mastication but also increased the lower third of the face for an overall improvement of facial proportions. Oral hygiene instructions were reviewed and the patients were placed on a 6-month recall period to ensure oral hygiene was maintained and did not experience any complications. At their 1-year recall (Fig. 8C and D), oral hygiene was deemed adequate and the gingival tissue was stable and healthy. Both patients were satisfied with esthetic and functional outcomes, but also had significant psychosocial improvements and overall better quality of life.

3. Discussion

Reconstruction of patients with AI can be challenging, not only from a structural standpoint, but psychosocial aspects as well. In order to improve overall quality of life, intervention should begin as early as possible [8] and also to prevent further deterioration of the natural dentition [10]. With hypoplastic (type 1) AI the enamel is typically altered quantitatively, which can range from pitting to a decrease in thickness, or complete agenesis of enamel [16]. In the sibling patients, a generalized decrease in enamel thickness and pitting of the enamel was observed. When determining proper timing of beginning prosthodontic intervention, patient maturity, ability to maintain oral hygiene, and occlusal positioning of the teeth must be taken into consideration. Despite having overall

Fig. 6. A, Preparation for complete coverage restorations, sibling A. B, Preparation for complete coverage restorations, sibling B. C, Relined interim restorations immediately after preparation, sibling A. D, Relined interim restorations immediately after preparation, sibling B.

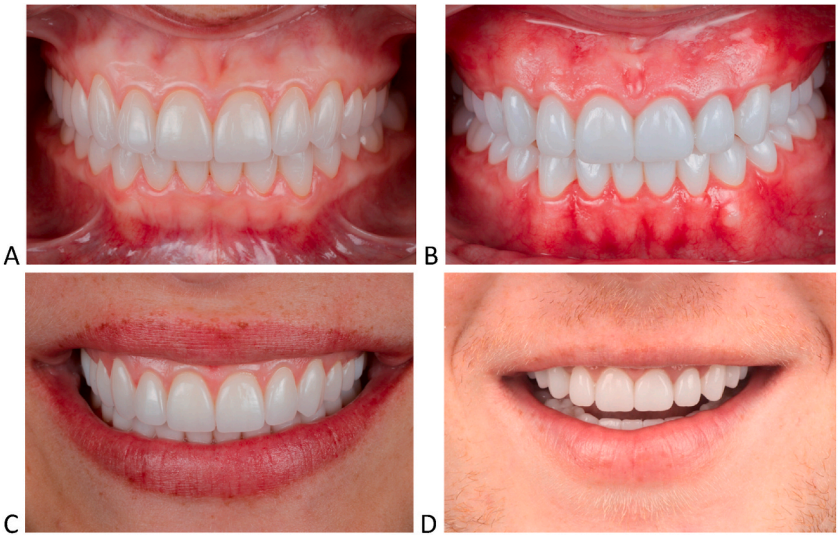

Fig. 7. Definitive zirconia restorations. A, Intraoral view, sibling A. B, Intraoral view, sibling B. C, Smile view, sibling A. D, Smile view, sibling B.

hypersensitivity, like most patients with AI [5], both patients maintained good oral hygiene habits prior to and throughout treatment. They both expressed excitement and motivation to have their reconstruction done, therefore the siblings were deemed good candidates to proceed with rehabilitation. Both patients had also previously completed orthodontic therapy and therefore no contraindications to treatment were found.

Pre-prosthetic orthodontic treatment is beneficial as patients with AI often have malocclusion, skeletal discrepancies, open occlusal relationships, or reverse articulation [11,13]. Patients with AI often require an interdisciplinary approach comprised of an orthodontist, pediatric dentist, prosthodontist, and in extreme cases an oral surgeon to correct skeletal discrepancies with orthognathic surgery [11,12]. Prior to completion of orthodontic therapy, a prosthodontic evaluation would be advantageous in determining final tooth positioning. Due to the hypoplastic nature of the teeth and missing enamel, the crowns of the siblings teeth were smaller and the teeth were ultimately moved together. If prosthetic evaluation had been incorporated prior to completion, the teeth could have been slightly spaced out with small diastemata to create room for the missing enamel which would be restored prosthetically. This is a circumstance that is unique to certain types of AI, where the patient presents with a loss of enamel thickness, and thus a smaller crown size. This commonly occurs in hypoplastic AI (although sometimes hypoplastic AI still presents with a normal thickness of the enamel), or with hypocalcified type 3 AI which is subject to extreme post-eruptive break down of the enamel. In our sibling patients, the overall tooth size, especially in the posterior, was inevitably made slightly smaller due to the final orthodontic positioning of the teeth. The

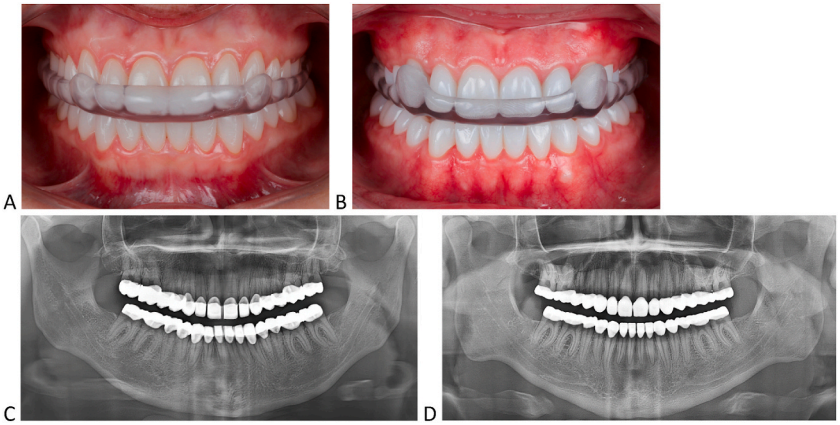

Fig. 8. Maintenance and follow-up. A, Delivery of nocturnal occlusal device, sibling A. B, Delivery of nocturnal occlusal device, sibling B. C, Panoramic radiograph, sibling A. D, Panoramic radiograph, sibling B.

interproximal contacts of the final restorations were also slightly longer with less height of contour in the cervical third of the teeth. An additional round of orthodontic therapy was presented and discussed with both patients, but ultimately declined. Overall, prosthetic, functional and esthetic outcomes were sufficient and the orthodontic therapy was still very beneficial for both patients.

Prosthodontic intervention on patients with AI can be very difficult with unique challenges. In both sibling patients with hypoplastic enamel, a generalized decrease in enamel and pitting was found. This ultimately led to a lack of tooth structure, worn dentition, a decrease in vertical dimension of occlusion, and overly tapered mandibular teeth. Crown lengthening had previously been completed on both patients in the maxillary anterior, which was then done on both patients in the mandibular posterior to achieve more ferrule in preparation for their full coverage restorations.

Hypoplastic AI presents with an alternation in enamel quantity which may present as a decrease in enamel thickness, loss of enamel structurally from pitting, or both. In both siblings, the overall decrease in enamel thickness caused a loss of vertical dimensions of occlusion which was then prosthodontically restored. The hypoplastic enamel in both siblings was also of sufficient quality which allowed for less removal of the enamel on the facial and occlusal surfaces, although more aggressive removal interproximally was done due to the orthodontic movement bringing the teeth together. This also allowed restorative margins to be left in enamel, as long as all pitted enamel was removed. In patients with altered quality of enamel, such as type 2 or 3 AI, if restorative margins are left in defective enamel, secondary caries is likely to occur as the soft enamel is chipped or further worn away.

Both siblings decided to proceed with full mouth reconstruction and full coverage indirect restorations. More conservative options such as direct restorations or composite veneers have presented in the literature [13,14], although typically is more useful in the pediatric population. When direct restorations like facial composite veneers are done at a young age in patients with AI, as the continued eruption occurs and gingiva matures, the restorative margins may become exposed (Fig. 3A) and need to be re-done later in life [14]. Both siblings continuously had to replace such restorations as they grew up and now into adulthood they were ready for a more definitive solution. Despite the composite restorations, the siblings also both still experienced hypersensitivity due to the exposed dentin and thin enamel, which was ultimately resolved with their full coverage restorations. Indirect full coverage restorations have generally been shown to perform well in patients with AI, and rehabilitation should start as early as possible [8] in order to decrease sensitivity, improve overall quality of life, restore function, and circumferentially protect the teeth from further deterioration and caries.

Consideration for future re-treatment of full mouth reconstruction [9] must also be discussed with patients and in the case of adolescent patients, their parents. Sibling B had full coverage restorations previously completed on some of his maxillary teeth, and fortunately the teeth were still deemed restorable upon removal of the existing crowns. Complications such as non-restorability, lack of sufficient tooth structure, recurrent caries [9], and pulpal involvement may be seen when patients are undergoing a second or third re-treatment in their lifetime. When more extensive treatment is completed as a younger age for patients with AI, the possibility of re-treatment may be inevitable, especially when improper diagnosis, management, or follow-up is done. For patients with AI to keep their natural dentition and avoid more invasive treatment such as extractions and implant therapy [9], close monitoring and meticulous oral hygiene is imperative.

Improvements from a psychosocial aspect were seen after initial tooth preparation in both patients. The siblings reported that their hypersensitivity resolved once the interim restorations provided a peripheral seal on all of the teeth. This enabled the patients to clean, eat, and function free of pain for the first time. Monitoring of functional, physical, psychological and social aspects throughout treatment are warranted and encouraged with this patient population. Patient selection and close maintenance monitoring is imperative as poor compliance, improper oral hygiene, and recurrent caries may occur. Further high level clinical research on the longevity and success of full mouth reconstruction in patients with AI is warranted.

4. Conclusion

Treatment of young adults diagnosed with AI can be challenging but should commence as early as possible in order to prevent further deterioration of the teeth and improve overall quality of life. This case report described the prosthodontic rehabilitation of 2 siblings affected by hypoplastic (type 1) AI and the unique challenges that are seen in patients with AI. Orthodontic therapy can be extremely beneficial for this patient population, although prosthodontic evaluation prior to completion of orthodontics may give rise to a better overall result in the rehabilitation. Esthetic, functional, and sociopsychological improvements were seen throughout treatment, but patient selection and close long-term maintenance is necessary to ensure longevity of restorations and compliance.

Data availability statement

This is a case report, so the data were detailed in the manuscript.

CRediT authorship contribution statement

Christina I. Wang: Writing – original draft. **Naif Sinada:** Writing – review & editing.

Declaration of competing 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.

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Appendix A. Supplementary data

Supplementary data to this article can be found online at https://doi.org/10.1016/j.heliyon.2023.e23939.

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