Ectodermal Dysplasia: An 11-Year Follow-up of Siblings With 2 Implant Treatment Approaches

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Purpose: *To describe 2 different treatment approaches for a 20-year-old Caucasian man and his 22-year-old sister who were affected by ectodermal dysplasia (ED) and compromised maxillary bone.*

Materials and Methods: *The sister had a history of an iliac crest transplant with 6 implants placed in the maxillary and mandibular arches, 6 years before complications. It was necessary to debride the right sinus, remove the failing infected bone graft and 2 implants. Her brother presented with missing teeth, few remaining deciduous teeth, and wearing all-acrylic resin temporary partials in both the maxillary and mandibular arches.*

Results: *Zygomatic and pterygomaxillary implants were used to rehabilitate the brother with a screw-retained fixed prostheses. His sister had 6 new maxillary implants. Both siblings' mandibular arches were restored with screw-retained implant-supported prostheses.*

Conclusion: *Although these siblings affected with ED had different treatment approaches, they both eventually obtained successful outcomes. The brother's treatment was expedited. Both siblings have been followed for a period of 11 years, and all postoperative evaluations have been uneventful. (Implant Dent 2014;23:387–393)*

Key Words: ectodermal dysplasia, dental implants, iliac crest graft, zygomatic implants, pterygomaxillary implants, osseointegration

Congenital anomalies are a broad category of health conditions that are present at birth and are a deviation from normal anatomical growth, development, or function. The congenital anomaly develops *in utero* and may have genetic origins.[1,2]

Ectodermal dysplasia (ED) is a congenital anomaly caused by a single abnormal gene or pair of abnormal genes.[2] In congenital ED, teeth are absent at birth, reduced in number or conical in appearance so that partial dentures are required from childhood. ED may present in various degrees of severity, ranging from a slight malformation of the coronal portion of a single tooth to the more commonly recognized "congenitally missing lateral incisors" or the advanced presentation of the syndrome with multiple missing teeth, thinning or absence of facial hair, and reduction of sweat glands.

The most commonly reported form is hypohidrotic ED (HED) (or Christ-Siemens-Touraine Syndrome).[2–4] The oral clinical phenotype of HED results in multiple missing or deformed teeth, lack of alveolar ridge development, and hyposalivation.[5] Because ED is often accompanied by an absence of adult teeth, it can also lead to underdevelopment of the jaws. A hypoplastic maxilla and mandible, having little if any dental support, produces bite collapse and narrowing of the alveolar ridges. This condition produces a diminished appearance of the lower third of the face. The reduction in size of the width of the jaws and supporting musculature is apparent in full-face view. In profile, the underdeveloped jaws create a facial disharmony.[6]

Congenital deformities in the craniofacial region are physically and psychologically devastating and can handicap the individual in many ways. In young adults, these anomalies can have a great impact on the psychosocial component of life.

In present society, young adults who have their academic, business, and social lives ahead of them are most likely to be affected by the psychological implications of esthetic deformities. A distracting appearance because of congenital defects may inhibit normal social interactions and can subconsciously create a negative impact on performance.

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Maxillofacial rehabilitation of young adults afflicted with severe ED can be successfully accomplished using modern treatment concepts. High on the list of therapeutic protocols is the use of osseointegrated dental implants to support nonremovable teeth.[7] Coupled with advanced prosthodontics and ceramic artistry to create natural looking replacement teeth, the overall treatment can yield an esthetically pleasing smile. In 1999, when the treatment of this report commenced, there was a paucity of literature on using dental implants in patients with ED. Ten years later, in 2009, Yap and Klineberg[8] reviewed the literature in patients with ED and found no randomized or case-controlled studies. They found from the literature that 12 case studies showed variable implant survival rates from 88.5% to 97.6% in patients with ED and 90% to 100% in patients with tooth agenesis.[8]

The implant success rate of the mandible was reported in 2005 by Sweeney et al[9] to be higher than the maxilla of a patient with ED. They reported the success rates of 80% in the maxilla and 91.3% in the mandible. In 2002, Guckes et al[10] reported that implant placement in the maxilla had a 2.8 times greater probability of failure than in the mandible.

This report describes the clinical treatment of a 20-year-old man and illustrates the biomechanical and esthetic advantages of modern implant prosthodontic rehabilitation. Paralleling his treatment was the revision treatment of his 22-year-old sister, who, like her brother, suffered with ED. Her original treatment began with surgical intervention by her previous treatment team, using an iliac crest bone transplant inlayed into the maxillary antrum. Subsequent degeneration of the graft required complex revision 7 years later. This 11-year follow-up review will evaluate both siblings and discuss the treatment protocol used for each patient, then evaluate the effect of this treatment over time.

SIBLING CASE REPORT

Brother's Patient History

The patient was a 20-year-old man (Fig. 1, A–D) who at the age of 4 was diagnosed with HED. He was in excellent general health, with occasional sinus problems and seasonal allergies when presenting for treatment related to the reconstruction of his dentition. His dental history revealed that he had undergone multiple tooth extractions and was wearing an all-acrylic resin maxillary overdenture that overlaid his deteriorated posterior dentition. An acrylic resin removable partial denture replaced the mandibular anterior teeth (Fig. 1, B). These temporary prostheses were fabricated at his existing decreased vertical dimension of occlusion.

His chief complaint was that he did not want to wear removable appliances and wanted to improve his appearance.

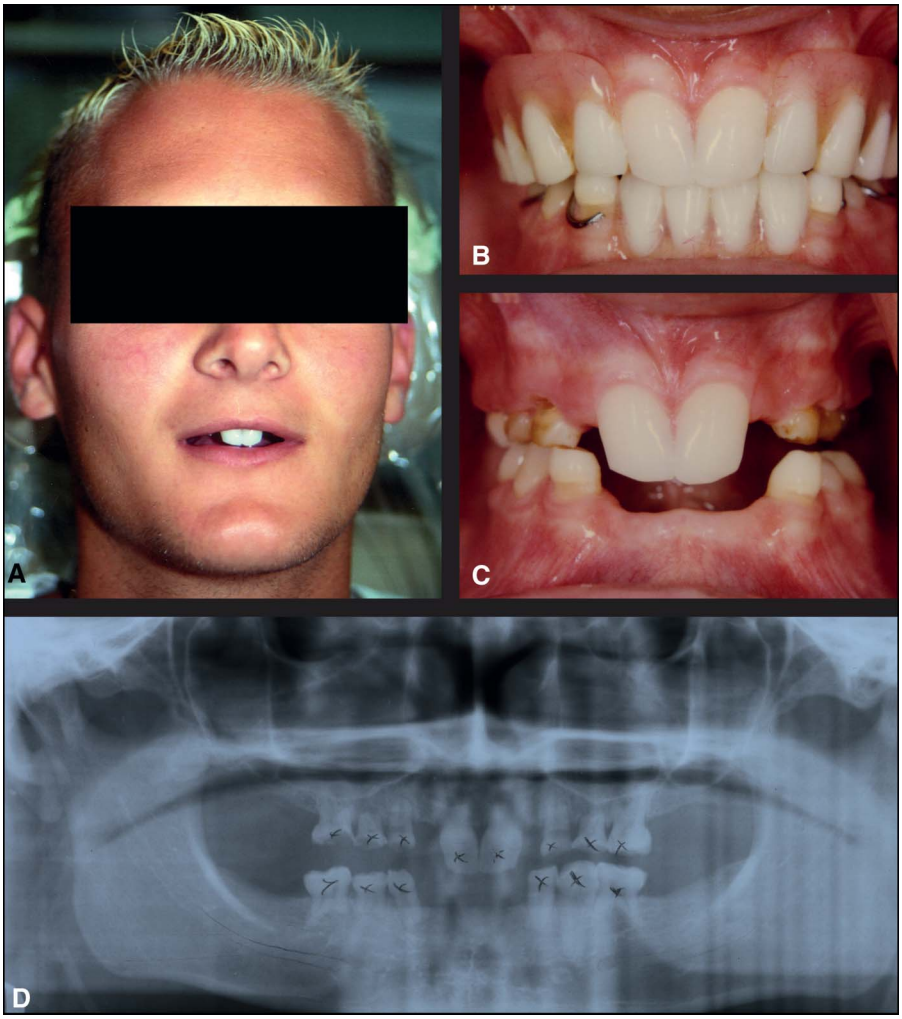

Fig. 1. Initial presentation of brother. **A**, Clinical presentation, (**B**) preoperative frontal view of all acrylic removable partial prostheses, (**C**) intraoral view of maximum intercuspation at decreased vertical dimension of occlusion, (**D**) panoramic radiograph.

Clinical Evaluation and Diagnosis

In 1999, the patient initially presented with the following teeth in the maxillary arch: #s A, B, C, 8, 9, H, I, J. In the mandibular arch, teeth #s K, L, M, R, S, and T were present.

His clinical examination revealed that he was in a skeletal Class I relationship. He showed severe decay and wear on occlusal surfaces of his posterior teeth, accompanied with loss of vertical dimension of occlusion (Fig. 1, C). Radiographic examination revealed horizontal and vertical bone loss, resorption of posterior roots, and significant underdevelopment of the alveolar bone, particularly in the posterior maxilla (Fig. 1, D).

His remaining dentition had a poor prognosis to serve as abutments for

Table 1. Implant Placement of Brother

Location of Implant Areas	Implant Size and Type
1, 16	18 × 3.75 mm; Brånemark
3	40 mm; Zygomatic
14	35 mm; Zygomatic
6, 11	10 × 3.75 mm; Brånemark
7, 10, 22, 24, 26, 28	15 × 3.75 mm; Brånemark
19, 30	10 × 4.0 mm; Brånemark

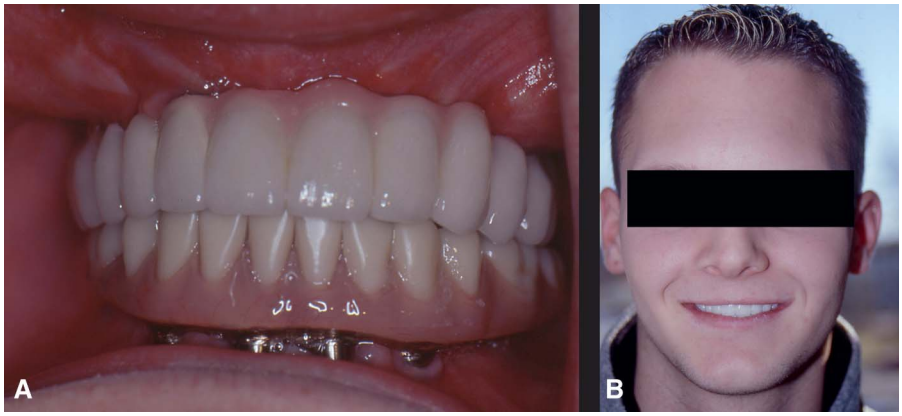

Fig. 2. Postoperative presentation of brother. **A**, Retracted frontal view of completed maxillary and mandibular prostheses, (**B**) clinical presentation showing increased vertical dimension of occlusion and good facial support.

fixed restorations. Other options included complete maxillary and mandibular dentures, or implant-retained overdentures. A complete maxillary denture would have presented retention challenges for the patient because of his shallow palate. Not replacing the teeth with implants would have allowed continued ridge resorption in both arches.[11,12]

A comprehensive treatment plan was presented to him and his family to include placement of Brånemark System dental implants (Nobel Biocare, Yorba Linda, CA) in the maxillary and mandibular arches immediately after removal of his remaining dentition. One zygomatic implant[13–17] bilaterally was included in the treatment plan to be placed in the molar region of the maxilla where bone volume was diminished. Other treatment options were presented to the patient to include iliac bone grafting and long-term use of complete removable prostheses. The patient expressed his desires for fixed prostheses. Ultimately, the family accepted the treatment plan for maxillary and mandibular screw-retained prostheses.

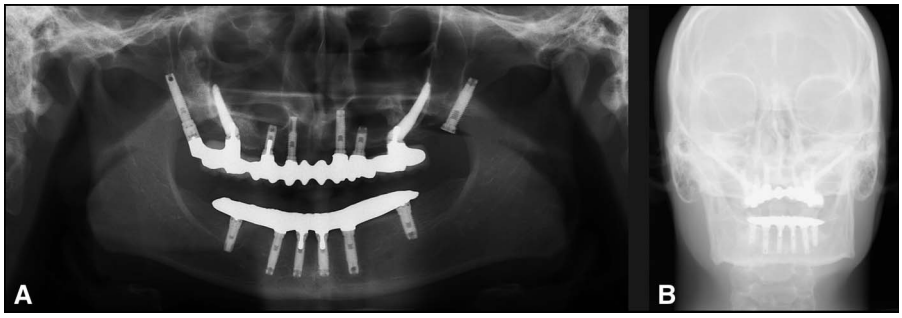

Fig. 3. Eleven-year postoperative follow-up of brother. **A**, Panoramic radiograph, (**B**) anteroposterior cephalometric radiograph.

Diagnostic casts were made and articulated at his existing vertical dimension of occlusion. The second set of diagnostic casts were made and articulated at a newly reestablished vertical dimension of occlusion giving the patient 3 mm of freeway space. The reestablished vertical dimension was recorded with an interocclusal bite registration (Regisil; Dentsply, York, PA). Diagnostic casts were articulated at this improved and increased occlusal vertical dimension, which was later used to fabricate transitional dentures for the maxillary and mandibular arches.

Clinical Treatment

General anesthesia with nasal intubation was administered in an outpatient setting. All maxillary and mandibular teeth were extracted. Crestal incisions were made along with flap elevation for both arches. The necessary anatomical landmarks in both arches were identified before making any preparations in the bone. Before the surgery, the determination on the position of the implants was made from panoramic radiographic analysis. Four 3.75 × 15 mm standard Brånemark System implants were placed in the anterior mandible between the mental foramen. Distal to the extraction socket of the last primary molar, 2 additional 4.0 × 10 mm Brånemark System implants were placed, 1 on each side. All the bones from the mandibular osteotomy sites were harvested for use in the maxilla. Cover screws were installed over the implants and tissue flaps were securely closed.

The maxilla was operated with bilateral antral lateral wall openings, which created directional visibility for placement of the zygoma implants. Eight implants in total were placed in the maxillary arch: 4 standard length Brånemark System implants in the anterior maxilla along with bilateral zygomatic and pterygomaxillary implants.[18,19] The autogenous bone harvested from the mandibular implant osteotomy sites was combined with a natural bovine-derived bone substitute (Bio-Oss; Luitpold Pharmaceuticals Inc., Shirley, NY) and with autologous platelet rich plasma gel treated to release growth factors.[20,21] This graft material was placed in the antral floor of the maxilla and around

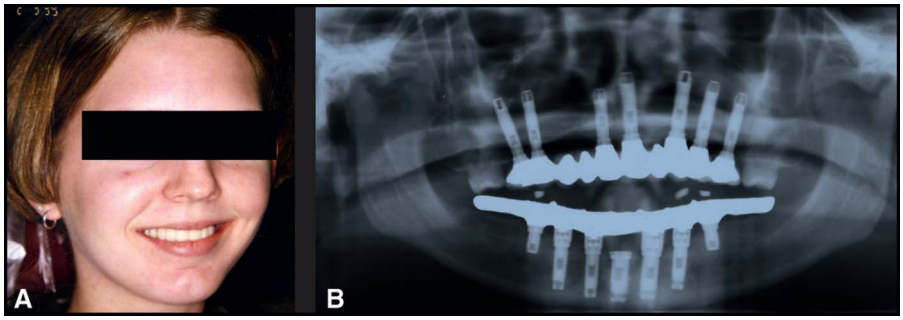

Fig. 4. Initial presentation of sister. **A**, Clinical presentation, (**B**) panoramic radiograph.

the zygoma implants to increase the bone-to-implant surface area. Similarly to the mandible, cover screws were installed over the 8 maxillary implants, and tissue flaps were securely closed. Table 1 shows the distribution of all 14 implants placed in both arches.

The patient returned 10 days postoperatively for suture removal. The maxillary and mandibular complete dentures were seated after a soft liner was placed. Second stage surgery[22] and final implant prosthodontic treatment commenced at 3 and 5 months, respectively for the mandible and maxilla. When the abutments were connected, the maxillary and mandibular conversion prostheses[23,24] were delivered. A screw-retained implant-supported fixed prosthesis was fabricated using acrylic on a gold framework for the mandibular arch (Fig. 2, A and B). In the maxilla, 1 pterygomaxillary implant was not connected because of sensitivity, despite the fact that it was clinically immobile. The definitive porcelain-fused-to-gold maxillary prosthesis was designed to fill the labial and buccal spaces, which had been void because of underdevelopment of the maxilla (Fig. 2, A and B).

Table 2. Initial Presentation of Sister

Location of Areas	Implant Size and Type
4, 5, 6, 8, 9, 12, 13	20 × 3.75 mm; Brånemark
21, 23, 24*, 26, 27	15 × 3.75 mm; Brånemark
20, 29	8.5 × 3.75 mm; Brånemark
3, 14	Natural teeth

*This implant was not connected to tissue-integrated acrylic-fused-to-gold prosthesis.

Follow-up care consisted of routine oral hygiene visits and periodic radiographic reevaluation. There have been no complications or negative physiologic changes in this patient for the past decade. Normal occlusal wear necessitated a "retread" 9 years after initial delivery of the final mandibular prosthesis. A retread is the process in which worn acrylic resin denture teeth are stripped from a metal framework and are replaced with new acrylic resin denture teeth at the original vertical dimension. His esthetic features related to the lower third of the face have remained stable. Hard and soft tissue response to the functional implants also remains stable. The patient is wearing a maxillary acrylic occlusal guard at night. The patient has been followed on a yearly basis for maintenance visits (Fig. 3, A and B) for the past 11 years.

SISTER'S TREATMENT

The sister's revision process began a month before her brother initiated treatment. According to the patient's mother, she underwent an iliac crest bone graft to augment the posterior maxilla 7 years before. After a prolonged healing period, implants were placed in both the maxilla and mandible, and fixed prostheses were constructed. The following 7 years found the patient in continuous treatment with several otolaryngology specialists for infected sinuses and failing bone grafts. The patient had been on and off antibiotics for several years.

Table 3. Implants After Retreatment of Maxillary Arch of Sister

Location of Implant Areas	Implant Size and Type
14, 16	20 × 3.75 mm; Brånemark
4, 5, 10, 11	4 × 15 mm; Ebon

The patient's initial visit was to diagnose and treat her pain and swelling in the right side of the maxilla (Fig. 4, A and B). Table 2 shows the implants and teeth at the patient's initial presentation. Under local anesthesia, a full flap was elevated on the maxillary right side. Two dehiscences were identified in the lateral wall of the sinus. The window was enlarged to facilitate debridement of the infected sinus tissues. Evaluation of the implants in the area of #4 and #5 found both implants without osseointegration. These implants were removed. The third implant in the area of #6 was visualized through the sinus window. The coronal third of this implant was well osseointegrated; however, the remaining portion was surrounded by infected failing graft material and granulation tissue. Partial resection of this implant was performed by using a high-speed diamond bur. The majority of the exposed implant was removed, leaving the stable coronal portion and a small area of the apex in healed bone.[25] The entire surgical site was irrigated with a mixture of Tetracycline (Bristol-Myers Squibb, Plainsboro, NJ) and Amoxicillin (GlaxoSmithKline, Philadelphia, PA) in sterile saline. One vial of Bio-Oss Bone (Luitpold Pharmaceuticals Inc.) was grafted into the area and stabilized with a resorbable membrane that was held with 9 titanium tacks (Biomet Microfixation LLC, Jacksonville, FL). Primary closure was then obtained where the implants were lost.

The prosthetic cylinders on the patient's existing implant-supported prosthesis were filled with acrylic in the areas of #4 and# 5. The lingual and occlusal surfaces were relieved on the right side so that excess pressure would not be exerted on the cantilevered portion of the restoration or the remaining portion of the implant in area #6. Postoperative instructions were given to the patient, and she was instructed to chew on the left side.

One month later, the patient presented with a similar complication in the maxillary left side. Again, sinus debridement was accomplished in conjunction with the removal of

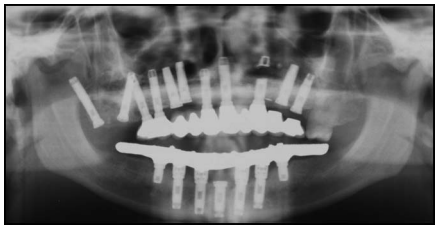

Fig. 5. Panoramic view of 6 new implants placed areas #4, 5, 10, 11, 14, 16 and partial resection of implant #6.

nonintegrated implants in the posterior area. After a 5-month healing period, the patient began the next phase of implant reconstruction.

Under general anesthesia, 6 additional implants were placed into the well-healed bone: 2 in the right posterior and 4 in the left posterior (Table 3). Tooth #14 was also removed to permit implant placement in the pterygomaxillary area. Cover screws were installed on the new implants, and the original prosthesis was fastened to the remaining 5 original implants (Fig. 5).

Five months from the time of implant placement, second stage surgery was performed under general anesthesia and the abutments were connected on the new implants. One pterygomaxillary implant in the area of #16 was not osseointegrated and removed. The primary tooth in the area of #3 was also removed at that time. A master impression for the maxillary and mandibular arches was made, and an all-acrylic conversion prosthesis[23,24] supported by 10 osseointegrated implants was delivered to the maxillary arch. An all-acrylic conversion prosthesis supported by the 6 previously placed implants in the mandibular arch was also fabricated. Two weeks later, the patient returned for suture removal and delivery of the definitive porcelain-fused-to-gold maxillary screw-retained prosthesis and the acrylic-fused-to-gold mandibular screw-retained prosthesis (Fig. 6, A–D). The implant in the area of #24 was not connected to the final prosthesis because of the close proximity of the other implants. The cantilever length of the final prosthesis was shortened to prevent mechanical overload and fatigue failure of the gold framework.

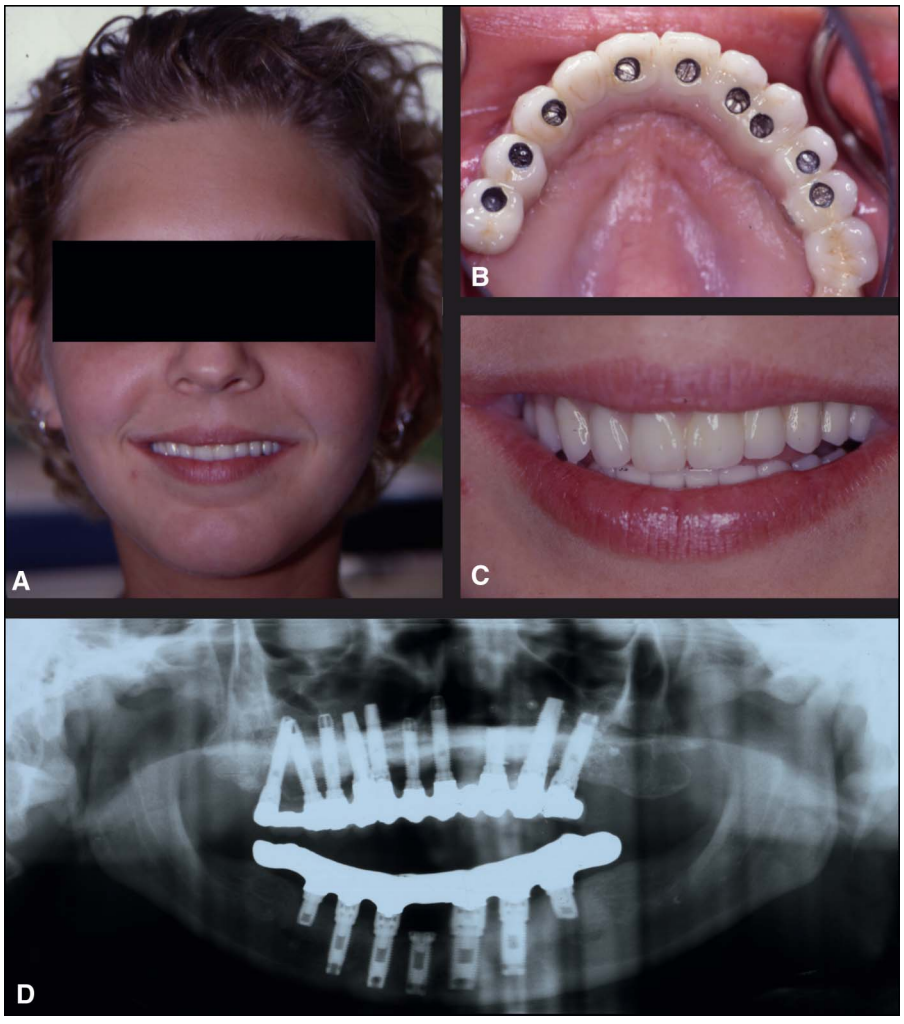

Fig. 6. Presentation of sister after revision treatment completed. **A**, Clinical presentation, (**B**) occlusal view of maxillary implant-supported fixed prosthesis, (**C**) frontal view of esthetic smile and newly fabricated definitive prostheses, (**D**) panoramic radiograph.

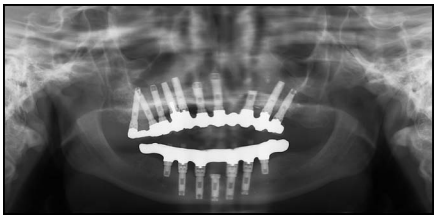

Fig. 7. Panoramic radiograph 11 years after revision treatment.

The patient has been followed uneventfully during routine oral hygiene visits for the past decade. Radiographically, bone levels remain consistent (Fig. 7). Clinically, the mucosal tissues have a healthy response to the titanium abutments and the prostheses are functional and stable. The apex of the implant in the area of #6 was ultimately removed at a subsequent surgical procedure.

DISCUSSION

Brånemark System implants were used successfully in treating siblings suffering from ED. Two different treatment approaches were described. The sister's revision treatment involved the resolution of an infected iliac crest graft and also loss of 3 implants, #4, #5, and #16. The graft was removed, and the area was allowed to heal and subsequently 6 new implants were placed. The brother's treatment was abbreviated because it bypassed the involvement of bone grafting. The combination of zygomatic and pterygomaxillary implants as well as implants in the anterior maxilla allowed for predictable treatment and the connection of a screw-retained prosthesis.

In general, clinicians have begun to explore the possibilities of shortening treatment periods by earlier delivery of implant-supported restorations or by placement of implants in extraction sockets at the time of extraction.[26–28]

As recent as 2011, D'Ambrosio et al[29] described a clinical case of ED treated with an immediate implant-loading protocol and until that time, there had been no clinical reports on implant-loading protocol in a patient with ED. Not only does immediate loading expedite treatment, it also minimizes hard and soft tissue loss and ensures a better esthetic outcome.[30,31] It also increases the psychological benefit, which can be important in patients afflicted with ED. At the time of implant placement for the siblings in this report, immediate loading in the maxilla was not a widely accepted and evidence-based concept. If the treatment in this report were performed today, both patients would have all the implants immediately loaded with an all-acrylic resin conversion prosthesis, the day of implant placement thereby expediting the overall rehabilitation. The use of immediately loaded pterygomaxillary implants eliminates the need for bone grafting, and recent research[32] has shown that these types of implants have a cumulative survival rate of 96.45%. Using zygomatic and pterygomaxillary implants to support a fixed prostheses is an effective and viable treatment option for patients with ED and eliminates the long-standing struggle with ill-fitting, uncomfortable, or unsightly removable prostheses. Given the findings from the current literature, an immediate loading protocol could be a predictable therapy for patients with ED.

Both the sister and brother had struggled odontologically from a combination of oligodontia and anodontia and also severely compromised bone volume. The removal of the brother's teeth without immediate reconstruction would most likely leave the patient with an edentulous state that would continue to deteriorate because of ongoing alveolar atrophy.[11,12]

CONCLUSION

The treatment described in this report provided the siblings with an excellent long-term prognosis and had a positive emotional impact on their lives. This treatment protocol is an improved standard of care for patients suffering from ED. In current treatment of patients with ED, an expedited treatment approach with immediate function can be employed.

DISCLOSURE

T. J. Balshi and S. F. Balshi declare that they have an ongoing relationship for lectures including service on speakers' bureaus with Nobel BioCare. The remaining authors claim to have no financial interest, either directly or indirectly, in the products or information listed in the article.

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