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CLINICAL REPORT

Amelogenesis imperfecta: Analysis of the genetic basis and treatment with a digital workflow: A clinical report

Patrick Klein, DMD, Dr med dent,[a] Johanna S. Hoppe, DMD,[b] Nuria C. Bramswig, MD, PD Dr med,[c] and Petra C. Gierthmuehlen, DDS, Dr med dent habil[d]

ABSTRACT

A 20-year-old woman with type I hypoplastic amelogenesis imperfecta (AI) experiencing esthetic concerns and hypersensitivity is presented. The treatment was aimed at both functional and esthetic aspects, focusing on complete mouth rehabilitation using a fully digital workflow. In an interdisciplinary approach, a multigene panel analysis was performed. Digital intraoral scans and a face scan facilitated precise virtual planning, guiding minimally invasive preparations. Long-term interim restorations milled from high-performance polycarbonate preceded definitive restorations in translucent zirconia. This approach resulted in a time- and cost-efficient treatment, helped the patient to understand her disease by highlighting the pivotal role of genetics, and demonstrated the success of collaborative dental interventions. A strict recall program will be most important for the long-term success of this patient with AI and a pathogenic frameshift variant in the *AMELX* gene. (J Prosthet Dent 2025;133:1387-1396)

Amelogenesis imperfecta (AI) represents a group of conditions characterized by enamel malformation associated with genetic disorders.[1] Therapeutically, early recognition is essential, followed by an interdisciplinary treatment plan involving prosthodontics and, in severe cases, endodontics, orthodontics, and maxillofacial surgery.[2–4] Prosthetic treatments such as complete coverage crowns, implants, fixed, or removable prostheses are often necessary.[3,5–7] Evidence-based guidelines for the treatment concepts of patients with AI are lacking, with guidance currently provided by case reports and case series.[8] However, a review recommended the early insertion of indirect restorations, which have better outcomes than composite resins. A twofold higher failure rate was observed for direct restorations on AI-affected teeth (63%) compared with unaffected teeth (27%).[9] The objective of this clinical report was to describe the genetic aspects of AI and to describe comprehensive treatment with a fully digital workflow.

CLINICAL REPORT

A 20-year-old woman with type I hypoplastic AI presented with significant esthetic impairment and pain associated with severe hypersensitivity (Schiff[10] score: 3) (Fig. 1). Overall, she was in good physical health and did not take any medication. A multigene panel analysis, including AI-associated genes, revealed a pathogenic

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[a]Assistant professor, Department of Prosthodontics, Medical Faculty and University Hospital Düsseldorf, Heinrich-Heine-University, Düsseldorf, Germany; Moorenstraße 5, Düsseldorf, Germany; and Assistant professor, Center for Rare Diseases, Medical Faculty and University Hospital Düsseldorf, Heinrich-Heine-University Düsseldorf, Germany; Moorenstraße 5, 40225 Düsseldorf, Germany.
[b]Assistant professor, Department of Prosthodontics, Medical Faculty and University Hospital Düsseldorf, Heinrich-Heine-University, Düsseldorf, Germany; Moorenstraße 5, Düsseldorf, Germany.
[c]Associate professor, Institute of Human Genetics, Medical Faculty and University Hospital Düsseldorf, Heinrich-Heine-University Düsseldorf, Germany; Moorenstraße 5, 40225 Düsseldorf, Germany; and Associate professor, Center for Rare Diseases, Medical Faculty and University Hospital Düsseldorf, Heinrich-Heine-University Düsseldorf, Germany; Moorenstraße 5, 40225 Düsseldorf, Germany.
[d]Professor and Chair, Department of Prosthodontics, Medical Faculty and University Hospital Düsseldorf, Heinrich-Heine-University, Düsseldorf, Germany; Moorenstraße 5, 40225 Düsseldorf, Germany; and Professor and Chair, Center for Rare Diseases, Medical Faculty and University Hospital Düsseldorf, Heinrich-Heine-University Düsseldorf, Germany; Moorenstraße 5, 40225 Düsseldorf, Germany.
[1]Petra C. Gierthmuehlen and Nuria C. Bramswig contributed equally to this case report.

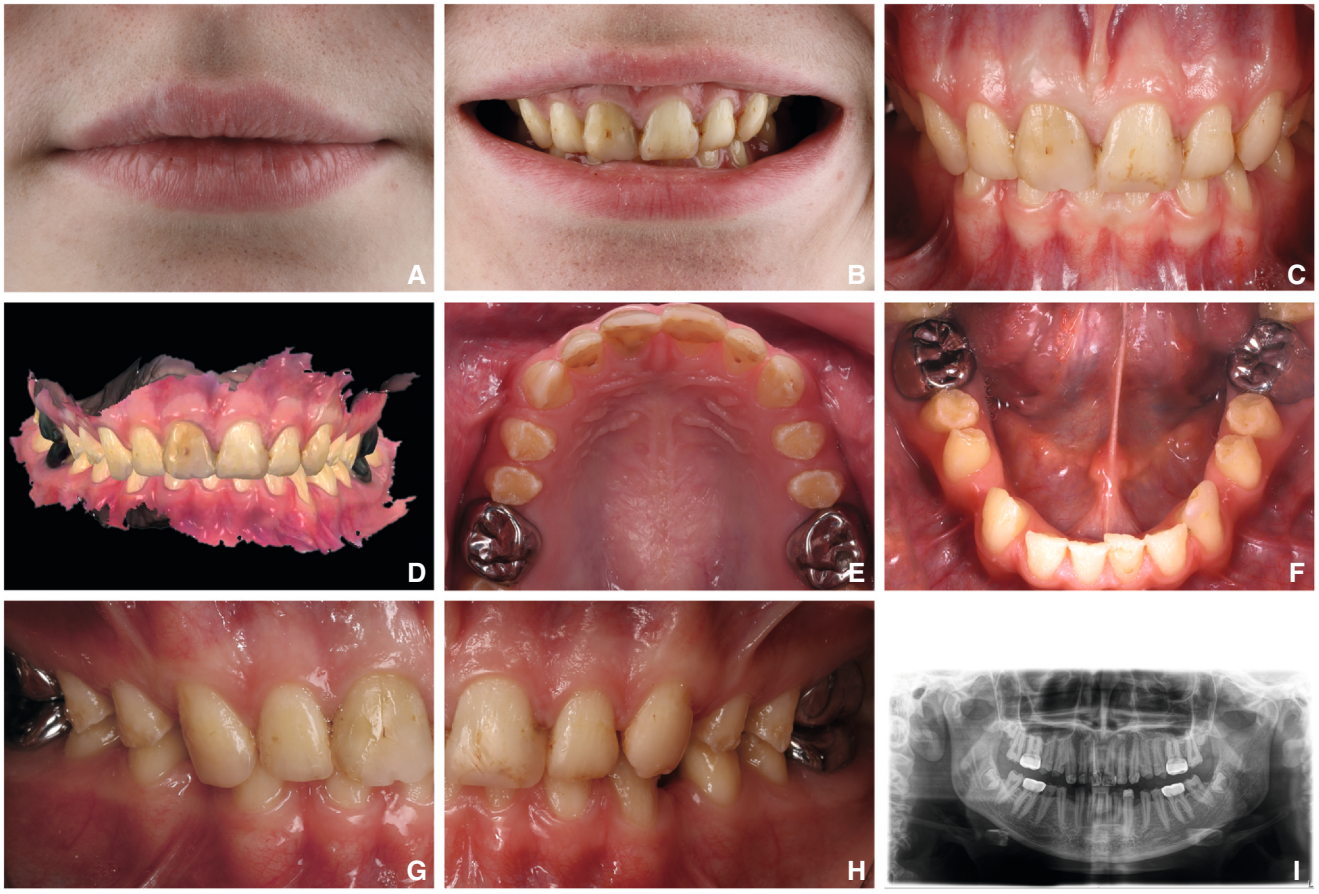


Figure 1. Preoperative presentation. A, Extraoral view (closed lips). B, Extraoral view (smiling). C, Intraoral view. D, Preoperative scan. E, Maxillary occlusal view. F, Mandibular occlusal view. G, Lateral view. H, Lateral view. I, Panoramic radiograph.

frameshift variant in the *AMELX* gene (NM_001142.2: c.431del, p.[Pro144Hisfs*31]), confirming and further specifying this diagnosis to type IE hypoplastic AI (OMIM #301200). Intraoral examination revealed partially discolored enamel with pits and grooves on all teeth, reduced enamel thickness, and wear facets. This clinical phenotype resulted from X-inactivation, a physiological process in which one of the two X-chromosomes is randomly and permanently inactivated in all cells in women (besides germ cells). The irregularly affected enamel in this heterozygous woman was attributed to random inactivation of the X chromosome with and without the pathogenic *AMELX* variant, leading to side-by-side production of dysfunctional and normal enamel, respectively. She had been referred by a general dentist and had received small restorations, metal crowns on the first molars, and endodontic treatment on her maxillary right central incisor. Her anterior teeth had deficient composite resin restorations. The metal crowns had been inserted shortly after eruption and exhibited restoration margins with excessive contours. Her periodontal findings included a bleeding on probing (BOP) index of 12% without clinical attachment loss and a plaque index of 7%. She maintained good oral hygiene and was diagnosed with localized plaque-induced gingivitis,[11] class II (division 2) Angle classification of occlusion,[12] and adequate transverse orthodontic relationships. The prognosis for all teeth was rated as

good,[13] her third molars were unerupted, and she exhibited excessive gingival display but declined correction through orthodontic treatment or periodontal surgery.

The aim of the treatment was to provide a complete mouth rehabilitation with an esthetic smile and to reduce her severe hypersensitivity. Three treatment options were considered: direct composite resin restorations, indirect partial coverage restorations, and complete coverage crowns. To meet the patient's requirements and restore the teeth proportionally with a new vertical dimension of occlusion (VDO), complete coverage crowns were recommended. The purpose of increasing the VDO was to enable minimally invasive tooth preparation and create adequate prosthetic space.

The treatment plan consisted of 4 digital phases: esthetic analysis, treatment simulation, trial restorations, and definitive restorations. Initially, oral hygiene instructions were provided, carious lesions were treated, and the deficient composite resin restorations were replaced. A fluoride varnish (Profluorid Varnish; Voco) was applied to promote remineralization, prevent dental caries,[14] and reduce hypersensitivity.[15,16] Her dentition

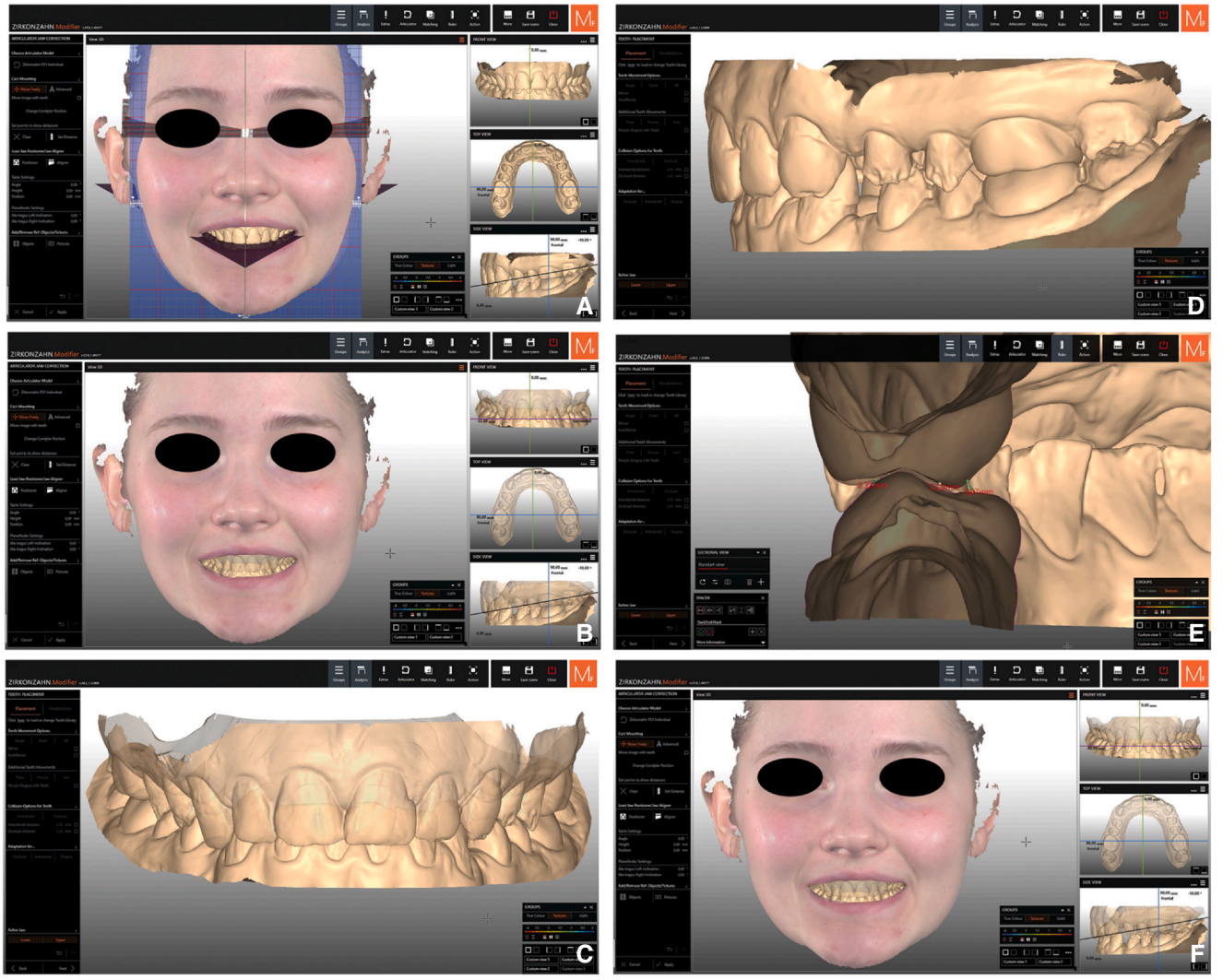
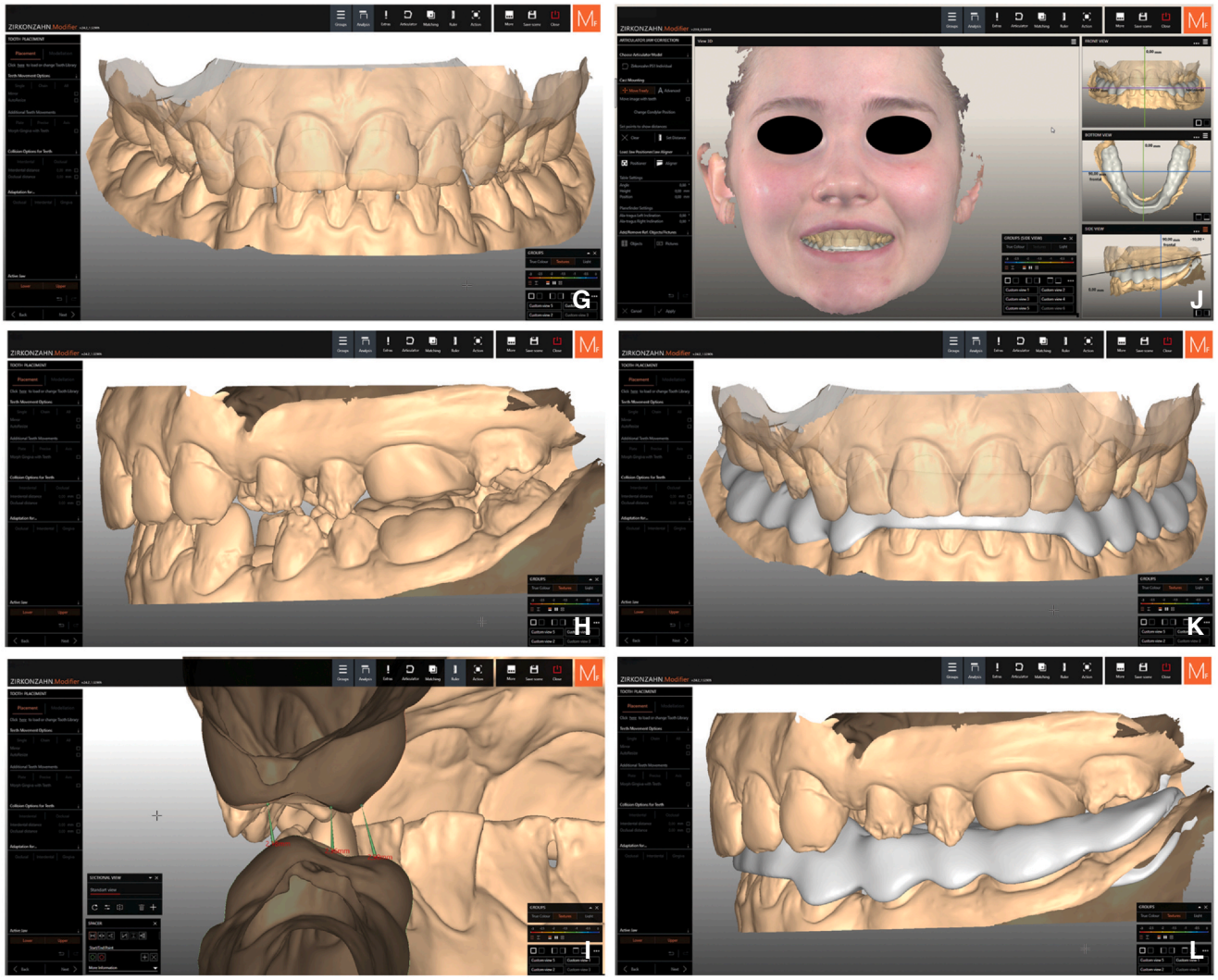


Figure 2. Screenshots of design software program with merged face scans and intraoral scans. A, Initial situation with reference planes. B-E, Initial situation without reference planes and existing VDO. F-I, Increased VDO. J-M, Increased VDO and mandibular device. N-Q, Digital overlap of initial situation and design for treatment simulation. R-U, Digital design of definitive restorations. VDO, vertical dimension of occlusion.



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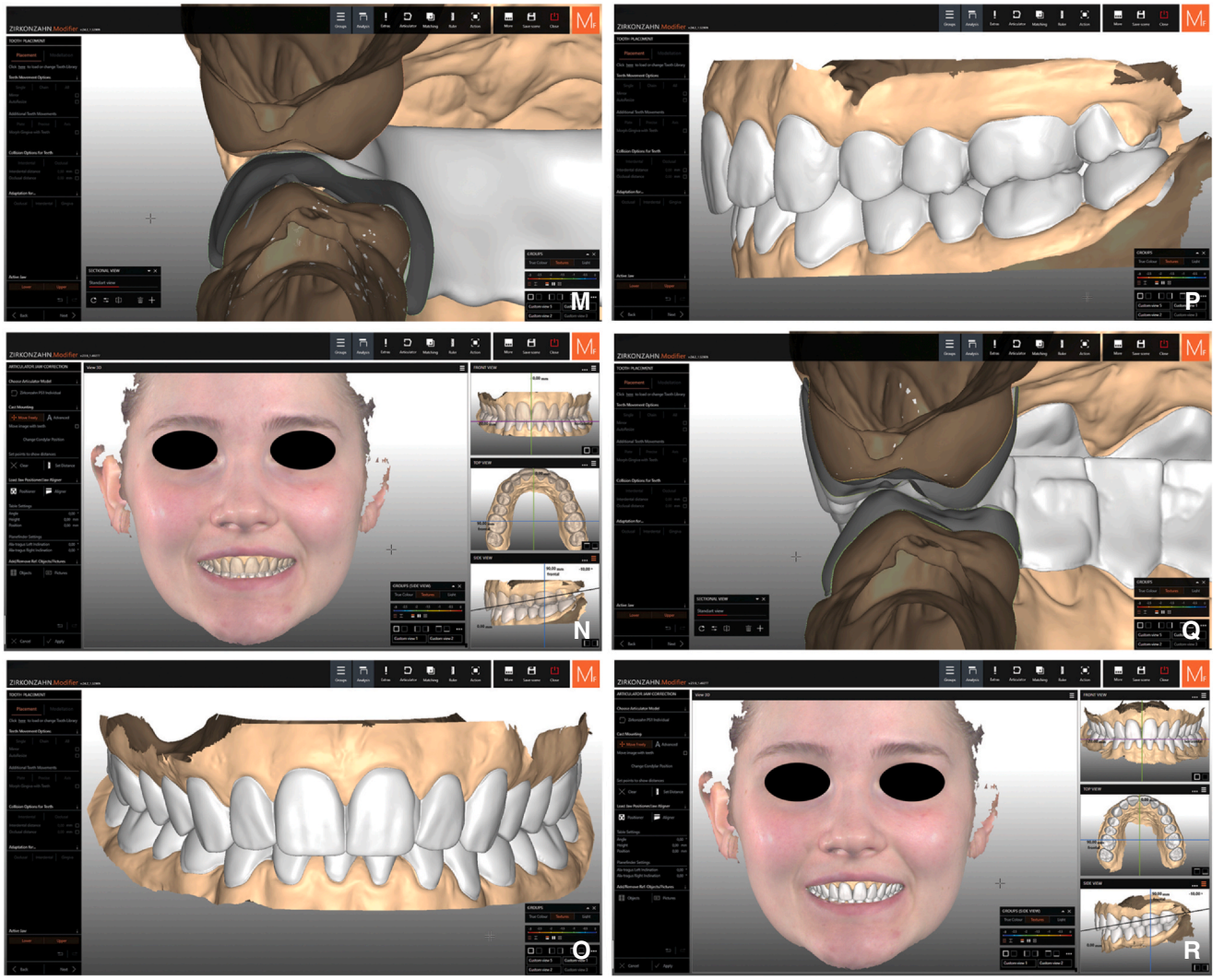
was then digitized using an intraoral scanner (TRIOS 3; 3Shape A/S). Anatomic landmarks of the face and reference planes were captured with a digital face scanning system (Face Hunter; Zirkonzahn). Subsequently, the digital data of the intraoral and facial scans were uploaded and merged into a design software program (Zirkonzahn.Modifier software program; Zirkonzahn) to visualize the initial situation (Fig. 2). The new VDO was developed with her input. Before determining the maxillo-mandibular registration in a centric relation position, an interim disposable diagnostic device (Aqualizer; Dentrade) was inserted followed by a device (Futar D fast; Kettenbach) placed in the anterior region (maxillary central incisors). The anterior device was adjusted until an optimal maxillo-mandibular relation for a correct speaking distance was achieved. The new VDO



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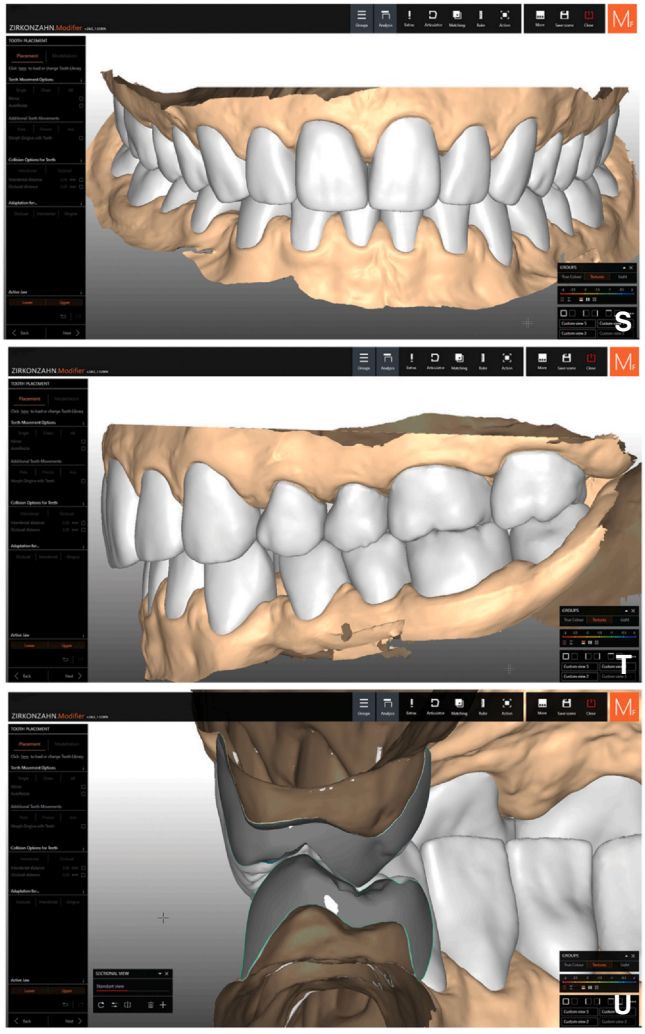
was captured with an intraoral scanner (TRIOS 3; 3Shape A/S) and an additional facial scan (Face Hunter; Zirkonzahn) while the device was inserted.

Both data sets were re-uploaded and merged into a design software program (Zirkonzahn.Modifier software program; Zirkonzahn) to visualize the new facial proportions. This alignment enabled the dental laboratory technician to make an esthetic analysis and to plan a referenced virtual treatment simulation. A mandibular device (Therapon Transpa; Zirkonzahn) was digitally designed, fabricated, and provided to assess the functional aspects of the increased VDO. After 6 weeks of use, she reported no concerns. The dental laboratory technician then digitally designed the treatment simulation at the increased VDO



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using a digital tooth library (Heroes Collection; Zirkonzahn). The 3-dimensional face scan facilitated the design of the teeth in relation to the smile line and to her face.

Maxillary and mandibular polycarbonate tooth-colored treatment simulation devices (Temp Premium Flexible; Zirkonzahn) were milled (M2 Teleskoper milling unit; Zirkonzahn) to visualize the function and esthetic of the planned rehabilitation (Fig. 3). The patient approved the esthetics and function of the treatment simulation devices. The teeth were then minimally prepared (Fig. 4), followed by an intraoral scan (TRIOS 3; 3Shape A/S) with a cross-mounting interocclusal registration scan. CAD-CAM interim restorations were milled (M2 Teleskoper milling unit; Zirkonzahn) from a high-performance polycarbonate (Temp Premium Flexible; Zirkonzahn) and inserted for 3 months with an interim cement (TempBond NE; Kerr Corp) (Fig. 5). A canine protected articulation was established, and she received thorough oral hygiene instructions. During the long-term interim phase, she approved the functional (occlusion and phonetic) and esthetic aspects. A definitive intraoral scan (TRIOS 3; 3Shape A/S) of the long-term interim restorations and abutment teeth was made. Occlusal schemes during the wearing phase of the interim restorations were incorporated into the design of the definitive restorations. All other aspects of the design were identical to the initial interim restorations, including the canine protected articulation, and no changes were made. Definitive crowns were milled from monolithic translucent zirconia (Prettau 3 Dispersive; Zirkonzahn) (M3Y-5Y Zirconia generation[17]) in a 5+1-axis milling unit (M2 Teleskoper milling unit; Zirkonzahn). The restorations were customized (ICE Stains 3D; Zirkonzahn), sintered, (Zirkonofen Turbo; Zirkonzahn),

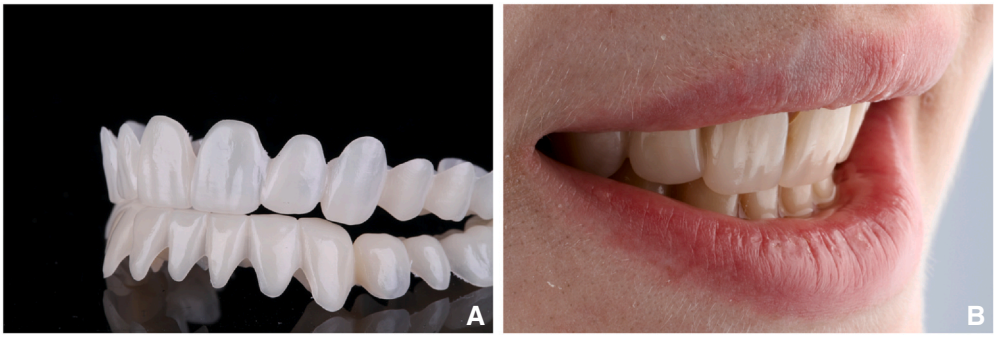

Figure 3. Treatment simulation. A, Maxillary and mandibular treatment simulation polycarbonate devices. B, Lateral view on inserted devices.

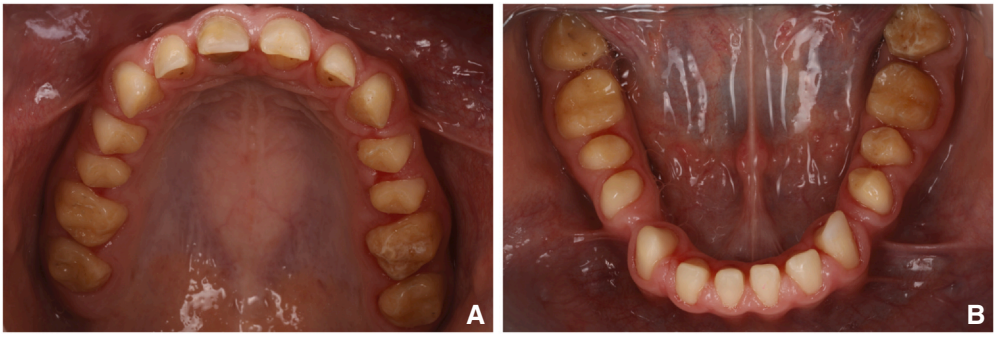

Figure 4. Minimal preparation. A, Maxillary occlusal view. B, Mandibular occlusal view.

Figure 5. Interim restoration. A, Intraoral view. B, Maxillary occlusal view. C, Mandibular occlusal view.

and then adhesively cemented with a resin cement (Panavia V5 opaque; Kuraray) according to the manufacturer's instructions (Figs. 6–8). She was provided with a CAD-CAM mandibular occlusal guard (Therapon Transpa; Zirkonzahn) to protect the restorations during sleep and ensure the long-term stability of the definitive restorations.[18] The marginal inflammation before treatment was most likely attributed to the AI related, rough tooth surfaces.[19] This factor should be eliminated after insertion of the definitive zirconia restorations.[20]

She was placed on a regular 6-month recall schedule to ensure effective oral hygiene and to conduct dental examinations focusing on maintaining stable dental and periodontal conditions.[21] Additionally, an amine fluoride gel (Elmex Gelée; CP GABA) for caries prevention was recommended for weekly use.[22]

DISCUSSION

Although enamel is quantitatively reduced in patients with type I AI, it remains mineralized,[23] while dentin exhibits a hypomineralized structure.[24] The caries susceptibility in patients with AI is controversial.[25,26] Patients with type IE AI tend to have low risk of dental caries, possibly because of the lack of proximal contacts and a high salivary pH.[25] A prospective clinical study showed no secondary caries in patients with AI after treatment with crowns in a 2-year follow-up.[20]

The complete rehabilitation of patients with AI poses challenges for both the dental team[27] and the patients, who are typically young and require extensive treatment. Esthetic and functional impairments as well as hypersensitivity can

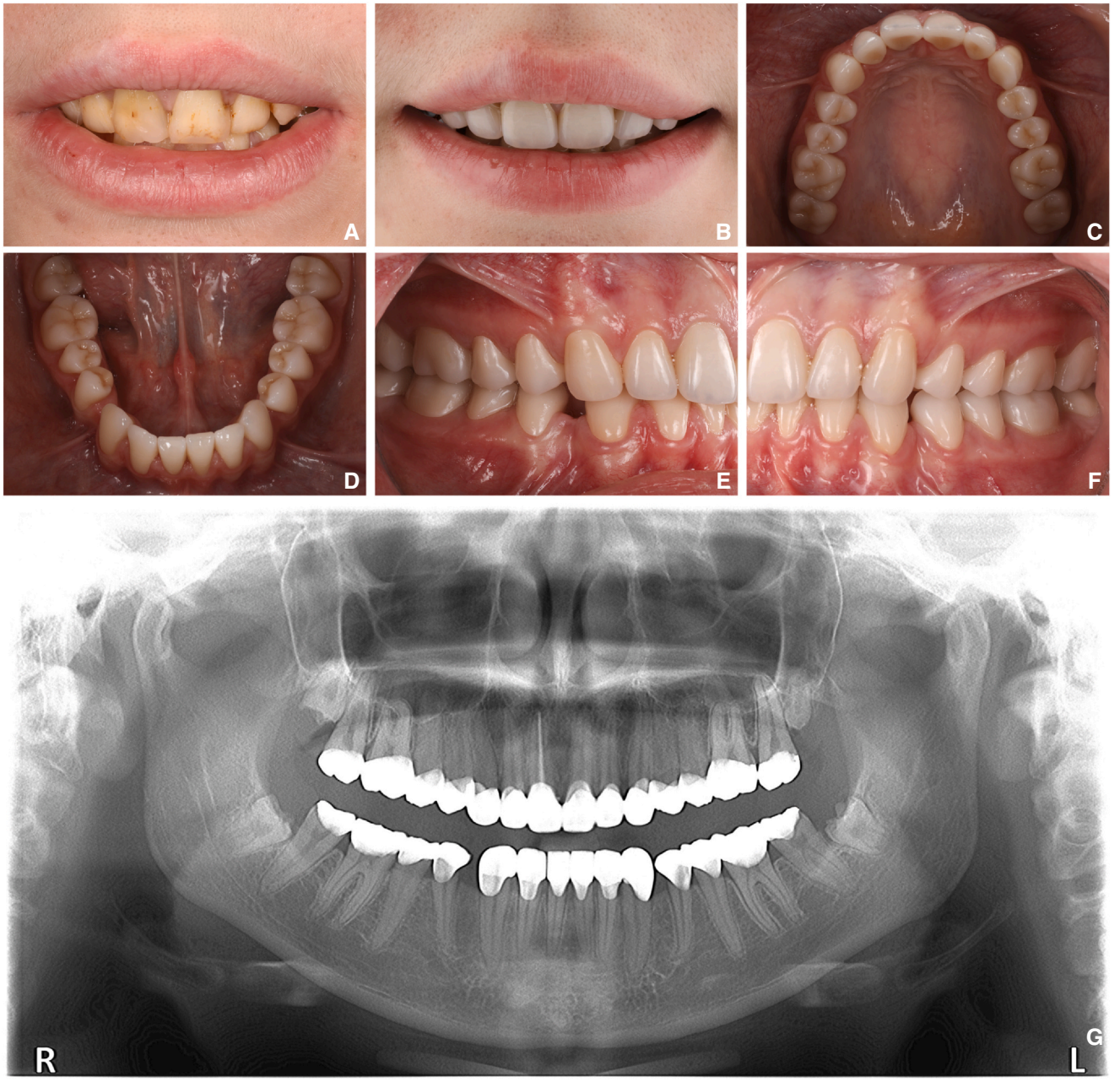

Figure 6. Clinical presentation. A, Initial extraoral view. B, Postoperative extraoral view. C, Postoperative maxillary occlusal view. D, Postoperative mandibular occlusal view. E, Postoperative lateral view F, Postoperative lateral view G, Postoperative panoramic radiograph.

be effectively remedied through the complete coverage rehabilitation of AI-affected teeth. Monolithic translucent zirconia has been reported to provide reliable single-unit restorations[28] with good esthetics and reliable long-term bonding with the APC protocol, which is based on air-borne-particle abrasion, primer application (containing 10-methacryloyloxydecyl dihydrogen phosphate), and the use of an adhesive composite resin.[29–35]

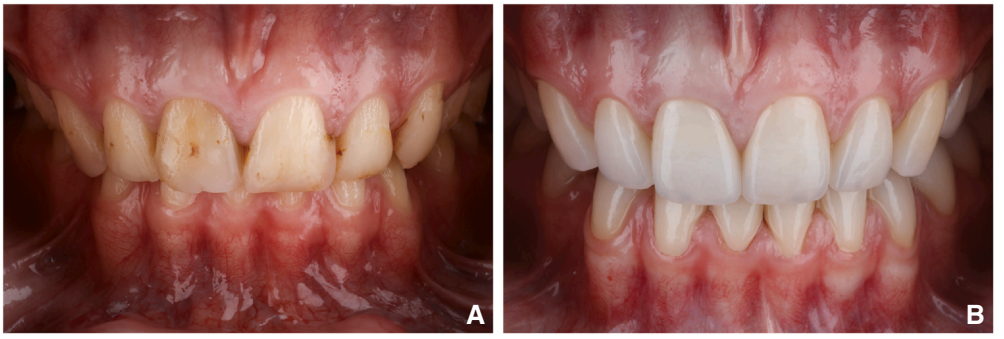

Figure 7. Treatment phases. A, Initial situation. B, Definitive restorations.

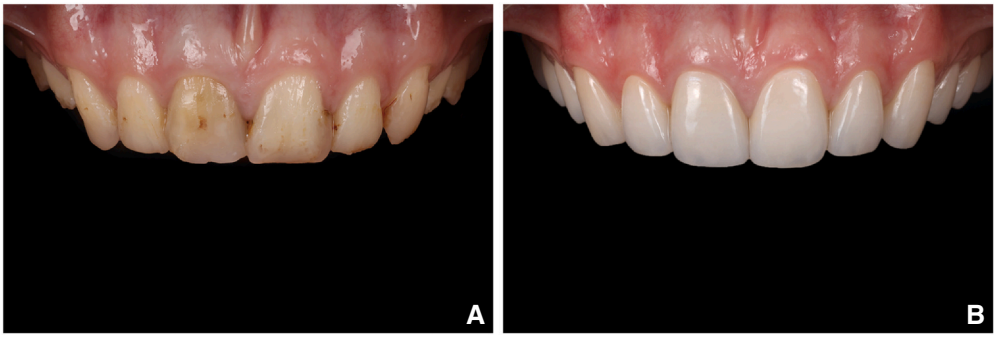

Figure 8. Maxillary frontal view. A, Initial situation. B, Definitive restorations.

SUMMARY

The digital workflow used for a woman with AI facilitated a time- and cost-efficient treatment with a predictable outcome. She adapted comfortably to her new oral situation, appreciating the convenience of digital scans over conventional impression making. A strict recall program comprising oral hygiene instructions, remotivation, wearing an occlusal guard, and fluoride application is imperative for the long-term success of people with AI. The genetic analysis confirmed and further specified the AI diagnosis, provided insight into its pathogenesis, and allowed for the assessment of recurrence risk in potential descendants, prompting earlier therapeutic intervention.

PATIENT CONSENT

Patient privacy and confidentiality were maintained in this clinical report. The patient was assured of the anonymization of data during publication. The patient provided written informed consent for the use of her images for educational and publication purposes.

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Corresponding author:
Dr Patrick Klein
Department of Prosthodontics
Medical Faculty and University Hospital Düsseldorf
Heinrich-Heine-University, Düsseldorf
Moorenstraße 5, 40225 Düsseldorf
GERMANY
Email: patrick.klein@med.uni-duesseldorf.de

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