Enamel Renal Syndrome: A Case History Report

Daiane Cristianismo Costa, DDS, MSc[1]/Maurício Rocha Dourado, DDS, MSc[2]/
Monize Ferreira Figueiredo de Carvalho, DDS, MSc[3]/Cássio Roberto Rocha dos Santos, DDS, MSc, PhD[4]/
Mauro Antônio da Cruz Batista, DDS, MSc[5]/Ana Terezinha Marques Mesquita, DDS, MSc, PhD[6]

Enamel renal syndrome (ERS) is a rare, commonly misdiagnosed condition that results in amelogenesis imperfecta and nephrocalcinosis and can lead to renal impairment in adulthood. This case history report describes a multidisciplinary dental management approach in a young adult patient with ERS. *Int J Prosthodont 2017;30:22–24. doi: 10.11607/ijp.4916*

Enamel renal syndrome (ERS) is a rare disease associated with recessive FAM20A mutations resulting in amelogenesis imperfecta (AI) and nephrocalcinosis.[1] Although presenting patients may be misdiagnosed as isolated AI, they usually show peculiar characteristics. The main features include generalized thin hypoplastic or absent enamel, flat cusps on posterior teeth, relative microdontia, spaced teeth, intrapulpal calcifications, delayed tooth eruption, impacted teeth, and gingival hyperplasia.[1,2] Nephrocalcinosis is typically observed in a renal ultrasound, although the patient may have no renal complications until late childhood or early adulthood.[3]

Depending on the severity of the disorder and the patient's age and socioeconomic status, dental management can be challenging. Moreover, robust information on such management and associated outcomes appears to be lacking. This report describes a multidisciplinary and inexpensive approach in a young adult male patient with ERS.

[1]*Clinical Dentist, Department of Dentistry, Federal University of Jequitinhonha and Mucuri Valleys (UFVJM), Diamantina, Brazil.*
[2]*PhD Candidate, Oral Pathology Section, Department of Oral Diagnosis, Piracicaba Dental School, University of Campinas (UNICAMP), Piracicaba, Brazil.*
[3]*PhD Candidate, Department of Dentistry, Federal University of Minas Gerais (UFMG), Belo Horizonte, Brazil.*
[4]*Professor, Department of Oral and Maxillofacial Surgery, Federal University of Jequitinhonha and Mucuri Valleys (UFVJM), Diamantina, Brazil.*
[5]*Professor, Department of Dentistry, Federal University of Jequitinhonha and Mucuri Valleys (UFVJM), Diamantina, Brazil.*
[6]*Professor, Oral Pathology Section, Department of Dentistry, Federal University of Jequitinhonha and Mucuri Valleys (UFVJM), Diamantina, Brazil.*

Correspondence to: *Dr Daiane Cristianismo Costa, Department of Dentistry, Federal University of Jequitinhonha and Mucuri Valleys (UFVJM), Fanor de Carvalho 75, Santa Rita, Montes Claros, MG, Brazil. Email: daiacosta02@gmail.com*

Case History Report

A 25-year-old man with reduced cognitive capacity for his age presented with a diagnosis of ERS. Extraoral examination revealed excessive lip seal associated with loss of occlusal vertical dimension. The intraoral assessment showed poor oral hygiene, retention of deciduous teeth, microdontia, spaced teeth, erupted teeth showing a yellow discoloration with thin and translucent enamel, occlusal and incisal wear, lack of permanent teeth, gingival hyperplasia, and severe irregularity of the maxillary alveolar ridge (Fig 1). Oral radiographs showed impacted permanent teeth with well-developed roots, some with pericoronal radiolucency and intrapulpal calcifications, while renal ultrasound revealed nephrolithiasis in the right kidney (Fig 2). The blood and urine testing performed (calcium, phosphate, parathormone, 25-hydroxy and 1,25-dihydroxyvitamin D, alkaline phosphatase, and creatinine) revealed no abnormal findings.

After diagnosis and consideration of diverse treatment possibilities, complete maxillary and mandibular dentures were prescribed. The first step was removal of erupted teeth because of their poor condition to support a prosthetic rehabilitation. Impacted teeth on the posterior maxilla were also removed, followed by osteoplasty and resection of the osseous structure to achieve acceptable gingival contour. All teeth and tissue removed were submitted for histopathologic analysis to confirm the diagnosis. The surgical interventions resulted in right maxillary sulcus depth loss, and a sulcoplasty was carried out to ensure a more stable prosthesis design. The single erupted mandibular molar was removed because of its poor stability and capacity to support the mandibular prosthesis. Conventional complete denture prosthetic treatment was started approximately 2 months postsurgery (Fig 3). The completed prostheses addressed the patient's functional and esthetic complaints.

Fig 1 **(a)** Extraoral view of a 25-year-old male ERS patient showing arch discrepancy and modification of occlusal vertical dimension as represented by excessive lip seal. **(b, c)** Intraoral views show missing teeth, microdontia, erupted teeth showing a yellow discoloration with thin and translucent enamel, occlusal and incisal wear, severe irregularity of the maxillary alveolar ridge, absence of interproximal contacts, absence of permanent teeth, retention of deciduous teeth, and gingival hyperplasia.

Fig 2 **(a)** Panoramic radiograph showing the presence of impacted permanent teeth with unusual location (L), intrapulpal calcifications in the molar crowns (IC), absence of the regular contrast between dentin and enamel, pericoronal hyperplastic follicles (PHC), and irregular alveolar ridge contour. Renal ultrasound revealed nephrolithiasis in the right kidney *(arrow)* **(b)**, but not in the left one **(c)**.

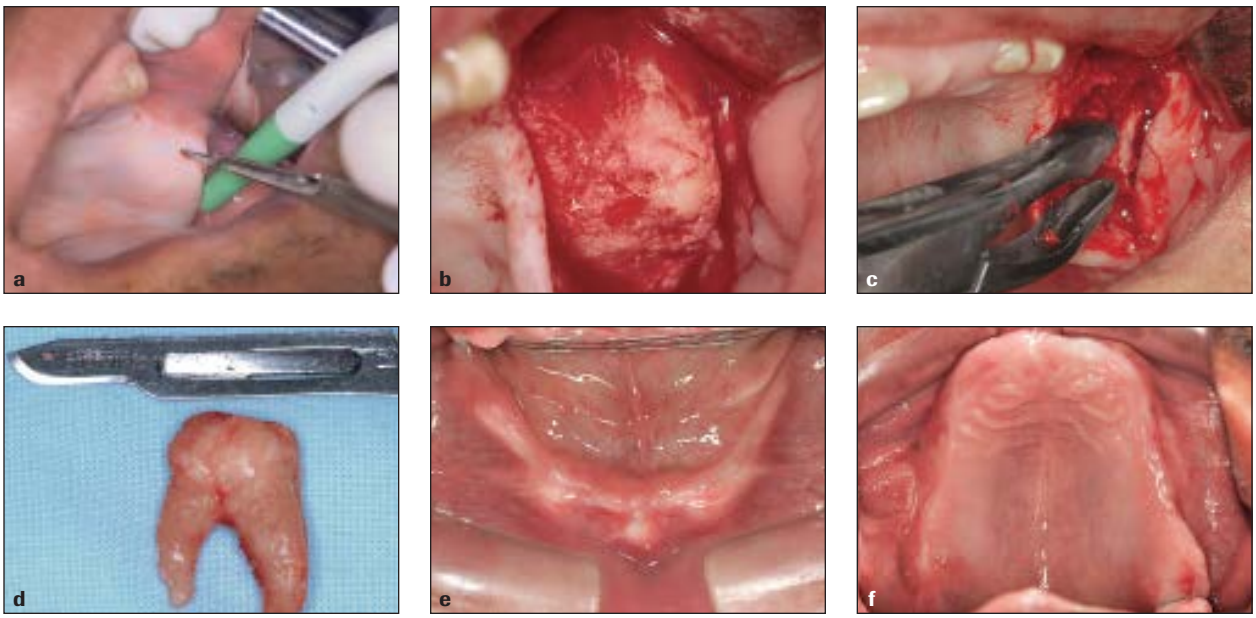

Fig 3 **(a–d)** Surgical procedure for extraction of the impacted maxillary left first molar and osteoplasty. **(e)** Mandibular and **(f)** maxillary postsurgical views at 9 months.

The number of impacted teeth, their ectopic location and poor structure, the presence of pericoronal radiolucencies, malocclusion, unknown bone quality, and the age and socioeconomic status of the patient presented challenging treatment considerations, such as expenses incurred for orthodontic traction or an implant-supported prosthesis. Direct composites or indirect restorations were precluded, since enamel

hardness and bond strength to permanent teeth affected by hypocalcified amelogenesis imperfecta are compromised compared with sound teeth.[4] Hence, the final choice was preprosthetic surgery to facilitate the fabrication and use of complete dentures. The clinical choice was for a simple, relatively inexpensive, multidisciplinary approach, which contributed to a successful outcome that improved the quality of life of the patient.

Conclusions

Since the oral features are the first to emerge—and the main cause for which patients seek professional help—dentists play a key role in the diagnosis and referral of ERS patients to nephrologists and geneticists.[2] Patients who have undergone this type of oral rehabilitation must be evaluated at regular recall appointments. The retained impacted teeth must be monitored since tooth eruption may occur in due course. Such changes will require modifications in the overlying prosthesis.

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Literature Abstract

Changes in Peri-Implant Bone Level and Effect of Potential Influential Factors on Dental Implants in Irradiated and Nonirradiated Patients Following Multimodal Therapy Due to Head and Neck Cancer: A Retrospective Study

This retrospective study evaluated changes in the marginal bone level of dental implants in irradiated patients (test group) and nonirradiated patients (control group) in an attempt to identify possible influential factors on implant success. In total, 36 patients (7 women and 29 men) diagnosed with squamous cell carcinoma participated in the study, with 194 implants (73 in the maxilla and 121 in the mandible) being placed. The mean age of the patients was 65.8 years (range 39–90 years). In all patients, a squamous cell carcinoma in the floor of the mouth involving the mandible or tongue was surgically removed. In the irradiated group, 17 patients underwent adjuvant radiochemotherapy that was completed a minimum of 6 months before implant placement. In the nonirradiated group, 19 patients underwent tumor resections and reconstructions using varying techniques before implant placement. Irradiated patients received an antibiotic regimen (clindamycin 600 mg orally 3 times daily, 3 days pre- and postoperatively). Nonirradiated patients were treated perioperatively with a single intravenous shot of antibiotics (clindamycin 600 mg). In both patient groups, the results showed that mean peri-implant bone loss (1 mm mesial and 0.9 mm distal) after 1 year had progressed after 3 years (1.4 mm mesial and 1.3 mm distal). Four implants were lost, with an overall success rate of 98.4% (maxilla, 100%; mandible, 96.7%). There was no statistically significant difference in changes in bone level related to age, sex, prosthetic superstructure, or whether an augmentation procedure was performed. However, radiation therapy was found to have a statistically significant effect on crestal bone loss ($P < .05$). The mean amount of crestal bone change in irradiated patients was twice as high in the mandible (2 mm mesial and 1.8 mm distal) compared to that in nonirradiated patients (0.8 mm mesial and 0.9 mm distal) at the 36-month follow-up. A similar trend was observed in the maxilla. Owing to the effect of radiation therapy, patients receiving the radiation dose in the region of the mandible may explain the observed differences in implant bone loss between the two groups.

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Reprints: Dr Nahles, Department of Maxillofacial Surgery, Navigation and Robotics, CVK, Charite, Augustenburger Platz 1, Berlin 13353, Germany; Email: Susanne.nahles@charite.de —*Loke Weiqiang, Singapore*