

Treatment of enamel hypoplasia in a patient with Usher syndrome

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Usher syndrome (USH) is a group of autosomal recessive diseases that are characterized by retinitis pigmentosa with sensorineural hearing loss. It is the leading cause of deaf-blindness, and its prevalence varies from 3.0 in 100,000 people in the Scandinavian population to 4.4 in 100,000 people in the U.S. population.¹

There are three types of USH. USH type I (USH1) is characterized by congenital, severe or profound hearing loss, vestibular dysfunction and retinitis pigmentosa that initiates during childhood. USH type II (USH2) is characterized by congenital, moderate or profound hearing loss and normal vestibular function, as well as development of retinitis pigmentosa after childhood. USH type III (USH3) is characterized by progressive hearing loss and a variable age of onset of retinal degeneration.² There is extensive genetic and clinical heterogeneity in patients with USH.³ Seven genetic loci have been identified for analyzing USH1, three genetic loci have been identified for USH2 and one genetic locus has been identified for USH3.⁴ Amelogenesis imperfecta is a clinically and

ABSTRACT

Background. Usher syndrome (USH) is a group of autosomal recessive diseases characterized by the association of retinitis pigmentosa with sensorineural hearing loss. There are three types of USH. In addition, in people with USH and hypoplasia, the thickness of the enamel is reduced.

Case Description. The authors describe a case of a patient with USH type II associated with severe enamel hypoplasia and multiple unerupted teeth. The authors placed direct composite crowns and extracted severely affected and impacted molars.

Clinical Implications. There is little information available on the oral pathologies of USH. Because the authors did not know how the patient's condition would progress and the patient still was growing, the authors treated the patient conservatively by placing direct composite crowns. The treatment has met both esthetic and functional expectations for 10 years.

Key Words. Composites; dental veneers; permanent dental restorations; amelogenesis imperfecta; Usher syndrome.

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genetically heterogeneous disorder that affects the enamel of the primary and permanent teeth. Cases of amelogenesis imperfecta can be classified into three groups: hypoplasia, hypocalcification and hypomaturation. The prevalence of amelogenesis imperfecta varies according to the population, ranging from one in 700 people in a study on the Swedish population to one in 14,000 in a study of the U.S. population.⁵ Hypoplasia is characterized by reduced enamel thickness of varying degrees, as well as pits and other irregularities, although the hardness and transparency of the enamel remain intact.⁵ We found one study in the literature whose authors described a patient who had USH and enamel hypoplasia.³

CASE REPORT

In 1994, one of the authors (V.A.P.) examined a 16-year-old boy in his private clinic (Clinica Dr. Victor Alonso de la Peña, Santiago de Compostela, A Coruña, Spain). The patient had received diagnoses of USH2, moderate sensorineural hypoacusis, normal vestibular function and developing retinitis pigmentosa. We found that the patient had nine primary teeth (teeth nos. A, B, C, H, J, K, M, R and T). All of his permanent incisors, as well as the maxillary left first premolar and both mandibular first premolars, had erupted. We observed generalized hypoplasia both on the primary teeth and on the permanent teeth (except for the primary canines), with thin enamel and almost no enamel at the occlusal contact area on all of the teeth (Figure 1). We did not see any alterations in the oral mucosa or pathology of the temporomandibular joint.

Radiographs showed that the canines and permanent premolars (except for tooth no. 29) were well positioned but had not erupted, and the molars were retained. Both the molars and premolars had alterations of the crown that were compatible with severe hypoplasia; the enamel could not be observed.

The patient came to the clinic for esthetic treatment to hide the hypoplasia of his anterior teeth. We created full crowns by using resin-based composite and strip crowns (Frasaco, Franz Sachs, Tettnang, Germany) for the matrix. All of the treatment was carried out in the dental clinic, and the patient received local anesthetic when necessary.

We selected a suitable strip crown and removed restorations from some of the maxillary central incisors. We used phosphoric acid to etch the teeth and then applied adhesive (Prime & Bond 2.0, Dentsply De Trey, Konstanz, Ger-

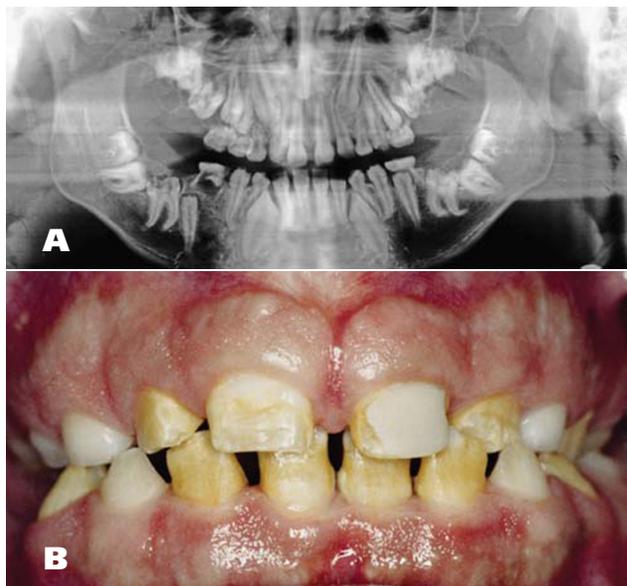


Figure 1. A. A panoramic radiograph from 1994 showing the primary and permanent canines, premolars and molars, all of which had reduced enamel thickness. B. A clinical photograph from 1994 showing severe hypoplasia on all of the teeth and that tooth no. 9 had been restored.



Figure 2. A clinical photograph from 1994 showing restored teeth nos. 8 and 9. The fit and shape of the strip crown can be seen on tooth no. 9.

many) to all remaining exposed tooth tissue. We filled the crowns with composite (Spectrum TPH composite, Dentsply De Trey) and placed them on the teeth. We removed excess composite and polymerized the whole restoration. We used an explorer to remove the strip crown, and then we polished and esthetically recontoured all of the restoration.

We used this technique to treat the maxillary and mandibular central incisors (Figure 2). After we saw how positive the results were, we decided to use this technique to treat teeth nos. 5, 12, 27, 28 and 29.

Since 1994, the patient received additional

ABBREVIATION KEY. USH: Usher syndrome. USH1: Usher syndrome type I. USH2: Usher syndrome type II. USH3: Usher syndrome type III.

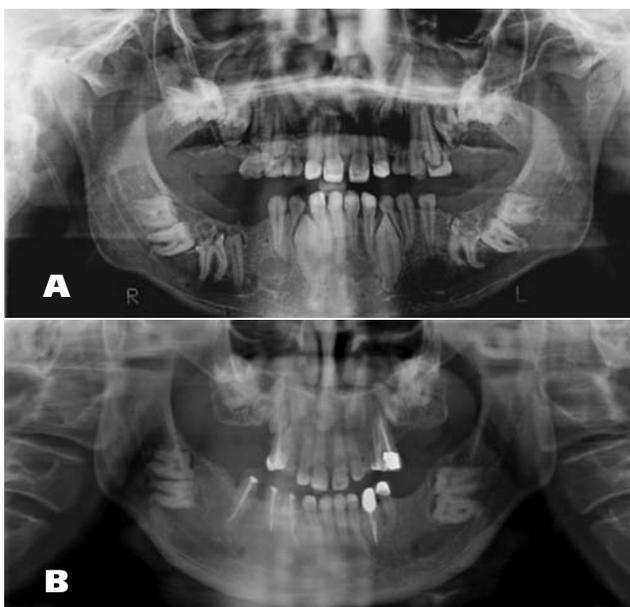


Figure 3. **A.** A panoramic radiograph obtained in 1996 shows that strip crowns were placed on the permanent incisors. **B.** A panoramic radiograph obtained in 2009 shows the treatment that had been performed between 1996 and 2009 (endodontic therapy in teeth nos. 5, 12, 13, 22, 27, 28 and 29; composite crowns placed on teeth nos. 5, 12, 27, 28 and 29; porcelain-fused-to-metal crowns placed on teeth nos. 13, 21 and 22).



Figure 4. Clinical photograph of the composite crowns placed on all of the permanent teeth after 15 years.

dental treatment. We extracted teeth nos. 19 and 30 because the patient had mandibular pain, and radiographs showed that the crowns of these unerupted teeth were almost completely missing. Tooth no. 20 had severe periapical pathology, so we extracted it. Because of mobility, we also extracted primary teeth (nos. A, B, J, M and R) that still were present. After teeth nos. 5, 12, 13, 22, 27, 28 and 29 erupted, pulp pathologies appeared in them, and we performed endodontic procedures on these teeth. We performed an apicoectomy on tooth no. 28. We placed porcelain-fused-to-metal crowns on teeth nos. 13, 21 and 22.

The patient received no further dental treatment after 2007. Radiographs from 1996 and 2009 and the clinical progress of the restora-

tions are shown in Figures 3 and 4, respectively. The second and third mandibular molars remained impacted. In the maxilla, the molars, canines and right second premolar had not erupted. As the patient was asymptomatic, no treatment was required for the unerupted teeth.

DISCUSSION

Many patients with hearing impairments, visual impairments or both have reported that they are not informed about their disease, treatment options and prognosis properly.⁶ A variety of factors can affect these patients' impairments, making it necessary for clinicians to treat patients in an individualized manner.⁶ These factors include the patient's age, how long the patient has had the impairment, the severity of the impairment (slight, moderate, serious, profound), any associated problems (learning difficulties), communication skills and preferences, family issues (parental hearing impairments and attitude) and type of education (oral, bilingual, integrated, special). The minor communication problems caused by our patient's progressive hearing loss and progressive blindness were resolved by the patient's guardian's being present during the treatment to help aid communication between the patient and the clinician.

There is little information regarding the oral pathologies of USH. The authors of only one article have associated enamel hypoplasia with USH.³ The authors of two other articles associate hypoacusis with enamel hypoplasia and blindness, but they made no specific reference to USH.^{7,8} An eight-year-old patient with USH1 and defects in the enamel is mentioned in another article, but its authors did not consider hypoplasia, because only the occlusal surfaces of the affected teeth had hypoplastic enamel.⁹

In addition, hypoplasia has been associated with Seckel syndrome¹⁰ or tricho-dento-osseous syndrome.¹¹ Taurodontism was associated with both of these syndromes.

Amelogenesis imperfecta has been treated with indirect restorations such as veneers or porcelain crowns,¹²⁻¹⁸ although the investigators of some studies used composite veneers, which is a more conservative treatment.¹⁹⁻²²

Because we did not know how the patient's condition would progress and the patient still was growing, we chose to treat the patient conservatively by placing direct composite crowns so as not to eliminate any dental structure and not to exclude other types of treatment. Another option we used for teeth nos. 13, 21 and 22 was to place porcelain crowns on all the permanent teeth.

As a result of the extractions and the impaction of the permanent molars, the patient could have had limited masticatory function. To avoid this outcome, the patient and his parents decided against his undergoing the extractions and prolonged restorative treatment.

CONCLUSION

Across 10 years, the treatment met our and the patient's esthetic and functional expectations. ■

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