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Enamel defects and ectopic eruption in a child with Usher syndrome and a cochlear implant

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Background. Usher syndrome is a genetic disorder consisting of progressive loss of vision and hearing.

Case Report. The paper describes an 8-year-old girl with Usher syndrome type I who presented with generalized defects of the permanent dentition and ectopic eruption of the right maxillary first permanent molar. A cochlear implant had been fitted for her hearing loss, and the report reviews the implications of this device for dental treatment. The impacted first permanent molar was encouraged to erupt into the correct position by shaving the distal surface of the second primary molar.

Conclusion. This is the first report to describe in detail an association between Usher syndrome and enamel defects.

Introduction

The Usher syndromes are a group of genetically inherited disorders. They are characterized by impaired ability of auditory nerves to transmit sensory input resulting in progressive (sensorineural) hearing loss and retinitis pigmentosa with progressive loss of vision[1]. Although very rare (a frequency of 4.4 in 100 000 has been suggested[2]), it has been estimated that half of all registered blind and deaf people in the USA have this condition[3]. Four types have been described, based on age of diagnosis and severity of symptoms, type I being the most severe. At least 10 genetic loci have been identified, type I being most commonly located to chromosome 11q13.5[1]. In recent years, hearing loss has been successfully treated with cochlear implants[4,5]. This device consists of two components – external and internal. The external component is a removable headpiece transmitter that fits just behind the ear. The headpiece converts sound to electromagnetic energy, which is transmitted to a surgically implanted receiver. The receiver, in turn, transmits directly to the cochlear, resulting in stimulation of the auditory nerve.

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There is only one report in the literature of a relationship between confirmed Usher syndrome and enamel defects[6]. This study reported the presence of enamel hypoplasia in two families with Usher syndrome. The teeth were described as small, brownish or peg-like, and were softer and more susceptible to dental caries.

Ectopic eruption of maxillary first permanent molars is a relatively common condition, although it has not been previously reported in relation to Usher syndrome. Most studies report a prevalence of 2–6%[7,8]. There is no difference in race or gender, and it occurs bilaterally as often as it occurs unilaterally. Although two-thirds of these teeth self-correct[9], the rest remain impacted, which may result in early loss of the primary molar and severe space loss as the permanent molar continues to drift mesially. The aetiology of the condition is unclear, with size[10], calcification time[8], angle of eruption[11] and length of maxilla[8] all being implicated.

Case report

The child was 8 years old when she was referred by her general dental practitioner (GDP). The letter indicated significant enamel problems that were attributed to amelogenesis imperfecta. It was also noted that she was registered deaf and blind, and had a cochlear implant that her GDP felt would prohibit panoral radiography.

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Medical history indicated that she had Usher syndrome, type I (OMIM #276900).

The child was in the mixed dentition stage. Two of the primary molars (64 and 75) had caries and two had small restorations (55 and 54). Numbers 84 and 85 had been extracted. Number 65 had exfoliated and there was marked mesial drift of 26. Number 16 was ectopic and impacted against the distal surface of 55 (Figs 1 & 2).

All four first permanent molars had enamel defects. These were well demarcated, brown/orange in colour and confined to the occlusal surfaces. Cuspal anatomy initially seemed normal, although the lower molars quickly developed occlusal lesions.

The upper permanent incisors were also affected. Numbers 12 and 22 had saucer-shaped deficiencies on the labial and palatal surfaces, with 22 demonstrating the brown/orange discoloration seen on the molars (Fig. 3). Close inspection of 11 and 21 also revealed enamel deficiencies of the labial surfaces. The lower permanent incisors seemed normal. Orthopantomograms and bitewing radiographs taken at initial assessment showed that all permanent teeth except for the third molars were present and at the correct stage of development. The whole of the permanent dentition – including the uneruptерd permanent teeth – showed evidence of reduced and irregular enamel. Number 55 had significant resorption associated with the impacted 16 (Fig. 2).

The child was monitored and treated over a period of 18 months. During this time, a number of the premolars erupted, all with significantly hypoplastic occlusal surfaces. These defects were also well-demarcated, had a uniform brown/orange colour, and extended onto the buccal and palatal surfaces of the cusps.

Included in the treatment was distal shave of 55, which was carried out early and again a few months later. Four months after this, the

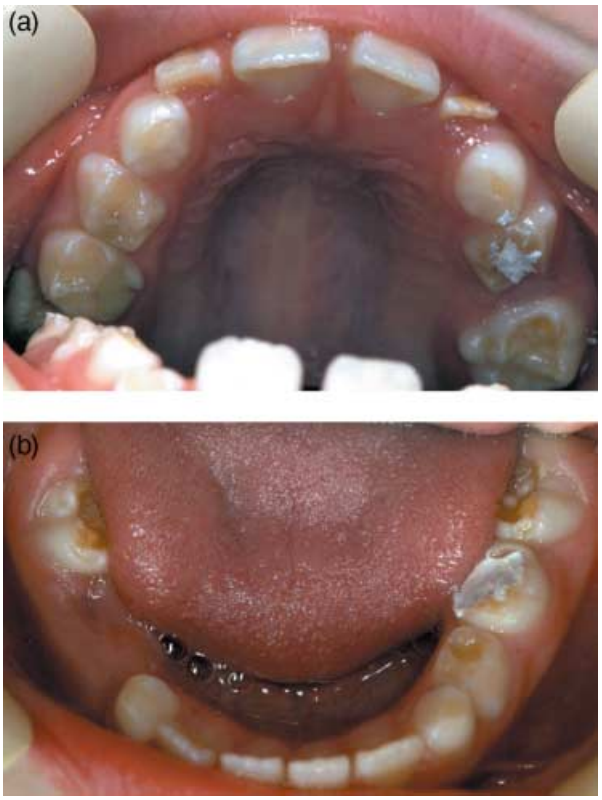

Fig. 1. (a) Intraoral occlusal view on presentation. Note the impacted 16 and the significant mesial drift of 26. (b) Lower occlusal view on presentation. Enamel defects are present on 36 and 46.

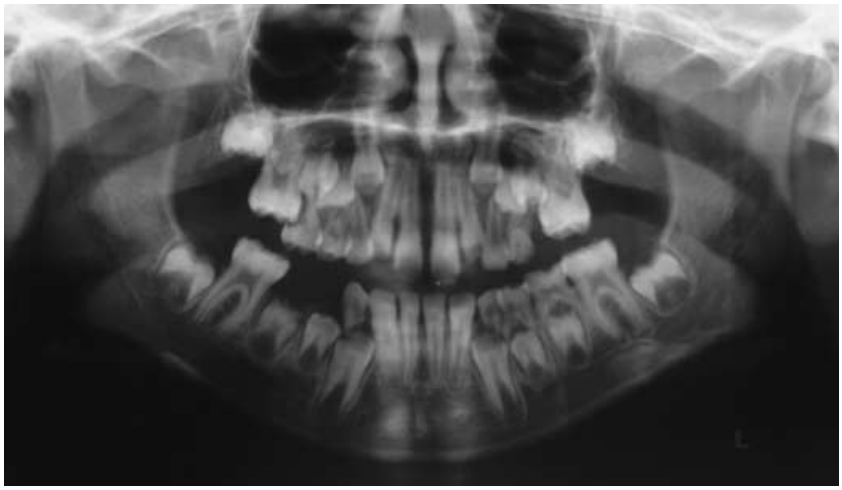

Fig. 2. Orthopantomogram on presentation. Number 16 is ectopic. The enamel of the unerupted permanent teeth is reduced and irregular.

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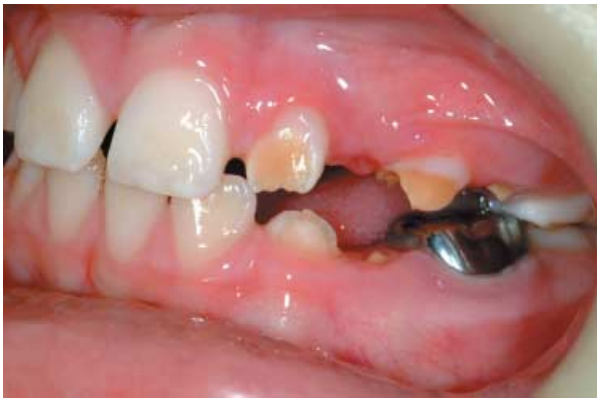

Fig. 3. Lateral view showing enamel defects of 22 and 24.

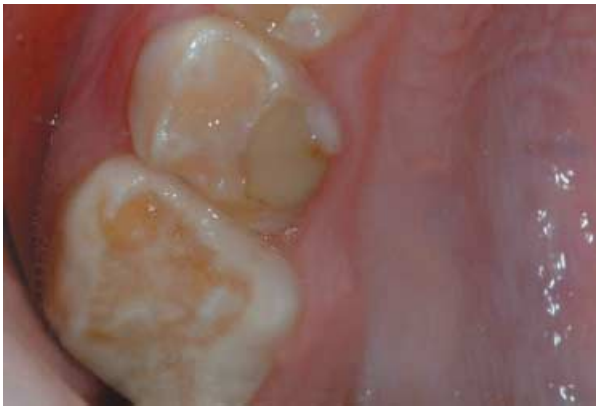

Fig. 4. Upper view showing 16 erupted into the occlusal plane following the distal shave of 55.

16 had erupted into the occlusal plane (Fig. 4). Number 55 exfoliated towards the end of the treatment period, allowing the 15 to erupt into the 55 space (Fig. 5).

Discussion

The child had been diagnosed with type I Usher syndrome, and at 6 years of age, she had received a cochlear implant. The presence of the implant had caused S.B.'s GDP to be concerned about using panoral radiography. Recent research, however, has concluded that radiography is safe in patients with cochlear implants[12]. This has been given official support by the Chief Medical Officer[13] in advice given to accident and emergency departments on the safety of patients with cochlear implants. Although this report advised that radiography was safe, it did identify that magnetic resonance imaging scans, surgical diathermy and electrical pulp testing could damage the implant, and therefore, should be avoided in these patients.

Initial clinical examination indicated that she might have had a type of molar incisor hypocalcification. First permanent molars and some of the incisors were affected and the primary dentition seemed to have been spared[14]. Radiographic examination, however, showed that the enamel of all the permanent teeth was thin and irregular. It was, therefore, concluded that the underlying defect was hypoplastic in nature, and this was confirmed as the premolars erupted.

According to the generally accepted definition of Witkop and Rao[15], it would be incorrect to diagnose the child with amelogenesis imperfecta. This is because their definition precludes enamel defects occurring in the presence of systemic disease. Recently, however, it has been argued that the definition of amelogenesis imperfecta should be changed to incorporate the statement that the enamel defects may '… be associated with morphologic or biochemical changes elsewhere in the body'[16].

The type of defect itself was unusual in that it seemed to affect only the occlusal surfaces of the teeth. This was noticeable on the first permanent molars, and to an extent, on the maxillary incisors, which were affected to varying degrees on the labial and palatal surfaces. It became much more obvious on the eruption of 24 and 25, which had severely hypoplastic occlusal surfaces surrounded by a collar of apparently normal

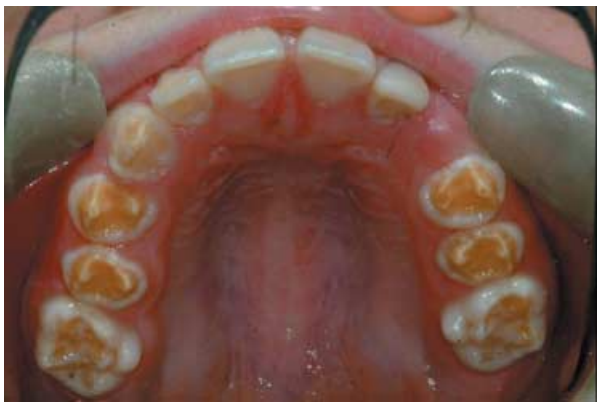

Fig. 5. Upper occlusal view following the eruption of the premolars. Hypoplastic occlusal surfaces are surrounded by a collar of apparently normal enamel.

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enamel. The mandibular permanent incisors were spared.

Although there is only one report in the literature relating Usher syndrome to enamel defects[6], two other studies have reported on the combination of hearing, visual and enamel defects without referring specifically to Usher syndrome. Bateman *et al.*[17] described a 15-year-old with retinitis pigmentosa, sensorineural hearing loss and enamel defects. They did not specifically identify this patient as having Usher syndrome, however. In 1998, Innes *et al.*[18] presented a family with a similar combination of hearing, visual and dental defects, and proposed that this was actually a new syndrome.

Ectopic eruption of first permanent molars is a common condition with estimated prevalence of 2–6%[11]. Given this prevalence rate, it is not possible, on the basis of this single case, to assume an association between the condition and Usher syndrome. Indeed, there are no other reports in the literature associating ectopic eruption with any other specific syndrome, although its association with cleft lip and palate, and therefore, by extension, with specific clefting syndromes, has been well documented[11]. Numerous treatments for ectopic permanent molars have been described in the literature. These include using stainless-steel crowns on the primary molars[19,20], separating elastics[21], brass ligature wire[22], push coil[23] and removable appliances[24]. Curiously, no description has been found of the approach taken in her case. This simple technique, which can be carried out without local anaesthetic, involves shaving the distal aspect of the primary molar to expose the mesial marginal ridge of the ectopic permanent molar. It simpler than most other techniques and has the added advantage that it does not require several visits. By encouraging the eruption of 16 into its correct position, and at the same time conserving 55, space was maintained for the eruption of 15.

What this paper adds

- No previous paper has given a detailed description of the relationship between enamel defects and Usher syndrome.
- This paper describes a previously unpublished method for the treatment of ectopic maxillarly first permanent molars.

Why this paper is important to paediatric dentists

- Paediatric dentists should be aware of the possible dental implications for children with Ushers syndrome.
- Specific investigations are contra indicated in children fitted with cochlear implants.
- Paediatric dentists should be aware of an alternative treatment for ectopic maxillary first permanent molars.

Conclusion

This paper has reported on a child with Usher syndrome who had been treated with surgical insertion of a cochlear implant and described the possible dental implications. The paediatric dentist should be aware of the possible association between Usher syndrome and generalized enamel defects. A simple method for the treatment of ectopic upper first permanent molars has been described.

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