The dental management of a patient with Nager syndrome: a case report

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Abstract

Patients with Nager syndrome have extensive craniofacial involvement resulting in restricted mouth opening. This case report is of a child with Nager syndrome who also had extensive dental caries. This patient required dental treatment to be carried out under general anaesthesia, due to her restricted mouth opening. Preventive advice is of vital importance.

Key words: Nager syndrome, dental caries, prevention

Introduction

Nager syndrome (NS), also termed Nager acrofacial dysostosis or preaxial acrofacial dysostosis, is a rare disorder which is characterised by aberration in the development of the 1st and 2nd brachial arches and the limb buds (Kavadia et al., 2004). It manifests as a mandibulofacial dysostosis with associated hand defects, specifically, abnormalities of the radial aspects of the forearms and hands (Daziger et al., 1999).

NAFD was first reported in 1908 by Slingenberg, but recognised as a specific entity by Nager and de Reynier in 1948 (Kavadia et al., 2004). The clinical features of NS include downward slanting palpebral fissures, mandibular hypoplasia, malar hypoplasia, a high nasal bridge, atretic external auditory canals, colobomas of the lower eyelids, micrognathia and a congenital absence of the soft palate. There is potential for significant difficulties with airway management and postoperative airway obstruction (Groeper et al., 2002).

Preaxial limb malformations include hypoplasia of the radius and shortened humeral bones, absent or hypoplastic thumbs (Vargervik, 1998), and occasional duplication of thumbs (Kavadia et al., 2004).

Associated abnormalities may include short stature, genitourinary malformations (Opitz, 2003) and congenital cardiac defects. Functional impairments resulting from NS include respiration and feeding problems, and varying degree of hearing loss. The speech is often affected, being hypernasal, as velopharyngeal closure cannot be achieved due to a hypoplastic or absent soft palate (Vargervik, 1998).

To date, less than 100 cases have been reported in the medical literature and cases appear to be sporadic occurrences. The mode of inheritance is unclear, although both autosomal dominant (Aylsworth, 1991) and recessive modes (Chmeke, 1988) have been suggested. Nager syndrome is of interest to the dental team due to the extensive craniofacial involvement which occurs with this syndrome and the resulting restricted mouth opening. This Case Report is of a child with NS, talipes and extensive dental caries who required dental treatment under general anaesthesia.

Case Report

A 15-month-old girl AB was referred to the multidisciplinary specialist cleft clinic at the Maxillofacial unit in Morriston hospital, Swansea, for management of her mandibulofacial defects. In addition, her parents reported that she had been suffering with pain from her teeth. The family history was essentially unremarkable although AB’s father had been born with talipes. Neither parent showed any dysmorphic features.

AB was born with significant multiple physical deformities. The head was unusual in shape, with a large cranium and she had a significantly small midface. AB exhibited micrognathia, downward slanting orbits, small external ears and skin tags in front of her ears (Figure 1 and 2). A CT scan (Figure 3) further revealed stunted condyles, coronoid processes and hypoplastic rami. The zygomatic arches were absent and the orbits appeared relatively large. There was a mild deficiency of the lower eyelids which constituted colobomas.

Examination of the upper limbs revealed very slender thumbs on both hands and limitation of elbow movement. Her toes were short, with webbing between the 2nd and 3rd toes. In addition, AB was born with talipes for which surgical correction had already been planned. A routine neonatal screening had detected congenital hypothyroidism for which AB had been placed on thyroxine. Her vision...
and hearing were normal. Due to the characteristic facies, Treacher Collins syndrome was initially suspected. The diagnosis was later revised in view of additional findings involving the limbs to Nager syndrome. AB had restriction of her mouth opening of 18mm (Figure 4), intraoral examination was therefore difficult. A brief examination revealed multiple carious primary teeth (Figure 5).

An orthopantomograph radiograph revealed the presence of all developing permanent teeth and confirmed the carious primary teeth. The patient required extensive multiple surgical procedures to improve her functional, aesthetic and developmental potential and continuing care is managed by a multidisciplinary team consisting of oral and maxillofacial surgeons, otolaryngologists, paediatric dentists, geneticists, psychologists, and speech and language therapists. Surgical treatment will involve correction of her maxillofacial deformity including the absent zygomatic arches and lower eyelid deficiencies, and possible use of distraction osteogenesis to correct her micrognathia.

Treating the dental caries was given priority, to render the patient free of pain and infection. On account of the restricted mouth opening and limited level of cooperation, detailed examination and dental treatment had to be carried out under general anaesthesia with parental consent. This proved to be challenging on account of limited mouth opening. Relevant findings of note were a significantly hypoplastic soft palate, and extensive caries involving several primary teeth (Figure 5).

The primary molars and lower left primary canine were found to be unrestorable and were extracted. The lower right primary canine was restored with a composite restoration (Figure 6). The patient made an uneventful recovery.

Maintaining good oral hygiene will be challenging due to restricted mouth opening and the preventive advice given had to be practical. Prevention was planned at home and at the chairside. AB’s parents were given detailed diet advice and educated on effects of frequency and consistency or refined carbohydrate consumption. Oral hygiene instructions were provided. AB’s parents were asked to help her in brushing her teeth using a toothbrush with a small head. A toothpaste with high concentration of fluoride was recommended. Fluoride varnish (22600 ppm F) will be applied topically on a four-monthly basis. Preventive advice provided will be regularly reinforced and arrangements made for regular dental review.

The patient remains under regular review on the multidisciplinary clinic where a definitive long term management plan will be made for correction of her maxillofacial deformity.
Discussion

Preaxial limb abnormalities that characterise NS are diagnostic and differentiate this syndrome from Treacher Collins. In addition, NS has a more extreme degree of mandibular hypoplasia and a higher incidence of lip and palate anomalies (Groeper et al., 2002). At initial presentation Treacher Collins syndrome was suspected in our patient. The diagnosis was finally established as NS based on the limb abnormalities and associated craniofacial anomalies.

Mandibular malformations and missing joint structures have contributed to extreme restriction in jaw movement, this together with coexisting limb abnormalities make maintaining satisfactory oral hygiene difficult. AB was not registered with a dentist and unfortunately so far had not received any preventive advice regarding her dental health. Prevention is important for all children and adults but there are certain circumstances which are indicative of an increased risk of dental disease and its consequences and this includes medically compromised children (Shaw, 1997).

The patient’s restricted mouth opening also made dental examination and treatment extremely difficult, necessitating treatment under general anaesthetic. Providing general anaesthesia can be challenging in NS. The combination of maxillomandibular hypoplasia with micrognathia, retroplaced tongue, and strabismus set the stage for severe airway obstruction (Friedman et al., 1996). Several of the primary defects that are characteristic of the syndrome, such as mandibular hypoplasia, as well as associated defects of other organ systems, may impact on the perioperative anaesthetic care of these patients. This is further compounded by the fact that often the affected children have associated verbal and hearing limitation due to involvement of the 1st and 2nd brachial arches, leading to possible problems in communication with the Surgical and Anaesthetic team (Groeper et al., 2002).
Every effort should be made to minimise the number of avoidable general anaesthetics including those for dental treatment. Failure to adopt preventive counselling may result in repeat general anaesthetics, which is undesirable in terms of morbidity and potential mortality, behavioural/emotional effects on the child as well as the cost (Davies et al., 2008). Emphasis should be made therefore on the prevention of dental disease. Patients diagnosed with NS should be referred to a specialist paediatric dental service for comprehensive oral hygiene instruction, dietary advice and fluoride prevention.

Conclusion

Nager syndrome is a rare but debilitating craniofacial syndrome which results in restricted mouth opening and potential difficulties with airway management during general anaesthesia. It is important that these patients receive comprehensive prevention and regular dental review in an effort to improve their oral health and therefore avoid when possible, the need for dental treatment under general anaesthesia.

References


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