Nager acrofacial dysostosis: a rare genetic disorder causing bilateral temperomandibular joint ankylosis in a 10-year-old girl

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DESCRIPTION

A 10-year-old girl presented to the oral medicine department, with restricted mouth opening since childhood. Her family history revealed that she was the third child born of a consanguineous marriage. She was born after an unremarkable pregnancy, with a birth weight of 2.3 kg. Physical examination revealed downsloping palpebral fissures, microretrognathia, malar hypoplasia, a sloping forehead, high nasal bridge with an upturned nasal tip and short philtrum (figure 1). She was also noted to have short forearms, proximal radioulnar synostosis (figure 2), hypoplastic thumbs and relatively short overlapping toes (figure 3A, B). Oral examination revealed extremely poor oral hygiene with a narrow V-shaped short palate; the mouth opening was 15 mm.

Lateral cephalogram showed an underdeveloped mandible with an extreme posterior inclination of the mandibular base and also complete obliteration of the temporomandibular joint (TMJ) space (figure 4). Panoramic radiographic examination showed reduced vertical height of ramus, and multiple impacted teeth in the maxilla and mandible, with complete obliteration of right and left TMJ space (figure 5). Hearing assessment by brain stem-evoked response audiometry showed a bilateral profound hearing loss. In this case, genetic analysis was not performed due to financial constraint. From the girl's overall clinical and radiographic features, she was diagnosed as having Nager acrofacial dysostosis. The patient's mother gave full informed written consent to this report being published.

Nager syndrome was first recognised by Nager and de Reynier in 1948. They used the term acrofacial dysostosis to describe their patient. The syndrome (preaxial acrofacial dysostosis) is due to aberrations in development of the first and second branchial arches and limb buds.¹

Nager acrofacial dysostosis is characterised by a variety of craniofacial features including malar hypoplasia, cleft palate and micrognathia. The limb anomalies associated with Nager acrofacial dysostosis are a significant feature of this syndrome and help the clinician to differentiate this entity from other craniofacial syndromes. Patients with Nager syndrome often require several surgical procedures for correction of congenital anomalies, including cleft palate repair and mandibular lengthening with distraction osteogenesis or orthognathic surgery, among others, including bone grafts and ramus reconstruction.²

Figure 1 Downsloping palpebral fissures with microretrognathia and malar hypoplasia.

Clinical management in these cases mainly depends on a multidisciplinary approach focused on treatment of symptoms. Panoramic radiograph and lateral cephalogram obtained during the initial diagnostic procedure revealed bilateral TMJ ankylosis. The patient was referred to the oral and maxillofacial surgery department and was motivated for surgical treatment.

Figure 2 (A and B) Short forearms with proximal radioulnar synostosis.

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Figure 3 (A) Hypoplastic thumb and (B) short overlapping toes.

Figure 4 Lateral cephalogram showing an underdeveloped mandible with an extreme posterior inclination of the mandibular base and also complete obliteration of temporomandibular joint (TMJ) space.

Figure 5 Panoramic radiographic examination showing reduced vertical height of ramus, and multiple impacted teeth in the maxilla and mandible, with complete obliteration of right and left temporomandibular joint (TMJ) space.

Total prosthetic TMJ replacement has been successfully used for various pathological, developmental and traumatic conditions, including inflammatory arthritis involving the TMJ, recurrent fibrous or bony ankylosis not responsive to other modalities of treatment.²

In this case, correction of the TMJ ankylosis was planned with a bilateral custom total prosthetic joint replacement; this surgery helps in re-establishing the form and function of the mandible and also significantly improves the patient's quality of life.

Learning points

- ▶ Nager acrofacial dysostosis is a rare congenital syndrome and nearly 94 cases have been reported in the medical literature. Nager syndrome is characterised by craniofacial, limb and musculoskeletal anomalies. The orofacial features are similar to those of Treacher Collins syndrome, but it can be distinguished from Treacher Collins syndrome by preaxial limb deformities.
- ▶ Nager acrofacial dysostosis is associated with various syndromic deformities, making it difficult to predict the treatment result. The clinical management for most cases mainly depends on a multidisciplinary approach consisting of maxillofacial radiology, oral and maxillofacial surgery, paediatrics and neonatology, otolaryngology, anaesthesia, plastic surgery and orthopaedic surgery.

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