Preaxial acrofacial dysostosis (Nager syndrome): a case report

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Abstract
This case report describes the surgical and dental management of a 12-year-old girl with preaxial acrofacial dysostosis also known as Nager syndrome. It highlights the importance of multidisciplinary treatment at an early age and the effect of parental noncompliance on the overall physical and psychological development of a medically compromised patient.

Key words: Nager syndrome, mandibulofacial dysostosis, multidisciplinary care

Introduction
Preaxial acrofacial dysostosis is a rare condition combining the features of mandibulofacial dysostosis and limb abnormalities. It was first recognised as a specific entity by Nager and de Reynier (1948), but was probably first reported by Slingenberg (1908).

The craniofacial deformities seen in Nager acrofacial dysostosis include downward slanting palpebral fissures, malar hypoplasia, high nasal bridge, micrognathia and external ear defects. Lateral extension of scalp hair onto the cheeks, reduced number of eyelashes and lower lid colobomas occur less frequently; while conductive hearing loss is seen frequently. Predominant oral findings include cleft palate and an absent soft palate. Typical upper limb abnormalities include absent radii, radioulnar synostosis and hypoplastic or absent thumbs. Lower limb anomalies also seen are absent tibia/fibula, talipes equinovarus and dislocated hips (McDonald and Gorski, 1993). Other structural malformations seen less frequently are tetralogy of Fallot, spina bifida, scoliosis, renal agenesis or malposition, duplicated calyces, bicornuate uterus. A few patients also exhibit microcephaly, hydrocephalus and mental retardation (Guigliani and Pereira, 1984).

The inheritance pattern of Nager syndrome is largely unknown. Most of the cases are sporadic. In offspring of mildly affected parents, the inheritance may be autosomal dominant, but some autosomal recessive cases have also been reported (Kavadia et al., 2004). Other structural malformations seen less frequently are tetralogy of Fallot, spina bifida, scoliosis, renal agenesis or malposition, duplicated calyces, bicornuate uterus. A few patients also exhibit microcephaly, hydrocephalus and mental retardation (Guigliani and Pereira, 1984).

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Nager syndrome is often misdiagnosed or not diagnosed at all. A differential diagnosis to distinguish it from Treacher-Collins, Miller and Pierre Robin syndromes is essential (Halal et al., 1983). Patients with Treacher-Collins syndrome have more severe forms of hypoplastic zygomatic, downward slanting palpebral fissures, lower lid colobomata and hypoplastic maxillae. In Nager syndrome patients, a higher degree of mandibular hypoplasia and greater frequency of palatal and limb abnormalities are seen as compared to Treacher-Collins syndrome, where severity is concentrated in the midface area (Danziger et al., 1990). Pierre Robin sequence presents with the characteristic features of retrognathia, glossoptosis, cleft palate and airway obstruction (Hunt and Hobar, 2002). Several variants of acrofacial dysostoses with varying levels of severity have been described such as Genée-Wiedemann syndrome and Rodríguez-type acrofacial dysostosis (Dimitrov et al., 2005). Miller acrofacial syndrome shows postaxial limb changes associated with facial deformities, while in Nager syndrome, the limb deformities are preaxial (OMIM, 2006).

Case report
A 12-year-old Caucasian girl was referred by the school nurse to the Craniofacial Center for treatment of severe dental and speech problems. She was initially diagnosed with Pierre Robin syndrome and received treatment for repair of the cleft palate in another institution. The patient was undergoing speech therapy at school with no marked improvement.

The patient's mother denied any significant medical problems and attributed her speech and intellectual defects to her congenital illness. However, records taken by the school nurse revealed that she had an asthma attack at 15 months...
of age and was treated in the emergency room. School records also showed in her past history that the patient was delayed in all the milestones of development. She also suffered from asthmatic attacks triggered by cold weather, which were managed by metered dose inhalers and nebuliser treatment. She had previously been diagnosed as having a 'third degree' curvature of the spine, which the parents were advised to leave untreated. However, the scoliosis could not be substantiated. She had also been evaluated for hearing loss several times in school as a possible reason for her poor academic performance and was diagnosed as having bilateral fluctuating conductive hearing loss. Tubes had been placed four times in her ears, the last one placed at 9 years of age.

At the time of her presentation to the Craniofacial Center, the facial examination revealed malar hypoplasia, slight downward slant of the orbits and left ear with a mild cup deformity (Figure 1). The retrognathic convex profile demonstrated a small mandible (Figure 2). Besides her unusual facial appearance, the patient also had anomalies in both hands with hypoplastic thumbs (Blauth I classification) (Figure 3). Her right arm appeared thinner and weaker than the left. This was confirmed by hand radiographs, which showed hypoplastic phalanges of both thumbs, with increased severity on the right thumb consistent with Nager syndrome. Oral examination revealed severe dental caries with plaque present, acute gingivitis and extremely poor oral hygiene (Figure 4). She also presented a narrow V-shaped, short palate with velopharyngeal insufficiency (Figure 5) diagnosed by the team’s speech pathologist. Orthodontic evaluation showed a retrognathic convex profile with a vertical growth pattern and an anterior open bite with increased overjet and an Angle’s Class II malocclusion. No lateral cephalogram radiograph was taken at the time of her examination.

Speech evaluation revealed nasal speech due to the velopharyngeal insufficiency. Audiological examination showed that her hearing was normal and the conductive hearing loss was due to accumulation of cerumen in the narrow canals. Psychological evaluation placed her within the Mentally Impaired Range with an intellectually deficient level of functioning. The speech defect contributed to difficulties in verbal expression and resulted in behavioural modification as an attempt to conceal her inadequacies.

Treatment was started for the patient with dental reha-
instructions in oral hygiene maintenance with regular brushing and flossing and periodic dental visits. Despite good treatment results, the patient failed to keep any further appointments and left the treatment incomplete.

Discussion

The combination of facial features with limb defects and the presence of cleft palate indicated Nager syndrome in this case. A common problem encountered in patients with Nager syndrome is misdiagnosis and lack of awareness on the part of the parents. There is some evidence of heredity in this syndrome but most cases are sporadic. Many of the earlier cases were reported as autosomal dominant; however some reports have found evidence of autosomal recessive mode of inheritance (OMIM, 2006). A detailed genetic counselling of family members of patients with Nager syndrome is important to make them aware of its heritability and increase their knowledge about this condition.

Clinical management in such cases has relied on a multidisciplinary approach for treatment. From birth and during infancy, multiple surgical procedures may be required to improve functional and aesthetic capabilities. In cases of decreased jaw size, a tracheotomy may be necessary to correct the airway obstruction and facilitate proper breathing (Friedman et al., 1996). Subsequently, feeding may rely on a gastrostomy tube. When present, clefts should be repaired in the early years. Hearing levels should be assessed at this time and, if necessary, ventilation tubes and hearing aids may be recommended if temporary or long-term hearing loss is present. In early childhood, speech and physical therapies may be needed to improve speech and hand movements, respectively. The patient must be referred to an orthopaedic surgeon for correction of limb defects. Approaching the teenage period, the facial appearance, including jaws and ears, can be improved by craniofacial and jaw surgery coupled with orthodontics. Orthodontic alignment of the teeth would be necessary for the presurgical dental decompensations. Orthognathic surgical procedure is indicated in most of these cases to correct the mandibular hypoplasia. Patients with minimal deformity require either no treatment or a genioplasty to improve facial appearance. Most of the patients have more severe forms of micrognathia and require surgery for mandibular advancement or mandibular distraction osteogenesis, which is typically performed after post-pubertal growth is completed (Stelnicki et al., 2002). A multidisciplinary approach must be adopted early on in the treatment to address the different problems faced by these patients. In this case, the patient failed to continue treatment and orthodontic/orthognathic care could not be attempted.

The provision of preventive and restorative dental care is an important aspect in the multidisciplinary management of these children. In medically compromised children, there is a risk of developing systemic complications from dental
infections (Foster and Fitzgerald, 2005) and a consultation with the primary health care provider is of absolute importance before undertaking any dental procedures. Antibiotic prophylaxis prior to commencement of dental treatment is essential to prevent any systemic complications from oral infections, especially in patients with cardiac abnormalities.

Low income and minority children and those with special health care needs are at a great risk for inadequate access to oral health care. Factors contributing to this in the United States include geographical maldistribution of clinicians and inadequate numbers of dentists treating children with government-sponsored insurance (Mouradian et al., 2004). To ensure adequate health care in such patients, an increase in parent motivation and improved access to oral health care in patients with special needs is essential.

References


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