Limbal dermoid in nager acrofacial dysostosis: A rare case report

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Nager syndrome, also called preaxial acrofacial dysostosis, comprises two groups of defects involving the limbs and craniofacial region, respectively. This syndrome is rare and only 70 cases have been reported in the literature. The exact cause of this syndrome is unknown, but there is indication that it is genetically based. Ocular manifestations of this syndrome include widely separated downward slanting eyes, absence of eyelashes, ptosis of upper eyelids and colobomas on the inner aspect of lower eyelids. We report limbal dermoid in a patient with Nager syndrome. We did not find such an association of “Limbal dermoid in Nager acrofacial dysostosis syndrome” on PubMed using Nager acrofacial dysostosis, limbal dermoid and ocular manifestations as the keywords.

Key words: Absent thumb, craniofacial defects, limbal dermoid, Nager syndrome

Nager syndrome, also called preaxial acrofacial dysostosis, was first described by Nager and Reynier in 1948.¹² It consists of a defect in the development of the first and second branchial arches and in the formation of cartilage.¹² Typically, Nager acrofacial dysostosis (NAFD) comprises two groups of defects involving the limbs and craniofacial region, respectively.¹² The former are mainly deficiencies mostly affecting the upper limbs. The latter form a complex indistinguishable from mandibulofacial dysostosis (MFD).²³

The prevalence is unknown; about 70 cases of Nager syndrome have been published. The MFD complex is unmistakable and comprises hypoplasia of the malar eminences and zygoma, hypoplasia of maxilla with cleft of secondary palate or highly-arched palate, absence of velum (rarely with choanal atresia), and extension of a “tongue” of temporal hair down the sides of the cheeks. The ocular features are important and include downward slant of palpebral fissures, ptosis of upper lids, coloboma of lower lids, and deficiency of eyelashes of the medial one-third to two-thirds of the lower eyelids. Hypoplasia or absence of thumbs is the most characteristic feature almost invariably associated with radio-ulnar synostosis. Triphalangeal thumbs and index finger are equally characteristic. Most NAFD individuals have normal intelligence, and after infancy most are healthy and are presumed to have a normal lifespan. All acrofacial dysostosis must be considered as genetic disorders until proven otherwise, and parents deserve careful scrutiny for mild manifestations.¹³⁻¹⁵

Limbal dermoids are benign congenital tumors that contain choristomatous tissue. They appear most frequently at the inferior temporal quadrant of the corneal limbus. We report limbal dermoid in a patient with Nager syndrome. We did not find such an association of “Limbal dermoid in Nager acrofacial dysostosis syndrome” on PubMed using Nager acrofacial dysostosis, limbal dermoid and ocular manifestations as the keywords.

Case Report

A 16-year-old female patient came to Department of Oral Medicine and Radiology with chief complaints of bleeding from gums. On examination, she was thin built. The right thumb of her right upper extremity was absent. Right forearm appeared short. All her vitals were within normal limits. Extraoral examination of face and skull revealed hypoplasia of the malar eminences and zygoma, hypoplasia of maxilla and underdevelopment of the mandible [Fig. 1]. Ears appeared low-set and rotated with presence of skin tags [Fig. 2]. Intraoral examination revealed inflamed gingiva due to poor oral hygiene. Radiological examination supported the clinical findings. Patient was then referred to ophthalmology department where ocular examination showed downward slant of palpebral fissures, ptosis of upper lids and limbal dermoid in right eye measuring around 1 × 1 cm in diameter [Fig. 3]. On the basis of examination and radiographic investigations, the patient was diagnosed as a case of Nager syndrome. The patient was uncooperative and refused any other treatment except for oral prophylaxis which we performed to relieve her from bleeding gums.

Discussion

This condition is described as a syndrome which involves the upper limbs and skull. The most ordinary type of acrofacial dysostosis is the Nager type (NAFD) of the so-called pre-axial acrofacial dysostosis. This disorder is categorized by the Office of Rare Diseases in the United States as a rare illness; thus, it has only affected less than 200,000 of the country’s population. This disorder is often sporadic although it can be inherited too. Some studies have shown that the gene which causes the illness might be located around 9q32 and it is possible that the ZFP-37 gene could also be involved.¹²⁻¹⁴

The major symptoms of Nager syndrome often include the underdevelopment of the jaw and cheek areas, a jaw that may appear smaller than the usual or normal jaw, hearing problems (and some defects on the external or internal ear area), cleft palate (which is a congenital defect of the mouth or the lip), the absence or underdevelopment of the thumb or thumbs, forearms that appear short (the forearms could...
altogether be absent of the radius bone), fusing of the bones of the forearms, poor elbow movements (or limited extension of this body part), ears that appear to be backward rotated, the ears manifesting some skin tags (or they could be low-set), and feeding and breathing problems (especially among infants who are afflicted with this syndrome). Our patient showed underdevelopment of jaws, hypoplasia of the malar eminences, zygomata and hypoplasia of maxilla. The ear showed presence of skin tags. There was absence of thumb of her right hand which also appeared short.

Ocular manifestations of this syndrome are rare and include downward slant of palpebral fissures, ptosis of upper lids, coloboma of lower lids, and deficiency of eyelashes of the medial one-third of the lower eyelids. Our patient showed downward slant of palpebral fissures, ptosis of upper lids and limbal dermoid in right eye.

Traditionally, the differential diagnosis of Nager syndrome is done with Treacher-Collins, Pierre-Robin and Goldenhar syndrome. Treacher-Collins syndrome is characterized by maxilla and mandible hypoplasia, downslanting palpebral fissures, colobomas of inferior eyelid and auricular malformation. Pierre-Robin syndrome presents with micrognathia, cleft palate and glossoptosis. In Goldenhar syndrome, we find hemifacial microsomy, flattening of maxilla, temporal and zygomatic bone, microtia, ocular and vertebral alterations, and in 50% of cases, Fallot tetrad.

Radiological findings are usually supportive of clinical examination revealing hypoplasia of facial structures. The echographic findings of these malformations become evident usually in the third trimester, therefore a systematic ultrasound exploration including visualization of fetal face line is essential for its detection.

The treatment is totally dependent on the symptoms manifested. If the patient suffers from speech retardation, then a speech therapist could help the patient. Physicians who specialize in ear problems can also help. They may not totally cure the condition, but they can help in managing the symptoms. Treatment of limbal dermoids may consist of periodic removal of irritating cilia, topical lubrication to prevent foreign body sensation, or excision of the lesion if it is causing significant cosmetic disfigurement or interfering with vision. Our patient refused to take any ocular treatment and insisted on taking only the dental treatment for which she visited our hospital.

**Conclusion**

Correct understanding of the underlying pathophysiologic pattern of this disease is necessary for optimal diagnosis and treatment planning. This should be supported with appropriate investigations, both systemic and radiological. Ophthalmological examination of these patients is a must as ocular manifestations are not uncommon in these patients. We found limbal dermoid associated with this syndrome.

**References**