Case Report

Nager Syndrome Involving Radicular Cyst: A Case Report

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ABSTRACT:

Nager syndrome, also known as preaxial acrofacial dysostosis, is a rare disorder characterized by malformations in the craniofacial region such as micronagthia, cleft palate, zygomatic hypoplasia and preaxial limb deformities. There is abnormal development of first and second branchial arches and limb buds. Besides these features, ankylosis of temporomandibular joint and downward slanting of palpebral fissures is also observed in such patients. There is presence of autosomal dominant and autosomal recessive inheritance for this syndrome, which further indicates genetic heterogeneity. It is a rare genetic condition with limb deformities such as missing thumbs and short forearm bones, due to mutation in the SF3B4 gene. Intraorally, a well defined dome shaped swelling was observed in the centre of hard palate. Radiographically, a well defined, roughly oval radiolucency with respect to 11 and 21 in the periapical region was observed.

Proper clinical assessment and genetic analysis can help in the diagnosis of this disorder. Treatment of this syndrome involves physical therapy, surgery and use of hearing aids. In early stages of management, breathing and feeding must be facilitated and then there should be assessment of oral and facial clefts. Cheek bone reconstruction and treatment of deformities in the eye should be considered for such patients. The purpose of this article is to focus on the clinical features of the patients, suffering from Nager Syndrome, and also evaluation of the orofacial manifestations which indicated the presence of radicular cyst in the patient. We concluded that such patients require clinical management based on multidisciplinary approach including several surgical procedures for correction of congenital abnormalities such as cleft palate repair and mandibular lengthening with orthognathic surgery.

KEY WORDS: syndrome, dysostosis, limb, micrognathia, cleft palate

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INTRODUCTION:

In 1948, Nager and de Reynier observed this syndrome and they used the term acrofacial dysostosis to describe their patient. Nager Syndrome, also known as preaxial acrofacial dysostosis, occurs due to variation the growth of first and second branchial arches and limb buds.[1] The major clinical features include micrognathia, cleft palate, limb deformities, downward slanting of the palpebral fissures, mandibular hypoplasia, hypoplasia or absence of thumbs, microretrognathia, absence of lower medial eyelashes and ankylosis of the temporomandibular joint.[2] An important feature of this syndrome is a limb deformity kown as micromesomelia with hypoplasia or

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Figure 1

Figure 2

Figure 3

Figure 4

Figure 5

Figure 6

Figure 7

Figure 1 shows front profile with orofacial features associated with Nager Syndrome, figure 2 shows deformity in upper limb, figure 3 shows deformity in lower limb, figure 4 shows dysplastic ears,figure5 shows intraoral swelling, figure 6 shows intraoral periapical radiograph with root displacement and root resorption associated with 11 and 21,and figure 7 shows occlusal radiograph with well-defined roughly oval radiolucency associated with 11 and 21 and other radiographic features of radicular cyst.

agenesis of the radius and thumb. There is bilateral hypoplasia of the facial bones, involving mandible and zygomatic complex. It is caused by mutations in the SF3B4 gene, and it typically occurs sporadically but can follow autosomal dominant inheritance.

There is microtia or atresia of the external ears and narrowing of the ear canal which causes conductive hearing loss.[3] It occurs at a frequency of 3 per 10 lac live births and studies indicate approximately 100 cases of such patients so far.[4] The clinical significance of presence of radicular cyst in a Nager syndrome patient is very high because such patients have compromised craniofacial structure. Such patients are already suffering from severe migrognathia and retrognathia, therefore any additional swelling or inflammation can obstruct oropharyngeal airway leading to life threatening respiratory distress.

CASE REPORT:

A 34 year old male patient reported to the department with the chief complaint of swelling in upper front tooth region since two months. Initially, the intraoral swelling was small in size but gradually the size increased and the intensity of pain also increased. There was a history of trauma to the upper front tooth region. There was also history of difficulty in mastication and swallowing but there was no history of bleeding or pus discharge. The family history of the patient was not contributory and no deleterious habits were present. On physical examination, the patient was found to be epileptic and had suffered trauma in the maxillary arch. Physical examination of the patient also showed micrognathia, microtia and mandibular hypoplasia (Figure 1). Malformed ear, hypoplastic mandible, hearing loss, limb deformity, speech deformity, speed difficulty and mental retardation was observed in the patient. There was presence of downslanting palpebral fissures, sloping forehead, short forearms, proximal radioulnar synostosis, hypoplastic thumbs and relatively small overlapping toes. The elbow articulation extension presented difficulties. There was presence of bilateral club feet. Dysplastic ears and auditory canal atresia was present (Figure 2, Figure3 and Figure 4). Hearing evaluation using brainstem evoked response audiometry (BERA) revealed bilateral profound hearing loss. Orofacial features included high nasal bridge with an upturned nasal tip, short philtrum, and high narrow hard palate was present. Hypoplastic zygomatic region and mandibular retrusion was observed with absence of soft

palate. The mouth opening was restricted and the interincisal opening was found to be 25 mm. Temporomandibular joint fibrosis and ankylosis was not present. Intraoral examination revealed discolouration with respect to 11 and 21. A well-defined dome shaped swelling was seen in the centre of the hard palate with respect to 11 and 21.Tenderness on vertical and lateral percussion was present in the associated teeth. The size of the intraoral swelling was approximately 2 X 2.5 cm in greatest dimension. The shape was roughly oval with raised and smooth surface. The colour was same as that of the adjacent mucosa (figure 5). On palpation, the intraoral swelling was found to be fluctuant with soft consistency. Tooth mobility was observed with respect to 11 and 21.A differential diagnosis of radicular cyst, periapical abscess and periapical granuloma with respect to 11 and 21 was given. A provisional diagnosis of radicular cyst with respect to 11 and 21 was given. An intraoral periapical radiograph (IOPA) and occlusal radiograph was taken and they showed well-defined, distinct, roughly oval radiolucency with respect to 11 and 21 in the periapical region with well-defined and corticated margins (Figure 6 and Figure 7). Other radiographic features include root displacement and root resorption with respect to 11 and 21.Based on the craniofacial features and the coexisting upper and lower limb preaxial deformities, a diagnosis of Nager Syndrome was given. Based on history and clinical features, another diagnosis of radicular cyst with respect to 11 and 21 was given. The patient did not undergo any further treatment.

DISCUSSION:

Nager syndrome is a malformation which occurs due to abnormal growth of first and second branchial arches and limb buds. It is mostly sporadic and occurs due to changes in the 9q32 chromosome.[5] There is haploinsufficiency of SF3B4 encoding a spliceosomal protein SAP49 and some families apparently with either dominant or recessive inheritance have be observed and assessed. The clinical features include down slanting palpebral fissures, micrognathia, malar hypoplasia, glossoptosis, deformity of palate, dysmorphic pinnae, middle ear abnormality, external auditory stenosis, microtia, deformity of limbs such as hypoplasia or absence of thumbs, triphalangeal thumbs, radial hypoplasia or aplasia and radioulnar synostosis. Facial dysostosis is a group of congenital craniofacial deformities. Nosology committee of the International Skeletal Dysplasia Society classified dysostosis with predominant craniofacial involvement which comprised of mandibulofacial dysostosis (Treacher Collins), mandibulofacial dysostosis with microcephaly, mandibulofacial dysostosis with alopecia, Miller syndrome (postaxial acrofacial dysostosis), Nager type, Rodriguez type and Cincinnati type. Approximately 60% of patients with characteristics of Nager syndrome have pathogenic variants in SF3B4 gene and these are frame shift and splice site mutations.[6] The differential diagnosis include Miller syndrome, Treacher Collins syndrome and Pierre Robin syndrome. The facial characteristics of Nager syndrome roughly resemble those of Treacher Collins syndrome but differentiation between the two can be made. Patients suffering from Treacher Collins syndrome have more severe type of hypoplastic zygoma, hypoplastic maxilla and downward slanting palpebral fissures. But in Nager syndrome, the severity of these features is comparatively less. Mandibular hypoplasia, lower eyelid colobomas, zygomatic hypoplasia, downslanting palpebral fissures, external ear malformation and genitourinary abnormalities are some of the common features seen in patients suffering from Nager syndrome. Other oral findings include absent soft palate, cleft lip, foreshortening of soft palate, temporomandibular joint fibrosis, ankylosis, oligodontia, microdontia, enamel hypoplasia, high arched palate and malocclusion.[7]

Sometimes, severe trismus, restricted mandibular movement and speech difficulties, due to impaired hearing and velopharyngeal insufficiency, are also present. Cleft palate can also occur in such patients due to influence of mandibular retrusion in respiration and feeding.[7] In some cases, cardiopulmonary anomalies such as severe aortic stenosis and right pulmonary bronchial stenosis can occur. In rare cases, ventricular septal fect can be seen.[8] The presence of trismus, maxillomandibular hypoplasia, and micrognathia can lead to severe airway obstruction in such patients. It can harm the perioperative anaesthetic care of such patients.[9] Previously reported cases included a case of 4 year old boy with feeding difficulties due to malformation of orofacial region like severe mandibular retrusion. Besides this, low set ears, duplication of the left thumb and cryptorchidism was observed but there was no evidence of mental retardation. The forearms were short and there was difficulty in elbow articulation. Hypoplastic zygomatic region and mandibular retrusion was present but there was no restriction of jaw movements. Anisopthalmia, epicanthus and downslanting palpebral fissures were observed but there were no hearing disturbances. Intraorally, there was microdontia of the primary dentition.[8] Another case of 3.5 year old girl was

reported with severe mandibular retrusion, cleft palate and tongue tie. Craniofacial features included malar hypoplasia, severe mandibular hypoplasia and retrognathia but there was no evidence of mental retardation. Bilateral club feet with short arms was observed. There was speech difficulty.[7] However, in our case, mental retardation was observed but tongue tie was not observed. Also, no abnormality was clinically detected in temporomandibular joint.

Early and aggressive surgical management for hemifacial disorders have been conducted in the past. Recommended rehabilitative treatment modalities include prespeech feeding activities, oral language stimulation, speech therapy and early audiological evaluation and amplification. Early intervention with tracheostomy is advised in case of severe airway obstruction. Gastrostomy is advised to improve nutritional status for patients suffering from difficulty in swallowing. Orthopedic management is suggested for such patients.[10] The patient's restricted mouth opening made dental examination and management very difficult and it was necessary to perform treatment under general anaesthesia but that too is challenging in such patients because maxillomandibular hypoplasia with micrognathia and slowly expanding intraoral swelling can lead to airway obstruction. It is important to decrease the number of avoidable general anesthetics including those for dental treatment. If the preventive counseling is not provided, it can lead to continuous use of general anesthesia which is undesirable in terms of morbidity and potential mortality, behavioural and emotional effects on the patient.

Radicular cysts are the most common cystic lesions present in the jaws and are mostly found at the apices of the nonvital teeth. The main causes are presence of dental caries, trauma and large faulty restoration. Facial asymmetry with slow growing, firm, non tender swelling is observed in such patients.[11] Mostly males in their third decade of life are affected. Mostly they are present at the apices of maxillary teeth than mandibular teeth. In maxilla, anterior region is mostly affected whereas in mandible, the premolar and molar region is more prone to the occurrence of this cyst. Rarely, these cysts are present in deciduous dentition. These cysts originate from epithelial residues in the periodontal ligament due to inflammation. Dental caries can cause pulpal infection and the bacteria travels from periodontal pockets to root canals through damaged periodontal blood vessels. Exposed dentinal tubules at cervical root surface can also cause pulpal infection. Autogenous oral microflora can infect tooth pulp and make it necrotic. There is development of mixed, predominantly anaerobic flora. The microbial invaders in the root canal have biological and pathogenic properties such as chemotaxis, antigenicity, mitogenic activity and activation of host cells. In response, the host activates its defense system comprising of antibodies and intercellular messengers and it results in destruction of periapical tissue causing chronic apical periodontitis which can sometimes develop into periapical or radicular cysts.[12] Most of these cysts are asymptomatic but patient can complain of slow enlarging swelling. Initially, the enlargement is bony hard but as the cyst increases in size, the covering bone loses its thickness. There is springiness in the swelling but fluctuation can also occur if the cyst has completely damaged the bone. Buccal or palatal enlargement occurs in maxilla but in mandible, enlargement is labial or buccal. These cysts are painless until infected but pain and infection can occur. Radiographic features include round or pear shaped unilocular radiolucency with well-defined cortex, more than 2cm in diameter, present at the apex of non vital tooth. There can be displacement and resorption of roots of adjacent teeth. On histopathological analysis, yellow mural nodules of cholesterol are found along with brown coloured fluid contents due to breakdown of blood and surrounded by non-keratinized stratified squamous epithelium. The presence of inflammatory cell infiltrate and hyaline bodies due to hemorrhage within inflamed cyst wall is often observed. Treatment involves conventional nonsurgical root canal therapy, if the lesion is localized or enucleation, marsupialization, if the lesion is large.[13] The choice of treatment is determined by factors such as extension of the lesion, association with anatomical structures, origin and clinical features. Disadvantages of surgery include damage to vital structures and scar formation,so endodontic therapy is preferred.[14] Early diagnosis and presence of multidisciplinary team including oral and maxillofacial surgeons, otolaryngologists, geneticists and psychologists can greatly enhance patient outcomes. Our patient refused treatment.

CONCLUSION:

Nager syndrome is a rare syndrome identified by facial deformity and limb defects.Nowadays,for diagnosis of genetic diseases,Whole Exome Sequencing(WES) or Whole Genome Sequencing (WGS) can be conducted.CT scans and other radiological methods are also useful in early diagnosis of this syndrome and treatment planning.The aim is to achieve functional,cosmetic and morphological improvement in such patients.Surgical management and physiotherapeutic techniques play an important

role in improving the quality of life of such patients.

DECLARATION OF PATIENT CONSENT:

The authors certify that they have obtained all appropriate patient consent forms. In the form, the patient(s)/guardian has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients/guardian understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

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Conflicts of Interest

There are no conflicts of interest.

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