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Nager Acrofacial Dysostosis: A Case Report

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Abstract

Nager syndrome is a genetic condition which causes a myriad of oro-facial and limb manifestations. Knowledge about the various clinical symptoms is essential to a pediatric dentist to provide proper dental care for the patient. This case report discusses the clinical manifestations seen in an eleven year old patient who was previously undiagnosed of the condition.

Introduction

Nager acrofacial dysostosis, (OMIM number: #154400) also known as preaxial acrofacial dysostosis is defined by mandibulofacial dysostosis and radial defects. It is a rare condition that mainly affects the development of the face, upper and lower extremities.(1) (2) The severity of the disorder varies among affected individuals. The syndrome is caused by mutations in the SF3B4 gene which was identified by the National Institutes of Health Centers for Mendelian Genomics. (3) Most cases are sporadic but some cases of autosomal dominant trait have been reported.(4) Nager syndrome was first described in medical literature in 1948 by Nager and DeReynier, but identified by Slingenberg in 1908.(5) Affected individuals may develop a variety of craniofacial and limb abnormalities, some of which are noticeable at birth. (2)

Case report

An 11 year old male child reported to the Department of Pedodontics and Preventive dentistry with the chief complaint of forwardly placed upper front teeth. The patient walked in, with a limp in his left limb. He appeared lucid and answered questions normally. On prenatal history, the patient's parent revealed that they had a non-consanguinous marriage and the mother had experienced two abortions prior to conceiving with the patient. In postnatal history, patient had shown delay in the milestones of development until three years of age. Patient did not develop suckling reflex and was bottle fed until two years of age. He had started walking and speaking only after three years of age.

On clinical examination, there was camptodactyly of the fourth phalange of the left hand, and shortening of the lower left limb.(Figure 1) Extra-oral features observed were, bilateral asymmetry of face, and downward slant of the palpebral fissures, partial absence of eyelashes in his lower eyelid, low set ears, micrognathia in relation to the

maxilla and mandible, a high nasal bridge and hypoplasia of the zygomatic region.(Figure 2) Patient's profile was convex and he had incompetent lips along with restricted mouth opening due to microstomia. Patient had retained infantile swallow pattern.

On intraoral examination, there was severe crowding present in relation to the upper and lower anteriors. Upper anterior proclination was seen in relation to 11, 21 with hypoplastic bands observed on the facial surface, 12 and 22 were placed palatally.(Figure 3) Orthopantomogram revealed the presence of all the permanent tooth buds beneath their respective primary counterparts. With the craniofacial clinical findings a differential diagnosis of Treacher Collins syndrome, Nager syndrome or Miller's syndrome was made. The final diagnosis of Nager's syndrome was made after careful examination of the preaxial symptoms.

The treatment plan devised was, extraction of all primary retained teeth. Followed by, maxillary arch expansion and twin block appliance therapy after eruption of the premolars. Finally, fixed appliance therapy for final alignment of teeth.

Discussion

Diagnosis of the syndrome had been made with the presence of clinical signs and symptoms seen in the patient. Malformation of the mandibulofacial structures involves the disruption of first and second brancial arches during the intrauterine life.(6) Taking into consideration the craniofacial abnormalities a differential diagnosis of Treacher Collins syndrome, Millers syndrome or Nagers syndrome was made. Though the orofacial findings mimicked Treacher Collins syndrome, it was excluded after taking the limb findings into consideration.(7) The classical feature of Miller syndrome is the absence of phalanges which was not seen in this patient.(8) The craniofacial abnormalities presented by the patient coincide with the findings presented by Nager in 1948.(9) Meyerson et al. 1977 has reported the presence of bilateral camptodactyly and clinodactyly of index and fifth fingers in his case report.(10) In this case unilateral camptodactyly was observed in the fifth finger of the left hand. Paladini et al. 2003 has reported a prenatal diagnosis of Nager syndrome which had led to better understanding of the condition by the caregiver's and immediate care.(1) In this case the patient was undiagnosed of the condition as the caregiver was not able to obtain proper medical attention for the patient during the prenatal and infancy period. Hence, parental counselling and guidance on the child's condition was lacking.

All the craniofacial abnormalities are to be corrected during childhood or adolescence. Thus, the dentist must have the proper knowledge of the clinical symptoms to work along with a Pediatric dentist, Oral surgeon and/or Orthodontist for a comprehensive care. Knowledge on the syndrome is important as the dentist may be the first person to diagnose a patient.

The management of craniofacial abnormalities should be done according to the nature of the defect with either surgical or orthodontic intervention at the appropriate stage.

Conclusion

Nager syndrome, though rare phenomenon if not diagnosed at an early age can lead to various complications in the child, especially in the orofacial complex. The diagnosis can be obtained from the craniofacial and limb findings. Proper diagnosis combined with surgical and orthododontic intervention can provide the best results for patients.

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Legends Figures

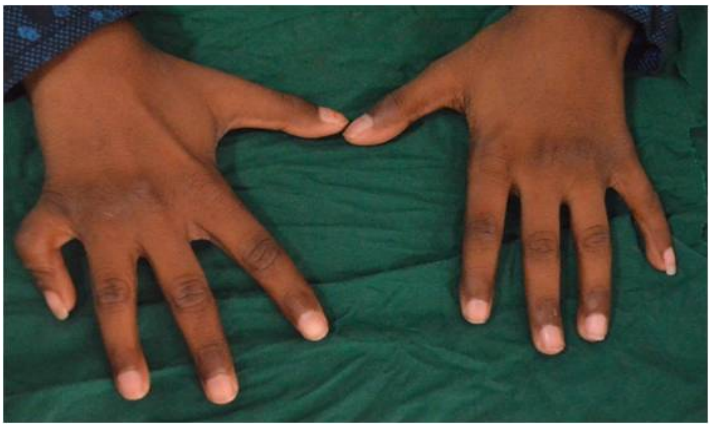

Figure 1

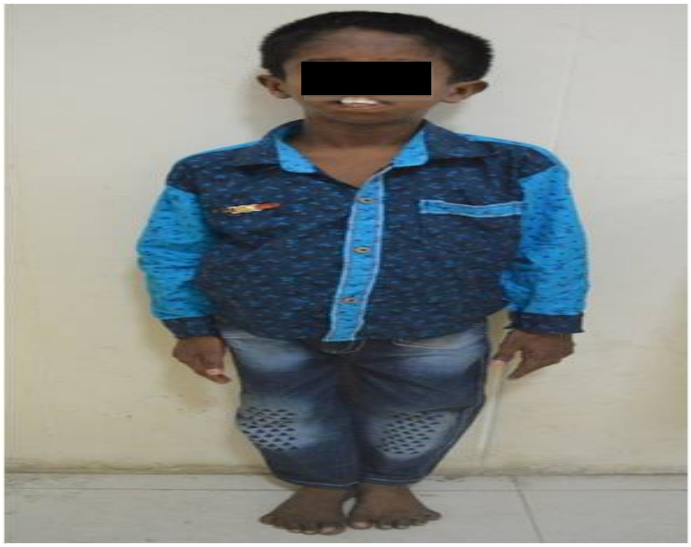

Figure 2

Figure 3

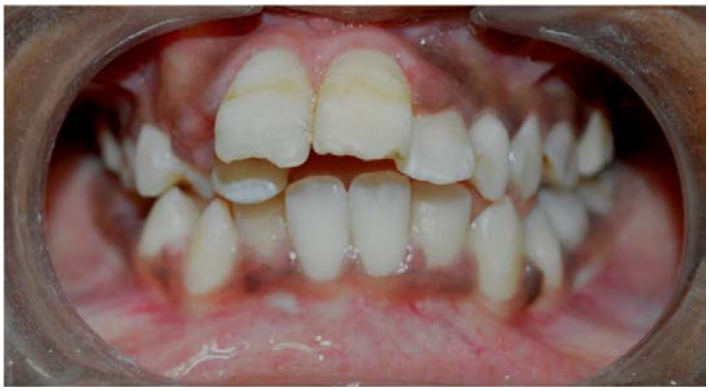

Figure 4