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Case Report

Dentistry Section

Orofacial Manifestations of Autosomal Recessive Robinow's Syndrome: A Rare Case Report

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ABSTRACT

Robinow's syndrome is a very rare genetic disorder which bears a resemblance to a foetal face. It is characterized by short-limbed dwarfism, defects in vertebral segmentation and abnormalities in the head, face and external genitalia. It has a genetic heterogeneity with autosomal dominant and recessive forms which relates to the severity of phenotype presentation. A rare case of an autosomal recessive form of Robinow's syndrome is presented with emphasis on, characteristic craniofacial and intraoral manifestations to aid in diagnosis and dental management of this patient.

Keywords: Absent tongue tip, Foetal face, Missing tooth

CASE REPORT

A 2.5-year-old male patient was referred for dental management prior to operation on genitalia. He was born to non consanguineous parents from the same geographic location. His parents and the other two siblings were normal. According to his medical history, the patient was diagnosed at birth as having Robinow's syndrome with the autosomal recessive form (diagnosis based on clinical features alone), as he had characteristic facial dysmorphism, mesomelic brachymelia and genital hypoplasia in form of buried penis for which he had been planned to be given three testosterone 25mg injections and is to be taken up for lengthening surgery for the same. No congenital heart disease has been diagnosed. On general examination, he was of a short stature with height of 74.1 cm and weight 9.3 kg, mesomelic dwarfism with UL:LL ratio of 1.6 and forearm rhizomelia with arm to forearm ratio of 1:1.5. Small feet with small hands were noticed. Gingival hyperplasia was seen in maxillary arch. There was no evidence of cleft and the lower left primary central incisor was missing clinically and radiologically [Table/Fig-1a]. Patient had mid thoracic scoliosis with multiple hemivertebrae [Table/Fig-1b].

Erupted teeth were hypoplastic. Follow up after 3 years revealed small feet with small hands with clinodactyly of 5th finger [Table/Fig-2a] macrocephaly with frontal bossing and low set ears [Table/Fig-2b]. Hypertelorism with prominent appearing eyes was noticed alongwith downslanting palpebral fissures. Mid facial hypoplasia was evident. Short upturned nose with broad nasal bridge was present [Table/Fig-2b]. Triangular mouth (bottom corners facing downwards) was seen. Tented upper lip with long philtrum was evident [Table/Fig-2b].

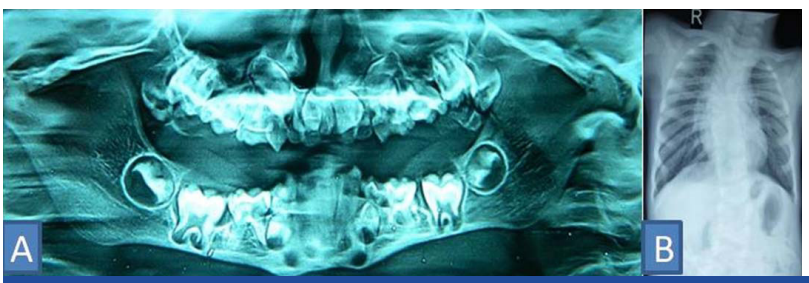
Intraoral examination revealed gingival hyperplasia [Table/Fig-3a]. Maxillary teeth were malaligned and high arched palate was present [Table/Fig-3b]. Early childhood caries was seen and the teeth were hypoplastic with missing left mandibular primary lateral incisor. Tongue tip was absent [Table/Fig-3c]. Radiographic examination showed that mandibular succedaneous left lateral incisor was missing [Table/Fig-3d].

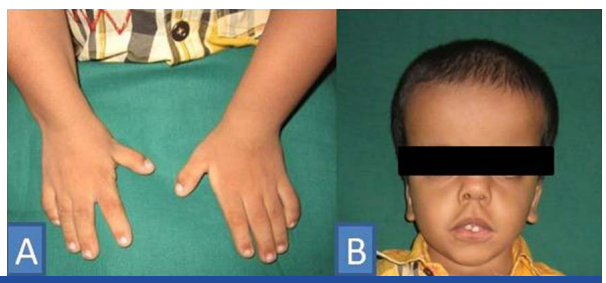
The dental treatment was restoration of carious teeth and flouride application (varnish) on teeth with enamel hypoplasia. Dietary and oral hygiene instructions were also given to the parents. Parents were informed about future needs of orthodontic/prosthodontic procedures and plastic surgery corrections. They were also provided information on speech therapy and surgical correction of bifid tongue if speech is found to be affected. Patient has been scheduled for 6-month follow up visits to monitor tooth eruption and enamel defects.

DISCUSSION

The fascinating resemblance of the affected individual to an eight-week-old foetus, also known as foetal face syndrome, was first described by Robinow et al., [1] It is a dwarfing syndrome, featuring mesomelic limb shortening, hemivertebrae and genital hypoplasia and is known as Robinow's syndrome. It is very rare, with an incidence of about 1:500000 and equal male to female ratio. However, the prevalence is slightly lower because 5% to 10% of the patients die in infancy or in early childhood [1]. The prognosis is poor especially when the patient presents with congenital heart disease [2]. It bears a genetic heterogeneity with autosomal dominant (AD) and autosomal recessive (AR) Robinow's syndrome forms [2]. The gene accountable for the autosomal recessive form was identified as the ROR2 gene on the 9q22 chromosome, a tyrosine kinase receptor [3], mutations in WNT5A, locus 3p14.3 are responsible for the AD form.

Diagnosis is based on the clinical picture and the characteristic foetal face appearance. Radiological examination is however necessary to confirm the presence of skeletal malformations. Only five cases of recessive Robinow's syndrome have been





[Table/Fig-1a & b]: Patient at the age of 2.5 years exhibiting: a) Absence of mandibular/maxillary cleft and absent primary left central incisor radiographically; b) Hemivertebrae.
[Table/Fig-2a & b]: a) Short hands with clinodactyly of 5th finger; b) Frontal bossing, low set ears, short upturned nose, tented upper lip.

[Table/Fig-3a, b, c, d]: a) Gingival hyperplasia b) High arched palate, malaligned maxillary teeth, missing left mandibular primary lateral incisor; c) Absent tongue tip; d) Follow up panoramic image showing absence of left mandibular succedaneous lateral incisor tooth bud.

published from India [4-6]. The aim of this article is to present a case of AR Robinow's syndrome with emphasis on craniofacial and oral abnormalities.

This syndrome is characterized by a large head with a prominent forehead, a flat nasal bridge associated with a short upturned nose, lower eyelids are S-shaped and ocular hypertelorism is present and such patients usually exhibit mandibular hypoplasia and in some cases facial nevus is also present. Indexed patient did not exhibit facial nevus and another interesting finding was evident midfacial hypoplasia in contrast to usually reported mandibular hypoplasia. The oral abnormalities, as also seen in the present case, include tented upper lip, exposed incisors and gingival hypertrophy [7]. These features are distinctive and abet in the diagnosis. Gingival hypertrophy may be present since birth. Teeth exhibit dental misalignment, crowded teeth, delayed loss of deciduous teeth, retained molar teeth and notching of teeth [7]. Our patient did not show these features. Also, Kantaputra et al., reported short teeth roots and narrow, thick floored pulp chambers in a patient with autosomal dominant type, which was not seen in the present case [8]. Missing left mandibular primary incisor and the succedaneous tooth bud, seen in the present case, has not been reported earlier. These patients are also reported to show rudimentary or absent uvula (18%), cleft lip and cleft palate (9%) [7]. The present case did not exhibit abnormal uvula. High arched palate was present. Tongue usually exhibits macroglossia, but the present case presented with absent tongue tip but his speech was not affected at this age.

The main differential diagnosis of AR form is AD pattern of inheritance. AD patients exhibit milder facial abnormalities and more prominent oral manifestations, whereas hemivertebrae and scoliosis are common in AR form. Syndrome that commonly involve dysmorphic features similar to Robinow's syndrome, particularly hypertelorism, along with genital hypoplasia that can be thought of as differential diagnosis are Aarskog syndrome (MIM305400) and Opitz G (MIM300000) syndrome [9].

Vertebral segmentation defects are seen in all patients with AR form. Most commonly it is seen as hemivertebrae and scoliosis, which was also seen in indexed patient. It is less commonly seen in dominant form. Patton and Afzal showed that patients with AR form have a height in the range of -2 SD or less, whereas AD cases have milder phenotype and could have height within normal ranges [3]. Mortality rate is 10% in AR cases, whereas AD cases do not show increased mortality [2]. Congenital heart defects are frequent in AR forms but were not present in the indexed patient.

The dental treatment that has been proposed includes restorations and fluoride application for hypoplastic teeth in serious caries risk. Follow-up visits to ensure monitoring of dental development have been scheduled. Parents have been advised for the possible need for orthodontic/prosthodontic treatment and orthopaedic and plastic surgery procedure for facial reconstruction in case of psychological problems caused by patient's craniofacial anomalies. Genetic counselling was provided for the family, because in the presence of family history, this can be relatively easy, and prenatal diagnosis is possible by means of foetal ultrasound at 19 weeks of pregnancy. The patient presented here has a good prognosis as there is no congenital heart disease and he has normal intelligence levels.

CONCLUSION

A rare case of autosomal recessive form of Robinow's syndrome with a 3 year follow up is discussed. Such patients need to be evaluated for caries risk and provided necessary preventive measures. They also need monitoring for dental development. Indexed patient also exhibited absent tongue tip and hypodontia which has not been reported earlier. A genetic counselling should be provided for the family and possibility of plastic surgery correction for specific facial deformities should be made known in case of psychological problem posed by the craniofacial anomalies in such cases. In case of parents belonging to highly consanguineous population, a detailed cardiac assessment is essential. Normal intelligence, adequate sexual functioning and reproduction, which are good prognostic indicators, occur in majority of the patients.

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