Robinow Syndrome and Fusion of Primary Teeth

Priti Sushil Jain, Tejashri Shreyas Gupte, Abdulkadeer M. Jetpurwala, Shely Pratik Dedhia

Department of Pediatric and Preventive Dentistry, Nair Hospital Dental College, Mumbai, Maharashtra, India

Abstract

Managing patients with rare genetic disorders is a challenge that dentists face often. Robinow syndrome (RS) is one such rare genetic disorder with <200 cases reported worldwide. RS demonstrates multiple craniofacial abnormalities and orodental disorders, which need to be taken into consideration by a dental practitioner while rendering dental care.

Keywords: *Craniofacial, fetal-facies, fusion*

Introduction

Robinow syndrome (RS), first described in 1969 by Robinow *et al*., is an extremely rare genetic disorder characterized by mesomelic dwarfism, abnormalities of the face, head, and external genitalia, as well as vertebral segmentation defects.[1] It has an incidence of 1:500,000 and a male to female ratio of 1:1.[2]

Both autosomal dominant RS (DRS1 OMIM#180700, DRS2 OMIM#616331 and DRS3 OMIM#616894) and autosomal recessive RS (RRS OMIM#268310) have been described.[3] The recessive form was previously known as costovertebral segmentation defects with mesomelia.[4] Due to high degree of consanguinity, relatively more number of RRS have been described in Turkey, Pakistan, Czechoslovakia, and certain Arab countries.[3] In this article, a case of an autosomal RRS child has been described.

Case Report

A 5.5-year-old male child diagnosed as RRS reported to the Department of Pediatric Dentistry for the treatment of decayed teeth. The child weighed 10 kg and had an average IQ. His history revealed that he was the third child born to a healthy 26-year-old mother in a consanguineous marriage, delivered by cesarean section at 36 weeks of gestation.

The patient had a short stature [Figure 1a] and his skeletal findings revealed that the patient had mesomelic as well as rhizomelic limb shortening, indicative of dwarfism. His fingers and toes showed brachydactyly and clinodactyly with dysplastic toenails [Figure 1b and c].

The patient had multiple vertebral segmentation defects showing hemivertebrae, butterfly-shaped vertebrae, as well as wedge vertebrae, which resulted in scoliosis of the spine with convexity to the right side [Figure 2a]. Rib fusion (pathognomonic for RRS)[5] and dysplastic sacrum were also observed in the patient [Figure 2b].

As per the patient's medical records, the patient also had micropenis but showed the presence of the both testes in scrotal sac. Ultrasonography of the abdomen showed the presence of 6-mm cyst in the lower pole of right kidney. Two-dimensional echo and color Doppler study showed no evidence of any congenital heart disease.

The patient's craniofacial features were characterized by macrocephaly, frontal bossing, micrognathia, and low set ears. There were marked hypertelorism and mid-facial hypoplasia due to a depressed and flat nasal bridge along with anteverted nares, giving an appearance of fetal-facies [Figure 1a]. His eyes were prominent with wide and down-slanting palpebral fissures giving an appearance of pseudo-exophthalmos due to lower eye lid deficiency.

The patient showed a triangular mouth giving an inverted 'V' (tented) appearance. The palate was deep, narrow and showed submucous clefting [Figure 3a].

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Address for correspondence:
Dr. Priti Sushil Jain, Department of Pediatric and Preventive Dentistry, Nair Hospital Dental College, Mumbai, Maharashtra, India. E-mail: drpritisolanki@gmail.com

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The presence of ankyloglossia and a bifid tongue was distinct [Figure 3b]. The patient showed malaligned teeth with gingival hyperplasia [Figure 3a]. Dental findings included multiple carious teeth and fusion of left primary mandibular lateral incisor and primary mandibular canine [Figure 3b]. Primary maxillary right central incisor was discolored and nonvital due to trauma 1 year back [Figure 3a]. Orthopantogram revealed that all the permanent teeth were present, but their root formation was found to be delayed [Figure 3c].

The child was already under growth hormone therapy which was later discontinued as the results were unsatisfactory. The patient had been given injection testosterone 25 mg once a month for 3 months for hypoplastic genitalia, and calcium supplements were also instituted for the patient as a part of medical management. The patient was referred to the Department of Paediatric Medicine for general systemic evaluation and routine hematologic investigations were advised which were within normal limits.

According to Frankl's Behavior Rating Scale, the child's behavior was positive and it was observed that the child was comfortable during dental treatment using desensitization behavior management technique. Protective stabilization was provided in the form of cushions around the waist and lower back as the patient had scoliosis. Initially, scaling and polishing of teeth along with topical fluoride application using 1.23% acidulated phosphate fluoride gel was done. The carious teeth (64, 82, 83, fused teeth 72 and 73) were restored with glass ionomer cement and pulpectomy of 51 was performed followed by obturation with zinc oxide-eugenol cement and postendodontic composite restoration. The patient was advised extraction of 81, but the parents refused; hence, the tooth was kept under observation. Oral hygiene instructions were given along with dietary counseling to the patient and parents. The patient was scheduled for regular fluoride applications every 6 months. The rationale for future orthodontic treatment was also explained to them. The management of ankyloglossia was explained, but the parent refused as the patient currently did not demonstrate any difficulty in speech.

Discussion

Autosomal RRS is caused by mutation in ROR2 gene. ROR2 gene is a tyrosine kinase-like orphan receptor on the 9q22 chromosome. The receptor tyrosine kinases play an important role in the control of most basic cellular processes including cell proliferation, differentiation, migration, and metabolism.[6] Specifically, ROR2 protein plays an essential role in many aspects of embryonic development. The protein is reported to be involved in the early formation of the chondrocytes and is required for cartilage and growth plate development.[6] It also appears to be critical for the normal formation of the skeleton, heart, and genitals.[6]

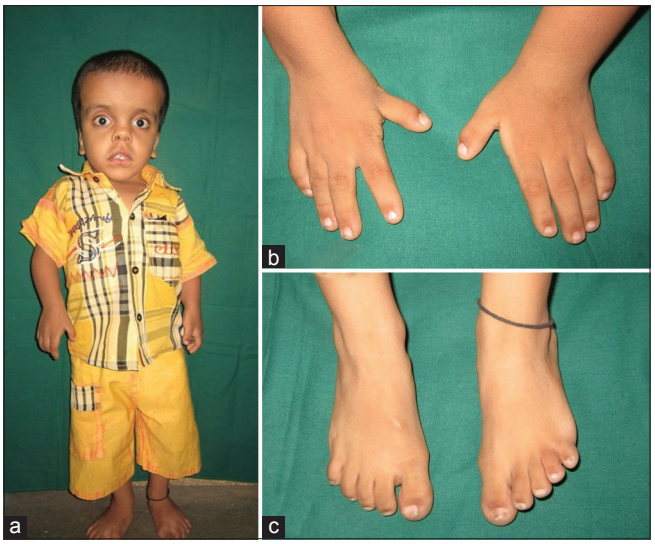

Figure 1: (a) Short stature, macrocephaly, and frontal bossing fetal-facies hypertelorism, pseudo-exophthalmos and mid-face hypoplasia, (b) brachydactyly and clinodactyly, (c) dysplastic nails of toes

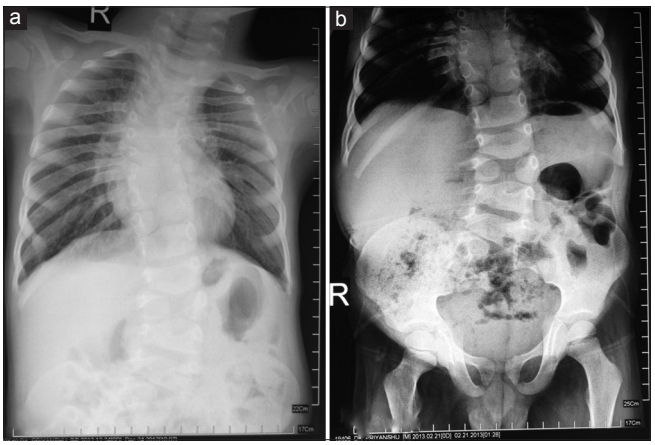

Figure 2: (a) Hemivertebrae, butterfly-shaped vertebrae, wedge vertebrae, rib fusion, (b) dysplastic sacrum

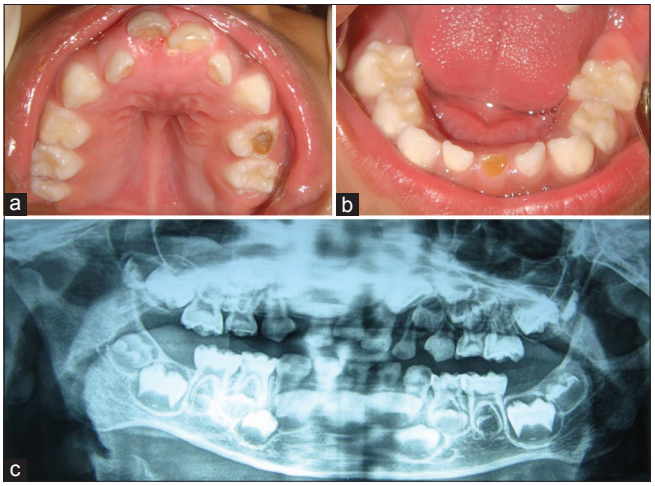

Figure 3: (a) Malaligned carious teeth and gingival hyperplasia, (b) fusion of left primary lateral incisor and canine and ankyloglossia, (c) orthopantogram showing delayed root formation of permanent teeth

Loss of function of ROR2 protein during early development disrupts embryonic development, leading to the skeletal abnormalities (characteristic of RS) and other phenotypic aspects of RS such as genital abnormalities and heart defects.

The presented case had most of the craniofacial and oral manifestations of RRS, craniofacial features being more prominent. This was similar to findings reported by Patton and Afzal and Beiraghi *et al*.[7,8] Craniofacial dysmorphology is more severe in RRS than DRS.[8] The craniofacial features of RRS also allows its differentiation from other syndromes of similar radiographic appearance such as Jarcho–Levin syndrome and spondylocostal dysostosis.[7]

Intraoral features such as crowding and malocclusion were predominant in the present case; however, it is reported to be more severe in DRS.[8] Gingival hyperplasia in the patient appeared more prominent due to short upper lip creating a gummy smile. Variations in the phenotypic characteristics of RS have been reported previously in literature such as short roots[9] and root malformations;[10] however, in this case, fusion of primary teeth in the lower arch was seen which gave an appearance of hypodontia in the lower arch. Fusion has not been reported previously in literature, and its association with RS is just a coincidence or an additional component of the syndrome needs to be evaluated, especially in cases where hypodontia have been reported.[8]

With such a wide array of clinical presentations, it is very important to understand the characteristic features of RS, for differentiation from other syndromes with similar radiographic and clinical presentations; to evaluate in the future whether fusion of primary teeth is a uniform feature observed in other RS cases; and to encourage the dental practitioners to provide optimal dental treatment for such patients.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients 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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