A case report on autosomal recessive Robinow syndrome

N. ERONAT¹, D. COGULU¹, F. OZKINAY²

ABSTRACT. Background Robinow syndrome or “foetal face” syndrome is an extremely rare genetic disorder with characteristic skeletal and orofacial findings. The purpose of the present case report is to describe the clinical findings of an 8 year-old female patient with autosomal recessive Robinow syndrome. Case report The patient was born to consanguineous parents and had anomalies typical of the recessive type of the syndrome such as short stature, mesomelic limb shortening, vertebral anomalies and dysmorphic facial features. Besides typical orodental findings, she also had root malformation in mandibular incisors, which is unusual finding of recessive type of Robinow syndrome. This case report emphasizes the importance of oral and dental manifestations of this syndrome.

Key words: Robinow syndrome; Autosomal recessive; Dental findings.

Introduction

Robinow syndrome, characterised by the “foetal face” appearance, mesomelic limb shortening, hemivertebrae and genital hypoplasia, was first described in 1969 [Robinow et al., 1969]. It is a genetic disorder with extremely variable appearances [Afzal et al., 2000b; Aksit et al., 1997; Atalay et al., 1993; Balci et al., 1993; Schorderet et al. 1992; Soliman et al., 1998; Teebi, 1990]. Both autosomal recessive and autosomal dominant inheritance have been described suggesting there is allelic heterogeneity [Palton and Afzal, 2002]. Robinow syndrome is very rare and estimated to have a frequency of 1:500,000 and a 1:1 male-to-female ratio [Robinow, 1990]. Parental age has not been found to be a significant factor in autosomal dominant form, although consanguinity seems to play an important role in the recessive form [Palton and Afzal, 2002].

The main clinical features of Robinow syndrome are the following.

- Craniofacial: frontal bossing, flat facial profile with large head, midfacial hypoplasia, hypertelorism, wide palpebral fissures, lower lid deficiency, ear abnormality, short upturned nose, broad nasal bridge.

- Oral: micrognathia, triangular mouth, long/short philtrum, crowding, congenitally missing teeth, gingival hyperplasia, abnormal uvula, cleft lip/palate, shortened tongue, ankyloglossia.

- Musculoskeletal: mild to moderate short stature (dwarfism), short lower arms, small hands with clinodactyly, small feet, vertebral defects, hemivertebrae, scoliosis.

- Genital/renal: hypoplastic genitalia, cryptorchidism, decreased fertility, renal tract abnormalities, hydronephrosis.

- Others: congenital heart disease, psychosocial problems related to appearance [Afzal et al., 2000b; Grothe et al., 2008; Robinow et al., 1969; Robinow, 1990; Robinow, 1993].

Intelligence is usually normal but developmental delay occurs in 10-15% of the cases. Macrocephaly is commonly found in Robinow syndrome and does not specifically indicate a risk factor for developmental delay [Guillen-Navarro et al., 1997].

Clinical presentation in autosomal recessive type of Robinow syndrome (RRS) tends to be more severe with fusions of vertebrae and ribs, hemivertebrae, radial head dislocation, and increased mortality [Patton and Afzal, 2002].

RRS is due to homozygous mutations in the ROR2 gene located on chromosome 9q22 [Afzal et al., 2000; van Bokhoven et al., 2000]. The RRS has been reported more often in societies that have high rates of consanguineous marriages such as Turkey, Pakistan, and a number of Arabic countries [Aksit et al. 1997; Balci et al., 1993; Glaser et al., 1989; Gulcan et al., 2005; Nazer et al., 1990].

The purpose of the present case is to describe the common and unusual orofacial findings in a female...
paediatric patient with autosomal recessive Robinow syndrome.

**Case report**

An 8-year-old female patient, previously diagnosed with Robinow syndrome was referred to our clinic in 2007 because of delayed eruption of the permanent teeth. She was born to a healthy 35-year-old mother at term after an uneventful pregnancy. She was the second child of consanguineous parents. The pedigree is given in Figure 1. The birth weight was 3200 g, length 46 cm and head circumference 34 cm.

At the age of 8 years her weight was 20 kg (at 10th percentile), height 103 cm (<3rd percentile), and head circumference 50 cm (2-50 percentile). She had a dysmorphic face characterised by frontal bossing, hypertelorism, remarkable exophthalmus, flat-saddle nose, anteverted nostrils, long philtrum, mesomelic shortening of the extremities and brachydactyly of the fingers (Fig. 2, 3). Abdominal, and renal ultrasound examinations and echocardiography were normal. She was mentally normal. Complete blood count, urine analysis and serum electrolyte levels were normal. Cytogenetic analysis revealed 46, XX karyotype. Homozygous loss-of-function mutations in the gene encoding receptor orphan receptor tyrosine kinase 2 (ROR2), located on chromosome 9q22 (the long arm of chromosome 9) were detected.

Oral examinations showed an absent uvula, bilobed tongue tip, gingival hypertrophy, high frenum attachments, operated cleft palate, ankyloglossia, delayed tooth eruption and exfoliation, crowding, microodontia and micrognathia (Fig. 4). Panoramic radiography revealed congenitally missing teeth: maxillary left and mandibular right and left permanent second molar teeth (Fig. 5). The teeth 74 and 84 were previously restored with resin restorations. Additionally, root malformations of mandibular permanent incisors were seen which are not usual features of the syndrome (Fig. 6, 7).

---

**FIG. 1** - The genealogy of the family.

**FIG. 2, 3** - The patient at the age of 8 years.

**FIG. 4** - Oral findings of the patient at the first visit.

**FIG. 5** - Panoramic radiograph of the patient.

**FIG. 6** - Periapical radiograph of the mandibular front region.
Clinical treatment

Informed consent was obtained from the parents. The dental treatment was planned as extraction of the carious maxillary primary central incisors and right lateral incisor in the first visit. The patient was instructed for oral hygiene, and sugar intake was limited for preventing caries. Topical fluoride application was performed and the patient was scheduled for regular fluoride applications every 6 months. Six-month follow-up of the patient revealed the continuous eruption of the maxillary left permanent central incisor (Fig. 8).

Discussion

The patient presented here was the second child of the parents with a history of consanguinity and none of the family members showed similar features of this syndrome. She had almost all characteristic craniofacial and oral manifestations of RRS with additional findings of root malformations in mandibular incisors. As reported in previous studies, typical oral and facial features of Robinow syndrome made possible the diagnosis of this rare condition. In the literature, a case with short tooth roots without root malformation has also been reported previously [Kantaputra et al., 1999].

The dental treatment proposed for the patient was the extraction of the carious maxillary primary central incisors and right lateral incisor and application of topical fluoride for prevention. Follow-up visits were scheduled to allow the continuous monitoring of dental development. The parents were advised of the possible need for orthopaedic/orthodontic treatment and plastic surgical procedure for facial reconstruction in case of physical and/or psychological problems caused by the patient’s craniofacial anomalies as previously reported in the literature [Cohen et al., 1997]. The hyperplastic gingival tissues will be rehabilitated in this case. The parents were informed about the unusual finding of this syndrome: root malformations that may produce complications during orthodontic treatment.

Robinow syndrome is an extremely rare congenital genetic disorder with multiple orofacial and dental anomalies which is usually diagnosed early in the childhood. The paediatric dentist may play an important role in the multidisciplinary treatment approach of this syndrome.

Conclusion

This case report highlights the common and unusual clinical findings, multidisciplinary dental management considerations, and genetic information necessary to knowledgeably care for dental patients with Robinow syndrome.

References