J Istanbul Univ Fac Dent 2017;51(2):43-48.
http://dx.doi.org/10.17096/jiufd.86251 *CASE REPORT*

*DENTAL MANAGEMENT AND OROFACIAL MANIFESTATIONS OF A PATIENT WITH ROBINOW SYNDROME**

Robinow Sendromlu Bir Hastanın Orofasiyal Bulguları ve Dental Tedavisi: Olgu Sunumu

Adil BAŞMAN [1], Gülsün AKAY [2], İlkay PEKER [2] ,Kahraman GÜNGÖR [2] , Zühre AKARSLAN [2], Suat ÖZCAN [3] ,Cemile Özlem ÜÇOK [2]

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ABSTRACT

Robinow syndrome (RS) is an extremely rare condition. Characteristic craniofacial findings of RS include a fetal facial appearance, ear abnormalities and oral findings. The aim of this case report was to evaluate the oral findings of a 26-year-old man with RS and to describe the dental treatments performed. The patient had short stature, vertebral anomalies, short and broad fingers, a fetal facial appearance, gingival hyperplasia, fissured tongue, caries and multiple impacted teeth. Periodontal and restorative dental treatments were performed under aseptic conditions with due precautions. No surgical treatment was performed to the impacted teeth because of the lack of symptoms.

ÖZ

Robinow sendromu (RS) oldukça nadir görülen bir hastalıktır. RS'nin karakteristik kraniyofasiyal bulguları; fetal yüz görünümü, kulak anomalileri ve oral bulguları içermektedir. Bu olgu sunumunun amacı RS olan 26 yaşındaki erkek hastanın orofasiyal bulgularını ve uygulanan dental tedavileri değerlendirmektir. Hastada kısa boy, vertebral anomaliler, kısa ve geniş parmaklar, fetal yüz görünümü, gingival hiperplazi, fissürlü dil, çürükler ve gömülü dişler izlenmiştir. Aseptik koşullarda ve gerekli önlemler alınarak hastanın, periodontal ve restoratif diş tedavileri yapılmıştır. Asemptomatik olmaları nedeniyle gömülü dişlere herhangi bir cerrahi tedavi uygulanmamıştır.

Keywords: *Robinow syndrome; gingival hyperplasia; gingivectomy; vestibuloplasty; impacted teeth*

Anahtar kelimeler: *Robinow sendromu; gingival hiperplazi; gingivektomi; vestibuloplasti; gömülü dişler*

[1] Department of Periodontology Faculty of Dentistry Gazi University
[2] Department of Dentomaxillofacial Radiology Faculty of Dentistry Gazi University
[3] Department of Restorative Dentistry Faculty of Dentistry Gazi University
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Introduction

Robinow and colleagues, was first described Robinow Syndrome (RS) in 1969 (1, 2). Prevalence of this rare syndrome has been reported as 1:500.000 but it is particularly higher in some regions of Turkey, Oman and the Czech Republic. This situation may be associated with the high percentage of consanguineous marriages in these countries (3). As a genetic disorder, RS may be inherited in an autosomal dominant or autosomal recessive manner, referred to as dominant Robinow Syndrome (DRS) or recessive Robinow Syndrome (RRS), respectively (1, 3). Dysmorphology, especially of the musculoskeletal system, is more severe in RRS than in DRS (1, 3). The disease affects many parts of the body, including the cardiovascular, skeletal and urogenital systems, cranium and face (4). Cardiovascular abnormalities consist of atrial septal defect, ventricular septal defect, aortic coarctation, Fallot tetralogy, and tricuspid atresia (3). Renal tract abnormalities and cystic dysplasia of the kidneys and genital hypoplasia were also reported (3). Skeletal abnormalities include vertebral malsegmantation, short stature, marked mesomelic limb shortening, rib fusion and hemivertebrae (5). Craniofacial features consist of a wide forehead, hypertelorism, midface hypoplasia, flattened and widened nose, depressed nasal bridge, down-slanted mouth corners, low-set ears, micrognathia and triangular mouth (3). Common oral findings include gingival hyperplasia, tongue anomalies, dental anomalies and dental crowding (4-6). Although RS indicates various orofacial and dental characteristics, published reports in literature regarding the findings and dental treatment protocol of these cases are relatively few (4, 7-10). Therefore, the aim of this case report was to present a case of RS with multiple misaligned impacted teeth and to describe the dental treatment of the patient.

Case Report

A 26-year-old male applied to the Department of Dentomaxillofacial Radiology of the Gazi University Faculty of Dentistry with complaints of gingival hyperplasia and dental caries. Medical anamnesis revealed that the patient had been diagnosed with RS (recessive type) at childhood (10 years-old). His parents were consanguineous. His three older siblings were unaffected. His mental development was normal. The patient reported the presence of mitral valve disease, hypertension, restrictive lung disease, glaucoma and loss of sight. He was taking antihypertensive, anticoagulant and antiarrhythmic drugs. Physical examination revealed a short stature, vertebral anomalies, scoliosis, mesomelic limb shortening, short and broad fingers, macrocephaly and a dysmorphic face (Figure 1), characterized by a prominent forehead, hypertelorism, flattened and widened nose and macrocheilia. The eyes were prominent and bilateral proptosis was observed. Intraoral examination showed insufficient width of an attached gingiva, gingival hyperplasia, macroglossia, cleft tongue, dental crowding, malocclusion, anterior cross-bite and caries (31, 32, 41, 42, and 35). The permanent maxillary and mandibular molars were absent. The patient had anterior and posterior cross-bites, a lack of interocclusal dental contacts and poor oral hygiene. (Figure 2). All procedures followed were in accordance with the ethical standards of the responsible committee on human experimentation (institutional and national) and with the Helsinki Declaration of 1964 and later versions. Additional informed consent was obtained from the patient for whom identifying information is included in this case report.

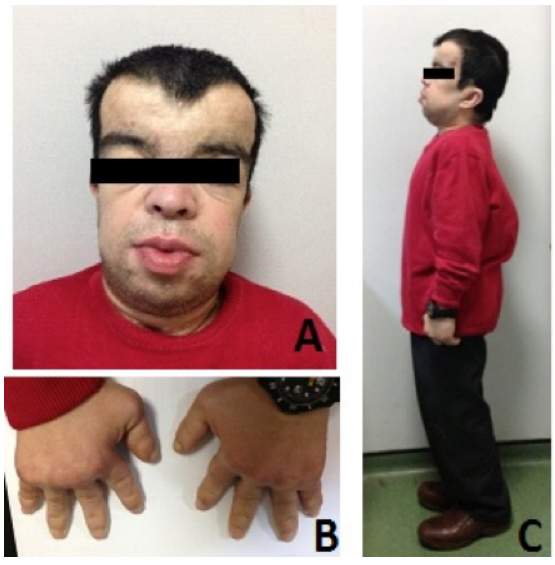

Figure 1. *Extraoral examination of the patient revealed a dysmorphic facial appearance, with (A) macrocephaly, prominent forehead, hypertelorism, flattened and widened nose and macrochelia, (B) short and broad fingers, (C) short stature, vertebral anomalies and scoliosis.*

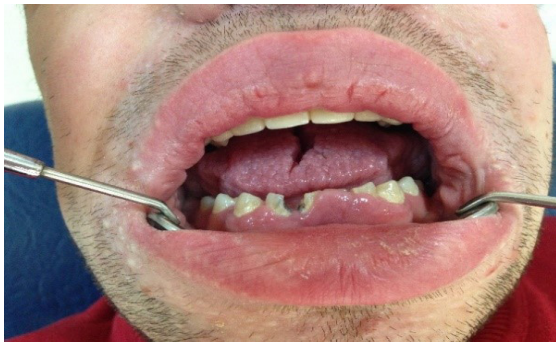

Figure 2. *Intraoral photograph of the patient before dental and periodontal treatment.*

Panoramic radiography revealed nine misaligned impacted teeth, including four mandibular and five maxillary molars and dental caries (Figure 3). Cone-beam computed tomography examination was carried out to assess the impacted teeth. The patient displayed two molar teeth with occlusal surfaces showing partial contact in the posterior region of the right quadrant of the maxilla, as well as three molar teeth having partial contact of their crowns in the left quadrant of the maxilla. (Figure 4) The mandible also exhibited bilateral impacted molars with occlusal contact (Figure 5).All radiographic examinations were carried out by oral and maxillofacial radiologists (IP, KG and ZA). The craniofacial and oral findings of the patient and cases reported in the literature are shown in Table 1.

The patient was referred to the Departments of Periodontology, Restorative Treatment and Oral Surgery for dental treatment. Periodontal and restorative treatments were planned to be performed under aseptic conditions taking due precautions. No surgical treatment was planned for the asymptomatic multiple misaligned impacted teeth, although periodic follow ups for these teeth were advised.

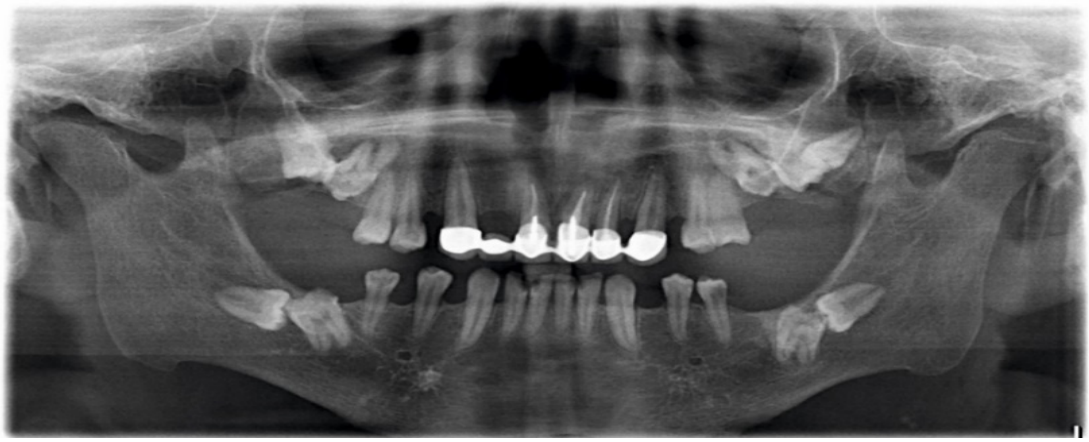

Figure 3. *Panoramic radiograph revealing dental caries and nine misaligned impacted teeth.*

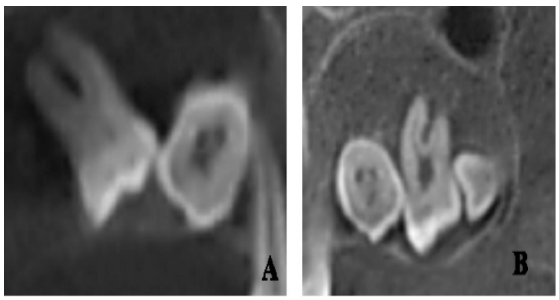

Figure 4. *Cone-beam computed tomography images of the maxilla, showing (A) two impacted molar teeth in the posterior region of the right quadrant and (B) three molar teeth in the left quadrant.*

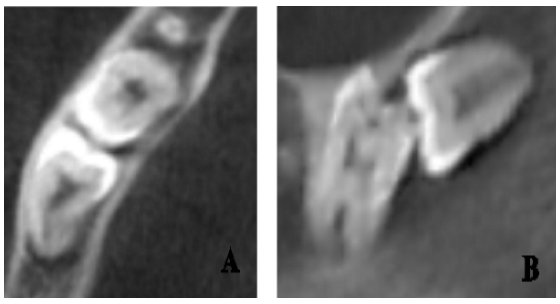

Figure 5. *Cone-beam computed tomography images of the mandible (A) in the right and (B)left quadrant.*

Table 1. *Craniofacial and oral features associated with RRS and DRS.*

CRANIOFACIAL FINDINGS	Mazzeu *et al.*(6) DRS(%)	RRS(%)	Beiraghi *et al.*(4) DRS(%)	RRS(%)	Present case RRS
Frontal bossing	79	78	33	67	+
Orbital hypertelorism	100	100	78	100	+
Midface hypoplasia	81	94	33	67	+
Depressed nasal bridge	78	49	44	100	+
Wide nasal bridge	97	95	89	100	+
Short nose	81	93	100	100	+
Upturned nose	87	97	78	100	+
Anteverted nares	96	96	89	100	+
Triangular mouth	65	86	44	100	+
Down-slanted mouth corners	63	95	44	100	+
Micrognathia	57	68	22	0	-
Retrognathia	44	37	33	33	-
ORAL FINDINGS					
Bifid tongue	39	59	67	67	Fissured tongue
Gingival hyperplasia	36	71	78	67	+
Dental malocclusion	49	94	89	67	+
Abnormal uvula	N/A	N/A	89	67	+
Wide retromolar ridge	N/A	N/A	100	33	-
Dental crowding	N/A	N/A	78	33	-
Flat palate	N/A	N/A	67	100	+
Short palate	N/A	N/A	67	67	-
Delay of dental eruption	N/A	N/A	56	67	+
Highly arched palated	51.5	14	0	0	+
Cleft lip/palate	35	13.5	56	0	-
Microdontia	N/A	N/A	0	0	-
Supernumerary teeth	N/A	N/A	0	0	-

N/A: Not available.
DRS: Dominant Robinow Syndrome
RRS: Recessive Robinow Syndrome

Before undergoing dental procedures, the patient was seen by a cardiologist and a pulmonologist because of his medical status. Full mouth scaling and root planning were performed and oral hygiene instructions (tooth brushing, interdental brushing and mouthwash) were given. Clinical periodontal parameters in the gingival enlargement areas were recorded before the treatment. Then, full mouth periodontal phase 1 treatment was completed and maintenance therapy was applied to the patient before the gingivectomy operation. Gingivectomy was performed with concomitant oxygen delivery by a standard nasal cannula. During periodontal treatment practice (Figure 6), the increase of the insufficient keratinized mucosa is considered an effective strategy in the prevention of advance periodontal diseases. Vestibuloplasty was carried out to augment the keratinized gingiva. The surgical site was covered by a periodontal pack. After 9 months, the gingiva appeared to be healthy, possibly due to good oral hygiene. After the healing phase, the patient underwent restorative dental treatment. Oral hygiene instructions (tooth brushing, interdental brushing and usage of mouthwash) were given and frequent dental visits (three times per year) were advised. At the 9 months follow-up visit, the patient's periodontal tissues did not show any signs of hyperplasia. All impacted molars were asymptomatic (Figure 7).

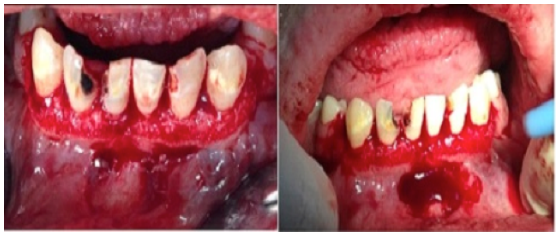

Figure 6. *Intraoral photographs of the patient during periodontal surgery.*

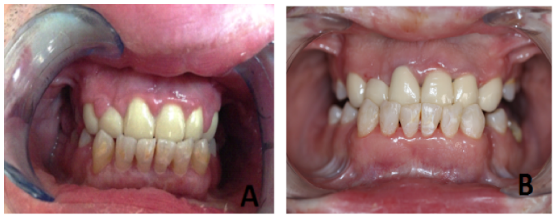

Figure 7. *Intraoral photographs of the patient (A) after and (B) 9 months after dental and periodontal treatment.*

Discussion

Higher prevalence of RS was reported in the regions of Turkey, Oman and the Czech Republic. This may be related to the high degree of consanguineous marriages in these countries (3). Diagnosis of RRS is not difficult because of the typical facial appearance, mesomelic limb shortening, costa and vertebral segmentation defects and genital hypoplasia (4, 5, 11). In contrast, clinical diagnosis of DRS can be difficult, because the musculoskeletal system is often minimally affected in this condition. In the present case, typical craniofacial findings of RRS were present. Patients with DRS and RRS usually have normal mental development, although developmental delays may be present in 10–15% of cases (3). The patient had normal mental development. Triangular mouth, lip/palate cleft, abnormal uvula, tongue anomalies (macroglossia, ankyloglossia, shortened tongue and bifid tongue) gingival hyperplasia, malocclusion and hypodontia has been reported in RS (3-5). Gingival hyperplasia and hypertrophy are often described as the major oral manifestations of RS. Dental crowding and irregular teeth have been observed in both deciduous and permanent dentitions (4). Kantaputra *et al.* (12) reported some new features, such as short-rooted teeth and narrow and thick-floored pulp chambers, in a male patient with DRS. Beiraghi *et al.* (4) reported the presence of anterior open-bite, deep-bite, as well as anterior and posterior cross-bites in RS. In the present case, triangular mouth, fissured tongue, macroglossia, gingival hyperplasia and anterior cross-bite were observed. Some authors reported delay of dental eruption in the patients with RS (8, 12). Grothe *et al.* (8) reported impacted maxillary incisor teeth in a 15-year-old female patient with RRS. Kantaputra *et al.* (12) reported eight misaligned impacted permanent molars (four in the maxilla and four in the mandible) in a 11-year-old male patient with DRS. In the present case, nine misaligned impacted molars; five in the maxilla and four in the mandible, were observed.

Surgical difficulties play an important role in the preoperative planning of impacted teeth. Several factors, such as the relative depth, relationship of the tooth to the ascending ramus, number, angulation/form of roots and their proximity to the inferior alveolar canal, and a lack of periodontal membrane space, all need to be carefully evaluated because they influence the treatment outcomes. Additionally, owing to the nature of impaction, general anesthesia could be necessary for extraction of impacted teeth (13, 14). In this case, we decided to follow up the misaligned impacted molars due to systemic condition of the patient and surgical difficulties. RS patients may have several systemic diseases (3). Thus, medical consultation of the patients is required prior to dental treatment. In the present case, the patient was consulted by a cardiologist and a pulmonologist. According to the consultation, gingivectomy was performed under oxygen delivery with a standard nasal cannula. Chronic inflammatory gingival enlargement originates as a slight ballooning of the interdental papilla and marginal gingiva. In the early stages, it produces a life preserver shaped bulge around the involved teeth. This bulge can increase in size until it covers part of the crowns. The enlargement may be localized or generalized and progresses slowly and painlessly, unless it is complicated by acute infection or trauma. Prolonged exposure to dental plaque may result in chronic inflammatory gingival enlargement (15). Factors that favor plaque accumulation and retention include poor oral hygiene, irritation of periodontal tissues by anatomic abnormalities and improper restorative and orthodontic treatment (15).

To assess the effectiveness of their plaque control, patients should perform their hygiene regimen immediately before the recall appointment. Plaque control must be reviewed and corrected until the patient demonstrates the necessary proficiency, even if additional instruction sessions are required. Patients instructed in plaque control have less plaque and gingivitis than uninstructed patients,

and the amount of supragingival plaque affects the number of subgingival anaerobic organisms (16). Therefore, dental plaque should be removed and chronic gingival enlargement should be treated with periodontal treatment in RS. Periodontal surgery, good oral hygiene practice and regular dental visits showed successful results for the treatment of gingival hyperplasia in the present case.

Conclusion

This case report adds information about misaligned impaction of the molar teeth to the oral findings of RS in the literature. Gingivectomy is useful for the treatment of gingival hypertrophy. Clinicians should be aware of the findings of RS and should take proper precautions during dental treatment.

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Conflict of Interest
None Declared

References

1. Robinow M. The robinow (fetal face) syndrome: A continuing puzzle. Clin Dysmorphol 1993;2(3):189-198.
2. Robinow M, Silverman FN, Smith HD. A newly recognized dwarfing syndrome. Am J Dis Child 1969;117(6):645-651.
3. Patton MA, Afzal AR. Robinow syndrome. J Med Genet 2002;39(5):305-310.
4. Beiraghi S, Leon-Salazar V, Larson BE, John MT, Cunningham ML, Petryk A, Lohr JL. Craniofacial and intraoral phenotype of robinow syndrome forms. Clin Genet 2011;80(1):15-24.
5. Cerqueira DF, de Souza IP. Orofacial manifestations of robinow's syndrome: A case report in a pediatric patient. Oral Surg Oral Med Oral Pathol Oral Radiol Endod 2008;105(3):353-357.
6. Mazzeu JF, Pardono E, Vianna-Morgante AM, Richieri-Costa A, Ae Kim C, Brunoni D, Martelli L, de Andrade CE, Colin G, Otto PA. Clinical characterization of autosomal dominant and recessive variants of robinow syndrome. Am J Med Genet A 2007;143(4):320-325.
7. Eronat N, Cogulu D, Ozkinay F. A case report on autosomal recessive robinow syndrome. Eur J Paediatr Dent 2009;10(3):147-150.
8. Grothe R, Anderson-Cermin C, Beiraghi S. Autosomal recessive robinow syndrome: A case report. J Dent Child (Chic) 2008;75(1):48-54.
9. Horbelt CV. Robinow syndrome, cockayne syndrome, and pfeiffer syndrome: An overview of physical, neurological, and oral characteristics. Gen Dent 2010;58(1):14-17.
10. Israel H, Johnson GF. Craniofacial pattern similarities and additional orofacial findings in siblings with the robinow syndrome. J Craniofac Genet Dev Biol 1988;8(1):63-73.
11. Al Kaissi A, Bieganski T, Baranska D, Chehida FB, Gharbi H, Ghachem MB, Hendaoui L, Safi H, Kozlowski K. Robinow syndrome: Report of two cases and review of the literature. Australas Radiol 2007;51(1):83-86.
12. Kantaputra PN, Gorlin RJ, Ukarapol N, Unachak K, Sudasna J. Robinow (fetal face) syndrome: Report of a boy with dominant type and an infant with recessive type. Am J Med Genet 1999;84(1):1-7.
13. Gbotolorun OM, Arotiba GT, Ladeinde AL. Assessment of factors associated with surgical difficulty in impacted mandibular third molar extraction. J Oral Maxillofac Surg 2007;65(10):1977-1983.
14. Jannu AA BS, Vivek GK, Veena GC, Kamath R. Missing molars caught kissing. J Dent Med Sci 2014;13:51-54.
15. Carranza FA, Hogan EL. Gingival Enlargement. In: Newman MG, Takei HH, Klokkevold PR, Carranza FA, editors. Carranza's Clinical Periodontology. 11th Ed.,St.Louis,Missouri: Elsevier Saunders, 2012, p.84-96.
16. Merin RL. Supportive periodontal treatment. In: Newman MG, Takei HH, Klokkevold PR, Carranza FA, editors. Carranza's Clinical Periodontology. 11th Ed.,St.Louis,Missouri: Elsevier Saunders, 2012, p.746-755.

Corresponding Author:
Adil BAŞMAN
Department of Periodontology
Faculty of Dentistry, Gazi University
06560, Emek-Çankaya-Ankara/Turkey
Phone: +90 312 203 42 52
e-mail: adilbasman@hotmail.com