Case Reports: Rieger Syndrome

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Abstract:

Background: Rieger syndrome is a rare autosomal dominant condition with at least three genetic forms. The main symptoms are umbilical cord anomalies, malformations of the anterior chamber of the eye, agenesis of certain teeth, and a hypoplastic mid-face. Case reports: In this paper three cases of Rieger syndrome are presented, focusing in particular on dental and craniofacial findings. Treatment: Treatments have been individualized and include temporary dentures and restoration of primary teeth to preserve them until their successors erupt. Follow-up: Patients with Rieger syndrome should be followed according to an individualized plan, depending on the severity of dental symptoms and general caries risk. Conclusion: It is important that the dental team have knowledge about this syndrome, as ocular complications can be prevented if the diagnosis is made early.

Introduction

Rieger syndrome (RS) is a rare, autosomal dominant condition with almost complete penetrance and variable expressivity. The syndrome is characterized by periumbilical, ocular, craniofacial, and dental abnormalities [Winter and Baraitser, 1996]. Failure of involution of periumblical skin has been reported to be a cardinal symptom [Jorgensen et al., 1978]. Ocular features comprise iris stromal hypoplasia causing the eyes to appear dark, and strands of iris tissue crossing the anterior chamber angle. Schwalbes line may be anteriorly displaced. Craniofacial features are dominated by an underdeveloped premaxilla and a relative mandibular prognathism. Oral anomalies comprise hypodontia, especially of maxillary front teeth both in the primary and the permanent dentitions, small teeth, peg-shaped front teeth, and hyperplastic upper labial frenulum. Other teeth may also be missing.

To date, RS has been associated with the genes PITX2 on chromosome 4q25 [Semina et al., 1996] and FOX-C1 on chromosome 13q14 (Phillips et al, 1996). Also one case report describing an association between the syndrome and PAX6 on chromosome 11p13 has been published [Riise et al., 2001]. This case is identical to one of the cases (Case 1) described in this report. Three cases seen at the TAKO-centre, Lovisenberg Diakonale Hospital, Oslo, Norway are presented. The TAKO-centre is a national resource centre aiming to provide multidiciplinary expertise in diagnostics and oral treatment planning in persons with rare medical conditions. Written consents to publish all photographs have been obtained.

Case reports

Case 1. A 14-year-old girl with healthy, unrelated parents had been diagnosed and treated for characteristic ocular features since she was 6 weeks old. There was also failure of involution of the periumblical skin. As she grew older, the ophthalmologist recognized her flat mid-face and missing maxillary central incisors. She was referred to the TAKO-centre when she was seven years of age for an examination with regard to a possible syndrome diagnosis. At that time she had the diagnosis partial aniridia. She had a retrognathic premaxilla and a mixed dentition with peg-shaped 52 and 62 as the only maxillary incisors present (Fig. 1a). A detailed medical and dental history revealed that teeth 51 and 61 had always been missing. Erupted permanent teeth were: 16, 26, 36, 32, 31, 41, 42 and 46. The lower incisors were peg-shaped and slender, and all teeth were small. The patient had an open bite with unilateral cross-bite. The palate was shallow and the maxillary labial frenulum was hyperplastic. Orthopantomogram and periapical radiographs revealed agenesis of 7 permanent maxillary teeth: 15, 13, 12, 11, 21, 23, and 25 (Fig. 1b).

Key words: Rieger syndrome, hypodontia, frenulum, treatment
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Treatment. Frenectomy was performed when she was 8 years old. After eruption of 22, composite restorations of 52 and 22 were made. Composite resin was also used on the buccal surfaces of 53 and 63 for retention of a partial denture, which replaced the central incisors. The patient was satisfied with the aesthetic result (Fig. 2a).

Follow-up. After completed treatment she has been seen annually in our clinic. A new partial denture was made after one and a half years due to loss of the first one. All permanent teeth are now erupted, but maxillary growth has been slow (Fig 2b), and the denture is still in use. Future planned treatment includes orthodontic therapy starting when 52 is lost, and 22 will be moved distally to give more room for replacement of 12, 11 and 21. Remaining primary teeth will be kept as long as possible. Dental implants are the most likely treatment for the patient at the age of 18-20 years of age and it may be necessary to perform orthognatic surgery because of the relative mandibular prognathism.

Case 2. A local dental hygienist participating in a post-graduate program first suspected a syndrome diagnosis in a 12-year-old boy and consulted staff at the TAKO-centre. He had previously been diagnosed with oligodontia (8 teeth missing), malocclusion, hypoplastic premaxilla (Fig. 3a), and hyperplastic upper labial frenulum by the local dentist. Earlier records from dental visits were obtained and sent to us. These included clinical photographs and an orthopantomogram. The radiographs revealed agenesis of 17, 12, 11, 21, 22, 25, 35, and 45 (Fig. 3b). His facial appearance and his dark coloured eyes suggested Rieger syndrome. He was also using glasses. Further information was obtained by the local dental hygienist and included reports of a rapid decline in sight. Despite this he had not been seen by an ophthalmologist for several years. He had been operated on for hernia umbilicatis when he was 3 months old.

Based on information revealed in taking the medical history and clinical findings, he was further referred to an ophthalmologist who reported hypoplasia of the iris stroma and an asymmetric pupil, thereby confirming the tentative diagnosis of Rieger syndrome. His intraocular pressure is now being measured annually, as glaucoma is a common complication in this condition.

The patient was also referred to a geneticist for consultation and genetic analysis. Results have still not been received.

Treatment. The patient has only been seen once at our clinic, at age 13 years, after the diagnosis had originally been made. At this time teeth 13 and 23 were erupting, 23 was erupting mesially for 63 and 53 had exfoliated. He had tried to use a partial denture, but it did not fit well. The maxillary labial frenulum was hyperplastic (Fig 3c). All teeth were small. He had a bilateral cross-bite and mesial occlusion with negative overbite of 4mm. OPG revealed taurodontia of all permanent molars. The roots on 75 and 85 were short.

Follow-up. The following recommendations were given based on the clinical findings. At this stage of tooth development it is challenging to make a successful denture. However, this was recommended after a frenectomy, which was necessary as he could not pout his lips, and the frenu-