Rieger Syndrome

Clinical Presentation of 3 Cases

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Introduction

Rieger syndrome (RS) is a rare, autosomal dominant condition with almost complete penetrance and variable expressivity. The syndrome is characterized by ocular, dental, skeletal and periumbilical abnormalities.

The incidence is estimated to be 1/200.000 persons, and about 30 % of the cases are new mutations.

To date Rieger syndrome has been associated with mutations in genes at chromosomes 4q25 (PITX2), 6p25 (FOX-C1), 13q14 and on chromosome 11 (PAX6). The association to the PAX6 gene on chromosome 11 with Rieger syndrome was first discovered on one of the patients presented (case 1).

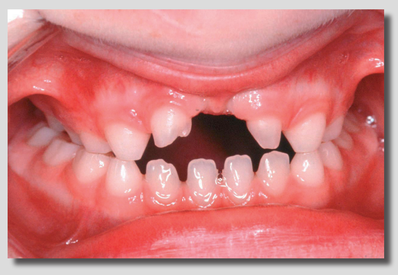
General symptoms

Ocular features comprise iris stromal hypoplasia and strands of iris tissue crossing the anterior chamber angle. Schwalbes line may be anteriorly displaced.

Cranio-facial anomalies are dominated by an underdeveloped premaxilla and a relative mandibular prognathism.

Dental anomalies comprise hypodontia, especially of maxillary front teeth in both primary and permanent dentition. Other teeth can also be missing.

Present teeth are small, and front teeth may be peg-shaped. Very characteristic is the hyperplastic frenulum labiale superior.



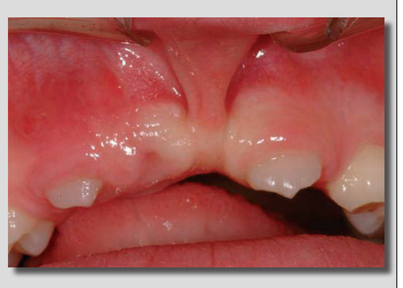
Redundant periumbilical tissue, with failure of involution.

The dentist's part in setting the diagnosis

Hypodontia (congenital absence of one or more teeth) may be the first recognizable symptom of Rieger syndrome.

First molars, maxillary central incisors and canines are regarded to be stable teeth. We therefore should have RS in mind, when central maxillary incisors are missing.

Ocular complications can be prevented with early



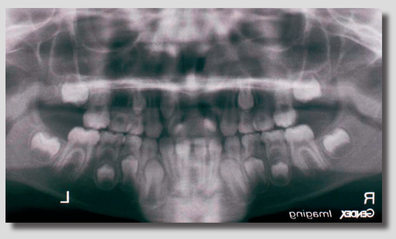
interventions. Therefore it is important that dentists are aware of the general symptoms of the syndrome. Glaucoma is estimated to occur in 50 % of patients by the age of 20 years.

Case presentations

1 Referred, 7 years old, from ophthalmologist

Girl born in 1992. Unrelated, healthy parents. Ocular features, characteristic for RS, were diagnosed 6 weeks old. Therapy was started, and intraocular pressure has been measured regularly. As the girl grew older the ophthalmologist recognized a flat midface and missing maxillary central incisors. The patient was then referred to the TAKO-centre for assessment.

Oral manifestations
Underdeveloped premaxilla. Primary teeth: Agenesis of 51 and 61 and hyperplastic frenulum labiale superior. Permanent teeth: Erupted lower incisors (peg-shaped) and small 1st permanent molars. Agenesis of 7 teeth: 15, 13, 12, 11, 21, 23, 25.



Treatment
Frenulectomi of frenulum labiale superior. 52 and 22 (peg-shaped): Rebuilding with composite. Partial denture replacing 11 and 21 (2001).

Follow-up, short-term
Maxillary growth has been slow, and the denture is still in use. All permanent teeth are now erupted (2006).

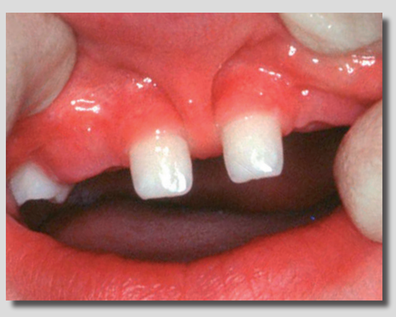
Follow-up, long-term
Prosthetic therapy, eventually combined with orthodontic treatment. Cranio-facial surgery will also be discussed in the treatment planing. Dental implants is the most likely treatment for the patient at the age of 18-20 years of age.

Conclusion
The oral manifestations confirmed the tentative clinical diagnosis Rieger syndrome. Later the genetic molecular result by use of FISH analysis showed a small deletion for the PAX6 gene one chromosome 11.

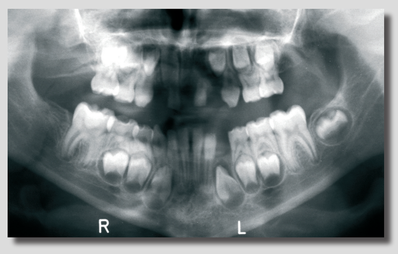
2 Referred, 2 years old, on family's initiative

Boy, born in 1998. Unrelated parents; his mother had recently realized that she had a syndrome, probably Rieger. She had all the typical symptoms of the syndrome, but without anyone mentioning this. She wanted to have her son examined because he had the typical dark eyes, like herself, and 4 teeth were missing in the maxillary front.

Oral manifestations
Underdeveloped premaxilla. Hyperplastic frenulum labiale superior and non-erupted 53, 52, 62, 63. Occlusal x-ray revealed agenesis of 52 and 62. We also noticed only one permanent central incisor germ.



Follow-up treatment
The boy has been seen regularly, and no treatment is given so far. He is late in tooth development. Orthopantomogram, 8 years old, reveals agenesis of following teeth: 17, 16, 12, 21, 22, 26, 27, 47.

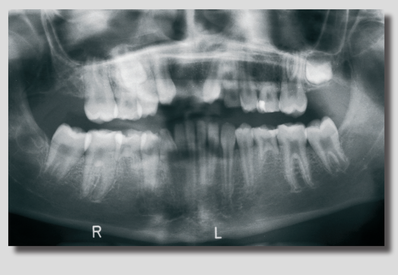


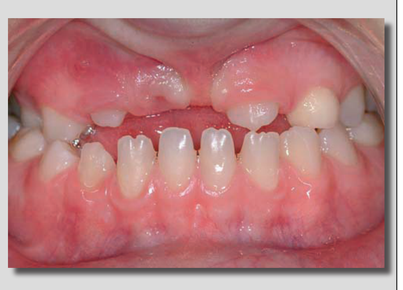
Conclusion
Results of the dental assessment were in agreement with symptoms of Rieger syndrome. The boy was referred to an ophthalmologist, who confirmed the clinical diagnosis.

3 Referred, 13 years old, from dental hygienist

Boy, born in 1992. Unrelated, healthy parents. Medical history: Hernia umbilicalis operated 3 weeks old, wearing glasses, multiple agenesis.

Oral manifestations
Underdeveloped premaxilla. Agenesis of 12, 11, 21, 22, 25, 35, 45. Microdontia. Hyperplastic frenulum labiale superior.





Conclusion
Dento-facial findings were in accordance with Rieger syndrome symptoms. Referral to ophthalmologist concluded with ocular findings confirming the clinical diagnosis.

Conclusions

- Dental professionals have a responsibility in setting the diagnosis Rieger syndrome.
- Dental characteristics may be the first recognizable symptoms of the syndrome.
- Early diagnosis is important since subsequent ocular complications may be prevented.
- Caries prophylaxis has highest priority.
- Interdisciplinary cooperation is essential in giving the best follow-up and treatment.

References

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