**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.

To date Rieger syndrome has been associated with mutations in genes at chromosomes 4q25 (PITX2), 6p25 (FOX1C1), 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 irides ternal hypoplasia and strands of iris tissue crossing the anterior chamber angle. Schwalbe’s 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 may 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.

**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 intracanal pressure has been measured regularly. As the girl grew older the ophthalmologist recognized a flat midface and no treatment is given so far.

**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.

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, but without anyone mentioning this. She wanted to have her son examined because he had the typical dark eyes, like her self, and 4 teeth were missing in the maxillary front.

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3. **Referred, 13 years old, from dental hygienist**


**DIAGNOSIS**

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**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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