UNIMAXILLARY DISTURBANCE OF TOOTH DEVELOPMENT AS A FEATURE OF ORAL-FACIAL-DIGITAL SYNDROME TYPE I - A CLINICAL CASE REPORT

C. BOYE KJØBSTED1, A.M. BERGMANN LØVSCHALL2, O. MOHR HOVGAARD1, H. LØVSCHALL1

1DEPARTMENT OF DENTISTRY AND ORAL HEALTH, AARHUS UNIVERSITY, 2AARHUS MUNICIPAL DENTAL CARE, 8000 AARHUS, DENMARK.

INTRODUCTION
Oral–Facial–Digital Syndrome Type I (OFD1) is an X-linked dominant disorder. Found to be caused by mutations in the CXORF5 gene. Nearly all carriers are female, and 75% of cases are sporadic. The syndrome is characterized by the following abnormalities: lobed tongue, hematomas or lipomas of the tongue, cleft of hard palate, thickened alveolar ridges, accessory gingival frenula, hypodontia, and abnormal dentition, facial, digital, brain, and kidney abnormalities. The patients therefore often need to undergo massive treatment, including dental care.

OBJECTIVES
We report on a girl with OFD1. In this case we observed striking manifestations of disturbed tooth development in the maxilla.

RESULTS - SYNDROMATIC OBSERVATIONS
We report on a girl with developmental deformities and mental retardation corresponding to oral-facial-digital syndrome type I. She has characteristic malformation of nose and upper lip without any cleft in upper jaw or lip, failure in tooth development, lobed tongue, hematomas and on her extremities she has clinodactyli, brachodactyli and syndactyli. The retina and hearing are normal. One kidney developed with invaginations. It is suggested that genetic mutations in this case lead to the syndromatic disorder (OFD1), which may include disturbance of genetic interactions responsible for the specification of maxillary permanent teeth. Both amelogenesis, cementogenesis, and dentinogenesis were affected. Apparently, the transition of the enamel organ to Hertwig's epithelial root sheet has been disturbed, leading to prolonged enamel formation, invaginations, and irregular root surfaces causing periodontal problems. In this case the transition between different sections of the dental healthcare system has been critical for the positive outcome of the treatment.

RESULTS - TREATMENT
As the dento-alveolar conditions were refractory to preventive and restorative approaches, a more radical treatment was performed. Optimal examination and proper treatment were only possible under general anaesthesia. The final treatment for this patient included extraction of maxillary teeth (Fig. 8) and full upper prosthesis (Fig. 9). The oral status and patient comfort improved significantly following prosthetic treatment (Fig. 10).

DISCUSSION AND CONCLUSION
RESULTS - DENTO-ALVEOLAR OBSERVATIONS
In the maxilla, disturbance of tooth development was seen in all examined permanent teeth. The observations (Fig. 1-7) included tooth aplasia, enamel hypoplasia, atypical pulp and root morphology, and periapical radio lucencies. Over time marginal periodontitis developed with vertical pockets, tooth loosening, gingival abscesses, and pain. Histological examination (Fig. 5) revealed disturbed tooth development with extended enamel coverage of the root surface, enamel inclusions in the dentin and invaginations. The chaotic morphology of the root surfaces explains the persistent periodontal complications and the patient discomfort (Fig. 2). In the mandibular, enamel hypoplasia was observed in the front teeth (Fig. 3). Panoramic radiographs revealed fewer manifest deviations in root morphology, primarily in the front (Fig. 6).

RESULTS - SYNDROMATIC OBSERVATIONS
In this case the oral manifestations of the syndrome were characterized by disturbed tooth development. The presence of atypical pulp and root morphology, as well as abnormalities of the enamel organ, suggests a genetic basis for the observed disturbances. The patients often require comprehensive treatment, including dental care, to address the complex range of symptoms associated with OFD1.

MATERIALS AND METHODS
Diagnostic examination of teeth was performed on histologic sections made by cutting-grinding technique for hard tissues.

ACKNOWLEDGEMENTS
The skilful assistance by lab technicians Anne Larsen and Birthe Gylling Jørgensen is kindly acknowledged.

REFERENCES