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

C. BOYE KJØBSTED[1], A.M. BERGMANN LØVSCHALL[2], O. MOHR HOVGAARD[1], H. LØVSCHALL[1]
[1]DEPARTMENT OF DENTISTRY AND ORAL HEALTH, AARHUS UNIVERSITY, [2]AARHUS 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 characterised by the following abnormalities: lobed tounge, hamartomas 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.[1] 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.

## MATERIALS AND METHODS

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

## 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 radiolucencies. 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 explained 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).

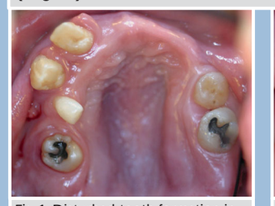

Fig. 1: Disturbed tooth formation in the maxilla.

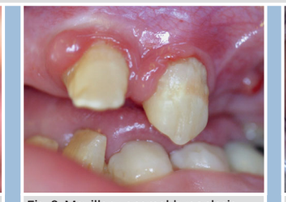

Fig. 2: Maxillary enamel hypoplasia causing periodontal complications.

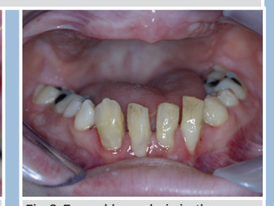

Fig. 3: Enamel hypoplasia in the manbibula.

## 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 tounge, hamartomas and on her extremities she has clinodactyli, brachodactyli and syndactyli. The retina and hearing are normal. One kidney developed with invaginations.

Fig. 4: Atypical pulp and root morphology in permanent teeth, showing extented enamal coverage of the root surfaces, enamel inclusions in the dentin and invaginations.

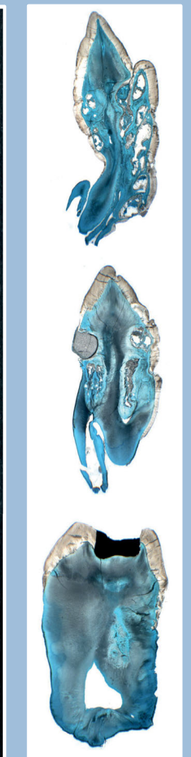

Fig. 5: Histological sections.

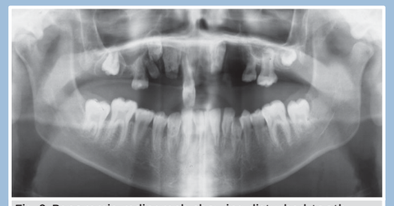

Fig. 6: Panoramic radiograph showing disturbed tooth development primarily occuring in the maxilla.

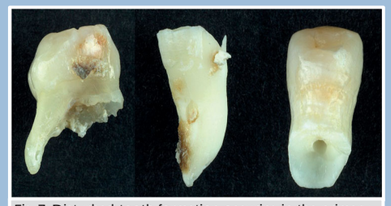

Fig. 7: Disturbed tooth formation occuring in the primary teeth.

## 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).

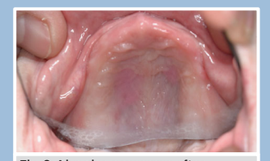

Fig. 8: Alveolar processes after extraction of maxillary teeth.

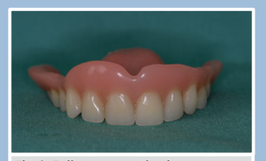

Fig. 9: Full upper prothesis as treatment for the patient.

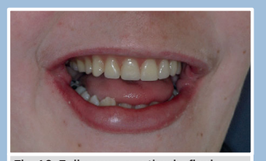

Fig. 10: Full upper prothesis, final treatment result for the patient.

## DISCUSSION AND CONCLUSION

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 syndromes dental manifestations has caused the patient a lot of pain and discomfort leading to a lowered quality of life. The combination of severe treatment needs and suboptimal cooperation make the patient a multidisciplinary case. The mental and general conditions of the patient made a radical treatment necessary. Highly specialized treatment has resulted in a clinical outcome that is acceptable and satisfying for the patient, even though ongoing prophylactics and maintenance are needed. The multidisciplinary treatment between different sections of the dental healthcare system has been critical for the positive outcome of the treatment.

## ACKNOWLEDGEMENTS

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

## REFERENCES

1. Ferrante MI, Giorgio G, Feather SA, et al. Identification of the gene for oral-facial-digital type I syndrome. *Am J Hum Genet*. 2001;68(3):569-576.