The Oro-Facial-Digital Syndrome - Manifestations in the Oral Cavity - Case Report

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Among the various features of the oro-facial-digital syndrome are characteristic malformations of the oral cavity. The syndrome is a genetic anomaly which also presents defects on the face and digits (upper and lower limbs). An early diagnosis is important for the treatment of oral manifestations from an odontologic viewpoint in order to minimize future problems. The treatment of malformations in the oral cavity should be carried out as a joint effort by Dental and Medical professionals.

Key Words: oro-facial digital syndrome, hypertrophied frenula, cleft lip-palate.

Introduction

The oro-facial-digital syndrome is a genetic anomaly presenting characteristic disorders of formation in the face, oral cavity and digits (upper and lower limbs). According to Gorlin and Psaume (1962), the first observations about this syndrome date back to 1883. Morton and Jordan (1935), Mohr (1941), Claussen (1946) and Hubinger (1952) also reported cases of children with malformations of the oral cavity, face and digits. In 1954, Papillon-Léage and Psaume, in a report of 8 cases, named these manifestations oro-facial-digital syndrome, and later Gorlin et al. (1961) suggested the name oro-facial-digital dysostosis.

The malformations of the oral cavity consist of multiple hypertrophied frenula which extend from the labial mucosa of the alveolar ridge and the tongue, resulting in clefts of the alveolar ridge and ankyloglossia, multilobulated tongue of irregular contours and with the presence of nodules, cleft lip-palate, supernumerary teeth, agenesis, poor dental position, and bone hypoplasia. The malformations of the face are aplasia of the alar cartilage, ocular hypertelorism, strabismus, seborrheic and granulose skin and hair, and alopecia. The digital malformations are quite variable, with syndactyly and brachydactyly being the most common. Brain malformation and mental retardation, as well as alterations of the stapes (causing conductive deafness) and polycystic renal disease are present in some patients (Gorlin and Psaume, 1962; Rimoin and Edgerton, 1967; Townes et al., 1976).

In view of the low incidence of this syndrome and of the limited number of reports on this subject in the Dental literature, the objective of the present communication was to
report a clinical case diagnosed at the Pediatric Dentistry Clinic of the Faculty of Dentistry of the University of Ribeirão Preto.

Case Report

C.F.G., a 6-year old white girl, came to the clinic for a routine dental examination. Clinical examination of the oral cavity revealed that the patient presented hypertrophied frenula which extended from the labial mucosa to the lower alveolar ridge, causing clefts in the ridge, and to the tip of the tongue, causing ankyloglossia (Figure 1A).

The tongue was divided into three lobules presenting whitish nodules at the extremities (Figure 1B). Bilateral partial clefts were observed in the upper arch between the deciduous canines and erupted supernumerary canines (Figure 1C). Radiographic examination revealed the presence of impacted upper supernumerary canines and agenesis of the lower permanent lateral incisors (Figure 1D).

Figure 1 - A. Hypertrophied frenula in the lower arch causing a cleft in the alveolar ridge and ankyloglossia; B, multilobulated cleft tongue with the presence of a nodule on the side; C, partial cleft of the upper arch located between the deciduous canine and the supernumerary canine; D, panoramic radiograph showing the presence of impacted supernumerary canines in the upper arch and agenesis of lateral incisors in the lower arch.

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The patient’s face showed a pseudocleft on the left side of the upper lip, with no other manifestations related to the oro-facial-digital syndrome. Clinically, there was a slight curvature of the fingers (clinodactyly).

The family pedigree did not show any similar case, except for two cases of cleft lip-palate.

The patient was referred to a Genetic Study Center for evaluation of the manifestations and characterization of the syndrome.

**Discussion**

A survey of the specialized genetic literature revealed six different types of this syndrome with small differences among them but quite similar and common general features (Mohr, 1941; Papillon-Léage and Psaume, 1954; Sugarman et al., 1971; Whelan et al., 1975; Váradi et al., 1980; Baraitser, 1986).

According to Gorlin (1973), the incidence of the oro-facial-digital syndrome is 1:250,000 births. Gorlin and Psaume (1962) reported that one in 100 patients with cleft palate had the oro-facial-digital syndrome.

This syndrome is transmitted genetically and there is evidence suggesting the existence of two genetic entities associated with the malformations: a) sex-linked autosomal dominant inheritance, with only females affected (a lethal condition for males), and b) recessive autosomal inheritance. This genetic heterogeneity in a clinical disease is a frequently observed important phenomenon (Rimoin and Edgerton, 1967).

Patients with this syndrome show similarities that can be easily identified, although some manifestations may not be present or may be of different intensity (Whelan et al., 1975). This case is rich in oral features, with few digital or facial manifestations.

Treatment of malformations of the oral cavity involves teamwork on the part of professionals specialized in different areas of Dentistry and Medicine. Hypertrophied frenula, clefts of the lip and palate and tongue defects must be corrected by plastic surgery. The Dentist should intervene to remove supernumerary and other teeth as needed, and to treat malocclusion using an orthodontic approach and oral rehabilitation, including restorations and prostheses. The objective of this multiprofessional treatment is to obtain rehabilitation of oral function such as mastication, deglutition, phonetics and aesthetic appearance. Other problems of the oro-facial-digital syndrome should be solved by professionals in the appropriate areas.

**References**


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