

# Oral-facial-digital syndrome, Type I: A case report

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*Oral-facial-digital syndrome is a group of congenital anomalies, which affects the face, oral structures and digits. There are nine subtypes. OFDS type I, is x-linked dominant trait mostly affecting females. Reports of OFDS type I in Asians are extremely rare. This paper shows a case of OFDS type I, in a southern Chinese girl, who in addition to most of the classic features, had fusion of the mandibular canine and lateral incisor teeth.*

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## INTRODUCTION

Oral-facial-digital syndrome (OFDS) is a group of congenital abnormalities, which affects the face, oral structures and digits. First described by Papillon-Leage and Psaume in 1954, nine different subtypes have subsequently been identified, which are based on a variety clinical findings.<sup>1</sup>

OFDS type I is an x-linked dominant trait, which occurs mostly in females and is lethal to males.<sup>2</sup> Although most males do not survive *in utero*, there has been a reported case of a male born with OFDS type 1, who survived for four hours.<sup>3</sup> The incidence of OFDS type I is estimated to be 1:250,000 live births.<sup>4</sup>

Oral manifestations of OFDS type I include the following: bifid tongue, multiple and hyperplastic frenae, hypodontia and supernumerary teeth and a cleft or highly arched palate.<sup>5-7</sup>

Deformities of the face include telecanthus or hypertelorism, median cleft lip and alar hypoplasia.<sup>8,9</sup>

The syndrome can manifest as clinodactyly, brachydactyly or syndactyly of the hands and polydactyly of the feet.<sup>6,7</sup>

Other conditions commonly associated with OFDS type I are alopecia, miliary skin lesions, polycystic kidney disease, variable central nervous system abnormalities, and increased skull base angle.<sup>2,9-13</sup> It has also been found that OFDS type I is associated with mental retardation in 30% to 50% of cases.<sup>6</sup>

The published dental literature regarding OFDS type I is relatively scarce, especially in respect of the Asian population. Therefore, the purpose of this paper is to present a case of oral-facial-digital syndrome, type I, in a southern Chinese girl.

## CASE REPORT

A five-year-old girl was referred from a genetics clinic to the Prince Philip Dental Hospital for treatment of multiple carious teeth. The child was diagnosed as having OFDS type I. According to her medical history, the girl had an operation in 1997 for the removal of tongue nodules and in 1999 to correct her limb deformity and polydactyly. The patient currently attends kindergarten and has a younger sister, who according to the mother does not have the syndrome. No other family member exhibited the condition.

The pregnancy was uneventful and the patient was born at full-term. The birth weight was 2.6 kilograms. Presently she weighs 16.4 kilograms and her height is 102.8 cm, which are both on the 10<sup>th</sup> percentile for Chinese girls in her age group.

Her hair is dry and sparse. She has a flat nasal bridge, hypoplastic alar cartilages and a prominent frontal bone (Figures 1 and 2). The patient also has brachydactyly of the left thumb and a scar on the medial surface of the left foot due to the surgery performed to correct the polydactyly (Figures 3 and 4). The left leg is approximately 2.5 cm shorter than the right leg, resulting in a limping gait.

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Figures 1 & 2. Frontal view and lateral view of showing sparse and dry hair, hypoplastic alar cartilages and a flat nasal bridge.

The maxillary arch is V-shaped, with a highly arched palatal vault. Multiple and hyperplastic buccal frenae are attached to the crest of the alveolar ridge (Figure 5). There is a supplemental primary lateral incisor on the left side of the maxillary arch and a supplemental primary canine on the right side of the same arch. Alveolar clefts are present distal to the supplemental lateral incisor and between the canines on the left side of the maxillary arch. Alveolar clefts are also present on the mandibular arch in the area of the primary lateral incisors (Figure 5).

The attachments of the frenae in the mandibular arch, which are hyperplastic and are attached to the crest of the ridge, also occur in the area of the primary lateral incisors. In addition, she has a bifid tongue.

The presented case is in the early mixed dentition with the mandibular left first molar being the only erupted permanent tooth. She also exhibits crowding in the maxillary anterior region. The primary lateral incisors and canine teeth in the mandibular arch clinically appeared to be fused together. The patient has a posterior cross bite on the right side and an anterior open bite, which is made more pronounced by the fusion of the mandibular primary canines and lateral incisors on both sides of the arch. In addition, she has multiple carious lesions in her primary teeth.

The panograph confirmed the presence of supernumerary teeth in the maxillary arch in the primary dentition and the fusion between the mandibular primary canines and lateral incisors (Figure 6).

There also appears to be supernumerary teeth in the maxillary arch in the permanent dentition, which are a supplemental permanent canine on the right side of the

maxillary arch and a supplemental premolar on the left side. In the mandibular arch, the permanent canine on the right side and the permanent lateral incisor appear to be congenitally missing. The successor of the three-rooted second primary molar on the left side of the mandibular arch is also missing.

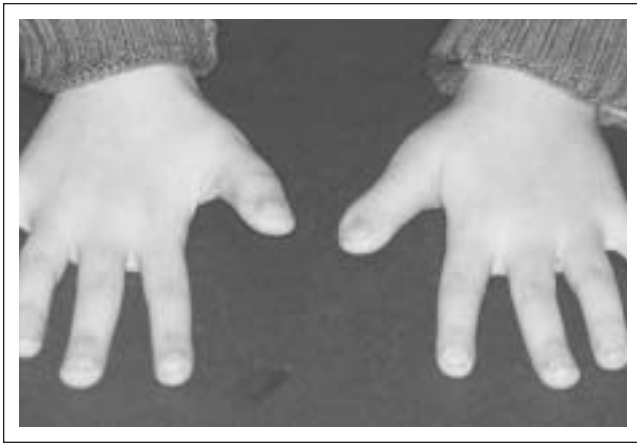
Lateral cephalometric measurements revealed that the skull-base angle is  $139.2^\circ$ ; while the SNA and SNB angles are  $73.9^\circ$  and  $68.2^\circ$  respectively (Figure 7).

## DISCUSSION

The patient presented in this paper had many features that are typical of OFDS type 1, as well as some features that are not commonly associated with this syndrome.

The syndrome has x-linked dominant trait, which makes it more common in females than in males. Based on our investigation of the present case, there appears to be no family history of the syndrome. However, it is known that the syndrome may show varying degrees of expressivity and may not be detected in some subjects, also it can skip several generations before recurring.<sup>5</sup> It may also be possible that the malformations in the present case may be due to a spontaneous mutation.

Flat nasal bridge and alar hypoplasia are commonly observed in OFDS type 1.<sup>6,8,12,15</sup> However, there have been cases reported with normal alar development.<sup>9</sup> Prominent frontal bones are also common features,<sup>16</sup> but may not be present in all cases.<sup>7-9</sup> Most cases reported in the literature reportedly exhibit alopecia and dryness of hair.<sup>6,7,9,12</sup> Facial features such as flat nasal bridge, alar hypoplasia and prominent frontal bones were observed in the present case. In addition to



**Figure 3.** Abnormally short left thumb (brachydactyly).



**Figure 4.** Scarring on the medial surface of the left foot as a result of the surgical correction of the polydactyly.



**Figure 5.** Multiple and hyperplastic buccal frenae attached to the crest of the ridge on the maxillary and mandibular arches. Maxillary anterior crowding due to the presence of supplemental teeth. Fusion of the lateral incisor and canine teeth on both sides of the mandibular arch. Cleft of the alveolar ridges on the areas of attachment of the buccal frenae.



**Figure 6.** Panelpipe radiograph showing supernumerary teeth in the maxillary arch in the primary dentition and fusion of the mandibular primary canines and lateral incisors. A supplemental canine on the right side and premolar on the left side of the maxillary arch can also be seen. Hypodontia of the permanent canine on the right side and permanent incisor and second premolar on the left side of the mandibular arch can also be seen.

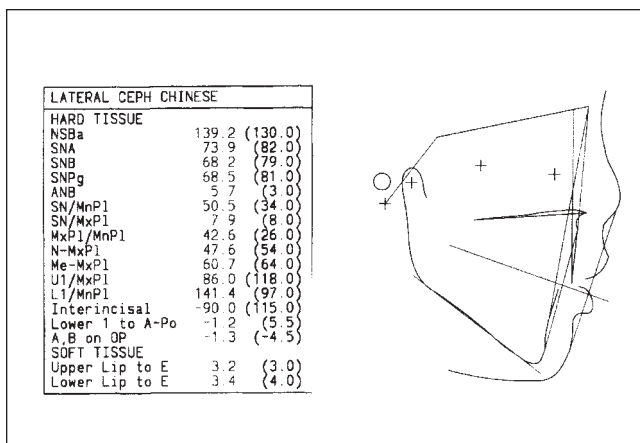
these features the presented case also exhibited dry and sparse hair.

A variety of malformations of the digits of the hands and feet are observed with OFDS type 1. According to Melnick and Shields, the presence of a multilobulated tongue and hyperplastic frenula with or without brachydactyly of the digits may be considered to be the minimum diagnostic criteria for oral-facial-digital syndrome type 1.<sup>6</sup> However, it is generally accepted that the presence of some form of digital malformation, such as clinodactyly, brachydactyly or syndactyly of the hands and polydactyly of the feet, in combination with the facial and oral abnormalities comprise the common manifestations of the syndrome.<sup>1</sup> Apart from the brachydactyly of the left thumb and the presence of an extra digit on the left foot, the presented case exhibited a deformity of the left leg, which was 2.5cm shorter than the right leg. Shaw and co-workers<sup>7</sup> also reported

malformations of the arms and legs. They presented a case with disproportionately short lower legs and forearms.<sup>7</sup>

Common intra-oral findings of patients with OFDS type 1 include tongue clefts, multiple and hyperplastic frenae, anomalies of tooth number and a cleft, or highly arched palate.<sup>5-7</sup> As with the present case, the case report by Lipp and Lubit also exhibited hyperplastic multiple buccal frenae attached to the buccal aspects of the alveolar ridges in the maxillary and mandibular arches.<sup>9</sup>

It has been reported that the incidence of OFDS type 1 in cleft lip and/or palate patients is 15:1,000.<sup>17</sup> Conductive hearing loss is reported to be a characteristic, which distinguishes OFDS type 1 from OFDS type 2.<sup>9</sup> However, it should be noted that this audiological condition is common in patients with cleft palate and may, or may not be directly related to the syndrome. The timing of



**Figure 7.** Lateral cephalometric measurements showing the increase in skull base angle (NSBa = 139.2) and decrease in the relative position of the maxilla to the anterior cranial base (SNA = 73.9) and relative position of the mandible to the anterior cranial base (SNB = 68.2).

\*values enclosed in parentheses are normal values for southern Chinese<sup>19</sup>

the surgical closure of the cleft lip and/or palate in patients with OFDS type 1 is similar to that of other cleft lip and palate patients.<sup>6,18</sup>

Abnormalities of the tongue such as clefting (bifid tongue), hyperplasia, ankyloglossia, multilobulations and presence of nodules have been reported in cases with OFDS type 1.<sup>9,12,16,18</sup> From the literature, the management of the various orofacial abnormalities appears to be based on the characteristics of the individual cases. Some authors have reported surgical correction of ankyloglossia and bifid tongue at ages ranging from 3 to 12 months.<sup>12,16</sup> In a case presented by Nagahara and co-workers, surgical correction of the ankyloglossia on a 14 year old Japanese girl was performed to accommodate the use of a myofunctional appliance.<sup>15</sup>

Tongue nodules were surgically removed for the presented case, but unfortunately no pathology report could be located. Tongue nodules that have been excised for the purposes of biopsy, were found to be lined by non-keratinized stratified squamous epithelium with an absence of filiform papillae and composed of irregularly arranged fibrous tissue interspersed with adipose tissue.<sup>8</sup> In many cases, asymptomatic tongue nodules have been left untreated.<sup>6,7,9,12</sup>

The presence of multiple and hyperplastic buccal frenae, result in the abnormal development of some structures in the oral cavity. For example, the crestal attachment of the frenae result in the formation of clefts of the alveolar processes, which are commonly observed in the area of the mandibular incisors.<sup>8,10,12</sup> The abnormally thick buccal frenae also result in the formation of a shallow buccal sulcus and a decrease in height of the alveolar ridge.<sup>9,12</sup> All of these features were observed in the reported case.

Anomalies of tooth number are common manifestations of this syndrome. An early report indicated that supplemental canines were usually present and that the missing teeth are usually the mandibular lateral incisors.<sup>5</sup> However, more recent literature has shown that a wide variety of teeth may be missing or there may be supplemental teeth.<sup>7-9,15</sup> Interestingly, none of the previously reported cases have exhibited fusion or gemination of the teeth. In the present case, if it is assumed that the lateral incisors were missing based on the common clinical findings of OFDS type 1, it could be speculated that there was gemination of the mandibular canines because of the presence of double crowns. However, careful radiographic investigation clearly shows the presence of two pulp canals. Hence, it can be concluded that there was fusion of the mandibular lateral incisors and canines.

Abnormalities in the quantity and quality of enamel have been reported, although it is difficult to accept these changes as being characteristic features of OFDS type 1. Axrup and co-workers reported enamel hypoplasia in the primary maxillary anterior teeth<sup>8</sup>, and there has been a case of generalized enamel hypoplasia in an eight year-old girl.<sup>16</sup> However, no enamel hypoplasia was observed in the presented case.

Lateral cephalometric analysis of this case demonstrated SNA and SNB angles that were decreased by more than three standard deviations and a skull-base angle, which was two standard deviations greater than the norm for southern Chinese.<sup>19</sup> A study of the cephalometric measurements of other patients with OFDS type 1 reveals that there is a tendency for an increase in the skull-base angle.<sup>10</sup> However, it may not occur in all cases.<sup>6</sup>

The oral manifestations of OFDS type 1 are not unique to the syndrome. These oral conditions can therefore be managed using conventional treatment methods. However, there are factors that can complicate the treatment.

Mental retardation, which is found in 30% to 50% of cases with OFDS type 1,<sup>6</sup> may require treatment under general anesthesia even for simple restorative procedures. The occurrence of various oral conditions such as a cleft palate, hypodontia and supernumerary teeth in a single patient requires comprehensive multidisciplinary treatment planning and dental care. Features such as multiple and hyperplastic frenae, shallow buccal sulcus and decreased alveolar ridge height and width may present difficulties in the design and fabrication of conventional removable prostheses to replace missing teeth. Removal of the high attachments of the frenae can be performed to increase the depth of the buccal sulcus.<sup>16</sup>

Dental caries can be a problem for patients with OFDS type 1.<sup>8,16,18</sup> The presence of crowding caused by the eruption of supplemental teeth and a constricted dental arch can result in areas where plaque can easily

accumulate,<sup>20</sup> and tooth brushing is likely to be further compromised by the multiple and hyperplastic frena. A preventive program consisting of oral hygiene instructions, dietary advice, regular fluoride mouth rinsing and the regular application of topical fluoride agents should therefore be recommended for these unfortunate children.

**SUMMARY**

Oral-facial-digital syndrome type 1 is x-linked dominant trait, which is more common in females than in males. The syndrome, which affects the face, extremities and the oral cavity, has varying degrees of expressivity. The case of a five-year-old girl with OFDS type 1 was presented.

She exhibited common clinical facial features of the syndrome such as a flat nasal bridge and hypoplastic alar cartilage, brachydactyly of the hands and polydactyly of the feet. Oral manifestations such as a bifid tongue, a highly arched palatal vault and multiple and hyperplastic buccal frena, hypodontia and supernumerary teeth, observed in the present case, are distinct features of this syndrome. Fused teeth were also observed in the primary dentition of the presented case. Furthermore, her lateral cephalometric measurements revealed an increased skull-base angle.

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