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Orofacial digital syndrome type i with vision impairment - A rare case report

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Abstract
Orofacial digital syndrome type I (OFDS I) is an X-linked dominant disease affecting the CXORF5 gene. There are 14 types of OFDS, of which OFDS type I accounts for 75% of cases. OFDS can be expressed differently from person to person and can manifest new oral/clinical observations or affect the new system. The current case describes OFDS I with vision impairment which is an additional observation reported for the first time. Managing an OFDS patient requires a comprehensive approach.

Keywords: Brachydactyly, orofacial digital syndrome, polydactyly, syndactyly

Introduction

Genetic disorders encompasses wide range of systemic manifestation that are passed down from one generation to another generation. Sometime that disorders manifest a rare manifestation or defect that can help in differential diagnosis and timely management. Orofacial digital syndrome was first described in 1954, is a manifestation of uncommon heterogeneous and hereditary condition involving bone and teeth [1]. There are 14 types of OFDS, of which OFDS type I accounts in 75% of cases [2]. It is an X linked dominant diseases affecting CXORF5 gene [3].

The syndrome is fatal in male. A de novo pathogenic variant can occasionally cause the disease in a female proband with OFDSI [4]. Common clinical manifestation are hyperplastic lingual and labial frenula, lobulated tongue, median cleft of the upper lip and palate, syndactyly, brachydactyly and polydactyly [5]. Distinctive characteristics features are adult-onset polycystic kidney disease (PKD) presenting in female.

OFDS can be expressed differently from person to person and can manifest new oral/clinical observations or affect new system. The current case describes OFDS I with vision impairment which is an additional observation reported for the first time which can be used in differential diagnosis when genetic testing not possible [6].

Patient information

A 10-year-old female patient reported to the Department of Pediatric and Preventive Dentistry, with chief complaints of irregularly placed teeth. On examination, the girl shows peculiar physical and cranial characteristics that necessitated a more thorough investigation of the case. On eliciting a thoroughly detailed history it was revealed that the mother had a normal delivery. Based on reports at the time of birth girl was present with a cleft palate, natal teeth, brachydactyly, syndactyl, and polydactyl. Chromosomal karyotype showed no abnormalities of the chromosome or the pair of sex chromosomes. No family history of congenital disorders was present.

Case report

Intraoral examination shows thick fibrous bands in the lower mucobuccal fold causing shortening of the sulcus [Figure 1] and alveolar ridge clefting, bifid tongue [Figure 2], missing lower lateral incisor, impacted lower right central incisor in OPG [Figure 3], 'V-shaped high arched constricted upper arch (surgery for cleft palate done when she was 1 year old) [Figure 4a], crowding in the upper and lower arch, anterior open bite and posterior crossbite [Figure 4b]. The extraoral assessment revealed a mildly flattened face, facial asymmetry, alopecic patch at the frontal region [Figure 4c], hypertelorism, depressed nasal bridge, ocular proptosis, strabismus, down-slanting palpebral fissures, frontal bossing, macrocephaly, midface hypoplasia, ross bow-shaped lips and parrot beak nose [Figure 4d].

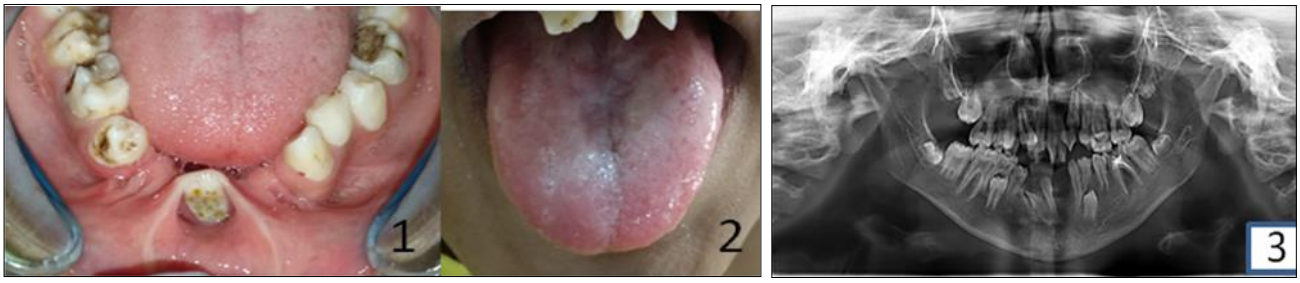

Fig 1: Thick fibrous bands in the lower mucobuccal fold
Fig 2: Bifid tong figure

Fig 3: OPG showing impacted lower right central incisor

Fig 4a: V-shaped high arched constricted upper arch. **Fig 4b:** Anterior open bite and posterior crossbite. **Fig 4c:** Localized alopecic patch. **Fig 4d:** Ross bow-shaped lips and parrot beak nose

Examination of upper limbs shows brachydactyly, complete syndactyly of the second and third fingers of both hands [Figure 5a], and the presence of postaxial polydactyl on the right side hand [Figure 5b]. Lower limb lateral deviation of the greatest toe and medial deviation of the first metatarsal [Figure 5c]. Vision impairment of both eyes seen. Vision impairment of both eyes important unusual observation we noted in this case which was not reported earlier in OFDS

Based on the clinical and radiographic findings OFDS I was diagnosed. Thorough systemic evaluation reveled no evidence of congenital heart defects, lung, liver, kidney and central nervous system disorders.

Diode laser excision of thick fibrous bands in the lower mucobuccal fold [Figure 6a]. restoration of carious teeth and oral prophylaxis is done. Follow up of patient was done after 3 month shows good healing of lower labial mucobuccal fold [Figure 6b].

Fig 5a: Complete syndactyly of the second and third fingers of both hands. **Fig 5b:** Postaxial polydactyl on the right side hand. **Fig 5c:** Lower limb lateral deviation of the greatest toe and medial deviation of the first metatarsal.

Fig 6a: Laser excision. **Fig 6b:** Follow up of patient after 3 month of labial mucobuccal fold

Discussion

OFDS I exhibits congenital malformations affecting the face, oral cavity, hands, and feet. Wide range of symptoms associated with OFDS makes the diagnosis challenging [7]. There are currently 14 different types of OFDS with high prediction for Type I. Although numerous variants of this condition have been reported, its complete spectrum is still unknown. This current report is a type I OFDS with an unique phenotypic characteristic.

Typical oral manifestation of OFDS I is, alveolar ridge notiching, hyperplastic frenulum, cleft lip and palate, ankyloglossia and cleft of tongue with hamartomas [8, 9]. Dental abnormalities such as, enamel dysplasia, impacted teeth, anterior open bite, and posterior cross-bite, hypodontia, hyperdontia and retained deciduous are commonly observed. Facial abnormalities in OFDS 1 include frontal bossing facial asymmetry, broad nasal bridge, hypertelorism, alar hypoplasia, unequal nostrils, low-set ears, flat mid-face, maxillary and mandibular hypoplasia, flat mid-face, vanishing milia on the face and ears, patterned alopecia of the scalp and dry brittle hair. Skeletal malformations include syndactyly, brachydactyly, polydactyly, clinodactyly and limb length variation. Abnormalities such as cerebellar anomalies, hydrocephalus, agenesis of corpus callosum have been noted and porencephaly [10, 11].

These clinical features not only overlap with different types of OFDS but also with Ellis Van Creveld syndrome, McKusick-Kaufman syndrome (MKK) and Weyers acrofacial dysostosis (WAD), so diagnosis of syndrome is quite difficult. However these are some peculiar characteristics that needs to be stressed on while differentiating ths syndrome OFDS I is distinctively associated with patchy alopecia which is absent in other types of OFDS [12, 13]. Presence of alopecia, cleft palate, bifid tongue, ocular hypertelorism, absence of dwarfism and hypoplastic finger nail help to differentiated from Ellis Van Creveld syndrome. Nail dystrophy is characterstic feature of WAD which is absent in OFDS I. Absence of hydrometrocolpos in OFDS I distinguishes MKK syndrome.

In the current case vision impairment is an additional observation that has not been reported in any of the syndrome. This additional feature helps in differentiating

OFDS I from other syndrome. Early eye examination should be done in these patient to prevent long term complications associated with vision.

Managing an OFDS patient requires a comprehensive approach. Prenatal ultrasonography can reveal anomalies of the face, brain, and digital or limbs. Due to compromised medical general health and oral manifestations, parental and child counselling is frequently necessary to reduce psychological trauma.

Declaration of patient consent

The authors certify that they have obtained all appropriate patient consent forms. In the form the patient(s) has/have given his/her/their consent for his/her/their images and other clinical information to be reported in the journal. The patients understand that their names and initials will not be published and due efforts will be made to conceal their identity, but anonymity cannot be guaranteed.

Conclusion

The diagnosis and management of OFDS I syndrome provides significant challenges to the oral health care professionals. Because of the genetic and environmental phenotype-modifying factors in syndromes, oral health care practitioners might experience some peculiar features. Present case reported unique feature that includes presence of vision blurriness which was not reported before in OFDS I. Early diagnosis and management is vital in preventing further complications associated with OFDS I. Treatment of OFDS I requires multidisciplinary approach involving orthodontic treatment and surgery performed hand in hand. By the means of extensive interdisciplinary approach the facial aesthetic of the patient can also be managed effectively.

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