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Case Report

ORO-FACIAL DIGITAL SYNDROME 1- IN ITS SUBDUED PRESENTATION

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ABSTRACT

Orofacial digital syndrome (OFDS) is an umbrella term for a group of apparently different genetic disorders, affecting the oral cavity, face and digits. OFDS-I is the most commonly encountered syndrome from this group and can present with multi-organ involvement. The case of OFD-I reported here is distinct, as it presents with minimal clinical features of OFDS-1 and will help the orophysicians in formulating the diagnosis.

INTRODUCTION

Orofacial digital syndrome (OFDS) is a multifaceted syndrome with 13 different subtypes reported till date, described as per the mode of inheritance and other organs and systems involvement (eye, brain, tibia, skeletal changes, and presence of millia) (1). Oral-facial-digital syndrome has an estimated incidence of 1 in 50,000 to 250,000 newborns, and type I, which is an x-linked dominant trait that has the highest incidence when compared with the other subtypes (2,3). It was first described by Papillon-Leage and Psaume in 1954 and further defined by Gorlin and Psaume in 1962 (3). Hence, the syndrome was also named Papillon — Leage and Psaume Syndrome (4). They described it under the heading of orofacial dysostosis, but as there was involvement of tissues other than the bone, the term OFDS was preferred (5).

Orofacial digital syndrome type I (OFDS-I) is transmitted as an X-linked dominant condition with lethality in males (3). Conversely, males have been reported in the literature with this disorder with the XXY genome (2). The other types of OFDS show autosomal recessive (AR) inheritance (2). Chromosomal aberrations have been demonstrated in only a minority of the patients (5). However, 75% of the cases have an apparently sporadic presentation (6).

OFDS I patients show a wide range of presentations with malformations of the face, oral cavity, and digits. It possess a high degree of phenotypic expressivity even within the same family, (3,6) owing mainly to the different degrees of somatic mosaicism (2). The present case introduces the subdued presentation of OFDS-I in a 3 years old girl.

Case Report

A 3 years old girl, who was referred by a private practitioner and accompanied by her mother reported to the department with a chief Complain of pain in upper front teeth region for the past 3-4 days. Pain was insidious in onset, intermittent and moderate in intensity. No aggravating or relieving factors were reported. On further questioning patient's mother reported that she had a small growth on the tongue region since the age of 6 months and was not normal regarding the daily activities as per the age of the child and had impaired speech and other developments as compared to children of the same age. The patient was never consulted to any physician previously for the same by her mother. Family history revealed the consanguineous marriage of parents and did not have any siblings. Parents and other family members were not affected.

On general physical examination, the patient had an altered gait and dolichocephalic head, Polydactyly noted with respect to the right hand, big toe was noted bilaterally on the feet and Clinodactyly was noted in the extremities. Extraoral

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examination revealed hypertelorism, Depressed nasal bridge retrognathic mandible.

On intraoral examination, tongue revealed 2 small nodular growths bilaterally on the lateral margin of the tongue, which were approximately 1.5 x1.5 cm in its maximum size. On palpation, these were sessile, soft, non-fluctuant and non-tender. Moderate ankyloglossia noted. Caries were present in relation to 54, 75 and 84. Temporary restorations were noted in maxillary anteriors.

Figure 1 Physical appearance of the patient along-with soft tissue growth on lateral border of tongue bilaterally. Hypertelorism with anti- mongoloid slant noted.

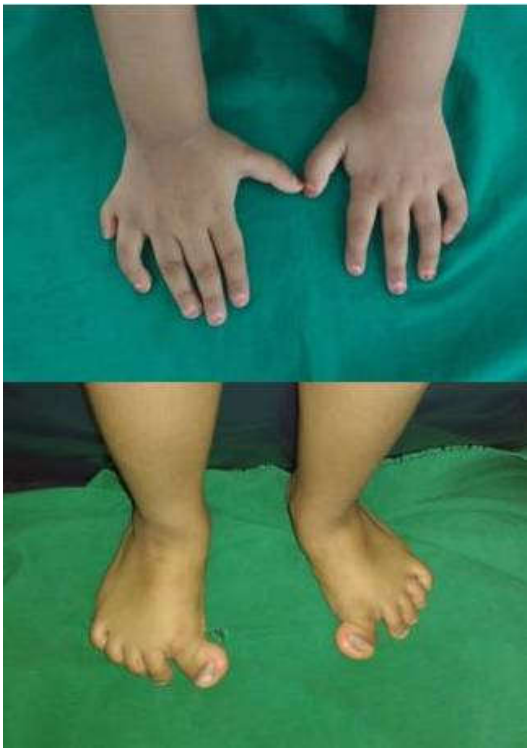

Figure 2 Polydactyly and Clinodactyly of hands and big toe with clinodactyly noted on feet

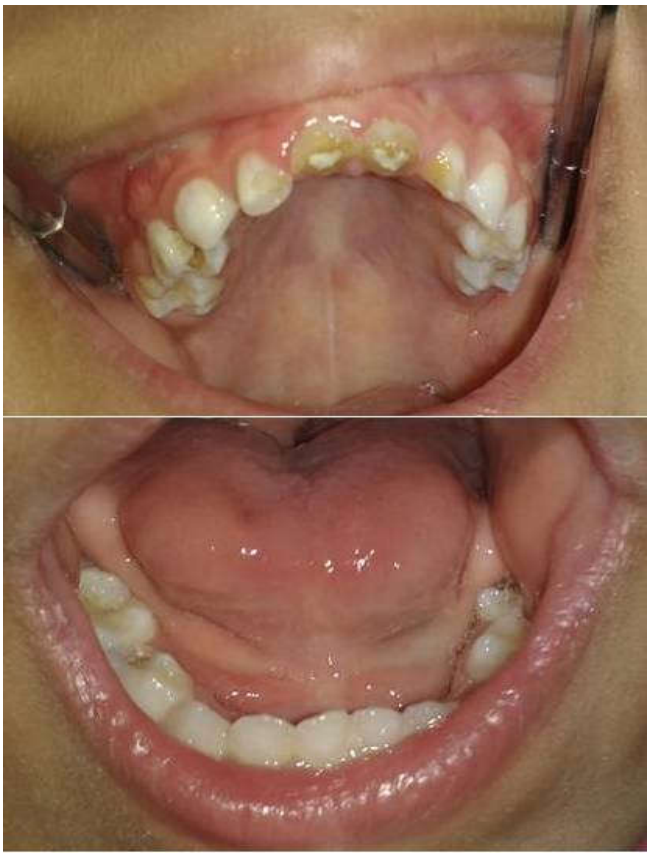

Figure 3 intraoral presentation depicting maxillary and mandibular arch with palate

The patient was referred to the pediatrician for further evaluation. The patient was not cooperative for the radiological examination of the head and neck region and hence was not done.USG abdomen revealed no abnormality. Karyotyping was not performed owing to the financial reasons by the patient's mother. This case is a subdued presentation of the orofacial digital syndrome as its features are depicted in table 1.

Table 1 Compares the features of the present case with the clinical features reported in the literature

Site	Clinical features	Present case
Intra-oral	Median pseudoclefting of the upper lip	Absent
	Clefts of the palate	Absent
	Hamartomas of the tongue	Present
	Bifid tongue	Absent
	Hyperplastic oral frenula	Absent
	Thickened alveolar ridges and abnormal dentition	Absent
	Irregular margin of the lips	Absent
Head and face	Facial asymmetry	Absent
	Hypertelorism	Present
	Micrognathia	Present
	Broadened nasal ridge	Present
	Hypoplasia of the malar bones and nasal alar cartilages, and frontal bossing	Present
	Dryness, brittleness, and/or alopecia of the scalp hair	Absent
extremities	Syndactyly	Absent
	Brachydactyly	Present
	Clinodactyly	Present
	Unilateral duplication of the hallux, and more rarely, pre- or postaxial polydactyly	Present

DISCUSSION

OFDS is considered to be a variant of ectodermal dysplasia and affects the development of the skin, nails, hair, eyes, oral cavity, face, fingers, and toes. The central nervous system, renal, cardiovascular and cutaneous abnormalities have been linked to this disorder (2) however, the present case has a subdued presentation of OFDS-I and did not present with such extensive presentation.

The gene responsible for OFDS-I is reported to be found on the 22nd band of the p-arm of the X chromosome (7) and mutation analysis has identified the gene as CXORF5 (2). In normally developing males i.e., XY males, mutation of OFD-I gene results in a total loss of OFD-I protein, which is critical in the early development of the brain, face, limbs, kidney, and another organogenesis. Thus, it causes disruption of normal growth and development and results in abortion of the fetus, suggesting that this syndrome usually predominates in females and males with altered karyotype. There is extensive phenotypic variability within this subgroup of OFDS owing to the somatic mosaicism (2).

Typical facial features of OFDS-I includes- ankyloglossia, lobulated tongue with the presence of nodular growths, alveolar ridge clefts, hypertrophied labial and lingual Freni, cleft lip-palate, tooth anomalies, etc. The facial abnormalities include aplasia of the alar cartilage and malar bones, hypertelorism, strabismus, vanishing milia on the face and ears, low-set ears, and dry brittle hair with zones of alopecia. Digital malformations of the hands and feet are syndactyly (fusion of digits), brachydactyly (shortened digits), clinodactyly (curved digits), and polydactyly (extra digits) along with radial and ulnar deviation and duplicated hallux (great toe) (2,7). The present case, being a subdued presentation, presented with only a few of these characteristics as summarised in Table 1.

Other systemic involvements reported in literature include- CNS malformations, Polycystic kidney disease, Pancreatic, ovarian and liver cysts, spasmodic movements, delayed motor and speech development may also be present in some patients (6,7). No such findings were observed in the present case.

It is recommended to perform a brain MRI, an abdominal ultrasound, a skeletal survey, an ophthalmologic evaluation, and an audiometric test in all consenting patients. All these investigations were carried out in the present case and no abnormality was detected.

Managing a patient with OFDS requires a multidimensional approach. If any history of a disease causing mutation in the family is known, antenatal diagnosis is possible by ultrasound examination which may detect structural brain malformations and/or duplication of the hallux. Reconstruction surgeries for the correction of clefts of the lip and/or palate, tongue nodules and accessory frenulae are required for the patients presenting with these defects. Besides these, Orthopedic surgery is often recommended to repair the defects of digits. Speech defects, ocular infections and defects; orthodontic treatment for malocclusion; if present should be addressed. Renal disease may require hemodialysis or peritoneal dialysis and renal transplantation (5). The degree of learning disabilities and other cognitive impairment should be evaluated along with speech therapy to provide appropriate support. The present case presented with dental caries for which restoration and endodontic treatment was done. However, patient mother did not give consent for the removal of lingual swellings.

Besides these active interventions, Regular follow-up for assessment of speech, periodic determination of blood pressure and serum creatinine concentration to monitor renal function is important. Annual assessment of renal function with follow-up by renal ultrasound, evaluation to assess cyst development if abnormalities are detected and periodic screening for ovarian, pancreatic, and hepatic cystic disease should be considered (3).

CONCLUSION

This case report is a subtle presentation of the OFDS-I syndrome and justifies the fact that it presents with great variation in the phenotypic expression. this case report also highlights the inevitable role played by orophysicians in proper documentation and reporting of these cases.

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References

1. Sukarova-Angelovska E, Angelkova N, Palcevska-Kocevska S, Kocova M. The many faces of Oral-facial-digital syndrome. Balkan Journal of Medical Genetics. 2012 Jan 1;15(1):37-44.
2. Duda PW, Singer SR, Strickland M. Oral-facial-digital syndrome type 1: Report of a case. Journal of Orofacial Sciences. 2014 Jul 1;6(2):118.
3. Adyanthaya A, Adyanthaya S. Oral facial digital syndrome type 1: a review. Bangladesh Journal of Medical Science. 2015 Apr 18;14(2):130-4.
4. Dhull KS, Acharya S, Mohanty M, Dhull RS, Panda S. oro-facial-digital syndrome Type 1: A case report. Journal of Indian Society of Pedodontics and Preventive Dentistry. 2014 Apr 1;32(2):152.
5. Dave KV, Patel SC, Dudhia BB, Panja P. Orofacial digital syndrome. Indian Journal of Dental Research. 2013 Jan 1;24(1):132.
6. Gurrieri F, Franco B, Toriello H, Neri G. Oral–facial–digital syndromes: review and diagnostic guidelines. American Journal of Medical Genetics Part A. 2007 Dec 15;143(24):3314-23.
7. Xavier AM, Rai K, Hedge AM, Joshi S. Hereditary oro-facial digital syndrome type 1: Diagnosis and management: Case report. Eur. J. Dent. Med., http://scialert. net/abstract. 2011.