ABSTRACT:
Orofacial Digital syndrome –Type I is an X-linked dominant disease affecting both male and female. It presents with distinct malformations in the oral cavity and defects in the face and digits of the upper and lower extremities. Early and Accurate diagnosis and multidisciplinary approach is required by allied health professionals to plan out a systematic management protocol in these victims in order to minimize future odontogenic problems, related skeletal and systemic problems. The objective of this article is to report a rare case of Orofacial Digital syndrome in a 6 year old female child, with characteristics features of Orofacial syndrome –type I and treatment undertaken to minimize the orofacial problems.

KEY WORDS: Orofacial Digital Syndrome, syndactyly

INTRODUCTION
Oral-facial-digital syndrome-type 1 (OFD 1) is an X-linked dominant condition with multiple distinct clinical features such as malformations of the face, oral cavity, and digits. It was first described by Papillon-League and Psaume. When it occurs in male it is lethal.1 The genetic background of the syndrome is found to be due to mutations in the gene OFD1. The expression of the gene begins at mid-gestation in the human kidney.2 strabismus and alopecia. Brachydactyly, syndactyly, clinodactyly of the fifth finger commonly, radial and ulnar polycystic renal disease, spasmodic movements/tics, brain malformation, delayed motor and speech development may also develop in the patient.3,4

The syndrome presents with multiple features such as ankyloglossia, multilobulated tongue with the presence of nodules, clefts of the alveolar ridge, multiple hypertrophied frenula, cleft lip, cleft palate. According to king and sanarees,5 clinical abnormalities seen are aplasia of the alar cartilage, ocular cartilage, ocular hypertelorism.

CASE REPORT
A 6 Year old female child patient reported to the Department of Oral medicine and Radiology with a chief complaint of regurgitation of liquid foods on swallowing, delayed speech, and multiple decayed teeth.

Patient gives a history of extraction of lower front teeth 6 months back following decay. Patient is not under any systemic medication, past medical history revealed no cardiac or renal abnormalities.

On extraoral examination there was scarcity of scalp hair, with areas of alopecia, skull appears brachycephalic. There was increased interpupillary space and depressed nasal bridge, the upper lip showed partial midline clefting (figure 1). The 6 yr old girl had a short stature with short upper limbs. Both the hands were short and broad as in brachydactyly, and right hand showed partially fused ulnar and radial finger as syndactyly (figure 2).

Other extraoral features include a broadened forehead, base of the nose and widely set eyes. Alopecia was noted at the central portion of head (Figure 1).

Intraoral examination revealed macroglossia with solitary tongue nodules of 3 in number roughly 1 cm in diameter and lesser were noticed on the lateral borders on the right of tongue, the surface of the nodules were smooth, featuring benign mass (figure 4). The palate showed partial clefting of primary palatal region with the secondary palate. There was midline clefting of soft palate. (figure 3)

Grossly decayed 51, 52, 63, 54, 73, missing 72,73,74, 82 was present. There was multiple labial frenum (6 in number) were present in maxilla, and another 6 labial frenum were present in mandible. Peusdoankylosis of the tongue was present (figure 5)

Radiological examination:
Maxillary Occlusal radiograph shows grossly decayed 51 52 53 561 62 63 with pulp exposure. The permanent tooth crowns of 11 12 13 21 22 23 27 17 was present.

No signs of any oroantral communication was present. (figure 6)
Complete skeletal survey was done for the patient which showed no abnormalities of the long bones, pelvis, rib cage, sternum, and clavicle. The right hand showed shadow of incomplete soft tissue union of the 4th and the 5th phalanges, with no evidence of any bony fusion of ring-small syndactyly is evident. (figure 7)

Treatment:

Surgical excision of all accessory frenum was carried out by laser under local anaesthesia, the post-operative period was uneventful, patient showed good healing in 2 weeks (figure 8), grossly decayed 51 52 61 62 73 were extracted and space maintained in form of a removable partial denture was fabricated. Patient was advised lip exercises and speech therapy following denture placement. After 2 months patient showed considerable improvement in speech, and regurgitation of fluids reduced due to improved lip activity. (figure 9) The patient was referred to orthopaedics for surgical correction of incomplete soft tissue union of 4th and the 5th digit.

Discussion:

Oral-facial-digital syndrome (OFD) is a type of ectodermal dysplasia involving the development of the eyes, skin, nails, hair, oral cavity, face, fingers, and toes. The diagnosis of OFD1 is primarily based on clinical features of oral, facial and digital abnormalities. It is transmitted as an X-linked dominant trait OFD1 has an incidence of 1/50,000-250,000 live births. Patients with this syndrome show a wide range of presentation due mainly to the different degrees of somatic mosaicism. The facial abnormalities seen in papillon-leauge-paucmsymdrome are nasal alar cartilage hypoplasia, frontal bossing, hypertelorism with broad nasal bridge and micrognathia with hypoplasia of mandibular ramsus. The oral aspects of OFD-1 syndrome frequently involves, tongue and frenum abnormalities. The case reported here showed many typical features of this syndrome.

The dental anomalies in OFD1 patients are supernumerary teeth, malposition of teeth, crowding, tooth fusion, absence of mandibular lateral incisors, and enamel hypoplasia. Cleft lip and/or cleft palate was observed in all individuals (100%); 88% of cases were palate cleft. The occurrence of OFD1 syndrome in cleft patients is estimated to be about 12 cases in every 1000 cleft individuals. Facial features such as flat nasal bridge, alar hypoplasia and prominent frontal bones were present in this patient. Common intra-oral findings include lobulated tongue, cleft lip and palate, numerous hyperplastic frenulas seen involving linguual, buccal, and labial frenulae, lobulated tongue with hamartomatous growths, narrow upper lip, and dental anomalies. The oral manifestations present in the case were hypoplastic teeth, multiple retained deciduous teeth, supernumerary teeth, multilobulated tongue, partial ankyloglossia, high arched palate. The case also presented hyperplastic multiple buccal and lingual frenulae attached to the buccal aspects of the alveolar ridges in the maxillary and mandibular arches.

OFDS type 1 presents with variety of malformation of the digits of the hands and feet. However, the common manifestations of the syndrome comprises of 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. The present case showed brachydactyly of both hands and feet partial syndactyly on her right hand. The malformations of arms and legs were reported in a case series by Shaw where the case presented with disproportionately short lower legs and forearms.

Other authors also presented patients with deformity of the left leg, which was 2.5 cm shorter than the right leg apart from brachydactyly of the left thumb and the presence of an extra digit on the left foot. Cutaneous abnormalities such as numerous milia, especially over the face and perina, can also be present as a patterned type of alopecia, sparse, fine or coarse, dry lustreless hair may also be present.

CONCLUSION:

Early diagnosis from odontogenic point of view will minimize the sequel of developing physical and dental abnormalities. Treatment for this syndrome requires multidisciplinary approach and role played by the dentist is vital in diagnosis, intervention and proper treatment to decrease morbidity and improve the quality of life for the patient.

References:


FIG 1

FIG 2