Oral-Facial-Digital Syndrome Type I

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Oral-facial-digital syndrome (OFD) is a collective term, which includes thirteen distinctive, genetic disorders. These syndromes are a form of ectodermal dysplasia, which affects the development of the skin, nails, hair, eyes, face, fingers, toes and the oral cavity. In addition, cardiovascular, renal and central nervous malformations are commonly associated with these conditions. The affected individuals may present with speech and learning disabilities, impaired mental ability, and seizures which are directly related to the severity of central nervous system involvement.

The inheritance pattern and phenotypic expression mainly distinguish the rare 13-subtypes. OFD type I has the highest incidence compared with the other subtypes, and equally distributed among different races and ethnicities. We present a thirteen-year-old female who presented with OFD syndrome, type I. OFD syndrome is a form of ectodermal dysplasia affecting the development of the skin, nails, hair, eyes, face, fingers, toes and the oral cavity.


Oral-facial-digital syndrome (OFD) is a form of ectodermal dysplasia affecting the development of the skin, nails, hair, eyes, face, fingers, toes and the oral cavity. The central nervous system, cardiovascular, renal, and cutaneous abnormalities have been linked to this disorder. Also, impaired mental ability, speech, learning disabilities, and seizures are directly related to the severity of central nervous system involvement. Some cases were reported with malformation of the stapes which led to conduction deafness.

Thirteen different subtypes of OFD were reported in the literature. OFD type I has the highest incidence compared to other subtypes; it affects all races and ethnicities in equal numbers. Approximately 75% of cases of OFD I are sporadic and approximately 25% are hereditary. OFD I is transmitted as an X-linked dominant condition, with prenatal mortality in homozygous males. Few male cases have been reported in the literature with the XXY genome.

The phonetic expressions vary in severity, as the naming indicates it is characterized by affecting the face, oral cavity, and the digits. The face of affected individuals may present with asymmetry, frontal bossing, hypertelorism, a broad and flat nasal bridge and different size nostrils. In addition, hypoplasia of the malar bones, cleft or pseudo-cleft lip, and vanishing milia on the face and ears. The hair is most commonly dry and brittle with zones of alopecia.

Abnormalities of the oral cavity occur in many types of the oral-facial-digital syndrome, which include cleft or highly arched palates, cleft or an unusual lobed shape tongue, growth of non-cancerous tumors on the tongue. Other characteristic features are the hyperplastic frenula that abnormally attach the lip to the gums; in addition to alveolar ridge notching and thickening. Furthermore, teeth present as enamel dysplasia, supernumerary teeth, missing teeth, impaction whether with or without retained deciduous teeth.

Gnathical variations which may present in these patients are an anterior open bite and maxillary or mandibular micrognathia. Digital malformations affect hands more frequently than feet, and it includes syndactyly (fusion of digits), brachydactyly (shortened digits), clinodactyly (curved digits), and polydactyly (extra digits).

The aim of this presentation is to report a case of oral-facial-digital syndrome, type I.

THE CASE

A thirteen-year-old female presented with oral-facial-digital syndrome for assessment of malocclusion. The patient was delivered by Cesarean section. At presentation, the patient had an average motor and sensory development. The parents were healthy and unrelated.

The patient’s mental and motor development was within the normal range and did not illustrate any evidence of conduction deafness. Upon extra-oral examination, the patient presented with brachydactyly (shortened fingers) of the index, middle and ring fingers of both hands, and clinodactyly (curved fingers) of the bilateral fifth fingers, see figure 1 (A). Head and neck examination revealed mild facial asymmetry, frontal bossing, broad nasal base, uneven nostrils, hypertelorism, pseudoclefing of the lips, vanishing milia on the face, dry skin and coarse hair, see figure 1 (B). Mildly low setting ears were seen from the lateral view, see figure 1(C-D).