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Oral-facial-digital syndrome- A case report

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Abstract
The oral-facial-digital (OFD) syndrome is a heterogeneous group of abnormalities that share anomalies of the oral cavity, face and digits of hands and feet. On the basis of other anomalies of brain, kidneys, limbs, eyes and other organs, at least 13 subgroups have been described. With the exception of OFD types I and VIII, which are X-linked, the majority of OFDS is trans- mitted as an autosomal recessive syndrome. A number of genes have already found to be mutated in OFDS and most of the encoded proteins are predicted or proven to be involved in primary cilia/basal body function. Here we present a three month old baby who was born at our department with clinical phenotype of oral facial digital syndrome.

Keywords: oral-facial-digital (OFD) syndrome; oral anamolies; polydactyly; genetic and clinical heterogeneity

Introduction

The oral-facial-digital syndromes (OFDS) represent a group of rare developmental disorders characterized by abnormalities of the face, oral cavity and digits. Additional signs involving the central nervous system (CNS), and visceral organs, such as the kidney, are also frequently observed. The first case presenting this condition was reported in 1941 [1] and since then a number of different OFDS types with overlapping phenotypes have been described [2, 3] (Table 1). Among the different types, OFD type I is the most frequently observed and can be easily recognized by its typical X-linked dominant male-lethal pattern of inheritance in familial cases. Most of the other OFDS are transmitted as autosomal recessive syndromes or represent sporadic cases. In the last few years, 11 genes responsible for OFDS have been identified allowing a better clinical and genetic definition for this heterogeneous condition. On the basis of the recent molecular data, we can distinguish (1) two more common types (OFDI and OFDVI), for which the causative genes have been identified; (2) four rare subtypes for which the causative gene has also been identified (OFDIII, OFDIV, OFDIX and OFDXIV), thus allowing molecular diagnosis; (3) two unclassified rare OFD subtypes whose causative genes have been identified but that still require further clinical and molecular validation and (4) additional unclassified OFDS which still await molecular characterization and further definition.

Case report

Term male child was born at our institution with abnormalities of mouth and extremities and respiratory distress at birth. There was no significant antenatal history with no abnormalities of the mouth or extremities in the parents. Her mother had no miscarriages or stillbirths. Relevant findings were as follows. The facial appearance was unusual with flat malar bones, slight hypertelorism, and small epicanthic folds. The upper lip had a slight, midline 'pseudocleft' of the vermilion border, and the nose was short and uptilted. There was frontal bossing. The hair, which was fine and dry, was sparse in both parietal regions but there was no frank alopecia. The hard palate was intact but had a high arch, and lateral ridges, and the soft palate was normal. There were three frenula on the upper lip, while three similar frenula on the lower lip appeared to divide the alveolar ridge. The tongue was cleft into four lobules, with small, yellowish, fibrous lumps between the clefts (Fig. 1). There was a partial web between the fourth and fifth digits of the left hand with bilateral postaxial polydactyly Radiography was normal and chromosome analysis showed a normal karyotype.

Fig 1

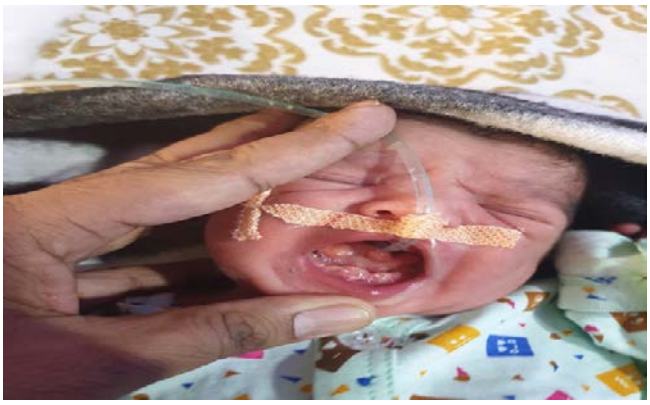

Fig 2

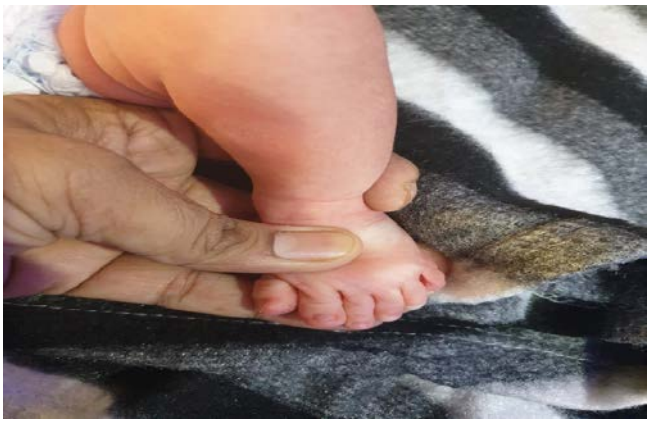

Fig 3

Discussion

There are now four separate oral facial digital (OFD) syndromes, designated as I to IV. Unfortunately, there is considerable overlap in the features and precise clinical identification is still difficult. The original description was by Papillon-Leage and Psaume in 1954 which described type 1 to have tongue nodules, bifid tongue, midline lip cleft, cleft palate, frenulae hypertrophy with thick lateral alveolar incisor bands absent. The original description of type 2 was given by Mohr in 1941.2 Mohr reported four males in a single sibship. Only one male was fully described; the three others had died previously. Subsequently, a male cousin was reported as being similarly affected (the parents were first cousins). One patient had unilateral syndactyly of fingers 3 and 4. The first and fifth toes were broad and the distal phalanges were duplicated. The designation of type III has been given to the sibship of Sugarman *et al*. They reported mentally retarded female sibs with a lobulated hamartomatous tongue (in one), a bifid uvula, extra small teeth with malocclusion, and a bulbous nose. Facial clefts and hypertrophied frenulae were not present. They had bilateral postaxial polydactyly of hands and feet. The Joubert-Boltshauser syndrome is characterised by cerebellar vermis aplasia, mental retardation, rotatory nystagmus, alternating apnoea, and hyperapnoea. Inheritance is autosomal recessive. The occasional patient has polydactyly and tongue tumours which make differentiation from OFD difficult.8 The sibship reported by Gustavson et att falls into the category but it is not yet resolved whether they form part of the OFD phenotype or that presence of the severe tibial aplasia that differentiates type IV from type I.

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