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Case Study

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CASE REPORT: TRANSITIONAL ASPECT OF ORAL-FACIAL-DIGITAL SYNDROMES

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ABSTRACT

The oral-facial-digital syndrome is a heterogeneous group of abnormalities that share anomalies of the oral cavity, face and digits of hands and feet. Here we report a case of neonate with oral-facial-digital syndrome diagnosed clinically as a transitional type between OFD II and OFD IV. The report points out the difficulty in classification of the subtypes of OFDS as significant overlap and variability in each type exists.

KEYWORDS: Oral-facial-digital syndrome, lobulated tongue, polydactyly, pseudo cleft lip.

INTRODUCTION

Oral-facial-digital syndromes (OFDSs) consist of a group of heterogeneous disorders characterized by abnormalities in the oral cavity, face, and digits and associated phenotypic abnormalities that lead to the delineation of 13 OFDS subtypes with different modes of inheritance.[1] The spectrum of anomalies includes variations in findings in the oral cavity: cleft palate/uvula, multiple frenulae, tongue nodules and hamartomas; in the face: hypertelorism, cleft lip,alar hypoplasia, bulbous nose and digital fi ndings; and in hands and feet: pre/postaxial polydactyly, syndactyly, and brachydactyly. Anomalies of brain, eye, heart, kidney, genitalia have been described, of which some are specifically connected with specific subtypes of the syndrome.[2] However classification into the subtypes is not always easy or clear, and additional subgroups has been proposed.[3-5] Here we report a case of OFDS showing several features of OFDS II with some overlapping feature of OFDS type IV.

CASE REPORT

A 2 day old male neonate born to a non-consanguineous marriage, healthy young parents with an unremarkable family history. The pregnancy was uneventful and the delivery was full term, birth weight 2700 grams.

On examination the following dysmorphic features were found: retrognathia, wide forehead, broad nasal root and low set ears as shown in Fig 1. The tongue was having multiple well to ill define round to oval lobules size ranging from 0.3cm X 0.3cm approx over the anterior surface of tongue as seen in Fig 2. The upper lip had a pseudo central cleft lip. The left hand showed preaxial and postaxial polydactyly (8 fingers), Fig 3. The right hand also showed pre-axial polydactyly (6 fingers). The left and right leg showed polydactyly with complete syndactyly between the thumb and extra finger, Fig 4.

Figure 1: Dysmorphic features including retrognathia and low set ears.

Cardiac, chest, abdominal examinations were all normal. Vision and hearing were also normal. Neurological examination showed normal tone and reflexes. Neurosonogram was grossly normal. Ultrasound abdomen and Echocardiography were also normal.

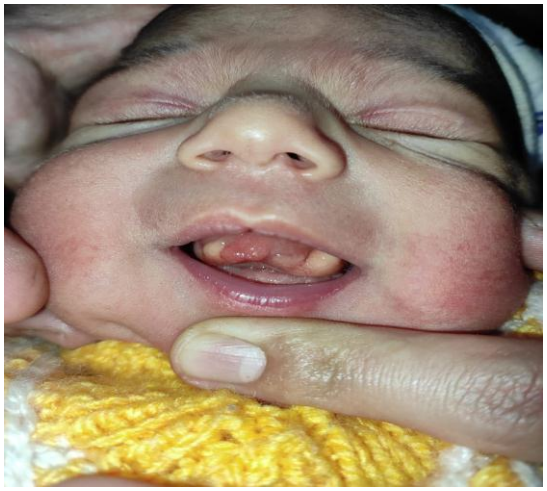

Figure 2: Facial features including lobulated tongue and pseudo cleft lip.

Figure 3: The left hand showing polydactyly (8 fingers) with partial syndactyly between the thumb and the extra finger.

Figure 4: b/l legs showing preaxial polydactyly with complete syndactyly between the great toe and extra finger.

DISCUSSION

The oral-facial-digital syndromes (OFDSs) result from the pleiotropic effect of a morphogenetic impairment affecting almost invariably the mouth, face and digits. Other organ systems can be involved, defining specific types of OFDS. Thirteen types have been distinguished based on characteristic clinical manifestations.[6]

Our patient had several typical and common features of OFDS II along with some overlapping features of OFDS IV. Typical feature included lobulated tongue, low nasal root and pre/post axial polydactyly. Typically in patients with OFDS type II, the hands shows postaxial polydactyly, and the feet show preaxial polydactyly.[7] Our patient showed preaxial and postaxial polydactyly in both hands and preaxial polydactyly in both feet. Our patient also had low set ears and micrognathia which were more commonly found in OFDS type IV.

Typical OFDS II patients have a midline pseudo cleft of the inferior vermilion border of the upper lip giving the upper lip a distinctive appearance. However asymmetric true clefts can occur. Also midline complete or submucous clefts in the primary or secondary palate are common in these patients.[8] Our patient also suffered from a central midline pseudo cleft lip.

OFDS II is usually inherited in an autosomal recessive manner. In our patient no consanguinity was present among the parents. Also there is no history of a similar condition in the family. So we consider our case as sporadic or inherited in an autosomal recessive manner as he is the first and the only child of his parents. Our patient may represent an additional subgroup of OFDS or a variant of OFDS II or a transitional type of OFDS II and OFDS IV.

CONCLUSION

Significant overlap exists between the types and patient may be classified as having a transitional type of OFDS when the feature of several types exists. Our patient support the hypothesis that clinical variability of OFDS II is wider than previously known and in order to find the correct diagnosis and offer a genetic counseling, it is necessary to search for more abnormalities associated with the OFDS spectrum of defects.

Conflict of interest

None.

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