Case Report

Mohr-Claussen Syndrome or Oro-Facial-Digital Syndrome (OFDS) Type-II

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Abstract

The Mohr-Claussen syndrome or oro-facial-digital syndrome type II (OFD-II)] is characterised by tongue lobulation, midline cleft lip, high arched or cleft palate, broad nasal root with wide bifid nasal tip, hypertelorism, micrognathia, brachydactyly, syndactyly and polydactyly, bilateral reduplicated hallux, conductive hearing loss and normal intelligence. In view of the different modes of inheritance and the different prognoses of the two oro-facio-digital syndromes, type 1 and type 2, it is important to establish a correct diagnosis in these patients. A neonate with features of oro-facio-digital syndrome, type-II is being reported and the distinguishing clinicoradiological features with type-I are compared.

Introduction

Oro-facial-digital syndromes are a heterogeneous group of developmental disorders in which at least nine different forms have been described. Oro-facial-digital syndrome type-II OFD-II is transmitted as an autosomal recessive condition and is characterized by malformation of face, oral cavity and digits.[1] Facial and oral features include frontal bossing, facial asymmetry, broad nasal bridge, cleft upper lip and cleft palate, lobulated tongue. Digital features include clinodactyly, syndactyly, brachydactyly, pre- and post-axial polydactyly and duplication of the first toe. Other systemic features include conductive deafness, congenital heart defects and renal abnormalities. in variable combination. Diagnosis is mainly clinical. We report a male neonate with Mohr-Claussen Syndrome or Oro-Facial-Digital Syndrome type-II (OFD-II). Our case has got certain features, which are uncommon and not reported earlier to the best of our knowledge, such as diagnosis in neonatal age, bilateral congenital talipes equinovarus (CTEV) defect and no cardiological or renal abnormalities.

Case Report

A full term male neonate was born out of non-consanguineous marriage (father 32 year, mother 27 years) to a first gravida mother by caesarian section (indication being less foetal movement). The family history was negative for genetic diseases and mental retardation. There was no history of radiation exposure or any significant drug intake, or any major illness during pregnancy period. The neonate did not require any active resuscitation at birth. At birth the APGAR score was 6 at 1min. and 9 at 5min.

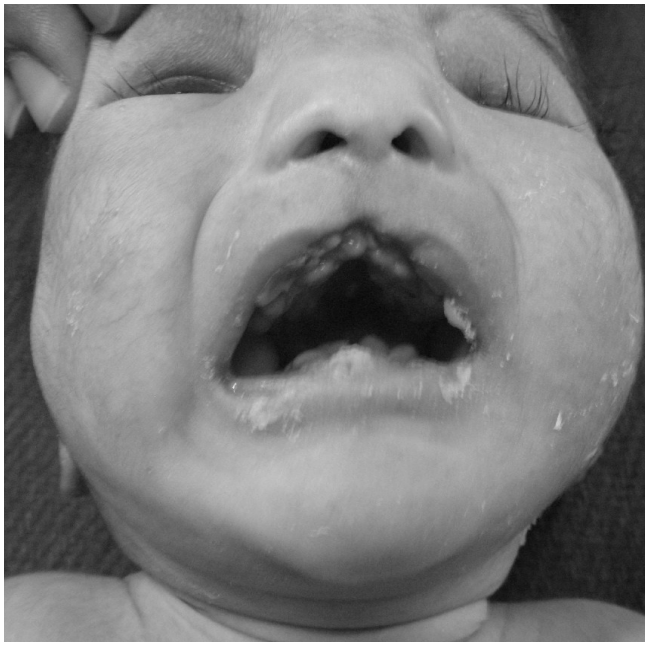

Figure-1: Facial and palatal abnormalities.

He came to our attention at birth. Anthropometric data showed weight 3200 g (> 90th centile), length 55 cm (> 90th centile), head circumference 36.5 cm (> 90th centile). Clinical examination revealed craniosynostosis, hypertelorism, unilateral micropthalmia, broad nasal bridge, low set ears, high arched palate (Fig-2), ankyloglossia,

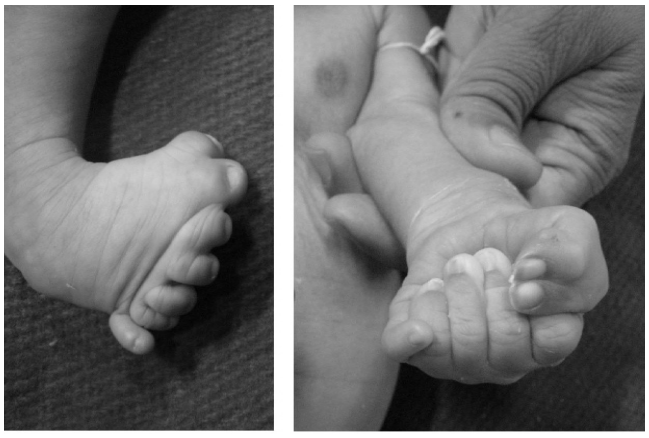

Figure-2: Polysyndactyly of both upper and lower limbs with bilateral CTEV.

partial central cleft lip (Fig-2), lobulated tongue, pectus excavatum, pre-axial polysyndactyly of both hands (Fig-1), pre-axial polysyndactyly of both feet (Fig-1); cardiac, respiratory and abdominal examination was unremarkable, the male external genitalia were normal, and the neuromotor examination was adequate for age.

X-ray revealed well develop calvarium, normal thoracic cage, bilateral pre-axial polysyndactyly of hands with duplication of the 1st finger, duplication of the 1st rays of both feet, bilateral enlargement of the distal metaphyses of humerus and femoral bone, no morphostructural abnormalities of the spine or other skeletal segments. Abdominal and cranial ultrasonography were normal. Echocardiography showed normal study except with thickened interventricular septum and mild narrowing of the aortic arch at the ductal level. ECG was normal.

Cranial CT scan was normal. Brainstem auditory evoked potentials showed reduced amplitude of the 5th wave bilaterally and on left side, increased latency, increased interpeak I-V and irregular morphology. Cytogenetic analysis on peripheral lymphocytes was 46XY.

We found it very difficult to initiate feeding in this baby. Due to the structural abnormalities of lip, palate and tongue, suckling of breast was futile and initial attempts of katori-spoon feeding were rewarded with multiple choking episodes suggestive of aspiration. We therefore started orogastric feeding via infant feeding tube with expressed breast milk. Later we again tried oral feeding with a long handled spoon and the baby tolerated well. The neonate having no other problem was managed conservatively and discharged subsequently with advice of further follow up for evaluation of surgically correctable abnormalities. Genetic counseling of the parents was done.

Discussion

Oro-facial-digital syndromes (OFDS) are a heterogeneous group of rare malformative diseases, characterized by abnormalities of the oral cavity, maxillo-facial region and digits. Such phenotypical pattern was first described by Mohr in 1941[1] and later defined as oro-digital-facial dysostoses by Papillon-Léage and Psaume in 1954[2] and finally named OFDS in 1967 by Rimoin and Engerton.

There are at least 9 different forms of OFDS on the basis of inheritance transmission pattern and phenotypical spectrum, of which the first two types are of common occurrence as compared with other varieties.[3,4]

Mohr-Claussen syndrome (OFDS II) is a rare autosomal recessive disease whose diagnosis is based only on clinical evidence. The molecular genetic basis is still unknown. Because of the variable clinical expression, even intrafamilial, the attribution of the correct diagnosis among the several forms of OFDS is often difficult.[5] In addition, the molecular genesis is still unknown for all OFDS except for the Papillon Léage - Psaume syndrome, which is related to the CXORF 5 gene (Xp22.2-22.3) coding for OFD1 protein.[6] Therefore, in order to achieve the correct diagnosis and offer adequate genetic counseling, it is necessary to search carefully for any possible abnormality associated with the oro-facial-digital spectrum of defects. In the present patient, the presence of transmission deafness and the characteristic facial, oral and limb abnormalities allowed us to recognize the diagnosis of Mohr-Claussen syndrome at neonatal age, considering the differential diagnosis with OFDS I.[7] Young LW,[8] et al have reported a case of short-rib polydactyly syndrome compounded with. Mohr oral-facial-digital syndrome. Our case does not have any associated rib cage anomaly.

Table: OFDS: Main clinical characteristics and inheritance transmission pattern.

Nomenclature	Inheritance	Clinical spectrum
OFDS I Papillon Léage Psaume 1954[2]	X-LD	Clinodactyly, syndactyly, brachydactyly, cleft lip, cleft palate, tongue nodules, micrognathia, alopecia, absent lateral incisors, cerebral abnormalities, renal dysplasia, mild mental retardation.
OFDS II Mohr-Claussen[1]	AR	Clinodactyly, syndactyly, brachydactyly, pre- and post-axial polydactyly, duplication of the 1st toe, absent medial incisors, bifid nasal tip, transmission deafness, mental retardation, congenital heart defects.

These patients apparently have normal intelligence and plastic surgery is indicated for cleft lip or palate, hypertrophied frenula and partial reduplication of the hallux. Ghossaini SN et al[9] has reported a case of Oral-facial-digital syndrome type II variant associated with congenital tongue lipoma. Our case however did not have tongue lipoma.

A surgical attempt to reconstruct the auditory ossicles should be made to improve the conduction deafness. Early accurate diagnosis is important from a genetic counselling point of view, since it implies a one in four risk of recurrence. We suggest that every newborn or infant with structural abnormalities of face, oral cavity and digits should be evaluated for oro-facial digital syndrome spectrum for future outcome and genetic counseling.

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