The Egyptian Journal of Medical Human Genetics (2013) **14**, 311–315

Ain Shams University

The Egyptian Journal of Medical Human Genetics

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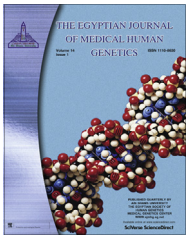

CASE REPORT

Oral-facial-digital syndrome type II: Transitional type between Mohr and Varadi

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Received 17 February 2013; accepted 12 March 2013
Available online 16 April 2013

KEYWORDS

Polydactyly;
Cleft lip;
Congenital heart;
Oral-facial-digital syndrome;
Mohr syndrome

Abstract We report a 2 months old boy, the first in order of birth of non-consanguineous parents, with several typical features of oral-facial-digital syndrome type II (OFDS II) including cleft lip, high arched palate, retromicrognathia, preaxial polysyndactyly of hands and feet, duplication of thumb and hallux. Interestingly, the patient also had mesoaxial polydactyly of the left hand with extra metacarpal bones characteristic of OFDS. VI, however mentality and MRI brain were normal. This unusual association may suggest an additional subgroup of OFDSs or a variant of OFDS II due to variable gene expression or a transitional type between OFDS II and OFDS VI.

1. 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]. However, the features of the various types significantly overlap, and some types are not yet well defined. The classification system for oral-facial-digital syndromes continues to evolve as researchers find more affected individuals and learn more about this disorder [2].

Oral-facial-digital syndrome II (OFDS II) is characterized by frontal bossing, broad nasal bridge, midline cleft upper lip and palate, lobulated tongue, clinodactly, syndactly, brachydactly, pre-and post axial polydactyly, and duplication of the first toe. Other systemic features include conductive deafness and congenital heart defects [3], however the molecular diagnosis is still unknown.

Here we report a case of OFDS showing several features of OFDS II (Mohr syndrome) with some overlapping features of OFDS VI (Varadi syndrome).

2. Case report

A two month old boy, the first in order of birth of non-consanguineous Egyptian parents, the age of the mother was 19 years, and the father was 27 years. The patient was 3.8 kg at birth (at 75th percentile) after normal vaginal delivery. During pregnancy the mother took effervescent tablets for treatment of urinary crystals between the 5th and 6th months of pregnancy, one injection for allergy at the 4th and again at the 8th month of pregnancy as well as vitamin supplementation.

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Peer review under responsibility of Ain Shams University.

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http://dx.doi.org/10.1016/j.ejmhg.2013.03.005

The patient was referred to the Genetics Clinics, Children's Hospital, Ain Shams University for his abnormal features, and allergic rash covering the whole body. There was no history of a similarly affected family member. At the age of two months, the patient had an average motor and mental development as he could support his neck, and started to recognize his mother. His weight was 3.4 kg (< 5th percentile), length was 53 cm (at 5th percentile) and skull circumference was 35 cm (< 5th percentile), with open anterior fontanel measuring 4×4 cm.

The patient had hairy forehead, transverse slanting palpebral fissures, with wide abnormal left nostril, broad nasal root, and small low set ears. The mouth showed non median left sided cleft lip and scar like tissue on the right side, hyperplastic frenula attaching the upper lip and the gum, with high arched narrow palate. There was retromicrognathia, microglossia, with small nodules on the under surface, and the right lateral side of the tongue, Fig. 1. The right hand showed preaxial thumb like polydactyly (6 fingers), with partial syndactyly of the thumb and the extra finger (duplication of the thumb), Fig. 2. The left hand also showed polydactyly (7 fingers), near complete cutaneous syndactyly between the 5th and 6th fingers, partial syndactyly between the 3rd and 4th, and between the 4th and 5th fingers (mesoaxial polydactyly), and duplication of the terminal phalange and nail of the thumb, Fig. 3.

The feet showed bilateral duplication of the big hallux with wide gap between them (6 toes), abnormal position of the second toe, medial deviations of the other toes, nail dysplasia with partial syndactyly between the 2nd and 3rd toes, Figs. 4 and 5.

Cardiac examination detected a harsh pansystolic murmur propagated all over the heart of grade 2. Chest, abdominal, and genital examinations were normal. Vision and hearing were also normal. Neurological examination showed normal tone and reflexes.

Feeding was normal at birth, and then the patient started to suffer from choking episodes, suggestive of aspiration at the age of 3 months. Therefore orogastric feeding via an infant feeding tube was started with expressed breast milk and artificial milk. The patient had repeated attacks of chest infection, and then he died at the age of 4 months with aspiration pneumonia.

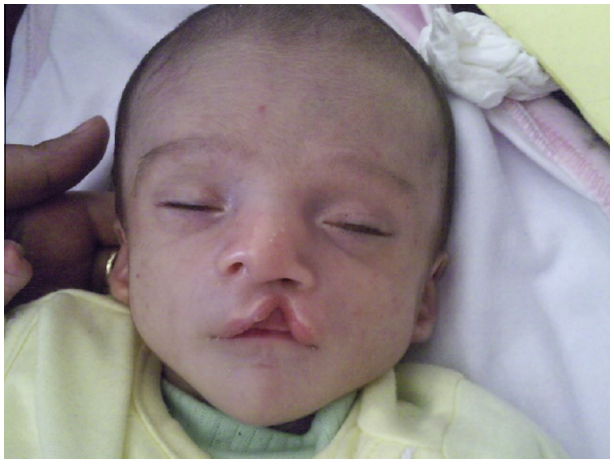

Figure 1 Facial features including hairy forehead, transverse slanting of palpebral fissures, with wide abnormal left nostril, broad nasal root, left lateral cleft lip and scar like tissue on the right side.

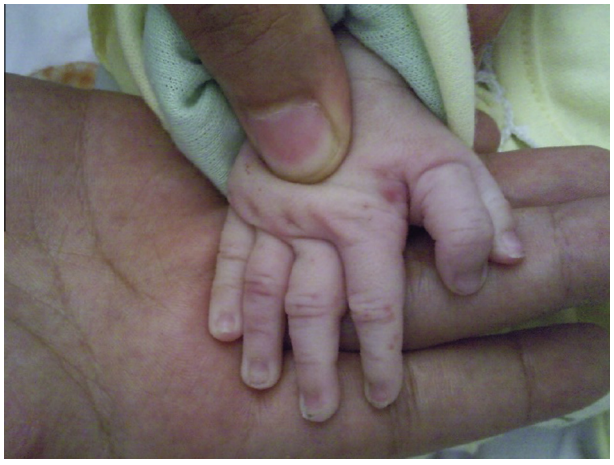

Figure 2 The right hand showed preaxial polydactyly (6 fingers), with partial syndactyly of the thumb and the extra finger.

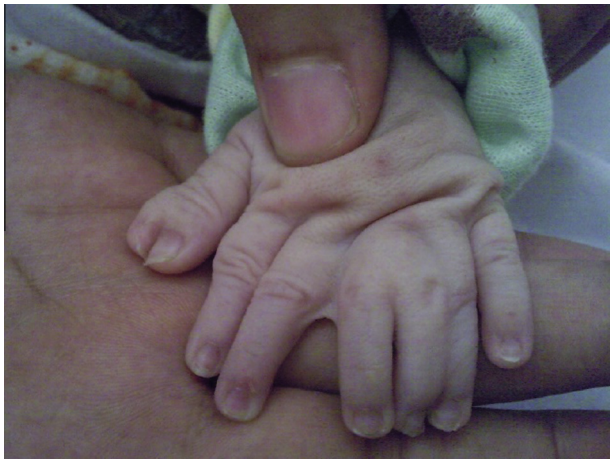

Figure 3 The Left hand showing polydactyly (7 fingers), near complete syndactyly between the 5th and 6th fingers, partial syndactyly between the 3rd and 4th, and the 4th and 5th fingers, as well as duplication of the terminal phalanges of the thumb.

X-ray of the right hand showed radial polydactyly in the form of duplication of the thumb, with its two metacarpal bones fused in their proximal three fourths, Fig. 6. X-ray of the left hand showed mesoaxial polydactyly (6 metacarpal bones), with the proximal phalanx of the middle finger fused with the duplicated one at its head. It also showed radial polydactyly with complete duplication of the thumb (extra metacarpal and phalanges are seen), Fig. 7. X-ray of both feet showed preaxial polydactyly of the first ray, in the form of the presence of extra metatarsal and phalanges of the big toe. The second metatarsal bone is relatively short (Fig. 8).

Echocardiography showed common atrioventricular canal (AVC) with pulmonary hypertension. Laryngoscopy showed epiglottic hypoplasia. Abdominal ultrasonography, and MRI brain showed no abnormalities. Karyotype revealed 46, XY normal male karyotype.