Mohr Syndrome- A Rare Case Report

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Abstract:

An orofaciodigital syndromes (OFDS) is a rare genetic disorder with diverse collection of abnormalities, mainly affecting the face, oral cavity and digits. Orofacial digital syndrome type II, also called the "Mohr syndrome" is a very rare subtype of OFDS. We report a case of 1-year-old male child presented to Paediatric outpatient department with complaints of syndactyly along with polydactyly associated with characteristic features of OFDS type II. Considering the overlapping clinical features, it is important to establish a correct diagnosis by distinguishing appropriate features.

Key words: Orofaciodigital syndrome, Mohr syndrome, Lobulated tongue, Typical facies, Post and preaxial polydactyly.

Introduction:

Oral-Facial-Digital Syndrome (OFDS) is a rare congenital condition with an incidence ranging from 1 in 50,000 to 1 in 250,000 live births. Presently, at least 13 clinical types of OFDS have been identified and distinguished according to the anomalies.[1-4]

Most of them share facial abnormalities, digital anomalies such as brachydactyly, syndactyly, clinodactyly, and polydactyly, and oral features such as cleft or lobulated tongue, oral frenula, and/or cleft palate. The remarkable variety for OFDS is reflected in cases with odd anomalies, even if it is frequently challenging to determine whether they represent other conditions or varied expressivity due to the presence of other abnormalities. There is phenotypic overlap among the OFDS.

Mohr syndrome (OFDS type II) (OMIM #252100), is characterised by poly, syn, and brachydactyly, lobulated tongue with papilliform protuberances, an angular form of the mandibular alveolar process, supernumerary skull sutures, and an episodic neuromuscular disturbance.[5] Cleft lip, cleft or severely arched palate, bifid uvula, cleft or hypoplastic maxillary and/or mandibular alveolar ridge, oral frenula, lingual hamartoma, and absent or hypoplastic epiglottis are just a few of the numerous oral abnormalities associated with this disease that have been documented. Anomalies of the teeth are frequent and usually involve variations in the number of teeth.[6-11] However, presently there is no specific test for identifying the other types of OFDS. OFDS patients required medical care and some surgeries such as the reconstruction of cleft plate and limb deformities.[12] In this case report a 1-year-old male child presented to Paediatric outpatient department is reported.

Case Report:

A 1-year-old male child presented to Paediatric outpatient department with complaints of polydactyly along with syndactyly. He is a full-term male baby born via normal vaginal delivery cried immediately after birth with birth weight of 1.8kg with history of NICU stay in view of cleft palate and was on orogastric tube feeds and was discharged on paladai feeding. He is the second child of healthy parents who were first cousins. No similar findings in previous child were present. Child has hypoplastic tongue, retrognathia (Fig.1), polydactyly-syndactyly (Fig.2), cleft palate, epicanthal folds, ear tag, bilateral congenital talipes equinovarus (Fig.3) and micropenis (Fig.4).

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As the proband's birth certificate indicates, she was born with a birth weight of 2450 gr, length 45 cm, and her head circumference was 32.5 cm (all <3rd centile). Her radiographs revealed an extra phalanx between the fourth and fifth fingers, bifid thumb, and postaxial digit in the right hand.

The hemogram and the urine report was normal. The chest radiograph, ECG, abdominal ultrasound, and cranial MR revealed no abnormalities. Hearing loss was detected in her left ear with auditory brain responses. Chromosome analysis using Gbanding technique revealed a 46,XX karyotype. At that time, he was diagnosed as having OFDS type IV.

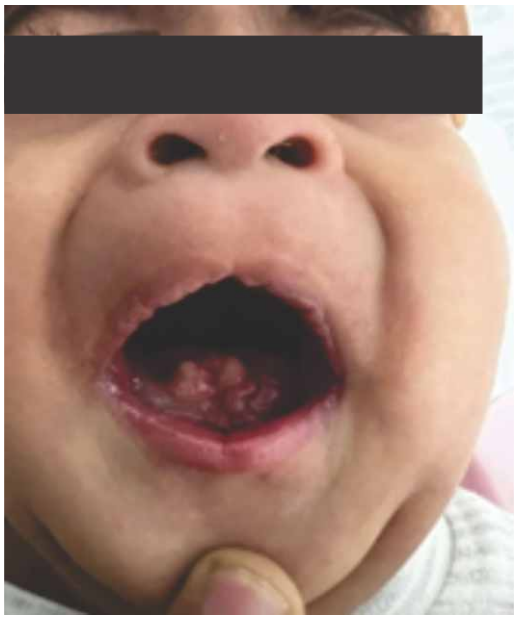

Figure 1: showing lobulated and hypoplastic tongue and retroganathia.

Figure 2: showing polydactyly and syndactyly

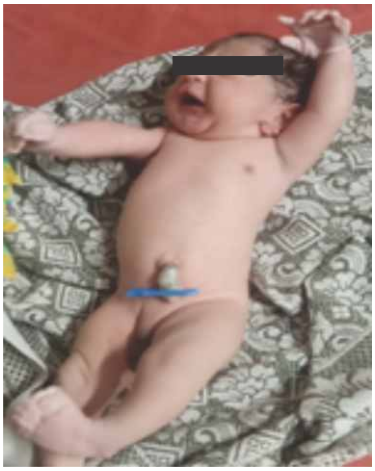

Figure 3: showing baby with bilateral congenital tallipes equino varus

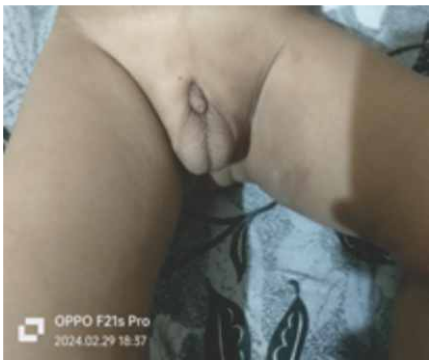

Figure 4: showing micropenis.

Discussion:

OFDS consists of wide variety of symptoms which make diagnosis confusing. The most common form, OFDS Type I, results from mutations in the Xp22 gene located on chromosome X (regions 22.3–22.2).[1,3,4] This gene is critical for the development of several organs, including the brain, face, limbs, and kidneys, and encodes a protein found in the centrosome and basal body of primary cilia. OFDS is classified as a ciliopathy due to its molecular basis and clinical presentation.[13]

It is essential to distinguish various OFDS types due to associated involvement of heat, kidney and central nervous system. OFDS type I is characterized by distinctive features such as small cysts (milia) on the face and ears, underdeveloped nostrils (alar hypoplasia), and hair loss (alopecia).[14] Renal anomalies, such as polycystic kidney disease, are frequently observed. This type often involves anomalies in the heart, kidneys, and central nervous system.[3, 15, 16]

Genetic testing can confirm the diagnosis of Type I, which is critical for appropriate management and genetic counselling.[16]

OFDS Type II (Mohr Syndrome) primarily presents with craniofacial anomalies such as a high-arched palate, a broad nose, wide-set eyes (hypertelorism), and nodules on the tongue. Dental anomalies like the absence of central incisors are common. Unlike Type I, renal involvement is rare in Type II, but it is important for anaesthesiologists to anticipate potential difficulties with intubation due to craniofacial anomaly and high arch palate in type II. Diagnosis relies on clinical features, as the associated gene remains unidentified.[12, 17, 18]

Mohr syndrome is an extremely uncommon form of OFDS. From infancy to adulthood, patients frequently require multiple surgical procedures, so the surgical team should be ready for a challenging intubation.

Conclusion:

It is recommended to look for OFDS in child with polydactyly along with syndactyly and then evaluate for other organ involvement for its appropriate management.

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