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CASE REPORT

ABLEPHARON - MACROSTOMIA SYNDROME: A CASE REPORT

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ABSTRACT

Ablepharon-macrostomia syndrome is an extremely rare congenital disorder apparent at birth. To our knowledge, this is a first case report in an Ethiopian family. The present case manifested unusual clinical features, which are important for better understanding of the cases. A 12-hour male neonate was born on 16, February 2017 from a 40 years old para-gravida and IX at gestational age of 41 weeks + 3 days. This male neonate with Ablepharon-macrostomia syndrome revealed dysmorphic features with absence of eyelids, eyelash and eyebrow. The neonate had an injected and opaque sclera. Bilateral eyeballs were extruded outside of eye socket. The neonate had a flat nasal bridge, deformed low-settled ears and wide "fish-like" mouth. In addition to previously reported cases of Ablepharon-macrostomia syndrome, the present case has wide anterior fontanel and suture lines with dilated 3rd and 4th ventricle (hydrocephalus). Moreover, he has a bilateral medially rotated foot, and absent prepuce and granular hypospadias. The present Ablepharon-macrostomia syndrome case is the first report from an Ethiopian non-consanguineous family with additional unusual features.

Key wards: *Ablepharon, macrostomia, hydrocephalus, medially rotated feet, Ethiopia*

INTRODUCTION

Ablepharon-macrostomia syndrome (AMS) is extremely rare congenital ectodermal dysplasias. AMS is first described in 1977 by McCarthy and West with characterized by absent eyelids, macrostomia, dysmorphic nose and ears, with abnormalities of nipple, genitalia and redundant skin (1). The word Ablepharon means absence of eyelids and macrostomia refers to wide "fish-like" mouth. A recent study identified that recurrent autosomal-dominant mutation in the basic domain of *TWIST2* (Twist Family BHLH Transcription Factor 2) cause AMS and barber-Say syndromes (2). Furthermore, studies suggested that AMS is acquired by autosomal dominant inheritance than autosomal recessive inheritance (3,4).

CASE PRESENTATION

A 12 hours- male neonate was born on 16, February 2017 from a 40 years old-para-gravida and IX (7 alive including this neonate and two deaths) at gestational age of 41 weeks + 3 days. The non-consanguineous father was 48 years old. The two deaths were the 2nd female infant at the age of 3 months and the 4th female child at the age of 4 years old died due to meningitis and malaria. The remaining six children did not have any congenital abnormalities. Mother had antenatal care follow up and was supplemented with iron and given tetanus toxoid vaccines twice. Her VDRL and serology for retroviral infection were negative but hepatitis B virus infection, ABO blood group and Rh No maternal fever or offensive vaginal discharge during her pregnancy. She did not take any teratogenic drugs, alcohol consumption and no exposure to radiation. There was no family history of ocular disorders and no history of diabetus mellitus, hypertension, renal or cardiac problems. Both parents were farmers with low socioeconomic status from rural areas. The labor started spontaneously and duration was 11 hours and membrane ruptured after was 2 hours. The mode of delivery was spontaneous vertex vaginal delivery at local health center. The newborn cried immediately after birth.

On the same day, the neonate was referred to FelegeHiwot Referral Hospital for ophthalmic and neonatal care. There was profuse bleeding from umbilical stump on the way to referral hospital as cord tie was loose and not accompanied by health professionals. There was no vomiting, history of fever, fast breathing or grunting. Breast feeding was not initiated as he had dysmorphic featured and mother did not come with the newborn.

The physical examination showed that the neonatal had hypothermic signs. His birth weight and height at 25th centiles were 3265 gram and 48cm, respectively. His head circumference at 75th centile was 36 cm. The neonate's pulse rate (PR) and respiratory rate (RR) were 116 beats and 26 breaths/ minute, respectively. Table 1 illustrates the dysmorphic features and detailed clinical features of the AMS.

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Table 1: Clinical manifestations and hematological values of Ablepharon-macrostomia syndrome.

Characteristics	Anomalies
Eyelids, Eyelash and Eyebrow	Bilateral absent
Eyeball	No movement and Both eyeballs are extruded outside of eye socket
Cornea	Bilateral Opaque
Conjunctiva	Bilateral sever chemosis
Pupil	Both not visualized
Mouth	Wide/"fish-like" mouth
Nose	Depressed nasal bridge and small nasal wing
Ears	Low-set and deformed
Scalp hair	Sparse
Nipples	Hypoplastic
Abdomen	Distended
Genitalia	Male type external genitalia, left testis undescended
Musculo skeletal findings	No prepuce, granular hypospadias, penile chordee and micro penis Congenital talipes equinovarus , camptodactyly and clinodactyly of both thumbs
Integumentary	Hypoplastic nails
CNS development	Conscious, depressed neonatal reflexes
Transfontanel ultrasound	Dilated 3rd& 4th ventricle (hydrocephalus)
Abdomen ultrasound	Normal
Total WBC/mm3	24,000
Neutrophils (%)	71.3
Hemoglobin and hematocrit	18g/dl and 54%
Platelet/ mm3	230,000

Ophthalmic examination showed that eyelid, eyelash, eyebrow and eyeball movement were absent (Figure 1). The neonate has bilateral injected and opaque sclera. His pupil was not visualized. Both eyeballs

The neonate had flat nasal bridge, small nasal wing and deformed low- settled ears. The neonate had wide mouth with no cleft lips or palate. Moreover, he has bilateral medially rotated feet (**figure 2**), and

Figure 1: A 12 hours-old neonate with lack of eyelids, eyebrow and eyelash and bilateral sever chemosis of conjunctiva and opaque cornea. The neonate has a depressed nasal bridge. His has a wide "fish-like" mouth and his nipples are hypoplastic. The neonate had medially rotated feet.

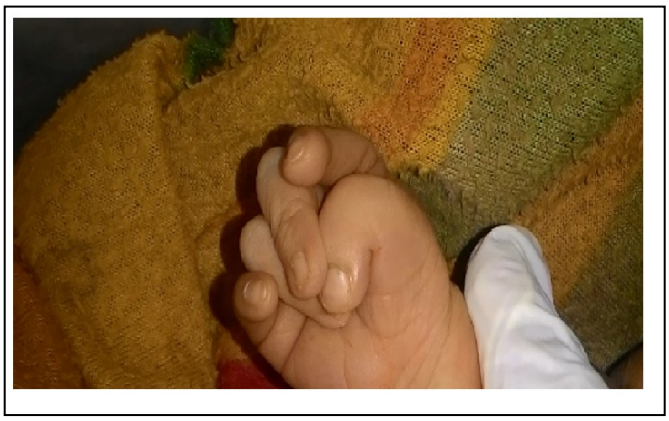

Figure 2: The neonate with camptodactyly and clinodactyly of both thumbs.

Therapeutic focus and assessment

The newborn was resuscitated with 20ml/kg of normal saline bolus and transfused with 20ml/kg whole blood over three hours, and also given vitamin K 5mg Intravenous stat. He was put maintenance fluid for 24 hours of life. He was given intravenous (IV) ampicillin and gentamycin for 7 days. Later he was on formula feeding for seven days. The ophthalmologist was consulted and decided to be given supportive treatment for the eye disorder with saline soaked gauze with tetracycline eye ointment and chloramphenicol eye drops. His father was counseled and given psychological support. The neonate was discharged from the hospital on seven days of age.

Ethics approval

The Research Ethics Review Committee of the College of Medicine and Health Sciences of Bahir Dar University has approved the case report for possible publication. Written informed consent was obtained from the patient for publication of this Case Report and any accompanying images. A copy of the written consent is available for review by the Editor of this journal. Written informed consent was taken from the father for possible case presentations and publication.

DISCUSSION

To our knowledge, Ablepharon- macrostomia syndrome (AMS) case from Ethiopian family is the first time report. AMS is an extremely a rare congenital disorder thus as of 2011 less than 20 AMS cases had been reported worldwide (4). The neonate was borne from non-consanguinity parents. Moreover, the six children of these parents are phenotypically normal for AMS. This indicates that the occurrence of AMS is mostly sporadic (3). In contrast; family occurrence of Ablepharon-Macrostomia Syndrome (AMS) was reported (5).

In the present case, the most common clinical features of AMS were manifested (1,3). In addition to the known clinical features of AMS, this neonate has anterior fontanel and suture lines with dilated 3^{rd}& 4^{th} ventricle (hydrocephalus). Likewise, patients with AMS showed dental malformation which was not present previously (6). However, in our patient chromosomal mutation analysis was not done because of limited laboratory setup.

In conclusion, Ablepharon-macrostomia syndrome (AMS) case is the first report from an Ethiopian non-consanguinity family with additional unusual features. Unusual characteristics of this neonate were the presence of wide anterior fontanel and suture lines with dilated 3^{rd} and 4^{th} ventricle (hydrocephalus), bilateral medially rotated feet, and non-prepuce and granular hypospadias.

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