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CASE REPORT

Otocephaly: A Rare Congenital Birth Defect

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ABSTRACT

Otocephaly is an uncommon congenital birth defect characterized by underdeveloped or absent mandible, misplacement of the ears, a small mouth, and absence or underdeveloped tongue.

We present a case of this congenital anomaly in a newborn baby born to a 27-year- third gravida at 31+2 weeks period of gestation (POG) spontaneously per vaginally. Unfortunately, the congenitally malformed preterm newborn did not survive due to severe respiratory distress. Early diagnosis of this condition is possible but difficult. Extensive research is required to better understand and manage this rare condition.

Key-Words: *Agnathia, antenatal scan, lethal congenital disorder, microstomia, otocephaly*

INTRODUCTION

Agnathia-otocephaly complex (AOC) is a first branchial arch defect responsible for a rare fetal facial aberration, mostly incompatible with life occurring in 1 in 70,000 births. It is characterized by agnathia (hypoplasia or the absence of a portion or the entirety of one or both the mandible), synotia (ventromedial and caudal displacement of the ears with or without the fusion of the ears, the union or approximation of the ears in front of the neck), melotia (congenital displacement of the auricle onto the cheek), microstomia (a small oral aperture) and aglosia (with or without a tongue hypoplasia).[1,2]

AOC has etiologies linked to teratogenicity and two genes disorder, the orthodenticle homeobox 2 (OTX2) gene and the paired related homeobox 1 (PRRX1) gene. The genetic analysis showed a novel 10 bp deletion mutation c.766_775delTTGGGTTTTA in the OTX2 gene, together with a missense variant c.778T>C in cis conformation.[1]

AOC has been diagnosed as early as first trimester on routine USG scan or when investigating for unexplained of polyhydramnios in late second trimester or third trimester.[1,3,4]

We hereby bring forth a case of AOC diagnosed after preterm birth and succumbed to death due to associated respiratory distress from prematurity on the top of lethal congenital malformation.

CASE

A 27- year married out of non-consanguineous marriage and a third gravida with two alive and well baby girls age 8 and 5 years, born out of vaginal delivery at home, was on random antenatal care.

A nondiabetic and normotensive, with B positive blood group, she tested negative for Hepatitis B antigen and both VDRL and HIV tests were non-reactive. She was on regular intake of iron and folic acid and was immunized against tetanus.

There was no history of radiation exposure or any harmful drug intake, neither family history of holoprosencephaly.

Obstetric scan around 28 weeks pregnancy showed features of bilateral fetal hydronephrosis. On repeat scan after a week, amniotic fluid index (AFI) was 30 cm indicating polyhydramnios.

Two weeks later on 27th October, 2023 at 7:36pm she was admitted in Paropakar Maternity and Women's Hospital, Thapathali, Kathmandu, at 31+2-week POG with establish labour pain, and as a sequela to spontaneous preterm labour, eventually had vaginal delivery giving birth to a premature male baby with poor APGAR score 2/10. Placenta was delivered spontaneously. Placenta was complete without any abnormalities. Mother's vital was normal and there was no PPH.

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Baby weighed 1700gm. On gross examination of the newborn baby (Fig1), there was distinctive gross anomalies on physical examination. The baby had absent mandible (agnathia), melotia (anteromedial malposition of ears), microstomia (small mouth) and microglossia. bilateral ears were positioned ventromedially on the neck and appeared fully developed, with proper helix and lobule. However, the lobules and concha of the ears were fused with the skin of the neck region at the ventromedial position. The baby's upper limb and thorax were normally formed. On systemic examination, baby had no other gross abnormalities or abdominal organomegaly. Umbilical cord had two arteries and one vein.

Unfortunately, within the next half an hour baby passed away.

DISCUSSION

It's sad to miss the diagnosis of otocephaly, a rare and lethal condition even at second trimester scan, amidst the presence of polyhydramnios, which is usually connected with this anomaly.[3,4]

Prenatal diagnosis using 3D USG has been utilized.[3] and MRI used to find details on fetal lungs.[4]

Although fetal survival appears futile because of extreme congenital deficit

impairing intubation and tracheostomy, yet efforts are continued, adapting ex-utero intrapartum treatment (EXIT) procedure only in selected few fetuses with high risk of airway obstruction immediately after birth.[4] As a hypoplastic lung cautions against severe respiratory failure and a tension pneumothorax.

Beyond this, virtual surgical planning (VSP) and a vascularized free fibula flap for further mandibular reconstruction has been planned in 10-year- boy, a survivor with milder disease form.[5]

CONCLUSION

Awareness of agnathia-otocephaly complex a fatal fetal facial malformation must be contemplated in conjunction with polyhydramnios to council mother and family, even we have though have very little to offer for the survival of newborn.

Fig 1: Preterm newborn male baby with Agnathia-otocephaly complex (AOC)

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