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Case report

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PATAU SYNDROME- An Unusual Case of Congenital Abnormality

Abstract

Single Eye Baby is a rare presentation of congenital anomalies. We observed a rare case of single eye in the centre of frontal bone of a live baby delivered by LSCS and priory diagnosed by Ultra Sonography.

Keywords: single eye, congenital abnormality, USG diagnosis, LSCS

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Introduction

Congenital malformations can be because of five reasons. These causes are chromosomal disorders, single gene disorder, multifactorially inherited disorders, teratogenically induced disorders and unknown causes. Chromosomal disorders accounts for 50% of all congenital malformations.[1] Single midline eye (Cyclops) with projection over the frontal bone (Proboscis) is a rare congenital anomaly. It is found 1in 10,000 live births. We observed one such case delivered by emergency LSCS at our Hospital.

Background

Patau syndrome, also referred to as Trisomy 13, was first described by Thomas Bartholin in 1657 and cytogenetically identified by Klaus Patau in 1960. It affects approximately 1 in 5000 live births. Typically, individuals with this condition have a median life expectancy of 7 to 10 days, with 90% not surviving beyond the first year of life. Survival beyond this timeframe is occasionally observed due to factors such as mosaicism and the diverse range of associated malformations, which vary in severity.

Case report

A 26 years old unregistered G3 P2 L1A0 D1 with 28 weeks pregnancy with Polyhydromnious was admitted on 4/11/2020 at 6 PM in the maternity ward of our Hospital. She had sudden abdominal pain at home 4 hours back. She was married 5 years back and there is a history of consanguinity. Patient had two children. Both delivered by LSCS at Esra Hospital, Hyderabad. First male baby died after one year due to Congenital Heart Disease and second 5 years old male child was alive and healthy. TIFFA scan was done on 23/10/20 showed smaller head with single midline eye and proboscis. There was evidence of Monoventricular cavity with partial fusion of Thalami. Cavum septum pellucidum was not visualized. Fetal spine, heart, stomach, kidney and urinary bladder were normal. Umbilical cord has two vessels. Another USG was done on 3/11/2020 showed Single Live Fetus of 25 wks and 6 days with cephalic presentation with small sized head, monoventricle and fused thalamus. Ultrasonologist reported as Semilobar holoprosencephaly with Cyclops with proboscis and the Expected Date of Delivery (EDD) was given 10/2/2021. On admission her Hb was 9.90gr% and her blood group was B+ve. The vitals were recorded hourly and first reading on admission was recorded as Blood Pressure was 130/90 mm of Hg, Pulse Rate was 80-86/min regular, Temperature was normal and urine output was 100 ml. On systemic clinical examination Chest was clear. On per abdomen examination height of the uterus was 28 weeks and foetal heart sounds were not clear. Patient was prepared for Emergency LSCS under spinal anesthesia (SA) on 4/11/2020 at 10.45PM. Patient delivered a grossly abnormal alive female baby with apgar score 2-4, wt 1 kg. The baby has single midline eye (Cyclops) with projection over the frontal bone (Proboscis).

Pt. was monitored after LSCS. Her condition was stable. The baby was expired on 5/11/2020 at 10.40 PM.[2]

Discussion

It is hypothesized that the anomaly may be Trisomy 13 or Trisomy-D (Patau Syndrome) which is nearly always fatal during fetal or early postnatal life. Affected infants have numerous malformations. These are defects of eyes, nose, lips and holoprosencephaly with incomplete development of forebrain, olfactory and optic nerve.[3]

They are small for gestational age and are microcepahlic, and midline facial anomalies including cyclopia, cebocephaly and clefts of the lip and palate are often seen. Midline defects of the brain, including alobar holopresencephaly may present. The forehead is sloping, the ears are small and malformed, and micropthalmia (Small eyes) or anopthamlmia (no eyes) may occur. The hands show postaxial polydactyly and abnormal palmar creases, skin defect in posterior scalp region and the feet are malformed. Males have hypospadias and cryptorchidism and females have hypoplasia of the labia majora. Internally numerous malformations are encountered, usual are congenital heart disease.[1] These features are found in Patau syndrome but one additional feature i.e presence of functioning eye in the centre of frontal bone has been observed, which is not matching with classical description of Patau syndrome.[4] It seems reasonable that condition might have been Trisomy 13, but it can't be said with certainty as chromosomal study could not be conducted (Figure 1 and 2).

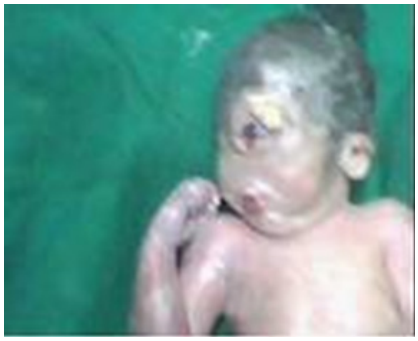

Figure 1 Single eye baby.

Figure 2 Single eye lies in the centre of frontal bone.

Conclusion

Each year, approximately 240,000 newborns worldwide succumb to congenital disorders within 28 days of birth, with an additional 170,000 children aged between 1 month and 5 years succumbing to these conditions. The impact of congenital disorders extends beyond mortality, often leading to long-term disability, placing substantial burdens on individuals, families, healthcare systems, and societies. Alarmingly, nine out of ten children born with serious congenital disorders hail from low- and middle-income countries, highlighting the pressing need for global attention and equitable access to healthcare resources. Some congenital disorders can be prevented. Vaccination, adequate intake of folic acid or iodine through fortification of staple foods or supplementation, and adequate care before and during pregnancy.

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Conflicts of authors

The authors declare that there are no conflicts of interest.

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