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A RARE CASE OF EXTREMELY LOW BIRTH WEIGHT NEWBORN WITH ALOBAR HOLOPROSENCEPHALY PRESENTING CYCLOPIA WITH SUPERIOR PROBOSCIS WITH HYDROCEPHALUS WITH MACROCEPHALY

Neonatology

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ABSTRACT

OBJECTIVE: To report a rare case of a Cyclopia baby with superior proboscis with hydrocephalus with macrocephaly and its importance in timely detection of anomalous babies.
INTRODUCTION: Cyclopia is a congenital disorder, a rare form of holoprosencephaly, characterized by the failure of embryonic prosencephalon to properly divide the orbits of eye into two cavities with grossly incomplete morphogenesis of fore brain. Typically, the nose is either missing or replaced with no functioning nose in form of a proboscis. A prenatal anomaly scan can help in the early detection of the condition and timely termination of pregnancy.
METHODOLOGY: A cyclopia baby was born from a 40 -year-old apparently healthy lady at SIMS Hapur on 25 June 2021. Mother was diagnosed as G5P4L4, PPROM, congenital anomaly, low lying placenta, moderate anemia, with no previous history of baby with congenital anomaly. Mother was presented at 25-week 6 days POG with complaint of lower abdominal pain and decreased fetal movements for 3 days.
RESULT: A ELBW, female fetus was delivered at SIMS ,Hapur and diagnosed as a case of alobar holoprosencephaly with cyclopia with superior proboscis with ill developed face with hydrocephalus with macrocephaly.
CONCLUSION: This case is presented because of its rarity. Timely detection of anomalous baby is necessary to decrease the suffering of parents and family.

KEYWORDS

INTRODUCTION-
Holoprosencephaly refers to a group of disorders arising from failure of normal forebrain development during embryonic life, with reported frequency of approximately 0.6 per 10,000 live births.[1]It includes a series of complex disorders with a broad range of severity varies from cyclopia which is most severe facial malformation to absent incisor teeth least one.

Cyclopia is a congenital disorder, a rare form of holoprosencephaly, characterized by the failure of embryonic prosencephalon to properly divide the orbits of eye into two cavities with grossly incomplete morphogenesis of fore brain. Typically, the nose is either missing or replaced with no functioning nose in form of a proboscis. A prenatal anomaly scan can help in the early detection of the condition and timely termination of pregnancy. Approximately 1.05 in 100,000 births are identified as cyclopia, including stillbirth.[2]

Case report-
A cyclopia baby was born from a 40 -year-old apparently healthy lady at SIMS Hapur on 25 June 2021. Mother was diagnosed as G5P4L4, PPROM, congenital anomaly, low lying placenta, moderate anemia, with no previous history of baby with congenital anomaly. There was no history of consanguinity, no exposure to known teratogens during pregnancy.

Mother was presented at 25-week 6 days POG with complaint of lower abdominal pain and decreased fetal movements for 3 days. Antenatally the baby, baby was fourth in birth order and product of a non-consanguineous marriage and was diagnosed as a case of hydrocephalus during intrauterine life. Baby delivered spontaneously by NVD at gestational age of 27 week +3 days by USG. Diagnosed as a case of alobar holoprosencephaly with cyclopia with superior proboscis with ill developed face with hydrocephalus with macrocephaly. Baby was extremely preterm with birth weight of 780 g, product of NVD, a female fetus was delivered at SIMS ,Hapur. On examination, baby was cyanosed. Heart rate was 146/minute, and respiratory rate 44/minute, Xiphoid retractions were present.

Apgar was not calculated because of abnormalities at birth i.e. dysmorphic facies, single eye shows duplex formation, absence of nose and eyebrows, low implanted ear, small mouth, a superior proboscis, umbilical cord with one artery one vein. Neither a cleft lip nor a cleft palate was noted. Extremities were normal in structure but cyanosed in appearance. The Newborn expired after 5 minutes of birth.

Fig: 1- Facial photograph of extremely low birth weight newborn with alobar holoprosencephaly shows cyclopia, dysmorphic facies, single eye with duplex formation, absence of nose, low implanted ear, small mouth, a superior proboscis with one umblical artery and one umblical vein.

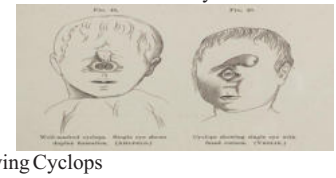

Fig: -2- Showing Cyclops
Investigation-
Obstetric Ultrasound scan was done at 25 week 6 days

USG Scan shows single fetus with breech presentation at the time of scan. Gross fetal cardiac activity and fetal movement was seen. Fetal heart rate was 140 bpm. Amniotic fluid was adequate.

BPD was 7.29 cm, HC was 25.64 cm, Abdo circumference was 19.93 cm, Femur length was 4.46 cm. Fetal weight was approx. 765.0 gram. There was evidence of large Holo ventricle with absence of cerebral cortex, falx and interhemispheric fissure. Macrocephaly was seen. Face was appeared dysmorphic with reduced interorbital diameter and orbito-orbital diameter. There was no proper USG definition of bilateral eyeball and nose. Placenta reaching up to the internal os (low lying) and was of grade 1 maturity.

USG findings are suggestive of Alobar holoprosencephaly with dysmorphic face with fetal hydrocephalus with macrocephaly.
X-Ray- normal
Brain MRI, Chromosomal analysis and post-mortem autopsy were not carried out as consent to these procedures was not given by father.

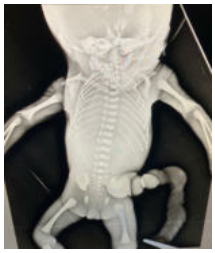

Fig:- 3 - X-ray Of Cyclopia Baby After Birth Was Normal

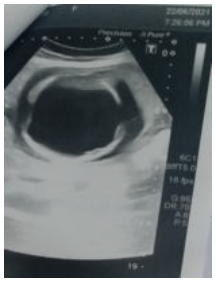

Fig: 4-Prenatal transabdominal USG Scan showing evidence of large holoventricle with absence of cerebral cortex, falx and interhemispheric fissure

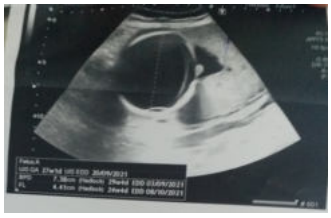

Fig-5- Prenatal transabdominal ultrasound of fetal head showing biparietal diameter. Alobar holoprosencephaly and macrocephaly was noted

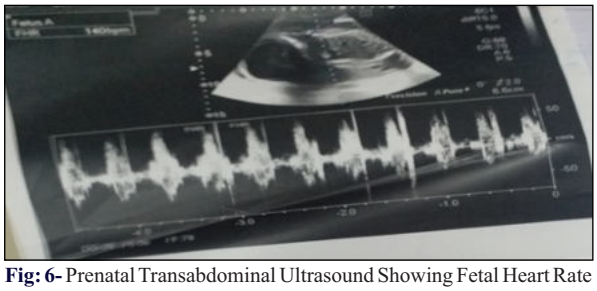

Fig: 6- Prenatal Transabdominal Ultrasound Showing Fetal Heart Rate

DISCUSSION-

A cyclopia with proboscis is a rare form of HPE with few more cases reported worldwide. To our knowledge, this was first case reported from our hospital. The most severe forms of holoprosencephaly frequently involve the face, resulting in severe anomalies include cyclopia, ethmocephaly , cebocephaly , and median cleft lip. Cyclopia refers to a single midline orbit that contains ocular structures that are anopthalmic, monophtalmic, or synophthalmic. A proboscis is usually present and may be doubled. It lies above the orbit, with the nasal and median facial bones missing. Ethmocephaly the next more severe malformation, is the least common facial subtype and consist of severely hypoteloric orbits, usually with marked microphthalmia and a proboscis with absent nasal structure.[3,4] The diagnosis of holoprosencephaly can be suspected in the presence of orbital hypotelorism and median facial anomalies.[2] During the 4th week of gestation, the neural tube forms the three primary brain vesicles (prosencephalon, mesencephalon, and rhombencephalon) and by the 5th gestational week, the prosencephalon further divides into the telencephalon and diencephalon. The two hemisphere and the lateral ventricle arises from the telencephalon, whereas the thalami, hypothalamus, and the basal ganglia arises from the diencephalon. There are three forms of holoprosencephaly: alobar, semi lobar, and lobar varieties, with alobar holoprosencephaly (cyclopia) being the most severe form and characterized by undifferentiated holosphere of the cerebral parenchyma with a central mono-ventricle, fused thalami, and absence of midline structures.[5] In semi lobar holoprosencephaly the two-hemisphere partial separated by a posterior cleft and the united previously observed in a horseshoe shape with a single ventricle and fused thalami. In lobar holoproscencephaly, two hemisphere two ventricles, two thalami but with midline defects and abnormalities of corpus callosum, septum pellucidum and olfactory bulbs. Sonography is the most useful in the prenatal diagnosis of cyclopia.[6] Holoprosencephaly can be expected to present in 16% or more of all cases of fetal hydrocephalus.[7] In our case fetus was diagnosed with hydrocephalus outside the hospital in the second trimester of pregnancy. In our institute USG was done at 25 week + 6 days and USG findings was suggestive of fetal hydrocephalus with macrocephaly. At birth, our case was found to have the typical facial feature of cyclopia.

The originality of our case was that it is the case report of live extremely low birth weight Newborn with cyclopia with, dysmorphic facies, single eye shows duplex formation, absence of nose, low implanted ear, small mouth, a superior proboscis, umbilical cord with one artery one vein, and our case was also present with hydrocephalus with macrocephaly. MRI, chromosomal analysis, and post-mortem autopsy can add to the diagnosis, but in our case, they were not carried out as consent to these was not given by father.

CONCLUSION-

We report a rare case of alobar holoprosencephaly, presenting cyclopia with superior proboscis with hydrocephalus with macrocephaly. The prenatal diagnosis of cyclopia can be made early by ultrasound. The awareness of the spectrum of USG findings of cyclopia can improve the accuracy of prenatal diagnosis and carried out timely detection of anomalous babies and preventing mothers from psychological trauma of carrying such fetus. Cyclopia is a very rare anomaly which one may never have the opportunity in a lifetime to witness.

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