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Case Report

A rare case report of a cyclopian malformation

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ABSTRACT

A rare form of median faciocerebral dysplasia, characterized by a single central orbital fossa with a tubular nose-like appendage above the orbit is known as cyclopian malformation/monster. It is the most severe form of alobar holoprosencephaly. Since most of these cases are sporadic, incompatible with life, and due to the limited literature knowledge, the exact etiology of this condition remains undetermined. However, various risk factors implicated include genetic factors and chromosomal anomalies (mostly trisomy D). Here we present a case of stillborn male cyclopian fetus born to a 34 year old 3rd gravida by caesarean section. There was no history of any drugs or alternative medicine intake (except iron-folic acid, calcium, thyroxin), radiation exposure, or a significant family history or consanguinity. Her only 33-week scan (done at a peripheral center) failed to identify any fetal abnormality. This case is reported because cyclopia is a rare/uncommon developmental anomaly especially with the advancement in antenatal ultrasonography to identify malformed fetuses early in pregnancy.

Keywords: Cyclopia, Holoprosencephaly, Cyclopian malformation/monster, Stillbirth

INTRODUCTION

A rare form of median faciocerebral dysplasia, characterized by a single central orbital fossa with a tubular nose-like appendage above the orbit is known as Cyclopian malformation/monster.[1] It is a lethal complex human malformation, where the affected embryos are either aborted or stillborn upon delivery or die shortly after birth.[2,3] Out of 100,000 births, approximately 1.05 are identified as newborn with cyclopia, including stillbirths.[4]

It is the most severe form of alobar holoprosencephaly, which result from incomplete cleavage of embryonic prosencephalon into bilateral hemispheres appearing between the 18th-28th day of gestation.[2] These developmental defects termed arhinencephaly, range from cleft palate and lip, atresia of the olfactory tracts with atresia of the olfactory tracts, single chambered prosencephalon to absence of median facial structures.[5] Extracranial malformations may include polydactyly, renal dysplasia, and omphalocele.[6,7]

CASE REPORT

A 34-year-old apparently healthy female $G_3P_{1+1}L_1A_1$ with no previous congenital anomalous birth, a homemaker, presented at 34-week period of gestation (POG) referred from primary health centre to our institute CNMCH, Kolkata in December 2021, with complaints of lower abdominal pain of increasing intensity. She had one previous caesarean section 10 years ago (no documents were available with regard to indication or type of incision) with a completely healthy child and an induced abortion 8 years ago. There was no history of any other drugs or alternative medicine intake (except iron-folic acid, calcium, thyroxin), radiation exposure, or a significant

family history or consanguinity. She was from a family with low socioeconomic status from a village near Kolkata, West Bengal. She had seen a gynaecologist (at a private clinic) only 2-3 times during this pregnancy and had not performed anomaly scan despite advice.

Her only 33-week scan (done at a peripheral center) failed to identify any fetal abnormality. Ultrasonography showed a singleton live pregnancy in breech presentation of approximately 32 weeks of POG, estimated fetal weight of 2 kg, with fundo-posterior placement of placenta and nearly absent amniotic fluid.

On examination, she was conscious with blood pressure of 122/74 mmHg, respiratory rate of 16/minute and tachycardiac (Pulse rate-116 bpm), and other vitals stable. On abdominal examination, the uterus was of 32-week size with increased intensity of contractions, tender on palpation. Fetal heart rate 130 bpm. On vaginal examination, cervical OS closed, soft posterior, uneffaced. All maternal laboratory investigations were within normal limits. Decision for emergency caesarean section taken in view of scar tenderness in a previous caesarean section with severe oligohydramnios and breech presentation.

The delivery was uneventful, but the infant was stillborn. The infant was 2.1 kg male with single centrally located ill-formed eye, no nose but a nose-like appendage located above the eye. Optic fissures had eyelid-like segments, as shown in Figure 1 and 2. The infant with crown-heel length of 44 cm had head, chest, and abdominal circumferences measuring 25, 27, and 29.5 cm, respectively. There was no other gross congenital anomaly like cleft lip/palate or mandibular-mental, limb defects etc. A cord sample was taken, but the parents refused to do a karyotype. Autopsy also couldn't be performed due to defiance by guardians.

Figure 1: Cyclopian infant just after delivery before umbilical cord clamping.

Figure 2: Same malformed infant (30 min after delivery).

DISCUSSION

Holoprosencephaly is a group of disorders arising from defective embryonic forebrain development between the third and fourth weeks of pregnancy. There are three forms of holoprosencephaly: alobar, semilobar and lobar type. Cleft lip is the mildest facial abnormality of holoprosencephaly, and alobar holoprosencephaly (cyclopia) is the most severe form, characterized by undifferentiated holosphere of cerebral parenchyma with a central single ventricle, fused thalami and absence of midline structures like corpus callosum and falx cerebri.[8-13]

Cardinal facial features of cyclopia may include a single orbit with a median single eye or partially divided eye (synophthalmia), absent nose, a proboscis above the eye, absent philtrum, otocephaly, astomia or microstomia. Extrafacial characteristics can include polydactyly, omphalocele and renal dysplasia. These obvious features of cyclopia or other forms of holoprosencephaly can be detected by antenatal USG (Ultrasonography) or MRI (Magnetic resonance imaging) if looked carefully.[9-13] In this case, intrauterine facial features were passed undetected.

Mostly the abnormalities of the holoprosencephaly are so severe that the fetuses either die intrauterine or are stillborn, because the brain as well as the other parts of the body that are necessary for the survival do not grow normally.[14]

Since most of the cases are sporadic, incompatible with life, and due to the limited literature knowledge, the exact etiology of this condition remains undetermined. However, various risk factors implicated include genetic factors and chromosomal anomalies (mostly trisomy D), multiple pregnancies (especially twin pregnancy), previous unexplained miscarriages, maternal diabetes, infections during pregnancy (toxoplasmosis, rubella, cytomegalovirus, and herpes simplex virus), exposure to UV light and ionic radiation, smoking, alcohol and certain drugs during pregnancy (such as salicylates, amidopyrine, corticosteroids, aspirin, lithium, anticonvulsants, retinoic acid, anticancer agents, cyclopamine [Veratrum californium, found in corn lily or false hellebore]).[1,3,5,13-17] However, in the presented case, no risk factors could be identified.

The awareness and education about the importance of antenatal checkups should be emphasised in all possible ways. Antenatal USG is the most helpful tool in diagnosing cyclopia, and in most of the reported cases, the anomaly has been recognized early in pregnancy with help of the anomaly scan. This early diagnosis permits timely termination of pregnancy and avoids maternal psychological trauma of giving birth to a malformed baby.[18,19] As there is no cure for this condition yet, legal abortion is the only way to prevent further harm to the newborn and the mother.

CONCLUSION

This case is reported due to cyclopia being an uncommon developmental anomaly characterized by fusion of the optic vesicle with a proboscis-like structure above the orbit. With the advancement in antenatal USG to identify malformed fetuses early in pregnancy, the incidence of malformed fetuses progressing to advanced gestations is decreasing. The awareness of the spectrum of cranial, facial and extra-facial sonographic findings of cyclopia can further improve the accuracy of prenatal diagnosis.

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