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SHORT COMMUNICATION

Cyclopia with spinal bifida, a rare major congenital anomaly: A case report

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Abstract

Cyclopia is, the most extreme form of holoprosencephaly, a rare and lethal complex human malformation resulting from incomplete cleavage of prosencephalon into the right and left hemispheres occurring between the 18th and 28th day of gestation(Dufresne and Jelks 2011). Cyclopia occurs approximately 1.05 in 100,000 births including stillbirths. Cyclopia typically presents with a median single eye or partially divided eye in a single orbit, absent nose, and proboscis above the eye. Extracranial malformations associated with Cyclops are polydactyl, renal dysplasia and omphalocele. The aetiology of Cyclopia is largely unknown. A 26-year-old primgravida with a gestational age of 22weeks plus 4 days delivered a 525gram female abortus with a single median eye, absent nose, 1.3cm by 1.5cm solid mass at the lumbar area at Ambo University Referral Hospital, Ethiopia. On prenatal ultrasound evaluation, severe hydrocephalus with spinal defect (Chiari II malformation) was considered, and the result was revealed to the couple and options for management, including termination of pregnancy, were discussed. The couple accepted the termination of the pregnancy. Prenatal diagnosis of Cyclopia can be achieved by detailed anatomical scan with ultrasound and MRI which, are usually followed by amniocentesis for fetal karyotype determination, which is impossible in our setup because of the lack of accessibility of the service.

Keywords: Cyclopia, Ultrasound, Prenatal diagnosis, Holoprosencephaly, proboscis

Background

The neural tube splits into three main brain vesicles (prosencephalon, mesencephalon, and rhombencephalon) by the fourth week of pregnancy, and the prosencephalon further divides into telencephalon and diencephalon by the fifth week of pregnancy(Dufresne and Jelks, 2011). The telencephalon gives rise to the two cerebral hemispheres and the lateral ventricles, whereas the diencephalon gives rise to the thalami, hypothalamus, and basal ganglia. Holoprosencephaly refers to a group of disorders arising from the failure of normal forebrain development during embryogenic life. There are three forms of Holoprosencephaly which include Alobar, lobar and semi-lobar(Funk and Siegel, 1988). Alobar Holoprosencephaly is the most severe form and is characterized by central monoventricle, fused thalami, and absence of midline structures like corpus callosum and falx cerebri (Dubourg et al., 2007). Cyclopia is an unusual anomaly in which anterior brain and mesoderm structures develop anomalously (Deftereou et al., 2013). The incidence of Cyclopia is approximately 1.05 in 100,000 births, including stillbirths (Rathod et al., 2015). The orbital region is grossly deformed, resulting in the formation of a central cavity 'pseudo orbit,' with the absence of a nasal cavity and the presence of a rudimentary proboscis above the pseudo-orbit (Barber and Muelling, 1950). It is more common in female foetuses and incompatible with life (Koregol et al., 2010). The aetiology of Cyclopia is unknown. The environmental factors that cause Cyclopia are the steroidal alkaloid jervine (found in the corn lily) and its deoxo form, called cyclopamine (Funk and Siegel, 1988).

Case report

26 year of age primigravida, a pharmacist, was referred to our hospital for a second-trimester ultrasound from a Health Centre (a small health institution that provides basic treatment to 25,000 people).

On history, she worked at one of the governmental pharmacies for the last three years as a pharmacist. She denied ingestion of drugs during pregnancy and for the 3 preceding months. The client declared no chronic medical conditions. At three months, she began antenatal care (ANC) follow-up at a Health Center, where she received two doses of tetanus toxoid vaccine, iron supplements, and was advised to undergo an ultrasound at five months.

Since there was no ultrasound machine at Health Center, where she started ANC follow-up, she referred to Ambo University Referral Hospital (AURH) for anatomical obstetric ultrasound scanning at 5 months of pregnancy, and the finding of obstetric ultrasound was a singleton with a gestational age of 22 weeks and 4 days, significantly dilated ventricles (17.3mm), mild polyhydramnios (AFI=10.8cm), and a 1.25x1.4 cm solid mass at the lumbar area. Accordingly, a diagnosis of severe hydrocephalus with spinal bifida and mild polyhydramnios was made.

The couple was counselled concerning the condition of the foetus and options for management. They agreed on the termination of the pregnancy, and then the client was admitted to the maternity ward. After 36 hours of 200 mg of mifepristone administration, misoprostol 400g was inserted vaginally every six hours, and on the fourth dose (cycle), she delivered a female abortus weighing 525 grams, with a single eye in the middle of the forehead, 1.5x1.3 cm mass in the lower lumbar, and missing nose (Fig 1 and 2).

Fig1

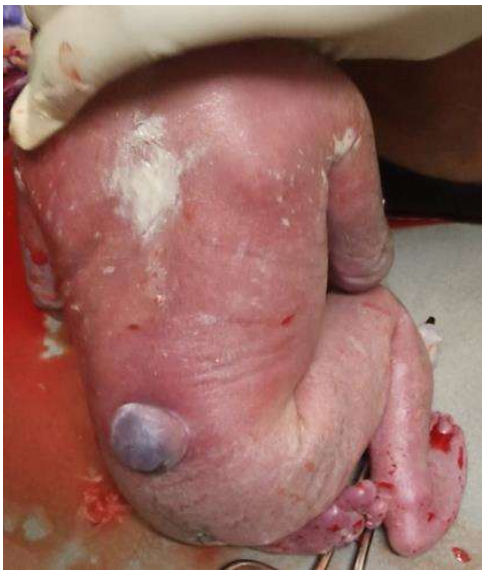

Fig2

Figure1 & 2. Anterior and posterior aspect of Cyclops abortus at Ambo University referral Hospital (AURH) respectively, Ambo, Ethiopia, 2022

Discussion

Holoprosencephaly refers to a group of disorders arising from the failure of normal forebrain development during embryogenic life (Münke 1989). There are three forms of Holoprosencephaly which include Alobar, lobar and semi-lobar (Nalam et al., 2018). Alobar Holoprosencephaly is the most severe form and is characterized by central mono-ventricle, fused thalami, and absence of midline structures like corpus callosum and

falx cerebri (Prayer et al., 2011). Cyclopia which is an anomaly of organogenesis of the eye results from the arrest of the development of the anterior end of the neural tube. Thus cyclopia is always associated with abnormality of the brain mainly with Alobar Holoprosencephaly (Filly et al., 1984).
One of the most significant tools in the prenatal diagnosis of cyclopia is obstetric ultrasonography. The bilateral choroid plexus (butterfly sign) is missing on first-trimester ultrasound, whereas fused thalami, lack of midline echo, mono-ventricle, and aberrant facial characteristics are the primary ultrasound findings of Alobar holoprosencephaly (Barkovich and Norman, 1989).

Severe hydrocephalus and spinal bifida were discovered in our instance during the second trimester of pregnancy, and the diagnosis of cyclopia was missed by prenatal ultrasound. Holoprosencephaly with cyclopia is a severe prenatal brain defect that cannot be corrected or cured. Chromosomal abnormalities affect 30-50 percent of foetuses diagnosed with holoprosencephaly, with Trisomy 13 accounting for 75 percent of chromosomal abnormalities (Funk and Siegel, 1988). Pregnancy management options include elective terminations if the diagnosis is made before the 24 weeks of pregnancy.

MRI imaging modality is very important for detecting the correct abnormality of the brain detected on prenatal ultrasound especially if the case of cyclopia suspected on prenatal ultrasound (Rathod et al., 2015) but in our case, severe hydrocephalus with a spinal defect was considered on prenatal ultrasound instead of holoprosencephaly and cyclopia and the diagnosis of the latter was missed. After the expulsion, post-mortem examination, pathology, and fetal chromosomal detection are also very important but they are not available in our setup.

Conclusion and Recommendations

Five hundred twenty-five-gram female abortus with a single median eye, absent nose, 1.3cm by 1.5cm solid mass at lumbar area expelled at our AURH and at post expulsion diagnosis of cyclopia with spinal bifida was made which missed by prenatal ultrasound and instead diagnosis of severe hydrocephalus with spinal bifida was made. The prenatal diagnosis of cyclopia can be made early with sonographics and the awareness of the spectrum of sonographic findings can improve the accuracy of diagnosis Cyclopia. In suspected cases, MRI can be determined depending on its accessibility. Prenatal fetal karyotype determination and post-mortem examination are also very important, in order to counsel the patient on the cause and possible recurrence of the case.

Consent

The patient and her spouse gave their written informed agreement to the publishing of this case and any associated image of the case without mentioning the patient's name or photograph. For publishing, an ethical review was also acquired from Ambo University Referral Hospital and College of Health Science.

Consent for publication

The patient and her spouse gave their written informed consent to publish this case and any associated image of the case without mentioning the client's name or photograph of the client or card number. Moreover, consent for publication was acquired from the ethical review board of Ambo University (Ref.No:PGC/153/2022, Date: 03/07/2022), College of Health Science.

Availability of data and materials

All data generated or analysed in this study are included in this case report.

Competing interests

The author declares that there are no competing interests.

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