ECRONICON OPEN ACCESS

EC GYNAECOLOGY
Case Report

Case Report of Cyclop Deformity

Shaista Zubair[1]*, Marie Lou Acevedo Villar[2], Zubair Anwar[3], Azimuddin Azim Siraj[4], Ved Vrat[3], M Zakir Hossain[4], Faizal Sharif[5] and Indulekha Anayath[6]

[1]Senior Registrar in Obstetrics and Gynaecology Good Hope Hospitals, University Hospitals of Birmingham NHS Foundation Trust, UK

[2]DPPS, Diplomate of Philippine Pediatric Society Medical Officer , Paediatric Suri Seri Begawan Hospital, Kuala Belait, Brunei Darussalam

[3]Dr ENT Department, Suri Seri Begawan Hospital, Kuala Belait, Brunei Darussalam

[4]Ophthalmology Department, Suri Seri Begawan Hospital, Kuala Belait, Brunei Darussalam

[5]Radiology Department, Suri Seri Begawan Hospital, Kuala Belait, Brunei Darussalam

[6]Obstetrics and Gynaecology, Suri Seri Begawan Hospital, Kuala Belait, Brunei Darussalam

***Corresponding Author:** Shaista Zubair MRCOG.UK, MRCPI. Ireland, FCPS, MCPS, Senior Registrar in Obstetrics and Gynaecology Good Hope Hospitals, University Hospitals of Birmingham NHS Foundation Trust, UK.

Received: March 15, 2021; **Published:** March 31, 2021

Abstract

Cyclopes are rare congenital abnormalities; a severe form of holoprosencephaly resulting in children being born with just one eye. It results from failure of the cerebral hemisphere to separate during fetal development. The incidence is 1 in 13000 live births but present in 1 in 2500 pregnancies that end up in miscarriage. It is incompatible with life. In this report we present a preterm vaginal delivery of Cyclop baby presented with Polyhydramnios and Spontaneous rupture of membranes.

Keywords: *Cyclopes; Holoprosencephaly; Polyhydramnios*

Introduction

The term cyclop is commonly used to describe either the abnormality of true cyclopia in which a single median eye is the only ocular structure present or synophthalmia in which two globes (right and left eyes) are partially fused in the median position. Cyclopia (also cyclocephaly or synophthalmia) is a rare and severe form of holoprosencephaly and is a congenital disorder (birth defect) characterized by the failure of the embryonic prosencephalon to properly divide the orbits of the eyes in to two cavities. Its incidence is 1 in 16000 in born animals and 1 in 250 in embryo [1,2].

Typically, cyclopia present with the nose either missing or replaced with a non-functioning nose in the form of a proboscis, Such a proboscis generally appears above the central eye and is characteristic of a form of cyclopia called rhinencephaly or rhinocephaly [3].

Suggested risk factors include maternal diabetes, infections during pregnancy (syphilis, cytomegalovirus, toxoplasmosis, herpes and rubella), drugs taken during pregnancy (alcohol, aspirin, lithium, anticonvulsants, hormones, retinoic acid, anticancer agents and fertility drugs) and physical agents like ultra violet light, previous pregnancy loss and first trimester bleeding [4].

Genetic problems or toxins can cause problems in the embryonic forebrain-dividing process [5]. One highly teratogenic alkaloid toxin that can cause cyclopia is cyclopamine or de-oxyjervine, found in the plant veratrum californicum (also known as corn lily or vetch weed).

The mistake of ingesting veratrum californicum during pregnancy is often due to the fact that hellebore, a plant with which it is easily confused, is recommended as a natural treatment for vomiting, cramps and poor circulation, there condition which are quite common in pregnant women [6].

Familial occurrences in twins and in consanguineous marriages have been documented and would be consistent with a single gene abnormality [7]. In normal differentiation of the cephalic midline structures there is increase expression of pax-2 gene and inhibition of pax-6 gene from the notochord. However, inappropriate expression of these genes may result in cyclopia [8]. Mutation of sonic hedgehog gene (SSH) has also been implicated in the formation of cyclopia. Other genes that play a role in the formation of these defects include ZIC2, TGIF and SIX3 [9]. In recent years several isolated case reports of Cyclops or holoprosencephaly in human and animals have been associated with abnormal chromosomes [10].

Case Report

A live infant (1.8 kg) delivered at 05:14 hours on 15 July 2013. Baby born in cephalic presentation in labor room of the Suri Seri Begawan Hospital Kuala Belait, Brunei Darussalam. Apgar score noted 7 and 9 at 1 and 5 minutes respectively. Noted Cyclops with multiple congenital abnormalities were apparent at birth (Figure 1). On subsequent examination findings were microcephaly, midline facial deformity single centrally located eye, two cornea seen and both the globes are fused in one. Micro cornea, non-reacting pupil's, eye lids cannot closed, proboscis (rudimentary nasal appendage).

Figure 1: *(Permission taken from both parents for images).*

The 34 years old Chinese Bruneian mother, married, primigravida was booked case at 11 weeks of gestation. She was on regular antenatal visit, non-consanguinity in marriage, antenatal screening Blood group B RH positive Group B streptococcus positive on high vaginal swab, while. HIV, VDRL, TPHA, Hepatitis B, OGTT all were normal. At 27+ weeks of antenatal scan shows polyhydramnios and multiple congenital anomalies with suspected cyclopia, later which was confirmed on subsequent ultrasounds and pediatrics counseling done about the outcome of baby but patient wanted to continue the pregnancy till term and normal delivery. She was admitted in antenatal

ward on 14-july-2016 at 35+1, IUGR, moderate polyhydramnios, preterm premature rupture of membranes and labor pain for 6 hours spontaneously progress to Normal vaginal Delivery at 05:14 hours on 15 July 2013. Counselling of parents done delivery and permission taken from both parents for images and publication without their identification.

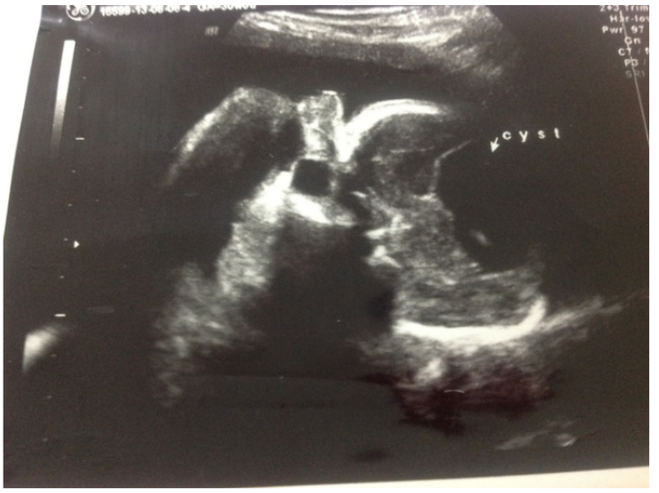

Figure 2: *Ultrasound findings of proboscis.*

This 1.8 kg female infant was with microcephaly, anterior fontanel open, head circumference 24 cm central eye, proboscis, mouth no cleft lip/palate. Normal ears, lungs clear, no murmur on auscultation, abdomen soft, umbilical cord one vein, two arteries, Hip and spines normal, anal opening patent, grossly normal extremities, Ballard: 36 weeks. Ultrasound brain done skull bones were fused and small fontanels. Crescent shaped mono ventricle with thin 1 cm cerebral mantle occupies anteriorly. No midline structure present. Large single mass midline thalamus and normal looking midbrain and cerebellum, both globes seen to be fused together with central septum visualized, two lenses visualized, no retinal detachment or vitreous bands observed, optic nerve could not be imaged. Normal liver, spleen and kidneys and no mass lesion found in abdomen. This baby was alive for 5 days in NICU and pronounced dead on 21-07-2013.

Chromosomal analysis/genetic testing done and its confirmed trisomy 13.

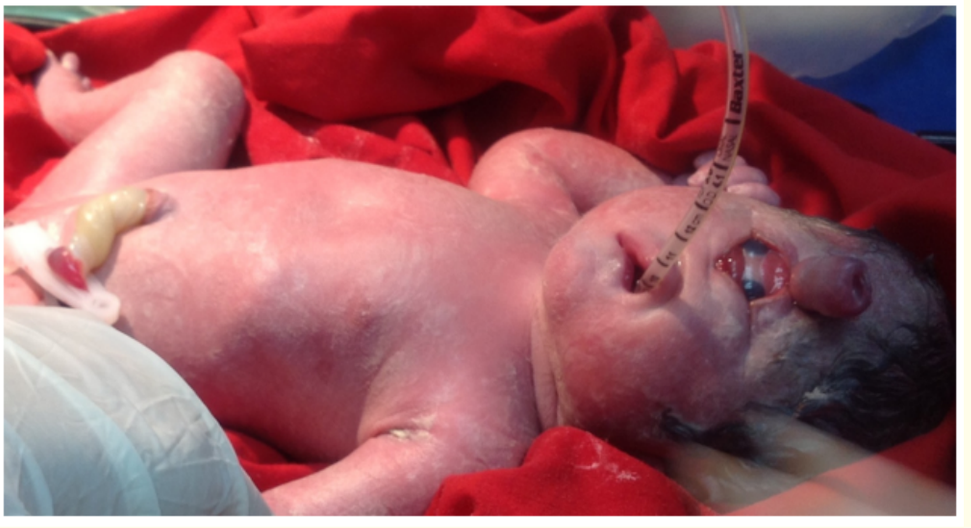

Figure 3

Discussion

Cyclop is usually associated with trisomy 13 or 18 and may be associated with other chromosomes abnormalities or occasionally with normal chromosomes. It arises early in gestational life causing lack of cleavage of the hemisphere into right and left cerebral hemisphere [11]. As noted in ultrasound report of brain that large single mass midline thalamus reported but normal midbrain and cerebellum. Its incidence is usually sporadic [12]. As at the time of this report, only 1 case delivered in Brunei Darussalam. However, this is the first case ever of Cyclops in Suri Seri Begawan Hospital reported and grossly different from the other cases.

The severity of facial abnormality reflects the severity of the brain abnormalities to a certain extent [13]. This is exemplified by the further investigation in this case. The total systemic deformity associated with cyclopean abnormality are generally inconsistent with fetal viability and only a few cyclopic individuals survive pregnancy to be still born or die shortly after birth [4]. One Cyclops were reported to live for ten years [14]. This female infant died 5 days after delivery.

Multitude of environmental agents (physical, chemical and biological) have produced experimental Cyclops because they alter chromosomes numerically and structurally [15]. Cyclopia has been reported in children's with diabetic mothers which support the multifactor theory of cyclopia development because diabetes is multifactor disease but this seems unlikely [16]. This 34 years old mother was not diabetic and gave no history associated with the risk factors mentioned above.

Conclusion

At present all the facts concerning cyclopia are consistent with some chromosomal abnormality, even if chromosomal observation represents only one group of several etiologies, cyclopia is a very rare anomaly which one may never have the opportunity in a life time to witness. This is the first case of cyclop to be reported in Suri Seri Begawan hospital Brunei Darussalam.

Acknowledgement

We remain grateful to the doctors of the Obstetrics and Gynecology, Pediatrics, Otolaryngology, Radiology, Ophthalmology of Suri Seri Begawan hospital for making this report possible by contributing in cyclopic baby examination and their opinion.

Bibliography

1. Gupta RC., *et al.* "Human cyclopia with associated microstoma and anencephaly". *Indian Journal of Ophthalmology* 29 (1981): 121-123.
2. Dubourg C., *et al.* "Holoprosencephaly Orphanet". *Orphanet Journal of Rare Diseases* 2 (2007): 8.
3. Dark Graham. "Rhinocephaly". In Online Medical Dictionary (2007).
4. 2007 case report.
5. Tapadia MD., *et al.* "Its all in your head: new insight into craniofacial development and deformation". *Journal of Anatomy* 207.5 (2005): 461-477.
6. Veratrum californicum. Available @ Teratology Society".
7. Ming JE and Muenke M. "Diseases and Holoprocencephaly". *American Journal of Human Genetics* 71.5 (2002): 1017-1032.

8. Kim JW and Lemke G. "Hedgehog-regulated localization of Vax 2 controls eye development". *Genes and Development* 20.20 (2006): 2833-2847.

9. Cordero D., *et al.* "Temporal perturbations in sonic hedgehog signaling elicit the spectrum of holoprosencephaly phenotypes". *Journal of Clinical Investigation* 114.4 (2004): 485-494.

10. RO Howard. "Chromosomal abnormalities associated with cyclopia and synophthalmia". *Transactions of the American Ophthalmological Society* 75 (1977): 505-538.

11. Chan A., *et al.* "Histogenesis of retinal dysplasia in trisomy diagnosis". *Pathology* 2 (2007): 48.

12. Cotran RS., *et al.* "Robbins pathologic basis of Diseases 5th edition". Philadephia, W.B.saunder (1994): 1302.

13. Poirer J., *et al.* "Manual of basic micropathology". Philadelphia: Saunders (1990):199.

14. Vare AM. "Cyclopic". *American Journal of Ophthalmology* 75 (1973): 880.

15. Pei W., *et al.* "Environmental and Genetic Modifiers of squint Penetrance during Zebrafish Embryogenesis". *Developmental Biology* 308.2 (2007): 368-378.

16. Dekabeu A and Maee K. "Occurrence of Neurological abnormalities in infants of diabetic mothers". *Neurology* 8 (1956): 193.

Volume 10 Issue 4 April 2021