



Vision Insight

Authors

Dr C. Renuka Devi¹, Dr Nalini.A², Dr N.S. Raghupathy³

¹3rd Year Post Graduate, ²D.M., Neonatology, ³HOD

Department of Paediatrics, Aarupadai Veedu Medical College, Pondicherry, India

Abstract

Anophthalmia means congenital absence of globe usually bilateral sometimes unilateral having incidence of 1:10000 worldwide. We report a neonate with right anophthalmia, periventricular leukomalacia and ventricular septal defect. Parents were counselled regarding vision and ocular prosthesis. Antenatal ultrasound can diagnose anophthalmia and be cost effective.

Keywords: Unilateral Anophthalmia, Congenital anomaly.

Introduction

Anophthalmia means congenital absence of globe. Microphthalmia means globe which is less than two standard deviation of mean for age. It is usually bilateral sometimes unilateral having incidence of anophthalmia/microphthalmia as 1:10000 worldwide^(1,2). Disease incidence occurs equally in both sexes. It can be an isolated finding or part of microphthalmia syndromes (MCOPS types 1,2&3), Lenz microphthalmia syndrome, microphthalmia with linear skin defects and anophthalmia-oesophageal-genital syndrome. Cause may include genetic mutations (SOX2, PAX6, FOXE3, OTX2, CHX10 and RAX)⁽³⁾, abnormal chromosomes, advanced maternal age, multiple births, low birth weight, TORCH (Toxoplasmosis, Rubella, Cytomegalovirus, Herpes simplex, Listeria, Parvovirus and HIV) infections, drugs (Alcohol, Hydantoin, Thalidomide and Vitamin A deficiency) and vascular disruption with the insults happening from third to eighth week of embryo. Diagnosis is

usually clinical, confirmed by Magnetic resonance imaging (MRI) and B SCAN. Antenatal ultrasound provides better and earlier diagnosis⁽⁴⁾. It is usually associated with central nervous system and cardiac anomalies. Early intervention is done with ocular prosthesis for cosmetic purpose and development of orbit than vision⁽⁵⁻⁷⁾. Treatment of comorbid conditions need to be addressed for optimal growth and development. In India, only one study done showed 1 out of 23,100 cases are reported so far⁽⁸⁾.

Case Report

We report a neonate with right anophthalmia. A 31-year-old primipara married non-consanguineously, had primary infertility for 5 years received infertility treatment but conceived spontaneously with no antenatal complications except multiple fibroids in the walls of uterus. There was no teratogenic drug intake. Aneuploidy screening result were in low risk. Targeted imaging for foetal anomalies (TIFFA) scan was

normal. There was no family history of foetal anomalies. She had spontaneous onset of preterm labour at 36 weeks of gestation and delivered a girl baby with a birth weight of 2.75Kg by Emergency Lower Segment caesarean section for cephalopelvic disproportion. Apgar scores were 8&9 at 1 & 5 minutes. She had transient tachypnoea of newborn which settled with oxygen by hood within hours. First physical examination showed right blepharophimosis with absence of eyelashes on lateral aspect of right eye. It was confirmed by B scan which showed absence of Right ocular globe and optic nerve. Neurosonogram done on 7th day of life revealed bilateral grade 2 periventricular leukomalacia and abdominal ultrasound showed minimal left adrenal gland enlargement. ECHO showed small subpulmonic Ventricular septal defect of 2mm. There was no hemodynamic instability. Baby had feeding difficulty with excessive weight loss which settled with correct positioning and attachment. Parents were counselled the need for right ocular prosthesis. This was the first case of anophthalmia in their family. Genetic studies for mutational analysis not done.



Discussion

Our patient presented with right anophthalmia with fusion of palpebral fissures on the lateral aspect, VSD, periventricular leukomalacia.

Anophthalmia will usually be not associated with adnexal anomalies because of the different embryonic origin of these structures⁽⁹⁾. Advanced maternal age has significant association with the disease. Our patient condition was diagnosed only after birth, TIFFA scan failed to pick up the anomaly but two dimensional and three-dimensional reverse face imaging of foetal ocular globe in axial and coronal planes in addition to detection of hyaloid arteries and lenses had diagnosed anophthalmia/microphthalmia spectrum in studies by Araujo and A. Searle^(1,10). This had led to early termination of foetus in those mothers. Periventricular leukomalacia of grade 2 had been diagnosed in our neonate. Usually central nervous system anomalies are associated with anophthalmia. Early intervention had been started. The long term neurodevelopmental outcomes need to be followed up.

Conclusion

Though Congenital anophthalmia is rare, it can be identified antenatally especially by anomaly scan. Hence detailed and meticulous assessment of antenatal scan can relieve the stress of parents and physician may be able to prepare the parents for further management. Thus, it can be a cost-effective intervention.

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