Case Report

Clinical anophthalmia and microphthalmia: a clinical case in a baby infected with TORCH

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Abstract

Anophthalmia is a very rare condition. Clinical anophthalmia is a term to describe cases of severe microphthlmia when the eyeball is severely hypoplastic or absent. Anophthalmos is complete absence of ocular tissue and is a histopathological diagnosis. The preciseaetiology of anophthalmos unknownbut is considered to be multifactorial. They include genetic mutations and environmental factors like viruses including the TORCH group of viruses. It may also occur as a part of a syndrome.This case report discusses a 2 days old child with clinical anophthalmia. It delves into the possible causes of this condition.

Keywords: anophthalmia, microphthalmos, TORCH, consanguinity.

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INTRODUCTION

True/Primary anophthalmos is very rare. The diagnosis of true anophthalmos can be established when there is a complete absence of ocular tissue within the orbit and is classically a histological diagnosis. Clinical anophthalmia is used to describe cases of severe microphthalmia when the ocular tissue is severely hypo plastic or absent.[1] Extreme microphthalmos is more common. In this condition, a remnant of the globe is usually present which may be missed on initial examination. Anophthalmia occurs when the neuroectoderm of the primary optic vesicle fails to develop properly from the anterior neural plate of the neural tubing. Genetic based cause for anophthalmia is due to mutation in the SOX2 gene. Without the SOX2 gene protein, the activity of genes important for the development of the eye is disrupted. Other recognised associations include Trisomy 13, Lenz syndrome, Goldenhar-Gorlin syndrome, Aicardi's syndrome and Warburg syndrome.[1] Environmental conditions have been associated with anophthalmos and microphthalmos, Viruses that can because anophthalmos include rubella, toxoplasma and influenza.

CASE REPORT

A 2 days old female baby was referred to the Department of Ophthalmology. The baby was born at 38 weeks of gestation with birth weight being 2.93 kgs by a lower segment Caesarean section. The indication for LSCS was a previous LSCS with frank breech presentation. Postoperatively, there were no complications. The mother's age is 22 years. Her marriage was of a 3rd degree consanguinity. She gives history of a previous pregnancy which was stillborn. The baby's general condition was stable with no signs of pallor, cyanosis, icterus and oedema. The general appearance of the head, back and spine and the extremities was normal. On initial examination, the orbital rim appeared small. The baby had no levator function. There was difficulty in retracting the eyelids to expose the globe. There was palpebral closure with absence of ocular globe of both eyes. On eyelid retraction with a Desmarre's retractor, no residual eyeball was seen with noconjunctival tissue. On diagnosing anophthalmia clinically, investigations were performed to confirm the diagnosis and to establish the possible cause. The baby tested positive for TORCH with high titres of rubella, cytomegalovirus and toxoplasma. The MRI scans showed rudimentary eye balls with

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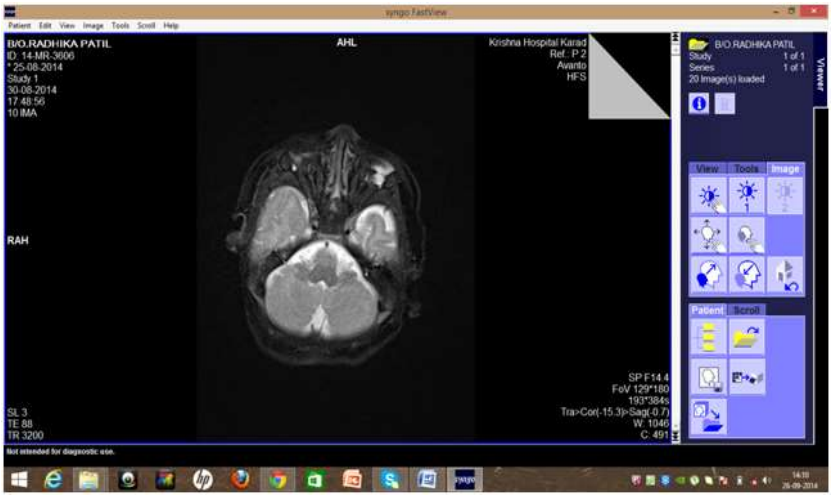
undeveloped optic nerve confirming the diagnosis of clinical anophthalmos.



DISCUSSION/CONCLUSION

Microphthalmia (abnormally small globe) and anophthalmia (complete absence of the globe) are rare congenital conditions.The birth prevalence of anophthalmia and microphthalmia has been generally estimated to be 3 and 14 per 100,000 population respectively.[2] The precise pathogenesis of these conditions remains unknown. Mann[3] suggested anophthalmia has its genesis early in gestation as a result of failure of development of the anterior neural tube (secondary anophthalmia) or optic pit (s) to enlarge and form optic vesicle (s) (primary anophthalmia) Epidemiological studies have suggested both genetic as well as environmental factors in causing microphthalmos and anophthalmos. Mutations in the SOX2 gene has been found to be a major cause for microphthalmia and anophthalmia[4] The strongest evidence for environmental causes is congenital infections like rubella, toxoplasmosis, cytomegalovirus, varicella, parvovirus and Coxsackie A9. Other environmental causes include maternal vitA deficiency.[5] In our case, the mother and the baby tested positive for TORCH infection which was requested for on diagnosing anophthalmos clinically. The mother has a history of a previous pregnancy which was stilborn. Such a past history should have demanded a detailed checkup in the antenatal period. However, the mother was not enrolled in any institution for antenatal care. This case report highlights the importance of antenatal care and monitoring, especially in mother's with a significant past history. It also emphasizes the strong evidence for congenital infections being a cause for microphthalmos and anophthalmos. The baby was born of a third degree consanguinous marriage. The influence of genetic factors hence cannot be overlooked. It is also possible to diagnose such a rare pathology in prenatal period due to advances in technology of ultrasound machines and the use of 3D images especially in high risk mothers'such as these.[6] Management of anophthalmia is mostly supportive. However, surgical intervention options like progressive conformers, balloon expanders, progressive orbital implants and hydrogel tissue expander implants are available.[7,8,9]

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