ANOPHTHALMOS — A Case Report from Mercy Eye Center, Abak, Akwa-Ibom State, Nigeria

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SUMMARY

Aim: To report a case of anophthalmia in an otherwise normal neonate in a rural eye hospital in Akwa Ibom State, Nigeria.

Case report: A one-day-old full-term female neonate, delivered by spontaneous vaginal delivery to a minor who had no proper antenatal care is reported. The mother had a history of attempts to abort the pregnancy by ingestion of traditional remedies. The only congenital anomalies present were primarily anophthalmic eyes. A brief review of the literature on its epidemiology, types and associated risk factors, associations and current concepts in management strategies is presented.

Conclusion: Anopththalmia is an uncommon, though interesting, condition whose management is extremely difficult. The motivation on the part of the relations has to be extremely high to make the child cosmetically acceptable.

Key words: anophthalmos, orbit, tissue expander, Nigeria

INTRODUCTION

Anophthalmia is a very rare condition[1] in which one or both eyes did not form during the early stages of pregnancy. It can be classified into 3 groups: primary anophthalmia which is the complete absence of eye tissue due to the failure of the part of the brain that forms the eye; secondary anophthalmia in which development of the eye is arrested, leaving very tiny residual eye tissue which can only be seen on close examination; and degenerative anophthalmia which occurs due to ischemia.

In some centres, it is so rare that cases are seen only once in twenty years.[2] Most cases seen are unilateral in nature.[2, 3] Cases of bilateral congenital anophthalmos were, however, reported about 30 years ago.[4] This is probably the first bilateral case reported in recent times. It has therefore generated much interest. It is for this reason that this rare case, seen in a rural, high volume referral centre is presented.

CASE REPORT

M.J.E., a one-day-old female child was brought on a routine clinic day to the Mercy Eye Center, Abak by her grandmother. She was delivered at home a few hours previously by traditional birth attendants. She was brought to the centre because it was noticed that she did not appear to have any visible eyes where they were supposed to be. The child was however breastfeeding normally.

The mother is a 16-year-old secondary school student who became pregnant while in school. An attempt to abort the pregnancy in the first trimester through the ingestion of all sorts of chemical and traditional remedies was unsuccessful. Her mother then advised her to keep the pregnancy and have the baby instead. She had an uneventful but unsupervised gestation period with no visits to a recognized hospital. Labour was also reported to be normal. Having just delivered, she remained at home while the child was brought to the centre.

On examination, the child appeared normal sized, with a birth weight of 2.5 kg, well proportioned limbs and normal head circumference (33 cm), and no visible physical or organic defects, except that the eyelids seemed to be shut as if the child was sleeping. All other systems were examined by a paediatrician and no other anomaly was observed.

On closer examination, following lid retraction with speculums, there were no clearly visible ocular tissues within. A diagnosis of presumed primary anophthalmia was made. The relations were now sent to a counselor for advice and reassurance. They were subsequently lost to follow-up.

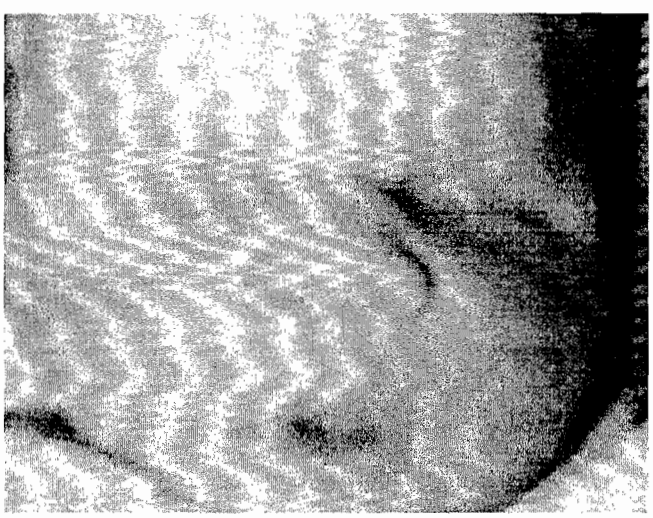
DISCUSSION

Anophthalmia is the congenital absence of one or both eyes. There can also be complete absence of eyeballs, optic nerves, optic chiasm, tracts and radiation.[5] True anophthalmia, where there is complete absence of ocular tissue within the orbit is very rare. Severe microphthalmos is more commonly seen and occurs in 1 in every 100,000 live births. Prevalence per 10,000 live births and stillbirths is 0.18.[1] Two-thirds are genetic and one-third is thought to be due to environmental factors,

i.e., pesticides, drugs, radiation, toxins, etc. A report from Nigeria also suggests maternal ingestion of drugs at 3 months gestation as a causative factor,[3] though some earlier reports did not find any such history.[4] In this report, there was a history of maternal ingestion of herbs to induce an abortion.

A study of 406 babies born between 1988 and 1994 in England showed that anophthalmos is common in congenital infection syndromes, e.g., measles.[6] Other viruses implicated are parvovirus B19 and influenza. Also in a study of 8 families in England, severe ocular malformations were seen in relation to heterozygous mutation of OTX2.[7] In a study carried out in Sweden, it was reported that there is no racial or sexual predilection with anophthalmia.[8] In the same study, it was also found to be associated with sub-fertility and maternal smoking in early pregnancy. In another study, thirteen cases were reported to be associated with oesophageal atresia.[9] The case reported in this paper had no difficulties with feeding as breastfeeding had been established before presentation. A case of an association with Goldenhar's syndrome has also been reported in a 3-day-old Nigerian neonate, with right anophthalmos and a lipodermoid in the same socket.[3] The risk of anophthalmia is also higher in children born by women aged 40 or more.[1]

The mother of this patient was just 16 years old. This condition may occur secondary to arrest of development of the neuroectoderm of the optic vesicle from the neural tube during embryological development. Early detection and treatment will help decrease the facial asymmetry and disfiguration which may result in these children, as growth and development of the bony orbit is directly dependent on the outgrowth of the globe. This asymmetry was already apparent in this patient from birth (see pictures a and b). Orbital volume is reduced by 35-58%, compared to the normal side.[10] A small bony orbit causes hemifacial hypoplasia which can result in significant cosmetic deformity.



A

B

Histopathology may reveal an extremely small or malformed globe with rudimentary ocular contents. Extraocular muscles are often absent or markedly decreased. This could not be carried out on this patient as she was immediately lost to follow-up.

In the management, the main aim is to encourage socket growth.[11] Medically, a solid conformer which can be progressively increased in size can be placed in the orbit to stimulate bony growth and to enlarge the orbital cavity in an attempt to attain normal proportions. A self-expanding, hydrogel tissue expander can also be employed.[12] Later, an ocular prosthesis may be fitted over the conformer to improve the appearance. If conformers cannot be tolerated, inflatable silicone expanders can be placed surgically deep in the orbit. This can be gradually filled with saline on a weekly or biweekly basis. It works best if placed early, in the first year of life. Eyelid surgery, such as lateral canthotomy/ cantholysis, can be performed to allow a large conformer. Also, expansion of the orbit can be done surgically in three directions – laterally, inferiorly and superiorly – particularly in cases of late referral or insufficient orbital volume. However, in this case, apart from the cost, the motivation for cosmetic treatment might have been better if the patient had at least one seeing eye.

Complications include significant cosmetic deformities, which are often seen if the anophthalmic orbit is not treated early, particularly in poorly motivated relations as in this report. Even after proper treatment, results are often cosmetically disappointing. Fitted prostheses are completely immobile, and the eyelids show significant malformation and are shortened and immobile. In this case, it is thought that the condition originated as a result of the ingestion of harmful traditional abortion remedies within the first three months of pregnancy. This indicates that the patient had a primary form of anophthalmia. A trans-vaginal scan at 14-16 weeks of gestation has been to pick

up a significant fraction of foetal eye anomalies,[13] and this is highly recommended to families with a history of congenital eye anomaly.

CONCLUSION

Anophthalmia is an uncommon condition whose management is extremely difficult and, in the particular terrain where this patient was found, almost impossible. Motivation has to be extremely high among the relations to make the child cosmetically acceptable – the child must be considered precious and not an unwanted baby, as in this case. It is recommended that proper and adequate counseling be offered to the parents of such children.

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