BILATERAL CONGENITAL ANOPHTHALMOS

Report of a Case

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Anophthalmos is a rare congenital anomaly. It means complete absence of one or both eyes. It is of interest to embryologists, ophthalmologists and pediatricians. With better understanding of the role of environmental factors in causation of congenital abnormalities, the interest of the pediatrician is growing. The condition was first noted in 1557, but for the first time it was reliably reported by Bartholin in 1657 (Sorsby 1934). A few cases have been reported since (Mann 1957, Warfel 1961, Michaels and Zugsmith 1963, Maria and Shukla 1965, Mehra 1965). Because of the rarity of this anomaly and scarcity of reports from the country, we report here a case of bilateral congenital anophthalmos.

Report of a Case

S., a 5-year-old male child was admitted to the pediatric ward of Medical College Hospital, Aurangabad, with the complaint of diarrhoea of one month's duration. On examination, the interesting feature observed was absence of both eyes. On questioning, the mother confirmed that the eyes were absent since birth.

The patient was the youngest sibling in a family of five, born after a normal delivery. The prenatal history was noncontributory. There was no other member in the family with a similar abnormality.

General health of the patient was poor due to undernourishment caused by chronic diarrhoea. Development was normal in other respects. His mentality was slightly above average. The bilateral anophthalmos was the only abnormality found (Fig. 1).

Both orbits were small, being larger in the transverse diameter than the vertical. They measured 30 mm. wide and 20 mm. in height, giving an orbital index of 66, normal value for coloured people being 84. Both eyelids and eye lashes were present. The palpebral fissures measured 14 mm. on either side. The fornices were shallow. Puncta were present. Lacrimal glands were present and functioning. The eyeballs could not be felt on either side. The conjunctival sac was reduced in size. Movements of the conjunctival sac were noticed, indicating the presence of extraocular muscles.

Discussion

Before discussing the mechanism, a brief recapitulation of the development
of the eye will not be out of place. The eye develops from three sources - the retina from the neural ectoderm, the lens from the surface ectoderm and the adnexa from mesoderm. Before the closure of the anterior neuropore, a stalk-like evagination approaches the ectoderm from the primary brain vesicle. This is the optic bulb. The surface ectoderm adjoining this bulb gets thickened and ultimately separates to form the lens. The optic bulb develops into a cup. This is the optic pit from which the retina develops. The mesoderm surrounding the optic bulb forms cornea, sclera, nonectodermal uvea and muscles.

Mann (1957) has classified anophthalmos into three types, depending upon the cause responsible for the abnormality.

1. **Primary anophthalmos**: In this there is failure of formation of the optic pits. Since the optic pits are recognised in the two mm. embryo, i.e., in the third week of intrauterine life, the cause operating must produce its influence before this stage. The defect is restricted to the optic pits only, since the rest of the brain tends to develop normally. Usually the condition is bilateral and cases occur sporadically. The orbit and other mesodermal structures develop normally though they may be malformed.

2. **Secondary anophthalmos**: This is secondary to non-development of the fore-brain. There are usually many congenital anomalies which are associated with this condition. As the catastrophe is associated with gross anomalies, it results in non-viable monsters and hence is not seen in clinical practice.

3. **Degenerative anophthalmos**: In this condition optic vesicles appear but they undergo destruction or degeneration. The evidence of this is the presence of a small fibrous nodule or rudimentary lens structure. There is usually no gross abnormality of the brain.

**Pathogenesis**

The pathogenetic mechanism of congenital anomalies of the eyes is of special importance for two reasons. Firstly, it leads to many blind and thus handicapped children and secondly, the role of preventable environmental factors in its causation has to be studied. Van Dyse postulated a theory according to which the pressure exerted by thickened amnion on the surface of the head leads to the suppression of the growth of optic vesicles. But there is no convincing evidence to support such a hypothesis. Association of anophthalmos with other congenital anomalies in which the germinal influence is established suggests that such a factor may also be influencing the occurrence of anophthalmos. Genetic and chromosomal abnormalities may be responsible in some cases.

Further, sporadic and isolated occurrence of anophthalmos suggests that the cause may be environmental. This is supported by experimental work wherein suppression of the optic vesicles in animals has been produced by teratogens like X-ray, hypothermia, antimetabolites, vitamin A deficiency, etc.

It may be said that there is no one cause, but multiple factors are responsible in the etiology of anophthalmos.

**Summary**

A case with bilateral congenital anophthalmos is reported. The subject is reviewed in brief.