Complicated Anophthalmos

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Holoprosencephaly is an early developmental defect of the brain in which there is a failure to form paired cerebral hemispheres. The cerebrum is made up of an unpaired sphere and the lateral ventricles are represented by a single midline cavity. Usually there is an associated arhinencephaly—absence of olfactory bulbs and tracts, cleft lip and microphthalmia or cyclopia. Affected children rarely survive past infancy(1). Complete failure of development of the primary optic vesicle results in anophthalmos(2). Complicated anophthalmos is a syndrome comprising of anophthalmos associated with craniofacial malformation, harelip, polydactyly, cardiac malformations and mental retardation(3). We report a similar case. This condition is extremely rare.

Case Report

A 21-day-old infant was admitted with facial abnormality, small eyes and inability to open the eyes. The baby was 3rd in order delivered at term normally. There was a history of antenatal drug
intake throughout pregnancy though the nature of drugs were not known. Natal and postnatal history was uneventful and there was no parental consanguinity. The family history was non contributory. The mother reported two still births but with no apparent congenital abnormality. She was married at the age of 21 years and was 24 years at the time of delivery.

On physical examination the baby weighed 2.2 Kg, head circumference was 26.5 cm, length 43 cm, chest circumference 29 cm, mid-arm circumference 8.5 cm, crownrump length 23 cm and rump to heel length 20 cm. All neonatal reflexes were normal. The head was globular microcephalic with normal black hair, no sutures and no fontanelle could be palpated. Craniotabes was present and transillumination was remarkable all over the skull. Both eyes were small in size with a slit like palpebral fissure (Fig. 1). The eyelashes, tarsal glands and puncta were present. The eyes were caved in. In both eyes no normal structures were seen or palpated. Palpebral aperture was smaller on the left side. On examination with the speculum there was a small rudimentary nodule in the right orbit which moved. The left pinna was normal sized, concha and scaphoid fossa were prominent and there was atresia of the external auditory canal. The right ear was normal. The dorsum of the nose was depressed, tip was bifid and right nasal vestibule was narrower than the left. Oral cavity was small in size but palate was high arched. There was no lymphadenopathy. Hands and feet did not reveal any abnormality. Scrotal sac was fully developed but testes were undescended bilaterally. Musculoskeletal system examination did not reveal any abnormality. The rest of the systemic examination was normal.

The investigations showed a hemoglobin levels of 11.0 g/dl, total leucocyte counts as 8,200/cu mm and differential count 38% polymorphs, 58% lymphocytes, 3% monocytes and 4% eosinophils. VDRL was non-reactive. X-ray skull showed premature closure of sutures. Ultrasound abdomen was normal. Echocardiogram did not show any abnormality. CT scan of the head showed absence of both hemispheres along with absence of falx and olfactory bulbs (Fig. 2). The thalamus, midbrain and cerebellum were normal. There was hypoplasia of left orbit along with hypoplastic intraorbital soft tissue contents. The right orbit was normal, hypoplasia of external auditory canal was also seen. Middle and inner ear were normal. A
Diagnosis of complicated anophthalmos was made.

**Discussion**

Complicated anophthalmos is a syndrome comprising of anophthalmos associated with craniofacial malformation, harelip, polydactyly, cardiac malformations and mental retardation. In most cases the cause of holoprosencephaly is unknown. Environmental factors, maternal disease and hereditary factors including consanguinity and chromosomal abnormalities have been associated with the disease. The most common chromosomal defect includes trisomy 13 and trisomy 18 short arm deletion syndrome. The association of holoprosencephaly with 13q deletion syndrome is well known. The gene responsible for the disease, however has not yet been identified. Five types of faces can be recognized that are pathognomonic and predict the malformed brain. Our case resembled type 3 cebocephaly. Bishop et al. have reported holoprosencephaly with no extracranial abnormality. Viney et al. have reported holoprosencephaly with retinoblastoma but no complicated anophthalmos. Very few reports are available on complicated anophthalmos.

**REFERENCES**