CYCLOPIA

by

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The anomaly consists of a single central orbital fossa with complete or partial fusion of the two eyes alongwith absence of a nose or a rudimentary nose situated above the eye and associated abnormalities of brain and other systems. Two cases of cyclopia met with in one year in Western Railway Hospital, Kota are reported.

Case 1

Mrs. K aged 21 years was admitted in the Hospital on 30-4-71 with the history of eight months amenorrhoea and labour pains. She was a second para, the first was a full term still birth, home delivery. There was no history of any specific infection, drug intake, rediation or bleeding during the present pregnancy. Examination showed that she was averagely built and nourished with moderate degree of hydramnios. There were no signs of toxaemia. Foetal heart sounds were not heard clearly. X-Ray abdomen was taken and diagnosis of hydramnios with anencephalic monster was made. Vaginal examination showed that the cervix was half dilated and membranes were very much tense and bulging. Artificial rupture was done and a good amount of clear liquor drained. After about half an hour of artificial rupture of membrane she delivered a live malformed female child with typical features of cyclops (Fig. 1 and 3). The child was gasping with open mouth and protruding tongue and remained alive for about two hours.

Weight of the cyclop was 1.1 KG., crown heel length was 48 cm. There was marked bending at the junction of trunk and lower extremities so much so that she took a sitting position immediately after birth without any support (Fig. 2). Single diamond shaped palpebral fissure was present in the centre of the forehead with nearly fused eye ball. Eye lids were rudimentary, eye lashes were present. Just above the central eye a tongue like process was seen. Ears were normal. Mouth was open, tongue protruding and palate closed. Skull bones, except in the lateral and part of posterior portion. were absent. On the posterior aspect a soft cystic mass covered with hair was present (Fig. 2).

Neck was very short and there was spina bifida throughout the vertebral column. Upper and lower extremities were bent inwards. Genitals were normal.

Post Mortum Findings

Nervous System: Entire brain was enclosed in cephalorachidian cavity. Cerebrum was small with smooth surface and with no evidence of convolutions and fissures. Cerebral hemispheres were not separated. On sagittal section of cerebrum a large single blown up cavity of lateral ventricle was seen to form a hydrocephalic sac. Corpus callosum, septum pellucidum and fornix were absent. Thalami were fused. The brain stem was small and deformed. The cerebellum was rudimentary. The spinal cord was lying opened up in rachidian cavity and only few nerves were attached with it. Olfactory bulb and tract were absent. Both optic nerves were fused to form single median atrophic nerve cord attached between the eye ball and the base of the brain. The other cranial nerves were also rudimentary.

Skeletal System: Vertebral column showed total spina bifida. Cervical vertebrae failed to fuse posteriorly to form a common cephalorachidian cavity and they were grossly deformed. Only three cervi-

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cal, eight thoracic and three lumbar vertebrae could be identified amidst the deformed irregular vertebrae. Sacrum and pelvis were apparently normal. The ethmoid and body of sphenoid were missing. Fused two halves of frontal bone formed the roof of the median orbit. Two maxillae were fused in median plane of face. From foramen magnum to the anterior fontanellae cranial vault was opened up to form cephalic part of cephalorechidian cavity.

Abdominal Cavity: There was complete failure of rotation of gut. The caecum and appendix were lying in the umbilical region. Uterus was subseptate. Rest of the viscera were normal. There was no evidence of diaphragmatic hernia.

Thoracic Cavity: The heart and lungs appeared normal. The upper part of left thoracic cavity was occupied by a rounded cystic swelling of 3 X 3 cm in size indenting the apex of the left lung. It was lying free and not connected with any neighbouring structure. It turned out to be dermoid exhibiting fibromuscular wall enclosing epithelium of squamous and columnar type. Microphotograph of the cyst wall, Fig. 4).

Endocrines: Hypophysis could not be located, probably damaged during removal of the brain. Contrary to the expectation surprarenals were well developed and about 1/3 of size of the kidney. Other endocrines were normal.

Case 2

Mrs. D. aged 26 years was admitted in the Hospital on 2-4-72 with the history of six and a half months amenorrhoea and pain in the adbdomen. She was a second para, first being full term normal delivery seven years ago. There was no history of abortion in the past, no history of taking any drug, bleeding or specific infection during present pregnancy. She never attended the hospital prior to this.

Examination revealed that she was averagely built and nourished with the pulse rate of 78 per minute and blood pressure 110/70 mm of Mercury. There were no signs of toxaemia or hydramnios. Uterus was twenty-eight weeks' size, head was presenting and foetal heart sounds were absent. She was having good uterine contractions and delivered prematurely soon after admission. The placenta was retained and manual removal was done. The child was still born, male, a typical cyclop (Fig. 5), weighing 1 Kg. and crown heel length was 31 Cms., head was hydrocephalic. The face was malformed with no eyes as such but a small transverse slit like structure in the centre at the margin of which soft eye lashes could be identified. Nose was absent, mouth looked normal in appearance. Palate was closed. Right ear was normal, left was smaller, half the size of the right. Neck was short. Upper extrimity—right was almost normal, left was very much shortened with lobster claw (Fig. 5). Left shoulder joint was dislocated. Humerus on this side was ill developed. Both the bones of forearms were absent. Only two phalanges were present. Lower extremity and genitals were normal. The cut end of umbilical cord examined showed opening of only two blood vessels. It was injected and presence of single umbilical artery demonstrated (Fig. 6). Placenta was kidney shaped and attachment of the cord was extreme marginal. Autopsy could not be performed.

Discussion

This malformation has been of much interest to the embryologist not only because it presents a striking deviation from the normal mode of human development, but also because of the frequency with which it is observed in other animals and ease with which it can be produced experimentally in lower forms of life. Though the importance has particularly been centred on the eyes, yet the associated abnormalities of the brain are equally significant.

This malformation has been observed in one member of a pair of twins and in one member of a double monster. Ballantyne (1904) found it in three members of a litter of eight pigs, an animal in which it seems to be particularly common. He stated that in many animals it ranks first

in frequency replacing anencephalus which is one of the most common malformations of the nervous system in human foetus.

The classical description of cyclop as described by Potter (1957), Morrison (1963), Daniel (1966) and also simulating with the present cases consists of a single centrally placed orbit in which the eye ball may be absent or rudimentary or show differentiation to a large almost a perfect eye. Alternatively, all degrees of doubling through a fused eye with two small eye balls which are nearly complete but lie adjacent may occur. Eyelids may be rudimentary or absent. Nose may be absent or represented by a short epithelial like structure situated above the eye

Cyclopia is commonly associated with other defects like aganthia, otocephaly, hiatus hernia, polydactyly, etc. (Ballentyne 1904) but its association with inencephaly (malformation of nape of neck and of the brain with reduction of number of vertebrae) as was present in our first case, is extremely rare. Anomalies of brain usually associated with cyclopia are—the hemispheres are fused into a single mass with a single open ventricle without olfactory nerves, corpus callosum or septum pellucidum, their substance is considerably reduced and the convolutional pattern simplified. Cerebellum is usually normal and the brain stem and cord are only reduced in size. There are usually areas of atypical proliferation of retinal tissues in the eyes and the optic nerve is single or absent

Presence of a short neck with webbing and incurving of little fingers—a combination of anomalies found in chromosomal aberrations—are not usually found in association with cyclopia as reported by Howorth *et al*, (1961), but incephaly in first and short neck in the second case were present in our cases. Similarly, association of hydramnios with congenital anomalies of foetus more so with anencephaly is not so important in cyclopia, though hydramnios of moderate degree was present in one of the above cases. Single umbilical artery which is also a common association with congenital malformations was found in one of the cases where umbilical artery was studied.

The pituitary glands may be absent. In a series of 53 cases, Edmonds (1950) described about the presence or absence of it in 11. Out of these 11 cases pituitary was found to be absent in 6 and in 2 cases of cyclops lacking pituitary glands, the adrenal cortex and thyroid were reduced in size. In case 1, hypophysis could not be located and contrary to the expectation endocrines were normal.

The etiology of this condition is obscure. Stockard (1909) showed that by temporarily reducing the metabolic rate at a time when a particular group of cells was at the peak of multiplication and differentiation subsequent development of that group of cells might be profoundly altered. He found that stimuli which would produce cyclops when acting at one time of embryonic development would produce partial or complete duplication of foetal body structure resulting in the formation of conjoined or separate twins if they act little later.

From experimental and comparative studies, it is now clear that the deficient part of a cyclopic embroyo can be traced to a wedge shaped defect in the anterior end of the forebrain and adjacent mesoderm at a very early presomite or even preneurular stage of development (Willis 1962). This defect is the result of the failure of primary head organiser to exert its inductive influence on the neighbour-

ing ectoderm and mesoderm. The exact cause of failure of primary organiser is not known. It may be a drug induced, as also suggested by Connaughton *et al.*, (1970) while reporting a series of three cases in one year or a genetically determined defect and Chromosomal studies may throw some light on this point.

Summary

Two cases of Cyclops occurring in one year are presented. The salient features associated abnormalities and possible etiological factors are discussed.

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See Figs. on Art Paper II and III