Cyclopia in Association with Trisomy D, Polydactyly, Accessory Spleen, and Meckel's Diverticulum

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Cyclopia is a rare congenital malformation characterized by the presence of a single orbital cavity in the area normally occupied by the root of the nose, and of a proboscis-like appendage located on the lower part of the forehead just above this cavity. There are three reports in known literature describing trisomy D in cyclopia[1~3]. *This report identifies a case of cyclopia in a female newborn, in association with trisomy D (47, XX D+), polydactyly, two accessory spleens and a small Meckel's diverticulum in ileum.*

Case Report

The infant was born at Taipei Municipal Jen-Ai Hospital on May 22, 1973 at 35 weeks' gestation. The mother was a 29-year-old Chinese high school teacher, gravida III, para II. The first pregnancy resulted in the delivery of a normal female infant 2 years and 4 months ago, the second pregnancy terminated by artificial abortion 2 years ago. She had been inserted a contraceptive loop since August 8, 1971. On November 12, 1972, her family physician conducted a routine examination of her because of amenorrhea. He could not find the loop in situ and revealed that a pregnancy test taken was positive. She had been given various medications including aspirin, antihistamine, barbiturate, tranquilizer and antitussives for occasional U. R. I. attacks during the fourth month of her relevant pregnancy. Polyhydramnios was noted approximately 2 weeks prior to delivery and abdominal X-ray examination was taken. The father, a 30-year-old Chinese had always been healthy. There is no consanguineous relationship between the father and mother. The family history did not reveal the occurrence of any congenital malformations or mental retardation.

The labor was uneventful but the infant lived only three hours. Examination of the baby revealed a premature female infant with cyclopia malformation (Fig. I). It's birth weight was 1250 gm, and length 40 cm. The circumferences of its head, chest and abdomen were 26, 21, and 18 cm, respectively. The Apgar score at one minute after delivery was 8. There was no jaundice, pallor, or edema but a slight peripheral cyanosis was noted on both hands and feet. The anterior fontanel was closed. There was a diamond-shaped

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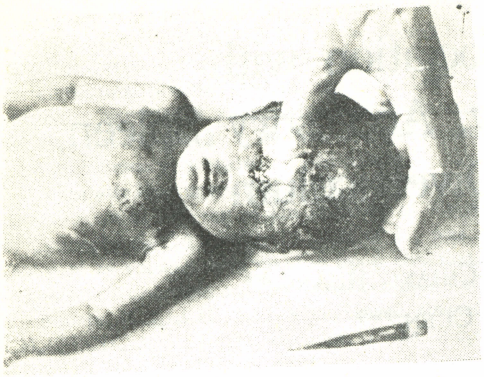

Fig. I

orbital cavity with two small fissures extending laterally from the angles and located centrally in the area corresponding to the root of nose. The lids were maldeveloped. A proboscis-like tubular appendage was situated on the low part of it's forehead just above the orbital cavity. The ears were low set but normal in shape. The mouth was normal, neither hare lip nor cleft palate was noted. The heart sounds were of good quality, no murmurs were audible. The breath sounds were slightly feeble. The abdomen was soft and flat. The liver was palpated one and half fingerbreadths below the right costal margin. The spleen was not palpable. Polydactyly with a sixth digit on the fibula aspect of each foot and on the ulnar aspect of the left hand were noted. The external genitalia appeared normal for a female infant. The remainder of the physical findings were unremarkable. The placenta measured 21×19×4.8 cm in size and weighed 500 gm. The maternal surface was covered by a litter of blood clots. The fetal surface was smooth and glistening. The umbilical cord measured 28 cm in length and 40 mm in diameter. It normally contained three blood vessels.

Cytogenetic Studies: Chromosomal analysis was performed according to a modification of the method of Moorhead and associates[4]. Cultures of peripheral leukocytes, obtained via cardiac puncture after death of the proposita showed 47 chromosomes with an extra chromosome in Group D (Fig. II). In the parents and sister, the peripheral leukocytes chromosomal analysis revealed normal karyotypes.

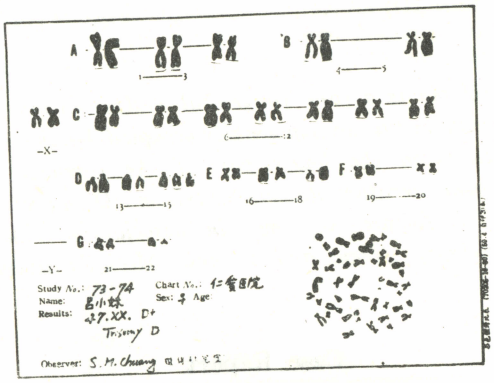

Fig. II

Postmortem Examination: The most striking findings are as follows: 1) Cyclopia[5] (centrally placed, a single orbital fossa containing two closely adjacent eye balls), 2) Superiorly placed polypoid nose with a small central cavity that ends blindly on the surface of the head, 3) Low set ears, 4) Polydactylia; There is a soft polypoid supernumerary finger attaching on the lateral aspect of metatarsal of the left little finger (postminimi without bone). Both feet are found to have six completely separated and well developed toes, a picture compatible with duplication of the fifth digit, 5) Two small accessory spleens measuring up to 1.4×0.7×0.7 cm in size, 6) A small Meckel's diverticulum measuring 1.6 cm, in length and 0.4 cm, in diameter is seen in the ileum, about 30 cm. proximal to the ileocecal valve, 7) The brain is hypoplastic and maldeveloped weighing 67 gm. The cerebral hemispheres are

fused with a single open ventricle without septum pellucidum and corpus callosum. The littoral of the ceredral hemispheres is directly connected with midbrain in the anterior portion and there is no third ventricle to be seen. But, the opening of the aqueduct of Sylvius can be identified on the lower dorsal portion of the hypertrophic midbrain and the fourth ventricle is normal in position but apparently enlarged. Two optic nerves are identified but the olfactory nerves are absent. The cerebral gyri and sulci are poorly developed and the blood vessels on the surface of the brain are apparently increased and markedly congested. The spinal cord is not remarkable.

Comments

There are various causative factors of cyclopia have been reported such as endocrines, anoxia, external compression of maternal uterus, advanced maternal age, hydramnios, etc. Hereditary factor has been considered by many authors where cyclopia had occurred in twins, in consanguineous parents, in same family and in family of arhinencephaly. Some authors are not satisfied with such an explanation because cyclopia are embryonic developmental defects. Harmful environmental agents such as maternal disease (rubella), drug ingestion, and radiation have been suggested. However, no clear environmental teratogen associated with cyclopia in man has been established.

Cyclopia is the severest malformation of arhinencephaly which has been used to describe a wide spectrum of faciocerebral malformation. Arhinencephaly and related malformations have been considered typical of trisomy D, but there are only three of eleven reports in literature known to have described cyclopia with chromosomal analysis were trisomy D (Table I); the other eight reports including three cases of normal chromosomes[6~8], two cases of deletion of short arm of chromosome 18[9,10], one of monosomy G mosaicism[11], one with 47 chromosomes in 30 per cent of cells and the extra chromosome being a fragment with satellities[12], and one of a ring chromosome 18[13]. Since this malformation has been found in either normal or various abnormal chromosomes, cyclo-

Table 1: Cytogenetics of Cyclopia

Author	Karyotype
Sohval (1961)	Normal
Lejeune (1965)	Normal
Jubery (1965)	Normal
Faint and Lewis (1964)	Deleted short arm of No. 18
Nitrowsky et al. (1966)	Deleted short arm of No. 18
Cohen (1966)	Normal/monosomy G
Pfitzer (1967)	Normal/normal plus fragment
Cohen et al. (1972)	Normal/ring chromosome No. 18
Toews and Jones (1968)	Trisomy D
Arakaki and Waxman (1969)	Trisomy D
Halbrecht et al. (1971)	Trisomy D
Present case	Trisomy D

pia has no direct relationship to the trisomy D or to other chromosomal abnormality.

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併有 Trisomy D, 多指(趾)症, 副脾, 和美克耳憩室的獨眼畸形兒之一例

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我們於1973年5月22日在臺北市立仁愛醫院遇見一位獨眼畸形的新生女嬰。在病人鼻骨上方有一個眼窩，在此眼窩的上方，即在前額下部中央處有一條象鼻樣的管狀附屬物，左手和双足各有多指(趾)畸形。染色體檢查發現有 Trisomy D (47，XX D+) 異常。解剖檢查除了腦、眼、鼻、手、足、等處有異常外，並發現有兩個副脾，且圳廻腸處有一個美克耳氏憩室。

獨眼畸形是一種很少見的先天性畸形，併有 Trisomy D 染色體異常的獨眼畸形症更爲罕見，在文獻上能找到的報告至今只有三例而已。