A PEDIGREE OF ANOPHTHALMOS*

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THE difficulties inherent in any assessment of the exact status of a case of anophthalmos is brought out by the following report of a family in which one child and two of its sibs were affected, but no ocular anomalies were found in the ascendants. The pedigree is shown in the Figure.

FIGURE.—Pedigree of a case of bilateral anophthalmos.

Case Report

A male infant aged 9 months (III, 7) was seen with a history of having been born blind. On examination he was found to have very small palpebral fissures with the eyelids very deeply set. The lower part of the lower lids and the upper part of the cheek showed a soft globular bluish swelling fairly well demarcated. On opening the lids there was no eye to be seen and the orbit appeared to be lined by the conjunctiva. The appearance was similar on both sides—the left side showing a more marked swelling. The child was otherwise normal. 6 months later he was brought back because the swelling

on the left side had suddenly increased. An exploratory puncture of the cystic swelling showed old blood which was evacuated.

The mother gave a history that two other boys (III, 4 and 6) born to her had been similarly affected, but had both died in infancy. A third child, a girl (III, 5), who also died in infancy, had normal eyes. No history of a similar condition in the ascendants or collaterals could be established.

The father (II, 2), who was aged 50 years, had full vision in each eye. The eyes were normal in size, emmetropic, and free of any abnormalities.

The mother (II, 4), who was aged 33 years, also had entirely normal eyes, normal in size, emmetropic, and with full vision.

Discussion

In view of the family history, causative environmental factors can be excluded from further consideration. A search for ocular anomalies in other members of the family proved difficult; only one sib (III, 3) of the affected boy could be seen, and she was found to be normal. The findings—observed anophthalmos in one child and reputed affection in two more sibs—suggest that anophthalmos in this family is recessive, which is consistent with other reported cases (Sorsby, 1951). The fact that the only child of the father's first marriage was normal fits this assumption, as does the fact that the three children born of the marriage of the father's brother and mother's sister were also normal. Presumably the only marriage where both parents were carriers was that of the parents of the affected sibship.

The available literature offers conclusive evidence for a sex-linked type as well as a recessive type. The sex-linked type is generally associated with mental deficiency and frequently with various neurological disturbances (Roberts, 1937; Sjögren and Larsson, 1949). Since in the present family the three affected sibs were all boys while three girls were all normal, recessive sex-linkage needs to be considered. This would postulate the mother and maternal grandmother being carriers and the mother's sister being normal. The absence of males in the mother's sibships precludes any test of this assumption.

The presence of a cystic swelling is common in anophthalmos. Such cysts are not always evidence of persistent choroidal cleft, for cysts may form without any developmental anomaly of the choroidal cleft, as is shown by the experimental observations of Chase and Chase (1941) on recessive anophthalmos in the mouse. In this condition the primary optic vesicle is formed normally and development proceeds normally until about halfway through pregnancy. At this stage, instead of further differentiation, regressive changes set in, leaving a variable degree of cyst formation. A cystic swelling can be taken as evidence of failure of closure of the choroidal cleft if microphthalmos is also present, but a microphthalmos is not in itself evidence of persistent choroidal cleft, since it may well be an abortive anophthalmos (as is seen in strains of anophthalmic mice). The significance

of anophthalmos and microphthalmos becomes still more complex when it is realized that these anomalies may be part of wide-spread defects bordering on the disorders seen in such conditions as the Laurence-Biedl syndrome (François, 1953). Anophthalmos corresponding to the experimentally observed anophthalmos in the mouse appears to be determined in the human foetus by anomalies at the end of the first month of pregnancy (Moretti, 1955). In these cases, as in the experimentally observed mouse, there is no gross maldevelopment of the brain. Gross cerebral anomalies would seem to occur if there is anomalous development at an even earlier stage, so that a monster is born rather than a defective viable child.

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