BUPHTHALMOS OVER THREE GENERATIONS*

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Case Reports

Case 1, a female child (IV, 3 in Pedigree) born on December 23, 1958, was noticed to have opaque corneae, of sufficient degree to prevent any estimate of the size of the pupils. There was corneal oedema but no appreciable abnormality of the corneal diameter. In the course of a few days the cornea of each eye had become obviously enlarged and a definite buphthalmos had become established.

The father (III, 3) informed me that he suffered from buphthalmos, and that his father (II, 2) also had the same condition, and I have since been able to examine them both.

Case 2 (III, 3) was first seen by an ophthalmic surgeon in March, 1942, at the age of 15. Both eyes then showed definite buphthalmos with both discs pale and cupped. He has had treatment and now shows a symmetrical condition with a corneal diameter of 14 mm. The visual acuity in the right eye is 6/6; in the left eye it is 6/60. Both optic discs are pale and cupped. During the last 2 months the left eye has developed a high tension with corneal oedema and a further drainage operation has been necessary.

FIGURE.—Family Tree.

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Case 3 (II, 2), born in 1902, was first seen by an oculist at the age of 35. Both eyes were then buphthalmic, and he gave a history of having noticed haloes at the age of 8. Both corneae are now markedly enlarged with deep anterior chambers and both discs pale and cupped.

Discussion

The literature has been extensively reviewed by Delmarcelle (1957), who found several instances in which it was claimed that inheritance had been established over several generations. He considered, however, that the evidence was not conclusive and in several cases he distinguished between buphthalmos and "juvenile glaucoma".

In the family here reported, both the grandfather and the father were discovered to have buphthalmos at the ages of 35 and 15 respectively, and there is undoubted enlargement of the corneal diameter in each of them.

It is now generally accepted that there is a genetic basis for buphthalmos; Sorsby (1951) pointed out that there were a number of pedigrees in which the affection had occurred over two generations in direct descent. François (1958) stated that there was no record of the affection over three generations. Westerlund (1947), who followed up 122 cases seen in Denmark, found 31 familial cases distributed over eleven families, and held that the gene was recessive and had a penetrance of about 40 per cent.

The pedigree published here shows the disease occurring in three generations with the grandfather transmitting his dominant gene to one of his three children.

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