BILATERAL CONGENITAL BUPHTHALMOS IN TWO SISTERS*

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THE relative rarity of familial cases of buphthalmos warrants the following record of two sisters suffering from severe bilateral buphthalmos at present under treatment at the Royal Eye Hospital (see Figure).

FIGURE.—Pedigree of affected sisters.

Case 1, a girl aged 3 years (III, 5), was born at full term on November 8, 1953.

At birth the corneae were noticed to be enlarged and cloudy, and she was transferred to the Royal Eye Hospital a few days later. On admission both corneae were enlarged and hazy. The anterior chamber was deep in each eye. Fundus details were not seen.

The corneal diameter was 11.5 mm. in the right eye and 12 mm. in the left. The ocular tension was 29 mm. Hg (Schiötz) in the right eye and 28 mm. Hg. in the left. The ocular tension was controlled in the right eye by goniotomy, using Barkan's method; this was repeated five times. The left eye required seven goniotomies and finally a trephine. Since May, 1955, when the child was 18 months old, the tension has remained controlled in both eyes, the corneae are clear, and no drops are required. The optic discs are pale and cupped.

Visual acuity, with illiterate "E" test, Right eye, with -7.5 D sph., 3/24.

Left eye, with -10.5 D sph., 3/60.

Binocular vision, with glasses, 3/18.

Case 2, a female infant aged 5 months (III, 6), was born at full term on February 24, 1957. When she was 5 days old it was noticed that both corneae were cloudy and she was referred to the Royal Eye Hospital. On admission both corneae showed dense annular opacities which made it difficult to estimate the depth of the anterior chambers and prevented ophthalmoscopy. The corneal diameter was 11 mm. in both eyes. The

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ocular tension was 60 mm. Hg. (Schiötz) in the right eye and 45 mm. Hg in the left. A right trephine was performed on April 4, 1957, and a left trephine on May 2, 1957. Both procedures were uneventful. The anterior chambers seemed deep though it was difficult to estimate the position of the limbus. At operation, the corneae were found to have an unusual consistency, the central portions especially feeling hard and fibrous. Despite the presence of satisfactory trephine blebs the ocular tension remains raised.

Discussion

Most cases of buphthalmos are "sporadic", but the available evidence makes it likely that the condition is not infrequently genetically determined. In a successive series of 172 cases, Bröns (1937) found nineteen familial examples, and Westerlund (1947) observed 31 familial cases in a series of 122. In the absence of any history of buphthalmos in the antecedents and with the lack of unusual sex distribution amongst the affected, autosomal recessive inheritance is a likely assumption—and this is supported by the high rate of parental consanguinity in both sporadic cases and affected sibs (Sorsby, 1951). In contrast there are several pedigrees suggestive of dominant inheritance. In the present sibship there is nothing to suggest dominant inheritance and, though parental consanguinity is lacking, the history is not inconsistent with recessive inheritance.

Summary

The occurrence of congenital buphthalmos in two sisters is recorded. The mode of inheritance of buphthalmos is discussed.

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