CONGENITAL TOTAL CATARACT—POSSIBLY RECESSIVE*

BY

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Since there are few cases on record of congenital cataract inherited in a recessive manner (Sorsby, 1951), the following report in an affected sibship is perhaps worth recording. The pedigree is set out in Fig. 1.

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**Fig. 1**

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**Case Report**

A girl aged 13 years (II, 1) had a history of having been born blind.

*Examination.*—She was well developed and intelligent. There were no signs of rickets. The eyes appeared slightly sunken and smaller than normal. Gross nystagmus was present. The corneae were clear, the anterior chambers of normal depth, and the pupils normal. The lens showed total cataract; no fundus view was possible.

*Family History.*—A younger sister was similarly affected. This child, aged 9 years (II, 2), was also found to be healthy and intelligent with no signs of rickets. The eyes appeared to be normal in size. Nystagmus was present. The corneae, anterior chambers, and

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pupils were normal. The lenses showed total cataract and no fundus view was possible. Apart from a younger brother (II, 3), who died at the age of 1 year and was affected—according to the mother’s statement—, there were no other affected members in the sibship or ascendants. The parents were not consanguineous nor did they come from neighbouring areas.

In the personal history of the mother there were no suggestion of rubella during any of the pregnancies.

The visual acuity of the father of the affected children (I, 3), who was aged 35, was as follows:

- **Right eye**: unaided 6/60; with -3.5 D sph., -1 D cyl., axis 180°, 6/9.
- **Left eye**: unaided 6/60; with -2.5 D sph., -1.5 D cyl. axis 180°, 6/6.

His eyes were normal in size. No lens changes were detected with the slit-lamp, and the fundi were normal.

The mother (I, 5), who was aged 30 years, was highly myopic; the visual acuity was as follows:

- **Right eye**: with -12 D sph., 6/18.
- **Left eye**: with -12 D sph., 6/18.

Her eyes were normal in size. There was no evidence of lens changes. The fundus showed typical changes of myopia. The discs were large and pale and there were large areas of choroidal atrophy all around them. The maculae showed dark pigmented areas.

The grandparents of the affected children were dead, but were reputed to have been normal.

It was possible to examine only one other member of the parent’s generation (I, 1), and his eyes were normal. (Visual acuity 6/6 in each eye, emmetropic, normal globe, fundus normal, no anomalies of the lens visible with the slit-lamp.)

The occurrence of cataract in two and possibly three sibs—the cataract being identical in the two observed sibs—makes it unlikely that an environmental factor came into play. Though there is no evidence of consanguinity, recessive autosomal inheritance is likely and could fit in with similar cases recorded. The alternative explanation, that in this family the cataract was inherited in an irregularly dominant manner, finds no support in the family history or in the literature.

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**REFERENCE**

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