equilibrium and in respect to sodium and chloride secretory energy must be postulated in the formation of the intra-ocular fluid.

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AN INVESTIGATION INTO THE MODE OF HEREDITY OF CONGENITAL AND JUVENILE CATARACTS*

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It has been generally accepted since the investigations of Nettleship (1905) and Bateson (1909), that congenital and juvenile cataracts are inherited as dominant characteristics. It is reasonable that this should be so, for the literature provides many examples of cases in which such cataracts have been traced through several generations, and it leaves no doubt about the existence of dominant types.

There are, however, many reported cases of congenital and juvenile cataracts appearing sporadically, or in small numbers within the same family, and in which they have not been traced to succeeding generations. Such cataracts are frequently recorded as "familial"; they are often of the congenital or juvenile variety, and the possibility of their hereditary nature has received little consideration.

Franceschetti (1930) makes the following statement in *Kurzes Handbuch der Ophthalmologie*: "Das häufige isolierte Auftreten bei Geschwistern liesse auch an recessive Vererbung denken. Da aber Konsanguinität selten ist, kommt unregelmässige Dominanz in Frage."

Waardenburg (1932) says: "Auch die angeborene Katarakt wird im allgemeinen in dominanter Weise vererbt. Es liegen aber

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einige Beobachtungen vor, wodurch die Möglichkeit, dass es vielleicht eine rezessiv-erbliche Typen gibt, nicht ganz in Abrede gestellt werden kann. Die Rolle der Blutverwandtschaft ist eine geringe."

In "Handbuch der Erbkrankheiten" (1938), Bücklers has given the following declaration: "Das Schriftum ist zwar reich an Mitteilungen über erbliche Stare, doch sind viele Veröffentlichungen nicht zu verwerten, weil ausführliche Beschreibungen und gute Abbildungen fehlen. Es soll deshalb nicht verschwiegen werden, dass unser Wissen gerade auf diesem Gebiete noch lückenhaft ist."

Emphasis has been placed on the rôle of exogenous factors, *e.g.*, spasmophilia and rickets in the aetiology of just these types of cataract, and more recently attention has been drawn to the influence of virus diseases, *e.g.*, rubella. The possibility of hereditary influences tends to be overlooked and calls for further investigation.

With this possibility in view, all cases of congenital cataracts and juvenile cataracts which came under the author's care during six years as eye-specialist in *Telemark*, Norway, have been re-examined. This country district is relatively isolated and intermarriage is frequent, so that grouping of recessive genes is to be expected, with a correspondingly high number of homozygotes. Although statistical proof is lacking, hereditary eye diseases appear to be very common in this district.

Patients with congenital and juvenile cataracts were examined. In the published pedigrees the individuals physically examined are marked thus ©. Information about their families was acquired from the patients themselves, their parents, their physicians, from parochial registers, and in some cases from other clinics.

The following are the more important points in the history of each patient:

Number of children in the family; number of affected individuals and their sex; convulsions or rickets in childhood; rubella or other diseases of the mother during pregnancy; consanguinity of the parents.

In the examination of the patient, the following points received attention:—

Type of cataract; comparison with types of cataract in affected siblings; uni- or bilateral; association with other defects or eye diseases, *e.g.*, nystagmus, microcornea; prognosis of the cataract.

The main object of this paper is to investigate the mode of inheritance in the reported cases of congenital and juvenile cataracts.

MATERIAL

(a) Survey

Within 17 families, 27 cases of congenital and juvenile cataracts are reported, 20 as congenital and 7 as juvenile cataracts.

In 8 sibships the cataract was evident in 2 or more individuals. In 7 sibships consisting of more than 2 individuals, only 1 was affected. In 2 cases the affected person was the only child.

Among the 27 affected individuals, 14 were males and 13 females. In 9 of the 17 families the parents of the affected individuals were related, and in 5 of these the parents were first cousins. In 4 families the relationship was more distant.

As to the diagnosis, 20 cases are recorded as congenital cataracts, with different sub-titles — cat. totalis, subtotalis, nuclearis, membranacea, polaris posterior et polaris anterior. In 7 of the patients the diagnosis is cat. juvenilis, with the sub-titles of cat. corticalis posterior in 1 (Case 15) and cat. zonularis in the other 6. Possibly some of these zonular cataracts were, in fact, also congenital.

The cataracts were bilateral in all cases, except in Case No. 7. In Case No. 5 the cataract was unilateral at birth, but appeared later in the other eye.

Within 7 of these families (Nos. 1, 4, 5, 8, 12, 14 and 17) cases of cataracta senilis were also found, but only in 1 family (No. 12) was the senile cataract recorded in the father of patients with congenital cataract. In the rest, the senile cataracts were spread over other generations or within other side-branches.

Two of the patients (Cases Nos. 12 and 13) also suffered from retinitis pigmentosa, as well as cataracta corticalis posterior and polaris anterior, and in Case No. 13 surdo-mutitas was also found. One of the patients (No. 14) was feeble-minded.

In 2 of the patients (Nos. 22 and 23) there was a history of convulsions and rickets in childhood. Case No. 23 also had enamel defects of the teeth.

None of the patients' mothers could remember having had rubella or other infectious diseases during pregnancy.

*Case Reports and Family Histories**Family 1: Cases 1 and 2.*

O.S., male, born November 12, 1927, and A.S., male, born August 12, 1942, are No. 1 and 3 in a fraternity of three. Born in Bø, Telemark.

Case 1, O.S., male. The cataract was noticed before the age of one year. Operation performed at age of 3 years in the right eye and at age of 3½ years in the left eye. A homogeneous greyish opacity covered all the pupillary area (3 mm. in diam.). No red reflex. Later on he was again admitted to a clinic for discissio cataractae o.u.

9 years old: No nystagmus. Corneal diameter normal o.u. Visual acuity o.d. +13 = 5/15; o.s. +13 = 5/30. Cat. sec. o.u. The patient had always been

healthy. No convulsions. No indications of rickets. His mother was healthy during pregnancy.

Diagnosis: Cataracta congenita (subtotalis).

Case 2, A.S., male. The patient's parents noticed the cataract immediately after birth. Otherwise he had been in good health. No convulsions. No indications of rickets. Mother healthy during pregnancy. Examined at age of 18 months and admitted to a clinic at age of 3 years.

3 years old: The patient was well-built and looked well. No sign of rickets. In both eyes there was an opacity in the central part of the lens and also considerable opacities in cortex. Discissio cataractae o.u.

5 years old: Repeated needlings. Vision: o.d. + 10 = 5/20; o.s. + 10 = 5/20.

Diagnosis: Cataracta congenita (subtotalis).

Pedigree information. See Fig. 1. The family was traced through four generations. In a fraternity of three (gen. IV) No. 1 and 3 had congenital cataract. No. 2, G.S., female, was normal (16 years old). The parents (G.S., male, 58 years, and

FIG. 1.

Family 1. Pedigree demonstrating that the parents of Case I, O.S., male, and Case II, A.S., male, both with congenital cataracts, were first cousins, H.O. (gen. I) aged 76, and H. S. (gen. III) aged 64, had senile cataracts.

I.S., female, 47 years) of the affected children were normal. G.S., male, was No. 3 in a fraternity of three. No. 1, H.S., male, 64 years old, had a cat. senil. incip. with sector-shaped opacities in the anterior cortex. Vision: o.d. = 5/15; o.s. = 5/20. No. 2, M.S., male, 61 years old, his wife M.S., 62 years old, and their daughter, A.S., 17 years old, were all normal.

The grandparents of the affected children (O.S., T.S., A.H. and O.H., gen. II) all died at an advanced age (69, 79, 59 and 83 years) and had had good vision. Their great grandparents (M.T. and A.T., gen. I) died aged 45 and 34 years respectively, and had had good vision. The paternal great-grandfather (H.O., gen. I) had cat. senil. and died at the age of 76 years.

From the pedigree it is seen, that the patient's parents were first cousins.

Summing up, there were in this family two cases of cat. congen. The patient's parents were first cousins. Furthermore there were two cases of cat. senil. one in the patient's uncle and one in their great grandfather.

Family 2: Case 3.

M.D., female, born May 12 in Skien, and was No. 1 among two sisters.

Case 3, M.D., female. History given by the parents of a healthy childhood. She had never had spasms or rickets. The cataract was noticed when 1 year old.

2 years old: Referred to a clinic for operation. At that time o.d.: Dense greyish

opacity in the pupillary area. No red reflex. O.s.: Grey opacity in the pupillary area with no shadow of the iris. She was operated on several times o.u. Attended elementary school and was able to read with the left eye. The right was almost blind. The patient's mother was said to have been healthy during pregnancy.

FIG. 2.

Family 2. Pedigree demonstrating that the parents of Case 3, M. D., female, with congenital cataract were third cousins. The consanguinity between the families 2 and 3 is also demonstrated.

FIG. 3.

Family 3. Pedigree demonstrating that the parents of Case 4, H. A., male, and Case 5, M. A., female, both with congenital cataracts, were first cousins.

21 years old: Nystagmus. Corneal diameter normal o.u. o. dext. in divergens. Total coloboma. Cat. sec. o.s. Pupillary area clear. Good red reflex. Vision o.d. P.L., O.s.=5/10.

Diagnosis: Cataracta congenita (subtotalis).

Pedigree information. See Figs. 2 and 3. The family was traced through six

generations. The patient's sister (B.D., female, aged 22 years old) was normal. Their parents (A.D., female, aged 51 years, and P.D., male, aged 53 years old), were also normal. Their grand-parents (generation IV) died at the ages of 70, 39, 47 and 87 years, and had normal vision. Their great grand-parents (gen. III) died at the ages of 90, 90, 87 and 70 years, and had normal vision. Information about the other generations was negative except that No. 2 and 3 in gen. II were brothers, and that they had good vision. Both of them died at an advanced age. The pedigrees of No. 2 and 3 show that there is consanguinity between the two families.

Hence, in this family there was one case of cat. congenita (subtotalis) in a fraternity of two. The patient's parents were third cousins. There was consanguinity also between this family No. 2 and the family No. 3.

Family 3: Cases 4 and 5.

H.A., male, born February 20, 1920, and M.A., female, born June 2, 1922, in Siljan, Telemark, were No. 1 and 3 in a fraternity of four.

Case 4, H.A., male. The cataract was noticed at the age of 6 months. No spasms. No rickets. Admitted to a clinic at the age of 3 years. His mother was healthy during pregnancy.

3 years old: It was impossible to identify the type of cataract from his record. Possibly it was a membranous cataract. Operation on both eyes. At the ages of 4 and 5 years he was operated again, and was then able to read and passed intermediate school examinations.

17 years old: Nystagmus present. Corneal diameter normal o.u. Cat. sec. o.u. Vision o.d. + 8 = 5/20; o.s. + 8 = 5/20.

The patient was otherwise healthy and well developed for his age.

Diagnosis: Cataracta congenita (membranacea).

Case 5, M.A. The cataract was noticed immediately after birth. No spasms or rickets. Her mother healthy during pregnancy. Her right eye was operated on at the age of 2 years. Had good vision in left eye. Also in this patient it was impossible to state the type of the cataract from the description, but most probably it was a total cataract.

24 years: Looked healthy. No nystagmus. Corneal diameter normal. O. dext in divergens. The pupillary area was clear with red reflex. O. sin. Central opacity in posterior cortex. Vision o.d. Counts fingers at $\frac{1}{2}$ m.; o.s. = 5/20. Operation still not advised in left eye, as reading was good.

Diagnosis: Cataracta congenita (totalis)?

Pedigree information. See Fig. 3.

Investigations through five generations. The patients were No. 1 and 3 in a fraternity of four. No. 2, J.A., male, born March 26, 1921, and No. 4, J.A., male, born July 30, 1923, were both normal. The patient's parents J.A., aged 56 years, and I.A., aged 55 years, were normal too. They were first cousins. The other members of the parents' group and their children were all normal. Three of the patient's grandparents (gen. III) died at advanced ages (72, 59, 82 years). No. 1, J.A., 92 years old, was still alive, and had normal eyes. Two of the patient's grandparents (M.A., female, and H.F.) were sister and brother. Their parents (gen. II) died aged 61 and 55 years, and had good vision. The connection between this pedigree and the previous one is obvious and demonstrates that the father of Case 3 (Fig. 2) was second cousin of the father and the mother of Cases 4 and 5 (Fig. 3). The side branches in the descendants after M.F. and I.F. (gen. II) were traced and no other cases of cataract found. In this family two patients of a fraternity of four had cat. congenita (?membranacea, totalis). The patient's parents were first cousins. Consanguinity between this family and the previous family No. 2 was noted.

Family 4: Case 6.

O.E., male, born August 26, 1939, in Rauland, Telemark, was No. 2 in a fraternity of two.

Case 6, O.E., male. The cataract was noticed at the age of 1 year. His mother was healthy during pregnancy. He looked well. No spasms. No rickets.

1 year old: Nystagmus. Corneal diameter 9 mm. o.u.

In both of the lenses there were opacities, most dense in the central part, but also in cortex. Faint red reflex in the peripheral areas of the lenses.

5 years old: Mydriasis. o.u. The lenses were opaque all over, more dense in the central part than in the periphery. Faint red reflex from the lens-periphery. Vision: eyesight enough to walk by himself. The patient was operated on: Discissio et evacuatio cataractae o.u. Vision after operation difficult to state (+11 = 25/20 o.u.).

Diagnosis: Cataracta congenita (totalis).

Pedigree information. See Fig. 4.

FIG. 4.

Family 4. Pedigree demonstrating that the parents of Case 6, O. E., male, with congenital cataract, were fifth cousins. O. K., gen. V, had senile cataract. (Aged 83).

The family was traced through eight generations. The patient's sister, M.E., was examined 18 years old, and was normal. His parents M.E., female, aged 49 years, and K.E., aged 68 years, were also normal. They were fifth cousins. The sisters and brothers of the patient's parents, and their children had normal eyes. The patient's grand-parents (gen. VI) of which only M.K., aged 78 years, was still alive, had good vision. Three of them died at advanced ages (70, 70 and 80 years old). From gen. V it was known that H.K., A.K., O.K. and K.K. died at advanced ages (81, 75, 83 and 96 years respectively) and, with the exception of O.K., who had cat. senilis, all had good vision. About the individuals in the other generations

I, II, III, and IV, it was only known that they died at advanced ages and probably had good vision. The descendency from gen. I was traced extensively, but no other cases of congenital or juvenile cataracts were found.

In this family, one patient in a fraternity of two had cat. congenita (totalis). The parents of the patient were fifth cousins. The patient's great grandfather had cat. senilis.

Family 5: Case 7.

I.L., male, born 1943, was the only child. Born in Mo, Telemark.

Case 7, I.L., male. His parents noticed the cataract at birth. No spasms. No rickets. His mother was healthy during pregnancy.

FIG. 5.

Family 5. Pedigree demonstrating that the parents of Case 7, J. L., male, with congenital cataract, were fourth cousins: G. L., gen. V, (aged 70), had senile cataract.

4 years old: The patient looked well. No nystagmus. Corneal-diameter normal o.u. o.s. In the central part of the lens there was a grey opacity formed as a disc. No riders. Red reflex around the opacity o.d. The lens was clear. Fundus normal. Vision: o.d. = C.F. at 1 m.; o.s. = 5/5.

Diagnosis: Cat. congen. (nuclearis) o.d.

Pedigree information. See Fig. 5.

Investigations into the family was carried out through seven generations. The patient's parents, K.L., male, aged 43 years, and A.L., female, aged 32 years, were normal. They were fourth cousins. The other members of the parents' fraternities and their children were normal. The patient's grandparents (gen. V) were still alive H.L. excepted, who died aged 58 years, and had good vision. The others

were G.L., female, aged 70 years, T.L., male, aged 62 years, T.L., female, 78 years old. The first one had cat. senilis with wedge-shaped opacities in cortex. Vision: o.d.=5/20; o.s.=5/15. The others had normal eyes. Information from the other generations disclosed no cases of cataract, and these individuals died at advanced ages and were supposed to have had good vision. Investigations into their descendants disclosed no congenital or juvenile cataracts.

In this family, the only child of normal parents, who were fifth cousins, had cat. congen. (nuclearis). One of the grandmothers had senile cataract.

Family 6: Cases 8 and 9.

E.M., male, born May 31, 1901, and K.M., male, born June 28, 1905, in Mo, Telemark, were No. 1 and 3 in a fraternity of three.

Case 8, E.M., male. The cataract was noticed at birth. Had always been healthy. No spasms or rickets. Mother healthy during pregnancy.

9 years old: He looked healthy. Nystagmus was present. Corneal-diameter normal o.u. Nuclear opacity in the lens, with a less dense zone around this. Good red reflex peripherally, o.u. Was operated in both eyes at the ages of 9 and 10 years, and was then able to read. Died 21 years old.

Diagnosis: Cat. congenita (nuclearis).

Case 9, K.M., male. The cataract was noticed at birth. No spasms or rickets. Was 5 years old when admitted to a clinic. No disease in mother during pregnancy.

5 years old: Looked healthy. Nystagmus present. Corneal-diameter normal. In the pupillary area a greyish, 4 mm. diam. disc-like opacity. Red reflex peripherally. Probably a nuclear cataract o.u. Operated on both eyes. Vision o.d.+11=6/36; o.s.+11=6/36.

25 years old: In later years the vision had diminished. Glaucoma diagnosed o.d.

43 years old: Right eye became blind at the age of 26 years. Then glaucoma developed in the left eye. Vision: o.d. amaurosis; o.s.+10=5/30. Intraocular tension o.s. 40 mm. Hg despite pilocarpine.

FIG. 6.

Family 6. Pedigree demonstrating that the parents of Case 8, E. M., male, and Case 9, K. M., male, both with congenital cataracts, were second cousins.

Diagnosis: Cat. congen. (nuclearis). Glaucoma.

Pedigree information. See Fig. 6.

Investigations through six generations were carried out. The patient's brother, K.M., aged 45 years, his wife, M.M., aged 28 years, and their child, K.M., 2 years old were normal. Their mother, B.M., aged 82 years, was No. 1 in a fraternity of five. All these and their children had normal eyes. The patient's father, P.M., aged 90 years, and No. 5, 6, 7 and 8 in his fraternity, and their children were all normal. No. 1, 3 and 4 of this fraternity died aged 86, 75 and 70 years old respectively, and had good vision. The paternal grandparents (gen. III) died aged 84 and 80 years, and had good vision. Three of their brothers and sisters died at an advanced age and had good eyesight. The maternal grandparents (gen. III), K.G. and E.G., died at the ages of 72 and 62 years and had good vision. In K.G.'s fraternity there were five other members who all died at advanced ages and had good eyesight. The patient's parents were second cousins.

In this family there were two cases of cat. congenita (nuclearis). The patient's parents were second cousins.

Family 7: Cases 10 and 11.

E.S., male, born 1885, and O.S., female, born 1889, in Siljan, Telemark, are No. 3 and 5 in a fraternity of nine.

Case 10, E.S., male. He stated that he always had poor vision. Could read at school. His mother was healthy during pregnancy.

50 years old: He looked well and had always been healthy. No nystagmus. Corneal-diameter was normal o.u. O. dext.: In the central part of the anterior lens capsule there was a whitish opacity of 2.5 mm. diameter. In the posterior cortex there was a central rosette-like opacity of about 5 mm. diameter. Otherwise this lens was clear. O. sin.: The same picture. Fundus normal o.u. Vision: 5/20 o.u. No operation was advised, as ability to read was fair. He was drowned at age of 60 years.

Diagnosis: Cat. congenita (Cort. post. et pol. ant.).

Case 11, O.S. She always had poor vision, but was able to read at school. Vision diminished within the last years. Mother healthy during pregnancy.

50 years old: Enjoyed good health. No spasms or rickets. No nystagmus. Corneal-diameter normal.

O. dext. In the central part of the anterior lens capsule a whitish spot of 2 mm. diameter was seen. In the posterior cortex there was a central rosette-like opacity of 4 mm. diameter. In addition widespread powder-like opacities were seen in the lens. O. sin. similar picture. Fundus normal o.u. Vision=5/20 o.u.

59 years old: Vision diminished, at that time=5/30 o.u. Also wedge-like opacities noted in anterior cortex. Extractio cataractae o.d. After operation, vision=5/6.

Diagnosis: Cat. congen. (cort. post. et pol. ant.).

Pedigree investigation. See Fig. 7.

The pedigree was followed through five generations. The patient's siblings were: No. 1, H.N., female, aged 65 years old. Normal eyes. Her husband died 45 years old, and had good vision. Their five children between the ages of 22-46 years were all normal. No. 2, O.S., male, aged 62 years, was normal. No. 4, B.O., female, and her four children, aged 26-31 years, were normal. No. 6, N.S., male, died 41 years old, but had good vision. No. 7, J.S., male, 52 years old, his wife and their three children, aged 20-23 years, were all normal. No. 8, P.O., female, and her husband died 40 and 60 years old respectively, and had good vision. Their four children, aged 23-30 years had normal eyes. No. 9, H.H., female, aged 46 years old, her husband and their three children, aged 10-20 years, were all normal. The patient's parents N.O., male, and B.O., died aged 77 and 62 years old respectively, and had good vision. The father had three siblings who all died at advanced ages and had good vision. The mother had seven siblings who also died at advanced ages and had good eyesight. The patient's parents were first cousins. The patient's grandparents O.H., A.H., O.O., and E.O., died aged 89, 60, 70 and 80 years old respectively, and had good eyesight. (A.H. and O.O. were siblings.) The parents of the last mentioned (H.H., male, and J.H.) died aged 70 and 94 years old respectively, and had good vision.

The descendants from these were all traced as far as possible, but only the two recorded patients with cataract were found.

In this family two cases of cat. congenita. (cat. cort. post et pol. ant.) among nine siblings were recorded. The patient's parents were first cousins.

FIG. 7.

Family 7. Pedigree demonstrating that the parents of Case 10, E. S., male, and Case 11, O. S., female, both with congenital cataracts, were first cousins.

Family 8: Case 12.

K.K., male, born March 13, 1908, in Drangedal, Telemark, was No. 1 in a fraternity of five.

Case 12, K.K., male. The patient was healthy during childhood. No spasms. No rickets. His eyesight had been poor as far back as he could remember. Difficulty in seeing in the darkness. His mother was healthy during pregnancy.

38 years old: General condition good. No nystagmus. Corneal-diameter normal o.u.

O. dext.: There was an anterior polar opacity of 2 mm. diameter in the lens. In the posterior cortex there was a rosette-like opacity. Otherwise the lens was clear. In the fundus retinitis pigmentosa was seen. O. sin.: Corresponding changes. Vision=5/20 o.u. Visual fields were constricted.

Diagnosis: Cat. congenita (Cort. post. et pol. ant.). Retinitis pigmentosa.

Pedigree information. See Fig. 8.

The pedigree information covered four generations. The patient's siblings were normal. They were aged 27-35 years. His parents O.K., male, aged 64 years, and A.K., who died 65 years of age of tuberculosis, were both examined, and had normal eyes. The patient's father was No. 1 in a fraternity of six. No. 2, I.K., female, was unmarried, and had normal eyes. No. 3, A.M., female, 57 years old, and her three children, aged 24-28 years, were all normal. No. 4, J.K., male, aged

FIG. 8.

Family 8. Pedigree demonstrating that the parents of Case 12, K. K., male, with congenital cataract, were first cousins. K. O. (gen. II), aged 71, had senile cataract.

54 years, had one child, 16 years old. Both had normal eyes. No. 5, A.M., female, aged 51 years, and her two children, 30 and 28 years old, were all normal. No. 6 died 20 years old, and had at that time apparently normal eyes. The patient's parents were first cousins.

The patient's mother had a sister who was still alive, T.L., 65 years old, and she had normal eyes.

His paternal grandfather, K.O., died 71 years old, and had then a senile cataract (from his physician). The grandmother, I.O., died 70 years old, and had good eyesight. His maternal grandfather, H.K., and grandmother, A.K., died aged 72 and 70 years respectively; and had good vision. The patient's paternal and maternal grandfathers were brothers.

In this family, one among five siblings were found to be affected. Diagnosis: Cat. congenita (cort. post. et pol. ant.). Retinitis pigmentosa. The patient's paternal grandfather had cat. senil. His parents were first cousins.

Family 9: Case 13.

A.K., female, born 1911, in Kragero, Telemark, and was No. 3 in a fraternity of three.

Case 13, A.K., female. She had always had bad eyesight. She was deaf and dumb, otherwise healthy. She was well proportioned. Her mother had no disease during pregnancy.

37 years old: No nystagmus. Corneal-diameter normal o.u.

O. dext. There was a greyish opacity of about 3 mm. diameter in the anterior polar region of the lens. In the posterior cortex there was a rosette-like opacity. The nucleus of the lens was opaque. The fundus could not be seen.

O. sin. Corresponding picture.

It was difficult to obtain information about vision. The right eye was operated upon: *Extractio cataractae*. After operation retinitis pigmentosa was found. Vision: with +14 she was able to read big print.

Diagnosis: Cat. congenita (pol. ant. et cort. post.), Retinitis pigmentosa. Surdo-mutitas.

Pedigree information. See Fig. 9.

The pedigree information covered six generations. The patient was No. 3 in a fraternity of three. Her eldest sister, K.P., 50 years old, her husband, H.P., 60 years

FIG. 9.

Family 9. Pedigree demonstrating that the parents of Case 13, A. K., female, with congenital cataract, were first cousins.

old, their daughter, S.H., 24 years old, and her husband, A.H. (28 years old) were all normal, together with their child, O.H., 4 years old. No. 2, T.K., female, died aged 31 years, but had good eyesight. The patient's parents, G.K., male, 70 years old, and S.K., 78 years old, had normal eyes. They were first cousins. The Patient's grandparents (gen. II) all died at advanced ages (64, 81, 84 and 83 years) and were said to have had good eyesight. Two of these, K.K. and A.T., were sisters. The descendants from these two were traced as far as possible and no other cases of cataract or other eye-disease were found.

In this family one sister among three had cat. congenita, (cat. pol. ant. et cort. post.). Retinitis pigmentosa and surdo-mutitas. The patient's parents were first cousins.

Family 10: Case 14.

S.B., female, born April 1, 1928, in Holla, Telemark. She was the only child.

Case 14, S.B., female. During pregnancy the patient's mother had eclampsia. Spontaneous delivery. No infectious disease (e.g., rubella). The mother noticed nystagmus in her child at birth. The child was admitted to a clinic 9 months old.

9 months old: Nystagmus. Total opacity of lens. Discissio cataractae o.u.

6 months old: Disc. cat. o.u.

9 years old: The patient was feeble minded. Not able to speak. Refused use of glasses. The pupillary-area was clear. Fundus appeared normal o.u. Nystagmus. Corneal diameter small ($8\frac{1}{2}$ mm.). Otherwise somatically normal.

Diagnosis: Cat. congen. (total). Imbecillitas.

Pedigree information. See Fig. 10.

The patient was the only child. Her parents, E.B., male, 45 years old, and K.B., 43 years old, were normal. The first one had four siblings, all normal. The second one had five siblings, all normal. There was no indication of relationship between the parents. The patient's paternal grandparents, R.B. and S.B., were both dead. The first one of pneumonia, 53 years old, and the second of apoplexia, 46 years old. Both of them had good eyesight. The patient's maternal grandparents were H.D., 80 years old, alive, with normal eyes, and M.D., who died 83 years old and had good eyesight. The paternal great-grandfather, H.D., 80 years old (gen. I) was still alive and had normal eyes. The other members of this generation died at

FIG. 10.

Family 10. Pedigree concerning Case 14, S. B., female, with congenital cataract. No consanguinity between the parents traced.

advanced ages and were supposed to have had good eyesight. The descendants from these were traced, but no other cases of eye-diseases were found.

In this family there was one case of cat. congenita (totalis) and Imbecillitas diagnosed. No indication of relationship between her parents.

Family 11: Case 15.

A.K., female, born May 2, 1924, in Gjerpen, Telemark, and was No. 2 of two siblings.

Case 15, A.K., female. No indication of spasms or rickets in childhood. The patient's mother was healthy during pregnancy. For many years her eyesight was poor specially in bright day-light.

24 years old: Looked healthy. No nystagmus. Corneal diameter normal. There was a posterior cortical opacity in the lens. Otherwise the lens was clear. Fundus normal. The opacities were alike in both eyes. Vision: o.d. = 5/10; o.s. = 5/10.

Diagnosis: Cat. juvenilis (cat. cort. post.).

Pedigree information. See Fig. 11.

Investigations through four generations were made. The patient's brother, K.K., 37 years old, his wife and their two children (aged 3 and 5 years) were all normal. Her parents, A.K., male, aged 67 years, and T.K., 70 years old, were both normal. Her father had two siblings, G.K., who died 74 years old, and K.K., 67 years old,

FIG. 11.

Family II. Pedigree concerning Case 15, A. K., female, with congenital cataract. No consanguinity between the parents traced.

both of which had good eyesight. Her mother had five siblings. Two of these died at advanced ages, but had good eyesight. Three were still alive, passed 60 years of age, and had normal eyes. Information indicated that the patient's grandparents died at advanced ages, and had good eyesight. The descendants were traced as far as possible, but no other eye-diseases were found. No consanguinity between the patient's parents was discovered.

One case of cat. juvenilis (cat. cort. post.) was diagnosed. No indication of relationship between the patient's parents.

Family 12: Cases 16, 17, 18, 19 and 20.

A.T., female, born April 24, 1903, I.T., female, born January 1, 1905, A.T., born June 30, 1907, S.T., male, born April 28, 1914, in Seljord, Telemark. They were No. 2, 3, 4 and 8 in a fraternity of eleven.

Case 16, A.T., female. The cataract was noticed at birth. Had otherwise been healthy. No spasms or rickets in childhood. Able to read a little. The patient's mother was healthy during pregnancy. She was admitted to a clinic when 6 years old and then operated upon.

45 years old: Well proportioned and healthy looking. Nystagmus present. Corneal-diameter normal o.u. Left eye in divergens.

O. dext.: There was a dense secondary cataract in which there were two holes about 3 mm. in diam. Fundus appeared normal.

O. sin.: The pupil was peripheral, as a small coloboma. Fundus could not be seen. Vision: o.d. + 14 = 5/30; o.s. + 14 C.F. at 4 m. With + 14 D lens she was able to read big print.

Case 17, I.T., female. The cataract noticed at birth. Otherwise she had been healthy. No spasms or rickets. Her mother healthy during pregnancy. Operated on at 3 years old in the left eye and 7 years old in the right eye. Able to read and carry out housework.

43 years old: Nystagmus. Corneal diameter normal o.u. Well-proportioned and healthy-looking.

O. dext.: Anterior chamber deep. Pupil was round and dark. Fundus appeared normal.

O. sin.: Divergent strabismus. Anterior chamber deep. The pupil was peripheral as a small coloboma. There was a secondary cataract. Vision o.d. + 12 = 5/5; o.s. + 12 = 5/20.

Case 18, A.T., female. The cataract noticed at birth. Otherwise of good health. No spasms or rickets. Her mother was healthy during pregnancy. The patient was operated on at 5 years old. Good vision post-operatively.

41 years old: Well-proportioned and looked healthy. Nystagmus present. Corneal-diameter normal o.u.

O. dext.: Ant. chamber deep. Round pupil with clear pupillary area. Fundus appeared normal.

O. sin.: Deep anterior chamber. Round pupil, with faint secondary cataract. Fundus looked normal.

Vision o.d. +12 = 5/6; o.s. +12 = 5/20.

Case 19, S.T., male. The cataract noticed at birth. Otherwise healthy. No spasms or rickets. His mother healthy during pregnancy.

His eyes were operated on at the ages of 5 and 15 years. His eyesight remained poor, but he was able to read big print with a magnifying glass.

FIG. 12.

Family 12. Pedigree concerning Cases 16, A. T., female, 17, J. T., female, 18, A. T., female, 19, S. T., male, all with congenital cataracts.

No consanguinity between the parents traced. S. T. (gen. III), aged 72, had senile cataract.

34 years old: He was well-proportioned and looked healthy. Nystagmus present. Corneal-diameter normal o.u.

O. dext.: Ant. chamber deep. Dense sec. cataract. Fundus was barely seen o.u.

Vision: o.d. +12, count fingers at 3 m.; o.s. +12 count fingers at 2 m. Discissio cataractae was advised.

Diagnosis: Cataractae congenitae.

Pedigree information. See Fig. 12.

The pedigree information covered five generations. The four patients had seven siblings, all normal. The patients were No. 2, 3, 4 and 8. No. 1 died 32 years old of tuberculosis. His wife and their two children (23 and 19 years old) were normal. No. 5, A.T. (twin of case 3) was normal. No. 6, M.S., 39 years old, and her husband and their children (4 years old) were normal. No. 7, K.T., his wife and their two children, aged 1 and 2 years respectively, were normal. No. 9, B.G., 33 years old, her husband, and their four children (twins), aged 6, 4 and 3 years, were normal. No. 10, S.T., 31 years old, his wife, and their three children aged 1 to 5 years, were normal. No. 11, H.T., 25 years old, was normal.

The patient's father, S.T., 72 years old, had cat. senil. incip. with wedge-shaped opacities in cortex.

Vision: o.d. = 5/6; o.s. = 5/10. Read easily. Seven of his siblings died between birth and 3 years. (Diphtheria, scarlatina, cholera, etc.) The patient's mother,

G.T., 68 years old, had normal eyes as also her three siblings, aged 66, 64 and 62 years. These three had thirteen children, all normal. The patient's paternal grandparents had good eyesight. The grandfather died 47 years old of pneumonia. The grandmother 38 years old of tuberculosis. The paternal great-grandparents died at the ages of 59, 91, 85 and 64 years, and had good vision. The maternal grandparents also had good eyesight. The grandfather died 64 years old (K.G.), and the grandmother 79 years old. The individuals in gen. I died all at advanced ages, 74, 64, 68 and 80 years old, and had good eyesight.

Investigations indicated no relationship between the patient's parents.

In this family four of eleven siblings had cat. congenita. Their father had cat. senil. incip. No consanguinity between the patient's parents was indicated.

Family 13: Cases 20 and 21.

T.H., female, born 1938, and O.H., male, born 1941, in Hjarstad, Telemark, were No. 2 and 4 in a fraternity of four.

Case 20, T.H., female. The cataract was noticed before the age of one year. No spasms or rickets in childhood. The patient's mother was healthy during pregnancy. The patient's eyes were operated on at the ages of $4\frac{1}{2}$ and $5\frac{1}{2}$ years. Afterwards she had good eyesight.

9 years old: Well-proportioned and looked healthy. Corneal-diameter normal o.u. In both pupillary areas there was a fine membrane of sec. cataract. Discissio cat. o.u. was performed. Vision o.d.=5/10; o.s.=5/10.

No indication of the type of cataract was obtainable.

Diagnosis: Cat. congenita.

Case 21, O.H., male. The cataract was noticed before the age of 1 year. He had always been healthy. No spasms. No indication of rickets. His mother was healthy during pregnancy.

4 years old: He looked healthy. Well-proportioned. Nystagmus present. The corneal-diameter normal o.u.

O. dext.: In cycloplegia a central, nuclear opacity was seen in the lens, measuring about 5 mm. in diameter. No riders. Red reflex in the periphery.

O. sin.: The same picture. Vision: C.F. at 2 m. o.u.

Diagnosis: Cat. congenita (nuclear).

Pedigree information. See Fig. 13.

The pedigree information covered three generations. The patients had two normal siblings, No. 1 and 3, 12 and 6 years old respectively. Their father, O.H., 40 years old, and his six siblings were all normal. Their mother, A.H., 35 years

FIG. 13.

Family 13. Pedigree concerning Cases 20, T. H., female, and 21, G. H., male, both with congenital cataracts. No consanguinity between the parents traced. A sister of T. H.'s father (gen. I) had senile cataract. (Aged 65 years).

old, and her seven siblings were also normal. The paternal grandfather, G.H., 81 years old, was normal, and his wife, T.H., who died 60 years old, had good eyesight. A sister of the latter's father had cat. senilis, was operated on and regained good eyesight. The maternal grandparents, S.H., male, aged 64 years, and A.H., 64 years old, were still alive, and had normal eyes.

No indication of consanguinity between the patient's parents. The descendants from first generation were traced, but no evidence of cataract was noted.

In this family there are two cases of congenital cataract (nuclear) in a fraternity of four. No consanguinity between the patient's parents was indicated. One case of senile cataract far back in the family.

Family 14: Case 22.

A.S., female, born August 4, 1911, in Solum, Telemark. The patient was No. 7 in a fraternity of nine.

Case 22, A.S., female. The patient's mother informed us that her daughter's eyesight was poor from childhood, and that she left school on that account at the age

FIG. 14.

Family 14. Pedigree concerning Case 22, A.S., female, with juvenile cataract. No indication of relationship between her parents.

of 11 years. Spontaneously the mother also had asked the patient to state that she had spasms as a child and later also had rickets. The mother was healthy during pregnancy.

25 years old: She looked healthy and was well-proportioned. No nystagmus. Corneal diameter normal o.u.

O. dext.: In cycloplegia a typical perinuclear opacity of 5 mm. diameter, with riders, was seen. Red reflex around the opacity. *O. sin.:* same picture. Vision: Counting fingers at 3 m. o.u. Discissio et evacuatio cataractae o.u. was performed.

Vision: o.d. + 12 = 5/10; o.s. + 12 = 5/10.

Diagnosis: Cat. juvenilis (zonularis).

Pedigree information. See Fig. 14.

The pedigree information covered four generations. Her husband, P.S., was normal, as were their two children, aged 16 and 9 years. These had always been healthy, and were well-proportioned. The patient was No. 7 in a family of nine. No. 1 and 2 of these, aged 47 and 44 years respectively were normal. No. 3 died 8 days old. No. 4, 5 and 6 were 40, 38 and 36 years old, and were normal. So also were No. 8 and 9, 34 and 29 years old respectively. The patient's parents, A.P., male, aged 70 years, and O.P., 69 years old, were normal. The patient's grandparents died at advanced ages, and had good eyesight. There was no indication of relationship between the patient's parents. In addition to this information, the ascendants and descendants were followed as far as possible, and no other case of eye-disease was discovered.

In this family one patient with cat. juvenilis (zonularis) in a fraternity of nine was found. No consanguinity between her parents was indicated.

Family 15: Case 23.

I.K., male, born November 24, 1919, in Porsgrunn, Telemark, was No. 4 in a fraternity of five.

Case 23, I.K., male. The patient's mother informed us that he had spasms as a nursing and later rickets. Was healthy later in childhood. Attended the elementary school, but had poor vision. His mother was healthy during pregnancy.

25 years old: Well-proportioned. Looked healthy. His teeth, however, demonstrated severe enamel defects. No nystagmus. Corneal diameter normal o.u.

O. dext.: Cycloplegia. A central perinuclear opacity of 5 mm. diameter in the lens was seen. Within this a lot of powdered opacities. Typical riders around the opacity. Fundus was normal.

O. sin.: Corresponding opacities. Vision: o.d.=5/15; o.s.=5/10. Able to read.

Diagnosis: Cataractae juvenilis (zonularis).

Pedigree information. See Fig. 15.

This pedigree information was followed through three generations. The patient's four siblings: No. 1, I.K., female, aged 39 years, No. 2, A.K., male, aged 35 years, No. 3, N.K., female, aged 32 years, and No. 5, B.K., female, aged 22 years, were

FIG. 15.

Family 15. Pedigree concerning Case 23, J. K., male, with juvenile cataract. No indication of relationship between his parents. J. K., gen. I, aged 91, had senile cataract.

all normal. They were all unmarried. The patient's parents (K.K., male, aged 64 years, and N.K., 64 years old) were normal. Of the patient's grandparents, only I.K., 91 years old, was still alive. He had cat. senil. incip. The other three died at advanced ages, 84, 82 and 87 years. They all had good eyesight. Their descendants were traced as far as possible and no other eye-diseases were discovered. No consanguinity between the patient's parents was found.

In this family there was one case of cat. juvenilis (zonularis), and one case of cat. senilis (in the paternal grandfather). No consanguinity between the patient's parents. The patient was said to have had spasms and rickets in childhood and demonstrated enamel defects of his teeth.

Family 16: Cases 24 and 25.

H.H., female, born April 11, 1927, and W.H., male, born 1925, in Brevik, Telemark, in a fraternity of two.

Case 24, H.H., female. The patient had always been of good health. No spasms. No rickets. She had good eyesight until 18 years old, when she noticed blurring before her eyes, and became tired on reading. Her mother was healthy during pregnancy.

20 years old: Looked healthy and was well-proportioned. No nystagmus. Corneal-diameter normal o.u.

O. dext.: In the central part of the lens, there was a perinuclear opacity corresponding to the foetal nucleus. (See Fig. 16a.) Around this was a layer of fine powder-like opacities. In the periphery there were more riders. These riders were supposed to be located around the infantile nucleus.

Fundus appear normal. *O. sin.*: Corresponding opacities in the lens. Vision = 5/6 o.u.

Diagnosis: Cat. juvenilis (cat. zonular).

Case 25, W.H., male. He had always been healthy. No spasms or rickets. Had good vision until recently, when he became tired on reading and noticed blurred vision at distance.

FIG. 16a.

Case 24, H. H., female, aged 20 years, with a particular type of zonular cataract. Her brother, Case 25, W. H., aged 16 years, demonstrates a similar cataract.

FIG. 16.

Family 16. Pedigree concerning Cases 24, H. H., female, and 25, W. H., male, with juvenile cataracts. No indications of relationship between their parents.

16 years old: He looked healthy. He was well-proportioned and had good teeth. No nystagmus. The corneal-diameter was normal o.u.

O. dext.: The clinical picture was here exactly the same as in his sister. There was a central perinuclear opacity with small powder-like opacities around this. Peripherally there were typical riders.

O. sin.: Similar opacities. Vision = 5/6 o.u.

Diagnosis: Cat. juvenilis (zonularis).

Pedigree information. See Fig. 16.

The pedigree information covered three generations. The patient's parents, W.H., male, aged 52 years, and A.H., 46 years old, were both normal. There was no indication of relationship between these, as the first mentioned came from Sweden and the other from Norway. The patient's father had two elder brothers, who both died 62 and 57 years old respectively, one of tuberculosis and the other of heart disease. They had good eyesight. The patient's mother was No. 5 in a fraternity of seven. The other six were all examined. They were 60, 56, 53, 51, 38 and 36 years old and had normal eyes. Their children were also examined and demonstrated normal eyes. The patient's paternal grandparents died aged 67 and 84 years old. They had good eyesight. The maternal grandfather died 71 years old and had good vision. The maternal grandmother was still alive, 85 years old, and had normal eyes. The descendants from these were examined and no other cases of eye-diseases were found.

In this family both children in the family had exactly the same type of cataract, cat. juvenilis (zonularis). Consanguinity between the patient's parents was excluded. No indication of spasms or rickets in childhood.

Family 17: Cases 26 and 27.

K.J., male, born 1901, and A.M., female, born 1902, in Bamle, Telemark, were No. 3 and 5 in a fraternity of five.

Case 26, K.J., male. He had always been healthy. No spasms. No rickets. He had always had poor eyesight. Able to read moderately well at school. From the age of 25 years he was convinced that something was wrong with his eyes. His mother was healthy during pregnancy.

36 years old: Looked healthy and was well-proportioned. No nystagmus. Corneal diameter normal o.u.

FIG. 17.

Family 17. Pedigree concerning the Cases 26, K. J., male, and 27, A. M., female, with juvenile cataracts. No indications of relationships between their parents.

O. dext.: Cycloplegia. A central perinuclear opacity of 5 mm. diameter. Some riders present. Red reflex around the opacity. Fundus appeared normal.

O. sin.: Corresponding opacity. Vision: o.d. +3 = 5/20; o.s. +3 = 5/15.

42 years old: The opacity more dense. Vision: +3 = 5/50 o.u. / Extractio cataractae performed on both eyes. Vision: o.d. = 5/15; o.s. = 5/10.

Diagnosis: Cat. juvenilis (zonularis).

Case 27, A.M., female. She had always been healthy. No spasms. No rickets. Poor vision from the age of 17 years, specially in bright light. Her mother was healthy during pregnancy.

38 years old: Looked healthy and was well-proportioned. No nystagmus. Corneal diameter normal o.u.

O.U.: The lens opacities showed the same picture as in the above brother. Fundus normal. Vision: -1 = 5/15 o.u. Extractio cataractae o.u. was performed. Vision: o.d. +11 = 5/10; o.s. +11 = 5/10.

Diagnosis: Cat. juvenil. (zonularis).

Pedigree information. See Fig. 17.

The pedigree data covered four generations. The patients were No. 3 and 5 in a fraternity of five. No. 1, I.J., male, aged 57 years, No. 2, H.J., male, aged 50 years, and his wife, 48 years old, had normal eyes. Case 26, K.F., had a wife 45 years old and two children, D.J., male, aged 25 years, and L.J., male, aged 14 years, all with normal eyes. No. 4, H.J., female, aged 42 years, her husband, 36 years old, and their three children, aged 13, 17 and 19 years, were all normal. No. 5 was the patient, Case 27, who was married, but had no children. The patient's parents died 80 and 77 years old and had good eyesight. Information about the patient's grandparents indicated no eye-disease. They died at advanced ages, and probably had good eyesight.

In this family two cases of cat. juvenilis (zonularis) were diagnosed in a fraternity of five. No indication of relationship between the patient's parents. No indication of spasms or rickets during the patient's childhood.

DISCUSSION AND CONCLUSION

A. Heredity

In the introduction to this paper the confusion about the mode of heredity of congenital and juvenile cataracts was emphasized, the dominant mode being considered the most common. The recessive mode, however, was not considered to be worth investigating.

The present material is numerically too small to prove recessivity statistically. There were, however, many indications of its existence: pronounced familial incidence, frequent relationship between the parents of the affected individuals, and the fact that the cataracts in most of the cases were congenital.

In this material the familial incidences were prominent. In 8 of 17 examined sibships, 2 or more individuals were affected. This cannot be accepted as proof of inheritance — exogenous factors must also be taken into consideration — but it is very suggestive.

Among the exogenous (environmental) factors giving rise to congenital cataracts, special attention during recent years has been paid to the virus diseases (rubella). A considerable literature already exists on this subject, and a rather complete

list is presented by Bardram and Brändstrup (1947). There is, of course, no doubt that rubella in the mother during pregnancy may cause a congenital cataract, but in the cases presented this etiological factor can be excluded.

From the available literature in this field, it appears that the whole clinical picture caused by rubella is different from the clinical picture of the above cases. The cataracts caused by rubella usually are accompanied by considerable somatic defects and malformations, and hence must be considered a more serious disease. It is also evident from the present material that rubella cannot be made responsible for many congenital cataracts within the same sibship.

In the Table I below, the frequency of relationship between the parents of the affected individuals is demonstrated. In 5 of the 17 families the parents were first cousins. In another 4 they were related further back. Consanguinity was found, therefore, in 9 of the 17 families. Taking into consideration the remarks made in the introduction about the rare occurrence of consanguinity, these facts are of great interest, and may indicate recessive heredity.

TABLE I

Number of children, number of cataracts, application of Weinberg's methods, and intermarriages in 17 families with congenital and juvenile cataracts.

Fam. No.	No. of Children (S)	No. of Catar. (T)	Weinberg's Meth.		Intermarriages in Parents
			T (S-1)	T (T-1)	
1	3	2	4	2	First cousins
2	2	1	1	0	Third cousins
3	4	2	6	2	First cousins
4	2	1	1	0	Fifth cousins
5	1	1	0	0	Fourth cousins
6	3	2	4	2	Second cousins
7	9	2	16	2	First cousins
8	5	1	4	0	First cousins
9	3	1	2	0	First cousins
10	1	1	0	0	No
11	2	1	1	0	No
12	11	4	40	12	No
13	4	2	6	2	No
14	8	1	7	0	No
15	5	1	4	0	No
16	2	2	2	2	No
17	5	2	8	2	No
Total :	70	27	106	26	

Unfortunately, the present cases are too few for statistical proof as to the frequency of first-cousin marriages. (Only first-cousin marriages are used for this purpose, as only information on the normal frequency of these is available.) In this survey the frequency is 29.4 per cent. Normally it is less than 1 per cent. among the West European population (Dahlberg, Dunn).

Special interest should be paid to the pedigrees belonging to the families Nos. 2 and 3 (See Figs. 2 and 3). The parents of the patient, Case 3, were third cousins, and the parents of the Cases 4 and 5 (family No. 3) were first cousins. There were also relationships between the two families, as the father of Case 3 was second cousin to both the father and the mother of patients Nos. 4 and 5. In both of these families the cataracts appear to have been of the same type. They were all congenital and were operated on at the same age (3, 2, 3 and 2 years, respectively). Hence, they must be considered genotypical.

Among 70 siblings 27 are affected. This represents 38 per cent. of the total number, and indicates recessivity, but the number affected was higher than expected from the Mendelian rules for mono-recessive heredity: Normal: affected = 3:1. The material here is, however, a selected one, and a so-called recessive-excess is to be expected.

If Weinberg's sibling-method is applied to this material (see Table I), the following results are obtained: The percentage of affected children then is $100 \frac{t}{t(s-1)}$ S. is the number of

siblings in a family, t is the number of affected children. $t(s-1)$ is the siblings of affected children.* The result is then 24.5 per cent., which is in accordance with expectations when mono-recessive inheritance is involved.

As mentioned, 20 of the 27 cases were recorded as congenital. Possibly some of the cases recorded as juvenile were also congenital.

The cataracts described were not sex-linked, as 14 of the affected individuals were males and 13 females. They must therefore be considered autosomal.

The material shows no evidence supporting the hypothesis that the genes are incompletely dominant.

As the parents of the affected individuals, with only one exception, were normal, and the cataracts for the greater part congenital, the familial incidence high, and the consanguinity among the parents of affected individuals frequent, the mode of heredity was probably autosomal recessive in nature.

* $t(t-1)$ is the affected siblings of affected children.

In 7 of the families (Nos. 1, 4, 5, 8, 12, 13 and 15) *cat. senilis*, as well as *cat. congenita* and *juvenilis*, was demonstrated. In one of these families, the father of 4 patients with congenital cataracts had senile cataract. Otherwise the senile cataracts were found in other generations or within side-branches. The patients with senile cataracts were all of considerable ages (76, 64, 83, 70, 71, 72, 65 and 91 years).

In the above-mentioned family (No. 12) the father of the 4 patients had an incipient senile cataract with wedge-shaped opacities in the anterior cortical layers of the lens. Visual acuity (72 years old) was 5/6 o.d.; 5/10 o.s. This cataract, accordingly, was entirely different from the congenital, total cataract in his 4 children.

The phenomenon of anticipation, in which cataracts appear at a younger age in every new generation, might explain the appearance of senile, juvenile or congenital cataracts within the same family. But this conception receives no support from the present investigations. The senile cataracts, Case No. 15 excepted, were all in families in which the patients had congenital cataracts. In cases of "Anticipation," intermediate types would be expected. The appearance of senile cataracts within these families therefore suggests a normal occurrence within the population.

In the material dealt with in the present paper there were 4 families (Nos. 14, 15, 16 and 17), representing 6 cases (Nos. 22, 23, 24, 25, 26 and 27), with zonular cataracts. It is mainly in this type that spasmophilia and rickets are supposed to be of etiological interest. This question, however, is not as yet solved. Franceschetti mentions that dominant inheritance is preponderant in zonular cataracts, but that recessive types (Baker, 1892, and Starbeck, 1877) also have been recorded. This writer also mentions spasmophilia and rickets as etiological factors.

In "Kurzes Handbuch der Ophthalmologie," Jess says (1930): "Nach allem ergibt sich dass eine einheitliche Schichtstargenese nicht angenommen werden kann."

In the present material, 2 patients (Nos. 22 and 23) among 6 suffered from spasmophilia and rickets in childhood. One of these (No. 23) demonstrated also considerable defects in the enamel of his teeth. In the other 4 patients with zonular cataracts (families Nos. 16 and 17) there was familial incidence, but neither the case histories nor the objective findings indicated spasmophilia or rickets.

The clinical picture of the 2 patients (Nos. 24 and 25 in family 16) is of great interest, as the zonular cataracts demonstrated peculiar arrangement of the lens opacities (see Fig. 16a, p. 26). The perinuclear central opacity probably is located in the foetal

nucleus, which develops between the 3rd and 8th months of foetal life. Peripherally in the lens some riders were seen. These riders are supposed to be located in the area of the infantile nucleus, which develops between the last weeks of foetal life and puberty. These findings may give the impression that some agents have been active at different periods. It does not disprove, however, that these cataracts may be due to genes.

In the present cases of zonular cataracts dominant mode of heredity is not indicated. Recessivity may be present, but cannot be proven. Neither can the effect of spasmophilia or rickets be assessed. Even if the aetiology of zonular cataracts is more obscure than that of other congenital or juvenile cataracts recorded in this paper, they are—with reservation—listed together in a common group.

As the patients were treated surgically in different clinics, and descriptions of the various types were often lacking, the differential diagnosis was rather difficult to make. With these reservations in mind, it would seem that the cataracts were of the same type within the same family. Thus, the type was considered to be similar (cat. subtotalis) in patients Nos. 1 and 2 in family No. 1. The cataracts were congenital and the two patients were both operated on at the same age (3 years). In the families Nos. 2 and 3, which were related, the cataracts also were congenital, and the patients were operated on at the same ages (2, 3 and 2 years). The diagnoses made were: cat. subtotalis, cat. membranacea and cat. totalis. In family No. 6 the diagnoses in Cases Nos. 8 and 9 were cat. nuclearis. Both were congenital and were operated on at the age of 9 and 5 years, respectively. In family No. 7 (Cases No. 10 and 11) the diagnoses were cat. corticalis and polaris ant., and the cataracts were of the same type. In family No. 12 (Cases Nos. 16, 17, 18 and 19) no diagnosis was made as to the special type, but they were all considered to be the same, as they were all congenital, and were operated on at the same ages (6, 3, 5 and 5 years, respectively).

In family No. 13 (Cases Nos. 20 and 21) the diagnosis was cat. nuclearis. Both of them had congenital cataracts. One of the patients was operated on when 4 years old, the other was ready for operation at a corresponding age. In family No. 16 (Cases Nos. 24 and 25) the zonular cataract was precisely of the same type in both brother and sister. In family No. 17 (Cases Nos. 26 and 27) there was a zonular cataract of the same type in both sister and brother. These cataracts were diagnosed and operated on at the same age. The type was, however, quite different from the one in the previous cases. Accordingly, the present

cases show that the types of cataracts vary from family to family, but within the same family the congenital or juvenile cataracts are much alike. These observations support the above-mentioned hypothesis that these cataracts are probably due to genes, and that there is no genetical connection established between these and the sporadic senile cataracts occurring in these families.

B. Occurrence of other diseases or symptoms in cataract patients

The occurrence of spasmophilia, rickets and enamel defects of the teeth has already been mentioned.

In one of the patients (Case No. 14) glaucoma was diagnosed. In two of them (Cases Nos. 12 and 13) the cat. corticalis posterior et polaris anterior was combined with retinitis pigmentosa. Case No. 13 also demonstrated surdo-mutitas.

As to the connection between the cataract and retinitis pigmentosa, two possibilities exist :

1. The cataract is correlated to retinitis pigmentosa.
2. The cataract is secondary to retinitis pigmentosa.

It is generally accepted that retinitis pigmentosa is as a rule a recessive condition. Kjerrumgard (1948) has found that 9 per cent. of all patients with retinitis pigmentosa demonstrated cat. pol. post., and that 5 per cent. of them had other types of cataract. He also demonstrated that 15 per cent. of the patients with retinitis pigmentosa had surdo-mutitas.

In our two cases of retinitis pigmentosa and cat. pol. post. there was also a cat. pol. ant., a type usually considered to be congenital. It is therefore reasonable to suggest that cat. pol. post., cat. pol. ant., retinitis pigmentosa and surdo-mutitas are correlated and that they are due to recessive genes.

Nystagmus was noted in 12 of the 27 patients and occurred only when combined with congenital cataracts.

Microphthalmos was found in 2 patients (Cases Nos. 6 and 14). Minor variations in the corneal diameters may have been more frequent, but exact measurements were not undertaken.

C. The prognosis

The results of the cataract operations were good, except in the following patients: Case No. 21, who was not operated upon; Case No. 14, who was feeble-minded; Case No. 9, with glaucoma; Case No. 19, with cataracta secundaria; Cases Nos. 12 and 13, with retinitis pigmentosa. Most of the operated patients were able to read and do full-time work in their different occupations.

In these conditions, therefore, the only advice to be given from

the eugenic point of view is to warn members of the families against inter-marriages. But as already noted, dominant types of congenital cataracts are likely to exist, and the mode of heredity in each case must be thoroughly investigated.

D. Conclusion

In the introduction it is shown how uncertain is the etiology and pathogenesis of congenital and juvenile cataracts. The dominant mode of inheritance is preponderant, and the recessive type of cataract is relatively rare.

Even if the material dealt with in this paper is too small to give proof by statistics, a series of cases of congenital and juvenile cataracts is cited which seems to indicate recessive inheritance.

Summary

The author of this paper has examined 17 families in which congenital or juvenile cataract occurred. Special attention was paid to their aetiology and mode of heredity.

In the introduction a review of the previous results from literature in this field was given. This showed that the dominant mode of inheritance is supposed to be the most frequent, with relatively rare examples of recessive inheritance. In recent years much attention has been paid to the virus diseases as causes of these cataracts, and inheritance has receded more into the background.

Exogenous factors, as spasmophilia and rickets, were suggested causes of zonular cataracts, but the rôle of heredity was also emphasized in this type.

Following this introduction, a review is given of the present material and the case reports with pedigrees.

In all, 17 families with 27 cases of congenital or juvenile cataracts have been examined. From these, 20 were recorded as congenital and 7 as juvenile. In each of 8 of these families, 2 or more individuals were affected. Among the 27 affected, 14 were males and 13 females.

In 9 of the 17 families, relationship between the parents of the affected individuals existed, and in 5 of them the parents were first cousins. In the other 4 families the relationship was less close.

In a following chapter, "Discussion and Conclusion," the results of these investigations were stated. Among the introductory remarks it was pointed out that the present material is too small to prove statistically the probable mode of inheritance in the cases investigated in this survey. As a eugenic measure,

the members of these families must be warned against inter-marriage, but, as dominant types also occur, it is emphasized that the mode of inheritance must be studied in each case.

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THE MOTOR IMPULSE ELICITED BY THE RETINAL STIMULUS AND THE BINOCULAR OPTICAL REFLEXES*

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IN order to study the problem of strabismus, it is necessary to investigate the disturbances of the normal binocular reflexes which occur in such patients. To do this, however, an exact knowledge of the essential features of these binocular reflexes in normal persons is required.

We started from the fundamental supposition that stimulation of a certain part of the retina not only produces a sensory effect, but is also active in a motor sense. In our opinion stimulation of the retina gives rise to a sensory and a motor impulse. There is evidence that in general these motor impulses may produce the following four physiologic phenomena:

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