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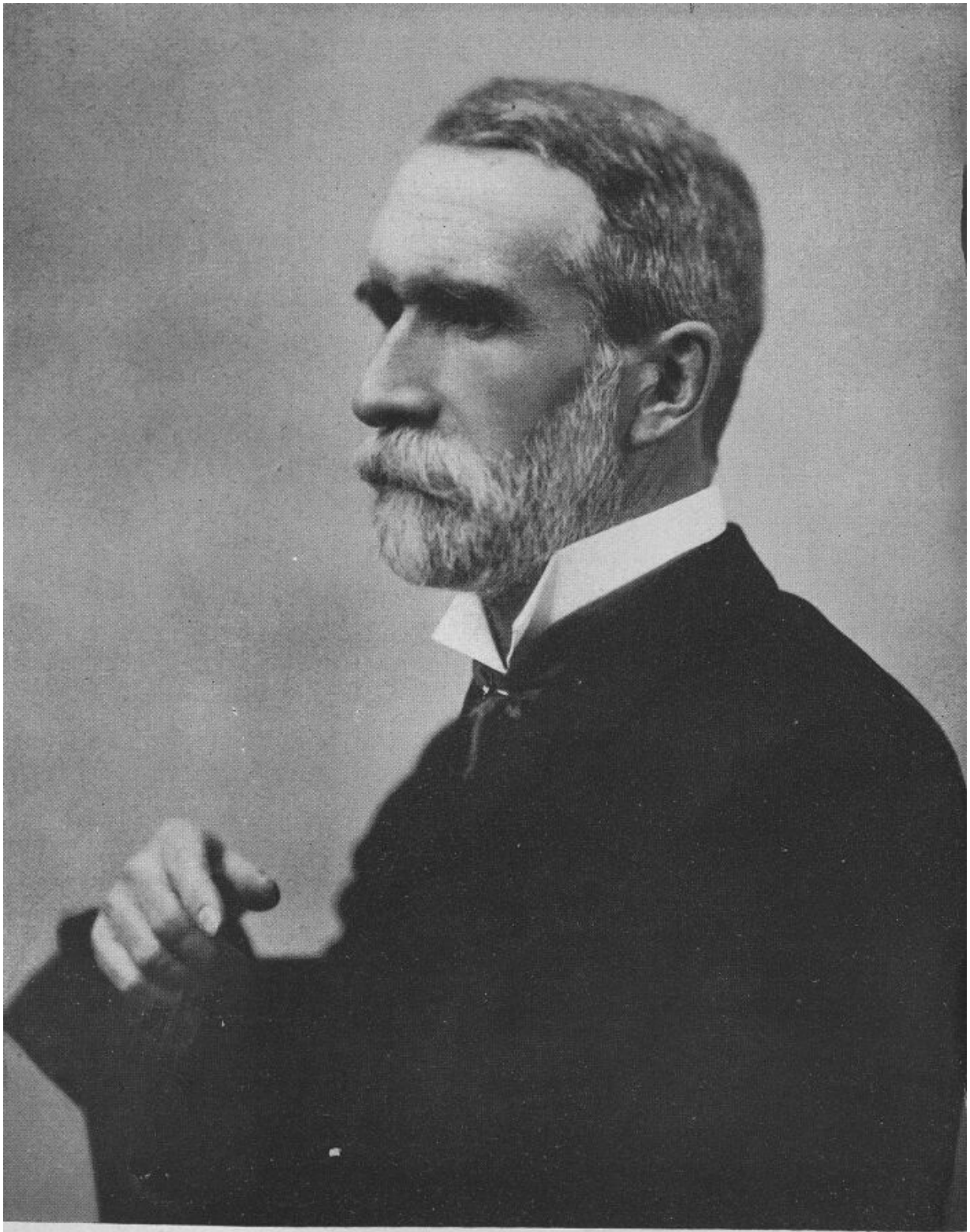

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A NOTE ON THE PROGRESS OF SOME CASES OF RETINITIS PIGMENTOSA SINE PIGMENTO AND OF RETINITIS PUNCTATA ALBESCENS.

By the late E. NETTLESHIP, F.R.C.S., F.R.S.

RETINITIS pigmentosa without pigmentation visible in ophthalmoscopic examination, and to be diagnosed therefore only by the condition of the disc and retinal vessels and the sometimes equivocal changes due to alterations of the pigment epithelium, is not common, and, even when seen, there appears to be some doubt as to its explanation. In continuation of what I wrote on the subject in a previous paper (these Reports, vol. xvii, p. 365 *et seq.*) I take the present opportunity of noting the two examples given below (Cases 1 and 2), each of which has been followed for a long period. One of these patients, whose fundi showed no pigmentation in childhood although he even then suffered severely from night-blindness, afterwards developed abundance of characteristic pigment; the other did not begin to recognise any difficulty in dull light until he was well past 20 and had qualified as a medical man. He was seen by Mr. Fisher at the age of 32 and at intervals until 45, and only one or two visible pigment deposits have developed, although the retinal atrophy has increased and sight become much worse.

In typical retinitis punctata albescens I have had the opportunity of examining the fundi in three cases over

periods varying from 8 to 27 years. In Case 3, watched for 27 years (to July, 1913), the changes appear to me to be exactly as they were at my first examination, but in the other two instances, in a brother and a sister (Cases 4 and 5), the appearances have altered. The dead white retinal dots are now less numerous in the sister (Case 5). In the brother, who is older and has been under observation for 26 years (to September, 1913), they have disappeared entirely between the ages of 39 and 49, and characteristic pigment collections have now begun to appear in the retina. Moreover, in this brother and sister the usual appearances at the optic discs are obvious, and in the sister there is now marked shrinking of the retinal vessels.

CASE 1.—When this patient was a boy, aged 7, I examined him in 1885 at St. Thomas's Hospital. He was even then very night-blind and the retinal vessels very small but no pigment could be seen at the fundi; he had myopia estimated by ophthalmoscope at 6 D.

In December, 1910, when he was about 32 and living in Dublin, Dr. Werner most kindly took the trouble to examine his eyes on my behalf and found "typical retinitis pigmentosa, the pigment reaching to within three disc diameters of the optic pupilla. The optic disc has the usual dirty yellowish-white colour; vessels, especially the arteries, thread-like"; the field extremely contracted, night-blindness very marked, V. with −5·5 D. 6/24 partly with the L., only 6/60 with the best attainable correction in the R. (some astigmatism with 0·5 D of myopia); nystagmus in both.

CASE 2.—Mr. B. began to notice that he saw badly in the dusk for some time before he first sought Mr. Fisher's advice, which was in 1898 when he was 32; the symptom had then been noticed for some years and the patient suspected that it had got rather worse latterly. Mr. Fisher found he had slight H. As. and that with correction V. was full; Fs. full both in bright and reduced illumination; the O.Ds. slightly pale and blurred and retinal arteries a very little reduced; no pigmentary changes of any kind seen anywhere and no retinitis punctata albescens.

In September, 1900, Mr. Fisher found V. a shade less acute; the O.Ds. about as before; retinal arteries and veins unmistakably reduced, all these changes being rather more marked in R. than L.; also in central part of R. fundus a whitish mottled and streaked appearance, and in L. on a smaller area half a dozen or more rather large greyish white spots; some irregularity or speckling of epithelium at periphery, but neither there nor elsewhere either white dots or pigment formations of any kind. Measurement of L. field on perimeter showed in dim light a pericentral scotoma in upper, temporal, and lower parts, reaching to within about 5° of the fixing point; but with full light on the 10 mm. test object the F. appeared n. and full in all parts. He gave a number of anecdotes showing that in a dim light the amblyopic annulus enclosing the minute normal central area made it very difficult for him to see small objects until he got them exactly into the centre of the field, when they suddenly became quite plain; and he said further that in a good light he did not experience this difficulty.

In October, 1902, I saw the patient with Mr. Fisher and we found but little alteration, a small round spot of pigment at the temporal part of L. fundus being the only obvious fresh feature, and it might possibly have been there before. He scarcely admitted any increase of his defect, but we felt sure he was somewhat worse.

In January, 1903, Mr. Fisher found V. of R. down to 5/12 and of L. 5/9, retinal arteries smaller than at the examination in September, 1900, and the pigment epithelium more stippled.

October, 1903, Mr. Fisher found him distinctly worse, V. R. only 5/18, L. 5/12: retinal vessels smaller than ever and choroidal vessels decidedly sclerosed.

On June 14, 1912, Mr. Fisher and I again saw him together. He was then 45. Sight had steadily fallen off and he could barely read ordinary print and that only with fatigue and distress; we avoided worrying him by testing distant vision. Fs. very defective with broad scotoma of zonular type, leaving a small central area of relatively good sight and also a zone of peripheral vision; he describes these features of his sight quite graphically and quotes instances. Ophthalmoscopic examination showed the discs pale and hazy and the retinal arteries very

small, and over a large area around the disc and reaching far towards the y.s. in each eye much stippling or irregularity of the hexagonal epithelium, with exposure and thickening of choroidal vessels; no general pigmentation, but in the L. eye to the nasal side between the O.D. and equator a single well-marked patch of bone-corpuscle-shaped pigment and close to it two or three minute but sharply defined pigment dots or grains, and we also saw but could not fix another spot of pigment in the temporal part of the fundus. No spots of pigment were seen in any part of R. fundus. *Thus during about* 20 *years of recognised night-blindness*, setting in comparatively late in life and characterised by steadily falling visual acuity, increasing damage to the fields and visibly progressive retinal atrophy, *only a minimal quantity of pigment has reached the inner layers of the retina.*

Mr. B. himself has no defects of terminal circulation, but his sister is subject to chilblains and his mother suffers badly from "dead fingers." His father became night-blind in his later years and got slowly worse; he consulted Sir Jonathan (then Mr.) Hutchinson, who I believe found ordinary retinitis pigmentosa; he married at about 28 and died of typhoid fever at 57; he had a sister with hare-lip but no other defects. Mr. B.'s mother, still living, aged about 78, was 27 when she married; she and her husband were not consanguineous, and she has never heard of any marriages of relations on either side. Mr. B. is one of six children as follows (ages as in 1913): 1, ♀ 51; 2, ♀ 50; 3, ♂ 48; 4, ♂ died aged 7 of typhoid fever; 5, ♂, the patient, 45; 6, ♂ 43. Mr. B. is the only one with any defect of sight (other than refractive errors, from which they all suffer), and neither he nor any of his siblings suffer from deafness or any affection of the nervous system. There is no known history pointing to retinitis pigmentosa in any generation before the patient's father. I believe Mr. B. has not had typhoid fever.

Case 3.—This lady is shown as IV, 1, in Fig. 144 of vol. xvii of these Reports, p. 385; she was then (1907) aged 39. I had seen her first in October, 1886, when she was 19, and recorded the case in O.T., vol. vii, p. 302. The account of the ophthalmoscopic appearances and symptoms is practically identical in both the above records; there had been no change. I examined her eyes under a mydriatic again in July of this

year, 1913, and found no alteration from the previous inspections. I thought the R. disc a trifle paler than the L., but could not say that the arteries were smaller in the R. as I thought they were in 1907, or that they were smaller than they should be or showed more white lining than is consistent with normality. She is now 46, and owns and personally manages a successful high-grade school for girls that formerly belonged to an aunt, now dead. The night-blindness appears to be no worse and no better than it was 27 years ago. She is and always has been very thin and frail looking and of anxious temperament, but says she is now stronger and better than some years ago, and has passed the age of greatest liability to phthisis, from which her younger night-blind sister died many years ago. But she has not the aspect of a person likely to live to a great age.

The next two cases are a brother (Case 4) and his younger sister (Case 5).

I first saw the brother (Case 4) in November, 1887, when he was 21, and recorded his case in O.T. vol. viii, p. 163 (1888), with a coloured illustration of the ophthalmoscopic appearances. I again examined him, under mydriatic, in 1905, and published additional particulars in these Reports, vol. xvii, p. 387 (Case 146); the fundus still showed the same white dots as at the first examination 18 years before; the retinal vessels were still of full size, and the slight haze of discs noticed on the first occasion had not become more marked. His symptoms had not become worse.

I saw him again on September 2, 1913, aged 47, and examined the eyes with great care; no mydriatic was used, but the pupils were not small, except when the y.s. was under inspection, and all details were well seen. The appearances had altered very much, so much that, at first sight, I thought my examination must be at fault, but this was not so. I could find *none of the characteristic white dots in any part of either fundus;* if any were present they must have been scanty and very ill-marked. Towards the equator in both eyes the hexagonal epithelium had a granular appearance as if disturbed. In parts the choroidal vessels were much exposed by absence of the epithelium, and perhaps chorio-capillaris; this was most marked

in the lower part of each fundus, and more so in the R., where the walls of the choroidal trunks were much thickened ("sclerosed"), and in this same part were *three definite pigment patches of characteristic bone-corpuscle pattern;* no other such patches seen in either eye, but in the L. were several smaller intensifications of pigment, less marked and not of typical shape. R.O.D. decidedly palish and hazy, L. not so much so; retinal trunk vessels might pass as within normal size, but are probably all slightly diminished. Refraction slightly H. Lenses and vitreous clear. V. not measured, but he expresses himself as no worse and has for some years held the office of Registrar of Births, Deaths, and Marriages, of a considerable town; his handwriting has not altered in size or neatness; he looks scarcely any older than in 1905, and though rather stout, shows no tendency to corpulence, and resembles his father, who is still alive, both in largeness of frame and general build. Nevertheless, one cannot but recognise that the alteration in the appearances is due to progressive atrophy of the inner layers of the choroid and outer layers of retina, and that the prognosis is by no means so favourable as I thought when I last saw him (1905).

The same apprehensiveness must be felt for his sister, the subject of Case 5, who is seven years junior to Case 4 and now aged 40. I first examined her eyes in 1905, when she was 32, when her fundus in each eye showed innumerable fine white dots like those in her brother (Case 4), but more abundant and the individual dots smaller; there was disturbance of epithelium at the periphery, but neither there nor elsewhere anything suggestive of retinal pigmentation. The discs were decidedly hazy and I thought the retinal arteries rather small.* She, like her brother, had been night-blind as long as she could remember, but had not got worse; V. 15/20, slight H.; Fs. to hand full in daylight, but decidedly contracted when the room was darkened.

* This description of the disc and retinal arteries was omitted in the account of the case in R. L. O. H. Reps., vol. xvii, p. 389, where she is marked as VI, 8, although it occurs in my original notes. In view of the present finding (September, 1913), the omission is important.

I examined her again, with her brother, on September 2, 1913, without a mydriatic, and found plenty of the characteristic white dots, especially between the y.s. region and equator, but they seemed to me to be much less abundant than at the previous examination; peripheral epithelial disturbance just as before; no pigment spots or patches found in either eye, but the discs markedly paler and more hazy than her brother's, the arteries very considerably shrunken and the veins also, but not so much. No choroidal sclerosis; media n. The changes in the discs and retinal vessels are certainly more advanced than at the previous inspection and the retinal dots less numerous, and one can hardly doubt that the morbid process is progressing in her as well as in her brother. V. and Fs. were not examined on this occasion, but she says that her sight, both for near and far, is as good as ever, and that the night-blindness is not worse. Both she and her brother have dark hair, pigmented irides, and moderately dark choroids, his fundus being darker than hers.

Case 4 is very important, as proving that the most typical picture of retinitis punctata albescens may remain to appearance unchanged for many years, at least 18 in this instance, and then give place to the well known changes of choroidal sclerosis and commencing typical retinitis pigmentosa. Although instances of typical retinitis punctata albescens and of retinitis pigmentosa have been recorded by several authors* as occurring in different members of the same family, I have not met with any account of a case such as the present one in which the characteristic appearances of the one condition have been observed to pass into those of the other in the same patient.

* Nettleship, R. L. O. H. Reps., vol. xvii, p. 378 *et seq.*, Cases 138 (Gayet), 148 (Groenouw), 149 (Wuestefeld), 76 (Treacher Collins).

ON THE INHERITANCE OF RETINITIS PIGMENTOSA, WITH NOTES OF CASES.

By C. H. USHER.

THAT retinitis pigmentosa is a hereditary disease has for long been known, but to what degree or in what percentage of cases inheritance can be found has yet to be determined, and not until a very large number of unselected pedigrees, including those in which only single cases occur, have been worked out can this be settled. To mention the opinions of only one or two authors on this subject Hutchinson expresses the view that there can be not the slightest doubt that retinitis pigmentosa is remarkably hereditary.* Landolt in the same year (1873) held that it was almost always hereditary.† More recently (1907) figures have been given by Mr. Nettleship which show that in rather more than 23 per cent. of the 976 families collected by him heredity continuous, discontinuous, or collateral, without consanguinity was proved and in between 3 and 4 per cent. there was heredity combined with consanguinity. In half of the recorded cases the available notes gave no information about these two factors and Mr. Nettleship suggests that had the records, which were often very imperfect, been fuller in these cases heredity would often have been evident.‡ There seems no reason to doubt the soundness of this suggestion which implies that retinitis pigmentosa pedigrees would show heredity in more than 27 per cent. On the other hand, possibly some of the pedigrees of retinitis pigmentosa were recorded just because they demonstrated heredity; these may therefore be regarded as selected, for they would otherwise not have been published, and as unduly increasing the percentage of pedigrees showing inheritance.

In constructing the charts of some new pedigrees a series

* R. L. O. H. Reports, 1873, vol. vii, pt. IV, p. 434.

† An. d'Oc., 1873, vol. lxix, p. 138.

‡ R. L. O. H. Reports, vol. xvii, pt. I, pp. 4 and 6; and Bowman Lecture, Trans. Oph. Soc., 1909, vol. xxix, p. xcv.

has been taken of unselected and nearly consecutive cases in which a diagnosis of retinitis pigmentosa had been made. A few cases were omitted because there were no records of the relatives and they cannot now be traced, or else the diagnosis was doubtful, and also a single pedigree already published.*

Through the courtesy of Dr. A. W. Falconer a Wassermann test was applied to a number of the cases and in each instance was carried out by himself. Broadly, the position as regards the value of this test appears to be (1) that when the test is positive syphilis is present, and (2) when the test is negative the patient may or may not have had syphilis.† The Wassermann test has been applied to cases of retinitis pigmentosa by Hessberg‡ who examined 5 cases, all were negative. Liégard and Offret§ examined 1 case and it was negative. Pöllot‖ 1 case, negative. Igersheimer¶ 7 cases, 6 negative and 1 positive. The case which was positive acquired syphilis after night-blindness had been observed. Gebb** 1 case, negative. Glanz†† 4 cases, all negative. Martin Cohen‡‡ 8 cases, 5 gave a positive and 3 a negative reaction. Fleischer§§ 4 cases, 1 positive and 3 negative. Casali and Pisani‖‖ 1 case, negative. Nakamura¶¶ 1 case, positive.

* Published by Mr. Nettleship in the Bowman Lecture, Trans. Oph. Soc., 1909, vol. xxix, p. cliii and p. xcvi with Fig. 37. I have since seen IV, 5, a boy, aged 8, he has typical retinitis pigmentosa. His mother has normal fundi. Wassermann reaction was negative in IV, 5. (This pedigree should strictly have been included in the present series.)

† McIntosh and Fildes, "Syphilis," International Medical Monographs. London, 1911.

‡ Klin. Monatsbl. f. Augenh., 1910, Beilageheft, p. 63.

§ An. d'Oc., December, 1912, vol. cxlviii.

‖ Arch. f. Ophth., 1912, vol. lxxx, p. 379.

¶ Arch. f. Ophth., 1910, vol. lxxvi, p. 279.

** Arch. f. Augenh., vol. lxiv, p. 204.

†† Berlin Ophth. Soc., 13th July, 1911; Ref. Centralbl. f. prak. Augenh., August, 1911, p. 228.

‡‡ Arch. of Ophthalmology, 1910, p. 91.

§§ Klin. Monatsbl. f. Augenh., July-December, 1910, vol. xlviii, p. 230.

‖‖ Annali di Ottalmologia, Anno XL, 1911.

¶¶ Nippon ganka Zahsl, Juli, 1910-Juli, 1911; Ref. Klin. Monatsbl. f. Augenh., February, 1912, p. 263.

Igersheimer* 13 cases, negative. The total is 38 negative and 8 positive. Komoto† found 16·66 per cent. of retinitis pigmentosa cases gave a positive reaction, but the number of cases is not stated in the reference.

In the present series of pedigrees, 35 individuals with retinitis pigmentosa were subjected to a Wassermann test. Of these, 27 are cases that originated the enquiry‡ into the family histories, 6 occur in the same childships with them and 2 are in other childships—28 were negative and 7 positive.

There are 40 pedigrees and these are arranged in three groups:—A, Wassermann test negative, 23 pedigrees, Figs. 1 to 23; B, Wassermann test positive, 4 pedigrees, Figs. 24 to 27 (it is the result of the test on the original case of retinitis pigmentosa that determines whether a pedigree belongs to group A or B); C, no Wassermann test made, 13 pedigrees, Figs. 28 to 40. The value of this grouping is diminished by the circumstance that a person may have had syphilis although the test gives a negative result. Then, an individual with retinitis pigmentosa may acquire syphilis, as seems to have occurred in Igersheimer's case§ of a married woman with retinitis pigmentosa, where there was night-blindness before she was infected with syphilis. It has been suggested that syphilis may give rise to cases which might be designated retinitis pigmentosa, brought out by syphilitic narrowing of choroidal arterioles.‖ A similar suggestion is made by Knape,¶ namely, that syphilis may be the exciting cause of retinitis pigmentosa where there is a predisposition on the part of the choroidal vessels to endarteritis obliterans due to a congenital

* Vereinigung der Augenärzte der Provinz Sachsen, 7th May, 1911; Ref. Klin. Monatsbl. f. Augenh., 1911, vol. xlix, 1, p. 741.

† Nippon ganka Zahsl, Juli, 1910–Juli, 1911; Ref. Klin. Monatsbl. f. Augenh., February, 1912, p. 260.

‡ Such cases will be referred to as "original cases," and the childships in which they occur as "original childships."

§ *Loc. cit.*

‖ Nettleship, R. L. O. H. Reports, vol. xvii, pt. II, p. 159.

¶ Knape, Ernst V., Arch. of Ophthalm., 1908, pp. 574–587.

anomaly. Again, sometimes ophthalmoscopic appearances may possibly be produced by retinitis pigmentosa, arising from whatever cause, and at the same time a syphilitic lesion such as choroiditis disseminata.

The following signs have been used in the pedigree charts :—

♂ and ♀, normal male and female.
○, normal individual, sex not known.
◯, several normal persons, sex not known.
♂ and ♀, male and female with retinitis pigmentosa.
○——○, twins.
s.p., married and without issue.
+, indicates that eyes were examined with ophthalmoscope.
~~~~, order of birth not known.

In the larger pedigree charts it has been necessary to group numbers of families into rectangular blocks as reproductions of the full detailed charts would have been unmanageable.

In some pedigrees a note was kept of all normals examined, as well as affected individuals; in these the number is stated on the chart. In others no note was kept of all the normals examined. Phthisis, though indicated in some pedigrees, was seldom particularly enquired for. The siblings are arranged in order of birth in the original childships, and as far as possible in parents' childships. Though the purpose of this paper is primarily with reference to heredity some data from the notes of cases are here briefly enumerated.

Sex.—Of the 69 examined cases of retinitis pigmentosa in these pedigrees 42 are males and 27 females—a preponderance of males as is usual. In Group A, males = 28, females = 19; Group B, males = 5, females = 1; Group C, males = 9, females = 7.

Age at Onset.—Information regarding the age at which symptoms were first noticed is frequently indefinite. In a number of cases the statement made is to the effect that sight was always defective or that they had never seen well,
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in others the defect was first noticed during the school period. A considerable proportion (10) believe that there was no defect until 20 or later. In four of the cases the age is given as older than 30 when vision was first affected. The figures are given in this table. Information available in 63 cases.

Age at onset of symptoms :—

	Cases.
From 1 to 5 years	10
„ 6 „ 12 „	24
„ 13 „ 19 „	5
„ 20 „ 30 „	6
„ 31 „ 50 „	3
After 50 years.....	1
"Always had bad sight" or "never saw so well as others in dim light"	12
Uncertain	2
Total	63

The case in which symptoms were first observed after 50 years (Fig. 22, III, 15) appears to belong to the group called senile retinitis pigmentosa,* and the clinical evidence points to a late onset of the affection.

An example of late onset of symptoms is recorded by Morton† in a case which he seems to have suspected was one of retinitis pigmentosa, in which the symptoms began about the age of 50.

Order of birth of retinitis pigmentosa cases :—

	Examined cases.		Examined cases.
1st born.....	9	8th born.....	4
2nd „	10	9th „	4
3rd „	12	10th „	1
4th „	8	11th „	1
5th „	5	12th „	1
6th „	5	13th „	2
7th „	6	14th „	1
		Total	69

* Nettleship, R. L. O. H. Reports, 1908, vol. xvii, pt. III, p. 359.

† Trans. Oph. Soc., 1885, vol. v, p. 142.

There are 17 childships in which two or more cases of retinitis pigmentosa occur; in seven of them the affected persons do not come consecutively in order of birth, being in each case separated by one or more siblings unaffected with retinitis pigmentosa. In Fig. 8 there are three persons with retinitis pigmentosa in a childship, and in Fig. 11 there are two, yet none of the affected individuals occur together. In the remaining 10 childships affected members occur together in groups of two, three, and four, and in six of these childships there are also single cases of retinitis pigmentosa, separated from an affected group by one or more normal individuals. Two of the groups of retinitis pigmentosa cases are formed by the first born siblings (Figs. 19 and 27), five of the groups by the last born (Figs. 7, 13, 23, 29, and 35). In the remaining three childships the affected groups are not at either end of the childship (Figs. 3, 4, and 21).

Thirteen, and possibly 15, of the 69 examined cases may be regarded as *mentally affected*. One was in a lunatic asylum (Fig. 19, V, 469); one was insane latterly (Fig. 37, III, 26); four were mentally slow (Figs. 7, IV, 9; 24, IV, 6; 26, V, 72; 35, IV, 28); two gloomy or sullen and morose (Figs. 3, III, 23; 34, V, 50); three mentally weak (Figs. 13, IV, 20; 19, V, 470; 38, IV, 20); one mentally weak with violent temper (Fig. 10, IV, 8); one timid (Fig. 17, V, 82); two with irritable temper or fidgety (Figs. 20, IV, 17; 25, IV, 11). There are eleven *deaf-mutes*, and, in addition to these, some degree of deafness is noted in 19 cases, no deafness in 19 cases, and in the remaining 20 cases there is no record. The *sense of smell* was tested in nine cases; eight of these are in Group A; in no instance was it defective. One of the unseen cases (Fig. 9, III, 35), a male, with night-blindness and deafness, is reported to have had defective sense of smell.

Six cases had no perception of light. When tested, their ages were 41, 47, 50, 52, 57, and 67. It was doubtful whether a seventh case, an old woman (Fig. 30, III, 12),

had perception of light. In eight cases the vision lay between perception of light and 4/60; 15 cases could see 6/60, or even 6/24; 20 read 6/18 or 6/12; 11 read 6/9 or 6/6, in the last group are some where the only record given is that they read 1 J, and in one case 2 J, or could read the newspapers. In six cases there is no record of the vision. The remaining two "see well in daytime." The refraction of the eye is stated in 48 cases. In 18 both eyes are myopic, in 28 they are hypermetropic or emmetropic. One case has mixed astigmatism in one eye and myopia in the other eye. Another case is emmetropic in one and myopic in the other eye.

Illnesses and other Conditions that may have a Bearing on the Etiology of Retinitis Pigmentosa.—From the notes of 52 cases there is evidence that in 21 there had been no severe illness. In other 2 cases only children's ailments are mentioned. Scarlet fever in childhood in 4 cases; "never strong" is reported of 2 cases; bronchitis and inflammation of lungs in 4; typhoid, "gastric," and "typhus" in 4; necrosed bone and abscess in 2; vomiting in 2; exposure to cold and wet 3; head injury 2; badly nourished 1; fright 1 (Fig. 11, IV, 24); hæmorrhage in six cases (Figs. 15, IV, 34; 20, IV, 17; 22, III, 15; 35, IV, 28; 26, V, 72; 40, III, 7); syphilis in cases giving a positive Wassermann reaction, Group B. Priestley Smith* has recorded cases of retinitis pigmentosa in a family which he believed were not improbably caused by maternal impression or shock. Instances of a pregnant woman experiencing shock or mental distress and bearing a child with retinitis pigmentosa are recorded in Figs. 10 and 39. In Fig. 11, IV, 24 is a woman with retinitis pigmentosa who had a fright at the age of 20, shortly before symptoms were noticed. Cases where the exciting cause may have been loss of blood, in other cases scarlet fever and other diseases, have been described by Nettleship.†

* Ophthalmic Review, 1882, p. 30.

† R. L. O. H. Reports, 1908, vol. xvii, pt. II, pp. 154–163.

Retinitis pigmentosa cases in subjects of cold hands and feet have been published by Hepburn.* Syphilis, as a cause of retinitis pigmentosa, Leber† believed occurred only in a few cases. Most published cases of retinitis pigmentosa, whose origin is attributed to syphilis, seem to be uniocular.‡ A question arises in Fig. 19 whether the site of the manifestations of congenital syphilis in retina and choroid of three sisters (V, 658, 659, 660) was determined by a hereditary susceptibility of those parts to syphilis acting as an exciting cause. They are descended from ancestors common to them and to two childships containing cases of retinitis pigmentosa. Gonzalez§ found that 36 per cent. of the children of syphilitic parents suffered from eye changes, but the proportion of ocular inherited syphilis was not the same in all of the families observed. Jaundice was present in a male (Fig. 28, III, 2), with retinitis pigmentosa. He says the defect of eyesight began at a time when he had much vomiting; he was not alcoholic. Cases of retinitis pigmentosa and night-blindness, associated with disease of liver, are mentioned by Parsons‖ and Hess.¶ In most of the night-blind cases with severe jaundice the ophthalmic signs are negative.**

The *fields of vision* are recorded in 53 cases. In five they are full or nearly full; in eight they are contracted, but none to 20°; 28 are contracted to 20° or less; eight of these extend beyond 20° in one meridian in one or both

* R. L. O. H. Reports, 1908, vol. xvii, pt. II, p. 238; and Ophth. Soc. Trans., vol. xxviii, p. 255.

† Handbuch der Gesammten Augenheilkunde, 1877, vol. v, p. 653.

‡ Reuter, Arch. f. Augenh., 1908, vol. lx, 1, p. 59 (Terrien, Galezowski, Antonelli, Streziminski, Tarnowsky and Fournier, Mannhardt, Leber, Hutchinson).

§ Special Report at the third annual meeting of the Mexican Ophth. Soc. and Anales de Oftalmologia, March, 1908; Ref. The Ophthalmoscope, 1910, pp. 230–1.

‖ The Pathology of the Eye, 1908, vol. iv, p. 1293.

¶ Arch. of Ophthalmology, 1910, p. 482; abridged translation from the German Edition, November, 1907.

** Parsons, Lancet, 22nd February, 1908, p. 556.

eyes; 12 cases had the fields measured by hand-movements: two of these fields are "contracted" and 10 "much contracted." In eight cases a ring scotoma was demonstrated,* and in four of these the peripheral part of the field was full or nearly full. Ring scotomata were sought for only in a proportion of the fields. Whether a real concentric contraction of the fields of vision takes place in retinitis pigmentosa appears, even now, to meet with opposite views. Gonin† maintained that the retraction of the field was not really concentric. Axenfeld,‡ on the other hand, says that it is not uncommon, not only for the ring scotoma to advance towards the periphery, but for the peripheral margin itself to contract centralwards. Bjerrum§ found in retinitis pigmentosa a considerable concentric contraction of the field of vision, although for large objects it had a normal margin. In the two cases of pigment formation in the retina with ring scotoma described by v. Graefe,‖ there was a full field in one and a contracted field in the other.

The fields of vision in an unaffected male (Fig. 23, IV, 55), age 48, who has three brothers with retinitis pigmentosa, are of particular interest in two respects, (1) presence of a partial second ring scotoma, (2) disappearance of an absolute complete ring scotoma and reappearance of part of it. It was thought possible when night-blindness and a ring scotoma were found that he had retinitis pigmentosa sine pigmento, and this may yet be found to be the case. *Double ring scotoma* is represented in the right field by an absolute narrow complete ring scotoma and, within this, a part of a smaller circle situated in the lower temporal quadrant (Fig. 3) between 50° and 60° for most of its extent. The only recorded example of double ring scotoma occurring in a case of retinitis pigmentosa,

* Fig. 6, V, 22; Fig. 14, IV, 114; Fig. 14, IV, 20; Fig. 16, III, 8; Fig. 17, V, 82; Fig. 22, III, 15; Fig. 33, IV, 76; Fig. 40, III, 7.

† An. d'Oc., 1902, vol. cxxviii, p. 105.

‡ Klin. Monatsbl. f. Augenh., Beilageheft, 1909, p. 57.

§ Verhandlungen des Xth Internat. Med. Congresses, Berlin, 1890, pt. x, pp. 66–70.

‖ Arch. f. Ophth., 1858, vol. iv, 2, p. 252.

known to me, is one reported by Hepburn,* where the outer ring scotoma was complete and the inner one almost complete, both were relative. In some other conditions double ring scotoma has been found. In choroiditis disseminata Crzellitzer's† Case 1 had in the left field a nearly complete ring scotoma close to fixation point, and four elongated scotomata nearer periphery. His Case 2 had incomplete double ring scotoma. Baas'‡ case of a syphilitic female, age 18, with atrophic spots in fundus and retinal pigmentation, had two partial ring scotomata in the left field. Ole Bull's case,§ M. A., a syphilitic female, had a nearly complete ring scotoma and within it a linear small scotoma with concavity towards fixation point, certainly suggestive of a second ring scotoma; fundus changes were present; in right field a nearly complete ring scotoma and several small scotomata nearer fixation point. In hysteria double ring scotoma has been found by v. Reuss.‖ Cases of inflammation of the ethmoidal and sphenoidal sinuses have been reported in which ring scotomata were present. Gjessing¶ found three complete ring scotomata in the right field and two in the left field of a man, aged 20, with optic atrophy. He explains the optic atrophy by the presence of ethmoidal empyema on both sides and the ring scotoma as functional, yet there were no hysterical symptoms. Ham** found a ring scotoma for white, connected with the blind spot, and a series of ring scotomata for colour in the right field of vision in a female, aged 35. Later, muco-pus was emptied from the anterior ethmoidal cells, and in a few

* R. L. O. H. Reports, January, 1908, vol. xvii, pp. 234 and 243.

† Arch. f. Augenh., 1900, vol. xl, p. 279.

‡ Das Gesichtsfeld, Stuttgart, 1896; and Arch. f. Ophthalmologie, 1897, vol. xliv, p. 642.

§ The Ophthalmoscope and Lues, Christiania, 1884, p. 60.

‖ Reuss, A. R., Das Gesichtsfeld bei functionellen Nervenleiden, Leipzig and Vienna, 1902, pp. 56–9.

¶ Arch. f. Ophthalmologie, 1912, vol. lxxx, p. 153.

** Nederlandsh Tijdschrift voor Geneeskunde, 11 Marti, 1911, pp. 918–936. Cited by Gjessing, Arch. f. Ophthalm., 1912, vol. lxxx, pp. 159 and 163, footnote.

days the ring scotoma for white disappeared and only a relative ring scotoma for colour remained. The origin of the ring scotoma was thought to be functional. Although

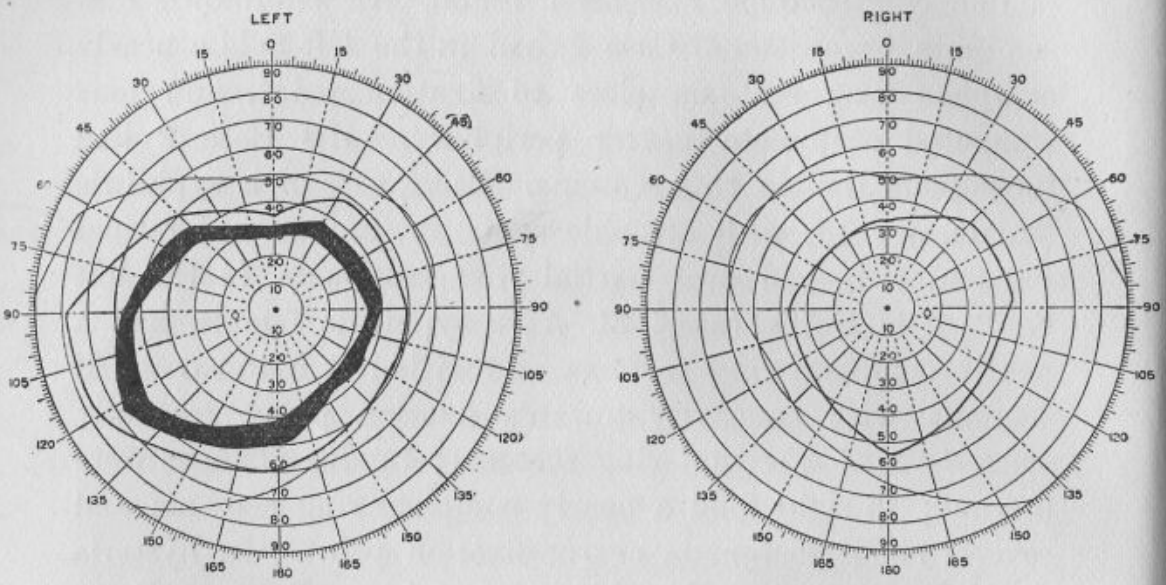


FIG. 1.—Fields of IV, 55, in Pedigree 23, on 1st May, 1909.

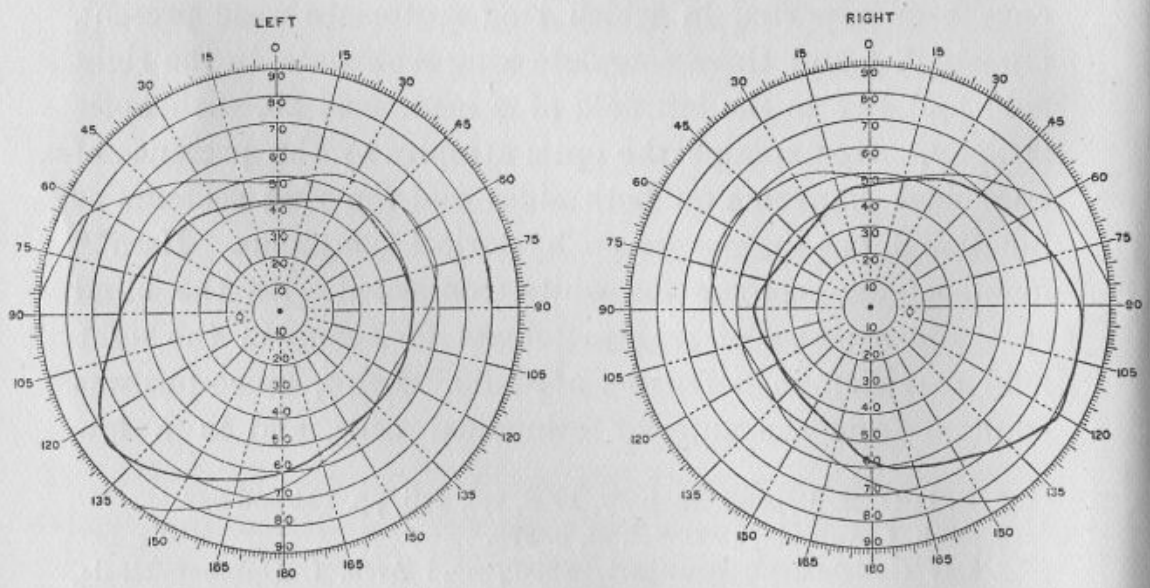


FIG. 2.—Fields on 19th August, 1910.

the next cases had no double ring scotoma they are of interest here for there was retinal pigmentation. Wood and Wallis,* in a case of bilateral papillœdema and ring

* Lancet, 24th September, 1910, pp. 937–939.

scotoma due to sphenoidal sinusitis, thought that retinitis pigmentosa had to be considered because there were spots of retinal pigment in the intermediate zone, but concluded that the papillœdema had in some way been the cause. De Kleijn,* in four cases of posterior sinusitis, saw pigmentation at the periphery of the retina recalling the appearance in retinitis pigmentosa. One, two, and even as many as five ring scotomata are figured by Wettendorfer† in cases of myopia.‡

The second point of interest in this case is the *vacillating or intermittent nature of the ring scotoma.* On May 1st, 1909, a complete absolute ring scotoma was present in the left field of vision, which was concentrically contracted (Fig. 1). The right field was more contracted than the left, but no scotoma could be found. Both fields were taken in good daylight with a 10 mm. white square. About 15 months later, August 19th, 1910, no trace of a scotoma could be detected in either field of vision, both fields again concentrically contracted (Fig. 2). This examination was made under the same conditions as the first one. Two and a half years later, on February 19th, 1913, in good daylight with a 10 mm. white square, both fields were practically full (Fig. 3). Scotomata were found and measured, a 5 mm. white object being employed. In the left field an absolute scotoma was once more present situated in the lower outer quadrant, and in form resembling part of a circle with concavity towards fixation point. Its site corresponded closely to the part of the ring scotoma that occupied the same quadrant of the field more than $3\frac{1}{2}$ years previously. The lower part of the scotoma was in exactly the same part of the field. In the right field was a complete narrow absolute ring scotoma and nearer the fixation point was a second absolute scotoma, occupying the lower temporal part of the field, which in shape

* Arch. f. Ophthalm., 1911, vol. lxxix, p. 497.

† Beiträge zur Augenheilkunde, 1902, vol. v, pt. 49, p. 17.

‡ In a woman, age 38, with high myopia and no retinal pigmentation I have seen a single complete ring scotoma near periphery of field; a similar case is IV, 38, in Pedigree 3.

and situation corresponded closely to the partial ring scotoma in the left field. He was seen a few months later (June 6th), and the fields were taken under the same conditions as regards light and size of objects used. The right field had again

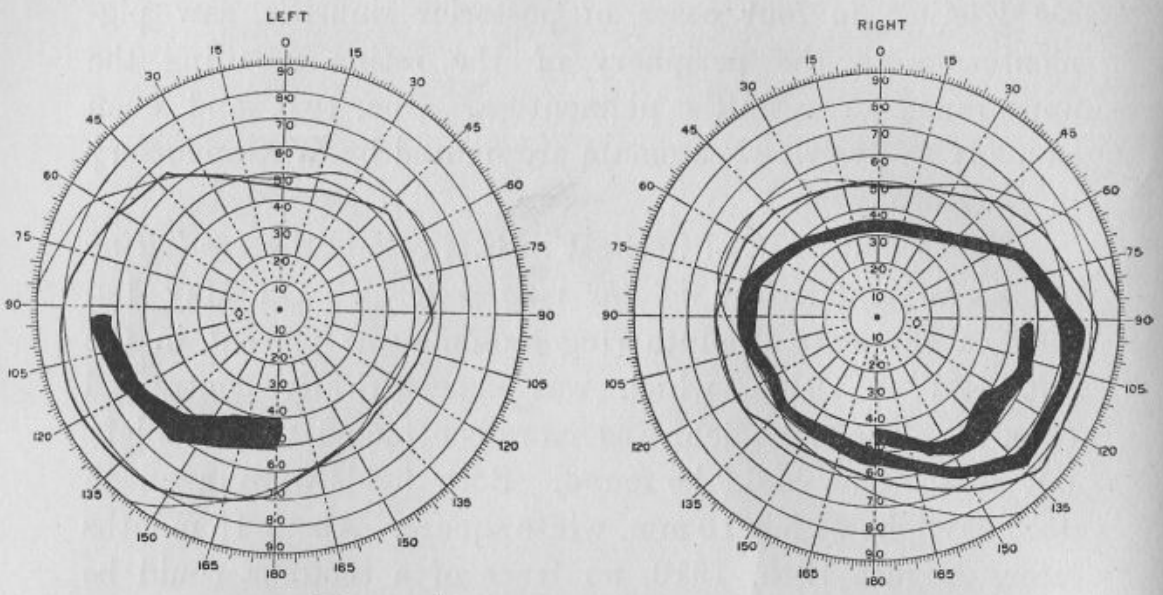


FIG. 3.—Fields on 19th February, 1913.

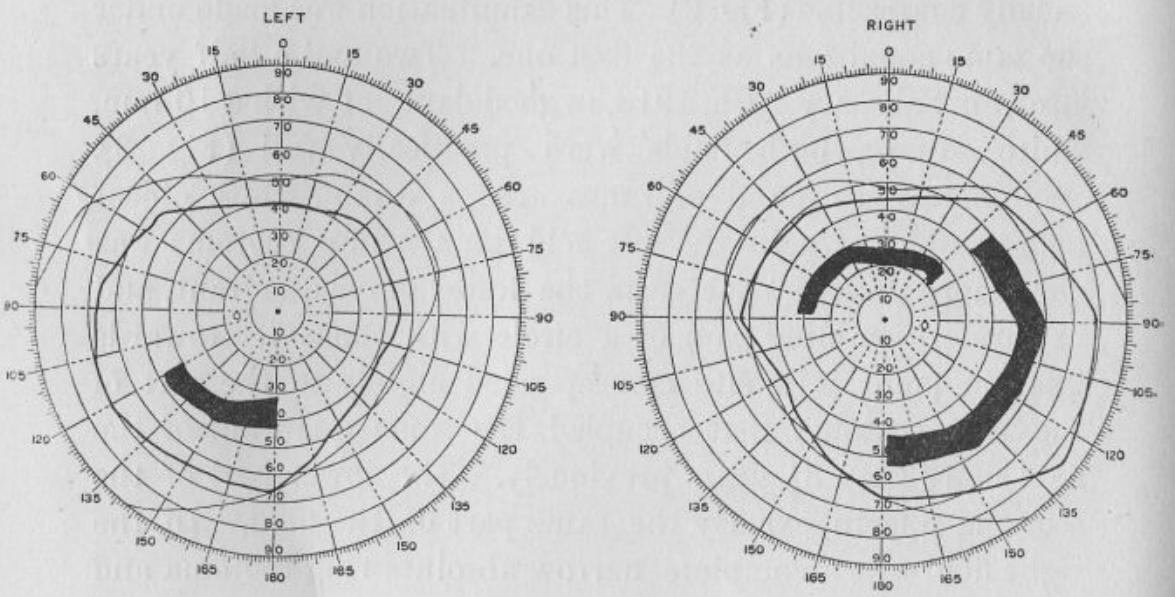


FIG. 4.—Fields on 6th June, 1913.

changed, there was no complete ring scotoma, but two partial ones (see Fig. 4). In the left field an absolute scotoma was still present in its lower temporal part, but it was smaller and did not quite correspond in situation to the one found in

February. A few days afterwards (June 13th), under the same conditions, the scotomata were found to have materially altered. A relative incomplete ring scotoma was present in the left field (Fig. 5), and in the right field a complete ring scotoma which was relative, except in lower temporal quadrant, where it was absolute at the black-shaded area in figure. The patient, an intelligent farmer, who acts as registrar of his district, is a satisfactory subject for examination, and in testing his fields of vision he was quite decided in his replies.

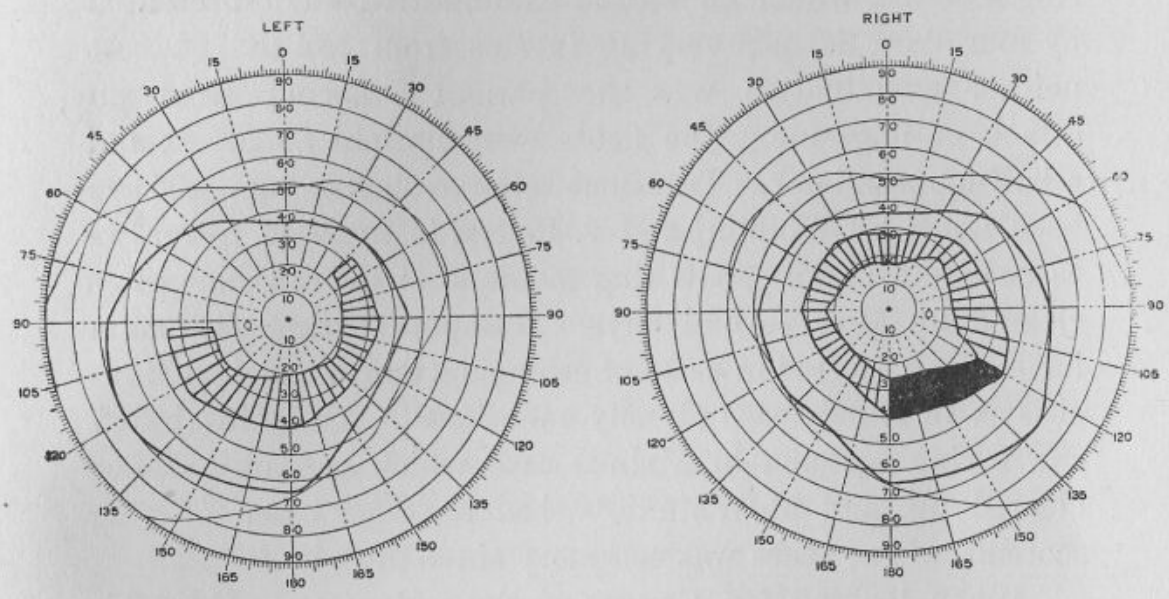


FIG. 5.—Fields on 13th June, 1913.

Hepburn's case, already referred to, with double ring scotoma, somewhat resembles this one in that after an interval of two years the outer relative ring scotoma was replaced by a visual area, but, so far as recorded, had not reappeared. A number of cases have been observed in which a ring scotoma has cleared up. Imre,* in a case of syphilitic choroido-retinitis, found a ring scotoma which eventually disappeared. Bull,† in syphilitic cases, saw ring scotomata alter in form and disappear. Compare also Baas'‡ case. Crzellitzer describes§

* Klin. Monatsbl. f. Augenh., 1876, p. 267.
† *Loc. cit.*
‡ Das Gesichtsfeld, Stuttgart, 1896, pp. 96 and 97.
§ Arch. f. Augenh., 1900, vol. xl, p. 283.

the case of a male with sudden loss of vision in one eye, with normal fundus, in which ringed scotomatous areas cleared up. Wilbrand and Saenger's* case of syphilitic chorio-retinitis had a complete ring scotoma in right field, which had entirely disappeared five months later. In a case of migraine Zentmayer† found an absolute complete ring scotoma in the right field, which five days later was reduced to the half in temporal part of field. On the same dates the first examination of the left field showed a temporal hemianopic ring scotoma, which on second examination was represented by four dark islands varying in size from 15° to 10°. At neither examination was the patient suffering from an attack of migraine. The fields were normal about six and a half months later. Transient relative ring scotomata have been noted by Wölfflin‡ and v. Reuss§ in hysteria. Jess‖ in cases of sun-blinding saw ring scotomata which in some cases cleared up after several days. Temporary ring scotomata have been reported in cases of ethmoidal and sphenoidal sinusitis as in Ham's case already mentioned. De Schweinitz¶ saw a ring scotoma in a sinus case which appears to have cleared up, and McWhinnie** has recorded cases of ring scotoma where there was accessory sinus disease.

When looking for a cause of the scotomata in the fields of vision of J. L. (Fig. 23, IV, 55) there is no reason to suspect syphilis, nor is there any evidence of sinus disease, hysteria, or sun-blinding. He is a subject of Raynaud's disease. Three brothers with advanced retinitis pigmentosa complete the childship. A vascular origin is suggested by the presence of Raynaud's disease, and it is tempting to attribute the vacillating character of the scotomata to spasm of ciliary arteries. This would accord with the vascular theory of

* Die Neurologie des Auges, 1909, vol. iv, 1, p. 15.
† Annals of Ophthalmology, 1912, vol. xxi, p. 279.
‡ Arch. f. Augenh., vol. lxv, p. 309.
§ *Loc. cit.*
‖ Muenchener Med. Woch. Mai, 1912, p. 1100.
¶ Trans. Amer. Ophth. Soc., 1910, vol. xii, pt. II, p. 477.
** New York Med. Journ., 13th August, 1900, vol. xcii, No. 7, p. 301.

retinitis pigmentosa that attributes the cause to diminished blood supply in the chorio-capillaris. In this case, however, the diminished blood supply would be caused, not altogether by sclerosis of choroidal vessels, but in addition by intermittent spasm of ciliary arteries lasting sufficiently long to cause a scotoma, but passing off before the rods and cones were irreparably damaged. It is important to remember, however, that at the present time the fundi of both eyes are normal. It would be difficult to understand how an absolute ring scotoma of long duration could entirely clear up, but in this case we do not know how long it had been present before the first examination or how long it had remained afterwards. The examinations of the fields made in June (1913) indicate that the scotoma can change in form and situation and even disappear altogether in a short time. Yet Bull* states in 'The Ophthalmoscope and Lues' that the form and situation of the scotomata can remain unaltered even for months. Should the disease change for the better the scotomata will diminish and gradually disappear. In a later publication† he states that in recent cases of syphilitic choroido-retinitis the scotoma never remains long unchanged.

The changes that occur in the fingers and toes in Raynaud's disease have been attributed to spasm of arterioles.‡ Spasm of the retinal arteries in Raynaud's disease and other affections is the explanation given by a number of authors for certain ophthalmoscopic appearances seen by them. If spasm of retinal arteries can occur it seems no less likely that spasm should occur in the ciliary arteries. Raynaud§ saw narrowing of the retinal arteries with sometimes local constrictions in a man with local asphyxia of the extremities and transient obscurations of vision. The examinations were confirmed by Galezowski. In another case, a man whose hands and face became

* *Loc. cit.*

† Perimetrie, p. 38. Bonn, 1895.

‡ Raynaud, Arch. Gén. de Méd., 1874, vol. i, p. 205 (VIe série, T. 23).

§ *Loc. cit.*, pp. 8, 11, and 814.

livid and who had transient attacks of dim vision, the arteries of the fundus of the eye became narrowed. He was examined by Dr. Panas. Some twenty years later Panas* states in his 'Maladies des Yeux,' that he was unable to make out any difference in the calibre of the vessels in any of the cases of local asphyxia of the extremities which he examined ophthalmoscopically at the request of Raynaud. Friedman† saw marked contraction of the small arteries of the fundus oculi in a woman, aged 23, with Raynaud's phenomena, when the asphyxia of the extremities was most pronounced. She had no disturbance of vision. Weiss‡ in a man, aged 54, subject to angiospastic attacks in left hand, saw, during transitory attacks of amaurosis in his right eye, the retinal arteries and then the veins empty themselves. After half an hour the vessels began to refill, at first the small atypical cilio-retinal vessels and the veins, and then gradually the larger arteries. The left was an injured blind eye. Kuffler§ in a student with transient attacks of blindness and evanescent erythema of the hands and face saw an appearance of embolism on the first ophthalmoscopic examination. Then in other affections Mauthner‖ in a man with sudden partial loss of vision which soon became complete saw perfect anæmia of the retinal arteries, evidently the result of embolism. But in a few minutes the blindness disappeared and the vessels were found to have returned to the normal condition. Benson¶ in a similar case where the ophthalmoscopic appearances during an attack were noted attributed the condition to spasm of retinal arteries. In migraine Walton** reports the case of a female with hysterical tendency and left-sided

* Traité des Maladies des Yeux, vol. i, p. 619. Paris, 1894.

† Amer. Journ. Med. Sci. Philad., 1910, vol. cxxxix, p. 238.

‡ Bericht 38 Versam. Ophth. Gesell. Heidelberg, 1912, p. 205.

§ *Ibid.*, p. 210.

‖ Amer. Journ. Med. Sci., 1874. Ref. Glasgow Med. Journ., April, 1874, from Stricker's medizin. Jahrbücher, No. 11, 1873.

¶ Trans. Eighth Internat. Ophth. Congress, Edinburgh, 1894, p. 81.

** Journ. of Nervous and Mental Disease, January–October, 1884, p. 429.

migraine in which there was an extreme contractility of the retinal blood vessels, which under ophthalmoscopic examination contracted to one-third their calibre. This was attributed to spasm of the blood vessels, which caused an intermittent retinal ischæmia. Narrowing of retinal arteries during an attack of migraine was observed by Pichler.* Landesberg† reports his own case in which marked narrowing of retinal arteries in both eyes was observed during the paroxysms. Knies‡ saw sudden narrowing of the retinal arteries from 10 to 20 seconds before an attack of epilepsy in a boy. In the cold stage of malaria Schnabel§ saw an enormous narrowing of the retinal arteries. Van Duyse‖ in a case of obstruction of retinal artery and sudden blindness attributes the ischæmia to spasm of retinal arteries. Galezowski¶ in a paper on spasm of the retinal vessels records a case of attacks of dimness of vision in which the arteries near the optic disc were in places invisible. Wagenmann** in a man with arterio-sclerosis and attacks of blindness found during the attack no blood column recognisable in the arteries. Ten minutes from onset a fine red line became visible in the arteries. Sachs†† in his case which resembled one of retinal embolism saw an appearance suggesting a slow peristaltic wave passing peripherally in a retinal artery which he attributed to cramp of the artery. Another example is Harm's‡‡ case with choked disc. If then temporary contraction of arteries in the extremities and in the retina can give rise to loss of function in these parts, there seems no reason to doubt the possible occurrence of spasm in the ciliary arteries.§§

* Prager Med. Woch., 1912, vol. xxxvii, No. 43, p. 607.
† Arch. f. Augen- und Ohrenheilkunde, 1878, vol. xvii, p. 268.
‡ Bericht Heidelberg Ophth. Gesellsch., 1877, p. 61.
§ Arch. f. Augenh., 1885, vol. xv, p. 377.
‖ Arch. d'Ophtalm., 1908, p. 129.
¶ Recueil d'Ophtalmologie, 1892, p. 69.
** Arch. f. Ophthalm., 1897, vol. xliv, p. 219.
†† Beiträge zur Augenh., 1902, vol. v, 4, p. 34.
‡‡ Bericht d. Ophth. Gesell., 33, p. 253. Heidelberg, 1906.
§§ In a recent discussion at the Ophthalmological Society on vascular

A point worth attention, in the case of J. L., is the absence of visual symptoms. He had not known that anything was wrong with his eyesight. His fields of vision were measured because he had come with a brother who had advanced retinitis pigmentosa. It is just possible, therefore, that intermittent ring scotomata may not be so very uncommon. Wittmer* in a case of retinitis pigmentosa with absolute ring scotoma in each field noticed that no inconvenience was caused to the patient—a tailoress. A gamekeeper (Fig. 40, III, 7) with ring scotoma had no difficulty in shooting birds, but he could not kill ground game.

This case of Raynaud's disease with ring scotoma may be compared with Mr. Nettleship's† case of senile gangrene in a woman with advanced retinitis pigmentosa and with Hepburn's‡ cases of retinitis pigmentosa with cold hands and feet, also with Bruner's§ case, a female with ring scotoma and cold hands and feet. In Zentmayer's case already referred to there was increased blood pressure. Grossmann‖ found retinitis pigmentosa associated with congenital heart disease and familial polydactylism. Guglianetti's¶ case of Raynaud's disease with sclerosis of choroidal vessels and pigment deposits at periphery of retina like retinitis pigmentosa and with night-blindness probably belongs to the same category as J. L. (Fig. 23, IV, 55). The optic discs and retinal vessels were normal. A sister had similar fundus appearances. In none of my cases of retinitis pigmentosa is there any record of cold hands or feet though this was enquired for in some of them. In some of the pedigrees there is evidence of arterial disease in individuals unaffected with retinitis

and other retinal changes in association with general disease, several speakers expressed a belief in the occurrence of spasm of retinal arteries (Trans. Ophth. Soc., vol. xxxiii, 1913).

* Arch. f. Augenh., 1911, vol. 68, p. 83.

† R. L. O. H. Reports, 1908, vol. xvii, pt. III, p. 360.

‡ R. L. O. H. Reports, 1908, vol. xvii, pt. II, p. 238.

§ Ophthalmic Record, 1898, p. 556.

‖ Wien. Med. Woch., 1908, No. 14, p. 742.

¶ Archivio di Ottalm., vol. xv, p. 469; Ref. Klin. Monat. f. Augenh., 1909, vol. xlvii, 1, p. 688.

pigmentosa. This is illustrated in Fig. 16, where a male (III, 35) with signs of early arterial degeneration and central retinitis suffers from cold hands and feet. His daughter (IV, 43) also is subject to cold hands and feet. Again, in Fig. 9, IV, 16, a medical man, reports that arterio-sclerosis especially of the arteries of the feet appears to be the family weakness, III, 17, died of acute gangrene of foot; III, 15, had arterio-sclerosis and III, 18, had defective circulation in feet for many years.

Night-blindness is noted as being present in 57 cases, there is no information about it in 10 and in two cases it was absent. One of those without night-blindness (Fig. 34, V, 50) has lens opacities and his fields are reduced to 35° on nasal side and 70° on temporal side. He complained of the sun dazzling him. The other case (Fig. 40, III, 7), when seen at first examination, said that he could see best in dim light. The fields of vision were full and there were ring scotomata. On second examination 17 years later fields were tested without a perimeter and found to be contracted. He maintained that he could see as well as other people in dim light. Leber* noticed that some cases of retinitis pigmentosa could see even better at night than in the day and that they were dazzled in bright daylight. Sometimes, but not always, the cause of dazzling was found in the presence of partial lens opacity. Nettleship† considers that the condition of those cases in which there is no particular difficulty at night is intelligible where the periphery of the retina is still tolerably healthy (ring scotoma with a wide belt of peripheral vision). Axenfeld‡ reports a case of retinitis pigmentosa in which no night-blindness had been complained of and in a second case no night-blindness was discovered by the use of Förster's photometer. The fields of vision are contracted in the second case and a ring scotoma is present in each field. He reported a third case§ of

* Handbuch der gesammten Augenh., 1877, vol. v, 5, p. 644.

† R. L. O. H. Reports, 1908, vol. xvii, pt. III, p. 360.

‡ Klin. Monats. f. Augenh. Beilageheft, 1909, pp. 54–5.

§ Bericht Ophth. Gesell. Heidelberg, 1906, p. 55.

retinitis pigmentosa without night-blindness and with ring scotoma. The papilla was not markedly atrophic and the retinal vessels were not narrow. Marlow's* case had very much contracted fields of vision. Kapuscinski† records three cases of retinitis pigmentosa with typical fundus appearances and ring scotoma in which there was no night-blindness.

Retinitis Pigmentosa sine Pigmento.—In this series of retinitis pigmentosa cases, five eyes in three individuals, examined ophthalmoscopically and found on first examination to be without pigment in the retina, developed typical appearances of retinitis pigmentosa, as seen on subsequent examinations. Case 1 (Fig. 32, IV, 26), a male, aged 14, with night-blindness, which had been noticed for five years. Vision in good light, with correction of myopic astigmatism, was R. 6/12, L. 6/18. In dim light V. 6/60, when a normal eye saw 6/9. Fields of vision slightly contracted. After homatropine with pupils dilated, no pigment could be found in the retina at its periphery or elsewhere. The optic discs looked redder and of a more uniform colour than usual, otherwise fundus of each eye was normal. No note of size of retinal arteries. Media clear. A diagnosis of retinitis pigmentosa sine pigmento was recorded. When examined 10 years later (1911), both fundi presented the usual appearance of well-marked typical retinitis pigmentosa with moss-like retinal pigment. Case 2 (Fig. 5, VI, 40), a female aged 8, when first examined in 1906. R. and L. V. 6/36 in good light, with correction of myopic astigmatism. Fields of vision much contracted. After using atropine no pigment was found in the retina of right eye. There was some pigment in retina of left eye. The retinal arteries were very narrow in both eyes, optic discs grey, semi-translucent, with blurred edges; choroidal vessels much exposed except at macula. When examined again in 1910 a considerable quantity of pigment was present in the

* Amer. Journ. of Ophthalmology, 1894, vol. xi, p. 191.

† Diss. Freiburg, 1909; Ref. Klin. Monats. f. Augenh., 1910, vol. xlviii, 2, p. 649.

retina of right eye, but there was much more in left eye. Retinal arteries and choroidal vessels of same appearance as on last examination; O.D.'s waxy; no white spots in fundus; night-blindness very marked. In light that normal eyes could read 6/12 she failed to see a large white board, though in good light she read 6/36 with correction. Case 3 (Fig. 27, III, 18), a farmer, aged 37 when seen in 1899. At that time V. = 6/24 in each eye; refraction emmetropic; fields contracted to within 10°. After homatropine, with pupils dilated, ophthalmoscopically: media clear; optic discs pale, edges blurred; retinal vessels much narrowed; retinal pigmentation chiefly at periphery; the pigment is mostly in very fine particles; choroidal vessels much exposed; a few circular grey spots 1/6 diameter of optic disc, with choroidal vessels passing uninterruptedly across them, are present in lower part of fundus; he denies having had syphilis. In 1912 vision was reduced to perception of light in each eye; optic discs were "waxy"; retinal arteries narrow; moss-like and granular pigment was present all over both fundi, except close round the optic disc. He has two brothers affected in the same manner. From hospital notes in his possession we learn that, in 1889, his vision was 6/9 in each eye; field in diminished light was not affected; that two brothers had retinitis pigmentosa, and that no pigment was seen in the retina of either of his eyes at that date. There is no note of the condition of the retinal arteries or optic disc.

Reports have been published of retinitis pigmentosa cases occurring with cases of retinitis pigmentosa sine pigmento in siblings, and also of several cases of retinitis pigmentosa in the same childship, in which the retinal pigmentation was found to be most marked in the oldest sibling, in less quantity in the others according to age, being most scanty in the youngest.*

* Snell, Ophthalmic Review, 1886, vol. v, p. 72; Derigs, Inaug. Diss., 1882, Ref. Nettleship, R. L. O. H. Reports, vol. xvii, III, p. 366; Cant, W. J., Ophthalmic Review, 1886, vol. v, p. 245; Gebb, Arch. f. Augenh., vol. lxiv, pp. 204–8.

An example of this sort occurs in Fig. 13. These cases are very suggestive, but do not actually prove that retinitis pigmentosa sine pigmento develops into typical retinitis pigmentosa. Peltesohn's* case, seen ophthalmoscopically before and after the deposit of pigment, had isolated pigment granules here and there in the retina at first examination. Mooren's† three cases all had a little retinal pigment when first examined. Shoemaker‡ saw a girl of 11 years with retinitis pigmentosa sine pigmento, who five years after the original record had a small amount of pigment around periphery in both eyes. Mr. Nettleship discusses the subject in these Reports.§

Sometimes there is difficulty in deciding whether to accept a case as one of retinitis pigmentosa. In such a case as Fig. 27, III, 18, in Group B, the presence of a few circular grey spots at one part of the fundus may well give rise to doubt as to whether the case is one of true retinitis pigmentosa or choroido-retinitis from congenital syphilis. The choroid is not completely atrophied at the spots referred to. The case is one of long standing. His two brothers were independently diagnosed in 1889 as having retinitis pigmentosa. Pöllot‖ has recorded a pedigree in which in certainly two, and perhaps three, generations, an anomalous chorio-retinitis pigmentosa not always having the same, but with similar appearances ("krankheitsbildes"), is directly inherited. Six, and possibly eight, persons were affected, and he considers it not wholly improbable that all of these cases belong to the group of retinitis pigmentosa. His Case 3 presented several round and oval atrophic white choroidal spots as in choroiditis disseminata. Compare likewise the two families with chorio-rétinite pigmentaire familiale seen by Dor,¶ and a familial fundus affection

* Centralbl. f. prakt. Augenh., vol. xii.
† Ophthalmiatrische Beobachtungen, 1867.
‡ Retinitis Pigmentosa, pp. 54 and 9. Philadelphia, 1909.
§ R. L. O. H. Reports, 1908, vol. xvii, Part III, p. 365.
‖ Arch. f. Ophthalm., 1912, vol. lxxx, p. 379.
¶ La Clinique Ophtalmologique, 1910, p. 60.

reported by Kuffler,* with changes at the periphery like hereditary syphilitic pepper and salt retinitis. Again, it was difficult to exclude a case with changes at the macula in Fig. 33 (V, 35), first cousin once removed of a woman with retinitis pigmentosa, for not only may the macula be involved in retinitis pigmentosa, but there may be central changes to the exclusion of the equatorial pigmentation.†

A female, L. G. (3, IV, 38), age 21, whose mother had advanced retinitis pigmentosa has, June, 1913, a complete ring scotoma for white in each field of vision (Fig. 6). The possibility of these being signs of retinitis pigmentosa at once suggested itself. The ophthalmoscopic appearances, however, were not those of retinitis pigmentosa, for the retinal arteries were of full size, optic discs of good colour, and there was no characteristic retinal pigmentation. There was a high degree of myopic astigmatism, and as ring scotomata occur in myopia‡ it is quite possible that this is the explanation of their presence in this case, notwithstanding the retinitis pigmentosa in the mother. The right field in Wettendorfer's§ case of myopia, his Fig. 29, Case 11, resembles the fields of vision in this case in that a single complete ring scotoma occupies a part of the field near the periphery.

In the retinitis pigmentosa cases mental affections, deaf-mutism and deafness have already been referred to (p. 135). Coloboma of iris was present in 1 case (Fig. 4, IV, 26), lamellar cataract in 1 case (Fig. 20, IV, 17), hyaline bodies‖

* Deutsche Med. Woch., 1910, No. 45, p. 2122.

† Nettleship, R. L. O. H. Reports, vol. xvii, pt. II, p. 158.

‡ Wilbrand and Saenger, Die Neurologie des Auges, 1909, vol. iv, p. 42.

§ *Loc. cit.*

‖ In one of two cases of hyaline bodies at optic disc reported by Morton and Parsons there was retinitis pigmentosa, and of 42 cases in the literature collected by them, 7 occurred in cases of retinitis pigmentosa (Trans. Oph. Soc., 1903, vol. xxiii, p. 135).

Doyne mentions a case of retinitis pigmentosa that had a *drusen* body overlapping the inner side of the right disc (The Ophthalmoscope, 1910, p. 626).

Topolanski has reported "Drusen am Sehnerveneintritte und Pigment

on optic disc in 3 cases (Fig. 7, IV, 10, and IV, 9; Fig. 14, IV, 114), dislocated lens in 1 case (Fig. 25, IV, 7),* nystagmus in 9. In brothers and sisters of the retinitis pigmentosa cases mental affections occurred in 3, Raynaud's disease in 1, chorea in 1, hydrocephalus 1, clubfoot, 1; of ocular conditions, ptosis 1, congenital nystagmus 2, myopia 3, senile cataract 2, coloboma of optic nerve 2. In childships unaffected with retinitis pigmentosa the conditions are given in this table.

Disease.	1. In childships unaffected with retinitis pigmentosa.	2. In unaffected members of childships in which retinitis pigmentosa occurs.	1 and 2 together.	3. In those with retinitis pig-mentosa.	Total, 1, 2, and 3 together.
Mental	71	3	74	13	87
Deaf-mutes	11	—	11	11	22
Epilepsy	4	—	4	—	4
Hysteria	1	—	1	—	1
Raynaud's disease	—	1	1	—	1
Exophthalmic goître	1	—	1	—	1
Phthisis	51	1	52	—	52
Deformed	2	1	3	—	3
Cleft palate	1	—	1	—	1
Digital deformities	1	—	1	—	1
Hare-lip	2	—	2	—	2
Spina bifida	1	—	1	—	1
Hydrocephalus	4	1	5	—	5
Myopia	37*	3	40	20	60
Ptosis, congenital	—	1	1	—	1
Lamellar cataract	1	—	1	1	2
Coloboma optic N.	—	2	2	—	2
Congenital nystagmus	—	2	2	—	2
Coloboma of iris	—	—	—	1	1
Hyaline bodies on O.D.	—	—	—	3	3

* Three of these are in children of retinitis pigmentosa cases.

Degeneration in der Retina," Wiener Ophth. Gesell., 26th October, 1908; Ref. Klin. Monats. f. Augenh., 1909, vol. xlvii, 2, p. 121.

* Herrlinger, in 92 cases, saw one with dislocated lens (Inaug. Diss. Tübingen; Ref. Nagel's Jahresb. for 1899, p. 355).

Lindner mentions a case (Wien. Med. Woch., 1895, No. 37; Ref. Die Neurologie des Auges, Wilbrand u. Saenger, 1909, vol. iv, p. 100).

It is not suggested that these figures can in any way represent the total number of individuals affected with these conditions. This applies particularly to Column 1. As stated, phthisis was seldom enquired for. The only abnormal ocular conditions found in the parents were: in mother, primary glaucoma one case, choroidal atrophy in one, myopia in one and possibly in two cases; in father one doubtful myopia. Of other conditions, several are affected mentally; one mother is "peculiar," another "peculiar and gloomy"; one father is alcoholic, another excitable, and a third one is mentally affected. None of the parents were epileptics as in Cant's* case, where the father of four children with retinitis pigmentosa was an epileptic. None of the retinitis pigmentosa cases had epilepsy. Such a combination occurred in Nolte's† atypical case.

Marriage and Fertility.—I. Of the 69 cases of retinitis pigmentosa 21 married, 11 males and 10 females. In Group A, 9, 5 males and 4 females; Group B, 4, 3 males and 1 female; Group C, 8, 3 males and 5 females. Nine of the 48 unmarried cases were under 18 years of age. Five of the 21 who married had no children. One of these is in Group B. The 16 married cases had altogether 90 children, 38 males and 51 females, and a child, sex unknown, that died young, and there were 11 miscarriages. This gives an average of 6·3 conceptions for the 16 fruitful marriages. One of the unmarried cases had an illegitimate son. Amongst the offspring twins occur once. In no instance has a parent with retinitis pigmentosa had a child affected with the same disease. II. Parents of the retinitis pigmentosa cases. As a result of the matings, there were 307 conceptions from 40 unions, the illegitimate child and his parents in Fig. 38 not being included, this is 7·67 for each union. This includes 9 miscarriages, 20 that were stillborn, or died in infancy, and 10 that died young. The largest family numbers 14, the smallest 1. At least 3 of

* Ophthalmic Review, 1886, vol. v, p. 245.

† Inaug. Diss., Marburg, 1896.

the retinitis pigmentosa cases were born out of wedlock (Figs. 17, 32, 38). Out of 711 idiots Mitchell* found that 108 were born out of wedlock.

Parents' Ages at Marriage.—The age at marriage of 20 of the fathers and 20 of the mothers was ascertained. Father's average age is 27·5 years; oldest age at marriage is 48, youngest 19. Mother's average age is 24·2 years, oldest age at marriage is 30, youngest 20.

Age at Death.—Average age of father (15 cases) is 68·06: oldest age at death is 87, another is past 70, youngest is 27. Average age of mother (18 cases) is 64·3; oldest age at death is 89, youngest 40.

Cause of Death of Parents.—Information available in only a few. In the father (seven cases): Cardiac disease in two cases; phthisis, abdominal tumour, bronchial affection, "inflammation of a rupture," "gradual illness," were each responsible in one case. In the mother (11 cases); "paralysis" in two cases, "change of life" two cases; phthisis, abdominal tumour, bronchial inflammation, pneumonia, cancer, "fever," rheumatism, were each the cause of one death.

Consanguineous Marriage in Parents and Grandparents of Retinitis Pigmentosa Cases.—In 41 unions between the parents of the original case† in each pedigree, consanguinity occurs nine times; eight are first cousin marriages and one a marriage of first cousins once removed. This gives 19·5 per cent. of first cousin marriages. Of the eight first cousin marriages the kind of cousin marriage is in two cases (Figs. 3 and 27) between children of brothers; in three cases (Figs. 4, 24, 34) between children of sisters; in three cases (Figs. 2, 6, 21) between children of brother and sister. The father in two cases (Figs. 6 and 21) is a child of the brother and the mother a child of the sister, whilst in Fig. 2 the father is a child of the sister and the mother a child of

* The Edinburgh Medical Journal, March, 1865, p. 792.

† The half brothers in Pedigree 25 are both included as original cases in this connection.

the brother. Adding to these, two marriages of first cousins, parents of children with retinitis pigmentosa, in other parts of the pedigrees—one in Fig. 19, marriage of the parents of IV, 163, where the father is child of a brother and the mother a child of his sister, the other in Fig. 21, marriage of the parents of IV, 20–23, where the father is child of a sister and the mother a child of her brother—there are 10 first cousin marriages in parents of retinitis pigmentosa cases. In five of these the father and mother are children not of two brothers or two sisters, but of a brother and a sister or a sister and a brother. Cases which show the kind of cousinship of consanguineous ancestors have been collected by Mr. Nettleship.* Consanguineous marriages are recorded five times in the grandparents of this series of retinitis pigmentosa cases—in paternal grandparents four times, and in maternal grandparents once. The degree of consanguinity in one of the cases is not known. The remaining four are first cousin marriages. Adding to these the maternal grandparents of an affected individual (Fig. 19, V, 660) outside the original childship, who are first cousins, there is a total of five first cousin marriages in grandparents of retinitis pigmentosa cases; 12 of the 40 pedigrees show consanguinity, that is, 30 per cent. In Group A the percentage is 39·13. This approximates to what was obtained from large numbers by Mr. Nettleship.† The proportion of cousin marriages in the general population of the districts‡ from which the cases came is not known. In a small fishing community on the north-east coast of Scotland Mitchell,§ in 1865, found blood relationship in 1 to 4·4 of all marriages. Darwin‖ thought it probable that 3 per cent. was a superior limit for the whole population in England, and said he would not be greatly surprised if the marriages of first cousins alone were as many as 4 per cent. in Scotland.

* R. L. O. H. Reports, 1908, vol. xvii, pt. III, p. 336.
† R. L. O. H. Reports, 1907, vol. xvii, pt. I, p. 4.
‡ North-east of Scotland.
§ Edin. Med. Journ., June, 1865, p. 1075.
‖ Journ. Stat. Soc., June, 1875, pp. 153–182.

Mitchell placed the proportion of cousin marriages in Great Britain at probably not more than 1 in 60 or 70.* Karl Pearson† from an inquiry addressed to doctors through the British Medical Journal estimated the percentage of cousin marriages to be 4·7, this proportion refers to the middle classes only, and among the parents of the patients at Great Ormond Street Hospital, where the Eugenics Laboratory made direct investigations into the matter, it was found to be 1·3.

The Inheritance of Retinitis Pigmentosa as shown in these Pedigrees.—It is exceptional to find affected individuals in more than one childship of a pedigree, and in a large proportion of pedigrees there is only a single case of retinitis pigmentosa present—21 in 40 pedigrees. Excluding Fig. 25, in which two half brothers are affected, only 4 of the 40 pedigrees show evidence of retinitis pigmentosa in persons outside the original childship, and these require careful consideration. In two of them the diagnosis rests solely on description, the fundus not having been examined. In Fig. 9 there seems good reason for believing that a first cousin once removed (III, 35) of the members in the original childship had retinitis pigmentosa, because he was well known to his first cousin (III, 24) to be night-blind in the same manner as his own two sons (IV, 58 and 60). He was deaf and known to lisp, and had defective sense of smell. Again, in Fig. 21 the evidence of retinitis pigmentosa occurring outside the original childship is satisfactory. Two brothers (IV, 20, and IV, 21), who died at age of 15, and their sister (IV, 22), who died at age of 5, are believed by their cousins, themselves affected, to have been subjects of the same disease with night-blindness, as well as another sister (IV, 23). She is an inmate of an asylum in Canada, and the medical officer's report says that "the patient you refer to is enjoying good health, but she is a deaf mute and the sight

* Edin. Med. Journ., April, 1865, p. 898.

† Elderton, Ethel M., "On the Marriage of First Cousins," Galton Laboratory for National Eugenics. London, 1911, p. 23.

is almost gone. She no doubt has retinitis pigmentosa."* The unseen cases marked black with a mark of interrogation in Fig. 30 have not been accepted, as the evidence of retinitis pigmentosa is insufficient and so the pedigree has been placed with those in which affected individuals are limited to a single childship; thus, a male (IV, 28), whose mother has retinitis pigmentosa, died in an asylum. His vision was "just the same as his mother's" (III, 12). Two males in generation II are reported to have been night-blind, one (II, 6) died at 50, the other is II, 10, in the pedigree chart.

The remaining pedigrees, two in number, are the only ones in which retinitis pigmentosa was proved by ophthalmoscopic examination to occur in individuals outside the original childship and even these require comment. In Fig. 14, IV, 20, although well known to her relatives to be night-blind, has exceedingly scanty moss-like pigmentation limited to the lower part of each retina. The optic discs would probably be passed as normal; retinal arteries show some narrowing; fields of vision contracted at upper part principally and there is a partial ring scotoma below. She is useless in dim light and always has been so, her age is 32. Wassermann test positive. This appears to be a nearly stationary case of retinitis pigmentosa at an early stage and might almost be designated sine pigmento. In the second pedigree, Fig. 19, a female (V, 660) with fundus appearances in each eye characteristic of retinitis pigmentosa, gave a positive Wassermann reaction and has sisters who are undoubtedly affected with congenital syphilis. IV, 163, a male, in the previous generation, now dead, had no blood test made. Fig. 33 has been placed with the single case pedigrees, because the fundus appearances in a female (V, 35), in a different childship from the original case, are so atypical, the changes being limited to the macular regions and a Wassermann test proving positive. It is remarkable that in these three pedigrees the three individuals

* In a letter, 6th May, 1911.

occurring outside the original childship whose blood was tested in each case gave a positive Wassermann reaction. It may seem quite unnecessary to say, when as it sometimes happens defective vision is attributed by a relative to the same condition, retinitis pigmentosa, that he or she or some other relative is affected with, that the statement should be received with great caution. Yet, it has so frequently occurred when working out these pedigrees that a description closely resembling that of retinitis pigmentosa has led to the supposition that this disease was present, until on examination some other condition was found, that the importance of accepting with reserve any case that has not been examined, it seems to me, cannot be too strongly emphasized. A father (IV, 4) and son (V, 6) in Fig. 30 were both reported to have the same condition, retinitis pigmentosa, as III, 7 and 12, yet in the case of IV, 4, nothing but myopia was found present, the son was not examined. In Fig. 2, an aunt (III, 8) of a case of retinitis pigmentosa has night-blindness which was proved to be due to gross symmetrical choroido-retinitis. Another good example is in Fig. 4, where a grandmother (III, 47), mother (IV, 109), and daughter (V, 98) were all supposed to be affected in the same way as their relatives, four siblings with retinitis pigmentosa (IV, 26 to 29). The daughter has myopia and no retinitis pigmentosa. In the case of the mother and grandmother, with complete cataracts, the history obtained from themselves when examined suggested myopia rather than retinitis pigmentosa. Any of these examples and also others that could be cited were liable to have been accepted as cases of retinitis pigmentosa had no examination been made.

Not one of the pedigrees contains many cases of retinitis pigmentosa and none illustrate continuous direct inheritance of the disease, unless in Fig. 30, a son (IV, 28) who died in an asylum is accepted as having had retinitis pigmentosa, with which his mother was affected. Nor is there discontinuous direct inheritance if we exclude in Fig. 36 a granddaughter of a case with retinitis pigmentosa in which the

evidence of retinitis pigmentosa is inconclusive. Collateral inheritance (Bollinger)* occurs in 14 of the pedigrees. Discontinuous or indirect inheritance (Nettleship)† is found in four (Figs. 9, 14, 19, and 21). In one pedigree (Fig. 25) half siblings are affected. Of the 35 pedigrees in which the disease is limited to a single childship, in 21 only a single case occurs, in 7 there are 2 affected individuals, in 4 there are 3 and in 3 there are 4 cases of retinitis pigmentosa.

When "equivalents" are considered by themselves in relation to the original case of retinitis pigmentosa continuous direct dissimilar inheritance occurs three times (Figs. 3, 6, and 26) as coloboma of optic disc, cleft palate and mental affection. Discontinuous or indirect dissimilar inheritance occurs in 27 pedigrees, and in 8 (Figs. 3, 10, 11, 14, 17, 19, 21 and 36) "equivalents" are found, both in the original childship and elsewhere in the pedigree. Thirteen of the pedigrees which show dissimilar inheritance are amongst the 21 pedigrees that do not show similar inheritance. Of these 13 pedigrees (Figs. 1, 10, 15, 16, 20, 24, 26, 31, 32, 34, 36, 37, 39), 12 contain mental cases, while other conditions found in these 13 pedigrees are epilepsy 1 case, deformity 1, hydrocephalus 1, deaf-mutism 5, lamellar cataract 1, harelip 2. On adding these 13 to the 19 pedigrees in which there is similar inheritance there is a total of 32 out of 40 pedigrees where there is either similar or dissimilar inheritance.

Excluding equivalents there is evidence of heredity in 19 pedigrees. Deducting those showing collateral inheritance and the pedigree with half-brothers affected, 4 out of 40 pedigrees are left which show inheritance, and, in two of these, only the cases in the original childships were seen. In one of the remaining two pedigrees the single individual

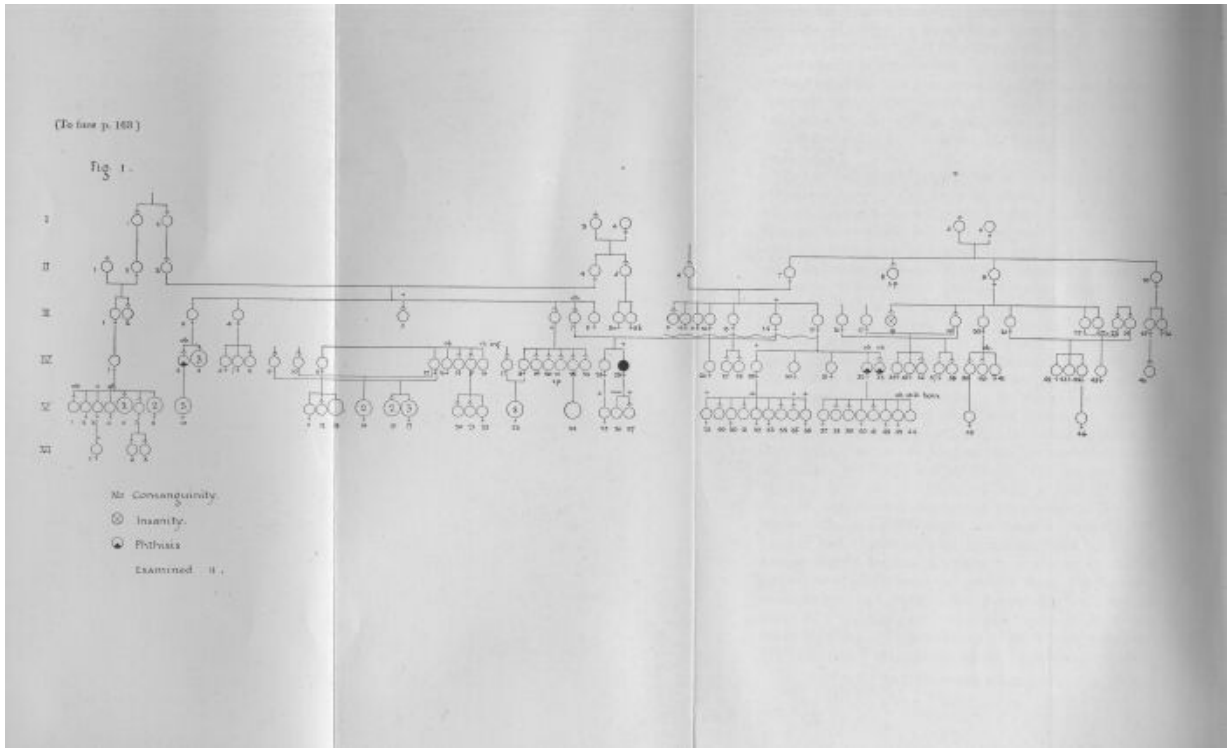
* Bollinger, O., Ueber Vererbung von Krankheiten (Vortrag gehalten in der anthropologischen Gesellschaft zu München am 29th April, 1881). Beiträge zur Biologie als Festgabe dem Anatomen und Physiologen, Th. L. W. von Bischoff.

† R. L. O. H. Reports, 1907, vol. xvii, pt. I, p. 5.

outside the original childship gave a Wassermann positive reaction. If objection is made to accepting it a single pedigree remains in which retinitis pigmentosa was found on ophthalmoscopic examination in individuals outside the original childship. In this remaining pedigree two outside childships each contain a case of retinitis pigmentosa. In one, some of the siblings are undoubtedly subjects of inherited syphilis, in the other, no Wassermann test was employed.

In the 40 pedigrees, in addition to 69 examined cases, 9 others are accepted, this gives an average of 1·95 cases of retinitis pigmentosa in each pedigree ; deducting the 21 single case pedigrees there is 3 per pedigree. On comparing with Mr. Nettleship's data, which are derived from his own cases and those collected from the literature and other sources, a lower average per pedigree is found in his records, for in his 976 families (pedigrees) 1,681 persons were known to be affected,* or 1·72 per family. Mr. Nettleship has kindly told me there are about 250 single case families in his records, and on deducting these there are 726 families with 1,431 cases of retinitis pigmentosa, giving an average of 1·99 per family. In his own cases, 106 families, the total number of retinitis pigmentosa cases is 216, with an average of 2 per family. On deducting 50 single case families there are 56 families with 166 cases of retinitis pigmentosa, an average of 3 per family. Of other cases than his own there are 870 families, with a total of 1,465 retinitis pigmentosa cases, giving an average of 1·7 per family. Deducting 200 single case families there are 670 families with 1,265 retinitis pigmentosa cases, giving an average of 1·88 per family. There is then a much smaller proportion of single case pedigrees in those 976 families, about 25 per cent., than in Mr. Nettleship's own cases, in which 47·1 per cent. are single case pedigrees, or in my series, in which 52·5 per cent. are single case pedigrees. The discrepancy may be explained by selection of cases

* R. L. O. H. Reports, vol. xvii, pt. I, p. 2.


(To face p. 168)
Fig. 1.
Consanguinity.
Insanity.
Phthisis.
Examined.

for publication. The single cases are more likely to remain unrecorded than those where several are affected in a family. If this be true, then the hereditary tendency of retinitis pigmentosa may not be so great as has been supposed.

FIG. 1.—An isolated case of retinitis pigmentosa in a childship of two. IV, 25 (M. C. or Mrs. M.), female, age 30 (1911), seen, a widow, quite intelligent, could never see so well as others in dim light, was never strong, but has had no illnesses or bleedings; hearing excellent, each ear distinguishes a feebly ticking watch at several feet; smell good, tested with cloves, peppermint, assafœtida, and iodoform; iris blue; fundus: retinal arteries narrow, O.D. pale and of waxy appearance, moss-like retinal pigmentation at periphery chiefly, though none seen at extreme periphery, choroidal vessels conspicuous, posterior cortical opacities, refraction R.H. 1·75 D. in vertical meridian and 2·5 D. in horizontal meridian, V. with correction = 6/12, L.H. 1·75 D. in vertical meridian and 2·75 D. in horizontal meridian, V. with correction = 6/12 partly, in dim light that allows 6/9 partly to be seen she gets 6/36 partly; fields of vision much contracted, R. is about 12°, except at outer part where it reaches 40° at one point, L. is 10°, except at outer part where it reaches 34° at one point; in 1898 both fields were very much larger; Wassermann reaction is negative; when seen 13 years ago (March 7, 1898), then unmarried, night-blindness was the same as to-day. She saw 6/36 partly in light that gave 6/9 partly to me, fundus note is as follows: R. and L. retinal pigmentation of usual but rather coarser type all round periphery of retina, in the pigmented area choroidal vessels are much more clearly seen than in other parts, O.D. too pale, edge has a blurred white appearance, retinal vessels markedly diminished in size, no white or yellow spots in retina, media clear. Mother (III, 14) 2nd youngest in sibship of seven, married at 30, had no miscarriages, fundus normal. Father (III, 7) 2nd youngest in sibship of six, married at 42, and *ob.* 70 of "fatty heart," had good vision and no night-blindness. II, 6, an only son and his wife, II, 7, had good vision, so had I, 5 and I, 6, II, 3 and II, 4, had good vision. V, 27, only child of IV, 25, fundus normal; V, 26, miscarriage. Information as regards I, 1

and 2, is from III, 2, who did not know of any of his relatives with defective sight.

IV, 24, sees well. V, 25, age 15, fundus normal. III, 9, *ob.* young. III, 10 and 11, unmarried, all in sibship III, 9 to 15, had good vision and none deaf. III, 8, unmarried. III, 3 to 8, all saw well. III, 5, is alive and knows of no visual defect in his relatives. II, 1, married at 19 and II, 2, at 30 and had two children (III, 1 and 2). III, 2, unmarried, and III, 1, both with good vision, the latter could thread a needle in old age, *ob.* 83. IV, 1, an only child, married at 18 and had nine children (V, 1–8) V, 1, *ob.* V, 3, no night-blindness, L.O.D. pale, arteries narrow, lamina cribrosa exposed, R. fundus normal, defect of L. came on suddenly, or at least rapidly, there is no retinal pigmentation, she has one child (VI, 1) age 15, fundus normal. V, 4, was very "short-sighted," *ob.* after 20. V, 7, has two children (VI, 2 and 3) aged 4 and 5. V, 8, two females, one wears spectacles. II, 5, has two daughters alive (III, 8*a* and 8*b*). II, 8, married, no children. II, 10, had two children, III, 26 and III, 25 with one son (IV, 46) abroad. Of the descendants of II, 9, III, 18 "went wrong in his mind," was in an asylum, unmarried. III, 19, married twice, had three children (IV, 34 to 36) by 1st wife (III, 17), and two children (IV, 37–38) by 2nd wife (III, 16). IV, 40, *ob.* young. IV, 39, had one child (V, 45). III, 21 and 22, married brothers, III, 23 and 24. IV, 42–45 and V, 46, said to have had good vision. III, 22*a*, died unmarried. IV, 2, *ob.* phthisis, had three daughters (V, 10). IV, 6 to 8, abroad. IV, 11, has a daughter (V, 11), and a son (V, 12), also some other children (V, 13). IV, 12, married twice, had two daughters (V, 14) by 1st husband (IV, 10), and five children (V, 15–17) by 2nd husband (IV, 9), none have defective vision. IV, 13, *ob.* 23. IV, 16, *ob.* infancy. IV, 15, has three young children (V, 20 to 22). IV, 18, married IV, 17, abroad, had eight daughters (V, 23), ages 20 to 6, one wears glasses. IV, 19, and IV, 20, unmarried. IV, 21, married, has no child. IV, 22, has large family (V, 24). IV, 23, unmarried. IV, 26, unmarried. IV, 27 and 28, see well. IV, 29, fundus normal, and all her children in sibship V, 28 to 36, hear and see well. V, 28, 35, and 36, fundus normal. V, 32, *ob.* age 6. IV, 30 and 31, unmarried. IV, 33, *ob.* phthisis, age 21. IV, 32, *ob.* phthisis,

has eight children (V, 37, age 14, to V, 44), all well known to IV, 29, see well, none deaf, no peculiarity. V, 44, stillborn. V, 43, *ob.*

FIG. 2.—One case of retinitis pigmentosa in a childship of three. IV, 3, a badly nourished and feeble female, never strong, no definite illness, face and teeth not suggestive of inherited syphilis, is very deaf, married, has no children, age 37 (1910), seen. Wassermann test negative. Vision was defective at age of 16; great difficulty in getting about in dim light. In both eyes much retinal pigmentation, less extensive at extreme peripheral parts than nearer the optic disc; retinal arteries narrow; pale optic discs; no atrophic white choroidal patches; posterior lens opacities; marked night-blindness; fields of vision contracted to about 10°; no vision found outside this, *i.e.*, no evidence of ring scotoma; refraction R. simple myopic astigmatism 3 D., V. with correction = 6/24; L. my. 2 D. in horizontal meridian and 4 D. in vertical meridian; V. with correction = 6/18. Mother (III, 4), *ob.* 54 of cancer. Father (III, 1), a 1st cousin of his wife, *ob.* 27 of phthisis. All his brothers (III, 2) *ob.* phthisis. II, 4 and II, 7 lived long; vision always good. I, 3 and I, 4, vision not affected. I, 2, died early. II, 2, was not night-blind. I, 1 and II, 1, are not known to have had any defect. III, 8, has gross symmetrical choroido-retinitis and night-blindness, probably syphilitic. IV, 21, still-born. IV, 20, *ob.* 19 days old. IV, 16–19 and V, 7–10, see well. III, 9, died young. All others in childship III, 3–9, had good vision, and none of their descendants (IV, 4–15 and V, 3–6) are known to have any defect. IV, 4, two females; one died age 6. III, 11–12 and IV, 23, are large families; no particulars. II, 5, 6, and 8, lived long and had good vision to the last. IV, 1, unmarried; drowned. IV, 2, drowned; had good sight and his children (V, 1, 2) see well.

(No note kept of number of normals seen.)

FIG. 3.—One case of retinitis pigmentosa, and possibly three cases, in a single childship, and one case of congenital ptosis. III, 23, female, married, age 30 (1897) when first seen; has always had bad sight; vision has got gradually worse; night-

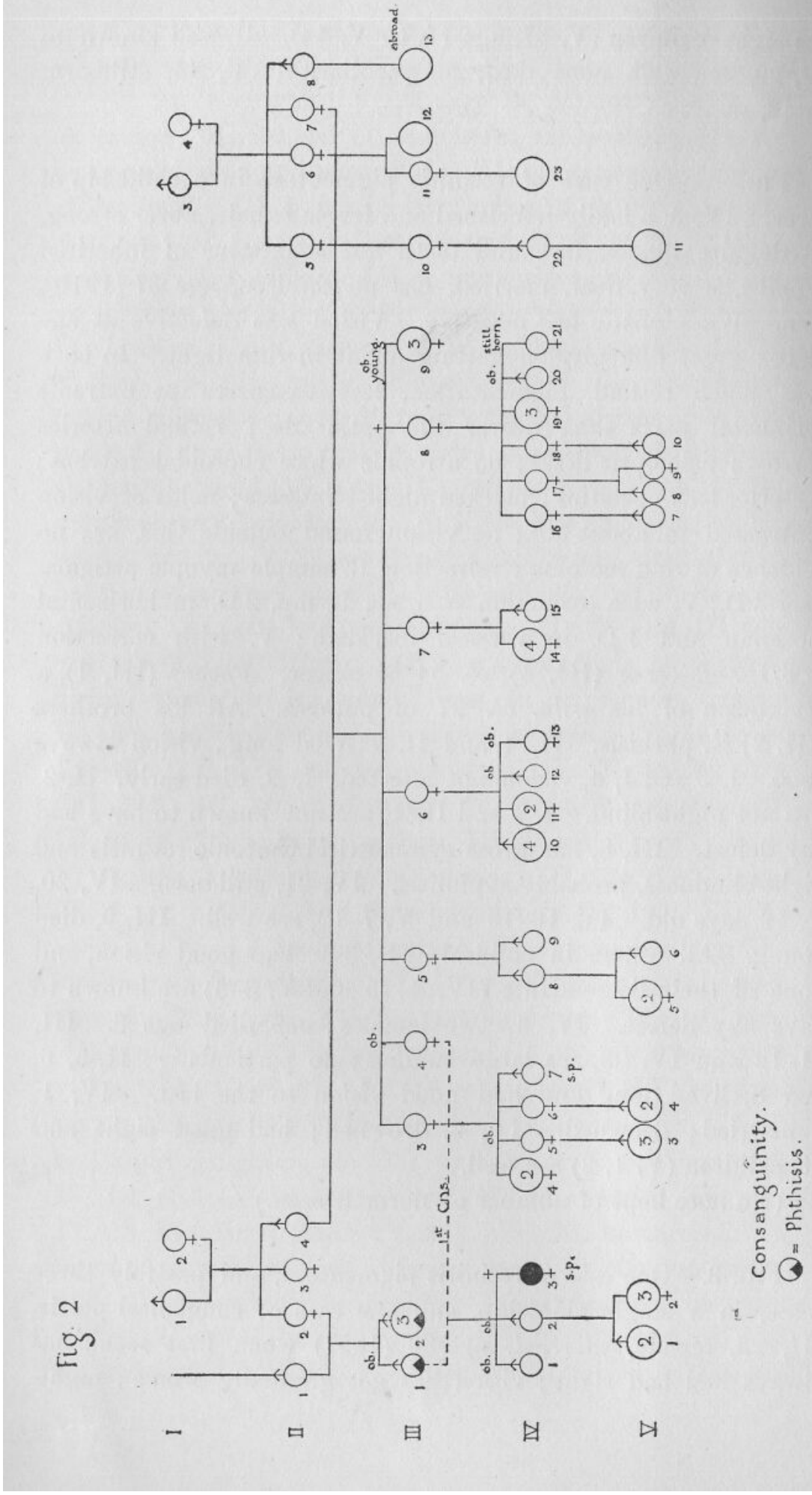
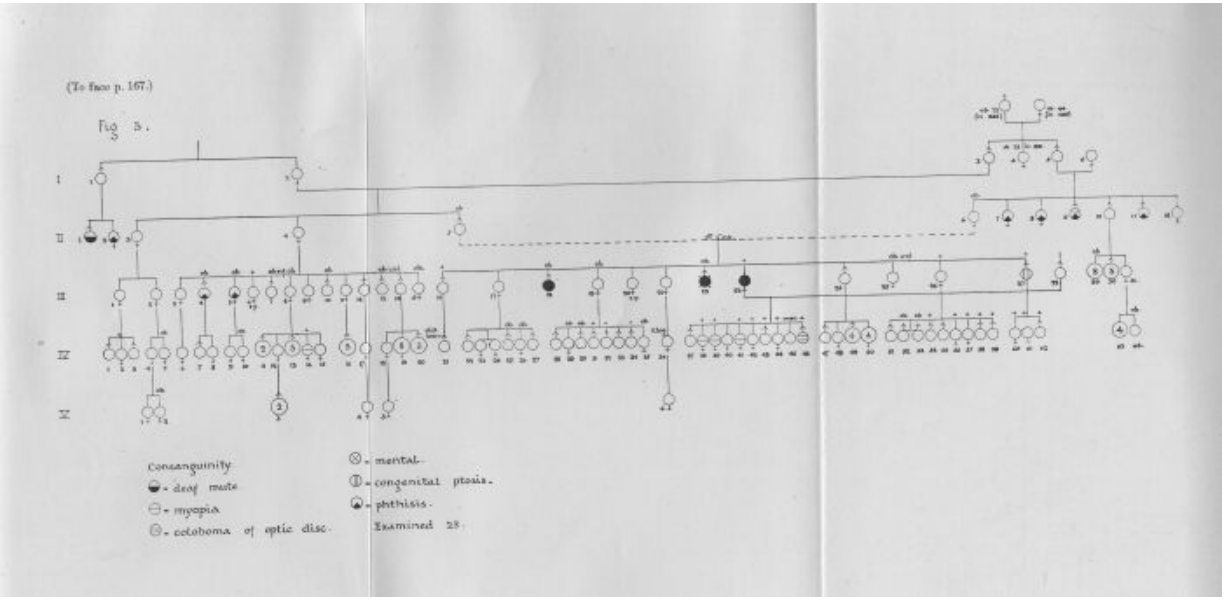

I
II
III
IV
V
1st Cous.
ob.
s.p.
abroad
still born.
young.
Consanguinity.
= Phthisis.

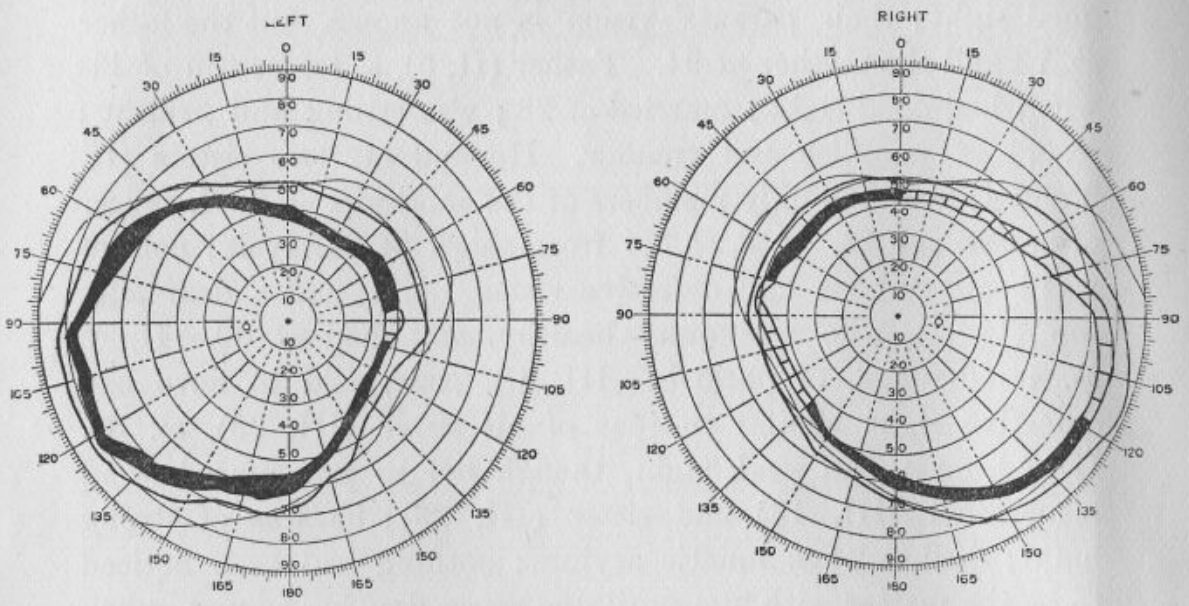
Fig. 2


(To face p. 167.)
Fig 5.
I
II
III
IV
V
Consanguinity
- deaf mute
- myopia
- coloboma of optic disc
- mental
- congenital ptosis
- phthisis
Examined 28

blindness; no deafness; diagnosis of retinitis pigmentosa made at this date. When seen again in 1912 R.V. = 1/60, L.V. = 2/60; no myopia; fields of vision very much contracted; appearance of each fundus that of typical retinitis pigmentosa; posterior cortical lens opacities. Wassermann test negative. No evidence of syphilis. She is a gloomy individual; no illnesses, no bleedings. Mother (II, 6) had good vision; married about 23; had no miscarriages; was strong and healthy; died of paralysis aged 82. I, 5, *ob.* 72 and I, 6, *ob.* 77; both healthy and had good sight; their parents' vision is not known, but the father *ob.* 73 and the mother at 94. Father (II, 5), a 1st cousin of his wife; had good sight; married at 28; was strong and healthy; *ob.* 71 of jaundice and tumour. He and his two sisters (II, 3 and 4) were the only members of the childship. I, 2, *ob.* cancer of breast, age 43. I, 3, *ob.* 74 from shock of paralysis; neither was heard to have had defective vision. I, 1, had a deaf-mute son (II, 1), who was quite healthy, and died at 80. II, 2, *ob.* middle age, of phthisis. III, 18, male, said to have had defective sight similar to that of his sister (III, 23), *ob.* 30. III, 22, male, had weak sight, though not so pronounced as in his brother (III, 18) and sister (III, 23); he was of strong build; died at 24 in lunatic asylum; nothing had been noticed to be the matter with him until one warm day in August, when working in the hayfield, he suddenly complained of pain in his head, and all at once became insane, and had to be removed to an asylum, where he died a few days afterwards. III, 16, had still-born child (IV, 21). III, 17, has some nose trouble; is abroad with her children (IV, 22–27); all see well. III, 19, *ob.* 37 of appendicitis; had good vision. IV, 28–35, all had good vision. IV, 29 and 30, *ob.* young. III, 20, not strong, sight good, married, has no child. III, 21, healthy, but has nasal trouble, unmarried, has a daughter (IV, 36), who has a child (V, 6). III, 24, not very strong, sees well, also his children (IV, 47–50), their ages are 19 and younger. III, 25, *ob.* infancy. III, 26, has ozæna and depressed bridge of nose, has had these for as long as she remembers, her children (IV, 51, 54–59) see well and are healthy. IV, 52, 53, male twins, *ob.* two weeks. III, 27, has congenital (partial) ptosis on right side, he and his children (IV, 60–62) have normal fundi. II, 7,

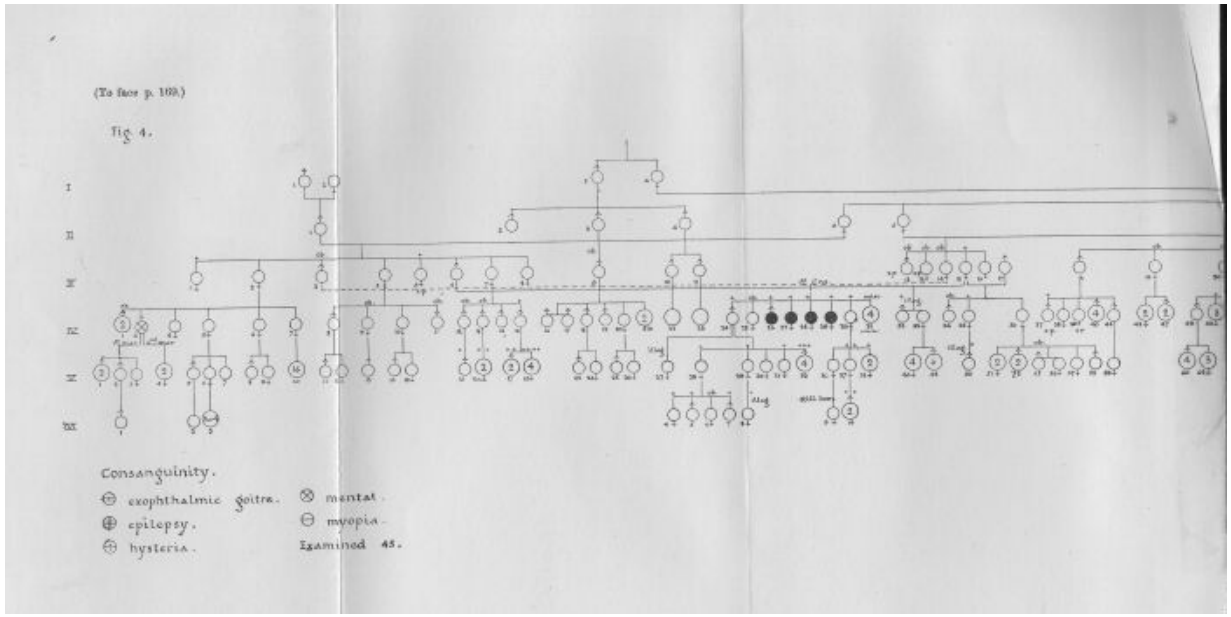
ob. 36 and II, 8, *ob.* 37, both of phthisis. II, 9, mentally deficient, *ob.* phthisis. II, 10, *ob.* 78, vision normal, eight of his children (III, 29), most were young, died during an epidemic of diphtheria. III, 30 and 31, and IV, 63, healthy. II, 11, *ob.* 36 of phthisis. II, 12, *ob.* after 70, vision normal. IV, 37 and 40, see well. IV, 38,* age 21, high myopia.

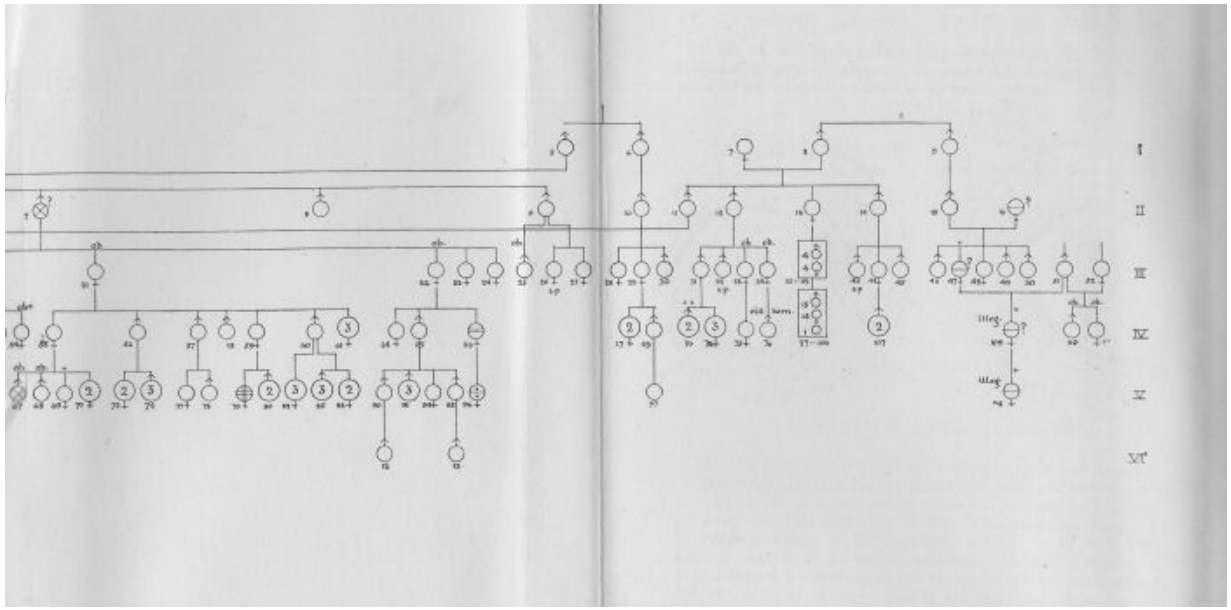
* IV, 38, seen again in June, 1913, at age of 22; fields taken without correction of error of refraction are nearly full, complete ring scotoma in each (see figure below), a narrow seeing area is left between it and periphery



Fields of IV, 38, in Pedigree 3, June, 1913.

of field; in right field the scotoma is absolute up in and down out, and relative up out and down in; in left field the scotoma is absolute, except at inner part at 90°; a 10 mm. white square was employed in good daylight; refraction same as in 1906. My. R. 10 D. in horizontal meridian, and 13 D. in vertical meridian. V. with correction = 6/12; L. 13 D. in horizontal meridian, and 16 D. in vertical meridian. V. with correction = 6/12. Night-blindness is not marked, but she and a younger sister say that she (IV, 38) cannot go into some places in dim light that others can. In dark room where normal people read 6/18 she could read some letters of 6/24; she takes a long time to get accustomed to the dark after coming out of the light. Oph.: R. and L. O.D. quite good colour; retinal arteries of full size; posterior staphyloma with complete choroidal atrophy; with the exception of a small piece of pigment at outer part of right retina, a spot merely and not bone-corpuscle like, no retinal pigment is present. This was searched for with Ps. dilated after homatropine. Has cold hands. Wassermann test positive. At age of 6 (1897) refraction of R. was My. 8 D. in one meridian, and 11·5 in opposite meridian; L. My. 8 D. in one, and 11 in opposite meridian. Atropine was used.


(To face p. 169.)
Fig. 4.
I
II
III
IV
V
VI
Consanguinity.
exophthalmic goitre.
epilepsy.
hysteria.
mental.
myopia.
Examined 45.



IV, 39* and 41,† myopic. Field of vision of IV, 39, measured on perimeter, is quite full. IV, 42–44, fundus normal. IV, 46, right fundus normal; left optic disc twice as large as right one, its outer part is pale and cupped, rest of it is normal (coloboma of optic disc), rest of fundus normal. Descendants of II, 3 (III, 1, 2; IV, 1–4; V, 1), are healthy and see well. IV, 2, pupils markedly eccentric inwards. Some in childship III, 3–15, *ob.* young, none deaf or night-blind. III, 4 and 5, *ob.* phthisis, III, 7 and 13, *ob.* infancy. III, 8, *ob.* cancer. Abroad IV, 7–9, 13, 17, and V, 4. IV, 11, age over 30. IV, 12, and her sons (V, 3) unaffected. IV, 14, has myopic astigmatism. IV, 15, good vision. IV, 16, three males with good vision. IV, 18, and child (V, 5) vision normal. IV, 19 and 20, all see well, none married. III, 12, IV, 17 and V, 4, normal vision.

FIG. 4.—Four cases of retinitis pigmentosa in one childship. IV, 26, male, unmarried, age 57 (1912), seen; vision very gradually failed until he became quite blind; saw sufficiently well for schooling; had night-blindness. R. and L. V. = no p.l.; in right eye retinal pigmentation can be seen, but in left eye fundus is not visible owing to complete cataract; congenital coloboma of iris in left eye (uniocular); Wassermann test negative; he and all others in childship, except IV, 28, had scarlet fever in childhood; none are deaf. IV, 27, female, unmarried, age 41 (1898), seen; *ob.* 1911, was never strong; night-blind; vision gradually failed; saw well enough for schooling; slow vertical nystagmus; refraction myopic; V. R. and L. = no p.l.; retinal vessels, especially arteries, extremely narrow; much pigmentation at periphery of retina of a moss-like pattern; O.D. pale yellow-red colour, edge not sharply defined, lamina cribrosa not exposed. IV, 28, female, unmarried, age 37 (1897), seen; had "gastric fever" at 13; vision began to fail before that time; R. and L. V. = p.l.; slow lateral nystagmus; posterior cortical lens opacities; choroidal vessels much exposed; very extensive retinal pigmentation which nearly reaches O.D.; retinal vessels very narrow. When seen

* IV, 39, in June, 1913. L., no ring scotoma found in left field, which is full; refraction My. R. 3 D.; L. 4·5 D.

† In June, 1913, My. not more than 1·5 D. in each.

again in 1912 V. R. and L. = no p.l.; much lens opacity; much retinal pigmentation, but no details of O.D. or retinal vessels could be seen. IV, 29, female, unmarried, seen 1912. V. R. and L. = no p.l.; much lens opacity; typical retinal pigmentation visible, but no details of O.D. and retinal vessels; vision had gradually failed; went to school; Wassermann test negative. IV, 25, *ob.* 8 years; saw and heard well. IV, 31; four miscarriages; order in which they came in relation to siblings is not known. IV, 24, a healthy farmer with normal fundus. All his descendants (V, 27–32, and VI, 4–8) have good sight. His 1st born (V, 27) is illegitimate. V, 31, a healthy looking woman though somewhat peculiar and possibly neurotic, age 18, said on being questioned that for two years she had found some difficulty in seeing at dusk. She had never told this to her parents, brothers, or sisters, and they had never noticed that she had any difficulty. At periphery of one fundus there is at least one piece of retinal pigment; O.D.'s good colour; retinal arteries not contracted; fields tested by hands and also by a 10 mm. square of white paper; no contraction of either field was found or a ring scotoma; no diminution in her light sense was found when tested with Snellen's types in a darkened room and by means of small objects placed on the floor. A second examination at an interval of some months revealed nothing more. At parts of periphery of each fundus is an indefinite yellowish dappled appearance behind retinal vessels. There is not sufficient to base a diagnosis of commencing retinitis pigmentosa. Both examinations made in the country without a perimeter. IV, 30, healthy married woman; fundus normal; her children (V, 36–38) and VI, 10, have good vision; one of the latter an infant in which fundus examination was very imperfect. IV, 24–30, live at a small farm in a rather remote part of the country. Mother (III, 14) had good vision; married at 25; *ob.* bronchial affection, aged 86. Father (III, 3), first cousin of his wife, had good sight; married at 25; *ob.* 84; bronchial affection is his doctor's report. II, 6, and II, 11, not cousins; the former *ob.* 92. II, 1, good vision; not deaf. II, 5, born 1795; *ob.* aged 90. I, 1, I, 2, I, 4, and I, 5, all had good vision and were known to III, 1, except I, 5. I, 7, and I, 8, saw well. III, 1, aged 90, unmarried; retentive

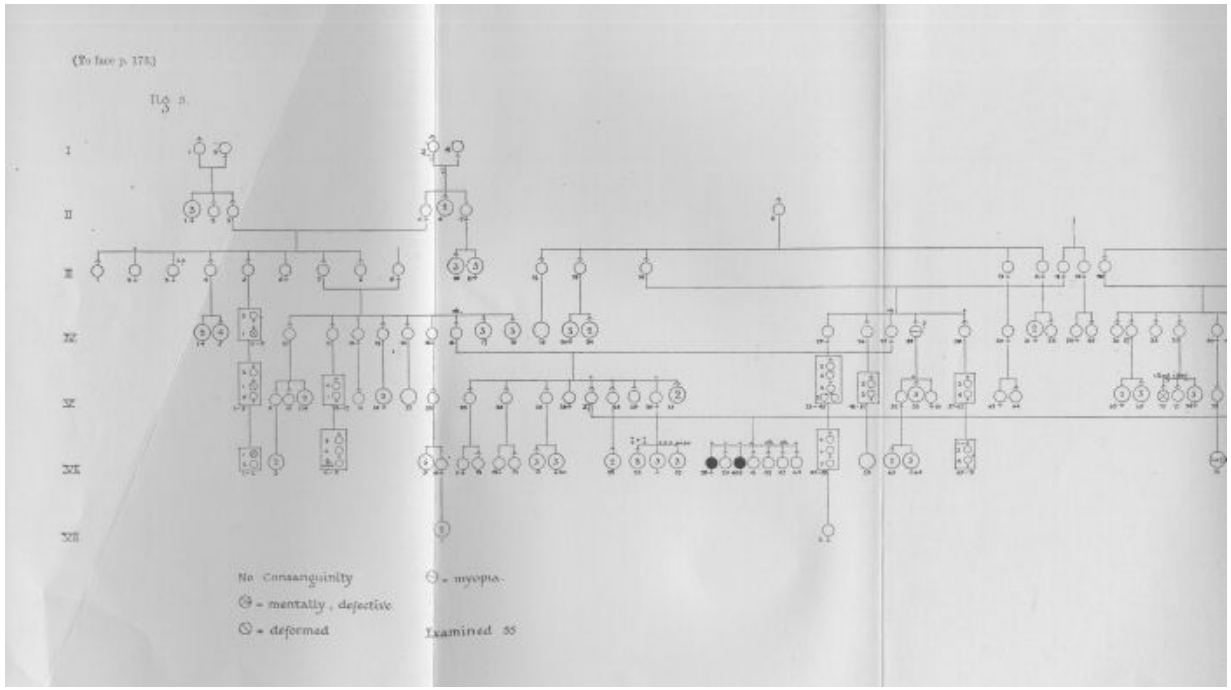
memory; fundus normal. III, 5, married; no issue; fundus normal. None in childship III, 1–8, had defective vision. III, 15, age 80; fundus normal; had illegitimate twins (IV, 32–33) by III, 6. She then married III, 17, and had three children (IV, 34–36). III, 12 and 13, married; no issue. III, 16, unmarried. IV, 32, strong healthy man, unmarried; fundus normal. His twin sister (IV, 33) and her children (V, 40 and 44) have good vision. IV, 34, *ob.* young. IV, 35, very nervous and refused fundus examination; her sight is good. Her illegitimate son (V, 50) is well developed; fundus normal. IV, 36, and her children (V, 51–58) all unmarried, see well. Many people in this pedigree are well known to IV, 37, and his brother, both medical men, and to the doctor of IV, 24–30, but they do not know of any others with night-blindness. III, 18, is alive. II, 7, was a "character"; went about in old fashioned costume, and was regarded as eccentric. Information about descendants of I, 3, is reliable. None had defective sight suggesting retinitis pigmentosa. III, 9, became blind, *ob.* 68, of Bright's disease; vision began to fail between 50 and 60; was treated for tobacco amblyopia. V. 67, mental, *ob.* V. 79, an epileptic; others from V, 59–95, normal. IV, 3, had violent temper which was attributed to insanity; latterly he became alcoholic. He married twice and had families (V, 1–4) that are reported with VI, 1–3, to see well. Others in childship IV, 1–7, and their descendants unaffected. IV, 50, one is *ob.*, one has normal fundus, and the 3rd has good vision. II, 9, married twice. His children (III, 25–27) had no issue. III, 18–24, none with defective vision or hearing. V, 97, a family abroad. The 4th born in childship V, 17–19, a female, age about 11, no evidence of hereditary syphilis, has normal fundi (O.D. good colour, retinal arteries of normal size) with the exception of a roughly circular, dark, nearly homogeneous patch several times the size of O.D., situated towards the periphery external to macula in left eye. Edge of this area is moderately well defined. The appearance suggests a patch from a very dark fundus let into a normally pigmented fundus. At its upper part only is the uniform darkness slightly interrupted by indications of darker and lighter lines which probably indicate the position of choroidal vessels. A single conspicuous small white spot is

situated on the area below its centre. It does not resemble an atrophic spot in choroid. The five others in the childship have normal fundi and so has their father (IV, 15), who has mixed astigmatism. V, 16, fundus normal. Part of the account of descendants of I, 8, and I, 9, was supplied by the wife of III, 46, aged 86, who has absolute glaucoma, and by the wife of III, 31. V, 98, illegitimate, age 25, has high myopic astigmatism. R. 10 D. in one meridian and 14 D. in opposite meridian, V. = 6/24 with correction. L. My. 15 D. in one meridian and 17 D. in opposite meridian, with correction V. 6/18 partly ; lenses clear ; posterior staphyloma in each ; the choroidal atrophy surrounds optic discs, and in left eye partial choroidal atrophy extends from optic disc outwards above yellow spot ; refraction and fundus examined after using a mydriatic ; no night-blindness. Her mother (IV, 109), illegitimate, had bad sight since school age ; could never read well ; no night-blindness ; never had her eyes examined ; age about 55 ; V. = feeble p.l. in each ; projection defective ; T.n. ; lens opacities mostly diffuse, some broad striæ ; fundus not visible ; no evidence of iritis. III, 47, age 80 ; V = p.l. in each with strong light ; projection faulty ; T.n. ; fundus no examination possible owing to lens opacities ; no history of night-blindness, was always shortsighted ; none of her brothers or sister (III, 46, 48–50) had defective vision, but her mother (II, 16) was shortsighted, her sight never got very bad. II, 15, not affected. It seems probable that the defect of vision in IV, 109, and III, 47, has been due to fundus changes in eyes with high myopia, and that it cannot be attributed to retinitis pigmentosa.

No defect in descendants of I, 3, I, 6, and II, 12–14. II, 2, and the twins IV, 16–17, are unmarried. II, 8, *ob.* unmarried. Children (IV, 110, 111) of III, 51, and III, 52, both *ob.* unmarried. IV, 37–66, all have good vision. IV, 66, a female, has exophthalmic goitre. Her daughter (V, 96) has hysteria. VI, 12 and 13, unaffected.

All deaths in Generations III, IV, and V have not been indicated on the chart.

Married and without issue : III, 5, 12, 13, 26, 32, 43, 35 (a male). IV, 37, 39.


(To face p. 173)
Fig 3.
I
II
III
IV
V
VI
VII
No Consanguinity
= mentally defective
= deformed
= myopia
Examined 55

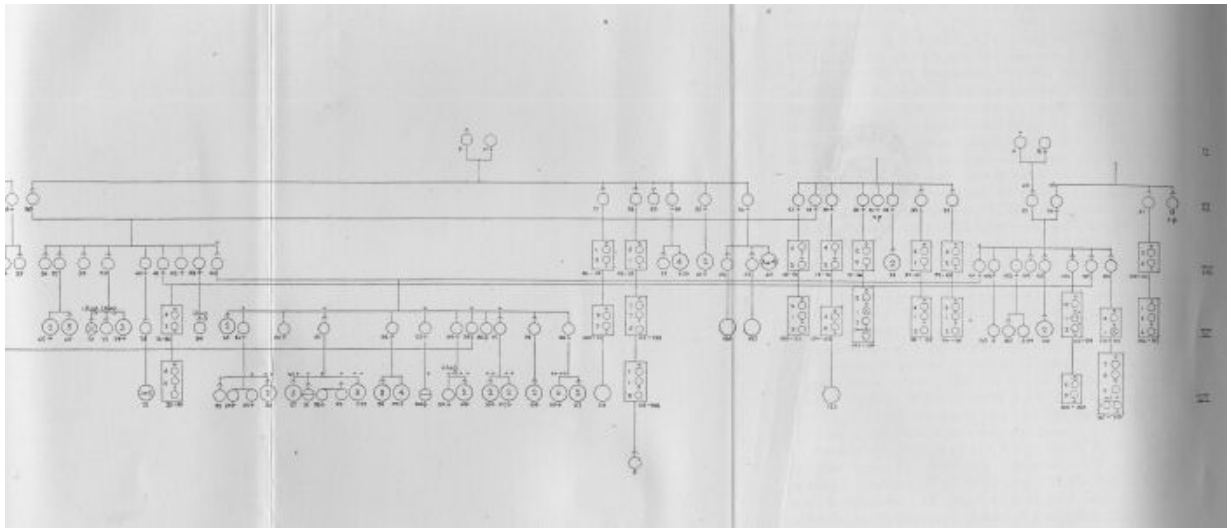
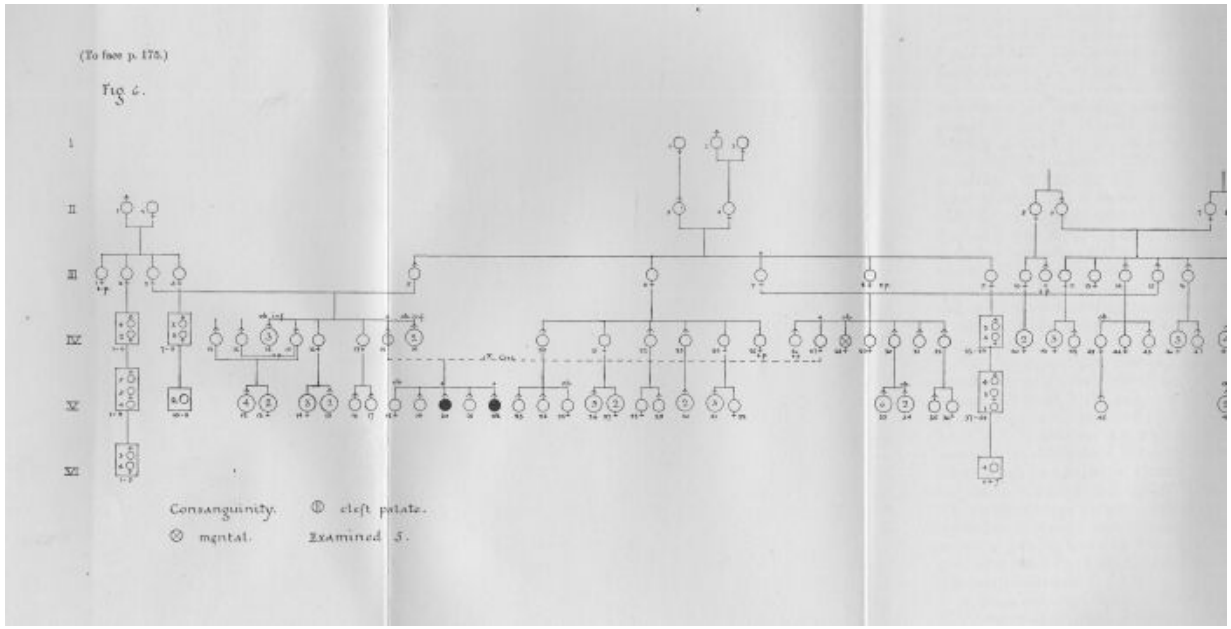
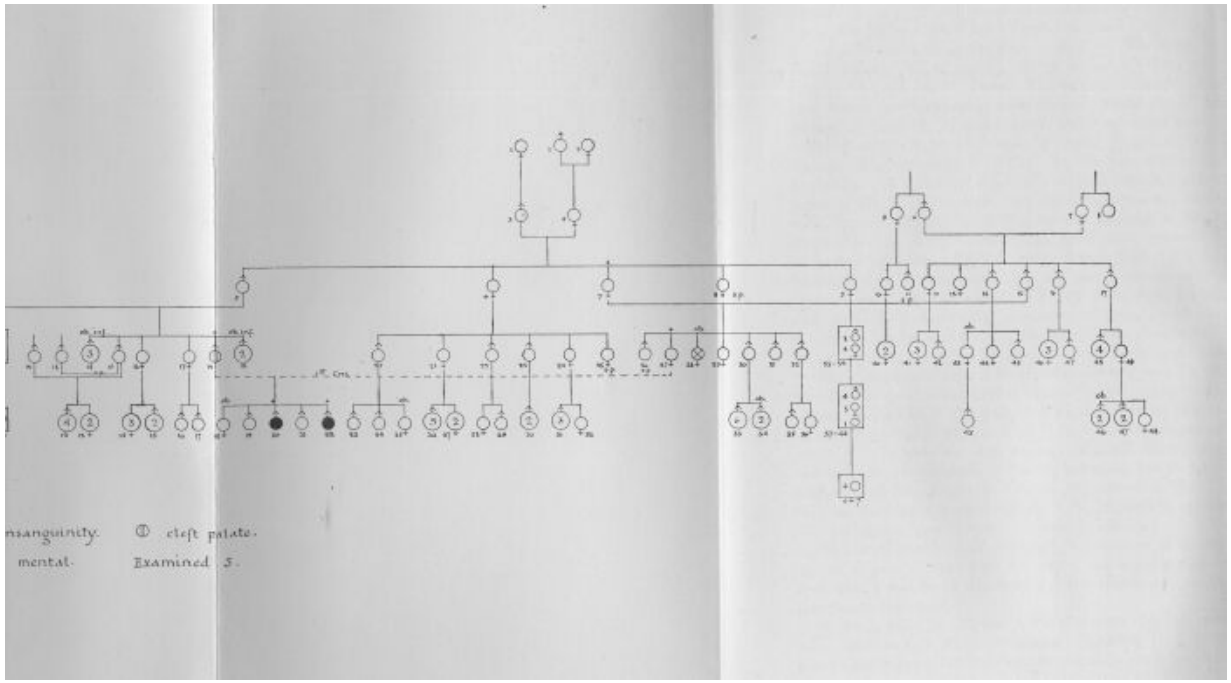


FIG. 5.—Two cases of retinitis pigmentosa in a childship of seven. VI, 38, female, age 18 (1911) seen, has typical appearance of retinitis pigmentosa in each eye; Wassermann test negative; hearing excellent; never had any serious illness; night-blindness noticed as soon as she could move about, as she could not pick up anything in dim light; refraction H.R. 0·5 D. in oblique meridian, 1·5 D. in opposite meridian, V. with correction = 6/18; L. 1 D., V. with correction = 6/18; small posterior cataracts; pale waxy discs; very narrow retinal arteries; choroidal vessels much exposed; much moss-like retinal pigmentation at periphery extending far towards macula and O.D.; seen first in 1906 on account of difficulty in seeing, aching of eyes and headaches; both fields of vision much contracted, about 10° in all directions. VI, 40, female, age 14 (1911) seen, no deafness, bronchitis and inflammation of lungs at age of 3, defect of vision noticed after that, has marked night-blindness; oph. (August 17th, 1910) retinal arteries very narrow; choroidal vessels much exposed; considerable amount of retinal pigmentation in right eye now, but much more in left eye, no white spots; on January 25th, 1906, there was retinal pigmentation, but only at periphery of left fundus, none in right; retinal arteries very narrow; O.D. grey and semi-translucent, edge blurred; choroidal vessels exposed except at macular region; fields of vision much contracted, left, more than right, is reduced to about 10°; right field in lower temporal part is larger extending to 42° on temporal side and has a scotoma continuous with blind spot, elsewhere 10°; refraction My. R. 1·5 D. in oblique meridian, 4 D. in opposite meridian, V. with correction = 6/36, L. 1·5 D. in oblique meridian, 4 D. in opposite meridian, V. with correction = 6/36; in light reduced to give normal eyes some letters of 6/9 she cannot see the board. Mother (V, 95), age 42, married at 22, fundus normal. Father (V, 27), married at 22, fundus normal, was 5th born of 10; his wife (V, 95), not a cousin, was the 9th born of 13. IV, 101, age 75, fundus normal. IV, 44, age 79, health excellent, fundus normal. IV, 27, *ob.*, saw well. IV, 16, 3rd youngest in childship, rather deaf after an accident, *ob.* from inflammation of bowels, age 52. III, 36, *ob.* 80. III, 35, *ob.* 84. II, 12 and II, 11, considered to be unrelated, nothing known as to vision of II, 11. II, 10 and II, 9, seen by IV, 44; II, 9,

had particularly good vision, was born in France, but parents were Scottish. III, 28, *ob.* 75. III, 20, 1st born, *ob.* 66. III, 18 and III, 14, seen by V, 30. III, 18, *ob.* 63. II, 8, good sight, *ob.* 93. III, 9, only child, parents not known to III, 2. III, 7, vision normal. II, 4, *ob.* 99, had use of all senses until one week before death; her husband (II, 3) not a cousin. I, 1 and 2 and I, 3 and 4 (informant III, 2), had good vision. I, 3, *ob.* 88. VI, 39, age 16, fundus normal. VI, 41, age 11, fundus normal. VI, 42, *ob.* 11 months. VI, 43, *ob.* 7 weeks. VI, 44, age 1 day, fundus examination not sufficiently satisfactory. *Mental defect* was present in five: IV, 9, could never walk, was silly, and an "object," *ob.* 21. V, 70, illegitimate, is "soft." V, 147, is "soft," his maternal grandmother is III, 30. V, 181, very deaf, was in an asylum. VI, 1, "silly," this child's mother's maternal grandfather is III, 5. V, 3, male, "an object" (deformed), cannot walk, but not silly, is the child of another daughter of III, 5. *Deafness* present in V, 4, female, deaf from birth, speaks only in broken sentences. VI, 23, male, always deaf. VI, 84, female, age 18, a twin, rather deaf, has chorea, catamenia first appeared at 17; fundus normal. VI, 93, male, twin, slightly deaf, hears the feeble tick of a watch at 1 inch in each ear; is delicate, fundus normal. VI, 12, male, "deaf from throat." VI, 97, female, deaf for 2 years, worse lately and with "colds," tonsils enlarged; a single pigment spot seen in left retina in front of vessel, rest of fundus normal; right fundus normal. *Myopia* in V, 170, male, and VI, 100, female. VI, 91, male, R. simple my. astigmatism 2·5 D., L. simple H. astigmatism 2·5 D. IV, 28, male, not seen, but his brother (IV, 29) says that he was always short-sighted, it has not got worse, has no night-blindness and uses glasses for distance only, age about 76. IV, 26, "has only one eye short-sighted." IV, 20, female, squints. VI, 97, female, age 9, conv. concom. strabismus; H. 4 D. approximately in each; fundus normal; a precocious child; had attacks of pain in head with flushing of face and watering of eyes, when 9 months of age, lasting a few seconds; upper jaw imperfectly developed.

V, 55, a male, age 25, has a "muslin patch" on left optic disc, otherwise both fundi are normal; refraction H. 7 D. in vertical meridian and 10 D. in horizontal meridian of R. and 9 D.


(To face p. 175.)
Fig. 6.
Consanguinity.
mental.
cleft palate.
Examined 5.


nsanguinity.
mental.
cleft palate.
Examined 5.

and 11 D. in oblique meridians of L. III, 2, unmarried, age 89 (seen), had good sight, now *ob.*, 2nd youngest in childship of 8 (III, 1–8); knew of no defect of sight in her relatives.

Married without issue: Males, III, 38. IV, 2, 4, 5, 56, 78, 86. V, 160. Females, III, 3, 31. IV, 47, 60, 61, 75, 85. V, 50, 51, 59, 39. *Died young:* Males, III, 23. IV, 72, 87. V, 31, 86, 78, 167. VI, 21, 22, 124. Females, IV, 1. V, 26, 90. VI, 26, 47, 83. Sex unknown, V, 41, 45 (4). Illegitimate males, V, 70, 71, 84, 134, 147, 160, 184. Females, V, 135, 142, 143, 146, 183. VI, 101. *Twins* occur five times, V, 41, 42; 43, 44. VI, 83, 84; 92, 93; 146. Many individuals in Generation VI are young and some also in Generation V. No one else marked in the figure besides those already indicated is known to have defective vision. Large open circles indicate families of which there are no particulars.

FIG. 6.—Two cases of retinitis pigmentosa in a childship of five. V, 20, male, unmarried, age 23 (1911) seen, has marked night-blindness, sees well in day time; fields of vision much contracted; retinal arteries narrow; optic discs pale and "waxy"; retinal pigmentation very extensive, it reaches to optic disc; refraction estimated emmetropic; no deafness. V, 22, fisherman, unmarried, age 19 (1911) seen, has night-blindness and vision is getting worse; sight was "always" defective. R. V. = 1/60; L. V. = 4/60, refraction low H. in each; optic discs pale; retinal arteries much narrowed; choroidal vessels exposed; much moss-like retinal pigmentation all over periphery of fundus; no white spots or patches; fields contracted and each has a complete ring scotoma; no deafness; Wassermann test negative. V, 18, *ob.* age 13. V, 19 and 21, unmarried, have no night-blindness and vision is good. Mother (IV, 27), considered peculiar, has normal fundus, married at 26, had no miscarriages, husband a 1st cousin. Father (IV, 18) has a cleft palate, married at 29, fundus normal. III, 7, fundus normal. III, 15, vision good. II, 3 and 4, not cousins, and I, 1, 2, and 3, none had defective sight. II, 6 and 7, vision normal. II, 8, others in childship, no particulars. III, 3, has same name as II, 7, but is said not to be related, she and her husband (III, 5) had normal sight. IV, 28, *ob.* 47, mentally

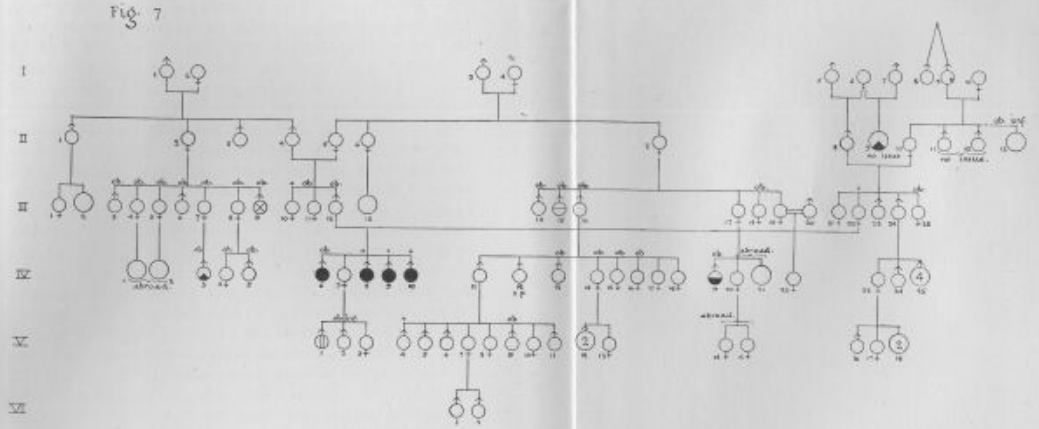
defective, did nothing for herself, never had good sight. IV, 31, fisherman, uses glasses for reading and mending nets, sight defective, getting worse, not deaf, has bad temper, formerly drank.* IV, 32, fisherman, with defective vision, which his doctor says is getting worse, so that he has to be taken to sea by his brothers for his sight is now so bad. IV, 29 and 30, and V, 33 (oldest 17), see well. IV, 15, married twice, had no children by second husband (IV, 13). IV, 15–17, and V, 12–17, see well. II, 1 and 2, not cousins, II, 5, III, 10, 11, and IV, 40, no visual defect. IV, 40, no issue. III, 2 and 4, and their descendants, IV, 1–11, V, 1–11, and VI, 1–5, not affected, but IV, 11, female, no information. V, 10 and 11, two families, numbers not given, vision said to be normal in all. III, 6, and III, 9, with descendants (IV, 20–25, 33–39, V, 23–32, 37–44, VI, 6, 7), unaffected. V, 23, age 23. V, 28–30, quite young. III, 12–17, saw well. III, 13, married abroad. Descendants of III, 12, 14, 16, and 17 (IV, 41–49, V, 45–48) have normal vision. No information as to whether IV, 41 and 42, had families, they saw well. V, 35 and 36, ages 5 and 3. V, 40–43 and 47 are young. Married without issue: Males, III, 11, IV, 2, 7, 9, 26, 33; females, IV, 25, 38, III, 1, 8.

FIG. 7.—Four deaf-mutes with retinitis pigmentosa in a childship of five, a deaf-mute in another childship of same generation, an imbecile in previous generation, and a boy with congenital deformities in next generation. IV, 10, male, unmarried, age 32 (1911), seen with typical retinitis pigmentosa; O.D.'s pale with 3 or 4 translucent bodies on them, probably hyaline bodies; retinal arteries markedly narrowed; retinal pigmentation of usual moss-like character; also pepper-like pigmentation in some places and small yellow spots; constant lateral nystagmus; is able to do his work as postman by holding the letters close to him; night-blindness; complete deafness; deaf-mute; refraction H.; iris, blue. When seen on May 1st, 1900, R. V. = 6/36 partly, and L. V. = 6/36 partly, neither improved by a spherical lens, now (1911) vision is 4/60 in each and not improved; fields much contracted, R. field not charted,

* Since going to press this man has been examined. There is no retinitis pigmentosa. Each eye is myopic.

(To face p. 176.)

Fig. 7



No consanguinity.
deformities, digital.
imbecile.
deaf-mute.
myopia.
phthisis.

field of L. is all within 20° ; has had children's ailments only ; Wassermann test negative. IV, 9, male, unmarried, age 34 (1911) seen, quite deaf and slow mentally, deaf-mute, cannot go about by himself in same way as his brothers, and this is not due to relatively worse vision ; R. and L. each reads type of about J. 2 at a few inches ; typical retinitis pigmentosa, more retinal pigmentation than in last case ; one large raised translucent body on right O.D., probably a hyaline body ; posterior cortical cataracts ; refraction H. R. and L. ; iris blue ; constant lateral nystagmus ; night-blindness ; does odd jobs. IV, 8, male, unmarried, age 36 (1911) seen, saddler, quite deaf ; deaf-mute, typical retinitis pigmentosa ; retinal arteries and veins very narrow ; O.D. pale ; much moss-like retinal pigment ; R. V. = 6/36 partly H.m. 2 D. ; L. V. = 6/60 H.m. 2 D., reads J. 4 with difficulty, with + 4·5 D. reads J. 1 ; night-blindness ; reads without glasses from May to August, requires glasses only in winter months ; fields of vision contracted to about 10° in every direction ; no nystagmus ; iris blue. IV, 6, male, unmarried, drowned in river in 1898, aged 27 ; subject to epileptic fits for seven years previous to death ; was deaf-mute ; night-blind, did odd jobs ; "his sight was a great drawback." In all these cases (IV, 6, 8, 9, and 10) deafness was noticed in very early life, but defect of vision only after they went to school. They never had any severe illness. IV, 7, has good vision, so have her children (V, 1 to 3), but the oldest (V, 1), age 7, has deformed left hand, thumb and forefinger only being complete, the other fingers terminate at the middle joint. V, 2 and 3, are said to be normal. Mother (III, 22) fundus normal, not night-blind. Father (III, 12) *ob.* was not night-blind. II, 8 and 10, good vision. II, 8, an only child of I, 5, an Italian, and I, 6 ; by her second husband (I, 7) I, 6, had a family (II, 9), all *ob.* phthisis, without issue. I, 8, in army. I, 9, 10, no defect. II, 1 and 2, *ob.* in old age. II, 3, *ob.* young. II, 4, *ob.* after 90. I, 1 and 2, not affected. I, 3 and 4, were not "short-sighted" as far as III, 19, knows. II, 5–7, good vision. III, 20, seen. III, 1, and III, 2, several others, all with good vision. III, 3–9, all *ob.* III, 3 and 6, were unmarried. III, 9, imbecile. III, 13, no particulars of this family. III, 14, blacksmith, single, *ob.* 80, not night-blind, wore glasses for reading only, was reported to

be short-sighted. III, 15, short-sighted, no night-blindness, unmarried, drowned. III, 16, *ob.* old. III, 18, *ob.* young. III, 21 and 23, unmarried. III, 25, *ob.* age 2. IV, 1 and 2, are families abroad, no particulars. IV, 3, *ob.* phthisis. IV, 4, *ob.* young. IV, 5, *ob.* 21, diabetes. IV, 12, abroad. IV, 13, drowned in boyhood. IV, 15 and 16, *ob.* young. IV, 17 and 18, single. IV, 19, deaf and dumb, good vision, *ob.* without issue. IV, 20, and V, 14 and 15, abroad. IV, 22, unmarried. III, 17, went to America, no information as regards number or condition of IV, 21. IV, 11, and V, 4–11, are living, except V, 9, *ob.* infancy, and are educated people, including schoolmaster and medical man, vision good in all. V, 6, a sea captain. VI, 1 and 2, young, nothing known to be wrong. V, 12, both in army. V, 13, at school, age 14. V, 16 and 17, IV, 23 to 25, and III, 24, have no defect known to III, 22. (April 25th, 1911.)

FIG. 8.—Three cases of retinitis pigmentosa in a childship of 11. V, 127, male, unmarried, age 19 (1902), seen, comes on account of night-blindness which was noticed by his mother when he was 10 years old; sight is getting worse; occasionally has aching in temporal regions; refraction, My. R. 2 D., V. with correction = 6/18, L. 1·5 D., V. with correction = 6/12; fields of vision contracted; right field, measured on perimeter, reduced to 12°; Ps. small, equal, contract sluggishly to light. R. and L. O.D. fairly good red colour, edge well defined; retinal arteries narrowed; typical fine retinal pigmentation most towards periphery where choroidal vessels become very conspicuous; vitreous opacities, only a few far forwards; hearing defective, right ear hears nothing, left ear watch on contact only; smell and taste normal; heart and lungs normal, also abdomen; he has had no illness. When seen again in 1909 he was a robust man, no mental defect; vision had got worse, fields of vision much contracted. Wassermann test negative. V, 124, male, stone-cutter, unmarried, age 25 (1902), seen, married later but had no issue (1910); is rather deaf; night-blindness; is strong and intelligent; no illness; now in Canada; vision getting weaker; particulars of fundus and fields of this case could not be found. V, 129, female, age 24 approximately (1910), seen, unmarried,

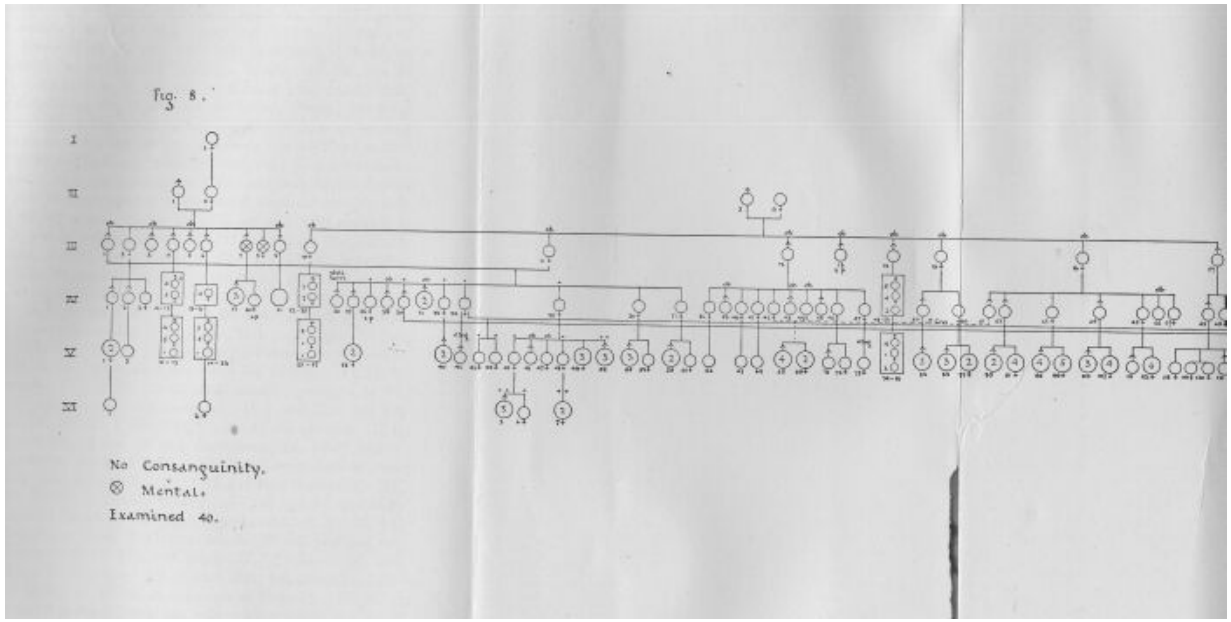
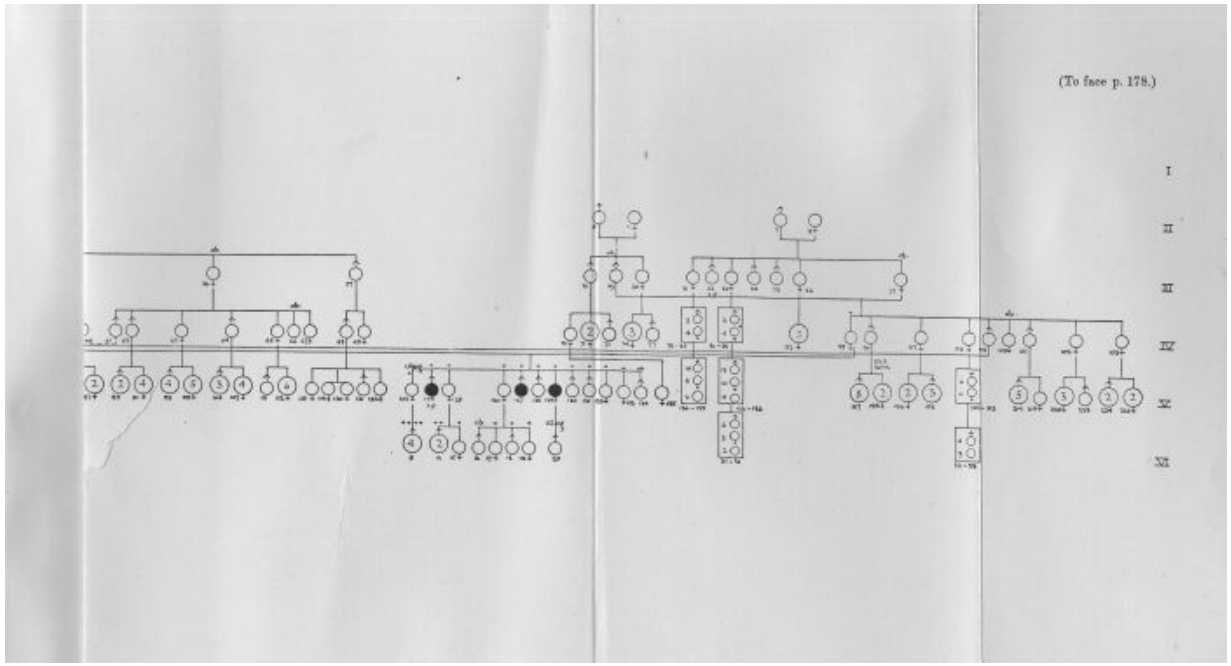


Fig. 8.
I
II
III
IV
V
VI
No Consanguinity.
⊗ Mental.
Examined 40.


(To face p. 178.)
I
II
III
IV
V
VI
VII

has illegitimate healthy male baby (VI, 20), is somewhat deaf; night-blindness; R. and L. retinal arteries narrowed; retinal pigmentation with usual characters, sparse in R.; O.D. pallor not marked; fields of vision contracted; vitreous opacities in R. These three (V, 124, 127, 129) have some difficulty in speaking; no evidence of syphilis. Mother (IV, 95), age 56 (1910), married at 23, has had 12 children; the first (V, 123) was illegitimate, IV, 30, was not her father. During the pregnancies and at the births of the three children with retinitis pigmentosa there was nothing abnormal. In 1902 the mother had no night-blindness; fields of vision full; V. = 6/6 in each; R. and L. O.D. good colour, at lower periphery of left fundus are two or three patches of superficial choroidal atrophy with well defined edges. Father (IV, 30), age 53 (1910), married at 21, R. and L. fundus normal. III, 27, healthy, *ob.* 75. III, 19, delicate, asthma, good vision, *ob.* 50. III, 11, *ob.* bronchitis. III, 1, *ob.* 75, quite blind, sight began to fail gradually in adult life, no night-blindness, had two operations for cataract 40 years ago. II, 1, *ob.* after 70. II, 2, *ob.* old age. I, 1, *ob.* 102, blind two years before death. II, 3 and 4, both *ob.* old age. V, 123, 125, 126, 128, 130 to 133, no night-blindness, fundi normal. V, 134, *ob.* 12 months, whooping cough and teething. VI, 16, *ob.* 14 months. VI, 9–15, 17–19, fundi normal; youngest of VI, 9, age 7 months, not fully examined. II, 7 and II, 8, and III, 21–27, IV, 78–103, V, 136–226, VI, 21–38 not affected. II, 5, II, 6, and III, 18 and 20 and descendants, IV, 70–77, and V, 135, have no defect of sight. IV, 74–77, some had families. No defective vision in III, 10, 12–17, and their descendants (IV, 22–25, 38–69, V, 23–37, 62–122). IV, 27, 33–37, and their descendants (V, 38–61, and VI, 3–7) are unaffected. IV, 34, had an illegitimate child, V, 42, then married IV, 99, and had two daughters, V, 43, 44. IV, 29, *ob.* young. IV, 31, *ob.* 5. IV, 32, *ob.* 9. V, 46 and 47, *ob.* young. III, 17, born 1841, last survivor in childship; others (III, 10–16), *ob.* from age of 58 to 64. IV, 67, in letter says "all the P. family had remarkable eyesight, that is my mother's (III, 16) family, her brothers were noted for excellent shots." All her brothers and sisters have good sight, and she says that IV, 42, who married her (IV, 42's) 1st cousin (IV, 61), her sisters and their families (V, 62–64, 71–73) have good eye-

sight. III, 2–9, and their descendants (IV, 1–20, V, 1–22, VI, 1 and 2) see quite well with the exception of V, 12, female, and 16, male, who wear glasses for "weak eyes," but are not affected like V, 124, 127, 129. IV, 21, a family, no information. III, 8, female, in lunatic asylum for a number of years. III, 7, male, was at one time in lunatic asylum.

Married and no issue: Males III, 13, 22; IV, 7, 17; V, 124; females IV, 11, 20, 28.

Letters from III, 21, IV, 64 and 67.

FIG. 9.—Two cases of retinitis pigmentosa in a childship of four and another case in the preceding generation. IV, 58, male, unmarried, age 32 (1909), seen, an exceptionally strong man, very deaf, defective vision noticed by his father before age of six; marked night-blindness; fields of vision contracted to 10°; refraction R. mixed astigmatism, V. reads 1 J. at 11 inches; L. low degree of My., V. reads 1 J. at 11 inches; optic discs pale; retinal arteries contracted; moss-like retinal pigmentation all round periphery; choroidal vessels much exposed; Wassermann test negative. IV, 60, male, unmarried, age 23 (1905), seen, is tall and of exceptional ability, first of his year at college; defective vision noticed before he was six; eyes often ache; he is very deaf; has night-blindness; fields of vision contracted to 10°; refraction R. emm. in horizontal meridian, My. 1·25 D. in vertical meridian, V. with correction = 6/12; L.H. 1 D. in horizontal meridian, My. 1·75 D. in vertical meridian, V. with correction = 6/18; opacity at posterior part of lenses; some vitreous opacity; much typical retinal pigmentation; choroidal vessels exposed, especially where retinal pigment is seen; P.'s equal, sluggish contraction to light; nystagmus; Wassermann test negative. IV, 59, media clear; fundus normal. IV, 61, male, age 19 (1905), has no night-blindness, reads as readily in dim light as a normal person; V. = 6/6 in each; fields full (perimeter); media clear; retinal arteries narrow, but not markedly so; choroidal vessels conspicuous at periphery of fundus; at places in periphery of right retina are fine granules of pigment, no typical retinal pigmentation; hearing excellent in both ears. Mother (III, 41), married at 22, was not night-blind; *ob.* 46, abdominal tumour, had no

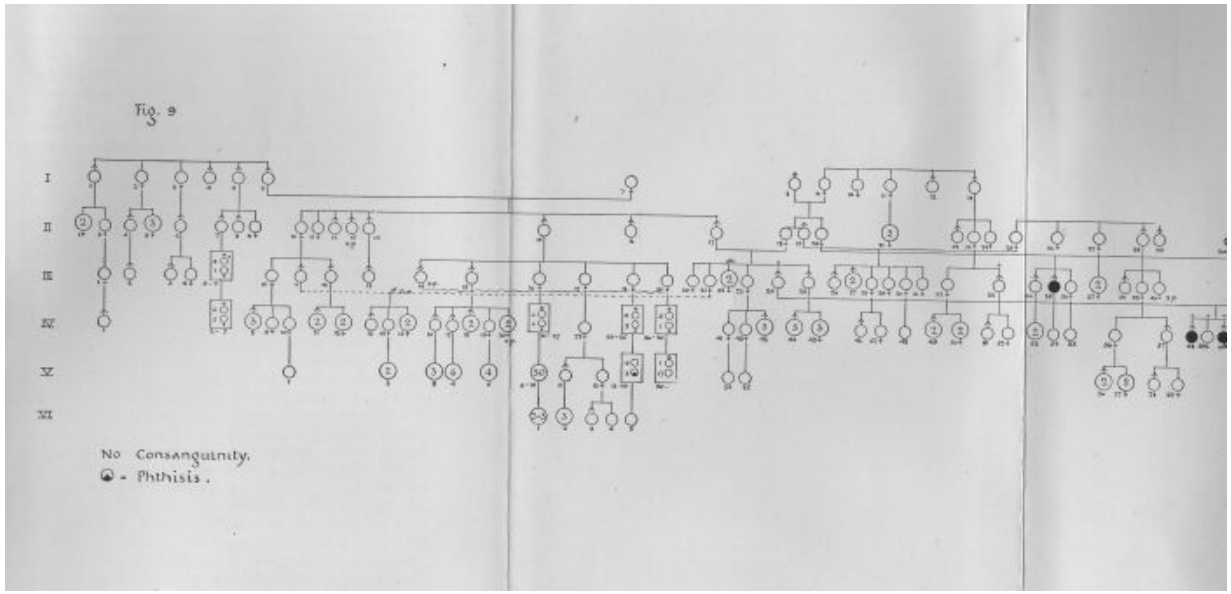
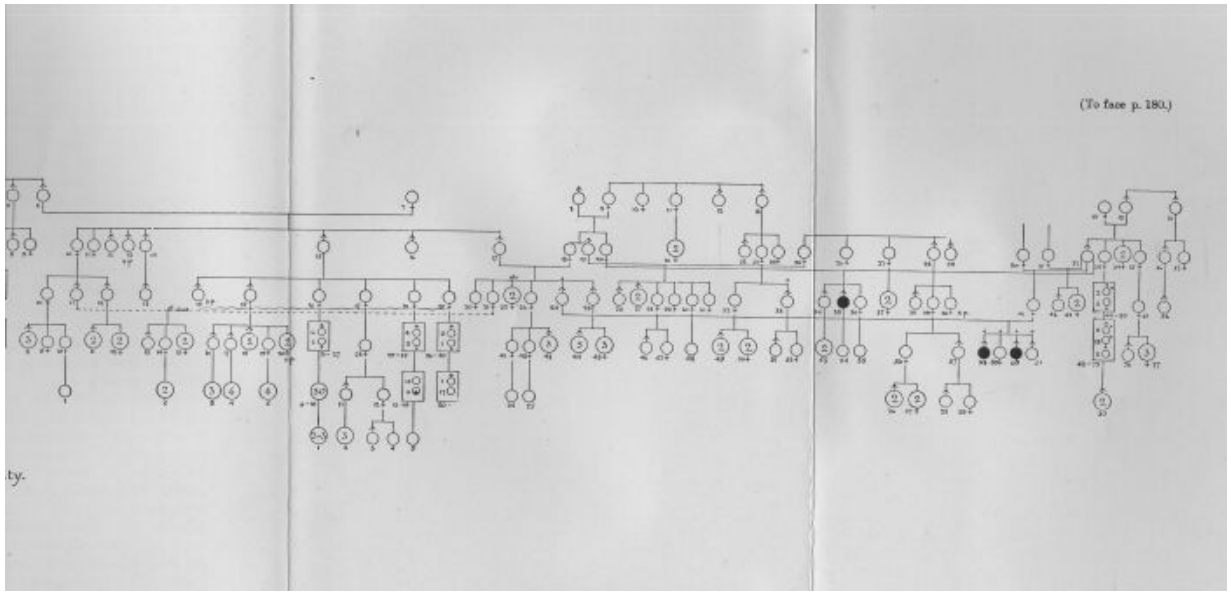


Fig. 9
I
II
III
IV
V
VI
No Consanguinity.
- Phthisis.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miscarriages. Father (III, 24) a healthy, robust, man; married at 28, fundus normal. II, 32, married three times and had three families. By first wife (II, 20), a sister of II, 18, the paternal grandmother of the two affected males (IV, 58 and 60), he had three children (III, 34–36), one (III, 35) was night-blind; by second wife (II, 30) a single child (III, 41), with good sight, which was the mother of IV, 58 and 60; by third wife (II, 31) three unaffected children (III, 42, 43). I, 14 and I, 15, are not known to have had any visual defect. II, 17, a very strong man, *ob.* 91, his wife (II, 18) was not a cousin. I, 6 and 7, had good sight. I, 8 and 9, normal vision, are the maternal grandparents of a night-blind male (III, 35) and the great-grandparents of the two males (IV, 58, 60) with retinitis pigmentosa. No night-blindness known in I, 1–5, or their descendants (II, 1–9, III, 1–9, IV, 1–7). IV, 3, male, quite deaf, caused by fever at 18 months. II, 5, unmarried, III, 2, no issue. II, 7, is alive (1911), age over 90. II, 11, 12, 16, unmarried. II, 10 and descendants (III, 10–12, IV, 8–15, V, 1–2) have good sight. II, 15 went to Canada with all his children (III, 14–19) except III, 19. An account of their descendants (IV, 16–40, V, 3–23, VI, 1–5) is given in a letter by IV, 16, a medical man in Canada. He does not know of night-blindness in any of them. III, 14, is alive, age 82. III, 17, *ob.* 91 of acute gangrene of foot. III, 15, *ob.* 73, apoplexy, had for years had arterio-sclerosis. III, 18, had defective circulation in feet for many years. IV, 16, remarks that early arterio-sclerosis especially of the arteries of the feet appears to be the family weakness. III, 19, *ob.* pneumonia. IV, 22, male, *ob.* sarcoma of kidney. VI, 3, age 12, had polio-myelitis at 2 years. V, 19, 3 children in 2 families, *ob.* phthisis. IV, 18, unmarried. I, 10–13 and descendants (II, 21–24, III, 32, 33, IV, 49–52) vision normal. III, 35, male, abroad, lost sight of, was well known to his cousin (III, 24) to be night-blind. He was deaf, lisped, and his sense of smell was defective. His sisters (III, 34, 36) have good sight. IV, 53–55, no particulars, but in a letter from III, 34, there is no mention of any blindness. III, 20, unmarried. Not known if III, 26 and 27 had issue. IV, 48, no particulars. II, 19, 25–29 and descendants (III, 26–31, 37–40, IV, 46–48, 56, 57, V, 26–29), no defect of vision heard of. IV, 56 and 57, have never heard of

night-blindness in their relatives. III, 37, *ob.* unmarried. I, 16, II, 33–37, and descendants (III, 44–52, IV, 62–77, V, 30) have good vision. III, 20–25, IV, 41–45, V, 24, 25, see well. II, 34, and IV, 76, unmarried.

The retinitis pigmentosa appears to have been transmitted by II, 32, or else by either I, 8, or I, 9, for they are ancestors common to the 3 males with night-blindness—2 proved to have retinitis pigmentosa.

FIG. 10.—A female with retinitis pigmentosa in a childship of eight. IV, 8, female, third in childship, unmarried, age 31 (1911) seen, always weak mentally and bodily and hearing imperfect; had diarrhœa in infancy; "inflammation of lungs" at 16 months; never spoke until age of 7; defective vision noticed first at age of 14, when she knocked against lamp-posts, it has gradually got worse; violent temper, especially during menstruation, once stabbed mother in arm, at other times morose. In 1903, posterior cortical cataracts; moss-like retinal pigmentation confined to a broad band at periphery of fundus, extreme periphery not pigmented; at pigmented band choroidal vessels more exposed than elsewhere, and a few small bright white bodies are scattered here and there in the band region; O.D. dirty white colour; retinal vessels very narrow; refraction R., My. 1·25 D. in oblique meridian, 2·25 D. in opposite meridian, V. with correction is 6/18 partly; L., My. 1·25 D. in oblique meridian, 2·25 D. in opposite meridian, V. with correction is 6/12 partly; fields of vision contracted. To-day (April, 1911) pigmentation of retina more marked, but O.D.'s and macular regions are free from pigment in retina; marked night-blindness; fields of vision much contracted; speech defective. R. V. with her glass is 6/24 in each. Wassermann test negative. Mother (III, 10) healthy woman, fundus normal, married at age of 21, had no miscarriages, and none of her children had snuffles or rash in infancy; when $4\frac{1}{2}$ months pregnant with IV, 8, she had a severe mental shock, thought husband intended to commit suicide; R. V. with +1 D. = 6/6; L. V. c. +0·5 D./+0·75 cyl. = 6/6. Father (III, 3) excitable, fundus normal, in army in India before marriage, and had moon-blindness, from which he recovered; after returning to England, contracted venereal disease

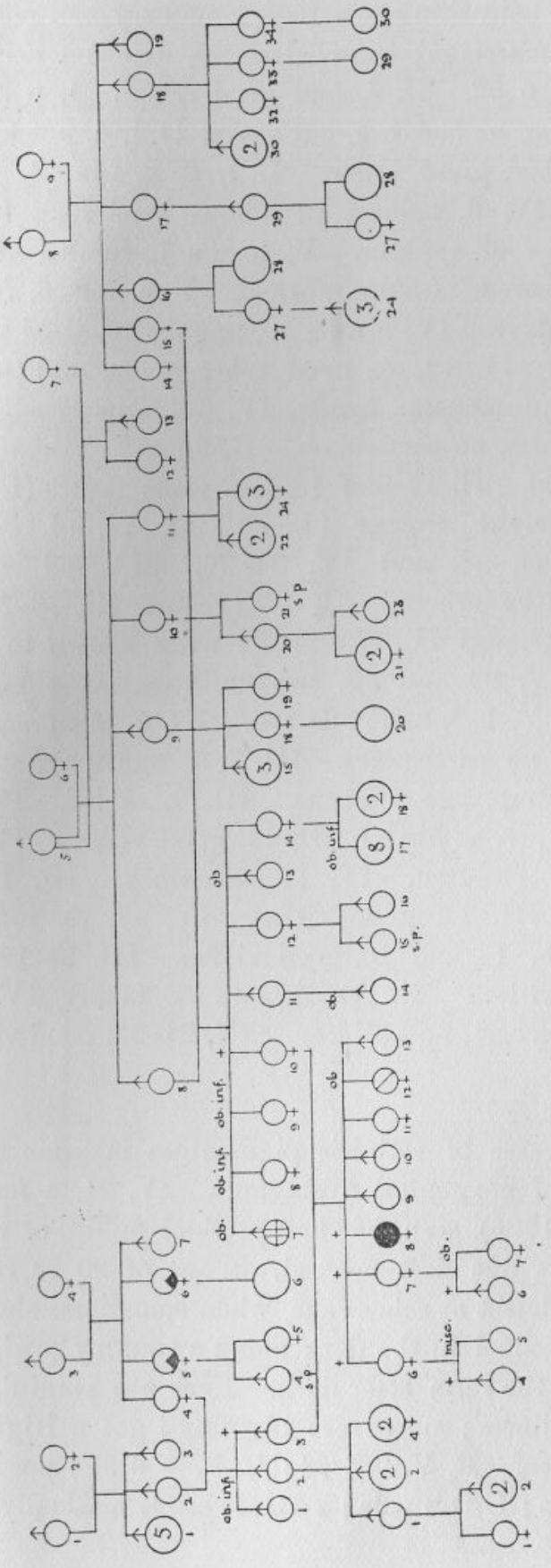

I
II
III
IV
V
No consanguinity.
deformed.
hydrocephalus.
Phthisis.

Fig. 10.

(sore throat, loss of hair, skin spots, took "mercury and potash") four years before marriage; married at 25, wife not a cousin, II, 15, still alive, age 92. II, 8, had good vision. I, 8 and 9, both had good vision, so had I, 5, but vision of I, 6, not known. II, 4, and II, 2, had good vision. I, 1, 2, 3, and 4, vision reported normal. IV, 6, subject to bilious headaches, fundus normal, no evidence of syphilis. V, 4, age 7, fundus normal. IV, 7, "quick tempered," fundus normal. V, 6, age 6, fundus normal, V, 7, *ob.* 2 days. IV, 9 and 10, unmarried, good vision. IV, 11, good vision. IV, 12, *ob.*, lived a few hours, said to have had club foot and a monster head. IV, 13, unmarried, good vision. II, 1, 5, males, no particulars. II, 3 and 7, vision good, the latter was killed. II, 12 and 13, had same father (I, 5) as II, 8–11, but different mother (I, 7). II, 14, *ob.* young. II, 16, III, 27 and 28, and IV, 24–26, all good vision. (No particulars of the others.) II, 17–19, and III, 30–34, all good vision. III, 29, and IV, 27 and 28, none known to have defective vision. IV, 29 and 30, two children, sex unknown. III, 2, IV, 1–5, and V, 1–3, none affected. III, 5, *ob.* unmarried. III, 6, large family, no particulars. III, 7, *ob.* age 4, "water in head." III, 8, *ob.* croup, age 6 weeks. III, 9, *ob.* inf. IV, 14, only child of III, 11, *ob.* at birth. III, 12, good vision. IV, 15, married 8 years, no children. IV, 16, schoolboy. III, 13, *ob.* unmarried, killed. III, 14, good vision. IV, 17, *ob.* infancy. IV, 18 and 19, ages 14 and 10, good vision. III, 15–19, and III, 22–26, good vision. No particulars of family IV, 20. III, 21, married 15 years, no children. IV, 21–23, good vision, youngest age 20.

FIG. 11.—Two cases of retinitis pigmentosa in same childship as two cases of congenital nystagmus. IV, 24, a female, unmarried, age 44 (1906), seen, has had gradual failure of vision since night-blindness was first noticed, at age of 20 or rather later. She had no defect at school age, when sometimes she had to walk home from school in the dark along a country road, and was often out about the farm after dark. Eyes are painful, and water; never any illness; no severe bleeding; got a fright at age of 20; is very deaf. R. V. = 6/24; L. V. = 6/36; posterior cortical lens opacities; O.D.'s pale; retinal vessels markedly con-

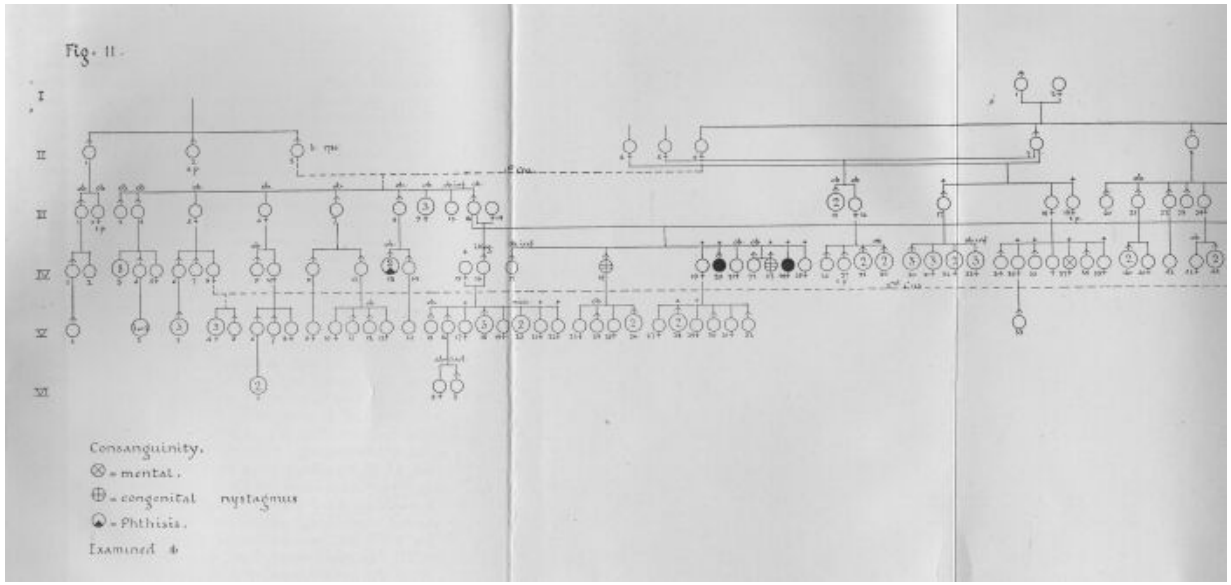
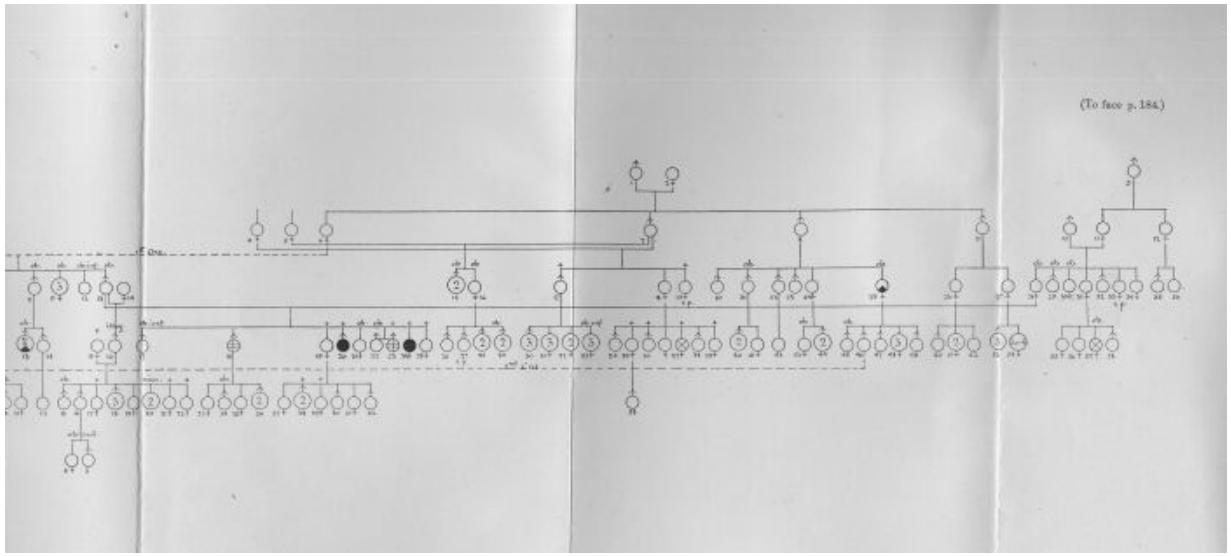


Fig. II.
Consanguinity.
⊗ = mental.
⊕ = congenital nystagmus
◒ = Phthisis.
Examined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tracted; choroidal vessels much exposed; much retinal pigmentation at periphery of each fundus; fields of vision contracted, left, taken on perimeter, is contracted to from 10° to 20°. When seen again in 1912 she could see to get about in daytime; fundus appearances similar to those on last examination; Wassermann test negative. IV, 20, male, unmarried, farmer, age 57 (1912), seen, very deaf, has had chronic blepharitis since measles; discharge and fragments of bone came away from clavicle about age of 16; no bleedings; can go about in daytime; marked night-blindness, which was noticed much earlier than in his sister (IV, 24); his mother said that he heard and saw well before he had measles; refraction My. R. and L. V., 1 J in each; nystagmus; posterior cortical lens opacities; optic discs pale; retinal arteries narrow; much retinal pigmentation at periphery of retina; appearances quite typical of retinitis pigmentosa. IV, 23, male, a twin, unmarried, age 53 (1912), seen, has no night-blindness, reads ordinary print readily, has constant lateral nystagmus in both eyes which has been "present from birth." R. and L. fundus normal; media clear; iris green with yellow parts; hair yellow. IV, 22, *ob.* 14 months. IV, 18, has constant nystagmus present since birth, it is not so marked as in IV, 23, seen by medical man, vision is good; no night-blindness. IV, 25, fundus normal, no deafness, no nystagmus. IV, 19, fundus normal, no nystagmus. IV, 21, *ob.* unmarried. Mother (III, 28), married at 28, *ob.* 75, had good sight. Father (III, 13) married at 29, *ob.* 76, was alcoholic, had exceptionally good vision all his life, no nystagmus; in order of birth he was in middle of the childship. III, 3–13, nearly all died after 70. III, 6, had cataract, no operation. IV, 16, is an illegitimate child of III, 13, and III, 14; his wife (IV, 15) says that he and all his children (V, 15–22) see well. II, 10, and II, 11, not cousins. I, 3, was a tailor. II, 3, a first cousin of his wife (II, 6), *ob.* 92, could read to the last. I, 1, and I, 2, not much known about them. II, 1, had two children whose vision became defective. III, 1, had an operation on eyes, and III, 2, got blind late in life. IV, 1, a joiner, "screws eyes up." II, 7, married twice; by first wife (II, 5) had three children (III, 15, 16) and by second wife (II, 4) three children (III, 17–19). IV, 26–39, none have defective vision. IV, 37, in lunatic asylum for a few

months at age of 22. II, 8 and 9, and descendants (III, 20–27, IV, 40–54), none known to have bad sight. IV, 53 and 54, most are married, some abroad. III, 25, a female, *ob.* phthisis. IV, 6–14, V, 3–14, and VI, 1, none affected; two males, IV, 13, *ob.* phthisis. V, 23–33, see well. II, 12, III, 29–36, and IV, 55–58, none known to have defective vision. IV, 57, died in lunatic asylum. IV, 3, abroad, most of them married, no particulars except that none have defective vision.

FIG. 12.—A single case of atypical retinitis pigmentosa in a childship of five. V, 20, female, age 6 (1911), seen. R. and L. uniform pallor of O.D.; physiological cup not filled in; retinal arteries and veins very narrow; all over periphery of fundus are numerous small black pigment specks and much delicate branched pigment in retina; little, if any, coarse moss-like pigment; no circular patches of choroidal atrophy; in some large areas at periphery choroidal vessels show more than elsewhere; no lens opacity; refraction H., R. 3·5 D. in oblique meridian and 6·5 D. in opposite meridian; L. 3 D. in oblique meridian and 6·5 D. in opposite meridian; she is illiterate; when she drops small things her mother says she does not see them, yet in badly lit room she seemed to have little difficulty in finding things on the floor. In 1906, when 17 months old, she did not see well and "groped about," there was constant lateral nystagmus, no gross fundus lesion, never had head-nodding. In December, 1907, ill defined changes were present at macula (R. and L.), with pepper-like pigmentation and choroid paler at one spot; no deafness; never had fits; Wassermann test negative. V, 19, 21, 22, 23, fundi normal, in V, 23, examination unsatisfactory, none are deaf. Mother (IV, 16) has myopic astigmatism; no retinitis pigmentosa; had no miscarriages; was alone when V, 20, was born. Father (IV, 2), fundus normal. III, 9, frail, is 2nd born in a childship of 18. III, 8, *ob.* 65, saw well. II, 8, lived longer than 60, and II, 9, *ob.* 73, both had good vision. II, 4, *ob.* cancer of bowel, he and his wife (II, 5), not cousins, saw well. I, 1, vision good. II, 2, by II, 1, had a daughter (III, 1) who became blind after 50, cause of blindness not ascertained, but history does not suggest retinitis pigmentosa. Her son (IV, 1) has absolute glaucoma in each eye, age 45. III, 2

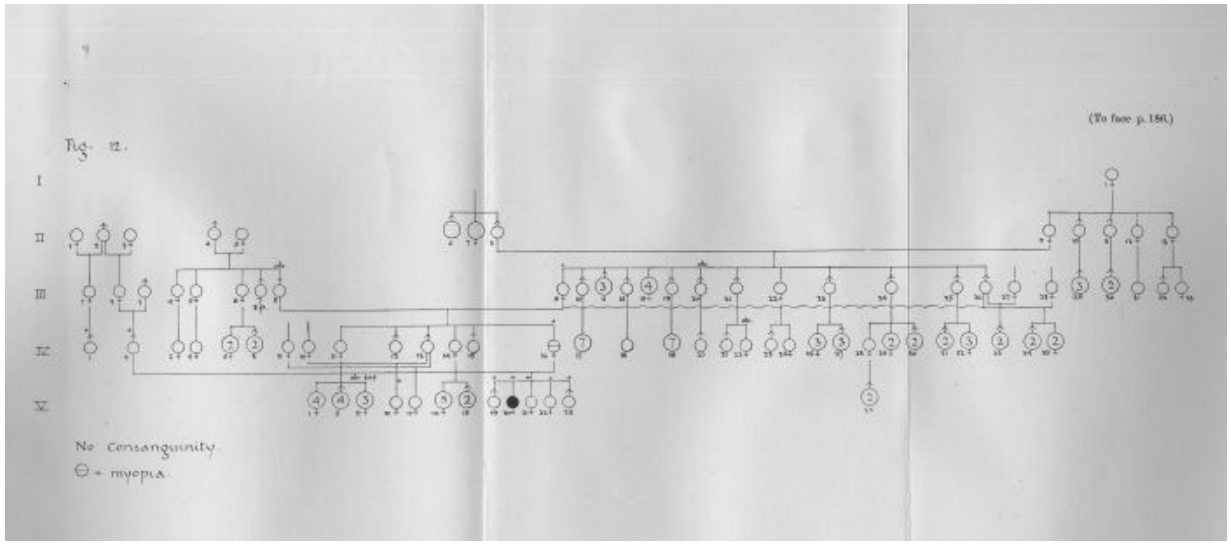
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Fig. 12.
I
II
III
IV
V
No consanguinity.
⊖ = myopia.

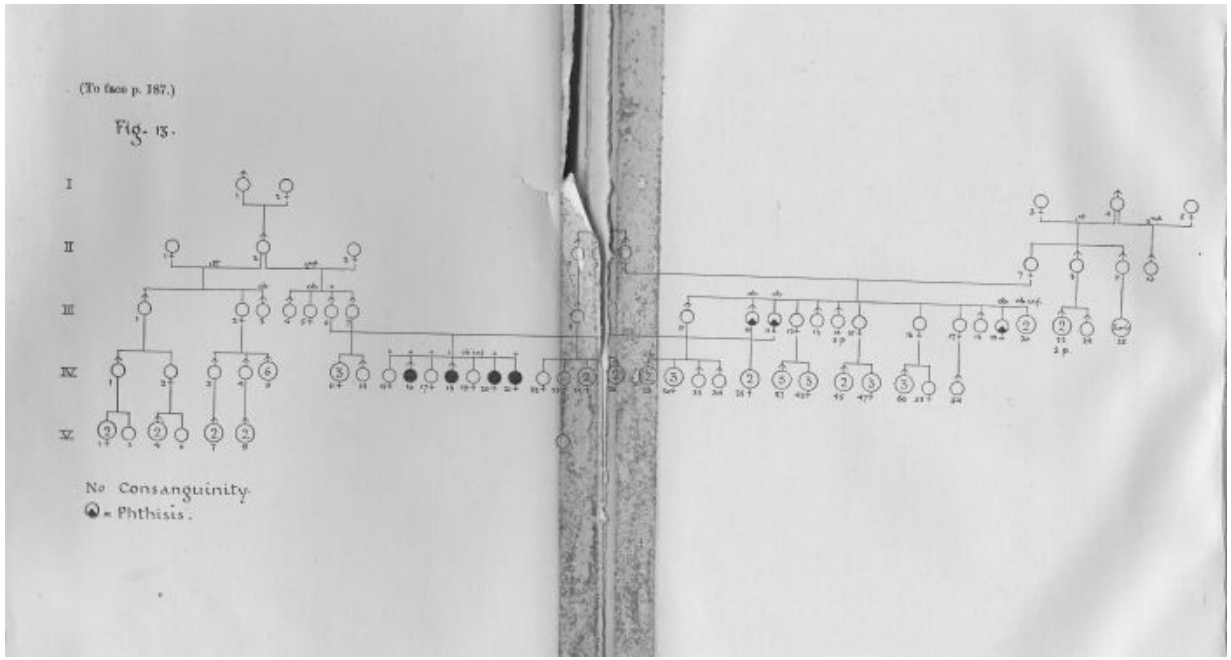
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Fig. 13.
I
II
III
IV.
V.
No Consanguinity.
= Phthisis.

and 3, excellent vision. II, 6 and 7, males and females, numbers not known, none had defective vision. III, 4–7, and IV, 3–8, vision good. IV, 5–8, were known only in early life. III, 7, had a "drawn-up" leg. No defective vision known in II, 10–13, III, 10–33, IV, 17–35. III, 15, a female, stillborn. III, 26, married twice. III, 31, a family, no particulars. IV, 12 and 15, unmarried. IV, 13, has two daughters, the first (V, 10) has normal fundus, the second (V, 11), a half sister of V, 10, no particulars. IV, 11 and 14, V, 1–8, none known to have any visual defect; oldest in childship V, 12–18, is aged 13. IV, 17, a family of 7, none known to be affected. IV, 18, no particulars. IV, 19, family of 7, state of vision unknown. V, 24, the elder age 3.

FIG. 13.—Four cases of retinitis pigmentosa with deaf-mutism in a childship of seven. IV, 16, male, age about 17 (1911), seen, deaf-mute with marked night-blindness, defect of vision not noticed until 7 or 8 years of age. V. R. and L. each read 1 J. in good light; O.D.'s pale, retinal vessels contracted, typical appearance of retinitis pigmentosa in each fundus, with characteristic retinal pigment; fields of vision contracted; pigment in relation to the retinal veins at parts where this was looked for. IV, 18, male, age about 15 (1911), seen, a deaf-mute with night-blindness, which was first observed when he went to an institution for deaf-mutes at age of 7. Oph.: R. and L. fundus changes are similar to those of his brother (IV, 16). IV, 20, female, age 9 (1911), seen, a deaf-mute mentally weak, is considered to be night-blind by those in charge at an institution for deaf and dumb, because in dim light she knocks against objects which other children avoid, but in a darkened room it was difficult to confirm the presence of night-blindness owing to her mental condition. Oph.: R. and L., retinal vessels narrow; O.D. pale; choroidal vessels exposed; some yellowish mottling towards periphery of fundus; no moss-like pigmentation; refraction, H. 2·5 D. in vertical meridian, 6 D. in horizontal meridian in each eye. Wassermann test negative. IV, 21, female, age 7 (1911), seen, more intelligent than her sister (IV, 20), is a deaf-mute, night-blindness believed to be present though not marked; tests made in darkened room gave

same result as in IV, 20. Oph.: R. and L., much yellowish mottling towards periphery of fundus, this is behind the retinal vessels; much pepper-like pigmentation of fundus, especially in left eye; only two or three moss-like pieces of pigment were detected, and these were at the periphery in front of the retinal vessels; refraction H. R. 2 D. in vertical meridian; 3·5 D. in horizontal meridian, L. 2 D. in vertical meridian, 5 D. in horizontal meridian. Wassermann test negative. None of these four children have had any illness or bleeding. IV, 15, age 18, no deafness, no myopia. Oph.: R., a dense white band extends from inner edge of O.D. to beyond its centre, probably congenital, otherwise both fundi are normal. IV, 17, R. and L. fundus normal; refraction R. emm., L. mixed astigmatism. IV, 19, *ob.* infancy. Mother (III, 11), the 3rd born, married at 29, was always delicate, *ob.* 44 of phthisis, had no miscarriages, she and her husband not cousins. Father (III, 6), 3rd born, an intelligent, robust farmer, age 54, never had venereal disease; married at 34, fundus (R. and L.) normal. It is from his account that the pedigree has been constructed. II, 2, married twice. None of the children (III, 1–3) of his first marriage with II, 1, or any of their descendants (IV, 1–5, V, 1–9) were affected, also none of his second family (III, 4–7) by II, 3, or IV, 11, 14, were affected. I, 1, had good sight. I, 2, *ob.* 94, seen by III, 6, no defect. III, 3, *ob.* young. III, 5, unmarried, *ob.* of kidney disease. V, 1–4, young. II, 6 and 7, not cousins. I, 4, married twice, none of his descendants (II, 7–10, III, 9–25, IV, 28–54) had defective vision except those already mentioned (IV, 16, 18, 20, 21) with retinitis pigmentosa. III, 22, two males, married, had no issue. II, 9, had a family of five or six (III, 25), all with good vision. II, 10, unmarried. III, 10 and 19, *ob.* phthisis. III, 13 and 18, unmarried. IV, 30–34, oldest is 14. No visual defect in II, 4, 5, III, 8, IV, 22–26. IV, 23 has a child (V, 11), no others in childship married.

FIG. 14.—A case of retinitis pigmentosa and two cases of coloboma of optic nerve in the same childship, and another case of retinitis pigmentosa in a different childship of the same generation. IV, 114, male, unmarried, age 25 (1910), seen, looks quite well, but mother says though his health is

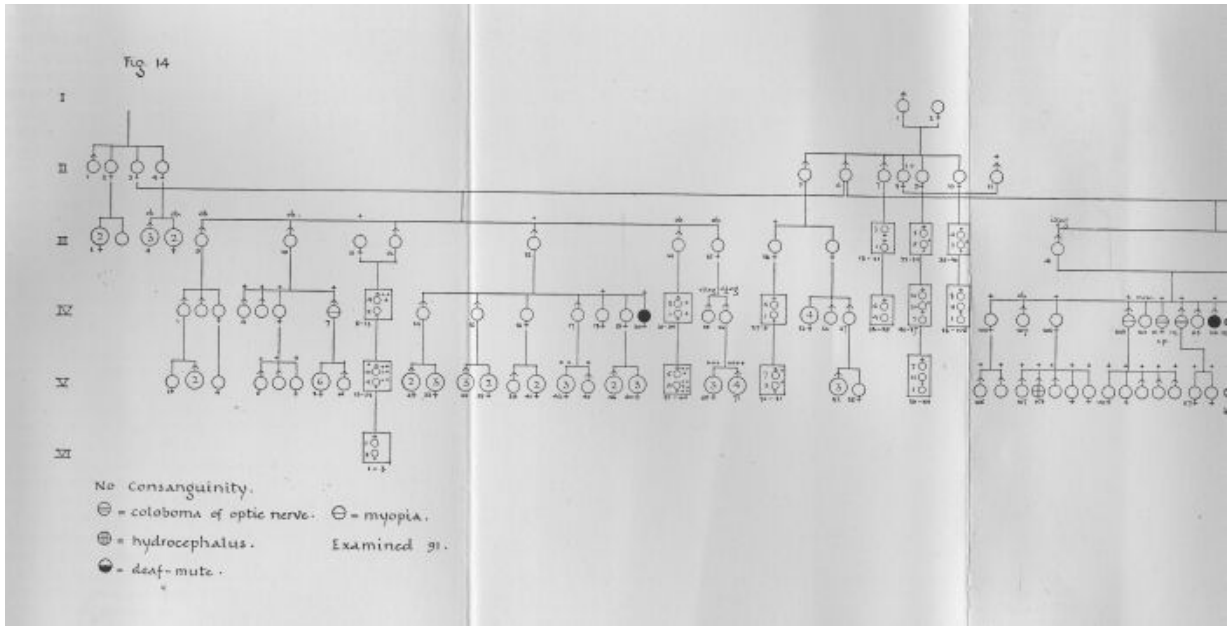
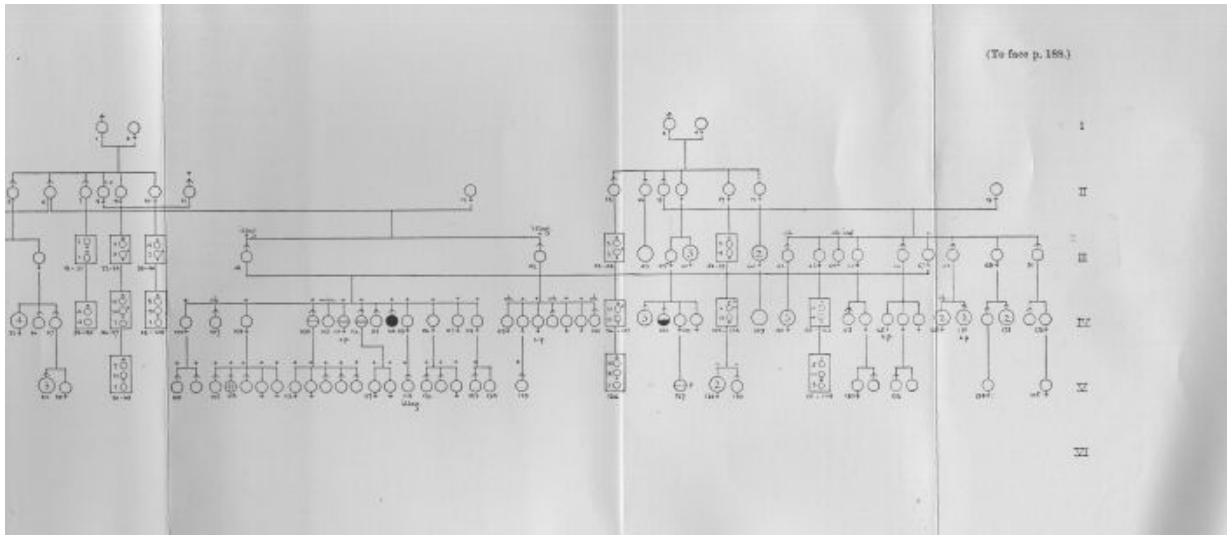


Fig 14
I
II
III
IV
V
VI
No Consanguinity.
= coloboma of optic nerve.
= myopia.
= hydrocephalus.
Examined 91.
= deaf-mute.

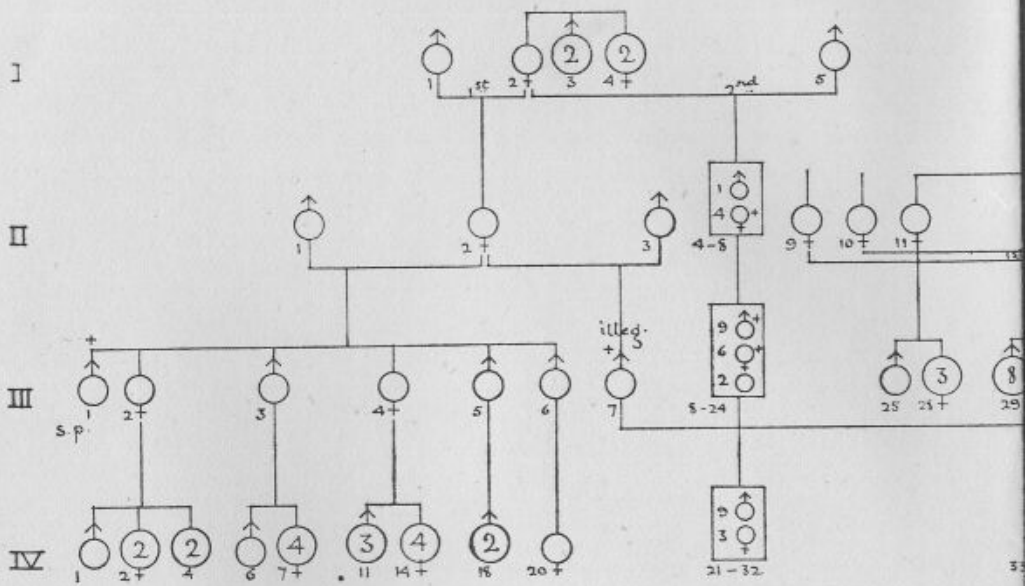

(To face p. 188.)
I
II
III
IV
V
VI

good he is not so strong as her other children, and he vomits readily things that do not agree with him; whooping cough in infancy; no bleedings; fingers and toes not subject to get cold; difficulty in dim light and in consequence cannot go from home in evenings; defect of vision first noticed after he went to school; R. and L. oph.: media clear, O.D. pale, retinal arteries very narrow, moss-like pigmentation of retina most abundant towards periphery, it is in relation to veins and not arteries; no spots or patches of choroidal atrophy, some yellow mottling; close to inner edge of R.O.D. are three small grey slightly raised bodies; refraction R.H. 2 D. in oblique meridian, emm. in opposite meridian, V. with correction 6/12, L.H. 2 D. in oblique meridian and 4 D. in opposite meridian, V. with correction = 6/18, fields of vision not contracted, complete absolute ring scotoma in each, Wassermann test negative. IV, 111, female, married for 6 years, is without child, age 30 (1910), examined by Dr. Wm. C. Souter; left eyeball smaller than right one; defective outward movement of each eye, lateral nystagmus on lateral movements of eyes; oph., central part of each fundus is occupied by a circular white area several times the size of a normal optic disc, it has well defined margin, retinal vessels converge to a part near upper edge of the area; two small white patches of choroidal atrophy are present below it in right eye and there is some choroidal atrophy at corresponding situation of left eye; edge of main area in each eye is not so well defined below as at other parts, very deep depression in each area, diagnosis is coloboma of optic nerve, rest of fundus (R. and L.) is normal, lenses and irides normal, refraction R.H. 3 D. in horizontal meridian, 5 D. in vertical meridian, V. with correction = 6/36; L.H. 3 D. in vertical meridian, emm. in horizontal meridian, V. with correction is $<6/60$; right field of vision has a large defect above extending from periphery nearly to fixation point. In the previous pregnancy her mother carried a dead 6 months' fœtus (IV, 110), for six weeks. IV, 112, male, age 31 (1912), seen, has constant lateral nystagmus. R. microphthalmic; O.D. replaced by a large crater-like depression 2 or 3 times the size of a normal optic disc, edge focussed with +7 D. lens, its depth with a +1·5 D., a few blood vessels pass over edge into crater, adjoining its lower part is some choroidal

atrophy, coarse opacities at outer and posterior parts of lens, refraction H. 9 D. V. with correction = counts fingers, L. O.D. larger than normal, cupped in lower $\frac{3}{4}$, edge focussed with − 6 D. lens, bottom of cup with − 9 D., choroidal atrophy adjoins lower part of O.D., refraction My. 6 D. V. with correction = 6/36, large sector absent at upper part of field of vision, no night-blindness, irides normal. IV, 106, a healthy woman with normal fundi. IV, 107, unmarried, *ob.* abroad of "fever," age 34. IV, 108, fundi normal. IV, 109, age 37 (1912), no night-blindness, fields full, media clear, fundi normal, refraction My. 1·5 D. in oblique meridian and 2·25 D. in opposite meridian, H. 2 D. V. with correction = 6/6 in each. IV, 113, unmarried, works on a farm, sees well. IV, 115, age 27 (1912), retinal vessels not contracted, no retinal pigmentation, O.D.'s good colour, refraction H.R.Hm. 1·75 D., L.Hm. 1 D., V. = 6/6 in each. IV, 116 and 118, have normal fundus (both), no myopia. IV, 117, age 21 (1912), refraction, R. and L. low H. V. = 6/6 in each, fundi quite normal, fields full, no difficulty at dusk. Mother (III, 67), healthy, fundus normal. II, 15, II, 19, and I, 3, had good sight; not much known about I, 4, name is not the same as I, 3. Father (III, 41), an old farmer; he and his brother III, 42, are illegitimate sons of II, 12, and II, 6, who both saw well. I, 1, and I, 2, had good vision, the latter *ob.* old. II, 6, married II, 3. Their children (III, 9–15) had good vision. III, 13, has 7 children (IV, 14–20), the youngest is night-blind. IV, 20, female, unmarried, age 32 (seen), 7th in childship, has always been quite useless in dim light, looks healthy, never had illness or bleeding. R. and L. retinal arteries show some degree of narrowing; O.D.'s no marked pallor; choroidal vessels exposed only at periphery of fundus; in each eye a small area below with retinal moss-like pigmentation; some vitreous opacity; fields contracted especially at upper part, at some parts below they are full, commencing ring scotoma below in each; refraction H. R. and L. 1 D. in vertical meridian, 5 D. in horizontal meridian, V. with correction = 6/12 in each; in dim light that gives a normal eye 6/18, she reads 6/60; Wassermann test positive; she lives in country far from a town; no appearance of syphilis. IV, 18, with exception of a large black pigment patch in upper part of left fundus both fundi are

(To face p. 191.)

Fig. 15.

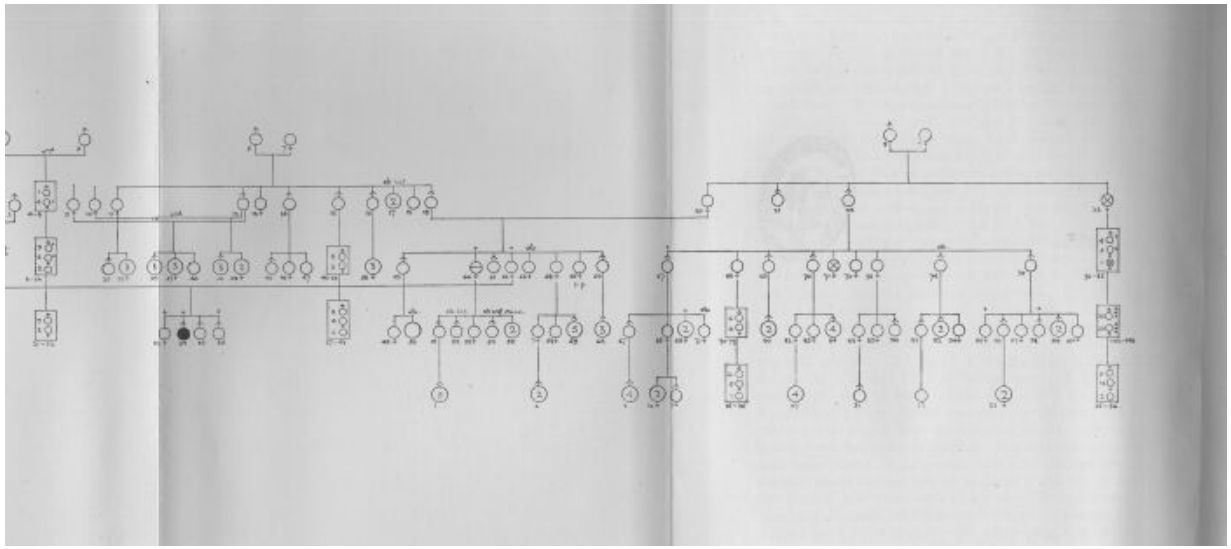


No Consanguinity.

⊗ = mental.

⊖ = myopia.

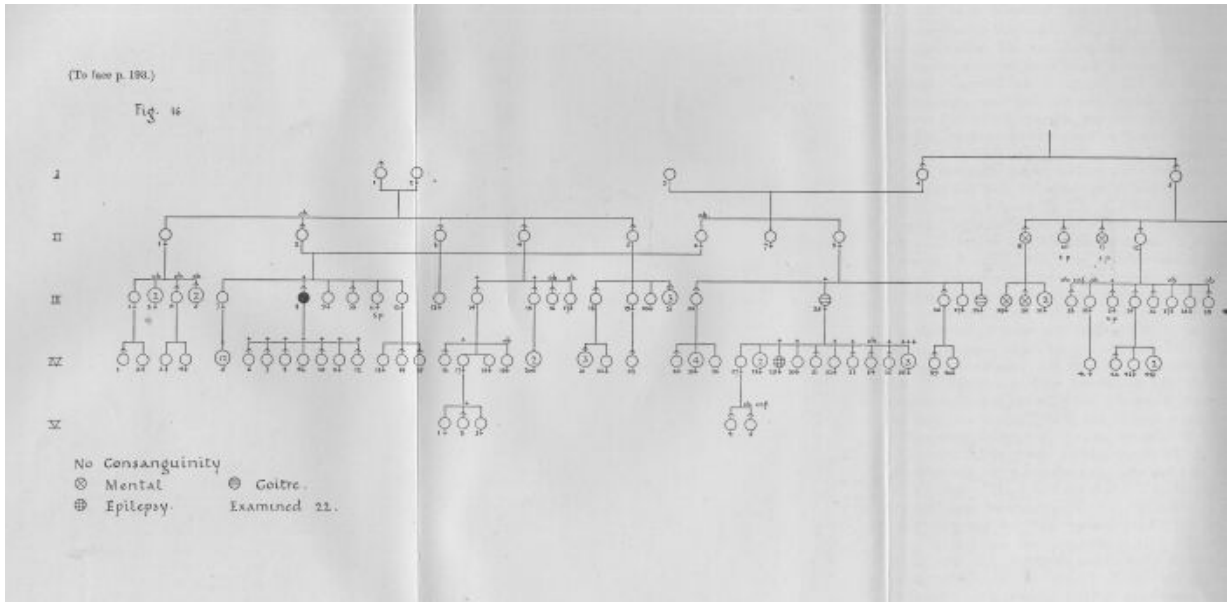
Examined 22.

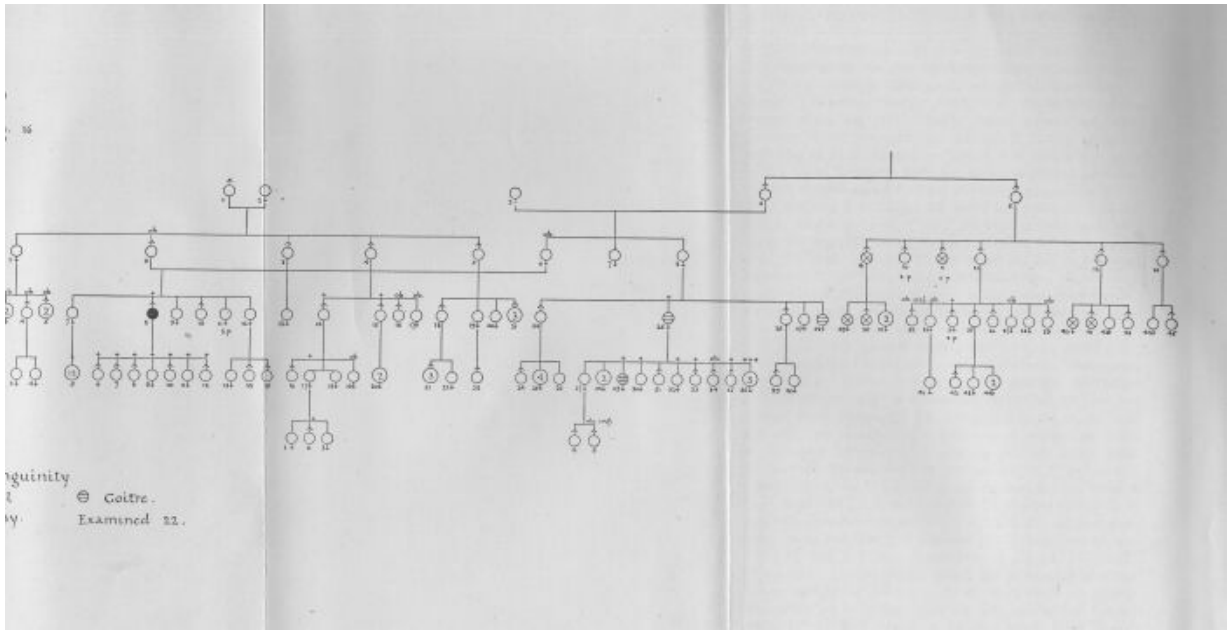


normal, no night-blindness. IV, 14–17 and 19 have good vision. The two cases of retinitis pigmentosa (IV, 20, and IV, 114) are children of half-brothers (III, 13, and III, 41). II, 3, suckled III, 41, the illegitimate son of her husband, at same time as she was suckling her legitimate son, III, 13. The night-blindness in offspring of III, 13, and III, 41, is attributed to the mental shock of II, 3, when she learned that III, 41, was her husband's illegitimate child. The fathers of the retinitis pigmentosa cases were suckled together by a distressed woman, the mother of one of them. III, 13, age 71, fundi normal, refraction R. emm., L. My. 0·5 D., V. = 6/6 in each. Wassermann test negative; his wife is alive and has good sight; they have been married for 50 years. II, 6, *ob.* 80. III, 15, unmarried, had 2 illegitimate children (IV, 25, 26). None of the other descendants of II, 3, nor of II, 2, 4, 5, 7–11 are known to have any visual defect except IV, 7, he is 37, fundi normal, fields full, no ring scotoma, R. My. 1·5 D. in oblique meridian and 2·25 D. in opposite meridian, V. with correction = 6/6, L. My. 2 D., V. with correction = 6/6. V, 124, a baby. V, 105–123, all have normal fundi except V, 108, boy, age 12 (1908), who has hydrocephalus and double optic atrophy. Information obtained from II, 11, a very old man, since dead. No defective vision known in descendants of I, 3 and 4, except in childship IV, 106–118, and V, 108, already noted, and also V, 127, a family in which every one is short-sighted, according to the medical man this is derived from their father, their mother (IV, 142) and her brothers and sister have good vision. IV, 141, male, age 55, a shoemaker, is a deaf-mute, unmarried, he goes about freely at night. His doctor has not been able to trace any deaf-mutism in his father's relatives; his father was illegitimate. Married females without children: II, 8; III, 27, 40, 47; IV, 111, 121, 160, 165. Males, married, with no children: III, 45; IV, 28, 158, 170, 171.

FIG. 15.—A single case of retinitis pigmentosa in a childship of four. IV, 34, male, unmarried, age 14 (1896) seen, farm servant, does not see well at night, ill for 6 weeks at age of $2\frac{1}{2}$ with "inflammation of brain," and his father believes that the sight has been defective since; nose bleeds easily, it bled daily when at school, no deafness, is intelligent, no liability

for hands and feet to become readily cold; no evidence of syphilis, is very healthy. R. and L. well marked retinal pigmentation with moss-like character, an unpigmented part of retina is present peripherally to the pigmented area at temporal and nasal sides, but not above and below, at nasal side of fundus are numerous black specks amongst the other forms of pigment; choroidal vessels exposed. O.D. of uniform reddish colour, edges not well defined; retinal vessels narrowed; posterior cortical lens opacities; refraction H. R. 2 D. in oblique meridian, 4·5 D. in opposite meridian, V. with correction = 6/12, L. 1·5 D. in horizontal meridian, emm. in vertical meridian, V. with correction = 6/9; fields of vision contracted to 20° at nasal side, and from 15° to 20° above and below, on temporal side L. reaches 30° and R. 70°. In 1901 L. reduced to 10°; R. to 10° below, 15° above and at nasal side, 50° on temporal side. In 1912, R.V. = 6/24, not improved; L.V. = 6/18, not improved; R. and L. O.D. waxy, moss-like retinal pigmentation does not extend to central region; retinal vessels very narrow; no white patches in choroid; he has to take care that night does not overtake him when in places unfamiliar to him; in dim light which gives a normal eye 6/12, he reads 6/36; hearing excellent; health has continued excellent; Wassermann test negative. Mother (III, 62), 4th in childship, and father (III, 7), illegitimate, both married at age of 20, are not cousins, fundi normal. II, 19, *ob.* 73, and II, 20, *ob.* 80, both saw well, not cousins. I, 8, was a good weaver, no defect of his vision known. I, 9, seen by III, 62, vision good. II, 3, went abroad, was not married to II, 2, and not a cousin of hers. I, 2, married twice, she and her 1st husband (I, 1), known to III, 7, had one child (II, 2) who married II, 1, they and their descendants (III, 1–6, IV, 1–20), all with good sight. I, 2, and her 2nd husband (1, 5) had five children (II, 4–8), they and their descendants (III, 8–24, IV, 21–32) all see well. III, 18, female (seen), has a dermoid at sclero-corneal margin in left eye. I, 6, *ob.* young, no reliable information as to vision. I, 7, *ob.* 100, vision and hearing good. IV, 33, high degree of H. astigmatism, fundi normal. IV, 35, sees well, no night-blindness. IV, 36, both fundi normal. II, 9–16, and descendants (III, 25–56, IV, 37–48) said

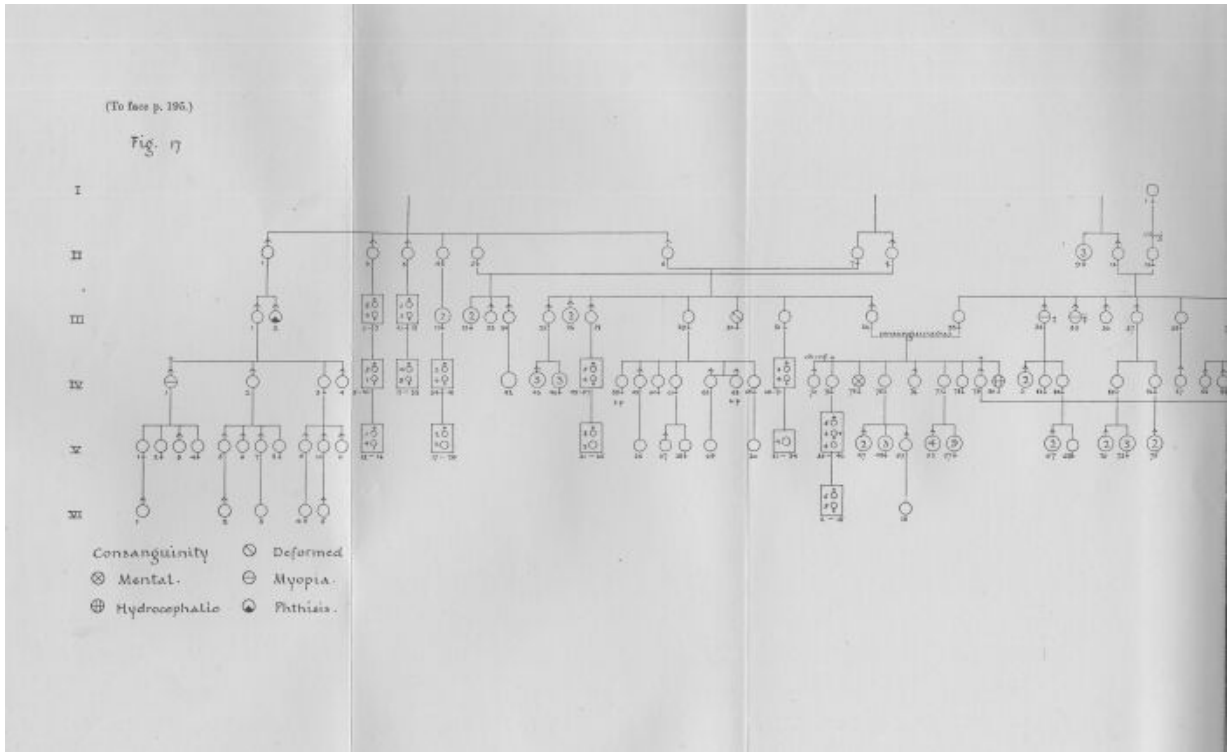

(To face p. 198.)
Fig. 16
I
II
III
IV
V
No Consanguinity
Mental
Epilepsy.
Goitre.
Examined 22.

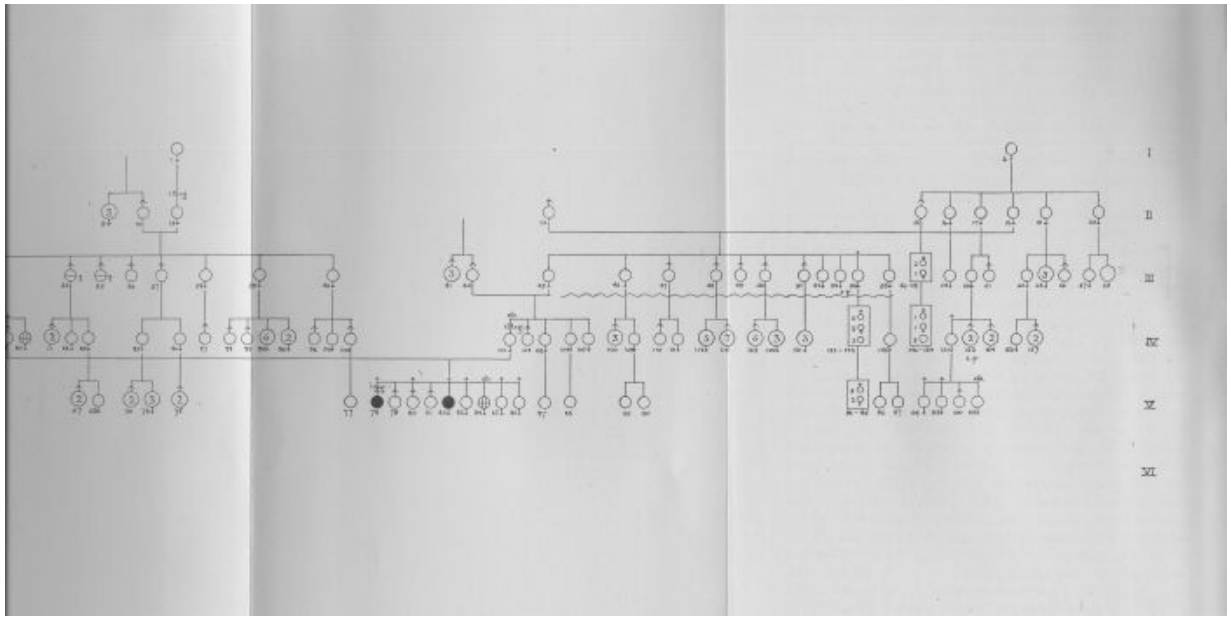

nguinity
⊖ Goitre.
Examined 22.

to have good sight. III, 59–66, all had good sight, none deaf. During last two years III, 60, female, age 61, has had difficulty in seeing at dusk, fields of vision full, no ring scotoma; My. 0·75 D. R. and L. V. with correction = 6/12 in each, in dim light giving 6/18 to normal eyes, she does not read 6/60; R. and L. O.D. good colour, retinal arteries not contracted; at macula an area, 2 O.D. in size, mottled with small yellowish spots which are interspersed and covered by small black pigment spots; in R. white crescent of choroidal atrophy next outer and lower edge of O.D., and at lower part of fundus two large oval yellowish spots with well defined and pigmented margins (probably syphilitic), rest of both fundi normal; she had two miscarriages (IV, 55) after her last child (IV, 54). Is subject to "influenza," after last two attacks in summers of 1911 and 1912 vision was left worse; Wassermann test positive. III, 63, *ob.* 5 diphtheria. IV, 49–64 and V, 1 and 6, none have defective sight. IV, 50, several *ob.* young. II, 21, unmarried. The only descendant of II, 22 with defective sight is IV, 98, a male, age 35, with symmetrical diffuse corneal opacities, dating from prolonged eye inflammation at age of 21 when a knee was also affected; fundi, no details visible. III, 71, a mental case requiring attention. II, 23, was inmate of lunatic asylum, temporarily, about age of 50, lived to over 80; a daughter (III, 85) drowned herself; IV, 116, male, age 33, has double primary optic atrophy, tabetic, R. and L. V. = no p.l. No other descendant of II, 23, has defective sight. Married and no children: Males, III, 1, 53, IV, 77, 106; females, II, 7, III, 65, IV, 115.

Fig. 16.—A single case of retinitis pigmentosa in a childship of six. III, 8 (farmer), age 54 (1912), seen, could never see well in the dark, uses his stick much in the evening when on a road to avoid the ditch, and dares not go about after dark in a strange place; remembers when a lad having difficulty in seeing a tennis ball towards sunset; had "inflammation of lungs" when a boy; some bleeding of nose when young, but never lost much blood; is not subject to cold hands or feet; radial arteries feel normal, and no increase of blood pressure; not subject to headaches; he is the 2nd born. Eyes: refraction

H. R. 1 D., V. with correction = 6/36; L. 1 D., V. with correction = 6/60; light sense much diminished; both fields of vision nearly full with symmetrical absolute ring scotoma; O.D.'s too pale, retinal arteries markedly narrowed, choroidal vessels much exposed, though not at the fundus periphery; retinal pigmentation, fairly characteristic, is limited to inner part of each fundus at an area no great distance from O.D., about 2 O.D. diameters, no white atrophic choroidal patches; Wassermann blood test negative. Mother (II, 6), married at 24, *ob.* 79, and father (II, 2), married at 48, *ob.* 62, not cousins, both saw well. I, 3, *ob.* 84, and I, 4, *ob.* 70, had good vision, not cousins. I, 1, *ob.* 92, and I, 2, *ob.* old, not cousins. III, 7, and 9 to 12, all abroad and have good vision, III, 9 and 10, unmarried. IV, 6 to 12, ages 21 to 11, none My., none deaf, no signs of syphilis, fundus normal. III, 1, and IV, 1–4, abroad, vision good. III, 2 to 6, all *ob.* grown up, saw well. IV, 5, and 13 to 15, abroad, it is said that if any had been defective in vision it would have been known to III, 8. II, 1, and 3 to 5, *ob.*, all saw well. II, 7, *ob.* young. II, 8, *ob.* III, 14, extensive rodent ulcer of left orbit, eye completely destroyed, age 74. III, 15, age 72, they and their descendants (IV, 16–20, V, 1–3), see well. III, 18–23, IV, 21–23, all abroad, often heard of, no defect. III, 24–28, and their children IV, 24–40, IV, 26, is an infant, and V, 4, have good sight. III, 25, has a goitre, no exophthalmos, tremors, or rapid pulse, her sister (III, 28) has a goitre. IV, 29, wears glasses for H., subject to epileptic fits. IV, 34, *ob.* 3, "meningitis." III, 35, a medical man, subject to cold hands and feet, and who has signs of early arterial degeneration, was seen by Mr. Marcus Gunn in 1896. From enquiries kindly made by Mr. Nettleship he seems to have had a central retinitis. III, 35, has supplied an account of the descendants of I, 5, his paternal grandfather. II, 9 and 11, were mentally affected, the latter had hæmaturia. II, 13, *ob.* angina pectoris. II, 14, *ob.* cardiac disease. II, 12, no disease remembered. II, 9 to 14, all *ob.* III, 29 and 30, mentally affected, the former in asylum. III, 31, unmarried. III, 33, *ob.*, albuminuria at birth of IV, 41, who is normal. III, 34, age 54, married, no offspring, has symmetrical cataract, his mother had cataract. IV, 43, a daughter of III, 35, has cold hands and feet like her father. IV, 42 and 44, normal.


(To face p. 196.)
Fig. 17
I
II
III
IV
V
VI
Consanguinity
Deformed
Mental.
Myopia.
Hydrocephalic
Phthisis.



III, 36, professional man. III, 37, *ob.* diabetes. III, 39, *ob.* 15 of diabetes. III, 38, has uterine fibroid. III, 40–43, all unmarried. III, 40 and 41, mentally affected. Married and without child: Males, II, 10, 11, III, 34; females, III, 3, 11.

FIG. 17.—Two cases of retinitis pigmentosa in a childship of nine. V, 78, male, unmarried, age 24 (1911), seen; illegitimate son of IV, 79, and IV, 101, who afterwards married; healthy looking farm servant; cannot see in dim light; night-blindness not noticed until he had left school, aged 14; has had no illnesses, is first born, and has incomplete deafness. When first seen on November 29th, 1902, there was fine black retinal pigmentation at extreme periphery of fundus; streaks of pigment here and there in front of retinal vessels; choroidal vessels much exposed at periphery and in patches here and there in other parts of fundus; retinal arteries at least in L. are too narrow; O.D. (?) too red, especially at outer part; very fine vitreous opacities focussed in R. only; V. = 6/12 in each; teeth well formed, some decayed molars; moderately deaf; field of vision contracted. On November 29th, 1911, very extensive moss-like pigmentation at periphery of fundus; retinal arteries much narrowed; pallor of O.D.; edge not well defined; posterior lens opacities; R. V. = 6/12 Hm. 0·5 D.; L. V. = 6/12 Hm. 0·5 D.; in dim light that gave 6/9 to a normal person, he could only read 6/60; fields of vision contracted to 18°, except at a point on horizontal line in outer parts of fields, where it reaches 22°; no ring scotoma; hearing, right ear does not hear a watch even on contact; left ear hears watch only on contact; smell normal (cloves, peppermint, assafœtida); Wassermann test negative; has had no bleedings, fevers or illness of any kind. V, 82, female, unmarried, age 18 (1911), seen; always deaf like her brother (V, 78); is timid; her father (IV, 79) says she is night-blind, but her brother (V, 80) thinks she is not; has had no illnesses or bleedings; oph.: R. and L. some vitreous opacity; retinal arteries much narrowed, veins to a less degree; O.D. colour certainly no marked pallor, hazy edge, outer margins focussed clearly, not other parts; at periphery of fundus are yellow-white spots and some pepper-like dots; in R. only two what might be called spots of pigment

are visible, one at outer, the other at inner part of periphery; these spots are black and have angles suggesting very short roots or commencing processes; no white atrophic spots or areas; in L. two similar black spots occur down out at periphery; choroidal vessels exposed; refraction; My. R. 3·5 D. in oblique meridian and Emm. in opposite meridian, V. with correction = 6/12; L. My. 1 D. in oblique meridian, 3·5 D. in opposite direction, V. with correction = 6/12; with correction in dim light that gives a normal person 6/12 she gets 6/36; fields contracted, most markedly above, though not nearly so small as those of V, 78; in R. is an incomplete narrow ring scotoma; iris blue; hearing: right ear not even watch on contact; left ear hears watch only on contact; smell normal (oil of cloves, peppermint, assafœtida); her night-blindness not noticed by her father until she was at school; seems of average intelligence; Wassermann test negative. V, 80, V, 83, V, 85, and V, 86, age 7, have normal fundus. V, 84, *ob.* age 3, of "water in head." V, 79, and V, 81, have no defect of vision. Mother (IV, 101), illegitimate, but having same parents as IV, 102–105, *ob.* about 10 years ago of pneumonia; had good vision; no miscarriages or difficulties at the birth of her children. Father (IV, 79), fundus normal. His sister (IV, 80) had "water in head," difficulty in walking, was deformed, unmarried, and probably silly. IV, 78, *ob.* age 2. IV, 76, unmarried. IV, 74, unmarried, *ob.* 25 in lunatic asylum; had fits. IV, 72, *ob.* infancy. IV, 73, fundus normal. IV, 75 and 77, had good sight. No defective vision in V, 35–57, or VI, 6–16. IV, 102 and 105, unmarried. IV, 103 and 104, had each a family of 1 or 2. III, 45, 8th in childship of 11, and her husband (III, 44) saw well. III, 41, had no issue. II, 18, 4th in childship, *ob.* 73, and II, 14, *ob.* 49 of dropsy; both had good sight. I, 2, *ob.* old, saw well. None of her descendants (II, 15–20, III, 45–68, IV, 101–157, V, 78–101) known to have defective sight, except III, 60, who became blind and deaf, vision got gradually worse, was advised to have one eye excised, vision failed within a year, and, as mentioned, V, 78 and 82. III, 68, several, some married, no particulars. IV, 144, female, deaf after inflammation of brain. III, 33, lived to over 70 and her husband (III, 32) *ob.* after 80; it is said they were related, but

not 1st cousins; both saw well. In childship III, 25–32, 25–31 saw well, and their descendants (IV, 43–71, V, 21–28) and families V, 29 and 34, are not known to have any affected members. III, 26, 27, and 30, unmarried. III, 30, deformed and small. III, 34 and 35, were reported as having been affected, but their nephew (IV, 89) says it was simply short sight, no night-blindness, sight never got worse. III, 34, *ob.* 77. III, 35, unmarried, *ob.* after 70. III, 36, unmarried. IV, 84, age 67, an active woman, good sight, was brought up with her grandparents (II, 12 and 13), and they often remarked that they did not know where the short sight in their sons (III, 34 and 35) came from. II, 13, an only child, illegitimate, vision good. Her mother (I, 1) *ob.* when II, 13, was born. II, 11 and 13, saw well. II, 9, no information regarding their children. No defect in III, 37–40, IV, 81–100, V, 67–77, except IV, 89, who reports that he has cataract which was first noticed when he was 4 or 5 years old; sees well in dim light. IV, 90, some have families abroad, no particulars. IV, 96, one had a "short-sighted eye, other eye long-sighted." V, 70–75, are children. II, 1–8, all well known to IV, 1. II, 1, *ob.* 90, II, 2, at 95, II, 3, at 82, and II, 4, lived to past 80 and became blind in old age. No visual defect known in III, 1–24, or their descendants (IV, 1–42, V, 1–20, VI, 1–5). IV, 1, age 65, has My. 4 D. in R.; L. H., fundi normal. Married and no issue: Males, III, 11, 13, IV, 63, 152, 153; females, III, 53, IV, 58, 137.

FIG. 18.—A case of retinitis pigmentosa in a childship of eight. III, 9, male, age 53 (1911), seen; factory worker, married, has difficulty in seeing in dim light and in seeing at either side of him; defect of vision and night-blindness not noticed until he was 45. R. and L. O.D. pale; retinal arteries much contracted; much retinal pigmentation, especially at periphery of retina; the appearances are those of typical retinitis pigmentosa with the exception of two patches at outer part of left fundus, which are suggestive of "choroiditis"; opacities at posterior part of lenses; R. V. 6/9, no Hm.; L. V. 6/18 imperfectly, Hm., 0·5 D.; fields of vision contracted to 10° in all directions; in 1903 fields were not so contracted, and there

was a narrow "seeing" area in each continued downwards to 45° in L. and 60° in R.; never had any venereal disease;

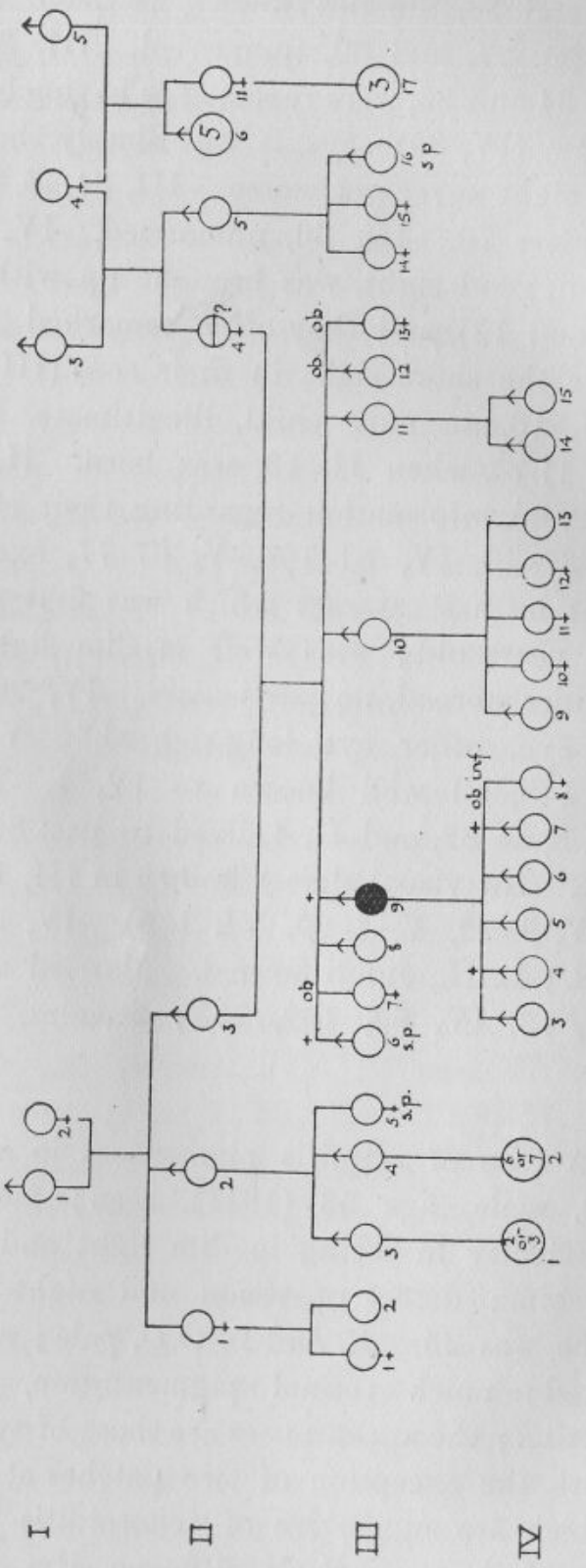
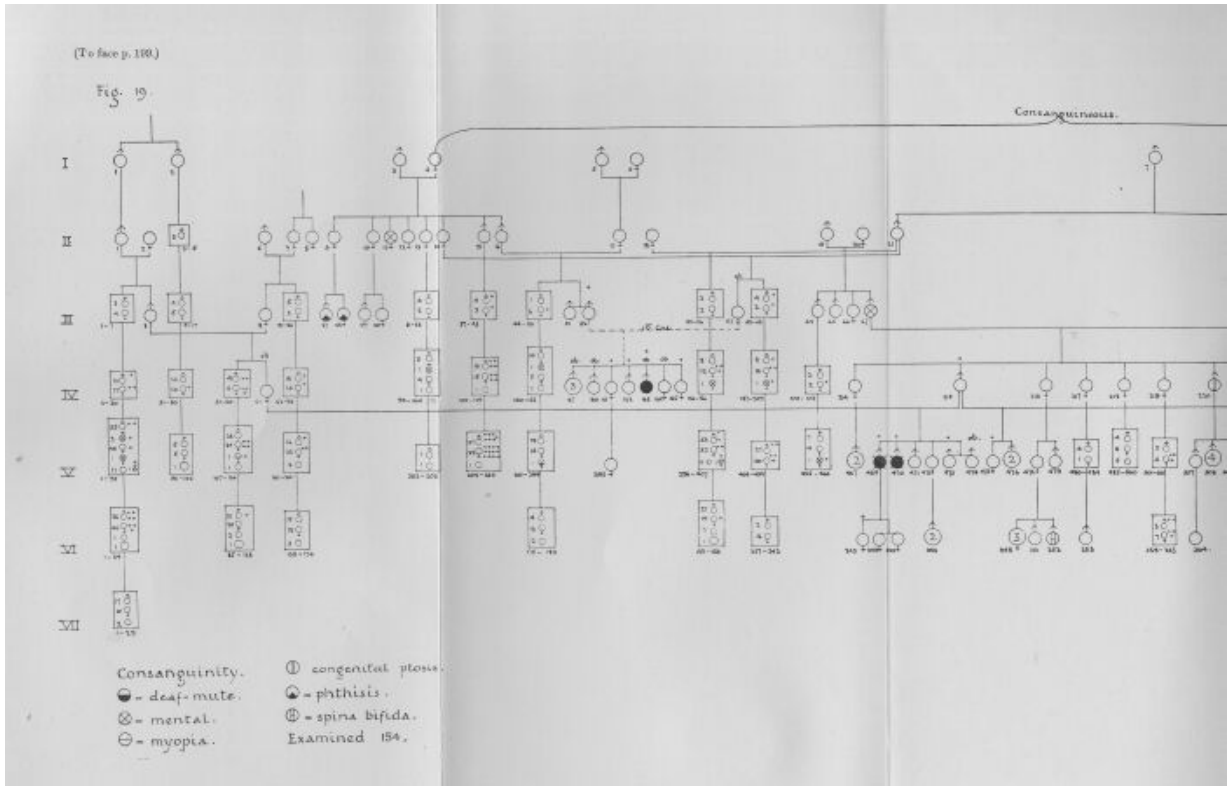
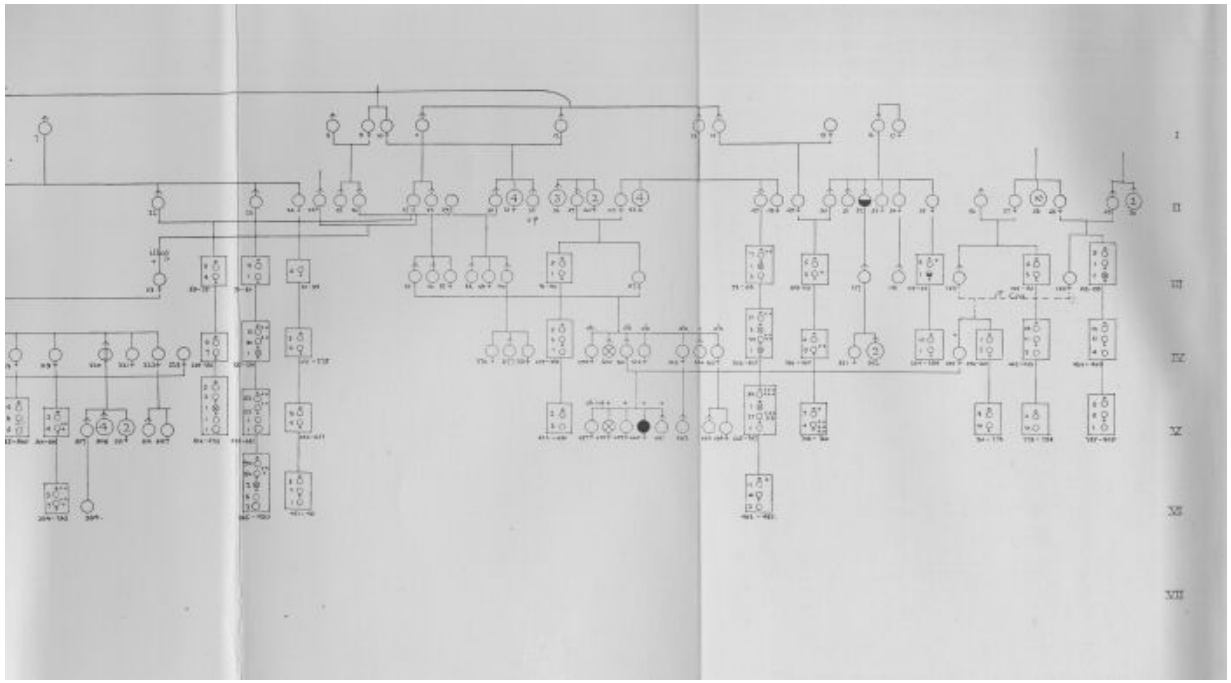


Fig. 18.

Wassermann test negative. III, 6, married without issue, vision good, no night-blindness, fundus normal. III, 7, *ob.* 5 "after


(To face p. 188.)
Fig. 19.
Consanguinity.
= deaf-mute.
= mental.
= myopia.
= congenital ptosis.
= phthisis.
= spina bifida.
Examined 154.



measles." III, 8, 10, 11, see well. III, 12, *ob.* young from accident. III, 13, *ob.* young. Mother (II, 4) *ob.* 89, was short-sighted, screwed eyes up, could see in old age, only had glasses for last few years. Father (II, 3), not a cousin of his wife, had good vision. I, 4, *ob.* 90, married twice, little known of first husband (I, 3) by whom she had 2 children (II, 4 and 5); by 2nd husband (I, 5) had 6 children (II, 6–11); all saw well. II, 11, had 3 children (III, 17) whose vision in childhood, at any rate, was good. III, 14–16, vision good. Nothing known of the families of II, 6–10. I, 1 and 2, had good sight even in old age. II, 1 and 2, and descendants (III, 1–5, IV, 1, 2) all saw well. IV, 3–7, 9–15, have good vision. IV, 4, high H. astigmatism; fundi normal. IV, 7, periodic internal strabismus; refraction R. and L. H. 8 D. in vertical meridian, 9 D. in horizontal meridian; fundus normal. IV, 13, one or two others. none with defective vision. Married without issue: Males, III, 6, 16; females, III, 5.

Fig. 19.—Two cases of retinitis pigmentosa in the same childship, a third case in another childship of same generation, and a fourth case in the previous generation. V, 469, fisherman, married, age 31 (1908), seen, with vision defective, especially at night, for three years. R. and L. marked retinal pigmentation, especially at inner part of fundus, retinal arteries narrow, fields contracted; R. V. 6/24; Hm. 1 D.; L. V. with +2 D. cyl. = 6/24. When seen again in 1912 he had just returned from a lunatic asylum; R. and L. O.D. pale; choroidal vessels much exposed; retinal arteries narrow; retinal pigmentation; no vitreous or lens opacities; no white atrophic patches in fundus; fields of vision extremely narrow; no illnesses or severe bleeding; Wassermann test negative; married, wife *ob.*, has 3 young children, VI, 243, fundi normal, and twins (VI, 244–245), no defect. V, 470, male, unmarried, age about 33 (1912), seen, is very weak mentally and physically, marked night-blindness noticed first when at school. V. each eye reads print about size of 1 J.; fields of vision much contracted; R. and L. retinal vessels narrow; O.D. pale; extensive retinal pigmentation of usual appearance; no white patches in fundus; media clear; Wassermann test negative. V, 471, fisherman,

unmarried. V, 472, and her two children (VI, 246) all see well. V, 473, fundi normal, his twin brother (V, 474) died. Mother (IV, 61), married at 26, *ob.* 40, had no defect. Father (IV, 215) married twice, age 27 at 1st marriage, by his 2nd wife (IV, 223) has 3 unaffected children (V, 475–477). III, 18, *ob.* after 70, paralytic stroke. II, 6 and 7, both lived to old age, the former was drowned. III, 8, drowned, age about 55. II, 1, *ob.* old, noted for bad temper. None of descendants of his father's brother (I, 2) have defective vision. III, 67, at age of 45 became mentally affected, recovered; *ob.* 82. III, 68, fresh old woman, age 90, fundi normal, is last survivor of 8. Her mother is II, 27, but it is doubtful whether II, 22, is her father, though no doubt exists as to his being the father of III, 69–75. It is said, and III, 61, a nephew of II, 22, firmly believes, that the father of III, 68, was not II, 22, but a ship's captain (II, 24A) from elsewhere; in support, they say that, whilst III, 68, is robust, the others in childship (III, 69–75) were feeble. The evidence is strongest in support of this view, so II, 24A, has been given as her father in the diagram. In any case, it is not through II, 22, but through II, 27, that the 2 cases of retinitis pigmentosa (V, 469, 470) are related to the other 2 cases in the pedigree, II, 27, being a daughter of I, 11, who is a brother of I, 13, from whom the 3rd case of retinitis pigmentosa (V, 660) is descended. These two brothers (I, 11 and 13) are consanguineous with, possibly 1st cousins of, I, 4, from whom IV, 163, the 4th case of retinitis pigmentosa, is descended. V, 660, female, age 16 (1912), seen, has always had difficulty in seeing at dusk, appearances of both fundi quite characteristic of retinitis pigmentosa. When examined in 1906, R. and L. extreme waxy pallor of O.D.; retinal vessels very narrow; much peripheral retinal pigmentation; no lens or vitreous opacity; refraction R. My. 1·5 D. in oblique meridian, H. 0·75 D. in opposite meridian, V. with correction = 6/60; L. H. 1·25 D., V. with correction = 6/24; fields of vision contracted, left one, taken on perimeter, is contracted to 15°, widest part is 20°, narrowest 10°. Wassermann test positive. V, 657, *ob.* 3 months. V, 658, female, mental, face shows evidence of hereditary syphilis; R. and L. retinal vessels narrow; O.D. pale; in R. moss-like pigmentation at periphery;

in L. a myopic eye, dense black pigment, not moss-like, at lower and outer part of fundus, scarcely any pigment elsewhere; R. V. = counts fingers; L. V. = 1 J.; no difficulty in getting about in dim light, quite different in that respect from her sister (V, 660). V, 659, female, had choroido-retinitis when seen in 1894 at the age of 3.* V, 661, fundi normal. Mother (IV, 395), age 44, had no miscarriages, but was treated at one time by her doctor for syphilis; fundi normal; V. = 6/6 in each; fields full. Father (IV, 301), *ob.* alcoholic, his brother (IV, 304) says he had venereal disease. IV, 300, in lunatic asylum. IV, 302, *ob.* cripple, result of an accident. IV, 303, *ob.* heart. IV, 305, *ob.* renal. IV, 396–401, a male and a female, *ob.* inf. III, 124, and III, 132, are 1st cousins, their mothers (II, 57, and II, 58) being sisters. III, 97, her maternal grandfather is I, 13, brother of I, 11, and consanguineous with I, 4. II, 29, mother of III, 85, married twice. Her 2nd husband (II, 30) is a son of I, 12, their 3 children (IV, 88–90) see well, and grandchildren (V, 276–278) abroad. IV, 163, fisherman, unmarried, age 30 (1897), seen, "gradual weakness in both eyes for 20 years." R. and L. retinal pigmentation of usual type at periphery of fundus, but not extending to extreme periphery; no white patches in fundus; O.D. dirty white colour; retinal vessels much contracted; choroidal vessels clearly visible; a few vitreous opacities; refraction My. R. 7 D. in oblique meridian, 9 D. in opposite meridian, V. with correction = 6/18; L. 5 D. in horizontal meridian, 6 D. in vertical meridian V. with correction = 6/18; fields of vision contracted, right field, taken on perimeter, contracted to 15°. In 1904 retinal vessels like threads; all round periphery of fundus spider-like pigmentation; fields of vision contracted; night-blindness very marked; posterior cortical cataract. In 1906 R. V. = 6/24, L. V. = 6/18 with correction. Fields R. contracted to 10°, L. 10° extending on temporal side to 30°.

* Seen again in 1913. R. V. = 6/18; the choroido-retinitis affects the whole fundus; macula is least affected; most of the pigment at the periphery; no spots of complete choroidal atrophy; O.D., pale; retinal arteries, narrow; pigment in large heavy masses and in circular forms as well as some moss-like: field much contracted. L. V. = no p.l.; much vitreous opacity; no fundus details.

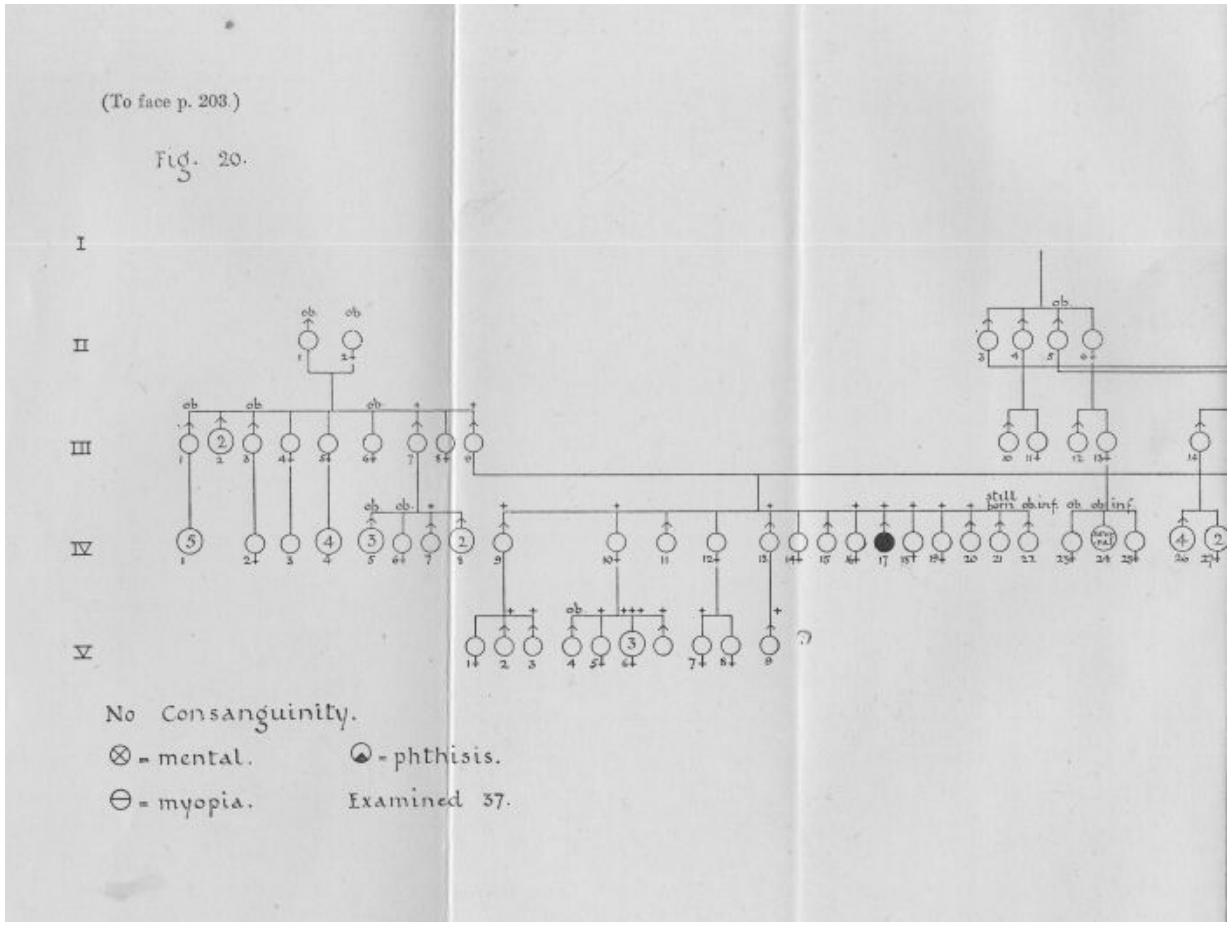
Dr. Souter noted that pigment comes well in from periphery; O.D.'s peculiar pallor, rather waxy than white. He is now (1912) dead. No Wassermann test. He was exposed to wet and cold at age of 18, sleeping in wet clothes. Never bleedings. IV, 157, 3 males, *ob.* young, croup. IV, 160, *ob.* 33, influenza. IV, 162, is peculiar, seldom does any work, wanders about with a gun aimlessly. Mother (III, 57), 24 at marriage, *ob.* 55 influenza; 1st cousin of husband. Father (III, 52), twin brother of III, 51, who *ob.* young, married at 26, age to-day 79 (1912), fundi normal. II, 21, brother of II, 22, who is the husband of II, 27, and possibly the father of III, 68, the paternal grandmother of two males with retinitis pigmentosa (V, 469, 470). II, 16, *ob.* 59, had a mental sister (II, 11), their mother (I, 4) has not only the same name as I, 11–14, but also the same nickname by which their branch of the family is distinguished, she is possibly their 1st cousin. It is remarkable that 12 of the 17 persons in Generation I have the same surname. None appear to have had defective vision, and no case of retinitis pigmentosa has been found in their descendants besides the 4 described above. The 4 affected individuals belong to a common stock. Three of these (V, 469; V, 660; IV, 163) came up independently of each other, and it was not known that they were related. They live at fishing places. V, 469 and 470, are at a distance of 11 miles from IV, 163, there is no railway to their villages. V, 660, is 16 miles from V, 469 and 470, and 4 or 5 miles from IV, 163.

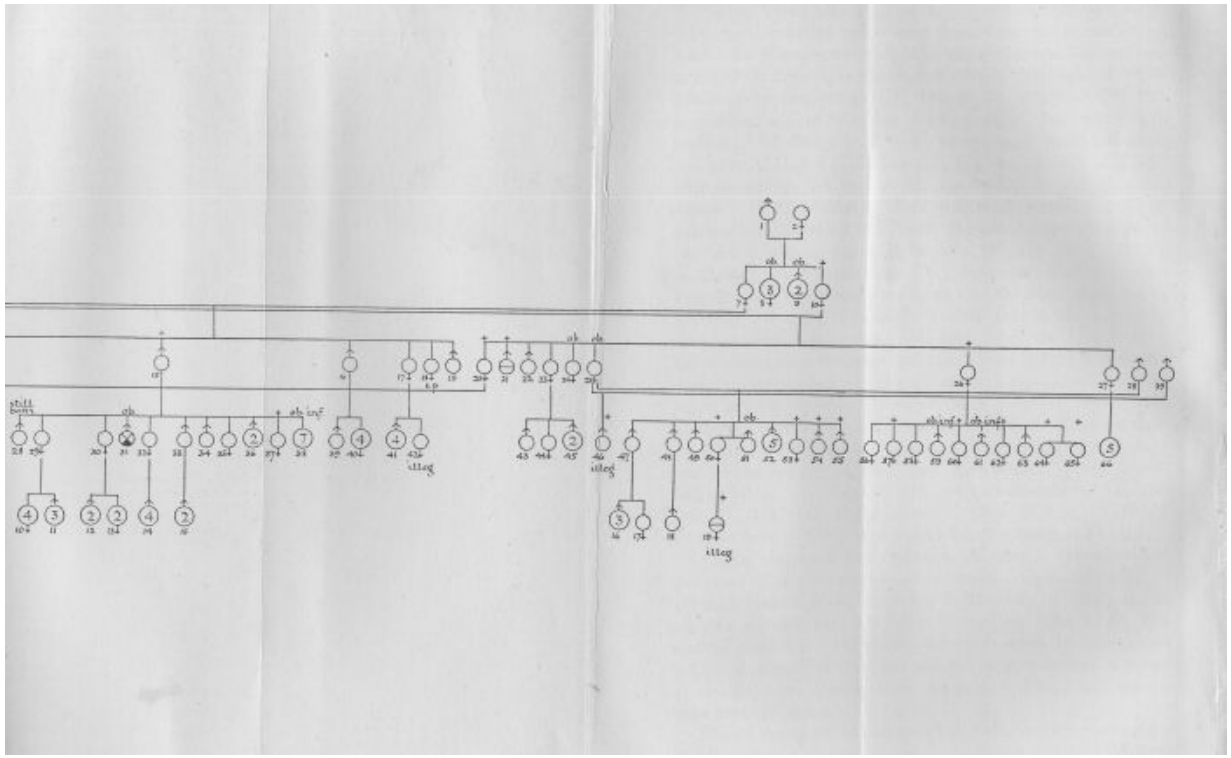
Cousin marriages are recorded 21 times. These are not indicated in the figure, necessarily much condensed, which is primarily intended to show the relationship of the cases of retinitis pigmentosa, as so many connecting lines would give rise to confusion. 1st cousin marriages occur 10 times; 2nd cousin marriages twice; 3rd cousins once removed 4 times (1 double); 1st cousins once removed 3 times; 2nd cousins once removed twice.

Mental cases—26 are known, but there are few particulars. 7 are mentioned as having been in a lunatic asylum. V, 717, described as "soft." V, 383, "wanders about." V, 36, didn't speak, his people had no control over him, they would not allow a fundus examination, as they thought it would excite him too much. VI, 440, imbecile. VI, 441, "mental, has big head,

(To face p. 203.)

Fig. 20.





cannot walk." III, 67, mentally affected at 45, recovered. III, 139, "silly." IV, 186, "simple," in poorhouse. IV, 270, mental, in a home. IV, 151, insane, in asylum.

Deaf-mutes, 2. II, 52, and III, 123.

Myopia in only 2 of the 154 examined cases. V, 52 and 108.

Congenital ptosis (R.). V, 220, ♂.

Married and no issue: Males, 14 (II, 35, III, 9, 23, 53, IV, 13, 32, 74, 94, 272, 315, 329, V, 305, 467, 525); females, 26 (III, 7, 35, 50, IV, 16, 42, 49, 55, 126, 127, 197, 221, 251, 256, 268, 274, V, 40, 44, 181, 282, 353, 359, 440, 579, 587, 588, 580).

Twins occur 11 times in 4 generations.

FIG. 20.—Retinitis pigmentosa and lamellar cataracts in same individual, a male, in a childship of 14. IV, 17, male, age 17 (1908) seen, well nourished, never had fits, is fidgety, teeth and palate normal, had mumps and measles when at school; when 4 years old fell against a broken pail and cut bridge of nose, is said to have lost much blood but the evidence is not convincing; defective vision noticed at age of 6 or 7, has been getting worse; typical symmetrical lamellar cataracts which were removed by curette extraction, the left one in 1908, the right one in 1909; after operation, in each, the course was uneventful, no iritis; retinitis pigmentosa was not detected until after removal of the lenses. In May, 1909, in each eye there was marked retinal pigmentation with moss-like character all over periphery of fundus; retinal arteries not markedly contracted; O.D. had a good colour; night-blindness; fields of vision markedly contracted; refraction before operation R. My. 2·5 D., V. with correction = 6/60; L. Emm. V. = 6/60; after operation R. H. 11 D., V. with correction = 6/36; L. H. 10 D., V. with correction = 6/60. In November, 1912, R. and L. central vision and refraction were unaltered, in dim light allowing a normal eye to read 6/12 he could read only 4/60; right field of vision was reduced to 10° and left field was rather less; retinal pigment appears to have increased; Wassermann test negative. Mother (III, 20), age 54, age at marriage 20, R. and L. media clear, fundus normal. Father (III, 9), age 64, age at marriage 35, R.

and L. media clear; fundus normal. His parents (II, 1, and II, 2), brothers and sisters (III, 1–8) and their descendants (IV, 1–8) have no defect of vision except III, 7, with senile cataract in right eye, no opacity in left, no mydriatic used. II, 10, media clear, fundi normal. II, 5, *ob.* 77, vision good. I, 1, and I, 2, both *ob.* about 50, had good vision. IV, 9–20, all alive, oldest 33, youngest 14, have good vision except IV, 17. IV, 21, stillborn. IV, 22, *ob.* 2 months. V, 1–3 and 5–9 see well. V, 5, age 12. II, 3, 4, 6 and descendants, III, 10–19, IV, 23–42, none with defective vision. IV, 23, *ob.* 18. IV, 31, *ob.* of phthisis in lunatic asylum; his insanity said to be derived from his maternal grandmother who was insane, and his mother at one time was queer. II, 7, age 80. II, 8, 2 of them and possibly the 3rd married, no defect known in descendants. III, 21, unmarried, R. My. approximately 5 D., L. traumatic cataract. III, 25, held things close to her, not night-blind. III, 23, 26, 27 and descendants and descendants of III, 25, namely IV, 43–66, and V, 16–19, see well except IV, 47, a bus driver, and IV, 49, who are short-sighted, hold objects near to them, require no glasses, no night-blindness, and V, 19, with R. My., L. mixed astigmatism.

FIG. 21.—Three males and one female, deaf-mutes, with retinitis pigmentosa in a childship of 13, and two males and two females, deaf-mutes, probably with retinitis pigmentosa, in another childship of same generation. IV, 34, male, in blind asylum, age 33 (1906), seen, a deaf-mute; R. and L. O.D. pale; retinal vessels extremely narrow; choroidal vessels much exposed; extensive retinal pigmentation extending to the O.D.; fields of vision very much contracted; has night-blindness; refraction, R. H. about 2 D., V. counts fingers at 6 feet. L. H. 1·5 D., V. reads 16 J. Married for 8 or 9 years, no offspring, Wassermann test negative; his wife (IV, 40) has good sight and hearing; sound teeth; sense of smell normal. IV, 35, male (1911) seen, farmer, deaf-mute, unmarried; posterior cortical cataracts; marked retinal pigmentation; O.D.'s pale; retinal vessels very narrow; night-blind; to-day (April, 1911) reads newspapers with high magnifying glass; in 1894 R. with +2 D. cyl. V. was 6/36; L. with +2 D. cyl. V. was 6/18; iris dark; upper central incisors absent, other teeth good; sense

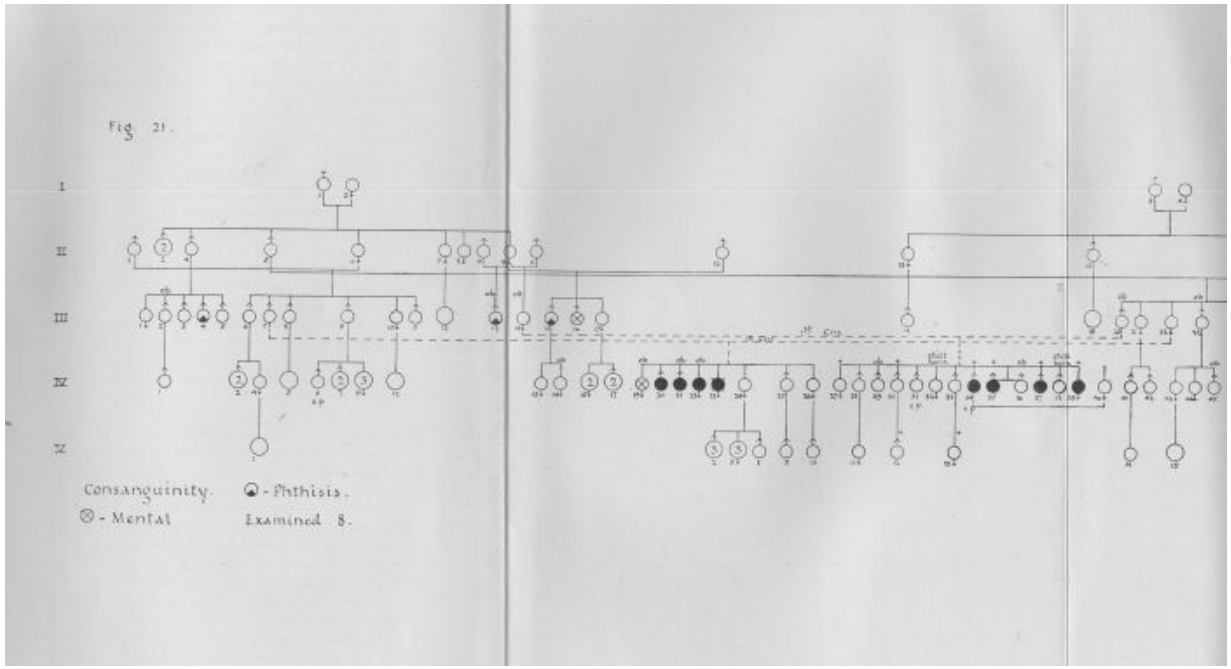
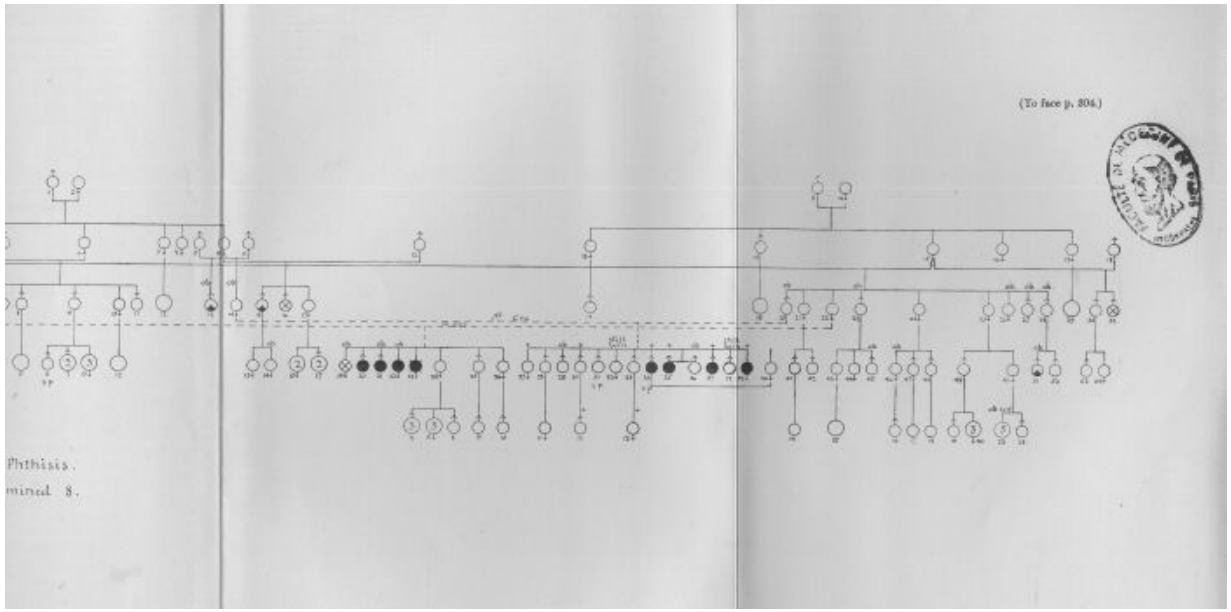


Fig. 21.
I
II
III
IV
V
Consanguinity.
◒ - Phthisis.
⊗ - Mental
Examined 8.


(To face p. 804.)
Phthisis.
mined 8.

of smell good; R. divergent strabismus. IV, 36, a twin with IV, 35, *ob.* 4 months and 10 days of "teething fever"; not known whether he was deaf. IV, 37, unmarried, seen in 1911, commencing posterior cortical cataracts; very extensive retinal pigmentation, more than in IV, 35. R. and L. O.D. pale; retinal vessels narrow; refraction of each low H., V. can get about by himself; with +3 D. sph. reads newspaper; night-blindness; is a deaf-mute. Defect of vision first discovered in IV, 34, 35, 37, and 39, during first year of school life. IV, 39, unmarried, age 30 (1911), seen, stout, healthy, very intelligent, deaf-mute; good teeth; R. and L. no cataract; retinal pigmentation confined to periphery of fundus, not much pigment; O.D. pale; retinal vessels narrow; night-blind; refraction R. H. 1 D. in vertical meridian and 2·5 D. in horizontal. L. H. 2 D. in vertical meridian and 2·5 D. in horizontal meridian, very defective though with strong glass reads newspapers. When seen in 1905 R. V. = 6/24; L. V. = 6/24; fields of vision both very much contracted; sense of smell good. None of these 4 cases (IV, 34, 35, 37, and 39) have had any severe illness or bleedings; no evidence of syphilis. IV, 38, stillborn. IV, 27, healthy, bilious headaches sometimes, age 52, fundus normal. IV, 28, and V, 11, abroad, vision good. IV, 28, is sometimes "livery." IV, 29, *ob.* 16, typhoid fever, heard and saw well. IV, 30, fundus normal. V, 12, infant, hearing normal. IV, 31, hearing and seeing good, married 7 or 8 years, no offspring. IV, 33, good sight and hearing. V, 13, baby, fundus normal, hearing good, healthy. Mother (III, 14) married young, was 3 years older than husband, excellent sight and hearing and smell; *ob.* 48 at "change of life," was weak for 2 or 3 years previously. Father (III, 20), married before 20, is 1st cousin of his wife, saw and heard well; *ob.* 56. II, 10, married 3 times, her husband II, 11, was no blood relation. She had one child (III, 13) by II, 9, that *ob.* 19 of phthisis; by II, 12, had 3 children (III, 15–17). III, 15, *ob.* phthisis, age 33. III, 16, single, weakly in body and mind, did odd jobs, was nearly silly. III, 17, *ob.* 50. IV, 13, not strong. IV, 14, *ob.* 18 of meningitis. IV, 15 and 17, unmarried. I, 2, *ob.* 80, probably saw well and not deaf, for IV, 27, often heard her mother (III, 14) speaking about her and her husband (I, 1) who predeceased his wife. II, 15, *ob.* 83,

good vision and hearing, first husband (II, 5) not a cousin, *ob.* 57 apoplexy, saw and heard well. Her second husband (II, 18), not a cousin, had 2 children (III, 30 and 31) by her, one (III, 31) was silly, the other (III, 30) has 2 children (IV, 53 and 54), ages 8 and 6, with good sight. I, 3 and 4, not known to have had defective sight or hearing. IV, 20 to 23, all deaf and dumb and have defective vision, abroad. IV, 20, a farmer. IV, 21, *ob.* age 15, when at school. IV, 22, *ob.* age 5. IV, 23, abroad, inmate of asylum. On May 6th, 1911, the medical superintendent kindly reported in reply to a letter: "The patient you refer to is enjoying good health, but she is a deaf-mute and the sight is almost gone. She no doubt has retinitis pigmentosa." IV, 19, single, *ob.*, "soft." IV, 24, 25, 26, and V, 2–10, have good sight and hearing. III, 22, *ob.* 66, influenza, married 1st cousin. III, 7, crippled with rheumatism, vision and hearing good. II, 1, and II, 6, saw well. II, 2, both unmarried, one *ob.* II, 4, and III, 1–5, good vision. III, 1, 3, and 5, unmarried. III, 4, *ob.* phthisis. No particulars of family (III, 12) of II, 7. II, 8, *ob.* young. II, 13, still alive (1911), and III, 18, see well. No particulars of III, 19 and 29—families of II, 14 and 17. No defect of vision known in III, 21, 23–31, and descendants IV, 41–54, V, 14–28. III, 23, drowned. IV, 45, *ob.* 5. III, 26, *ob.* young. IV, 51, *ob.* phthisis. V, 15, a family, no particulars. V, 16–18, infants. V, 19 and 20, ages from 8 to 2. V, 28, age 7. III, 6, and IV, 1–4, abroad, none affected. III, 9, and IV, 6–9, abroad, all see well. IV, 12, and V, 1, families, no particulars. IV, 5, large family, no deaf-mutes, no defect of vision known, abroad.

FIG. 22.—An isolated case of retinitis pigmentosa commencing late in life. III, 15, female, 7th in childship of 8, age 64 (1905), seen, not deaf, night-blindness first noticed about age of 55; no miscarriages; severe bleeding at time of menopause confining her to bed. In light allowing 6/12 to normal eyes she cannot see 6/60; refraction H. R. 1 D., V. with correction = 6/9, L. 1 D., V. with correction = 6/9; right field of vision taken on perimeter is full or nearly so; R. and L. O.D. pale, rather yellowish; lamina cribrosa much exposed; retinal arteries narrow, superior main artery on right O.D. has a short constriction; typical

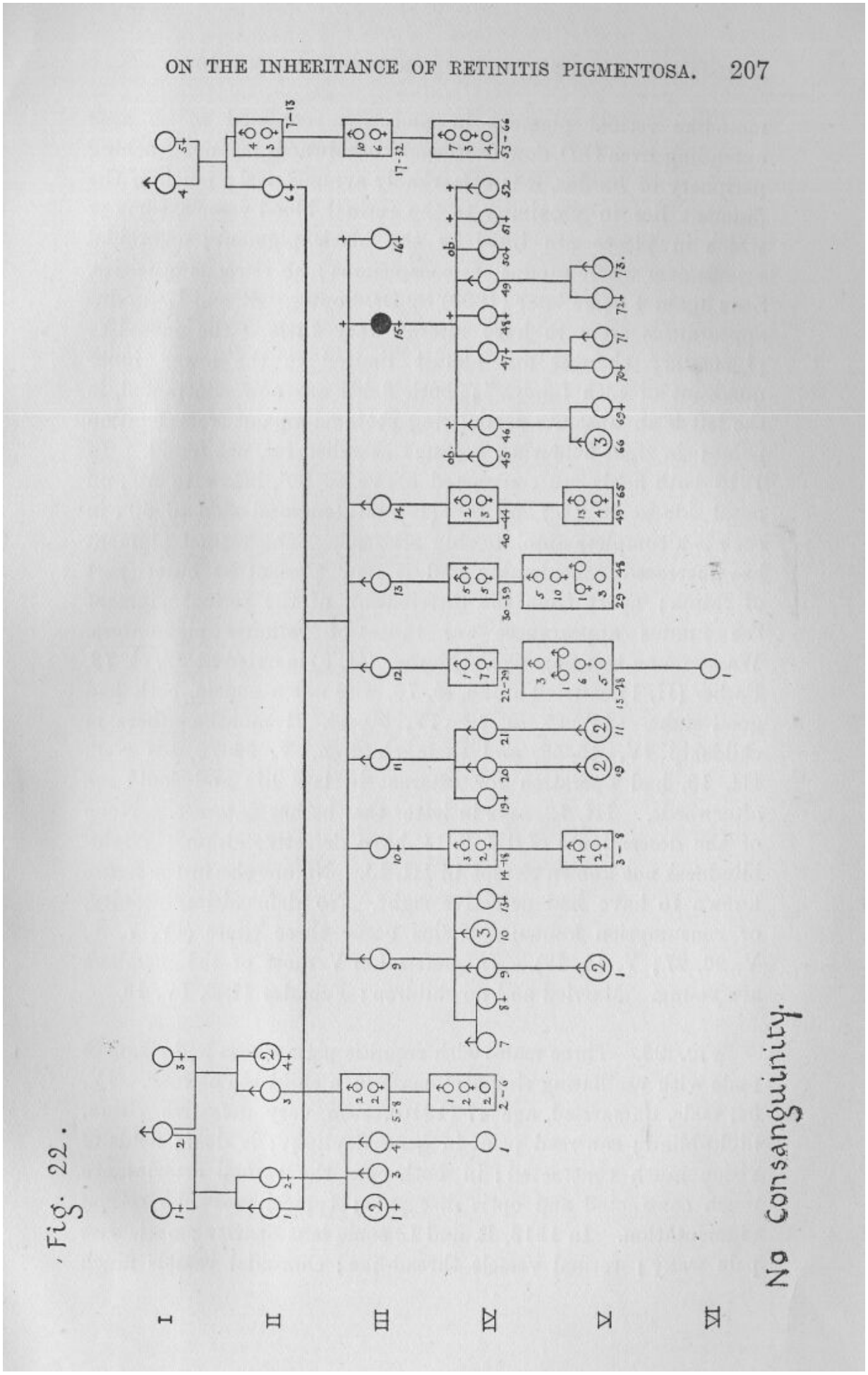
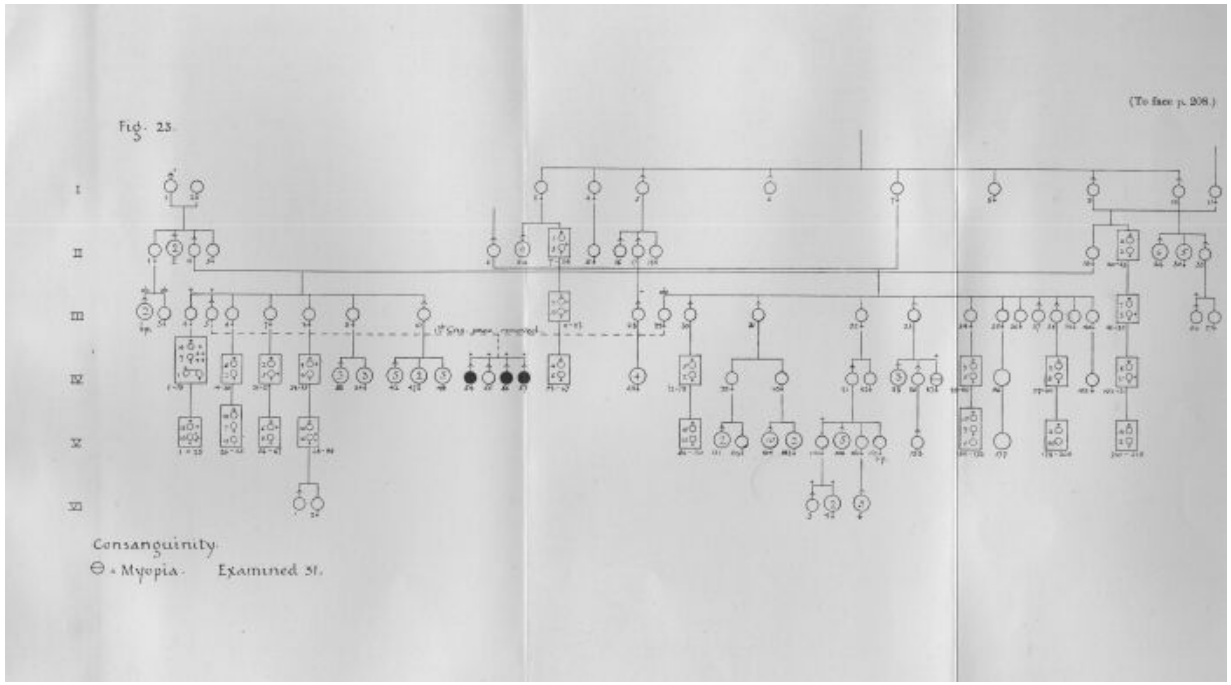


Fig. 22.

moss-like retinal pigment in each eye confined to an area extending from O.D. downwards and nasalwards but not reaching periphery of fundus, it is remarkably symmetrical; much of the pigment lies in proximity to the retinal blood vessels, some of which in places are lined by the black pigment; choroidal vessels over whole fundus are conspicuous; no vitreous opacities. Seen again 4 years later (1909) by Dr. Souter. R. and L. fundus appearances seem to have altered very little, "the moss-like pigmentary changes are almost limited to the lower inner quadrant of each fundus"; both fields are now contracted, in the left is an absolute partial ring scotoma up-out from fixation point; in right field ring scotoma searched for, not found. In 1913 both fields are contracted above to 30°, below to 50°, on nasal side to 40° (L.) and 50° (R.), on temporal side to 60°, in each is a complete absolute ring scotoma. The retinal pigment has increased considerably and is now present at outer part of retina; apart from the distribution of the retinal pigment the fundus appearances are those of retinitis pigmentosa. Wassermann test negative. Mother (II, 6) married at 22, *ob.* 73. Father (II, 1) married at 26, *ob.* 70, wife not a cousin, both had good sight. IV, 45, *ob.* 4. IV, 50, *ob.* 21 months, others in childship IV, 45–52, and their children, V, 66–73, see well. III, 13, had operation for cataract in late life and could see afterwards. III, 12, says in letter that he has cataract. None of the descendants of III, 9–14, have defective vision. Night-blindness not known except in III, 15. No one else in the figure known to have had defective sight. No deformities, insanity, or consumption known. Twins occur three times (IV, 7, 8; V, 26, 27; V, 41, 42). In generation V most of the members are young. Married and no children: Females II, 5, IV, 16.

FIG. 23.—Three males with retinitis pigmentosa and a fourth male with vacillating ring scotomata in a childship of four. IV, 54, male, unmarried, age 47 (1910), seen, very defective vision, night-blind; can read print in good daylight; is deaf; fields of vision much contracted; in both eyes the retinal arteries are much contracted and optic disc pale; typical moss-like retinal pigmentation. In 1913, R. and L. some lens opacity; optic disc pale waxy; retinal vessels thread-like; choroidal vessels much


(To face p. 208.)
Fig. 23.
I
II
III
IV
V
Consanguinity.
⊖ = Myopia. Examined 31.

exposed; much retinal pigment towards periphery; refraction My. R. 3 D., V. with correction = 6/24, L. 3 D., V. with correction = 6/24 in good daylight; night-blindness is very marked; fields of vision could not be measured on perimeter; no defect of smell. Wassermann test negative. IV, 55, see notes below. IV, 56, male, unmarried, age 39 (1910), seen, is deaf; fields of vision much contracted; fundus (both) has typical appearance of retinitis pigmentosa; posterior cortical cataracts; marked night-blindness. IV, 57, male, unmarried, age 34 (1910) seen on May 1st, 1909, is deaf and has defective vision, gradually getting worse. R. and L. V. of each is hand-movements at 5 metres; pupils equal and sluggish to light; oph. R. and L., with dilated pupils after mydriatic; posterior cortical cataract; retinal pigmentation most marked midway between optic disc and periphery of fundus, not much of it is moss-like, some is arranged in circles or round patches like choroidal pigmentation; no white spots; retinal arteries very narrow, a mere streak of blood is visible in them; fields of vision reduced to less than 5°. Mother (III, 29), *ob.* 62, married at 20, at age of 35 had an illness, in bed for 2 years, "paralysis of voice, one side of body paralysed, headaches and vomiting, vision became affected at that time and gradually got worse, quite blind in one eye at time of death" is the account of this illness from relatives. Father (III, 5), fundus normal; married at 25; his maternal grandfather (I, 9) was the brother of his wife's mother (I, 7). II, 19, *ob.* 78, never saw well, not night-blind, could see far objects, did not hold objects near, never wore glasses, was cut for cataract, saw well afterwards. II, 4, 4th in childship, *ob.* 52. I, 1, *ob.* 95. II, 6, not a cousin of his wife (I, 7). I, 2, had good vision. I, 9 and 11, not cousins, saw well. None in generations I and II known to have had defective vision except II, 19. II, 2 and 3, unmarried, *ob.* about 60. II, 5, unmarried, *ob.* 83. I, 8, and II, 16, unmarried. III, 3, *ob.* unmarried. No descendant of II, 20–35, known with defective vision. II, 24 and 30, have descendants, no defect of vision known in them. None of the father's brothers and sisters (III, 4, 6–10) or their descendants (IV, 1–53; V, 1–95; VI, 1, 2) have defective sight. III, 4, fields of vision full, retinal arteries of good size, both fundi normal, lens opacities, age 73. The two sisters, III, 6 and 8, married twin

brothers not shown in figure. IV, 14, male, had 8 children, sex not known (V, 26), none lived longer than a few hours. All *ob.* in childship III, 29–40, but wife of III, 33, seen, III, 36, 37, 39, unmarried, none of them had bad sight nor descendants of III, 30–35, 38, 40, except IV, 87, who has a low degree of myopia, V, 193, 194, two sisters, abroad, "near-sighted," not night-blind, do not use glasses and IV, 81, age 65, vision always defective, his daughter (V, 145) says when sowing corn she had to walk beside him to let him know where he had already sown; no particular difficulty in dim light; reads with glasses. IV, 96, and V, 177, abroad, no particulars. V, 151, abroad, with 5 sons (VI, 6), no information as to their vision. II, 6*a*, all had issue (not shown in figure), 12 male and 14 female children, and 4 male and 2 female grandchildren, none affected. III, 11–28, and IV, 58–68, defective vision unknown. Married and no child: Males, III, 1, 2, IV, 83, V, 146; females, IV, 6, 7, V, 152. Letters from III, 8, IV, 34, 72, 80, 88, 100.

IV, 55, male, unmarried, age 45 (1910), seen, "nothing was thought about his sight" although the parents knew it was not right. The defect of vision in his brothers was noticed as soon as they could walk. On May 1st, 1909, his fields of vision were contracted though not markedly and there was an absolute complete ring scotoma in the left field of vision (Fig. 1, p. 140); oph. with dilated pupils after homatropine, no retinal pigmentation; optic disc of good colour, retinal vessels of good size. On August 19th, 1910, R. V. with + 1 D. cyl. = 6/9: L. V. with + ·5 D. sph. and + 0·75 D. cyl. = 6/9; with correction, in dim light he read 6/36 when two normal people saw 6/12; fundus as on last examination; fields of vision still contracted, but there is no trace of the ring scotoma in left field (Fig. 2, p. 140). In 1913 (February 19th), R. V. = 6/12 with correction, L. = some of 6/9; in darkened room, 6/60 when others read 6/18; R. and L., no retinal pigmentation; O.D. good colour; retinal arteries not contracted. Fields of vision not much contracted, 10 mm. white square used; in right eye is a complete absolute ring scotoma (Fig. 3, p. 142), and on temporal side, between the scotoma and fixation point, is a partial absolute ring scotoma (these were measured with a 5 mm. white square); in left field is a partial absolute ring scotoma, corresponding

in situation to that in right field; fingers white to-day; no defect of smell. Is subject to cold hands and feet, his fingers become white; gets frontal headaches. Wassermann test negative. Dr. A. W. Falconer has kindly reported that the patient apparently has typical local syncope (Raynaud's disease), affecting the three outer fingers of both hands. His vessels are perhaps not so good as they should be. There is no definite cardiac hypertrophy, and his urine contains neither sugar nor albumen. In a later note (June 9th, 1913) he says: Heart: Apex beat in 5th space just inside nipple line. Well-marked, somewhat musical, apical systolic murmur. Second sound at the aorta not markedly accentuated. Cardiac rhythm normal. Radial vessels moderately degenerated. Blood pressure in each 150 mm. Lungs normal. Abdomen: Scar in epigastrium from operation 5 years ago. Some general tenderness over whole of epigastrium, but especially over aorta. Liver and spleen not enlarged. Mouth: marked pyorrhœa in lower jaw, upper teeth mostly gone. Nervous system: nothing abnormal. Urine: no sugar, no albumen.

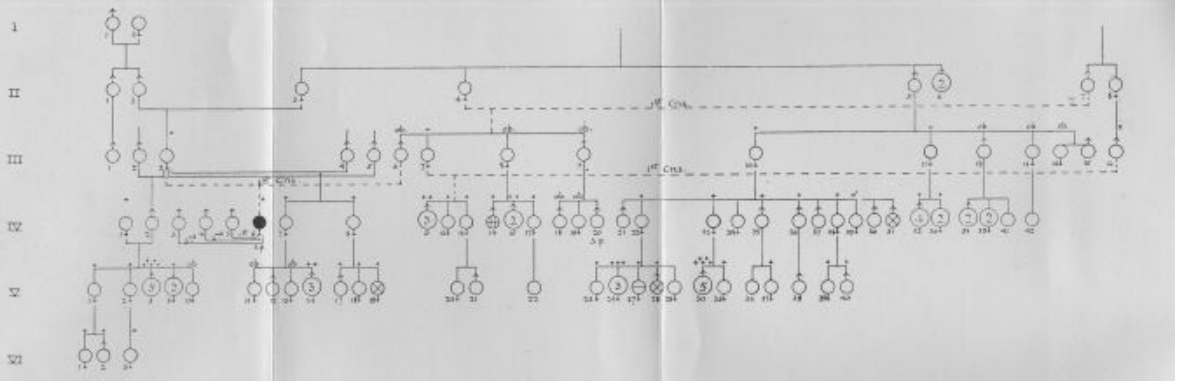
Note.—IV, 55, appears to have had a transient ring scotoma. He works on a farm with his three brothers. He takes them to and from their work. He says that he has no difficulty in dim light, yet examination showed that he has some night-blindness. When examined in 1909 he made no complaint, and had come merely to look after his brother, IV, 57. He was examined because he was a brother of a case of retinitis pigmentosa. The presence of a ring scotoma in one eye at that time was unexpected; its absence 15 months later was more surprising. Both results were obtained under similar conditions; a 10 mm. white square was used and daylight, about midday, the former in May, the latter in August. The reappearance of a partial absolute ring scotoma in left field of vision, and a similar one in right field, with, in addition, a complete absolute ring scotoma, suggests that these scotomata may in some cases come and go. The partial ring scotoma of February, 1913, in left field of vision is in same part of field as the corresponding part of the complete ring scotoma of 1909. A 5 mm. white square was used to mark the ring scotomata in February, 1913. In looking for an explanation of this transient scotoma, the liability of this man to cold

hands and feet, with whitening of the fingers, makes a vascular origin seem probable, and Dr. Falconer's report shows that the man is the subject of Raynaud's disease.

June 6th, 1913.—Several weeks ago he was asked to notice any variation in his sight. He says that the vision does vary. He sees better on some days than on others; sees best on warm days; vision is worst when fingers are white, and this may last for 2 or 3 days; has sometimes to place his hands in warm water to relieve the pain, which may keep him from sleeping; toes also affected. Apparently the vision does not invariably get worse when his fingers are white, for to-day he says his sight is not so good, it began to get worse last night when his fingers, which had been white for 2 days, were recovering. His fingers just now are swollen and reddened, and there is puffiness at back of right wrist. The following additional information has been obtained:—The angio-spastic attacks have been worse for 5 years since an abdominal operation for ulcer of stomach, but he had attacks before this for at least several years. Professor Marnoch, in October, 1908, did gastro-enterostomy for gastric ulcer. He believes that his vision has been definitely defective since the operation. *Migraine-like Attacks.*—For several years he has had attacks beginning with giddiness and then headache on left side, and a mist which, beginning on left side, comes down over his eyes. He has to lie down during the attacks, which last for some hours. These occur about once in 2 weeks; formerly they were less frequent. He has had no fits; is not nervous, "the very opposite," he says; no nose trouble. Two years ago, when driving home, he lost consciousness. On recovering, 48 hours later, he had "paralysis," evidently not complete, of both arms and legs; could not walk for a month. His doctor says that he had a similar, but less severe, attack 2 years before the second attack. It seems doubtful to-day whether retinal arteries are at all narrowed or optic discs too pale. Fields of vision taken again to-day (Fig. 4, p. 142). The right field has again changed. There is no complete ring scotoma, but two partial ones, one in temporal half of field approximately occupying the same situation as the inner partial ring scotoma of February 19th, and extending into upper part of field; the other, in upper part of field, lying between 20° and 30°. About an hour later, his hands

(To face p. 213.)

Fig. 24.



Consanguinity.
⊗ - Mental ⊕ - Hydrocephalus.
⊖ - Myopia Examined 54.

in the interval having been placed in ice-water for 5 minutes, the second or upper scotoma had disappeared, and the lower scotoma had altered and become smaller, being limited to the lower outer quadrant. The left field has an absolute scotoma in lower outer quadrant, but smaller and not quite corresponding in situation to the one found in February. Taken about 1 hour later, after hands were in ice-water for 5 minutes, no scotoma could be detected in any part of the field, either absolute or relative. There is no reversal of the blue and red colour fields, and no gradual contraction of the white field was seen when trying for the presence of a spiral field. Both fields for white are somewhat contracted. June 13th, 1913, fields (Fig. 5, p. 143) both have scotomata, which have again changed in size and position. The scotomata are relative, except part in lower outer quadrant of right field. Each fundus examined with wide pupils after homatropine appears to be quite normal, O.D. good colour, retinal arteries not narrow, and no retinal pigment; some fine translucent opacities far forwards in each vitreous. R. V. $\bar{c}+1$ D. cyl. = 6/9; L. V. $\bar{c}+1$ D. cyl. = 6/9.

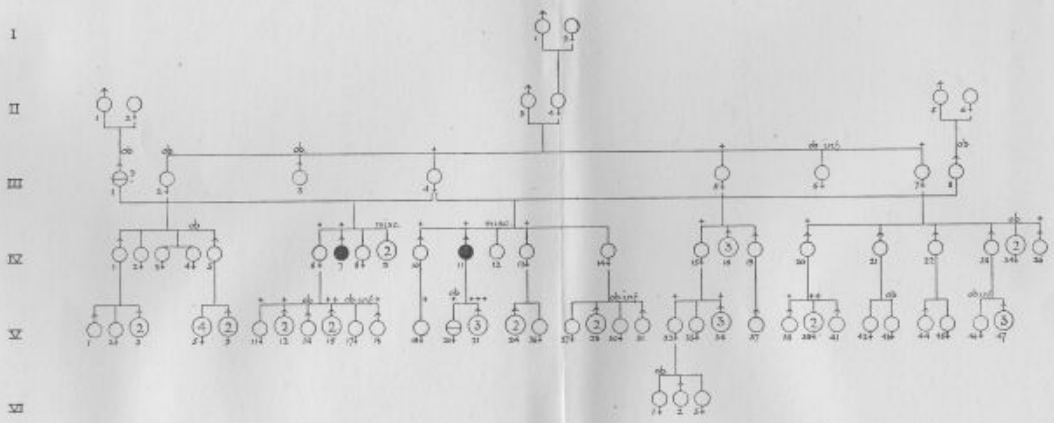
FIG. 24.—An isolated case of retinitis pigmentosa. IV, 6, female, age 24 (1898), seen, with night-blindness and difficulty in reading and sewing for 12 years. R. and L. marked retinal pigmentation at periphery of fundus except at outer part, no white patches; O.D. dirty reddish colour, paler than normal; retinal vessels, especially arteries, very narrow; at nasal part of left fundus, between O.D. and pigmented area, is a speckled, pepper-like distribution of pigment; refraction, My. R. 2 D., V. with correction = 6/12, L. 1·75 D., V. with correction = 6/12; fields of vision peripherally contracted, left to within 20°, right at temporal part to 30°, at other parts to within 20°. Seen again in 1907: R. and L. small posterior cortical cataract; O.D. waxy; extremely narrowed arteries; retinal spider-like pigment at periphery and approaching central region of fundus, especially to nasal side of O.D., but not involving macula; R. V. with correction = 6/12, L. V. with correction = 6/18; right field of vision is more contracted, left one unaltered; she is slow mentally; married three times, no issue; after third marriage both ovaries were removed; she was an only child and delicate;

when 6 weeks old her mother had "spotted (black) typhus fever." Wassermann test positive. Mother (III, 3), age 65 (1911), quite blind from primary glaucoma, an only child, at age of 27 married her first cousin (III, 6) when he was 21; she had no children by second husband (III, 5), but has an illegitimate male child (IV, 2) by III, 2, and two illegitimate children (IV, 7 and 8) by III, 4. II, 2, *ob.* 78, II, 3 and 4, both *ob.* old. II, 4 and 7, were first cousins. In childship II, 7 and 8, there were no others so far as is known. I, 1, was in army. I, 2, heard and saw well, *ob.* old. III, 1, sailor. III, 7, married her first cousin, III, 16, has retinal hæmorrhages, no retinitis pigmentosa; no defective vision in her descendants or those of III, 8 and 9 (IV, 9–20, V, 20–22), except IV, 14, age 29, hydrocephalic with double optic atrophy; no retinitis pigmentosa; never worked. IV, 20, abroad, married 15 years, no child. II, 6, and IV, 2, soldiers. III, 15, whereabouts unknown. No descendants of II, 5, known to have had defective vision (III, 10–15, IV, 21–42, V, 23–40), but V, 23, corneal opacities, and V, 27, congenital crescent in left fundus; right eye myopic. V, 28, male, mental, fundi normal. IV, 32, four males, two in army; a third one seen, fundi normal. IV, 36, one abroad, other fundi normal. IV, 31, in lunatic asylum. IV, 21, 24, 27, abroad, have good vision. V, 1–18, no defective vision. VI, 1–3, fundi normal. V, 19, age 4, imbecile, slavers, does not speak, has to be fed, nystagmus, gross choroido-retinitis in each eye.

FIG. 25.—Two cases of retinitis pigmentosa in half brothers with posi ive Wassermann reaction. IV, 11, male, age 24 (1901), seen. "Can't see when it becomes dark" noticed since early childhood; had convulsions in childhood; refraction, My. R. 4 D., V. with correction = 6/36. L. V. = counts fingers at 1 metre. L. has corneal nebula (central) and an anterior polar cataract; marked divergent strabismus; fields of vision much contracted, R. to 10°. Oph., R. O.D. pale yellowish colour with soft looking margins; retinal vessels much contracted; retinal pigmentation as delicate branched lines forming, at places, a network; the pigment is well away from posterior pole of eye; macular region of more uniform dark red than surrounding parts of fundus; vitreous opacities; L. appearance of fundus similar to that of

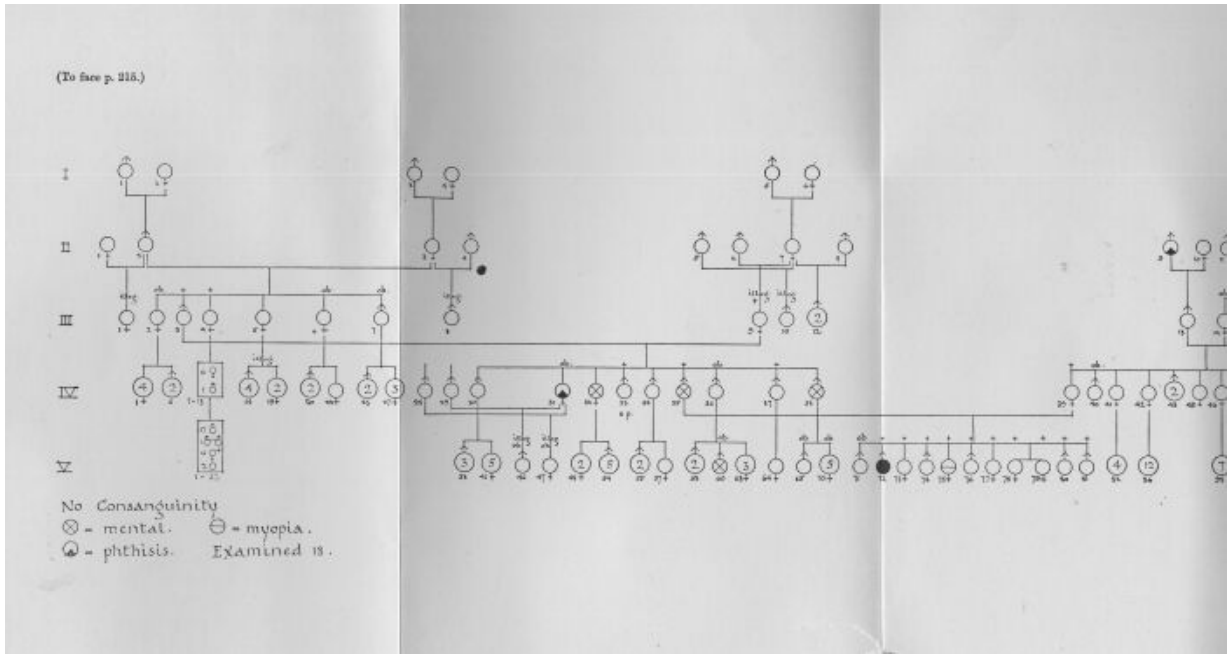
(To face p. 214.)

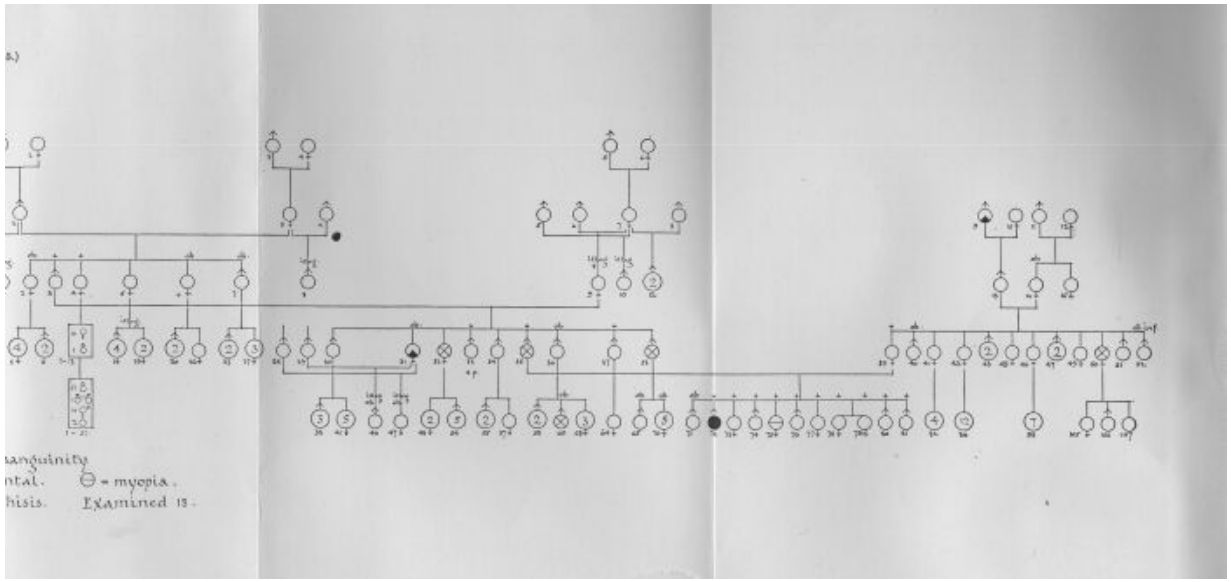
Fig. 25.



No Consanguinity.
⊖ = Myopia.
Examined 26.

(To face p. 215.)




anguinity
ntal.
hisis.
⊖ = myopia.
Examined 13.

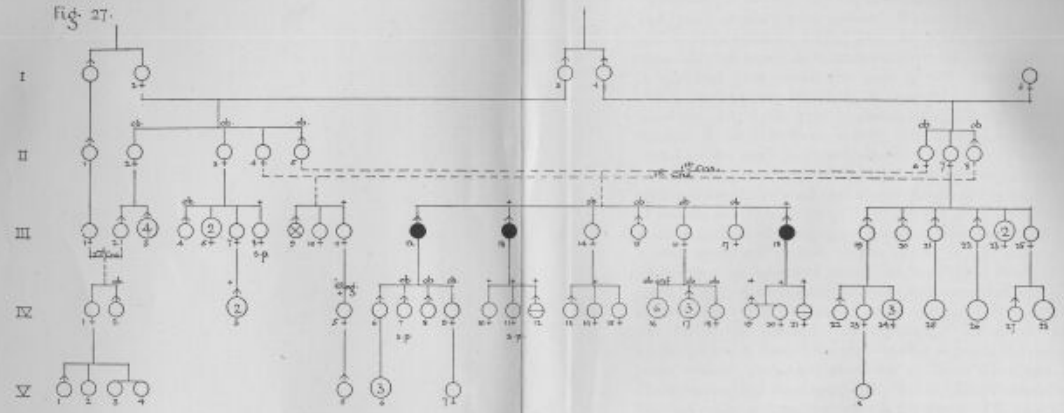
R., though there is more retinal pigment; some small yellowish dots. Is now (1912) working in a blind asylum, says vision has got no worse, night-blindness troublesome; oph., no white atrophic choroidal patches, moss-like retinal pigmentation not in large quantity, choroidal vessels exposed, O.D.'s pale waxy. Had bronchitis when 18 months old; has irritable temper; Wassermann test positive. IV, 7, male, unmarried, age 40 (1905), seen, "always short-sighted," gradually got worse. R. V. = p.l., iris tremulous, retinal pigmentation over whole of periphery of fundus, appearance of typical retinitis pigmentosa; posterior cortical cataract, choroidal vessels much exposed; O.D. has a good colour. L. V. = p.l., projection bad; lens opaque and dislocated into A.C., this had caused pain for one month, and on account of this he sought advice; no history of syphilis, is an abstainer from alcohol and tobacco. Now (1912) there is diffuse total grey opacity of right lens, preventing another examination of fundus. V. = no p.l. in either. L. lens was extracted in 1905, much corneal opacity is present. Is in good health and spirits, and works daily. Wassermann test positive. Mother (III, 4), age 75, always healthy, fundi normal. III, 1, a joiner, *ob.* 60, result of a chill, held things near, sight did not get worse, not night-blind. III, 8, *ob.*, had good vision. II, 3, lived long, *ob.* paralytic stroke. II, 4, *ob.* 44. I, 1, and I, 2, lived long, saw well. II, 1 and 2, and II, 5 and 6, all had good vision, and *ob.* old. IV, 6, and her children (V, 11–18) hear and see well. V, 20, *ob.*, when seen at age of 6, refraction was My. R. 10 D. in horizontal meridian, 12 D. in vertical, L. 8 D. in horizontal meridian, 11 D. in vertical; small myopic crescents. V, 19, 21–31, good vision. III, 3, soldier, *ob.* No defect of sight in III, 2, 5, 7, and their descendants, IV, 1–5, 15–26; V, 1–9, 32–47; VI, 1–3. IV, 16, three males, one married, has no children, another *ob.* infancy. III, 5, married twice, IV, 19, is by her 2nd husband. IV, 20, married twice. V, 41, is by his 2nd wife. No deafness and no insanity in this pedigree.

FIG. 26.—A single case of retinitis pigmentosa in a childship of 11. V, 72, male, age 14 (1911), seen, vision failing gradually for 4 or 5 years, 2nd born in childship, unable to see at dusk; had no illnesses; always very healthy; 4 years ago cut

right forefinger with an axe and lost about a teacupful of blood; wound healed readily; states that thereafter sight began to fail; fairly intelligent though somewhat slow mentally; memory good; hair very dark and stubbly, tends to encroach laterally on eyebrows; some dark hair on upper lip; forehead narrow; ears normal; no deformities in arms, legs or trunk; no bony thickenings; high palate; teeth good, but irregular; no enlarged glands in neck, axillæ or groins, pulse 75, regular; systolic pressure (Riva-Rocci) = 120 mm. Hg.; heart sounds pure; hearing, smell, and speech normal; urine normal; Wassermann test positive and when repeated on August 16th, 1912, was again positive; eyes: iris dark hazel; light sense markedly defective, reads 6/60 in light that gives normal eyes some of 6/9; refraction H. R. 0·25 D. in one meridian, 1 D. in opposite meridian, V. with correction = 6/18; L. 1 D. in one meridian, Emm. in opposite meridian, V. with correction = 6/18; oph. R. and L. media clear, retinal arteries much narrowed, choroidal vessels much exposed, no white atrophic spots, O.D. pale, its edge well defined, retinal pigmentation only at nasal part of right fundus and in left fundus at two places: (1) nasal part, (2) at outer periphery; in all these places pigment is very scanty and has moss-like arrangement and is not very dark, it requires careful looking for; in addition in left eye is a not very dark pepper-like mottling in some places, this is behind retinal vessels; fields of vision contracted especially upper halves. This boy was probably vaccinated from arm to arm for it was customary at that time. The doctor who vaccinated him is dead. The boy's mother says that the pustule healed without trouble and there was no illness following. V, 73–81, have normal fundi, good vision and no night-blindness has been noticed by parents; refraction is low degree of H. except in V, 75, with low degree of My. V, 81, age $2\frac{1}{2}$, refraction H.; fundus not seen, crying. V., 71, *ob.* 19, pneumonia, worked at a farm, vision good, no night-blindness. Mother (IV, 39) had no miscarriage or stillborn child, R. and L. fundus normal, vision normal. Father (IV, 35) was in lunatic asylum for depression at age of 18; fundi normal, good sight. III, 13, age 68, farm servant, sees well. III, 14, *ob.* 57 "water all over her," had good vision. II, 11 and 12, both *ob.* "cholera." II, 9, *ob.* phthisis. II, 10, *ob.* age 90. III, 3, leg amputated above knee for white

(To face p. 217.)

Fig. 27.



Consanguinity
⊗ = mental.
⊖ = myopia.
Examined B.

swelling, after marriage, age is 76, 2nd born in childship, fundi normal. III, 9, illegitimate child of II, 6, age 75, fundi normal, was brought up by her grandparents (I, 5 and 6), her mother (II, 7) had another illegitimate child (III, 10) by II, 5, and two sons (III, 12), unmarried, of her marriage with II, 8, all see well. II, 7, *ob.* 87, had a watery eye, but saw well. I, 5, *ob.* 58. I, 6, *ob.* 66, 3rd born of 10, both saw well. III, 1,* and III, 8, both illegitimate, are half-brothers of III, 3. II, 2, *ob.* 75. II, 3, *ob.* 92, both had good sight and hearing. I, 1, 2, and 3 were known to III, 3, but I, 4, *ob.* when he (III, 3) was young. I, 1, lived to nearly a hundred. IV, 40–51, and V, 82–107, all with good vision. IV, 40, *ob.* boyhood. IV, 50, in lunatic asylum, "went wrong" after marriage, has 3 children (V, 105–107). IV, 30–38 and V, 38–70, none with defective vision. IV, 38, in lunatic asylum, his children (V, 65–70) *ob.* young. IV, 32, weak minded, very quiet, never in asylum. IV, 33, leg amputated at knee for white swelling. V, 60, *ob.* 11, mentally weak, had large head and small body, not paralysed. IV, 31, *ob.* phthisis, age 23, had two illegitimate children, V, 46, and V, 47, both *ob.* young. III, 2, 4–7, and descendants, IV, 1–27, V, 1–37, have good vision. III, 5, had 6 children (IV, 14–19) all illegitimate, 3 *ob.*, a son and two daughters. Died infancy or still born: Males, IV, 6, 17, 52; females, IV, 1 and 2, 27.

FIG. 27.—Three males with retinitis pigmentosa in a childship of seven. III, 18, farmer, married, age 37 (1899), seen, no history of syphilis, scarlet fever in childhood, no other illness, no bleeding, no deafness; Wassermann test positive. R. and L. oph. after homatropine, pupils dilated, media clear; O.D. pale; retinal vessels much narrowed; retinal pigmentation mostly at periphery and largely in form of fine granules; choroidal vessels much exposed; a few circular grey spots 1/6 diameter of O.D. in lower part of fundus, choroidal vessels pass uninterruptedly across them; refraction both eyes emm., V. = 6/24 in each; fields of vision contracted to 10°. In 1912 V. reduced to perception of light in each; R. and L. posterior cortical lens opacities; moss-like retinal pigmentation except at macula; O.D.'s pale and waxy; retinal arteries narrow. He has preserved his hospital

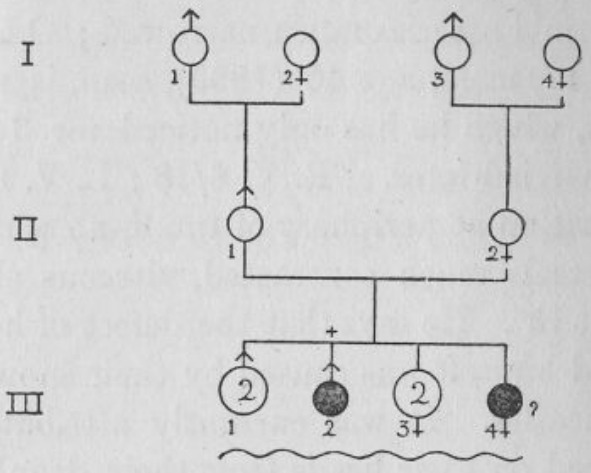
* Wrongly given as female in chart.

notes (not mine) of 1889, when he was 27; these state: "R. and L. V. = 6/9, fields in dim light not affected; no pigment seen in either eye. Two brothers have retinitis pigmentosa." III, 13, farmer, married, age 67 (1912), seen, is mentally keen, no deafness, not robust in appearance; never any illness, unless possibly scarlet fever, no bleeding. R. and L. posterior cortical cataract; O.D. waxy; retinal arteries and veins very narrow; moss-like retinal pigmentation except at macula; choroidal vessels much exposed; R. and L. V. = no p.l.; refraction H. low degree; for 30 years has been quite blind; he is very uncertain as to time of onset of visual defect, but places it between his 20th and 30th year. III, 12, male, abroad, believed to be affected in same way as his brothers, no deafness, is totally blind; failure of vision noticed first about age of 30, gradually progressed. Mother (II, 6), *ob.* 85, and father (II, 5), *ob.* 87, had good sight, first cousins. I, 3 and 4, brothers, had good vision; possibly others were in childship. I, 2 and 5, good vision. IV, 6, and his family (V, 6), abroad, reported to have no defect. IV, 7–9, and V, 7, had good vision. No defect of sight in II, 1–4, and descendants III, 1–11, IV, 1–5, V, 1–5. IV, 12, My. corrected in each with 0·75 sph. and 0·75 cyl. IV, 13–15, see well. IV, 14, congenital crescent of right optic disc. III, 14, *ob.* typhoid. III, 15, *ob.* "gastric fever," age 23. III, 16, *ob.* "hæmorrhage," her first six children (IV, 16), *ob.* infancy, and the remaining four (IV, 17 and 18), adults, *ob.* typhoid. III, 17, *ob.* 23, "brain fever." IV, 21, age 14, R. My. 0·5 D. in vertical meridian, H. 5 D. in horizontal meridian, L. H. 3·5 D. in oblique meridian, 5 D. in opposite meridian, fair hair; blue iris, no nystagmus, fundi normal; her mother has H. 3 D. in each eye. III, 9, child of first cousins (II, 4 and 8), mental, shy, would not be seen, unmarried, good vision and hearing. III, 3, abroad. IV, 2, *ob.* age 6. II, 7, and her family (III, 19–25), all see well. IV, 27, always been bed-ridden, hearing defective, no information regarding vision. IV, 28, others in childship. IV, 22–26, none known to have defective vision. Married and no children: Males, IV, 7; females, III, 6, 8, 24, IV, 11.

FIG. 28.—One, possibly two cases of retinitis pigmentosa in a childship of six. III, 2, male, basket maker, unmarried, age 52

(1897), seen, with gradual failure of vision since age of 24. In each eye extensive retinal pigmentation; choroidal vessels much exposed; posterior lens opacities and vitreous opacities in left eye; no note kept of appearance of O.D. or retinal vessels, but diagnosis is retinitis pigmentosa. R. V. = p.l.; L. V. = no p.l.; refraction My. in each. The eyes were examined at the request of Professor D. W. Finlay, who has kindly allowed me to use his notes, from which the following is taken. Patient is subject to indigestion; at age of 24 had headaches and vomited large quantities, and since then has been much troubled by these symptoms; is temperate; has had no venereal trouble.

Fig. 28



No Consanguinity.

Admitted to hospital for acute pain in epigastrium and vomiting, which had been present for a few days. Skin had marked icteric tint; conjunctivæ light greenish yellow; lateral nystagmus; no abdominal tumour felt; urine contained no albumen and no sugar. He was treated for dyspepsia and left the hospital much improved in about 3 weeks. He has since died, and no trace of his relatives could be found. He had formerly lived in the country. His sister (III, 4) has "same sight" as his own. Two other sisters (III, 3) and two brothers (III, 1) alive and healthy, vision normal. The ages of his brothers and sisters are from 42 to 62. Mother (II, 2), *ob.* 50, "fever," had good sight. Father (II, 1), not a cousin of his

wife, *ob.* 47, from "inflammation of a rupture," vision good. Grandparents (I, 1–4) had normal vision.

FIG. 29.—Retinitis pigmentosa in a sister and brother, the youngest members in a childship of 14. IV, 16, female, age 45 (1895), seen, a widow, rather deaf, never saw well in dim light, R. V. = 3/36; L. V. = 3/36; extensive retinal pigmentation with moss-like character extending to periphery of fundus, pallor of O. D., retinal vessels much diminished in size. She married twice and had no children. In 1910 she came to get glasses. R. V. distinguishes hand-movements at 8 feet; L. V. counts fingers at 6 feet. Her vision has very gradually got worse; refraction: R. and L. Emm. or low H. In each slight opacity of posterior part of lens; much moss-like retinal pigmentation over practically the whole of fundus, choroidal vessels conspicuous, retinal arteries much narrowed; O.D. pale, "dirty-looking." IV, 17, male, age 40 (1895), seen, is rather deaf, has night-blindness, which he has only noticed for 3 years, single at this date but married later. R. V. 6/18; L. V. 6/24; extensive retinal pigmentation at periphery of fundi, no white spots, O.D.'s pale, retinal vessels much contracted, vitreous clear, fields contracted to about 15°. He says that the defect of hearing and sight in his sister and himself was caused by their snow-balling during an attack of measles. It was currently attributed, however, to the slaps received on their heads from their drunken father, who latterly came down in the world. Mother (III, 30) had glasses for reading, had no night-blindness; hearing excellent, *ob.* 69, was a "clever" woman. Father (III, 2), a postman, had good sight and hearing, *ob.* 69, of a gradual illness, had no night-blindness. V, 27,* *ob.* 17 months, sight and hearing normal. V, 28–31, good sight and hearing, also their offspring (VI, 82–87). II, 18, not night-blind and not deaf. II, 4, vision and hearing normal. IV, 6, never saw II, 2, or II, 3, or heard of any visual defect in them. I, 5 and I, 4, are believed not to have been night-blind. I, 3, *ob.* unmarried. I, 1 and 2, believed to have had normal vision and hearing. IV, 6, age 80, healthy, keen mentally, knew a large part of the pedigree; fundus

* Information respecting descendants of IV, 17, was given in a letter from himself (IV, 17) on 25th January, 1910.

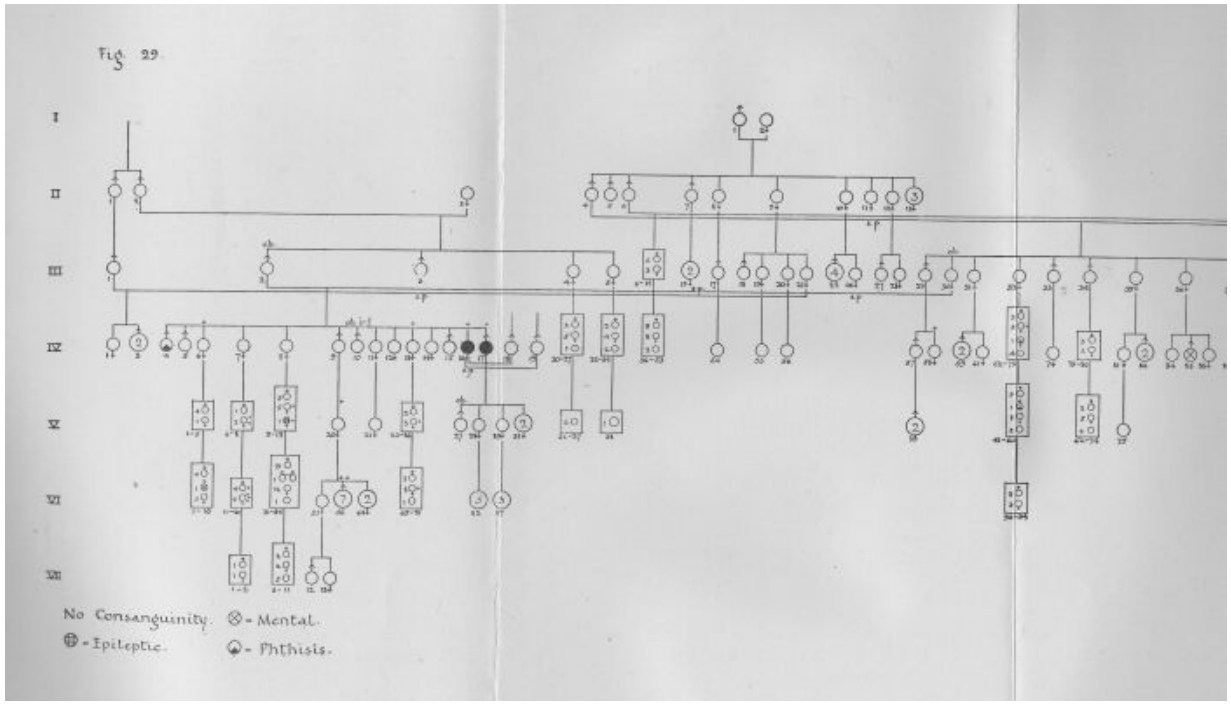
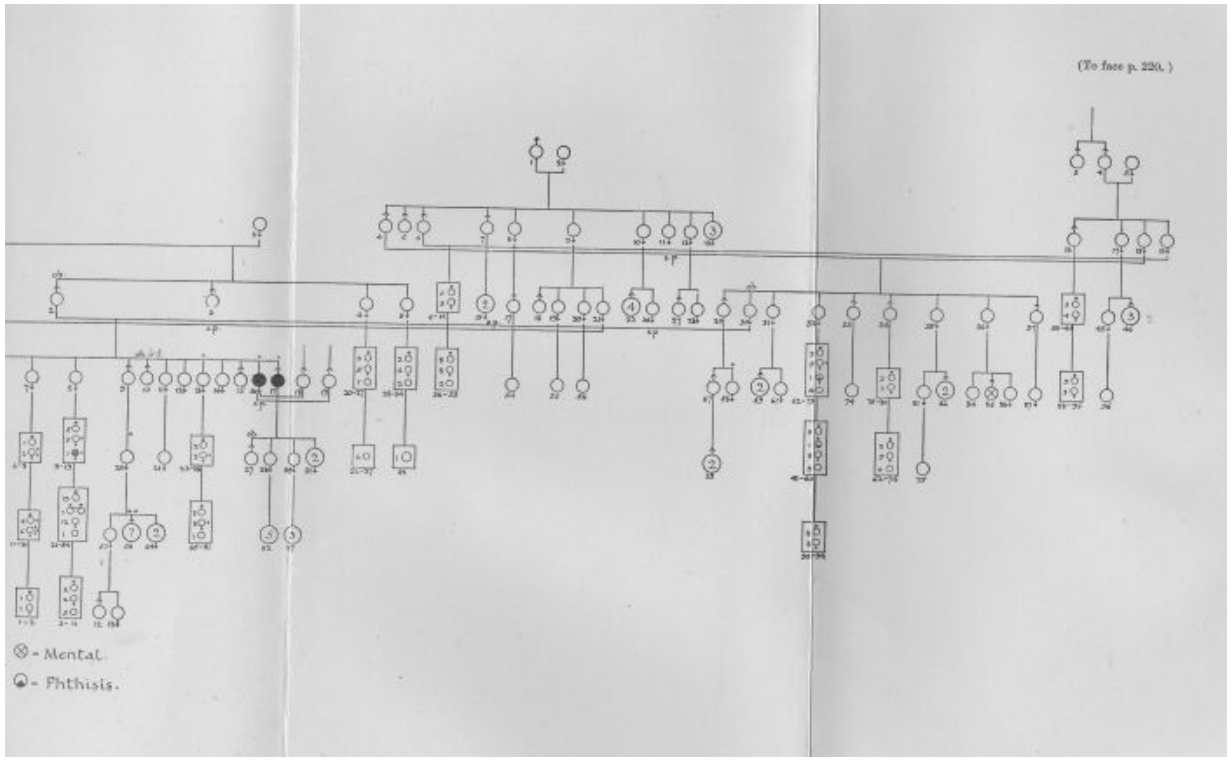
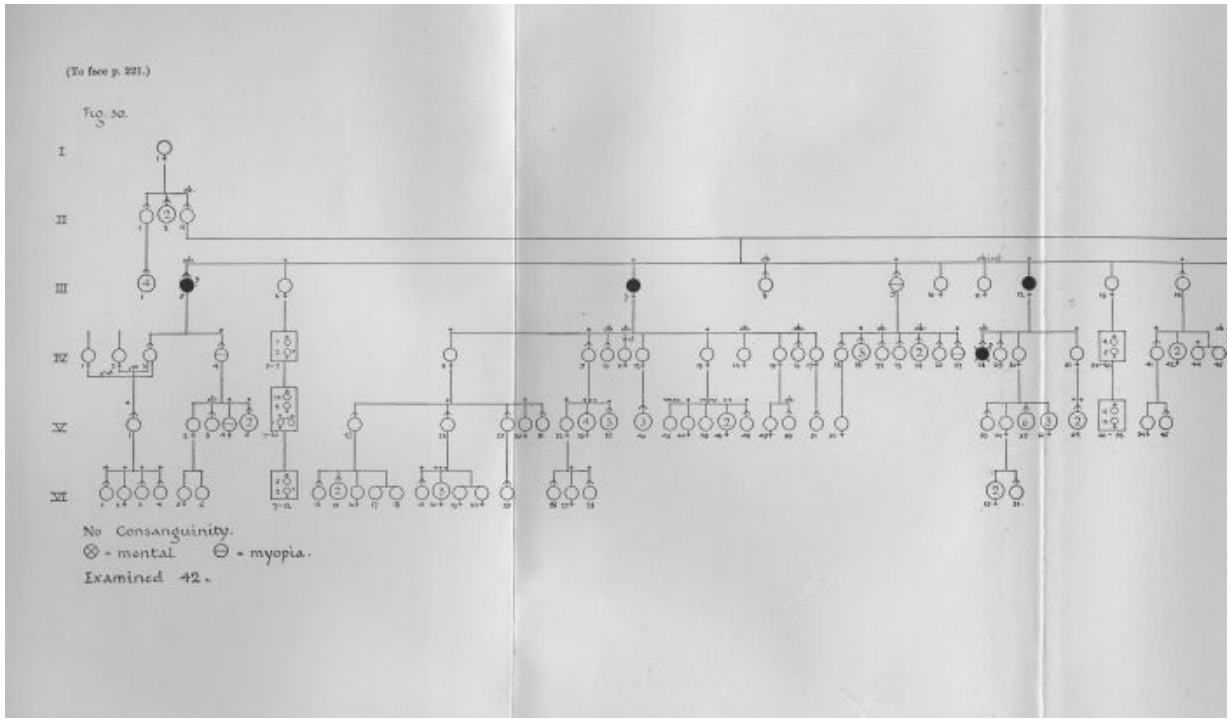
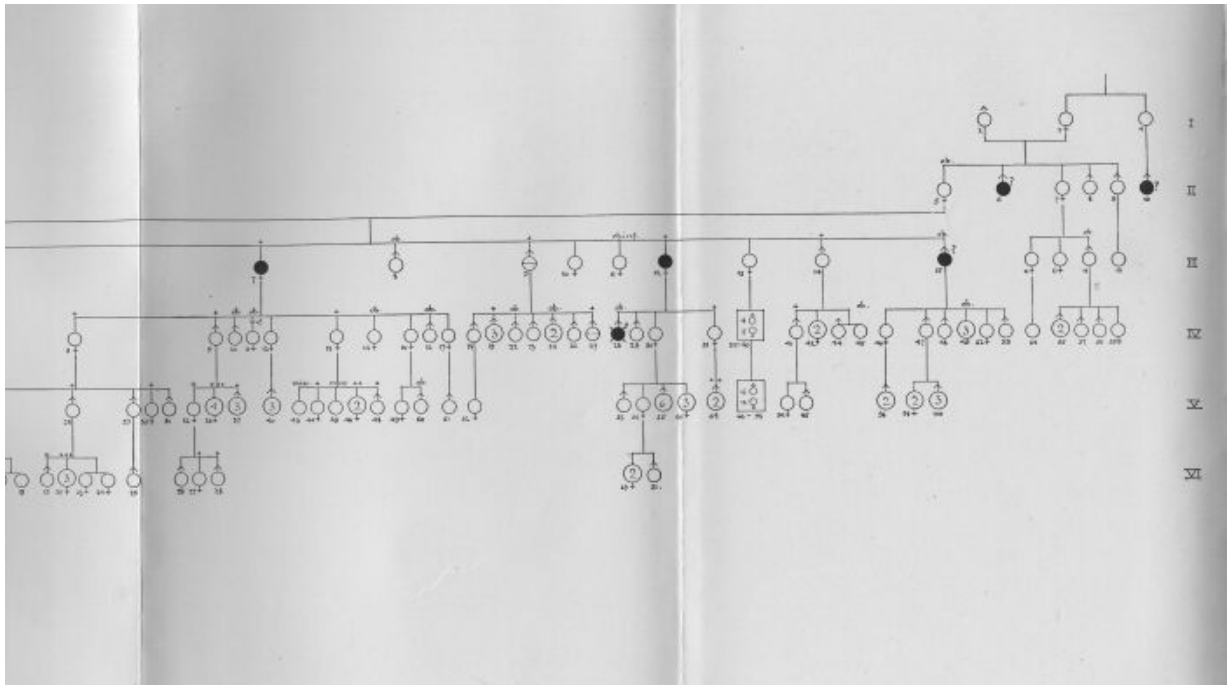


Fig. 29
No Consanguinity.
⊗ - Mental.
⊕ - Epileptic.
◒ - Phthisis.


(To face p. 220.)
⊗ = Mental.
◐ = Phthisis.


(To face p. 221.)
Fig. 50.
I
II
III
IV
V
VI
No Consanguinity.
⊗ = mental Θ = myopia.
Examined 42.

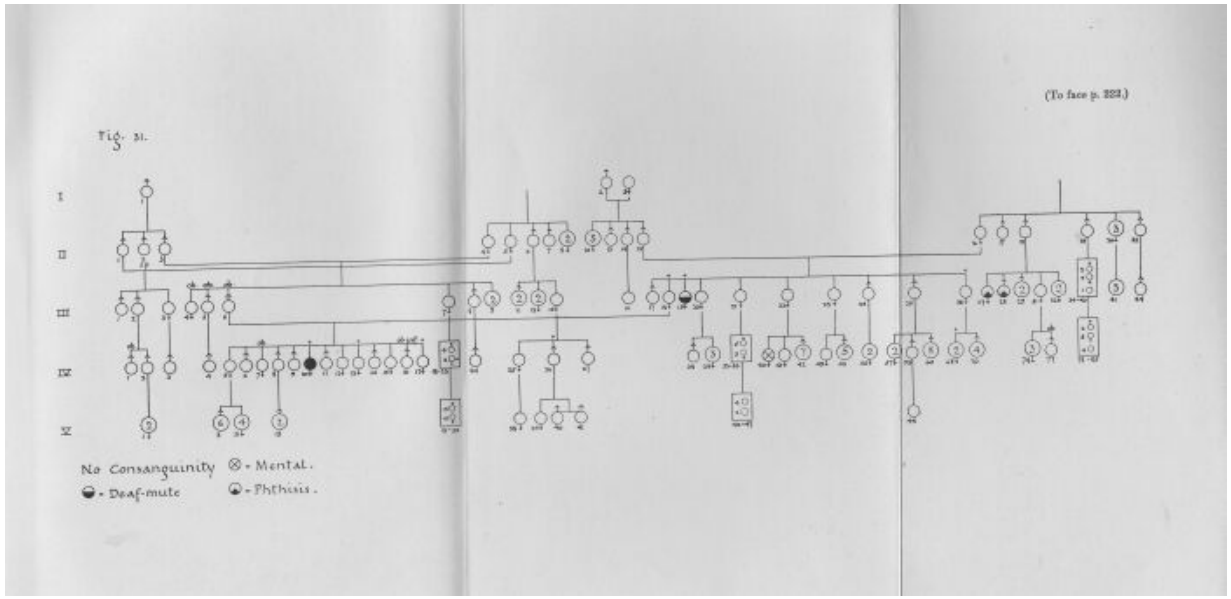


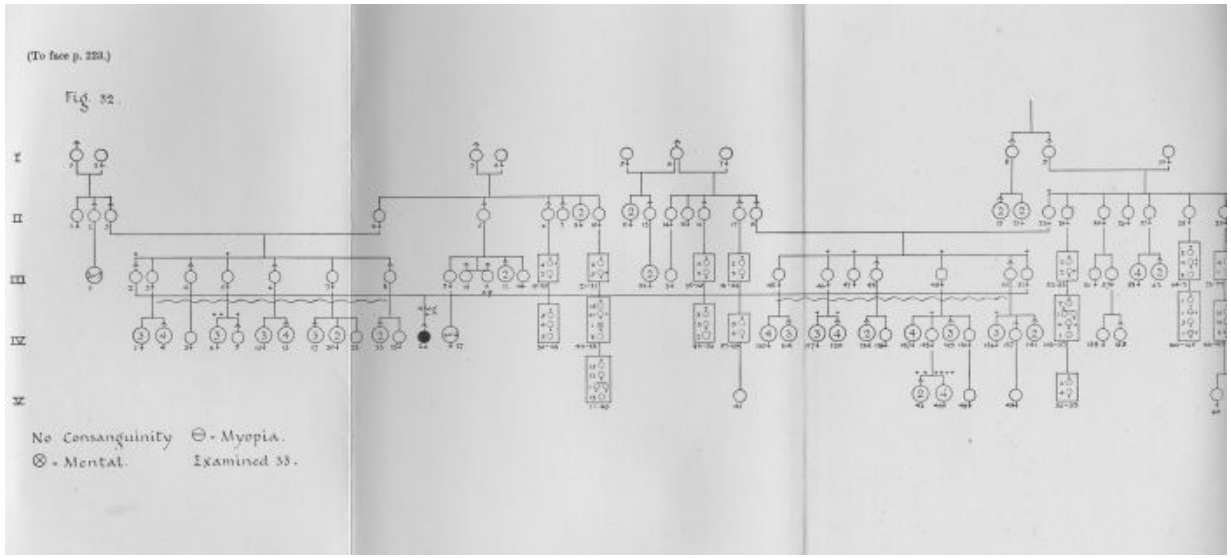
normal, has no night-blindness. A number of the descendants of IV, 6, 7, 8, and 9, were examined by Dr. Souter, in no case was the fundus abnormal. IV, 64, female, well acquainted with the family tree, has normal fundi, her daughter (V, 41), a teacher, is short-sighted, wears glasses for distance, has no night-blindness. IV, 27, by letter said he had never heard of any others besides IV, 16 and 17, with night-blindness. From information obtained there is no reason to suppose that anyone else shown in the figure has retinitis pigmentosa. Married and no child: Males, III, 3, IV, 28; females, II, 11, III, 15, 16, 26, 43, IV, 16, 65, 67, 95. This family history was worked out by Dr. Souter.

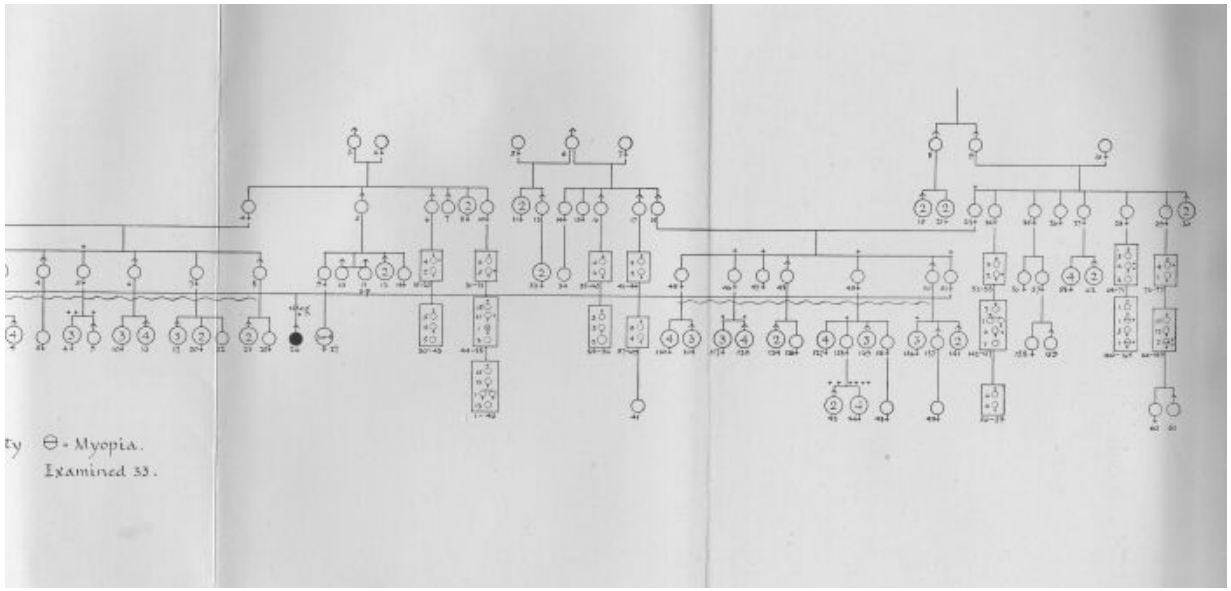
FIG. 30.—Two, possibly four, cases of retinitis pigmentosa in a childship of 11, three other supposed cases, one in the next generation and two in the previous generation. III, 7, female, age 66 (1898), seen, not deaf, now *ob.*, no difficulty in seeing until 25 years of age. R. and L., extensive retinal pigmentation at periphery, but extreme periphery is free from pigment; retinal vessels much narrowed; O.D. yellowish white colour; posterior lens opacities; refraction My. R. 9 D., V. with correction = 6/18, L. 4 D., V. with correction = 6/36; fields of vision contracted to about 10° in every direction. III, 12, female, in her dotage (1910), seen; not deaf; R. and L. fundus seen with difficulty, small pupils and probable lens changes, retinal pigmentation could be recognised at periphery; no atrophic patches or choroidal pigmentation; vision failed gradually, she is now nearly if not quite blind; had night-blindness. III, 15, male, *ob.* 58, was "always short-sighted; got glasses, but did not benefit by them"; night-blind. III, 5, male, *ob.*, said to have been affected in same way as III, 7, and III, 12. III, 6, bed-ridden, has senile cataract, neither fundus visible. III, 8, drowned, age 32. III, 9, age 74, myopic; R. fundus normal, L. has myopic crescent. III, 10, unmarried. III, 11 *ob.* infancy. III, 13, has good sight. III, 14, age 63, each fundus normal. Mother (II, 5), *ob.* 48, rheumatism, and father (II, 4), *ob.* 90, not cousins, both had good vision. I, 1, *ob.* 103, no particulars of her husband. Nothing known of parents of I, 2 and 3. None of the descendants of those with retinitis

pigmentosa in the childship III, 5–15, or of their unaffected brothers and sisters are affected with retinitis pigmentosa unless possibly a son (IV, 28) of III, 12. IV, 28, was silly, *ob.* in lunatic asylum, vision "just the same as his mother's." From reports given of IV, 4, and his son V, 6, it was expected that they had retinitis pigmentosa; examination proved otherwise for IV, 4, age 62, is myopic, no night-blindness; deaf; lens opacities; no retinal pigmentation; posterior staphyloma; always short-sighted; vision is not getting worse; his only difficulty is in seeing distant objects. V, 4, his daughter, is myopic, no night-blindness; fields full; no retinitis pigmentosa and a son V, 5, has normal fundi. V, 36, female, in bed with spinal disease. V, 40 and 51, young. IV, 18–27, 10 sons of III, 9, 3 *ob.* (IV, 22, 24), their father (III, 9) and IV, 27, were supposed to have the same disease as III, 7 and 12. IV, 27, R. and L. fundus normal, refraction low My., fields full; no night-blindness. V, 54, wears glasses, her children (VI, 29, 31) see well. IV, 42, *ob.* 7. VI, 23, 24, infant twins. II, 6, male, *ob.* 50, and II, 10, male, without issue, are both said to have been night-blind. II, 8, unmarried. No particulars of family III, 19. IV, 57, says in letter that he and IV, 55–59, have good vision and III, 18, *ob.* 11 years ago. His aunt (III, 16) and cousins (IV, 54) had good vision. III, 1, 4 males, 1 married, no children, they and II, 1–3, all had good sight. IV, 11, *ob.* infancy. IV, 14, *ob.* 5. IV, 10, *ob.* 4. IV, 16, *ob.* 15.

FIG. 31.—A single case of retinitis pigmentosa in a childship of 13, and a case of deaf-mutism in the previous generation. IV, 10, female, unmarried, age 28 (1911), seen, has defective vision especially in dim light; vision failing for 13 years; has frontal headaches, feels tired, was in a situation until 18 months ago; no deafness; abscess in right leg 2 years ago; diphtheria in 1909, in hospital 5 weeks, a few months later found difficulty in doing her work which she had not experienced before; frequently been bloodless since age of 18; no other illness or bleedings; extensive scar on bridge of nose, result of a burn at age of 6 months; R. V. counts fingers at 10 inches; L. V. sees hand movements at 2 metres; night-blindness, not very marked as tested in dark room; R. and L. retinal vessels very narrow;


(To face p. 223.)
Fig. 31.
I
II
III
IV
V
No Consanguinity
⊗ - Mental.
◒ - Deaf-mute
◔ - Phthisis.


(To face p. 223.)
Fig. 32.
No Consanguinity
⊗ = Mental.
⊖ = Myopia.
Examined 33.


ty ⊖ - Myopia.
Examined 33.

O.D. of a homogeneous dirty red colour; choroidal vessels much exposed; retinal pigmentation only at extreme periphery, it is moss-like and at some places there is pepper-like pigment; no white patches; media clear; fields of vision contracted to within 10°, left rather smaller than right. Mother (III, 18), second born in childship of 10, healthy, intelligent, both fundi normal. Father (III, 6), *ob.* 67, had weak heart. IV, 7, *ob.* "nervous breakdown, lung and kidney trouble." IV, 5, 6, 8, 9, 11–15, 17, see well, none deaf, healthy. No defective vision amongst II, 16–23, and descendants of II, 18–23 (III, 27–44; IV, 74–85). I, 2 and 3, saw well, also II, 10–15. II, 15, and II, 16, not cousins, though name is the same. None affected in III, 20–26, and descendants, IV, 28–70; V, 42–48. III, 19, female, unmarried, deaf-mute, fundi normal, refraction H. IV, 40, deformed, hunchback, legs swollen, she is mentally defective (description given by III, 18). III, 26, *ob.* recently, and daughter (IV, 68), fundi normal. IV, 73, male, *ob.* 6 months. III, 27 and 28, *ob.* phthisis. IV, 77, *ob.* young. No suspicion that I, 1, or his descendants, excluding IV, 10, were night-blind. II, 1 and 3, two brothers, married II, 5 and 4, two sisters. IV, 18, female, reported affected, has H. for which glasses are worn, no night-blindness, each fundus normal. She volunteered the statement that her own vision has been defective since profuse bleeding from nose at age of 14. No defect known in descendants of II, 6.

FIG. 32.—A single case of retinitis pigmentosa—an only child. IV, 26, male, age 24 (1911), seen, unmarried, a healthy farm labourer, no illness but measles and whooping cough, is deaf, not completely, night-blindness has been noticed for 15 years, was born illegitimate, his parents married subsequently. When first examined on September 7th, 1901, a diagnosis of retinitis pigmentosa sine pigmento was made; O.D. of a uniform redness, no retinal pigmentation seen even after mydriatic was used; night-blindness evident; media clear; no nystagmus; fundus normal excepting possibly a too uniform colour of O.D.; refraction R. My. 2 D. in oblique meridian, 2·5 D. in opposite meridian, V. with correction = 6/12; L. My. 2 D. in oblique meridian, 2·5 D. in opposite meridian, V. with correction = 6/12

partly; in light giving a normal eye 6/9 he could read 6/36 partly; both fields of vision contracted only to a small extent. When examined 10 years later, on September 7th, 1911, after mydriatic, both fundi presented the usual appearance of well-marked retinitis pigmentosa with moss-like retinal pigmentation; night-blindness troublesome, can work in day time, has sometimes got lost in the fields at dusk, "must not attempt to walk at night as he would trip over anything near his feet and looking down can see nothing"; he can read almost the smallest print in day time. Mother (III, 51), youngest of 7, robust, fundi normal; refraction low H. Father (III, 2), oldest of 7, good health, fundi normal; refraction low H. II, 23, old woman, sees well, but memory failing, her sister, II, 29, knows of (letter) neither deafness nor defective eyesight in any of her relations. I, 10, an only child, *ob.* 84, and husband (I, 9), not cousins. No information regarding vision of II, 19–21, descendants of I, 8. No night-blindness in II, 24–30, and their descendants. IV, 172, female, myopia 6 D. in each eye, her sister, IV, 169, has My. in L.; H. in R. IV, 160, male, My. R. 11 D. and 13 D. in opposite meridians, L. 9 D. IV, 161, female, anisometropic, R. My. 10 D., L. H. 0·5 D. I, 1, 2, 3, and 4, were known, and had good sight, descendants, excepting IV, 26, all said to see well. IV, 69, female, mentally affected, had to leave school, got worse, *ob.* 20. I, 6, married twice, by his first wife (I, 7) had 5 children (II, 14–18), by his second wife (I, 5), 3 children (II, 11–13), none deaf or see badly. III, 32–50; IV, 89–141; V, 42–49, no defect of sight or hearing. III, 46, R. fundus normal, L. near periphery of fundus on temporal side is an area three times the size of O.D., with well-defined margin, roughly circular, in which choroidal vessels are well seen, rest of fundus is dark and choroidal vessels hardly visible. Married and no issue: Males, III, 11, 12, 16, 22, IV, 70, 130; females, II, 8, III, 47, 52, 65.

FIG. 33.—A single case of retinitis pigmentosa in a childship of 12, the only case in a large pedigree, when a doubtful case in the succeeding generation, with changes limited to the macular areas, is excluded. IV, 76, female, age 39 (1908), seen, difficulty in seeing at a distance for 2 years, slight frontal headaches.

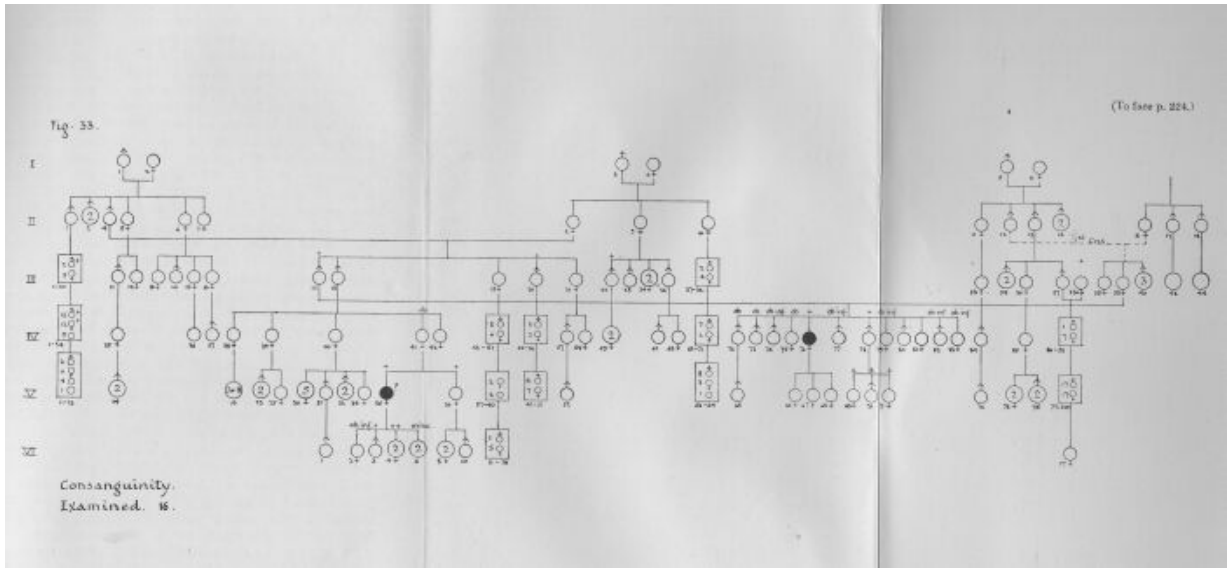


Fig. 33.
(To face p. 224.)
Consanguinity.
Examined. 16.

R. and L. retinal arteries narrowed, O.D. not much pallor; well-marked symmetrical retinal pigmentation at inner part of fundus between periphery and O.D.; media clear; fields of vision full; absolute symmetrical ring scotoma; refraction, R. H. 0·5 D. in oblique meridian, My. 1·5 D. in opposite meridian, V. with correction = 6/9; L., Emm. in oblique meridian, My. 2 D. in opposite meridian, V. with correction = 6/9; no complaint of night-blindness; liable to indigestion; typhus fever when aged 12; had inflammation of bladder and congestion of kidneys; urine contains no albumen or sugar. Has three unaffected daughters (V, 66–68), the eldest is 19. Mother (III, 39), good vision. Father (III, 17), fundi normal. IV, 72, *ob.* 37, "apoplexy." IV, 73, *ob.* 21, typhus. IV, 75, *ob.* 15, "abscess." IV, 80, at sea. IV, 81, a nurse. V, 35, female, first cousin once removed of IV, 76, age 31 (1908), seen, vision defective as long as she remembers; R. and L. O.D., slight pallor; retinal vessels little, if at all, narrowed; retinal pigmentation is limited to macular region; some pigment on superior and inferior temporal vessels, none is present further from fovea than inferior temporal vessels, and what pigment is above macula lies immediately beyond superior temporal vessels; it is in branched forms coarser than is usual in retinitis pigmentosa; choroidal vessels seen indistinctly in pigmented area; light sense markedly defective; fields of vision full; paracentral absolute symmetrical scotoma below fixation point; refraction, My. R. 1 D., V. = counts fingers at 3 feet, L. 1 D., V. = counts fingers at 3 feet; Wassermann test positive; no deafness; in 1908 in hospital with pelvic cellulitis, "no history of syphilis." Her first child (VI, 2) *ob.* 4 months; next three children are living (VI, 3–5), fundi normal. None of her children had snuffles or rash. VI, 6 and 7, two miscarriages. V, 36, fundi normal; her mother (IV, 41), *ob.* age 27, "decline"; her father, not in figure, *ob.* 35, kidney disease. II, 8, *ob.* 66, blind for 6 years before death. II, 16–18, saw well; no night-blindness in III, 43, 44, and their descendants. I, 1, *ob.* 85. I, 2, *ob.* 90; no visual defect in them or their children (II, 1–7) or in III, 1–16, IV, 1–31, 33–37, and V, 1–14. IV, 32, male, large patch of choroido-retinitis at periphery of nasal half of fundus, dense black pigment, some in front of retinal vessels, and nearly complete choroidal atrophy; O.D.'s normal; retinal arteries of

normal size; mixed astigmatism. I, 3 and 4, and II, 9 and 10, good vision. No visual defect known in III, 19, 21–32, IV, 43–71, V, 37–64, VI, 11–16. III, 20, has senile cataract, fundi not clearly seen, no pigmentation visible, nothing suggesting retinitis pigmentosa in his history. III, 18, had good vision, and IV, 38–42, V, 16–34, VI, 1, 8, 10. I, 5 and 6, II, 11–14, and descendants of II, 11 and 13, are not known to have defective sight. III, 38, *ob.* young. III, 40, two drowned, none with issue.

FIG. 34.—One and probably two cases of retinitis pigmentosa in a childship of four. V, 50, fisherman, unmarried, age 31 (1899), seen; is sullen and morose, never had good sight, which has failed gradually; does not see well in bright light; sight is best at sunset and sunrise; sight is bad for some considerable time when he goes from a light to a dark place and *vice versâ*; the sun dazzles him; no serious illness; influenza in 1899 followed by rheumatic pains in hips; R. and L. retinal arteries very narrow; scanty retinal pigmentation at periphery of fundus; O.D. of a uniform red colour; choroidal vessels readily seen; refraction H. R. 1 D., V = 6/36, L. 1 D., V. = 6/36; fields of vision contracted. Eleven years later (1909) R. and L. V. each = counting fingers at 7 feet; fields of vision reduced to 35° on nasal side, 70° temporal side, 35° below, 10° above (R.), 5° (L.); R. and L. marked posterior cortical cataract; retinal arteries extremely narrow; O.D. good colour; retinal pigmentation over whole of periphery of fundus; no white spots. V, 51, *ob.* 30, "consumption," unmarried, vision defective like her brothers; night-blind and fears were entertained that she would fall over the pier. V, 48 and 49, have normal fundi. Mother (IV, 53) *ob.* at age of 29, married her 1st cousin, had good vision. Father (IV, 8), 1st born in childship, married at 28; R. and L. fundus normal. III, 30, no visual defect. II, 13, had same name as II, 8–10, but is not known to be related. III, 26, saw well. III, 29, vision good, married her 1st cousin, III, 18, he had good sight and hearing and was 2nd born in childship. II, 8, married twice; by 1st wife (II, 6) had 6 children (III, 17–22), by 2nd wife (II, 7) had a family (III, 23) of which there are no particulars. None in Generation II (1–13)

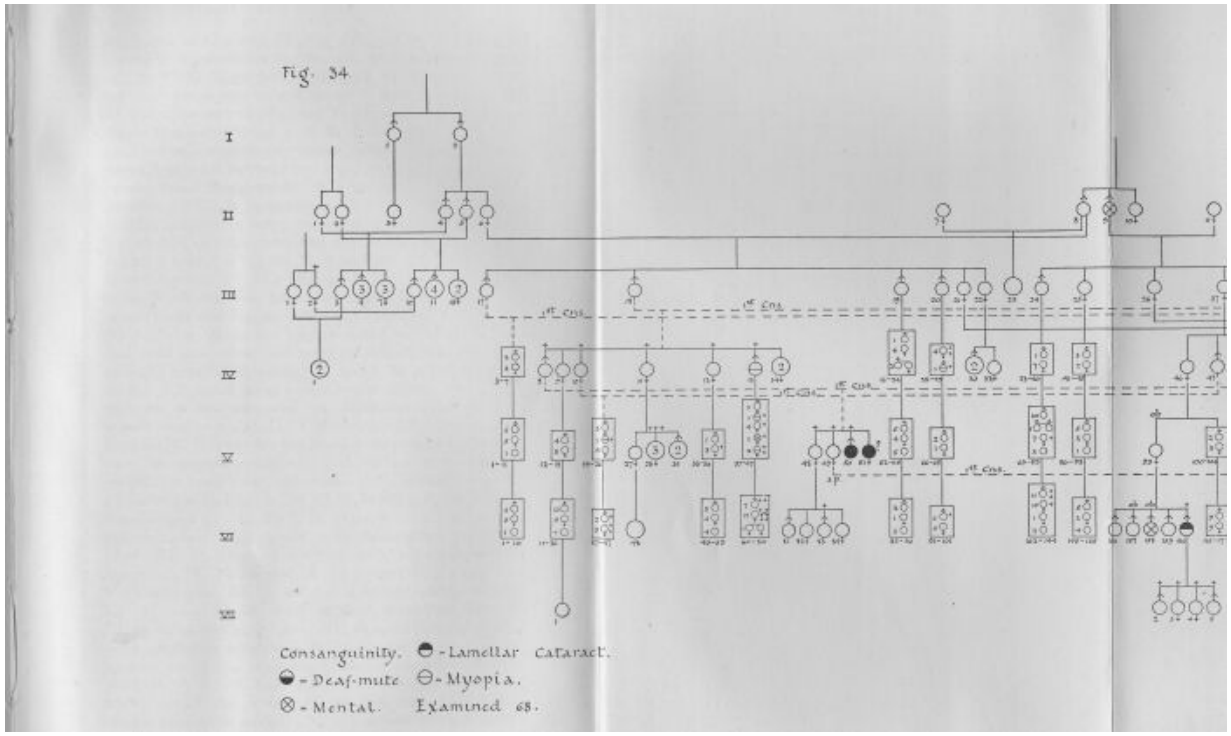
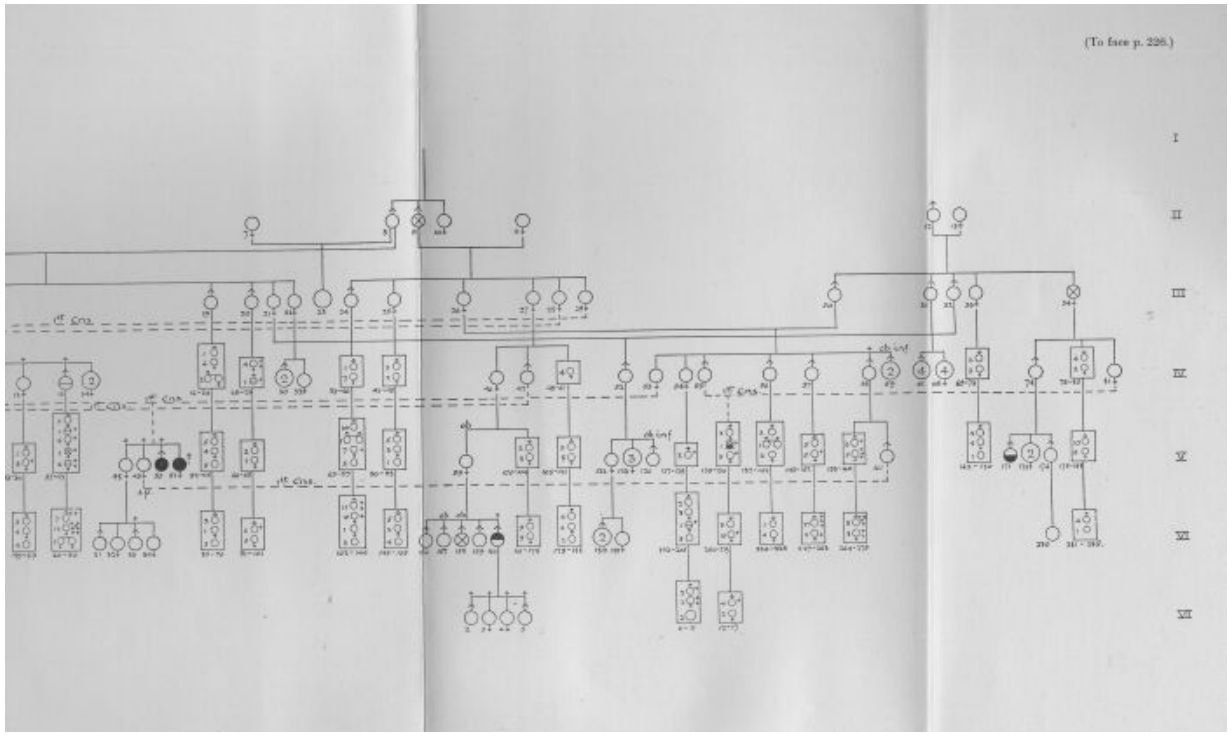


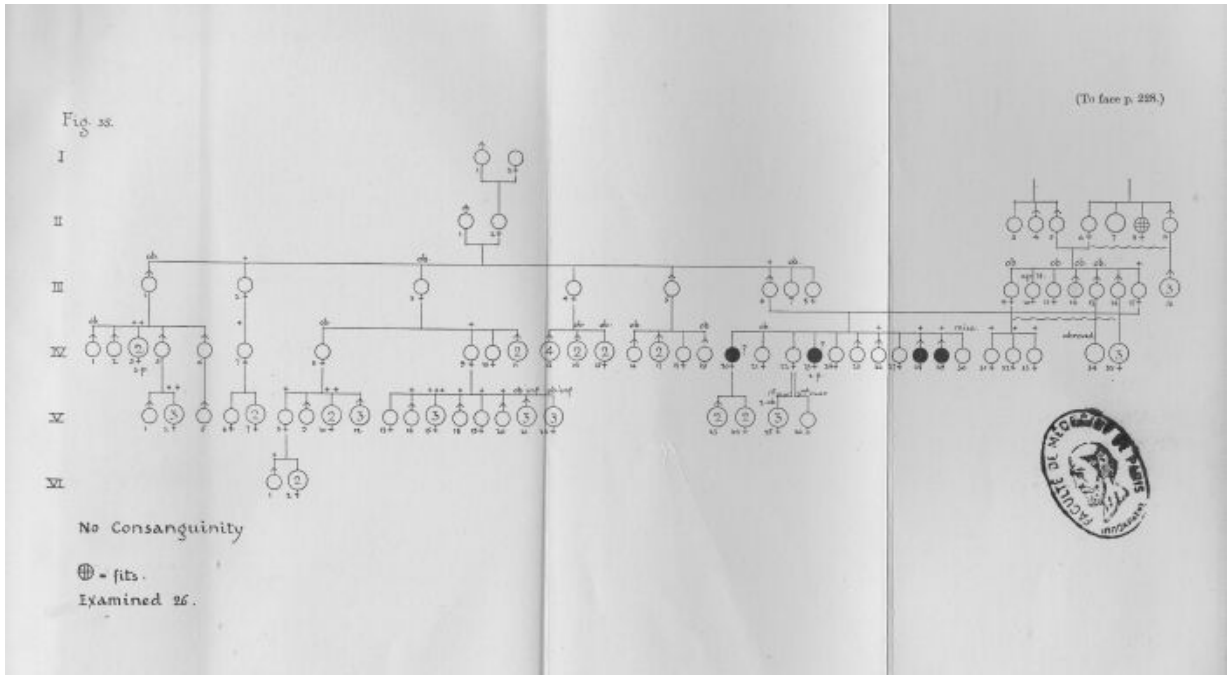
Fig. 34.
I
II
III
IV
V
VI
VII
1st Cous.
Consanguinity.
◒ - Deaf-mute
⊗ - Mental.
◓ - Lamellar Cataract.
⊖ - Myopia.
Examined 68.


(To face p. 226.)
I
II
III
IV
V
VI
VII

are known to have had defective sight. II, 9, was mentally affected for 8 years before he died. No information as to vision of I, 2. VI, 81–84, see well. Among III, 31–34 and IV, 54–58 and their descendants only 2 known to have vision affected, VI, 192, a female with My., and V, 161, male, with uniocular simple optic atrophy, age is 44, L. V. 6/36 Hm. 1 D., O.D. much too pale, edge sharply defined, lamina cribrosa exposed; retinal arteries not narrowed; no white lines along retinal blood vessels; rest of fundus normal. R. refraction H. 1 D. in vertical meridian, 3·5 D. in horizontal meridian; V. with correction = 6/24; no history of injury; no central scotoma; T.n.; fundus n.; left field of vision contracted, especially nasal side; vision was never good; he married V, 49, no issue. III, 34, mentally affected latterly. IV, 58, a cripple. V, 130, male, age judged to be 50, in poorhouse, 1st cousin of the retinitis pigmentosa cases (V, 50 and 51), his father (IV, 55) and their mother (IV, 53) being brother and sister, is a deaf mute and silly; can read small print when held near; no difficulty in dim light; refraction mixed astigmatism; R. and L. no pigmentation of retina; O.D. good colour; examination difficult even after mydriatic, as eyes were often kept in extreme lateral position; is quite clean; his mother's (IV, 81) brother (IV, 74) has a deaf-mute son (V, 171) who is a 2nd cousin of the retinitis pigmentosa cases, his paternal grandmother (III, 34) and their maternal grandfather being sister and brother. No defect known in descendants of II, 4 and 5. Of III, 17–22 and their descendants 6 are myopes (IV, 13, 27, V, 19, 40, 44, 47). III, 24 and 25 and descendant VI, 110, said to be short-sighted, not night-blind. IV, 13, fisherman, for years took large quantities of laudanum. IV, 10, married her 1st cousin IV, 47, their son V, 19, is myopic. III, 17, no defect of sight or hearing, married his 1st cousin III, 28, none of their descendants affected. III, 24–29, all had good vision. IV, 46 and 48–51 and descendants unaffected, except VI, 160, male, age 31, with slight stammer and typical symmetrical lamellar cataract, R. V. with +1·25 D. cyl. = 6/12, L. V. with +1 D. cyl. = 6/12, R. and L. fundus normal; his mother (V, 99) is a 2nd cousin of the retinitis pigmentosa cases (V, 50, 51) for her maternal grandmother (III, 27) and their paternal grandmother (III, 29) were

sisters. VI, 158, imbecile, *ob.* age 9. VI, 157, *ob.* age 2. To avoid confusion the following three consanguineous marriages have not been indicated in the figure:—(1) IV, 71, ♀, and IV, 77, ♂, 1st cousins, children of two sisters (III, 33, and III, 34), they had 1 child (V, 168), sex unknown, mother *ob.* at child's birth. IV, 77, married again and had 3 sons and 1 daughter (V, 183–186). (2) V, 150, ♂, married VI, 192, ♀, his 1st cousin once removed, his father (IV, 57) being his wife's paternal grandmother's (IV, 54) brother, they had 2 daughters and 2 sons (VII, 6–9). (3) V, 53, ♂, married his 2nd cousin (V, 21), their maternal grandfathers (III, 19, and III, 18) being brothers, they had 4 daughters and 1 son (VI, 41–45). V, 43, a widow, was in a lunatic asylum for some time after the death of her husband. A large number in this pedigree are fisher folk, especially those in the earlier generations.

FIG. 35.—Two, possibly four, cases of retinitis pigmentosa in a childship of eleven. IV, 28 (male) hairdresser, unmarried, age 21 (1911) seen, some deafness, short-sighted as long as he can remember, great difficulty in seeing at night; mentally slow; chorea when at school; operation on an ear 8 years ago, lost much blood; is strong. Refraction, R. My. 3 D. in one meridian, 4 D. in opposite oblique meridian, V. with correction = 6/12; L. My. 3 D. in one meridian, and 4 D. in opposite oblique meridian, V. with correction = 6/12; both fields of vision contracted to within 20°, except outer part of field, which extends at one part nearly to 30°; R. and L. typical retinitis pigmentosa; O.D. waxy; retinal vessels very narrow; extensive moss-like pigmentation of retina; choroidal vessels exposed. IV, 29, male, age 17 (1911), seen, has marked night-blindness; deafness: right ear quite deaf, left ear hears watch at 1 inch; never had any illness, but is not very strong; R.V. 6/9 some letters, no Hm., not improved by lenses. L.V. 6/9 some letters, no Hm., not improved by lenses. R. and L. typical retinitis pigmentosa; retinal arteries narrow; O.D. pale and waxy; choroidal vessels exposed; much moss-like retinal pigmentation over greater part of fundus; fields of vision resemble his brother's (IV, 28), are contracted to 20°, except at outer part, where they approach 30°. IV, 20, according to her mother, was affected like her


(To face p. 228.)
Fig. 33.
I
II
III
IV
V
VI
No Consanguinity
⊕ = fits.
Examined 26.

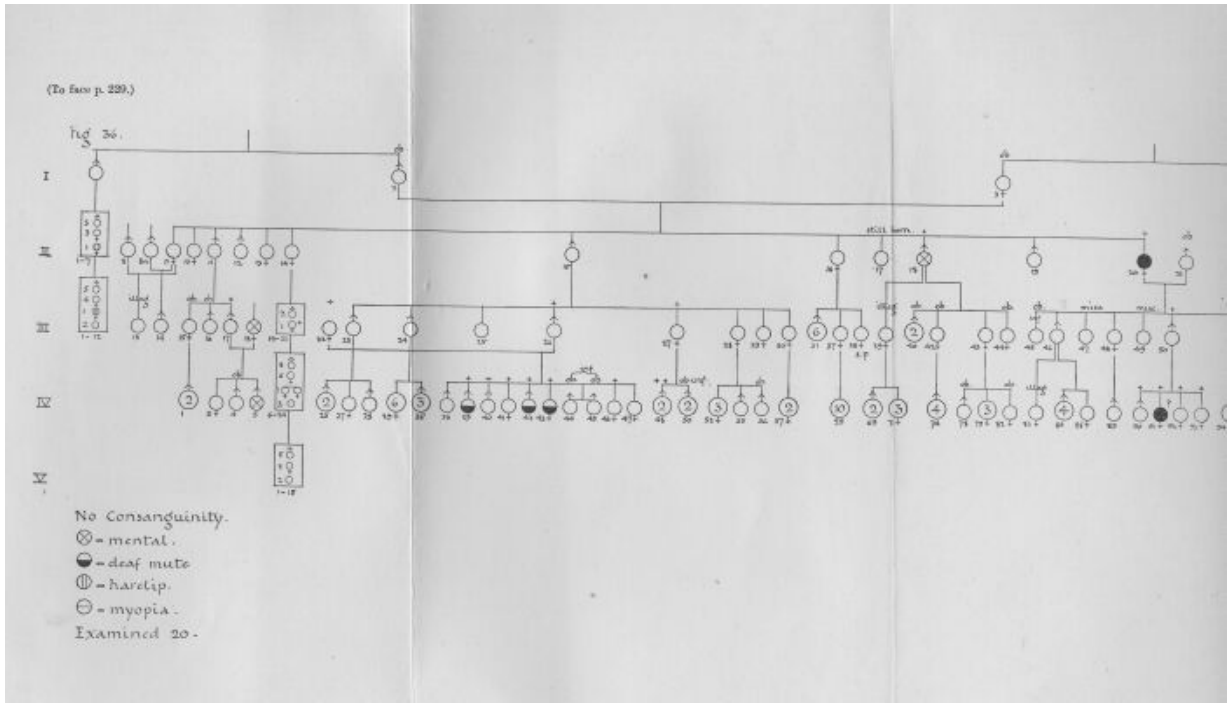

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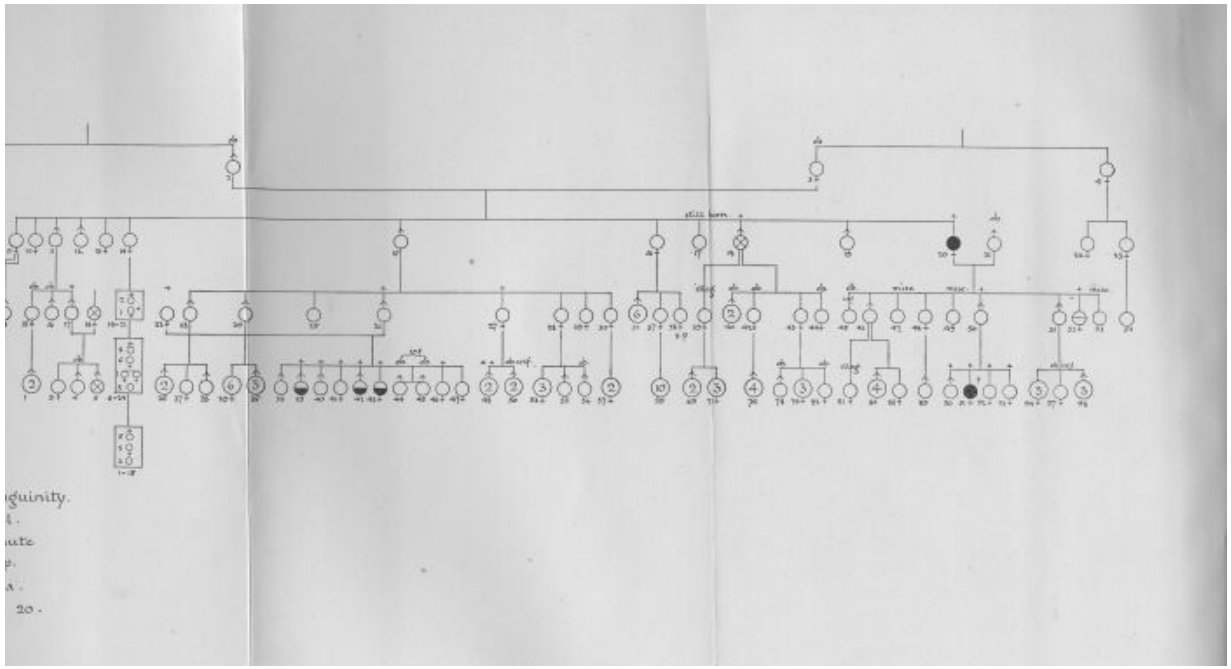
Fig 36.
I
II
III
IV
V
No Consanguinity.
⊗ = mental.
◒ = deaf mute
◍ = harelip.
⊖ = myopia.
Examined 20.



brothers. She was obstinate and left home, and has not been seen for many years. IV, 23, female, married, no children, reported to be deaf and night-blind; not on speaking terms with mother for 16 years. IV, 21, *ob.* croup, age 2. IV, 22, had chorea and is very nervous; married twice. Two of her 3 children in 1st family (V, 25) are dead; has 1 child (V, 26) by 2nd husband. IV, 24, unmarried. IV, 25, age 27, unmarried. IV, 26, fundi normal. IV, 27, had rickets, is not strong. Mother (III, 15), youngest in childship, married at 19, fundi normal, not deaf, mentally peculiar, gloomy. Father (III, 6), retired sea captain, age 63, married at 25, fundi normal. II, 5 and 6, II, 1 and 2, had good vision. I, 1 and 2, saw well. All in childship II, 6–9, had good vision. II, 7 = other members, all saw well. II, 8, paralysed at 15 and took fits. II, 6, was youngest in childship. II, 3–5, noted for excellent vision. III, 7, *ob.* young, all in childship III, 1–8, had good sight and none deaf. III, 4 and 8, abroad. IV, 17, one in Navy, other abroad. IV, 31–33, hypermetropic, no night-blindness, fundi normal. IV, 33, has strabismus, wears glasses. IV, 31, wears glasses. IV, 34, no information, all abroad. IV, 35, 2nd one, age 24. III, 9, *ob.* 70, not night-blind, was "not so smart" as others. III, 10, age 78, unmarried, can thread smallest needle, is not night-blind. III, 11, *ob.* unmarried, blind at 20 from small-pox, vision good previously. III, 12, *ob.* unmarried, had good sight. None of Gen. IV, 1–19, known to have defect of sight or hearing, or their descendants. Married and no issue: females IV, 3, 4, 23.

Fig. 36.—A single case of retinitis pigmentosa in a childship of 12 and a doubtful case in a grandchild. II, 20, female, age 54 (1896), seen, youngest in childship, never had good sight, which has been getting worse for some years, bothered by bright light; in early life received poor nourishment and was taken to an orphanage at age of 8; measles 3½ years ago; R. and L. extensive peripheral retinal pigmentation; retinal vessels narrow; O.D. pale; opacities at posterior part of lens; vitreous opacity; R. V. = 6/60, L. V. = 6/60; fields of vision contracted to 12° and 10° at some parts. In 1912, at age of 70, R. and L. V. = no p.l.; complete cataract; she is healthy, alert, and has

good memory; refused blood examination. Husband (II, 21), a strong, healthy man, *ob.* suddenly age 65, was supposed to have been suffocated. Mother (I, 3) and father (I, 2) believed to have died comparatively young. No visual defect known in I, 1, and his descendants. II, 5, and her niece (III, 7) had hare-lip. I, 4, *ob.* 86, no defect of sight known in his descendants. II, 18, is in lunatic asylum, delusions, fundi normal. II, 16, abroad, only other member of childship II, 9–20, alive. II, 10, 12, 13, and 19, *ob.* young. III, 52, age 18 (1896), R. and L., myopia 8 D., myopic crescents, otherwise fundus normal; with her glasses, has no difficulty to-day (1912). III, 50, and his children (IV, 90, 92, 93) have normal fundi. IV, 91, a bright girl, age 12, no complaint of vision; R. and L., a few particles of pigment at periphery of retina at one part, questionable whether retinal arteries are narrowed and O.D. too pale or whether both are within normal limits. Seen again a few months later (December 26th, 1912) under better conditions: R. V. 6/24, with +1 D. cyl. = 6/9, not every letter; L. V. 6/18, with +1 D. cyl. = 6/12, not every letter; in dim lights which gave normal eyes 6/60 and again 6/18 she could read exactly the same as they; fields of vision definitely contracted except down and in; no ring scotoma in either. R. and L. O.D. just appreciably too pale; retinal arteries narrowed to a small extent, more in left eye than right; choroidal vessels exposed towards periphery, where fundus has a mottled appearance, and there is here and there a small deposit of pigment in retina, which is very sparse; no spots or patches in choroid with defined margins, but in places choroidal vessels become more evident; upper central incisors normal, no evidence of syphilis in parents or any of their children. IV, 90, age 15, bedridden. III, 45, lived one day. III, 46, and IV, 83–88, no visual defect. III, 48, delicate, nervous, had faintings, she and her child (IV, 89) have good vision, so has III, 51, and his children, IV, 94 and 98. II, 2, male, *ob.* after 70, blind for 20 years before he died. All in childship II, 1–7, are dead. III, 18, was in lunatic asylum. III, 17, believes that his son (IV, 5) is mentally affected, for he is morose and has sometimes assaulted him. II, 15, *ob.* (1910) cerebral hæmorrhage, his son (III, 26) has three deaf-mute children (IV, 39, 42, 43) with normal fundi. The twins (IV, 44–45) were born prematurely and

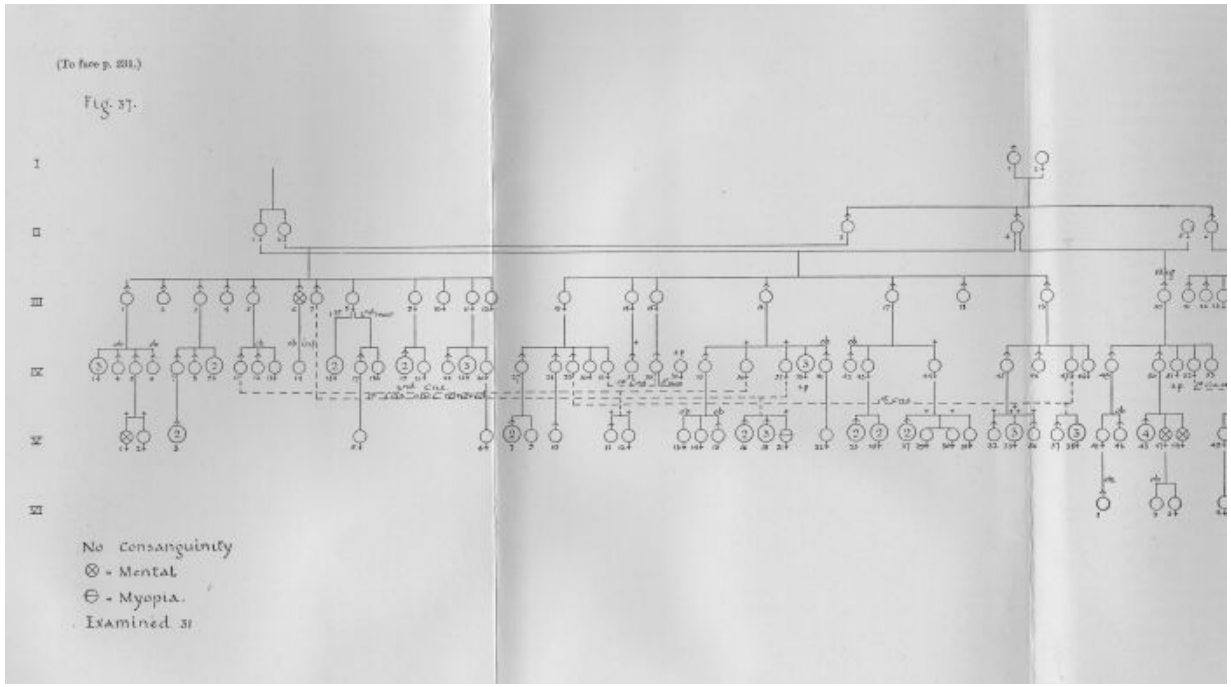

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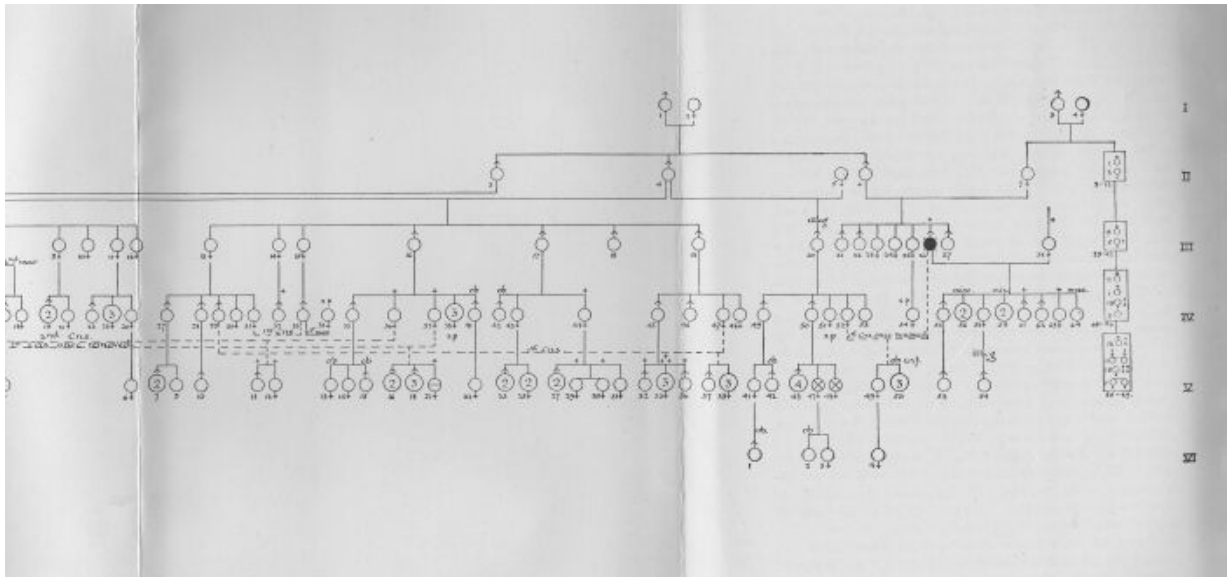
Fig. 37.
I
II
III
IV
V
VI
No consanguinity
⊗ = Mental
⊖ = Myopia.
Examined 31



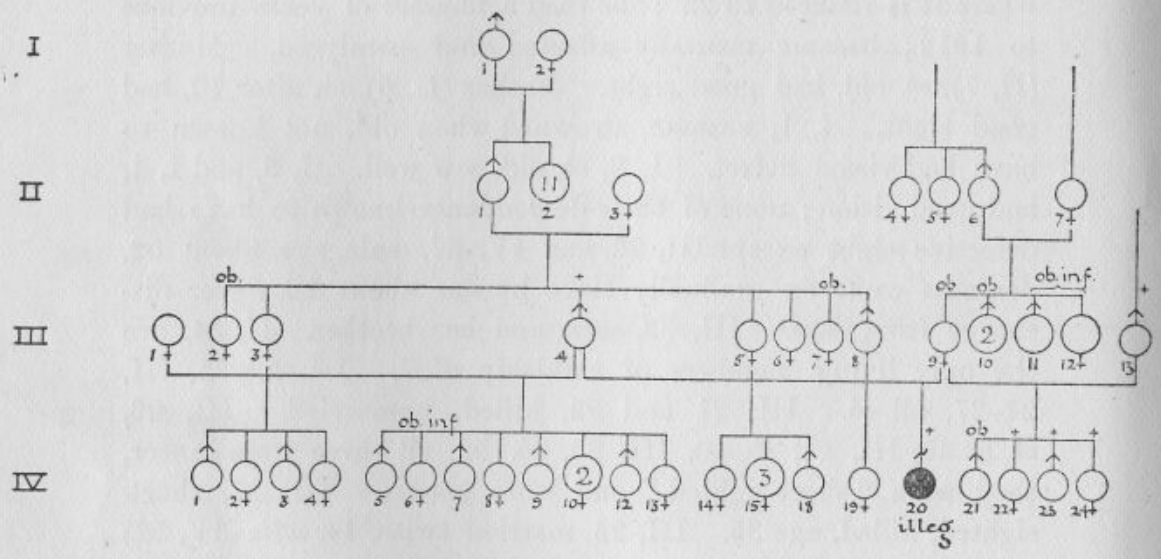
died within three weeks of birth. Wife of II, 15, had four deaf-mute cousins, children of her father's brother. III, 25 and 29, *ob.* diphtheria at ages of 2 and 21. IV, 59, five of them married. IV, 4, a soldier. IV, 12 and 13, in Navy. No defect of vision known in any descendant of II, 9, 11, 14–16, 18.

FIG. 37.—A single case of retinitis pigmentosa in a childship of seven. III, 26, sailor, age 48 (1897), seen; gradual failure of vision for 12 years; could never see so well as most people; in 1884, one evening he walked off a plank into the sea; denies venereal disease; R. and L. retinal pigmentation at periphery with moss-like arrangement, pigment is scanty; choroidal vessels exposed; no white atrophic choroidal patches; O.D. dirty white colour; lamina cribrosa visible; retinal vessels contracted to mere threads; vitreous opacities; R. V. = p.l., L. V. = 3/36, left field of vision contracted peripherally, most at upper part, where it is reduced to 22°; he died a number of years previous to 1912; became mentally affected and paralysed. Mother (II, 7), *ob.* old, had good sight. Father (II, 6), *ob.* after 70, had good sight. I, 1, seaman, drowned when old, not known to have had visual defect. I, 2, *ob.* old, saw well. I, 3, and I, 4, had good vision; none of their descendants known to have had defective sight except III, 26, and IV, 67, male, age about 52, deafness came on gradually since he was about 20, never discharge from ears. III, 33, seen, and her brother, III, 34, are the only living members of childship of 7. III, 29–35, III, 21–27, all *ob.* III, 21 and 22, killed, unmarried. III, 23, *ob.* at 45, III, 24, *ob.* 40, III, 25, *ob.* 52, all three from cancer, they were "short-sighted," and wore glasses. III, 27, short-sighted, killed, age 35. III, 26, married twice, 1st wife (IV, 52) was his 1st cousin once removed, they had a daughter (V, 49), and 3 other children (V, 50) that *ob.* infancy; his 2nd wife (III, 28), still alive, had 10 pregnancies, 5 miscarriages (IV, 56, 59, 64) and 5 that lived to adult life; IV, 55, 58, and 62 see well, none deaf. V, 53, infant. The two brothers II, 3, and II, 4, married two sisters (II, 1, and II, 2); in their descendants visual defect known in two, namely, high myopia with large posterior staphylomata in V, 21, and much defect of vision from corneal opacities in IV, 32. V, 1, has some mental defect;

fundi normal. The mental defect in V, 47 and 48, is believed to be derived from their mother, as she has a sister in a lunatic asylum. III, 6, only survivor of childship III, 1–12, is or was in a lunatic asylum. Married and no issue: Males, III, 40, IV, 33 (he is quite recently married to IV, 31); females, IV, 38, 39, 40, 51, 54, 70, 79, 82, 84, 92, 93. The living members of this family reside in a small township on the Banffshire coast, where the family has been for many generations engaged principally in shipping and fishing. Their medical man says that 30 years ago, or so, syphilis was rife in the town.

FIG. 38.—An illegitimate male with retinitis pigmentosa. IV, 20, farm worker, age 18 (1902) seen, lazy, mentally feeble,

Fig. 38.



No Consanguinity.

always short-sighted, night-blindness; R. and L., much typical retinal pigmentation at periphery of fundus; retinal arteries distinctly contracted; O.D. good colour, right one rather paler than left; choroidal vessels much exposed; fields of vision peripherally contracted; no white spots or patches in fundus; no vitreous or lens opacities; nystagmus on lateral movement of eyes; refraction My. R. 3 D., V. with correction = 6/60, L. 3 D.,

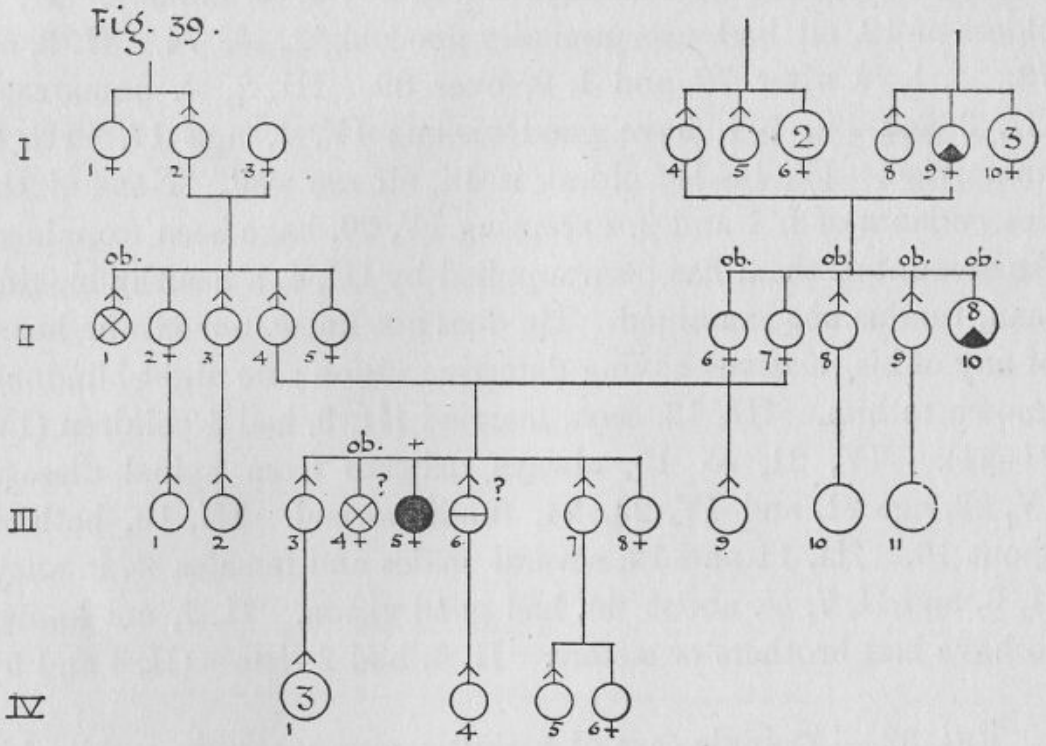
V. with correction = 6/60, each eye reads 1 J. at 6 inches; with light reduced so as to allow a normal eye 6/12 he does not read 6/60. In 1911 his doctor reported that the vision was not markedly worse and that he could do, at least, ordinary work. Mother (III, 9), *ob.* 44 at "change of life." III, 4, acknowledges that he is possibly the father of IV, 20, and III, 13, says that he (III, 4) certainly is. III, 4, has good vision and has subsequently married III, 1, and had 9 children all with good sight; age of oldest (IV, 5), however, is only 13 and IV, 6, *ob.* infancy. II, 1, oldest of 12, all had exceptionally good sight, *ob.* 74. II, 3, *ob.* 72. I, 1, *ob.* after 70, and I, 2, over 80. III, 2, *ob.* unmarried. III, 3 and IV, 1–4, have good vision; IV, 1, age 17. III, 6, unmarried. IV, 14–19, oldest is 14, all see well. None of the descendants of I, 1 and 2, excepting IV, 20, have been examined. An account of them has been supplied by III, 4, a healthy-looking man, fundus not examined. He does not know, nor has he heard of any of his relatives having defective vision; no night-blindness known to him. III, 13, seen, married III, 9, had 4 children (IV, 21–24). IV, 21, *ob.* 15, always delicate from spinal disease. IV, 22, age 21, and IV, 23, 24, fundi normal. III, 10, both *ob.* about 16. III, 11 and 12, several males and females *ob.* infancy. II, 6, and II, 7, *ob.* about 65, had good vision. II, 7, not known to have had brothers or sisters. II, 6, had 2 sisters (II, 4 and 5).

FIG. 39.—A single case of retinitis pigmentosa in a childship of six. III, 5, female, unmarried, age 27 (1907), seen by Dr. John R. Levack and Dr. Wm. C. Souter, at Aberdeen Royal Infirmary. Her sight has always been defective, worse last 12 months; can hardly see at night, and stumbles against things in the dark; never saw so well as her sisters in dim light; night-blind as long as she can remember; an aunt says it was not noticed until she was 14; anæmia 2 years ago; typhoid fever age 6, was very ill; teeth good; hearing excellent; R. and L., well-marked retinitis pigmentosa; O.D. pale; retinal vessels small; posterior cortical cataract; refraction My. R. 1·5 D., V. with correction = 6/36, L. 2·5 D., V. with correction = 6/18; fields of vision contracted to 10°. Mother (II, 7), only survivor of 12, 9 died of phthisis;* when II, 7, was pregnant with III, 5,

* The 9th death from phthisis was either II, 6 or 8 or 9.

her, at that time, youngest child (III, 4) was sickly, became totally blind, and died age 2, "the doctor stating that had she lived it would have been as an imbecile"; the distress of the mother is supposed to have caused the defective vision in III, 5. Father (II, 4), vision splendid. I, 9, *ob.* phthisis, age 47. She and I, 4, had good sight. I, 2, and I, 3, healthy people with good vision. III, 6, "short-sighted," probably myopic, for his glasses enable III, 5, who is myopic, to see at a distance; not

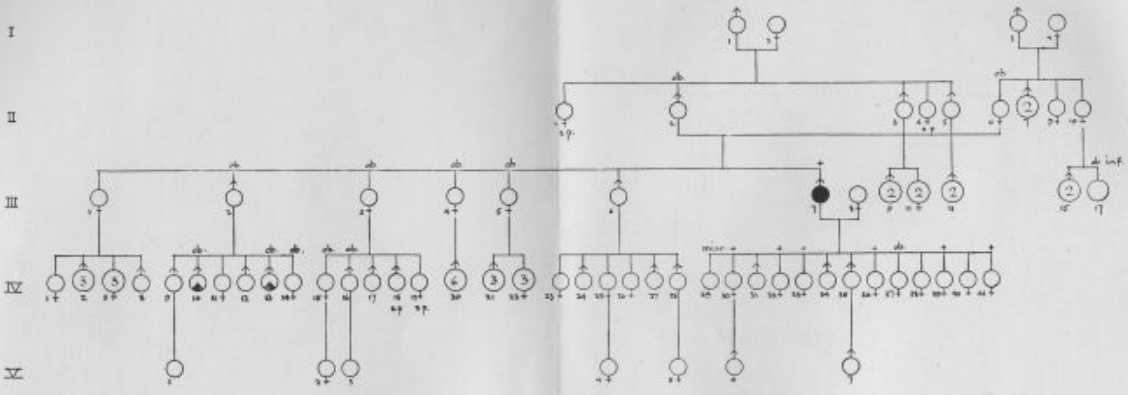
Fig. 39.



night-blind. III, 3, 7, and 8, no night-blindness, deafness, or mental defect, see well. IV, 1–6, healthy and see well. III, 10 and 11, two large families of males and females, none have defective vision. II, 6, *ob.* 70, was very deaf. III, 9, and 1 and 2, good vision. II, 1, *ob.* in lunatic asylum, age past 40. II, 2, 3, 5, and I, 8 and 10, vision good. Most of the account of this family is contained in a letter from the patient's father (II, 4) and mother (II, 7). The wife of II, 3, was interrogated.

(To face p. 236.)

Fig. 40.



No Consanguinity.

◒ = phthisis.

FIG. 40.—An isolated case of retinitis pigmentosa in a male in a childship of seven. III, 7, gamekeeper, age 43 (1894), seen; vision failing rapidly in both eyes for 3 months, sees best in dim light. R.V. 6/18, not improved; L.V. 6/24, not improved; fields of vision full, with absolute ring scotoma complete in L. and R., though very narrow at upper nasal part of right field; optic discs have waxy appearance, retinal vessels diminished in size, especially in left eye; edge of right optic disc more sharply defined than that of left; moss arrangement of pigment in retina between optic disc and extreme periphery, there is no pigment immediately around optic disc or at extreme periphery of retina; patient says he cannot shoot ground game, but birds he can; has always shot off left shoulder, using left eye, though not left-handed for anything else. When seen again after 17 years (1912), he was working in a mill as timekeeper (bell-ringer) having had to give up his position as gamekeeper; he says vision is not so strong as 17 years ago, yet with glasses he reads 1 J. with each eye readily; no history of syphilis; had "gastric fever" at age of 14; was badly shot 38 years ago (1876), had 78 pellets in him; up to age of 20 was subject to nose bleeding, as much as a cupful he thinks was sometimes lost; does not bruise readily; has no more difficulty in dim light than other people; fields of vision taken with 10 mm. square of white paper on end of a stick have an absolute ring scotoma below and at nasal part, but not above or in temporal parts where field is much contracted to perhaps 10° or 15°; retinal arteries are narrow, optic discs pale, choroidal vessels exposed, much retinal pigment extending nearly to optic disc, no white spots or patches of choroidal atrophy. No note of media; light sense not tested. I have seen the fundus of only 6 others, namely, his 6 children, IV, 30, 32, 33, 36, 39, and 41, and these are quite normal. The following has been supplied in 1912 by the patient (III, 7), who is intelligent, his wife (III, 8), and his children. Mother (II, 6) and father (II, 2) had good vision and both died between 80 and 90. I, 1, and I, 2, both noted for their good vision, *ob.* old. I, 3, and I, 4, had no defect of vision, *ob.* old. All the brothers and sisters (III, 1–6) of patient (III, 7) saw well, only III, 1, a widow, and III, 6, are alive. IV, 29, one miscarriage was followed by 12 living children (IV, 30–41).

IV, 30, age 37, has a young child, V, 6, fundus examination was impossible. IV, 34, is gamekeeper and IV, 35, a porter with 1 child (V, 7). IV, 37, *ob.* age 7. IV, 39, age is 21. IV, 41, age is 13. All in this childship have excellent vision. His wife (III, 8) looks particularly robust. II, 1, and II, 4, married, but had no children. II, 3, had 4 children (III, 9–12), and II, 5, had 2 sons (III, 13 and 14), but nothing known of their offspring if any. II, 7, 8, and 9, unmarried. II, 10, had 2 sons (III, 15 and 16) and some children (III, 17) that *ob.* infancy. IV, 1–8, most are married. IV, 9, abroad, no information as to his family (V, 1). IV, 10 and 13, *ob.* phthisis. IV, 11, unmarried. IV, 12, ? unmarried. IV, 14, *ob.* in girlhood. IV, 15 and 16, *ob.* V, 2, infant. V, 3, sex not known. IV, 17, unmarried. IV, 18 and 19, married, neither have progeny. IV, 20, family of 6 males, not married when known. IV, 21 and 22, vision good. IV, 23, 26 and 27, unmarried. IV, 24, ? unmarried. V, 4, age 3. V, 5, age 8. None of his (III, 7) relatives are known to have defective vision.