THE BRITISH JOURNAL

OF

OPHTHALMOLOGY

MARCH, 1938

COMMUNICATIONS

THE SIGNIFICANCE OF HEREDITY IN OPHTHALMOLOGY. PRELIMINARY SURVEY OF HEREDITARY EYE DISEASES IN TASMANIA*

BY

J. BRUCE HAMILTON

HOBART, TASMANIA

(Concluded)

Optic Atrophy—Hereditary (Leber's)

Although previous mention has been made of this disease in the literature, it was first described in detail with pedigrees, by Leber in 1871, since when considerable research has been done in many countries. Consequently Julia Bell was able in 1931 to publish 237 pedigrees, two of which were recorded by Hogg from Tasmania, and are included—with his permission—with my pedigrees. To Hogg's two pedigrees I am able to add one complete (No. 31) and one very incomplete one (No. 32) of my own. The cause of the incompleteness of No. 32 was the fact that propositus was endeavouring to receive compensation, and until taxed, repudiated all suggestions that his condition was hereditary, which is a further illustration of the difficulties of a pedigree recorder.

I propose to discuss the four Tasmanian pedigrees together under the following headings, but have nothing new to add. So

* Continued from p. 108.

much has been written about Leber's disease, and so little new elucidated that I have found this section of my work the most difficult to write.

Sex Incidence

At once it became apparent that males greatly predominate in all four pedigrees, and the percentage of males affected is given in the following table:—

Pedigree No.	Males	Females	Percentage of Males
10	19	0	100 per cent.
39	3	0	100 per cent.
42	25	2	92.55 per cent.
43	3	1	75 per cent.
Totals	50	3	94.3 per cent.

This is certainly a higher rate than Julia Bell found in European and Japanese pedigrees, which were respectively 84.8 per cent. and 59.1 per cent. of males.

Age of Onset

Hogg, when describing his two pedigrees (Nos. 33 and 34), gives no indication of the age of onset in cases examined by him; and as in my 2 pedigrees only 4 of the cases were examined by me this question with regard to Tasmania cannot be answered definitely. In passing I might mention that in pedigree No. 31 the ages of onset were:—In case III/43 at 27 years, in case IV/7 at 40 years, in case IV/45 at 25 years, in case IV/47 at 20 years, while in pedigree No. 32, in case II/13, the onset was at 19 years.

Signs and Symptoms

In three of my cases the onset was sudden, while in the fourth the mode of onset was doubtful. In all four cases both eyes were affected simultaneously, and in all eight cases there were central scotomata and no peripheral constriction. In two of the eight eyes the central scotomata were only partial, and I was able to correct the sight in one eye to 6/24 and in the other eye to 6/12 vision. This shows that the prognosis was not altogether hopeless in two cases out of four.

Causation

In none of the four cases was there any suspicion of pituitary dysfunction, nor were the visual fields obtained, in the slightest degree, suggestive of this condition; and Dr. Hogg reports that

■ - AFFECTED MALES
● - AFFECTED FEMALES
◊ - NO HISTORY
✕ - DEAD
X - EXAMINED
✎ - PROPOSITUS

No. 31. LEBER'S DISEASE.

his X-ray findings of the sella turcica were negative in all cases. Gowers in 1904 declared that the disease was an abiotrophy (Swab, 1934). I feel certain he came nearer the truth than any previous worker, and research during the subsequent 32 years has not been able categorically to contradict his assertion. If Gowers is right—and there seems little doubt that he is—then the correct line of treatment is prophylactic, that is, the prevention of breeding by all carrier females.

Mode of Transmission

In the four Tasmanian pedigrees there is no variation in transmission from those quoted from England and Scotland. In every case the transmission was through an unaffected female who frequently has affected brothers; and in no case did an affected male or female appear to transmit the disease, as was found in the pedigree of congenital optic atrophy reported by Thompson (1935). The disease in Tasmania is therefore of the sex-linked recessive type of Lossen. (Macklin, 1927).

There is no history of consanguinity in any of the four Tasmanian pedigrees, and Julia Bell (1931) records consanguinity in only 18 of her 237 pedigrees; so that this factor does not appear to be a very important one. Nevertheless Russell (1931) reported a pedigree in which a consanguineous marriage appears to have precipitated the disease in three brothers, in an otherwise clean stock, with no affected members in previous generations.

Treatment

Kuhn (1931) reported surgical exposure of the chiasma of a male with symptoms of four weeks' duration, no abnormality was detected, and no visual improvement resulted. A further proof that to date the only available treatment is prophylactic.

Comments on Pedigrees

Pedigree 31.—I consider this pedigree accurate, and fairly complete as it was first given to me by III/43, and later was verified through IV/7. As many of the family are living in New Zealand, and some in Fiji, as well as many on the Continent of Australia, it has been impossible to examine the majority of those affected. V/31, a male who lives in Fiji, and is in his early twenties, is the most recent member to become affected.

III/23. Normal.

III/43. Optic Atrophy. R. and L. R.V.C. correction = 6/12.

IV/7. Optic Atrophy. R. and L. R. and L.V. = <6/60.

IV/45. Optic Atrophy. R. and L. R.V. = 6/24.

Pedigree No. 32.—As explained this pedigree is grossly incomplete, but I hope at a later date to glean more details of further affected members who undoubtedly exist. The one member examined, II/13, had <6/60 R. and L.E.

Pedigrees Nos. 33 and 34.—These are published by kind permission of Dr. G. H. Hogg of Launceston, Tasmania; but I am unable to give any details at all. Dr. Hogg in private communication regrets that he has no details available (other than those given when he published the pedigree in the Medical Journal of Australia, on March 24th, 1928), except that the members came from the northern end of this island, and are unlikely to be included in my complete pedigree (No. 31).

REFERENCES

- BEDELL, A. J. (1934).—*Amer. Jl. Ophthal.*, Vol. XVII, No. 3, p. 195.
 BELL, J. (1931).—*Treasury of Human Inheritance*. Vol. II, Part IV. Cambridge.
 CORDES, F. C. (1933).—*Trans. Amer. Ophthal. Soc.*, Vol. XXXI, p. 289.
 HOGG, G. H. (1928).—*Med. Jl. Austral.*, March 24, p. 372.
 HOGG, G. H. (1936).—Personal Communication.
 KUHN, A. S. (1931).—*Arch. of Ophthal.*, March. (Abstract.)
 MACKLIN, M. T. (1927).—*Canad. Med. Assoc. Jl.*, Vol. XVII, p. 1339
 RUSSELL, W. R. (1931).—*Trans. Ophthal. Soc. U.K.*, Vol. LI, p. 187.
 SWAB, C. M. (1934).—*Nebraska Med. Jl.*, Vol. XIX, p. 184.
 THOMPSON, A. H. (1935).—*Proc. Roy. Soc. Med.*, Vol. XXVIII, p. 1415.

Pterygium

Tasmania, which is famous throughout the Antipodes for its delightful climate, is practically free from progressive pterygium, trachoma, hyperkeratosis and rodent ulcer of the lids. During the last 5½ years out of 4,880 private case records I find the following statistics:—

Pterygia	...	...	...	44 cases=0.9 per cent.
Trachoma (old)	...	...	...	4 cases=0.08 per cent.
Trachoma (active)	...	...	...	4 cases=0.08 per cent.
Rodent ulcer of lids	...	...	...	5 cases=0.1 per cent.

Of these 44 cases of pterygium—10 cases have had to be dealt with surgically, so that it is evident that pterygium *per se* is a disease of little consequence in Tasmania, although on the Continent of Australia it is a constant source of concern.

Of these 44 cases I can find an hereditary influence in only 2 cases—2 brothers whose maternal parent suffered from the same condition.

Sex Incidence

Reference to hereditary pterygium in the literature is very scanty. Macklin (1927) reports two pedigrees from the literature

—one of 3 affected males and 5 affected females in 4 generations; and another of 10 affected males and 2 affected females in 6 generations.

My pedigree consists of two males and one female, and this predominance of males on the whole is possibly influenced by greater outdoor activities of males.

Mode of Transmission

Macklin considers the transmission as dominant, being transmitted by both parents to offsprings of either sex.

Comments on Pedigrees

Pedigree No. 35.—I/2 is dead, but definitely had a pterygium which her sons think was on her right eye.

II/1 has a right active pterygium as also had II/2. I marsupialised both after the McReynold's method, with satisfactory result.

REFERENCES

- GATES, R. R. (1929).—*Heredity in Man*. London.
 MACKLIN, M. T. (1927).—*Canad. Med. Assoc. J.*, Vol. XVII, p. 698.

Hereditary Ptosis

In a limited pedigree such as the one reported by me, the author should be extremely careful that the described condition is not due to birth injury. In this pedigree I certainly cannot be dogmatic, but as the uncle of the patient examined by me has definitely a bilateral ptosis, it is strongly suggestive that the condition is hereditary in nature. On the other hand, first the fact that birth injuries to the eye are prevalent in Tasmania (and I hope to report a series shortly), secondly that Briggs (1919), Usher (1925) and Macklin (1927) all assert that ptosis is dominant in transmission (which is not so in this pedigree), and thirdly, that in my pedigree only males are affected (see sex incidence), make me somewhat doubt the authenticity of the family record. Nevertheless the propositus volunteered the facts at the first examination of his son, and I have been able to confirm his assertions in one instance at least.

Sex Incidence

According to Macklin (1927), the sex incidence is about equal taking all reported pedigrees collectively, but in my pedigrees only males have been affected to date.

Mode of Transmission

In at least five of the authors referred to, the mode of transmission has been dominant, but my pedigree does not confirm this.

Associated Defects

Epicanthus.—Usher (1925) and Clausen (1923) surveyed the literature on hereditary epicanthus with ptosis—they found them frequently reported together, and although ptosis occurs frequently without epicanthus, yet they could only find one instance of hereditary epicanthus *per se*.

High Refractive Error, Nystagmus, and Paralysis of Superior Rectus.—These also have been frequently reported in pedigrees of hereditary ptosis. Usher (1925) reports that in 18 eyes examined, only two had normal vision. My examined case had high refractive error, and affected superior recti.

Squint with Amblyopia ex Anopsia.—This likewise is a fairly frequent concurrent sign, and was present in my case, in the form of a convergent strabismus of 20° and partial amblyopia in his left eye.

Pathology

According to Briggs (1919), the defective action of the levator may be due to five causes:—

- a Defective development.
- b Adhesion to superior rectus.
- c Abnormal insertion.
- d Replacement by connective tissue.
- e Entire absence.

Treatment

In view of this pathology, to attempt advancement of the levator appears to be futile, and therefore some other muscle must be attached to the lid, and used to elevate it. This can be carried out by utilising the superior rectus as in the classical Motaïs operation, or the more recent operation of Greeves (1933), or by utilising the frontales by the classical operation of Hess.

Comment on Pedigree

- Pedigree No. 36*.—II/1. Male. Not examined or confirmed.
 III/8. Female. Examined. No ptosis.
 III/7. Male. Not examined but confirmed. Patient has only a slight degree of bilateral ptosis.
 IV/3. Male. Examined. No ptosis.

IV/4. Female. Examined. No ptosis.

IV/18. Male. Not examined or confirmed.

V/1. Male, aged 5½ years. Marked bilateral ptosis with head tilting, and contraction of both frontales. Marked by defective movement of right globe up, and up and out, and slight defective movement of left globe up, and up and in. Left convergent concomitant strabismus of 20° with 6 dioptres of left hyperphoria.

An interesting point in this child is this:—Having corrected horizontal muscle error by exercises and operation, I attempted to correct his vertical error with 6 prism dioptres, combined with his hypermetropic lenses. The wearing of this prism increased his vertical error, so that without the prism this stood at 12 prism dioptres, while with it the vertical error was stationary at 6 prism dioptres. Apparently he has an abnormal vertical correspondence of 6 prism dioptres, an unusual but not unreported condition.

REFERENCES

- BRIGGS, H. H. (1919).—*Amer. Jl. Ophthal.*, Vol. II, p. 408.
 CLAUSEN, W. (1923).—*Zentralbl. f. die ges. Ophthal. und ihre Grenzgebiete*, Vol. XI, No. 11, p. 481.
 MACKLIN, M. T. (1927).—*Canad. Med. Assoc. Jl.*, Vol. XVII, p. 55.
 MCILROY, J. H. (1929).—*Proc. Roy. Soc. Med.*, Vol. XXIII, p. 285.
 USHER, C. H. (1925).—*Ann. Eugenics*, Vol. I, p. 128.
 USHER, C. H. (1935).—*Trans. Ophthal. Soc. U.K.*, Vol. LV, p. 164.

TREATMENT

- GREEVES, R. A. (1933).—*Brit. Jl. Ophthal.*, Vol. XVII, No. 12, p. 741.

Retinal Detachment

While perusing the available literature, it became apparent that much confusion exists in medical circles regarding the terms familial and hereditary. According to Gould's Medical Dictionary, 4th Edition, the meaning of these two words is as follows:—

Familial.—Characteristic of a family.

Hereditary.—Transmitted from parent to offspring.

I must at once admit guilt myself in including in this survey at least two pedigrees which are only familial, and not hereditary in nature; but I plead privilege in so much as this is only a preliminary survey, and I hope in a final survey to prove their hereditary nature.

The Librarian of the Royal Society of Medicine has been able to secure reports of six pedigrees of hereditary detachments of the retina for me, but one of these is purely familial in character, and therefore not pertinent to the subject under discussion. Furthermore, five of the pedigrees are in myopic families, so that really these pedigrees should be under the heading of—myopia with detachment as a concurrent complication, which conforms to the classification of other myopic defects, such as posterior staphylomata. These points are strongly exemplified in

Schmelzer's (1929) pedigree, in which both parents, and every member of the only affected sibship were myopic, and 3 of the 5 sibs developed detachment, although neither parent had the complication. Again in Bogatsch's (1911) pedigree of 38 members of a family, 11 had myopia, and of these 11, 4 had bilateral detachments, and three unilateral detachments. The one non-myopic hereditary detachment reported, is that described by Pagenstecher in 1913 and reported in the Graefe-Saemisch-Hess Handbuch (1916). Here a grandfather and two grandsons suffered from detachments, with evidence of hypermetropia only. The last-mentioned pedigree bears a definite similarity to the pedigree I am about to present and discuss, in so much that the detachments were definitely hereditary, and non-myopic in nature.

Arruga (1933) has clearly pointed out in his paper the rarity of the hereditary factor in detachments in Spain; and the pedigree he quotes of a father, two sons and a daughter is the most extensive pedigree he found in the examination of 682 cases of detachment of the retina between 1910 and 1932. Anderson (1931) in his monograph dismisses the influence of heredity in half a page.

Treatment

Macklin (1927) quotes all authors as describing the mode of transmission as dominant. Treacher Collins described it as recessive, and Pagenstecher found it a sex-linked recessive character. Macklin explains this disparity as possibly due to detachment of the retina, being a defect associated with several eye diseases. I think this is a most probable explanation.

Comments on Pedigrees

Pedigree No. 37.—The first member of this pedigree to come under observation five years ago, was III/6. He was then 11 years of age and had a complete right retinal detachment, with a hole at 7.30 o'clock, 2 D.D. from the ora. On this eye he had had three Gonin operations performed elsewhere, without success. The left eye had a complicated cataract obscuring all fundus details, and in this eye also was a partial aniridia. By partial I mean an almost complete aniridia of the mesoderm of the iris, with loss of half the ectoderm. The condition on inspection was from centre outwards:—A very dilated pupil, wide ring of iris pigment followed by narrow ring of iris stroma. (See Figure 1.)

Four years later I had to see this boy again for pension purposes, and he then had a mature complicated cataract in his right eye, obscuring all details, but his left eye was most enlightening. The left lens had become shrunken and dislocated upwards revealing a complete detachment of the retina which was white, opaque

FIG. 1.

and studded with glistening crystalline deposits. The detachment in this eye resembled very much the bilateral detachment of III/1, about to be described. The right vision was now doubtful perception of light, and with the left eye no perception of light was obtainable.

The same year I saw III/1, aged 5 years, with bilateral detachments of each retina which were semi-solid in nature, *i.e.*, pseudogliomatous in type. At that time I had no idea she was a cousin of III/6. There were gross vitreous opacities in the right eye, but no hole in either retina. Three years later I saw this case again as an inmate of the Blind Institution. The right eye now had a complete detachment, while the left detachment was unaltered. In the upper half of the left retina was gross choroidal scarring but no hole was discernible in this eye either. The vision in her right eye was now counting fingers at 6 inches, and in the left eye counting fingers at 3 feet.

Case III/8, aged 11 years, came under observation last year. His left sight failed two years previously, and had been treated by the family doctor who can give me no details of his past history. Later he was seen by Dr. Hogg (1936) who found his eye in the same condition as that about to be described. When I examined him his right eye was normal, but took a correction of $+3.0$ D.S. which gave 6/6 and J.1 vision. His left eye had $+0.5$ D.C. $\rightarrow 180^\circ$

a mature cataract with perception of light but no projection. I therefore can only conjecture that a detachment is present in this eye; and if this is so, the high hypermetropia in the right eye is an interesting factor.

Case III/6 and III/8 had a negative W.R. but III/1 had a doubtful reaction at first, which rapidly became negative with

antispecific treatment, so that syphilis is certainly not the causal factor; and this is confirmed by the authors of pedigrees given in the references. But I feel that the unilateral aniridia of III/6, which according to Bell (1932) is a most uncommon condition *per se* may be suggestive. Such aniridia, according to Croll (1929) is supposed to be due to late differentiation of the lens delaying true iris formation. In this eye there was a definite lack of associated developmental determinant in the iris mesoderm and ectoderm. May not this lack also be exhibited in the retina with deficient cohesion between the retinal layers, and ultimate detachment? I think this theory is worthy of consideration, and should other members of this family become affected, I may be able to offer further substantiation.

I am unable to find any members with aniridia in the other pedigrees of hereditary detachment studied and quoted in the references, nor does Bell (1932) find detachment associated with any of the 118 cases of aniridia which she has analysed, but Clausen (1923) mentions that it does occur.

REFERENCES

ANIRIDIA

- BELL, J. (1932).—*Treasury of Human Inheritance*. Vol. II, Part V. Cambridge.
 CROLL, J. J. (1929).—*Archiv. of Ophthalm.*, Vol. II, No. 6, p. 699.
 MACKLIN, M. T. (1927).—*Canad. Med. Assoc. Jl.*, Vol. XVII, p. 937.

DETACHMENT OF RETINA

- ANDERSON, J. R. (1931).—*Detachment of the Retina*. Cambridge.
 ARRUGA, H. (1933).—XIV Concilium Ophthalm., 1933. Hispana, T. 11/1 (abstract).
 BOGATSCH, G. (1911).—*Klin. Monatsbl. f. Augenheilk.*, Vol. XLIX, p. 431.
 CLAUSEN, W. (1923).—*Zentralbl. f. die ges. Ophthalm. und ihre Grenzgebiete*, Vol. XI, No. 11, p. 481.
 CARTER, J. L. R. (1936).—Personal Communication.
 GRAEFE-SÄEMISCH-HESS (1916).—*Handb. d. ges. Augenheilk.* 2nd Edition. Bd. VII/2, Kap. X.a/2.
 HOGG, G. H. (1936).—Personal Communication.
 MANN, I. C. (1928).—*Development of the Human Eye*. London.
 MARSHALL, J. C. (1936).—*Detachment of the Retina*. London.
 MACKLIN, M. J. (1927).—*Canad. Med. Assoc. Jl.*, Vol. XVII, p. 1336.
 SALZMANN, M. (1921).—*Wien. Med. Wochenschr.*, Vol. LXXI, p. 1082.
 SCHMELZER, H. (1929).—*Arch. f. Augenheilk.*, Vol. C-CI, p. 268.

Retinitis Pigmentosa

It is obvious that the elucidation of the nature of retinitis pigmentosa immediately succeeded the invention of the ophthalmoscope in 1851, for while the symptoms were somewhat typical, the actual diagnosis rested with the fundus examination. In 1853 the first case was described by van Trigh, at Utrecht, and about the same time von Graefe described a case in Berlin, but the credit

of classifying all available information must remain with Nettleship. (Bell, 1922).

Under the above heading Nettleship included the following 6 diseases as belonging to one group of hereditary defects of sight :

- (1) Retinitis pigmentosa.
- (2) Retinitis pigmentosa sine pigmento.
- (3) Retinitis punctata albescens.
- (4) Gyrate atrophy of the choroid and retina.
- (5) Stationary night blindness.
- (6) Choroideremia.

Julia Bell (1922) finds no inclination to depart from this classification, and gives her reasons, while Usher in his Bowman Lecture (1935), also gives ample pedigrees to substantiate this contention.

In my investigation of hereditary eye diseases in Tasmania during the past 5½ years I have been able to find only 14 fresh cases of retinitis pigmentosa, eleven of which I have been able to examine. In not one case could I find an hereditary history, but in seven cases—2 in each of 2 families and 3 in another—there was a familial history, and these are depicted in pedigrees 38, 39, 40. In one family (pedigree 38) the parents were first cousins, and I was able to examine only the female member as the male member refused to be submitted to any examination. The female member in this family also refused all treatment except change of glasses.

Concurrent Anomalies

Ring scotoma.—In none of the 22 eyes examined was there a complete ring scotoma, but in three eyes there was a suggestion of one, though nothing more.

Wassermann reaction.—This was done in 6 cases out of 11, but all 6 tests were negative. One of these 6 cases had a Kahn test done and another of the 6 a Kline test, but both of these were negative also.

Deafness.—Two out of 11 patients complained of deafness—these both occurred in pedigree 39. The elder sister was examined by Dr. Hiller (1933) who reported a bilateral nerve type of deafness.

Obesity, hypogenitalism, polydactylism, or mental diseases (Laurence-Moon-Biedl-Syndrome)—Sorsby (1932)—Savin (1935)—did not appear in any of the 11 cases, neither was there evidence of hyperactivity of the sympathetic system such as Raynaud's disease or chilblains. In view of the number of new pedigrees of this syndrome, reported since Sorsby's article on Laurence in 1932, I have asked my medical and surgical colleagues in Tasmania to draw my attention to any cases they may see.

Pathological Examination

None of the 22 eyes examined has been submitted to pathological examination, but I feel that little new would have been revealed by such examination, as the signs and symptoms of the disease conform very closely those reported from England, and those seen by the author while in London. Therefore I have nothing to add to the theories of causation of this disease, except that the theory of vaso-constriction of the retinal vessels does not appear to hold good in the light of recent surgical treatment. (See treatment.)

Consanguinity

This was apparent in only 2 cases out of 14, *i.e.*, 14.28 per cent. compared with Julia Bell's (1922) estimation of 27.2 per cent. in 817 cases, but I shall say more of this under economic significance.

Sex Incidence

Of the 14 cases discovered no less than 10 were females, giving an incidence of 71.35 per cent., but Bell (1922) found a predominance of males in her series. Dickson and Mitchell (1927) report a predominance of females, but quote no figures or references to substantiate this.

Mode of Transmission

Usher (1935) in his Bowman Lecture describes retinitis pigmentosa as a "Mendelian Recessive through a few large pedigrees with many affected individuals showing predominance." He also gives a pedigree of sex-linked retinitis pigmentosa. My 14 cases show no evidence whatever of being hereditary—all pedigrees given (38, 39, 40) being familial in type. Dr. G. H. Hogg (1936) of Launceston, Tasmania, in a personal communication says that he is of the opinion that there are several cases of retinitis pigmentosa related to one another in Northern Tasmania, but that he has not tabulated them to date, and is unable to give me details.

Economic Significance

On closely examining the first 250 pedigrees of this disease given by Julia Bell (1932), I find the following facts:—

Consanguinity. 95 pedigrees.

No consanguinity. 102 pedigrees.

No record of consanguinity. 53 pedigrees.

Total of accurate records. 197 pedigrees.

Percentage of consanguinity in 197 accurate records = 48.5 per cent.

And so it appears that in 197 pedigrees 48.5 per cent. could have been prevented by stricter laws controlling consanguinity. As at the present time there is no dependable treatment for this progressive and disabling malady, the obvious mode of prevention lies in revised legislation, and it behoves all Ophthalmological Societies to bring this fact to the attention of their respective State Governments, and urge action.

J. B. S. Haldane (1936) quotes Usher's cases when asserting that 27 per cent. of retinitis pigmentosa cases are due to consanguineous marriages; and even at this figure legislation is urgently required.

Education

This should be pursued along normal lines with regular ophthalmological examinations (Report of the Committee on Partially Sighted Children, 1934), until such time as the ophthalmic surgeon considers admission to a sight-saving class or blind institution advisable. But the above report stresses the necessity of repeated and close observation of the vision, fields and fundi of each case. From the onset these patients should never be allowed abroad alone between sunset and sunrise, as their disability renders them unsafe in busy traffic at night.

Treatment

Barrett (1934) and Royle (1930 and 1932) both report retardation of this disease from cervical sympathectomy, while Meighan (1935) and Walch (1935) each report three failures from the same operation. In the discussion which followed this paper by Meighan, Hepburn expressed the opinion that early cases might derive more benefit from the operation—a point stressed by Barrett (1934) and Kerr (1935). Recently I have had 1 patient in this series of 14, operated on the right side only. She is a very early case, aged 10 years, with an incomplete ring scotoma in the right field. I watched the case for three years to confirm the fact that it was progressive, and when the pigment commenced to take on a definite bone-corpuscle formation, and when the field at the same time commenced to show early scotomata, I advised operation. The operation has produced a right Horner's syndrome, but I cannot detect any increase in the calibre of her right retinal vessels nor could Walch (1935) in his three cases. After six months both fields of vision show progressive deterioration, and both fundi increase in pigmentation. We must now patiently await results, in my case, but I fear they will be disappointing.

To date I have not tried acetylcholine on any patient with retinitis pigmentosa.

Allied Diseases

With regard to the other diseases of this group, namely, retinitis pigmentosa sine pigmento, retinitis punctata albescens, gyrate atrophy of choroid and retina, stationary night blindness, and choroideremia, no cases of these diseases have come under my notice in Tasmania, neither—to my knowledge—have any cases been reported. Nevertheless this is only to be expected, as in the British Isles with a population 200 times that of Tasmania, cases of these diseases are of very rare occurrence. (Milner, 1932.)

Comments on Pedigrees

Pedigree No. 38.—II/1. Typical case with temporal island in right field, and moderate myopic astigmatism. Parents were first cousins.

Pedigree No. 39.—III/1. Fields reduced to fixation. Complicated cataracts. Vision down to hand movements. Nerve deafness.

III/4. Sister of the above. Fields reduced to fixation. Left eye still 6/18 with myopic astigmatism correction.

IV/6. Examined for myopic astigmatism, but not affected with retinitis pigmentosa.

Pedigree No. 40.—III/1. Not examined. Affected but no details.

III/5. Fields reduced to 10° right and left. Complicated cataracts. Right and left vision with correction = 6/12.

III/6. Affected, but now dead.

Pedigree No. 41.—This pedigree Dr. Carter of Launceston was good enough to send me, but it did not arrive until after this section of my paper had been completed. Every member has been examined by him and his comments are as follows:—

I/1, aged 46 years. Normal.

I/2, aged 44 years. A typical variety of retinitis pigmentosa. Slight field changes.

II/1, aged 21 years. Typical case.

II/2. Normal.

II/3, aged 16 years. Very advanced case. Fields reduced to 10° from fixation.

II/4. Normal.

II/5, aged 5 years. Typical advanced case. Fields contracted to 30° from fixation.

II/6. Normal.

At the moment he is investigating other branches of the mother's family for evidence of the disease.

REFERENCES

- BEDDELL, A. J. (1936).—*Brit. Med. Jl.*, No. 3944, p. 296.
 BELL, J. (1922).—*Treasury of Human Inheritance*. Vol. II, Part I. Cambridge.
 CARTER, J. L. R. (1936).—Personal Communication. Launceston.
 CRAWLEY, R. H. (1934).—Report of Committee of Enquiry into Problems of Partially Sighted Children.
 DICKSON and MITCHELL (1927).—Supplement to *Med. Jl. Austral.*, December 3 p. 462.
 HALDANE (1936).—*Brit. Med. Jl.*, February 8, p. 281.
 HILLER, B. (1933).—Personal Communication.
 HOGG, G. H. (1936).—Personal Communication.
 MACKLIN, M. T. (1927).—*Canad. Med. Assoc. Jl.*, Vol. XVII, p. 1341.
 MILNER, J. G. (1932).—*Brit. Jl. Ophthalm.*, Vol. XVI, p. 418.
 USHER (1935).—*Trans. Ophthalm. Soc. U.K.*, Vol. LV, p. 164.
 WHISHAW, R. (1936).—Personal Communication.

TREATMENT

- BARRETT, J. (1934).—*Trans. Austral. Med. Congress*, p. 206.
 CAMPBELL, GLEN. (1931).—Personal Communication. Vancouver, Canada.
 KERR, H. H. (1935).—*Amer. Jl. of Surgery*, Vol. XXVIII, p. 364 (abstract).
 MEIGHAN, S. S. (1931).—*Trans. Ophthalm. Soc. U.K.*, Vol. LI, p. 124.
 MEIGHAN, S. S. (1935).—*Trans. Ophthalm. Soc. U.K.*, Vol. LV, p. 93.
 ROYLE, N. D. (1930).—*Med. Jl. Austral.*, September 13, p. 364.
 ROYLE, N. D. (1932).—*Med. Jl. Austral.*, July 23, p. 111.
 WALCH, F. B. (1935).—*Arch. of Ophthalm.*, Vol. XIV, p. 699.

LAURENCE MOON-BIEDL SYNDROME

- PATON, L. (1936).—*Proc. Roy. Soc. Med.*, Vol. XXIX, p. 751.
 SAVIN, L. H. (1935).—*Brit. Jl. Ophthalm.*, Vol. XIX, p. 597.
 SORSBY, A. (1932).—*Brit. Jl. Ophthalm.*, Vol. XVI, p. 727.

Strabismus

There is no doubt that strabismus stands pre-eminent as associated with monocular blindness, as the result of loss of binocular fusion. The loss of monocular vision in itself is not so very devastating nor is the loss of binocular fusion, but with only one functioning eye all squinters are in a most precarious position, and any injury or disease to the non-squinting eye may lead to blindness. To the lay mind the cosmetic effect is paramount but to present day ophthalmologists the acquired amblyopia is the main issue, or at least it should be. To Worth (1929) we must do honour, for he constantly urged that permanent amblyopia ex anopsia could be avoided if taken in hand before the patient reached the age of seven. Gradually the old practice of allowing the squinters to grow out of their squints has passed away, and parents are now bringing their children to oculists at a much earlier age. Even in the past 5 years a great awakening has occurred in the parents of Tasmania, so that some bring their children for advice before they are twelve months old. I feel that this has been largely brought about by the introduction of orthoptic exercises, for as Parsons (1936) says, "It (orthoptic training) has one overwhelming argument in its favour, viz., that

when it is successful it cures the squint." Pugh (1936) and Travers (1936) substantiate this view. Most parents are loath to submit their young offspring to operations, but will submit them to orthoptic training almost indefinitely if they observe even slow progress. It therefore appears to me that amblyopia ex anopsia in adults will have almost vanished in the next generation, and thus another milestone in the prevention of blindness will have been passed.

Hereditary Factor and Mode of Transmission

With the increase in patients seeking early advice, the hereditary nature of strabismus has become more apparent (Mayou, 1935), and in many families a history of cousins, etc., suffering from the same complaint can be frequently elicited. In practically every instance the mode of transmission is recessive, nor does there appear to be any sex linkage. Czelletzer quoted by Travers (1936) asserts that the transmission depends on two recessive Mendelian factors. Worth (1929) found amongst 1,373 cases of squint a history of squint in either parent, grandparent, brother or sister of the patient in no less than 711, *i.e.*, about 51 per cent., while Franceschetti (1930) believes that heredity plays but a small part in the causation of concomitant squint. In Tasmania I should roughly estimate the percentage to be somewhat lower than Worth's figures, but I have no statistics to substantiate this view, as heredity not being a potent factor in the individual diagnosis, had not been inquired into by me in every case before I commenced this paper.

Sex Incidence

There seems to be a preponderance of females in both the heterotropias and heterophorias in my pedigrees. In the cases of hereditary heterotropias I find 21 females and 11 males, *i.e.*, 65.6 per cent., and 34.4 per cent. respectively. While in my cases of hereditary heterophorias I find 5 females and 3 males, *i.e.*, 62.5 per cent. and 37.5 per cent. respectively. In the available literature I am unable to find any reference to this factor.

Unilateral Incidence

From my reported pedigrees of heterotropia it would appear that it is usual for the inherited defect to be transmitted to the same sided eye from the parents to sibs, and although I shall give no details of refractive errors it is remarkable how constantly these are transmitted also.

Comments on Pedigrees

Heterophorias

Pedigree No. 42.—The two affected members are mother (I/2) and daughter (II/2). Each suffered from high hypermetropia, the maternal refractive error being the higher. Each has also marked esophoria—the daughter's being greater for distance, while the mother's error was more marked for near vision—a fact to be expected considering the mother was almost at the age of presbyopia.

Pedigree No. 43.—Here the whole family—mother, father, son and daughter have been examined. The mother's examination was negative, the father showed high hypermetropic astigmatism with 2 prism dioptres of right hyperphoria, and 2 prism dioptres of exophoria, the elder child (the son), had no refractive error, but up to 18 prism dioptres of right hyperphoria on fatigue; while the daughter showed moderate hypermetropic astigmatism, with a right convergent concomitant strabismus with amblyopia ex anopsia.

Pedigree No. 44.—In this pedigree a mother (I/2) with moderate hypermetropic astigmatism had from 4-8 prism dioptres of esophoria, while the youngest daughter (II/3) who was slightly myopic has such a high exophoria that it is not accurately measurable by ordinary tests. With orthoptic exercises this was gradually reduced to 14-18 prism dioptres, and finally she became absolutely orthophoric for near and distance.

Pedigree No. 45.—Here a father (I/1) and daughter (II/3) have marked exophoria. The father has no refractive error of consequence, but has from 7-12 prism dioptres of exophoria. The daughter is hypermetropic but had 18-22 prism dioptres of exophoria both for distance and near, and 4 prism dioptres of right hyperphoria for distance. With orthoptic treatment the exophoria has been reduced to 10 prism dioptres for distance and 1 prism dioptre for near, while the hyperphoria has been reduced to 1 prism dioptre. The mother (I/2) is believed also to have some muscle error but to date has not been examined.

It will be noted in the foregoing four pedigrees that the parental muscle error was less in every case than that of the offspring.

Heterotropias

Pedigree No. 46.—Here the mother (I/1) and daughter (II/1) have amblyopic eyes. (I/1) has grown out of her squint, but has marked esophoria with a cover test. While II/1 with 65° of concomitant convergent strabismus has been successfully operated on by me.

Pedigree No. 47.—There is a history of consanguinity in this

family, and also of squint on both maternal and paternal sides of patient (IV/1) who has congenital amblyopia in the right eye. At least one paternal uncle is amblyopic also, but of course this may be ex anopsia and not congenital in origin. II/5, II/6, III/31 and IV/10 have also been examined. II/5 has hypermetropic astigmatism, but no squint; II/6 has senile cataract with hypermetropia; III/31 has right concomitant convergent strabismus, with high hypermetropia and amblyopia ex anopsia; while IV/10, aged 5 years, has left concomitant convergent strabismus, with amblyopia ex anopsia now under treatment. The left sidedness of the strabismus of IV/10 can be accounted for by a birth injury to that side of her face. The other two squinting members of this pedigree who were examined (IV/1 and IV/10) have right-sided squints.

Pedigree No. 48.—This records two sisters and a distant cousin with squint. IV/1 has been under treatment since 1917, and was first examined by me in 1931 with 15° right concomitant convergent strabismus with correction and high hypermetropia and right amblyopia ex anopsia. In 1936 she had 7° right concomitant convergent strabismus, and 6/36 and J.16 vision with correction. After a course of synoptophore exercises this angle was reduced to 4° and the right vision improved to 6/24 and J.8 with correction. Her sister (IV/4) has been under treatment since 1928, *i.e.*, since 6 years of age. She has also straight visual axes, and 6/9 and J.2 vision with correction in the eye which previously squinted. In this pedigree both are right-sided squints.

Pedigree No. 49.—As only IV/2 has been examined no comments are permissible.

Pedigree No. 50.—Here again squinting is apparent both on the maternal and paternal side of the two examined children (III/10 and III/11). III/10 came under treatment when 8 years of age, with a high hypermetropic astigmatism, 45° left concomitant convergent strabismus and left amblyopia which would not respond to prolonged occlusion. Her post-operative result supplemented with orthoptic exercises was perfect. Her brother III/11, had extremely high pure hypermetropia with 55° concomitant convergent strabismus of alternating type. Despite his abnormal correspondence, after two operations he had a perfect cosmetic result. The fusional result it need hardly be said was poor.

Pedigree No. 51.—In this pedigree six members have been examined. I/2 has low grade hypermetropia, but no squint. II/3 has hypermetropic astigmatism and 2 prism dioptres exophoria for distance, while II/5—her sister who is dead—is reputed to have had crossed eyes. II/6 has high mixed astigmatism but no squint.

III/1, the daughter of II/3, has high hypermetropic astigmatism, 35° left concomitant convergent strabismus and left amblyopia ex anopsia. Occlusion followed by exercises and operation have restored both visual axes and vision to normal. III/4 has high hypermetropic astigmatism alone, while III/5 has right concomitant convergent strabismus, high hypermetropic astigmatism, and amblyopia ex anopsia. Both are right-sided squints.

Pedigree No. 52.—The paternal responsibility for strabismus is here clearly shown. Only IV/1 and IV/2 has been examined, and both are under early treatment. Both have squints and high hypermetropia and amblyopia ex anopsia in the left eye, and the onset in each was under 2 years of age.

Pedigree No. 53.—I have only examined two members of this pedigree, neither of whom has a squint, but the propositus III/9 assures me that every squinting member of this pedigree, except II/10, squints with the right eye.

REFERENCES

- CANTONNET, A. (1934).—*Strabismus*. London.
 DOBSON, M. (1933).—*Binocular Vision and Modern Treatment of Squint*. London.
 FRANCESCHETTI, B. (1930).—*Kurze. Handb. d. Ophthal.*, p. 685.
 MAYOU, S. (1935).—*Brit. Jl. Ophthal.*, Vol. XIX, p. 37.
 PARSONS, J. H. (1936).—*Diseases of the Eye*. 8th Edition, p. 558. London.
 PETERS, A. (1924).—*Zeitschr. f. Augenheilk.*, Vol. LII, p. 369.
 PETER, L. (1928).—*The Extra-ocular Muscles*. 1st Edition. London.
 PUGH, M. A. (1936).—*Squint Training*. London.
 RODIN, F. (1934).—*Arch. of Ophthal.*, Vol. XII, p. 874.
 TRAVERS, T. A'B. (1936).—*An Essay on Concomitant Strabismus*. London.
 WILKINSON, O. (1928).—*Strabismus. Its Etiology and Treatment*, p. 57.
 WORTH, C. (1919).—*Squint*. 6th Edition. London.

I should indeed be ungrateful if first I did not acknowledge the great assistance I have received from my colleagues and friends in the compilation of this work. On this side of the Equator I extend my sincere thanks to the following:—G. H. Hogg, M.D., F.R.A.C.S., and J. L. R. Carter, M.B. (both of Launceston); W. D. Counsell, M.B.; Joseph Pearson, D.Sc.; Ralph Whishaw, M.B., M.R.C.P.; A. W. Shugg, M.B.; F. Short, B.A., LL.B., M.B.; D. H. E. Lines, M.B., F.R.A.C.S.; the late L. S. Miller, M.B.; J. Heyer, M.A.; R. S. Waring, M.A.; L. H. Payne, D.S.O.; Mrs. A. McIntyre; Miss K. Waring; Miss N. Gibson (all of Hobart); and especially to Miss L. Gilchrist (Hobart) who so skilfully drew the pedigree charts for me. On the other side of the Equator I must tender my acknowledgements to H. B. Stallard, F.R.C.S., and my most grateful thanks to the Librarian of the Royal Society of Medicine, without whose co-operation this paper could never have been written.

Secondly, I would appeal for a wider co-operation of Ophthalmologists throughout Australia, and the Empire. In Australia there is little or none. Such co-operation can only result in greater cohesion between, and greater knowledge of, the integral parts of our great Empire.

A profound ignorance of the nationality, situation, and population of Tasmania which exists throughout the world is probably mostly due to the remoteness of the island, and if this paper serves no other purpose than to bring Tasmania to the notice of Ophthalmologists throughout the Empire, then I shall be more than satisfied.

And lastly I must offer my gratitude to the Surgeons of Moorfields, whose stimulating guidance in past years has made this work possible to-day.

ACCOMMODATION IN THE EYES OF MAMMALS

BY

SIR JAMES BARRETT

MELBOURNE

THE excellent account of the theories of accommodation published by Edgar F. Fincham opens up, incidentally, the old question of whether mammals, other than primates, can accommodate at all or if so only to a negligible extent.

A great deal of experimental work has been done, and in some cases by experienced physiologists on the eyes of animals which certainly have no useful accommodation, and some erroneous conclusions have been drawn from such experimental work. Nothing is more dangerous in dealing with biological problems than the argument from analogy and also from the assumption of a supposed uniformity of type in the animal kingdom.

I venture therefore to set out briefly the history of the efforts made to ascertain what takes place in the eyes of mammals, other than primates. When the ciliary muscle was discovered by Bowman and Bruecke after 1840, and Helmholtz had in consequence developed the classical theory of accommodation, those interested (possessing the mentality of the period) assumed that the eyes of all mammals behaved just as the eyes of man did. A spate of experimental work began on the eyes of cats, dogs, and other animals which has not quite ceased yet and has sometimes, when dealing with eyes which have practically no accommodation, been misleading. But at an early date observers noted the difference between the ciliary muscle of monkeys and of cats and dogs.