UNILATERAL RETINITIS PIGMENTOSA*

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THE rare cases of unilateral retinitis pigmentosa raise the question whether they represent a distinct clinical entity unrelated to the classical type of retinitis pigmentosa or are abortive forms of this affection.

CASE REPORT

Miss D. L. (Royal Eye Hospital, patient No. 49/5135), aged 52 years (II, 3), first seen on May 14, 1949.

Personal History.—The patient complains of a defective left eye of which she has been conscious all her life. She has been wearing glasses since the age of 11 years. Her general condition is good. She is a well nourished woman with no history of any serious illnesses. She is not conscious of night blindness. Wassermann reaction negative.

FIG. 1.—Left fundus, showing retinitis pigmentosa with characteristic type and distribution of pigment, discoloured disk and narrow arteries.

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Examination	Right	Left
Vision	6/24	6/60
Refraction	+ 1.50 D sph. $\bar{c}$ + 0.5 cyl. axis 90	—0.25 D sph.
Vision with Correction	6/9	Not improved
Ophthalmoscopic Examination	Media and fundus normal, except for a simple melanoma below and nasal to the disk	Slight posterior lenticonus with typical picture of retinitis pigmentosa. The disk is waxy, and the vessels narrow. Typical bone corpuscle pigment is present towards the periphery (Fig. 1)
Fields	Normal	Irregular contraction (Fig.2)
Tests for Dark Adaptation	On testing with the Crookes adaptometer, almost identical curves were obtained for the two eyes (Fig. 3, overleaf). There is some slight lag in dark adaptation, but nothing suggestive of night blindness.

FIG. 2.—Left field of vision, showing marked constriction—object 3/330 white.

FIG. 3.—Graph of dark-adaptation curves as determined by the Crookes adaptometer. The affected eye shows substantially the same curve as the normal eye.

Family History.—The family history revealed that one maternal aunt was registered as a blind person and that another aunt was blind in one eye. The patient's parents were not consanguineous. Fig. 4 shows a pedigree chart of the family.

FIG. 4.—Patient's pedigree chart.

I, 5. Mr. L., father, died aged 69. Reputed to have had normal sight.

I, 4. Mrs. L., mother, died aged 70 of heart disease. Reputed to have had normal sight.

I, 3. Miss H., maternal aunt, aged about 65. Blind in left eye. Normal media and fundus right eye. Seclusio pupillae, secondary cataract of left eye; case of old iritis, ? secondary.

I, 2. Miss H., maternal aunt, aged about 70 years. Has had

bad sight since she was a girl. Found to have diffuse corneal opacities. Anterior segment of sclera thinned in both eyes. Ciliary staphyloma of the left eye. Probably a case of old interstitial keratitis. No retinitis pigmentosa. Wassermann reaction negative.

II, 1. Mrs. B., sister, 53 years old. Found to have normal fundi and vision. Has one son (III, 1), aged 24 years, who is reputed normal.

II, 3. Miss D. L., subject of this report.

II, 5. Mrs. A., sister, aged 43. Found to have normal sight and fundi. Has one son (III, 3) and one daughter (III, 2).

II, 6, 7, and 8. Two brothers and a sister. Examined and found normal.

III, 2. Niece. Found normal.

III, 3. Nephew. Found normal except for a small patch of congenital pigment in upper part of left fundus.

Comment.—This would appear to be a case of unilateral retinitis pigmentosa. It is noteworthy that :

(1) A suggestive family history of blindness proved to have no bearing on the patient's affection.

(2) There were no other affected members nor was there any history of consanguinity in the parents.

(3) The fundus appearances in the left eye were typical of retinitis pigmentosa, a finding supported by the field but not by dark-adaptation tests.

(4) There was nothing in the personal history to account for the lesion.

REVIEW OF LITERATURE

(1) **CASES RECORDED.**—Those reported as unilateral retinitis pigmentosa are summarized in the Table (pp. 102 to 109). Of these a number are clearly inadmissible or of doubtful validity for the following reasons :

(a) Since injury may produce a picture very similar to retinitis pigmentosa, the cases recorded by Hine (1924) and Schupfer (1937), with a clear history of injury, are questionable. It should, however, be stressed that in none of these three cases was the injury direct to the eye.

(b) Night blindness was recorded in a number of cases (Wilbrand and Saenger, 1909 ; Bakker, 1925) ; it was probably also present in several more (Günther, 1913 ; Fricke, 1917 ; Mohr, 1921 ; Štěpka, 1933). It is improbable that a patient with only one eye affected would complain of night blindness. The presence of night blindness therefore raises the possibility of unequally developed retinitis pigmentosa in the two eyes.

(c) A history of preceding inflammation of the eyes was present in some cases (Steffan, 1874 ; Neuffer, 1893 ; Moon, 1904 ; Reuter, 1908 ; Leber, 1916). There is therefore the possibility of an inflammatory reaction producing a picture simulating retinitis pigmentosa.

(d) Diagnosis in some cases is questionable because of poor vision in the apparently unaffected eye, as in the cases of Dumont (1886), Schmidt (1890), Reinecke (1894), and Barrett (1898).

(e) In the case recorded by Mooren (1867) the retina and optic disk of the apparently normal eye were poorly developed, so that the possibility of a congenital anomaly should here be considered.

(f) The cases of Guibor (1933) and Yasutake (1933) are clearly instances of retinitis pigmentosa in unequal degrees in the two eyes.

TABLE
REPORTED CASES OF UNILATERAL RETINITIS

Case No.	Author and Year	Sex	Age	Affected Eye	Vision and Refraction	State of Other Eye	Features other than Retinitis Pigmentosa in Affected Eye	Field of Vision of Affected Eye
1	Pedraglia 1865	M	36	Right	6/18	Normal	Divergent from childhood	Contracted
2	Mooren 1867	F	35	Left	—	Scantly developed retina and optic disk	—	Contracted
3	de Wecker and de Jaeger 1870	F	15	Left	Perception of light —9.0 D sph.	Vision = 2/3 —9.0 D sph. with correction	Divergent	Constricted
4	Baumeister 1873	M	44	Left	Emmetropic Blind since age 12	Normal	—	—
5	Steffan 1874	—	—	—	—	Normal	Choroiditis Vitreous opacities	Concentric contraction
6	Derigs 1882	F	46	Right	1/10	No details given	Posterior polar cataract	Contracted but not concentrically
7	Ancke 1885	F	15	Left	1/3	Normal	Typical retinitis pigmentosa at periphery of fundus, but equator thickly sown with bright confluent dots	Contracted
8	Dumont 1886	F	—	Left	1/6	Vision = 1/3	—	Concentric contraction
9	Guensburg 1890	M	42	Left	1/15	Normal	Posterior polar cataract	Contracted to 15°
10	Schmidt 1890	F	29	—	20/70	20/70	Incipient cataract Unilateral retinitis pigmentosa	—
11	Neuffer 1893	M	16	Left	Amblyopic since childhood	Normal	Vitreous opacities Detached retina Chorio-retinitis Atrophic disk	—

SHOWING

PIGMENTOSA (CHRONOLOGICAL ORDER)

History of Trauma	Family History		General Defects	Period of Observation (years)	Remarks
	Affected Members	Consanguinity			
—	None	No	Deaf in both ears	26	Deutchmann performed pathological examination of affected eye and confirmed findings 26 years later
—	—	—	—	—	Field of vision constricted in both eyes. Poorly developed retina and optic disk in apparently normal eye
—	None	No	—	—	—
—	None	No	Deaf in left ear	—	—
—	—	—	—	—	Had been treated by Steffan for syphilitic irido-choroiditis and vitreous opacities for several years
—	Brother with bilateral retinitis pigmentosa	No	—	—	No history of syphilis
—	—	No	—	—	Defective sight for 8 years No history of syphilis
—	—	No	—	—	Vision becoming progressively worse. No cause mentioned for poor vision in right eye. Was treated with quinine for malaria
—	None	No	—	—	Sudden hemiplegia at age 40. Acute inflammation of right eye recently—kerato-iritis. No evidence of syphilis
—	Brother with bilateral retinitis pigmentosa	—	—	—	—
—	Two sisters with bilateral retinitis pigmentosa, and irregular pigmentation in choroid	Yes—parents	—	—	—

(Table continued overleaf)

TABLE—

Case No.	Author and Year	Sex	Age	Affected Eye	Vision and Refraction	State of Other Eye	Features other than Retinitis Pigmentosa in Affected Eye	Field of Vision of Affected Eye
12	Reinecke 1894	F	44	Left	Counting fingers	20/100	—	—
13	Barrett 1898	M	24	Right	6/12 partly	Blind—fundus appears normal	—	Contracted
14	Rosenbaum 1900	M	20	Right	—	Normal	—	Contracted
15	Schmidt-Rimpler 1901	—	—	—	—	—	—	—
16	Gonin 1902	M	25	Left	1/20	Normal	Convergent squint	Incomplete annular scotoma
17	Moon 1904	M	11	Left	5/4 with correction	5/4	—	Contracted below
18	Nettleship 1908	F	30	Right	6/18 with —2.0 D	Normal	Divergent	Not examined
19	Reuter 1908	M	65	Left	Blind	Normal	Posterior synechiae	—
20	Wilbrand and Saenger 1909	M	46	Right	Counting fingers	Normal Vision = 6/9	Posterior polar cataract Reduced light sense	Concentric contraction
21	Jennings 1911	F	35	Right	5/15 with —0.75 = 5/9	5/5 — normal	—	Contracted
22	Hayden 1912	—	—	—	—	—	—	—
23	Günther 1913	F	15	Left	5/25 Refraction + 2.0 D	Completely normal Vision = 5/7.5 Refraction +2.0 D	Divergent Posterior polar cataract Light sense 1/40 of good eye	Contracted to 10°

continued

History of Trauma	Family History		General Defects	Period of Observation (years)	Remarks
	Affected Members	Consanguinity			
—	—	—	—	—	—
—	—	—	—	—	—
—	—	—	—	—	Night blindness
—	—	—	—	—	This author mentions seeing a case of unilateral retinitis pigmentosa
—	Four siblings died young	No	Hemiatrophy right side of face after abscess of jaw	—	Father died suddenly at 34. Patient died of tuberculosis and the eye was examined pathologically. No evidence of syphilis
—	—	—	—	—	Pigment resembled bone corpuscles. Optic disk pale, not atrophied, vessels normal
—	Not investigated	—	—	—	—
—	—	—	—	—	At age 30 had iritis with normal fundus—left eye (old records). Seen again at age 47 for reduced vision, left eye had typical retinitis pigmentosa
—	One sister aged 51 almost blind and rather deaf	No	—	—	Noticed poor vision in right eye at age 14 Light sense of good eye also affected Night blindness
—	—	No	—	—	—
—	—	—	—	—	Case not described
—	—	—	—	—	—

(Table continued overleaf)

TABLE—

Case No.	Author and Year	Sex	Age	Affected Eye	Vision and Refraction	State of Other Eye	Features other than Retinitis Pigmentosa in Affected Eye	Field of Vision of Affected Eye
24	Leber 1916	F	17	Right	Counting fingers	Normal	Discrete irregular spots with pigmented edges and white centres, and marked black area at macula Vessels normal	—
25	Fricke 1917	F	46	Left	0.2	Vision full Fundus normal	Pupil does not react to light Fundus shows typical picture of retinitis pigmentosa except in periphery which is irregularly mottled	—
26	Bentzen 1917	F	30	Right	Less than 6/36	Normal	Anterior polar cataract	Contracted to 15°
27	Mohr 1921	M	38	Left	6/18	Normal Vision = 6/18	Cortical cataract Night blindness	Constricted
28	Hinc 1924	M	34	Right	6/9	Normal	Posterior polar cataract	Contracted to 5°
29	Bakker 1925	F	—	Left	—	Normal	Light sense reduced	—
30	Rossi 1926	F	—	Right	5/5	Normal	Posterior polar cataract	Reduced
31	Shoji 1926	M	24	Left	0.9 Refraction — 2.0 D	Normal	Reduced light sense	Reduced to 5°
32	Beigelman 1931	F	36	Left	20/25—20/40	Normal	Posterior polar cataract	Reduced to 10°
33	Guibor 1933	M	46	Right (more)	20/70	Vision = 20/40 Bone-corpuscle-like pigment over veins	Posterior synechiae	—
34	McLean 1933	M	48	Left	20/40	Normal	Bone-corpuscle-like pigment and white spots typical of retinitis albicans	—

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continued

History of Trauma	Family History		General Defects	Period of Observation (years)	Remarks
	Affected Members	Consanguinity			
	Mother had bilateral "chorio retinitis pigmentosa"	—	—	—	—
—	—	No	—	—	Night blindness. Husband contracted syphilis (Wassermann reaction positive). Thirteen children : 2, 3, 4, and 6 alive ; 1, 5, and 7 died before 1 year. Six other pregnancies : 3 abortions, 3 stillbirths. Three children examined showed various physical defects (Wassermann reaction negative)
—	—	—	—	—	Quoted by Dreisler
—	None	No	Bilateral middle-ear deafness	—	History of illness for 5 years
Yes	—	—	—	—	Wound over right frontal sinus—kick from mule
—	—	No	—	—	Wassermann reaction negative. Light sense abnormal both eyes : right eye 1/6, left eye 1/10 of normal
—	—	—	—	—	—
—	—	Yes—grand-parents	—	10	Steady increase of symptoms. Wassermann reaction negative
—	—	—	—	6	No history of syphilis
—	—	—	—	—	A bilateral case, unequally marked in the two eyes
—	None	No	—	—	One kidney removed for calculus. Later operation for papillary carcinoma of ureter

(Table continued overleaf)

TABLE—

Case No.	Author and Year	Sex	Age	Affected Eye	Vision and Refraction	State of Other Eye	Features other than Retinitis Pigmentosa in Affected Eye	Field of Vision of Affected Eye
35	Yasutake 1933	M	48	Left	—	Normal	Anomalies of light sense	Concentric contraction
36	Yasutake 1933	M	54	Right	—	Nephritic and diabetic retinopathy	Anomalies of light sense	—
37	Yasutake 1933	M	49	Right	0.4 both eyes	Retinitis pigmentosa pigmento	—	Annular scotoma
38	Štěpka 1933	?	35	—	—	Normal	Reduced dark adaptation	Contracted to 15°
39	Schupfer 1937	F	55	Right	5/7.5	Keratitis when young Corneal nebulae	Lens slightly opaque	—
40	Schupfer 1937	M	24	Left	5/7.5	Normal Vision = 5/5	Poor vision in twilight Light sense reduced	Peripheral vision poor in twilight
41	Agatston 1939	M	53	Right	Hand movements	Normal	Posterior cataract polar	Only temporal field left
42	Agatston 1939	F	45	Left	Perception of light	Normal	Posterior cataract polar	Only temporal field left
43	Lowegren 1940	F	63	Left	0.2	Normal	Posterior cataract polar	Contracted to 10°
44	Dreisler 1948	F	53	Left	No perception of light	Normal	Posterior cataract polar Convergent squint	—
45	Gordon 1949	M	26	Right	20/70	Normal Vision = 20/15	Posterior cataract polar	Constricted

The remaining cases, 25 out of a total of 45 recorded in the literature, are not all equally convincing. Some of them have been observed only once or over short periods so that there is no

continued

History of Trauma	Family History		General Defects	Period of Observation (years)	Remarks
	Affected Members	Consanguinity			
—	—	Yes	—	—	History of syphilis (Wassermann reaction positive). Good response to treatment
—	—	No	Nephritis and diabetes	—	Wassermann reaction positive
—	—	No	—	—	Annular scotoma also in left eye
—	—	No	—	—	Physical condition good. History dates back to age 15. Burn of face and right hand in aeroplane accident, not involving eyes
Yes	—	—	—	—	History of head injury. Light sense reduced both eyes, more in right
Yes	None	No	—	—	Noticed one week after injury to left temple that he saw little in twilight with left eye. Vision later improved from 5/7.5 to 5/5. No change in fundus
—	—	—	Pulmonary tuberculosis	—	—
—	—	—	Deaf in right ear	—	—
—	—	—	—	—	Quoted by Dreisler
—	None	No	—	2	—
—	None	No	—	—	—

record of any possible subsequent changes in the other eye. Others have been recorded with little or no information on the apparently healthy eye. However, in the fifteen cases (Pedraglia, 1865 ;

de Wecker and de Jaeger, 1870; Baumeister, 1873; Guensburg, 1890; Gonin, 1902; Nettleship, 1908; Jennings, 1911; Günther, 1913; Bentzen, 1917; Beigelman, 1931; McLean, 1933; Agatston, 1937; Dreisler, 1948; Gordon, 1949), the fellow eye is clearly normal, ophthalmoscopically and functionally. It may therefore be taken that the clinical picture of unilateral retinitis pigmentosa is a genuine entity, but as only four (Pedraglia, 1865; Shoji, 1926; Beigelman, 1931; and Dreisler, 1948) have been followed for two years or more, it is still an open question whether changes develop in the fellow eye at a later date.

The cases appear to be equally distributed over the two sexes.

(2) ASSESSMENT.—It is generally accepted that the cardinal features of retinitis pigmentosa, apart from its bilateral and symmetrical distribution, are its hereditary character, the generally relentless course, and the presence of night blindness and characteristic field defects. In addition, deaf-mutism is present in some 10 per cent. and mental deficiency in some 4 per cent. of cases. Applying these criteria to the cases of apparent unilateral retinitis pigmentosa, the following considerations emerge :

HEREDITARY CHARACTER.—As can be seen from the Table, in only 23 out of a total of 45 cases recorded is there any mention of a family history ; in seventeen of these 23 the family history is recorded as negative, and in six positive findings are given as follows :

(a) *Cases with Affected Sibs*

(i) *Derigs*.—A brother is recorded as having bilateral retinitis pigmentosa. Since no details are given on the apparently normal eye of Derig's patient, it is open to question whether unequally developed retinitis pigmentosa was present.

(ii) *Schmidt*.—A brother is recorded as suffering from bilateral retinitis pigmentosa. Whether Schmidt's patient is a genuine case of unilateral retinitis pigmentosa is uncertain, as the vision in the apparently normal eye was as poor as that of the affected eye.

(iii) *Neuffer*.—Two sisters had bilateral retinitis pigmentosa and the parents were consanguineous. Here, however, the patient with unilateral retinitis pigmentosa is recorded as showing disease of the vitreous with opacities, detached retina, chorio-retinitis, and optic atrophy. It is therefore possible that this patient was suffering from some incidental inflammatory disease rather than from retinitis pigmentosa.

(iv) *Wilbrand and Saenger*.—A sister was "rather deaf and almost blind". No details are given as to the nature of the blindness. Moreover, the patient himself complained of night blindness, so that the validity of a finding of unilateral retinitis pigmentosa is questionable.

(b) *Cases with Consanguinity*. Information is given in 23 cases, with a positive record in three.

(i) *Shoji*.—Consanguinity in the grandparents is recorded.

(ii) *Yasutake*.—"Consanguinity" is noted.

(iii) *Neuffer*.—This case, already discussed, is of doubtful validity.

(c) *Affected Parents*

There is only one case, that of *Leber*. The patient's mother is recorded as having had "chorio-retinitis pigmentosa". The account given of the condition of the patient suggests that she suffered from an inflammatory lesion in one eye.

RELENTLESS COURSE.—It has already been noted that in the total of 45 cases, only four seem to have been followed for over two years or more. That of *Pedraglia* (1865) was followed for 26 years, when *Deutchmann* confirmed histologically pigmentary changes in the retina. *Shoji* (1926) followed his case for 10 years and noted a steady increase in symptoms. *Beigelman* (1931) followed his case for 6 years, vision slowly deteriorating in the affected eye from 20/25 to 20/40. *Dreisler* (1948) observed his patient over two years, the affected eye being blind. In none of these cases did the other eye become affected. The information given in the remainder is not enough to judge whether the cases are progressive, and whether some of them may not have been early cases of unequal retinitis pigmentosa in the two eyes. This possibility is suggested by the facts that generally in unilateral retinitis pigmentosa vision is recorded as good, and that the cases were presumably fairly early ones.

NIGHT BLINDNESS AND FIELD DEFECTS.—Unilateral loss of dark adaptation appears to have been present in three cases (*Shoji*, 1926; *Yasutake*, 1933; *Schupfer*, 1937), and such information as is given on field defects is not of pathognomonic significance.

SUBSIDIARY FEATURES: DEAF MUTISM AND MENTAL DEFICIENCY.—In only one case (*Pedraglia*, 1865), is deafness, apparently of the acquired type, recorded. In two cases (*Baumeister*, 1873; *Agatston*, 1939), there was unilateral deafness, on the same side as the affected eye and on the opposite side respectively. There is no record of mental deficiency.

DISCUSSION

Much of the recorded material comes from older literature wherein the distinction between retinitis pigmentosa and pigmentary changes in the fundus was not firmly established. But even the more recent cases do not add up to a clear picture. The only point that emerges is that some patients show unilateral fundus changes resembling retinitis pigmentosa, but lack almost entirely the other features of the affection.

In this analysis there is not a single acceptable case with a definite family history of retinitis pigmentosa.

The two acceptable records of consanguinity (*Shoji*, 1926; *Yasutake*, 1933) both refer to Japanese cases and may well require

a local interpretation. Even this is minimized by the fact that in one a syphilitic aetiology is not unlikely. Evidence of unilateral defective dark adaptation was present in only three cases, and in no case was there evidence of deaf-mutism, or mental deficiency. The case reported here is no exception in lacking such essential or subsidiary features. Until such evidence is produced there is nothing to suggest that the picture of unilateral retinitis pigmentosa has any relevance to the classical bilateral affection with its clear-cut genetic aspects and invariable complaint of night blindness. The causative factors in unilateral retinitis pigmentosa are still to be established. It is likely that they will prove to be of environmental origin, though there is a possibility that some may arise as a somatic mutation.

SUMMARY

A case showing the features of unilateral retinitis pigmentosa is recorded. The negative family history conforms to the findings in the literature. In spite of a typical fundus reaction there was no defective dark adaptation. It is suggested that unilateral retinitis pigmentosa is unrelated to the classical bilateral affection with its clear-cut genetic aspects and characteristic functional disturbances.

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