Lattice degeneration in a family

With retinal detachment and cataract

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Nearly a century ago Lang (1885) described the occurrence of retinal detachment in a brother and his sister. Since that time many pedigrees have been presented to illustrate the hereditary nature of some cases of simple (syn. serous, idiopathic, rhegmatogenous, perforated) retinal detachment. Most of the earlier authors related the occurrence of the detachment to the presence and degree of co-existing myopia. Braendstrup (1941) and Cuendet, Streiff, and Dufour (1958) have described a myopic family of five generations in which dominant transmission of retinal detachment was apparent. The hereditary nature of certain cases of detachment is evident from those pedigrees in which a detachment occurs in non-myopic individuals of a myopic sibship (Millikin, 1896; Edmund, 1961). François (1968) has described retinal detachment occurring in an emmetropic family in which dominant transmission occurred. Although this is the usual mode of inheritance in familial cases, recessive and sex-linked (Vogt, 1936) inheritance have also been observed.

Vogt (1924) indicated that it is the predisposing retinal or vitreoretinal degeneration rather than the retinal detachment which is transmitted, and in recent years family studies of certain types of retinal degenerative disorders, especially lattice degeneration, have been reported.

Recently two brothers with retinal detachments received treatment at the High Holborn branch of Moorfields Eye Hospital, and as their mother and other close relatives were also known to have developed a retinal detachment it was decided to examine as many members of the family as possible in order to detect the presence of any asymptomatic predisposing retinal degenerative lesions which might benefit from prophylactic therapy.

Methods

A family tree comprising 110 members was constructed. The progenitors married in 1889 and there is good reason for assuming that they were cousins. An attempt was made to contact as many surviving members of the family as possible. Most were aware of the family history of retinal detachment and welcomed the opportunity to be examined; a small number declined the offer or lived too far away to attend. In the latter case, the local ophthalmologist was informed and asked to examine the patients. Children under the age of 5 years were excluded from the survey.

59 patients were examined either at Moorfields Eye Hospital, or at the Eye Department of the Kent and Sussex Hospital, Tunbridge Wells. An ophthalmic and general medical history were obtained. Routine ophthalmic examination included direct and indirect ophthalmoscopy, and the Goldmann 3-mirror gonioscope was used whenever possible.

Information regarding a further six patients was obtained by studying case sheets of patients examined and treated elsewhere.

Findings (see Pedigree)

A retinal detachment is known to have occurred in eight members of the family (5 bilateral) and there are good reasons for assuming that three other members also developed retinal detachment (2 bilateral), making a total of eighteen retinal detachments in eleven patients.

FIGURE Pedigree of family investigated

In five of the eleven patients, lattice degeneration of the retina was known to be present, and the retinal detachment was bilateral in four of these cases. In the fifth, discovered as a result of the survey (Patient III₂₁), a retinal detachment developed in an eye which had been subjected to trauma some years previously.

Two further patients with lattice degeneration of the retina and two patients with vitreous degenerative changes were encountered as a result of the survey. One other patient had received prophylactic photocoagulation for a retinal hole elsewhere (Patient III₂₄). A summary of the findings is presented in Table I (overleaf).

REFRACTIVE ERROR

None of the affected members of the family had a spherical refractive error greater than —3 dioptres (Table I).

Discussion

The precipitating factor in the formation of a retinal detachment is the development of a retinal break, either tear or hole. Although the break may occur in a region of apparently healthy retina (Dumas and Schepens, 1966), it usually appears within an area of peripheral retinal degeneration. The significance of the various types of peripheral degeneration in predisposing to the formation of a break and subsequent detachment has been reviewed by Dumas and Schepens (1966) and Blach and Bedford (1966). It is generally agreed that lattice degeneration is of special aetiological importance, whereas

Table I Particulars of 16 patients in four generations

Generation	Patient no.	Sex	Predisposing lesion	Age first seen (yrs)	Side affected	Age when detachment first occurred (yrs)	Side affected	Refractive error	
								Right	Left
I	I ₂	F				51	Bilateral	Not known	
	II ₄	F				33	Left	Not known	
II	II ₆	F				50	Bilateral	Not known	
	II ₈	F				53	Bilateral (Aphakic)	Not known	
	II ₉	M				41	Left (Aphakic)	Not known	
	II ₁₂	F	Lattice	54	Bilateral	54	Bilateral (Aphakic)	Not known	
III	III ₈	M				28	Right	Nil	
	III ₁₉	M	Lattice	38	Bilateral	38	Bilateral	$\frac{-3}{-1}$ 90°	Not known
	III ₂₀	M	Lattice	32	Left	32	Bilateral	-1.5 90°	-1.5 90°
	III ₂₁	F	Lattice	29	Bilateral	30	Right (Trauma)	$\frac{-2.75}{-3.25}$ 45°	+0.5 Dsph
	III ₂₄	M	Retinal hole	33	Right			$\frac{-1}{-1.75}$ 60°	$\frac{-1.75}{-1.5}$ 50°
	III ₂₇	F	Lattice	32	Bilateral	32	Bilateral	Not known	
	III ₂₈	F	Lattice	28	Bilateral			-1 90°	$\frac{-0.5}{-1}$ 90°
	IV ₇	M	Vitreoretinal degeneration	23	Bilateral			Nil	
IV	IV ₂₁	M	Vitreoretinal degeneration	14	Bilateral			Nil	
	IV ₂₉	F	Lattice	21	Bilateral			Nil	

cobblestone (paving stone), microcystoid, and pigmentary degeneration are of less serious prognostic significance.

Lattice degeneration of the retina has been well described by Straatsma and Allen (1962) as a sharply demarcated, circumferentially orientated lesion that is located at, or somewhat anterior to, the equator and characterized by retinal thinning, an arborizing network of fine white lines which are often continuous with blood vessels, collections of pigment, and vitreous abnormalities. In addition, they noted the frequent association of multiple round, punched-out areas of retinal thinning or holes, and a tendency for tears to develop along the posterior margin of the lesion. Traction by vitreoretinal adhesions is believed to be important in the pathogenesis of these retinal tears.

Straatsma and Allen (1962) found lattice degeneration to be present in 6 per cent. of a series of 202 autopsy cases, the finding being commonest in middle life. In a similar study Everett (1966) found an incidence of 9.5 per cent. In a clinical study, Byer (1965) found the incidence of lattice degeneration to be 7.1 per cent. in a relatively unselected series of 1,300 patients. The importance of lattice degeneration in relation to retinal

detachment is apparent from the observations of Dumas and Schepens (1966), who found it in 30 per cent. of a selected group of their patients treated for retinal detachment. Straatsma and Allen (1962) reported a similar incidence, 31 per cent., in patients with retinal detachment, or requiring prophylactic treatment. Few population studies to establish the incidence of retinal detachment have been conducted, but Michaelson and Stein (1969) found an annual incidence of 8.9 new cases of retinal detachment per 100,000 population in Israel. As lattice degeneration occurs in about 6 per cent. of individuals, it is obvious that only a small proportion of patients with this condition will develop a detachment.

That a hereditary factor may be operative in determining the development of lattice degeneration is suggested by the reports of its presence in families. Cibis (1965) described the occurrence of lattice in a myopic family in which the development of cataract was also a prominent feature. Everett (1968) described a family of 88 members, none of whom were myopic, in which lattice degeneration occurred in 19 patients (22 per cent.). Unilateral retinal detachment developed in five patients (6 per cent.). An autosomal dominant mode of inheritance of lattice with 80 per cent. penetrance was observed.

In the family described here, lattice was found in six of the 59 patients (10 per cent.) examined by us, and was present in at least five of the eleven patients with retinal detachment. Lattice was bilateral in six of our seven cases (86 per cent.) and was generally associated with a slight degree of myopia (Table I).

Although the progenitors were probably cousins, the occurrence and distribution of the lattice degeneration suggests an autosomal dominant form of transmission with incomplete penetrance rather than an autosomal recessive mode of inheritance.

The role of trauma in relation to retinal detachment is not disputed. Schepens and Marden (1966) found trauma to be a factor in 232 (11.5 per cent.) of a series of 2,016 unilateral cases of retinal detachment. The occurrence of a retinal detachment in one eye of Patient III₂₁ with bilateral lattice degeneration, who received an injury to that eye many years previously, is therefore not surprising.

Three patients in Generation II developed retinal detachment after surgery for cataract. Two further patients in this generation also required cataract extraction, although no retinal detachment followed. One other patient with bilateral cataract was not operated upon. The occurrence of, and age at surgery for, cataract is recorded in Table II.

Table II *Incidence of cataract in Generation II (age at time of cataract surgery)*

Patient no.	Eye	
	Right	Left
II ₁		
II ₂		
II ₃	Cataract (? age at time of surgery)	Cataract
II ₄	Cataract (43)	Cataract (38) (after retinal detachment)
II ₅		
II ₆	Cataract	Cataract
II ₇		
II ₈	Cataract (53)	Cataract (56)
II ₉	Cataract	Cataract (41)
II ₁₀		
II ₁₁		
II ₁₂	Cataract (57)	Cataract (51)

It is of interest to note the early age at which cataract surgery became necessary in four of these patients.

The introduction of photocoagulation and cryotherapy has facilitated the treatment of predisposing retinal degenerative lesions. Colyear and Pischel (1960) reduced the rate of retinal detachment in their patients with retinal holes from 29 per cent. when left untreated to 4.6 per cent. when photocoagulated. Abraham and Shea (1968) report an incidence of retinal detachment of only 0.42 per cent. in 475 eyes treated prophylactically by cryotherapy. However, there is at present no unanimity of opinion regarding when predisposing lesions should be treated. As has been indicated, lattice degeneration rarely progresses to retinal detachment, and many retinal breaks remain asymptomatic. Careful selection of patients for prophylactic treatment is, therefore, necessary. There are, however, several definite indications for prophylaxis; these include the presence of predisposing retinal changes in the fellow eye after the occurrence of a retinal detachment in the first, and the presence of such changes in a patient with a known family history of retinal detachment.

As a result of our survey, three patients have so far received prophylactic treatment, and others have come under observation.

The overall incidence of familial cases of retinal detachment is small. Cuendet (1965) found a definite history in 3.5 per cent. of 454 cases of detachment. There seems little doubt, however, that some cases of retinal detachment could be prevented by regular screening of relatives of affected patients, and the timely use of prophylactic treatment. With the continuing improvement in ophthalmic services, family clinics for retinal scrutiny could assume increasing importance.

Summary

The occurrence of lattice retinal degeneration in a family consisting of 110 members is reported. At least eight members of the family, five with lattice changes, developed a retinal detachment.

It is suggested that careful scrutiny of the retinae of relatives of patients with simple retinal detachment would be of value in detecting predisposing retinal degenerative conditions which could be treated prophylactically before the development of a detachment.

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