LATTICE DYSTROPHY OF THE CORNEA*

OCCURRENCE IN FOUR GENERATIONS OF A YORKSHIRE FAMILY

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DARK and Thomson (1960) reviewed the earlier literature on lattice dystrophy of the cornea and described two new families, one in Leicestershire and the other in Oxfordshire. In each family the condition was inherited as an autosomal dominant, but the authors found certain differences between the families clinically; for example, the age at onset was in the first decade in family "O" and in the third decade in family "L". Also, the fine punctate stromal opacities seen in family "O" were not seen in any case in family "L".

It is the object of the present paper to place on record a further family showing dominant transmission of the lattice type of corneal dystrophy. All members reside in the West Riding of Yorkshire and as many as possible were visited at home, or asked to attend the Out-Patient Department of the General Infirmary, Leeds. At each home visit a limited ocular examination was made and those thought to be affected were asked to attend at the hospital for a more complete examination. This included refraction and examination by the naked eye, loupe, slit lamp, and ophthalmoscope, but did not include the full range of tests used in the admirable and very thorough investigation of Dark and Thomson (1960). For instance, neither intravital methylene blue staining nor examination with ultraviolet light was carried out, nor was corneal sensitivity determined.

The pedigree of the Yorkshire family, covering five generations (Figure, overleaf) is discussed under the following headings:

- (a) Affected members.
- (b) Possibly affected members.
- (c) Members reputed normal but not examined by the authors.

(a) Affected Members

Of the eight members shown as "affected", seven were examined by the authors, the eighth (II. 3) having died before this study began.

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FIGURE.—Pedigree of a family with lattice dystrophy of the cornea.

Case II. 3, a woman born in 1883, died in 1949 at the age of 65 with coronary thrombosis.

Ocular History.—Hospital records show that this woman was an in-patient in 1944 with bilateral lattice keratitis and hypopyon ulcer of the left eye. The visual acuity was not recorded.

The age given below of each of the other seven affected members is the age at the time of examination.

Case III. 2, a man born in 1915 (aged 46), was a woollen worker by trade.

Ocular History.—In childhood a firework had exploded striking the right eye, and vision in that eye had been defective ever since.

Examination.—The visual acuity was 6/9 with correction in the left eye and counting fingers at 2 ft in the right.

The slit lamp showed lattice and punctate opacities in the anterior third of the cornea in each eye.

Case III. 8, a woman born in 1905 (aged 56), was a housewife.

Ocular History.—She had had recurrent attacks of ocular inflammation since early childhood, and had “always had bother with her eyes”. She was first referred to the hospital at the age of 10 from the school clinic, and had been attending ever since.

A right keratoplasty was performed in 1952, and a left keratoplasty in 1960, when the corneal disc was sent for histological examination.

Histological Report.—“Sections of the full-thickness disc show an irregular epithelium separated from Bowman’s membrane, which is fragmented, by eosinophilic degenerative material, some of which is also present in between the parenchymal lamellae. Descemet’s membrane and the endothelium are normal.”

Examination.—The visual acuity was 6/12 with correction in each eye.

Case III. 12, a man born in 1914 (aged 47), was a jam boiler.

Ocular History.—He had had recurrent attacks of ocular inflammation since he was a small child. A left corneal graft was performed in 1957.

Examination.—The visual acuity was counting fingers at 2 feet in the right eye and 6/24 partly with correction in the left (since the lamellar keratoplasty).

The slit lamp showed lattice and punctate opacities in the anterior third of both corneae, except in the region of the lamellar keratoplasty, which appeared translucent and not invaded by lattice material.

Case IV. 3, a woman born in 1926 (aged 34), was a housewife.

Ocular History.—She had had recurrent attacks of ocular inflammation since childhood and defective vision since her late ‘teens. She described several eye injuries during childhood, for instance one at the age of 7 with a shuttlecock, and another caused by a knitting needle.

Examination.—The visual acuity was 6/12 with correction in each eye.

The slit lamp showed typical lattice dystrophy in the anterior third of each cornea.

Case IV. 6, a man born in 1943 (aged 18), was a clerk.

Ocular History.—He had had recurrent attacks of ocular inflammation since childhood with gradual deterioration of vision. At age 2, he had an eye injury said to “result in squint for which he had operation at age 7”. He had recently given up night school because of difficulty with reading.

Examination.—The visual acuity was 6/18 with correction in the right eye, and 6/24 with correction (Reads J2) in the left.

The slit lamp showed typical lattice and punctate opacities in the anterior two-thirds of both corneae.

Case IV. 15, a schoolgirl born in 1947 (aged 13).

Ocular History.—The mother stated that she had had “attacks” lasting about 3 days, from when she was a small child. During an attack the eyes “clouded up” and even closed, she had photophobia and had to retire to bed.

Examination.—The visual acuity was 6/9 (reads J1) in the right eye and 6/9 (partly) in the left.

The slit lamp showed the early appearance of typical lattice opacities in the anterior third of the corneae. No punctate opacities were seen.

Case V. 2, a schoolgirl born in 1949 (aged 11).

Ocular History.—Her mother stated that, for the last year or two, she had had difficulty in reading music when playing the violin—"The notes run into each other."

Examination.—The visual acuity was 6/6 (reads J1) in the right eye and 6/6 in the left.

The slit lamp showed very early commencement of lattice dystrophy in both corneae. The striae were fine with a radial orientation. The axial area of each cornea was unaffected.

(b) Possibly Affected Members

Case I. 2, a woman, had died before the first World War.

Several of her relations described her as "having always had poor eyesight". Her son (II. 6) believes this was first discovered after an eye injury, when his mother was about 16. To quote his own description, "on removing the bandages after this injury, it was seen that something had appeared on her eyes." He remembers she "held herself very erect and staring—like a blind person" and was considerably handicapped, though able to move about the house. Another relative thought she "had cataracts".

Case I. 1, a brother of I.2, was described as "having had eye trouble".

Case II. 1, a man born in 1879, lost one eye through an injury at the age of 14, and is said to have attended hospital because of poor vision in the remaining eye. However, no record of such attendance could be traced, and there is no mention of ocular defect in the case notes relating to his death in hospital following prostatectomy at the age of 60.

Case II. 2, a woman now about 80 years of age, was said to "have cataracts" and to be nearly blind. No examination could be made, however, since she had never sought medical aid and was unwilling to take any part in this survey, even to be visited at home.

(c) Members Reputed Normal but not Examined by the Authors

I. 3, another brother of I.2, is described as having been unaffected.

II. 4, a man now dead, is reported as having had "good eyes".

II. 5, a man now aged about 71 years, could not be traced but is reputed normal, as are his two children (III. 13 and 14).

III. 1 and 3 were examined at the General Infirmary, Leeds, when their affected brother (III. 2) first attended; they were both recorded as normal.

III. 4-7, are believed to be unaffected.

III. 9 and 10 were unwilling to be interviewed but both wrote saying that neither they nor their children had had trouble with their eyesight.

III. 15 has good vision according to his father (II. 6).

IV. 2, a boy aged about 8 years, and **IV. 17 and 18** have not been examined but are thought to be unaffected.

Discussion

Clinically there is a remarkable uniformity in the age at onset and in the progress of the dystrophy in the seven surviving affected members of this

family, and in each case symptoms date from early childhood. The age at manifestation is in the first decade and it is very interesting to note that even at this early age the lattice dystrophy is bilateral. The condition is often put down to an injury by the patient, whereas in fact a minor eye injury focuses attention on the eyes and reveals the developing dystrophy. The fine punctate stromal opacities make their appearance when the abiotrophy becomes firmly established; they were absent in the two schoolgirls aged 11 and 13, who showed little change in their vision, but were present in the 18-year-old clerk with defective vision.

The pattern of inheritance is that of an autosomal dominant gene with complete penetrance and remarkably constant expression. There are six affected among a total of fourteen offspring of "certainly affected" members, giving good agreement with the one in two risk to children which is to be expected in such transmission. Every person showing the typical corneal changes has a parent similarly afflicted or a parent in whom the condition has been suspected, and the fact that two sibs in the first recorded generation are reported to have had "bad eyes" indicates possible involvement even earlier in the pedigree.

The Yorkshire family, therefore, shows a similar mode of inheritance and the same "striking intrafamilial constancy in the age at onset and evolution of the corneal changes" noted in the two families reported by Dark and Thomson. Further, the clinical picture, including the consistently early age at onset, type of corneal opacity, etc., is indistinguishable from that of Family "O" of the earlier report. The underlying chromosome abnormality is probably identical in the Yorkshire and Oxfordshire families, and it would not be unreasonable to suggest that the two families may even share a common affected ancestor.

Summary

The pedigree of a Yorkshire family afflicted with lattice dystrophy of the cornea is presented. Of the eight "certainly affected" members, seven were examined by the authors. Clinical aspects are described and details are given of the histopathology of a corneal disc.

The pattern of inheritance is that of an autosomal dominant gene with complete penetrance and constant expression.

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The author of the histology report quoted for Case III. 8 is Dr. René Barry, Assistant Pathologist, Institute of Ophthalmology, London.

REFERENCES

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