Familial occurrence of dot (microcystic), map, fingerprint dystrophy of the cornea.

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The clinical description of corneal microcysts, map-like changes, and fingerprints has led investigators to the conclusion that these changes may represent a corneal dystrophy. The familial or hereditary evidence which is usually necessary to label a corneal disease a dystrophy has been lacking. This paper describes a familial pattern of disease in two families where three generations were involved and in eight families with corneal changes in at least two generations.

Interest in pathologic alterations of the anterior cornea increased after Cogan and co-workers described cystic changes of the corneal epithelium in five women aged 44 to 72 years old. Bilateral, nonvascularized, central, epithelial, cystic corneal changes were noted in their first four cases. In the fifth case, these changes were present in the patient's only remaining eye. The patient's other eye was removed 30 years before. The pathology of this eye was reviewed for their paper, and a solitary corneal epithelial microcyst was noted as well as extension of basement membrane into the epithelium. Although observations were not systematically made on other members of the family, the authors referred to their findings as a "Microcystic Corneal Dystrophy."

One year later, Guerry reported on nine cases of microcystic dystrophy adding his observations of a background of "map-like changes," to the "small, grayish, puttylike dots," previously mentioned. Eight of Guerry's nine cases had these dots. He felt the dots occurred in association with the map-like dystrophic changes. There were seven women and two men in his series and all were adults. He did not comment on any hereditary pattern to this dystrophy. Others have confirmed these findings, but investigation of families for similar pathology has not been reported.

Fingerprint-like lines in the superficial cornea were described in 1950 by Guerry. He saw "peculiar wavy lines . . . seen best by retroillumination and of such a whorl-like contour as to suggest fingerprints." These changes were not present in three children of one case, nor in three siblings of a second case. DeVoe added three similar cases to the literature but could not assign an etiologic cause to this condition. He felt trauma could be responsible for the changes in some cases. While commenting on this paper Guerry mentioned that one of his cases reported 12 years before (1950), had shown absolutely no change in the intervening years and still had fingerprint-like lines in the epithelium.

Kaufman and Glower also noted these changes and described both the fingerprint-like lines and maps in the same cornea. They felt these changes were due to irregularities of Bowman's membrane or the epithelial basement membrane. Some of their cases were symptomatic with repeated painful corneal ulceration and reduction in vision. Family studies were not reported.

The combination of fingerprint lines, maps, and dots with or without corneal erosion was also reported by Bron and Brown and Trobe and Laibson. In neither report was there identification of a familial pattern to these superficial corneal changes.

We wish to report eight families where the dot, map, and fingerprint changes occurred in two generations, two families in which three generations were involved, and two families with affection of multiple siblings.

Case reports.

C. M. family. A five-year-old child was referred to the Cornea Service at Wills Eye Hospital because of a recurrent corneal ulcer. She had been well until 4.5 years of age when she began having a series of corneal ulcers. Her referring ophthalmologist thought these recurrences were due to herpes simplex virus because of their geographic appearance, but when she did not respond to antiviral and antibiotic drugs she was sent for evaluation. Her past and family history were normal. Examination of the cornea of her left eye revealed a healing corneal erosion and whorl-like lines in the epithelium resembling the map-like pattern of changes described by others. These map-like changes were noted away from the healing erosions in her left eye and were also seen in the asymptomatic right eye. Her mother, who accompanied her, was also examined by slit lamp on the initial visit. Although she had no ocular complaints and had never had a corneal ulcer, map-like changes were noted in her left eye with several intra-epithelial microcysts scattered in the map-like area. Only map-like changes were found in her right eye. We requested that all the members of the family be examined and fortunately they were available and cooperative (Fig. 1). No corneal changes were noted by slit lamp examination in two of the mother's siblings. The three-year-old brother of the propositus was also negative. The 63-year-old grandmother of the propositus, who had never had a corneal or ocular problem was discovered to have map-like and
fingerprints in each eye. These changes were noted centrally without corneal scarring or signs of past recurrent corneal erosion. Vision of the mother and grandmother was normal. When the recurrent erosion healed in the child, her vision also returned to 6/6. The corneal sensitivity of all members of the family was normal.

The corneal erosion was treated and responded to patching. Five per cent sodium chloride ointment was applied at bedtime for one month following the last erosion episode and the child has remained asymptomatic in a six-month follow-up period.

S. G. Family. Sharp pains had awakened a 63-year-old woman from sleep three or four nights a week for three months. When she was examined by her ophthalmologist he found a corneal erosion of the right eye with peculiar changes in the superficial cornea of both eyes which could not be classified. This led to referral and examination by the Cornea Service at Wills Eye Hospital. Prior to the onset of her complaints three months before, she had been comfortable and had visual acuity of 6/7.5 in the right eye and 6/6 in the left eye with best correction. Her right eye had always been slightly blurred compared to the left. Slit lamp examination revealed a healed corneal erosion in the pupillary area of the right cornea with minute punctate epithelial staining spots in the area of the erosion, demonstrated with both fluorescein and Rose Bengal dyes. In addition to the central corneal changes, map-like lines were seen in the corneal epithelium in the mid-periphery. In her left eye there were fingerprint lines in the superior half of the cornea. She had experienced intermittent pain in the left eye during sleep which awakened her several times in the past. These lines were noted in the epithelium, but were not present in the stroma. The remaining ocular examination was normal, including corneal sensitivity and the Schirmer test. Because of our experience with the first family, we requested that her son and daughter be examined as well as her grandchildren. She had no siblings and her parents were deceased. As far as she could recall, there had been no previous ocular problems in either parent. When her thirty-four-year-old son was examined, map-like changes were noted in his right eye. He had been asymptomatic and had never experienced corneal ulcers, erosions, or even a history of pain or foreign body sensation in either eye. His thirty-year-old sister was normal. We requested that his children be examined even though they were asymptomatic too. A twelve-year-old son had map-like changes in one eye which were only visible in retroillumination with the red reflex of the fundus as background illumination. These changes were noted on examination by two different observers at two separate examinations and were stationary. Corneal sensitivity was normal in each member of the family. No other positive corneal findings were found in two other children of the son of the propositus.

In addition to these two families with either map-like lines, fingerprints, microcystic epithelial changes, or combinations of each in three generations, we examined eight other families and found similar corneal changes in two generations in these families. Most of these changes were map-like intra-epithelial lines and microcystic epithelial lesions. The pathology was seen in the epithelium in each case but not in the stroma. Fingerprint lines were present in some members of these families, however, the more consistent finding was the map-like change with or without epithelial microcysts.

In three families, multiple siblings were noted to have these changes, however, examination of all family members was not possible because of geographic problems.

Discussion. Previously, the changes described by various authors and called microcystic corneal dystrophy, fingerprint dystrophy of the cornea, map-like corneal dystrophy, or dystrophic changes in the anterior cornea did not link family members with similar pathology. Although it has been suspected that these conditions may represent the changes of a corneal dystrophy, the familial evidence to show this has not been published to our knowledge. These corneal changes have the characteristics of a corneal dystrophy in that they are more central than peripheral, bilateral in many cases, are not accompanied by vascularization or severe inflammation, and are noted now to be hereditary.

The pathology in similar cases has been described by Cogan and co-workers and by Rodrigues and co-workers. The defect causing this clinical picture may relate to production of aberrant basement membrane or misdirection of the corneal epithelial basement membrane.

Similar anterior corneal changes, particularly the map-like and fingerprint changes have been seen in the superior half of the cornea in several
patients following cataract extraction. These changes were found only in the operated eye and family studies were negative in two instances. All cases with this pathology, therefore, may not necessarily be familial and surgery may play a role in some cases.

The pattern of heredity in our nonsurgical cases is apparently dominant. These changes have been seen in children and adults, but are most common in the middle-aged and elderly population. The relative frequency of these changes is unknown and under investigation. A more general term based upon the familial appearance and pathology might be epithelial basement membrane dystrophy of the cornea.


Key words: microcystic dystrophy, corneal epithelial dystrophy, anterior corneal dystrophy, fingerprint lines.

REFERENCES

Electron diffraction study of asteroid bodies. WAYNE F. MARCH AND DAVID SHOCH.

Electron diffraction studies of the crystalline particles in human vitreous asteroid bodies indicate that they contain calcium oxalate monohydrate and calcium hydroxyphosphate (hydroxyapatite).

Asteroid bodies appear as numerous but innocuous, spherical opacities suspended in the vitreous. Their composition has intrigued investigators for one hundred years, and minute crystals have been noted to be embedded in the larger body.1 A previous report2 has described the appearance of these crystals surrounded by an amorphous matrix, which make up a “satellite” asteroid body. These “satellite” asteroids probably conglomerate to form the macroscopic bodies.2 Analyses of macroscopic bodies have shown them to contain a variety of materials,1, 3 but no prior study has determined the chemical composition of the individual crystalline particles. By localizing a crystal under the beam of a transmission electron microscope then adding an electron diffraction aperture, diffraction patterns for selected crystals were obtained.

Methods. The eye of a 79-year-old man was enucleated because of a malignant melanoma and placed in formaldehyde. Asteroid bodies in the vitreous appeared typical with polarized light, oil red O, and alcian blue stains. Sections for transmission electron microscopy were prepared as previously described.4 Individual crystals were selected with the direct beam of an AER electron microscope before using the diffraction aperture. A gold standard was employed to obtain the camera constant (2\(\lambda\) = 38.26 to 38.76). A

Fig. 1. An electron diffraction pattern.