HEREDITARY EPITHELIAL DYSTROPHY OF THE CORNEA* MEESMANN TYPE

BY

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IN 1963 our out-patients department was visited by a 27-year-old man with a corneal foreign body. On slit-lamp examination, both corneae appeared to be studded with numerous tiny, grey-white points, which were situated in the epithelium. In retro-light they appeared as fine, round, or somewhat irregular vesicles or droplets spread over the entire corneal surface, apart from the limbal area. Towards the periphery, the vesicles were somewhat larger and, at several points, islets of corneal epithelium had remained unaffected. Bowman's membrane, the parenchyma, Descemet's membrane, and the endothelium showed no alterations. The patient had never had eye-complaints. In 1958, a brother of the propositus had been treated for a unilateral traumatic corneal erosion. A diagnosis had been made of the rare bilateral vesicular corneal affection which may be caused by dye-stuffs. This patient had done some painting in his home.

The occurrence of an identical picture in these two brothers called to mind the hereditary epithelial dystrophy of the cornea first described by Pameijer (1935) and subsequently by Meesmann (1938), and to verify this diagnosis, a family investigation was carried out, in which 32 persons (16 men and 16 women) were examined. Five men and five women appeared to be affected by this rare hereditary corneal dystrophy (Figure.)

FIGURE.—Pedigree showing affected members of the family of a man with Meesmann's dystrophy.

Curiously enough, these patients did not complain of the occasional irritation of the eye which is a common symptom of the disease. Although the picture was essentially the same in all patients examined, there were variations in the density, site, number, size, and shape of the epithelial vesicles, irrespective of the age and the sex of the patients. In some cases, the corneal surface was slightly irregular and some of the

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vesicles stained with fluorescein. Visual acuity was slightly diminished (0.8–0.9), probably owing to the presence of a considerable degree of corneal astigmatism in nearly all cases. Intra-ocular pressure was normal in all cases. Apart from convergent squint in one case, there were no other ocular anomalies.

Pameijer (1935) reported a family five members of which showed bilateral vesicular epithelial corneal alterations. The stroma and endothelium were normal and corneal sensitivity was unimpaired. Only the older members suffered from ocular discomfort.

Meesmann and Wilke (1939) described three families affected by an epithelial dystrophy of the cornea, characterized by small dots and vesicles in the epithelium together with slight irregularities of the corneal surface. As in Pameijer's cases, the cornea would repeatedly stain with fluorescein. The condition was inherited as an autosomal dominant. The average visual acuity was about 0.5 and never below 0.33. The disease began in infancy, progressed slowly, and was accompanied by attacks of irritation.

Other families with this dystrophy have been described by Böck (1941), Bürki (1946), Paufigue and Étienne (1950), Stocker and Holt (1955), François (1957), Stokes (1960), Snyder (1963), Kuwabara and Ciccarelli (1964), and Kornzweig (1964). These reports show that this form of corneal epithelial dystrophy is transmitted in a dominant manner. Both sexes are affected and the sex ratio is 50 : 50. Most authors mention that the majority of the patients complain from early childhood of attacks of photophobia, itching, burning, redness, and watering of the eyes. These complaints tend to decrease with advancing age. In many cases, however, subjective symptoms are lacking. The objective picture given by later observers corresponds on the whole to the first descriptions of Pameijer and Meesmann. Moreover, delicate, serpiginous opacities at the level of the deepest epithelial layers of the cornea were frequently seen, especially in elderly patients. The youngest patient observed was a 5½-month-old girl. With regard to the sensitivity of the cornea opinions differ: it is said to be either normal or more or less diminished. Visual acuity, though originally normal, diminishes gradually; but rarely becomes less than 0.4.

Pathology

According to Meesmann and Wilke (1939), the main lesions are located in the epithelium of the cornea and that of the adjacent conjunctiva. Apart from uncharacteristic degenerative alterations of the epithelial cells, such as swelling, pycnosis, and nuclear disintegration, there was vacuolation of the epithelial cells due to accumulation of glycogen (Best staining method). Furthermore, there was irregularity of the palisade-like arrangement of the basal layer. Bowman's membrane was normal.

Stocker and Holt (1955), likewise reported changes in the epithelial structure, degeneration, vacuolation, and cyst-formation. By means of the modified periodic acid-Schiff staining method, they demonstrated the presence of an intracellular and intercellular substance which stained a dark red. In addition, they found, instead of a normal thin basal membrane, an amorphous dark-red-stained layer of irregular thickness with pedunculated excrescences in the epithelium. Between this substance and the normal Bowman's membrane there was sometimes a somewhat less dark-red-stained material. The corneal stroma, Descemet's membrane, and the endothelium were perfectly normal. This shows that the affection is limited to the epithelium and that the pathological material is presumably the product of the diseased epithelial cells. Kuwabara and Ciccarelli (1964) carried out a histological and electron-microscopical study of the epithelium, and showed that the superficial cyst-like structures contain degenerate products of epithelial cells. Marked thickening of the basal membrane of the epithelium was present. The presence of glycogen deposits in all epithelial cells may be due to the rapid turnover of these cells.

Differential Diagnosis

The conditions which can be easily excluded include the transient corneal epithelial oedema occurring in glaucoma, in diffuse vesicular keratopathy, and after instillation of cocaine. Fuchs's dystrophy is characterized by both epithelial vesicles and bullae as well as by marked alterations of the endothelium and stroma.

The familial corneal epithelial dystrophy of Kraupa (1934) is characterized by epithelial vesicles, white dots, and diffuse stromal opacities in the centre of the cornea.

The picture of the superficial corneal degeneration (the *tâcheté* type) described by Rollet (1933) resembles Meesmann's dystrophy and is also transmitted as an autosomal dominant. However, in Rollet's type the alterations are grossly visible and consist of small white epithelial spots composed of numerous points in the central corneal area. Since typical vesicle formation has not been mentioned, it is doubtful whether this can be identified with Meesmann's dystrophy.

Gifford (1932) reported an epithelial vesicular corneal dystrophy accompanied by decrease in the intra-ocular pressure which occurred mainly in elderly women. Heredity was not mentioned.

Cornea verticillata, characterized by the familial occurrence of superficial whorl-shaped opacities, does not show vesicular alterations in the corneal epithelium.

Treatment

If subjective complaints are present conservative measures are recommended. If visual acuity is much reduced keratoplasty must be taken into consideration. However, in the case in which Stocker and Holt carried this out, a relapse of the dystrophy was noted. Stokes (1960), as well as Kuwabara and Ciccarelli who removed the corneal epithelium, observed that the epithelial lesions recurred after some months.

Summary

In a family of 32 persons examined, ten cases of dominantly inherited corneal epithelial dystrophy of the Meesmann type were found. None of the patients had ever had any ocular complaints. The clinical appearance, differential diagnosis, treatment, and pathology are discussed.

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