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A rare case of total hereditary sclerocornea is presented along with clinical history, histopathology, and a review of the possible pathogenesis and current literature on the subject. It is quite possible that imperfect cleavage of the anterior chamber angle may be the predisposing factor in scleralization of the cornea due to a failure of neural crest cell differentiation. If surgical intervention to correct for the corneal opacification is indicated, corneal transplantation should be performed as soon as possible after birth, similar to the indications for removal of dense congenital cataracts to avoid deprivation amblyopia.

Sclerocornea is a rare, intriguing congenital abnormality that may be the extreme form of anterior chamber angle dysgenesis (1,2). Normally the cornea is crystal clear at birth; in sclerocornea, it is opaque (scleralized). The corneal opacification may be only peripheral with a clear but relatively flat central cornea, and the vision is fairly normal. If the corneal opacification is diffuse (total sclerocornea), vision is profoundly impaired, and suppression amblyopia is common.

Current embryologic concepts hold that the corneal endothelium is formed by the neural crest cells surrounding the optic cup, which migrate between the primitive corneal stroma and the lens vesicle (3-5). Neural crest cells also give rise to the anterior 2/3 of iris stroma, including the iris melanophores (6). Failure of normal differentiation of the neural crest cells therefore may account for a spectrum of congenital anomalies ranging from sclerocornea to congenital glaucoma, including Peter's and Rieger's anomalies.

With only slightly more than 100 cases reported, sclerocornea is relatively poorly understood by clinicians. Peripheral sclerocornea, which is dominantly inherited, is more frequently encountered. However, hereditary total sclerocornea is extremely rare because it is a recessive trait. Its effect on vision is devastating. The following case report illustrates total hereditary sclerocornea that was successfully treated surgically by corneal transplantation. Unfortunately, the patient's vision failed to improve because of longstanding deprivation amblyopia.

Case Report

A 53-year-old woman was first seen on February 15, 1983, because of poor vision in each eye. She had had bilateral opaque corneas since birth. This condition was present unilaterally in three of her brothers and one niece, while a great niece had bilateral opaque corneas. The patient's parents, aunts, and uncles had normal eyes, but one sister had bilateral Rieger's anomaly. There was no history of intermarriage among the family and relatives.

The patient's visual acuity for the right eye was the ability to count fingers at one foot, and for the left eye, at six inches (Fig 1). Slight enophthalmos was present as were random eye movements and unsteady fixation. The pupils were not visible although a small area of iris could be seen in the right eye supranasally. Scleralization of both corneas, centrally and peripherally, was present, and fine vessels extended centrally from the limbus at the level of the anterior stroma. The anterior chamber was not clearly visible. A small area of iris was seen peripherally in the right eye between 12 and two o'clock.

Intraocular tension was 15 mm Hg in the right eye and 18 mm Hg in the left eye. Ultrasound examination showed normal axial length with normal appearing posterior segment. Both lenses could be seen anteriorly, but whether the cornea and lens were attached was uncertain. The patient had accurate color perception in both eyes for red, green, and blue and had good two-point and intensity discrimination. Entoptic phenomena appeared to be very good.

Penetrating keratoplasty was recommended as a means to produce significant improvement in vision. Initial surgery was chosen for the left eye because it had a denser scar and better entoptic phenomena.

A penetrating keratoplasty was successfully performed (Fig 2), but the vision was only slightly improved in the left eye postoperatively even though the media were relatively clear. The persistent poor vision was attributed to suppression amblyopia secondary to the congenitally opaque cornea.

Pathology

The following microscopic findings were present. The corneal epithelium was thinned, flattened, and atrophic (Fig 3). There was edema of the basal cell layer of the epithelium and hyaline subepithelial deposits, which were lightly PAS-positive and of variable thickness. Bowman's membrane was absent (Fig 4). Irregularly arranged stromal

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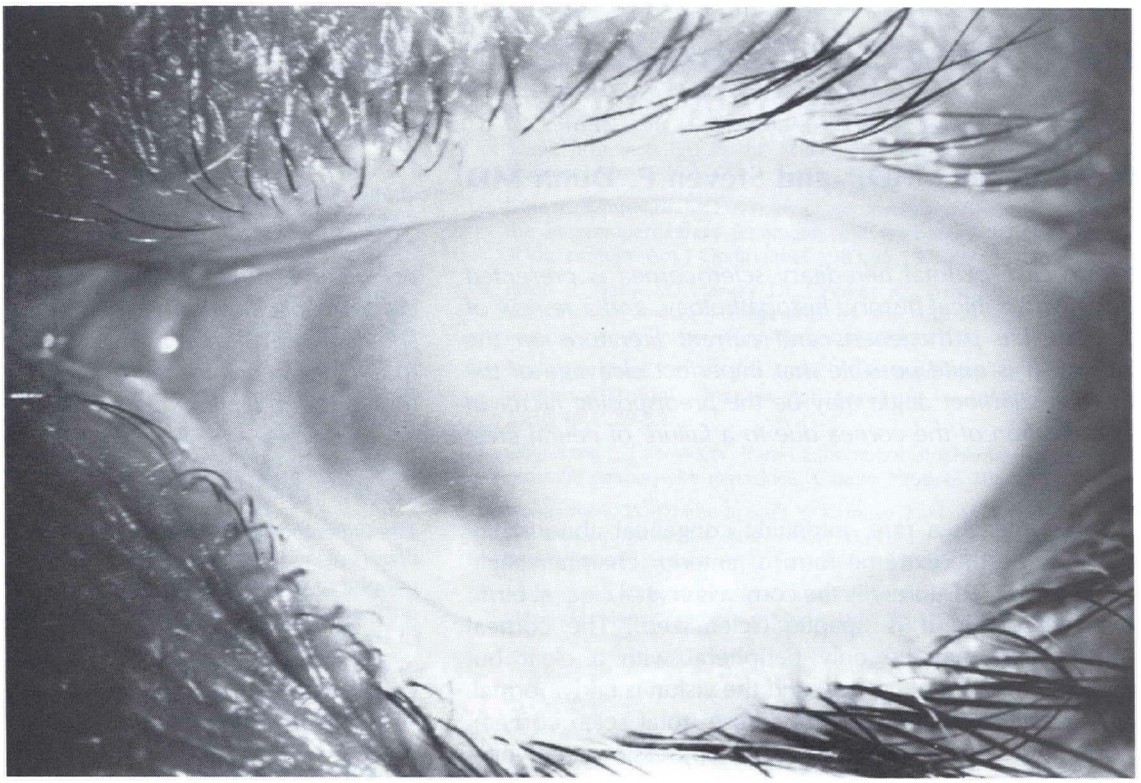

Fig 1
Total sclerocornea. Almost complete opacification of entire cornea with occlusion of visual axis.

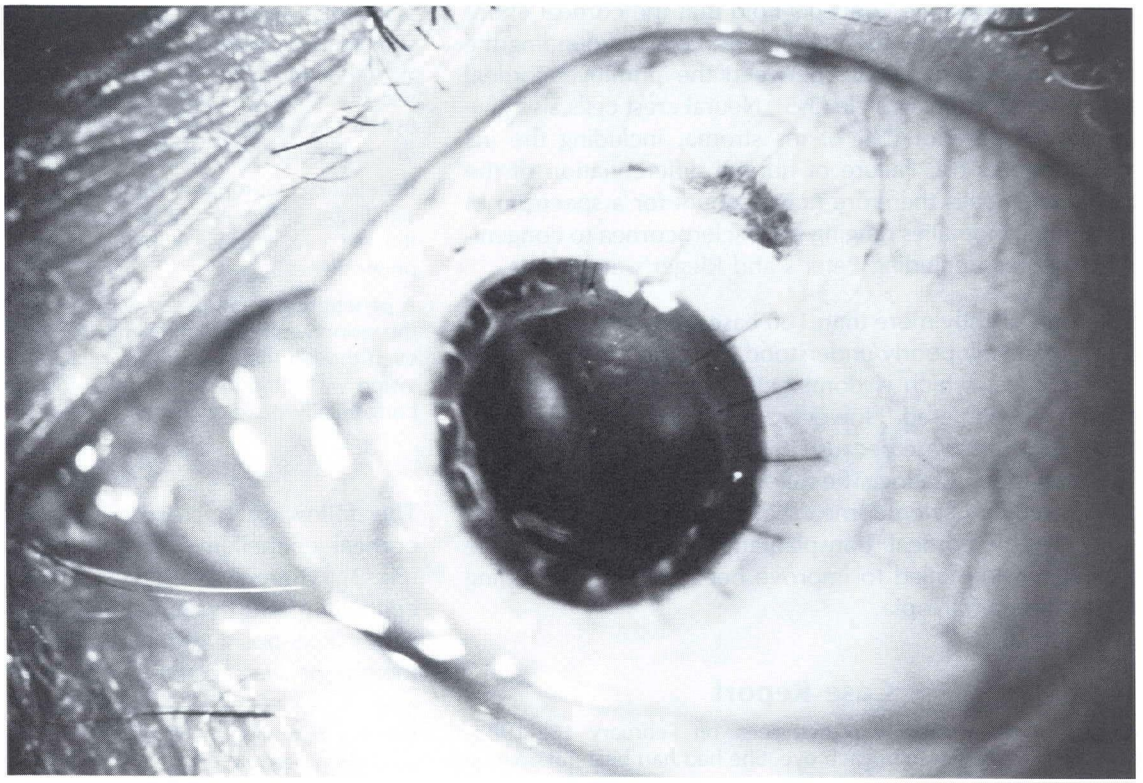

Fig 2
Postoperative appearance following penetrating keratoplasty. Note clarity of graft and relatively normal central zone in visual axis.

collagen fibers, thicker superficially, were similar to those seen in the sclera (Fig 5). Vascularization of the corneal stroma was present at all levels, but there was no inflammation in the stroma. The interlacing collagen stromal fibrils had variable diameters, the narrowest bundles in the deeper stroma. Descemet's membrane and the endothelium were absent centrally, but peripherally there was a thin layer of Descemet's membrane with sparse, flattened, endothelial cells. Flattened pigmented cells were seen in the deeper stroma centrally, particularly where Descemet's membrane and the endothelium were absent (Fig 6). The findings were compatible with the clinical diagnosis of sclerocornea totalis.

Discussion

Hereditary sclerocornea probably develops about the seventh week of gestation (20 mm embryo stage) (7-9). Corneal development begins at the 18 mm to 20 mm embryo stage, and corneal collagen appears at the 20 mm stage. The limbus becomes evident at the 28 mm embryo stage. Scleral collagen fibers in the superficial layers are thicker, probably because they are pushed out by the newer fibers forming in the deeper layers. In sclerocornea the collagen fibers are arranged like scleral fibers, and there is no Bowman's membrane, suggesting that the stimulus to scleralization originates in the corneal stroma. Descemet's membrane is approximately one micron, the thickness present at a seven-month gestation and about 1/10 normal thickness of the adult cornea. The endothelium is often discontinuous or absent. The few endothelial cells present have high density cytoplasm and produce the abnormally thin Descemet's membrane peripherally. The pathologic examination of the sclerocornea has shown elastic fibers in the anterior stroma, a decrease in the size of collagen fibers from the anterior to the posterior layers of cornea, and failure of Descemet's membrane with immature endothelium (10,11). The condition may be related to incomplete cleavage of the anterior chamber angle and therefore may be associated with posterior embryotoxon (Axenfeld's syndrome), Peter's anomaly, Rieger's syndrome, and congenital glaucoma.

According to Bloch (12), total sclerocornea in its severe form is inherited recessively, yet the peripheral type of sclerocornea results from a dominant gene. Both sexes are equally affected, and at least 50% of the cases are sporadic. Aside from the ocular anomalies related to imperfect angle cleavage and glaucoma, cornea plana and cataracts have been noted, along with coloboma of the choroid and retina. Associated extraocular anomalies include defects in the skin, face and central nervous system (13).

The differential diagnosis of sclerocornea includes inflammatory corneal scarring (eg, interstitial keratitis), mucopolysaccharidosis (14), and congenital hereditary endothelial corneal dystrophy (15), along with Peter's anomaly and Rieger's anomaly.

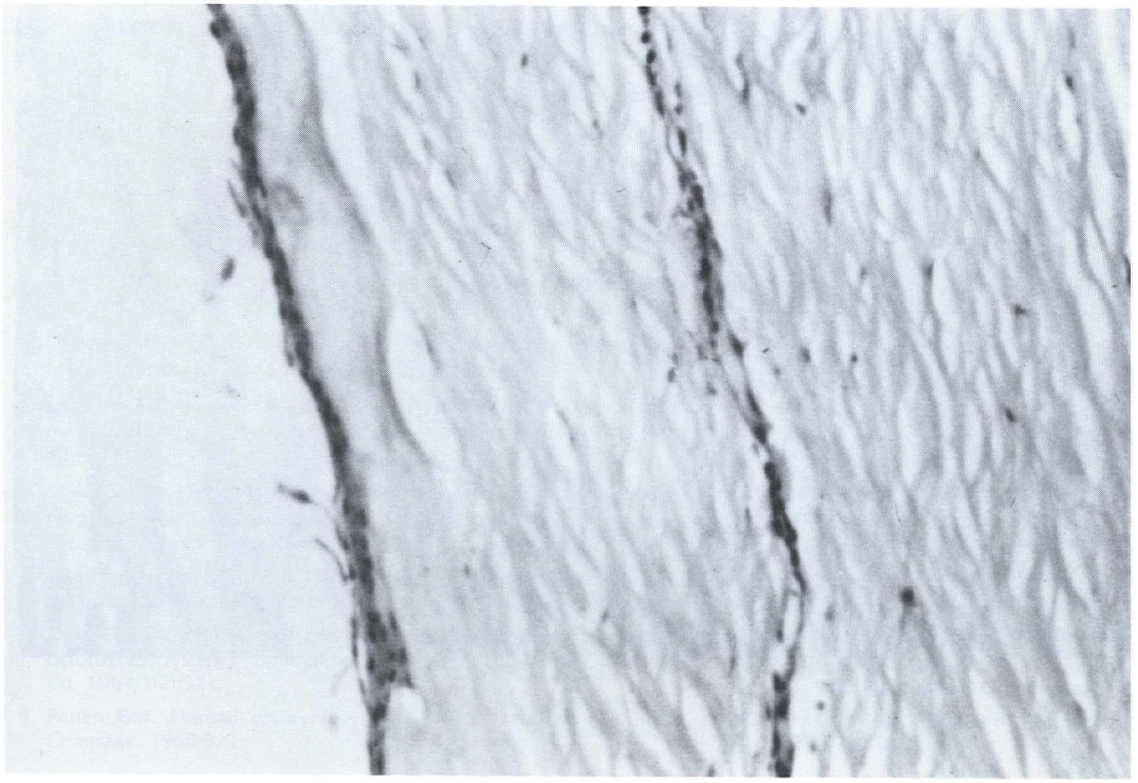

Fig 3
Photomicrograph of sclerocornea illustrates atrophic epithelium with subepithelial hyaline deposits (PAS x 100).

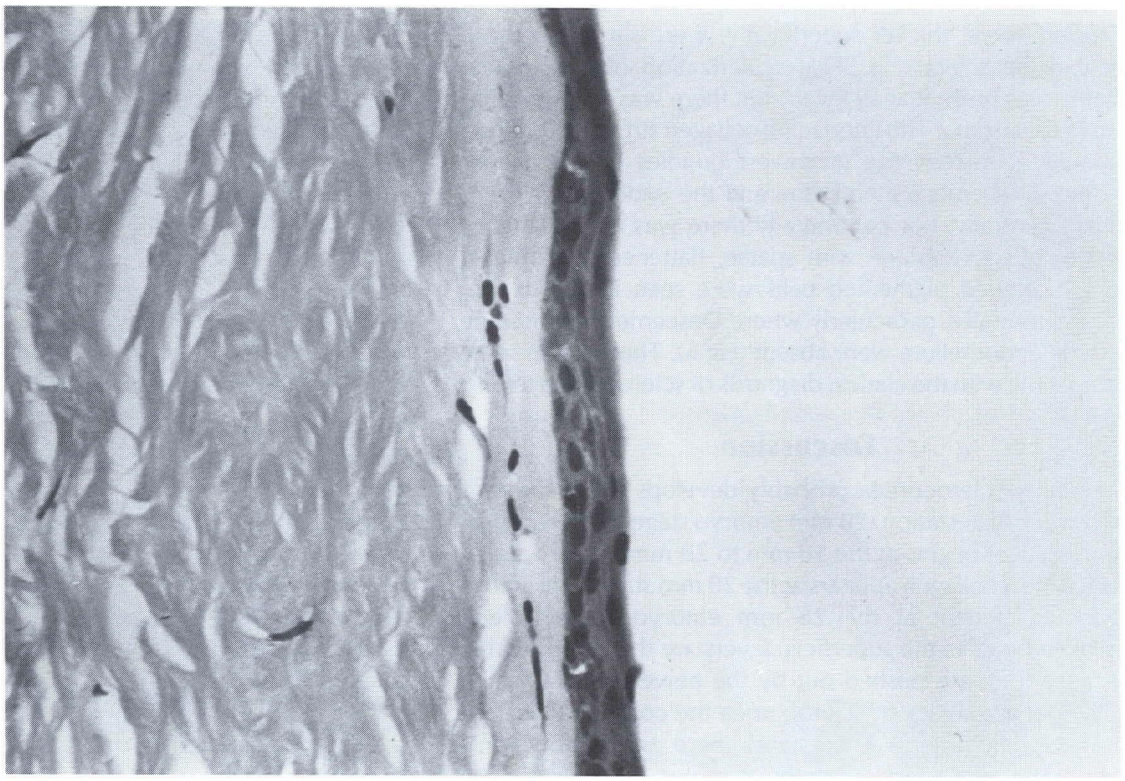

Fig 4
Photomicrograph of sclerocornea illustrates absence of Bowman's membrane (PAS x 200).

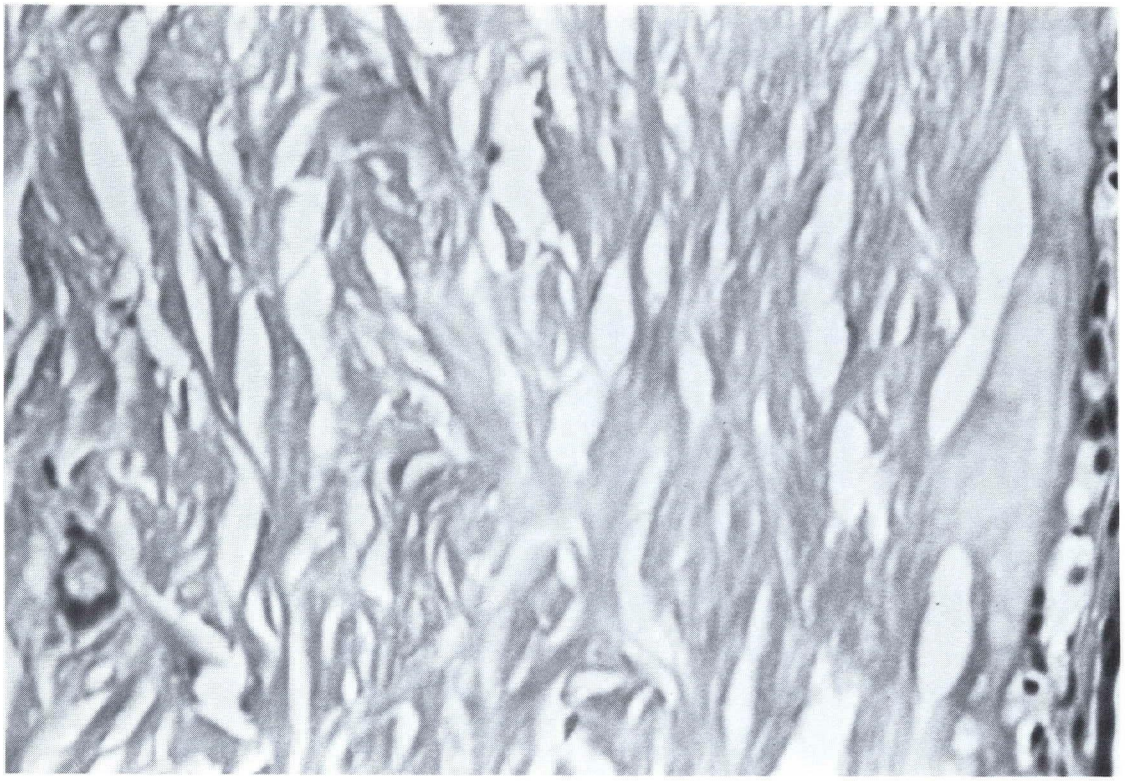

Fig 5
Photomicrograph of sclerocornea. Note irregular arrangement of stromal collagen fibers, thicker superficially, and thinner in deeper layers (PAS x 100).

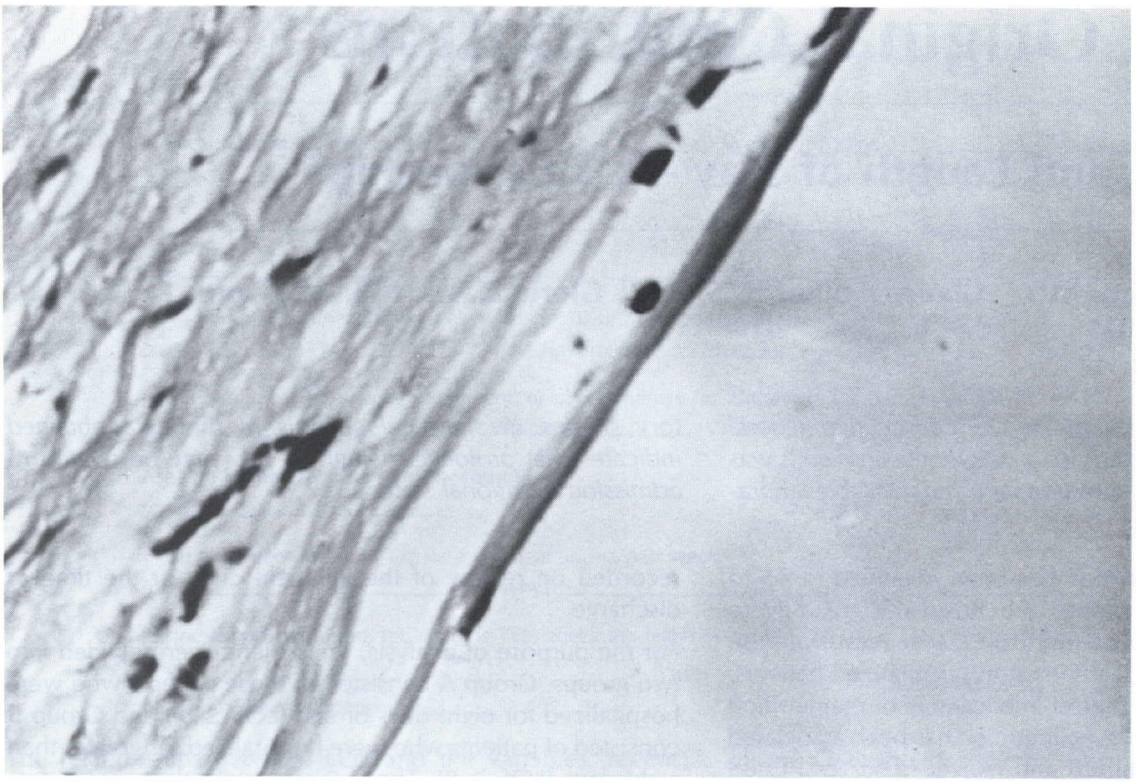

Fig 6
Photomicrograph of sclerocornea. Pigmented cells in deeper stroma centrally where endothelium and Descemet's membrane are absent (PAS x 200).

Treatment by corneal transplantation must be performed early to avoid suppression amblyopia due to the opaque cornea (16,17). The graft sutures should be removed early postoperatively because infantile corneas heal quickly, and it is important to prevent suture abscesses and vascularization of the graft. The prognosis for recovery of visual function is poor (18-20). Perhaps, as in the case of congenital cataract, early surgery will offer a better chance for the patient to develop useful vision and to avoid the handicap of near blindness.

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