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SCLEROCORNEA*
(in 3 siblings)

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Sclerocornea is an uncommon anomaly characterized by scleralization of the peripheral, central, or entire cornea[1]. The opacities are present at birth and mostly bilateral. It has been described most frequently in conjunction with cornea plana and various other anomalies[2,3] but may also occur as an isolated congenital anomaly[4]. At present approximately 50 cases with 50% dominant or recessive inheritance have been reported[4,5].

In this report the ocular findings in three siblings with sclerocornea are described.

Case Reports

Case 1. Z.K., a 12-year-old girl, was seen at the Hacettepe Children's Hospital because of loss of vision due to a cloudy cornea. She was the second born of four affected siblings (Figure 1) whose parents were first cousins****. Only their fifth child, a 3 1/2 year old boy, had no corneal opacities. Z.K. was the product of an uneventful pregnancy and delivery. Corneal opacity and enlargement of the cornea had been noticed at birth, and she had had an unsuccessful operation, presumably for glaucoma, at the age of five. Her growth and development were unsatisfactory.

Physical examination revealed that height and weight were below the third percentile. The rest of the physical findings were unremarkable.

Ophthalmic examination revealed bilateral hydrophthalmia secondary to glaucoma, more pronounced on the right side, and staphyloma anteriorly (Figure 2).

Laboratory tests including CBC, urinalysis, NPN, total lipids and cholesterol, and total protein were normal.

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**** The first child, a boy, who had the same type of eye anomaly, had died at the age of one.

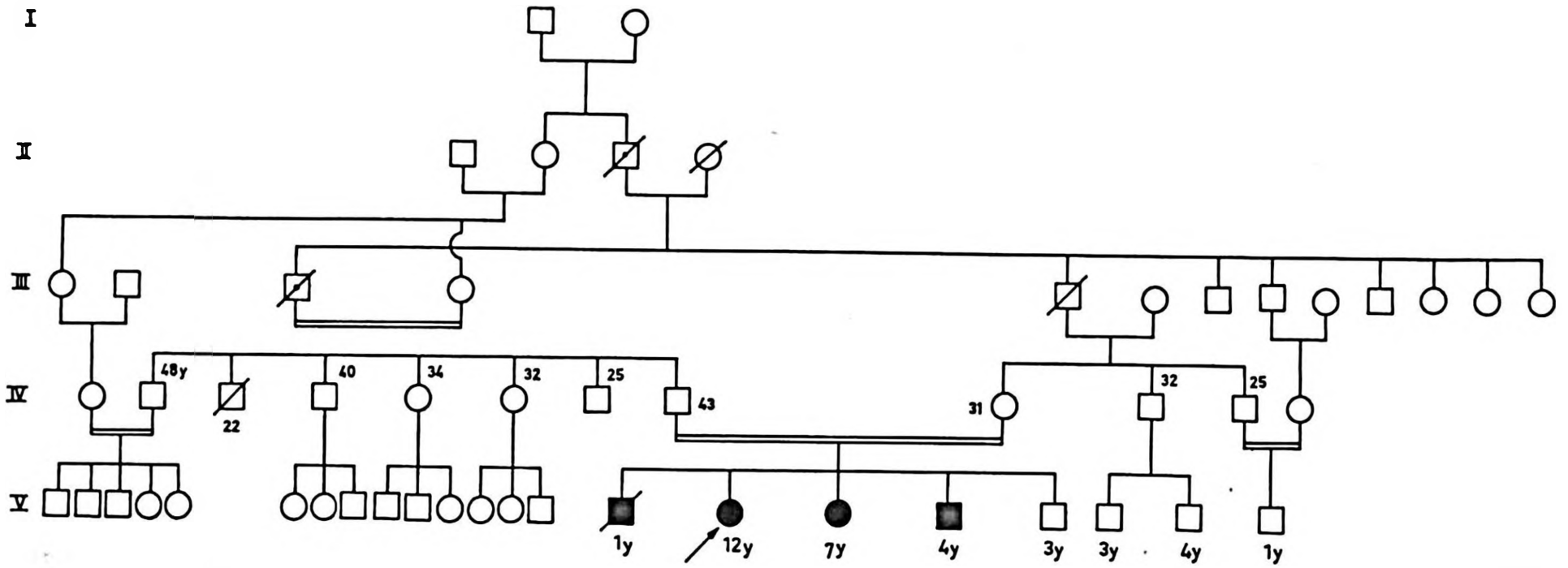

Figure 1.

Five generation pedigree pattern of the sclerocornea patients.
Family Legend: squares = males, circular = females, solid symbols = affected persons, open symbols = unaffected persons.

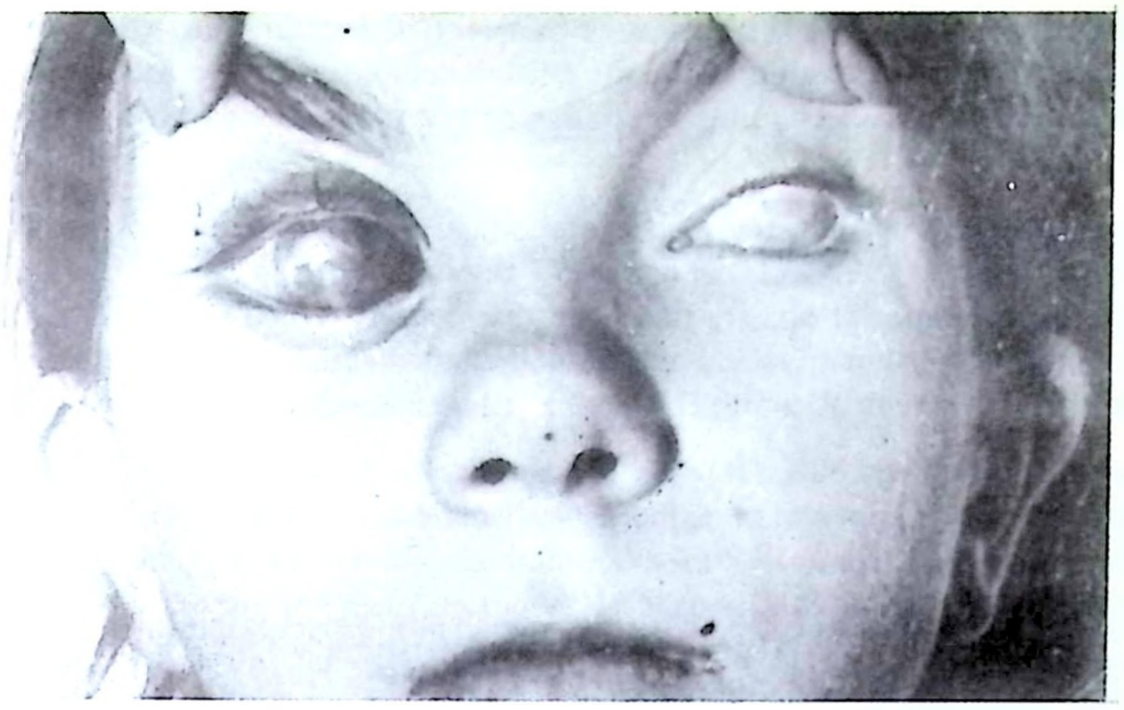

Figure 2.

Case 1. 12-year-old girl with unsuccessful operation for glaucoma on the right eye and densely opaque cornea on the left eye.

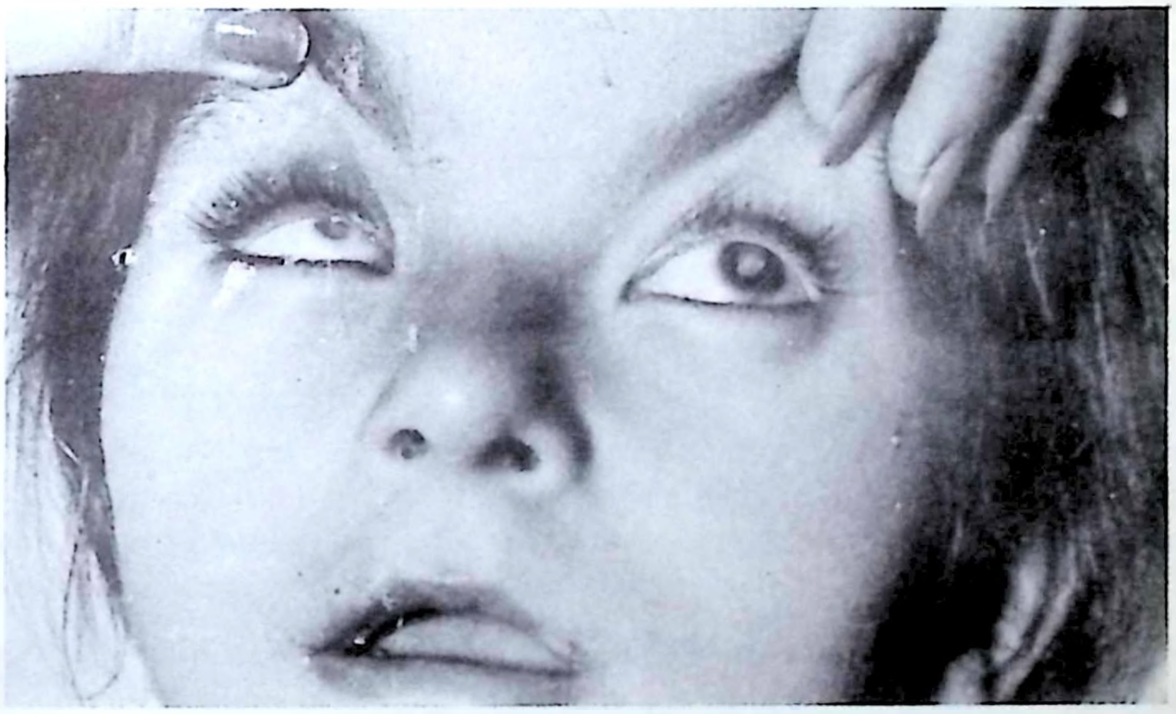

Figure 3.

Case 2. 7-year-old girl with microcornea on the right eye and retrocorneal opacity at the center of the cornea of the left eye, associated with sclerocornea.

Case 2. G.K., the 7-year-old sister of Case 1, was admitted to the Hacettepe Children's Hospital for an operation to correct earlier surgery performed elsewhere. She had had total opacity of the right eye and central opacity of the left since birth (Figure 3).

Ophthalmic examination revealed microcornea and total opacity of the cornea of the right eye, and retrocorneal opacity at the center of the cornea of the left eye. Left-side retrocorneal opacity had been operated on twice with inferior nasal iridectomy. The operations had not been successful in improving visual acuity, which remained only finger counting at 50 cm. She had bilateral horizontal nystagmus.

Laboratory findings including CBC, urinalysis, NPN, total lipids and cholesterol, total protein, calcium and phosphorus and amino acids in the urine and blood were all within normal limits.

Case 3. S.K., a 4-year-old boy, was examined at the Hacettepe Children's Hospital with the chief complaint of loss of vision since birth. He was the fourth child and fourth affected sibling.

His eye examination revealed microphthalmic and total scleralization of the cornea of the right eye, and iris colobomata at the superior portion of the left pupil with density due to a cataract at the inferior portion of the pupil (Figure 4).

Figure 4.

Case 3. 4-year-old boy with total sclerocornea on the right eye and iris colobomata at the superior portion of the left pupil with density due to cataract at the inferior portion.

Laboratory findings including CBC, urinalysis, total lipids and cholesterol were all within normal limits.

Discussion

Sclerocornea totalis which presents in a severe form is recessive, and the peripheral type is dominant according to Bloch[1]. Both sexes are affected in equal numbers, and 50% of the cases are sporadic[5].

Extraocular defects described with sclerocornea include abnormalities of the central nervous system, skin, face, ears and testes[2].

The various ocular abnormalities have been documented with some reservations in previous publications. Blepharoptosis, flattened cornea, shallow anterior chamber, anterior synechia, hypoplastic iris, cataractous lens, elevated intraocular pressure and fundus changes are the main anomalies associated with sclerocornea[5]. Sclerocornea is probably the result of maldevelopment of the limbal mesodermal tissue during the seventh week of gestation when an abnormality develops in the paraaxial mesoderm, anterior to the optic cup[6,7].

Results of light and electron microscopy revealed increased and variable diameters of collagen fibers with elastic fibers in the anterior stroma, a decrease in the magnitude of collagen fibers from the anterior to the posterior layers, and an underdevelopment of Descemet's membrane with abnormal endothelium[8].

The differential diagnosis of hereditary sclerocornea must consider genetic disease such as mucopolysaccharidosis[9] or congenital stationary corneal dystrophy[10]. There is also similarity between some of the corneal findings in sclerocornea, Peter's anomaly, and Rieger's anomaly[1]. These conditions may represent variations of the same basic defect. Common findings include blending of the cornea and sclera, anterior synechia, posterior embryotoxin, prominent iris processes in the iridocorneal angle, and congenital glaucoma. Differences include mode of inheritance, site of corneal involvement, extent of iris abnormality, and possibly associated systemic anomalies[5].

Surgery, such as penetrating corneal grafts, in patients with bilateral sclerocornea, is advised especially at an early age in an effort to prevent irreversible amblyopia[11]. The prognosis of useful vision after surgery is often guarded because of the known association of sclerocornea with shallow anterior chamber, glaucoma, high refractory errors and colobomas of the choroid and retina[11].

The first patient described in this paper represents an extreme complication of sclerocornea, bilateral glaucoma at an early age, with an unsuccessful operation at the age of five. The second patient showed total opacity of the right eye, and surgery on the left eye, which had only central opacity, had been performed twice without success at the age of six.

These three siblings present severe cases of sclerocornea with autosomal recessive inheritance. Surgical approaches for better vision were unsuccessful because of the additional underlying severe ocular congenital anomalies.

Summary

In this report autosomal recessively inherited sclerocornea in three siblings with additional severe ocular anomalies is described, and the surgery attempted on two of the patients is reported.

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