SJÖGREN-LARSSON SYNDROME*†

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SJögren and Larsson (1957) described 28 patients with spastic diplegia, low grade oligophrenia, and ichthyosiform erythrodermia. Three of the patients had ophthalmological abnormalities consisting of patches of atrophy of the pigment epithelium of the macular area, and two had, in addition, increased macular pigmentation. Wallis and Kalushiner (1960) and Heijer and Reed (1965) have reported other patients with the SJögren-Larsson syndrome and macular lesions, but so far there have been no reports in the literature of corneal changes in this rare condition.

The patient presented here had a severe spastic diplegia (Fig. 1) and ichthyosiform erythrodermia, a variety of autosomal recessive ichthyosis (Wells, 1966). He was mentally...
very retarded, could understand only simple instructions and was unable to indicate ideas beyond signifying assent or denial. The corneal complications were dramatic and demanded repeated emergency surgery.

Case Report

He was born on June 11, 1944, to working-class parents in whom there was no history of consanguinity or any particular family disorder.

He was seen at the age of 13 years at an Eye Infirmary and attended repeatedly with corneal ulceration, affecting the right eye more than the left. He was admitted to hospital at the age of 16 years with severe corneal ulceration; the visual acuity after treatment, mainly with topical steroids, was 6/18, and 3 months later he had developed a descemetocoele of the right cornea, while the left cornea showed a thinned central area.

He was admitted to hospital for investigation and keratoplasty, and was seen by Dr. M. E. R. Stoneman for a paediatric opinion. She made the provisional diagnosis of Sjögren-Larsson syndrome and thought that he was fit for surgery. At that time the visual acuity in the right eye was 6/60 (E test). Slit-lamp examination showed the right cornea to be deeply vascularized from above and below. The corneal epithelium was clear except in its central region and the stroma appeared normal. The central area was thinner with an intact epithelium and obvious descemetocoele formation (Fig. 2). The left cornea showed a thinned central area with a scar occupying all layers, but it was not bulging (Fig. 3). On ophthalmoscopic examination at a later date, when the cornea had been grafted, the right fundus appeared normal. The details of the left fundus have never been seen clearly.

Operation.—A 6.0 and 6.1 mm. penetrating keratoplasty was performed on the right eye on July 5, 1961, and convalescence was uneventful. On discharge the visual acuity was 6/24 (E test) (Fig. 4).

The non-ulcerative thinning process continued in the left cornea for the next 8 months and a descemetocoele developed. The left cornea perforated and a 5.0 and 5.1 mm. penetrating keratoplasty was performed with iridectomy to free the anterior synechiae. The eye settled and he was discharged 4 weeks later.
Progress.—The right eye remained quiet for about 2 years until October 1, 1963, when he was reported to have a recurrence of ocular irritability with extreme thinning of the centres of both grafts. He was re-admitted and was found to have central thinning of the right graft. A similar, though more advanced, change had occurred both at the centre of the graft and near the graft edge at 9 o’clock in the left eye. A left descemetocoele developed and ruptured 4 days later, and this required another emergency penetrating 5-0 and 5·1 mm. keratoplasty instead of the cold repair as planned. The graft failed to heal firmly and became slightly ectatic, requiring re-suturing and reinforcement with an overlay of further donor corneal tissue. This last graft settled into place and is still reasonably healthy 3 years later.

In the meantime, the right thinned cornea was unchanged, enabling the repair to be carried out as planned. A 6·0 and 6·1 mm. penetrating keratoplasty was performed on November 29, 1963. The eye settled uneventfully and the visual acuity is now 6/24 with correction.

Result.—The slit lamp shows that the right graft is clear in all its layers with good apposition at the host-graft junction and no vascularization.

The left graft shows oedema of the epithelium with some bullous formation. The stroma is opalescent but avascular, and through it features of the anterior chamber can be seen. The visual acuity is hand movements.

The series of recurrent erosions of both host and donor corneal tissue was noteworthy and no obvious cause for it was seen. The suggestion that there was a mechanical factor due to the patient rubbing his eyes in a compulsive way was not observed in this case, neither was there any infection nor relative avitaminosis.

Pathology.—The following reports were received from Prof. Norman Ashton.

1961 (right graft) and 1962 (left graft)
The corneal epithelium is keratotic and very irregular with areas of hyperplasia and attenuation. Bowman’s membrane is partially destroyed and thrown into folds, while there is scarring and vascularization of the superficial substrata. The central area of one specimen (right graft) is occupied by a descemetocoele which is covered by epithelium; in the other specimen (left graft) the central area has been disorganized in preparation but here also the appearances suggest a descemetocoele.

1962 (left re-graft)
Centrally the cornea is thinned and scarred with deep proliferation of epithelium and Bowman’s membrane; Descemet’s membrane is absent over the scar. There is no evidence of a descemetocoele.

1963 (right re-graft)
Section shows a disc of corneal tissue which appears to consist of either an extremely atrophic cornea or of the superficial layers only. The corneal epithelium itself is extremely attenuated, Bowman’s membrane is intact centrally but is lost at the scars of the old graft. Descemet’s membrane is not seen and the inner layers of the disc are fragmented. There is no evidence of a descemetocoele.

Prof. Ashton reviewed the slides of the specimens with the full history and established the diagnosis.

Discussion

The appearances in this case are those of a non-specific keratitis with marked keratosis and superficial stromal scarring and vascularization. This is consistent with the keratitis associated with ichthyosis described by Cordes and Hogan (1939), and presumably is secondary to the associated chronic blepharo-conjunctivitis. From the description their patient may have had ichthyosiform erythrodermia which is seldom associated with spasticity or oligophrenia. It is possible that the descemetocoeles are caused by rubbing the eyes over a long period of time, a parallel being found, perhaps, in the keratoconus associated with vernal conjunctivitis.
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Summary

A patient with the Sjögren-Larsson syndrome is described in whom recurrent corneal ulceration necessitated multiple corneal grafts in order to save his sight.

I am indebted to Mr. T. Stewart-Black Kelly, F.R.C.S., for referring the case, and to Prof. N. Ashton for reporting in detail on the fragments of cornea removed at keratoplasty and for providing the conclusion; Dr. R. S. Wells encouraged me to report this case, having read the clinical notes. Mr. V. Herbert of Weymouth kindly processed the photographs.

REFERENCES

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