Corectopia and lipid proteinosis

LENWORTH N JOHNSON¹ AND ROBERT S HEPLER²

From the ¹Neuro-Ophthalmology Unit, the Milton S Hershey Medical Center, Pennsylvania State University, Hershey, Pennsylvania, and ²Jules Stein Eye Institute, University of California at Los Angeles, Los Angeles, California, USA

SUMMARY Lipoid proteinosis (Urbach-Wiethe disease) is a rare autosomal recessive disorder associated with deposition of hyalinised material in the skin, mucous membrane, and brain. Corectopia has not been described in this disorder. A case is presented of lipoid proteinosis with bilateral corectopia.

Lipoid proteinosis (Urbach-Wiethe disease) is a rare autosomal recessive genetic disorder characterised by hoarseness beginning in infancy and multiple lesions of mucous membrane and skin.¹ The skin lesions are composed of hyalinised material and appear as clusters of white to yellow papules on the elbows, knees, axillae, perineum, and scrotum. Persons with lipoid proteinosis can present with psychomotor seizures. In some cases intracranial calcification of the hyalinised material has been documented within the hippocampal gyri.² Ocular manifestations include papules along the lid margins and retinal drusen. To our knowledge pupillary abnormalities have never been reported in this disorder. We report a case of lipoid proteinosis with bilateral corectopia.

Case report

A 32-year-old man was seen for complaints of bilateral monocular diplopia of two months' duration. The patient had had a persistent hoarse cry from infancy. Psychomotor seizures began at the age of 16 years, and secondary generalised tonic-clonic seizures began at 21. The seizures were successfully controlled with carbamazepine (Tegretol). The patient had several episodes of rage reaction in which he would destroy objects valued by himself and his loved ones. There was no past history of meningoencephalitis, hydrocephalus, stroke, or injury to the head. The patient was born of a full-term uncomplicated pregnancy and delivery. Forceps were not used in the delivery. His parents are first cousins and

Correspondence to Lenworth N Johnson, MD, Neuro-Ophthalmology Unit, Milton S Hershey Medical Center, Pennsylvania State University, Hershey, Pennsylvania 17033, USA.

are in good health. There are no known ocular abnormalities in members of his family, nor are other relatives known to have lipoid proteinosis.

The general physical examination revealed confluent waxy papules on the eyelid margins and face (Figs 1, 2), elbows, knees, and body. The voice was hoarse. The neurological examination gave normal results. Best corrected visual acuity was 20/30 in each eye. Monocular diplopia disappeared when tested with a pinhole. Quantitative automated perimetry was normal. The corneas and gonioscopic examination of the anterior chamber angles were normal.

Fig. 1 Typical confluent waxy papules on the eyelid in lipoid proteinosis. Corectopia with displacement of the pupil superiorly and nasally is present.

Fig. 2 Typical confluent waxy papules noted on the face and eyelids in lipoid proteinosis. Corectopia is noted.

Bilateral nuclear lens opacifications were noted. There was no evidence of posterior synechiae or lens dislocation. The pupils were oval and displaced upwards and nasally (Fig. 1). (Similar corectopia was recorded at 9 months old). The irides were otherwise normal. Pupillary reactions were brisk to light and near stimulation. There was no iris sector palsy or atrophy. No afferent pupillary defect was present. Ocular motility and funduscopy examination were normal. Retinal drusen were not apparent on ophthalmoscopic examination.

A computed tomographic scan, magnetic resonance imaging, and cerebrospinal fluid analysis were normal. Electroencephalography showed bitemporal seizure foci. Skin biopsy had findings characteristic of lipoid proteinosis (Figs 3, 4).

Fig. 3 Photomicrograph of verrucous lipoid proteinosis lesion. The hyaline material deposited in the dermis consists of homogeneous bundles staining pale pink with haematoxylin and eosin. $\times 17$.

Discussion

Our patient's clinical history and the findings from the physical examination were suggestive of lipoid proteinosis, with skin biopsy confirming the diagnosis. Histological studies of the dermis in lipoid proteinosis usually show diffuse deposits of amorphous hyalinised material composed in part of neutral mucopolysaccharides³ round blood vessels.

Fig. 4 'Onion skin' appearance round blood vessels. (Haematoxylin and eosin, $\times 100$.)

There may be an associated increased number of fibroblasts in the dermis. Lipid deposits are not a notable feature of this disease.

Corectopia had been present in our patient at least since the age of 9-months. It has been shown pathologically to occur in patients with significant rostral midbrain injury, possibly due to partial destruction of the parasympathetic pupillary fibres.^{4,5} Our patient did not have a history of severe injury to the head. In addition the seizure disorder began several years after the corectopia was noted. However, hyalinisation of the hippocampal gyri of the temporal lobe has been observed histopathologically in cases of lipoid proteinosis with temporal lobe seizure disorder.² Indeed, our patient had bitemporal seizure foci. Although the corectopia may be a consequence of consanguinity, it is conceivable that the hyalinised material noted on skin biopsy was also present in the rostral midbrain, accounting for the

corectopia. Although to our knowledge this is the first reported case of pupillary abnormality associated with lipoid proteinosis, this disorder should be added to the differential diagnosis of corectopia.

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