Principles and Practice of Clinical Electrophysiology of Vision

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Sorsby's Fundus Dystrophy

Thomas A. Berninger

In 1949 Sorsby et al.[9] described a progressive fundus dystrophy with three key features: autosomal dominant inheritance, loss of central vision in the fifth decade, and loss of ambulatory vision in the seventh decade. The authors described "a central retinal lesion showing edema, hemorrhage and exudates developing into generalized choroidal atrophy with massive pigment proliferation." The autosomal inheritance was shown in four pedigrees: the C., E., K., and R. families. Two sisters from a further family were included in the first report. But there was no proof of dominant inheritance. The large hemorrhagic exudative lesion gave rise to the term "Sorsby's pseudoinflammatory macular dystrophy."[4] This term, however, is misleading for two reasons. The macular lesion is due to subretinal neovascularization with a subsequent disciform reaction.[3, 6] Furthermore, the extramacular fundus is frequently involved in addition to the macular lesion. Therefore the disorder has been termed *Sorsby's fundus dystrophy* (SFD).[3] In recent years three of the original four families have been examined in more detail. Although the three key features were present in all three families, significant interfamilial variations were found, which suggests that SFD is more than one disorder.

FUNDUS

Prior to loss of vision the first fundus changes are fine drusenlike deposits at the level of Bruch's membrane, angoid streaks, and plaquelike deposits of yellow subretinal material in the macular region.[3, 6, 8] In advanced cases two reactions of the macula have been described: atrophy and subretinal vascularization. Subretinal vascularization occurred in several families, being universal in E.[6] and common in K.,[3] although atrophy was seen in some family members. By contrast, atrophy was the predominant response in the C. family.[8] Common to all families was peripheral retinal atrophy in the final stage of the disease.

RETINAL FUNCTION

Visual Acuity

The loss of central vision occurred at a similar age, but the progression of loss varied. In the E. family[6] the loss of central vision was sudden and profound, in the K. family[3] the deterioration usually occurred over months, and in the C. family[8] reading vision was often retained until patients were in the seventh decade of life.

Color Vision

Color vision has been frequently examined. However, the results were inconsistent. A mild deuteranomaly was observed in some members of the R. family,[5] no color defects were found in the E. family,[6] while a tritan defect was found in the K. family.[3] With a new color test a tritan defect was observed in 16 of 34 patients with 50% risk to develop SFD.[1]

Other Symptoms

Prior to the loss of central vision all affected members of the C. family reported difficulties in adapting to changes in ambient light.[8] By contrast, progressive difficulties with night vision for up to 30 years before the loss of visual acuity is reported for the K.

family,[3] while the E. family was asymptomatic before the loss of central vision.[6]

ELECTROPHYSIOLOGY

There are no complete reports about electrophysiological tests. Normal electro-oculographic (EOG), electroretinographic (ERG), and macular ERG values were found in the E. family,[6] while in the K. family[3] a reduced light rise was observed in the EOG.

HISTOLOGY

There has been up to now only one histological report from a member of a family with proven SFD. The dominant histopathological feature in this patient was the confluent, lipid-containing, amorphous deposit found between the basement membrane of the retinal pigment epithelium (RPE) and the inner collagenous layer of Bruch's membrane. The deposit is much thicker (30 μm) and more widespread than in age-related changes. It is weakly eosinophilic, but lipid-positive.[2]

PATHOGENESIS

There is so far no known pathogenesis. An explanation may be found in the early changes of the disease. In the C. family abnormalities in the choriocapillaris precede accumulation of the yellow material. If the primary abnormality arises in the choriocapillaris, a second accumulation of phagosomal debris in Bruch's membrane can be expected. Alternatively, the yellow material may be the primary response, followed by atrophy of the choriocapillaris as a secondary event. The integrity of choriocapillaris itself is dependent on the RPE.[7]

DIFFERENTIAL DIAGNOSIS

The differential diagnosis of SFD should include all retinal and choroidal dystrophies. However, there is no other dystrophy with really marked similarities to SFD.

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REFERENCES

1. Berninger TA, Polkinghorne PJ, Capon MRC, Gündüz K, Arden GB, Bird AC: Color vision defect: An early sign in Sorsby's fundus dystrophy. *Invest Ophthalmol Vis Sci* 1988; 29(suppl):3.
2. Capon MRC, Marshall J, Krafft JI, Bird AC: Sorsby's fundus dystrophy: A light and electron microscopic study. *Ophthalmology* 1989; 96:1769–1777.
3. Capon MRC, Polkinghorne PJ, Fitzke FW, Bird AC: Sorsby's pseudoinflammatory macula dystrophy—Sorsby's fundus dystrophies. *Eye* 1988; 2:114–122.
4. Duke-Elder S, Perkins ES: Diseases of the uveal tract, in *System of Ophthalmology*, vol 9. London, Kimpton, 1966, pp 715–719.
5. Fraser HB, Wallace DC: Sorsby's familial pseudoinflammatory macular dystrophy. *Am J Ophthalmol* 1971; 71:1216–1220.
6. Hoskin A, Sehmi K, Bird AC: Sorsby's pseudoinflammatory macular dystrophy. *Br J Ophthalmol* 1981; 65:859–865.
7. Korte GE, Reppucci V, Henkind P: RPE destruction causes choriocapillary atrophy. *Invest Ophthalmol* 1984; 25:1135–1145.
8. Polkinghorne PJ, Capon MR, Berninger TA, Lyness AL, Sehmi K, Bird AC: Sorsby's fundus dystrophy: A clinical study. *Ophthalmology* 1989; 96:1763–1768.
9. Sorsby A, Mason MEJ, Gardener N: A fundus dystrophy with unusual features. *Br J Ophthalmol* 1949; 33:67–97.