Electroretinographic findings in macular dystrophy

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Abstract. The flash and pattern electroretinogram were investigated in a group of families with rare forms of inherited macular dystrophy, which included Sorsby’s fundus dystrophy, X-linked retinoschisis and macular dystrophy of uncertain classification and variable expression. Flash electroretinograms, under both photopic and scotopic conditions, were attenuated in both Sorsby’s fundus dystrophy and X-linked retinoschisis—with some effect on implicit time being noted in the latter condition—but in the unknown group the effect was less demonstrable, only 50% having attenuated flash electroretinograms. Pattern electroretinograms were reduced in all three conditions and in almost all cases. The study demonstrates that some so-called macular dystrophies also have widespread abnormalities affecting the peripheral retina. These findings may contribute to a better understanding of the underlying pathophysiologic mechanisms in these rare forms of retinal dysfunction.

Abbreviations: SFD—Sorsby’s fundus dystrophy; XLJR—X-linked juvenile retinoschisis.

Introduction

Retinal dystrophies have traditionally been classified as either rod (retinitis pigmentosa) or cone (central or macular) dystrophies. Clinical classification has been based on inheritance, natural course and fundus appearance.

This classification is inadequate, as both rod and cone function are affected, even in the early stages, in many progressive retinal dystrophies. Classification is increasingly based on the identification of the underlying genetic abnormality, but this is not always easy to correlate with the clinical and electrophysiologic findings. Clinically indistinguishable eye diseases can be caused by more than one genetic abnormality, and one genetic abnormality may give rise to different eye diseases in different individuals within a family. For example, mutations in the rds gene can give rise to retinitis pigmentosa or macular dystrophy [1–3].

The gene for one macular dystrophy (Sorsby’s fundus dystrophy [SFD]) has been identified and codes for TIMP-3, an inhibitor of metalloproteinase
involved in the remodeling of extracellular matrix. Little is known about the electrophysiologic signals in the condition, apart from two studies on two of the families referred to below. One family did not show any effect on the electroretinogram (ERG) or electro-oculogram [4], while another family demonstrated a reduced electro-oculogram light rise [5]. Jacobsen et al. [6], in a study of the effect of vitamin A on night blindness in SFD, noted rod ERG abnormalities in six of seven members of one family, and one of two patients tested after treatment showed improvement in the maximum b-wave amplitude.

X-linked juvenile retinoschisis (XLJR) was first described by Haas in 1898 [7]. The basic defect in XLJR is thought to be in the Muller cell [8,9], as there is a characteristic reduction in ERG b-waves and oscillatory potentials in all affected patients [10]. Older patients may also show reduction in the a-wave. Recently, Murayama et al. [11] have demonstrated that the ERG scotopic threshold response is more affected than either the b-wave or oscillatory potentials. The ERGs of obligate carriers are normal.

The pattern ERG (PERG) is a retinal potential that conveys information on both the middle/outer receptoral layers of the retina as well as the proximal (ganglion cell) layer, their contribution being distinguished, it is suggested, in the P50 and later N95 components, respectively [12]. The response is known to be strongly dominated by that of the central retina [13] and is affected in various conditions of the macula. Its value in studying disease of the central retina has developed over the last 10 years or so, although it was first reported to be affected in a macular condition more than 20 years ago [14]. To our knowledge it has never been studied in SFD and XLJR.

This report describes our investigations into patients with SFD and XLJR using flash ERGs and PERGs and in an additional family with an unknown macular dystrophy that caused a clinical picture similar to retinitis pigmentosa in some family members.

**Subjects and methods**

**Patients**

All patients, other than three with X-linked retinoschisis referred from other ophthalmologists, were examined by the same ophthalmologist (M.P.C.). A full clinical ocular examination was performed, including slit-lamp examination and fundus biomicroscopy, supplemented with fluorescein angiography where appropriate.

Five of the investigated individuals had SFD (Table 1). Two individuals were from the Ewbank family, one of the original families described by Sorsby