Correlation of ERG and pigment epithelium changes in external progressive ophthalmoplegia (EPO)

P. STEINDLER, A.P. TORMENE, G.F. MICAGLIO* and A. GALAN

Department of Ophthalmology and Neurology*, University of Padova, School of Medicine, Padova, Italy

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Abstract. Six out of 17 patients with progressive external ophthalmoplegia (EPO) were found to have pigment anomalies with alterations in the electroretinographical (ERG) tracings. However, fluorangiography demonstrated alterations of the retinal pigment epithelium in patients with normal fundus and ERG examinations. We conclude that in our series there was no correlation between retinopathy and tapetoretinal degeneration.

Introduction

Progressive external ophthalmoplegia (EPO), the progressive symmetrical weakening of the external eye muscles leading to their total paralysis, can be found in subjects of all ages, but is more frequent in 10- to 20-year-olds. In the majority of cases the elevator muscle of the upper lid is first affected.

This disease has been studied by several authors; the myopathy can spread and affect the pharyngeal (Victor, 1962) or the skeletal (Kiloh and Nevin, 1951) muscles. In some cases EPO may be associated with organic diseases affecting the heart (Kearns and Sayre, 1958; Daniele and Corea, 1981) or endocrinal glands (Pellock et al., 1978; Pierson, 1979; Lundberg, 1962), and can be associated with peripheral neuropathy (Drachman, 1968); it has also been involved in various biochemical disorders (Di Mauro, 1973).

An association between EPO and retinitis pigmentosa was reported by Croft et al. (1977) in about 11% of his cases of EPO; additionally, he found retinitis pigmentosa in 13 cases among the 348 reported in the literature.

The numerous associations found between EPO and other diseases on the one hand attest to the interest in EPO but on the other have prompted authors to describe numerous 'new syndromes'. The descriptions, based on clinical and laboratory findings, make it difficult to work out a nosologic picture of EPO, because conditions reported as new syndromes by some authors are considered new variants by others.

Various classifications have been proposed such as those of Drachman (1968), Glaser (1978), and Bastiaensen et al. (1982), the last following on the
new etiopathogenetic causes of EPO based on mitochondrial alterations and
the classification used in this paper.

**External Progressive Ophthalmoplegia (EPO)**

1. Mitochondrial EPO
   = Ocular myopathy and descending ocular myopathy
   = Kearns disease (or ophthalmoplegia plus in a restricted sense)
   Infantile form (‘Kearns–Sayre syndrome’)
   Juvenile form
   Adult form
2. Oculopharyngeal dystrophy
3. EPO in heredoataxias
4. EPO in dystrophic myotonia
5. Ocular myositis

**Materials and methods**

Our series consisted of 17 patients aged from 16 to 68 years (10 females and
7 males) referred to us from the Regional Center of Padua University, which
specialized in epidemiology and the prevention of neuromuscular diseases, in
whom biohumoral, electromyographic, bioptic, electroencephalographic, and
otofunctional examinations were performed.

We studied these patients, paying particular attention to electroretinogram
findings, graphic findings, examination of visual acuity, ophthalmic
visual field, light adaptation, and fluorangiography.

Standard ERG was performed on patients in dark adaptation with
pupillary dilatation obtained by tropicamide 1%. A white 1.5-joule flash was
used as stimulation. After a preadaptation of 2000 lux for 5 min an ERG
during dark adaptation was registered with 3-joule flash and a red filter
Wratten Kodak number 92. The amplitude from electric baseline of B1
(photopic) and B2 (scotopic) waves were noted at 3°, 6° and 15° min of
dark adaptation. We did not perform EOG to study Arden’s index, as the
patients were unable to make the eye movements necessary for recordings.

The findings of the ophthalmologic examinations are summarized in Table
1.

In six of our patients (Cases 5, 7, 10, 11, 13, 14), we found retinal pig-
mamentary changes that were ophthalmoscopically visible, although we observed
no characteristics typical of a tapetoretinal degeneration: the alterations
mainly involved the pigment distribution with areas of pigmentary clumping
that was localized (cases 5, 7, 13) or diffuse (cases 10, 11, 14); pigment
rerrangement ranged from a simple pigment dispersion to accumulation of
different dimensions to hyperpigmentation. No typical bone corpuscles were
observed (Figure 1).