DIFFICULTIES IN DIAGNOSING LEBER'S OPTIC ATROPHY

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Keywords: Leber's optic atrophy, Tobacco-alcohol neuropathy, Quinine intoxication

ABSTRACT

The lowered vision of a patient was attributed to an acute quinidine intoxication, although his condition differed from that of another patient with an acute quinine intoxication. Later, the uncle of the first patient showed lowered vision too, possibly due to a tobacco-alcohol neuropathy. Both uncle and nephew belonged to a family, in which Leber's hereditary optic atrophy occurred.

A 40-year-old patient was referred to the electro-ophthalmological department for the investigation of the lowered visual acuity of his right eye. Vision of this eye was 5/50 with a centrocoecal scotoma of approximately 40°, the other eye had normal visual acuity with a relative centrocoecal scotoma of 10° (Fig. 1). The retinal electrical responses of both the rod system and the cone system were at the lower limit of the normal value, while the occipital potentials to a pattern reversal stimulus of 2 per second with a check size of 1° and 80% contrast were low for the right eye and could hardly be detected for the left eye.

Apart from optic neuritis, tobacco-alcohol neuropathy belonged to the possible diagnoses, since the patient was accustomed to drink alcohol daily and smoked more than 20 cigarettes a day. Some doubt, however, remained because the alcohol intake was not as large as we usually see in these neuropathies.

In the family history there was a nephew, who also had bad vision, which however was attributed to an overdosage of quinidine for an attempted suicide. This patient had been seen at the department some years before. He was 25 years old. Vision of his right eye was 0.01, that of the left eye 0.5. Visual fields showed, apart from centrocoecal scotomas, peripheral field defects also (Fig. 2). Electro-ophthalmological examination revealed very low b-waves of the rod system, while the responses of the cone system combined normal a-waves with very low b-waves. The occipital potentials, both to pattern stimulation and to light flash stimulation, were non-detectable.
Fig. 1. 'Visual acuity, visual fields, scotopic and photopic ERGs, and occipital potentials of a patient, with possible tobacco-alcohol neuropathy. LE: left eye; RE: right eye. The occipital potentials have been evoked with a 2 Hz pattern reversal stimulus. Lower limits of the positive peaks of the scotopic and photopic ERG are 200 μV and 100 μV respectively.

This type of retinal electrical response is typical for a nearly total occlusion of the retinal circulation and is also seen in acute quinine intoxication.