

3.15 VEP follow-up of a mitochondrial optic neuropathy

J. Bastien, F. Rebelo, C. Riger, T. N'guyen, A. Denoyer, C. Arndt

Reims University Hospital, Reims, France

Purpose: In early stages of optic neuropathy, visual acuity, fundus examination and visual field testing may remain unremarkable. Color vision changes and VEP abnormalities may be early signs, especially in toxic optic neuropathy. The purpose of this clinical case presentation is to report the value of VEP in diagnosis and follow-up in a case of paucisymptomatic optic neuropathy.

Methods: A 25 year-old female patient was referred for bilateral optic neuropathy. An initial complete assessment of visual function was performed, including best corrected visual acuity, static automated perimetry, and pattern VEP. Cranial and medullary MRI were normal. There was no evidence for

autoimmune disease or inflammation. Serologies for human immunodeficiency virus, syphilis and Lyme disease were negative.

Results: On presentation, the patient complained of diffuse visual discomfort. Visual acuity was 25/20 in both eyes, the anterior segments were normal, and optic nerve head pallor was observed in both eyes. The OCT RNFL displayed axonal loss. Visual fields were normal and the visual evoked potential revealed axonal dysfunction. VEP the responses to both 60' and to 15' checks were normal. However, no P100 wave could be identified in response to 7' checks. A mitochondrial optic neuropathy was suspected. There was no history and no clinical evidence of alcohol abuse. Genetic testing was performed and the blood levels of common mitochondrial micronutrients (Vitamin B1, B3, B6, and carnitine) were determined. The tests revealed carnitine deficiency and PP vitamin deficiency. Visual symptoms and the responses to 7' checks progressively improved after 6 months and 12 months of supplementation.

Conclusions: Improvement of visual function after supplementation with mitochondrial micronutrients can be monitored with VEPs.