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Full-field Electroretinographic Findings in Children with Neuronal Ceroid Lipofuscinosis

Fábio Ejzenbaum, Adriana Berezovsky, Paula Yuri Sacai, Ana Paula Hamad, Solange Rios Salomão Purpose : Neuronal ceroid lipofuscinosis (NCL) is the most common neuro-degenerative disease in children and young adults. Vision loss is the first sign in the majority of NCL cases, and retinal function deterioration can precede fundus abnormalities in this condition.The purpose of this study was to investigate retinal function by the full-field electroretinogram (ERG) of children with neuronal ceroid lipofuscinosis. Methods: A group of 15 children with NCL were evaluated by standard full-field electroretinography (ISCEV protocol), after 30 minutes of dark adaptation. Patients were sedated with cloral hydrate and had their ERG recorded through a bipolar Burian-Allen contact lens electrode filled with methyl-celullosis eyedrops placed on the corneal surface after local anesthaetic drops. The following responses were obtained in one eye from a fully dilated pupil: scotopic rod response, scotopic maximal response (rods and cones); oscillatory potentials; photopic single-flash cone response and photopic flicker 30 Hz. ERG parameters analyzed were peak-to-peak amplitude (μV) and b-wave implicit time (ms) and compared to normative values to the patient's age. Results : Rod and cone function abnormalities were found in all patients. In 60% of the patients substantially decreased cone response (flicker amplitude ranging from 1.5μV to 35.9μV ; mean=13.75μV ±12,52μV; photopic single-flash cone amplitude ranging from 0 to 49.1μV ; mean=22.43μV ±18.75μV) and moderately decreased rod response (rod b-wave amplitude ranging from 19.2μV to 171.3μV; mean=74.75μV ± 50.27μV; b-wave implicit time ranging from 99ms to 126 ms; mean=110.5ms ± 10.05ms) were found. In with more advanced disease (40%) both cone and rod responses were non-detectable. Conclusions : ERG abnormalities were found in all patients with NCL. In this small NCL case series there was a trend to find initially decreased cone responses, followed by decreased rod response and finally non-detectable ERGs. These results support that ERG is a useful non-invasive clinical tool to confirm NCL when associated with other clinical findings.