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Carbohydrate-deficient glycoprotein syndrome and progression in electrophysiological results

J Fr Ophtalmol 2002, 25, 6, 584-9

PURPOSE: To document the progression of clinical and electrophysiological abnormalities in an infant with carbohydrate-deficient glycoprotein syndrome type Ia (CDGS Ia) over a period of 5 years. **Patient and methods:** A 12-month-old male underwent clinical ophthalmic and electrophysiological examination at the age of 1, 2, and 6 years. Neurological examination, magnetic resonance imaging of the brain and a genetic study were also undertaken.

RESULTS: Clinical examination revealed hypotonia and ataxia with a convergent squint, nystagmus, myopia with pontocerebellar hypoplasia on the MRI of the brain. The initial electroretinogram showed reduced amplitude of the scotopic and photopic b wave. At 3 years of age, biological and genetic evaluations confirmed the diagnosis of CDGS Ia. At 6 years of age, the electroretinogram was flat with a marked delay in psychomotor development.

CONCLUSION: Carbohydrate-deficient glycoprotein syndrome type Ia is a recently described cause of progressive retinal degeneration and an electroretinogram should be taken in patients with psychomotor retardation of unknown etiology. In addition, strabismus associated with electrophysiological signs of anomaly should raise the possibility of CDGS Ia.