Doc Ophthalmol. Abstracts of the 56th Annual Symposium of the International Society for Clinical Electrophysiology of Vision

(ISCEV, Reims 18-13 June 2018)

9.13 Autosomal recessive bestrophinopathy: a case series

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Purpose: Autosomal recessive bestrophinopathy (ARB) is a recently described clinical entity which is consecutive to the presence of two mutations in BEST1 gene. The presentation is uneven and the diagnosis may be difficult in children. Electrophysiology allows elimination of differential diagnoses and support of the clinical presumption.

Methods: We performed a retrospective observational study of cases diagnosed as ARB. Patients were examined in the ophthalmology department of Necker-Enfants Malades University Hospital. Multimodal imaging had been performed for each patient. Full-field ERG and sensory EOG helped the diagnosis of ARB, which was confirmed with a molecular study.

Results: Four patients presented with ARB, two adults and two children. No anomaly was identified in the parents of the two children. Patients presented with acuity ranging from "counting fingers" to 1.0 (decimal). Lesions were associated with macular yellowish deposit of material at the posterior pole, patchy atrophy areas, or pigmented lesions. On spectral domain ocular coherence tomography (SD-OCT), a pathological triad was noticed: hyperreflective subretinal material, subretinal fluid, and macular schisis. In all cases, the full-field ERG was normal but the EOG showed a severely reduced Arden ratio, eliminating retinal dystrophies or enhanced s-cone syndrome, which are the main differential diagnoses. There was no correlation between clinical presentation and ERG or EOG results. Electrophysiology alone could not differentiate between typical Best macular disease and these four cases of ARB, but the clinical presentation was unmistakable. For each patient, two mutations were identified in BEST1 gene.

Conclusions: We present the ophthalmological features of a series of four patients with ARB. A normal ERG associated with an impaired EOG confirmed the clinical diagnosis. Molecular analysis with two mutations in BEST1 gene enabled a formal diagnosis. The visual prognosis of ARB is very variable, from normal central acuity to low vision due to various retinal complications.