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Is electrophysiology necessary for the diagnosis of inherited retinal dystrophies at the time of multimodal imaging?

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Introduction: Inherited retinal dystrophies (IRD) are rare disorders characterized by an apoptosis of either retinal epithelium cells, cones, rods or cones and rods. More than 250 genes with or without a purely retinal expression are associated with these IRD. To add, different mutations in one gene can lead to a macular phenotype as a retinal one (PRPH2, BEST1, CRX, CRB1) as to dominant or recessive disorders (BEST1, NR2E3). An inherited retinal dystrophy is to be considered in any patient with a symmetrical bilateral involvement, particularly if there is at least one of the following signs: a family history, a photophobia with color vision anomalies, a night blindness, or a reduction of the peripheral visual field.

Methods: To establish a final clinical diagnosis, patients have multimodal imaging including color and autofluorescence images, optical coherence tomography with macular and optic nerve head scans and mapping. Fluorescein angiography is not required in IRD.

Results: Why electrophysiological explorations are essential despite multimodal imaging?

1. To confirm a disease at a preclinical or incipient stage, especially in cone dystrophies where multimodal imaging can be subnormal but cone driven responses are decreased.
2. To confirm if a patient is a non-affected carrier in Best's disease with altered EOG or in X linked retinitis pigmentosa with reduced cone driven responses.

3. To quantify the disease topography or severity in Stargardt's disease with three prognostic subgroups, in "localized" retinitis pigmentosa (pericentral, inferior forms).
4. To distinguish photoreceptor loss versus dysfunction (prolonged dark adaptation time) in fundus albipunctatus and in EMAP, a severe macular atrophy of middle-aged patients.
5. To confirm the mode of inheritance, for example in congenital stationary night blindness disorders or in recessive forms of Best's disease with impairment of both EOG and full-field ERG.
6. To specify an ERG phenotype that redirects to an enhanced S-cone syndrome, a congenital stationary night blindness, a neuronal ceroid lipofuscinosis, or an X-linked retinoschisis at an atrophic stage.

Conclusions: In this lecture, the place of electrophysiology will be illustrated through clinical cases in which its role has been essential to correct the clinical diagnosis, the mode of inheritance and the prognosis of the disease.

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