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8.04 Does unilateral retinopathy pigmentosa exist? Report of four cases

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Purpose: To manage the follow-up of unilateral retinopathy pigmentosa to confirm the diagnosis and to discuss its etiologic opinions.

Methods: Four cases of unilateral retinopathy pigmentosa were selected according to their similar unilateral anatomic and functional ocular anomalies. Full field ERG, mfERG, multimodal imaging, visual field, visual acuity and genetic testing were performed.

Results: Three women and one man, ranging from 25 to 33 years old, were studied. In all cases their visual acuity was bilaterally optimal. In contrast, their visual field was unilaterally very constricted, along with a strict unilateral decrease of both full field ERG and mfERG. Their fundus appearance was also characterized by strict unilateral anomalies, ranging from narrowed retinal vessels, mottling and granularity of the retinal pigment epithelium, migration of pigment in various sizes of clumps or bone spicule formations, atrophy of the retinal epithelium to papillary atrophy. Fundus autofluorescence imaging revealed unilateral central and midperipheral autofluorescence signal decrease, associated with an abnormal parafoveal ring arc of increased autofluorescence. Spectral domain optical coherence tomography scans showed one-sided photoreceptor disruption; cystoid oedema was described in one case. Infectious, neoplastic, and vascular causes were excluded for all four patients, but one had a possible history of eye trauma. Genetic test results are pending. We consider the etiologic hypotheses from the literature where mosaïcism or somatic mutations are discussed.

Conclusions: Unilateral retinopathy pigmentosa is a rare tapeto-retinal dystrophy affecting strictly only one eye, the fellow eye being completely unaffected. All infectious, traumatic, vascular and neoplastic causes have to be excluded and a long visual electrophysiologic follow-up is required for this diagnosis where mosaïcism or somatic mutation may be suspected.