Mystery of the blinding tunnel: compressive optic neuropathy with atypical retinitis pigmentosa in a paediatric patient

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ABSTRACT

Cases of atypical retinitis pigmentosa (RP) in paediatrics were not well documented in literature. We report an unusual case of atypical RP with concurrent compressive optic neuropathy. A 7-year-old healthy boy presented with right eye sudden unprovoked vision loss and night blindness. Right relative afferent pupillary defect was positive, with visual acuity (VA) of hand movement in the right eye (RE) and 6/30 in the left eye (LE). Anterior and posterior segment was normal, neurological assessment was unremarkable. Visual field (VF) assessment was unable to be performed for RE, while LE showed a constricted VF. Visual evoked potential is suggestive of chiasmal and right optic tract lesions. Magnetic resonance imaging (MRI) brain and orbit noted compression of the optic chiasm and intracranial right optic nerve by internal carotid artery. Surgical decompression noted intraoperatively very minimal compression. Post-operatively, the VA was 6/7.5 in both eyes, VF improved with residual tunnel vision. After a year, his bilateral VF worsened, but VA remained 6/7.5. Repeated MRI was unremarkable. From a multi-disciplinary discussion, he was treated as optic neuritis with systemic corticosteroids. Serum vitamin B, folate, autoimmune workup, anti-aquaporin-4, and Leber hereditary optic neuropathy gene analysis were normal. After 2 years of follow-up, multiple hypopigmented spots appeared over bilateral peripheral retina with attenuated retinal vessels. Fundus florescence angiogram revealed multiple hyperfluorescent spots. Diagnosis of atypical RP was made. The unusual presentation in this patient poses a diagnostic challenge as the symptoms of atypical RP were shadowed by the compressive optic neuropathy and initial normal fundus appearance.