Wyburn Mason Syndrome: A Rare Phacomatosis

An 18-year-old man presented with gradual diminution of vision in his left eye, a whooshing sound in his head, and headache. Ophthalmoscopy (Fig 1A) evinced a dilated, tortuous retinal artery arising from the left optic disc, passing temporally toward the macula, communicating with a dilated retinal vein, and traversing back to the disc. Arterial phase of fundus fluorescein angiography (Fig 1B) showed rapid transit of dye through the arteriovenous malformation (AVM) without leakage. Optical coherence tomography through the fovea (Fig 1C) documented malformed vessels. Wyburn-Mason syndrome was confirmed after AVMs were identified in the brain with magnetic resonance imaging.

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