Optical coherence tomography angiography in Leber hereditary optic neuropathy

A 17-year-old man had acute vision loss OS (visual acuity 20/40), having lost vision OD months before (20/400). There were cecocentral scotomas OU. Mitochondrial DNA testing revealed a T14484C mutation, suggestive of Leber hereditary optic neuropathy (LHON), which results in painless sequential vision loss. Acutely, there is hyperemia of the optic nerve, circumpapillary microangiopathy, and dilated, tortuous vasculature; the pathognomonic finding is nerve swelling without leakage on fluorescein angiography. Chronically, the optic nerve develops atrophy, sometimes with cupping. Optical coherence tomography angiography provides rapid, high-resolution visualization of retinal vasculature without dye injection, and is a novel imaging modality in LHON (figure).

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