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## NEW FINDINGS FROM MULTIMODAL FUNDUS IMAGING OVER 3 YEARS OF A PATIENT WITH MICROCEPHALY, CHORIORETINOPATHY, AND KIF11 MUTATION

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### Abstract

**Purpose:** To describe ocular features and long-term follow-up in a patient with microcephaly, chorioretinopathy, and KIF11 mutation.

**Methods:** Multimodal imaging including fundus color photography, autofluorescence, spectral-domain optical coherence tomography, visual fields, electrophysiologic assessment, and neuroimaging.

**Results:** A 44-year-old man with microcephaly and long-standing poor vision in his right eye presented for general ophthalmic review. Fundus examination revealed bilateral, symmetrical, inferior, atrophic chorioretinal lacunae, outer retinal and retinal pigment epithelial atrophy, curvilinear streaks, and retinal arteriolar sheathing. These findings were documented over a 3-year period by multimodal imaging and showed slow progressive deterioration in visual acuity, visual field testing, and fundus autofluorescence appearance. Genetic testing confirmed a KIF11 gene mutation.

**Conclusion:** Curvilinear streaks and retinal arteriolar sheathing in this patient expand on the more typical fundus findings of KIF11 mutations. The outer retina is preferentially involved, and there is anatomical sparing of the macula until later in the disease state when multifocal electroretinography indicates functional impairment. Lacunae represent scleral depressions with the loss of overlying choroid and outer retina. Slow atrophic progression with loss of vision may occur over time.

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