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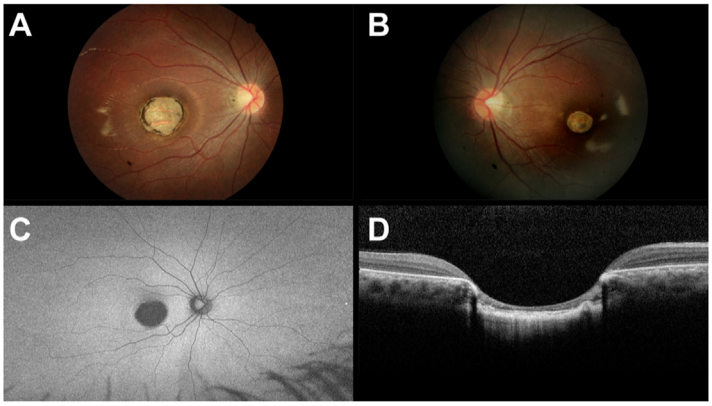
Pictures & Perspectives



Macular Pseudocoloboma in Achromatopsia

A 13-year-old girl with a history of nystagmus from 4 to 18 months of age developed intense photophobia. Examination showed poor vision (0.8 logarithm of the minimum angle of resolution in both eyes). Fundus photographs (**A**, **B**), autofluorescence imaging (**C**), and spectral domain OCT (**D**) showed stable, deep, coloboma-like central macular lesions in both eyes. We observed absent cone responses but normal rod responses on full-field electroretinogram. Molecular genetic testing detected a pathogenic c.1561C>T (p.Gln521*) homozygous mutation in the exon 13 of *ATF6*. The presence of clinically extended lesions of the macula, usually a sign of cone-rod dystrophies when not postinfectious, does not always preclude the diagnosis of achromatopsia (Magnified version of Figure **A**-**D** is available online at www.aaojournal.org).

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