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Abbreviations and Acronyms:

CV = coefficient of variation; ECD = endothelial cell density;

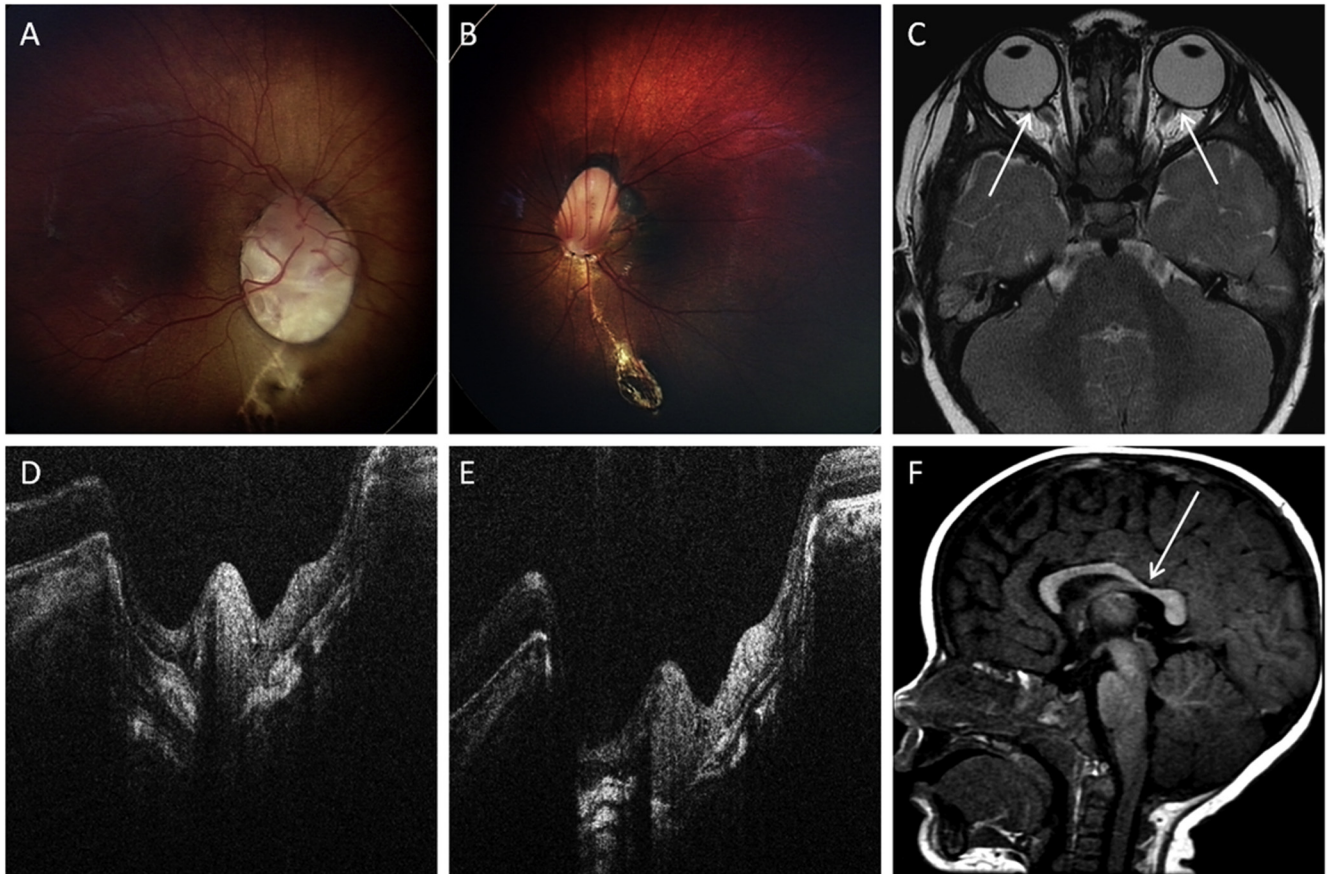
HLA = human leukocyte antigen; IOP = intraocular pressure;

UCLA = University of California, Los Angeles.

Correspondence:

Gary N. Holland, MD, UCLA Stein Eye Institute, 100 Stein Plaza, Los Angeles, CA 90095-7000. E-mail: uveitis@jsei.ucla.edu.

Pictures & Perspectives



Aicardi Syndrome

A 10-month-old girl was referred for “wandering eye.” Ophthalmoscopy (A, right; B, left) revealed scalloped, excavated retinal areas and pathognomonic peripapillary chorioretinal lacunae. Optical coherence tomography (D, E) confirmed absence of normal tissue. Magnetic resonance imaging showed small colobomas (C, arrow) and thinning of the corpus callosum (F, arrow). “Morning glory syndrome” associated with callosal dysgenesis and seizures constitutes Aicardi syndrome. Only manifest in girls (or boys with Klinefelter syndrome), Aicardi syndrome was considered a dominant X-linked condition, although recent evidence suggests that mutations in the *TEAD1* gene (chromosome 11) may explain some or all of the cases. All cases are due to de novo mutations.

ROBERT J. YOUNG, MD¹

JASMINE H. FRANCIS, MD²

DAVID H. ABRAMSON, MD²

¹Neuroradiology Service in the Department of Radiology, Memorial Sloan Kettering Cancer Center, New York, New York; ²Ophthalmic Oncology Service, Memorial Sloan Kettering Cancer Center, New York, New York