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Pediatric retinal dystrophies and their characteristic findings in spectral-domain optical coherence tomography (SD-OCT) and fundus autofluorescence (FAF)

Chung, Daniel C
University of Pennsylvania, USA

The diagnosis of inherited retinal degeneration in children can been challenging, but in recent years, major progress has been made in the knowledge of disease presentation, clinical course, retinal imaging and molecular genetic testing. The knowledge of the clinical presentation, coupled with recent advances in retinal imaging (SD-OCT, FAF) has enabled the clinician to narrow the potential genes for molecular genetic testing. Therefore it is important to be able to recognize key clinical characteristics of disease, as well as understanding the distinct imaging patterns of the retina as seen on SD-OCT and FAF imaging. This presentation will allow physicians to:

- Identify the clinical presentation and disease course of the most common pediatric retinal dystrophies
- Correlate phenotypic characteristics to specific genotypes
- Discuss retinal phenotypic characteristics found on spectral domain optical coherence tomography (SD-OCT) and fundus autofluorescence (FAF) in pediatric retinal degenerations
- Technical pearls in OCT/FAF imaging in young children with nystagmus, poor central fixation and other difficult presentations