

- A thorough ocular/medical history and pedigree documenting family history of eye disease should be obtained at the initial visit and updated during subsequent visits.
- Understanding the patient's mood and affect is important, being sensitive to signs of depression that may accompany progressive vision loss.
- **Molecular genetic testing** (genotyping) of the patient can be valuable to confirm the diagnosis and optimize management.
- **Clinical evaluation:**
 - best-corrected visual acuity with manifest refraction using standardized eye charts,¹
 - biomicroscopy with measurement of intraocular pressure (assessment of cataracts and anterior segment anomalies),
 - dilated ophthalmoscopy to document features potentially related to vision loss (optic nerve and other retinal diseases, deposits, vessels, atrophy, schisis and macular edema).
- **Imaging**
 - **Standard color or wide-field fundus photography** may be performed at the initial visit to provide documentation of disease state and provide the context to align and compare data from other fundus modalities such as fundus autofluorescence (AF) images.
 - For patients with nyctalopia and/or peripheral visual field loss, wide-field imaging has advantages since the primary site of disease is not in the macula in early disease.
 - Fundus photos should be used sparingly in Stargardt disease and other maculopathies due to the risk of light toxicity. A test should be ordered only if it will be useful to monitor disease progress or determine if a patient is eligible for a clinical trial.
 - Macular or wide-field AF fundus imaging using reduced illumination (25%), longer exciting wavelengths, infrared AF or near infrared fundus reflectance are good alternatives to short-wavelength AF in patients with retinitis pigmentosa and Stargardt disease to possibly reduce the risk of phototoxicity, although near-infrared imaging provides distinct information.^{2,3}
 - Near infrared fundus reflectance is a separate imaging modality from short-wavelength AF that also provides insight on RPE health.
 - **Optical Coherence Tomography (OCT)** provides cross-sectional imaging of the photoreceptors, retinal pigment epithelium, and inner retinal layers including the retinal nerve fiber layer.
 - High-density volume scans with documentation of central retinal thickness provide a useful baseline for monitoring progression in structural features and helping to monitor CME, epiretinal membranes or macular schisis.
- **Visual field testing** is important to document the functional extent of vision from central to the far periphery. This is essential for determination of legal blindness, disability, and to counsel patients on visual limitations.
 - **Static visual field testing** has advantages of automated indices of sensitivity loss and performance parameters to assess reliability.
 - Newer perimeters test the entire field, and digital data can be exported into other applications for specific purposes such as modeling of sensitivity, which is useful for quantitative measurement in clinical trials.
 - Although static perimetry using the Humphrey visual field (HVF) 30-2 protocol is acceptable in the federal registry for the determination of