Hereditary Optic Neuropathies

Etiology/Pathophysiology

- exact pathophysiology unknown, mitochondrial dysfunction thought to play a part

Signs/Symptoms

- symmetric, bilateral, painless, usually permanent, central visual loss
- affects papillomacular nerve fiber bundle → central/cecocentral scotomas
- May present with other ophthalmologic, neurologic, or systemic symptoms/signs/diseases such as hearing loss, ophthalmoplegia, cataracts, myopathy, diabetes mellitus, paraplegia, skin abnormalities, ataxia

Differential

Optic Neuropathies WITHOUT other systemic signs

- Leber Hereditary Optic Neuropathy
  - point mutations in mitochondrial DNA, predominately males age 15-35, second eye affected within 1 year of first eye affected, pseudoedema and optic atrophy can be seen on exam
- Dominant Optic Atrophy
  - mutations in OPA1 gene of chromosome 3, 4-10 years of age, unaware of visual problem and discovered as consequence of examination of other family members, mild to moderately reduced visual acuity in both eyes, on exam optic atrophy may be subtle, temporal only (triangular excavation), or involve entire disc
- Autosomal Recessive Optic Atrophy
  - discovered before 3-4 years old, consanguinity between parents, severely reduced visual acuity (may have sensory nystagmus), optic discs atrophic and deeply cupped

References

Taylor and Hoyt's Pediatric Ophthalmology and Strabismus, Fifth Edition. *Hereditary Optic Neuropathies* Chapter 54, 581-591