Leber Hereditary Optic Neuropathy

Genetics

-MTND1, MTND4, MTND6 (complex I subunits of the mitochondrial respiratory chain)

-Mitochondrial inheritance

Clinical findings/Dysmorphic features

-Develops during young adult life --> visual blurring in central visual field in one eye

-Similar symptoms appear in the other eye an average of two to three months later

-Visual acuity is severely reduced

-Visual field testing shows an enlarging dense central or centrocecal scotoma

-After the acute phase, the optic discs become atrophic

Etiology

-In Northern Europe: 1:10,000 – 1:50,000

Pathogenesis

-Focal degeneration of the retinal ganglion cell layer and optic nerve

Genetic testing/diagnosis

-Bilateral, painless, subacute visual failure that develops during young adult life

-Common variants: m.3460G>A in MT-ND1, m.11778G>A in MT-ND4, m.14484T>C in MT-ND6

Others

-Males 4x more likely affected