**Condition:** Leber hereditary optic neuropathy (LHON)

**Inheritance:**
Mitochondrial (maternal transmission).

**Genetic etiology:**
Mutation in various mitochondrial genes involved in complex I of respiratory chain; the three most common mutations are G11778A in *MT-ND4*, T14484C in *MT-ND6*, and G3460A in *MT-ND1*.

**Frequency:**
Clinical disease about 1/30,000.

**Clinical features:**
LHON presents in young adults, particularly in males with subacute onset of blindness in one or both eyes. Vision progressively deteriorates, though some spontaneous improvement may occur.

**Management:**
Supportive care.

**Genetic counseling:**
Transmitted maternally, with affected individuals generally homoplasmic for mutation. There is a high rate of nonpenetrance, especially in females, for unknown reasons. Molecular genetic testing is available.