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.

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