

Leber hereditary optic neuropathy (LHON) is mainly characterized by bilateral, painless subacute loss of central vision during young adult life. In most cases, symptoms begin with one eye first, followed a few weeks later by visual failure in the other eye. Extremely rarely there may be neurologic abnormalities, such as peripheral neuropathy, postural tremor, nonspecific myopathy, and movement disorders. LHON is caused by mutations in mitochondrial DNA and it is transmitted by maternal inheritance. LHON affects approximately 1:50,000 people. Many carriers never suffer significant visual loss; males are about four to five times more likely than females to be affected. LHON affects both males and females and can only be inherited from a female. The birth prevalence of LHON is approximately 1 in 50,000 people. LHON is diagnosed based on ophthalmologic findings, which include specialized visual testing. The testing involves dilated fundus examination to identify characteristic changes in the optic disc and vascular changes during the acute phase, electrophysiologic studies, and neuroimaging. Molecular genetic testing for mitochondrial genes associated with LHON can be used to confirm diagnosis. Most affected individuals know if their family members also are affected by LHON.