LHON and Cardiac Issues

Some individuals with an LHON mutation, with or without vision loss, experience cardiac issues.

LHON is a mitochondrial disease that impacts cellular energy production. The optic nerve consumes a large amount of energy, and its function can be disrupted when an individual has an LHON mutation. For many such individuals, sudden-onset vision loss is the only recognized symptom.

The heart is also a high-energy-dependent tissue. Abnormalities of the heart rhythm (arrhythmias) or heart muscle (cardiomyopathy) are seen in a number of different mitochondrial diseases. Some studies suggest that people with an LHON mutation have a higher-than-usual likelihood of having a cardiac issue, though additional research is required.

Until additional research better characterizes the nature of potential heart disease in LHON, those with an LHON mutation may wish to consult their doctor or cardiologist about this potential connection. A baseline cardiac assessment including some or all of the following tests may be recommended:

Electrocardiogram

Also referred to as an ECG or an EKG: a quick test performed in the clinic to evaluate the heart's rhythm and electrical activity.

Echocardiogram

An ultrasound to evaluate the heart structure.

Holter monitor

A portable, continuous ECG worn for a day or longer to evaluate the heart's rhythm over time.

Relevant Research

These articles may be of interest to you and your health care providers:

Hey TM, Nielsen SK, Eriksen U, Hansen F, Mogensen J. Leber's Hereditary Optic Neuropathy and Hypertrophic Cardiomyopathy. CJC Open. 2022. In press. doi:10.1016/j. cjco.2022.06.005

Orssaud C. Cardiac Disorders in Patients With Leber Hereditary Optic Neuropathy. Journal of Neuro-Ophthalmology. 2018. 38(4): 466-469. doi:10.1097/WNO.0000000000000623

Newman NJ, Yu-Wai-Man P, Sadun AA, Karanjia R, Carelli V. Management of ophthalmologic manifestations of mitochondrial diseases. Genetics in Medicine. 2017. 19(12): 1380-1381. doi:10.1038/gim.2017.171

Finsterer J, Zarrouk-Mahjoub S. Leber's hereditary optic neuropathy is multiorgan not mono-organ. Clinical Ophthalmology. 2016. 10: 2187-2190. doi:10.2147/OPTH.S120197

Sorajja P, Sweeney MG, Chalmers R, Sachdev B, Syrris P, Hanna M, Wood ND, McKenna WJ, Elliott PM. Cardiac abnormalities in patients with Leber's hereditary optic neuropathy. Heart. 2003. 89(7): 791-792. doi:10.1136/heart.89.7.791