LHON Carrier Checklist

Who's at risk?

Everyone on the maternal bloodline	Both males and females	Onset at any age
Someone with genetic confirmation of an LHON mutation[1] will carry the same mutation.	25% of those affected by LHON are female. LHON is not just a "young man's disease."	Those carrying an LHON mutation are never too old or too young to become affected.

Be Prepared

Onset of vision loss happens quickly. If it does, there will be many issues to address and obstacles to overcome all at once, and it will be overwhelming. Preparation makes onset less traumatic.

Take the following actions now if you or a loved one carries an LHON mutation by reviewing the LHON Carrier Checklist

1 Find one or two LHON doctors

- ☐ Identify a local neuro-ophthalmologist[2] or ophthalmologist who understands LHON and is on your insurance plan.
- ☐ Get established as a patient.
- ☐ Consider identifying an LHON expert to collaborate with your local LHON doctor.
- ☐ Complete a baseline vision exam with either LHON doctor and obtain a copy of your exam results (especially the Optical Coherence Tomography (OCT[3]) and Visual Field Test (VFT[4]).

2 Know your LHON mutation

- [ ] Ask a maternal relative or consider getting tested to determine your mutation.
- [ ] If you don't have genetic test results, develop a plan for how you'll get it done quickly if needed.
    - [ ] Consider Target Mutation Analysis, otherwise known as "known familial variant" (KFV[8]) testing, which can save time and money.
    - [ ] Evaluate lab options based on cost and turnaround time.
    - [ ] Determine who will order the test (LHON doctor, genetic counselor, or primary care provider - PCP [9]); be prepared to provide them necessary forms to order the test.

Store your medical records

- [ ] Maintain your baseline Optical Coherence Tomography (OCT) and Visual Field Test (VFT) in a paper or electronic file.[5]
- [ ] If you have genetic test results, store them with your other tests, or obtain and store a copy of a maternal relative's test results.

Know how to find clinical trials

- [ ] ClinicalTrials.gov; LHON patient advocacy organizations (i.e. LHON Collective, UMDF, LHON Canada, LHON Society).
- [ ] Understand inclusion or exclusion criteria[10], such as mutation, time since onset, and age.

Locate counseling services

- [ ] Consider meeting with a genetic counselor[6] to discuss LHON issues, such as discussing it with family.
- [ ] Look into meeting with a mental health therapist.

Be aware of factors that can impact LHON

- ☐ Don't smoke and avoid all environmental smoke
- ☐ Read the Proactive Considerations short list [11]
- ☐ Read the following journal article about Toxic Medications [12]

Know how to evaluate onset if you experience vision changes

- ☐ Evaluate each eye individually: Is one blurrier or cloudier than the other? Do you see a small blurry spot (scotoma[13]) in one or both eyes?
- ☐ Look at something bright red, first with one eye then the other: Is the red "muddier," darker, or pale orange with one eye?
- ☐ Visit an LHON doctor immediately if you answered "Yes" to any of these questions.

Definitions, Abbreviations, and Additional Resources

1 Mutation: In LHON, this refers to dysfunctional mitochondria that are passed on to children 100% of the time from the mother. There are 3 primary mutations in LHON that are the most common (11778, 14484, 3460), as well as many other rare mutations.

2 NANOSweb.org: Find a Neuro-Ophthalmologist

3 OCT: Optical Coherence Tomography

4 VFT: Visual Field Test

5 To understand LHON vision tests such as Optical Coherence Tomography (OCT) and Visual Field Test (VFT), watch: bit.ly/2Xa8dpR

6 Genetic counselor: A person who advises those carrying or affected by genetic disorders on the medical, psychological, and familial implications of that particular disease.

7 NSGC.org: Find a Genetic Counselor

8 KFV: Known Familial Variant; to learn more about genetic testing, watch: bit.ly/2BL49Vp

9 PCP: Primary Care Provider

10 Inclusion/exclusion criteria: Inclusion criteria are characteristics that potential participants must have in order to be eligible to participate in a clinical trial. Exclusion criteria are characteristics that would disqualify potential participants from participating in a clinical trial.

11 New to LHON

12 Toxic medications in Leber hereditary optic neuropathy (Kogachi et al., Mitochondrion, 2018). Available at: bit.ly/2JAJohp

13 Scotoma: A blurry spot in the visual field that will gradually increase in size with LHON onset. It differs from a floater, which appears as a speck of dust or a cobweb that moves across the field of vision and remains stationary.

This document should be considered general information only and should not be considered medical guidance or professional advice. Always direct any questions concerning your personal health to your doctor or another appropriate health care professional. December 2025