

LHON Data Collection Program (in Partnership with RARE-X)

Tips and Recommendations

Before you Start:

Direct Link to LHON Portal on RARE-X = <https://lhon.rare-x.org>

Web Browser - Use only Google Chrome or Apple Safari version 14 or higher

Questions / Concerns / Feedback – Malinda Marsh at MalindaLHON@yahoo.com AND support@rare-x.org

Tips and Tricks (Items that may seem a little tricky):

What Role to Assign Yourself - Whether you are affected by LHON vision loss, an unaffected carrier, or LHON Plus, enroll as a “Patient”. In other words, if you carry an LHON genetic mutation, enroll as a “Patient.”

What Role(s) to select at the initial point of Registration - Consider that you may one day want to enroll a deceased loved one or a minor child - select ALL potential role(s) on the Request Access page at the beginning of your enrollment and at the end of the Informed Consent. This will give you that flexibility at a later date to add others.

Second E-mail may be needed to enroll - If you have enrolled in the mitoSHARE Registry, you must use a different email for this LHON Data Collection Program (DCP).

General Information Section - In the General Information section, the bottom of the form is not immediately visible. To complete this page, keep scrolling down until you reach and complete all the required fields and then click on the “Save” button to save your answers.

For Caregivers or those adding information of a Deceased Loved One - After you have enrolled and created an account you will be brought to the RARE-X homepage. There is a tab that says, “Add Patient”. Click on that tab to get started. If you have more than one person to add to the DCP, you will do this for each person you register.

Expanding Surveys for participants entered by a Caregiver - Once the patient is added and you are back on the home screen, next to the patient’s name all the way to the right, there will be a book icon that you will need to click on. This will populate all the available surveys to be filled out.

Important Notes and Instructions (Keep handy while in the DCP):

Health and Development Survey – This is a very important survey. Everything you answer “Yes” to will then create a more detailed survey called a Level 2 Survey for that identified issue. The Level 2 surveys can then be found on your dashboard after completing this survey. Any symptom or diagnosis you have can be included in this section, even if you don’t think it is related to LHON. You never know what

commonalities we could learn when we put all the data together, looking at each person from head to toe.

Where to Input Specific Non-Vision Loss Symptoms that are fairly commonly reported with LHON -

- Migraine - answer “yes” to “Brain and/or Nervous System”
- Central Auditory Processing Disorder - answer “yes” to “Ears and/or Hearing” and describe in Other

Addition of a Genetic Report to substantiate your LHON Diagnosis - If a genetic report is available, please upload it to the LHON Data Collection Program so health information can be associated with genetic information.

Consenting – If you are a patient reporting just for yourself, you will consent just once. If you are a caregiver you will be asked to consent for yourself and then also for your minor child or deceased loved one once you add them as a patient. If you have multiple registrants, you will need to consent for each of them.

Frequently Asked Questions:

How long will this take? It may take around 45-60 minutes for account creation, enrollment, consenting, then getting into all the surveys.

Can we do it on our mobile phones or tablet? Not at the moment. We suggest using a computer. There are future enhancements to the platform that will improve the functionality on mobile devices.

Do I have to live in the United States to participate? No. This is global and you can fill this out anywhere in the world, though if you are outside the US, you will have to agree to an additional consent question. The goal of this is to be a global database to include all who have LHON.

What languages are available? Currently English. If you understand English (even if it is not your primary language) or have someone that can go through this with you and translate, we encourage you to participate.

When will other languages be added? We don't have firm dates. We expect Spanish to be next and RARE-X is targeting the end of 2022, with more major languages to follow.

Why aren't there specific questions about LHON? The RARE-X platform is built for all rare diseases and uses standard surveys. The data is in a format that is research-ready and can be understood across diseases. This gives us an opportunity to attract greater interest from researchers and pharmaceuticals to solve for our symptoms when greater numbers of people across different diseases can share their diagnoses in a combined format to create a critical mass.

How often are we expected to enter data? The goal is for everyone to go into the LHON DCP once a year and review and make any updates, especially if a new symptom has presented itself. If specific surveys that are relevant to LHON are added, we will communicate that with you and hope you would go in and

fill out that specific survey. Once you have enrolled, every subsequent log-in will be significantly easier and less time consuming since you no longer need to enroll or consent.

Who owns the data? You (the patient or caregiver) always have full control of your data. It is owned by you and enabled by RARE-X.