USHER SYNDROME COALITION

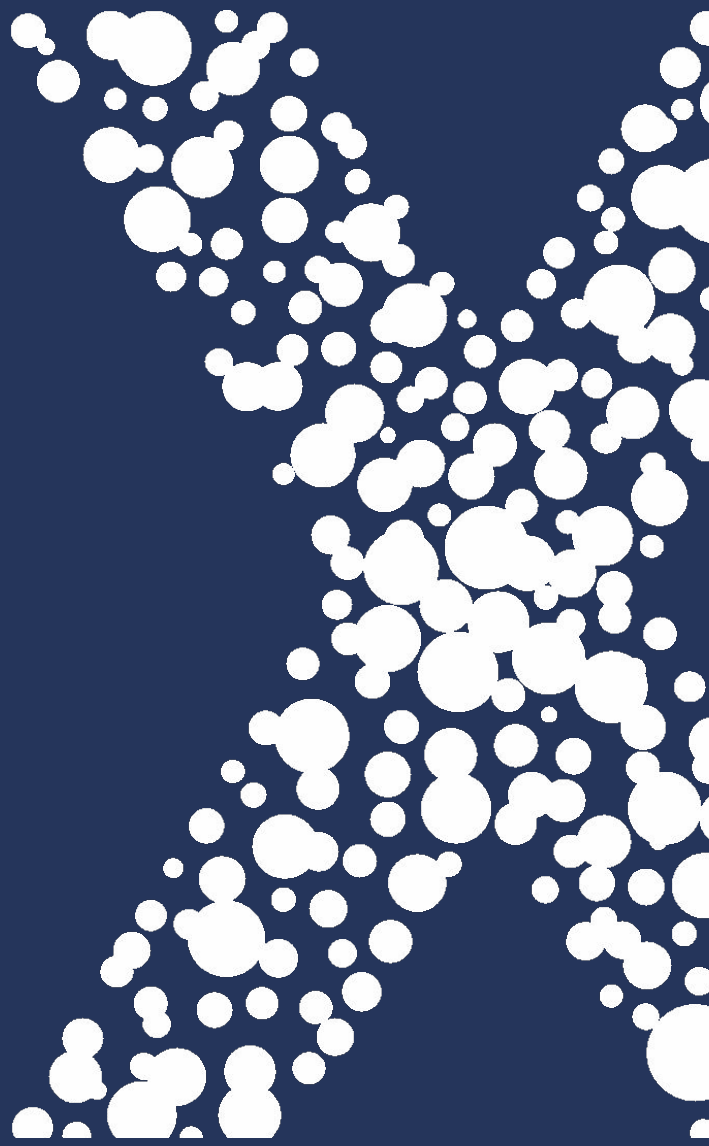
usher syndrome society

USHER 1F COLLABORATIVE

Usher Syndrome

Data Collection Program

November 16, 2022



Agenda

- What is RARE-X?
- Mission & Values
- What are we solving for?
- The RARE-X Data Collection Portal
- Data Governance

Agenda

- Benefits of the Data Collection Program
- Getting Started
- DCP User Journey
- Other Tips
- How this data collection is different

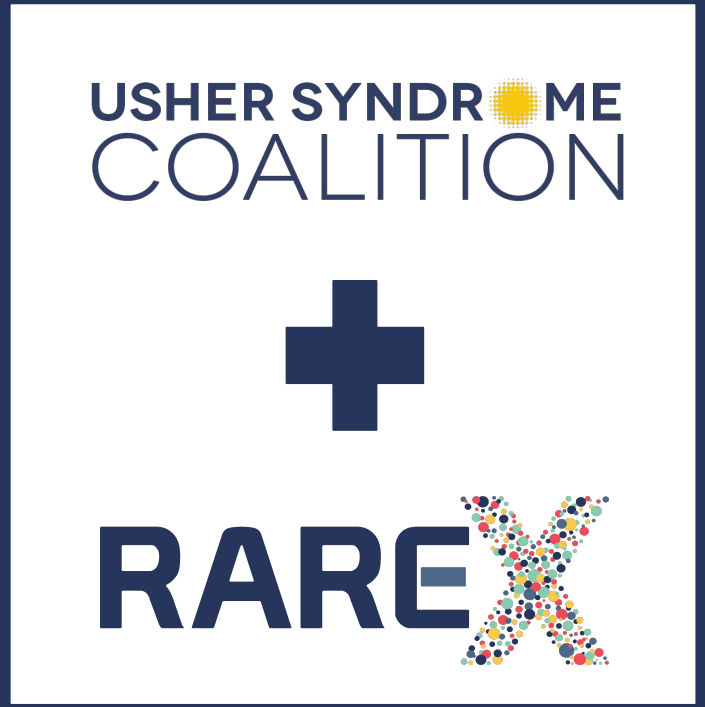
What Is RARE-X?

- RARE-X is a NONPROFIT created to accelerate rare disease research, treatments, and cures by removing barriers for data collection and sharing
- RARE-X is a platform to **collect, connect,** and **share** data

- *RARE-X does not own, sell, or do research with the data they collect*

Why did Usher Leadership Choose RARE-X?

- Participant owned
- Data security/privacy
- No cost to participants
- No cost to researchers
- Structured, standardized
- Data on all body systems
- Streamlines researcher access
- Speeds treatment development
- Ability to connect to existing data sources



What is the benefit to YOU?

- You will get data BACK to you in de-identified summary
- You may have the chance to participate in clinical trials
- Reach more researchers worldwide
- Update symptoms at any time
- Manage who uses your data
- Speed up treatment development

RARE-X Data Collection Portal

Participants & Communities

FOXP1

Usher

LHON

Ring14

SYNGAP1

ADCY5

RARE-X Website participant Community Pages

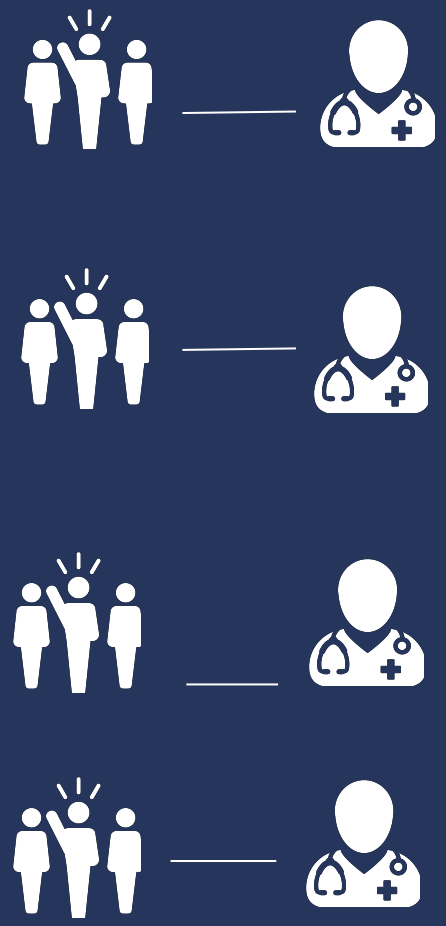
Data Collection Program with standardized domain modules

Consent | Data Sharing | General Info | Health & Dev. | Ethnicity | Quality of Life

Researchers

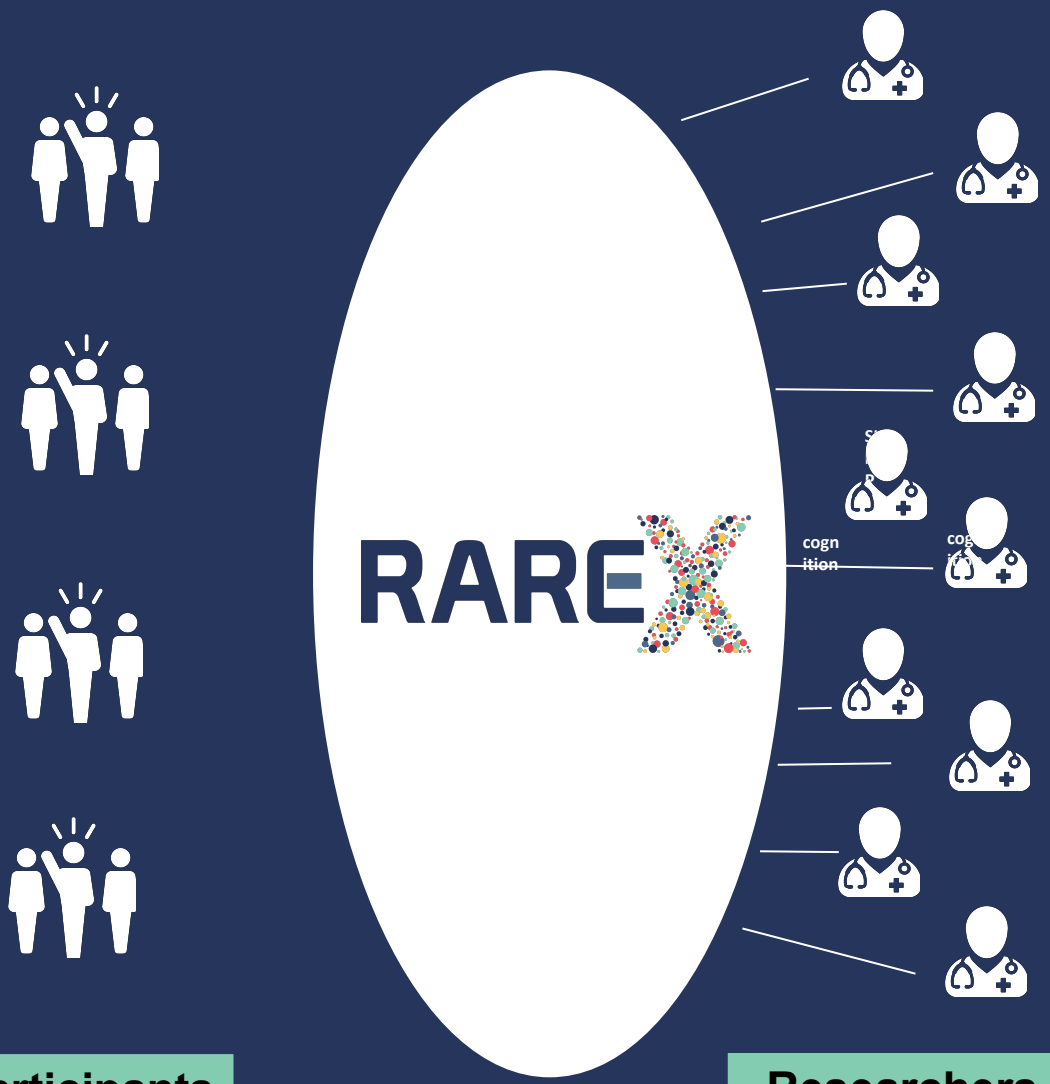
Intl

Why Providing The Broadest Data Sharing Is Critical



Participants

Researchers



Participants

Researchers

What Do You Need To Get Started?

- Email address and Create a password
- An approved Browser
  - Google Chrome, or
  - Apple Safari version 14 or higher
  - Microsoft Edge
- No need to finish it all at once

Participant Journey in the Data Collection Portal

Community

Community Page - Welcome, Privacy, Terms of Use, Start

Data Collection Program User ID Page

Verification eMail is sent to you - letting you add a password and complete your registration

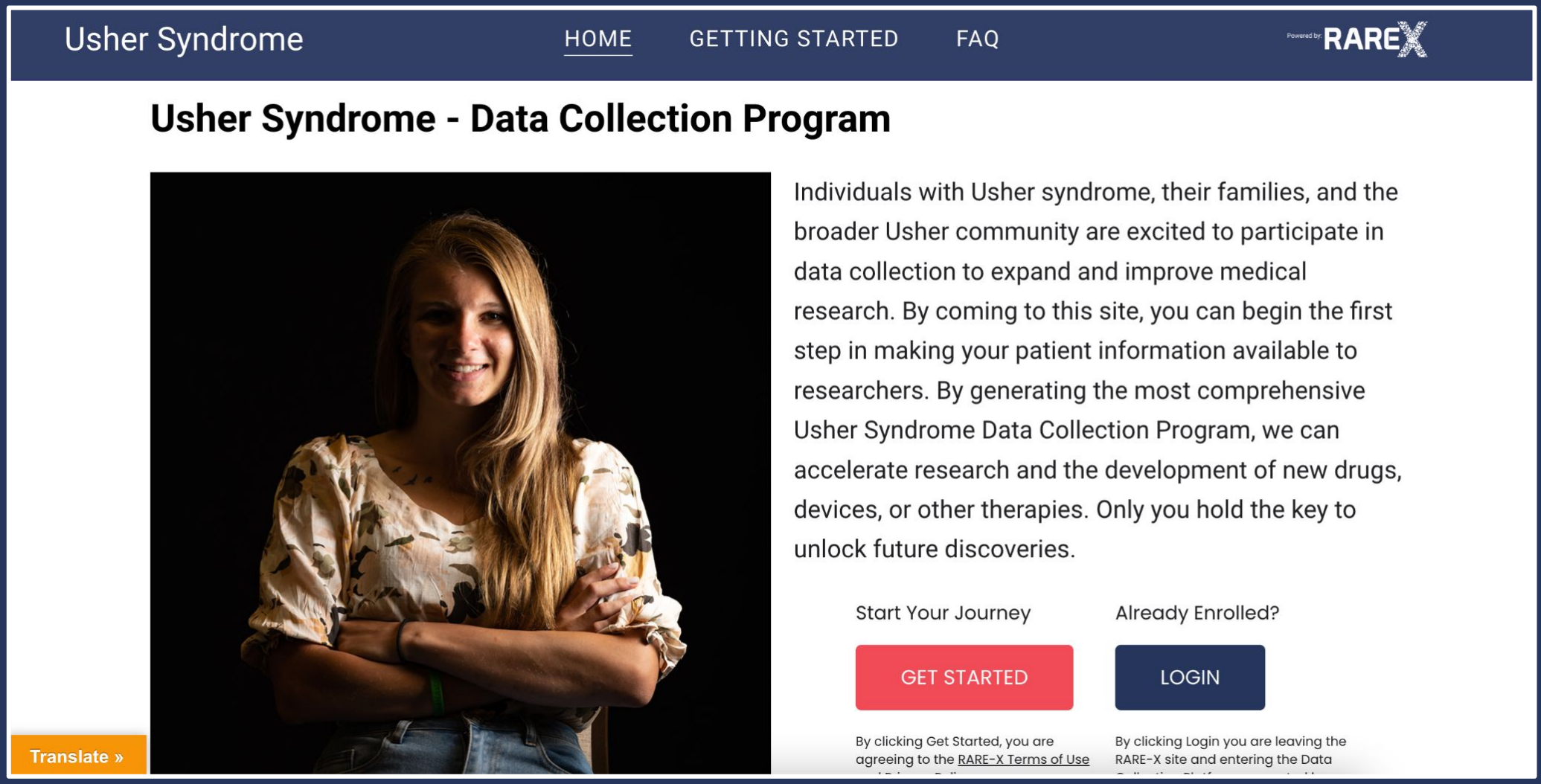
Participant Consent

Data Sharing Preference Survey

Survey Topics (Domains)

Participant Dashboard

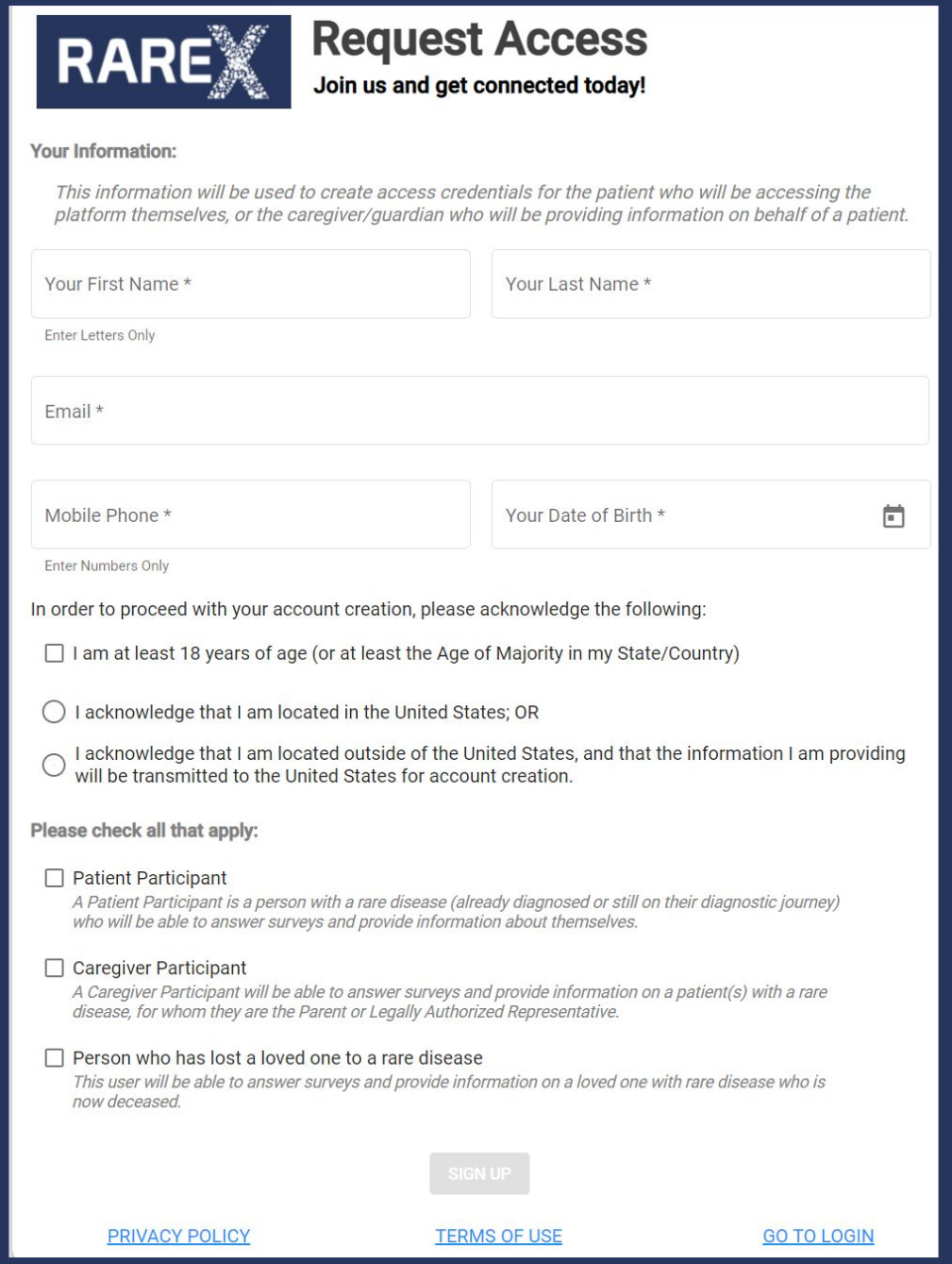
Usher Syndrome Community Page on RARE-X



https://ushersyndrome.rare-x.org/

First-time Login Page
(Pre-Qualifications Page)

All biological parents of underaged children with Usher syndrome would be considered “carriers” and should also choose “Patient Participant” in addition to “Caregiver”

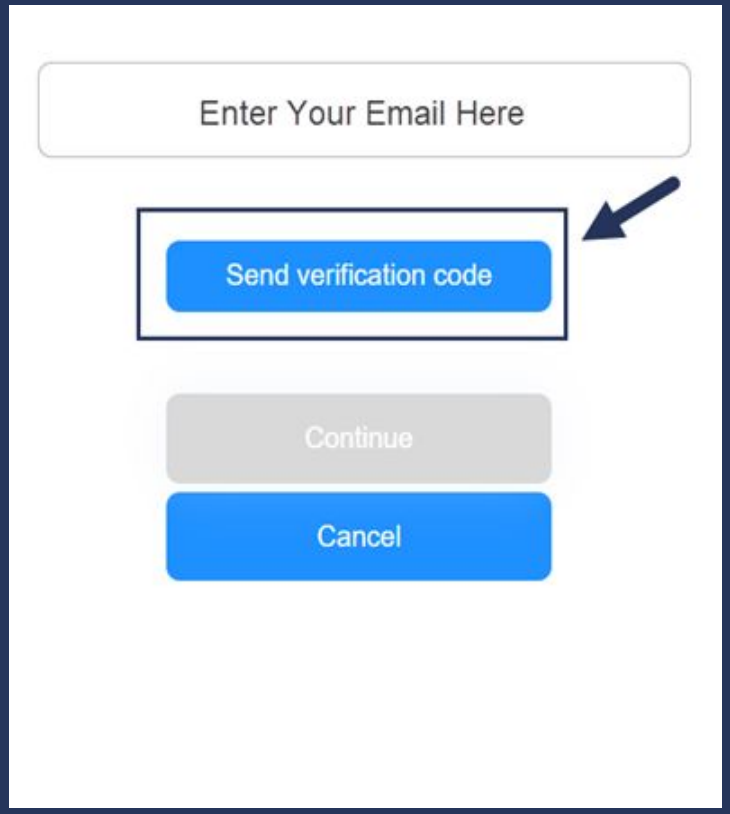


Receive Account Creation Email

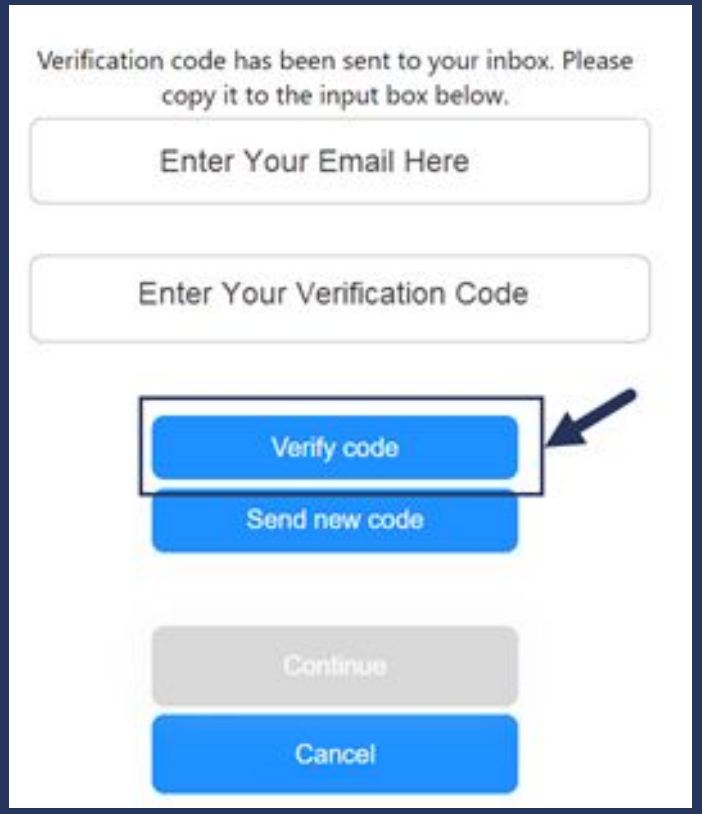
If you do not receive a response within a few minutes, check your spam/junk folder

Email Verification (multi-factor for your privacy)

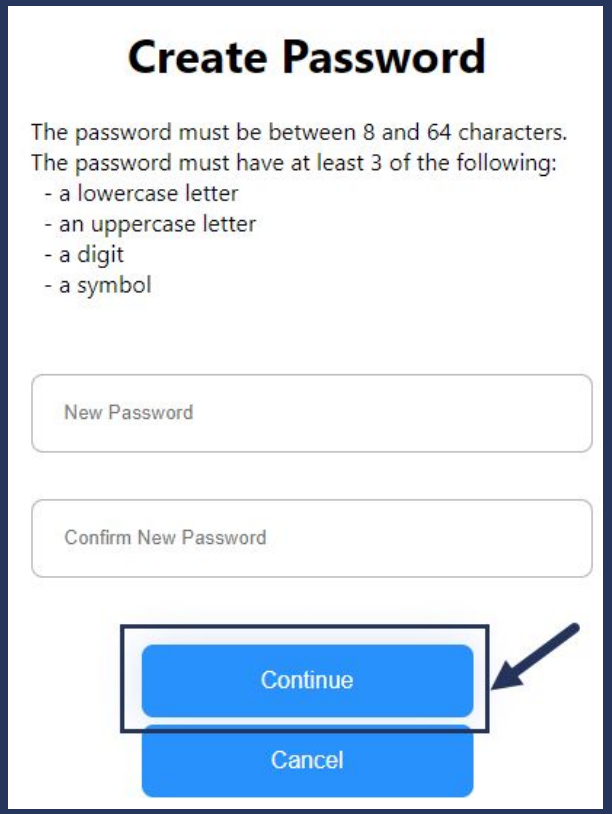
1. Request your verification code



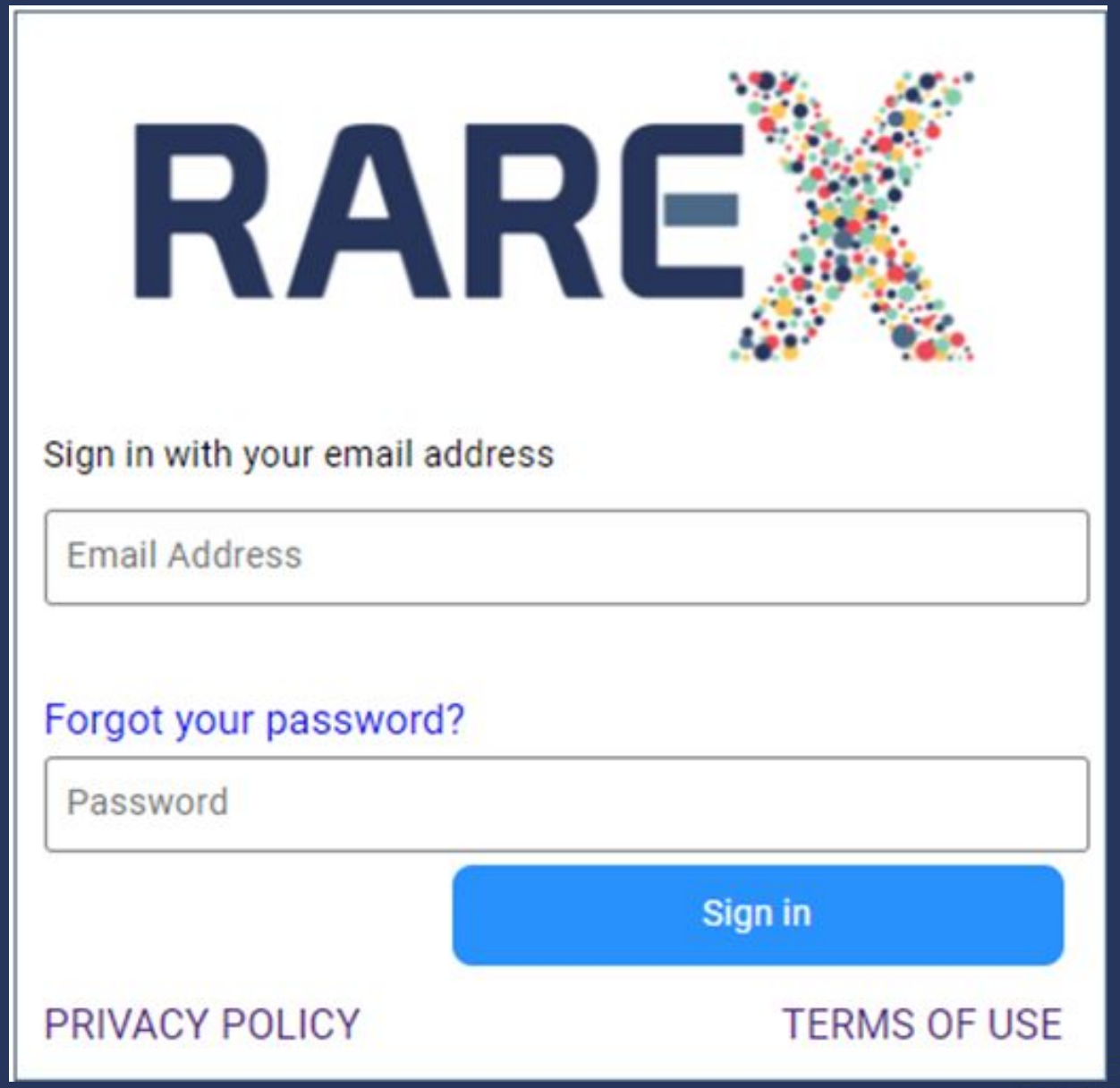
2. Confirm your verification code



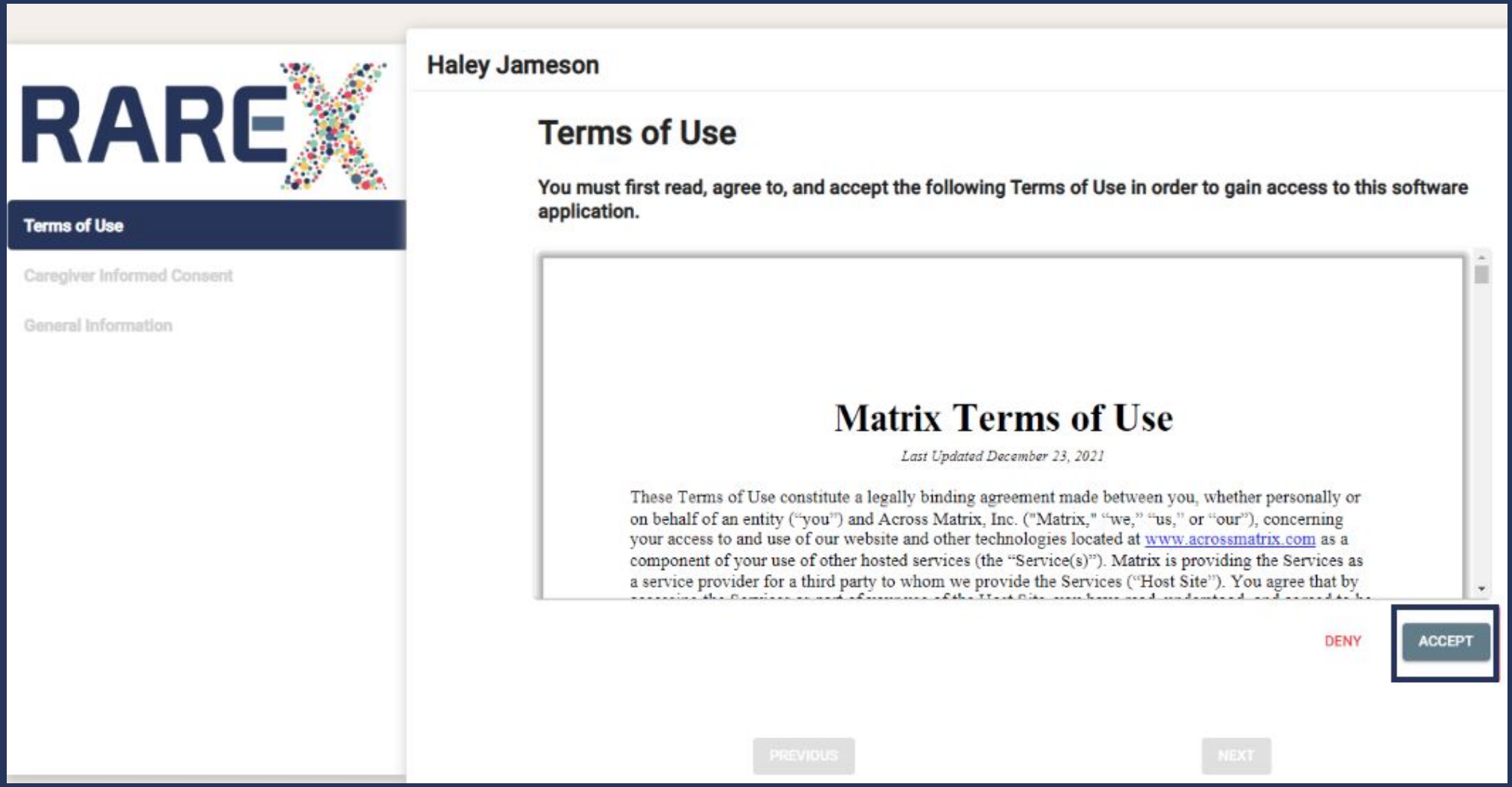
3. Create your password



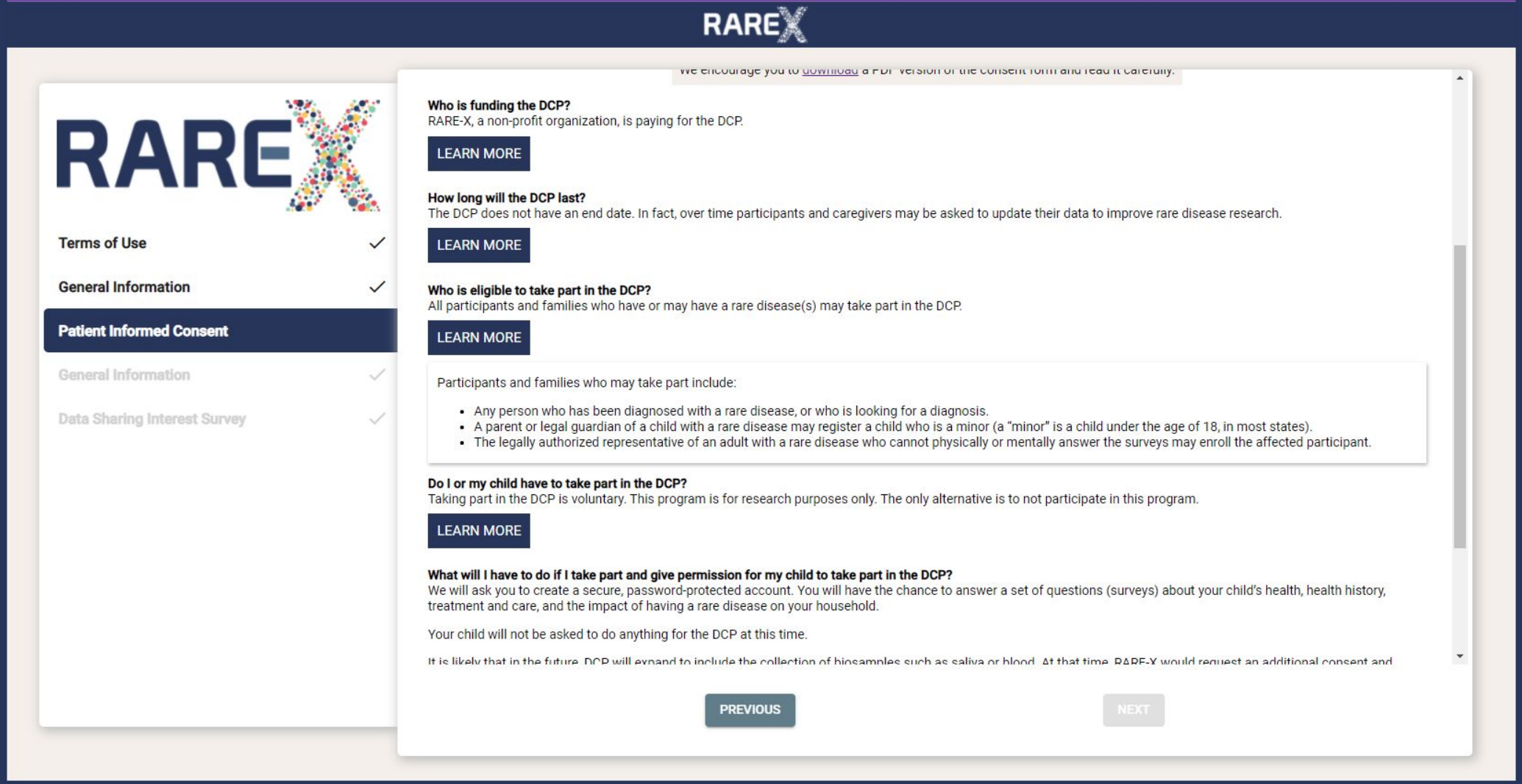
Login to the DCP



Terms of Use


RARE X
Terms of Use
Caregiver Informed Consent
General Information
Haley Jameson
Terms of Use
You must first read, agree to, and accept the following Terms of Use in order to gain access to this software application.
Matrix Terms of Use
Last Updated December 23, 2021
These Terms of Use constitute a legally binding agreement made between you, whether personally or on behalf of an entity ("you") and Across Matrix, Inc. ("Matrix," "we," "us," or "our"), concerning your access to and use of our website and other technologies located at www.acrossmatrix.com as a component of your use of other hosted services (the "Service(s)"). Matrix is providing the Services as a service provider for a third party to whom we provide the Services ("Host Site"). You agree that by
DENY
ACCEPT
PREVIOUS
NEXT

Informed Consent - 8 Pages of Detailed Q&A to Ensure Understanding



Check all that apply

Informed Consent

I am taking part in the RARE-X DCP for one or more of the following reasons (Check all that apply) *

- [ ] You have stated that you have or may have a rare disease.
- [ ] You are the Parent or Caregiver of a person who has or may have a rare disease.
- [ ] You are the legally authorized representative of a person who has or may have a rare disease.
- [ ] You are the family member, other than a parent, caregiver or legally authorized representative of a person who has or may have a rare disease.
- [ ] You have lost a person who had or may have had rare disease.

Check the boxes below to indicate if you agree to the following options. *If you check "no" to any given option, you can still take part in the DCP.*

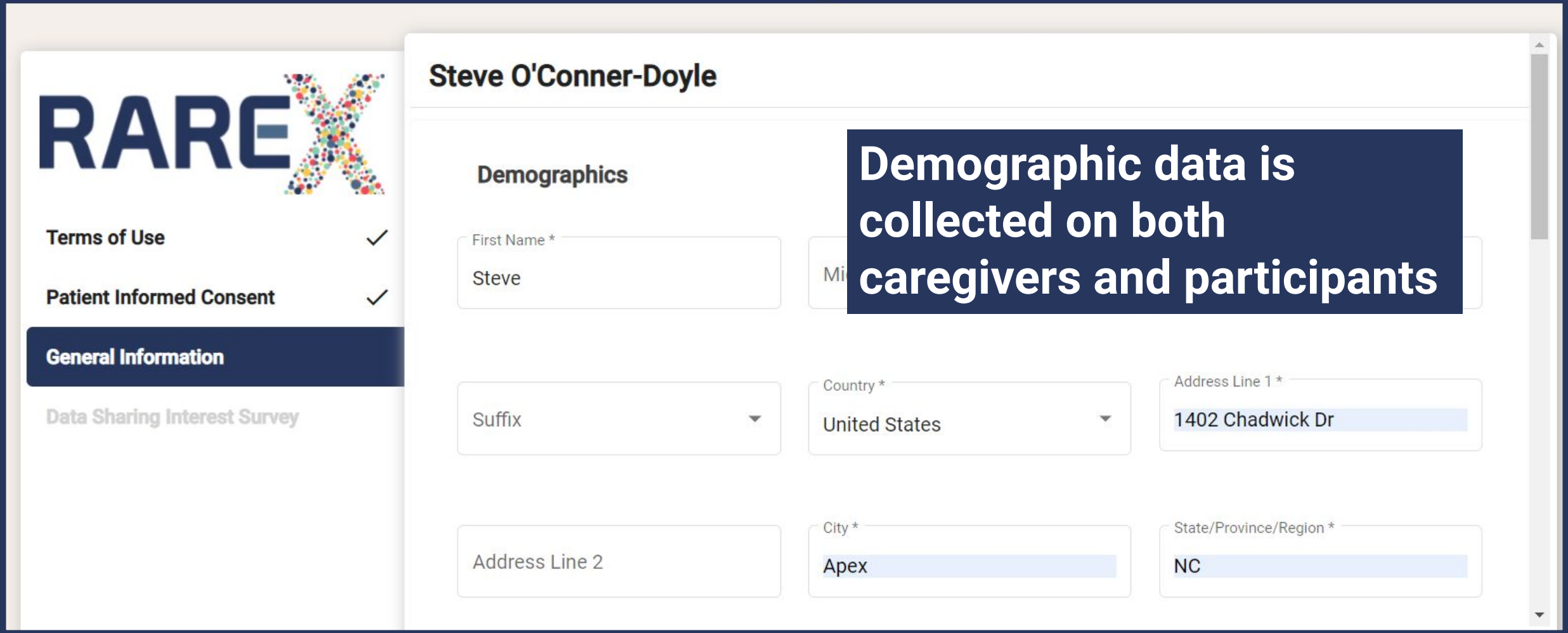
RARE-X may contact me with follow-up research surveys and invitations to take part in additional studies. I may choose to ignore these surveys/invitations. *

- ○ Yes
- ○ No

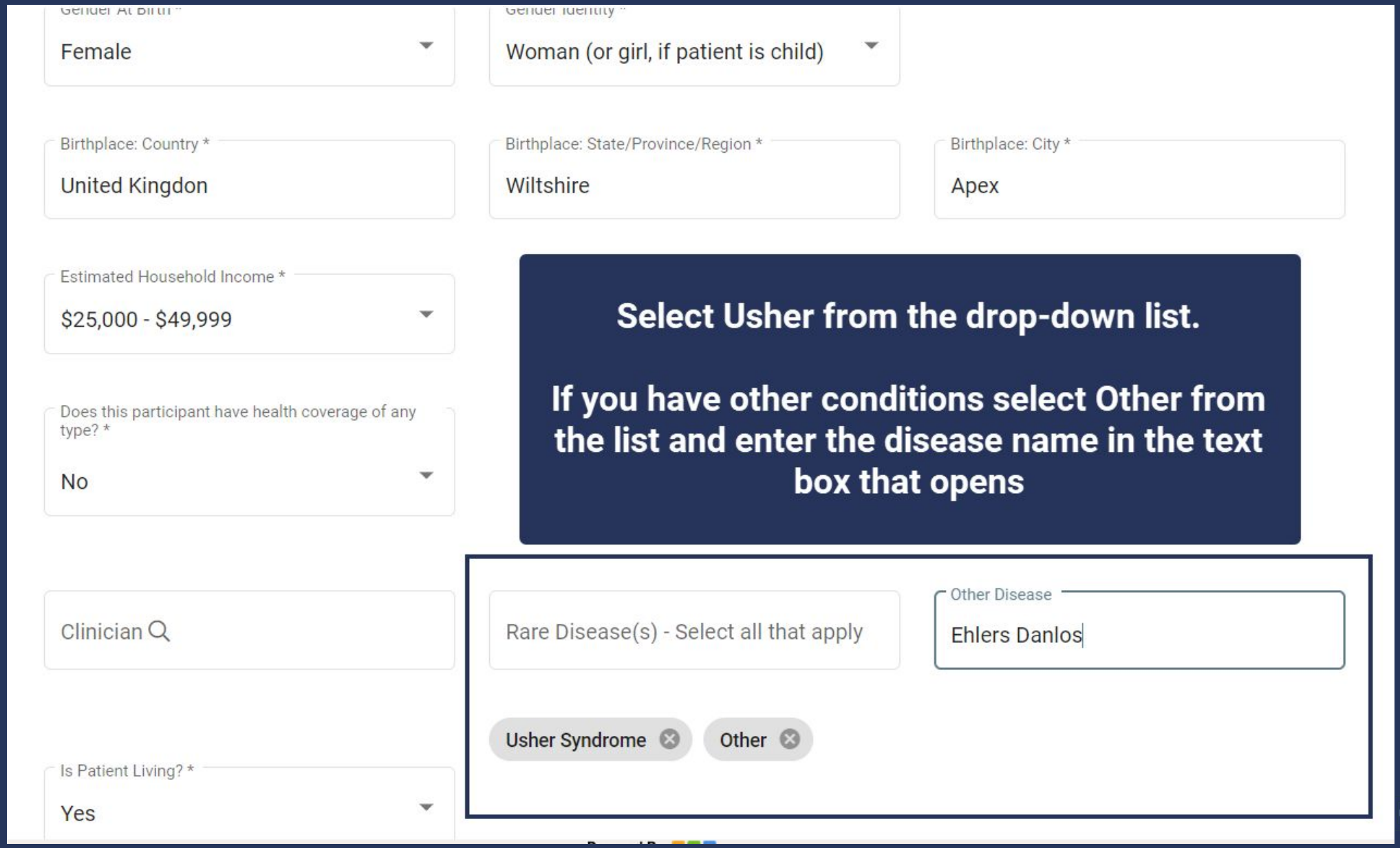
RARE-X or a qualified patient organization may contact me if a researcher thinks that I qualify to be part of a clinical trial/study. *

- ○ Yes
- ○ No

General Information - *Demographics*

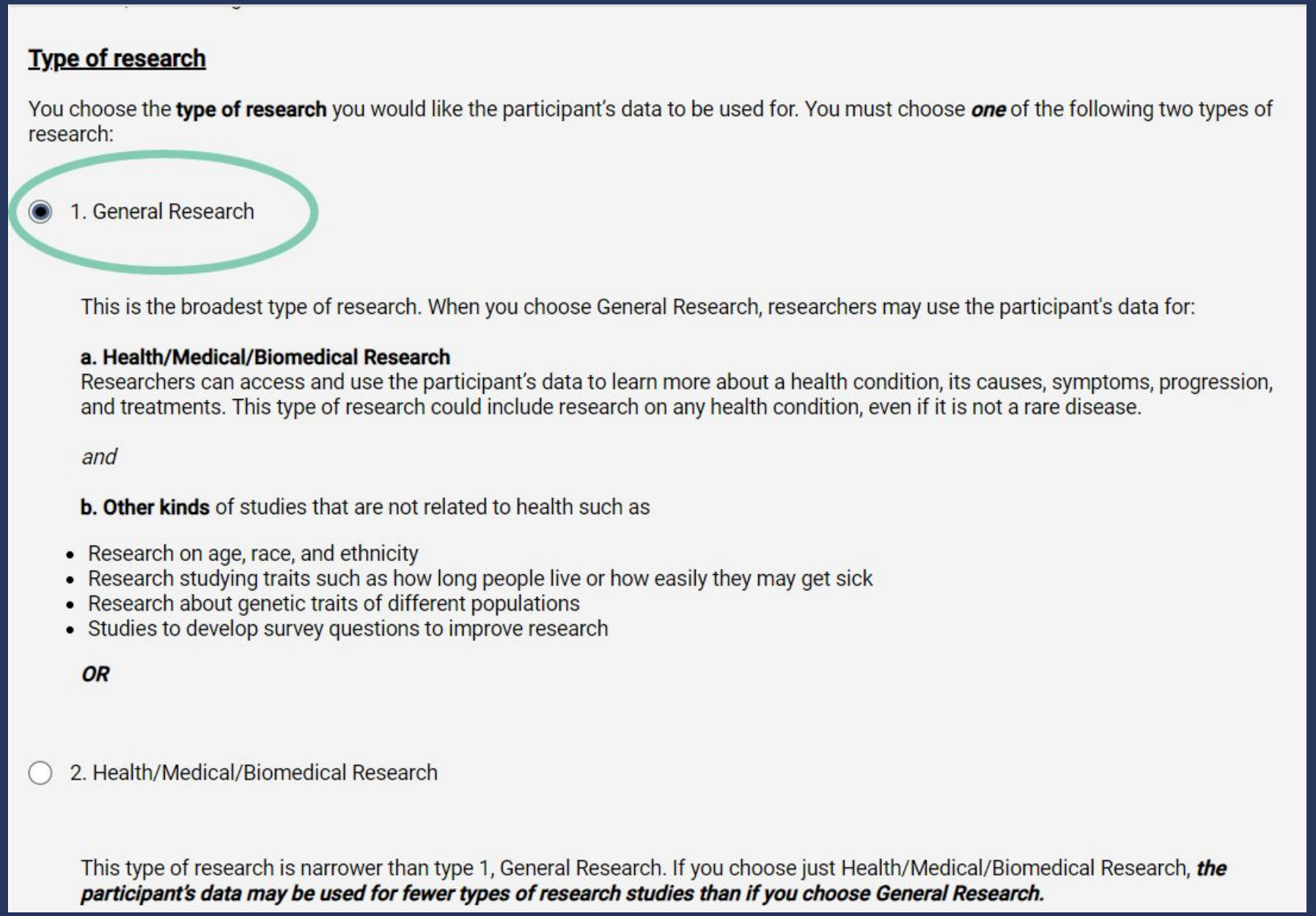


General Information – *Other Information*



Data Sharing Preference Agreement

By selecting ***General Research*** your participant's data will reach the most researchers (recommended)



Type of research

You choose the **type of research** you would like the participant's data to be used for. You must choose ***one*** of the following two types of research:

(●) 1. General Research

This is the broadest type of research. When you choose General Research, researchers may use the participant's data for:

a. Health/Medical/Biomedical Research
Researchers can access and use the participant's data to learn more about a health condition, its causes, symptoms, progression, and treatments. This type of research could include research on any health condition, even if it is not a rare disease.

and

b. Other kinds of studies that are not related to health such as

- Research on age, race, and ethnicity
- Research studying traits such as how long people live or how easily they may get sick
- Research about genetic traits of different populations
- Studies to develop survey questions to improve research

OR

( ) 2. Health/Medical/Biomedical Research

This type of research is narrower than type 1, General Research. If you choose just Health/Medical/Biomedical Research, ***the participant's data may be used for fewer types of research studies than if you choose General Research.***

Data Sharing Preference Agreement

Other Limits on Research - Optional

You do *not* have to put any additional limits on how the participant's data is used for research. If this is your choice, you can stop now and go to the end of this form.

But if you would like, you may choose to further ***limit*** how the participant's data is accessed and used for research. You can select *one* or *both* options below.

☐ Research solely for non-commercial purposes.

If you choose this limit, it means the participant' data may **NOT** be used by any researcher to do studies to develop a drug, treatment, or device that might later be sold to make a profit. For example, if you choose this limitation, a drug development company (biotech or pharmaceutical) would not be allowed to access or use the participant's data for research to develop a drug, treatment, or device that they will sell.

☐ Only research that has been approved by an Institutional Review Board (IRB).

If you choose this limit, it means that only researchers that have had their studies reviewed by an Institutional Review Board (IRB) may access the participant's data for their research. An IRB is a type of committee that reviews research studies and methods to make sure they are not harmful to people. Most of the people who are on an IRB have professional expertise to be able to review the research. The IRB has scientists and nonscientists as part of the committee. When you make this choice, a researcher must present written proof of the IRB's approval, or proof of exemption, of their study before they can access the participant's data for their research.

Page 2 of 3

PREVIOUS NEXT

Data Sharing Preference Agreement

Mary Lucus

GLOSSARY

Data Sharing Interest Survey

RAREX

Biospecimen(s)

Do you know if there are biological samples that you have given for research purposes? *

- ◯ Yes
- ◯ No

Are you interested in the collection of biological samples for research (saliva/spit, blood, bodily fluids, etc)? *
You will be contacted when this option is available.

- ◯ Yes
- ◯ No

RARE-X

Data Sharing Preference Agreement

Medical Records

Are you interested in having your medical records connected to the data you provide on RARE-X? *
You will be contacted when this option is available.

- (●) Yes
- ( ) No

Other Possible Data Sources

We know that you may have participated in other studies/data collection. Are you interested in having the data from those studies connected to your data in RARE-X? *
You will be contacted when this option is available.

- (●) Yes
- ( ) No

Data Sharing Preference Agreement

Patient Community Connections

Do you want to share your contact information with patient advocacy groups that support your diagnosis(s)? *

- (●) Yes
- ( ) No

Contact Information = Name and email only

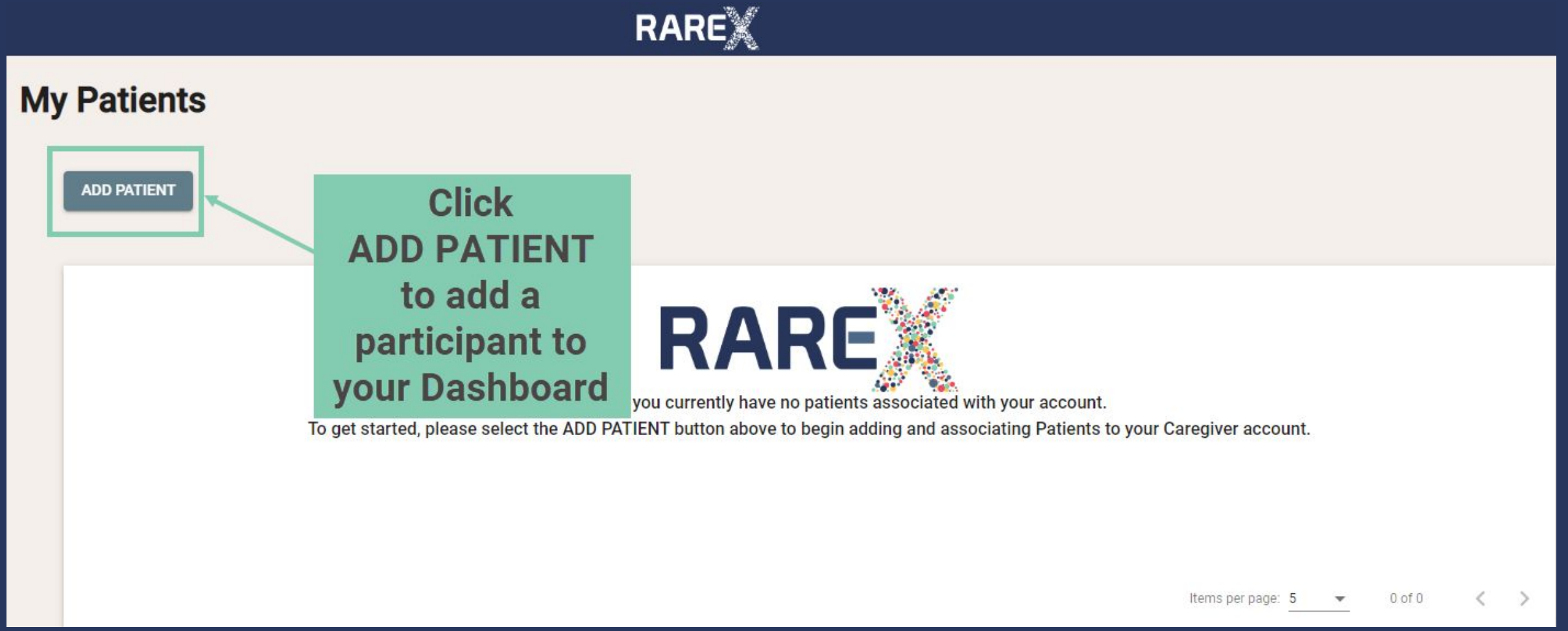
Including Your Data in Summaries on the RARE-X DCP Data Dashboard

We combine data from Participants, remove all identifiers from the combined data, summarize it and present the summary data to others on a "dashboard" that is used to display RARE-X DCP data. This summary might be made available to users of the RARE-X DCP and the general public. May we include your information in the dashboard data? *

- (●) Yes
- ( ) No

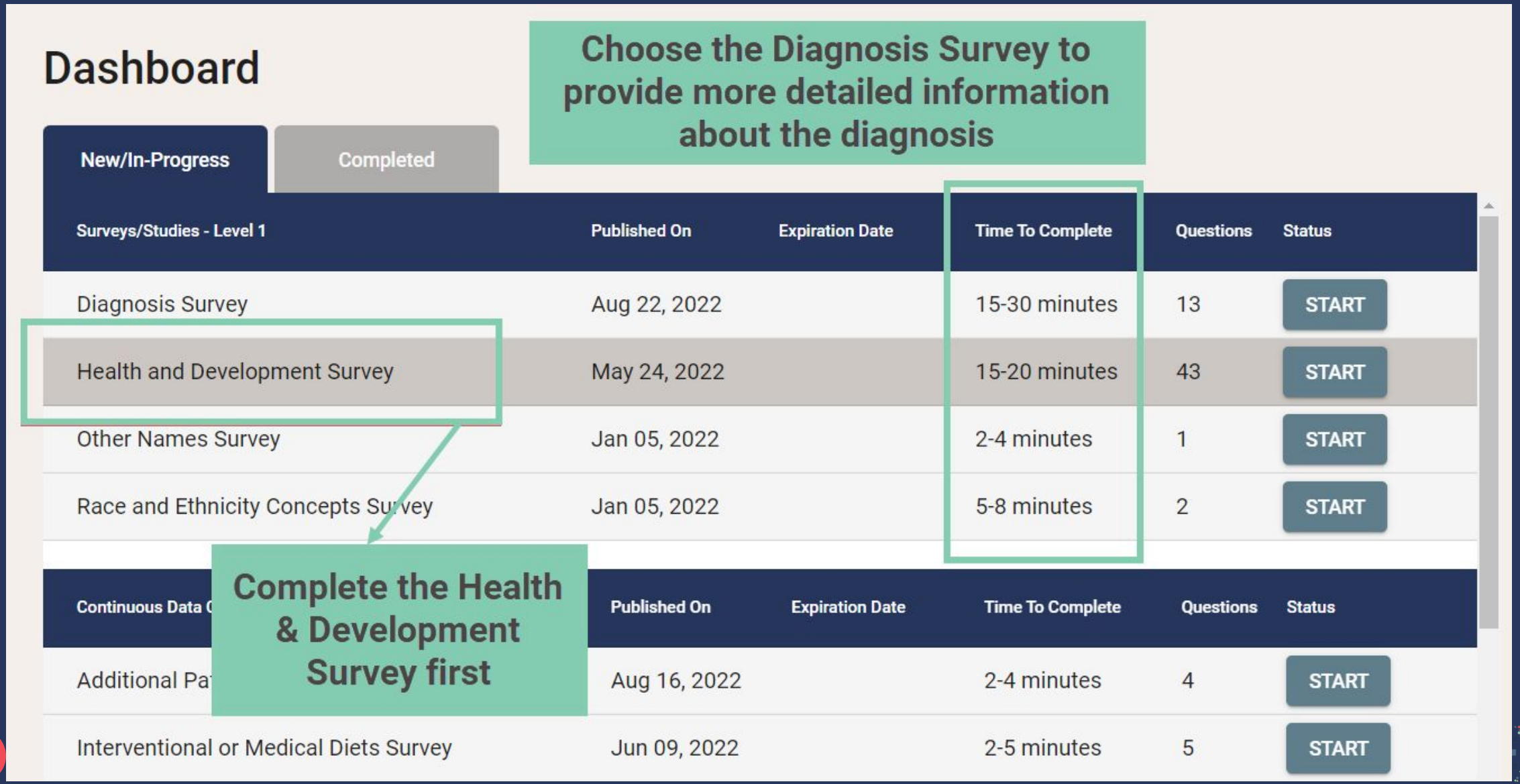
Caregiver Dashboard

Adding a participant to Your Caregiver Dashboard

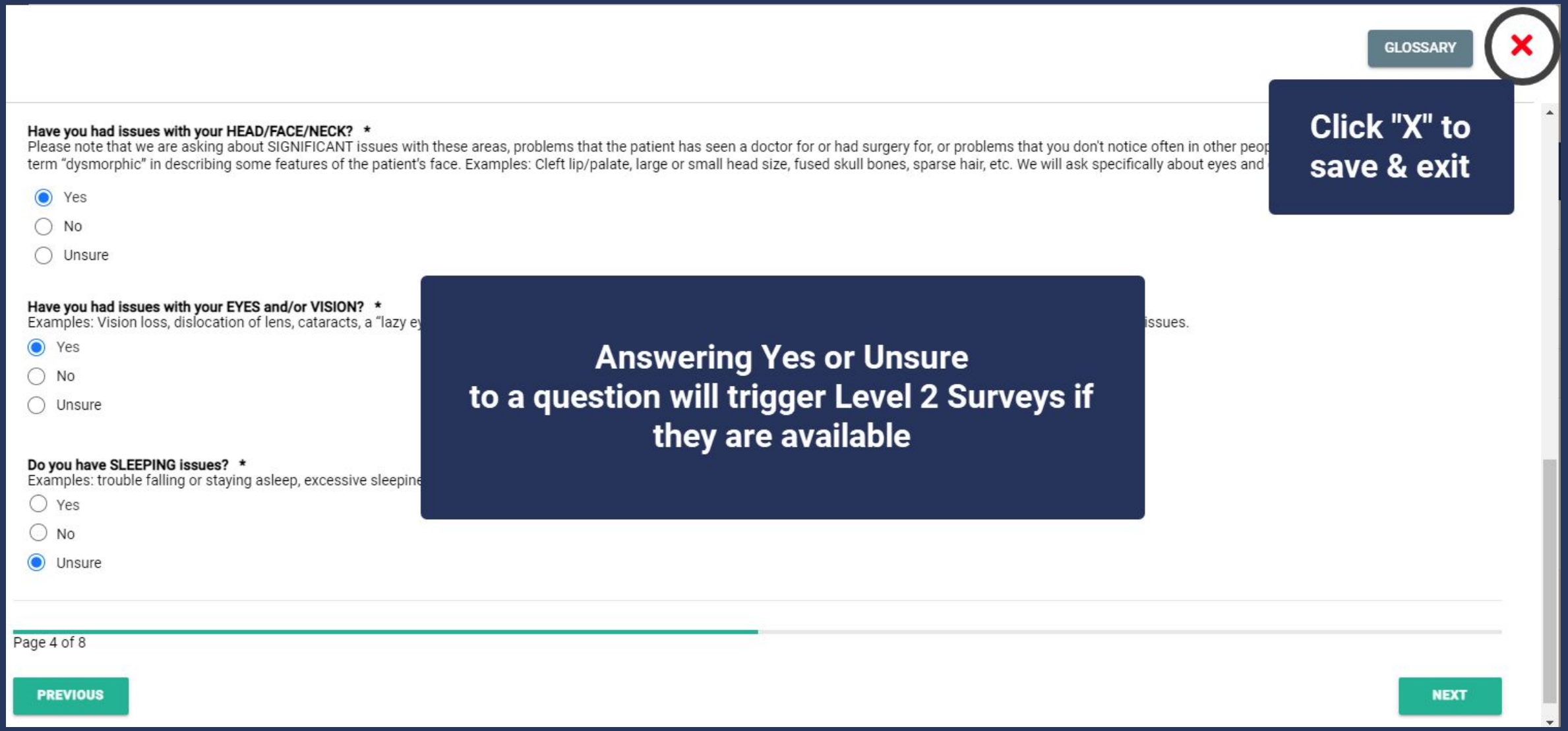


Caregiver Dashboard

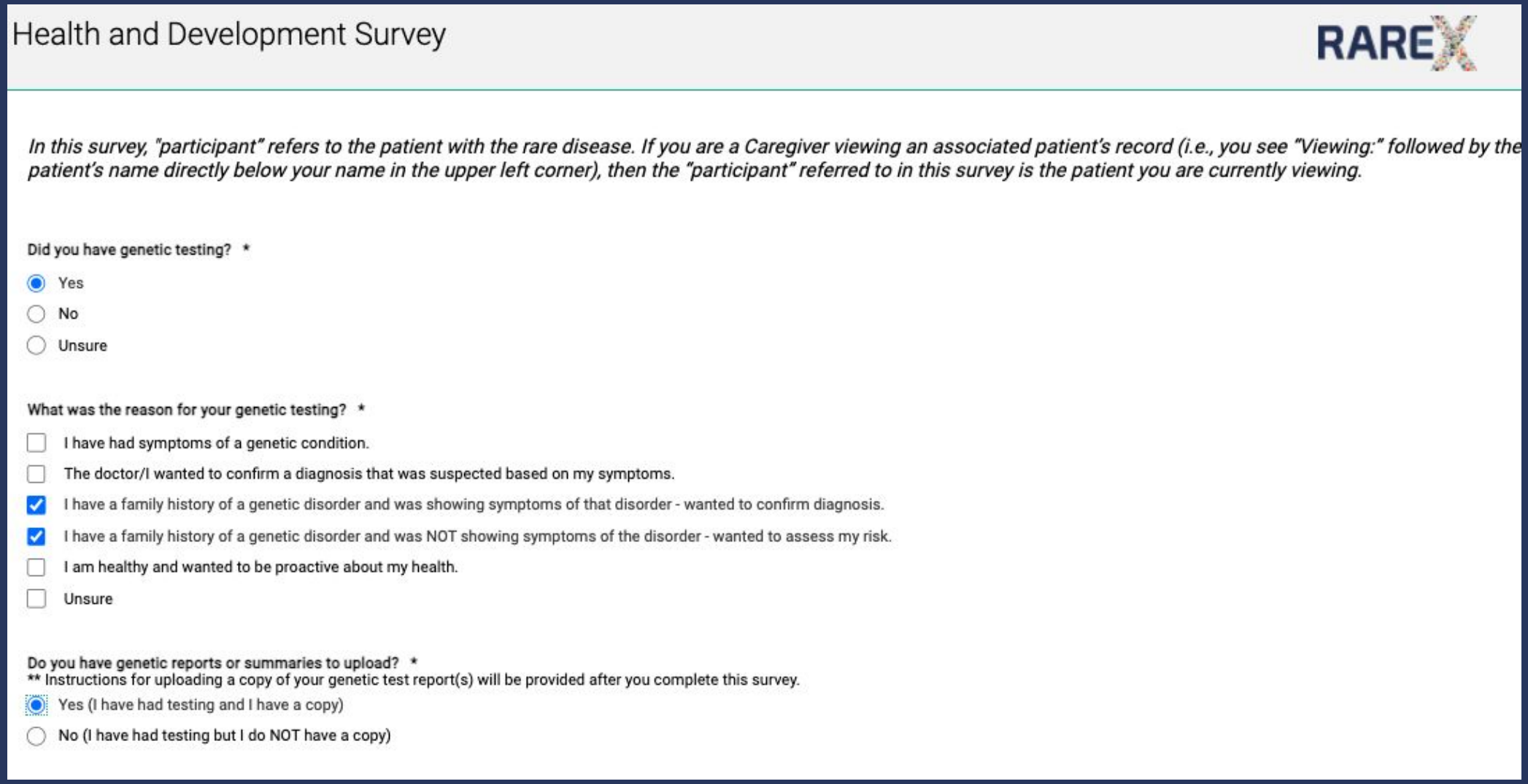
My Dashboard



Health and Development Survey



Genetic Testing Information



Health and Development Survey

In this survey, "participant" refers to the patient with the rare disease. If you are a Caregiver viewing an associated patient's record (i.e., you see "Viewing:" followed by the patient's name directly below your name in the upper left corner), then the "participant" referred to in this survey is the patient you are currently viewing.

Did you have genetic testing? *

- (●) Yes
- ( ) No
- ( ) Unsure

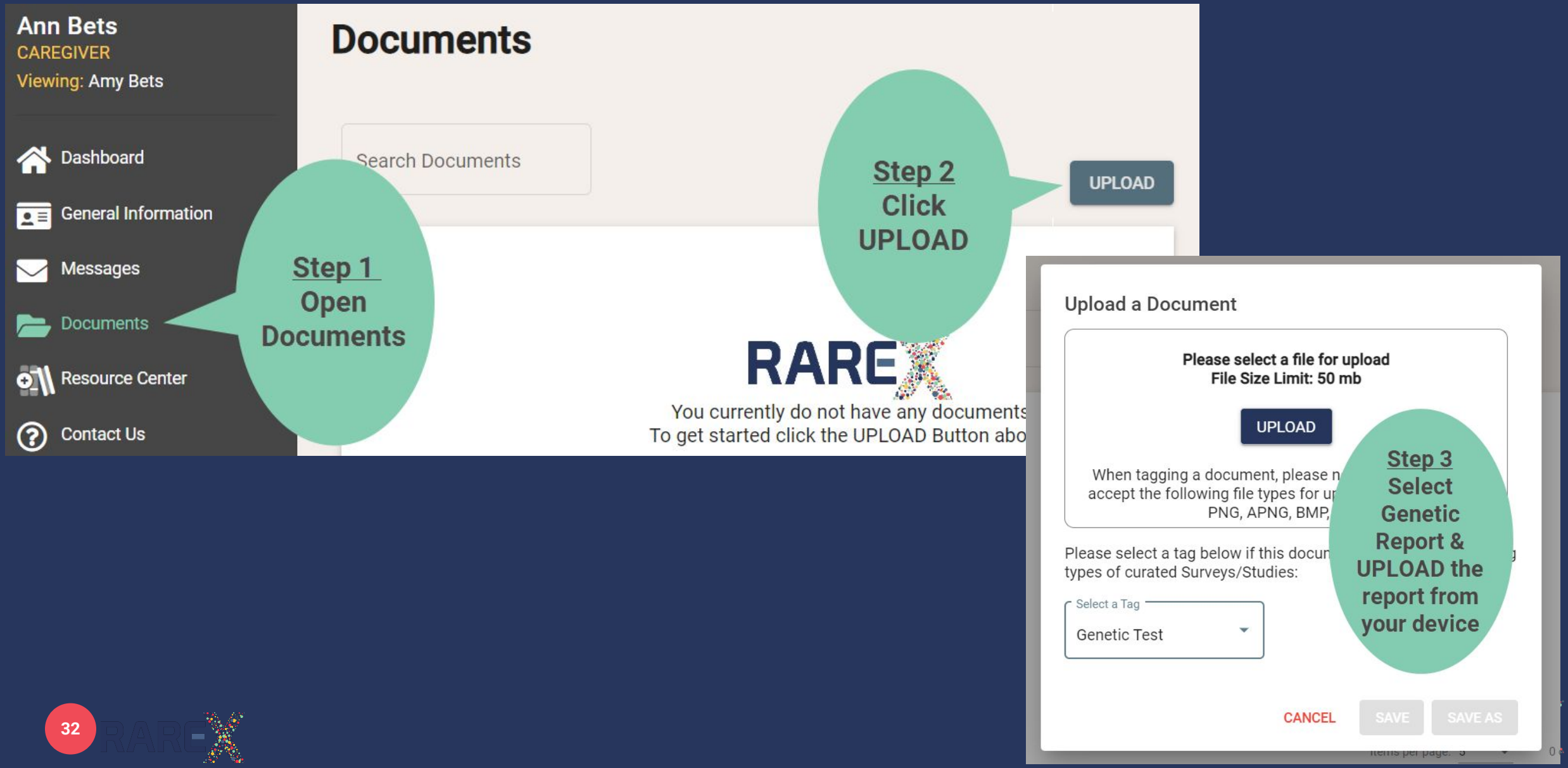
What was the reason for your genetic testing? *

- [ ] I have had symptoms of a genetic condition.
- [ ] The doctor/I wanted to confirm a diagnosis that was suspected based on my symptoms.
- [x] I have a family history of a genetic disorder and was showing symptoms of that disorder - wanted to confirm diagnosis.
- [x] I have a family history of a genetic disorder and was NOT showing symptoms of the disorder - wanted to assess my risk.
- [ ] I am healthy and wanted to be proactive about my health.
- [ ] Unsure

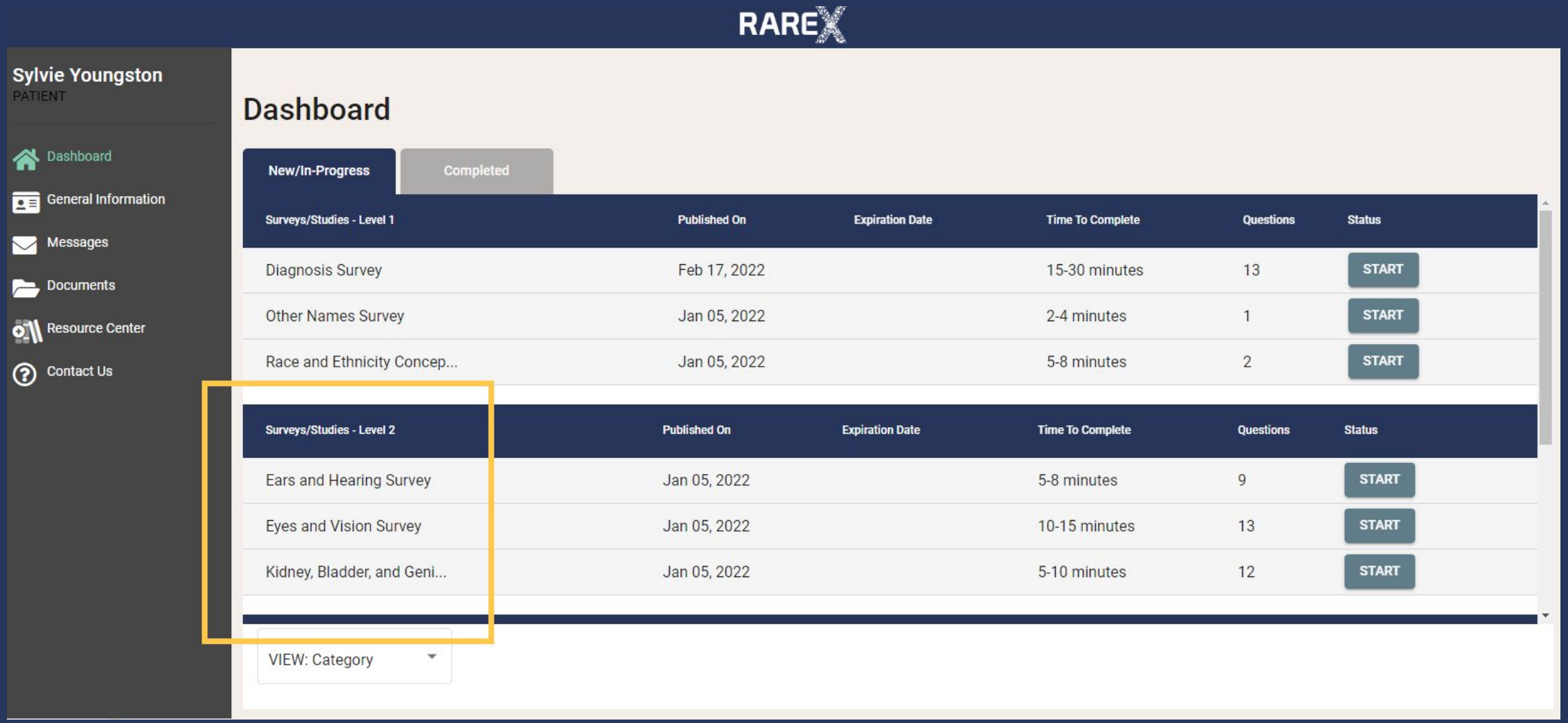
Do you have genetic reports or summaries to upload? *
** Instructions for uploading a copy of your genetic test report(s) will be provided after you complete this survey.

- (●) Yes (I have had testing and I have a copy)
- ( ) No (I have had testing but I do NOT have a copy)

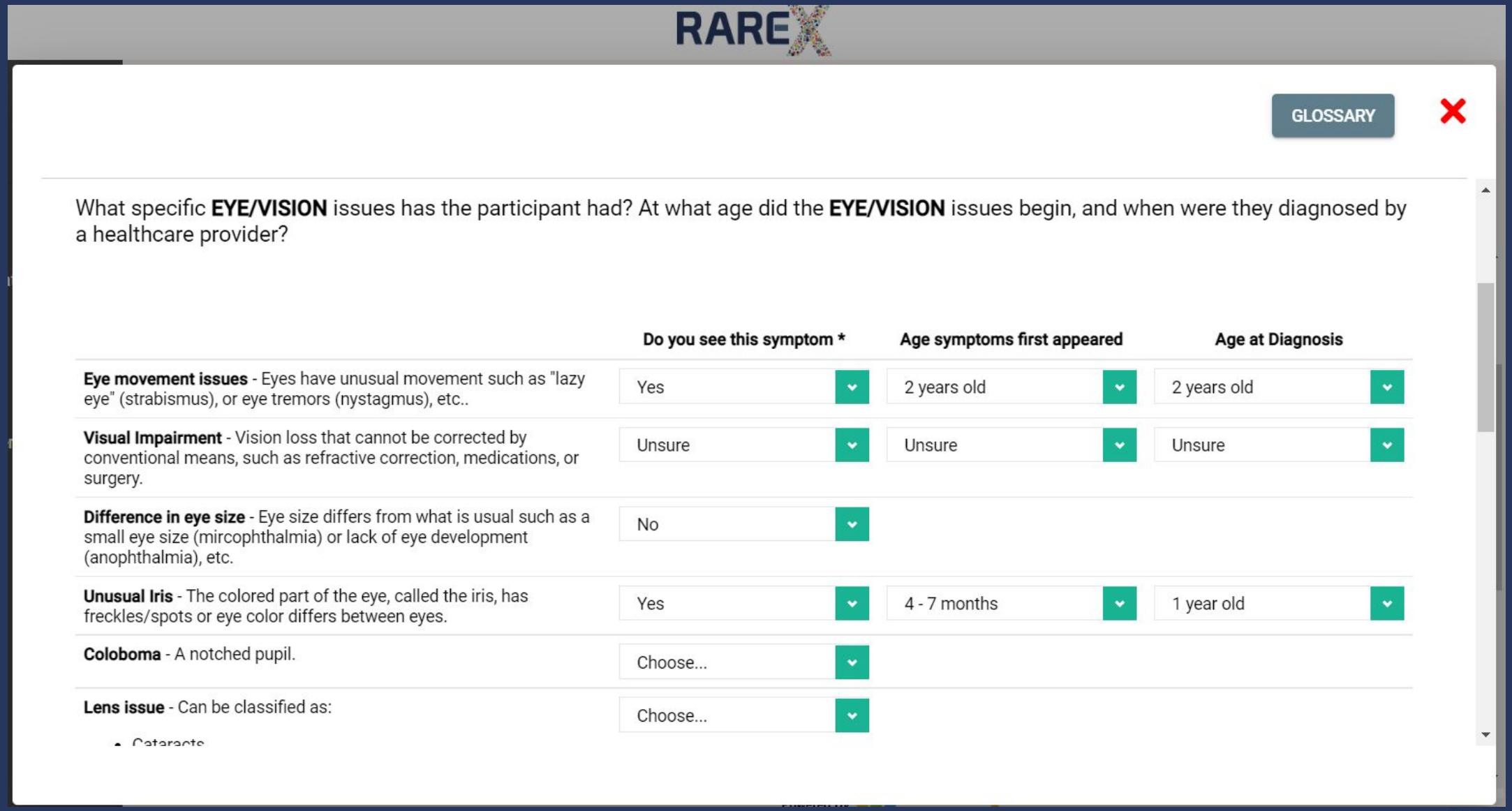
Uploading a Genetic Test Report



Level 2 Surveys



Answering Level 2 Surveys

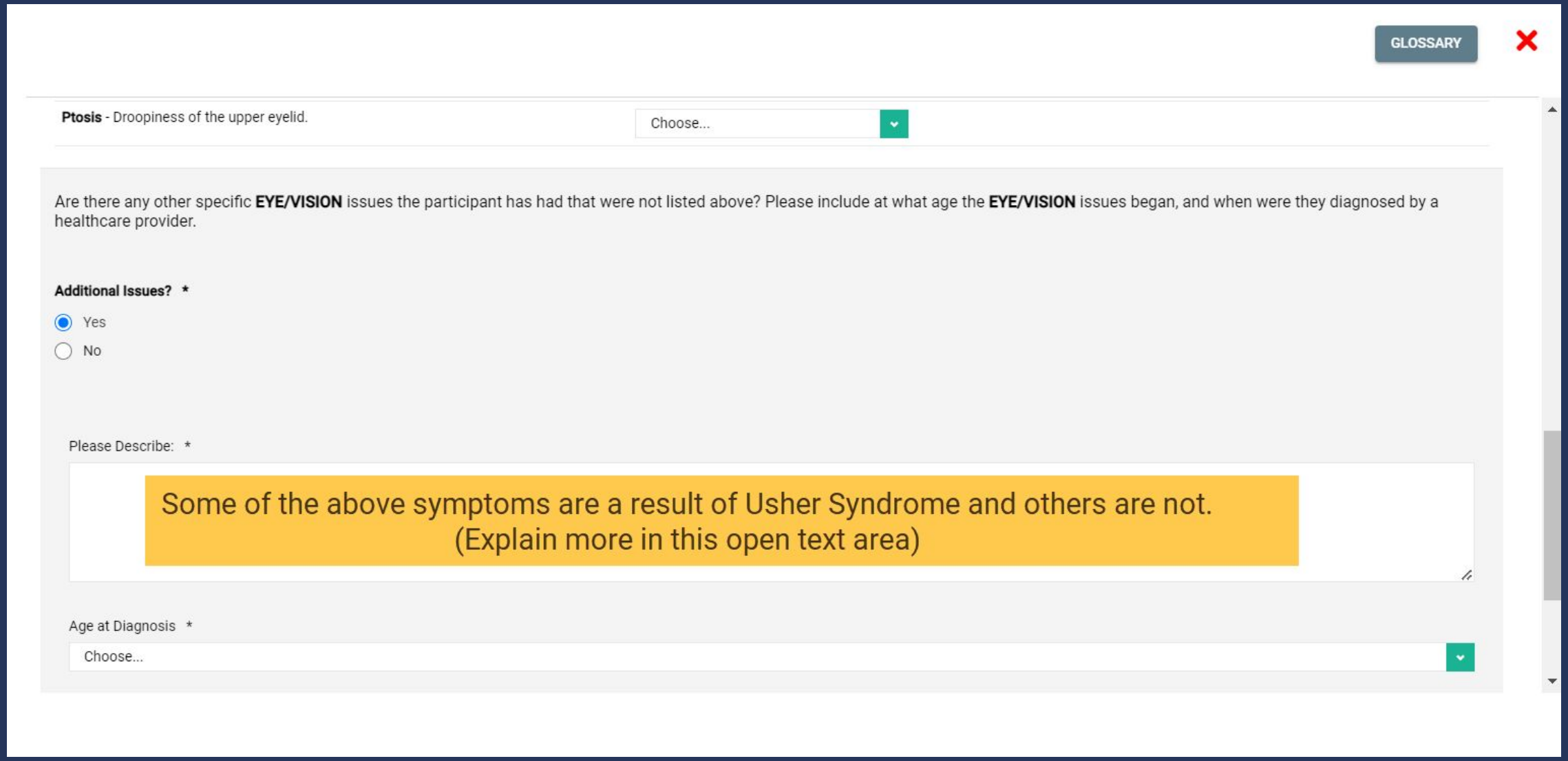


Level 2 - Eyes/Vision Survey (partial)

What specific **EYE/VISION** issues has the participant had? At what age did the **EYE/VISION** issues begin, and when were they diagnosed by a healthcare provider?

	Do you see this symptom *
Eye movement issues - Eyes have unusual movement such as "lazy eye" (strabismus), or eye tremors (nystagmus), etc..	Choose...
Visual Impairment - Vision loss that cannot be corrected by conventional means, such as refractive correction, medications, or surgery.	Choose...
Difference in eye size - Eye size differs from what is usual such as a small eye size (mircophthalmia) or lack of eye development (anophthalmia), etc.	Choose...
Unusual Iris - The colored part of the eye, called the iris, has freckles/spots or eye color differs between eyes.	Choose...
Coloboma - A notched pupil.	Choose...
Lens issue - Can be classified as: • Cataracts • Abnormality of lens shape • Aplasia/Hypoplasia of the lens • Ectopia lentis • Pseudophakia • Phakodonesis	Choose...
Major vision issues - Issues may include blindness, color blindness, night vision problems, floaters, or light sensitivity, etc.. Please do NOT include minor nearsightedness or farsightedness.	Choose...
Farsightedness - Also called hyperopia or hypermetropia; things up close look fuzzy.	Choose...
Nearsightedness - Also called myopia; things in the distance look fuzzy.	Choose...

Level 2 Surveys - Adding Additional Symptoms at the Bottom



Level 2 Ears - Hearing Survey

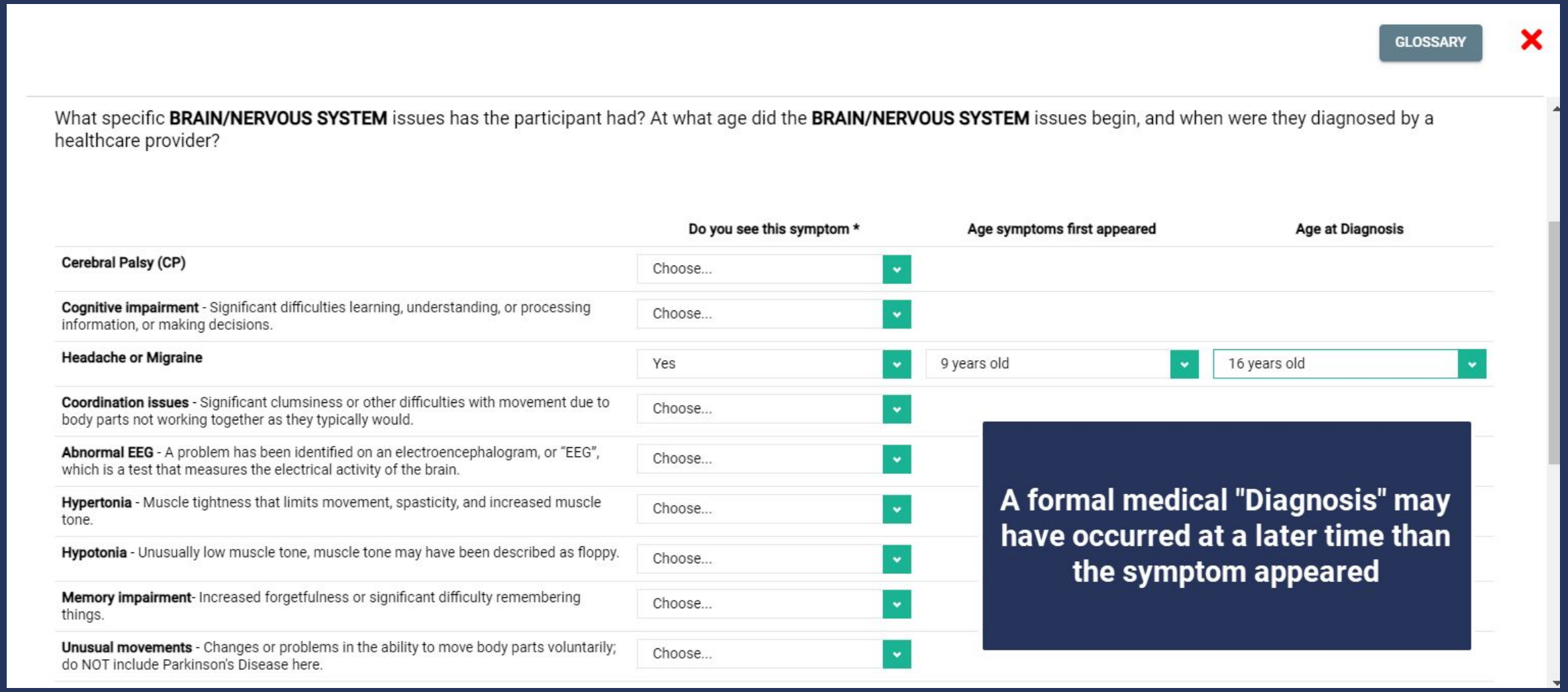
GLOSSARY

What specific **EAR/HEARING** issues has the participant had? At what age did the **EAR/HEARING** issues begin, and at what age were they diagnosed by a healthcare provider?

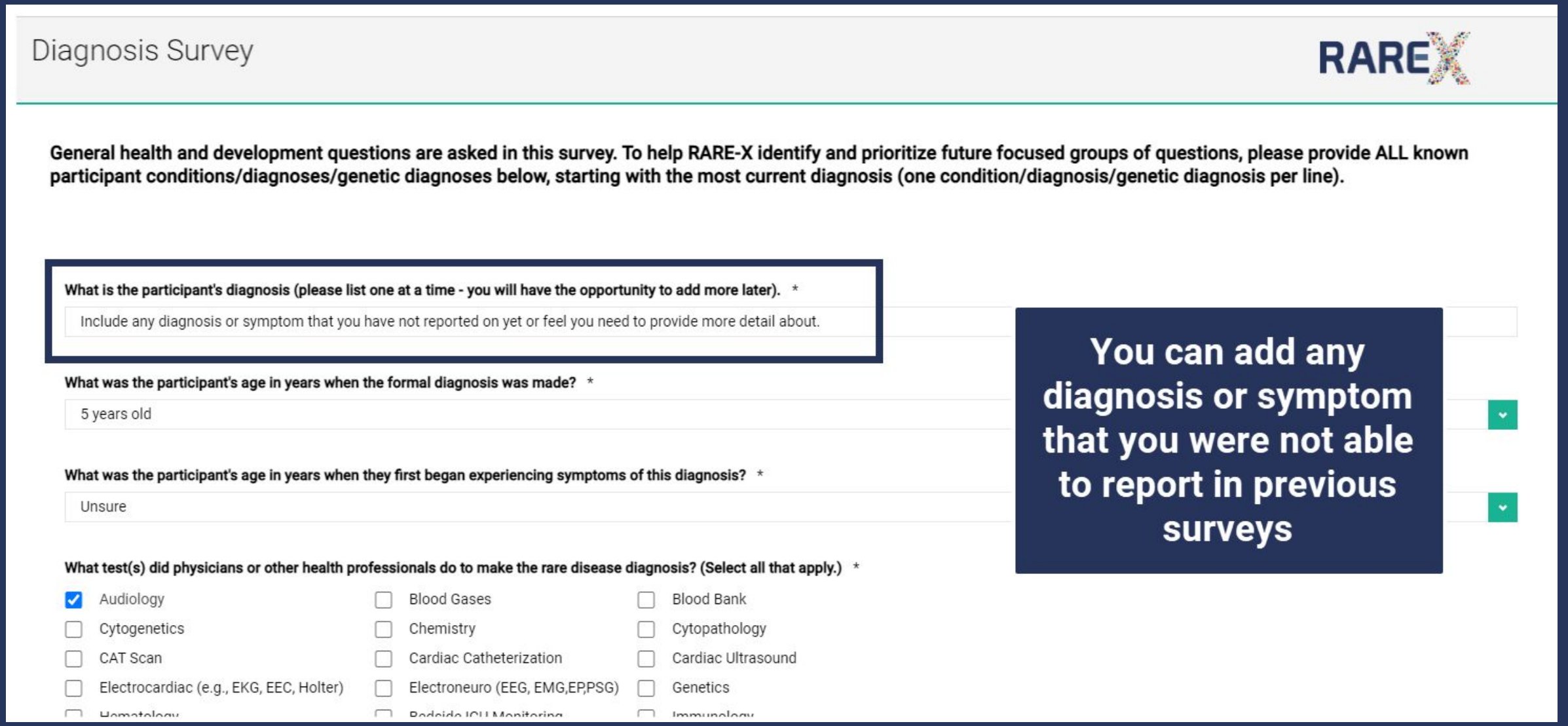
	Do you see this symptom *	Age symptoms first appeared	Age at Diagnosis
Ear shape difference (includes outer or inner ear) - Part or all of the ear may be over or under developed; can include issues such as an ear structure that is missing or underdeveloped, a cupped ear shape, or an overly-large appearance of the ear, etc.	Yes	1 year old	1 year old
Conductive hearing loss - Hearing loss or impairment due to an issue with the ear canal, ear drum, or the bones in the middle ear	No		
Sensorineural hearing loss (SNHL) or impairment - Hearing impairment or loss in one or both ears due to an issue with the nerves in the inner ear or connecting ear to the brain	No		
Mixed hearing loss or impairment - A combination of both conductive and sensorineural hearing loss.	Choose...		
Ringing in the ears - Also called tinnitus.	Choose...		
Hyperacusis - Reduced tolerance and increased sensitivity to everyday sounds in your normal environment.	Choose...		
Vertigo - Abnormal sensation of spinning/dizziness while the body is not moving	Choose...		

Are there any other specific **EAR/HEARING** issues the participant has had that were not listed above? Please include at what age the **EAR/HEARING** issues began, and at what age were they diagnosed by a healthcare provider.

Level 2 Brain - Nervous System Survey



Diagnosis Survey



Quality of Life - Examples of Questions

During the past 30 days, for about how many days have you felt WORRIED, TENSE, or ANXIOUS? *
Number of Days

10

- None
- Don't know/Not sure
- Prefer not to answer

During the past 30 days, for about how many days have you felt you did NOT get ENOUGH REST or SLEEP? *
Number of Days

- None
- Don't know/Not sure
- Prefer not to answer

During the past 30 days, for about how many days have you felt VERY HEALTHY AND FULL OF ENERGY? *
Number of Days

20

- None
- Don't know/Not sure
- Prefer not to answer

Anxiety

In the past 7 days...

I felt fearful... *

- Never
- Rarely
- Sometimes
- Often
- Always
- Prefer not to answer

Are you LIMITED in any way in any activities because of any impairment or health problem? *

- Yes
- No
- Don't know/Not sure
- Prefer not to answer

What is the MAJOR impairment or health problem that limits your activities? *

Eye/vision problem

For HOW LONG have your activities been limited because of your major impairment or health problem? *
Please select below which unit of time your response will be in:

- Days
- Weeks
- Months
- Years
- Don't know/Not sure
- Prefer not to answer

Number of Months *

3

Dashboard – Completed Tab

How is this data collection different from other data collection programs?

- Comprehensive across all body systems
- This is YOUR community's data collection program
- Ability to do cross-disease research
- Return of de-identified summary data to community
- Complement other studies
- Ability to update over years and years - showing progression of disease

https://www.perkins.org/accessibility-statement/

Use TalkBack to browse the web with Chrome

You can get spoken feedback when you use TalkBack in the Chrome browser.

To learn more, complete the TalkBack tutorial.

Contents

Explore pages with reading controls

To cycle through the reading controls:

1. In Chrome, open a page.
2. Swipe down then up, or up then down, until you reach the setting that you want.
   - You can choose a setting like **Headings**, **Links**, or **Words**.
   - On devices with multi-finger gestures, you can also three-finger swipe.

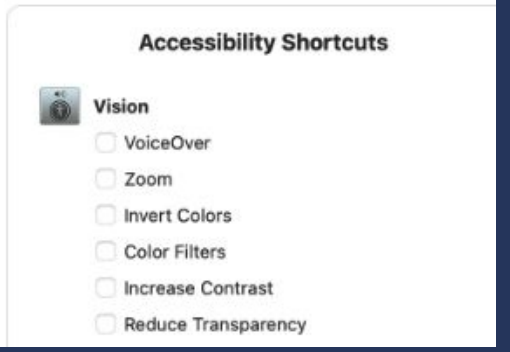
Use the Accessibility Shortcuts panel on Mac

The Accessibility Shortcuts panel offers shortcuts to quickly turn on or turn off common accessibility features such as Zoom, VoiceOver, and Sticky Keys.

Open the Accessibility Shortcuts panel

Press these keys together: Option (⌥), Command (⌘), and F5. On laptop computers with a Touch Bar, you might need to press the Fn (Function) key as well.

On laptop computers with Touch ID, triple-press Touch ID. If you press and hold the Command key while doing this, VoiceOver turns on or off.



Accessibility— for everyone

Windows 11 built-in accessibility features empower every person to discover and do the things they love.

If you have any questions, experience and technical issues, or want to provide any feedback please send an email to:

support@rare-x.org

and cc: n.odonnell@usher-syndrome.org

Thank you!

Questions?