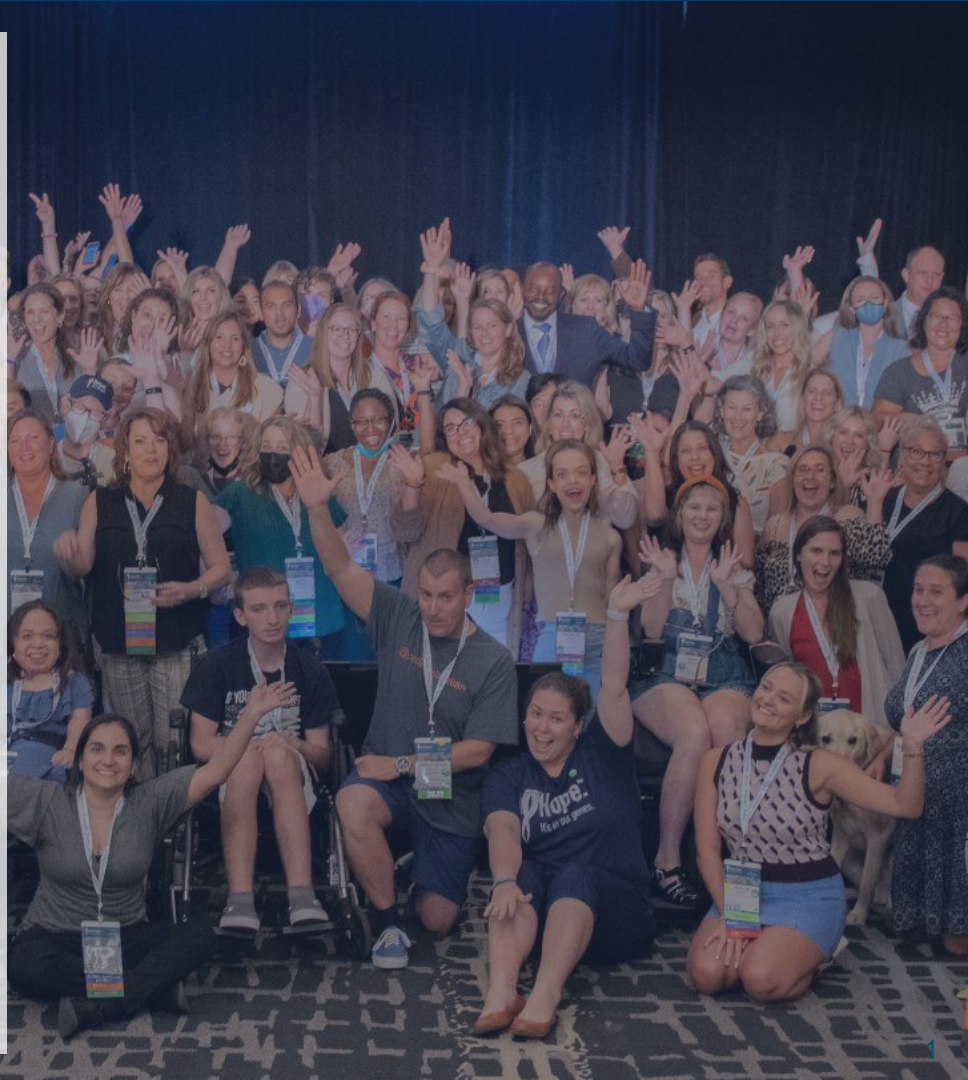
The DESSH Foundation

Global Genes®
Allies in Rare Disease

RARE-X

DeSanto-Shinawi Syndrome (DESSH) Data Collection Program

November 18, 2023



What Is RARE-X?

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

RARE-X is **not** a replacement for any current research or clinician-sponsored patient registries.

What Are We Solving For?

The speed and productivity of innovation in rare disease is limited by cost and lack of access to standardized, structured patient data

Data exists, but is captive within silos

Data is not in a structured, standardized format that is useful to research/patient communities

Data doesn’t yet exist; most groups don’t have the resources to collect data properly

Why did DESSH Leaders Choose RARE-X?

- GLOBAL data collection
- Patient owned and managed - not organization managed
- No cost to patients or organizations
- No cost to researchers (scientists/pharma)
- Structured, standardized Q&A
- Governance is handled
- Streamlines researcher access to data - de-identified data is made available in Federated Data Access Platform
- Speeds up research and drug development
- Ability to connect to existing data sources

The DESSH Foundation

+

RARE-X

What is the benefit for patients/caregivers/families?

- Collecting data on ALL systems of the body will allow a better understanding of the disease
- Summary data returned to the community so you can compare your symptoms to others with the same disease
- The chance to participate in clinical trials
- Ability to update a change in symptoms at any time
- Reach more researchers worldwide
- Ability to manage who uses your data

Patient Journey in the Data Collection Portal (DCP)

Patient Community

Community Page Get Started

Welcome
Privacy Policy
Terms of Use

Enroll in DCP

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

Matrix Terms of Use

Patient Consent & Data Sharing Preference Survey

Patient Dashboard

Potential Survey Topics (Domains)

- General Info
- Gen Medical
- Neuro Gen
- Quality of Life
- Eye
- Kidney
- Heart
- Lung
- Skin
- Endocrine
- Med Usage
- Ear

How To Access Data Collection Program

DESSH HOME GETTING STARTED FAQ Powered by RAREX

DESSH - Data Collection Program

DeSanto-Shinawi Syndrome (DESSH) patients, families, and communities are excited to participate in data collection to expand and improve medical research. By coming to this site, you can begin the first step in making your patient information available to researchers. By generating the most comprehensive DeSanto-Shinawi Syndrome (DESSH) Data Collection Program, we can accelerate research and the development of new drugs, devices, or other therapies. Only you hold the key to unlock future discoveries.

Start Your Journey — GET STARTED

Already Enrolled? — LOGIN

The DESSH Foundation

Why Should You Participate?

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

What Do You Need To Get Started?

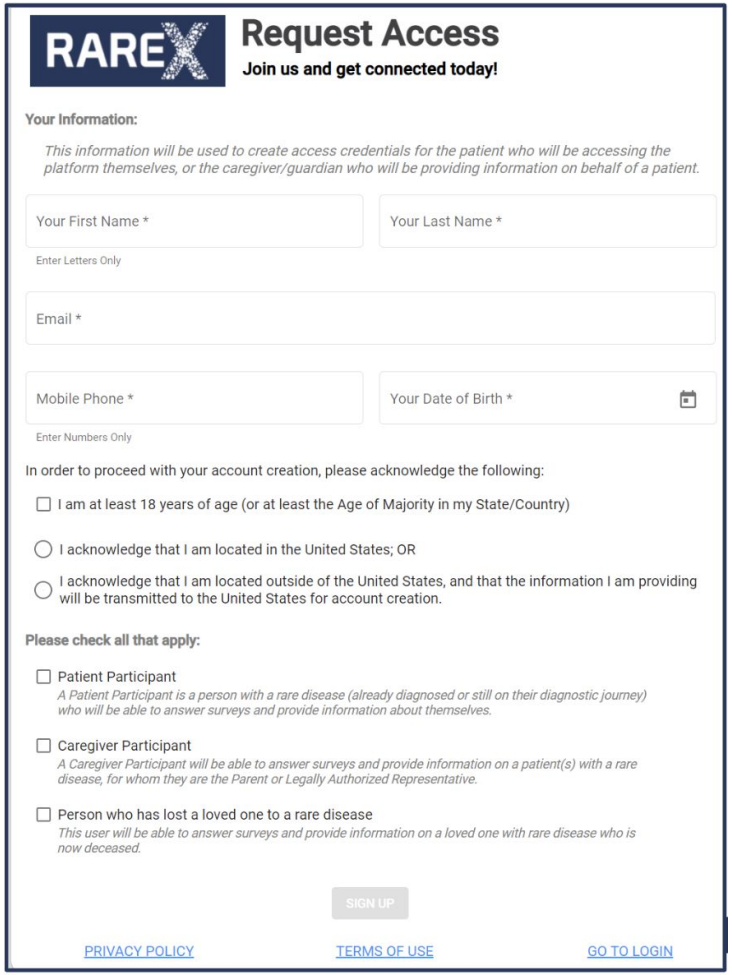
- Email address and Create a password
- An approved Browser
  - Google Chrome, or
  - Apple Safari version 14 or higher
  - Microsoft Edge
- Do not use an unapproved Browser
  - Internet Explorer
  - Mozilla
- What you do NOT need to get started?
  - A Lot of Time
  - To Finish It All At Once

First-time Login Page

(Pre-Qualifications Page)

Caregivers should also check the "Patient Participant" box if:

- **they are diagnosed with the disease**
- **they are a "carrier" of the disease**

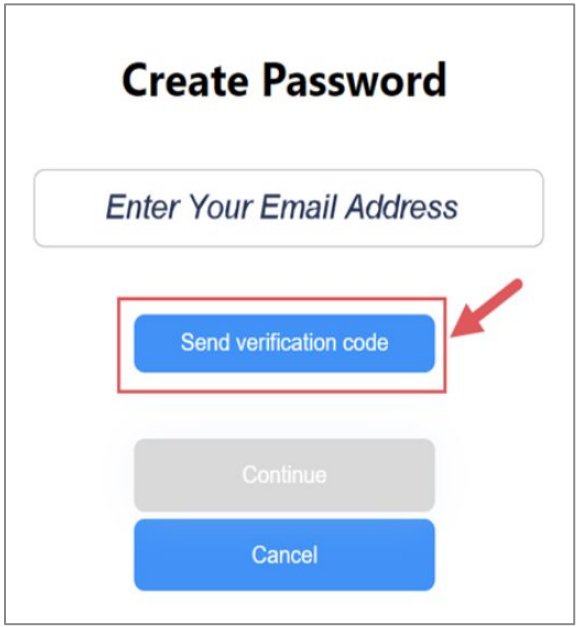


Receive Account Creation Email

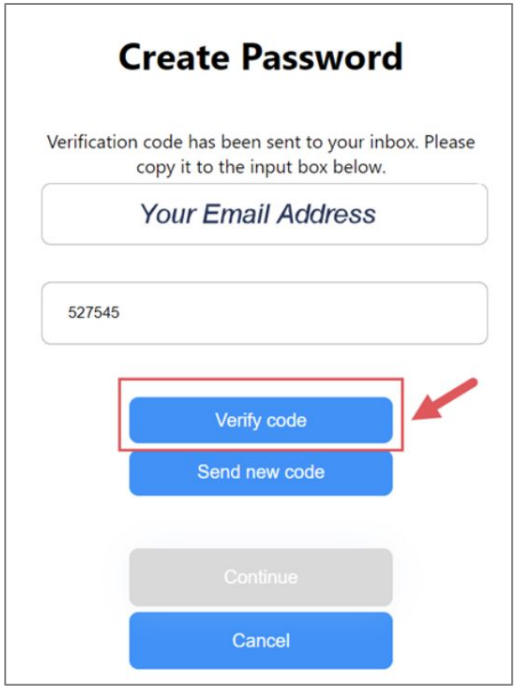
If you do not receive a response within a few minutes, check your spam/ junk folder

Email Verification (multi-factor for your privacy)

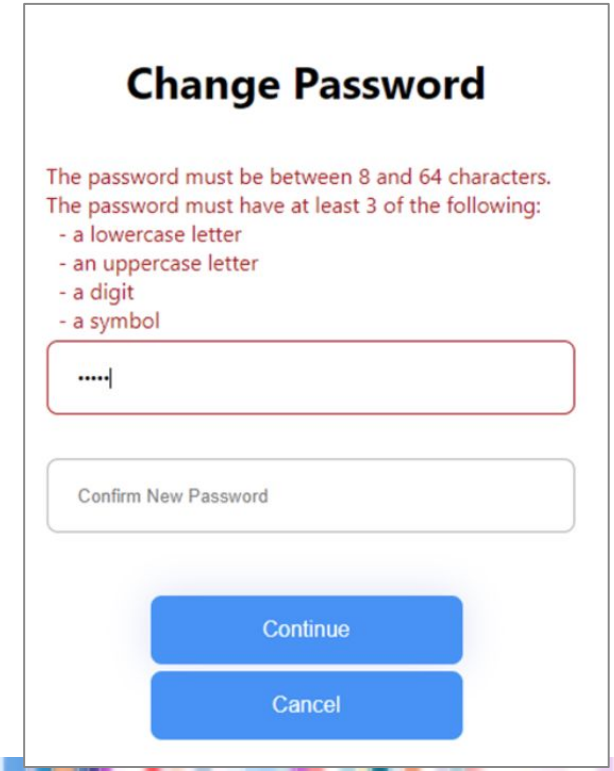
1. Request your verification code



2. Confirm your verification code

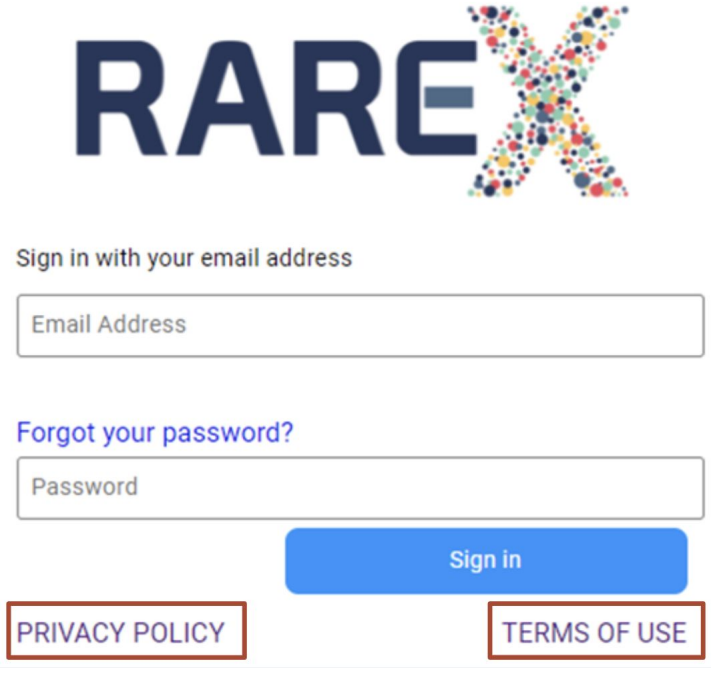


3. Create your password



Privacy Policy & Terms of Use for RARE-X

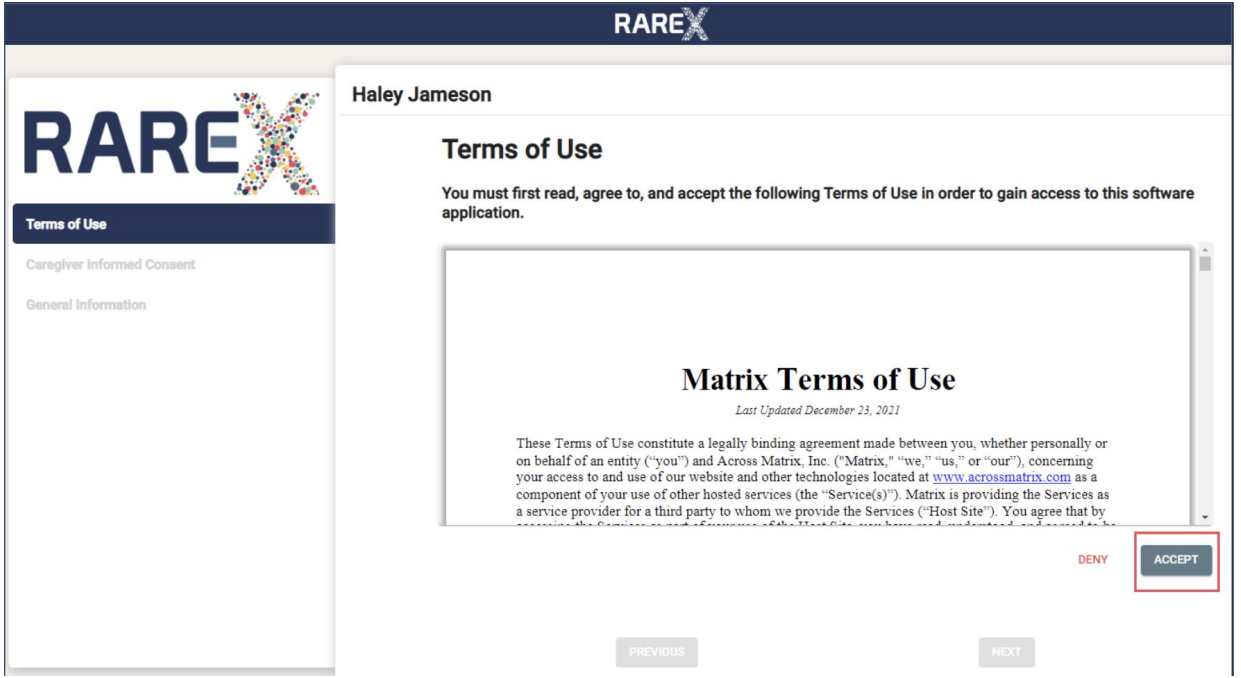
Login to the DCP



You can access these documents anytime by clicking on them

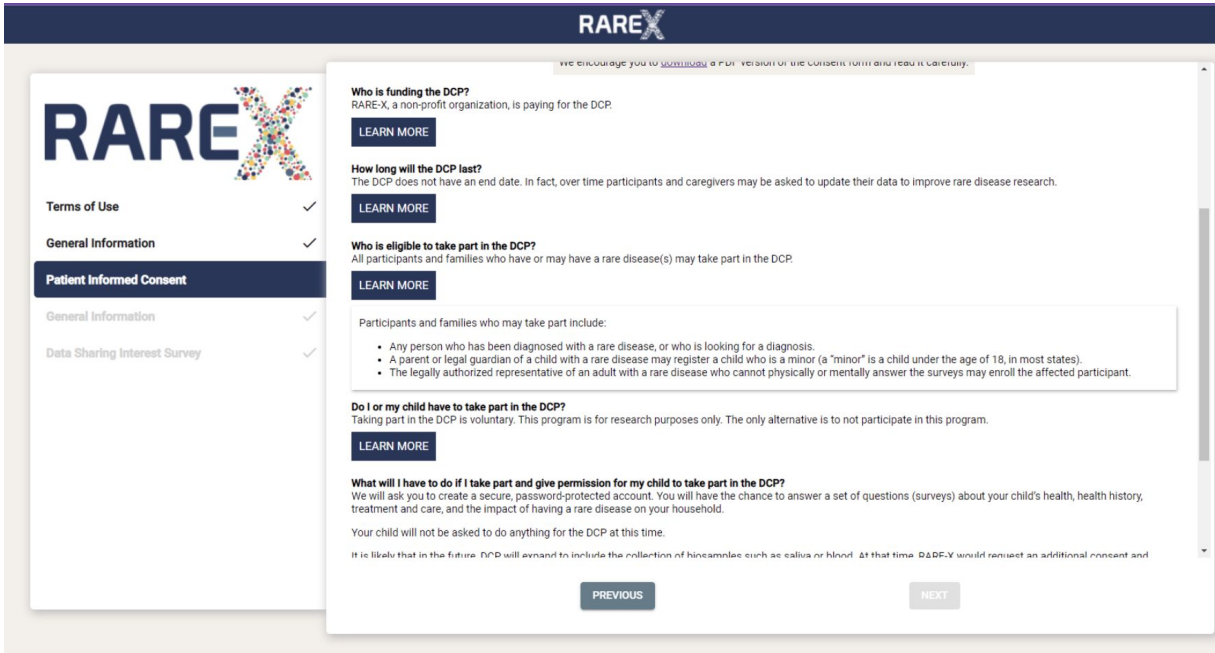
Technology Platform

Terms of Use



Informed Consent

8 Pages of Detailed Q&A to Ensure Understanding



Informed Consent

Check all that apply

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. *

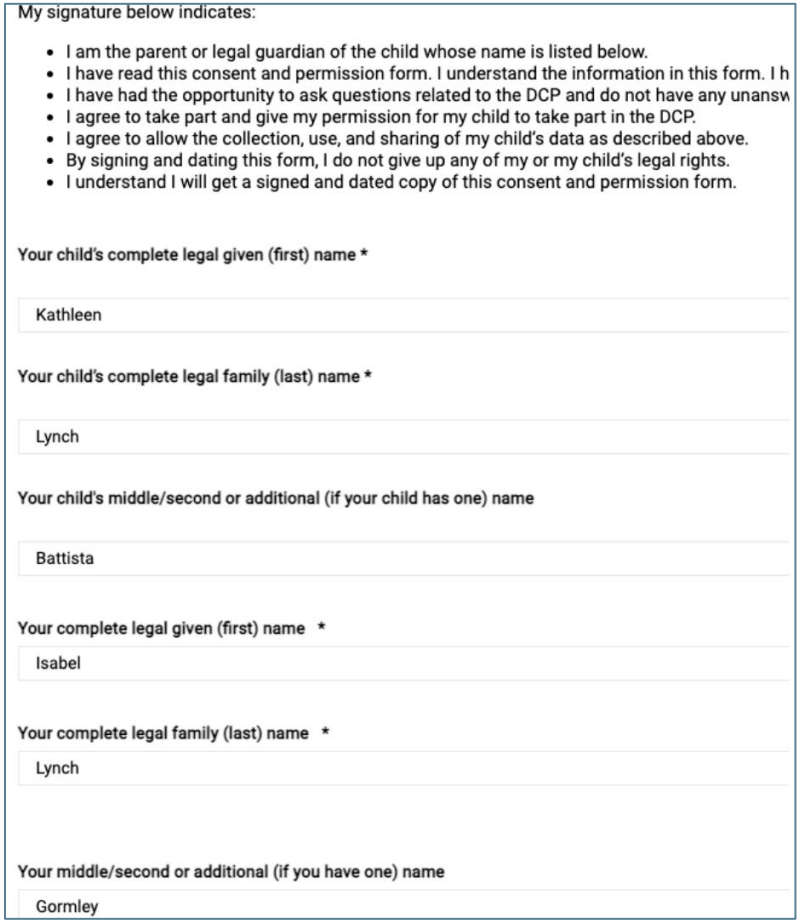
- ◯ Yes
- ◯ No

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

Informed Consent

If you are a caregiver of a patient, be sure to provide your info and the child's info in the correct places



My signature below indicates:

- I am the parent or legal guardian of the child whose name is listed below.
- I have read this consent and permission form. I understand the information in this form. I h
- I have had the opportunity to ask questions related to the DCP and do not have any unansw
- I agree to take part and give my permission for my child to take part in the DCP.
- I agree to allow the collection, use, and sharing of my child's data as described above.
- By signing and dating this form, I do not give up any of my or my child's legal rights.
- I understand I will get a signed and dated copy of this consent and permission form.

Your child's complete legal given (first) name *

Kathleen

Your child's complete legal family (last) name *

Lynch

Your child's middle/second or additional (if your child has one) name

Battista

Your complete legal given (first) name *

Isabel

Your complete legal family (last) name *

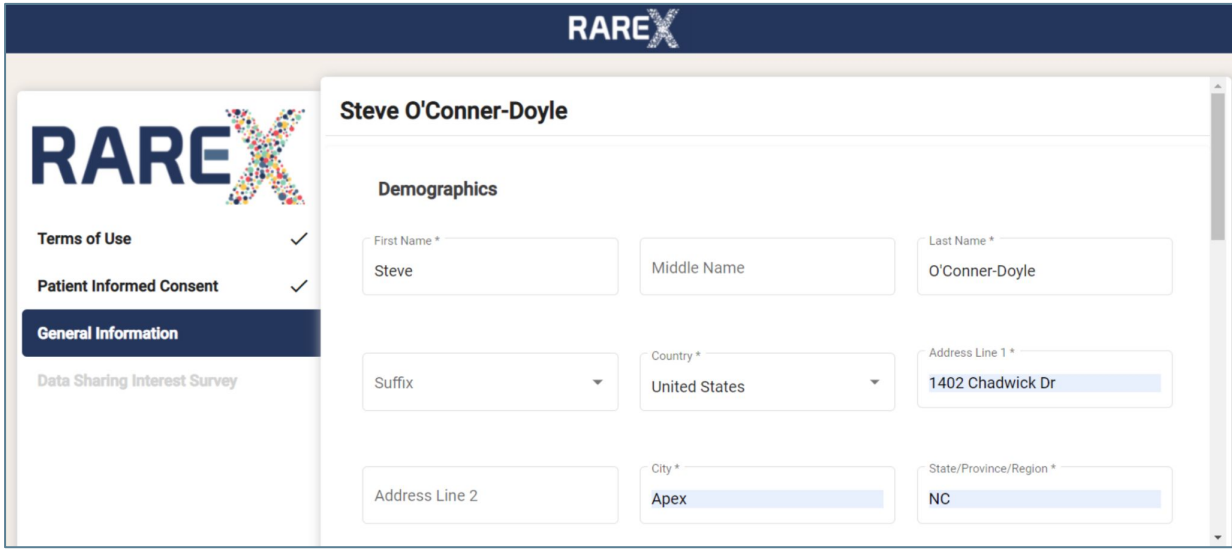
Lynch

Your middle/second or additional (if you have one) name

Gormley

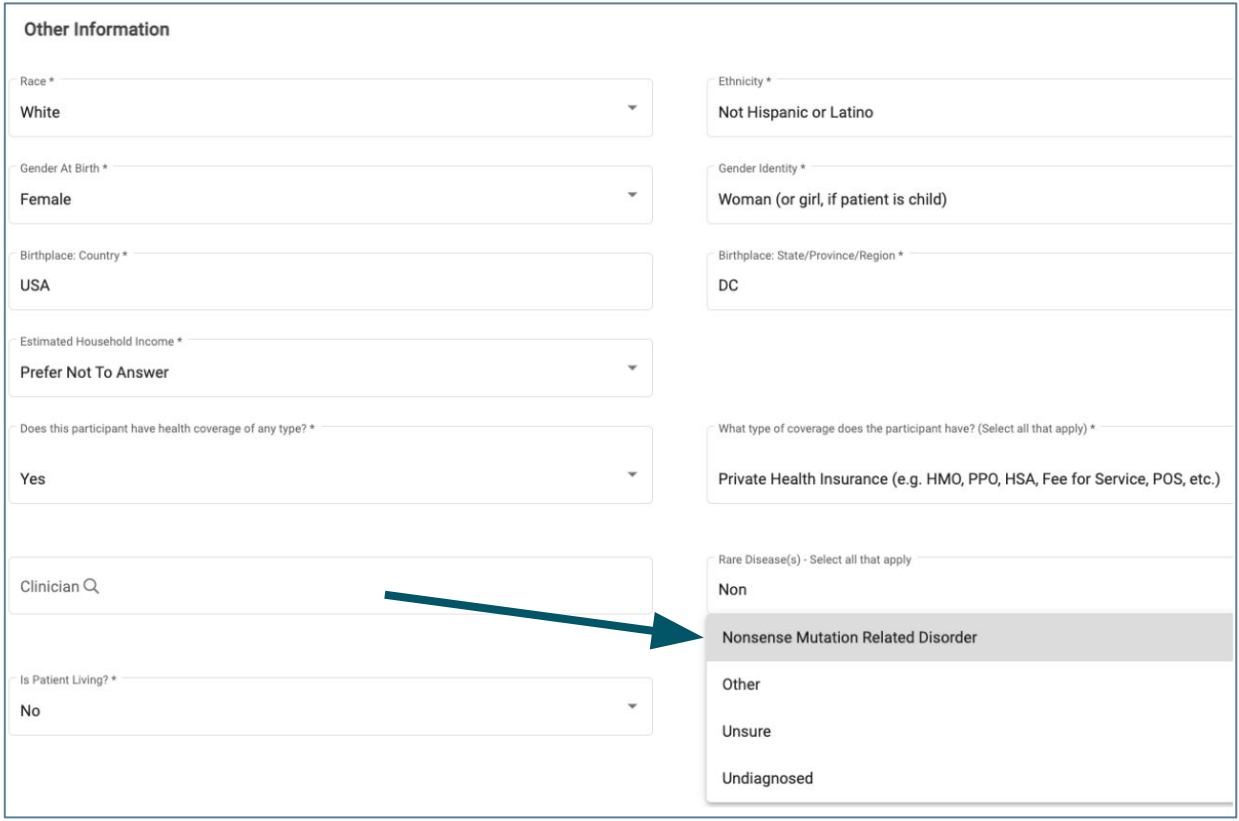
General Information *Demographics*

Demographic information collected on both patients and caregivers



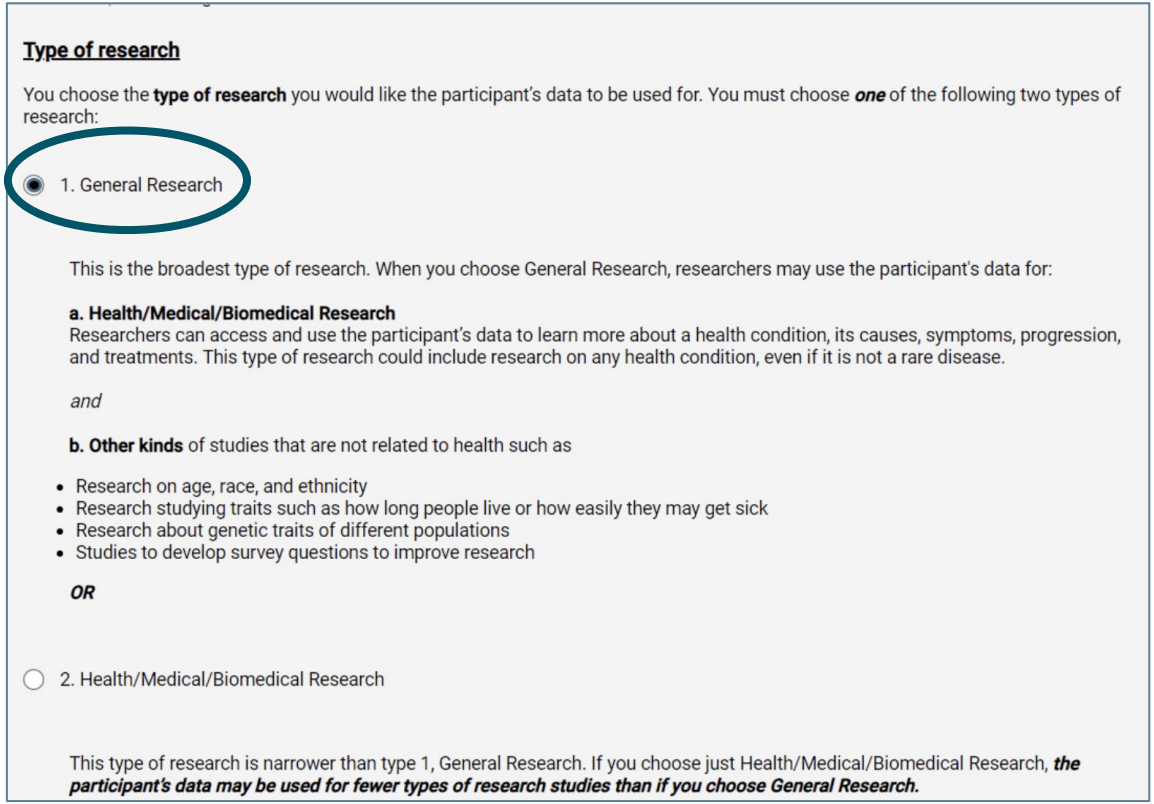
General Information *Demographics*

Demographic information collected on both patients and caregivers



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

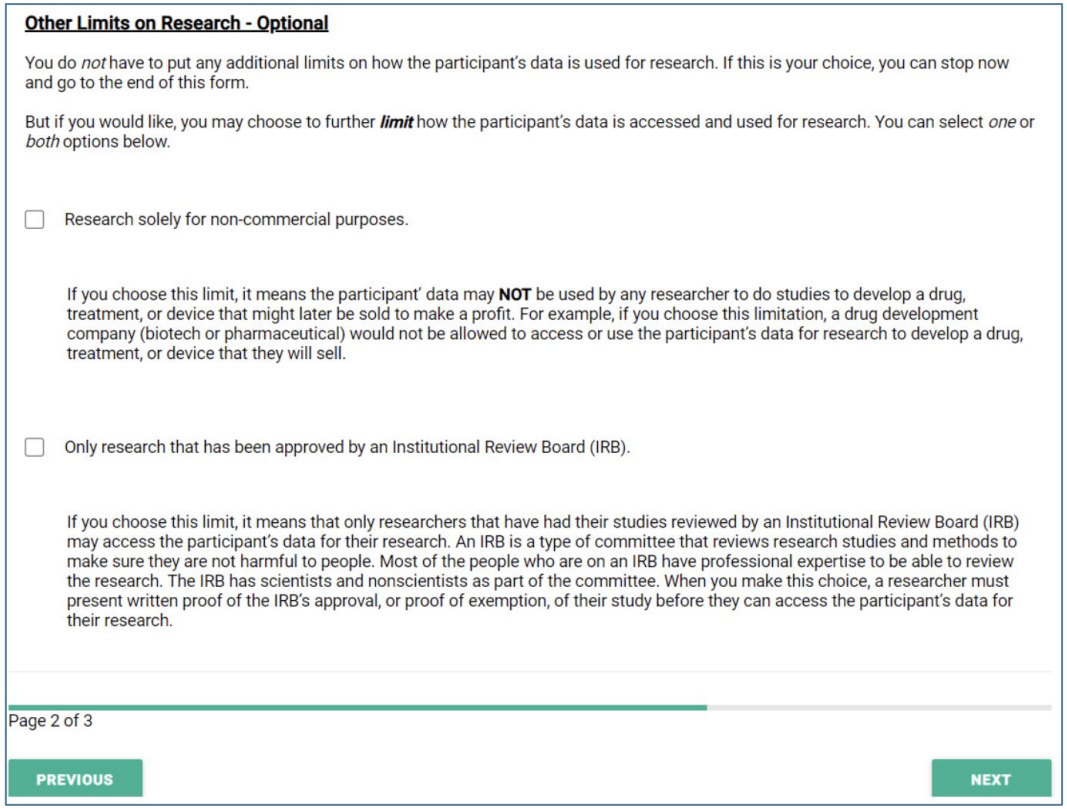
OR

( ) 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*

Optional: Setting Restrictions



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.

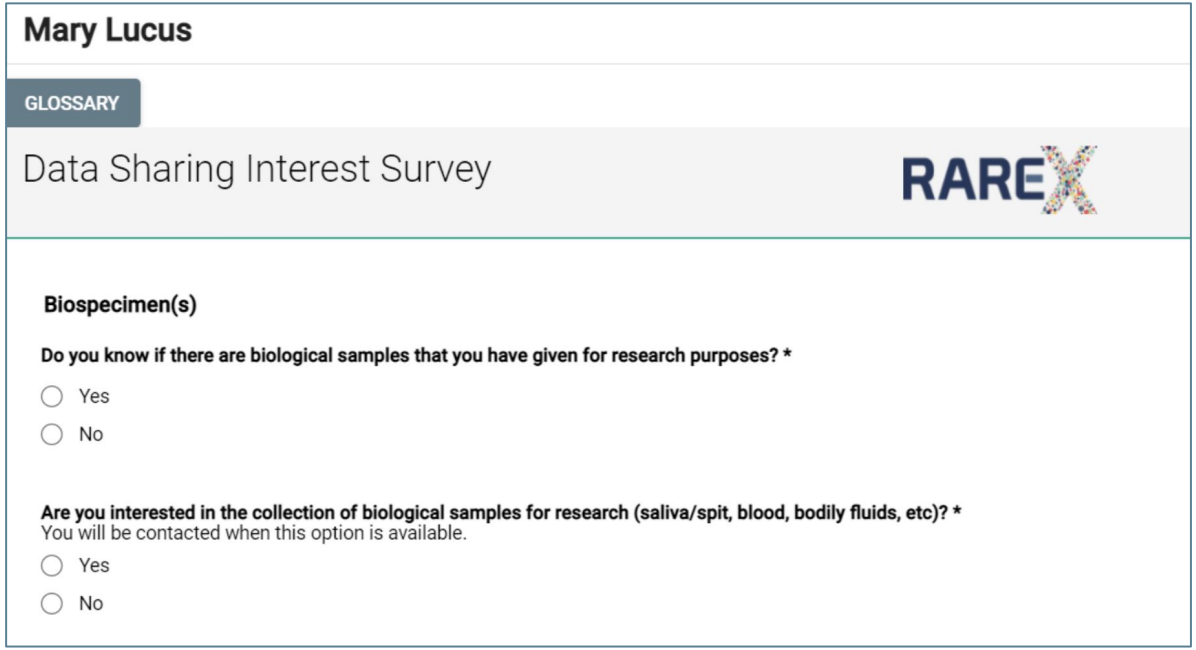
☐ Only research that has been approved by an Institutional Review Board (IRB).

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)

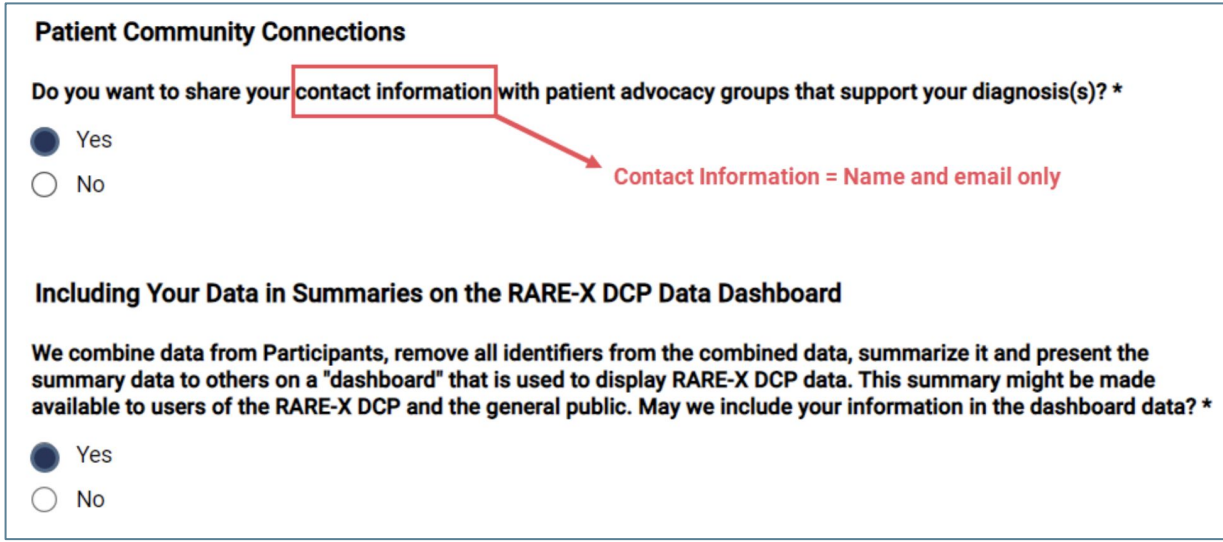
Do you know if there are biological samples that you have given for research purposes? *

- ◯ Yes
- ◯ No

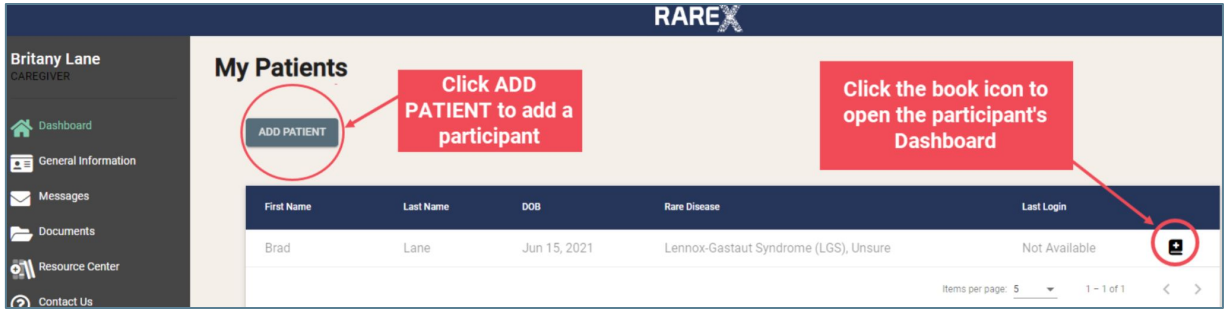
Are you interested in the collection of biological samples for research (saliva/spit, blood, bodily fluids, etc)? *
You will be contacted when this option is available.

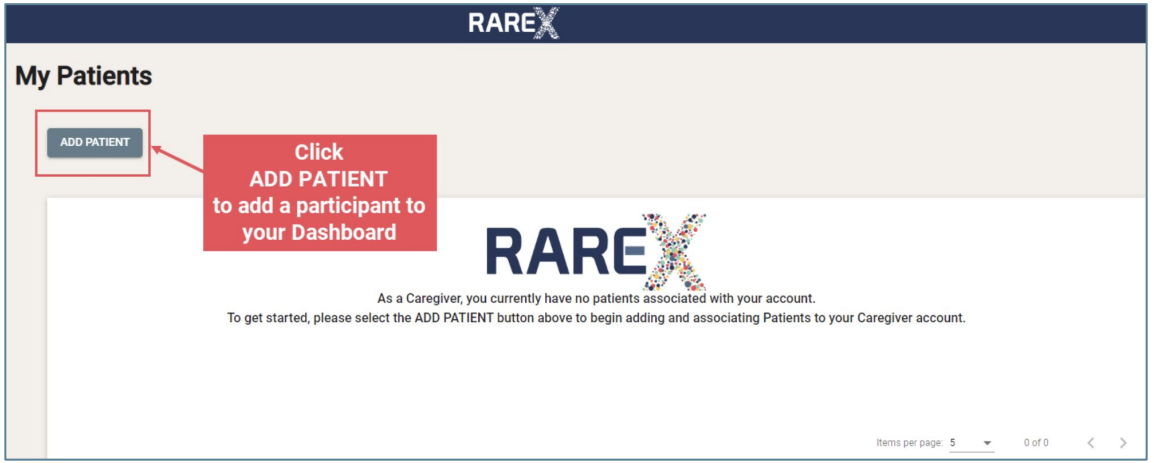
- ◯ Yes
- ◯ No

Data Sharing *Preference Agreement*



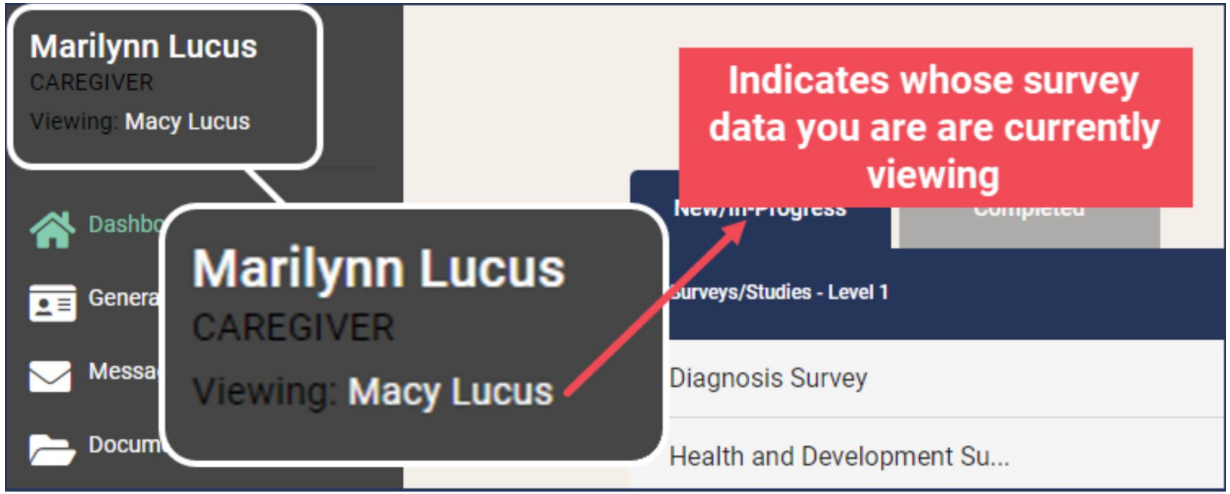
Caregiver Dashboard *Adding a Patient*





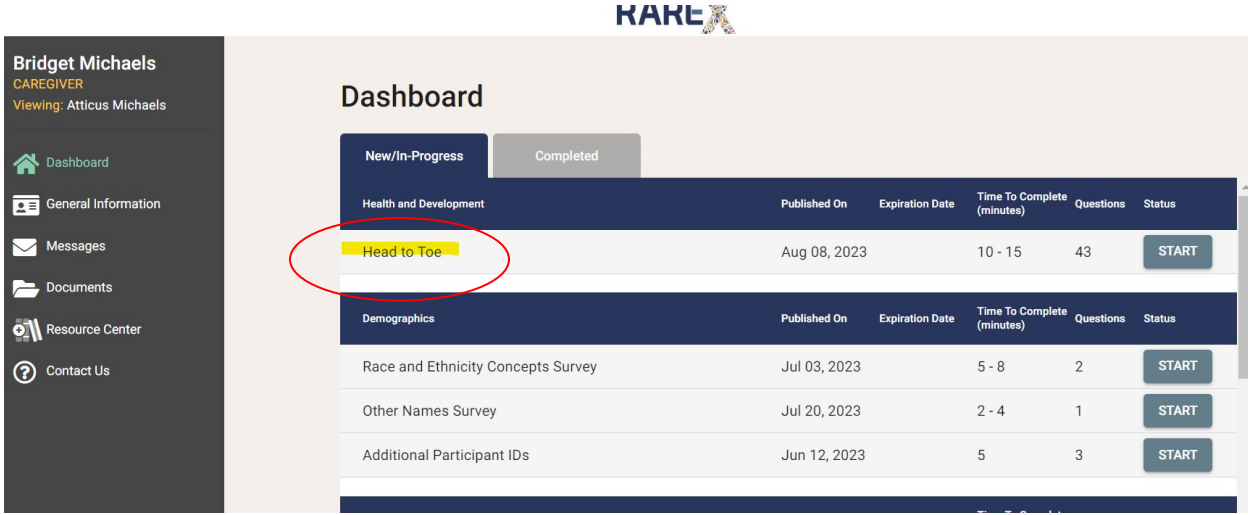
Caregiver Dashboard

Navigating Between Caregiver & Patient



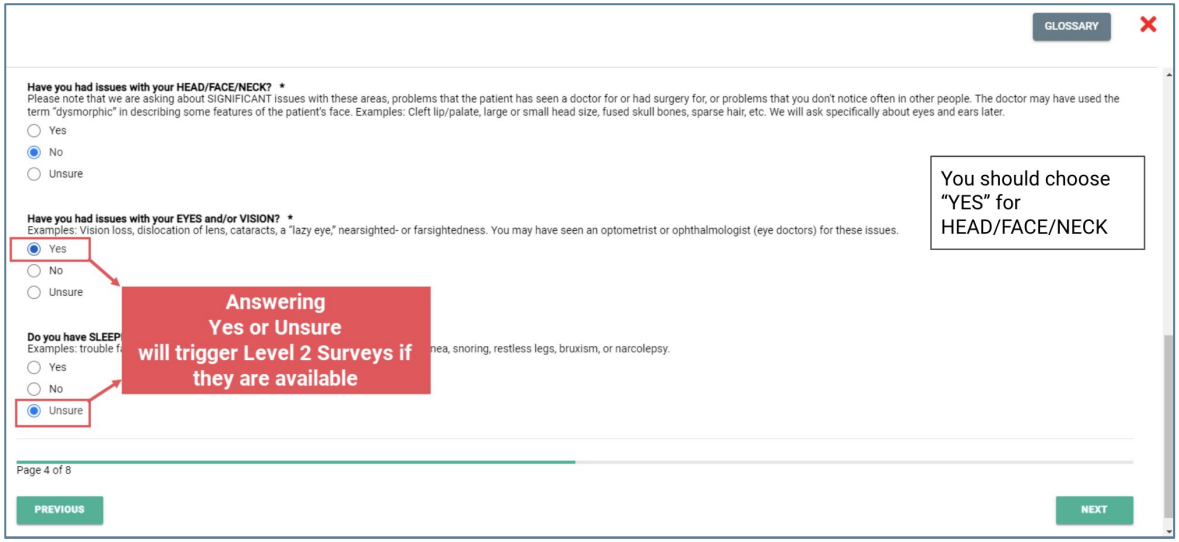
Surveys
Health & Development

You should choose the Diagnosis Survey to provide more detailed information about the diagnosis

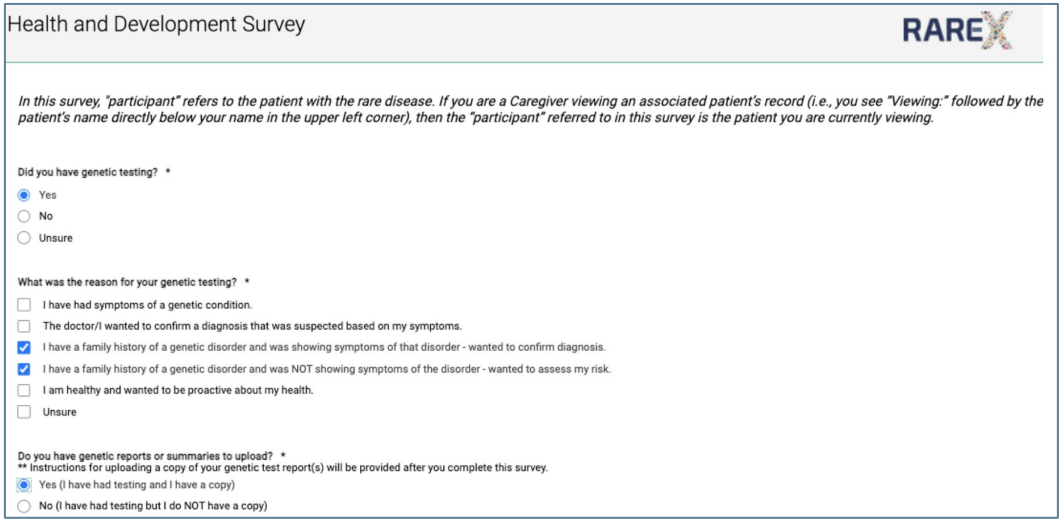


Surveys
Head to Toe

Click on the “X” to save & exit



Genetic Testing Information



Health and Development Survey

RAREX

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

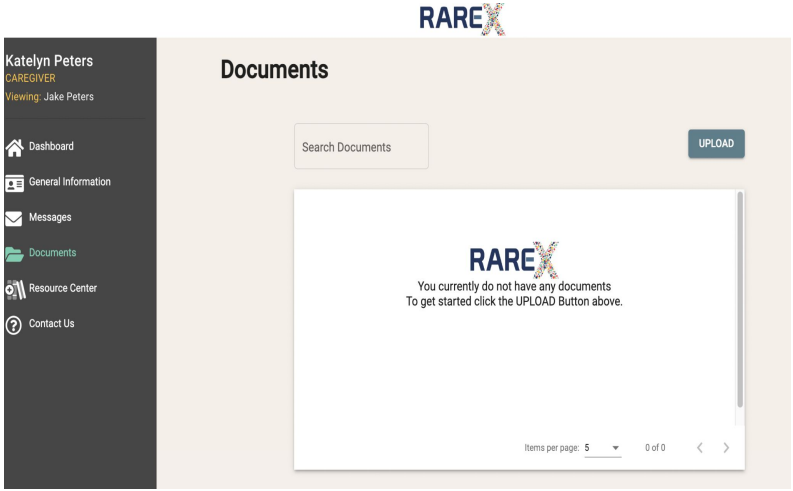
Do you have genetic reports or summaries to upload? *
** Instructions for uploading a copy of your genetic test report(s) will be provided after you complete this survey.

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

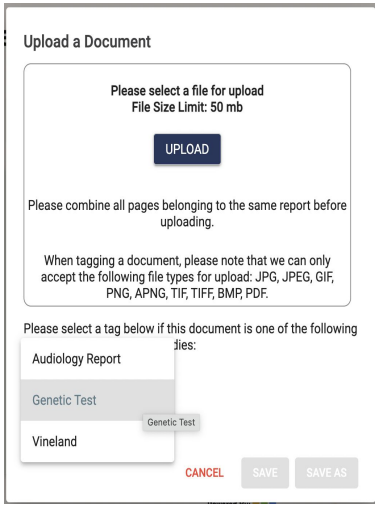
Genetic Testing Information

Updating a Genetic Report

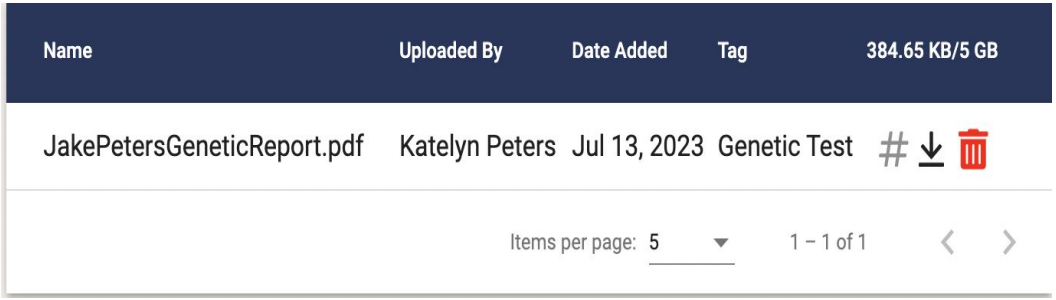
1- Documents



2- Upload & Tag



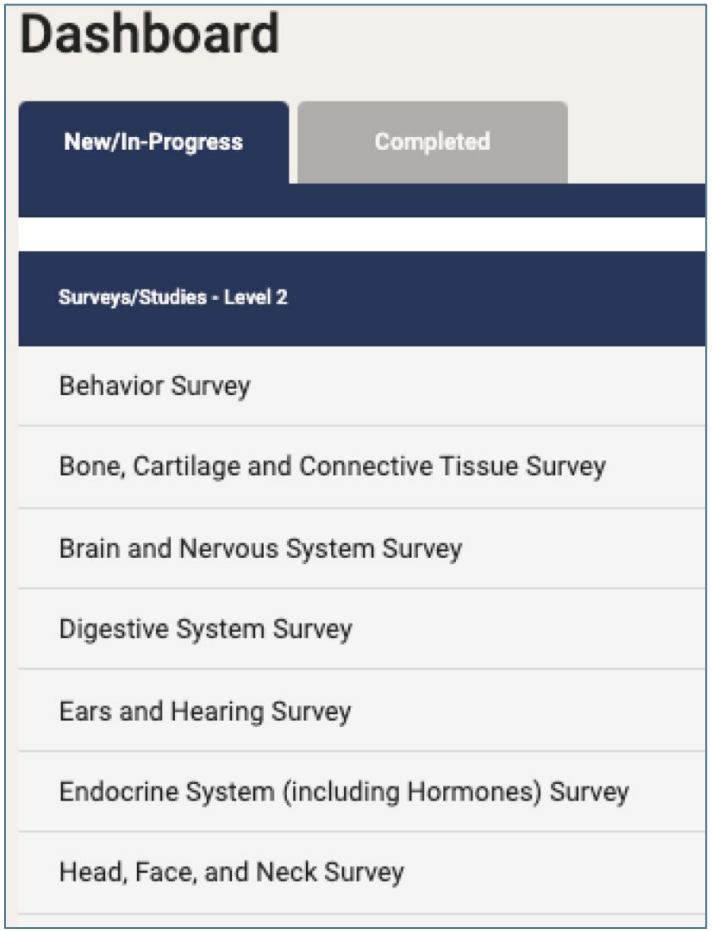
3- Save & View



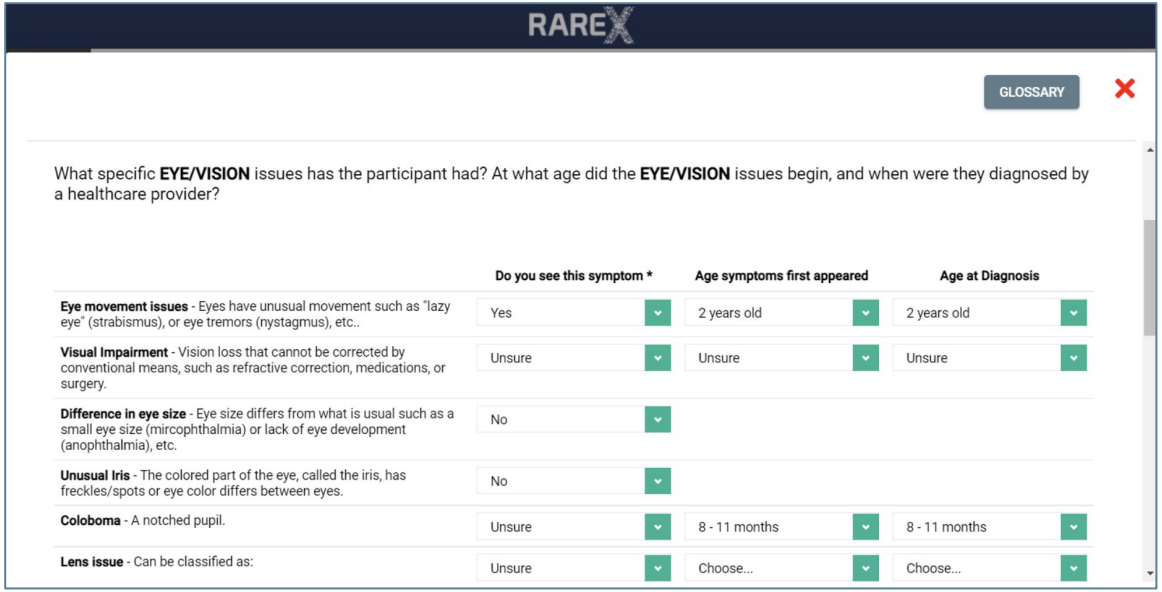
Surveys
Level 2 Survey Example

More detail than Head to Toe, but not a “deep dive”

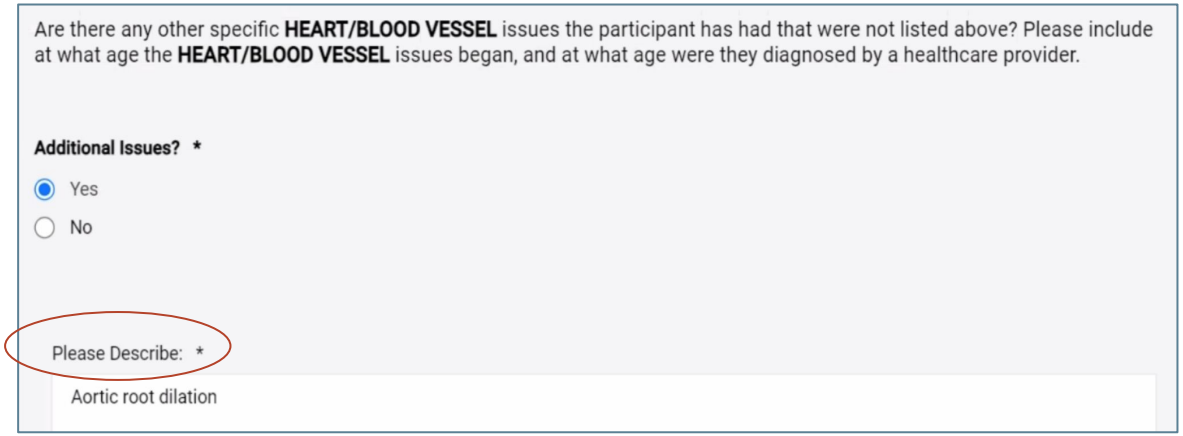
Always able to provide additional details in “free text” section at the bottom (do not use patient’s name or other identifiers)



Surveys
Answering Level 2 Surveys



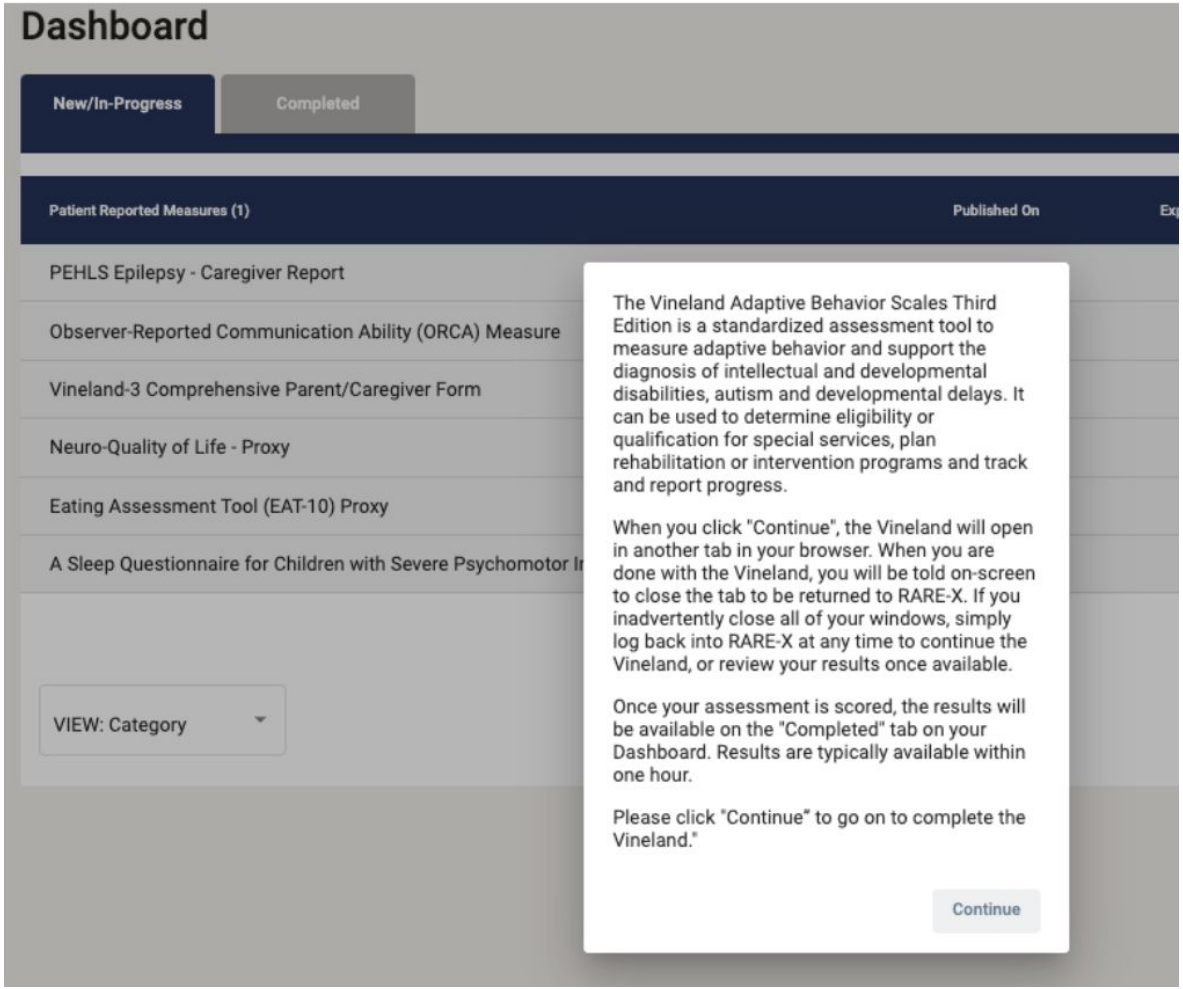
Surveys
Adding Symptoms Not Mentioned in Level 2 Surveys



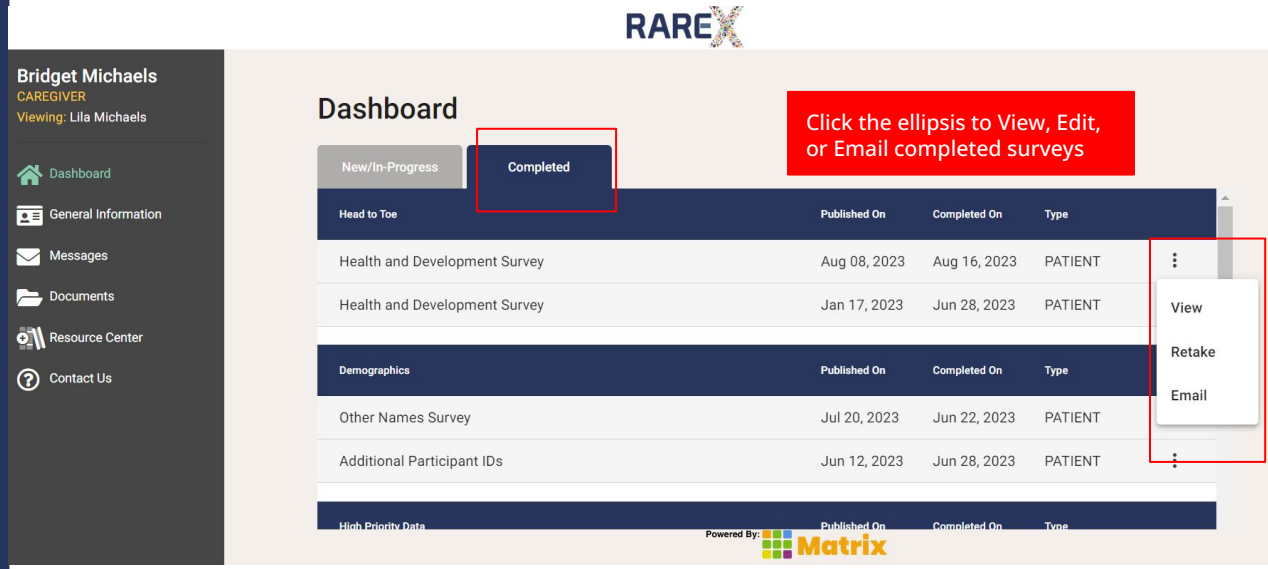
Please try not to put an identifying information in the free text area!

Surveys
Vineland Survey

You do not have to complete the Vineland Survey in 1 sitting, but you do have to complete it within 30 days of starting



Dashboard
Completed Tab



If you have questions or technical issues…

Email:

- rarexsupport@globalgenes.org

Thank You!