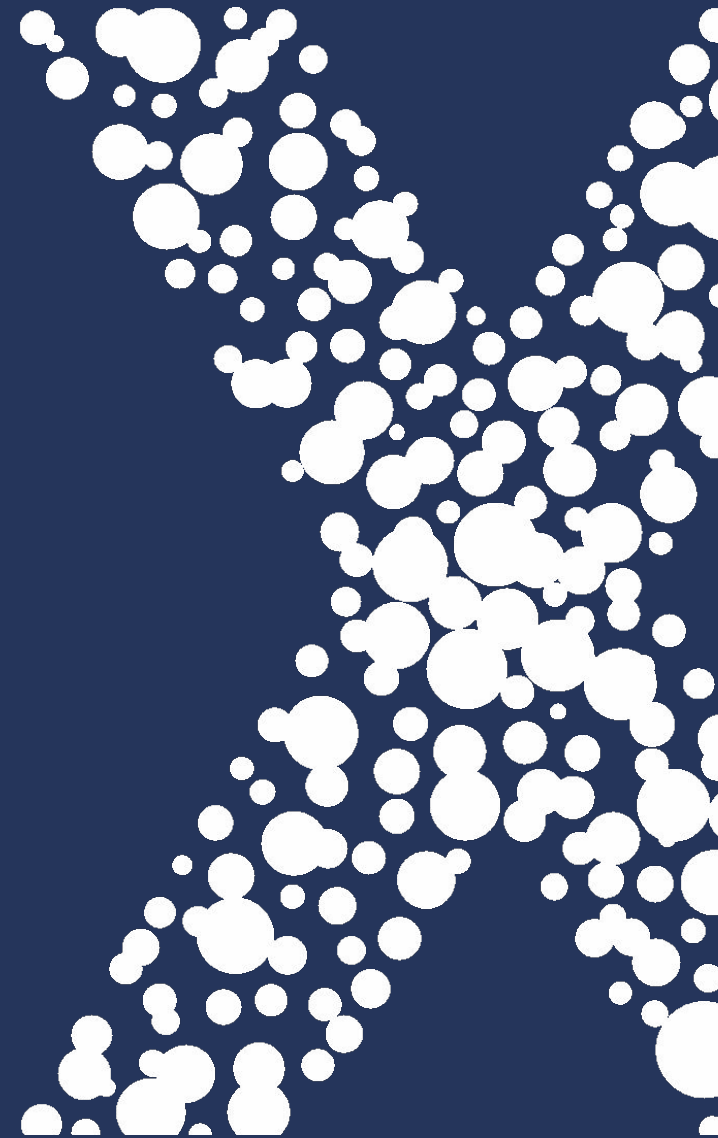


# USHER SYNDROME COALITION



## 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



**RARE**X



cogn

ition



Participants

Researchers

Participants

Researchers



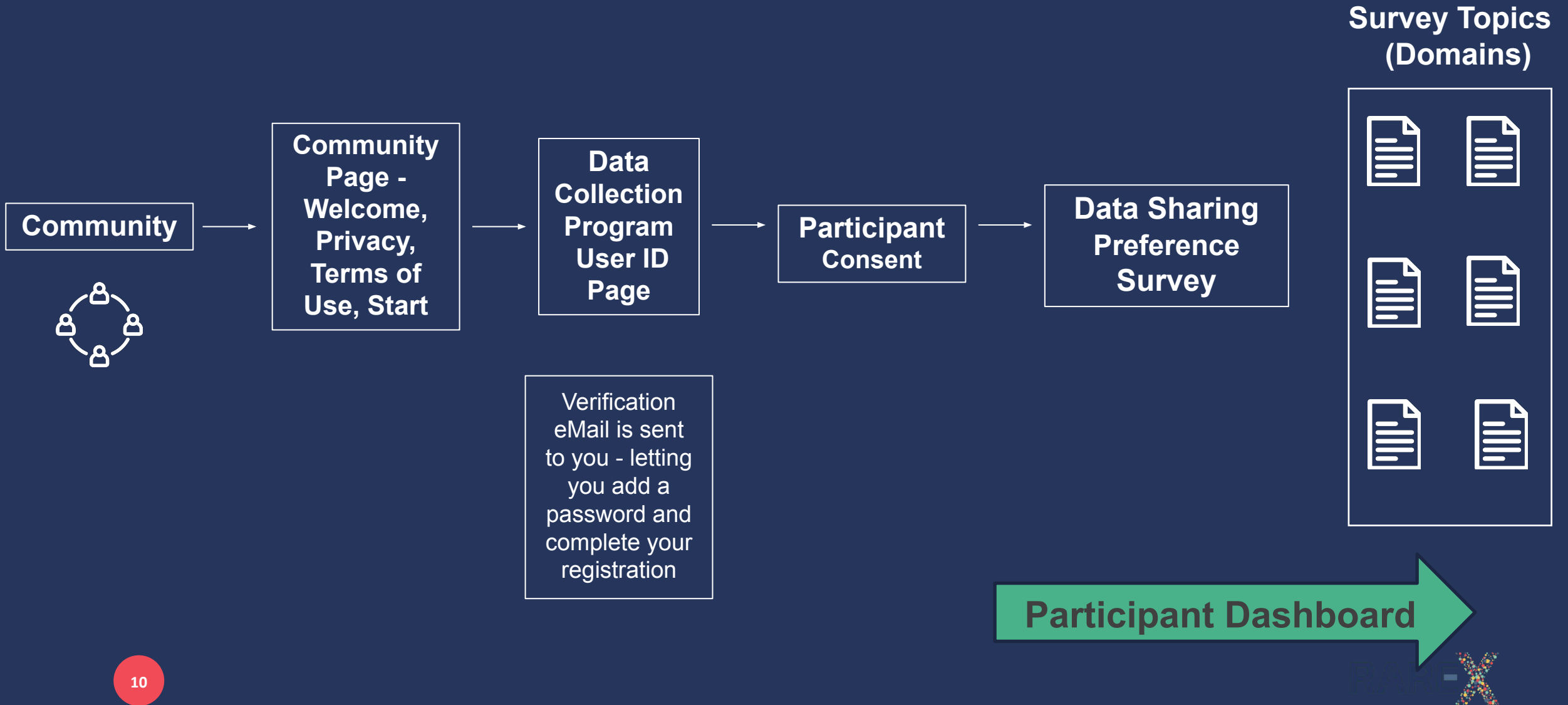


# What Do You Need To Get Started?

- **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




# Usher Syndrome Community Page on RARE-X

Usher Syndrome

[HOME](#) [GETTING STARTED](#) [FAQ](#)

Powered by **RARE-X**

## Usher Syndrome - Data Collection Program



Individuals with Usher syndrome, their families, and the broader Usher community 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 Usher Syndrome 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

Already Enrolled?

**GET STARTED**

**LOGIN**

By clicking Get Started, you are agreeing to the [RARE-X Terms of Use](#)

By clicking Login you are leaving the RARE-X site and entering the Data Collection Site


[Translate »](#)



# 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”





## Request Access

Join us and get connected today!

**Your Information:**

*This information will be used to create access credentials for the patient who will be accessing the platform themselves, or the caregiver/guardian who will be providing information on behalf of a patient.*

Your First Name \*

Your Last Name \*

Enter Letters Only

Email \*

Mobile Phone \*

Your Date of Birth \*

Enter Numbers Only

In order to proceed with your account creation, please acknowledge the following:

I am at least 18 years of age (or at least the Age of Majority in my State/Country)

I acknowledge that I am located in the United States; OR

I acknowledge that I am located outside of the United States, and that the information I am providing will be transmitted to the United States for account creation.

**Please check all that apply:**

Patient Participant  
*A Patient Participant is a person with a rare disease (already diagnosed or still on their diagnostic journey) who will be able to answer surveys and provide information about themselves.*

Caregiver Participant  
*A Caregiver Participant will be able to answer surveys and provide information on a patient(s) with a rare disease, for whom they are the Parent or Legally Authorized Representative.*

Person who has lost a loved one to a rare disease  
*This user will be able to answer surveys and provide information on a loved one with rare disease who is now deceased.*

[PRIVACY POLICY](#) [TERMS OF USE](#) [GO TO LOGIN](#)

# 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

Enter Your Email Here

Send verification code

Continue

Cancel

## 2. Confirm your verification code

Verification code has been sent to your inbox. Please copy it to the input box below.

Enter Your Email Here

Enter Your Verification Code

Verify code

Send new code

Continue

Cancel

## 3. Create your password

### Create Password

The password must be between 8 and 64 characters.  
The password must have at least 3 of the following:

- a lowercase letter
- an uppercase letter
- a digit
- a symbol

New Password

Confirm New Password

Continue

Cancel



# Login to the DCP



Sign in with your email address

[Forgot your password?](#)

Sign in

[PRIVACY POLICY](#)

[TERMS OF USE](#)



# Terms of Use

**RAREX**

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](http://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

**RAREX**

we encourage you to [download](#) a PDF version of the consent form and read it carefully.



- Terms of Use ✓
- General Information ✓
- Patient Informed Consent**
- General Information ✓
- Data Sharing Interest Survey ✓

**Who is funding the DCP?**  
RARE-X, a non-profit organization, is paying for the DCP.  
[LEARN MORE](#)

**How long will the DCP last?**  
The DCP does not have an end date. In fact, over time participants and caregivers may be asked to update their data to improve rare disease research.  
[LEARN MORE](#)

**Who is eligible to take part in the DCP?**  
All participants and families who have or may have a rare disease(s) may take part in the DCP.  
[LEARN MORE](#)

Participants and families who may take part include:

- Any person who has been diagnosed with a rare disease, or who is looking for a diagnosis.
- A parent or legal guardian of a child with a rare disease may register a child who is a minor (a "minor" is a child under the age of 18, in most states).
- The legally authorized representative of an adult with a rare disease who cannot physically or mentally answer the surveys may enroll the affected participant.

**Do I or my child have to take part in the DCP?**  
Taking part in the DCP is voluntary. This program is for research purposes only. The only alternative is to not participate in this program.  
[LEARN MORE](#)

**What will I have to do if I take part and give permission for my child to take part in the DCP?**  
We will ask you to create a secure, password-protected account. You will have the chance to answer a set of questions (surveys) about your child's health, health history, treatment and care, and the impact of having a rare disease on your household.  
  
Your child will not be asked to do anything for the DCP at this time.  
  
It is likely that in the future, DCP will expand to include the collection of biosamples such as saliva or blood. At that time, RARE-X would request an additional consent and

[PREVIOUS](#) [NEXT](#)



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



# General Information - *Demographics*



Terms of Use



Patient Informed Consent



**General Information**

Data Sharing Interest Survey

Steve O'Conner-Doyle

## Demographics

First Name \*

Steve

Mi

**Demographic data is collected on both caregivers and participants**

Suffix

Country \*

United States

Address Line 1 \*

1402 Chadwick Dr

Address Line 2

City \*

Apex

State/Province/Region \*

NC



# General Information – *Other Information*

Gender At Birth \*  
Female

Gender Identity \*  
Woman (or girl, if patient is child)

Birthplace: Country \*  
United Kingdom

Birthplace: State/Province/Region \*  
Wiltshire

Birthplace: City \*  
Apex

Estimated Household Income \*  
\$25,000 - \$49,999

Does this participant have health coverage of any type? \*  
No

Clinician Q

Is Patient Living? \*  
Yes

**Select Usher from the drop-down list.  
If you have other conditions select Other from the list and enter the disease name in the text box that opens**

Rare Disease(s) - Select all that apply

Other Disease  
Ehlers Danlos

Usher Syndrome ✕ Other ✕



# 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's 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.



# Data Sharing Preference Agreement

Mary Lucus

GLOSSARY

Data Sharing Interest Survey



## 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

## 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



**Ann Lane**  
CAREGIVER

- Dashboard
- General Information
- Messages
- Documents
- Resource Center
- Contact Us

## My Patients



Click ADD PATIENT to add a participant

Click the book icon to open the participant's Dashboard

First Name	Last Name	DOB	Rare Disease	Last Login
Brad	Lane	Jun 15, 2021	Lennox-Gastaut Syndrome (LGS), Unsure	Not Available

Items per page: 5    1 - 1 of 1    < >



# Adding a participant to Your Caregiver Dashboard

RAREX

## My Patients

ADD PATIENT

Click  
ADD PATIENT  
to add a  
participant to  
your Dashboard

RAREX

you currently have no patients associated with your account.

To get started, please select the ADD PATIENT button above to begin adding and associating Patients to your Caregiver account.

Items per page: 5

0 of 0



# Caregiver Dashboard

The image shows a screenshot of a caregiver dashboard. On the left is a dark sidebar with navigation icons for Dashboard, General, Messages, and Documents. The main content area is light-colored and displays user information and a list of surveys. Two callout boxes highlight the user information: one in the top-left corner and a larger one in the center. A green callout box with an arrow points to the 'Viewing: Macy Lucus' text in the center callout, explaining that it indicates whose survey data is currently being viewed. The survey list includes 'Surveys/Studies - Level 1', 'Diagnosis Survey', and 'Health and Development Su...'. A red circle with the number '28' is in the bottom-left corner, and a colorful 'X' logo is in the bottom-right corner.

**Marilynn Lucus**  
CAREGIVER  
Viewing: Macy Lucus

**Marilynn Lucus**  
CAREGIVER  
Viewing: Macy Lucus

Indicates whose survey data you are currently viewing

Dashboard

General

Messages

Documents

Surveys/Studies - Level 1

Diagnosis Survey

Health and Development Su...



# My Dashboard

## Dashboard

New/In-Progress

Completed

Choose the Diagnosis Survey to provide more detailed information about the diagnosis

Surveys/Studies - Level 1

Published On

Expiration Date

Time To Complete

Questions

Status

Diagnosis Survey

Aug 22, 2022

15-30 minutes

13

START

Health and Development Survey

May 24, 2022

15-20 minutes

43

START

Other Names Survey

Jan 05, 2022

2-4 minutes

1

START

Race and Ethnicity Concepts Survey

Jan 05, 2022

5-8 minutes

2

START

Complete the Health & Development Survey first

Continuous Data C

Published On

Expiration Date

Time To Complete

Questions

Status

Additional Pa

Aug 16, 2022

2-4 minutes

4

START

Interventional or Medical Diets Survey

Jun 09, 2022

2-5 minutes

5

START



# Health and Development Survey

GLOSSARY



Click "X" to  
save & exit

## Have you had issues with your HEAD/FACE/NECK? \*

Please note that we are asking about SIGNIFICANT issues with these areas, problems that the patient has seen a doctor for or had surgery for, or problems that you don't notice often in other people. We will ask specifically about eyes and term "dysmorphic" in describing some features of the patient's face. Examples: Cleft lip/palate, large or small head size, fused skull bones, sparse hair, etc.

- Yes
- No
- Unsure

## Have you had issues with your EYES and/or VISION? \*

Examples: Vision loss, dislocation of lens, cataracts, a "lazy eye" or strabismus, or other eye issues.

- Yes
- No
- Unsure

## Do you have SLEEPING issues? \*

Examples: trouble falling or staying asleep, excessive sleeping

- Yes
- No
- Unsure

Answering Yes or Unsure  
to a question will trigger Level 2 Surveys if  
they are available

Page 4 of 8

PREVIOUS

NEXT



# 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.
- I have a family history of a genetic disorder and was showing symptoms of that disorder - wanted to confirm diagnosis.
- 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

**Ann Bets**  
CAREGIVER  
Viewing: Amy Bets

- Dashboard
- General Information
- Messages
- Documents**
- Resource Center
- Contact Us

## Documents

Search Documents

UPLOAD

**Step 1**  
Open  
Documents

**Step 2**  
Click  
UPLOAD



You currently do not have any documents.  
To get started click the UPLOAD Button above.

### Upload a Document

Please select a file for upload  
File Size Limit: 50 mb

UPLOAD

When tagging a document, please note that we only accept the following file types for upload: PDF, PNG, APNG, BMP, GIF, JPEG, JEPG, SVG, TIFF, XLS, XLSX, ZIP

Please select a tag below if this document is related to the following types of curated Surveys/Studies:

Select a Tag

Genetic Test

CANCEL SAVE SAVE AS

**Step 3**  
Select  
Genetic  
Report &  
UPLOAD the  
report from  
your device



# Level 2 Surveys



Sylvie Youngston  
PATIENT

- Dashboard
- General Information
- Messages
- Documents
- Resource Center
- Contact Us

## Dashboard

New/In-Progress

Completed

Surveys/Studies - Level 1	Published On	Expiration Date	Time To Complete	Questions	Status
Diagnosis Survey	Feb 17, 2022		15-30 minutes	13	START
Other Names Survey	Jan 05, 2022		2-4 minutes	1	START
Race and Ethnicity Concep...	Jan 05, 2022		5-8 minutes	2	START

Surveys/Studies - Level 2	Published On	Expiration Date	Time To Complete	Questions	Status
Ears and Hearing Survey	Jan 05, 2022		5-8 minutes	9	START
Eyes and Vision Survey	Jan 05, 2022		10-15 minutes	13	START
Kidney, Bladder, and Geni...	Jan 05, 2022		5-10 minutes	12	START

VIEW: Category



# Answering Level 2 Surveys



GLOSSARY












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 *	Age symptoms first appeared	Age at Diagnosis
<b>Eye movement issues</b> - Eyes have unusual movement such as "lazy eye" (strabismus), or eye tremors (nystagmus), etc..	Yes	2 years old	2 years old
<b>Visual Impairment</b> - Vision loss that cannot be corrected by conventional means, such as refractive correction, medications, or surgery.	Unsure	Unsure	Unsure
<b>Difference in eye size</b> - Eye size differs from what is usual such as a small eye size (microphthalmia) or lack of eye development (anophthalmia), etc.	No		
<b>Unusual Iris</b> - The colored part of the eye, called the iris, has freckles/spots or eye color differs between eyes.	Yes	4 - 7 months	1 year old
<b>Coloboma</b> - A notched pupil.	Choose...		
<b>Lens issue</b> - Can be classified as: • Cataracts	Choose...		



# 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 (microphthalmia) 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: <ul style="list-style-type: none"><li>• Cataracts</li><li>• Abnormality of lens shape</li><li>• Aplasia/Hypoplasia of the lens</li><li>• Ectopia lentis</li><li>• Pseudophakia</li><li>• Phakodonesis</li></ul>	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

[GLOSSARY](#) ✕

---

**Ptosis** - Droopiness of the upper eyelid. Choose... ▼

---

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

**Additional Issues? \***

Yes  
 No

Please Describe: \*

Some of the above symptoms are a result of Usher Syndrome and others are not.  
(Explain more in this open text area)

Age at Diagnosis \*

Choose... ▼



# 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
<b>Ear shape difference (includes outer or inner ear)</b> - 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
<b>Conductive hearing loss</b> - Hearing loss or impairment due to an issue with the ear canal, ear drum, or the bones in the middle ear	No		
<b>Sensorineural hearing loss (SNHL) or impairment</b> - 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		
<b>Mixed hearing loss or impairment</b> - A combination of both conductive and sensorineural hearing loss.	Choose...		
<b>Ringling in the ears</b> - Also called tinnitus.	Choose...		
<b>Hyperacusis</b> - Reduced tolerance and increased sensitivity to everyday sounds in your normal environment.	Choose...		
<b>Vertigo</b> - 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

GLOSSARY



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

	Do you see this symptom *	Age symptoms first appeared	Age at Diagnosis
<b>Cerebral Palsy (CP)</b>	Choose...		
<b>Cognitive impairment</b> - Significant difficulties learning, understanding, or processing information, or making decisions.	Choose...		
<b>Headache or Migraine</b>	Yes	9 years old	16 years old
<b>Coordination issues</b> - Significant clumsiness or other difficulties with movement due to body parts not working together as they typically would.	Choose...		
<b>Abnormal EEG</b> - A problem has been identified on an electroencephalogram, or "EEG", which is a test that measures the electrical activity of the brain.	Choose...		
<b>Hypertonia</b> - Muscle tightness that limits movement, spasticity, and increased muscle tone.	Choose...		
<b>Hypotonia</b> - Unusually low muscle tone, muscle tone may have been described as floppy.	Choose...		
<b>Memory impairment</b> - Increased forgetfulness or significant difficulty remembering things.	Choose...		
<b>Unusual movements</b> - Changes or problems in the ability to move body parts voluntarily; do NOT include Parkinson's Disease here.	Choose...		

**A formal medical "Diagnosis" may have occurred at a later time than the symptom appeared**



# Diagnosis Survey

## Diagnosis Survey



General health and development questions are asked in this survey. To help RARE-X identify and prioritize future focused groups of questions, please provide ALL known participant conditions/diagnoses/genetic diagnoses below, starting with the most current diagnosis (one condition/diagnosis/genetic diagnosis per line).

What is the participant's diagnosis (please list one at a time - you will have the opportunity to add more later). \*

Include any diagnosis or symptom that you have not reported on yet or feel you need to provide more detail about.

What was the participant's age in years when the formal diagnosis was made? \*

5 years old

What was the participant's age in years when they first began experiencing symptoms of this diagnosis? \*

Unsure

What test(s) did physicians or other health professionals do to make the rare disease diagnosis? (Select all that apply.) \*

- |  |  |   |
|--|--|---|
| <input checked="" type="checkbox"/> Audiology                    | <input type="checkbox"/> Blood Gases                     | <input type="checkbox"/> Blood Bank         |
| <input type="checkbox"/> Cytogenetics                            | <input type="checkbox"/> Chemistry                       | <input type="checkbox"/> Cytopathology      |
| <input type="checkbox"/> CAT Scan                                | <input type="checkbox"/> Cardiac Catheterization         | <input type="checkbox"/> Cardiac Ultrasound |
| <input type="checkbox"/> Electrocardiac (e.g., EKG, EEC, Holter) | <input type="checkbox"/> Electroneuro (EEG, EMG, EP/PSG) | <input type="checkbox"/> Genetics           |
| <input type="checkbox"/> Hematology                              | <input type="checkbox"/> Bedside ICU Monitoring          | <input type="checkbox"/> Immunology         |

You can add any diagnosis or symptom that you were not able to report in previous surveys



# 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



Ann Bets  
CAREGIVER

- Dashboard
- General Information
- Messages
- Documents
- Resource Center
- Contact Us

## Dashboard

New/In-Progress	Completed			
Surveys/Studies - Level 1		Published On		
Health and Development Su...		May 24, 2022		
Consents and Data Sharing Preference		Published On	Completed On	Type
Data Sharing Interest Sur...		Feb 18, 2022	May 27, 2022	PATIENT
Consents and Data Sharing Interest		Published On	Completed On	Type
Informed Consent		Feb 10, 2022	May 27, 2022	INFORMED CONSENT

Click the ellipse to  
View, Edit or Email  
completed surveys

- VIEW
- EDIT
- EMAIL



# 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](#)

[Explore pages with a keyboard](#)

[Read articles in simplified view](#)

[Related resources](#)

[Get help](#)

## 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.

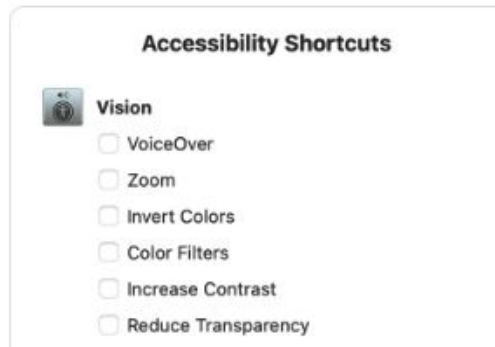
## Use the Accessibility Shortcuts panel on Mac

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](mailto:support@rare-x.org)

and cc: [n.odonnell@usher-syndrome.org](mailto:n.odonnell@usher-syndrome.org)



**Thank you!**

**Questions?**

