



CHANNELING HOPE
FOUNDATION

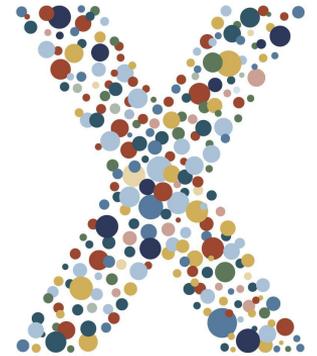
Data Collection Launch

November 20, 2024



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



Patient Journey in the Data Collection Platform (DCP)

Patient Dashboard

Potential Survey Topics (Domains)

Patient Community



Community Page
Get Started

Enroll in the DCP

Matrix Terms of Use

Patient Consent & Data Sharing Preference Survey

Head to Toe Survey



Diagnosis



Neuro Gen



Quality of Life



Kidney



Heart



Lung



Endocrine



Med Usage



Ear

NALCN Channel-related disorders (including IHRPF)- Data Collection Program



NALCN Channel-related disorders (including IHRPF) 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 NALCN Channel-related disorders (including IHRPF) 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



<https://rare-x.org/nalcn/>

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**
 - **Firefox**
- **What you do NOT need to get started?**
 - **A Lot of Time**
 - **To Finish It All At Once**



Request Access

Join us and get connected today!

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.

First Name

Your First Name *

Enter Letters Only

Last Name

Your Last Name *

Enter Letters Only

Email

Email *

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 review each type of account below and select the one (or multiple, if applicable) that describes **YOU**, the person creating the account:

Patient Participant

Select this option if you are enrolling to **answer surveys and provide information about yourself**. You will select this option if **you** are:

- A person diagnosed with a disease or condition, or still on your diagnostic journey
- A person with a connection to a person with a disease or condition (for example, a family member), providing consent to donate **your own** biosamples and/or complete your own surveys

Caregiver Participant

Select this option if you are enrolling to **answer surveys and provide information for another person**, including:

- A person with a disease or condition, for whom you are the **Parent** or **Legally Authorized Representative**
- A family member (such as a healthy sibling) of a person with a disease or condition, for whom you are the **Parent** or **Legally Authorized Representative**

Once you have created your own Caregiver account and logged into the application, you will then be able to add the person with the disease or condition, or family member who is participating.

Person who has lost a loved one

You will be able to answer surveys and provide information about a loved one who is now deceased. Please create an account in your own name, and once logged in, you will be able to add the deceased person about whom you are providing information.

Please note: Within the application you will be referred to as a "Caregiver", as this role allows you to enter information on behalf of your loved one.

SIGN UP

[PRIVACY POLICY](#)

[TERMS OF USE](#)

[GO TO LOGIN](#)

First-time Login Page

(Pre-Qualifications
Page)

Receive Account Creation Email

Thank You for Signing Up for RARE-X! Inbox x  



RARE-X
to bridget.michaels+sleep ▾

3:21PM (0 minutes ago)   



Dear Bridget,

Thank you for taking the time to sign up for the RARE-X Data Collection Program (DCP). Your account has been created. Please follow the link below to complete the verification process and create your new password.

[Create Password](#)

[Create Password](#)

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

Password Creation

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

Continue

Cancel

Privacy Policy & Terms of Use for RARE-X

Login to the DCP



Sign in with your email address

Forgot your password?

Sign in

[PRIVACY POLICY](#)

[TERMS OF USE](#)

You can access these documents anytime by clicking on them

Technology Platform 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 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

RARE X

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

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 biocamples such as saliva or blood. At that time, RARE X would request an additional consent and

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

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

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.*

My signature below indicates:

- I am 18 years or older.
- I have read this consent form. I understand the information in this form. I have had enough time to read the consent form and think about agreeing to take part in the DCP.
- I have had the opportunity to ask questions related to the DCP and do not have any unanswered questions at this time.
- I am willing to take part in the DCP.
- I agree to allow the collection, use, and sharing of my data as described above.
- By signing and dating this form, I do not give up any of my legal rights.
- I will get a signed and dated copy of this consent form.

Your complete legal first (given) name *

Bridget

Your complete legal last (family) name *

Michaels

General Information

Caregiver Demographics

Demographic information collected on both patients and caregivers

The screenshot shows a web form for caregiver registration. At the top right is the RAREX logo. On the left is a summary card for 'BRIDGET MICHAELS' with sections for 'Terms of Use' (checked), 'CAREGIVER PARTICIPANT', 'Caregiver Informed Consent' (checked), and 'General Information' (highlighted in blue). Below this is a link for 'Data Sharing Agreements and Other Research'. The main form area contains several input fields: 'First Name*' (Bridget), 'Middle Name', 'Last Name*' (Michaels), 'Suffix' (dropdown), 'Country*' (dropdown, required), 'Address Line 1*' (required), 'Address Line 2', 'City*' (required), 'State/Province/Region*' (required), 'Zip/Postal Code*' (required), 'Date of Birth*' (calendar icon, required), and 'Phone*' (required). At the bottom are 'PREVIOUS' and 'NEXT' buttons.

General Information

Demographics

Demographic information collected on both patients and caregivers

Other Information

Relationship/Marital Status*
Married

Race*
Required field. Please enter Race

Gender Identity*
Woman (or girl)

Birthplace: Country*
Required field. Please enter Birthplace Country

Estimated Household Income*
Prefer Not To Answer

Ethnicity*
Prefer Not To Answer

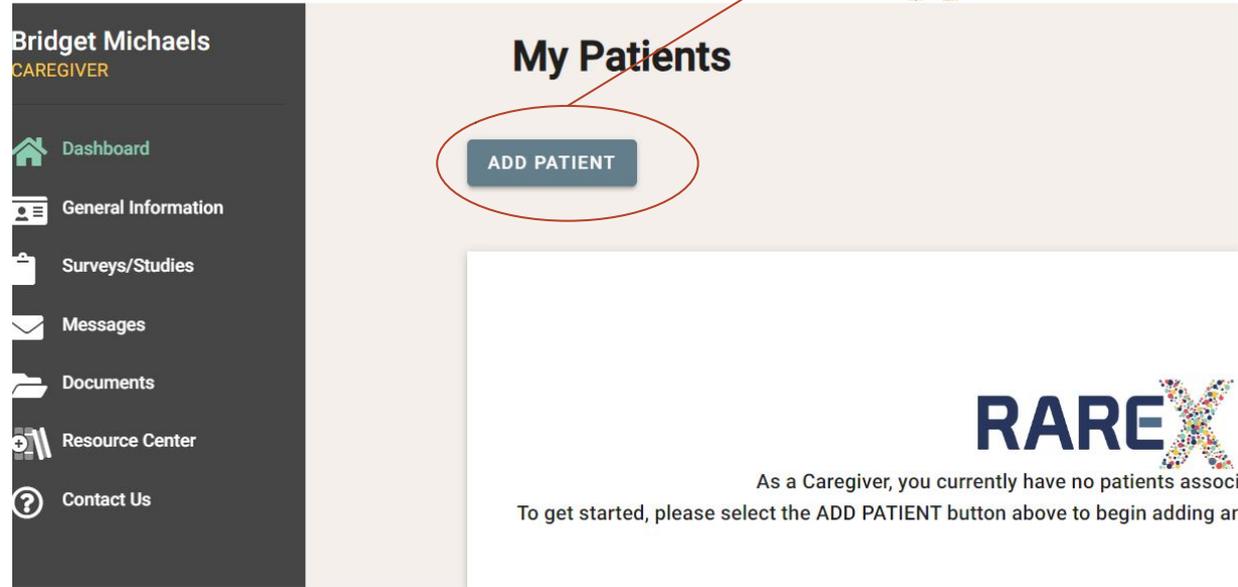
Biological Sex at Birth*
Female

Birthplace: State/Province/Region*
Required field. Please enter Birthplace State/Province/Region

Birthplace: City*
Required field. Please enter Birthplace City

Caregiver Dashboard *Adding a Patient*

Click ADD PATIENT on your Caregiver
Dashboard to add your patient
participant



The screenshot displays the RAREx Caregiver Dashboard. On the left is a dark sidebar with the user's name 'Bridget Michaels' and role 'CAREGIVER'. Below the name are navigation icons and labels for 'Dashboard', 'General Information', 'Surveys/Studies', 'Messages', 'Documents', 'Resource Center', and 'Contact Us'. The main content area is light gray and features the 'RAREx' logo at the top right. Below the logo is the heading 'My Patients' and a blue button labeled 'ADD PATIENT' which is circled in red. A red line connects the text box above to this button. Below the 'My Patients' section is a white box containing the RAREx logo and the text: 'As a Caregiver, you currently have no patients associated with you. To get started, please select the ADD PATIENT button above to begin adding a patient participant.'

Informed Consent

This time, you will
complete the
consent form on
behalf of the
patient.

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.*

My signature below indicates:

- I am 18 years or older.
- I have read this consent form. I understand the information in this form. I have had enough time to read the consent form and think about agreeing to take part in the DCP.
- I have had the opportunity to ask questions related to the DCP and do not have any unanswered questions at this time.
- I am willing to take part in the DCP.
- I agree to allow the collection, use, and sharing of my data as described above.
- By signing and dating this form, I do not give up any of my legal rights.
- I will get a signed and dated copy of this consent form.

Your complete legal first (given) name *

Bridget

Your complete legal last (family) name *

Michaels

General Information

Patient Demographics

Select the Rare Disease Community in the patient demographics section

Other Information

Race *
White

Gender At Birth *
Female

Birthplace: Country *
USA

Estimated Household Income *
Prefer Not To Answer

Does this participant have health coverage of any type? *
Yes

Clinician Q

Is Patient Living? *
No

Ethnicity *
Not Hispanic or Latino

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

Birthplace: State/Province/Region *
DC

What type of coverage does the participant have? (Select all that apply) *
Private Health Insurance (e.g. HMO, PPO, HSA, Fee for Service, POS, etc.)

Rare Disease(s) - Select all that apply
Non
Nonsense Mutation Related Disorder
Other
Unsure
Undiagnosed

Your disease will be listed here!

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'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.

Page 2 of 3

PREVIOUS

NEXT

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

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

Genetic Testing Information



CAREGIVER PARTICIPANT

Caregiver Informed Consent ✓

General Information ✓

PATIENT PARTICIPANT

Patient Informed Consent ✓

General Information ✓

**Data Sharing Agreements and
Other Research**

Genetic Test Upload

Genetic Testing

Did the participant have genetic testing? *

Yes

No

Unsure

PREVIOUS

Genetic Report Upload

CAREGIVER PARTICIPANT

- Caregiver Informed Consent ✓
- General Information ✓

PATIENT PARTICIPANT

- Patient Informed Consent ✓
- General Information ✓
- Data Sharing Agreements and Other Research**
- Genetic Test Upload

Do you have genetic reports or summaries to upload? *

Upon completion of this survey, you will be prompted to upload your genetic reports or summaries.

If you do not currently have a copy, you may upload the report at any time by navigating to "Documents" in the left navigation menu, uploading your report or summary, and tagging it as a "Genetic Test".

Yes (the participant has had testing and I have a copy)

No (the participant has had testing but I do NOT have a copy)

Unsure

PREVIOUS NEXT

If you select "No" you will still be able to upload your report later!

Head to Toe Survey

*All Level 2 surveys
"branch" from the
Head to Toe*

Has the participant had issues with their HEAD/FACE/NECK? *

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

Yes

No

Unsure

Has the participant had issues with their EYES and/or VISION? *

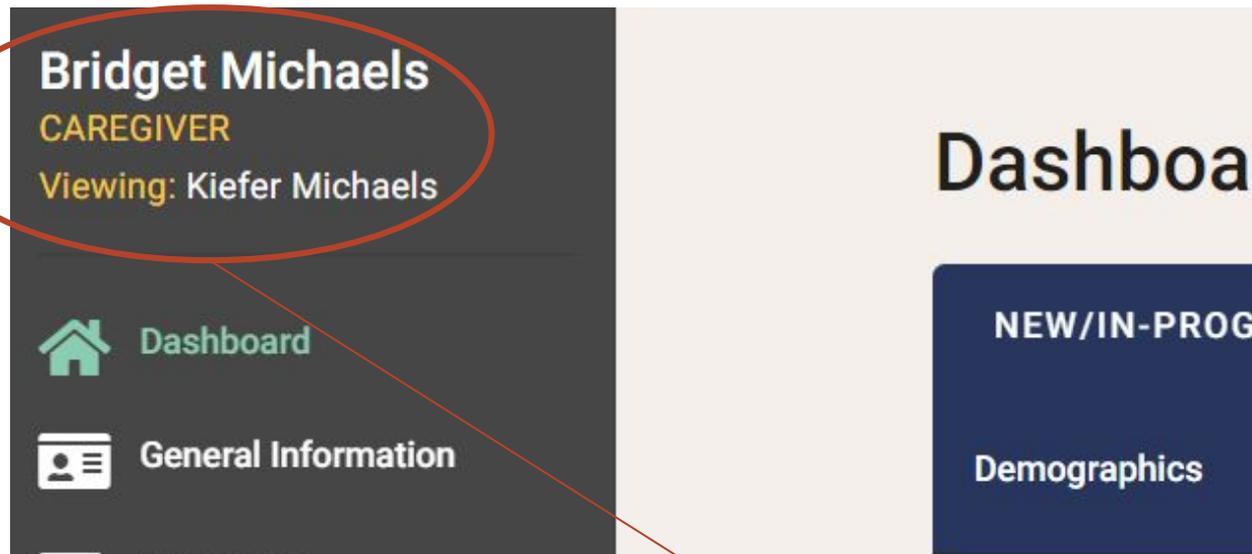
Examples: Vision loss, cataracts, a "lazy eye," nearsighted- or farsightedness. The participant may have seen an optometrist or ophthalmologist (eye doctors) for these issues.

PREVIOUS

NEXT

Answering "Yes" or "Unsure" will trigger Level 2 Surveys

Caregiver Dashboard *Navigating Between Caregiver & Patient*



The top left corner will always indicate whose dashboard you are viewing!

Surveys Level 2 Surveys



Dashboard

New/In-Progress		Completed				
Clinical Data Collection	Published On	Expiration Date	Time To Complete	Questions	Status	
Surveys/Studies - Level 2						
Behavior Survey	Jul 20, 2023		20-25 minutes	21	CONTINUE	
Bone, Cartilage and Connective Tissue Survey	Jul 19, 2023		10-15 minutes	13	CONTINUE	
Brain and Nervous System Survey	Jul 20, 2023		10-15 minutes	12	START	
Eyes and Vision Survey	Jul 20, 2023		10-15 minutes	13	START	
Patient Reported Measures (1)	Published On	Expiration Date	Time To Complete	Questions	Status	

Powered By:  Matrix

[Privacy Policy](#) | [Terms of Use](#)

Surveys Answering Level 2 Surveys

RAREX

GLOSSARY X

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
Eye movement issues - Eyes have unusual movement such as "lazy eye" (strabismus), or eye tremors (nystagmus), etc..	Yes <input type="button" value="v"/>	2 years old <input type="button" value="v"/>	2 years old <input type="button" value="v"/>
Visual Impairment - Vision loss that cannot be corrected by conventional means, such as refractive correction, medications, or surgery.	Unsure <input type="button" value="v"/>	Unsure <input type="button" value="v"/>	Unsure <input type="button" value="v"/>
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.	No <input type="button" value="v"/>		
Unusual Iris - The colored part of the eye, called the iris, has freckles/spots or eye color differs between eyes.	No <input type="button" value="v"/>		
Coloboma - A notched pupil.	Unsure <input type="button" value="v"/>	8 - 11 months <input type="button" value="v"/>	8 - 11 months <input type="button" value="v"/>
Lens issue - Can be classified as:	Unsure <input type="button" value="v"/>	Choose... <input type="button" value="v"/>	Choose... <input type="button" value="v"/>

Surveys

Adding Symptoms Not Mentioned in Level 2 Surveys

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

Additional Issues? *

Yes

No

Please Describe: *

Aortic root dilation

Try not to include any identifying information in the free text boxes!

Surveys Diagnosis Survey

Dashboard

NEW/IN-PROGRESS		COMPLETED			
Demographics	Published On	Expiration Date	Complete (minutes)	Questions	Status
High Priority Data					
Diagnosis Survey	Apr 17, 2024			0	START
Quality of Life Survey	Aug 22, 2022		15 - 20	43	START
Medication and/or Supplement Usage	Jul 25, 2023		10 - 15	20	START
Interventional or Medical Diets Survey	Jul 25, 2023		2 - 5	5	START
WHODAS 2.0 - World Health Organization Disability Assessment Schedule	Feb 17, 2023		15 - 18	39	START

Powered By: 

Diagnosis Survey

You chose the following condition as the rare disease diagnosis you affiliate with: **NALCN channel-related disorders (including IHRPF)**

Please note: If you chose more than one diagnosis when you set up your account, you will be asked these questions about each diagnosis separately.

There are multiple specific conditions that can be diagnosed in individuals in this disease community. Does one of these more specific conditions apply to you? *

- CLIFAHDD syndrome, NALCN-related
- IHRPF1 (Infantile Hypotonia syndrome), NALCN-related
- IHRPF2 (Infantile Hypotonia syndrome), UNC80-related
- NALCN Channel-related disorder, unknown type

The Diagnosis survey will ask about any sub-diagnosis you may have!

Please note that throughout this survey, "you" refers to the participant with the rare disease. If you are filling this survey out as a caregiver, please respond based on your observations of the person you care for.

Diagnosis Survey

Do you know the genetic cause of your disease? *

If you are not sure of the genetic cause, please say no.

Yes

No

Family History

Please remember that these questions pertain ONLY to this diagnosis: Sleep-related disorders.

The Diagnosis survey will also ask about genetic testing, newborn screening, and family history

Dashboard Completed Tab

Bridget Michaels
CAREGIVER
Viewing: Lila Michaels

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



Dashboard

Click the ellipsis to View, Edit, or Email completed surveys

New/In-Progress		Completed		
Head to Toe	Published On	Completed On	Type	
Health and Development Survey	Aug 08, 2023	Aug 16, 2023	PATIENT	⋮
Health and Development Survey	Jan 17, 2023	Jun 28, 2023	PATIENT	View Retake Email
Demographics		Published On	Completed On	Type
Other Names Survey	Jul 20, 2023	Jun 22, 2023	PATIENT	
Additional Participant IDs	Jun 12, 2023	Jun 28, 2023	PATIENT	
High Priority Data		Published On	Completed On	Type

Powered By: Matrix

If you have questions or technical issues...

Email:

- rarexsupport@globalgenes.org

Questions?

