




FGFR Syndromes Research Registry Participant User Guide

Register for an Account

- Step 1: Read the Terms and Conditions and Privacy Policy and attest to the statements provided. When you are finished with this page, click “Next”.

Featuring



Registration

Terms & Conditions Contact Info Notifications Review & Submit Confirmation

Below are links to the IAMRARE Terms of Use and Privacy Guidelines. The purpose of these documents is to outline your rights and responsibilities when using the platform. These documents include: 1) Standard policies for all studies on this platform, 2) A privacy statement that details how your data can be used, 3) Information outlining the unacceptable uses of the platform, and 4) Information about how to address questions and issues.

Acknowledgements:

- You are at least 18 years of age, the age of majority in your state, province or country, and able to consent on behalf of yourself and/or an individual that you have legal responsibility for. *
- You agree to support the Platform's research activities by providing truthful, appropriate information and to not do anything that will put the Services or the information in the Platform at risk. *
- You understand that NORD will use reasonable efforts to keep the information you enter on the Services safe, but no data transmissions over the Internet can be guaranteed to be 100% secure. The information you provide will be available to authorized users at NORD for platform maintenance and research activities, as well as to the sponsor of the studies you consent to participate in. *
- You agree to the [Terms and Conditions & Privacy Policy](#) *

[Return to login](#) Next

- Step 2: Enter your personal information in the spaces provided. When you are finished with this page, click “Next”.

Featuring

Registration

Terms & Conditions Contact Info Notifications Review & Submit Confirmation

Country of Residence *

First Name * Last Name *

E-mail *

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- Step 3: Select whether you are interested in being contacted by NORD regarding available studies. When you are finished with this page, click “Next”.

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Registration

Terms & Conditions Contact Info Notifications Review & Submit Confirmation

I am interested in NORD contacting me regarding available studies. *

Yes No

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- Step 4: Select “Next” so that an activation link is sent to your e-mail to complete registration.

Featuring

Registration

Terms & Conditions Contact Info Notifications Review & Submit Confirmation

An activation link will be sent to test@test.com. Click "Next" to send this e-mail and continue.

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- Step 5: Click the link you are sent via e-mail. Please check your Spam folder if you do not see the e-mail. You will be taken to the following screen in a new tab within your browser. Set your password and click “Submit”.

E-mail Validation

Your e-mail your.email@email.com has been successfully validated.
Please create your password below.

Password

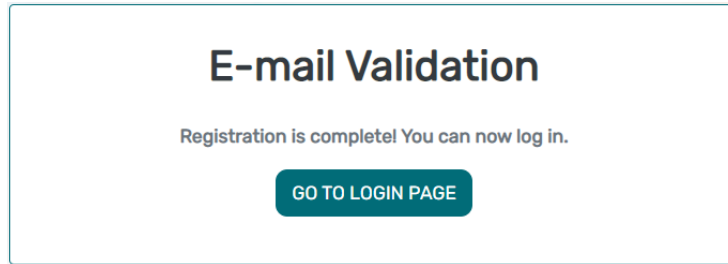
A password must be at least 8 characters long: ✘

- contain 1 uppercase letter ✘
- contain 1 lowercase letter ✘
- contain 1 digit ✘
- not contain text from top 1000 commonly used passwords ✘

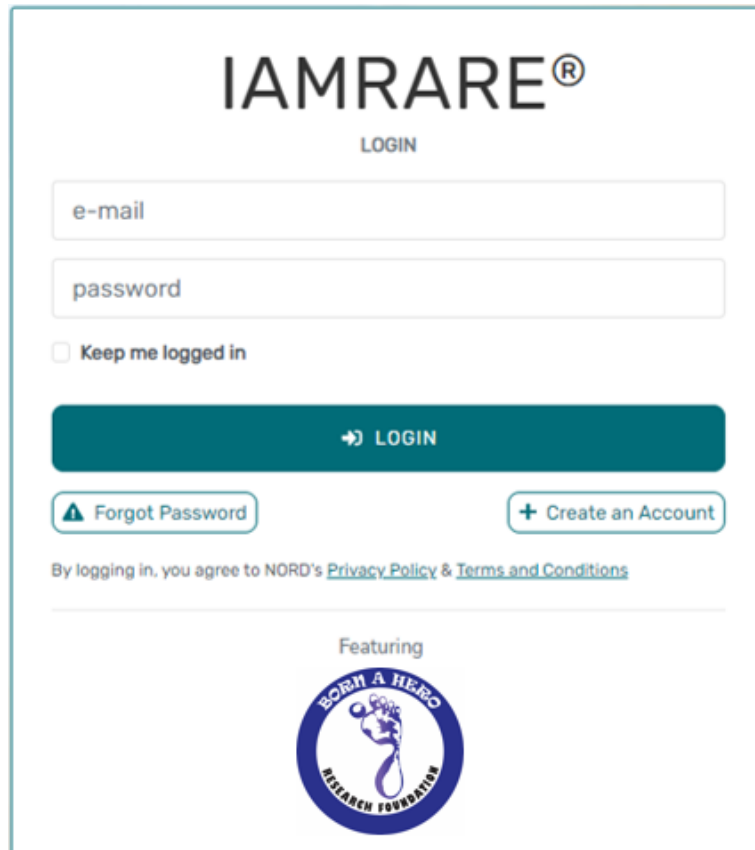
Repeat Password

[SUBMIT](#)

- Step 6: Your validation is now complete. Select “Go to Login Page”.

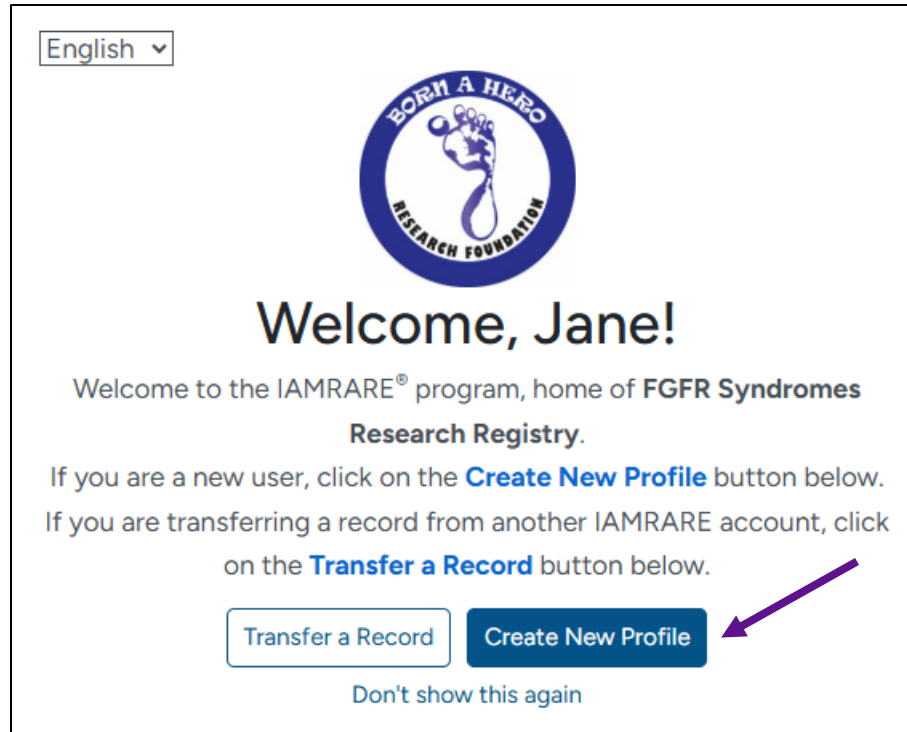


- Step 7: Log in using your new e-mail and password.




Add a Participant

- Step 1: To start, click Create New Profile



English ▾



Welcome, Jane!

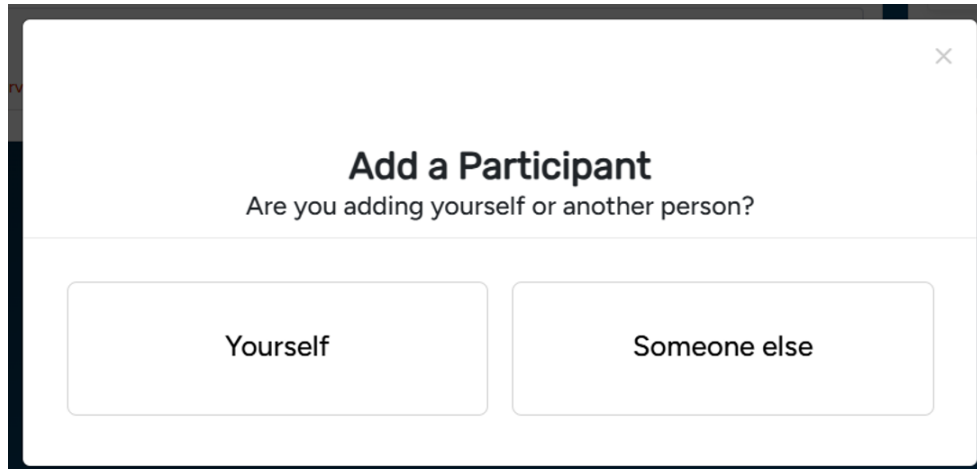
Welcome to the IAMRARE® program, home of **FGFR Syndromes Research Registry**.

If you are a new user, click on the **Create New Profile** button below.
If you are transferring a record from another IAMRARE account, click on the **Transfer a Record** button below.

[Transfer a Record](#) [Create New Profile](#)

[Don't show this again](#)

- Step 2: Select who you will be providing information about.



Add a Participant

Are you adding yourself or another person?

[Yourself](#) [Someone else](#)

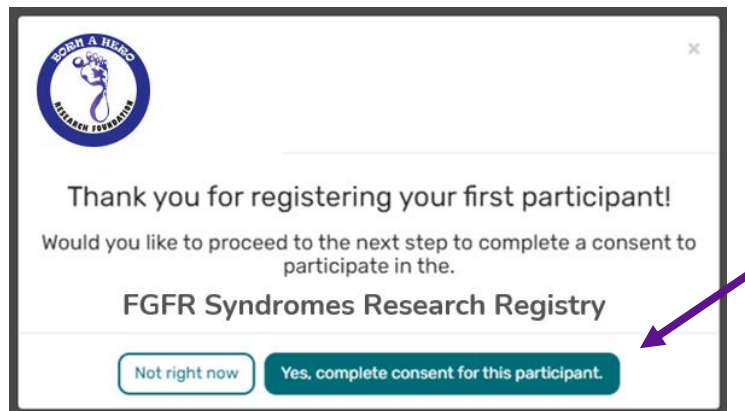
- Step 3: Fill out the Participant’s information.

The screenshot shows a web form titled "Add Participant" with a close button (X) in the top right corner. The form is divided into two columns and contains the following fields:

- Who Is Being Added as a Participant?**: Radio buttons for "Self" and "Other" (selected).
- Preferred First Name ***: Text input field.
- Current Last name ***: Text input field.
- First Name on Birth Certificate ***: Text input field.
- Middle Name on Birth Certificate ***: Text input field with a placeholder "Type 'NA' if none".
- Last Name on Birth Certificate ***: Text input field.
- Date of Birth ***: Date picker field with a calendar icon and a placeholder "mm/dd/yyyy".
- Sex Recorded on Birth Certificate ***: Dropdown menu.
- Country of Residence ***: Dropdown menu.
- State/Province/Region of Residence ***: Text input field with a placeholder "State/Province/Region".
- Country of Birth ***: Dropdown menu.
- City/Municipality of Birth ***: Text input field with a placeholder "City/Municipality of Birth".
- What is Your Relationship to ? ***: Dropdown menu.

Consent to the Study

- Step 1: Click on “Yes, complete consent for this participant.”



- Step 2: Scroll down and read through the consent form thoroughly. Once you finish each page, click the “Next” button. Once you reach the Authorization form, read through the statements thoroughly. If you are comfortable consenting to participate in the study, please read each statement and authorize your consent. After checking the boxes, click “Next.”

Consent to FGFR Syndromes Research Registry

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Consent Overview

Those eligible to participate in our study include:

Participant: An individual diagnosed with an FGFR Syndrome who is at least 18 years of age, the age of majority in their state, province or country, and able to provide consent for themselves.

Legally Authorized Representative: an individual (such as a family member or guardian) who is legally responsible for the healthcare of the Study Participant who is a minor (child under the age of 18) or an adult who is unable to contribute their own data. This individual must also be at least 18 years of age and the age of majority in their state, province or country.

Designated Representative: A legal adult who was the caretaker of an individual who passed away from an FGFR Syndrome, defined as a spouse, parent, sibling, offspring, close relative, close friend, guardian and/or significant other of the individual who had an FGFR Syndrome and who had knowledge and participated in their medical care. This individual must also be at least 18 years of age and the age of majority in their state, province or country.

1. Please tell us about the Participant you would like to enroll in this study. [Clear](#)

They are a minor or an adult who is unable to contribute their own data. I am currently their caregiver.

They were a patient with an FGFR Syndrome. I participated in their medical care.

[Next](#)

Consent to FGFR Syndromes Research Registry

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Title: FGFR Syndromes Research Registry

Principal Investigator: Carolina Sommer, Founder/CEO

Phone: 425-894-8448

Email: registry@bornahero.org

Sponsor: Born a Hero Research Foundation

Key Information

You are invited to take part in a research study for individuals with a syndrome caused by a mutation in an FGFR gene on behalf of the person in your care. We hope that this form will help you decide whether or not to participate, but you can also call or e-mail the study staff at the contacts above if you have any other questions.

Things you should know:

We are doing this research to gain a more comprehensive understanding of the approximately 15 FGFR Syndromes involving mutations in FGFR1, FGFR2, or FGFR3 genes, including the relationship between disease symptoms and genes. The hope is to gather information that will help researchers and clinicians develop better ways to treat affected patients. The syndromes included are:

1. Antley-Bixler syndrome without genital anomalies or disordered steroidogenesis
2. Apert syndrome
3. Beare-Stevenson cutis gyrata syndrome
4. Bent bone dysplasia syndrome
5. CATSHL syndrome (Camptodactyly, tall stature, and hearing loss syndrome)
6. Crouzon syndrome
7. Crouzon syndrome with acanthosis nigricans
8. Hartsfield syndrome (holoprosencephaly, ectrodactyly, and bilateral cleft lip/palate)
9. Hypogonadotropic hypogonadism 2 with or without anosmia (Kallmann syndrome with anosmia)
10. Jackson-Weiss syndrome
11. LADD syndrome (Lacrimalauriculodentodigital syndrome)
12. Muenke syndrome
13. Osteoglophonic dysplasia
14. Pfeiffer syndrome, type 1, 2 and 3
15. Thanatophoric dysplasia, type I and 2

If you choose to participate on behalf of the participant, you will be asked to complete the survey initially, and to update approximately once a year, or when there are new events to report. The initial report may take an average of 2 hours to complete, depending on how much history and updates should take less than one hour each. The survey does not need to be completed in real-time. You can save your responses and return at your convenience.

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Consent to FGFR Syndromes Research Registry

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Authorization

The following statements are intended to:

- Make sure that you have had the time and opportunity to consider whether you and the Study Participant want to participate in this registry;
- Have had your questions answered; and
- Agree to participate in the study as described.

You will be asked to acknowledge:

- That you have read the consent form and have no further questions about the registry and the Study Participant's participation;
- That you wish to provide the Study Participant's personal data to the registry for the purposes of the Study;
- That you allow for this data to be used for future research;
- That you have explained the study to the Study Participant to the extent they are able to understand; and
- That you are of legal age.

This is a web-based form. Your digital signature is the same as if you had signed your name to a paper document. By answering "Yes" to all of the following statements, you are giving your consent to participate in the FGFR Syndromes Registry on behalf of the Study Participant. After signing, a copy of the consent form will be e-mailed to you. If you cannot comfortably answer "Yes" to these statements, please do not check the consent boxes in the following section.

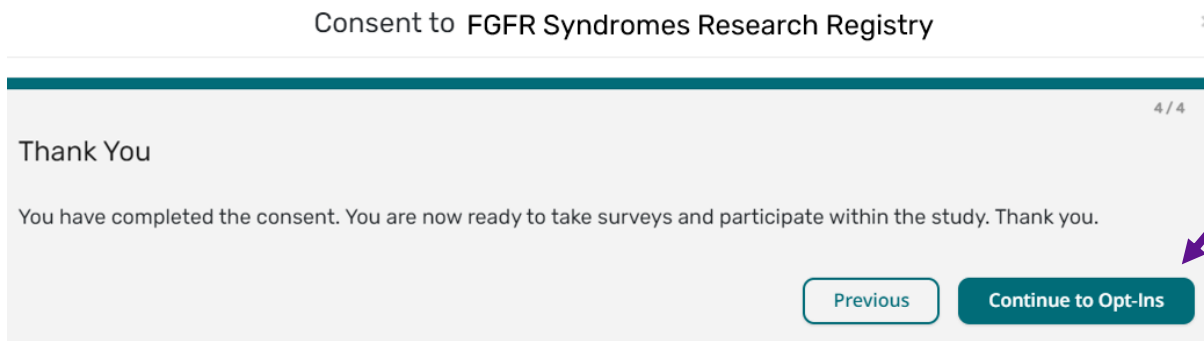
I have read this Consent and Authorization Form to provide the Study Participant's personal and medical data to be shared for the purpose of research. All my questions about the FGFR Syndromes Registry have been answered to my satisfaction, and I understand the purpose of the registry and the risks of participation.

I wish to provide the Study Participant's research data to the FGFR Syndromes Registry for the purposes described above under Study Aims.

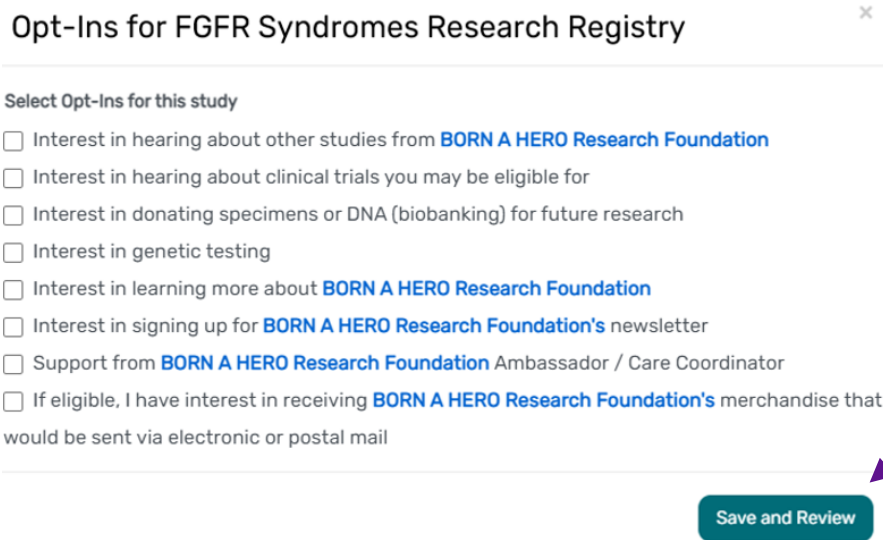
I wish to provide the Study Participant's research data to the FGFR Syndromes Registry for future research within recognized ethical standards for scientific research, as described under How We Use The Data.

[Previous](#) [Next](#)

- Step 3: Once you click “Next” and reach the Thank You page, click “Continue to Opt-Ins”.

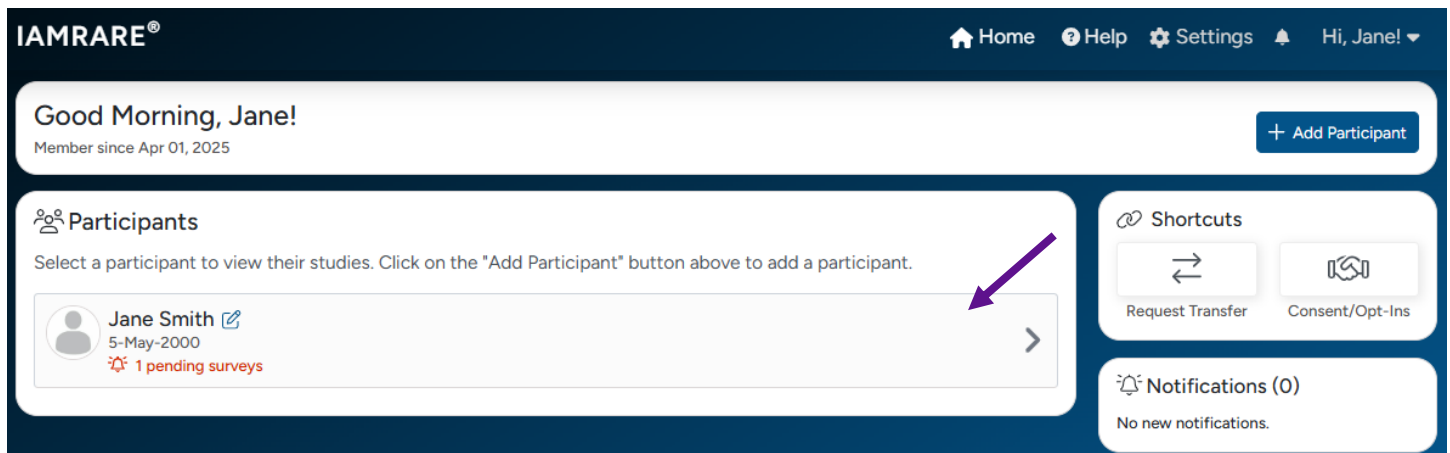


- Step 4: Once you click “Continue to Opt-Ins” read through the opt-ins thoroughly. If you would like to receive information about the topic, check the box, and click “Save and Review”.



Taking Surveys

- Step 1: Click on your Participant.



- Step 2: Click on the appropriate study.

Back to participant list

Jane Smith 5-May-2000

Search Studies

Enrolled Studies

Click a study to see the list of surveys. Click the **i** icon to see more information about the study. Click "Search Studies" above to find additional studies.

English

FGFR Syndromes Research Registry **i**

Born a Hero, Research Foundation

Consented

1 pending surveys

Shortcuts

Request Transfer Consent/Opt-Ins

Notifications (0)

No new notifications.

- Step 3: Click "Take Survey" for an available survey.

Back to study list

Jane Smith 5-May-2000

FGFR Syndromes Research Registry

Surveys **1 pending**

All (1) Complete (0) Pending (1)

0% Getting Started Not Started

Take Survey

View Responses and Reports

- Step 1: Once you have submitted a survey, you are able to view your responses to that survey as well as the graphs for any questions that are programmed to show graphs. Click "View Responses" to see your completed survey. Click "Reports" to see any available graphs.

Jane Smith 5-May-2000

FGFR Syndromes Research Registry

Surveys **14 pending**

All (15) Complete (1) Pending (14)

Getting Started Completed on 1-Apr-2025

View Responses (1)

Reports

View Consent and Opt-Ins

- Step 1: Once you have consented to the study, you are able to view your consent at any time. Click “Consents/Opt-Ins” to see your consent and opt-ins. You may revoke your consent at any time by clicking “Revoke”. You may also edit your Opt-Ins by clicking “Opt-Ins”.

← Back to participant list

Jane Smith 5-May-2000

Search Studies

Learn about searching for studies

Enrolled Studies

Click a study to see the list of surveys. Click the ⓘ icon to see more information about the study. Click "Search Studies" above to find additional studies.

Shortcuts

Request Transfer

Consent/Opt-Ins

- Step 2: You may revoke consent at any time by clicking “Revoke”. You may also edit your Opt-Ins by clicking “Opt-Ins”.

← Back to study list

Jane Smith 5-May-2000

Consents/Opt-Ins

Study Name	Consent Status	Consented On	Actions
FGFR Syndromes Research Registry	✓ Consented	1-Apr-2025	View Consent Revoke Opt-Ins

Dark Mode Settings

- Step 1: You can view the platform in Dark Mode. First, click Settings.

IAMRARE®

Home Help Settings Hi, Jane!

Good Morning, Jane!

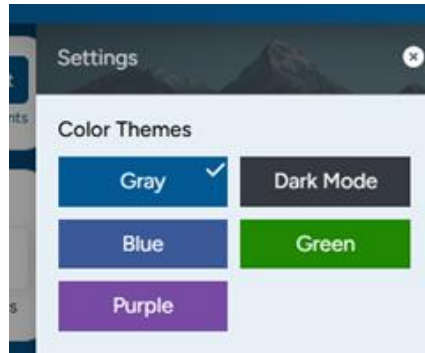
Member since Apr 01, 2025

+ Add Participant

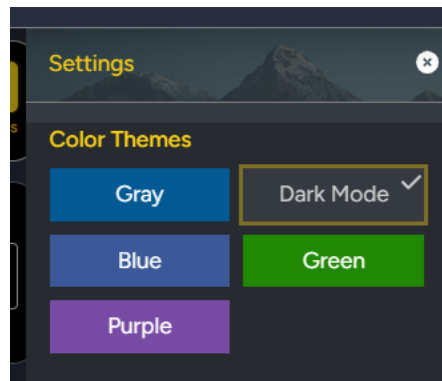
Participants

Shortcuts

- Step 2: Select Dark Mode.



- Step 3: Exit the Settings menu, and your selection will be saved.

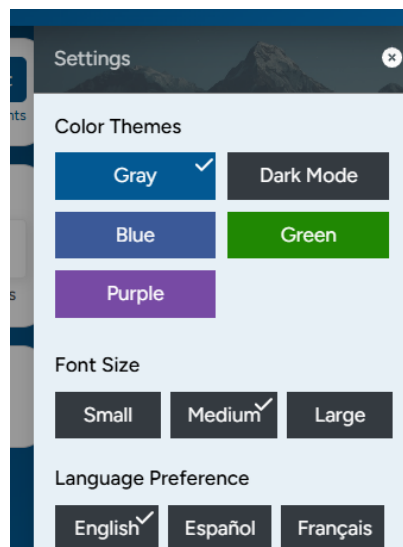


Display Settings

- Step 1: You can change the platform display settings. First, click Settings.



- Step 2: Select a color theme, a font size, or language preference.



Microsite Visibility

- Step 1: You can change how you view the microsite (fgfr.iamrare.org) using an Accessibility menu. Click the icon of a person at the bottom of the screen. You are able to change the settings such as the contrast, text sizing, and text spacing.



For Researchers

Drive Research

This is a unique rare disease patient registry. Are you interested in using our data to further your rare disease research?

LEARN MORE

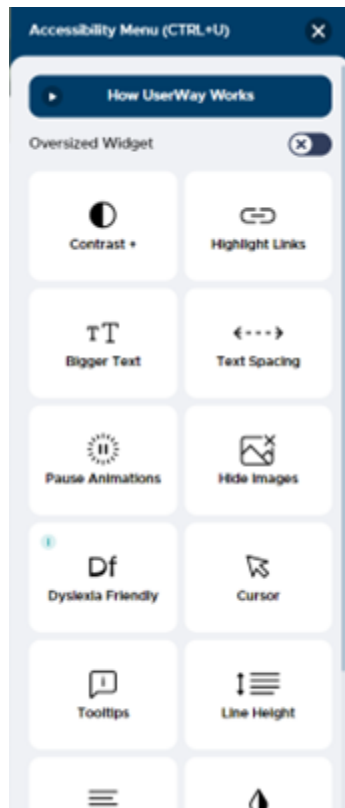


For Patients

Get Involved

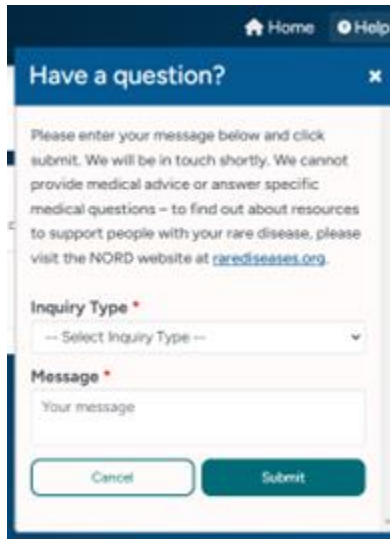
Information collected during this study may be used to help provide opportunities for patients and researchers to collaborate in the rare disease community.

LEARN MORE



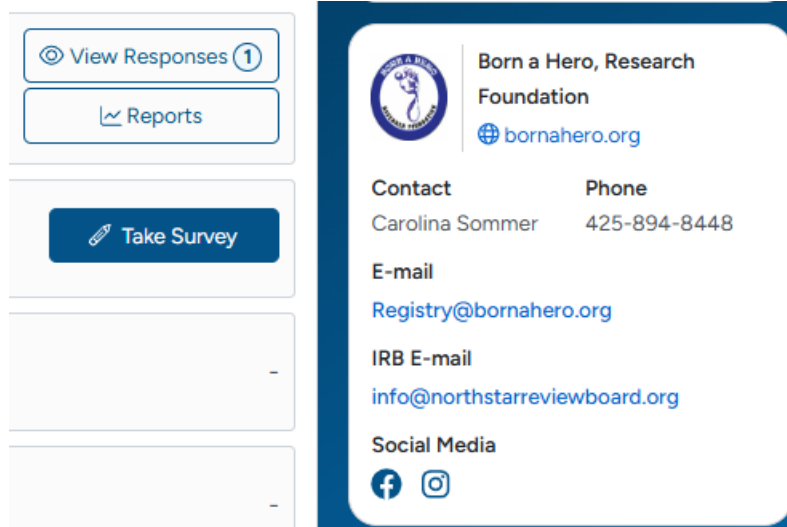
Need Assistance?

- Step 1: If you need help while using the platform, click Help.
- Step 2: Select an Inquiry Type and type a message.



The screenshot shows a mobile application interface with a blue header containing 'Home' and 'Help' icons. Below the header is a modal window titled 'Have a question?' with a close button. The modal contains a text area for a message, a dropdown menu for 'Inquiry Type' (currently showing '-- Select Inquiry Type --'), and a 'Message' field with the placeholder text 'Your message'. At the bottom of the modal are 'Cancel' and 'Submit' buttons.

- Step 3: Click Submit.
- You may also contact the study sponsor directly by using the contact information shown on your dashboard or the study website.



The image shows a dashboard with navigation buttons on the left and contact information on the right. The navigation buttons include 'View Responses' (with a notification badge '1'), 'Reports', and 'Take Survey'. The contact information is for the Born a Hero, Research Foundation, with the website bornahero.org. It lists contact details for Carolina Sommer (425-894-8448), an email address Registry@bornahero.org, and an IRB email address info@northstarreviewboard.org. Social media icons for Facebook and Instagram are also present.

Contact	Phone
Carolina Sommer	425-894-8448

E-mail
Registry@bornahero.org

IRB E-mail
info@northstarreviewboard.org

Social Media
Facebook, Instagram