




Chromosome 17q12 Deletion & Duplication Syndromes Patient Registry (17q12 Patient 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



17q12
FOUNDATION

Registration

Terms & ConditionsContact InfoNotificationsReview & SubmitConfirmation

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

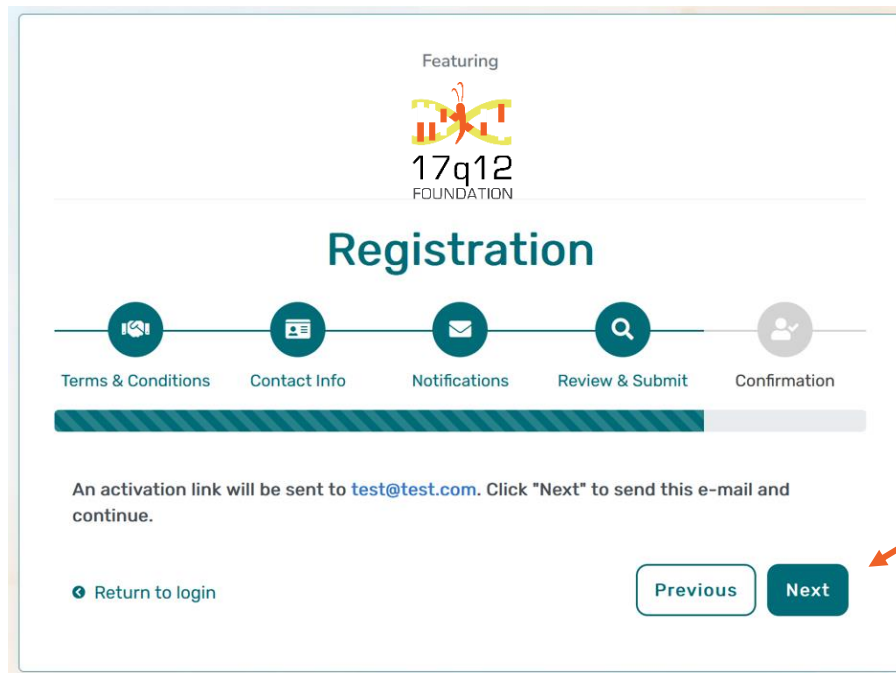
- Step 2: Enter your personal information in the spaces provided. When you are finished with this page, click “Next”.

The screenshot shows the 'Registration' page for the 17q12 FOUNDATION. At the top, it says 'Featuring' above a logo of a DNA double helix with a red ribbon. Below the logo is the text '17q12 FOUNDATION'. The main heading is 'Registration'. A progress bar with five steps is shown: 'Terms & Conditions', 'Contact Info', 'Notifications', 'Review & Submit', and 'Confirmation'. The 'Contact Info' step is currently active, indicated by a blue bar. Below the progress bar, there are form fields for 'Country of Residence' (a dropdown menu), 'First Name' (a text input field), 'Last Name' (a text input field), and 'E-mail' (a text input field). At the bottom, there is a 'Return to login' link and two buttons: 'Previous' and 'Next'.


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

The screenshot shows the 'Registration' page for the 17q12 FOUNDATION, specifically the 'Notifications' step. The progress bar at the top shows five steps: 'Terms & Conditions', 'Contact Info', 'Notifications', 'Review & Submit', and 'Confirmation'. The 'Notifications' step is currently active, indicated by a blue bar. Below the progress bar, there is a question: 'I am interested in NORD contacting me regarding available studies.*'. Below the question, there are two radio buttons: 'Yes' (selected) and 'No'. At the bottom, there is a 'Return to login' link and two buttons: 'Previous' and 'Next'.

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



Featuring


17q12
FOUNDATION

Registration

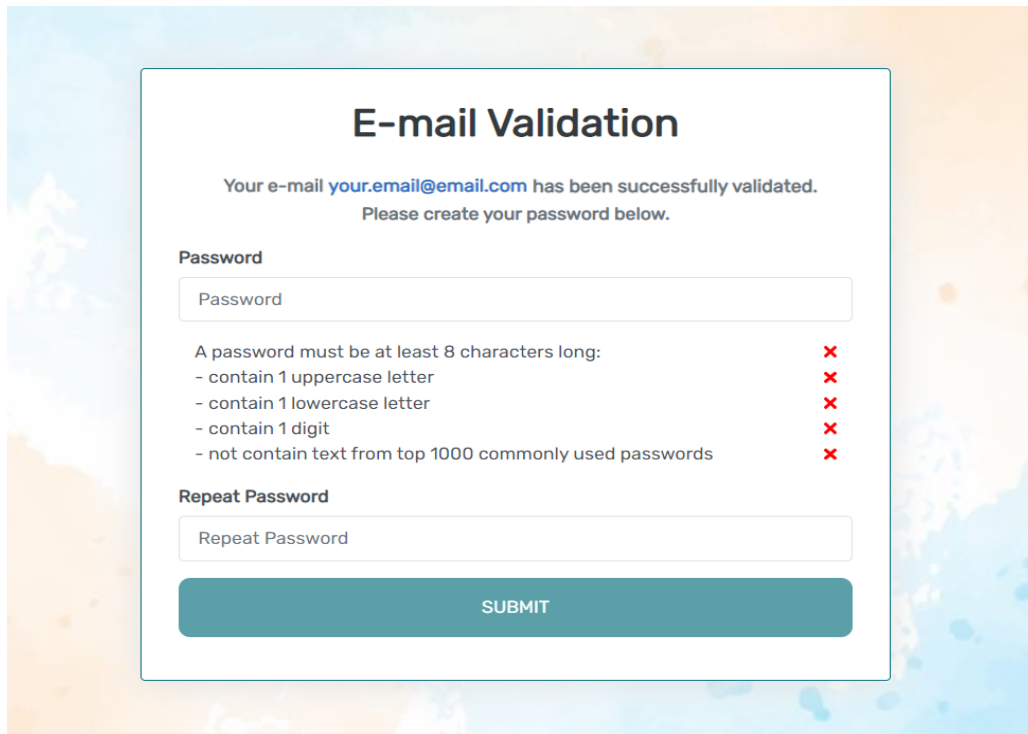
Terms & Conditions Contact Info Notifications Review & Submit Confirmation

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

[Return to login](#) [Previous](#) [Next](#)

An orange arrow points to the "Next" button.

- 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

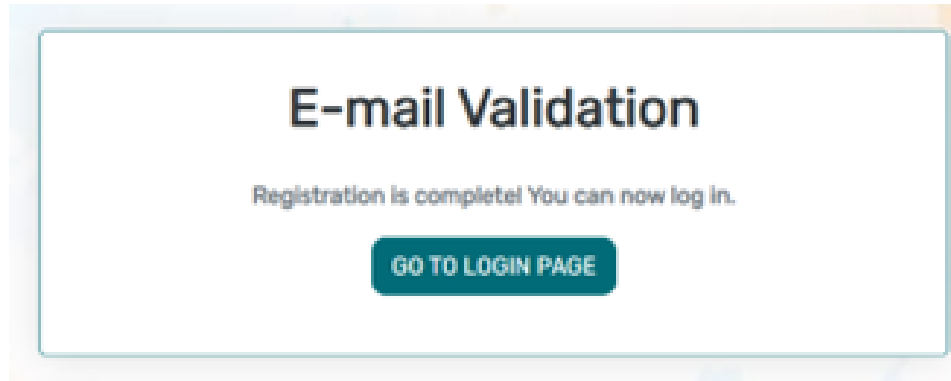
A password must be at least 8 characters long: ✗

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

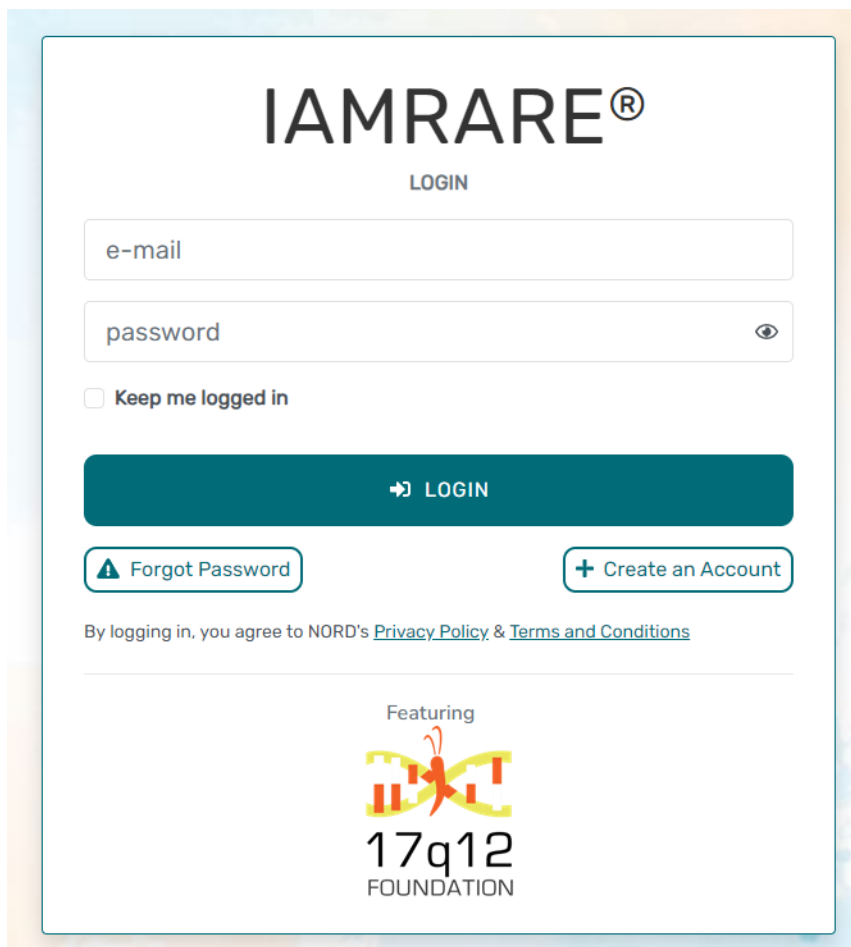
Repeat Password

[SUBMIT](#)

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




- Step 8: Log in using your new e-mail and password.

A screenshot of the IAMRARE LOGIN page. The page has a white background with a teal border. At the top, it says "IAMRARE®" in large black letters, followed by "LOGIN" in smaller black letters. Below this are two input fields: "e-mail" and "password". The "password" field has a teal eye icon to its right. Below the input fields is a checkbox labeled "Keep me logged in". Below the checkbox is a large teal button with a white right-pointing arrow and the word "LOGIN". Below the button are two smaller buttons: "Forgot Password" (with a teal warning icon) and "Create an Account" (with a teal plus icon). Below these buttons is a line of text: "By logging in, you agree to NORD's [Privacy Policy](#) & [Terms and Conditions](#)". At the bottom, it says "Featuring" above a logo for the 17q12 FOUNDATION. The logo consists of a stylized DNA double helix in yellow and red, with the text "17q12" in large black letters and "FOUNDATION" in smaller black letters below it.

Add a Participant

- Step 1: To start, click Create New Profile.

English ▾


17q12
FOUNDATION

Welcome, Jane!

Welcome to the IAMRARE[®] program, home of **Chromosome 17q12 Deletion & Duplication Syndromes Patient Registry (17q12 Patient 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.


17q12
FOUNDATION

Add a Participant

Are you adding yourself or another person?

Yourself

Someone else

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

Add Participant

Who Is Being Added as a Participant? ?

☐ Self
☒ Other

Preferred First Name *

Current Last name *

First Name on Birth Certificate *

Middle Name on Birth Certificate *

Last Name on Birth Certificate *

Date of Birth * ?

Sex Recorded on Birth Certificate * ?

Country of Residence * ?

State/Province/Region of Residence * ?


Country of Birth *

City/Municipality of Birth *

What Is Your Relationship to ? * ?

Consent to the Study

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



Thank you for registering your first participant!

Would you like to consent to participate in the **Chromosome 17q12 Deletion & Duplication Syndromes Patient Registry (17q12 Patient Registry)** ?

- 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.”

John Smith

Consent to Chromosome 17q12 Deletion & Duplication Syndromes Patient Registry (17q12 Patient Registry)

Consent Overview

Those eligible to participate in our study include:

Participant: An individual diagnosed with chromosome 17q12 deletion or duplication 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 chromosome 17q12 deletion or duplication syndrome, defined as a spouse, parent, sibling, offspring, close relative, close friend, guardian and/or significant other of the individual who had chromosome 17q12 deletion or duplication 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.

Please tell us about the Participant you would like to enroll in this study. *

☐ 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 chromosome 17q12 deletion or duplication syndrome. I participated in their medical care.

Next

John Smith

Consent to Chromosome 17q12 Deletion & Duplication Syndromes Patient Registry (17q12 Patient Registry)

Consent for a Person with a Legally Authorized Representative (Caregiver)

Title: Chromosome 17q12 Deletion and Duplication Syndromes Patient Registry (17q12 Patient Registry)

Principal Investigator: Margo Casados, 17q12 Foundation Board Member

Co-Investigator: Elizabeth Fourie, Vice President, 17q12 Foundation

Phone: 515-329-5877

Email: info@chromo17q12.org

Sponsor: 17q12 Foundation

Key Information

You are invited to take part in a research study for individuals with chromosome 17q12 deletion or duplication syndrome 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 email the study staff at the contacts above if you have any other questions.

Things you should know:

We are doing this research to collect information about chromosome 17q12 deletion and duplication syndromes to learn more about how the 17q12 patient community is affected.

If you choose to participate on behalf of the participant, you will be asked to fill in personal information, submit a genetic lab report, and complete surveys over the internet to a secure database. The amount of time the surveys take will vary Participant to Participant. You can save your progress and come back to it as needed.

Participating in the Chromosome 17q12 Deletion and Duplication Syndromes Patient Registry does not pose any physical risks, but some questions in the surveys may be unpleasant or uncomfortable for some participants.

Participating in our study may not help the Study Participant directly, but your time and information may help others with chromosome 17q12 deletion and duplication syndromes in the future. The

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

John Smith

Consent to Chromosome 17q12 Deletion & Duplication Syndromes Patient Registry (17q12 Patient Registry)

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 17q12 Patient 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 17q12 Patient 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 17q12 Patient Registry for the purposes described above under Study Aims.

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

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

John Smith

Consent to Chromosome 17q12 Deletion & Duplication Syndromes Patient Registry (17q12 Patient Registry)

Please continue to select your opt-ins. Once you have made your selections, please click Save and Review. You will then be ready to take surveys and participate in this study.

[Previous](#) [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".

Opt-Ins for Chromosome 17q12 Deletion & Duplication Syndromes Patient Registry (17q12 Patient Registry) ×

Select Opt-Ins for this study

- ☐ Interest in hearing about other studies from [17q12 Foundation](#)
- ☐ Interest in hearing about relevant clinical trials
- ☐ Interest in donating specimens or DNA (biobanking) for future research
- ☐ Interest in genetic testing
- ☐ Interest in learning more about [17q12 Foundation](#)
- ☐ Interest in signing up for a [17q12 Foundation](#) newsletter

[Save and Review](#)

- Step 5: Once you've reviewed your consent, click "Close". You will then have access to start taking surveys.

Taking Surveys

- Step 1: Click on your Participant.

IAMRARE®
Home
Help
Settings
Hi, John!

Good Morning, John!
Member since Mar 27, 2025
+ Add Participant

Participants
Select a participant to view their studies. Click on the "Add Participant" button above to add a participant.

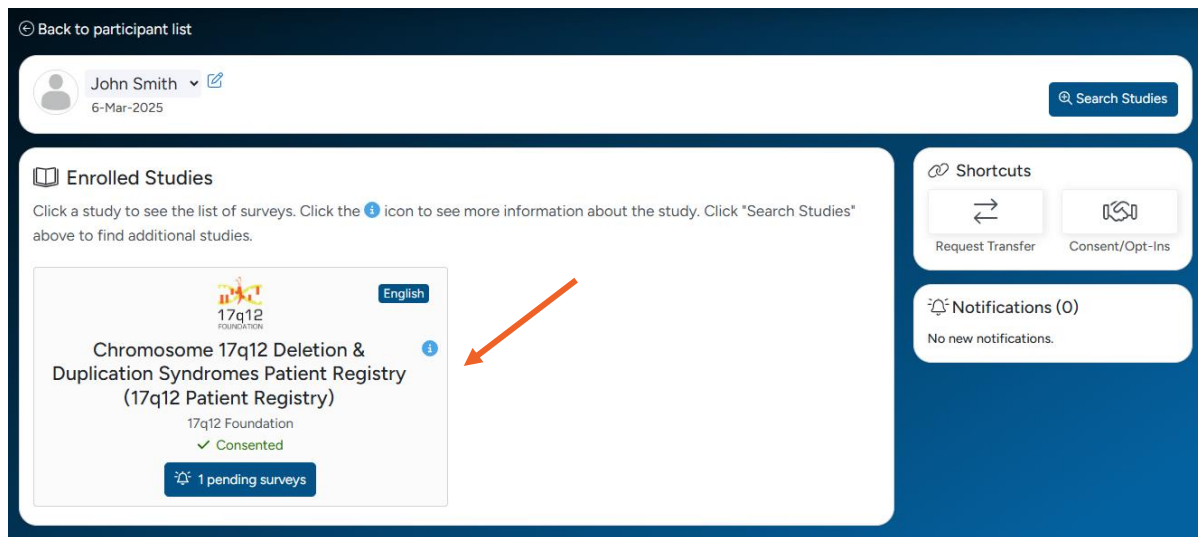
John Smith
6-Mar-2025
1 pending surveys

Shortcuts

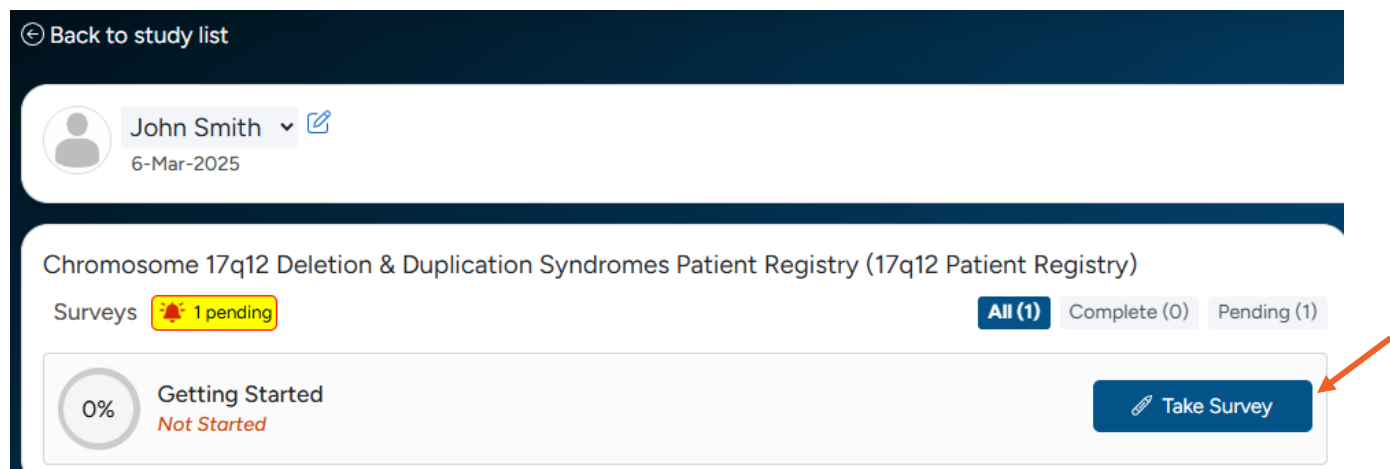
Request Transfer
Consent/Opt-Ins

Notifications (0)
No new notifications.

- Step 2: Click on the appropriate study.

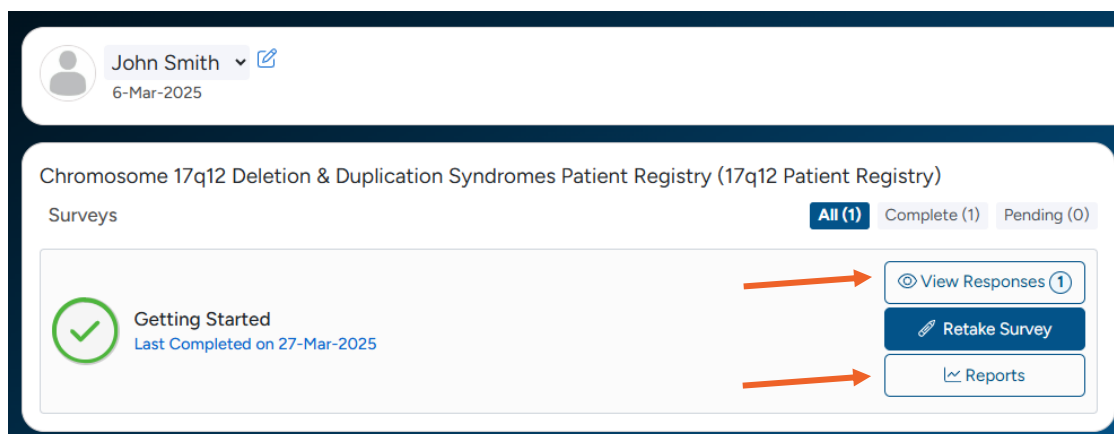


- Step 3: Click “Take Survey” for an available survey.



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.



View Consent and Opt-Ins

- Step 1: Once you have consented to the study, you are able to view your consent at any time. Navigate to the Enrolled Studies page. Then, click “Consents/Opt-Ins” to see your consent and opt-ins.

Back to participant list

John Smith 6-Mar-2025

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.

Chromosome 17q12 Deletion & Duplication Syndromes Patient Registry (17q12 Patient Registry)

17q12 Foundation

Consented

No Pending Surveys

Shortcuts

Request Transfer

Consent/Opt-Ins

Notifications (0)

No new notifications.

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

Back to study list

John Smith 6-Mar-2025

Consents/Opt-Ins

Study Name	Consent Status	Consented On	Actions
Chromosome 17q12 Deletion & Duplication Syndromes Patient Registry (17q12 Patient Registry)	Consented	25-Oct-2024	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, John!

Good Morning, John!

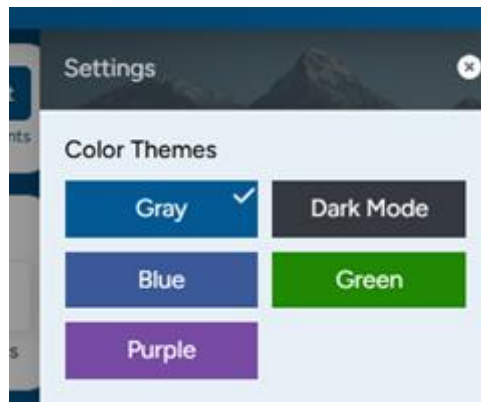
Member since Mar 27, 2025

Add Participant

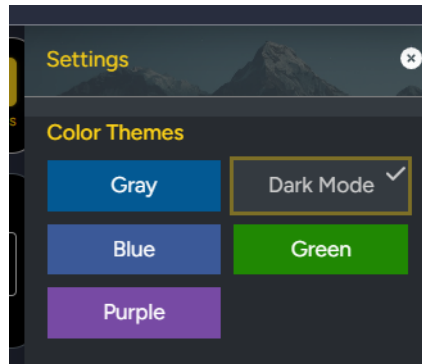
Participants

Shortcuts

- Step 2: Select Dark Mode.



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

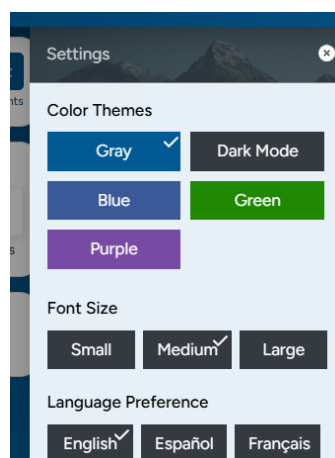


Display Settings

- 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 (17q12registry.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

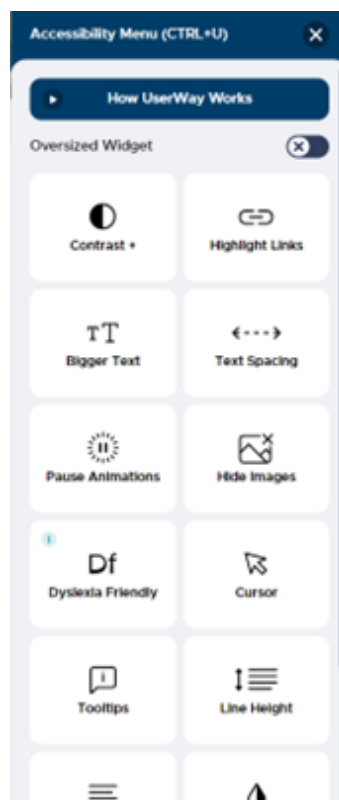


For Patients

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.

Have a question?

Please enter your message below and click submit. We will be in touch shortly. We cannot provide medical advice or answer specific medical questions – to find out about resources to support people with your rare disease, please visit the NORD website at rarediseases.org.

Inquiry Type *

-- Select Inquiry Type --

Message *

Your message

Cancel Submit

- 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.

Registry

Complete (1) Pending (0)

View Responses (1)

Retake Survey

Reports

Notifications (0)

No new notifications.

17q12 Foundation

chromo17q12.org

Contact	Phone
Liz Fourie	515-329-5877
E-mail	
info@chromo17q12.org	
IRB E-mail	
info@northstarreviewboard.org	
Social Media	
f t i	