




Chromosome 17q12 Deletion & Duplication Syndromes Patient Registry (17q12 Patient Registry) Participant User Guide

Register for an Account

- Step 1: Select the appropriate Account Type. If you need more information to help you choose, click “Not sure? Help me choose”.
 - If **you** have a diagnosis of 17q12 Deletion of Duplication Syndrome, select **Participant Account**.
 - If you are entering information for **someone else** who has 17q12 Deletion of Duplication Syndrome, select **Caregiver Account**.
 - If you are entering information for a 17q12 Deletion of Duplication Syndrome **patient who has passed away**, select **Caregiver Account**.

Featuring



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Select Account Type

I have a rare disease, condition, and/or diagnosis.

Participant Account

I am a family member or guardian of someone with a rare disease.

Caregiver Account

[Return to login](#) [Not sure? Help me choose.](#)

- Step 2: Read the Terms and Conditions and Privacy Policy and attest to the statements provided. When you are finished with this page, click “Next”.

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Caregiver 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 3: Enter your personal information in the spaces provided. When you are finished with this page, click “Next”.

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Caregiver Registration

Terms & Conditions Contact Info Notifications Review & Submit Confirmation

Country of Residence *

First Name * Last Name *

E-mail *

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Step 4: 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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I am interested in NORD contacting me regarding available studies. *

Yes No

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

- Step 5: Select “Next” so that an activation link is sent to your e-mail to complete registration.

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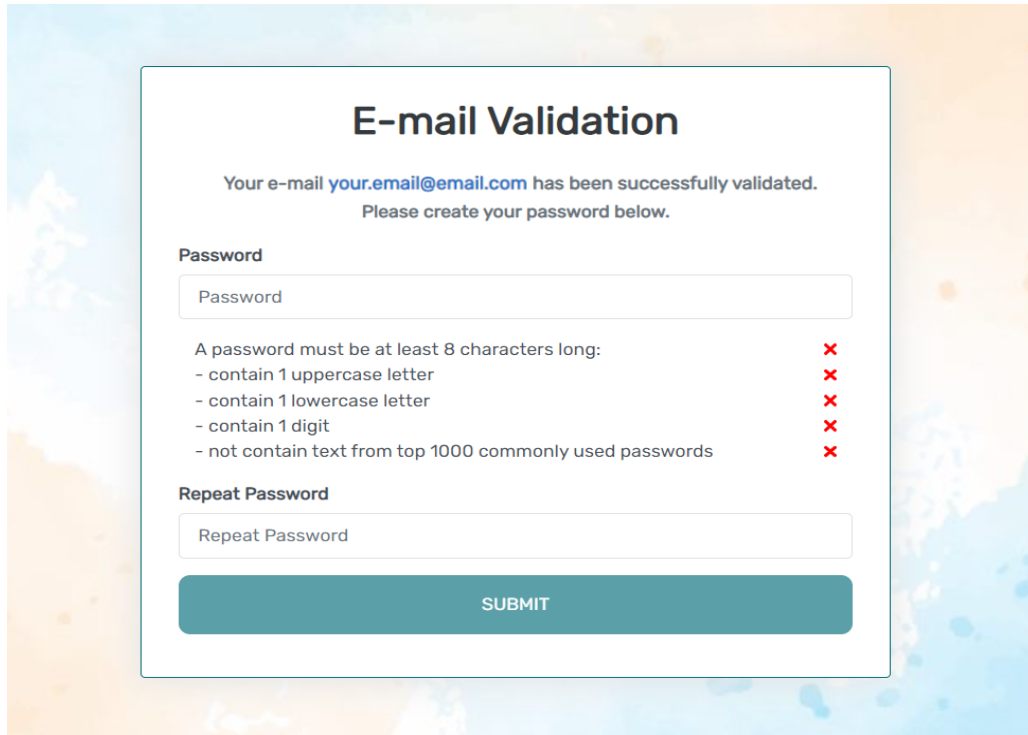
Caregiver Registration

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An activation link will be sent to [your.email@email.com](#). Click “Next” to send this e-mail and continue.

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

- Step 6: 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.

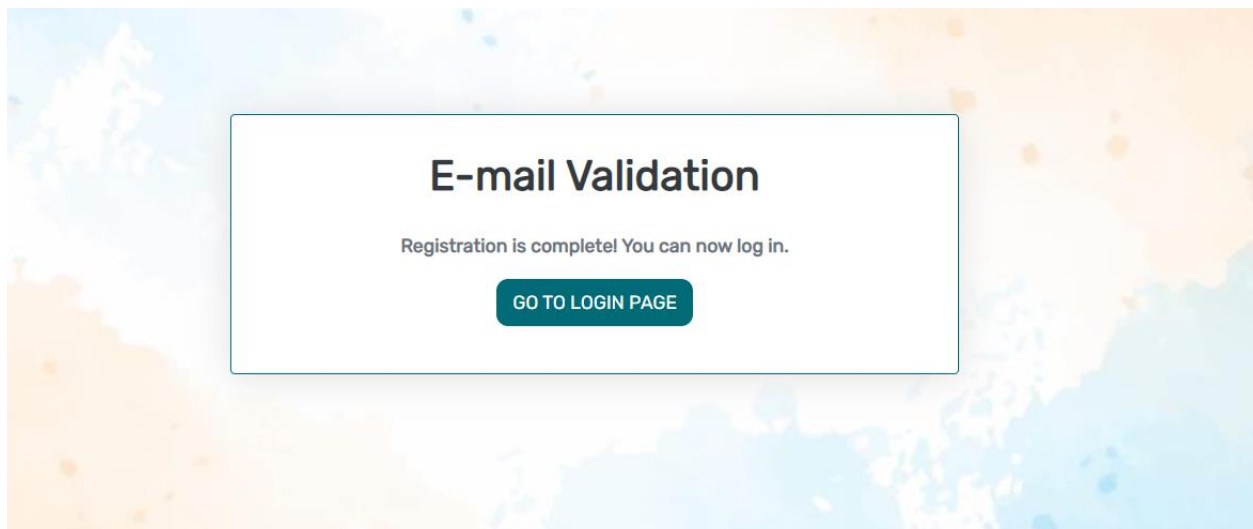
Password

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 7: Your validation is now complete. Select “Go to Login Page”.

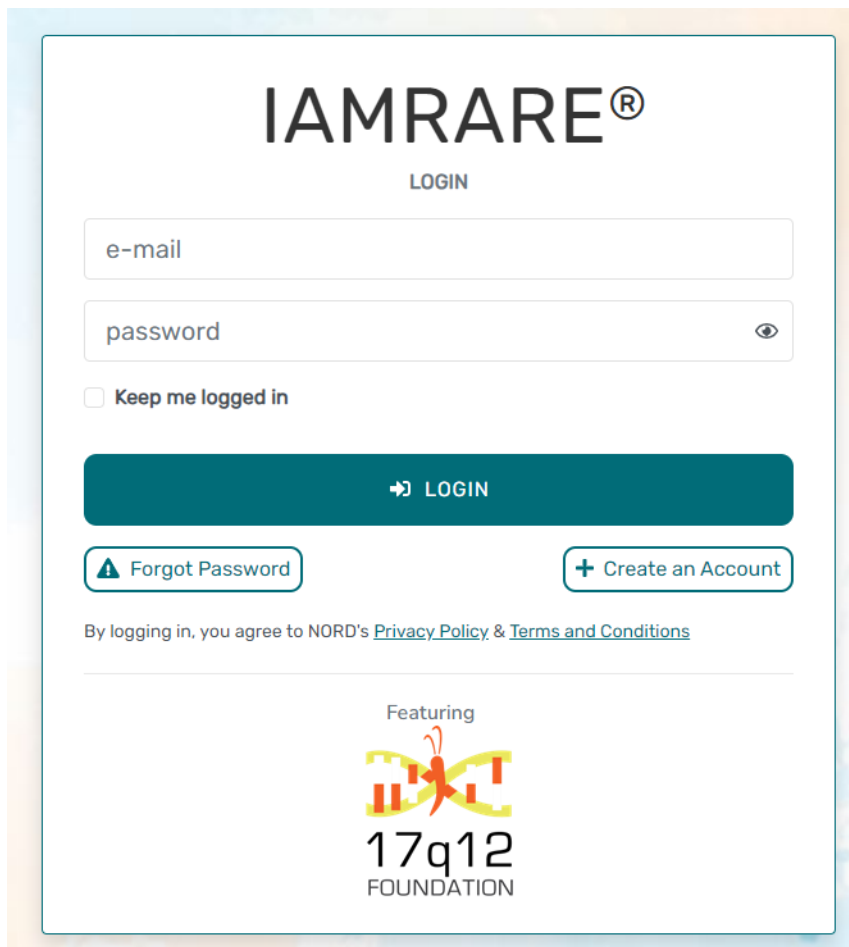


E-mail Validation

Registration is complete! You can now log in.

GO TO LOGIN PAGE

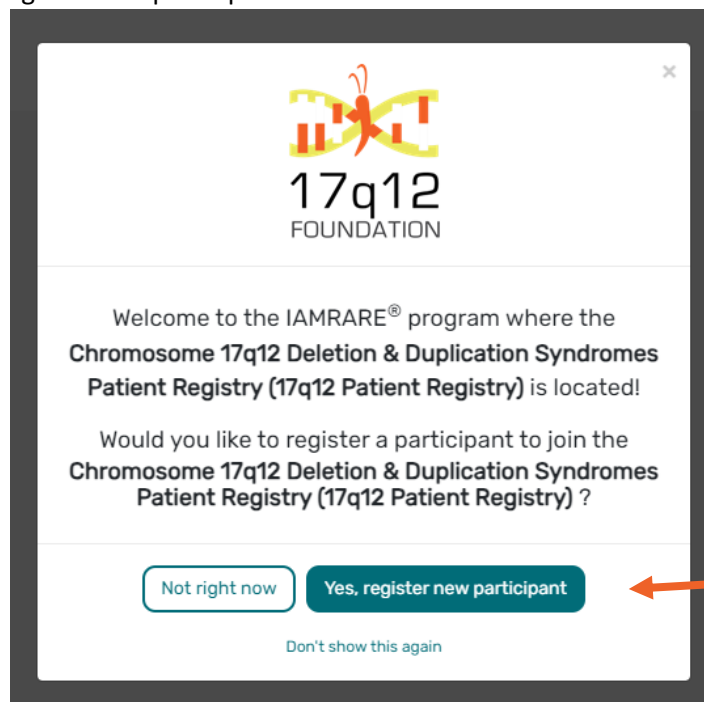
- Step 8: Log in using your new e-mail and password.



The image shows the IAMRARE LOGIN page. At the top, the text "IAMRARE®" is displayed in a large, bold, black font, with "LOGIN" centered below it in a smaller font. There are two input fields: "e-mail" and "password". The "password" field has an eye icon on the right side. Below the input fields is a checkbox labeled "Keep me logged in". A large teal button with a right-pointing arrow and the text "LOGIN" is centered. Below the button are two smaller buttons: "Forgot Password" with a warning triangle icon and "Create an Account" with a plus icon. At the bottom, there is a line of text: "By logging in, you agree to NORD's [Privacy Policy](#) & [Terms and Conditions](#)". Below this is a section titled "Featuring" with the 17q12 FOUNDATION logo, which includes a stylized DNA double helix with a red ribbon and the text "17q12 FOUNDATION".

Add a Participant

- Step 1: To start, click Yes, register new participant.



The image shows a registration dialog box for the 17q12 FOUNDATION. At the top, there is the 17q12 FOUNDATION logo, which consists of a stylized DNA double helix with a red ribbon and the text "17q12 FOUNDATION". Below the logo, the text reads: "Welcome to the IAMRARE® program where the **Chromosome 17q12 Deletion & Duplication Syndromes Patient Registry (17q12 Patient Registry)** is located!". This is followed by the question: "Would you like to register a participant to join the **Chromosome 17q12 Deletion & Duplication Syndromes Patient Registry (17q12 Patient Registry)** ?". At the bottom, there are two buttons: "Not right now" and "Yes, register new participant". An orange arrow points to the "Yes, register new participant" button. Below the buttons is a link that says "Don't show this again".

- Step 2: Fill out the Participant’s information.

Add Participant ✕

Acknowledgement*

By checking this box, you acknowledge that information collected on this platform will only be used for research purposes by NORD and in ways that will not reveal who you are. Federal or state laws may require us to show information to university or government officials (or sponsors) who are responsible for monitoring the safety of any studies running on this platform. You will not be identified in any publications.

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 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.”


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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.”

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

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. This will take approximately _____.

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 direct benefits of participation are that you can access charts and graphs of the combined data contributed to the registry by all of the study participants. This data does not reveal identities. Being included in the registry will give you access to ongoing 17q12 community opportunities.

It is up to you whether to participate in this study, and you can stop at any time. Please take time to read this entire form and ask questions before deciding whether to take part in this research project on behalf of the person in your care. As the guardian or legally authorized representative for the Study Participant, we encourage you to discuss the registry with the Study Participant to the extent compatible with their understanding. Detailed information about your participation in this study follows.

Purpose of this Informed Consent Document

This document will give you the information so you can decide if you want to join this study on behalf of the participant or not. This consent document is structured to follow the framework provided by federal regulations. While we hope the information we provide will answer most of your questions, it may not answer them all. If you have any remaining questions, please contact the Principal Investigator at the phone number or email listed above.

Definitions

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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.

I wish to provide the Study Participant's research data to the 17q12 Patient Registry for future research within recognized ethical standards for scientific research, as described under How We Use Your Data.

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

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.

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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 [17q12 Foundation's](#) newsletter

Save and Review

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



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


Consented

● You have 1 pending surveys.

[Search Studies](#)

Surveys 🔔 1 pending All (1) Complete (0) Pending (1)

● **Getting Started**
Not Started

[Take Survey](#) 

View Responses

- 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.

The screenshot shows the user's profile for the "Chromosome 17q12 Deletion & Duplication Syndromes Patient Registry (17q12 Patient Registry)". The status is "Consented" and it notes "You have no pending surveys." Below this is a "Search Studies" button. A "Surveys" section lists two completed surveys: "Getting Started" and "Demographics", both completed on 5-Jun-2023. For each survey, there are "View Responses" and "Reports" buttons. Two orange arrows point to the "View Responses" buttons for the "Demographics" survey.

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”.

The screenshot shows the "CONSENTS/OPT-INS" page for a participant named Jane Smith (DOB: 5-May-2000). On the left, a sidebar menu has "Consents/Opt-Ins" selected, indicated by an orange arrow. The main table lists the study: "Chromosome 17q12 Deletion & Duplication Syndromes Patient Registry (17q12 Patient Registry)". The consent status is "Consented" (with a green checkmark), and the date is "26-Jul-2023". In the "Actions" column, there are buttons for "View Consent", "Revoke", and "Opt-Ins". An orange arrow points to the "View Consent" button. The page is labeled "Page 1 of 1" and has a navigation bar at the bottom.