




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
KAT6
Foundation

Registration

Terms & Conditions Contact Info Notifications Review & Submit Confirmation

Country of Residence *

First Name * Last Name *

E-mail *

[Return to login](#) [Previous](#) [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”.

Featuring
KAT6
Foundation

Registration

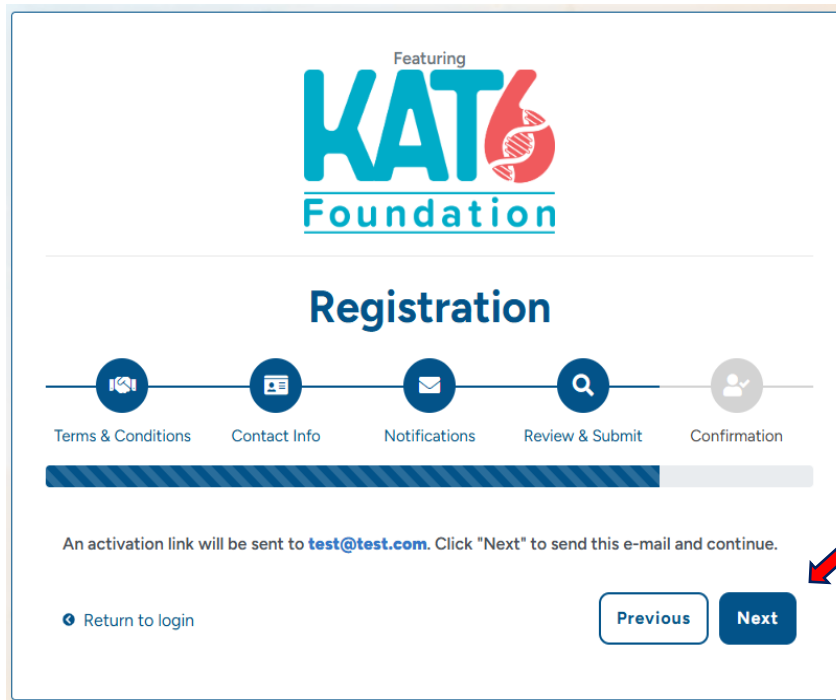
Terms & Conditions Contact Info Notifications Review & Submit Confirmation

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

Yes No

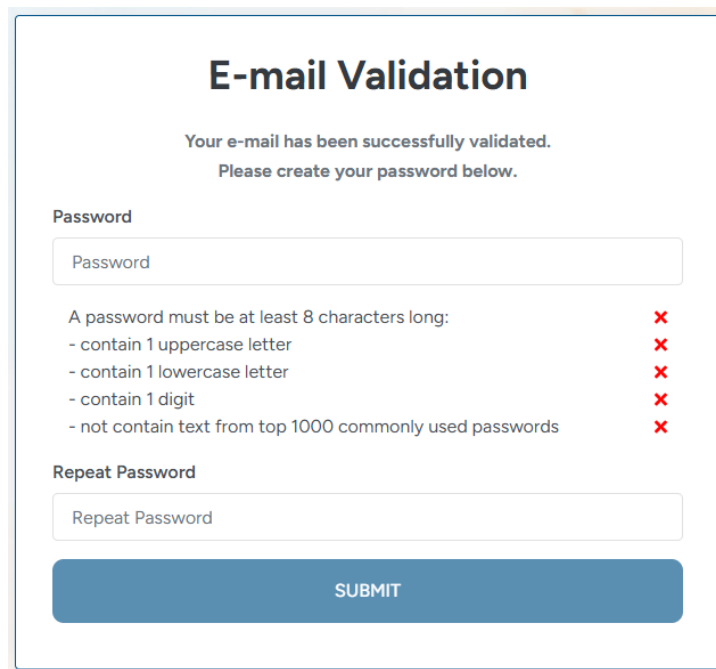
[Return to login](#) [Previous](#) [Next](#)

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



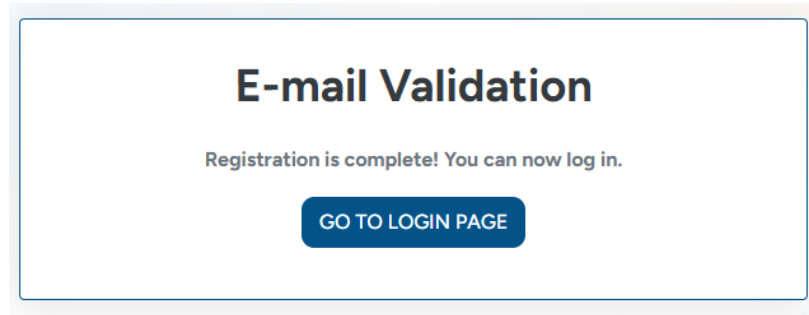
The image shows a registration progress screen for the KAT6 Foundation. At the top, it says "Featuring KAT6 Foundation" with a logo. Below that, the word "Registration" is centered. A progress bar shows five steps: "Terms & Conditions", "Contact Info", "Notifications", "Review & Submit", and "Confirmation". The "Review & Submit" step is currently active. Below the progress bar, a message states: "An activation link will be sent to test@test.com. Click 'Next' to send this e-mail and continue." At the bottom, there is a "Return to login" link, a "Previous" button, and a "Next" button. A red 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”.

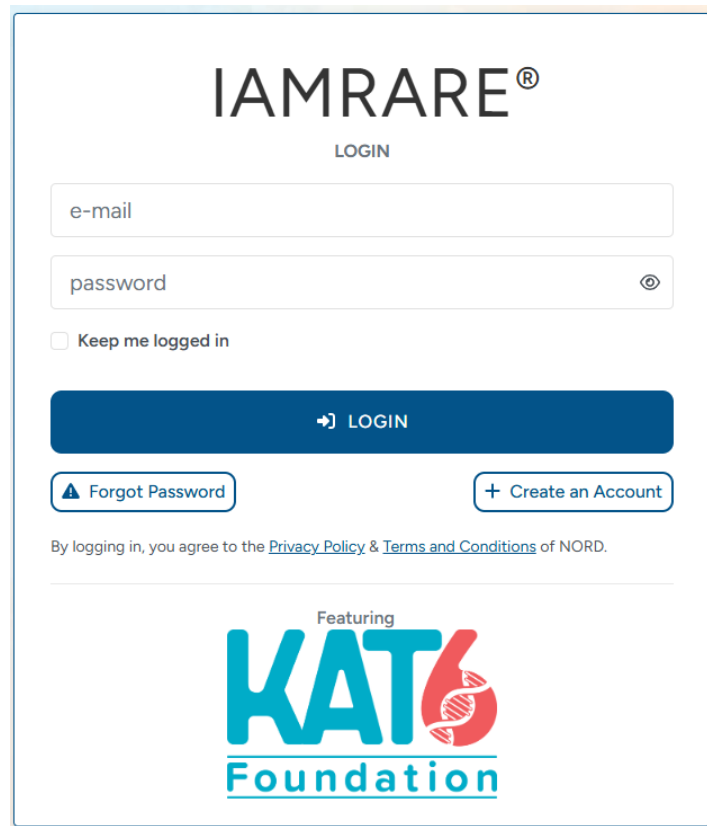


The image shows an "E-mail Validation" screen. It says "Your e-mail has been successfully validated. Please create your password below." There are two password input fields: "Password" and "Repeat Password". Below the "Password" field, there are four requirements listed with red 'X' marks indicating they are not met: "A password must be at least 8 characters long:", "- contain 1 uppercase letter", "- contain 1 lowercase letter", "- contain 1 digit", and "- not contain text from top 1000 commonly used passwords". At the bottom, there is a large blue "SUBMIT" button.

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

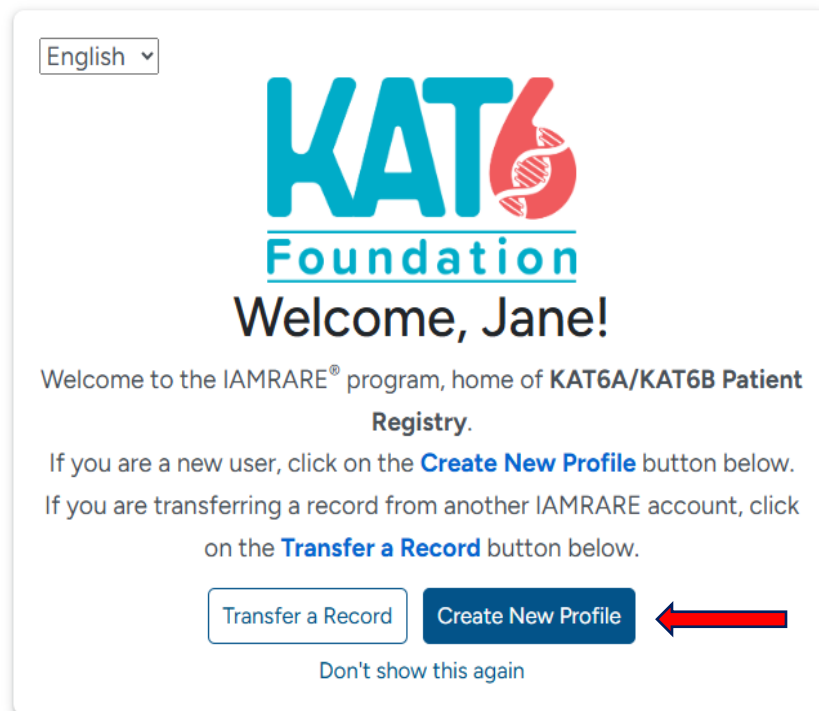


- Step 7: Log in using your new e-mail and password.

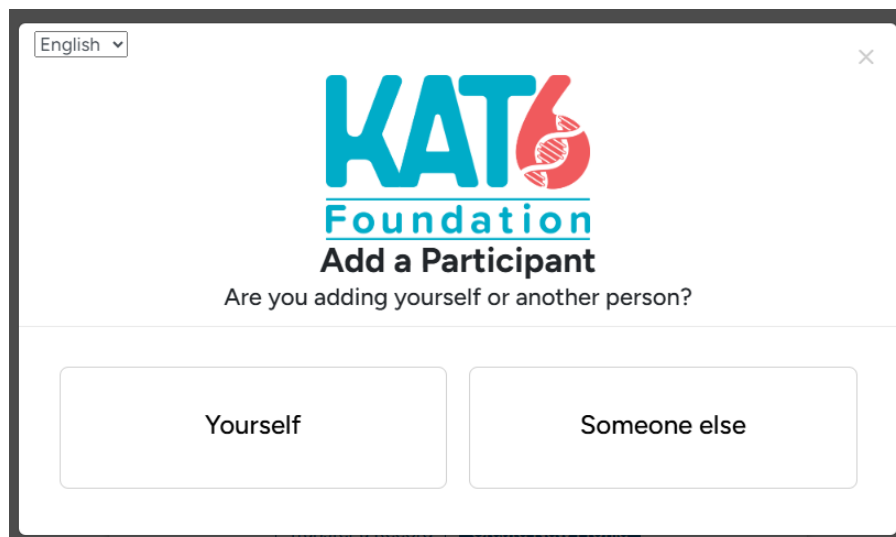


Add a Participant

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



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



- 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.”



- 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.”

Jane Smith

Consent to KAT6A/KAT6B Patient Registry

Consent Overview

Those eligible to participate in our study include:

Participant: An individual diagnosed with KAT6A/KAT6B 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 with KAT6A/KAT6B Syndrome who is a minor (child under the age of 18) or an adult who is unable to contribute their own data. The Legally Authorized Representative must 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. The Designated Representative is defined as a spouse, parent, sibling, offspring, close relative, close friend, guardian and/or significant other of the individual who had KAT6A/KAT6B Syndrome and had knowledge of and participated in their medical care. This Designated Representative must 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 KAT6A/KAT6B Syndrome. I participated in their medical care.

Next



Jane Smith

Consent to KAT6A/KAT6B Patient Registry

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

Consent to Participate in the KAT6A/KAT6B Patient Registry and to Allow Data to be Shared for Future Research

Title: KAT6/KAT6B Patient Registry

Principal Investigator: Susan Hartung

Phone: 240-372-3106

E-mail: registry@kat6.org

Sponsor: KAT6 Foundation

Key Information


You are invited to take part in a research study for people with KAT6A/KAT6B Syndrome on behalf of the person in your care who is not able to provide their own consent. Your role is called Legally Authorized Representative (LAR). This form will help you decide if you want to participate. You can contact the study staff above if you have any other questions.

Things you should know:

Patient registries have been shown to help speed the development of better treatments and ultimately improve patient outcomes. The KAT6A/KAT6B Patient Registry will:

- Provide new insights into the disease and identify areas for additional study.
- Facilitate partnerships with university researchers, medical, food and pharmaceutical companies.
- Guide the development of standards of care across the globe.
- Generate a greater number of clinical trials.
- Allow participants to store and access their medical information in one place.

Previous Next



Authorization

The following statements are intended to:

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

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 KAT6A/KAT6B 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 KAT6A/KAT6B 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 KAT6A/KAT6B Patient Registry for the purposes described above under Study Aims.
- I wish to provide the Study Participant's research data to the KAT6A/KAT6B Patient Registry for future research within recognized ethical standards for scientific research, as described under How We Use Your Data.
- I have explained the study to the Study Participant to the extent they are able to understand, and the Study Participant has given their assent to participate in this study.

Previous Next



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

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 KAT6A/KAT6B Patient Registry

Select Opt-Ins for this study

- Interest in hearing about other studies from [The KAT6A 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 [The KAT6A Foundation](#)
- Interest in signing up for a [The KAT6A Foundation](#) newsletter
- Support from [The KAT6A Foundation](#) Ambassador / Care Coordinator
- If eligible, I have interest in receiving [The KAT6A Foundation](#) merchandise that would be sent via electronic or postal mail
- Support from other Patient Advocacy Groups
- Interest in learning about upcoming events such as webinars and conferences

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 "Take Survey" for an available survey.

Back to the study list

Jane Smith 5-May-2000

KAT6A/KAT6B Patient Registry

Surveys 🔔 14 pending All (14) Complete (0) Pending (14)

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

KAT6A/KAT6B Patient Registry

Surveys 🔔 13 pending All (16) Complete (3) Pending (13)

Getting Started
Last Completed on 25-Oct-2024

View Responses 1
Retake Survey
Reports

Diagnosis
Completed on 25-Oct-2024

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. Navigate to the Enrolled Studies page. Then, click “Consents/Opt-Ins” to see your consent and opt-ins.

Back to the 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.

Shortcuts

Request Transfer Consent/Opt-Ins

- 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 the study list

Jane Smith 5-May-2000

Consents/Opt-Ins

Study Name	Consent Status	Consented On	Actions
KAT6A/KAT6B Patient Registry	✓ Consented	26-Jun-2025	View Consent Revoke Opt-Ins

Dark Mode Settings

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

IAMRARE®

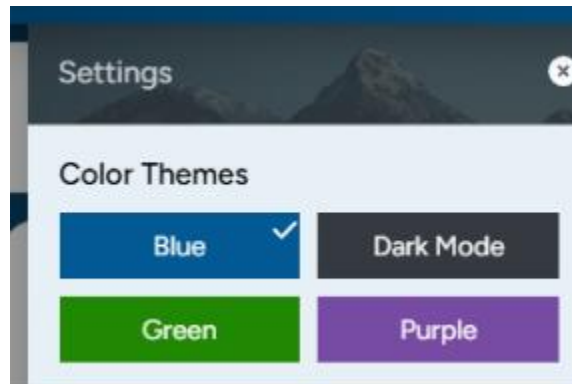
Home Help Settings Hi, Jane!

Good Afternoon, Jane!
Member since Jun 26, 2025

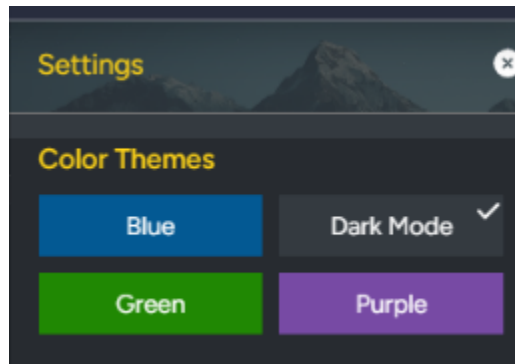
Add Participant

Participants Shortcuts

- Step 2: Select Dark Mode.



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

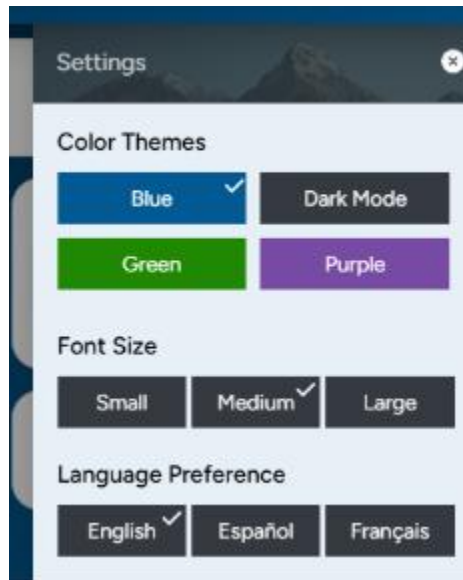


Display Settings

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



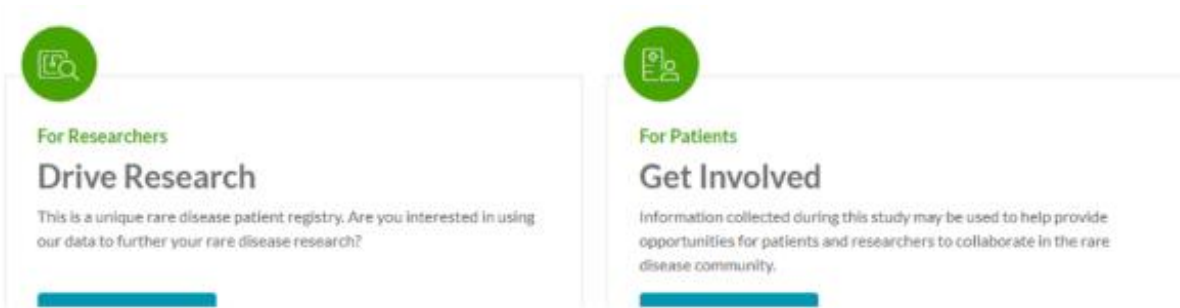
- Step 2: Select a color theme, a font size, or language preference.

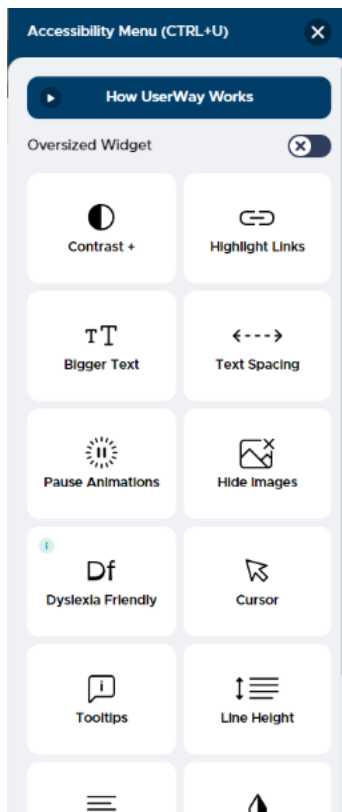


- Step 3: Exit the Settings menu, and your selection will be saved.

Microsite Visibility

- Step 1: You can change how you view the microsite (<https://kat6a.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.





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 image shows a mobile application's help form. At the top, there are "Home" and "Help" icons. The form title is "Have a question?" with a close button. Below the title is a paragraph of text: "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." Below this text are two required fields: "Inquiry Type *" with a dropdown menu showing "-- Select Inquiry Type --" and "Message *" with a text input field containing "Your message". At the bottom, there are two buttons: "Cancel" and "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.

The image shows a dashboard on the left with four buttons: two labeled "View Responses 1" and two labeled "Take Survey". To the right is a contact information card for the KAT6 Foundation. The card includes the logo, the text "The KAT6A Foundation" and "kat6a.org", and contact details for the foundation, an email registry, an IRB email, and social media links for Facebook, Twitter, and Instagram.

View Responses 1

View Responses 1

Take Survey

Take Survey

KAT6 Foundation | The KAT6A Foundation
kat6a.org

Contact
The KAT6A Foundation

E-mail
registry@kat6.org

IRB E-mail
info@northstarreviewboard.org

Social Media
f t i