



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”.

A screenshot of the NPKUA registration page. At the top, it says "Featuring" above the NPKUA logo. Below the logo is the word "Registration" in a large, bold, blue font. A progress bar with five steps is shown: "Terms & Conditions" (active, highlighted in blue), "Contact Info", "Notifications", "Review & Submit", and "Confirmation". Below the progress bar, there is a paragraph of text explaining the purpose of the Terms of Use and Privacy Guidelines. This is followed by a section titled "Acknowledgements:" with four checkboxes, each followed by a statement of agreement. At the bottom left is a link "Return to login" and at the bottom right is a blue button labeled "Next".

Featuring

NPKUA

national PKU alliance

Registration

Terms & Conditions Contact Info Notifications Review & Submit Confirmation

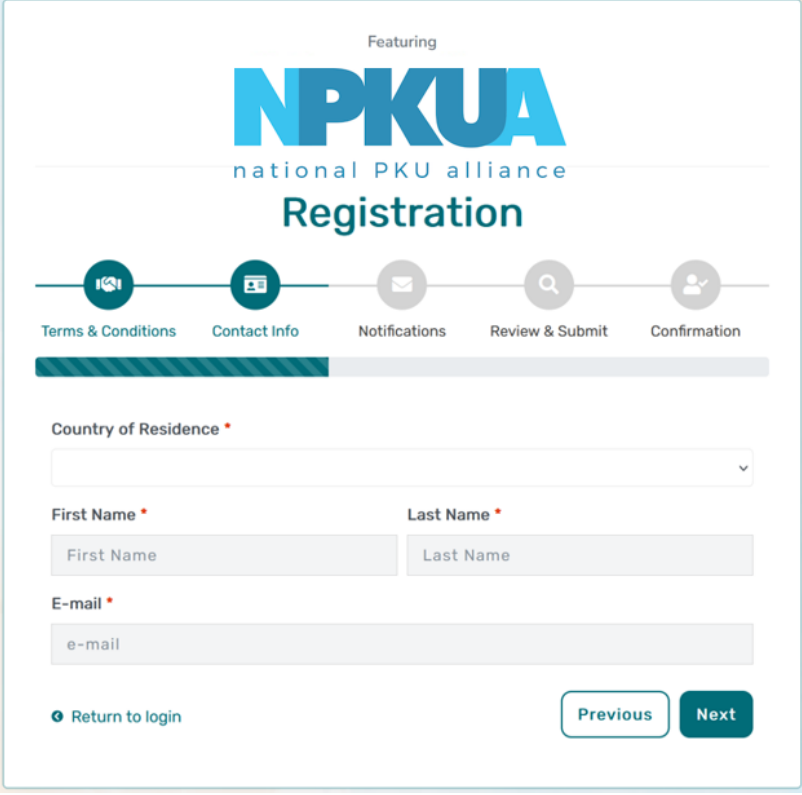
Below are links to the IAMRARE Terms of Use and Privacy Guidelines. The purpose of these documents is to outline your rights and responsibilities when using the platform. These documents include: 1) Standard policies for all studies on this platform, 2) A privacy statement that details how your data 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

NPKUA

national PKU alliance

Registration

Progress bar: Terms & Conditions (active), Contact Info, Notifications, Review & Submit, Confirmation

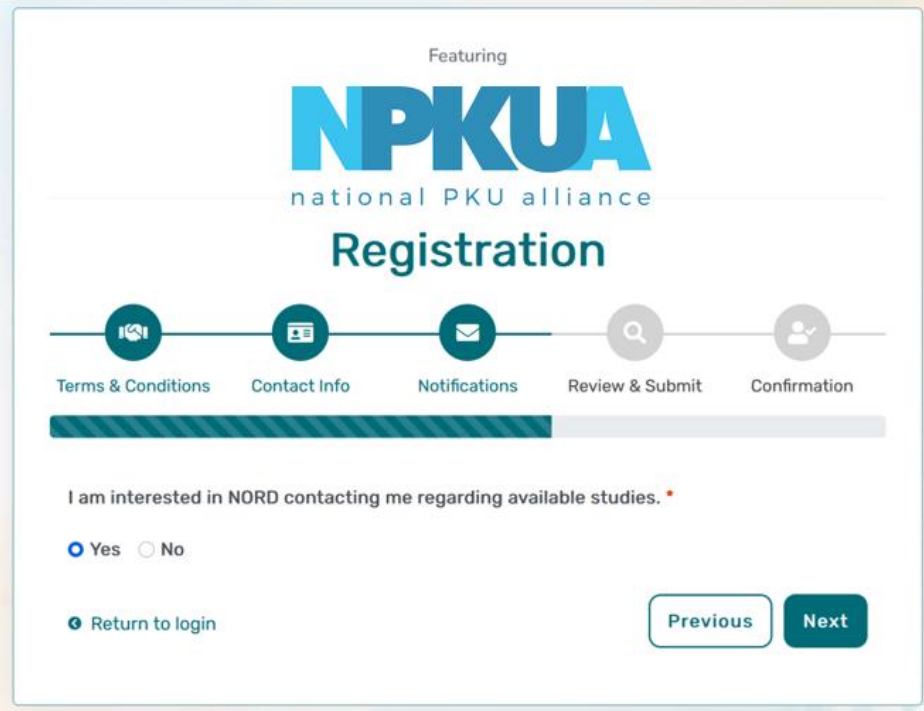
Country of Residence *

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

NPKUA

national PKU alliance

Registration

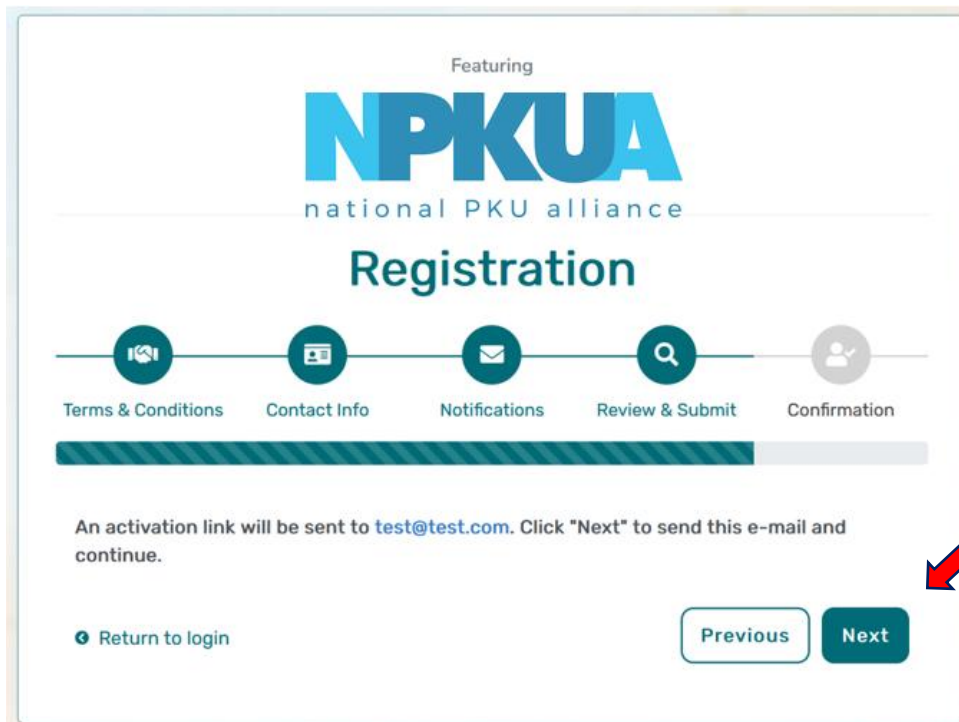
Progress bar: Terms & Conditions, Contact Info, Notifications (active), Review & Submit, Confirmation

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.



Featuring

NPKUA

national PKU alliance

Registration

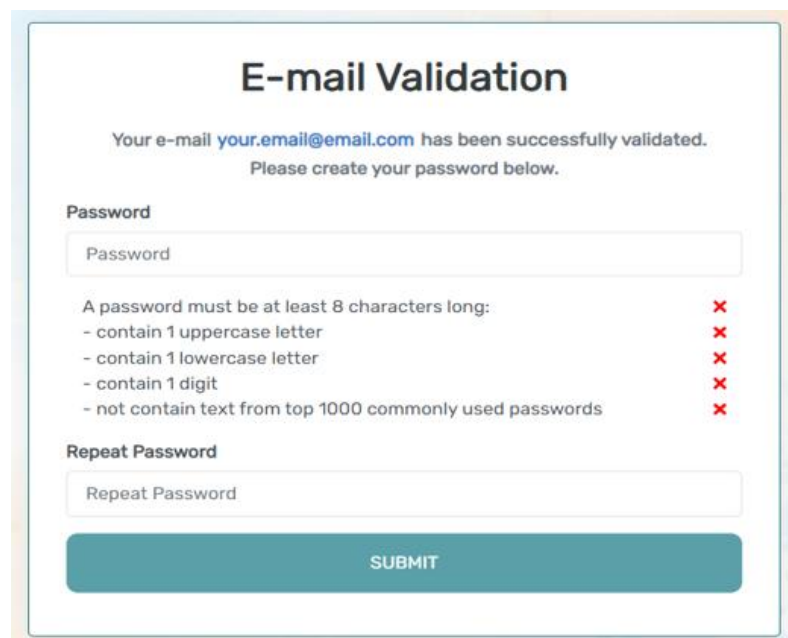
Terms & Conditions Contact Info Notifications Review & Submit Confirmation

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

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

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”.



E-mail Validation

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

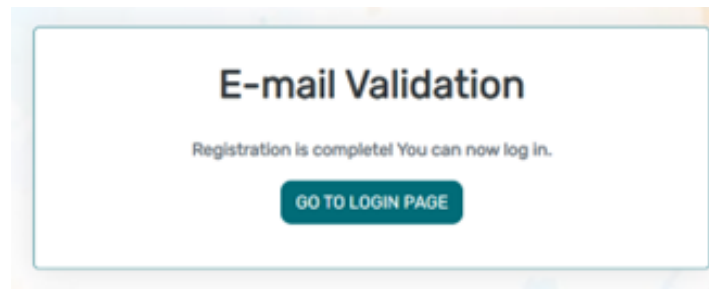
Password

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”.

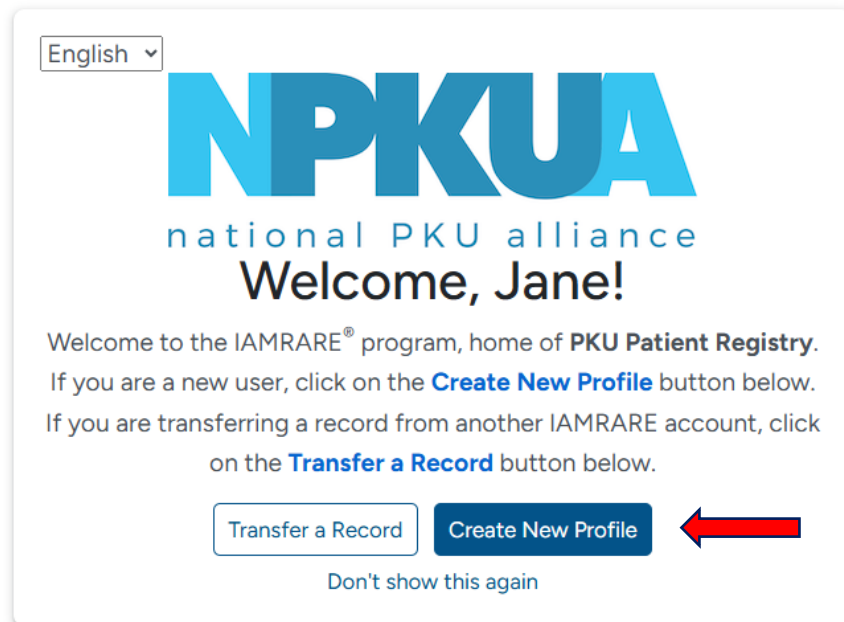


- Step 7: Log in using your new e-mail and password.

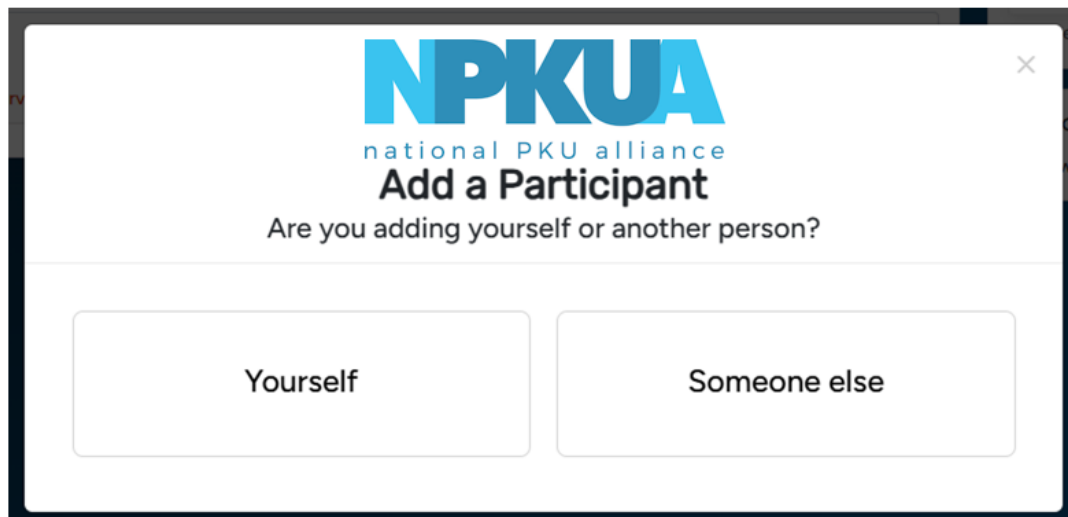


Add a Participant

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



- 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 *

First Name on Birth Certificate *

Last Name on Birth Certificate *

Sex Recorded on Birth Certificate * ?


Country of Residence * ?

Current Last name *

State/Province/Region of Residence * ?

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 PKU 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 PKU Patient Registry

Answered 0/6 questions

Consent Overview

Those eligible to participate in our study include:

Participant: An individual diagnosed with Phenylketonuria (PKU) 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.

Next

Consent to PKU Patient Registry

Answered 0/6 questions

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

Title: PKU Patient Registry

Principal Investigator: Susan A. Berry, MD

Phone: 612-624-5965

Email: registry@npkua.org

Sponsor: National PKU Alliance

Key Information

You are invited to take part in a research study for individuals with Phenylketonuria (PKU) 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 provide data about individuals to create a more comprehensive understanding of PKU and its impact on the individuals affected by it collected in the real-world setting. This information can then be applied to research to assist in creating new therapies and an eventual cure, in an effort to improve the quality of life of those with PKU.
- If you choose to participate on behalf of the participant, you will be asked to answer questions in online surveys in the Registry that ask about your lived experience with PKU. This will take approximately 5-30 minutes to complete depending upon the survey.
- You may experience the following risks, discomforts or inconveniences from participating: no risks of physical harm associated with participation in the PKU Patient Registry. Participation in the PKU Patient Registry does involve the potential risks of a breach of confidentiality of medical information and associated privacy of the participants, but measures are taken to minimize this risk.
- Participating in our study may not help the Study Participant directly, but your time and information may help others with PKU in the future. There are no direct benefits of participation.
- 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.

Previous

Next

Consent to PKU Patient Registry

Answered 2/6 questions

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 PKU 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 PKU 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 PKU 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".

Consent to PKU Patient Registry

Answered 6/6 questions

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 PKU Patient Registry

Select Opt-Ins for this study

- ☐ Interest in hearing about other studies from [National PKU Alliance](#)
- ☐ 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 [National PKU Alliance](#)
- ☐ Interest in signing up for [National PKU Alliance's](#) newsletter
- ☐ 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 on your Participant.

The screenshot shows the IAMRARE dashboard. At the top, there's a navigation bar with 'Home', 'Help', 'Settings', and a user profile 'Hi, Jane!'. Below the navigation bar, a greeting says 'Good Afternoon, Jane!' with 'Member since Apr 22, 2025'. On the right, there's a '+ Add Participant' button. The main section is titled 'Participants' and includes instructions: 'Select a participant to view their studies. Click on the "Add Participant" button above to add a participant.' Below this, there's a list of participants. The first participant is 'Jane Smith', with a date '5-May-2000' and a status '20 pending surveys'. A red arrow points to this participant card. To the right of the participants list, there are 'Shortcuts' for 'Request Transfer' and 'Consent/Opt-Ins', and a 'Notifications (0)' section stating 'No new notifications.'

- Step 2: Click on the appropriate study.

The screenshot shows the 'Enrolled Studies' section for Jane Smith. At the top, there's a 'Back to participant list' link. Below it, the participant's name 'Jane Smith' and date '5-May-2000' are shown, along with a 'Search Studies' button. The main section is titled 'Enrolled Studies' and includes instructions: '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.' Below this, there's a list of studies. The first study is 'NPKUA national PKU alliance', with a status 'Consented' and a button '20 pending surveys'. A red arrow points to this study card. To the right of the studies list, there are 'Shortcuts' for 'Request Transfer' and 'Consent/Opt-Ins', and a 'Notifications (0)' section stating 'No new notifications.'

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

The screenshot shows the 'PKU Patient Registry' survey page. At the top, there's a 'Back to study list' link. Below it, the participant's name 'Jane Smith' and date '5-May-2000' are shown. The main section is titled 'PKU Patient Registry' and includes a 'Surveys' section with a status '20 pending'. Below this, there's a progress bar showing '0%' and a status 'Getting Started Not Started'. On the right, there's a 'Take Survey' button. A red arrow points to this button. At the top right, there are tabs for 'All (20)', 'Complete (0)', and 'Pending (20)'.

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.

The screenshot shows the PKU Patient Registry interface for Jane Smith (5-May-2000). Under the 'Surveys' section, there are 18 pending surveys. Two completed surveys are listed: 'Getting Started' and 'About the Participant', both completed on 22-Apr-2025. For each completed survey, there are two buttons: 'View Responses' (with a magnifying glass icon) and 'Reports' (with a bar chart icon). Red arrows point from the survey titles to these buttons.

Survey	Status	Completed On	View Responses	Reports
Getting Started	Completed	22-Apr-2025	View Responses (1)	Reports
About the Participant	Completed	22-Apr-2025	View Responses (1)	Reports

View Consents 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.

The screenshot shows the 'Enrolled Studies' page for Jane Smith. A red arrow points to the 'Back to participant list' link at the top left. Another red arrow points to the 'Consent/Opt-Ins' shortcut button in the 'Shortcuts' section on the right. The 'Search Studies' button is also visible at the top right.

- Step 2: 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 Jane Smith. It displays a table with the following data:

Study Name	Consent Status	Consented On	Actions
PKU Patient Registry	Consented	22-Apr-2025	View Consent, Revoke, Opt-Ins

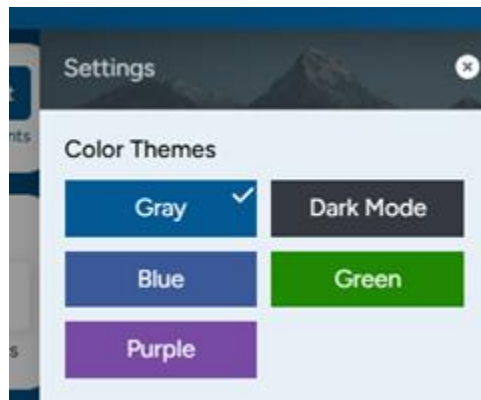
Red arrows point to the 'Revoke' and 'Opt-Ins' buttons in the Actions column.

Dark Mode Settings

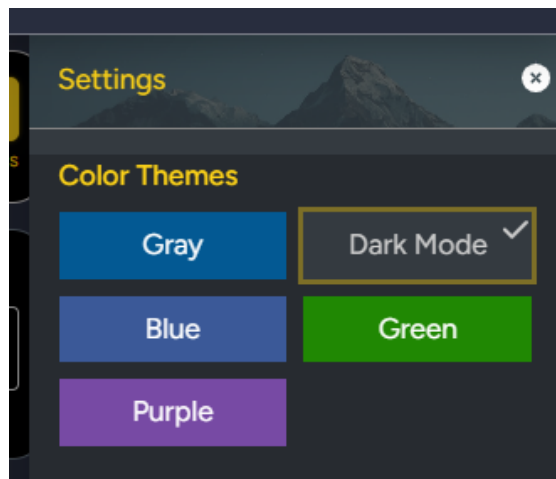
- Step 1: You can view the platform in Dark Mode. First, click Settings.



- Step 2: Select Dark Mode.



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

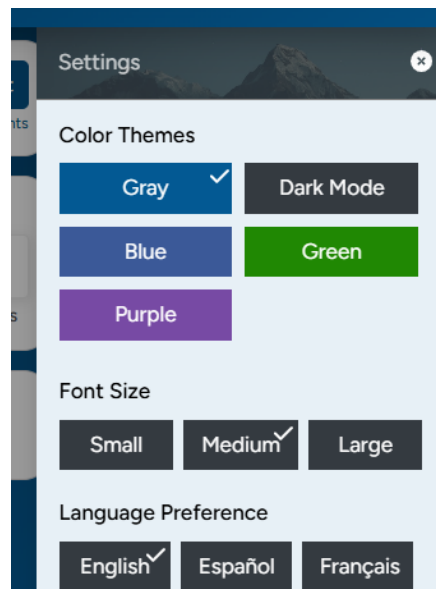


Display Settings

- 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://pku.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?

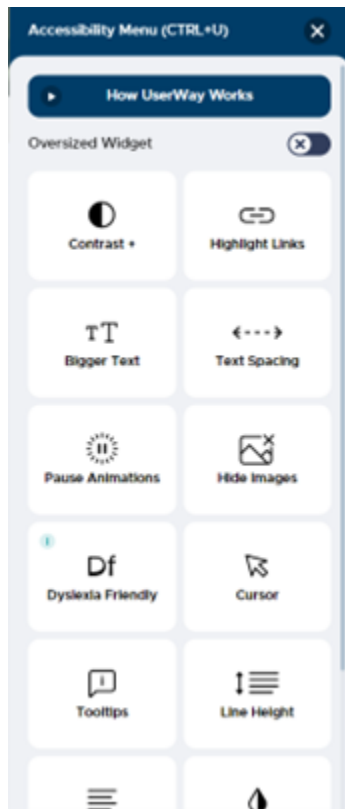


For Patients

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.





Need Assistance?

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

A screenshot of a mobile application's "Have a question?" form. The form has a blue header with the title "Have a question?" and a close button (X). Below the header 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](\"http://rarediseases.org\")." Below this text are two form fields: "Inquiry Type *" with a dropdown menu showing "-- Select Inquiry Type --" and "Message *" with a text input field containing "Your message". At the bottom of the form 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.

View Responses 1

Reports

View Responses 1

Reports

Take Survey

Take Survey

NPKUA
national PKU alliance

National PKU
Alliance
npkua.org/

Contact
Kelsey McQueen

Phone
855.675.8211

E-mail
registry@npkua.org

IRB E-mail
info@northstarreviewboard.org

Social Media
[!\[\]\(4fc2f7cc81639d6310529267a2ee602c_img.jpg\)](#) [!\[\]\(714d70875eaf0e2f34d0a261eaf96dad_img.jpg\)](#) [!\[\]\(dc7d17b015a4a5f15a29473bc04652a8_img.jpg\)](#)