

Kabuki Syndrome Registry

Procedure for Registering, Consenting and Completing Surveys

1. Go to the **Kabuki Syndrome Registry** website at: <https://kabuki.iamrare.org>

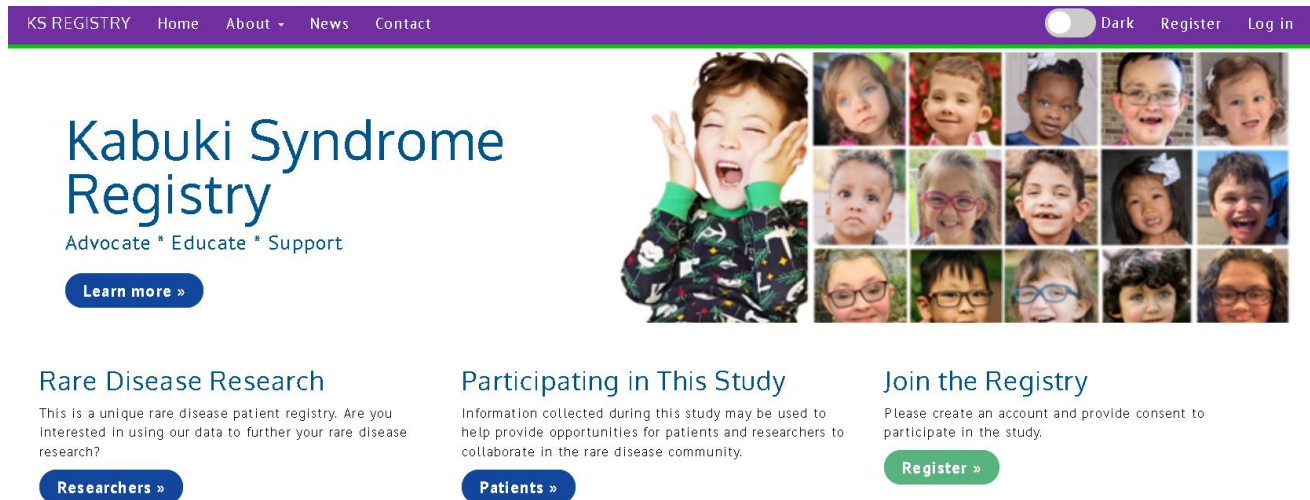


Figure 1. Landing Page

2. Click on the green **Register** button.
3. Complete the age attestation.

The image shows the 'Register' page on the website. At the top, there is a heading 'Register' and a message: 'You must be a legal adult (at least 18 years of age, or the age of majority in your state, province, or country) to register.' Below this is a breadcrumb trail 'Home / Register'. A note says 'Please fill out the form until all red symbols turn into a green symbol.' The main question is 'Before we begin, are you a legal adult (at least 18 years of age, or the age of majority in your state, province, or country)? *' with radio buttons for 'No' and 'Yes'. The 'Yes' button has a red error symbol. Below the question is a link to 'Terms and Conditions' and two buttons: 'View Terms and Conditions' and 'Download as PDF'.

Figure 2. Age attestation

4. Read the **Terms and Conditions** document.
5. Fill in the requested information.
6. **Opt in or out** of reasons to be contacted by Study Staff.
7. Agree to the Terms and Conditions.
8. Click **Create Account** button.

Register


You must be a legal adult (at least 18 years of age, or the age of majority in your state, province, or country) to register.

[Home](#) / Register

Please fill out the form until all  symbols turn into a .

Before we begin, are you a legal adult (at least 18 years of age, or the age of majority in your state, province, or country)? *

Yes 

First Name * 


Middle Name

Last Name * 

Country of Residence * 

Username (Email Address) * 


Confirm Username * 

A password must be at least 8 characters long: 

- contain 1 uppercase letter 

- contain 1 lowercase letter 

- contain 1 digit 

- contain 1 special character 

- not contain text from top 1000 commonly used passwords 

Password * 

Confirm Password * 

Additional Contact Information (Optional)

Home Phone

Work Phone

Mobile Phone

Contact Preferences

Yes, I would like to be contacted about...


☐ Reminders to update my survey responses

☐ Clinical trials I may be eligible for †


☐ Potentially donating biospecimen and/or DNA for future research studies ††

☐ Facebook Group

Terms and Conditions

 [View Terms and Conditions](#)

 [Download as PDF](#)

☒ I have read and agree to the terms and conditions. 

[Create Account](#)

Figure 3. Creating an account – Blank Registration Form

Register

You must be a legal adult (at least 18 years of age, or the age of majority in your state, province, or country) to register.

[Home](#) / Register

Please fill out the form until all ❌ symbols turn into a ✅ symbol.

Before we begin, are you a legal adult (at least 18 years of age, or the age of majority in your state, province, or country)? * Yes ✅

First Name * ✅

Middle Name

Last Name * ✅

Country of Residence * ✅

Username (Email Address) * ✅

Confirm Username * ✅

A password must be at least 8 characters long: ✅

- contain 1 uppercase letter ✅
- contain 1 lowercase letter ✅
- contain 1 digit ✅
- contain 1 special character ✅
- not contain text from top 1000 commonly used passwords ✅

Password * ✅

Confirm Password * ✅

Additional Contact Information (Optional)

Home Phone

Work Phone

Mobile Phone

Contact Preferences

Yes, I would like to be contacted about...

☒ Reminders to update my survey responses

☒ Clinical trials I may be eligible for †

☐ Potentially donating biospecimen and/or DNA for future research studies ††

Terms and Conditions

[View Terms and Conditions](#) [Download as PDF](#)

☒ I have read and agree to the terms and conditions. ✅

[Create Account](#)

Figure 4. Creating an account – Completed Registration Form

9. A confirmation email will be sent to the email address that was provided during registration.
 - a. Confirm registration by copying the **Confirmation Token** from your email.
 - b. You can also confirm the email address by clicking the link in the email.

10. Verify Registration

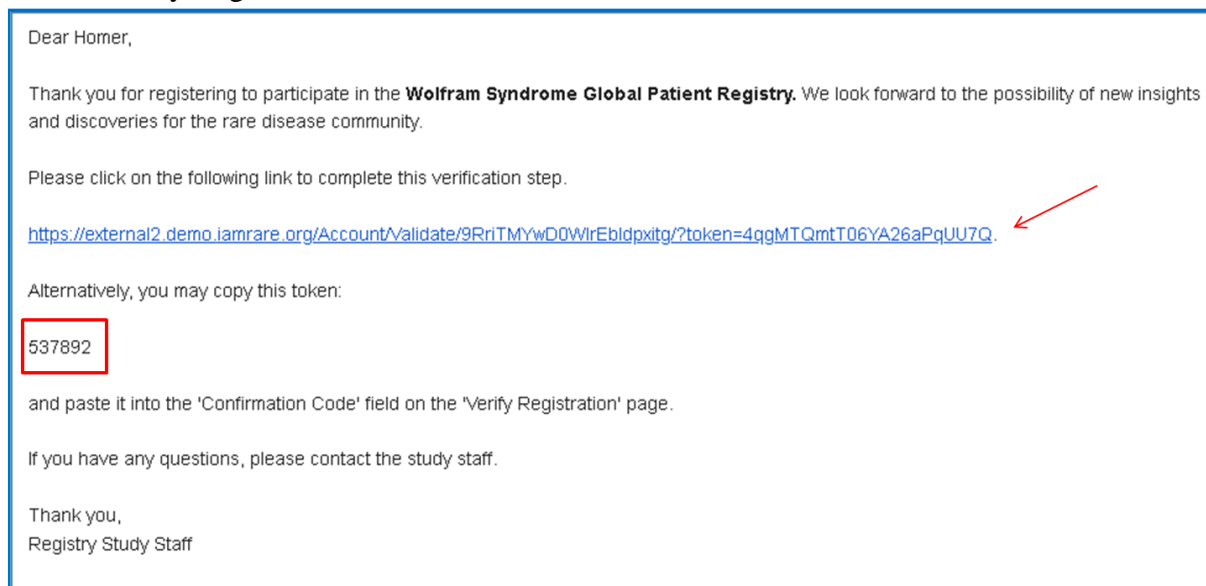


Figure 5. Registration confirmation email

Note: If the confirmation email has not been received after a few minutes, click **Resend confirmation email** at the bottom of the page. Be sure to check spam folders for the email confirmation.

Verify Registration

Home / Verify Registration

Thank you

A new user account has been created.

A confirmation message has been sent to XXXXXXXXXX@rarediseases.org.

You must verify receipt of this email before participation in the 'External Demo Registry 2' registry.

Activate your account by entering the confirmation code from the welcome email you received. Alternatively, you may click on the link within the email. Please note that using the link will require that you re-enter your username and password.

Confirmation Code

Note:

If you do not see this email, please ensure that the provided email address is correct. Please add regisadmin@rarediseases.org to your contacts so that messages from us do not get directed to your email spam.

[Resend confirmation email.](#)

Figure 6. Entering the confirmation code to complete registration

11. Click on **Participant Enrollment**.

12. Select the appropriate **Option**. You can add yourself, or someone else. **Note: the Study Participant is the person who has the condition or diagnosis.**

Participant Enrollment

Welcome! You are in good company. There are currently 89 Participants in this registry.

[Home](#) / Participant Enrollment

I have a rare disease diagnosis and would like to answer surveys on my experience.

[Add Yourself as a Participant](#)

I am a parent or other legally authorized representative of a person with a rare condition and I would like to answer surveys on their behalf.

[Add Someone Else as a Participant](#)

I would like to start answering surveys for a participant that has already been enrolled by another legally authorized representative.

[Start Participant Transfer](#)

You have no active participant enrollments.

You haven't added any survey participants yet.

[Newsletter](#)

Figure 7. Adding a Participant

Alternatively, you can begin the process of transferring a Participant from one party to another. Speak to your study administrator if you have questions.

13. To enroll a Participant, fill in the fields as they apply to the **Study Participant**.

Add Yourself as a Survey Participant

[Home](#) / Participant Enrollment / Add Yourself as a Survey Participant

Before you can add yourself as a survey participant, we need a bit more information about you.

Participant Birthdate (MM/DD/YYYY) *

Participant Age

Country of Residence *

Preferred Contact Method

Preferred Contact Language

Home Phone

Work Phone

Mobile Phone

Address Line 1

Address Line 2

City

State, Province, or Prefecture

Postal Code

[Add Yourself](#) [Cancel](#)

Add Yourself as a Participant

Please verify that your birth date is **February 9, 1997**.

If this is correct, click **Add Yourself** to add yourself as a participant.

[Cancel](#) [Add Yourself](#)

Figure 8. Adding Participants – Adding yourself as a Participant

Add a New Survey Participant

[Home](#) / [Participant Enrollment](#) / Add a New Survey Participant

If you are adding someone other than yourself as a participant in the registry, please ensure you are legally able to do so as the caregiver, guardian, or legally authorized representative.

Your relationship to the participant *

Participant First Name *

Participant Middle Name

Participant Last Name *

Participant Birthdate (MM/DD/YYYY) *

Participant Date of Death (MM/DD/YYYY) (if applicable)

Country of Residence *

Can we contact the participant directly? ☒ Yes ☐ No

Preferred Contact Method

Preferred Contact Language

Email

Home Phone

Figure 9. Adding participants - Adding another person as a Participant

14. Grant Consent

Participant Enrollment

Welcome! You are in good company. There are currently **110** Participants in this registry.

[Home](#) / [Participant Enrollment](#)

Active Participants

	Consent Granted		
Bart Simpson	No	Grant Consent To Take Surveys	Edit Delete
Lisa Simpson	No	Grant Consent To Take Surveys	Edit Delete

[Add Self](#)
[+ Add Participant](#)
[Start Transfer](#)

Figure 10. Accessing the Consent

Note: You must agree to all terms outlined in the Consent Form.

Do you confirm that you have read the Informed Consent Agreement? ☒ ←

I give permission on behalf of the study participant to provide research data to the Power of Patients Registry only for the purposes described above.
☒ Yes ☐ No

I give permission on behalf of the study participant to provide research data that has been de-identified to the Power of Patients Registry for future, undefined research.
☒ Yes ☐ No

[I Agree with the Terms of Consent](#)
[Cancel without Agreeing to the Terms of Consent](#)

Grant Consent?

Please confirm that you agree to all Terms in the Consent document by clicking below:

[Cancel](#) [I Agree with the Terms of Consent](#)

Figure 11. Granting Consent

15. Access Surveys by clicking on **Take Surveys**.

Participant Enrollment

Welcome! You are in good company. There are currently 9 Participants in this registry.

[Home](#) / Participant Enrollment

Active Participants

[+ Add Participant](#) [⇌ Start Transfer](#)

Name	Consent Granted	Actions
Jed Clampett	Yes	Take Surveys Edit Delete Manage Consent

Figure 12. Participant Dashboard

16. Complete the **surveys** applicable to the **Study Participant**. Click the **Take** button next to the **Survey Title** to open the survey.

- NOTE: The Reporter is listed in the top right hand corner of the page and the Study Participant is listed on the left side of the page.** If someone is answering surveys for themselves, both the Reporter and Study Participant fields will reflect the same name.

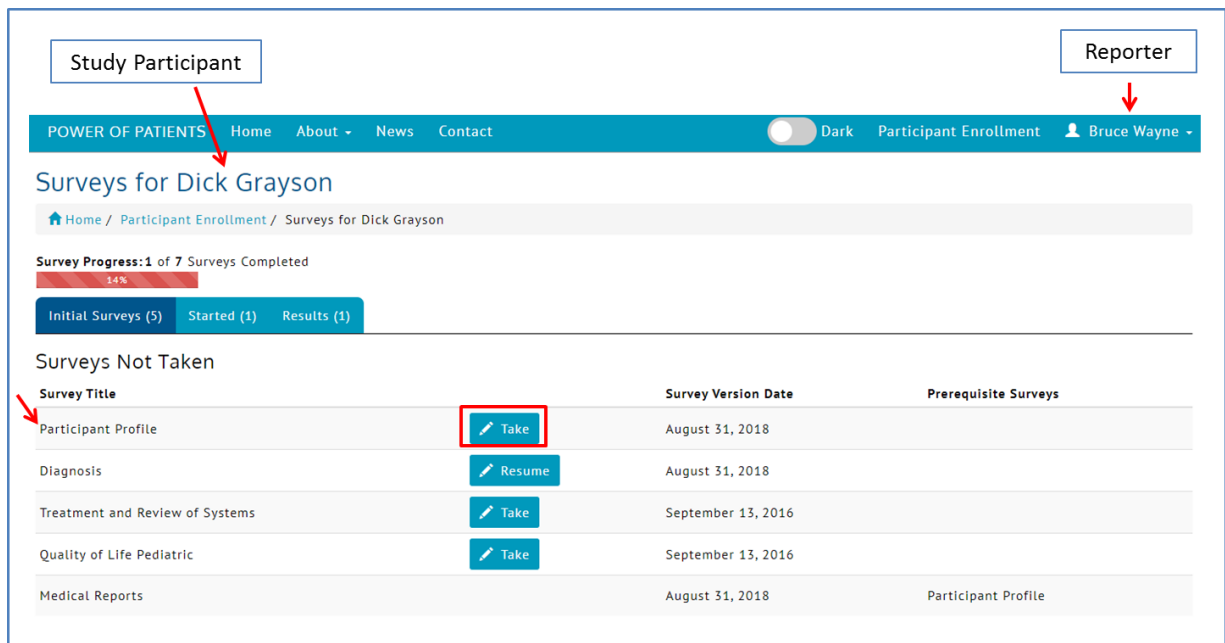


Figure 13. Completing surveys

17. Complete the survey. Questions marked with a red * require a response.

In which country was the Participant born, as shown on their birth certificate? *

United States

In which state or territory was the Participant born, as shown on their birth certificate?

What is the name of the city, town or village of the Participant's birth (as it appears on the birth certificate)?

Help Cancel Finish Later **I'm Finished**

Figure 14. Completing questions and submitting surveys

18. When a survey has been completed, click on the **I'm Finished** button. If not completed, surveys can be saved as drafts by clicking the **Finish Later** button. Click the **Cancel** button to leave the survey and return later. Note that if you click Cancel, no changes will be saved.

19. Using the Calendar Feature

The registry platform supports date entry via a calendar feature.

Participant Birthdate (MM/DD/YYYY) *	11/08/1990
Participant Age	
Participant Date of Death (MM/DD/YYYY)	
Can we contact the participant directly?	
Preferred Contact Method	
Preferred Contact Language	Spoken English

Figure 15. Date Field Displaying Calendar Widget

To use the calendar feature, click on the date field.

Participant First Name *

Participant Middle Name

Participant Last Name *

Participant Birthdate (MM/DD/YYYY) *

Participant Age

Participant Date of Death (MM/DD/YYYY)

Can we contact the participant directly?

☒ Yes ☐ No

Preferred Contact Method

Preferred Contact Language

Address Line 1

Address Line 2

City

State, Province, or Prefecture

Postal Code

Click on Birthdate (or Date of Death) field to engage Calendar Picker:

Participant First Name *

Participant Middle Name

Participant Last Name *

Participant Birthdate (MM/DD/YYYY) *

Participant Age

Participant Date of Death (MM/DD/YYYY)

Can we contact the participant directly?

☒ Yes ☐ No

Preferred Contact Method

Preferred Contact Language

Address Line 1

Address Line 2

City

State, Province, or Prefecture

Postal Code

Calendar Picker (August 1988):

Su	Mo	Tu	We	Th	Fr	Sa
31	1	2	3	4	5	6
7	8	9	10	11	12	13
14	15	16	17	18	19	20
21	22	23	24	25	26	27
28	29	30	31	1	2	3
4	5	6	7	8	9	10

Figure 16. Using the Calendar Feature

To easily scroll through the calendar options, click on the header field. Clicking on the month will populate all the months. Clicking on the year will produce more years, etc. Use the left and right arrows to navigate to the desired time point.

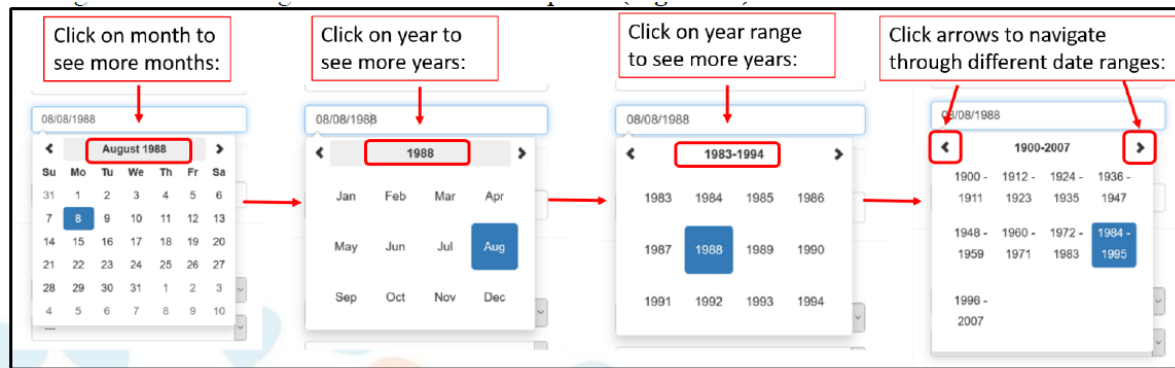


Figure 17. Calendar scrolling options

Note that dates can also be typed directly into the calendar field.

20. Updating and Retaking Surveys

Some surveys will be available to edit after submission and others will be set to a regular retake, or *longitudinal*, schedule. This supports the study of the disease over time. You may opt in for email reminders when retakes are due.

Surveys for Homer Simpson

Home / Participant Enrollment / Surveys for Homer Simpson

Survey Progress: 1 of 7 Surveys Completed

Initial Surveys (6)

Updatable (1)

Surveys to Retake (1)

Surveys Not Taken

Survey Title		Survey Version Date	Prerequisite Surveys
Wolfram Syndrome Diagnosis and Treatment	Take	February 2, 2022	
Wolfram Syndrome Associated Diagnoses	Take	February 2, 2022	
Additional Medical and Family History	Take	February 1, 2022	
Adult Quality of Life - Social/Emotional	Take	February 2, 2022	
Adult Quality of Life - Physical/Medical	Take	February 2, 2022	
Genetic Reports	Take	February 2, 2022	

Figure 18. Participant Survey List

To update a survey, click on the **Updatable** tab and click the **Edit** button next to the survey. The survey will populate with the previous answers. Simply edit any information that has changed.

Surveys for Homer Simpson

Home / Participant Enrollment / Surveys for Homer Simpson

Survey Progress: 1 of 7 Surveys Completed
14%

Initial Surveys (6) **Updatable (1)** Surveys to Retake (1)

Updatable Responses

You have completed these surveys and may now view your answers by clicking on the **View** button next to the survey title. You can update a previously completed survey by clicking on the **Edit** button.

Survey Title		Survey Version Date	Submitted Date	Last Updated Date	
Participant Profile	View	January 13, 2020	March 6, 2020	Never	Edit

Figure 19. Accessing Updatable Surveys

To retake a survey, click on the **Surveys to Retake** tab and click the **Retake** button next to the survey. Longitudinal surveys will be blank each time, to capture fresh unbiased information.

Surveys for Homer Simpson

Home / Participant Enrollment / Surveys for Homer Simpson

Survey Progress: 1 of 7 Surveys Completed
14%

Initial Surveys (6) Updatable (1) **Surveys to Retake (1)**

Surveys to be Retaken

These surveys are a part of the study schedule and require completion at varying intervals. A survey can be completed according to the date displayed in the Next Follow-Up column.

Survey Title		Last Taken Version Date	First Taken	Last Taken	Last Updated	Next Follow-Up
Participant Profile	Retake	January 13, 2020	March 6, 2020	March 6, 2020	Never	March 7, 2020

Figure 20. Retaking Longitudinal Surveys