

PMS
DataHub
Step-by-Step
User Guide

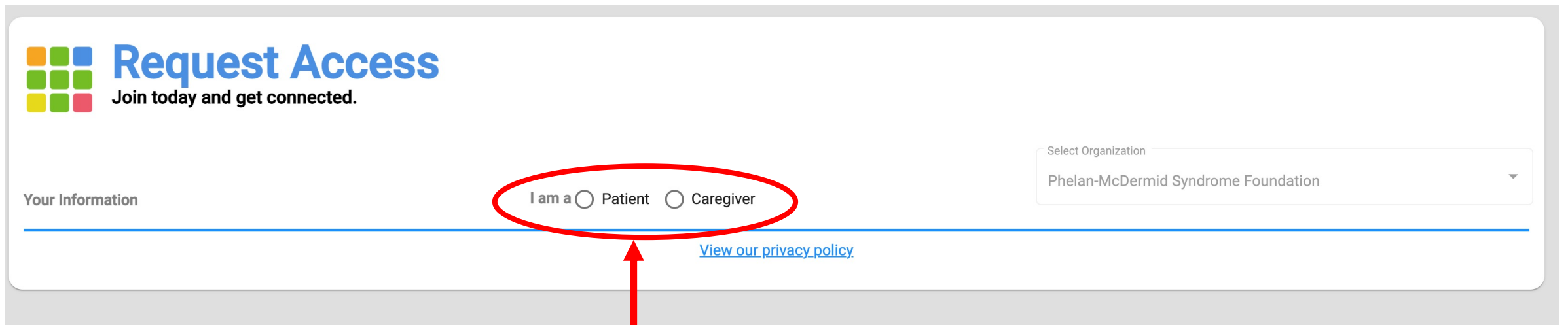


DataHub

**Google Chrome and Safari are the preferred browsers for the DataHub.
Firefox is not supported**

To create an account:

Go to: <https://pmsfx.acrossmatrix.com/#/user-request>



Request Access
Join today and get connected.

Your Information

I am a Patient Caregiver

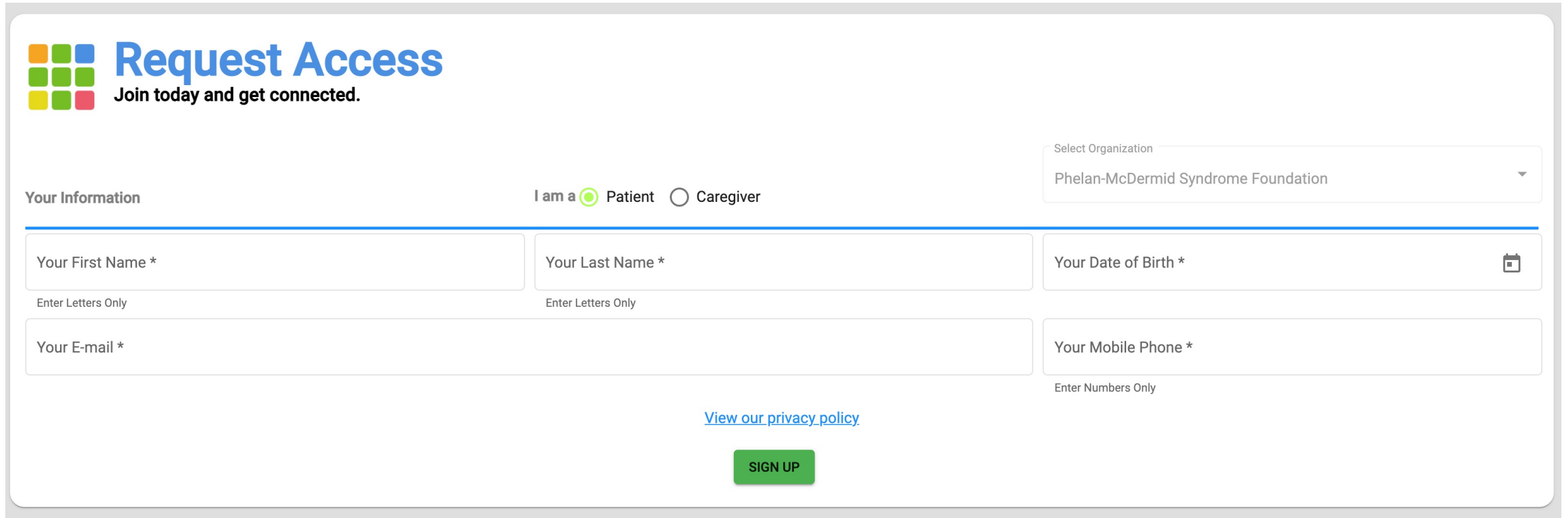
Select Organization
Phelan-McDermid Syndrome Foundation

[View our privacy policy](#)

Choose an option

***this applies to the person filling out the information**

The following applies to continuing to create a new account
If you are the person with PMS (patient), this screen will
come up:



Request Access
Join today and get connected.

Your Information Patient Caregiver

Select Organization
Phelan-McDermid Syndrome Foundation

Your First Name *
Enter Letters Only

Your Last Name *
Enter Letters Only

Your Date of Birth *
Calendar icon

Your E-mail *

Your Mobile Phone *
Enter Numbers Only

[View our privacy policy](#)

SIGN UP

Fill out your information and hit “Sign Up”.

If you are a caregiver or someone representing the person with PMS, this screen will come up.

Enter information for both the caregiver (top) and patient(s) (bottom):



Your Information

Caregiver info

I am a Patient Caregiver

Select Organization
Phelan-McDermid Syndrome Foundation

Your First Name *
Enter Letters Only

Your Last Name *
Enter Letters Only

Your Date of Birth *

Your E-mail * Enter Letters Only

Your Mobile Phone *
Enter Numbers Only

Patient Information One Patient Two Patients

Patient One

First Name *
Enter Letters Only

Last Name *
Enter Letters Only

Date of Birth *

Additional Information

[View our privacy policy](#)

SIGN UP

Caregiver DOB

Person with PMS DOB

Your Information

I am a Patient Caregiver

Select Organization
Phelan-McDermid Syndrome Foundation

Your First Name *
Enter Letters Only

Your Last Name *
Enter Letters Only

Your Date of Birth *
📅

Your E-mail *

Your Mobile Phone *
Enter Numbers Only

Patient Information One Patient Two Patients

Patient One

First Name *
Enter Letters Only

Last Name *
Enter Letters Only

Date of Birth *
📅

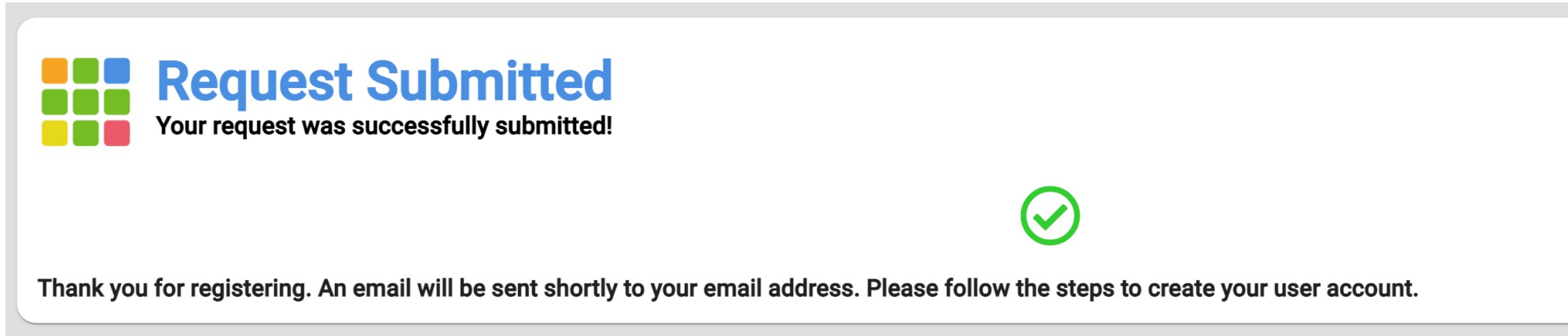
Additional Information

[View our privacy policy.](#)

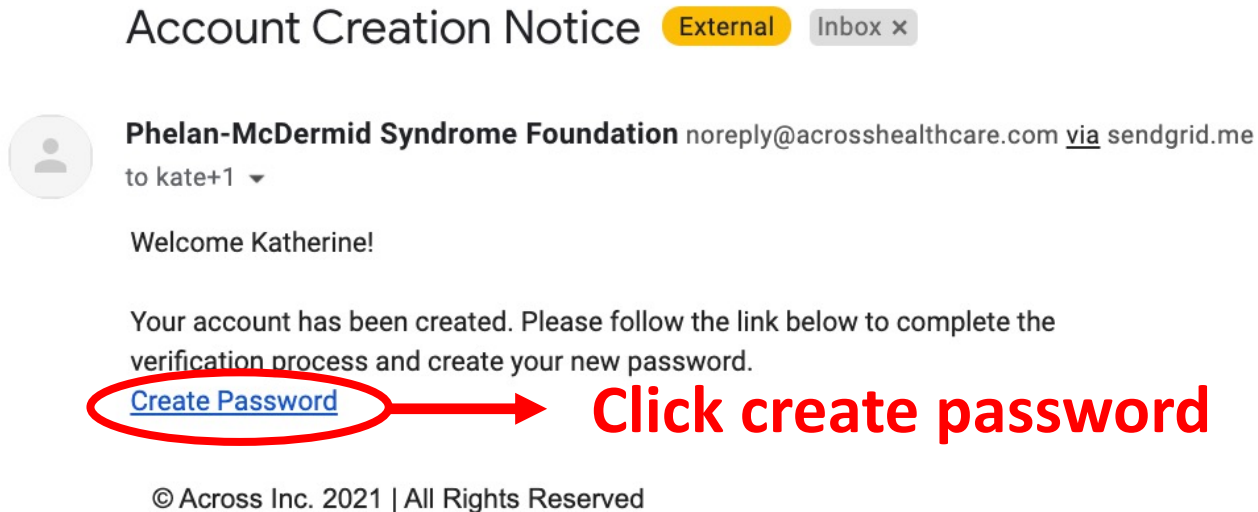
SIGN UP

Don't forget to select the number of people with PMS and add their information.

After signing up, you will receive this screen:



You will receive an email like this: (***)this may take anywhere from 1-10 minutes!)



Enter in your email and click “Send verification code”.



DataHub

Enter your email address below and click send verification code. Then your code will be emailed to you.

This information is required.


Send verification code

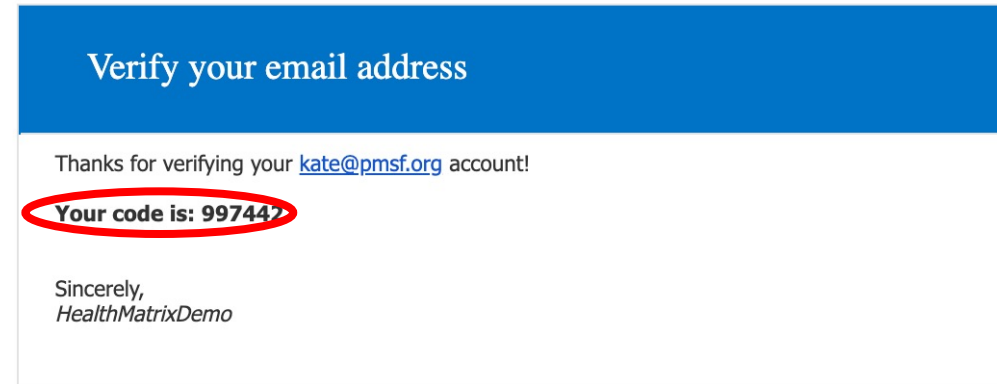
Continue

Cancel

You should receive an email like this:

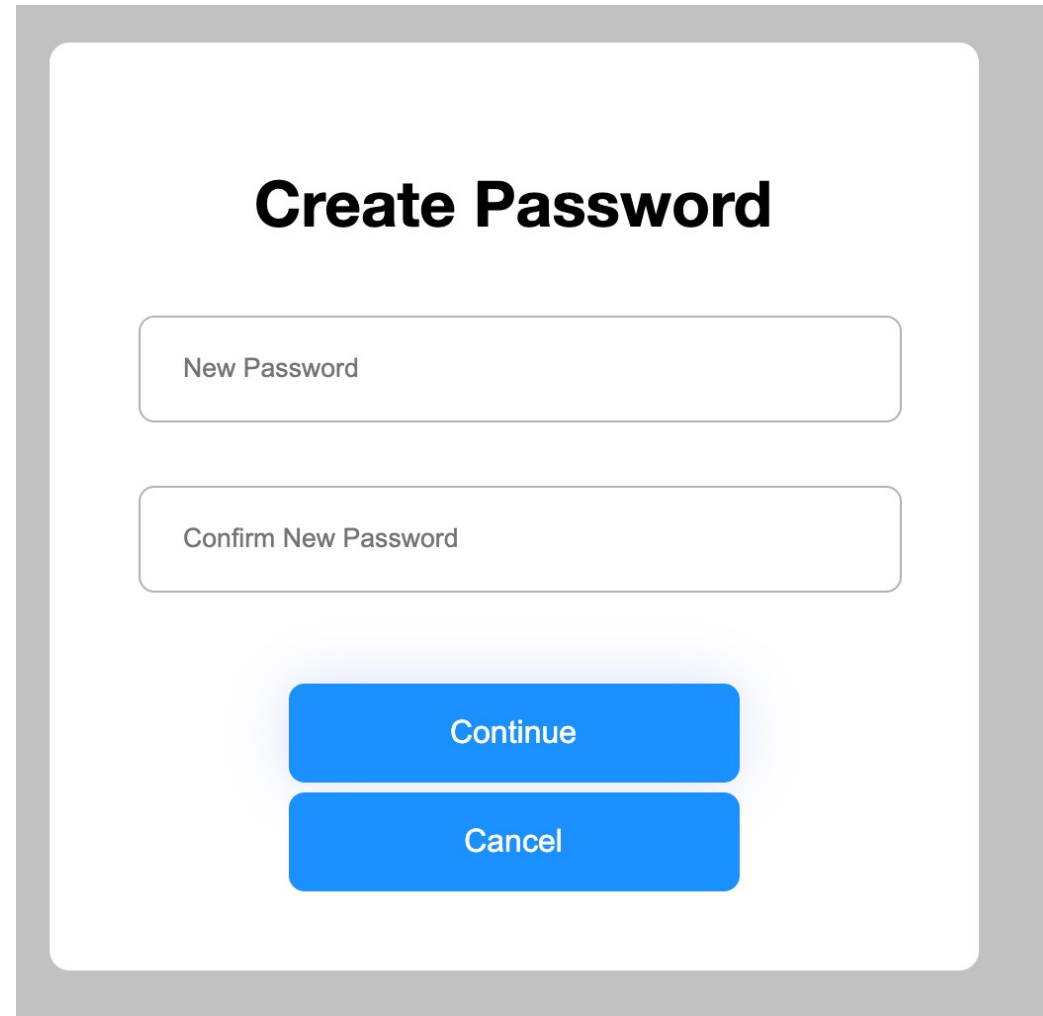
HealthMatrixDemo account email verification code External Inbox x

 **Microsoft on behalf of HealthMatrixDemo** <msonlineservicesteam@microsoftonline.com>
to me ▾



Enter the code on the previous DataHub screen.

Create a new password and click “Continue”.



Create Password

New Password


Confirm New Password

Continue

Cancel

The image shows a 'Create Password' dialog box with a white background and a grey border. It features a title 'Create Password' in bold black text. Below the title are two text input fields: 'New Password' and 'Confirm New Password'. At the bottom, there are two blue buttons with white text: 'Continue' and 'Cancel'.

Enter your email and new password and click “Sign In”.



The logo for Phelan-McDermid Syndrome DataHub features a circular emblem with green and light green curved lines and dots, resembling a stylized DNA helix or a network. The text "PHELAN-McDERMID SYNDROME" is written in a circular path around the emblem.

DataHub

Sign in with your email address

[Forgot your password?](#)

All progress can be saved on the DataHub platform!

If you are logging back in after creating an account signing out of the DataHub, please go to:

<https://pmsf.acrossmatrix.com/>

Read through the End-User License Agreement. Click Accept or Deny

End-User License Agreement (EULA)

Please open and sign this agreement in order to gain access to this application. This agreement outlines how this application can be used and explains obligations and restrictions.

END USER LICENSE AGREEMENT

Across Matrix, Inc. provides the Across Matrix Platform (as defined herein) solely on the Terms and Conditions set forth in this Agreement and on the condition that you Accept and Comply with them. By Clicking the “ACCEPT” Button. You: (A) Accept this Agreement and Agree that You are legally bound by its Terms; and (B) Represent and Warrant that: (I) You are Legal Age to enter into a Binding Agreement; And (II) If You are a Corporation, Governmental Organization, or Other Legal Entity, You have the Right, Power, and Authority to enter into this agreement on behalf of such Entity and bind such Entity to its Terms. If You DO NOT AGREE to the Terms of this Agreement, Across Matrix, Inc. WILL NOT and DOES NOT grant You Access to the Across Matrix Platform and You MUST Not Use the Across Matrix Platform or submit ANY DATA through the Across Matrix Platform.

DENY

ACCEPT

If you clicked Accept, a document will appear which explains the benefits and risks in participating in the DataHub (informed consent).

Test Patient

PMSF Informed Consent

"Understanding Your Participation" & Informed Consent

Sponsor / Study Title: Phelan-McDermid Syndrome Foundation / "Phelan-McDermid syndrome DataHub"

Protocol Number: Pro00006079

Principal Investigator: Katherine Still, Ph.D.

Telephone/email: 941-485-8000; datahub@pmsf.org

Address: Phelan-McDermid Syndrome Foundation 8 Sorrento Drive Osprey, FL 34229

For the purpose of this document "you," "your," "me," and "I," refers to:

-the individual affected by Phelan-McDermid syndrome (PMS) or related condition

OR

-in cases where the individual with PMS cannot legally consent, the parent, guardian, or family member providing the information on behalf of the individual with PMS.

In cases where the participant's representative gives consent, the individual with PMS should be informed about the study to the extent possible given his/her understanding. During the use of this platform, if the individual with PMS gains the capacity to consent, informed consent will be obtained from the individual with PMS, and they will be offered the ability to remove their information if desired.

Please read this form carefully. Take your time to ask the principal investigator or DataHub staff (datahub@pmsf.org) as many questions as you would like. The principal investigator or DataHub staff can explain words or information that you do not understand. Reading this form and talking to the principal investigator or DataHub staff may help you decide whether to take part or not. If you decide to take part in this study, you must sign your name at the end of this form and date it.

Please scroll (along the righthand side) and hit "Next".

You will be asked a series of questions about you understand the consent and agree to participate in the DataHub:

Informed Consent Form
Phelan-McDermid syndrome DataHub
Informed Consent for Participation

For the purpose of this document "you," "your," "me," and "I," refers to the person filling out this form, either the individual affected by PMS, or the parent, guardian or family member providing the information on behalf of the individual with PMS.

1. Please indicate that you have reviewed and understand the "Understanding Your Participation" page, and do not have any questions about your participation in the DataHub. *

- I have reviewed the "Understanding Your Participation" information. I understand it, and I do not have any questions.
- I have not reviewed or do not understand the "Understanding Your Participation" information.

2. When you create an account in the DataHub, you will be assigned a unique numeric identifier that will be used in place of your personal, identifying data. This code allows researchers to access data you have entered into the DataHub without knowing who you are. *
I give permission for the data I share in the DataHub to be provided to researchers using my unique numeric identifier.

- Yes
- No

3. I would like to be contacted by the DataHub staff if researchers learn anything new about PMS.

- Yes
- No

4. I would like to be contacted by the DataHub staff if I, or my family member, becomes eligible for a clinical trial. (Please note that even if the coordinators of a clinical trial believe that you might be eligible for the trial based on the data about you stored in the DataHub, it is still possible that later on it will turn out that you do not meet the trial inclusion criteria after all. Please also be aware that if we inform you about the existence of a trial, this does not imply that we endorse it. In order to participate in any trial, you will need to fill out a separate informed consent form.

- Yes
- No

Please answer the questions and click "Next" when ready. If you answer "No" to understanding/agreeing to key points of the consent (ex – questions 1 & 2), you will not have consented to the DataHub and the platform will not proceed. This is to protect your rights since this is a voluntary exercise.

If you consent to joining the DataHub, please select who you are in order to sign.

Test Patient

PMSF Informed Consent

CONSENT

I have read and understand the information in this informed consent document. I have had the opportunity to ask questions and all of my questions have been answered to my satisfaction. I voluntarily agree to participate in this research until I decide otherwise. I do not give up any of my legal rights by signing and dating this consent document. I will receive a copy of this signed and dated consent document.

10. Please select a signature option; I am a person who is a(an): *

- Individual with PMS
- Parent/Legal Guardian
- Legally Authorized Representative

This applies to you, the person filling out this information – either the person with PMS, or the person representing someone with PMS.



Page 3 of 3

PREVIOUS

COMPLETE

Please sign and date if you consent to join the DataHub. Both the printed name and signature will be typed in.

10. Please select a signature option; I am a person who is a(an): *

- Individual with PMS
- Parent/Legal Guardian
- Legally Authorized Representative

11. Printed Name of Parent/Legal Guardian *

if the person with PMS is under age 18, or cannot legally consent to this form

12. Signature of Parent/Legal Guardian *

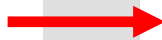
if the person with PMS is under age 18 or cannot legally consent to this form

13. Date *

Please fill out some general information about yourself. If you are a caregiver, this section will be about you, and not the person with PMS.

Your name will already be populated from login.



General Information

Demographics

| | | |
|--|--|--|
| First Name Kate | Middle Initial | Last Name Still |
| Suffix | Country * <small>Required field. Please enter Country</small> | Address Line 1 * <small>Required field. Please enter Address Line 1</small> |
| Address Line 2 | City * <small>Required field. Please enter City</small> | State/Province/Region * <small>Required field. Please enter State/Province/Region</small> |
| Zip/Postal Code * <small>Required field. Please enter Zip/Postal Code</small> | Date of Birth * | Phone * |
| Email Address katherinestillconsultancy@gmail.com | Preferred Language | |

Other Information

Relationship/Marital Status *
Required field. Please enter Relationship/Marital Status

Associated Patient Information

| First Name | Middle Initial | Last Name | Relationship to Patient |
|------------|----------------|-----------|--|
| Test | | Patient | Relationship to Patient <small>Required field. Please enter a Relationship to Patient</small> |

CANCEL SAVE

Enter in information about the person with PMS:

Their name will already be populated from login.



General Information

Demographics

| | | |
|--|--|--|
| First Name Test | Middle Initial | Last Name Patient |
| Suffix | Country * <small>Required field. Please enter Country</small> | Address Line 1 * <small>Required field. Please enter Address Line 1</small> |
| Address Line 2 | City * <small>Required field. Please enter City</small> | State/Province/Region * <small>Required field. Please enter State/Province/Region</small> |
| Zip/Postal Code * <small>Required field. Please enter Zip/Postal Code</small> | Date of Birth * <small>Required field. Please enter Date of Birth</small> | Phone * |
| Email Address katherinestillconsultancy@gmail.com | Preferred Language | |

Other Information

| | | |
|---|---|---|
| Race * <small>Required field. Please enter Race</small> | Ethnicity * <small>Required field. Please enter Ethnicity</small> | |
| Gender At Birth * <small>Required field. Please enter Gender at Birth</small> | Gender Identity * <small>Required field. Please enter Gender Identity</small> | |
| Birthplace: Country * <small>Required field. Please enter Birthplace Country</small> | Birthplace: State/Province/Region * <small>Required field. Please enter Birthplace State/Province/Region</small> | Birthplace: City * <small>Required field. Please enter Birthplace City</small> |

Powered By:

The
“clinician”
field should
be grayed out
and you do
not need to
enter
anything
here.



Other Information

Required field. Please enter Race

Required field. Please enter Gender at Birth

Required field. Please enter Birthplace Country

Required field. Please enter Estimated Household Income

Required field. Please enter Ethnicity

Required field. Please enter Gender Identity

Required field. Please enter Birthplace State/Province/Region

Required field. Please enter Insurance Coverage

Required field. Please enter Rare Disease

Required field. Please enter Birthplace City

Qualified independent research groups who are engaged in rigorous rare-disease and other similar types of studies are only allowed to request access to the patient tool data that we store in Matrix; this excludes data stored or collected by any of the forms in our Surveys/Studies section, which is governed by the organization's Informed Consent.

With regard to your permission, for the benefit of rare-disease and other similar types of studies, please select from one of the following privacy settings:

- You may share my patient tool data
- Please don't share any of my patient tool data
- Please ask me before sharing any of my patient tool data

Please add or update any information to continue.

CANCEL

SAVE

At the bottom of the screen, it will ask you if you want to share “patient tool data.” This is any information aside from genetics and general health history covered under the consent. This will include any additional information you want to enter on the platform menu, such as medications, symptoms, activities, and other categories you want to track. These areas are included for the benefit of rare disease research, but the most important pieces of information are genetic reports and general health information.

Other Information

| | | |
|---|---|--|
| Race * | Ethnicity * | |
| Required field. Please enter Race | Required field. Please enter Ethnicity | |
| Gender At Birth * | Gender Identity * | |
| Required field. Please enter Gender at Birth | Required field. Please enter Gender Identity | |
| Birthplace: Country * | Birthplace: State/Province/Region * | Birthplace: City * |
| Required field. Please enter Birthplace Country | Required field. Please enter Birthplace State/Province/Region | Required field. Please enter Birthplace City |
| Estimated Household Income * | Insurance Coverage for Patient? * | |
| Required field. Please enter Estimated Household Income | Required field. Please enter Insurance Coverage | |
| Clinician | Rare Disease(s) * | |
| | Required field. Please enter Rare Disease | |

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With regard to your permission, for the benefit of rare-disease and other similar types of studies, please select from one of the following privacy settings:

- You may share my patient tool data
- Please don't share any of my patient tool data
- Please ask me before sharing any of my patient tool data

Please add or update any information to continue.

CANCEL **SAVE**

Please click “Save” to continue.

Please upload any genetic reports for the person with PMS.

Document Upload

Please upload a document for Genetic Test by selecting the button below:

No documents to show

UPLOAD +

I do NOT have a document for Genetic Test available to upload

Next

Please fill out the general health survey. This includes general questions about major body systems. It is designed to be general to be easy to analyze, especially across diseases.

Questions refer to the person with PMS

[GLOSSARY](#) ✕

General Health Survey

Demographics, Testing, and Development

For the purpose of this survey "you," "your," "me," and "I," refers to the individual affected by PMS/SHANK3 (the patient).

1. How do you identify your gender/sex? *

Female

Male

Intersex

MTF/Transwoman/Transgender Female

FTM/Transman/Transgender Male

Unknown

Other

2. Have you had genetic testing? *
Examples: Chromosome microarray (CMA single gene sequencing), single gene test, gene panel test, whole exome sequencing (WES), whole genome sequencing (WGS)

Yes

No

Unsure

3. What type(s) of genetic testing have you had? Please select all that apply. *

Chromosomes (Also called chromosome analysis, karyotype, karyotype analysis, or G-band karyotype)

Direct-to-consumer (DTC) genetic test (Genetic testing that is ordered by and returned directly to the person tested rather than through a doctor, also called DTC; please specify name of the company and/or test below)

Single gene or gene panel test (Testing of ONE or MULTIPLE genes with a single test; please specify the name of the test or condition being tested for below)

Microarray (Also called chromosomal microarray, CMA, cytogenomic array, array, or array CGH)

Exome sequencing (Testing that targets a person's complete set of genes; also called whole exome sequencing or WES)

You are currently not tracking streaks. To start tracking your medicine/supplement streaks, navigate to your Medicine and Supplements page and turn on the streak toggle. The actual increment of medicine/supplement streaks will show once you set a reminder and take the medicine/supplement.

If you answer yes to having issues in a category, specific questions will be asked about that category later, including diagnoses, and when they were diagnoses.



[GLOSSARY](#) ✕

General Health Survey

Health & Development

*Additional pages of the survey are assigned based on responses to this set of questions.
For the purpose of this survey "you," "your," "me," and "I," refers to the individual affected by PMS/SHANK3 (the patient).

6. Have you been told that there were issues during your biological mother's PREGNANCY with you and/or your DELIVERY/BIRTH? *
Examples: Mother had to be put on bed rest, there were concerns on ultrasound, you were born early (prematurely), or you had to stay in the neonatal intensive care unit (NICU).

Yes
 No
 Unsure

7. Have you had issues with your GROWTH? *
Examples: You are much shorter or taller than expected for your family, you have had significant and/or lifelong problems gaining weight, or you have significant and/or lifelong problems with obesity. You may have seen an endocrinologist or other specialist for these issues.

Yes
 No
 Unsure

8. Have you had issues with your HORMONES and/or ENDOCRINE SYSTEM (hormone-producing glands)? *
Examples: Diabetes, issues with estrogen or testosterone levels, or function of the thyroid, parathyroid, or adrenal glands. You may have seen an endocrinologist for these issues.

Yes
 No
 Unsure

9. Have you ever been diagnosed with cancer, colon polyps or a non-cancerous tumor? *
Examples: Breast cancer, prostate cancer, skin cancer, lung cancer. You may have seen an oncologist for these issues.

Yes
 No

You are currently not tracking streaks. To start tracking your medicine/supplement streaks, navigate to your Medicine and Supplements page and turn on the streak toggle. The actual increment of medicine/supplement streaks will show once you set a reminder and take the medicine/supplement

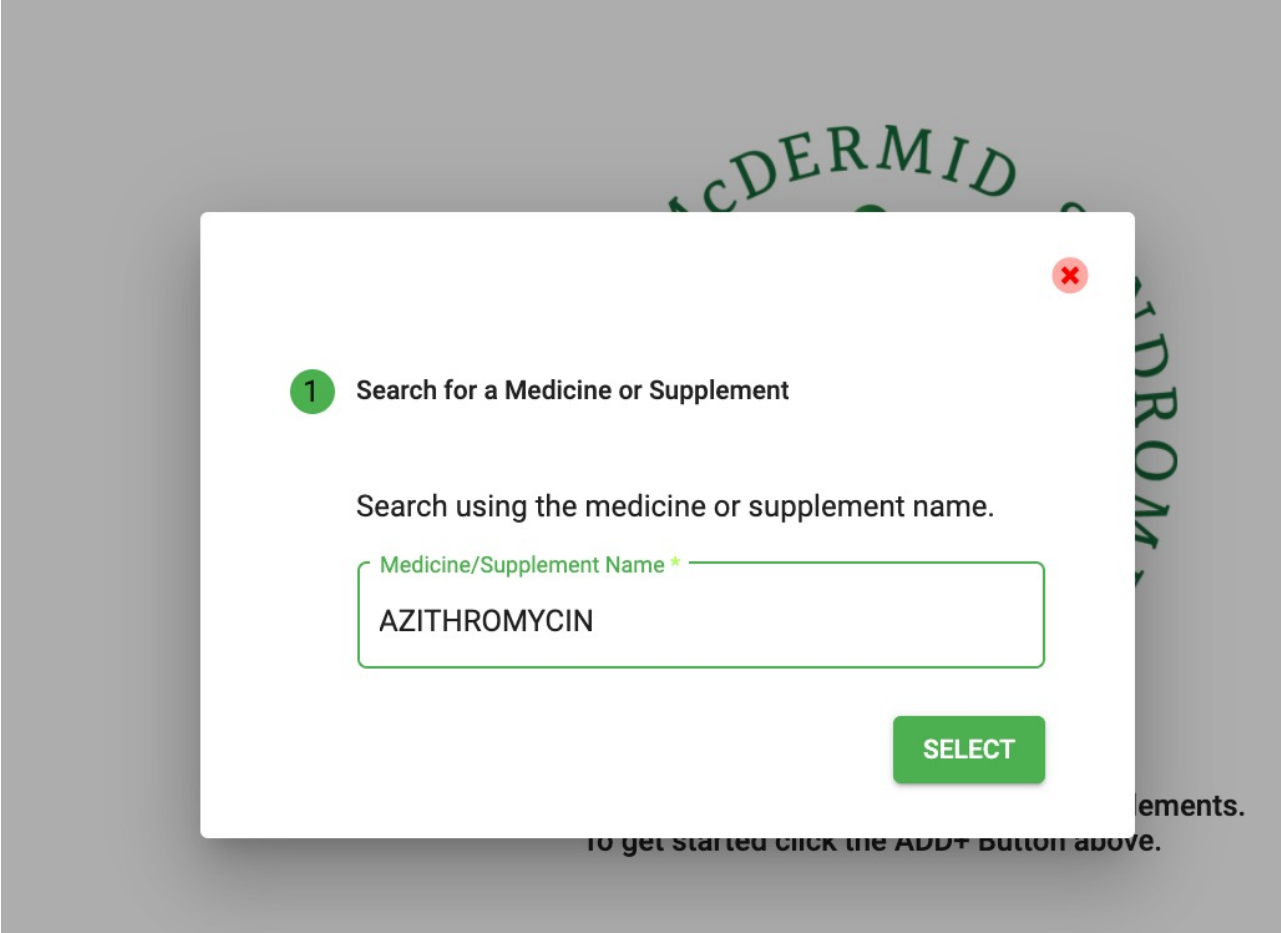
Powered By:

Why does the General Health Survey seem non-specific to PMS? Is there a place for me to add more detail about medical issues?

This survey is standardized so it can be used across diseases, to learn more about similarities and differences between PMS and other neurodevelopmental disorders, and other rare diseases. We have received feedback from researchers that this is a powerful method to understand underlying factors in disease. More surveys specific to PMS will be added in the future to dig deeper. If you wish to add a condition that is not listed, or add more detail, there are free text fields where you can add this information at the bottom of the page for each specific system – e.g., digestive system.

When you are done with all questions in the General Health Survey, please click “Complete”.

Please fill out which medications the person with PMS is on, by typing in the name and finding it on the list. If your medication does not pop up, don't worry! Fill in as many as you can. This is a government-generated list and is not perfect.



Please include information on the type of medication and dosage. If the type of medication or correct dosage is not listed, please select the closest answer. As mentioned previously, this list is automatically generated and is not perfect, but can still be very helpful for research. The medication itself is the most important piece of information.

The “Refills” section is optional for those who want reminders.

In “Special Instructions”, you can enter more details on dosage, etc.

AZITHROMYCIN ✕

1 Search for a Medicine or Supplement

2 Form and Dosage

Select a Form first and then a Dosage.

Form *
TABLET, FILM COATED

Dosage *

BACK NEXT

3 Frequency

4 Refills

5 Special Instructions

To view your completed surveys, click Surveys/Studies on the left menu, and click “View” for the specific survey.

The screenshot shows the DataHub interface with a dark header bar containing the DataHub logo. The main content area is divided into two sections. The left section, titled 'Surveys/Studies to be Completed', has two tabs: 'New' (selected) and 'In Progress'. Below the tabs is a white box containing the PHELAN-McDERMID SYNDROME DataHub logo and the text: 'There are currently no surveys/studies to be completed. When your organization creates a survey/study, it will appear here.' The right section, titled 'Completed Surveys/Studies', has three tabs: 'Patient' (selected), 'Clinician', and 'Curator'. Below the tabs is a table with a dark green header. The table has two columns: 'Title' and 'Completed On'. There are three rows of data, each with a 'VIEW' button and an 'ANALYZE' button. At the bottom of the table, there is a pagination control showing 'Items per page: 5' and '1 - 3 of 3' with navigation arrows.

| Title | Completed On | VIEW | ANALYZE |
|--------------------|--------------|------|---------|
| PMSF Informed C... | 9/30/21 | VIEW | ANALYZE |
| PMSF Membership... | 9/30/21 | VIEW | ANALYZE |
| General Health ... | 9/30/21 | VIEW | ANALYZE |

Items per page: 5 1 - 3 of 3 < >

You can analyze de-identified results of the General Health Survey. You can see how many people answered Yes/No alongside you for specific questions.

To do this, click the Surveys tab on the lefthand menu



Click the completed tab

Click the three dots next to the General Health Survey

Surveys/Studies

New/In-Progress **Completed**

| Title | Published On | Completed On | Category | Type | |
|------------------------|--------------|--------------|-----------------|----------------|---|
| PMSF Membership Survey | Aug 25, 2021 | Oct 22, 2021 | Surveys/Studies | PATIENT | ⋮ |
| General Health Survey | Sep 01, 2021 | Oct 22, 2021 | Surveys/Studies | PATIENT | ⋮ |
| PMSF Informed Consent | Oct 08, 2021 | Oct 22, 2021 | Surveys/Studies | GLOBAL CONSENT | ⋮ |

Items per page: 5 1 - 3 of 3

VIEW: Title

- VIEW
- ANALYZE
- EDIT

Click "Analyze"

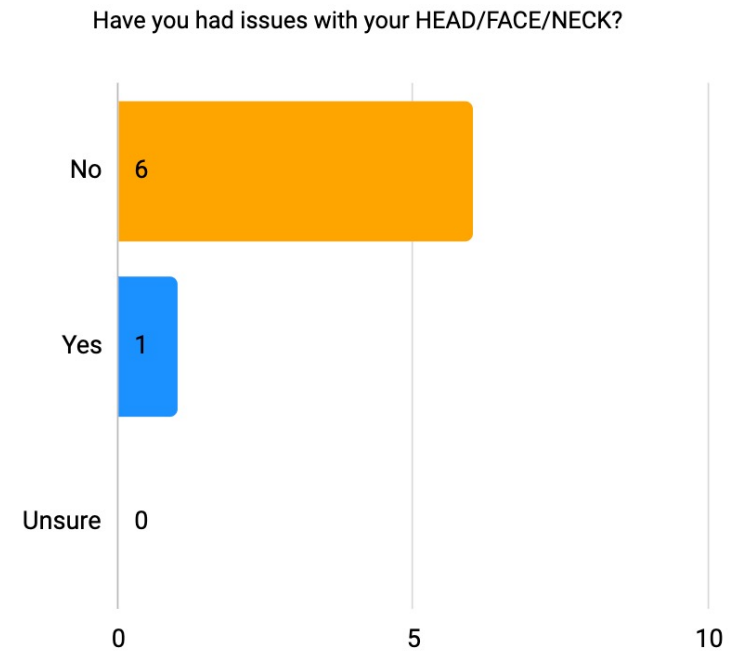
Go to the question that you want to analyze and look at the data!

Analytics

General Health Survey

| |
|---|
| Have you had issues with your GROWTH? |
| Have you had issues with your HORMONES a... |
| Have you ever been diagnosed with cancer... |
| Have you had issues with your HEAD/FACE/... |
| Have you had issues with your EYES and/o... |
| Have you had issues with your EARS and/o... |
| Have you had issues with BRAIN and/or NE... |
| Have you had BEHAVIOR and/or PSYCHIATRIC... |
| Have you had issues with your SKIN? |
| Have you had issues with your BONES, CAR... |

Your Response



This concludes the data which is most helpful for researchers

Kate Still
CAREGIVER
Viewing Test Patient

Sep 21, 2021

Test Patient

Incomplete Surveys/Studies

PHELAN - McDERMID SYNDROME
DataHub

You currently have no new/in progress surveys/studies. When your organization assigns a new survey/study, it will appear here.

Items per page: 5 0 of 0

Medicine and Supplement Reminders
No available reminders for today

Quick Add Symptoms & Activities

Quickly add any occurrence of your currently tracked symptoms or activities.
When an item in the section below is marked it will appear here for today's occurrences.

Powered By: Matrix

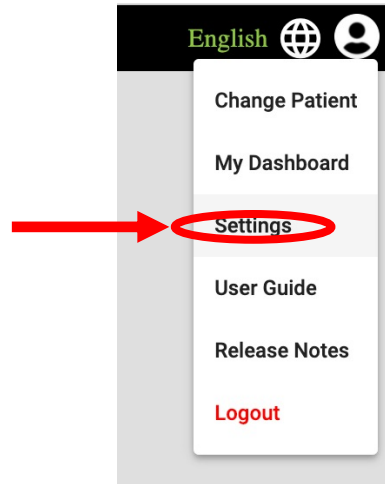
Feel free to look along the menu for other sections you are interested in filling out. This information can be helpful for your own tracking, and helpful across rare disease research. Anything in addition to genetic reports, general health, and medications, is considered a bonus.

To stay up to date on survey updates and recruitment for new research studies:

Click the person icon in the upper righthand corner.



Click “Settings” and then “Edit” at the bottom of the page.



Turn on notifications for new surveys and messages by toggling on text and/or email.

Notification Settings:

Medicine and Supplement Adherence Reminders

Get a reminder when it is time to take your medicine(s) and/or supplement(s).

Text Email

Prescription Refill Reminders

Get a reminder when it is time to refill your prescription.

Text Email

New Survey/Study Notifications

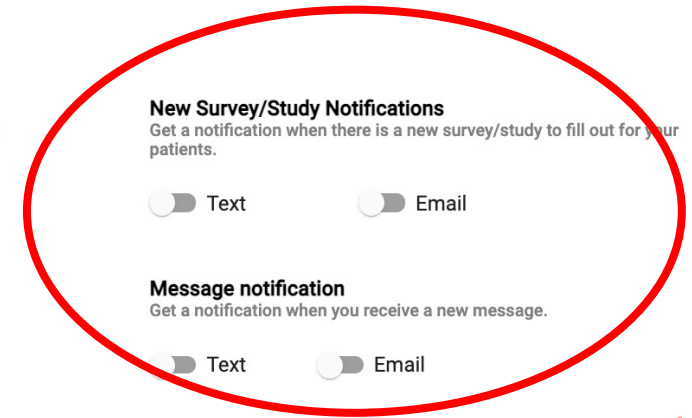
Get a notification when there is a new survey/study to fill out for your patients.

Text Email

Message notification

Get a notification when you receive a new message.

Text Email



Your progress can be saved at any time!

If you are logging back in after creating an account signing out of the DataHub, please go to:

<https://pmsf.acrossmatrix.com/>

For more information, please go to:

pmsf.org/datahub

**For questions, please contact
Datahub@pmsf.org**

Thank you for
helping advance
research in
PMS!



TM
pmsf.org