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](https://pmsfx.acrossmatrix.com/#/user-request)

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:

Fill out your information and hit “Sign Up”.

[Image of a form for signing 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):

### Caregiver info
- **Your First Name** * 
- **Your Last Name** * 
- **Your E-mail** * 
- **Your Date of Birth** *

### Patient Information
#### One Patient
- **First Name** * 
- **Last Name** * 
- **Date of Birth** *

#### Two Patients
- **First Name** * 
- **Last Name** * 
- **Date of Birth** *

---

Caregiver DOB

Person with PMS DOB
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!**)

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

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

This information is required.

Email Address

Send verification code

Continue

Cancel
You should receive an email like this:

Enter the code on the previous DataHub screen.
Create a new password and click “Continue”.

Create Password

New Password

Confirm New Password

Continue

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

Sign in with your email address

Email Address

Forgot your password?

Password

Sign in
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
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
- 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:

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.

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

This applies to you, the person filling out this information – either the person with PMS, or the person representing someone with PMS.
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 *
    mm/dd/yyyy
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.

<table>
<thead>
<tr>
<th>General Information</th>
<th>Demographics</th>
<th>First Name: John</th>
<th>Middle Initial:</th>
<th>Last Name: Smith</th>
</tr>
</thead>
<tbody>
<tr>
<td></td>
<td>Gender: Male</td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>City: New York</td>
<td>State: New York</td>
<td>Zip/Postal Code: 12345</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Email: <a href="mailto:johnsmith@gmail.com">johnsmith@gmail.com</a></td>
<td>Preferred Language: English</td>
<td></td>
<td></td>
</tr>
<tr>
<td>Other Information</td>
<td>Relationship/Marital Status: Single</td>
<td>Associated Patient Information</td>
<td></td>
<td></td>
</tr>
<tr>
<td></td>
<td>First Name: Jane</td>
<td>Middle Initial:</td>
<td>Last Name: Doe</td>
<td></td>
</tr>
<tr>
<td></td>
<td>Relationship to Patient: Mother</td>
<td></td>
<td></td>
<td></td>
</tr>
</tbody>
</table>
Enter in information about the person with PMS:

Their name will already be populated from login.
The "clinician" field should be grayed out and you do not need to enter anything here.

<table>
<thead>
<tr>
<th>Race *</th>
<th>Ethnicity *</th>
</tr>
</thead>
<tbody>
<tr>
<td>Required field. Please enter Race</td>
<td>Required field. Please enter Ethnicity</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Gender At Birth *</th>
<th>Gender Identity *</th>
</tr>
</thead>
<tbody>
<tr>
<td>Required field. Please enter Gender at Birth</td>
<td>Required field. Please enter Gender Identity</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Birthplace: Country *</th>
<th>Birthplace: State/Province/Region *</th>
</tr>
</thead>
<tbody>
<tr>
<td>Required field. Please enter Birthplace Country</td>
<td>Required field. Please enter Birthplace State/Province/Region</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Estimated Household Income *</th>
<th>Insurance Coverage for Patient? *</th>
</tr>
</thead>
<tbody>
<tr>
<td>Required field. Please enter Estimated Household Income</td>
<td>Required field. Please enter Insurance Coverage</td>
</tr>
</tbody>
</table>

<table>
<thead>
<tr>
<th>Clinician</th>
<th>Rare Disease(s) *</th>
</tr>
</thead>
<tbody>
<tr>
<td>Required field. Please enter Clinician</td>
<td>Required field. Please enter Rare Disease</td>
</tr>
</tbody>
</table>

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.
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.

Please click “Save” to continue.
Please upload any genetic reports for the person with PMS.
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.
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.
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.
To view your completed surveys, click Surveys/Studies on the left menu, and click “View” for the specific survey.
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

Click “Analyze”
Go to the question that you want to analyze and look at the data!
This concludes the data which is most helpful for researchers.

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.
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!