HOW TO REGISTER TO THE

PHELAN-McDERMID SYNDROME

INTERNATIONAL REGISTRY
1. To register your child in the Phelan-McDermid Syndrome International Registry (PMSIR), you can go to [http://22q13.org](http://22q13.org) and click on the big green button with the PMSIR logo on it (identified with the red arrow below). You then will be redirected to the PMSIR as shown on the second screen of this page.

2. You can also go directly to the PMSIR at [https://pmsiregistry.patientcrossroads.org/](https://pmsiregistry.patientcrossroads.org/). Once you are at the home page of the PMSIR, you have to click on "Click here to register now!". You may need to scroll down a little to see the button (pointed by the red arrow).

At any time, logged in or not, member or not, registered or not, you can access the "Home", "About", "Glossary" and "Clinical Trials" tabs, as pointed by the green arrow.
3. The following page will appear. This is the “Understanding Your Participation” section and you need to carefully read each paragraph before you can register. Here is the top of the page:

At the end of the page, you need to click on the "Next" button to access the "Registration and Consent" section.
4. The following page will appear. This is the "Registration and Consent" section. You need to carefully read all information given to you and answer each question by clicking in the appropriate (or only) box as shown by the red arrow. Here is the top of the page:

You need to fill every box followed by a yellow "!". If you point your mouse on a blue "i", a text box will appear, giving you additional information about the field. For example, there is a box to help you to choose an appropriate username or password (numbers of character required, etc.)

When you enter a username, a message will appear to confirm you if the one you entered is available or not for registration. If not, you will have to enter a new one. Once you enter a username free for registration (look at the message pointed by the green arrow), you will be authorized to click on the "Register" button at the bottom of the page.
5. There you go! Your registration is complete. To access your account, you need to log in, using the username and password previously given for registration.

6. Now you need to create a profile. You will automatically be directed to the "My Profile" tab when logging in for the first time. You need to fill in the form. Here is the top of the page:
7. At the end of the form, you need to carefully read each paragraph and answer every question of the "Consent" section.

8. At the bottom of the page, once you answered all questions, you have to click on the "Update" button and your profile is created.
9. You are now at the beginning of THE questionnaire. In fact, there are two of them. The first one is the “Clinical Questionnaire” and the second one is the “Developmental Questionnaire”. You can start now or leave the PMSIR to come back at another time. If you start now, you can stop anytime you want and come back at another time. Once you start answering the questions, after each completed page, you will be informed of the % completed. Here is the top of the first page of the Clinical Questionnaire:

At the bottom of the page, click on the “Next” button to save your answers in the PMSIR. You can leave some questions unanswered if you’re not sure and want to verify something in your papers for instance. You may want to skip a part of the questionnaire by clicking the “Next” button and go answer another section. The important thing is to make sure that when your answer is “no” or “unsure” or “not applicable”, you MUST select the appropriate option. If you leave a question unanswered, the PMSIR cannot consider the blank as a “no” or “unsure” or “not applicable”. The PMSIR then consider that you just didn’t answer the question and doesn’t count it in the statistics until you update and save a valid answer.
If you decide to leave before answering any question, the next time you will log in your account, this window will appear:

When your annual update will be due, you’ll receive a reminder about it in the "Notifications" section in the "My Profile" tab, as pointed by the red arrow.

You can select the Clinical or Developmental Questionnaire while you are in the "Profile" tab, as shown by the green arrow.

At any time, questionnaire completed or not, you can take a look or update your “Contact Info”. 
At any time, questionnaire completed or not, you can make your genetic report available to the researchers by attaching it (or “them” if you have a CGH and FISH report for instance) to your account in the “Attachments” tab. You need to select the type of document you want to attach (Genetic test results) and then browse your computer to upload the appropriate file. When the document is correctly identified in the “Choose a file to upload” field, you just have to click on the “Upload” button and your document will be saved in your account of the PMSIR. If you have a second genetic report, you can select “Other” as the type of document (because you cannot select twice the same type of document) and send an email to coordinator@pmsiregistry.org to inform them.

Once you completed the questionnaire, when you come back to “My Profile” tab or if you log in your account, the page will look like this:

(You may have a message in a “Notifications” section that will appear when needed.)
If you want to look at some statistics of the PMSIR, you just have to click on the "Explore" tab and from there, you will be able to have an overlook of some demographic data. You can also point and click on any section of both questionnaire to access statistics of some questions. Here is the top of the page when you access the "Explore" tab:

Here is the middle of the page. You can see the beginning of the Developmental Questionnaire sections on which you can click to look at their statistics:
On the next screen, you can see how statistics show up when we click on the "Diagnosis" section of the Clinical Questionnaire. You can also unroll additional graphics by clicking on the green categories appearing here and there between some graphics.

The other tab you can access while you are logged in your account is the "Forums" tab. There, you can ask a question in the appropriate category and/or read the comments other posted. Here is how it looks like:
The other secret the "Forums" tab holds is that if you leave your mouse on the "Forum" tab, another option will roll down and it is called "Ask an Expert". If you select this option, you will see the next window appear. You just have to select the reason for contact, write your question in the text box below it and click on "Send Contact Request". You will soon receive an answer by email (the one you previously gave in the "Contact Info" part of the "My Profile" tab).

If you need to update one or a few questions or if you need to do your annual update, just go in the "My Profile" tab and click on the questionnaire you need to re-do or update some questions from.
On the next screen, the Clinical Questionnaire was selected. From there, you can select a section heading to view and update one or a few questions. If you need to do your annual update, you just click on the “Re-take the survey” link as pointed by the green arrow.

Here is what you see when you click on a section heading. The “Diagnosis” heading was used in the next screen. Click on the paper and pencil image to upgrade this particular response. When available, you can click on the graphic image to see how your response compares to other members of the community (look at the green arrows).

Congratulations! The PMSIR has no more secrets for you!

Thank you for participating in this important project for our children.