Resource Summary Report

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Phelan-McDermid Syndrome International Registry

RRID:SCR_004230
Type: Tool

Proper Citation

Phelan-McDermid Syndrome International Registry (RRID:SCR_004230)

Resource Information

URL: https://pmsiregistry.patientcrossroads.org/

Proper Citation: Phelan-McDermid Syndrome International Registry (RRID:SCR_004230)

Description: International registry that consolidates information from individuals with Phelan-McDermid Syndrome into a single database, which will be utilized by researchers to understand Phelan-McDermid Syndrome better. In order to accelerate translational efforts (moving from basic laboratory research to meaningful health outcomes, such as therapies and treatments) related to Phelan-McDermid Syndrome, PMSF is funding the Phelan-McDermid Syndrome International Registry. The Registry is important for characterizing and understanding the syndrome better. Not only will the Registry provide valuable information for families and doctors to make the best care decisions possible, it will be important to help researchers decide what are the most important challenges to address. The Registry will also help scientists find out if there are any PMS patients who might be a good match for their research studies. Collecting information from PMS patients is very important, but protecting the privacy of people affected by PMS is also extremely important. In order to protect your privacy, Patient Crossroads (the company that designed the registry software) has designed many safeguards. Your child''s information will be de-identified so no one who looks at the data can identify you or your child. Your child''s information will be assigned a code. If a researcher is interested in learning more about your child, the researcher will ask the Patient Crossroads/PMSIR genetic counselor to contact you. A scientist will not be able to receive any identifying information about your child unless you give explicit consent for your child''s identity to be released to that researcher. BE PART OF OUR INTERNATIONAL REGISTRY The Registry will provide valuable information for families and doctors to make the best care decisions possible, and it will help researchers decide what are the most important challenges to address in PMS. Establishing the registry addresses two important scientific needs. First, scientists studying PMS need accurate, firsthand information to understand how PMS affects people. Second, scientists who are ready to start studies, such as those that test new treatments, will be able to access The Registry to identify people that may be eligible to participate in studies. In either case, your privacy is assured while the cause of

research is advanced. While raw data about PMS will be available to scientists, they won''t have access to any identifying information about your child unless you agree to have your child''s identity released.

Abbreviations: PMS International Registry

Resource Type: patient registry, people resource

Keywords: phelan-mcdermid syndrome, clinical trial, registry, therapy, treatment, child

Related Condition: Phelan-McDermid Syndrome

Funding: Phelan-McDermid Syndrome Foundation

Availability: The community can contribute to this resource

Resource Name: Phelan-McDermid Syndrome International Registry

Resource ID: SCR_004230

Alternate IDs: nlx_143654

Record Creation Time: 20220129T080223+0000

Record Last Update: 20260806T054400+0000

Ratings and Alerts

No rating or validation information has been found for Phelan-McDermid Syndrome International Registry.

No alerts have been found for Phelan-McDermid Syndrome International Registry.

Data and Source Information

Source: SciCrunch Registry

Usage and Citation Metrics

We have not found any literature mentions for this resource.