

Phelan-McDermid Syndrome Data Network (PMS_DN)

Principal investigator

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PCORI funded the development of PCORnet®, the National Patient-Centered Clinical Research Network, to make it easier and more efficient to conduct research. PCORnet is made up of Partner Networks that harness the power of large amounts of health data and patient partnerships.

Patient-Powered Research Networks (PPRNs) are networks operated and governed by patients, families, caregivers, and community members in close collaboration with clinical researchers, with the goal of shaping and conducting research to better serve their communities and improve their health outcomes. Several PPRNs, including the Phelan-McDermid Syndrome Data Network (PMS_DN), participated as Partner Networks in PCORnet.

PCORI funded PMS_DN’s participation in PCORnet from 2015 to 2018. This report outlines PMS_DN’s achievements in building its research infrastructure capacity to

1. Create ways to involve patients, families, and caregivers in decision making about the network
2. Create a way to collect and share data reported by patients for research
3. Involve patients, caregivers, families, and communities in the research process, including recruiting people to participate in research studies
4. Lead or join in research studies that focus on the community’s priorities

Title	Phelan-McDermid Syndrome Data Network (PMS_DN)
Network design	Patient-Powered Research Network (PPRN)
Lead partner	The Phelan-McDermid Syndrome Foundation
Other partners	Harvard Medical School Department of Biomedical Informatics Boston Children’s Hospital
Participants	519 individuals with Phelan-McDermid Syndrome
Area(s) of focus	Phelan-McDermid Syndrome and related genetic conditions

How does the network operate?

PMS_DN gave each partner oversight of different parts of the network. The Phelan-McDermid Syndrome Foundation (the Foundation) continued to collect and store all patient data, including genetic reports, electronic health records, and information about quality of life. It stored the data in a database called a registry. Boston Children’s Hospital took the electronic health records collected by the Foundation and turned them into usable data for researchers. The Harvard team made sure data were high quality and oversaw data analysis. Together the Harvard and Boston Children’s Hospital teams created ways to share data with researchers using software. The network developed rules and processes to share data with researchers with participants’ permission.

How did the network involve patients and other partners?

A parent of a young adult with PMS led the network. The network trained families on how to collect medical records and genetic information and how to submit that data to the registry. It also connected families who were eligible to participate in research with scientists who were leading studies. A person who specializes in working with families worked to understand their needs and involve them in the research process. The specialist led a group of up to 10 parents of individuals with PMS who helped identify research priorities and review and approve research studies. The parent group also helped improve systems for sharing patient health information and recommended ways to communicate with families. The network paid members of the parent group for their time.

The network used social networking sites like Facebook to connect families with researchers, who shared information about PMS and research activities. The network offered webinars for families that described PMS research topics and helped the network learn what research topics were most important to families. The Foundation hosted a conference every two years to bring together families affected by PMS from around the world to connect with one another and learn about PMS research.

Who is in the network?

The PMS_DN contained data on 519 participants as of March 30, 2019. The registry contains medical records, genetic testing results, and information about patient experiences with PMS. While a Partner Network in PCORnet, PMS_DN participated in three studies. The network participated with other PPRNs and other networks with similar research interests. The network has a publicly available website. Users who have special permission can browse and analyze data from all data sources, and those who have the highest level of approval can request access to individual-level data.

Population (as of 03/30/19)

Race/ethnicity

- 58% white
- 5% other
- 3% black
- 3% Asian
- 1% American Indian/Alaska Native
- 17% no information
- 12% Hispanic

Sex

- 53% female
- 47% male

Age

- 74% 17 years or younger
- 25% 18–44 years
- 1% 45–64 years

How does the network support future research?

The Foundation continues to build the PMS International Registry and the PMS_DN and actively involve families in all stages of the research process. Partners in PMS_DN continue to do analyses to help understand the associations between different genetic changes and characteristics of PMS. They are also comparing what can be uniquely learned about the condition among different data sources, including patient billing codes, electronic health records, and patient surveys.

To learn more about this network, visit www.pcori.org/RI-O'Boyle026.

Glossary

Institutional Review Board (IRB): A group that follows federal regulations, state laws, and institutional policy to review, monitor, and approve research in order to protect the ethical rights and privacy of the subjects involved.

Patient-Powered Research Networks (PPRN):

PPRNs are operated and governed by patient groups and their partners and are focused on particular conditions or populations.

PCORnet: PCORnet is a network of networks that brings together patients, clinicians, researchers, and healthcare systems to share information and participate in research.