Phelan-McDermid Syndrome Foundation
INTERNATIONAL REGISTRY

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Engaging Families in Research To Drive Progress: The Phelan-McDermid Syndrome International Registry and the PMS Data Network

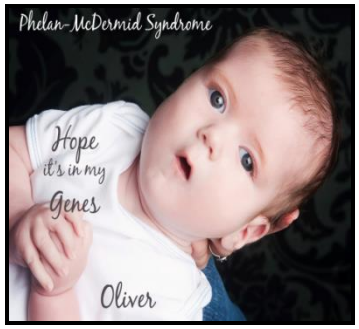
What does PMS look like?
Fast facts about PMS…

Features

- Developmental disability of varying degrees
- Autism
- Neonatal hyptonia
- Absent or delayed speech
- Impaired motor skills
- Sleep Issues
- Gastrointestinal issues
- Seizures
- Sensory issues

Genetic causes

- Deletions of 22q13
- Mutations of SHANK3



PMS International Registry

- Founded in 2011 by PMS Foundation parent volunteers and hosted by Patient Crossroads
- Collects contact info (for Foundation use only)
- Collects genetic reports (curated by a genetic counselor)
- Includes several questionnaires
  - Clinical (medical) questions (organized by organ) (100+ questions)
  - Developmental questions (100+ questions)
  - Adult specific questions (100+ questions)
  - Researcher-specific questions
- Provides de-identified data to qualified researchers
- Returns aggregate data to families
- >1000 families participating from 43 countries

Enabling Family Participation in Research

- We have our families' …
  - **Trust**
  - Contact information
- Families can consent to…
  - Share their data with other databases
  - Update their information yearly
  - Be notified of additional research opportunities
- We have relationships with researchers and industry and are effective liaisons

Maximize data about PMS → Return aggregate data to families → Families become engaged and comfortable participating in research

How are the data collected?

- All of the PMSIR data comes from patients and their caregivers → Patient Reported Outcomes

- Registrants complete or provide:
  - Demographics Form (age, sex, etc.)
  - Questionnaires (surveys)
  - Attachments (genetic test reports, etc.)

Phelan-McDermid Syndrome
INTERNATIONAL REGISTRY

Select English

Username ••••••••

Login

Forgot login?

Home | About | Glossary | Clinical Trials | Help

Welcome to the Phelan-McDermid Syndrome International Registry. The purpose of this registry is to consolidate information from individuals with Phelan-McDermid Syndrome into a single database, which will be utilized by researchers to understand Phelan-McDermid Syndrome better.

Click here to register now!

Researchers register here!

Newsflash

NEW

NEW RESEARCH OPPORTUNITY!

The Phelan-McDermid Syndrome Foundation is pleased to announce the first researcher-sponsored project via our PMSI patient registry. This study will explore medical problems such as sleep disturbances, attention deficit disorder, autism spectrum disorder and mitochondrial disorders for the purpose of identifying treatable aspects of medical disease related to Phelan-McDermid Syndrome.

Login and click the My Profile button to join these surveys today!

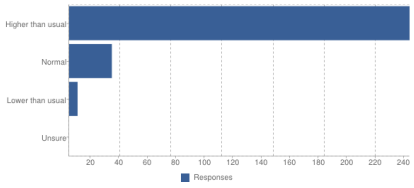
PMS International Registry FACTS

- Launched in 2011 by the PMS Foundation
- Registrants can consent to:
  - share data
  - be re-contacted
  - be contacted for clinical trials
  - be reminded to update data annually
- Collects contact info (for Foundation use only)
- Collects Genetic Reports (curated/de-identified by a trained genetic counselor)
- Includes several questionnaires:
  - Clinical - 100+ questions
  - Developmental - 100+ questions
  - Adult onset issues - 100+ questions
  - Researcher Initiated

PMS International Registry FACTS

- Provides de-identified data to qualified researchers
- Returns aggregate data to families (charts and statistics)
- IRB approval (annually reviewed)
- Year 2 - 50% of the global diagnosed families were registered.
- Year 5 - 71% of global diagnosed families were registered. (1,000+ registrants / 420+ with genetic data)
- Registrants are from 43 countries

Why does the Foundation Sponsor our Registry?



- For Our Families
  - Empower them to advocate and participate in research
  - Decrease survey fatigue *(by having ONE central registry for surveys)*
  - Provide access to aggregate data (charts)
  - Become better informed about the syndrome. This makes them better advocates in all arenas
  - Convenience of an on-line platform
  - New Q&A can be added as needed based on input from the families at conferences and social media
  - Build trust and experience in research. This is the “baby step” into the “research world” for many families. A positive experience will engage them in future projects/studies/clinical trials.
  - The data can be printed out for new doctors to review. Improves “training” of clinicians and caregivers

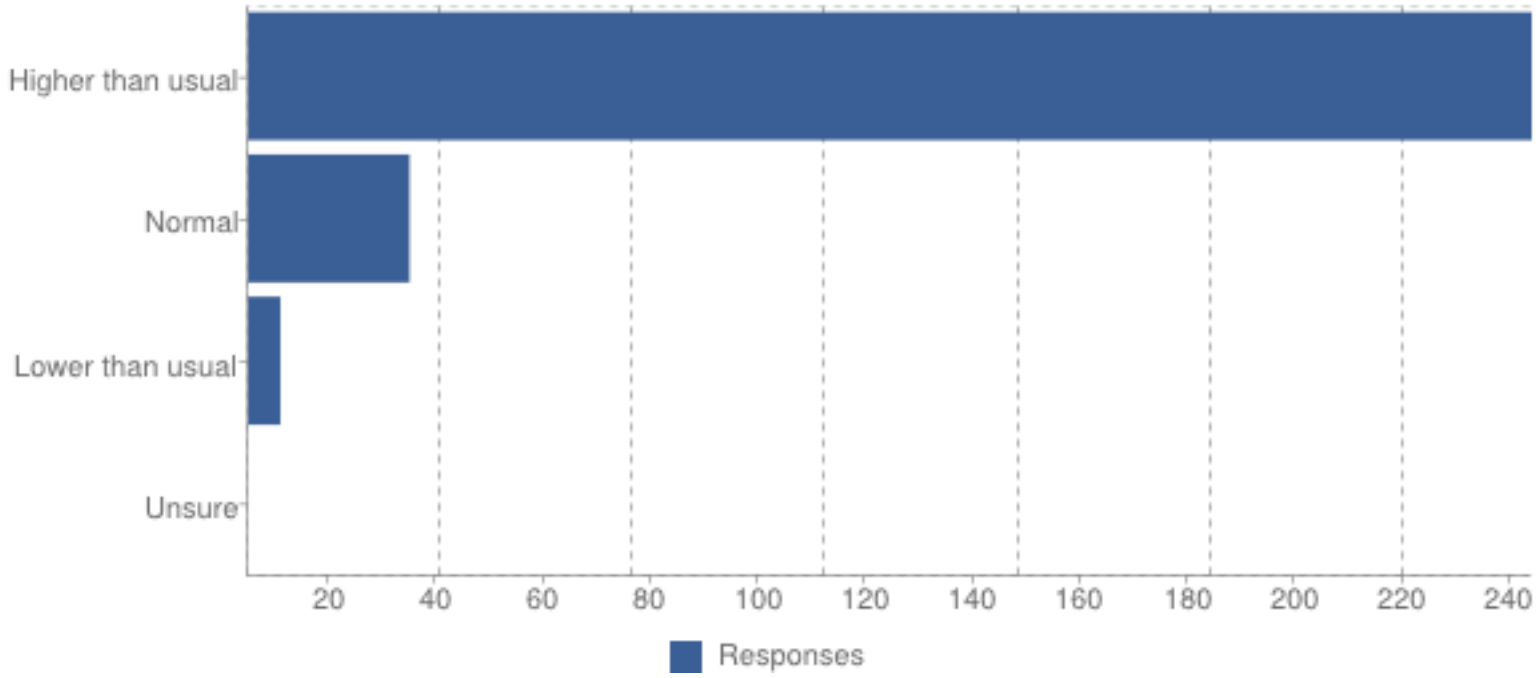
Why does the Foundation Sponsor our Registry?

- For the research community
  - Families are already recruited, **consented** and have completed the surveys.
  - Data can be shared with more researchers
  - **Data can be shared faster and for less money**
  - New surveys can be added at the request/cost of researchers and families will be directed by the foundation to answer them.
  - **Families can be re-contacted +++**

- For industry partners
  - Targeted (based on genotype or phenotype) recruitment for clinical trials
  - New surveys can be added at the request/cost of industry.

Aggregate Data Helps Families

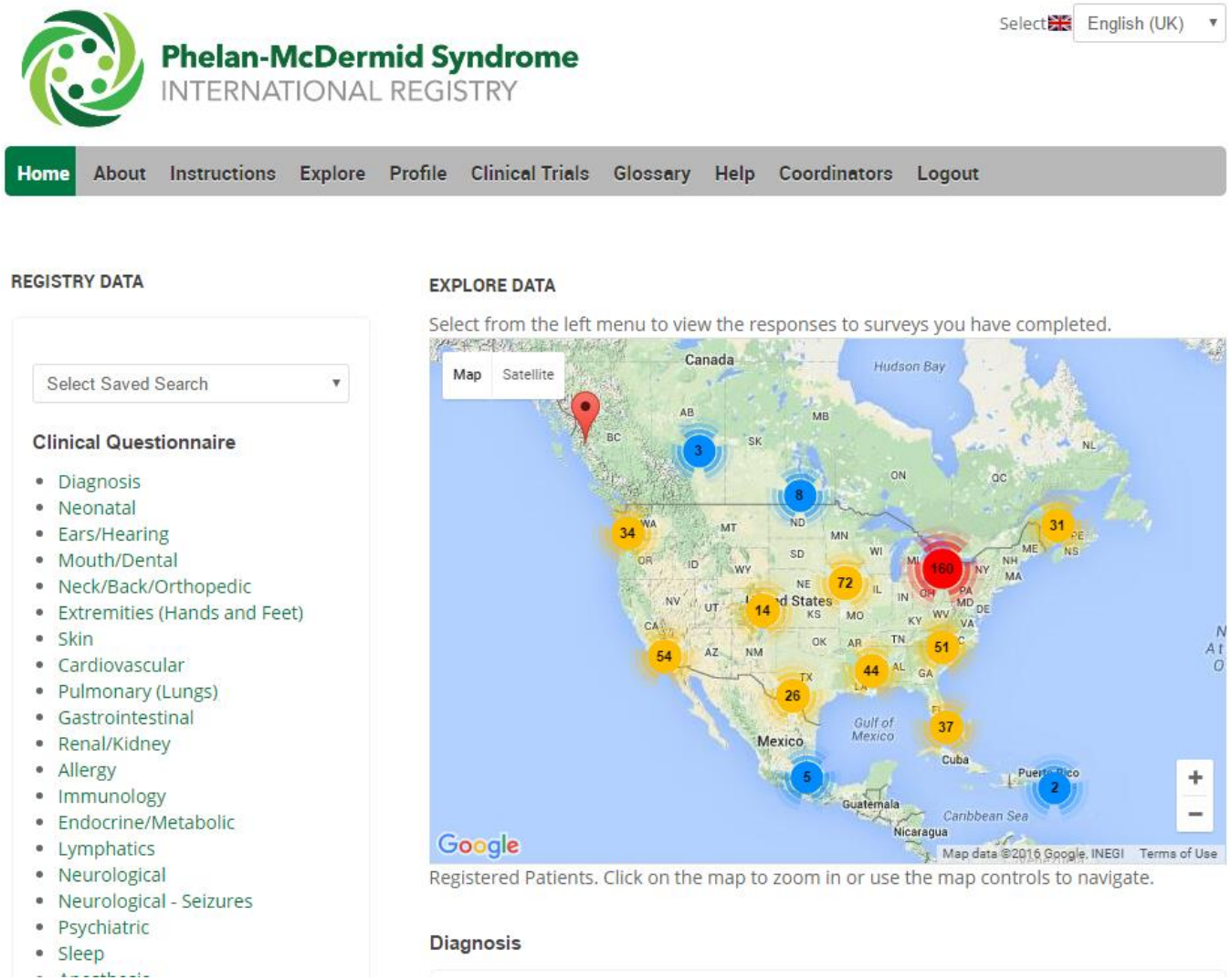
What is the patient's pain tolerance?



* The majority of patients with PMS are non-verbal and do not feel pain appropriately. This can be a very dangerous combination in a school setting if there is not sufficient supervision. Parents can use data from the PMS International Registry to make their case for on-to-one supervision in the school setting.

You can *Explore* anytime:

Allows users to see some of the data we have collected, in overall (aggregate) format



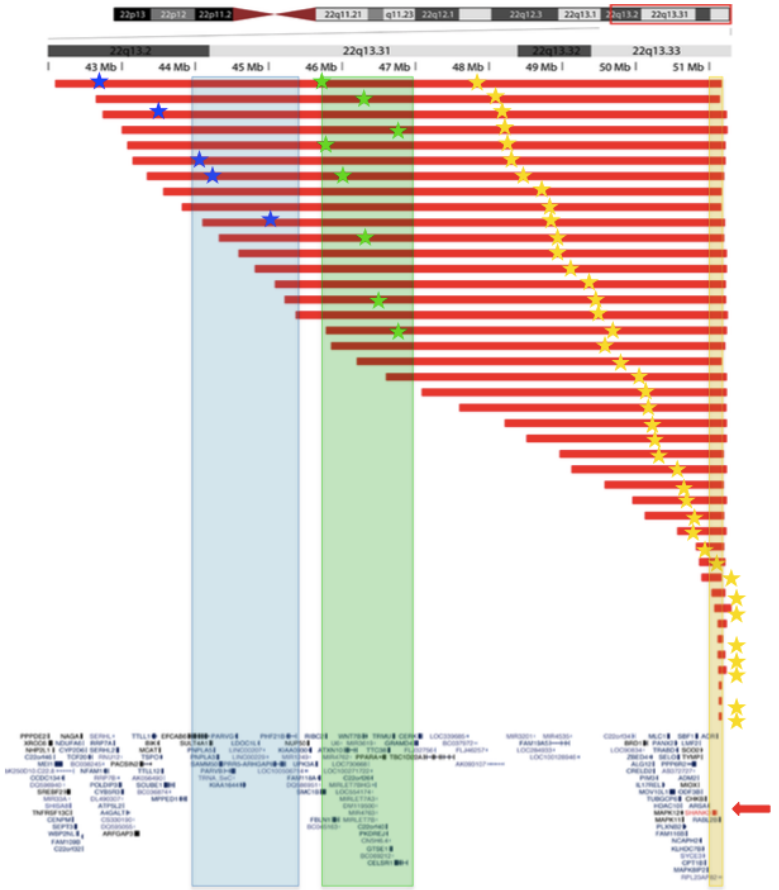
Genotype-phenotype correlations

Catalina Betancur, July 2014

Genotype-phenotype correlations

Examples:

- ★ abnormal language
- ★ heart defects
- ★ renal defects



Aims

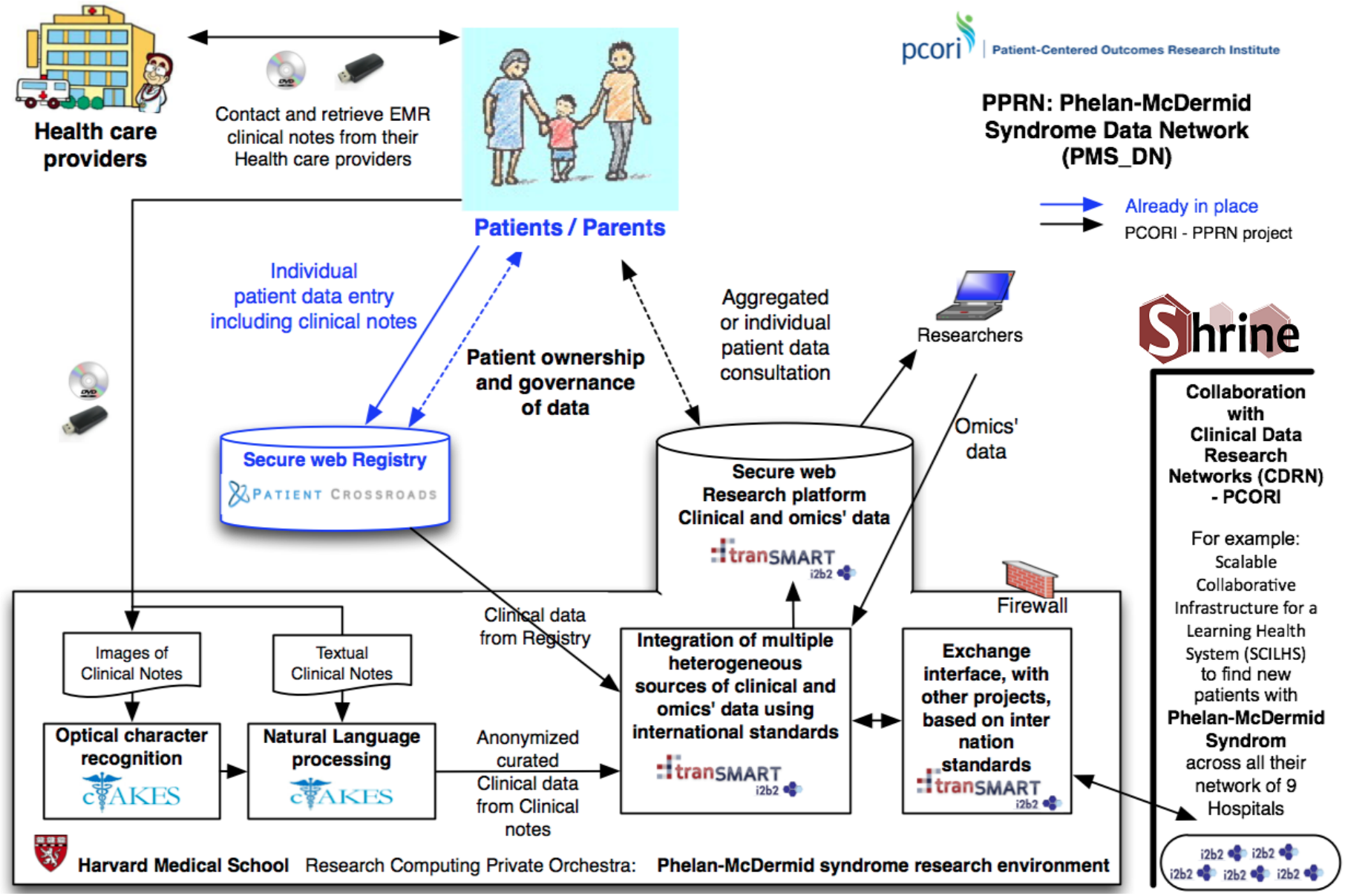
- identify genes/regions involved in specific phenotypes
- inform clinical practice

Success Stories

- Successful recruitment for several studies based on geographic location, genetotype, phenotype or age.
- Biosamples have been collected from very targeted patient groups with specific type of mutation.
- Families have gained additional educational or social services based on the data from the PMSIR.
- Families are excited about research exceeded expectations of biosample (blood & skin) collection.
- Our Families are “research ready”

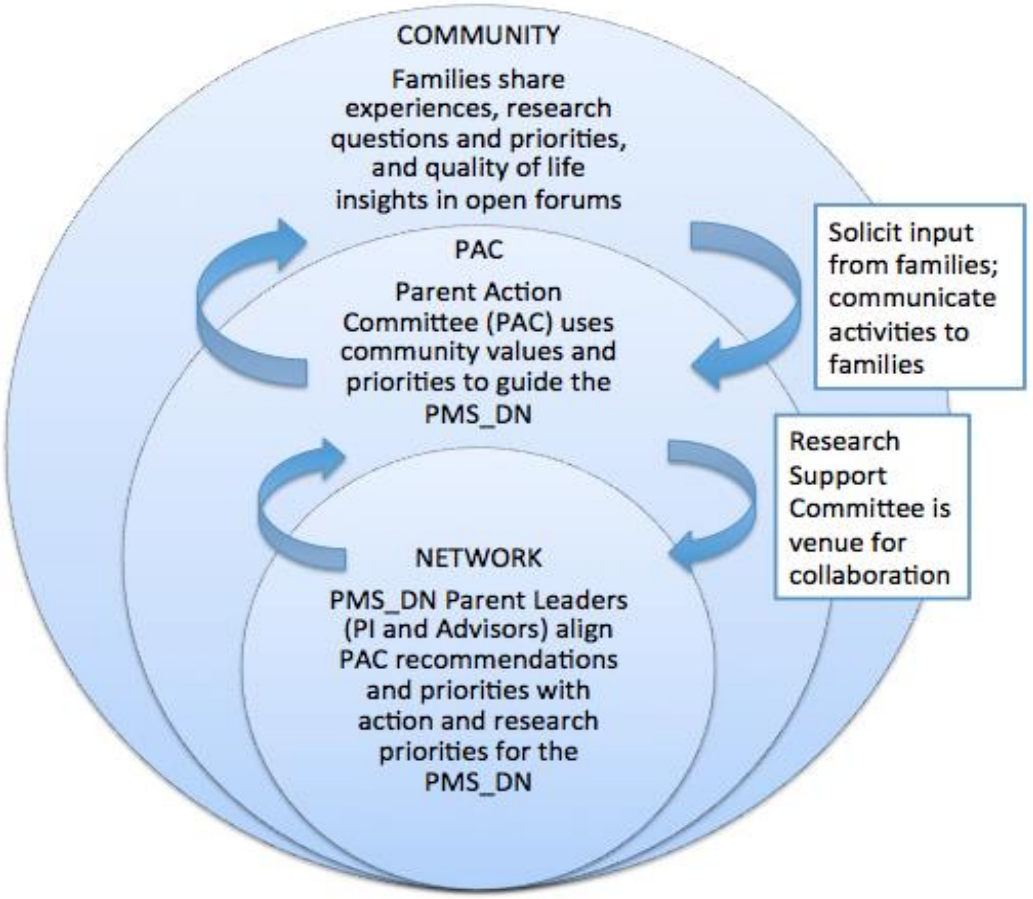
PMS_Data Network (PCORI Project)

- Through a PCORI contract the PMSIR data is being shared with a larger i2b2/tranSMART data warehouse and web interface.
- All data from the PMSIR and knowledge extracted from clinical notes provided from patient Electronic Health Records (EHR) will be integrated into one interface.
- Additional data from other sources (from wearable devices, medical collection apps, etc) could also be added to this data warehouse and interface.
- Researchers and industry will be able to access data from multiple sources, on one patient from one portal, AND there will be extensive analytical tools available to do real-time analysis of the data.


Health care providers
Contact and retrieve EMR clinical notes from their Health care providers
Patients / Parents
pcori
Patient-Centered Outcomes Research Institute
PPRN: Phelan-McDermid Syndrome Data Network (PMS_DN)
Already in place
PCORI - PPRN project
Individual patient data entry including clinical notes
Patient ownership and governance of data
Aggregated or individual patient data consultation
Researchers
Shrine
Collaboration with Clinical Data Research Networks (CDRN) - PCORI
For example: Scalable Collaborative Infrastructure for a Learning Health System (SCILHS) to find new patients with Phelan-McDermid Syndrom across all their network of 9 Hospitals
Secure web Registry
PATIENT CROSSROADS
Secure web Research platform Clinical and omics' data
tranSMART i2b2
Omics' data
Firewall
Clinical data from Registry
Images of Clinical Notes
Textual Clinical Notes
Optical character recognition
cTAKES
Natural Language processing
cTAKES
Anonymized curated Clinical data from Clinical notes
Integration of multiple heterogeneous sources of clinical and omics' data using international standards
tranSMART i2b2
Exchange interface, with other projects, based on inter nation standards
tranSMART i2b2
Harvard Medical School
Research Computing Private Orchestra:
Phelan-McDermid syndrome research environment
i2b2 i2b2 i2b2 i2b2 i2b2

PMS Data Network

- Nearly 600 families participating in the PMS_DN (320 US)
- Genetic Reports from nearly 300 families
- Clinical notes from 114 families and counting



Parents standing in line to re-consent to PMS_DN

What is the Value Proposition for Families?

- Who will benefit from these data collections?
  - PMS patient: more research that may lead to better therapies or treatments
  - PMS Family: more understanding of the syndrome
  - Medical team: more understanding of the disease progression
  - Education team: more understanding of the syndrome
  - Researchers: faster, less expensive access to patient data
  - Industry: faster, less expensive access to patient data going back years before clinical trials and continuing through post market

Final Thoughts

- PCORI funding has enabled the Foundation to build the PMS_DN in collaboration with Harvard Medical School
- Input from families has provided key guidance to this project
- Authentic engagement of families has allowed us to populate this resource
- Together, these have created new opportunities and research collaborations for the Foundation
- We couldn’t do it without our PMS families

Engage patients and families early and often in research!

Thank you