



Phelan-McDermid Syndrome Foundation  
INTERNATIONAL REGISTRY

Alison Turner, PMSF Regional Rep UK & Ireland  
and Gibson's mum

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# Phelan-McDermid Syndrome International Registry – PMSIR Patient Advocacy Group Model



# Gibson's story





# Regional Links & Global Partnership

The screenshot shows the PMSF UK website. At the top is the PMSF UK logo, which consists of a stylized circular design with dots inside, followed by the text 'pmsf UK'. Below the logo is the text 'Phelan-McDermid Syndrome Foundation UK'. To the right of the logo are social media links for Facebook, Twitter, and YouTube. Below these links is a text box that says 'Text PMSF22 £2 / £5 / £10 to 70070 to donate now eg PMSF22 £5'. Below this is a yellow button that says 'Donate Now'. Below the logo and text is a navigation bar with links: Home, About Us, What Is PMS?, PMS Support, PMSF & Registry, Shop, Fundraising, and Contact Us. Below the navigation bar is a large photo of a group of people sitting together. Below the photo is a section titled 'Parental Support' with a sub-header 'Parental Support, Social Services and Respite Care'. To the right of this section is a blue button that says 'Useful Links'. Below the 'Parental Support' section is a paragraph of text. To the right of this paragraph is a purple box with the title 'New diagnosis' and the sub-header 'What happens next?'. Below this title is a paragraph of text. Below this paragraph is another paragraph of text. Below this paragraph is a list of bullet points.

www.pmsf.org.uk/parental-support/ — Parental Support – PMSF UK | Phelan McDermid Syndrome Foundation UK

Top Sites

Parental Support – PMSF UK | Phelan McDermid Syndrome Foundation UK

 **pmsf UK**

Phelan-McDermid Syndrome Foundation UK

Facebook Twitter YouTube

Text **PMSF22 £2 / £5 / £10 to 70070**  
to donate now eg **PMSF22 £5**

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**Parental Support**

**Parental Support, Social Services and Respite Care**

It takes time to understand the Phelan-McDermid Syndrome diagnosis and what that means for the family. Coming to terms with a child's disability can be very emotional. The process of completing forms for, Disability Living Allowance, Education Health and Care plans on top of medical appointments can be overwhelming. Most boroughs in the UK should have support in place for parents with special needs and disabled children. All parents of disabled children have the right to ask for an assessment by Social Services. (in Scotland this is known as the Social Work Department, and in Northern Ireland it is the Health and Social Services Trust). Your local council has a duty to provide these services under the Children Act 1989. You can ask to be referred by your Paediatrician or the school or self-referral through a GP.

**Useful Links**

**New diagnosis**  
**What happens next?**

Here is a check list for the areas you may find helpful to look at for the person who has been diagnosed with Phelan-McDermid Syndrome.

All of these areas will provide the individual with PMS and their family with additional support. Please don't feel like you are alone in this diagnosis.

There is a worldwide community of families affected by Phelan-McDermid Syndrome and we are here to support you.

Take a deep breath and keep reading.

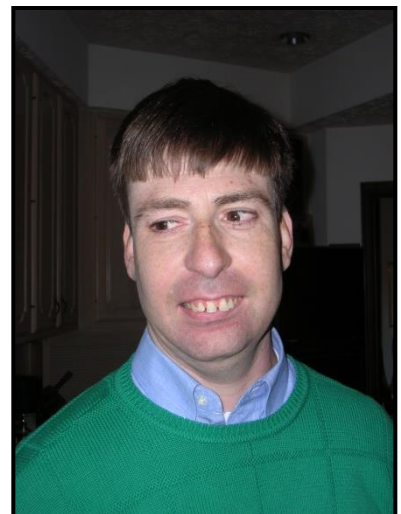
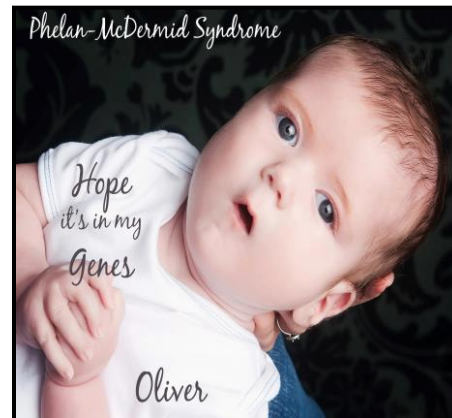
- Medical appointments necessary lead by Paediatrician

www.pmsf.org.uk



# What does Phelan-McDermid Syndrome (22q13 Deletion) look like?

- Global delays
- Absent or delayed speech
- Sleep Issues
- Gastrointestinal issues
- Seizures
- Sensory issues
- Inappropriate Behaviours
- Associated with ASD (80%)
- Hypotonia (97%)
- Cardiac and Renal issues (less 50 %)







# PMSF Who Are We?

- Young (founded 13 yrs ago) we are primarily parent volunteers
- Small (1,200 known diagnosed globally)
- Passionate and determined to move research forward (Registry, Repository, Animal models, etc.)
- 2011 launched the PMSF International Registry
- 2013 Awarded the Patient Centered Outcome Research Institute contract (PCORI)





# Patient Advocacy Group

The value of Patient Advocacy Groups (PAG) in Research

- We have our families ...
  - Trust & Contact information
- We have...
  - Ability to inform and recruit patients easily and quickly
  - Ability to collect patient-reported data through our registry
  - Consent from families to be re-contacted



# PMS **International** Registry FACTS

- Founded in 2011 by the PMS Foundation
- Hosted by Patient Crossroads (they have international clients)
- Collects contact info (for Foundation use only)
- Collects Genetic Reports (curated/de-identified by a trained genetic counselor)
- Includes several questionnaires
- Provides de-identified data to qualified researchers
- Returns aggregate data to families (charts and statistics)
- IRB approval



# Why do **we** Sponsor our Registry?

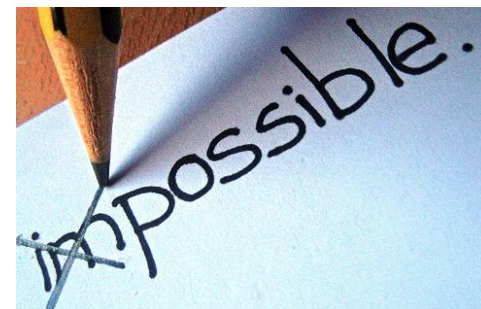
- For Our Families
  - Build trust and experience in research
  - **Empower them to participate in research**
  - **Decrease survey fatigue** (by having ONE central registry for surveys)
  - Provide access to aggregate data (charts)
  - Opportunity to contribute (feel helpful)
- For the research community
  - Data can be shared with more researchers
  - **Data can be shared faster and for less money**
  - More researchers can access the data
  - New questions and answers can be added as needed (Lymphedema experience)
  - The data can be printed out for the doctors to review







# How did we accomplish what others said we couldn't?



We...

- Found the right vendor
- Compiled potential Q&A (including Autism Speaks AGRE)
- Consulted with researchers about the Q&A
- Created necessary documents: Informed Consent, IRB protocol, marketing materials, etc. (with the help of outside advisers)
- Families Beta tested
- Marketed to families
- Returned data to patients/families whenever possible
- Will re-assess the registry after 4 years and change as needed overhaul



# Why did we put our limited time & resources into building a patient registry?

- Better characterize syndrome
- Educate and **empower** families
- Improve diagnosis and clinical care
- Provide data for pre-clinical and clinical research
- Identify cohorts
- Facilitate recruitment & reduce enrollment lag
- Connect families to research opportunities
- Collecting patient contact information (for communications and recruitment)





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.....

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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.

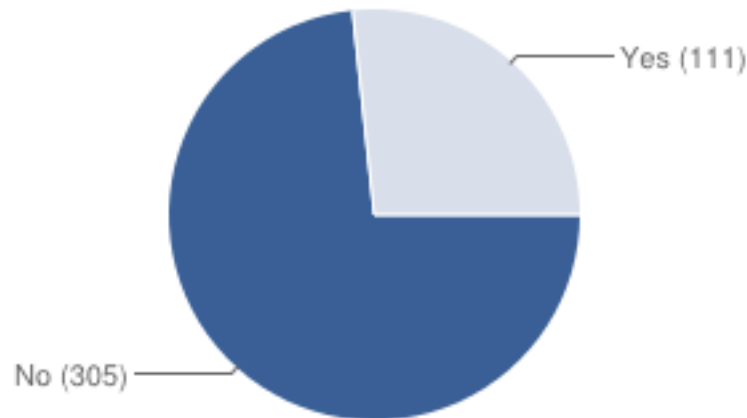
Login and click the My Profile button to join these surveys today!



# Aggregate Data Helps Families

- Returning data to families is very helpful in communicating with the care team

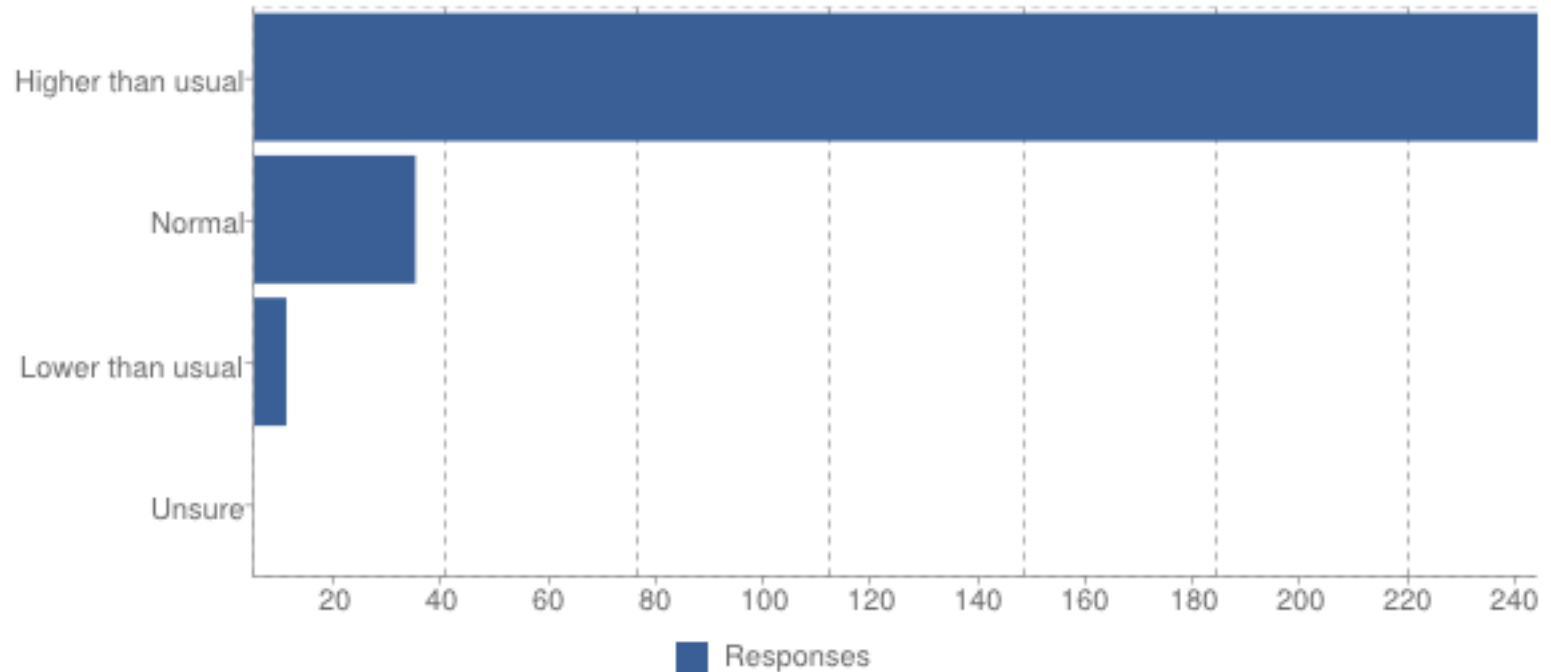
*Does the patient have seizures?*





# Aggregate Data Helps Families

*What is the patient's pain tolerance?*







# PMSIR Accomplishments

In 4 years the PMSIR has...

- Registered 750+patients of known 1,200 foundation (diagnosed) members worldwide
- >97% of families agree to be recontacted and notified about clinical trials
- Translated registry questions into 3 additional languages (Spanish, French, Italian)
- Crafted 3 surveys driven by parent feedback and priorities (Clinical, Developmental, Adolescent and Adult)
- Expanded collaborations with the research community
- Registered families from 43 countries





# Success Stories

- Posted recruitment message to families about a behavior study at noon; the next day by 9 am the study was filled
- Invited families to an regional gathering in the UK, and now many of the families are enrolled in clinical trials in Europe (EUAIMS)
- Recruited for biosamples at 2012 Family Conference - 50 families volunteered to donate blood, skin punches, and buccal swabs; Had to turn controls (parents) away and ran out of punch biopsy kits



# Bio SAMPLES

- Collection of blood and skin from PMS patients and parents at the 2012 PMSF family conference. The PMSF coordinated the collection, Stanford Univ. provided the IRB and the Nat'l Institute of Mental Health (NIMH) paid for the processing, storage and management of the collection.
- Held in centralized repository (Rutgers/New Jersey).
- the NIMH owns the collection and will release samples to any legitimate researchers (in the world) who request it.
- All researchers must be IRB approved, sign off on agreement.
- There are at least 3 researchers that have accessed samples in the first year for studies re: Autism, Gastro issues and other related symptoms.



## What we've learned...

- Patients/families have limited time:
  - Short, specific surveys are best
  - Ability to go back to finish is preferred
  - Too many questions can be overwhelming
  - If the first experience took too long then they are less likely to update annually – diminishing the longitudinal data
- Participating in the registry **empowers** parents/families.
- Families are using PMSIR to **inform their physicians** about the syndrome and specific conditions





## Parting Thoughts ...

- Parents are in this for life so you've got very committed participants
- Parents/carers have a lot of VAULABLE DATA
- Once you have correlated the data, research and government bodies will follow.
- Don't let perfect be the enemy of good





# Thank you

- Autism Speaks – Andy Shih & Amy Daniels
- NUI Galway – Geraldine Leader
- Trinity College Dublin – Louise Gallagher & June O'Reilly
- Phelan-McDermid Syndrome Foundation Global Partners

