pmsf UK

Phelan-McDermid Syndrome Foundation UK

This rare genetic disorder affects speech, mobility and cognitive development.



What we do

The Phelan-McDermid Syndrome Foundation UK (PMSF UK) is a registered charity in the United Kingdom. PMSF UK is about providing a supportive inclusive community for those who have or know someone with Phelan-McDermid Syndrome (PMS). We provide family days and get-togethers and some limited support for additional therapies for the person with PMS.

There are almost 250 people who are diagnosed with Phelan-McDermid Syndrome in the UK & Ireland, and over 2200 worldwide. All people with PMS have difficulty communicating; many are non-verbal and have issues around cognitive development and mobility. People diagnosed with PMS need additional care to support their daily lives.

PMSF UK is a global partner with the Phelan-McDermid Syndrome Foundation that has members around the world in over 59 countries. Although it is a rare disease it affects people across the globe. PMSF UK is bringing that community together and we can put UK families in touch with other local families. Support is critical when you are caring for someone who has the syndrome.

Thank you for taking an interest in learning about Phelan-McDermid Syndrome.

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Phelan-McDermid Syndrome Foundation UK

Registered Charity No. 1157423



What is Phelan-McDermid Syndrome?

Phelan-McDermid Syndrome (PMS) is a rare genetic disorder that affects speech, mobility, and cognitive development.

The syndrome, which affects over 2200 people worldwide, is a result of a deletion of chromosome 22q13 or a SHANK3 mutation. SHANK3 is a type of protein that is missing or mutated in many cases of PMS resulting in global developmental delay.

Some of the features of people with Phelan-McDermid Syndrome are:

Physical

- Absent or severely delayed speech
- Hypotonia (low tone)
- Global developmental delay
- Normal or accelerated growth
- Minor dysmorphic features, including wide brow, wide nasal bridge, ptosis, large fleshy hands and flaky toenails

Behaviour

- Autistic or ‘autistic like’ behaviour
- Persistent mouthing and chewing, sometimes teeth grinding or tongue thrusting
- Decreased perception of pain
- Aggresiveness

Neurological

- Seizures
- Anachroid cysts
- Sleep disturbance

People with Phelan-McDermid Syndrome will need additional care their entire lives.



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

- Disability Living Allowance (DLA) or Personal Independence Payment (PIP) ☑
- Register with PMSF and PMSF UK ☑
- Education, Health and Care Plan (children 0-25 years) ☑
- Blue Badge / Mobility scheme ☑
- Complete International Registry (PMSIR) ☑
- Connect with other families on the closed Facebook family pages ☑
- Support for you and register with local disability groups ☑

Phelan-McDermid Syndrome Foundation

The mission of PMSF is to improve the quality of life of people affected by PMS worldwide by providing family support, accelerating research and raising awareness.

PMSF facilitates connections among families through their website, monthly newsletter, Facebook, Twitter, regional gatherings and a biennial international conference. PMSF are working hard to help find effective therapy for people with Phelan-McDermid Syndrome. PMSF fund important basic and translational science, an international web-based patient registry, and scientific meetings.

PMSF has a global partnership with PMSF UK. A Global Partner with the same mission to improve the quality of life of people affected by PMS worldwide.

www.pmsf.org

GLOBAL PARTNER

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International Registry (PMSIR)

The Phelan-McDermid Syndrome International Registry is a patient-driven registry that consolidates information from individuals with Phelan-McDermid Syndrome into a single database, which is utilised by researchers to better understand PMS.

Not only does the Registry provide valuable information for families and doctors to make the best care decisions possible, it is important to help researchers decide what are the most important challenges to address.

The PMSF funds the PMSIR in order to accelerate basic laboratory research to meaningful health outcomes, such as therapies and treatments related to Phelan-McDermid Syndrome.

The registry has members from over 59 countries, bringing together this one community.

https://pmsiregistry.patientcrossroads.org

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JEANS FOR GENES DAY

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