pmsf UK

Phelan-McDermid Syndrome Foundation UK



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





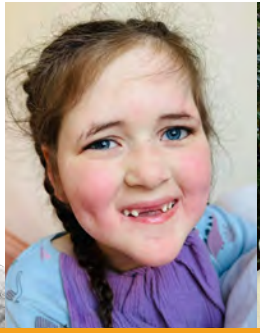
What we do

The Phelan-McDermid Syndrome Foundation UK (PMSF UK) is a registered charity in the United Kingdom. We have built a supportive inclusive community for those who have or know someone with Phelan-McDermid Syndrome (PMS) in the UK & Ireland. We provide family days and regional get-togethers and some limited financial awards for the person with PMS and their family.

There are almost 500 people who are diagnosed with PMS in the UK & Ireland, and 5000 worldwide. All people with PMS have difficulty communicating; many are non-verbal, autistic, and have issues around cognitive development and mobility. People with PMS need additional care to support their daily lives.

PMSF UK is a global partner with PMSF that has members around the world. 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.




What is Phelan-McDermid Syndrome?

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

The syndrome is a rare disease. It is a result of a deletion of chromosome 22q13 or a SHANK3 mutation. SHANK 3 is a type of protein that is missing or mutated in many cases of PMS that affects brain development and functioning. This results in intellectual and physical disabilities that vary from person to person. Most people with PMS require long term care and medical attention.

It is estimated that approximately 1 in 10,000 people may have this genetic condition. Many medical professionals are unfamiliar with the syndrome, and it's underdiagnosed.

Features of 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

Behavioural

- Autistic or 'autistic like' behaviour
- Persistent mouthing and chewing, sometimes teeth grinding
- Decreased perception of pain
- Behaviours of concern (aggressiveness, withdrawn, oversensitive to touch)

Neurological

- Seizures
- Anachroid cysts
- Sleep disturbance







New diagnosis, what happens next?

Please find below a checklist that you may find helpful for your family and the person who has been diagnosed with PMS.

- Disability Living Allowance (DLA) or Personal Independence Payment (PIP) ☑
- Education, Health and Care Plan (children 0-25 years) ☑
- Blue Badge / Mobility scheme ☑
- Register with PMSF and PMSF UK ☑
- Connect with other families on the closed Facebook support groups ☑
- Register with the international DataHub ☑
- Register with local disability groups ☑
- Support from your local council with respite care, short breaks, medical equipment, their local offer ☑



Phelan-McDermid Syndrome Foundation

PMSF UK is a global partner with PMSF, which is the international organisation for people affected by PMS and has members in over 78 countries around the world. We share a common mission to improve the lives of those affected with PMS.

PMSF is based in the USA, manages all new membership worldwide and facilitates connections among families through their website, monthly newsletter, Facebook, and a biennial international conference. PMSF also works closely with researchers, pharmacological companies, biotechnology, academic institutions and other external scientists with the goal of finding effective treatments for Phelan-McDermid Syndrome.



www.pmsf.org

Phelan-McDermid Syndrome DataHub

The DataHub is an online data collection platform where families can enter medical information about PMS that assists researchers, pharmaceutical companies, and others who are working to unlock the many aspects of the syndrome.

The DataHub is also a place for families to organize their medical information and compare it with others by viewing summarized de-identified data to inform medical decisions, and for their own empowerment.

www.pmsf.org/datahub

Contacts

- **PMSF UK**
  www.pmsf.org.uk
  info@pmsf.org.uk

- **CureSHANK** www.cureshank.org
- **Unique** www.rarechromo.org
- **Genetic Alliance** www.geneticalliance.org.uk
- **Gene People** www.genepeople.org.uk
- **ERN-ITHACA** www.ern-ithaca.eu

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