



support

Family Support

The Phelan-McDermid Syndrome Foundation understands the challenges and rewards of raising a child with the Syndrome. Through resources such as our website, quarterly newsletter, family conference, patient registry and webinars, we are working to make sure families have access to credible and up-to-date information about the Syndrome.

We believe in the strength of an international community of support where those affected by the Syndrome can find acceptance, comfort, information and practical advice. Through Yahoo Groups, Facebook, Twitter and regional networks, we are empowering families to be effective advocates in the medical, educational and research communities. Families can get connected by registering as a member with Phelan-McDermid Syndrome Foundation. To register go to www.pmsf.org. There is no cost to join!

Our Pledge

The Phelan-McDermid Syndrome Foundation offers information, family support, and hope to those affected by Phelan-McDermid Syndrome. We address the needs of our families in many ways including a global network of communication and significant investments in research. We offer hope that one day the debilitating effects of the Syndrome may be minimized or reversed.

fundraising

Donate Now!

Your support is critical in achieving our goals of improving the quality of life for both individuals and caregivers affected by Phelan-McDermid Syndrome. It is through the generous support of our donors that we are able to fund important initiatives such as:

- Scientific Symposia - to advance scientific knowledge about the Syndrome and SHANK3 and to foster collaboration among scientists
- Scientific Grants and Research Fellowships - to accelerate the development of effective therapeutics through scientific research
- International Registry and Biorepositorires - to build resources to stimulate and sustain research interest in the Syndrome
- International Conference – allows families and researchers to gather biennially to learn more about the Syndrome
- Family Scholarships – given on an as-needed basis so families can attend the Foundation's international conference
- Coordinate support for families around the world through outreach and awareness

The Phelan-McDermid Syndrome Foundation is a 501c3 non-profit organization. The funds raised by the Foundation are used to cover many areas of need including research, support for our families, conference costs, grant writing and administration. Donations are tax deductible and should be made payable to Phelan-McDermid Syndrome Foundation.

For more information or to make an on-line donation now, visit www.pmsf.org

outreach

Our Mission

The mission of Phelan-McDermid Syndrome Foundation is to improve the quality of life of people affected by Phelan-McDermid Syndrome worldwide by accelerating research, providing family support and raising awareness.

For more information, visit:

www.pmsf.org

or call:

1-941-485-8000

Family support provide through Facebook, Twitter and Yahoo! Groups.


Phelan-McDermid SYNDROME FOUNDATION

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Phelan-McDermid SYNDROME







medical

What is Phelan-McDermid Syndrome?

Phelan-McDermid Syndrome, sometimes called 22q13 Deletion Syndrome, is an underdiagnosed genetic condition caused by deletion of the terminal end of chromosome 22 or mutation of the SHANK3 gene or Ring chromosome 22, in which the 22nd chromosome forms a ring. In most cases, the condition is not inherited, but results from a *de novo* (spontaneous) mutation.

Core Features

The most common characteristics in Phelan-McDermid Syndrome include:

- Intellectual disability of varying degrees
- Delayed or absent speech
- Symptoms of Autism or Autism Spectrum Disorder

Deletion Size

Deletion sizes are highly variable, as are the clinical features of Phelan-McDermid Syndrome. Individuals with larger deletions may be more likely to have dysmorphic features, neonatal hypotonia, neonatal feeding problems, atypical reflexes, and greater delays in meeting developmental milestones.

Diagnosis

Array comparative genomic hybridization (CGH), also called microarray, is currently the most common method for diagnosing Phelan-McDermid Syndrome. Fluorescence in situ hybridization (FISH) or chromosome analysis may detect larger deletions. If a diagnosis of Phelan-McDermid Syndrome is suspected, but no deletion of 22q13 is detected through CGH, DNA sequencing may detect mutations of the Shank3 gene. Deletions, rings, translocations and mutations can be detected from blood samples as well as from skin cells or cells used for prenatal testing.



For more information, visit www.pmsf.org



Dr. Katy Phelan and child diagnosed with Phelan-McDermid Syndrome.

research

Our Vision

Phelan-McDermid Syndrome Foundation's commitment to improving the lives of individuals with the Syndrome is affirmed by our scientific and medical research support initiatives.

Our Research Initiatives

The Foundation funds grants and fellowships that target the following research priorities:

- Identification of the molecular pathophysiology of Phelan-McDermid Syndrome that can inform translational research for drug discovery
- Development and evaluation of novel treatments that can address the core symptoms and associated conditions of Phelan-McDermid Syndrome throughout the lifespan
- Development of evidence-based clinical care recommendations for Phelan-McDermid Syndrome through clinical research and clinical trials

Through initiatives that bring together the scientific community, such as Phelan-McDermid Syndrome Foundation organized symposia, we are working to raise awareness of the Syndrome in the scientific community, fostering collaboration among scientists, and helping to coordinate efforts among stakeholders.

Initiatives such as the Phelan-McDermid Syndrome International Registry and biorepositories are important ways the Foundation is helping to bring together patients, doctors, and scientist to accelerate the development of effective therapeutics for Phelan-McDermid Syndrome.




Learn more about our research, at www.pmsf.org