



**Phelan-McDermid Syndrome
F O U N D A T I O N**

Strategic Framework

2024-2027



Transforming today's barriers into tomorrow's breakthroughs

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Letter from Leadership

Dear Friends,

I am very proud to serve and partner with our families, donors, board of directors, and staff each and every day to make today better and the future brighter for everyone living with Phelan-McDermid syndrome—from the moment of diagnosis to the delivery of treatments and cures.

As the pre-eminent global organization for families affected by the rare genetic condition Phelan-McDermid syndrome, the Foundation has positioned itself to play a leading role in patient advocacy and family support and driving research breakthroughs. Approved by the Foundation’s board of directors, this new strategic plan enables us to focus the organization’s efforts toward maximizing the impact we can have on this complex syndrome.

The plan was built on a strong belief that a new vision for the Foundation should be co-created. Our families and key stakeholders provided input into a path that addresses the needs and concerns of the different segments that make up the tapestry of the Foundation. For example, early diagnosis; research; treatments; family supports; and building a networked community emerged as priorities. This inspired our four key priority areas in the strategic plan: Connect, Care, Cure, and Cultivate.

I am immensely proud of the new strategic plan that our Foundation family has co-created and believe that the Foundation is positioned to have a significant impact in benefiting the field of rare disorders and improving lives.

With respect and gratitude,



Ronni Blumenthal, CEO

Board of Directors

Denise Croden, *President*
Lisa Brown, *Vice President*
Dr. Heidi Grabenstatter, *Secretary*

Michael O’Boyle, *Treasurer*
Dr. Alycia Halladay, *Past President*
Michael G. Fraunces, *Esq.*

Chris Jauch
Eden Rivera
Cat Valcourt-Pearce

Vision

Phelan-McDermid syndrome is treatable and curable.

Mission

To make today better and the future brighter for everyone living with Phelan-McDermid syndrome—from the moment of diagnosis to the delivery of treatments and cures.



Values

- **We put families first**
We put families at the center of every decision we make and action we take, leaving no stone unturned to provide help for today and answers for tomorrow.
- **We lead with strength**
Although living with Phelan-McDermid syndrome brings extraordinary obstacles, we celebrate the challenges and rewards that having a child with PMSF brings. We believe every person with PMSF is beautiful and meaningful, and we strive to honor and elevate their value every day in every way.
- **We act with urgency**
“Someday” is too far away when you’re living or caring for someone with Phelan-McDermid syndrome. We approach every situation as creative problem-solvers, moving quickly yet responsibly to meet the needs of our community.
- **We collaborate for change**
The pace of progress is faster when we work together. That’s why we seek knowledge and input from families, scientific experts and community partners, prioritizing collaboration to deliver answers.
- **We take an all-in approach**
Because Phelan-McDermid syndrome changes everything, it requires a big-picture approach to delivering solutions. Rather than tackling single issues or symptoms, we work one-on-one with families to ensure every person and system is whole—from siblings and spouses to schools, social workers and scientists.

Strategic Priority Areas



Connect	
Care	
Cure	
Cultivate	

Programs & Services



CONNECT - Family Support Program

The Family Support program provides support and resources to help families build resilience and increase their ability to care for their child while they build a life that includes someone with significant medical and educational needs. We welcome new families to membership and provide support and resources that align with their immediate needs through our family support specialist. The Foundation offers families opportunities to receive emotional support through our focused mental health groups, access to a genetic counselor for families who have long wait times, and expert support for families experiencing neuropsychiatric crises. A network of peer supporters enables family interaction and assists in locating local resources to increase the caregivers' ability to secure services and provide support for their child or adult. Parents of children and adults with Phelan-McDermid syndrome have a wealth of knowledge about lived experience to share; our network keeps families in contact with one another to exchange ideas, practical solutions, and information about education, therapies, legal, medical issues and life experience as they face new challenges and milestones. A sense of belonging is vital to our families as it provides a connection to others, even though they may be hundreds or thousands of miles away. The Family Support program promotes interaction and provides resources in many ways including a biennial international family conference, monthly newsletters, a website, social networking and regional events.

CARE - Clinical Care Network

While securing medical care for someone with a rare condition is both critical and challenging, the Foundation programs provide the most comprehensive resources for information on clinical care for people diagnosed with Phelan-McDermid syndrome. The Foundation is supported by medical advisors who have extensive clinical experience with Phelan-McDermid patients. This body assists the Foundation by creating medical advisories with the Phelan-McDermid community and by providing guidance on complex and novel clinical questions. The Foundation is engaged with the PMSF-Neuropsychiatric Consultation Group - a body of medical experts who share experiences and guide other clinicians on best known practices in the treatment of neuropsychiatric conditions in Phelan-McDermid syndrome. Our programs facilitate family participation in many aspects of enhancing clinical knowledge of Phelan-McDermid syndrome by providing travel support for families to participate in the natural history study, alerting families and clinicians about pharmacologic guidelines, engaging families in the development of clinical care guidelines, and we establish relationships with clinics in major medical centers. Through a well-structured communication strategy comprising quick resource cards, comprehensive resources on the Foundation website, events, publications, and targeted social media outreach, we disseminate vital information to medical professionals, families, supporters, and the public, bolstering awareness and support for those living with Phelan-McDermid syndrome.

CURE - Research Program

Our research program is two-fold: we focus on identifying solutions to help families manage the many symptoms of Phelan-McDermid syndrome today while simultaneously investing in research to uncover treatments and cures for a brighter tomorrow. The program pillars include funding critical research, collecting and leveraging patient data, lowering barriers to effective clinical trials, driving pharmaceutical development, and improving symptom management. We prioritize communicating research breakthroughs in a variety of formats. The Foundation is supported by an international Scientific Advisory Committee and has developed strategic partnerships in the research and rare disease community to find ways to generate, support, and sustain the advancement of research. Central to the Research program, the Foundation maintains a robust Grants Program to direct research funds towards community interests. The Foundation also developed and maintains the largest Phelan-McDermid patient registry in the world- the PMSF DataHub. The Foundation financially supports the Phelan-McDermid syndrome Natural History Study - one of the most long-standing research projects in the syndrome. Finally, the Foundation breaks down barriers to patient participation in research by providing educational materials, amplifying the family voice in research studies, providing research opportunities to families, and providing data gathered from families to researchers.

Strategic Goals & Imperatives



Strategic Priority **Connect**

Goal 1

Strategically grow support programs and resources to respond to the needs of the PMSF community

Families need one another. The founders of PMSF started the Foundation to help families whose child had been diagnosed with the rare genetic condition, Phelan-McDermid syndrome. This remains the first priority of the organization; to connect with families from diagnosis through their life journey, creating the services and supports identified by families that strengthen our members' ability to navigate supporting their family member and gaining knowledge and skills from their peers and Foundation resources..

Strategic Imperatives:

1.1	Integrate mental health, and social and emotional wellbeing into support services
1.2	Build connections through regional and international gatherings
1.3	Expand and disseminate a library of online and print resources



Strategic Priority Care

Goal 2

Strengthen an expert clinical network

Families struggle to find care from medical professionals familiar with Phelan-McDermid syndrome. We are committed to recruiting, educating, and ensuring a robust network of physicians and clinicians (who are familiar with and educated about the unique aspects of the multidisciplinary care needs involved with Phelan-McDermid syndrome) are available to families. We continue to create and provide multimedia materials to enhance understanding and educate healthcare professionals.

Strategic Imperatives:

2.1	Share and discuss research and medical findings, exchange information and insights about the lived experience
2.2	Promote better coordination of care through evidence-based information and translate research and innovation into clinical care
2.3	Raise awareness with healthcare professionals and families to ensure patients get the right diagnosis faster and connect to the available information and resources
2.4	Build a medical expertise network to enhance the quality and coordination of care



Strategic Priority Cure

Goal 3

Facilitate research and data collection to advance treatments and cures

Families continue to hope for the future, through better symptomatic treatment, and ultimately, a cure. The successful treatments or cures for this condition will only be possible through continued participation in research and clinical trials. We leverage powerful patient data to fuel these efforts.

Strategic Imperatives:

3.1	Center patients and their families in conversations about external research studies and Foundation funding programs
3.2	Attract more clinical studies and prepare families to participate
3.3	Enable coordinated and collaborative research and data collection to facilitate the monitoring and cross cutting knowledge of rare disorders, informing care management, treatments and health system planning
3.4	Increase participation in the PMSF DataHub; market to science, research, and pharmaceutical companies



Strategic Priority **Cultivate**

Goal 4 Organizational Excellence

Families rely on us for current information, support, and help navigating the many ways life is impacted by having a family member with Phelan-McDermid syndrome. To achieve the goals of this strategic plan we will make intentional investments in our own capacity as an organization for the long-term.

Strategic Imperatives:

4.1	Grow revenue through a strategic and diversified funding model focused on financial sustainability
4.2	Create a vested community with a culture of philanthropy that generates engagement, donor loyalty, and results
4.3	Invest in robust, coordinated marketing, and communications to build a recognizable, respected brand
4.4	Optimize systems, planning, and internal process and procedures to improve operations
4.5	Manage talent as a strategic asset and ensure leadership continuity



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