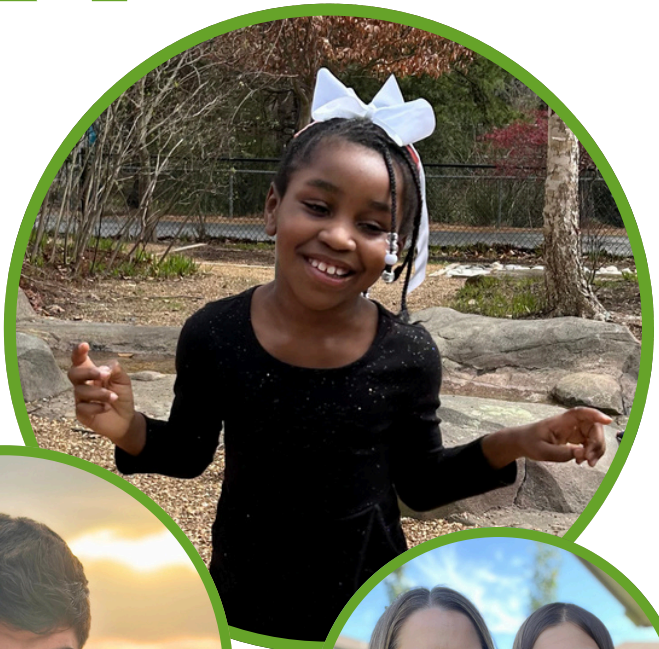


impact REPORT

20
25

our 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.



our vision

Phelan-McDermid syndrome is treatable and curable.

Letter from the CEO



Dear Friends,

As we reflect on 2025, I invite you to explore the extraordinary, measurable progress your commitment and generosity have made possible for the Phelan-McDermid syndrome community. The pages ahead share the impact you helped create, and the lives you are changing.

Because of you, PMSF advanced our mission across three core priorities: Connect. Care. Cure.

CONNECT

We deepened engagement across our community, creating meaningful opportunities for connection, education, and shared experience. By bringing families, clinicians, and researchers together, we strengthened collaboration and mutual support. For families like Dawn's, we stood by her in the first 100 days, and every day after.

CARE

We expanded access to expert clinical support through our growing referral network and strengthened resources for families navigating diagnosis, complex medical needs, and long-term care. At critical moments, families received trusted guidance from medical experts and trained peers, giving Paige and her family the clarity and confidence to navigate care.

CURE

We accelerated research through strategic grant investments, strengthened partnerships across academia and industry, and advanced critical data collection through our expansion of the Phelan-McDermid Natural History Study. Promising clinical trials and breakthroughs in gene therapy are moving the field forward. And alongside dedicated researchers, family members like the Van Weelde family bring passion and purpose to this work, helping turn hope into real progress.

These advances are made possible through community-driven fundraising efforts, partner events, and donor generosity that power every program across PMSF.

The Phelan-McDermid Syndrome Foundation was founded in 2002 by volunteers: families, scientists, and community members who wanted to take action on behalf of those living with Phelan-McDermid syndrome. Volunteers remain the engine that drives our work forward, and I warmly invite you to get involved. If you would like to learn more, please feel free to reach out to me directly at robbie@pmsf.org.



Robbie Baker

Chief Executive Officer
Phelan-McDermid Syndrome Foundation

Supporting and connecting families. Supporting and connecting families is central to our mission. A diagnosis of Phelan-McDermid syndrome can be overwhelming, but no family should face it alone. From the very beginning, PMSF surrounds families with guidance, connection, and community through programs like First 100 Days, caregiver support groups, specialized referrals, and personalized outreach. These aren't just resources; they are lifelines, offering reassurance, shared experience, and the confidence that someone is always there to help navigate what comes next.

FAMILY STORY (Caregiver Support Groups)

"The 'First 100 Days Program' is like a warm hug, offering genuine support and valuable resources for new families eager to learn more about Phelan-McDermid syndrome following diagnosis. The program provided a wealth of information and a sense of community from Day 1. It was extremely helpful to receive a walkthrough of the Phelan-McDermid Syndrome Foundation website, and I find myself visiting this resource any time I have a question or want more information about important news, upcoming research studies, or other general guidance. The standout feature of the program, however, was meeting the Director of Family Support, Carla.

I cannot say enough positive things about Carla; she is welcoming, shares extensive knowledge about PMSF, and put me at ease from the very first time we spoke. Being able to connect with other families, learn about the diagnosis, and ask questions of someone who has lived this experience is truly an invaluable offering that I would encourage all families to take advantage of as soon as they are ready! I know my first 100 days of settling into our life after diagnosis was filled with a number of questions and yet many more answers given the individualized touch this program offered. Thank you, PMSF, for making this resource available to newcomer families such as ours. We are grateful."

- Dawn Moschberger
Daughter - Leah, age 10



2025 CONNECT IMPACT

PMSF welcomed nearly **300 new families** and provided **2,400+ connections** through our family support programs.

- **First 100 Days**
- **Caregiver Support Groups**
- **Pathways Education Series**
- **1:1 Outreach & Support**
- **Regional REPs**
- **Global Partnerships**

Engagement totals reflect participation across programs; individuals may engage in multiple offerings.

Improving medical care. We are improving medical care by educating and activating the medical community to ensure every person with Phelan-McDermid syndrome receives an accurate diagnosis, compassionate care, and coordinated treatment. We built critical connections to launch the first Phelan-McDermid Clinical Center and Healthcare Provider Resource, developed with clinical and research partners and shaped by our community. Because care extends beyond a single appointment, PMSF equips families and clinicians with expert guidance, clinical resources, and real-time support to strengthen decision-making, especially in complex or urgent situations.

FAMILY STORY (Clinical Care Center Partnership)

“After our son was diagnosed with autism and global developmental delay at age 2 and was not making progress in his therapies, we kept wondering if there was something more we could do. Six months later, we received his genetic results showing a SHANK3 Class I deletion. We immediately started researching and found the list of Phelan-McDermid Syndrome Clinics on the PMSF website. We were thrilled to see that Cincinnati Children’s was within driving distance for us.

Going into the appointment, we didn’t have many expectations since most of Walker’s providers had little experience with Phelan-McDermid syndrome. But Dr. Shillington made us feel welcome from the moment we arrived. She spent nearly an hour listening to Walker’s story and truly took the time to understand him.

My husband and I left the appointment feeling incredibly grateful. It was so worth the trip. We’re already planning our next visit, where we’ll meet additional specialists through their multidisciplinary clinic, a true ‘one-stop shop.’ We hope to return each year for follow-up care.

I’m so grateful that this clinic exists and that we were able to find it through the PMSF website.”

- Paige Montgomery
Son - Walker, age 3



2025 CARE IMPACT 3,600+ clinical care resources accessed in 2025.

- Physician consultation cases through Phelan-McDermid syndrome-Neuropsychiatric Consultation Group
- Genetic counseling referrals
- Neuropsychiatric consultation referrals
- Educational views through Pathways
- Phelan-McDermid syndrome clinics added to the clinical referral network
- Clinical documents downloaded
- Website visits accessing care resources
- Quick resource cards

Totals reflect engagement across multiple care resources.

Driving research breakthroughs. We are accelerating the path to treatments and cures by aligning families, researchers, and industry. Families are at the heart of this work, helping build a more informed and connected community.

PMSF advances research through our DataHub registry, the Phelan-McDermid Natural History Study, and our grants program, while partnering with industry to expand clinical trial access and readiness. We don't just fund research, we connect it, shape it, and move it forward faster. By empowering families to participate, we ensure research reflects their lived experience and leads to meaningful, real-world impact.

PRIMARY INVESTIGATOR STORY (Natural History Study Funding)

"The Natural History Study (NHS) has been critical to advancing our understanding of Phelan-McDermid syndrome. The insights gained from this work have directly informed updates to the clinical consensus guidelines. Participation from families across the country has been essential, allowing us to study a diverse group of individuals and build a more complete picture of the syndrome. This work would not be possible without the financial support of the Phelan-McDermid Syndrome Foundation, which enables us to reach and learn from so many families."

- Siddharth Srivastava, MD | Boston Children's Hospital

FAMILY STORY

"People often say you learn who your true support team is during difficult times, and we are so grateful for those who have supported and guided us on this journey. We've felt that our son's story matters, not only to our family, but also to the research community. Knowing that his experiences may help advance research through the Natural History Study, improve care for other families, and provide a story others can relate to means so much to us.

We are especially thankful to Dr. Sid and the PMSF team for sharing their knowledge with our local medical team, service providers, and school district, helping bring greater understanding of Phelan-McDermid syndrome and moving away from a 'one size fits all' approach."

- The Van Weelde family (Asher)

2025 CURE IMPACT **850+** research contributions accelerating Phelan-McDermid syndrome science.

- Strengthened a key research asset by completing **429 DataHub patient records**
- Administered a competitive grant program, with **100% increase in program interest**
- Invested **\$100K to advance neuropsych biomarker research**
- Enabled **Natural History Study participation through \$20K in travel stipends and \$50K in program support**
- Expanded **global research awareness, reaching audiences in 30 countries**

Totals reflect engagement across multiple research initiatives.

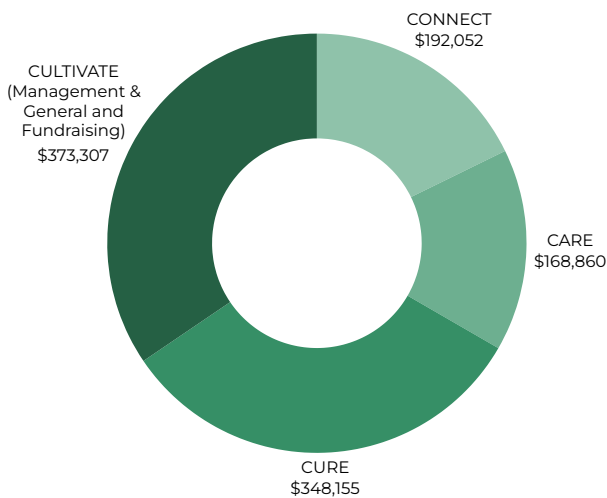
You fuel progress... and the mission

The progress highlighted throughout this report is only possible because of the dedicated community of donors, families, partners, and fundraisers who believe in a better future for individuals with Phelan-McDermid syndrome. Backed by our experienced staff and more than 20 years of established infrastructure, donor support powers every program across PMSF.

We are committed to responsible stewardship by ensuring every dollar is used strategically to advance care, strengthen connections, and accelerate research toward effective treatments.

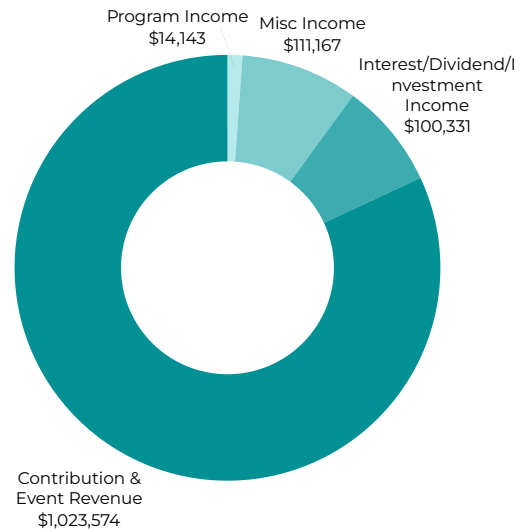
EXPENSES

Total Expenses: \$1,082,374



REVENUE

Total Revenue: \$1,249,215



Ending Net Assets \$2,066,185

PMSF is a tax-exempt, nonprofit organization pursuant to Section 501(c)(3) of the Internal Revenue Code with Federal Tax ID# 04-3673104. All gifts and donations are tax deductible in accordance with IRS regulations.



PMSF demonstrates excellence in governance, accountability, and transparency by earning the highest ratings on the following industry standards:



“2025 was a year of tangible growth and meaningful milestones, bringing us closer to effective treatments and, ultimately, a cure.”

— Denise Croden, Board President

Looking ahead: building what comes next

The momentum of 2025 has positioned PMSF for an even stronger year ahead. As we enter 2026, our focus remains clear: strengthening community, improving care, and accelerating the research that will lead to treatments and cures for Phelan-McDermid syndrome.

Guided by our expert team, together with families, clinicians, researchers, and partners around the world, we are advancing systems, knowledge, and collaborations needed to move the field forward.

CONNECT — 2026

Bringing the community together and strengthening support for families at every stage.

- Host the largest global gathering of the Phelan-McDermid syndrome community at the 2026 PMSF Family Conference
- Expand caregiver programming and peer connection opportunities
- Strengthen regional and international family engagement

CARE — 2026

Improving access to knowledgeable clinicians and practical tools that support better medical care.

- Develop additional clinical tools for emergency and hospital settings
- Expand physician education and clinical resource adoption
- Grow the network of Phelan-McDermid Syndrome Clinics
- Expand opportunities for family engagement and connection
- Elevate the lived experience of the Phelan-McDermid syndrome community

CURE — 2026

Accelerating the research ecosystem that will lead to treatments and clinical trials.

- Fund new translational research through the next PMSF grant cycle
- Strengthen partnerships with academic and industry collaborators
- Increase family engagement in research design and priorities
- Enhance the quality and readiness of data shared through the PMS DataHub

The progress ahead depends on a connected and empowered community.

Every program, every research initiative, and every partnership described in this report exists because families living with Phelan-McDermid syndrome continue to show extraordinary resilience, generosity, and commitment to moving the field forward.

With the continued support of families, donors, clinicians, and partners, we are building a future where every person with Phelan-McDermid syndrome has access to expert care, meaningful support, and effective treatments.

about us



The Phelan-McDermid Syndrome Foundation is the largest and most trusted nonprofit working 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.

Phelan-McDermid syndrome is a rare genetic condition that can compromise many critical functions in a person's body, from learning and communicating to eating and sleeping. Started by families who understand both the beauty and barriers of having a child with this life-altering condition, the Foundation is doing everything it takes to support and connect families, improve medical care, and drive research breakthroughs to deliver treatments and cures faster.

Powered by the collective strength of families, scientific experts and community partners, we'll achieve a world where Phelan-McDermid syndrome is treatable and curable, and every person and family affected are thriving.

Learn more and join us at pmsf.org.

our vision

Phelan-McDermid syndrome is treatable and curable, and every person and family affected are thriving.



Follow us on social media!



Check out our website!



Contact us

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PMSF's progress is made possible by dedicated volunteers, board members, clinicians, researchers, and families who give their time and expertise to advance our mission.