ANNUAL REPORT

2025

CURE MFM 13

Uniting against rare challenges

Contents

What is Cure MFM13 and MFM13?

Cure MFM13 is the only charitable project dedicated to overcoming the challenges posed by Myofibrillar Myopathy type 13 with Rimmed Vacuoles (MFM13). We have brought together a multidisciplinary team of patients and families, advocates, researchers and healthcare professionals to treat and eventually to cure this debilitating condition.

MFM13 is an ultra-rare, adult-onset muscle-wasting condition that progresses slowly. It is inherited in an autosomal dominant manner and belongs to the broader group of Myofibrillar Myopathies (MFMs). MFM13 was first described by Ghaoui et al. in 2016, and its molecular mechanism was further explored by Tedesco et al. in 2023 and 2025. The disease is caused by frameshift mutations in the *HSPB8* gene, resulting in a toxic gain of function, with around 60 cases reported worldwide.

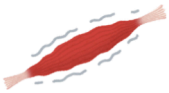
The symptoms of MFM13 vary between individuals but generally involve progressive muscle-related issues, often beginning between adolescence and the 40s, with rare cases in childhood (Yang et al., 2024). Common features include weakness and wasting of skeletal muscles, which can begin either proximally or distally. This often leads to difficulty walking, getting up from a chair, or raising arms. Symptoms usually progress slowly, although the rate of progression varies among patients.

MUSCLE ATROPHY

loss of strength due to degeneration

FATIGUE

general tiredness caused by muscle inneficiency and overcompensation



MUSCLE WEAKNESS

which can start proximally or distally

SLOW PROGRESSION

symptoms worsen gradually and vary greatly between individuals

HSPB8 stands for heat shock protein beta-8 and is essential for cellular stress response. A molecular multitasker, HSPB8 maintains protein stability and protects proteins during stress. HSPB8 is expressed across several tissues but is especially abundant in skeletal muscle, neurons, brain, and heart. Its role is especially prominent during cellular stress, ensuring vital functions remain intact.

Cure MFM13

Our mission is to improve the lives of all people affected by MFM13 and their families. We do this by accelerating the drug development process, building a strong and empowered community, and advocating for the community.

Our vision is life free of MFM13 and all its burden.

Our purpose is to alleviate suffering and empower the patients.

Our strategy is to invest in R&D, support MFM13 families, advocate for the community and grow as an organization.

Our values are urgency, transparency, evidence-based.

What differs us: we are the only charitable project dedicated to addressing challenges of MFM13.

Our goal is to find a treatment and a cure using state-of-the-art technologies and advances in science.

Words of Founder

Dear Friends, Supporters, and Fellow Travelers,

As we look back on 2025, I'm struck less by any single achievement and more by something quieter and more powerful: continuity. Cure MFM13 exists because people refused to let curiosity fade, refused to let ultra rare mean invisible, and refused to accept that slow and steady progress is the same as no progress.

This year was about laying deeper foundations. We asked better questions and dug deeper. We strengthened relationships with researchers and clinicians who share a long-term view, knowing that meaningful breakthroughs are rarely sudden, but often cumulative. And through our team's diligence, we achieved an important milestone: securing an official name for the disease, prompting our transition from Cure HSPB8 to Cure MFM13. Names matter. They give shape to understanding, legitimacy to inquiry, and visibility to people who might otherwise remain unseen.

At Cure MFM13, we believe that hope is not passive. It is built through shared effort, careful science, and a willingness to invest in understanding—both the biology of MFM13 and the full community of people affected by it—before outcomes are obvious. An essential part of that work is actively finding and connecting with every person living with this condition, so that no one remains undiagnosed, unsupported, or isolated by rarity. Every conversation, every collaboration, and every contribution moves the work forward, even when the finish line is not yet in sight.

To everyone who stood with us this year: thank you. Your trust and engagement make this work possible. We remain committed to advancing research with rigor, transparency, and urgency, and to supporting a future where MFM13 is better understood, better treated, and ultimately overcome.

With gratitude and resolve,

Todd King
Founder & President, Cure MFM13

The Team

Todd King

Founder

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Program Director

Sylwia Szwec

Research Program Manager

Matt McLeod

Scientific Advisor

Dr. Karolina Chwalek

Strategic Advisor

Khosiyat Makhmudova

Outreach Manager

Julia Mielcarz

Brand Manager

Vania Fortes

Bookkeeper

Accomplishments of 2025

Since its founding under the auspices of Social and Environmental Entrepreneurs in April 2024, Cure MFM13 continued its rapid growth and impact in 2025, building on the progress made in its inaugural year. This year we made tangible advances in research, advocacy, and community engagement, reflecting our commitment to patients and the broader rare disease field.

Research and Development

Research efforts have largely focused on expanding the models and resources needed to understand the biology of MFM13 and to build a solid foundation for translational research.

- **Supporting fundamental research:** We awarded funding for Professor Angelo Poletti and Assistant Professor Barbara Tedesco at the University of Milan in Italy, for a project: Modelling HSPB8 pathology: assessing the molecular mechanisms regulating the RQC/PQC and HSPB8 axis and HSPB8 mutant dynamics. Understanding the mechanisms of MFM13 is crucial to developing future effective treatments.

- **Cell models:** We finalized the development of induced pluripotent stem cells (iPSCs) derived from a MFM13 patient carrying *HSPB8* frameshift mutations, along with matched isogenic control lines. Following molecular validation, these models will be shared with the research community to accelerate scientific discovery. IPSCs provide a valuable addition to cell models already available for the research community in Coriell Repository.

- **Mouse Model:** In collaboration with the International Institute of Molecular and Cell Biology (IIMCB) in Poland, we initiated the generation of the first partially humanized MFM13 mouse model - a major milestone for the field. Initial results are eagerly awaited, with detailed phenotypic characterization studies planned to start in 2026.

- **Antibody development:** In partnership with XYZ Laboratory we have developed a mutation-specific HSPB8 antibody targeting the c.515dupC | p.P173Sfs*43 frameshift variant, a tool critical for studying disease-specific protein behavior and toxic gain-of-function mechanisms. We aim to make the antibody openly available for scientific use, prioritizing access for researchers worldwide.

Advocacy and Community:

- **Disease classification:** Securing an OMIM number (#621078) and an official disease name for the condition caused by HSPB8 frameshift mutations significantly increased recognition and visibility for MFM13.

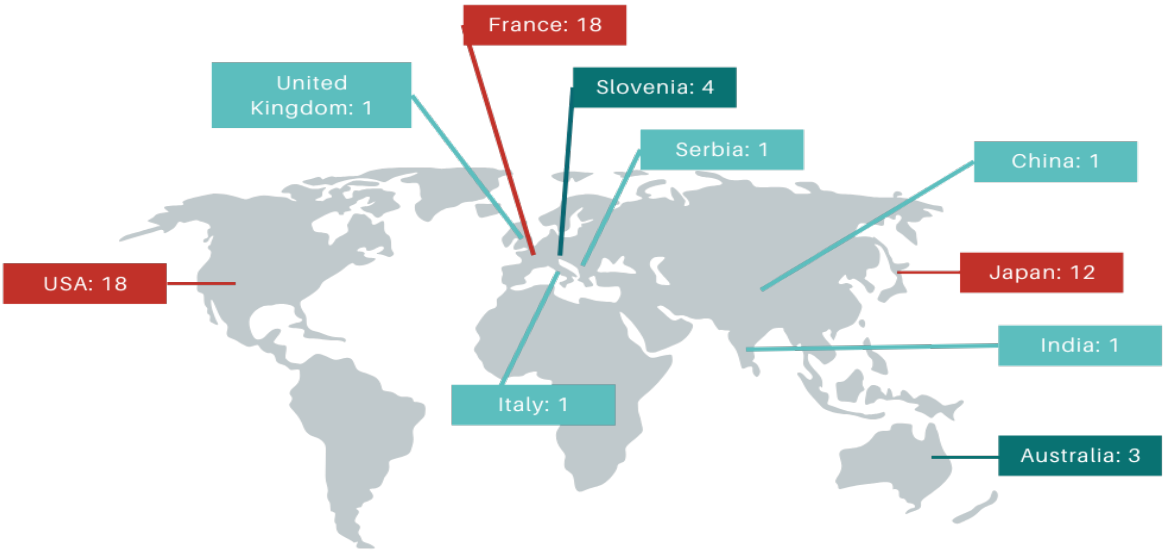
OMIM #621078

- **Rebranding:** To reflect this formal classification and improve clarity, we rebranded from Cure HSPB8 to Cure MFM13.

CURE HSPB8

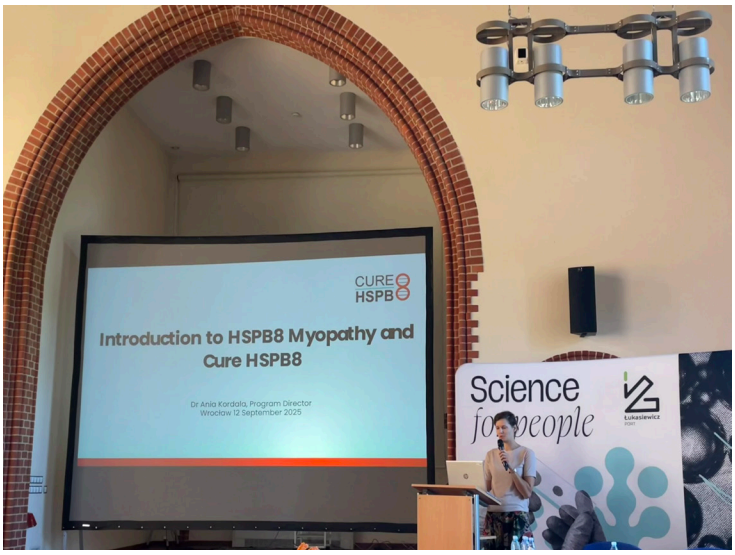
CURE MFM13

- **New Patients:** In 2025, we learned about several new individuals affected by MFM13, who now have access to our growing knowledge base, resources, and community.



- **Regular Cure MFM13 Research Meetings** grew, strengthened and continued to bring together patients and researchers to share the latest findings and foster collaborations in MFM and HSPB8 research. With 30 regular attendees, among others, we discussed ways to manipulate HSPB8 with small molecules and ASOs, explored the role of protein quality control in MFM13 pathology, and learned about possibilities of using 2D and 3D cell systems for distal Hereditary Motor Neuopathy (dHMN)/Myopathy.

- **Building network through conference attendance:** We strengthened our presence in rare disease and myopathy communities through participation in several scientific and patient-focused conferences. We presented posters at World Orphan Drug Congress (WODC) in Boston in April, and at WODC Europe in Amsterdam, in October. We were Keynote Speakers at Genomica Conference of Genetics, Cracow, Poland.



- **Spreading the word**: Our Founder, Todd King, and Director, Anna Kordala, were featured in RAREatives in the article "CureMFM13: How a Rare Disease Diagnosis Built a Community," sharing the story of how patient-driven research can unite and empower families.

- **Podcast**: We have launched a podcast focused on MFM13, discussing disease mechanisms, recent publications, and case studies to help in knowledge dissemination and raising awareness.

YouTube
Spotify
iHeart
ApplePodcasts

- **MFM13 Library:** We launched a curated scientific library that consolidates key publications on MFM13 molecular mechanisms, the CASA complex, autophagy-related processes, and therapeutic approaches, making complex information more accessible.

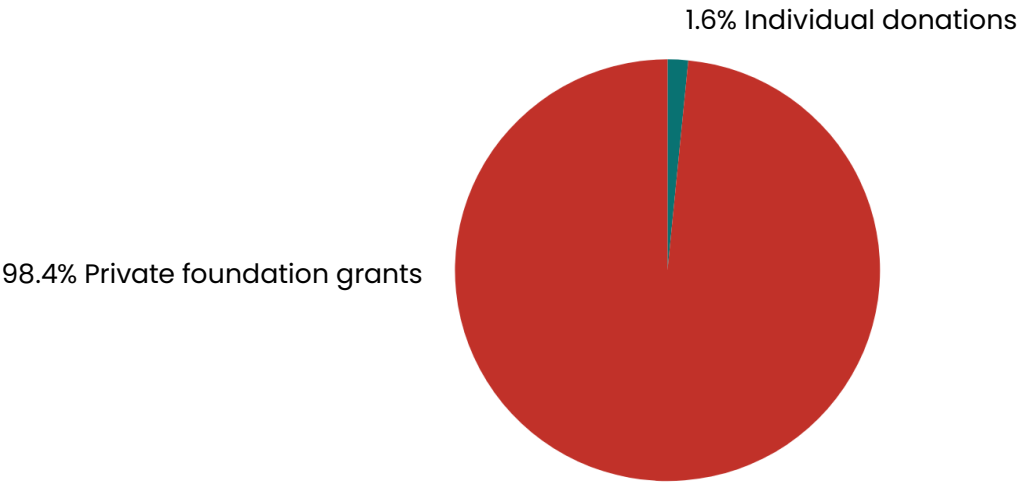
Priorities for 2026

Last year was full of important achievements in awareness, advocacy, and research development, but it also highlighted ongoing challenges faced by the MFM13 community. We are still advocating for *HSPB8* to be included in more myopathy panels, as it is currently mostly available only in neuropathy panels, which severely delays accurate diagnosis for MFM13 patients.
At the same time, we are continuing our efforts to identify more patients, develop reliable biomarkers, and to lay the groundwork for translational research.

Financial summary

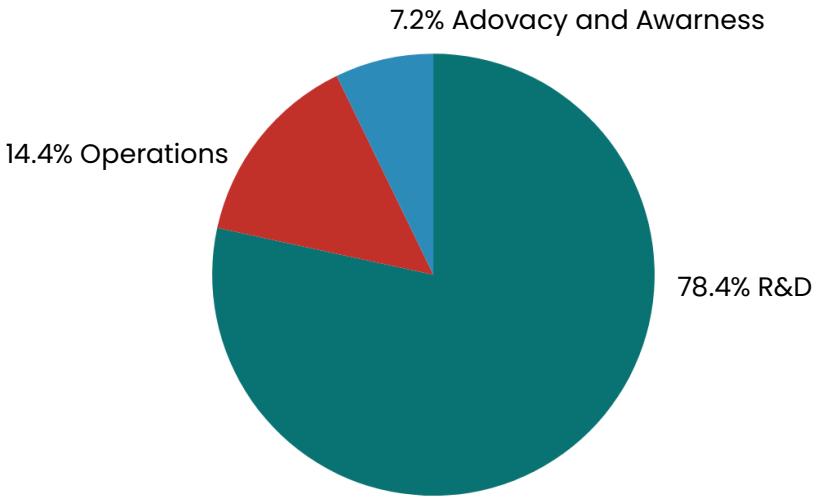
Total income: $380 950

- Private foundation grants: $375 000
- Individual donations: $5 950



Total expenses: $417 797

- R&D: $308 132
- Operations: $56 636
- Advocacy & awareness: $28 306



Connect with us

Thank you for reading and for your support of the MFM13 community! Stay connected by subscribing to our newsletter at curemfm13.org. Follow us on LinkedIn, X, and Facebook for the latest news, updates, and ways to get involved.

- Visit our website: curemfm13.org
- Contact us: ania@curemfm13.org
- Make sure you sign up for our newsletter
- Follow us: @curemfm13
- Follow us: https://www.linkedin.com/company/cure-mfm13
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