Community Activation Roadmap

Achieving accelerated and accurate genetic diagnosis for people with mitochondrial disease

March 2025

Partners

Association contre les maladies mitochondriales (AMMi), International Mito Patients (IMP), MitoAction, MitoCon, Lily Foundation, The Jeremiah Gracen TK2d Foundation, UMDF and UCB

Our ambition is to empower the mitochondrial disease (Mito) advocacy community to improve early access to accurate genetic testing for every member of the Mito community.

We have developed this tool to inspire community organizations, large and small, who support people with mitochondrial diseases to **take action now** to improve the current challenges surrounding genetic diagnosis. We know that an accurate and accessible genetic mitochondrial disease test, and ultimately a qualified and clearly understood diagnosis, is the door that remains closed before other doors can open.[1] So we designed this roadmap in a way that all of the community can take action through small changes and consider new paradigms and collaborations that will impact the future.

Why?

Advancing the diagnostic journey will:

- Illuminate pathways to specialised, personalized care and targeted treatments.
- Help guide families by providing valuable information about health and genetic risk factors.
- Improve clinical care by delivering more meaningful interventions for Mito patients now and in the future.
- Enable the advocacy community to reach the right people with the right support.
- Improve the quality of life for Mito patients and their loved ones, allowing them to live the lives they want.

Our Vision

By working together, we can achieve a future where every member of the Mito community has access to genetic testing and diagnosis as a fundamental standard of care.

We aspire to create a world where genetic diagnosis in mitochondrial and rare diseases is routine, eliminating the need to advocate and fight for it.

By supporting and connecting with each other, we can expand our reach and enhance our voices to support patients far and wide.

The Activation Roadmap

The following roadmap has been shaped through the collaboration of **patient advocates, patient experts, and patient organizations**, who provide insights for the broader advocacy community to address barriers in genetic testing and inspire change for the future.

Barriers

Three fundamental barriers need to be overcome to improve genetic testing:

1

Low clinician knowledge of mitochondrial diseases such as TK2d and pathways to genetic testing[2]

Healthcare professionals have insufficient training in diagnosing mitochondrial diseases, limited exposure to rare Mito patients, limited experience in navigating the complex diagnostic landscape, and limited coordination between specialists. So, they often have low knowledge of which tests to order, and when and how to order them.

2

Insufficient patient education on testing pathways and results[3]

Patients have low disease awareness and appreciation of the importance of genetic testing pathways. They often receive inadequate support in navigating the complex diagnostic process and have limited assistance in interpreting complicated test results, understanding their implications, or knowing the potential next steps to take.

3

Heterogenous access to/ funding for genetic testing and genetic counsellors[4]

Access to and funding for genetic testing and counselling is highly variable and scarce across many regions. High costs and inconsistent insurance coverage, particularly in the US, limit access. Ordering genetic tests can be time-consuming and can require consensus in Europe. In some countries, such as the UK, detailed testing is often reliant on research studies or public systems.

Activities

The following priority activities were identified for activation by the advocacy community:

	Low clinician knowledge of TK2d and pathway to genetic testing	Insufficient patient education on testing pathways and results	Heterogenous access to/ funding for genetic testing and genetic counsellors
Short term	 **Clinician awareness programs and training support** • Develop and launch an educational campaign. • Convert diagnostic guidelines for mitochondrial disease into a simple resource. • Develop and launch training opportunities to upskill genetic counsellors. • Support the training of the next generation of newly qualified healthcare professionals.	 **Patient awareness and education resources** • Increase local community-based educational events, such as webinars or videos, and genetic testing awareness campaigns. • Develop a comprehensive brochure and one-page factsheet on the 'what/ when/how' of genetic testing. • Help patients and families make sense of diagnoses that include genetic variants of unknown significance.	 **One-to-one navigation guide** • Support patients at the point of diagnosis by providing a first point of contact representative to act as a navigation guide.
Medium term	 **Rare disease collaboration and standardization** • Advocate for and influence policies that will cement genetic testing as a standard of care, especially for newborns. • Develop and finalize clinical care guidelines through collaboration and consensus. • Develop and launch rare disease training programs for HCPs, covering genetic testing for multiple conditions.	 **Enhanced patient support at point of diagnosis** • Support the provision of patient community experts to assist healthcare professionals in delivering diagnoses and providing support. • Create and share animations to explain more complex genetic concepts so patients and caregivers are empowered to ask questions rather than be intimidated by the science of genetics. • Implement a "train the trainer" program, where the patient community can educate HCPs/genetic counsellors.	 **Fund improvements in genetic counselling** • Fund genetic testing services and help employ more genetic counsellors for rare diseases.
Long term	 **Plan for the application of technology and AI advances** • Explore and implement AI applications for patient identification through symptom matching and diagnostic suggestions. • Identify, support or develop grant programs to support newborn testing research programs.	 **Raise public interest in mitochondrial health** • Reframe mitochondrial disease as mitochondrial health to emphasize its importance to everyone. • Launch a large-scale "Think Mito" campaign to raise awareness of the most common metabolic disorders. • Increase visibility of Mito diseases through talk shows, TED Talks, YouTube, and TV shows and unite in the call to action that genetic diagnosis must be the new standard.	 **Push for rare disease genetic testing as standard of care** • Establish a rare disease umbrella organization to connect and support various stakeholders where improving genetic diagnosis and access to clinical trials are key aims, among others. • Work together to advocate for genetic testing as standard of care, e.g., by conducting a multi-stakeholder study to provide insights for cost analysis and value assessments, to further emphasise the need for testing.

What you can do

As we continue our mission and strive for impactful change in genetic diagnosis, we encourage all those who play a role in the Mito patient community to support in the delivery of this roadmap. Which of these activities can you deliver as an individual, as an organisation or as part of a collaboration with other advocates?

Join us so that we can unite our voices, share our efforts, and amplify our resources to tackle barriers and improve access to genetic testing for all.

Together, we can make rapid, accurate and seamless genetic testing an integral part of standard care, offering hope and options to mitochondrial disease patients and families.

For further information, resources or to discuss this further, please contact: The International Mito Patients Organization at info@mitopatients.org.

Should you need assistance in your local language, please reach out to your regional association.

References: 1. Kassardjian CD, Amato AA, Boon AJ, et al. The utility of genetic testing in neuromuscular disease: A consensus statement from the AANEM on the clinical utility of genetic testing in diagnosis of neuromuscular disease. Muscle Nerve. 2016;54(6):1007-9. **2.** Grier J, Hirano M, Karaa A, et al. Diagnostic odyssey of patients with mitochondrial disease: Results of a survey. Neurol Genet. 2018;4(2):e230. **3.** Delikurt T, Williamson GR, Anastasiadou V, et al. A systematic review of factors that act as barriers to patient referral to genetic services. Eur J Hum Genet. 2015;23(6):739-45. **4.** Gatto EM, Walker RH, Gonzalez C, Cet al. Worldwide barriers to genetic testing for movement disorders. Eur J Neurol. 2021;28(6):1901-9.

This Community Activation Roadmap has been funded and supported by UCB. The content was co-created during a series of workshops by a patient steering committee. Patient organizations were compensated at fair market value for their time and contributions.

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