CURE MITO FOUNDATION

Dear Friends, Partners, and Followers of the Cure Mito Foundation,

We are thrilled to share with you that our paper: **"Leigh Syndrome Global Patient Registry: Uniting Patients and Researchers Worldwide"** is published in the Orphanet Journal of Rare Diseases!

This has been a truly patient driven effort with Cure Mito Foundation leading this project including doing data analysis and building a diverse and collaborative team of 11 authors who worked together on the paper! We would like to thank each one of the authors who brought their talents, time, and dedication to this project.

To our knowledge this is the first paper about a global patient registry for Leigh syndrome, with many of the reported outcomes reported for the first time. It may also be a first truly international registry in mitochondrial disease with 70% of participants residing outside the United States.

Please note that data analyzed in the paper includes only the first 7 months since our registry was live (September 2021-April 2022). Even at that that time, the amount of information and insights gained from the registry was enormous. Now, nearly 300 participants are enrolled, there's a lot more data and much more is coming up, including an additional paper about data interoperability with CDISC standards which is still undergoing peer review.

Thank you to many of our partners who have supported and encouraged us along the way, and to those medical doctors who shared our registry with their patients.

Special thank you to Dr. Matthew Klein, MD, CEO of PTC Therapeutics for his invaluable input into the design of Leigh syndrome survey.

Finally, thank you to all the patient families who enrolled, took time to share their information, and helped share the registry with their doctors and other families. This registry and paper would not have happened without them.

Plese read the full paper HERE
Hear updates on this and more at Empower and Inspire: 2nd Annual Leigh Syndrome Symposium

The Cure Mito team

"They didn't know it was impossible so they did it."
- Mark Twain

"Cure Mito Foundation's Leigh syndrome global patient registry is foundational – it will catalyze basic research while also helping to get medicines approved by the FDA and other regulatory agencies."

-Dr. Vamsi Mootha, MD

Harvard Medical School, Massachusetts General Hospital, Broad Institute

RESEARCH Open Access

Leigh syndrome global patient registry: uniting patients and researchers worldwide

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Abstract

Background Leigh Syndrome (LS) is a rare genetic neurometabolic disorder, that leads to the degeneration of the central nervous system and subsequently, early death. LS can be caused by over 80 mutations in mitochondrial or nuclear DNA. Patient registries are important for many reasons, such as studying the natural history of the disease, improving the quality of care, and understanding the healthcare burden. For rare diseases, patient registries are significantly important as patient numbers are small, and funding is limited. Cure Mito Foundation started a global patient registry for LS in September 2021 to identify and learn about the LS patient population, facilitate clinical trial recruitment, and unite international patients and researchers. Priorities were to allow researchers and industry partners to access data at no cost through a clear and transparent process, active patient engagement, and sharing of results back to the community.

Results Patient registry platform, survey design, data analysis process, and patient recruitment strategies are described. Reported results include demographics, diagnostic information, symptom history, loss of milestones, disease management, healthcare utilization, quality of life, and caregiver burden for 116 participants. Results show a high disease burden, but a relatively short time to diagnosis. Despite the challenges faced by families impacted by Leigh syndrome, participants, in general, are described as having a good quality of life and caregivers are overall resilient, while also reporting a significant amount of stress.

Conclusion This registry provides a straightforward, no-cost mechanism for data sharing and contacting patients for clinical trials or research participation, which is important given the recruitment challenges for clinical trials for rare diseases. This is the first publication to present results from a global patient registry for Leigh Syndrome, with details on a variety of patient-specific and caregiver outcomes reported for the first time. Additionally, this registry is the first for any mitochondrial disease with nearly 70% of participants residing outside of the United States. Future efforts include continued publication of results and further collaboration with patients, industry partners, and researchers.

Keywords Leigh syndrome, Leigh disease, Mitochondrial disease, Patient registry, Patient driven, Real world data, Clinical trials, Rare disease, Research, Hope

Cure Mito Foundation is a 501(c)(3) nonprofit organization led by parents who volunteer their time to search for a cure. 100% of your donations are tax-deductible and will go directly to research dedicated to mitochondrial diseases.

Your donation matters!

For many ways to give please visit:
https://www.curemito.org/ways-to-give

Many volunteer opportunities are also available!

Skills and expertise currently needed: fundraising, marketing, writing (scientific writing, grant writing, blog writing). Please contact us at info@curemito.org to learn more.

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