Leigh Syndrome Global Patient Registry

CURE MITO FOUNDATION

About Leigh Syndrome

Leigh syndrome is a most common pediatric mitochondrial disease. It is a neurodegenerative disease causing the loss of abilities to walk, talk, swallow. Leigh syndrome can be caused by 110+ nuclear and mitochondrial DNA mutations. Approximately 1 in 40,000 are affected.

About Cure Mito

We are a leading patient advocacy organization dedicated to advancing research of Leigh syndrome and empowering and supporting affected families worldwide. Our mission is to unite the global Leigh syndrome community to accelerate patient-centered research, treatments, and cures.

Registry Collaborations

SANFORD RESEARCH

SUMPTUOUS DATA SCIENCES

RDCA-DAP Rare Disease Cures Accelerator Data and Analytics Platform

CRITICAL PATH INSTITUTE

HOPE FOR PDCD RESEARCH FOR A CURE

Registry Milestones

September 2021 - Registry launched in collaboration with Sanford CoRDS

November 2021 - Cure Mito collaborates with C-Path on data sharing into C-Path's RDCA-DAP® platform

May 2022 - Cure Mito collaborates with Sumptuous Data Sciences on data interoperability to CDISC

February 2023 - FDA listening session about the registry

July 2023 - Leigh syndrome registry data is shared with C-Path

September 2023 - First peer-reviewed paper published in Orphanet Journal of Rare Diseases

January 2024 - Cure Mito launches Mitochondrial and Metabolic disease taskforce with C-Path and seven other stakeholders

January 2024 - Second peer-reviewed paper published in Journal for the Society of Clinical Data Management

Feburary 2024 - Cure Mito collaborates with Hope for PDCD foundation to launch PDCD registry on the same platform in alignment with Leigh syndrome registry

2021-2024 - 4 posters presented at multiple conferences

Registry Highlights

300+ participants

37+ countries represented

5 y.o. median age at joining registry

57% nuclear DNA mutation

33% mitochondrial DNA mutation

60% diagnosed within 1 year of symptom start

2 y.o. median age at diagnosis

95% genetic testing used to confirm diagnosis

To learn how this Leigh syndrome registry can support your clinical trial or research study, please contact us at info@curemito.org

ClinicalTrials.gov (NCT01793168)
curemito.org/leighsyndromeregistry

Check out our published papers!

Leigh Syndrome Global Patient Registry: Uniting Patients and Researchers Worldwide

DOI:10.1186/s13023-023-02886-0

Interoperability of Leigh Syndrome Patient Registry Data with Regulatory Submission Standards

DOI:10.47912/jscdm.244