



# Leigh Syndrome Global Patient Registry

## 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 Milestones

- Global Leigh syndrome registry launched in collaboration with **Sanford CoRDS** in 2021
- Registry data integrated into **C-Path's RDCA-DAP® platform**
- Data interoperability completed across raw data, **CDISC, and OMOP** formats
- **Three peer-reviewed publications** published and one additional manuscript under review
- Research presented at multiple **international conferences**
- Two **FDA listening sessions** held discussing the registry
- **Mitochondrial & Metabolic Disease Taskforce** launched with C-Path (20 participating organizations)
- Collaboration expanded to support **additional rare disease registries**, including PDCD



## Registry Highlights

**450+**

participants

**48**

countries

**63%**

nuclear DNA mutation

**37%**

mitochondrial DNA mutation

**62%**

diagnosed within 1 year of symptom start

**2 y.o.**

median age at diagnosis

**5 y.o.**

median age at joining registry

**ClinicalTrials.gov (NCT01793168)**  
**curemito.org/leighsyndromeregistry**

**To learn more and collaborate, please contact us at [info@curemito.org](mailto:info@curemito.org)**

## Check out our published papers!

Leigh Syndrome Global Patient Registry: Uniting Patients and Researchers Worldwide

DOI:[10.1186/s13023-023-02886-0](https://doi.org/10.1186/s13023-023-02886-0)

Interoperability of Leigh Syndrome Patient Registry Data with Regulatory Submission Standards

DOI:[10.47912/jscdm.244](https://doi.org/10.47912/jscdm.244)

Expanding research and care for Leigh syndrome: efforts of a patient-led advocacy organization

DOI:[10.1186/s40900-025-00808-x](https://doi.org/10.1186/s40900-025-00808-x)