Sophia Zilber, MA [1]
Kasey Woleben [1]
Danielle Boyce,DPA,MPH [1,2]
Kevin Freiert, MBA [1]
Souad Messahel,PhD [3]
Joana Matos, PhD [1]
Courtney Boggs, RN [1]
Saima Kayani, MD [4]

1 - Cure Mito Foundation
2 - Department of Neurology, Johns Hopkins School of Medicine
3 - Peter O'Donnell Jr. Brain Institute, University of Texas Southwestern Medical Center
4 - University of Texas Southwestern Medical Center

LEIGH SYNDROME GLOBAL PATIENT REGISTRY

CURE MITO FOUNDATION

Corresponding authors:
sophia@curemito.org
kasey@curemito.org

INTRODUCTION

Leigh syndrome (LS) is a rare genetic neurometabolic disorder, leading to the degeneration of the central nervous system and death. The onset of symptoms usually occurs between the ages of three months and two years, but some patients have later onset of symptoms. LS can be caused by a number of mutations in mitochondrial or nuclear DNA.

Cure Mito is a parent-led, volunteer-run foundation dedicated to advancing education and research for Leigh syndrome and mitochondrial disease.

OBJECTIVES

Cure Mito Leigh Syndrome registry was started in September, 2021 to meet the following goals:

- Internationally available
- Identify and collect comprehensive information about LS patients population
- Facilitate clinical trials recruitment
- Share results and findings on an ongoing basis
- Build stronger patients and researchers community

METHODS

Cure Mito Leigh Syndrome Global registry was started in partnership with Coordination of Rare Diseases at Sanford (CoRDS). CoRDS is a disease agnostic platform with data for 1,901 rare diseases, 87 partner groups, 14,034 participants, 50 states, 84 countries represented - as of February, 2022.

Participants provided informed consent to share their data with researchers and the Cure Mito Foundation

Participants are asked to respond to 2 surveys

- General survey - Common Data Elements (CDE) advised by NIH
- LS specific survey

Information collected

Demographic
Genetic mutation
Time to diagnosis
Specialists seen
Symptoms
Loss of milestones
Disease management
Healthcare utilization
Infections
Quality of life
Caregiver burden

Data analysis

- Data analysis was done in SAS 9.4.
- QC of selected variables have been performed.

Current poster represents data collected between September 23 - December 31, 2021

RESULTS

Participants Enrollment

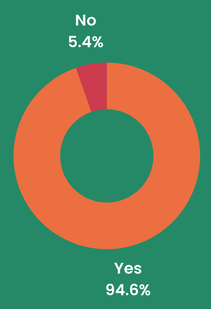
Enrolled: 134; Excluded: 25 - did not complete surveys, 8 - did not respond to question about sharing data with Cure MIto, 6 - did not give permission to Cure Mito to view data, 2 - confirmed to be asymptomatic carriers of mutation; Included in analysis: 93.

Table 1. Participants Characteristics (N=93)

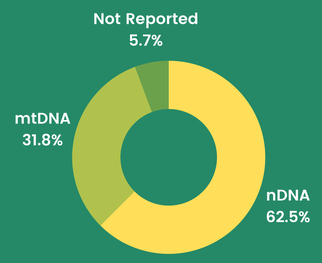
Participant is living, n (%)	85 (91.4)
Female, n (%)	49 (52.7)
White, n (%)	72 (77.4)
Age at survey submission [a] Mean (SD); Median (Q1, Q3); Min, Max	8.1 (10.7); 5.0 (3.0, 8.0); 0, 68
Age at death [b] Mean (SD); Median (Q1, Q3); Min, Max	2.8 (2.8); 2.0 (1.0, 3.5); 0, 9
Age at diagnosis [c] Mean (SD); Median (Q1, Q3); Min, Max	3.1 (5.8); 2.0 (1.0, 3.0); 0, 46

a. Calculated for living participants only (N=85)
b. Calculated for deceased participants only (N=8)
a. Calculated for participants with non-missing response (N=78)

Diagnosis determined by genetic testing (N=93)

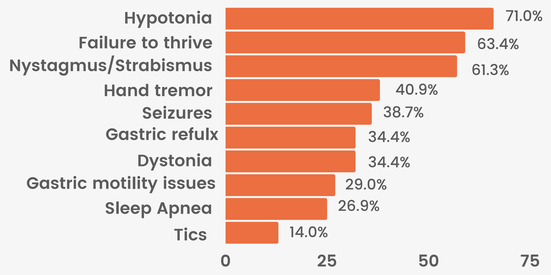


Mutation Type (N=88) [a]

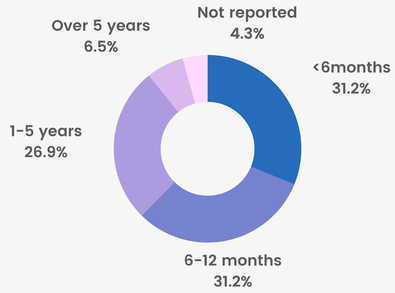


a. Calculated for participants whose diagnosis was determined by genetic testing (N=88)

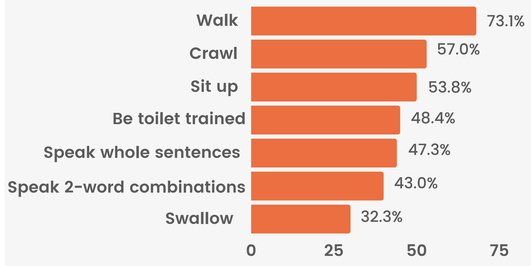
History of Symptoms (N=93)



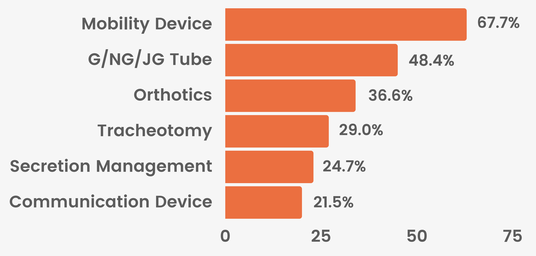
Time from Symptom Onset to Diagnosis (N=93)



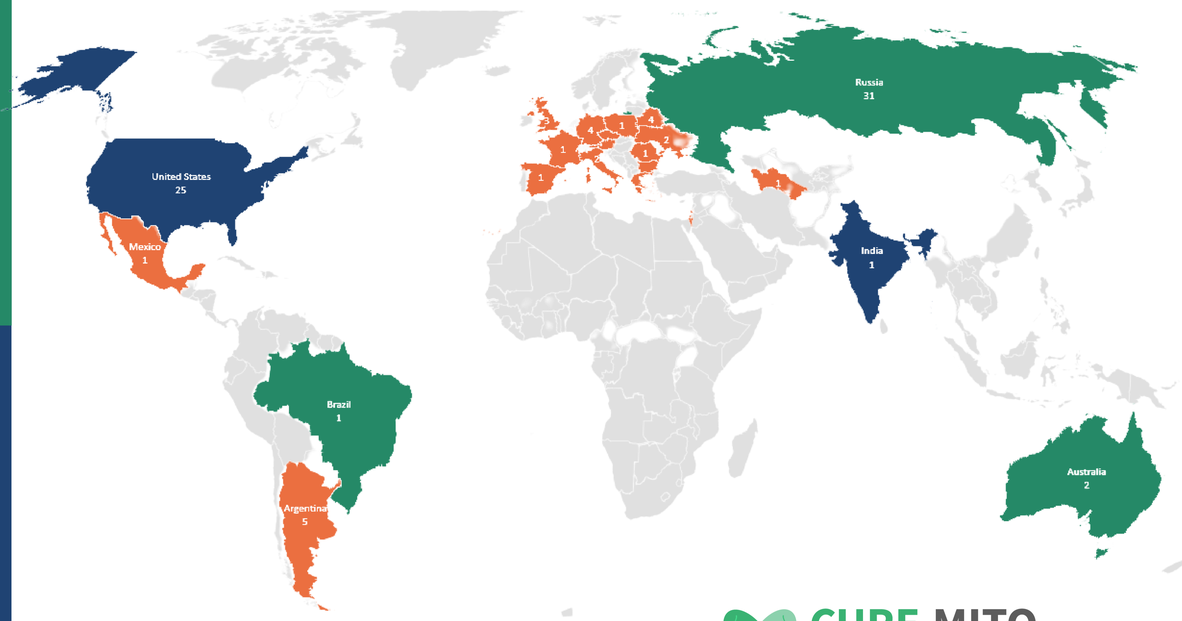
Lost or Never Reached a Milestone (N=93)



Disease Management (N=93)



Participants Country of Residence



CONCLUSIONS

Data collected during the first three months since starting the registry is comprehensive and describes the Leigh Syndrome population's diagnostic information, symptoms, and other clinical characteristics.

Participation from 22 countries in the first 3 months, with over 70% international patients, suggests that this might be the first truly global patient registry for LS.

Future efforts include:

- Continued recruitment of patients from around the world
- Publication of further results and findings
- Integrating de-identified data into RDCA-DAP platform by Critical Path Institute
- Collaboration with AllStripes for additional Leigh Syndrome insights

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This study is registered with ClinicalTrials.gov, Identifier NCT01793168.