



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

We are thrilled to share that we have our 2nd published paper about Leigh syndrome patient registry! Our paper, "**Interoperability of Leigh Syndrome Patient Registry Data with Regulatory Submission Standards**", is published in the *Journal of the Society for Clinical Data Management*.

The Clinical Data Interchange Standards Consortium (CDISC) is dedicated to helping to improve clinical research by driving meaningful and efficient research through data standardization. CDISC standards are mandatory for submissions to the FDA and PMDA.

Establishment of interoperability of registry data with regulatory submission standards like CDISC is essential to accelerate the development of therapies. The project described in our paper has been undertaken to establish such interoperability for Leigh syndrome patient registry data.

To our knowledge, this is the first such project for any mitochondrial disease patient registry, and we hope our experience will serve as a model for other mitochondrial disease and rare disease patient registries.

Additionally, we are proud to see that our work on the registry is very well aligned with the 2 recent FDA guidances: [Real-World Data: Assessing Registries To Support Regulatory Decision-Making for Drug and Biological Products](#) and [Data Standards for Drug and Biological Product Submissions Containing Real-World Data](#).

This project is a result of our partnership with Sumptuous Data Sciences, LLC. Sumptuous Data Sciences provides biostatistics and data operations services in healthcare sector with vision to work towards bridging the gap between healthcare and clinical data analysis and management. We are extremely grateful to Parag Shiralkar and Pallavi Bakare and their team for their partnership on this project and for their support of the Cure Mito Foundation and Leigh syndrome community.

We also thank all patient families who enrolled into the registry so far and all the doctors who share the registry with their patients.


Please read the full paper [HERE](#)

Please join the registry [HERE](#).

The Cure Mito team

**"In a gentle way, you can shake the world."**  
- Mahatma Gandhi





Shiralkar P, Bakare P, Woleben K, Zilber S. Interoperability of Leigh Syndrome Patient Registry Data with CDISC Standards. *Journal of the Society for Clinical Data Management*. 2023; 4(1): 1, pp. 1-6. DOI: <https://doi.org/10.47912/jscdm.244>

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**ORIGINAL RESEARCH**

## Interoperability of Leigh Syndrome Patient Registry Data with CDISC Standards

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**Introduction:** Leigh syndrome (LS) is a rare, severe neurometabolic disorder and a type of primary mitochondrial disease. The Cure Mito Foundation is a nonprofit foundation founded in 2018 by parents of affected children. The organization’s mission is to unite the global LS community to accelerate patient-centered research, treatments, and cures. Cure Mito has launched a global patient registry for Leigh syndrome in September 2021 to better understand the disease, facilitate clinical trials recruitment, and build a strong global community.

**Objectives:** Patient data is collected in registries for getting a real-world view of patient reported outcomes, and to improve the understanding of the disease. Establishment of interoperability of registry data with regulatory submission standards like CDISC is essential to accelerate the development of therapies. The project described here has been undertaken to establish such interoperability for Leigh syndrome patient registry data.

**Methods:** Establishing interoperability consisted of assessment of all data elements and collected data, alignment of data elements and variables with CDASH domains and variables, terminology alignment, data transformation, and compliance assessment.

**Results:** Data assessment has been done, domain map has been developed, and data has been converted to CDASH standard.

**Conclusion:** Cure Mito Foundation and Sumptuous Data Sciences, LLC completed a project focused on interoperability of Leigh syndrome patient registry data with CDISC standards, converting the data to CDASH standards, with SDTM conversion in progress. This project is a key milestone for all stakeholders who collect real world data in rare diseases.

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**Keywords:** Leigh syndrome; mitochondrial disease; patient registry; CDISC; CDASH; SDTM; patient advocacy; cure mito; data standards



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](mailto:info@curemito.org) to learn more.



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