PP04

# PP04: Alignment and Interoperability of Leigh Syndrome Registry Data with Regulatory Submission Standards

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**Abstract:** The patient data is collected at registries for getting a real- world view of patient reported outcomes, and to know about the diseases more. Establishment of interoperability of registry data with acceptable submission standards like CDISC is essential in order to accelerate the development of therapies and is a critical milestone in case of rare diseases.

This is a pilot project for establishment of such interoperability of patient registry data with CDISC standards. This poster presentation provides an overview of an outcome of this pilot project. The poster provides overview of the current data collection practices of patient registry data collected through Sanford CoRDS patient data registry provides methodical steps executed to convert such data into CDISC requirements and provides further assessments and recommendations pertaining to modification of patient registry data collection instrument.

## Background of Patient Registry Data

Leigh Syndrome (LS) is a rare genetic neurometabolic disorder resulting into resulting into degeneration of central nervous system and death. LS can be caused by a number of mutations in mitochondrial or nuclear DNA. Patient registries are particularly important in rare diseases, where patient numbers are small, and funding is limited.

Coordination of Rare Disease at Sanford (CoRDS) platform to host the data

NIH Common Data Element Repository- Survey Questions Design

→ Patient Registry data in the form of Questionnaire response from patients

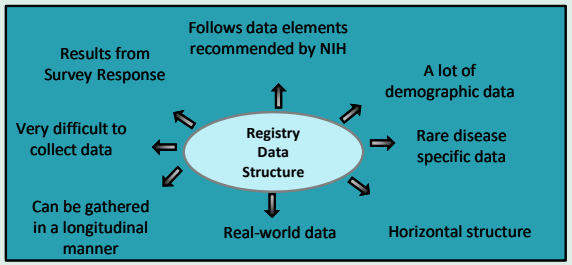
## Need for Establishment of Interoperability

CoRDS Patient Registry contains real-world data specific to Leigh Syndrome

Such securely managed data is easily verifiable at its 'source' or collection point.

The data can be used for interaction with regulatory agencies if it is aligned in right standards/formats

Establishment of interoperability can help academia, researchers and industry:
- To understand the data for its use for clinical research
- To analyze and report the data in commonly accepted standards and models.




## Approach in Establishing Interoperability of the Data

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| Step-1: Assessment of NIH data elements | Step-4: Variable alignment with CDASH domain variables |
| Step-2: Review and assessment of collected data | Step-5: Terminology alignment |
| Step-3: Alignment of data elements with CDASH Domains | Step-6 : Data transformation and compliance assessment |

**Key Observations –**

- Patient registry data is a single dataset with all information together. Domain map needs to be developed to split the data into 'domain' specific to CDASH (data collection standards of CDISC) requirements.
- Variables need to be differentiated as relevant and irrelevant from clinical significance point of view. Additional variables to be kept from 'sufficiency' point of view. Requires Data Cleaning.
- Horizontal structure of data; needs to be pulled into CDISC compliant format. Data from single survey form field is split into multiple variables.

**Current Status:**

- Patient registry data is analyzed, and domain map created to split data as per CDASH standards.
- Significant portion of data (>90%) can be mapped to key CDASH domains of AP, CE, DD, DM, DS, PF, MH, and QS – rest to map to supplemental domains.
- A lot of data pertaining to Pharmacogenomics/Genetics Findings, Medical history, and Questionnaire domains.
- Programming development of CDASH specific domains in progress.

**Next steps:**

- Complete the transformation of registry data to CDASH standards and then convert the data to SDTM standards.
- Based on the assessments of the data, and the CDISC specific needs, give feedback to data collection instrument design team for possible design of data elements to adhere to standards.
- By taking CDISC360 degree approach convert CDASH based data into SDTM for submission readiness (as per US-FDA requirements) through P21 compliance processes.
- Provide a synopsis of the process along with compliance assessments to researchers, as well as to industry to foster, promote, and accelerate the data analysis, reporting, and representation processes in a regulatory compliant manner.
- Promote, foster, and accelerate the development of novel therapies and treatments to cure Leigh Syndrome

**References:**

1) Clinical Data Acquisition Standards Harmonization (CDASH), CDASH | CDISC
2) NIH Common Data Element repository, NIH Common Data Elements (CDE) Repository

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