Best Data Practices for Rare Disease Patient Foundations and Researchers – PHUSE Working Group Project

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Introduction

- Leigh syndrome global patient registry – Cure Mito Foundation

- PHUSE working group: [Best Data Practices for Rare Disease Patient Foundations and Researchers]

- Patient registry office hours, conferences, podcasts, collaborations (UPenn Orphan Disease Center)

- *“I hope that something comes of this. I have done so many surveys and questionaires [sic] and NOTHING has ever come of it, NOTHING”.*

- *"I am hopeful that I can make useful contributions through my participation that will help others in the future. Otherwise, my participation is in vain. "*

- *"The one thing I wish was to receive the results of the study. Even if the study didn’t show how exercise helps delay the onset of HD, it would have been great to at least be shown the results or receive a link to the publication. Letting the patients know about the results is something that all research studies should require. Without the patients, there are no studies!”*

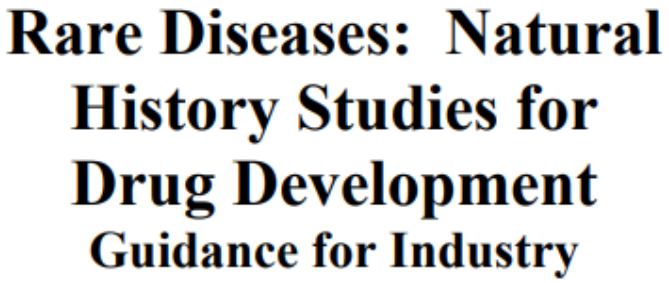
Quotes from patients

Challenges

There's no guidance (from FDA, industry) for registries developed by patient groups.

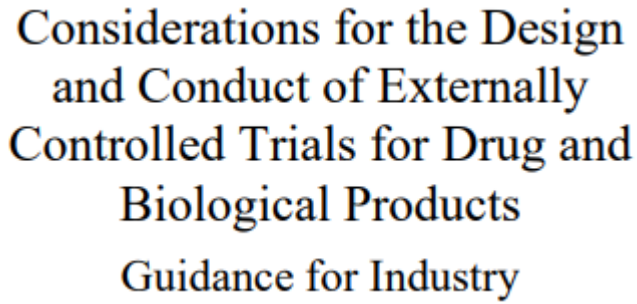
Many patient groups are fully volunteer or have some staff, but not data analysis/technical knowledge expertise.

Existing guidance documents are for the industry:



https://www.fda.gov/media/122425/download

https://phuse.s3.eu-central-1.amazonaws.com/Deliverables/Real+World+Evidence/WP-074.pdf



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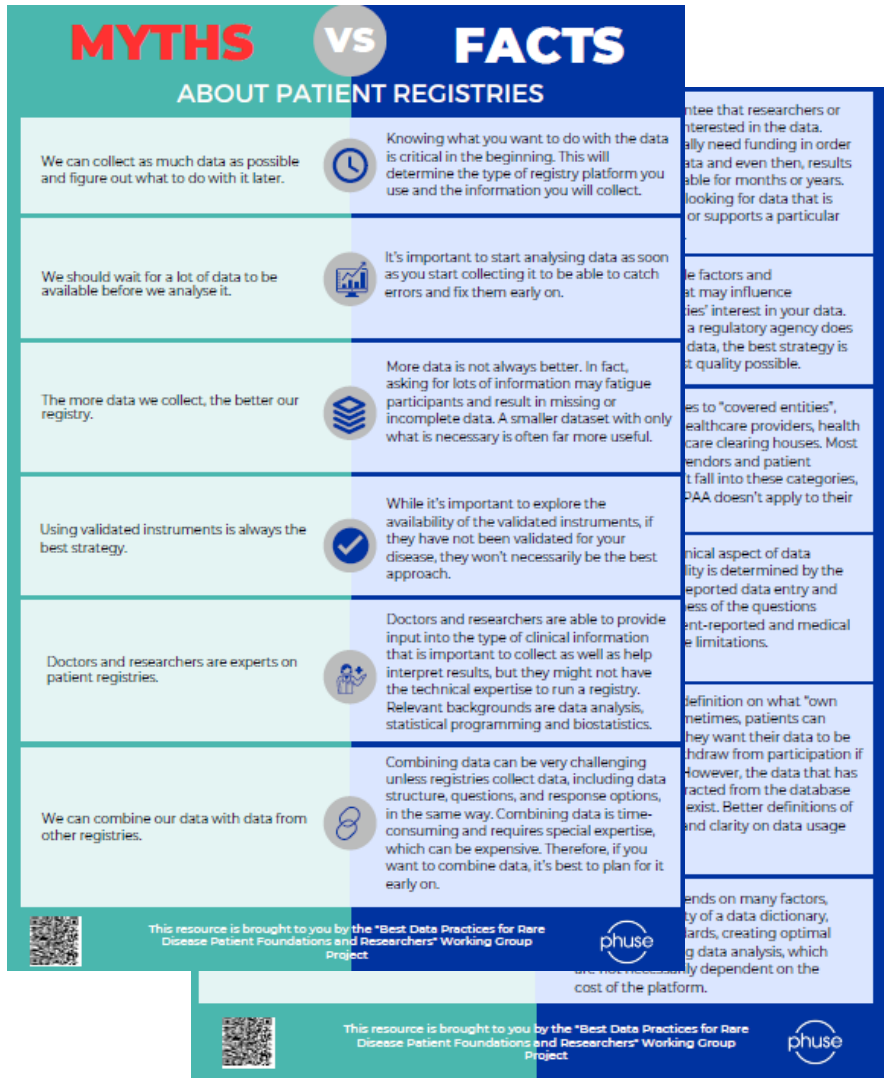
PHUSE working group: best data practices for patient foundations and researchers



Common misconceptions about registries among patient groups

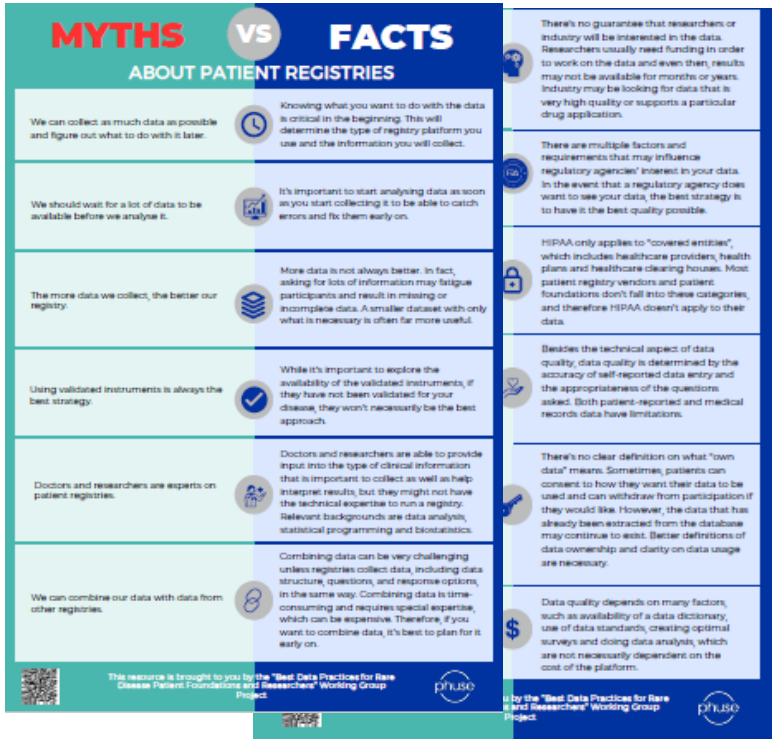
Industry, FDA, and researchers would find the data highly valuable

Data can be used as natural history data in a clinical trial (for example as control arm)

Data can be easily combined with any other data or registry can “plug in” to another registry

Data privacy (HIPAA), etc..

UPenn survey - 17/22 responded that patient foundations are required to follow HIPAA
https://www.orphandiseasecenter.med.upenn.edu/patientregistryfaqs



Registry vendor considerations

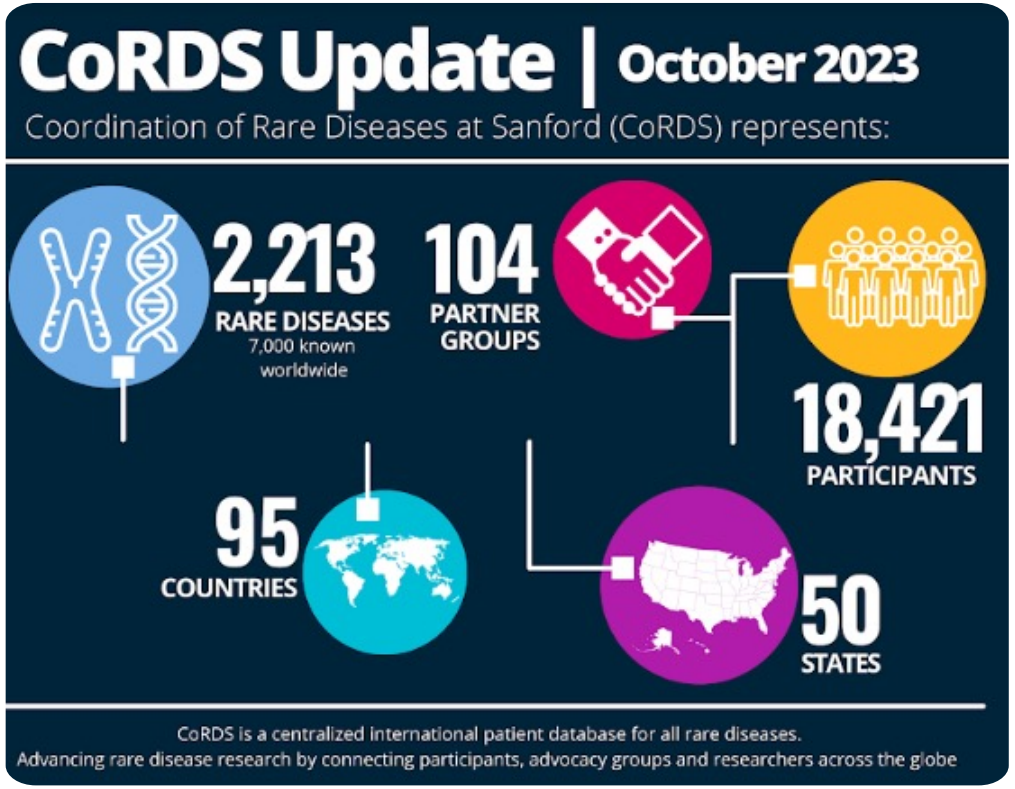
- Price – free vs. paid (price can be per patient, per survey, per question in survey)
- Data access – patient group has access to all of data vs. doesn't have access
- IRB approval – patient group has to submit IRB proposal vs. registry vendor has IRB
- Registry PI – patient group representative is the PI vs. registry vendor representative is a PI
- Data ownership – patient group "owns" data, can move all data to another platform if desired vs. patient group doesn't have this ability
- Data structure, number of surveys that can be created, availability of surveys "built-in"

Patient groups considerations when choosing registry platform

- Common consideration by patient groups: Patient registry vendor has similar values/beliefs about patient advocacy, patient engagement, “Patient owns data”, etc..

Rarely considered by patient groups:

- Data structure
- Raw data availability
- Being able to easily obtain basic information from the data (Example: How many patients have seizures?)
- Ability to share insights and what else they (as an organization) can do with the registry



Leigh syndrome patient registry – Cure Mito foundation

- **Considerations in choosing this platform**
- Easy to contact patients with clinical trial opportunities
- Raw data is available
- Data dictionary is available
- Data structure (NIH common data elements)
- Well designed general survey provided by CoRDS
- Free
- Cure Mito could use data from the very beginning to bring value, regardless if researchers/industry/FDA interest in data

Leigh syndrome patient registry- Cure Mito foundation

Launched by Cure Mito Foundation in September 2021

Partnership with Coordination of Rare Diseases at Sanford (CoRDS) - 100+ patient groups, 2000+ rare diseases

Global registry, ~ 300 participants, 35+ countries

95% - diagnosis genetically confirmed (60% diagnosed within 1 year since symptoms start)

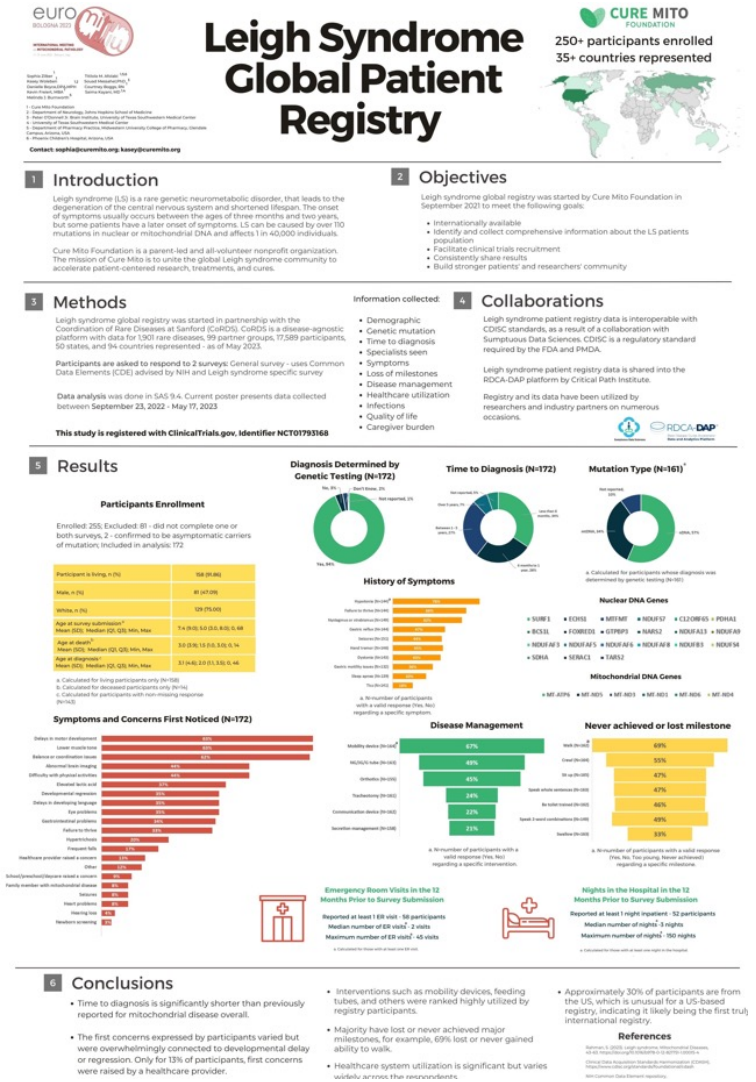
Data is interoperable with CDISC standards (CDASH/SDTM)

Data is shared integrated into RDCA-DAP platform by Critical Path Institute (C-Path)

FDA listening session February 2023

10+ conferences, 4 posters, 2 papers (1 published, 1 in peer review)

Data was shared with researchers, used to share clinical trial opportunities multiple times



Medical records collection

Medical records are collected with patient's consent (Citizen, Picnic Health) mainly to build natural history data to use in clinical trials as a control arm

- Can this data actually be used as a control arm? (vs. prospective natural history study)
- What are other possible uses of such data by industry?
- How does it compare with claims data

- Implementing standard, consistent, and up-to-date definitions of medical conditions and treatments. These elements employ uniform medical language and are typically provided in advance in study protocols and procedure manuals.
- Providing a consistent schedule of medical visits for the patient.
- Providing standard operating procedures for investigators, which provides for greater consistency in the information collected (e.g., using the same clinical outcome assessments with comparable instructions for use).
- Collecting additional data that may elucidate the pathogenesis and manifestations of a disease and the patient's concomitant treatments.

Source: Rare Diseases: Natural History Studies for Drug Development (FDA guidance)

How data collected by patient groups can help

Make it easy to share clinical trial opportunities with patients

Make data available to industry and researchers

Build partnerships between patient community and industry

Understand burden of the disease

Build strong global community

Understand patient and caregiver perspectives

Thank You!

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