



Patient Registries – Challenges and Opportunities

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Introduction/Bio

Sophia Zilber is a Statistical Programming Lead in Pfizer, where she's managing a team of statistical programmers, preparing the study and asset programming deliverables for submission. She has 20 years of experience with clinical data analysis. Her experience includes programming, creating study documentation, and establishing strategy, timelines, and resourcing of statistical programming and analysis deliverables, following CDISC and regulatory standards.

External to Pfizer, Sophia applies her passion for patient advocacy and data sharing through her involvement with rare disease community. Sophia is currently a board member for Cure Mito Foundation where she's also a patient registry director and has developed a global patient registry for Leigh Syndrome. She's also a board member for New England Regional Genetics group and a scientific advisor to Rare-X. Sophia has authored papers and has done multiple presentations on the topic of patient registries and data sharing.

Sophia believes that patients are critical to advancing treatments forward and that sharing knowledge, and open, honest and transparent communication are a key to a successful partnership.



Disclaimer

The views expressed in this presentation are the personal views of the author and may not be understood or quoted as being made on behalf of or reflecting the position of the regulatory agency/agencies or organizations with which the author is/are employed/affiliated.

Agenda

Background

Patients-driven research: advantages, challenges

Patient registry for patient reported data – example

Program for electronic health records data collection – example

Limitations of both methods and discussion topics

CDISC and Real World Data

Opportunities for pharma industry

Q&A

Background

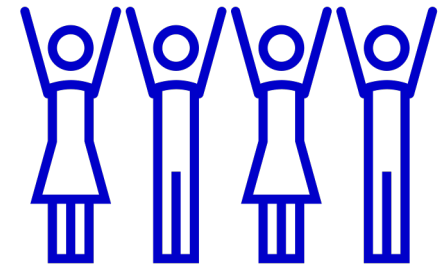
Real World Data examples¹:

- Electronic Health Records (EHRs);
- Medical claims and billing data;
- Data from product and disease registries; biobanks;
- Patient-generated data, including from in-home-use settings; and data gathered from other sources that can inform on health status, such as mobile devices

Patient Registries:

- Used to be fully maintained by hospitals or academic institutions
- As patients play a greater role in research, patient registries are frequently run by patients themselves, usually through patient advocacy foundations

¹ - <https://evs.nci.nih.gov/ftp1/CDISC/Glossary/CDISC%20Glossary.pdf>



Advantages



Patients have a voice in research



Collecting data that's meaningful to patients



Often quicker to collect data, easier outreach



Data can be shared much easier than data collected in other ways (hospital, and other)



Overall faster to advance clinical trials, treatments

Challenges

Quote from patient: *"I hope that something comes of this. I have done so many surveys and questionnaires [sic] and NOTHING has ever come of it, NOTHING"*

Lack of expertise (survey design, data management, data analysis)

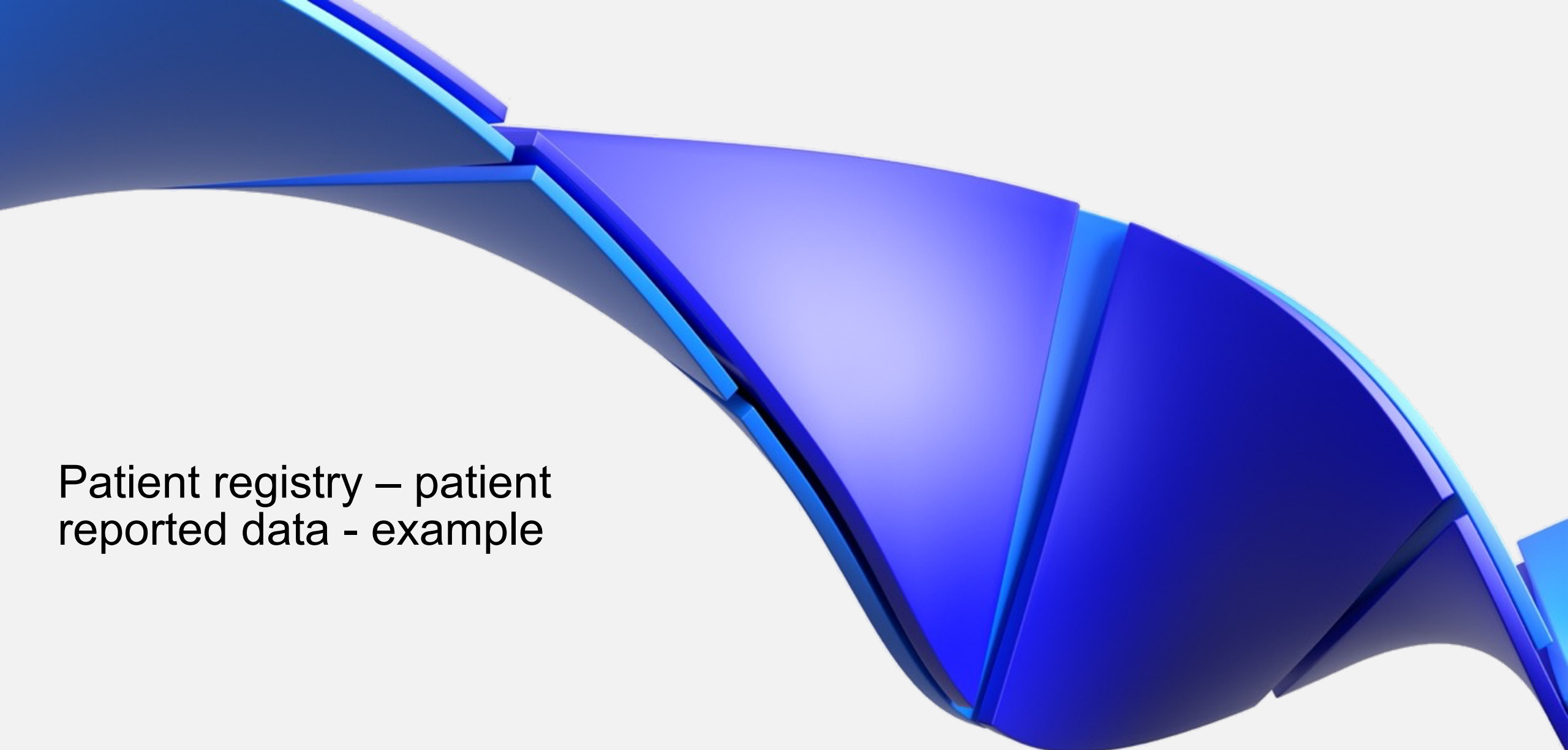
Poor data quality

Data sometimes never analyzed or used

Lack of clarity regarding data privacy and data ownership

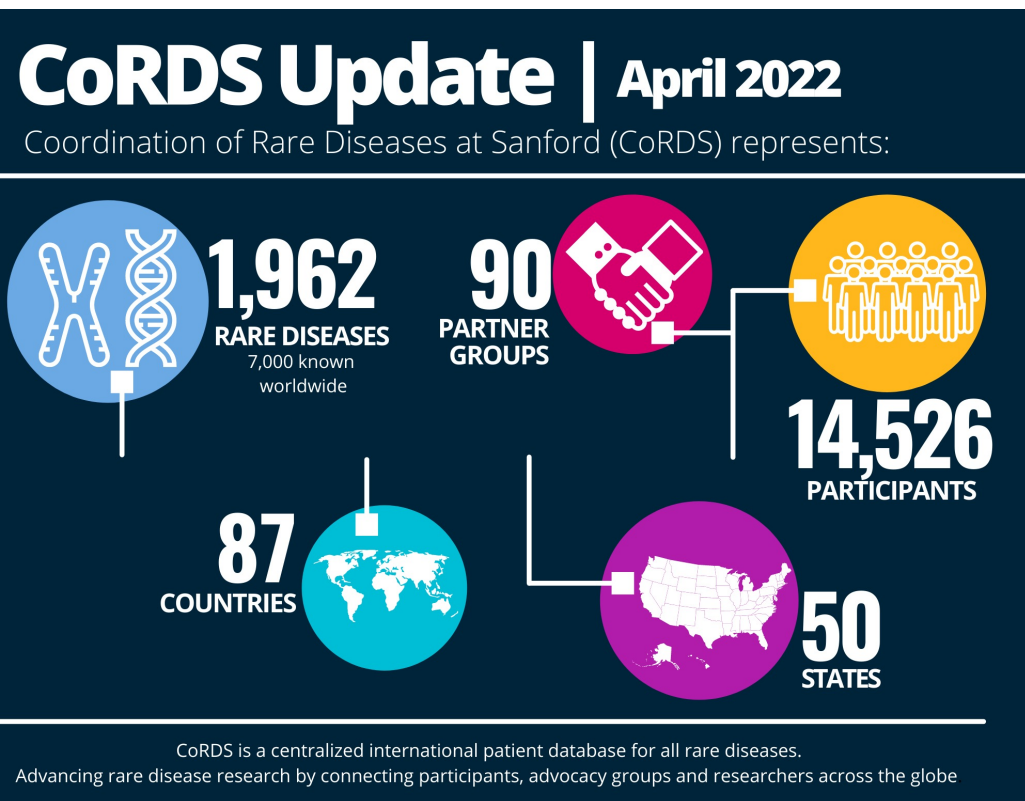
Inaccurate beliefs about data (examples: registry=cure, FDA/industry have high interest in the data, different datasets can be easily combined)

Challenges result in slowing down research, patients discouraged

An abstract, three-dimensional graphic composed of several blue, curved, and faceted planes that overlap and curve across the top and right side of the slide, creating a sense of depth and movement.

Patient registry – patient reported data - example

Leigh Syndrome Global Registry – Cure Mito Foundation



Based at Sanford Research, a nonprofit research institution, CoRDS is a centralized international patient registry for all rare diseases.

We coordinate the advancement of research into 7,000 rare diseases. Here's how:

- We work with patient advocacy groups, individuals and researchers.
- We capture health information from individuals with a rare diagnosis, undiagnosed patients, unaffected carriers or at-risk patients.
- We connect researchers and patients and notify our participants of emerging clinical trials.
- We make the registry accessible. Participants can enroll for free and researchers can access it for free.

[Source:https://research.sanfordhealth.org/rare-disease-registry](https://research.sanfordhealth.org/rare-disease-registry)

Leigh Syndrome Global Registry – Cure Mito Foundation

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

Leigh Syndrome Global Registry – Cure Mito Foundation

General survey – based on NIH survey for rare diseases (Common Data Elements recommended by NIH)

Exactly the same general/demographic survey for each rare disease supported by CoRDS

Easy to harmonize data across rare diseases

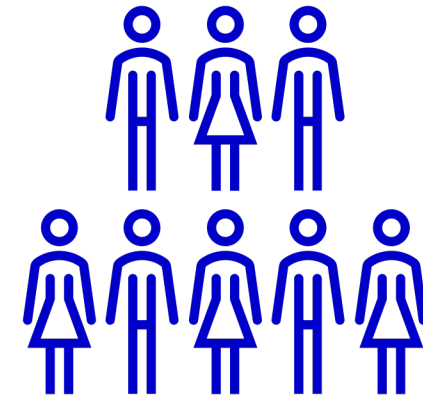
Disease specific survey – developed separately for each disease

Linked to Orphanet list of rare diseases

Patient can join even if no advocacy group represented in CoRDS

Joining based on disease

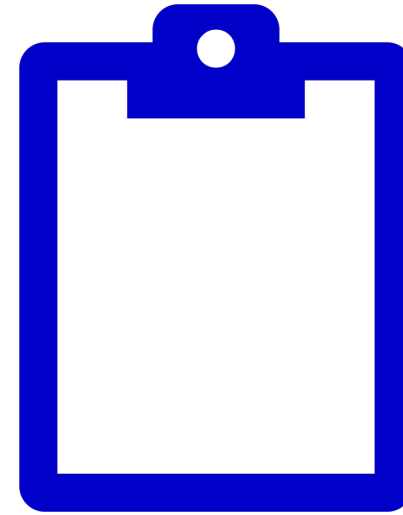
Multiple family members with same disease can be linked together



Leigh Syndrome Global Registry – Cure Mito Foundation

Information collected:

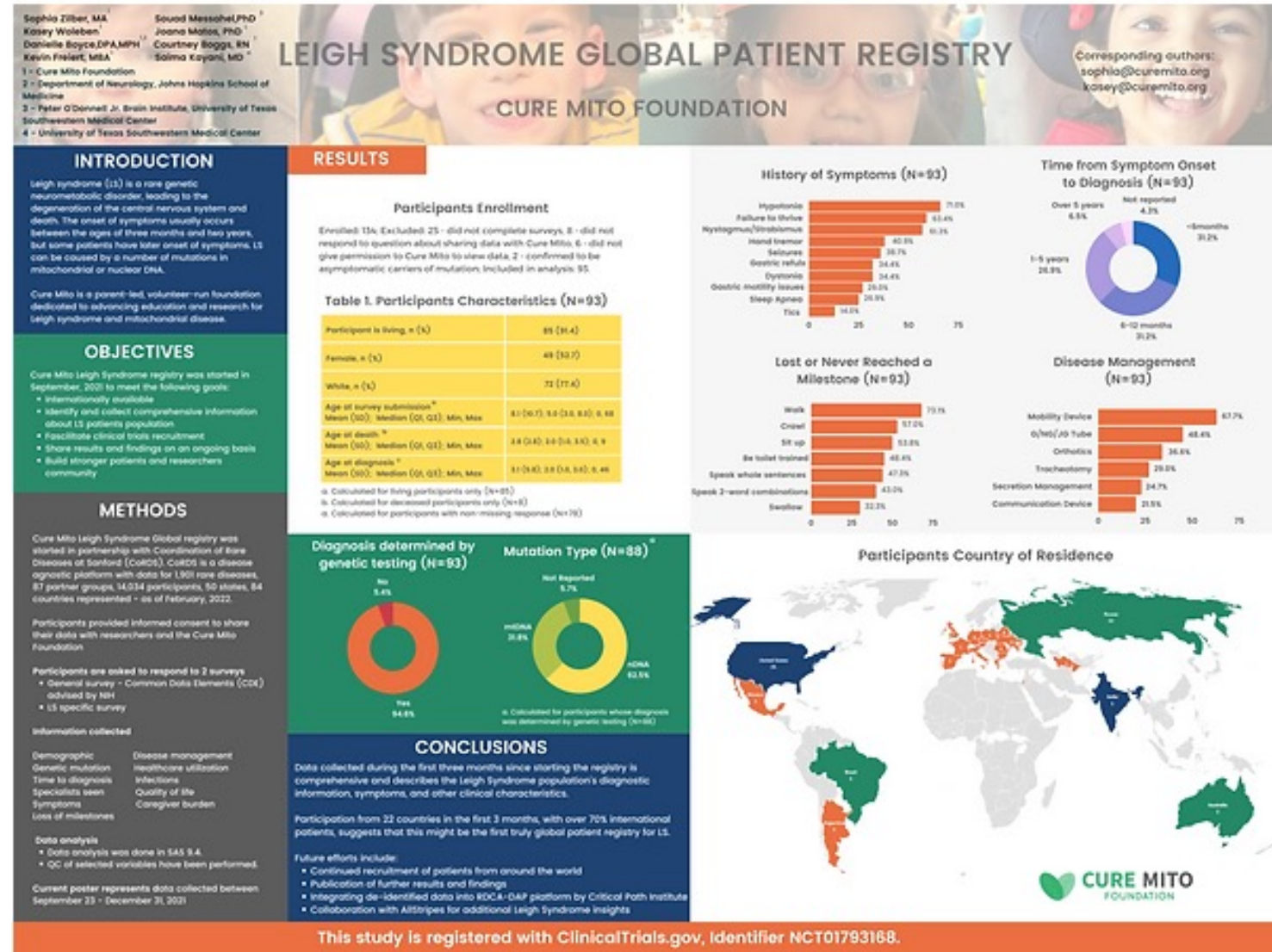
- Diagnosis and genetic mutation information
- Symptoms/concerns first noticed
- Loss of milestones
- Disease management
- Symptoms experienced
- Healthcare utilization
- Quality of life
- Caregiver burden



Poster

Mitochondria-Targeted Drug Development Summit, February 22-24, 2022

<https://www.curemito.org/results>



Poster

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)

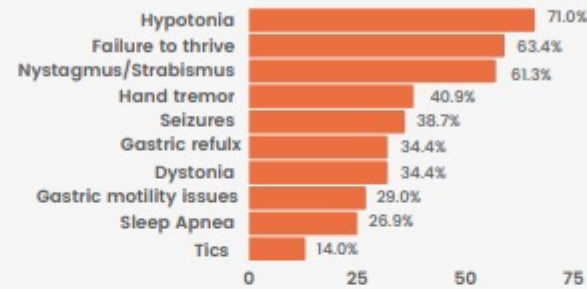
Participant is living, n (%)	85 (91.4)
Female, n (%)	49 (52.7)
White, n (%)	72 (77.4)
Age at survey submission ^a Mean (SD); Median (Q1, Q3); Min, Max	8.1 (10.7); 5.0 (3.0, 8.0); 0, 68
Age at death ^b Mean (SD); Median (Q1, Q3); Min, Max	2.8 (2.8); 2.0 (1.0, 3.5); 0, 9
Age at diagnosis ^c Mean (SD);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

c. Calculated for participants with non-missing response (N=78)

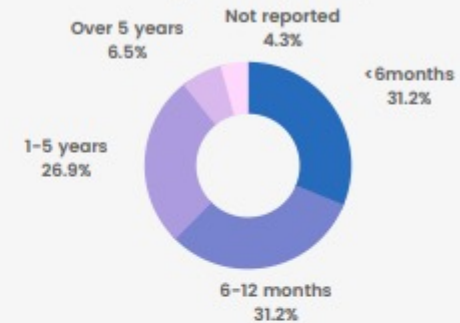
History of Symptoms (N=93)



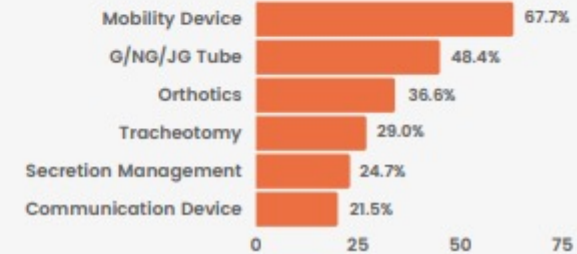
Lost or Never Reached a Milestone (N=93)



Time from Symptom Onset to Diagnosis (N=93)



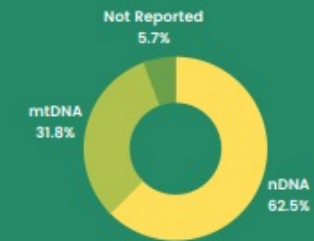
Disease Management (N=93)



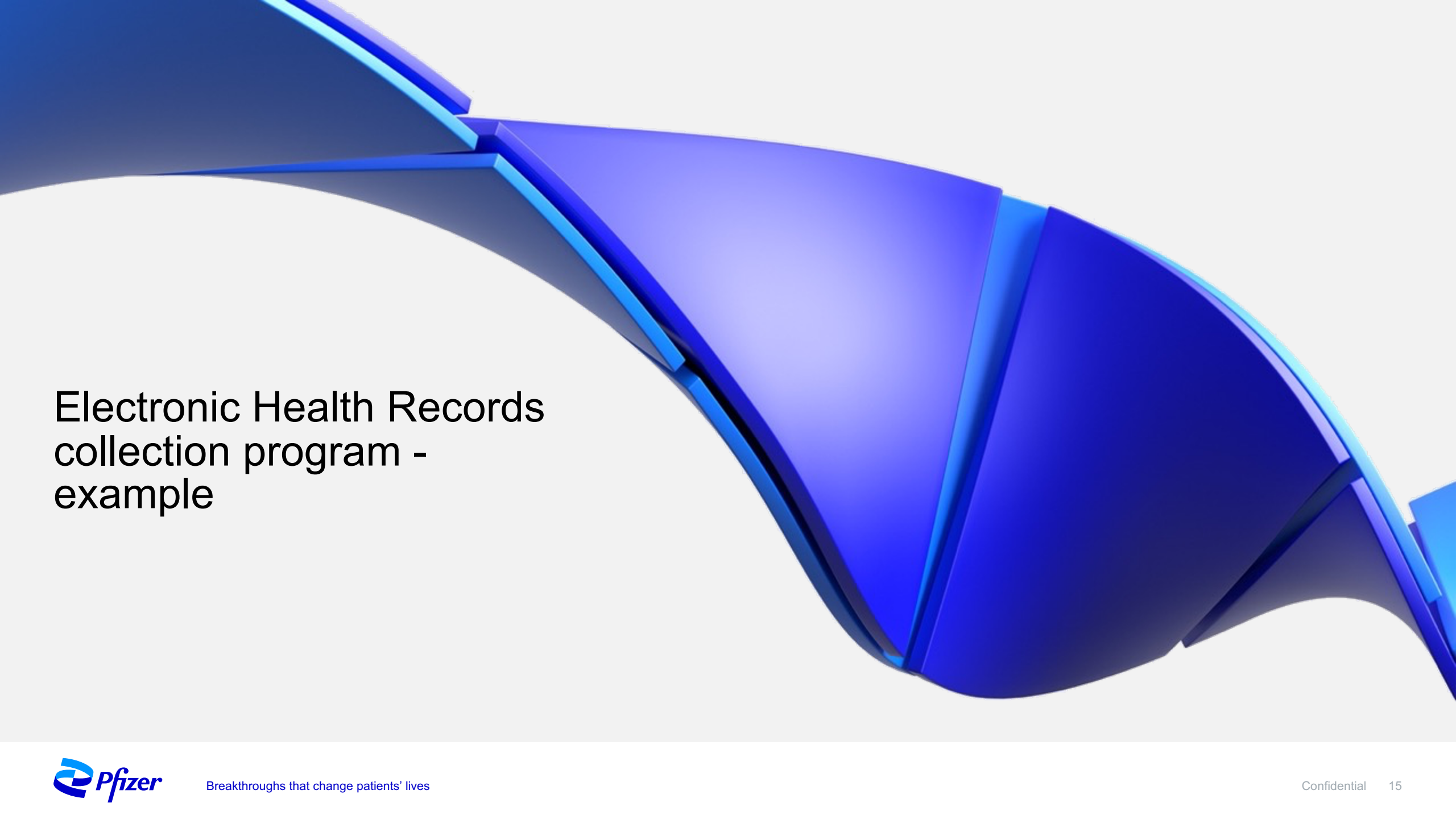
Diagnosis determined by genetic testing (N=93)



Mutation Type (N=88)^a



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



Electronic Health Records collection program - example

Allstripes – Electronic Health Records – Cure Mito Foundation

- AllStripes collects medical records on behalf of patients, removing all burden from patients and caregivers to connect their health portal accounts or upload their files.
- AllStripes digitizes, structures and abstracts information from the records, including handwritten notes from doctors that may contain valuable insights.
- The data in AllStripes is longitudinally updated. AllStripes looks at data from the entire medical history since pre-diagnosis and updates data each year to create a much more complete picture.
- Countries: US, UK, Canada

BENEFITS TO JOINING

Have a voice in research

Your de-identified records, and your input through surveys, can help guide treatment research.

Access your medical records

We collect your records, at no cost, in your secure account. View doctors' notes and your care journey.

Learn from your community

Receive updates on what we're learning from your community's data, and your research impact.

Limitations and discussion topics



Patient-reported data:

Works well for clinical trials recruitment, engaging patient community, raising awareness of disease

Research value of data?

Support for regulatory submission?

Lacking clinical information (labs, procedures, medications)



Electronic health records

Provides clinical information

Is information complete?

What is missing?

Research value of data?

Support for regulatory submission?



Critical Path Institute (C-Path)

Data can be combined using GUID

Concern regarding patient re-identification



Use of Clinical Data Interchange Standards Consortium (CDISC) Standards for Real-world Data: Expert Perspectives From a Qualitative Delphi Survey

Facile R, Muhlbradt EE, Gong M, Li Q, Popat V, Pétavy F, Cornet R, Ruan Y, Koide D, Saito TI, Hume S, Rockhold F, Bao W, Dubman S, Jauregui Wurst B; Use of Clinical Data Interchange Standards Consortium (CDISC) Standards for Real-world Data: Expert Perspectives From a Qualitative Delphi Survey; JMIR Med Inform 2022;10(1):e30363; doi: [10.2196/30363](https://doi.org/10.2196/30363) PMID: [35084343](https://pubmed.ncbi.nlm.nih.gov/35084343/)

<https://medinform.jmir.org/2022/1/e30363>

Reasons why CDISC should prioritize EHR¹:

- EHRs are one of the most available and largest data sources
- EHRs are already in electronic format
- EHRs contain important and essential information directly relevant to the patients' health status

- Real World Data plays increasingly important role in clinical research
- Data should be well defined and structured
- Consistent across stakeholders
- Clinical Data Interchange Standards Consortium (CDISC) data standards are globally recognized and used by pharmaceutical industry for regulatory submissions
- Can help link clinical trial data to other data sources
- Improve data quality
- Survey included at least 66 experts globally (criteria: knowledge of CDISC and experience with RWD)
- Barriers to using CDISC for RWD include
 - CDISC standards considered complex
 - Insufficient resources to implement
 - Developed for different purpose
 - Insufficient knowledge of value of data standards

Opportunities for pharma industry

Guidance for patients and patient advocacy groups

- Understanding of drug development process
- Understanding of data and how it's used
- Limitations of data
- Explain data management, data analysis
- Explain value/possible use of patient-reported data, electronic health records

Guidance for researchers

- Explain data from industry perspective
- Explain CDISC
- Value of CDISC standards
- Individual use cases

Why is this important?

- Patients are highly motivated
- A lot of money, time, effort already invested into collecting data
- By identifying and addressing the gaps, we can put patients into the center and get to treatments and cures faster



Q&A

Thank You

