Mapping Mitochondrial Disease Global Registries: A Comprehensive Review of Publicly Listed Patient-Driven and Clinical Umbrella Registries Related to Mitochondrial Disorders

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Poster **171**

Cristy Balcells,[1] Sophie Housiaux,[2] Kira Mann[3]

[1]UCB Pharma, Smyrna, GA, USA; [2]UCB Pharma, Brussels, Belgium; [3]International Mito Patients, The Netherlands

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Background

- Mitochondrial diseases are a heterogeneous group of rare, debilitating, and often fatal multi-systemic conditions that can severely impact daily living and quality of life[1-3]
- Low clinician awareness of rare diseases can delay diagnosis and patient access to novel therapies[4]
- Patient registries contain real-world scientific, clinical, and policy information that can be utilized to support research and capture patient-reported outcomes
  - However, information may remain siloed within individual registries with differing structures, purposes, and data schema; additionally, patients with mitochondrial diseases may belong to different support networks[5]
- In partnership with International Mito Patients (IMP), a global consortium of mitochondrial disease patient advocacy organizations, the need for improved sharing of information and opportunity for cohesion amongst various stakeholders interested in mitochondrial disease registries was acknowledged
  - Global organizations and networks that host umbrella mitochondrial disease registries were surveyed to learn about their goals and the challenges they face

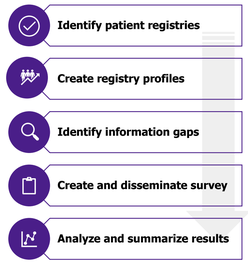
Objective

- To comprehensively describe the mitochondrial disease patient registry environment utilizing a web-based survey and additional online desk research

Methods

- An overview of the study process is provided in **Figure 1**
- A list of 15 mitochondrial disease registries known to UCB/IMP was combined with additional information obtained through desk research to generate a list of *umbrella* mitochondrial disease registries; disease-specific registries were excluded
  - Desk research was conducted via websites, patient advocacy groups, government agencies, academic institutions, rare disease portals, and a review of academic papers
- The information was used to build a database structure profiling the registries and information gaps were identified
- A 10-minute, online survey was designed to fill gaps in information with a series of dichotomous, and closed- and open-ended, questions (**Figure 1**)

Figure 1. Summary of Global Mitochondrial Disease Patient Registry Mapping Process



Example survey questions

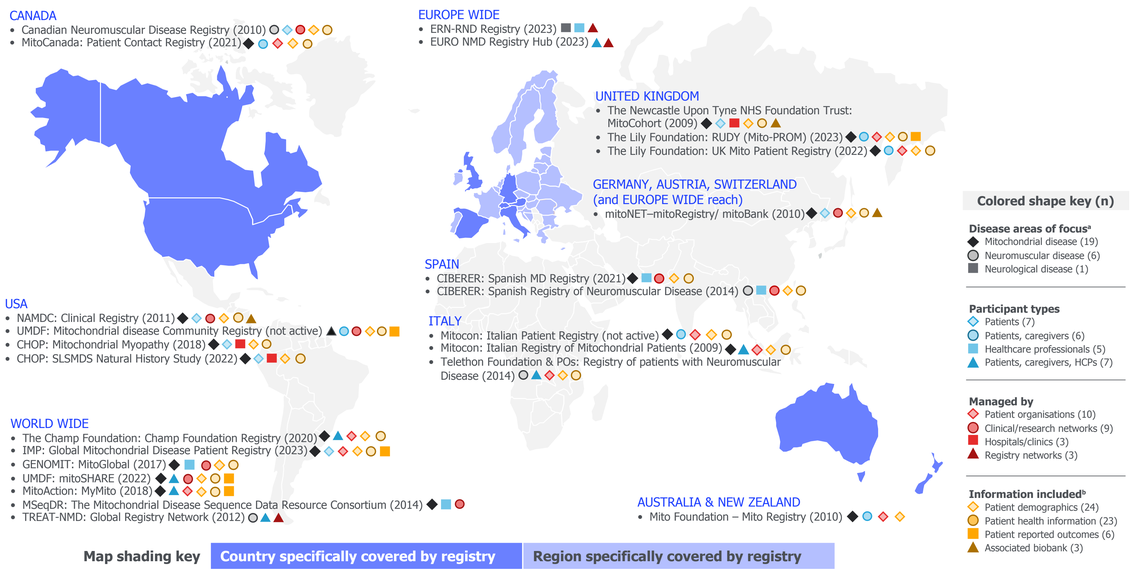
Does your registry collect identifiable patient information?
- Yes
- No

Does your registry collect any of the following patient health information?
- Choose from variables related to diagnostics, symptoms, treatment, and health records

Overall, what would you consider the biggest challenges or sources of concern about running a registry?
- Free text response

Do you expect your organization's registry to expand in scope or depth in the next 3-5 years? Will its purpose or reach change?
- Free text response

Figure 2. Global Map of Umbrella Mitochondrial Disease Patient Registries (N=25)



Note, mapping includes umbrella mitochondrial disease registries and excludes disease specific registries. We would welcome further input on patient registries to include in this mapping.
[a]Includes one registry that falls into two disease areas of focus (see **Table 1** for detailed data).
[b]Overlap exists between umbrella registry networks and individual registries (see **Table 1** for detailed data).

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Table 1. Umbrella Mitochondrial Disease Patient Registries Summary Table (N=25)

Region	Organisation: Registry name (date founded)	Disease area of focus	Participant types	Managed by	Information included: Patient demographic data	Patient health data	Patient reported outcomes	Biological material/ biobank
WORLD WIDE	The Champ Foundation: Champ Foundation Registry (2020)	Mitochondrial	Patients, caregivers, HCPs	Patient organisation	YES	YES		
WORLD WIDE	IMP: Global Mitochondrial Disease Patient Registry (2023)	Mitochondrial	Patients	Patient organisation	YES	YES	YES	
WORLD WIDE	GENOMIT: MitoGlobal (2017)	Mitochondrial	Healthcare professionals	Clinical/research network	YES	YES		
WORLD WIDE	UMDF: mitoSHARE (2022)	Mitochondrial	Patients, caregivers, HCPs	Clinical/research network	YES	YES	YES	
WORLD WIDE	MitoAction: MyMito (2018)	Mitochondrial	Patients, caregivers, HCPs	Patient organisation	YES	YES	YES	
WORLD WIDE	MSeqDR: The Mitochondrial Disease Sequence Data Resource Consortium (2014)	Mitochondrial	Healthcare professionals	Clinical/research network				
WORLD WIDE	Treat-NMD Registry Network (2012)	Neuromuscular	Patients, caregivers, HCPs	Registry network	*Network of individual, independent patient registries*			
AUSTRALIA & NEW ZEALAND	Mito Foundation: Mito Registry (2010)	Mitochondrial	Patients, caregivers	Patient organisation	YES			
CANADA	Canadian Neuromuscular Disease Registry (2010)	Neuromuscular	Patients	Clinical/research network	YES	YES		
CANADA	MitoCanada: Patient Contact Registry (2021)	Mitochondrial	Patients, caregivers	Patient organisation	YES	YES		
EUROPE WIDE	ERNRND Registry (2023)	Neurological	Healthcare professionals	Registry network	*Network of individual, independent patient registries*			
EUROPE WIDE	Euro NMD Registry Hub (2023)	Neuromuscular	Patients, caregivers, HCPs	Registry network	*Network of individual, independent patient registries*			
GERMANY, AUSTRIA, SWITZERLAND (and EUROPE WIDE reach)	mitoNET: Global Registry/mitoRegistry (2010)	Mitochondrial	Patients	Clinical/research network	YES	YES		YES
ITALY	Mitocon: Italian Patient Registry (not active)	Mitochondrial	Patients, caregivers	Patient organisation	YES	YES		
ITALY	Mitocon: Italian Registry of Mitochondrial Patients (2009)	Mitochondrial	Patients, caregivers, HCPs	Patient organisation	YES	YES	YES	
ITALY	Telethon Foundation and Patient Organisations: Italian Registry of Patients with Neuromuscular Disease (2014)	Neuromuscular	Patients, caregivers, HCPs	Patient organisation	YES	YES		
SPAIN	CIBERER: Spanish MD Registry (2021)	Mitochondrial	Healthcare professionals	Clinical/research network	YES	YES		
SPAIN	CIBERER: Spanish Registry of Neuromuscular Diseases (2014)	Neuromuscular	Healthcare professionals	Clinical/research network	YES	YES		
UNITED KINGDOM	The Newcastle Upon Tyne NHS Foundation Trust: MitoCohort (2009)	Mitochondrial	Patients	Hospitals/clinics	YES	YES		YES
UNITED KINGDOM	The Lily Foundation: RUDY (Mito-PROM) (2023)	Mitochondrial	Patients, caregivers	Patient organisation	YES	YES	YES	
UNITED KINGDOM	The Lily Foundation: UK Mito Patient Registry (2022)	Mitochondrial	Patients, caregivers	Patient organisation	YES	YES		
USA	NAMDC: Clinical Registry (2011)	Mitochondrial	Patients	Clinical/research network	YES	YES		YES
USA	UMDF: Mitochondrial Disease Community Registry (not active)	Mitochondrial	Patients, caregivers	Clinical/research network	YES	YES	YES	
USA	CHOP: Mitochondrial Myopathy (2018)	Mitochondrial/ Neuromuscular	Patients	Hospitals/clinics	YES	YES		
USA	CHOP: SLSMDS Natural History Study (2021)	Mitochondrial	Patients	Hospitals/clinics	YES	YES		

Color key

Disease areas of focus[4]
- Mitochondrial disease
- Neuromuscular disease
- Neurological disease

Participant types
- Patients
- Patients, caregivers
- Healthcare professionals
- Patients, caregivers, HCPs

Managed by
- Patient organisations
- Clinical/research networks
- Hospitals/clinics
- Registry networks

Information included
- Patient demographics
- Patient health information
- Patient reported outcomes
- Associated biobank

Results

Overview

- Manager overlap was identified among the registries; therefore, a total of 18 registry managers or teams were contacted for participation in the online survey
  - Responses were received from 15/18 umbrella registry managers (83%) regarding 19/25 registries (76%)
  - Three of the remaining 6 registries were not contacted due to lack of available up to date contact information; survey responses were not received from the other 3 registry managers
    - For these 6 registries, desk research provided data related to registry demographics such as disease areas of focus and participant types, but objectives, plans, and challenges faced could not be obtained
- Desk research and survey results revealed information relating to registry demographics, priorities, plans for growth, and challenges (**Table 1**, **Figures 2, 3,** and **4**)

Registry Objectives & Plans

- Most registries are primarily focused on collecting and tracking clinical data (**Figure 3**)
- Support of scientific research and advancing disease understanding were reported as top objectives by 89% (17/19) and 84% (16/19) of registries, respectively (**Figure 4**)
- 63% (12/19) of registries have plans for growth or change in the next 3-5 years including increased enrollment and participation in global registry networks (**Figure 4**)
  - The global registry mitoSHARE indicated plans to expand its scope and capabilities to better capture post-marketing data
  - GENOMIT, the most comprehensive global mitochondrial disease registry, supports clinical information and patient data; expansion plans include a patient-facing equivalent across EU-based registries
    - Driven by International Mito Patients (IMP), the expansion will be hosted on one common platform and managed by respective patient organizations

Challenges

- Time, resources (ie, staffing), and/or securing funding were commonly reported challenges for 47% (9/19) of registries (**Figure 4**)
- Additional work is needed to identify synergy between registries and ways to increase the number of recognized patients

Figure 3. Mitochondrial Disease Registry Demographics (N=25)

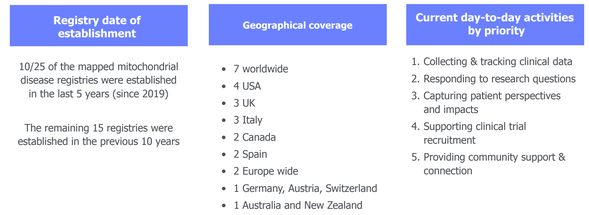

Figure 4. Key Insights Into Objectives, Growth Plans, and Challenges Reported by Mitochondrial Disease Registries (N=19)

Conclusions

- Mitochondrial disease registries are important for tracking patient data
- In a landscape where FDA- or EMA-approved therapies for ultra-rare genetic mitochondrial disorders are lacking, this fragmented infrastructure and limited funding remain challenging barriers to cohesion to aid the advancement of care for patients
- This mapping project provides a central resource of currently available data within the global umbrella disease registry environment
- For stakeholders, clinicians, and patients alike, these results will aid future research and the development of therapies for patients with mitochondrial diseases

For additional questions, contact: Cristy Balcells; Cristy.Balcells@ucb.com