

# Outcomes of an expert working group in Pompe Disease | Efforts towards building a responsible online research platform

## Background

The Pompe Consortium is a multi-perspective, collaboration-based, infrastructure building effort to support the Pompe community. A key area of interest of the consortium is accelerating research through collaboratively setting direction for a widely accessible research platform.

In partnership with the nonprofit organization Global Genes and its research platform RARE-X, the Pompe Consortium is creating a data collection and sharing initiative to support knowledge development in Pompe. This IRB-approved research study collects data from participants to aggregate and organize research-grade data. Data contributing participants retain full discretion over the use of their individual data, while the dataset is made available on an open science platform to encourage broad research use.

### Why Pompe Community?

Community is everything in rare disease, and this community - architected by the Pompe Consortium - is formed to accelerate understanding of the unknown or underrecognized.

### Who is it for?

Whether you're a newly diagnosed or experienced individual/family, advocacy org leader, clinician, researcher, or drug developer: this community has a place for you.

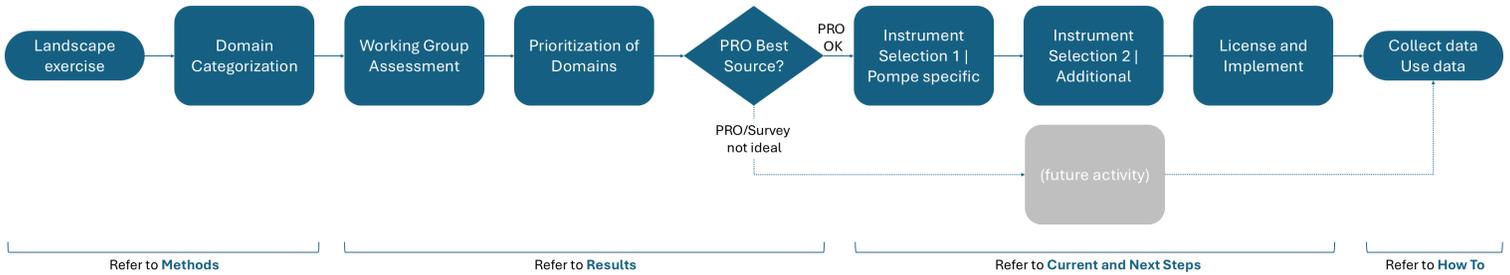
## Objective

Data is only useful if used.

The goal of the Pompe Consortium data collection and sharing initiative is to accelerate understanding of Pompe by reducing barriers to clinical research. This includes making it easier for patients and families to contribute data and making it easier for researchers to access and use that data.

A broad cross section of researchers, limited only by their ability to demonstrate proper data use and protection, have access to the dataset which is aggregated from contributors who have authorized sharing.

## Process



## Methods

Data can be sourced from various streams, including directly from contributing participants via patient-reported outcome measures (PROs) that address multiple domains, including (but not limited to) physical functioning, mental health, quality of life, and activities of daily living.

A landscape exercise identified **100+ variables** with a history of use in Pompe research across **19 domains/categories**.

Each member of a **multi-perspective\***, 17 person working group individually assessed:

- Importance of the domain (by age demographic)
- PRO\*\* appropriateness
- Best source of information

Potential Clinical Outcome Assessments (COA) instruments measuring the symptoms that are both critical and appropriate to collect via patient report were identified.

\*The working group includes diagnosed individuals and caregivers across a spectrum of age/progression/diagnosis path, physicians and researchers from academia and industry

\*\*Patient reported data can include patient reported symptoms & diagnosis, disease and treatment burden, and standardized clinical outcome assessment measures (COAs).

## Results Thus Far

Represented below is the summary scoring outcome from the prioritization/source assessment, as an aggregate for all age demographics\*.

Symptoms/ Domains	Total Importance Score
Respiratory	189
Muscular/Movement	176
Cardiac	164
Metabolic labs	159
Digestive (s/f)	152
Genetic data	148
ADL (Activity of Daily Living)	143
Nutrition	142
Prescriptions / treatment	141
Treatment Logistics & Challenges	134
Cognition	133
Physical Activity/ exercise	131
Pain	129
QoL (Quality of Life)	125
Psychological	121
Communication	108
Fatigue	107
Digestive (GI)	105
Vision/ocular	102

Symptoms/ Domains	Appropriateness for PROs
Psychological	9
Fatigue	9
ADL (Activity of Daily Living)	10.5
Pain	10.5
QoL (Quality of Life)	11
Physical Activity/ exercise	15
Treatment Logistics & Challenges	21.5
Digestive (GI)	32
Vision/ocular	50.5
Cognition	53
Nutrition	53.5
Communication	70
Prescriptions / treatment	70.5
Digestive (s/f)	72
Muscular/Movement	91
Respiratory	127.5
Cardiac	129.5
<b>Genetic data**</b>	<b>133</b>
Metabolic labs	144

Most Critical  
PRO is less ideal

Carry forward to instrument selection  
Carry forward to future activity

\*individual results by age demographic are considered in instrument selection step.

\*\*Genetic Data (from test reports) submitted by patients, curated by experts is already implemented on RARE - X

## Current and Next Steps

In consideration of participant burden, the working group is considering instrument selection in two segments:  
 1 | Pompe specific instruments  
 2 | Additional instruments that would cover areas of current or future lived experience/research interest that are not well captured by the Pompe specific instruments.

The working group is currently scoring each instrument option on the following criteria:

- **Applicability to Pompe research:** Is this measure useful for Pompe research? Will the data captured meaningfully contribute to areas of research you are interested in conducting/reviewing?
- **Resolution of Pompe Experience:** What is the level of detail this insight provides across a range of progression?
- **Coverage / Uniqueness of Insight:** Are the insights provided through this measure unique and is this the best way to capture them? (For example, only the Pompe Disease Severity Scale captures "morning headache".)
- **Historical Importance:** Is the historical use of this measure (e.g., in multiple and/or important studies) significant enough that it should be included to provide continuity of data?

Instruments that are selected will be licensed and implemented on the RARE-X Platform

## How to participate/contribute

**Participate in research**  
 Visit <https://rare-x.org/pompe>



**Stay up to date**  
 Visit <https://PompeCommunity>



**Interested in an active role?**  
 Visit <https://PompeConsortium.org>



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