# Closing the loop for Fabry disease and Pompe disease in the UK (RARE)

**First published:** 07/02/2023

**Last updated:** 23/04/2024

Study Ongoing

## Administrative details

**EU PAS number**

EUPAS103426

**Study ID**

103466

**DARWIN EU® study**

No

**Study countries**

☐ United Kingdom

**Study description**

This study uses digital biomarkers in EMR data to support general practices with improving earlier diagnosis of patients with Fabry disease and Pompe disease help meet the objectives of the UK Rare Disease Framework and improve

patient outcomes.

**Study status**

Ongoing

# Research institutions and networks

## Institutions

### Observational & Pragmatic Research Institute Pte (OPRI)

United Kingdom

**First published:** 06/10/2015

**Last updated:** 19/08/2024

**Institution** | **Educational Institution** | **Laboratory/Research/Testing facility** | **ENCePP partner**

# Contact details

**Study institution contact**

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**Study contact**

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**Primary lead investigator**

David Price

Primary lead investigator

## Study timelines

**Date when funding contract was signed**
Actual: 14/12/2021

**Study start date**
Actual: 27/07/2022

**Data analysis start date**
Actual: 23/12/2022

**Date of interim report, if expected**
Planned: 07/02/2023

**Date of final study report**
Planned: 14/12/2023

## Sources of funding

- Non-for-profit organisation (e.g. charity)
- Other
- Pharmaceutical company and other private sector

## More details on funding

Optimum Patient Care Global Ltd, Sanofi Global, Volv Global

## Regulatory

**Was the study required by a regulatory body?**

No

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**Is the study required by a Risk Management Plan (RMP)?**

Not applicable

# Methodological aspects

## Study type

## Study type list

**Study type:**

Non-interventional study

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**Scope of the study:**

Disease epidemiology

Other

**If ‘other’, further details on the scope of the study**

Using digital biomarkers within EMR (OPCRD) to detect potentially undiagnosed patients with rare or difficult-to-diagnose disease in population-scale data

**Main study objective:**

The key objectives: 1. Ensuring patients get the right diagnosis faster 2. Increasing awareness of rare diseases among healthcare professionals 3. Better coordination of care

## Study drug and medical condition

**Medical condition to be studied**

Fabry's disease

Glycogen storage disease type II

# Population studied

**Age groups**

Children (2 to < 12 years)

Adolescents (12 to < 18 years)

Adults (18 to < 46 years)

Adults (46 to < 65 years)

Adults (65 to < 75 years)

Adults (75 to < 85 years)

Adults (85 years and over)

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**Estimated number of subjects**

19993084

# Study design details

**Outcomes**

The primary outcome of this QI study is to use digital biomarkers for the purpose of identifying patients at risk of rare and difficult to diagnose diseases and help clinicians to diagnose disease in population-scale data. (1) To provide educational material within the QI reports that targets the importance of diagnosing rare disease and includes specific content about individual cases. (2) To highlight patients at risk of the disease to GPs for consideration of further evaluation and investigation, therefore, helping patients to access the specific

and coordinated care and treatment that they need early on.

**Data analysis plan**

The QI programme runs the machine learning algorithm through EMR data to identify patients flagged as at risk of either Fabry disease or Pompe disease against 500 features within the EMR. The key analysis metrics: • Number of patients flagged at risk of Fabry or Pompe disease at baseline • Number of patients with a diagnosis of Fabry or Pompe disease at baseline • Number of patients flagged at risk of Fabry or Pompe disease at quarterly intervals • Number of patients with a diagnosis of Fabry or Pompe disease at quarterly intervals • Number of practices that opt in • Number of referrals made for a diagnosis The analysis runs the algorithm monthly on the EMR data to learn lessons from the initial cycle of QI and barriers are addressed by the study team and the programme adapted going forward. A learning healthcare system is created that combines cyclical QI reports for GPs for external validation and to generate knowledge that can be used to achieve change in practice.

# Data management

## ENCePP Seal

**Composition of steering group and observers**

Optimum Patient Care_QI Protocol_Rare Disease_Steering Committee Composition_September 2022_V1.1_CLEAN VERSION.pdf(123.81 KB)

## Data sources

**Data source(s)**

Optimum Patient Care Research Database

**Data sources (types)**

Disease registry

Electronic healthcare records (EHR)

## Use of a Common Data Model (CDM)

**CDM mapping**

No

## Data quality specifications

**Check conformance**

Unknown

**Check completeness**

Unknown

**Check stability**

Unknown

**Check logical consistency**

Unknown

## Data characterisation

## Data characterisation conducted

No