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: 20/08/2026

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

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

Not applicable

Methodological aspects

Study type

Study type list

Study type:

Non-interventional study

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

- Adolescents (12 to < 18 years)
- Children (2 to < 12 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)

Estimated number of subjects

19.993.084

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

The use of the ENCePP Seal has been discontinued since February 2025. The ENCePP Seal fields are retained in the display mode for transparency but are no longer maintained.

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