OPCRD ADEPT application Study Protocol

First published: 02/07/2024

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Study Finalised

Administrative details

EU PAS number

EUPAS1000000164

Study ID

1000000164

DARWIN EU® study

No

Study countries

United Kingdom

Study description

Rare diseases are individually uncommon, affecting less than 1 person in 2000, however with more than 6,000 diseases they are collectively common. A feature shared by many rare diseases is a long path to diagnosis, typically measured in years or even decades. During this ‘diagnostic odyssey’ patients

experience the many challenges of not having an accurate diagnosis; repeated investigations and referrals; a lack of explanation for their problems, and a lack of expert care and/or treatments. Further, until a diagnosis is made, affected individuals cannot benefit from the support of patient advocacy groups. MendelScan is a rare disease case finding tool that uses patients' GP records to identify patterns that suggest they may have an undiagnosed rare disease. Identified patient records are then reviewed and a targeted report returned to their GP for suggested next steps included. In this study we will use patients' GP records in the large primary care research database, OPCRD, to examine the performance of MendelScan for a range of diseases and use the database to support the development of other rare disease detection tools.

Study status

Finalised

Research institutions and networks

Institutions

Mendelian

Contact details

Study institution contact

Amanda Worker amanda@mendelian.co

Study contact

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Primary lead investigator

Hadley Mahon

Primary lead investigator

Study timelines

Date when funding contract was signed

Planned: 11/11/2022

Actual: 11/11/2022

Study start date

Planned: 11/11/2022

Actual: 11/11/2022

Data analysis start date

Planned: 11/11/2022

Actual: 11/11/2022

Date of final study report

Planned: 12/04/2024

Actual: 12/04/2024

Study protocol

OPCRD_protocol_amendment.pdf (162.68 KB)

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 topic:

Disease /health condition

Study type:

Non-interventional study

Scope of the study:

Hypothesis generation (including signal detection)

Method development or testing

Data collection methods:

Primary data collection

Study Design

Non-interventional study design

Case-control

Data management

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.

Data sources

Data source(s)

Optimum Patient Care Research Database

Use of a Common Data Model (CDM)

CDM mapping

No

Data quality specifications

Check conformance

Yes

Check completeness

Unknown

Check stability

Unknown

Check logical consistency

Unknown

Data characterisation

Data characterisation conducted

Not applicable