European Porphyria Registry

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Data source **Human** **Disease registry**

Administrative details

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PURI

https://redirect.ema.europa.eu/resource/47557

Data source ID

47557

Data source acronym

EPR

Data holder

Norwegian Porphyria Centre (NAPOS)

Data source type

Disease registry

Main financial support

European public funding

Funding by own institution

Care setting

Hospital inpatient care

Hospital outpatient care

Primary care – GP, community pharmacist level

Primary care – specialist level (e.g. paediatricians)

Secondary care – specialist level (ambulatory)

Data source qualification

If the data source has successfully undergone a formal qualification process (e.g., from the EMA, ISO or other certifications), this should be described.

No

Data source website

http://www.porphyriaregistry.org/

Contact details

Aasne K. Aarsand

Main

porfyri@helse-bergen.no

Data source regions and languages

Data source countries

Belgium

France

Italy

Netherlands

Norway

Spain

Sweden

United Kingdom

Data source languages

English

Data source establishment

Data source established

15/06/2012

Data source time span

First collection: 15/06/2012

The date when data started to be collected or extracted.

Publications

Data source publications

J.C. Deybach, S. Parker, M. Badminton, S. Sandberg. European Porphyria Network (EPNET) for information, epidemiological data, quality and equity of service. Orphanet J Rare Dis. 2010, 5(Suppl 1): P16.

Data elements collected

The data source contains the following information

Disease information

Does the data source collect information with a focus on a specific disease? This might be a patient registry or other similar initiatives.

Yes

Disease details

Porphyria

Rare diseases

Are rare diseases captured? In the European Union a rare disease is one that affects no more than 5 people in 10,000.

Yes

Pregnancy and/or neonates

Does the data source collect information on pregnant women and/or neonatal subpopulation (under 28 days of age)?

No

Hospital admission and/or discharge

Yes

ICU admission

Is information on intensive care unit admission available?

Yes

Cause of death

Captured

Prescriptions of medicines

Captured

Dispensing of medicines

Not Captured

Advanced therapy medicinal products (ATMP)

Is information on advanced therapy medicinal products included? A medicinal product for human use that is either a gene therapy medicinal product, a somatic cell therapy product or a tissue engineered products as defined in Regulation (EC) No 1394/2007 [Reg (EC) No 1394/2007 Art 1(1)].

No

Contraception

Is information on the use of any type of contraception (oral, injectable, devices etc.) available?

No

Indication for use

Does the data source capture information on the therapeutic indication for the use of medicinal products?

Not Captured

Medical devices

Is information on medicinal devices (e.g., pens, syringes, inhalers) available?

No

Administration of vaccines

No

Procedures

Does the data source capture information on procedures (e.g., diagnostic tests, therapeutic, surgical interventions)?

Not Captured

Healthcare provider

Is information on the person providing healthcare (e.g., physician, pharmacist, specialist) available? The healthcare provider refers to individual health professionals or a health facility organisation licensed to provide health care diagnosis and treatment services including medication, surgery and medical devices.

Yes

Clinical measurements

Is information on clinical measurements (e.g., BMI, blood pressure, height) available?

Yes

Genetic data

Are data related to genotyping, genome sequencing available?

Captured

Biomarker data

Does the data source capture biomarker information? The term “biomarker” refers to a broad subcategory of medical signs (objective indications of medical state observed from outside the patient), which can be measured accurately and reproducibly. For example, haematological assays, infectious disease markers or metabolomic biomarkers.

Captured

Patient-reported outcomes

Is information on patient-reported outcomes (e.g., quality of life) available?

No

Patient-generated data

Is patient-generated information (e.g., from wearable devices) available?

No

Units of healthcare utilisation

Are units of healthcare utilisation (e.g., number of visits to GP per year, number of hospital days) available or can they be derived? Units of healthcare utilisation refer to the quantification of the use of services for the purpose of preventing or curing health problems.

No

Unique identifier for persons

Are patients uniquely identified in the data source?

Yes

Diagnostic codes

Captured

Diagnosis / medical event vocabulary

ICD-10

Orphacode

Medicinal product information

Captured

Medicinal product information collected

Brand name

Quality of life measurements

Not Captured

Lifestyle factors

Captured

Lifestyle factors

Alcohol use

Diet

Tobacco use

Sociodemographic information

Not Captured

Quantitative descriptors

Population Qualitative Data

Population age groups

Paediatric Population (< 18 years)

Preterm newborn infants (0 – 27 days)

Term newborn infants (0 – 27 days)

Infants and toddlers (28 days – 23 months)

Children (2 to < 12 years)

Adolescents (12 to < 18 years)

Adults (18 to < 46 years)

Adults (46 to < 65 years)

Elderly (≥ 65 years)

Adults (65 to < 75 years)

Adults (75 to < 85 years)

Adults (85 years and over)

Population

Population size

289

Data flows and management

Access and validation

Biospecimen access

Are biospecimens available in the data source (e.g., tissue samples)?

No

Access to subject details

Can individual patients/practitioners/practices included in the data source be contacted?

Yes

Description of data collection

EPR participants are recruited by the International Porphyria Network's (Ipnet, former Epnet) porphyria centres of expertise or by physicians with a special interest in porphyria. Participants were recruited when their diagnosis was established (symptomatic or as part of predictive testing), for treatment and for follow-up. Data for AIP patients was collected until 2019 during regular and emergency visits to the hospital or primary health care. The registry recorded data such as gender, date of birth, porphyria diagnosis, laboratory test results, symptoms and treatment. Physicians were able to enter consecutive visits for the same participant. Recruitment is presently not ongoing

Event triggering registration

Event triggering registration of a person in the data source

Disease diagnosis

Data source linkage

Linkage

Is the data source described created by the linkage of other data sources (prelinked data source) and/or can the data source be linked to other data source on an ad-hoc basis?

No

Data management specifications that apply for the data source

Informed consent for use of data for research

Required for general use

Possibility of data validation

Can validity of the data in the data source be verified (e.g., access to original medical charts)?

Yes

Data source preservation

Are records preserved in the data source indefinitely?

Yes

Approval for publication

Is an approval needed for publishing the results of a study using the data source?

No

Common Data Model (CDM) mapping

CDM mapping

Has the data source been converted (ETL-ed) to a common data model?

No