

Norwegian Porphyria Registry

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Data source

Human

Disease registry

Administrative details

Administrative details

Data source ID

47555

Data source acronym

Norwegian Porphyria Registry

Data holder

[Norwegian Porphyria Centre \(NAPOS\)](#)

Data source type

Disease registry

Main financial support

National, regional, or municipal public funding

Other

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

<https://www.kvalitetsregistre.no/registeroversikt/norsk-porfyriregister/>

Contact details

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Main

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Data source regions and languages

Data source countries

Norway

Data source languages

Norwegian

Data source regions

Agder
Innlandet
Møre og Romsdal
Nordland
Oslo
Rogaland
Troms og Finnmark
Trøndelag
Vestfold og Telemark
Vestland
Viken

Data source establishment

Data source established

15/06/2002

Data source time span

First collection: 15/06/2002

The date when data started to be collected or extracted.

Publications

Data source publications

[Health-related quality of life in porphyria cutanea tarda: a cross-sectional registry based study. Andersen J, Thomsen J, Enes ÅR, Sandberg S, Aarsand AK. Health Qual Life Outcomes. 2020 Mar 30,18\(1\):84.](#)

[Porphyria cutanea tarda increases risk of hepatocellular carcinoma and premature death: a nationwide cohort study. Baravelli CM, Sandberg S, Aarsand AK, Tollånes MC. Orphanet J Rare Dis. 2019 Apr 3,14\(1\):77.](#)

Sick leave, disability, and mortality in acute hepatic porphyria: a nationwide cohort study. Baravelli CM, Aarsand AK, Sandberg S, Tollånes MC. Orphanet J Rare Dis. 2020 Feb 21,15(1):56.

Porphyria cutanea tarda and patterns of long-term sick leave and disability pension: A 24-year nationwide matched cohort study. • Baravelli, C.M., Aarsand, A.K., Sandberg, S., Tollånes, M.C. Orphanet J Rare Dis. May 4,17(1):180. (2022).

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 (other)

The Norwegian porphyria register has the following inclusion criteria: • Everyone who has a confirmed diagnosis (active) or by a predictive test has been shown to have a genetic predisposition (latent disease) for one of the porphyria diseases: o acute intermittent porphyria – AIP (ORPHA: 79276, ICD-10: E.80.2) o porphyria variegata – PV (ORPHA: 79273, ICD-10: E.80.2) o hereditary coproporphyria – HCP (ORPHA: 79473, ICD-10: E80.2) • Anyone who has a confirmed diagnosis of one of the porphyria diseases: o porphyria cutanea tarda – PCT (ORPHA: 101330, ICD-10: E.80.1) o erythropoietic protoporphyria – EPP (ORPHA: 79278, ICD-10: E80.0) Some of the participants with acute porphyria have had a liver transplant and have practically been cured of the

disease. These are not included in the analyses.

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

No

ICU admission

Is information on intensive care unit admission available?

No

Cause of death

Not 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)?

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?

Yes

Patient-generated data

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

Yes

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

Captured

Quality of life measurements vocabulary

other

Quality of life measurements, other

RAND-12

Lifestyle factors

Captured

Lifestyle factors

Alcohol use

Diet

Tobacco use

Sociodemographic information

Captured

Sociodemographic information collected

Age

Education level

Gender

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 (\geq 65 years)

Adults (65 to < 75 years)

Adults (75 to < 85 years)

Adults (85 years and over)

Estimated percentage of the population covered by the data source in the catchment area

95%

Description of the population covered by the data source in the catchment area whose data are not collected (e.g., people who are registered only for private care)

Nation-wide

Population

Population size

1394

Active population size

1160

Data flows and management

Access and validation

Biospecimen access

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

Yes

Access to subject details

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

Yes

Description of data collection

The data collection consists of questionnaires that the participants complete themselves, and from 2015 also questionnaires that are completed by the treating physician, as well as some specific variables retrieved from laboratory information systems and health records (data validation). Information is collected on diagnosis, symptoms, treatment, control and follow-up, laboratory results, as well as other health information, socio-demographic variables, quality of life and lifestyle. The variables registered in the Norwegian porphyria register are the result of many years of work in the porphyria field, both nationally and internationally.

Event triggering registration

Event triggering registration of a person in the data source

Disease diagnosis

Other

Event triggering registration of a person in the data source, other

Patients filling in a questionnaire

Event triggering de-registration of a person in the data source

Other

Event triggering de-registration of a person in the data source, other

If the person refuse to participate (active reservation).

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?

Yes

Linkage description, possible linkage

Data can be linked to other sources using the Norwegian national identity number.

Linked data sources

Pre linked

Is the data source described created by the linkage of other data sources?

No

Data source, other

Central Norwegian health registers (such as the Medical Birth Register or the Cancer Register)

Linkage variable

Data can be linked to other sources using the Norwegian national identity number.

Pre linked

Is the data source described created by the linkage of other data sources?

No

Data source, other

Other medical quality registers

Linkage variable

Data can be linked to other sources using the Norwegian national identity number.

Pre linked

Is the data source described created by the linkage of other data sources?

No

Data source, other

Registers located at Statistics Norway (SSB)

Linkage variable

Data can be linked to other sources using the Norwegian national identity number.

Pre linked

Is the data source described created by the linkage of other data sources?

No

Data source, other

The General Practitioner Register (HELFO)

Linkage variable

Data can be linked to other sources using the Norwegian national identity number.

Pre linked

Is the data source described created by the linkage of other data sources?

No

Data source, other

The Norwegian Labor and Welfare Administration (NAV)

Linkage variable

Data can be linked to other sources using the Norwegian national identity number.

Data management specifications that apply for the data source

Data source refresh

Yearly

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?

Yes

Common Data Model (CDM) mapping

CDM mapping

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

No