REGISTRY: a study by the European Huntington’s Disease Network

First published: 01/02/2024

Last updated: 17/10/2024

Data source

Human

Biobank

Disease registry

Administrative details

Administrative details

Data source ID

30505

Data source acronym

EHDN - REGISTRY

Data holder

University Hospital of Ulm

Data source type

Biobank

Disease registry

Main financial support

Funds from patients organisations, charity and foundations

Care setting

Hospital inpatient care

Hospital outpatient care

Other

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.ehdn.org

Contact details

Katrin Barth katrin.barth@uni-ulm.de

Main

katrin.barth@uni-ulm.de

Data source regions and languages

Data source countries

Austria

Belgium

Czechia

Denmark

Finland

France

Germany

Italy

Netherlands

Norway

Poland

Portugal

Russian Federation

Slovenia

Sweden

Switzerland

United Kingdom

Data source languages

English

Data source establishment

Data source established

15/06/2004

Data source time span

First collection: 17/08/2004

The date when data started to be collected or extracted.

Last collection: 15/08/2017

If data collection in the data source has ceased, the date new records last entered the data source.

Publications

Data source publications

Jacobs M, Hart EP, van Zwet EW, Bentivoglio AR, Burgunder JM, Craufurd D, Reilmann R, Saft C, Roos RA REGISTRY investigators of the European Huntington’s Disease Network. Progression of motor subtypes in Huntington’s disease: a 6-year follow-up study. J Neurol. 2016 Oct263(10):2080-5. doi: 10.1007/s00415-016-8233-x. Epub 2016 Jul 19. PMID: 27435968

Genetic Modifiers of Huntington’s Disease (GeM-HD) Consortium. Identification of Genetic Factors that Modify Clinical Onset of Huntington’s Disease. Cell. 2015 Jul 30 162(3):516-26. doi: 10.1016/j.cell.2015.07.003. PMID: 26232222

Moss DJH, Pardiñas AF, Langbehn D, Lo K, Leavitt BR, Roos R, Durr A, Mead S TRACK-HD investigators REGISTRY investigators, Holmans P, Jones L, Tabrizi SJ. Identification of genetic variants associated with Huntington’s disease progression: a genome-wide association study. Lancet Neurol. 2017 Sep16(9):701-711. doi: 10.1016/S1474-4422(17)30161-8. Epub 2017 Jun 20. PMID: 28642124

van Duijn E, Craufurd D, Hubers AA, Giltay EJ, Bonelli R, Rickards H, Anderson KE, van Walsem MR, van der Mast RC, Orth M, Landwehrmeyer GB the European Huntington’s Disease Network Behavioural Phenotype Working Group. Neuropsychiatric symptoms in a European Huntington’s disease cohort (REGISTRY). J Neurol Neurosurg Psychiatry. 2014 Dec 85(12):1411-18. doi: 10.1136/jnnp-2013-307343. Epub 2014 May 14. PMID: 24828898

Sun Z, Li Y, Ghosh S, Cheng Y, Mohan A, Sampaio C, Hu J. A Data-Driven Method for Generating Robust Symptom Onset Indicators in Huntington’s Disease Registry Data. AMIA Annu Symp Proc. 2018 Apr 162017:1635-1644. eCollection 2017. PMID: 29854234

Studies

List of studies that have been conducted using the data source

Survey on the collection of data on adverse events related to medicinal products through registries

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

Huntington's disease

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

Captured

Cause of death vocabulary

Not coded (Free text)

Prescriptions of medicines

Not Captured

Dispensing of medicines

Captured

Dispensing vocabulary

ATC

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)].

Yes

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?

Captured

Indication vocabulary

MedDRA

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.

No

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

Genetic data vocabulary

Other

Genetic data vocabulary, other

Not coded (free text)

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.

Not 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

Read

Medicinal product information

Captured

Medicinal product information collected

Active ingredient(s)

Brand name

Dosage regime

Dose

Route of administration

Medicinal product vocabulary

WHO Drug

Quality of life measurements

Captured

Quality of life measurements vocabulary

SF-36

Lifestyle factors

Captured

Lifestyle factors

Alcohol use

Other

Tobacco use

Sociodemographic information

Captured

Sociodemographic information collected

Age

Education level

Ethnicity

Gender

Living in rural area

Marital status

Type of residency

Quantitative descriptors

Population Qualitative Data

Population age groups

Paediatric Population (< 18 years)

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)

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

Estimation of population covered by the data source: 22%

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)

N/A

Family linkage

Family linkage available in the data source permanently or can be created on an ad hoc basis

Ad hoc

Population

Population size

14777

Population by age group

Age group	Population size

Paediatric Population (< 18 years)	74
Children (2 to < 12 years)	10
Adolescents (12 to < 18 years)	64
Adults (18 to < 46 years)	5792
Adults (46 to < 65 years)	6535
Elderly (≥ 65 years)	2376
Adults (65 to < 75 years)	1782
Adults (75 to < 85 years)	535
Adults (85 years and over)	59

Median observation time

Median time (years) between first and last available records for unique individuals captured in the data source

2.00

Data flows and management

Access and validation

Governance details

Documents or webpages that describe the overall governance of the data source and processes and procedures for data capture and management, data quality check and validation results (governing data access or utilisation for research purposes).

http://www.ehdn.org/hd-clinicians-researchers/data-mining-projects-2/

Biospecimen access

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

Yes

Biospecimen access conditions

All details relating to biosamples access procedures are provided on the EHDN website (https://www.euro-hd.net/html/projects/proposals/scipro)

Access to subject details

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

No

Description of data collection

Data was collected either as a paper CRF and entered onto an online EDC system or entered directly into the EDC

Event triggering registration

Event triggering registration of a person in the data source

Other

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

Consent to study participation

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

Death

Loss to follow up

Other

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

Participant request

Event triggering creation of a record in the data source

Specialist visit

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

A unique 9 digit identifier (HDID) is assigned to each participant. The HDID is used in multiple studies of Huntington's disease to link datasets (e.g Enroll-HD, Track-HD, clinical trials)

Linked data sources

Pre linked

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

No

Data source, other

Enroll-HD

Linkage strategy

Deterministic

Linkage variable

Unique 9 digit identifier (HDID)

Linkage completeness

Complete

Pre linked

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

No

Data source, other

Track-HD

Linkage strategy

Deterministic

Linkage variable

Unique 9 digit identifier (HDID)

Linkage completeness

Complete

Data management specifications that apply for the data source

Data source refresh

Yearly

Informed consent for use of data for research

Other

Possibility of data validation

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

No

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

Informed consent, other

All data have been consented by participants for Huntington's Disease research and development.

Data source last refresh

15/08/2017

Common Data Model (CDM) mapping

CDM mapping

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

No