European registry and network for intoxication type metabolic diseases

First published: 01/02/2024

Last updated: 17/10/2024

Data source | Human | Disease registry | Other

Administrative details

Administrative details

PURI

https://redirect.ema.europa.eu/resource/46312

Data source ID

46312

Data source acronym

E-IMD

Data holder

European registry and network for Intoxication type Metabolic Diseases (E-IMD)

Data source type

Disease registry

Other

Data source type, other

Post-authorisation safety studies (PASS) database

Main financial support

European public funding

Care setting

Hospital inpatient care

Hospital outpatient care

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.eimd-registry.org/

Contact details

Stefan Kölker

Main

stefan.koelker@med.uni-heidelberg.de

Data source regions and languages

Data source countries

Austria

Belgium

Croatia

Czechia

Denmark

France

Germany

Hungary

Italy

Netherlands

Poland

Portugal

Spain

Switzerland

Taiwan

United Kingdom

United States

Data source languages

English

Data source establishment

Data source established

14/06/2011

Data source time span

First collection: 14/06/2011

The date when data started to be collected or extracted.

Publications

Data source publications

Early prediction of phenotypic severity in Citrullinemia Type 1

From genotype to phenotype: Early prediction of disease severity in argininosuccinic aciduria

Networking Across Borders for Individuals with Organic Acidurias and Urea Cycle Disorders: The E-IMD Consortium

The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 1: the initial presentation

The phenotypic spectrum of organic acidurias and urea cycle disorders. Part 2: the evolving clinical phenotype

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

Analysis of patients with arginase 1 deficiency treated with Loargys in standard clinical care enrolled in a European, non-interventional, multicentre registry

A European, non-interventional, multicentre, registry-based post-authorisation safety study to evaluate the long-term safety of Loargys treatment in arginase 1 deficiency patients in standard clinical care

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)

Organic acidurias: Methylmalonic aciduria, Propionic aciduria, Isovaleric aciduria, Glutaric aciduria type 1 Urea cycle defects: N-Acetylglutamate synthase deficiency, Carbamylphosphate synthetase 1 deficiency, Ornithine transcarbamylase deficiency, Citrullinemia type 1 / Argininosuccinate synthetase deficiency, Argininosuccinate lyase deficiency, Arginase 1 deficiency, Hyperornithinemia-hyperammonemia-homocitrullinuria (HHH) syndrome / mitochondrial ornithine transporter 1 deficiency

---

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)?

Yes

Hospital admission and/or discharge

Yes

ICU admission

Is information on intensive care unit admission available?

Yes

Cause of death

Captured

Cause of death vocabulary

ICD-10

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?

Yes

Indication for use

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

Captured

Indication vocabulary

ICD-10

Medical devices

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

Yes

Administration of vaccines

Yes

Procedures

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

Captured

Procedures vocabulary

ICD-10

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

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.

Captured

Biomarker data vocabulary

Other

Biomarker vocabulary, other

Not coded (free text)

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?

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

Medicinal product information

Captured

Medicinal product information collected

Active ingredient(s)

Dosage regime

Dose

Route of administration

Medicinal product vocabulary

Not coded (Free text)

Quality of life measurements

Captured

Quality of life measurements vocabulary

other

WHOQOL

Quality of life measurements, other

PedsQoL

Lifestyle factors

Not Captured

Sociodemographic information

Captured

Sociodemographic information collected

Age

Country of origin

Education level

Ethnicity

Gender

Marital status

Socioeconomic status

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)

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

We estimate 89.6% of the population in 28 EU MS according to Eurostat 2013 (see also doi: 10.1007/8904_2015_408)

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)

Regional sub-set - E-IMD members are metabolic specialist centres with varying degrees of coverage of the respective countries.

Family linkage

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

Permanently

Family linkage available between the following persons

Father-child

Mother-child

Sibling

Population

Population size

1435

Active population size

1435

Population by age group

Age group	Population size	Active population size
Paediatric Population (< 18 years)	1046	1046
Preterm newborn infants (0 – 27 days)	19	19
Term newborn infants (0 – 27 days)	19	19
Infants and toddlers (28 days – 23 months)	132	132
Children (2 to < 12 years)	603	603
Adolescents (12 to < 18 years)	292	292
Adults (18 to < 46 years)	361	361
Adults (46 to < 65 years)	22	22
Elderly (≥ 65 years)	6	6
Adults (65 to < 75 years)	3	3
Adults (75 to < 85 years)	3	3
Adults (85 years and over)	0	0

Median observation time

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

4.96

Median time (years) between first and last available records for unique active individuals (alive and currently registered) capt

4.96

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

Observational clinical data from regular patient care is entered into a web-based registry.

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

Enrollment by the treating health care provider. Patient has to be affected by target disease and sign an informed consent (parents/legal guardians in case of underage patients).

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

Patient withdrawing consent.

Event triggering creation of a record in the data source

Visist are created according to the regular follow-up of a patient as decided by the treating physician.

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

In case of UCDC the linkage strategy was deterministic since the samples are strictly geograhpically seperated. In case of E-HOD / U-IMD the process would be deterministic due to overlapping consortia with local ID code lists.

Linked data sources

Pre linked

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

No

Data source, other

Data source could in principle be linked to other data sources, provided a linkage mechanism (e.g. Spider) exists and is legally and technically feasible. The data source was succesfully merged to the UCDC dataset (doi: 10.1002/jimd.12031). Data source can also be merged with E-HOD (https://www.ehod-registry.org/) and U-IMD (https://u-imd-registry.org/). Other data source would need previous evaluation.

Linkage strategy

Deterministic

Linkage variable

Personal identifier.

Linkage completeness

Complete for the above mentioned.

Data management specifications that apply for the data source

Data source refresh

Yearly

Informed consent for use of data for research

Required for all studies

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?

No

Data source preservation length (years)

10 years

Approval for publication

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

Yes

Data source last refresh

30/05/2023

Common Data Model (CDM) mapping

CDM mapping

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

Yes

CDM Mappings

CDM name (other)

EU CDE

Data source ETL specifications (link)

https://eu-rd-platform.jrc.ec.europa.eu/set-of-common-data-elements_en

CDM name (other)

ICD10 Codes