Galactosemia Patient Registry

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

Administrative details

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PURI

https://redirect.ema.europa.eu/resource/18473

Data source ID

18473

Data source acronym

GalNet

Data holder

Maastricht University Medical Center (MUMC)

Data source type

Disease registry

Main financial support

Funds from patients organisations, charity and foundations

Care setting

Hospital outpatient care

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.galactosemianetwork.org

Contact details

M. Estela Rubio-Gozalbo

Main

estela.rubio@mumc.nl

Data source regions and languages

Data source countries

Australia

Austria

Belgium

Croatia

Estonia

France

Germany

Greece

Ireland

Israel

Italy

Lithuania

Netherlands

Portugal

Spain

Switzerland

United Kingdom

United States

Data source languages

English

Data source establishment

Data source established

01/01/2014

Data source time span

First collection: 01/01/2014

The date when data started to be collected or extracted.

Publications

Data source publications

The galactosemia network (GalNet)

International clinical guideline for the management of classical galactosemia: diagnosis, treatment, and follow-up

Galactokinase deficiency: lessons from the GalNet registry

The natural history of classic galactosemia: lessons from the GalNet registry

Galactose epimerase deficiency: lessons from the GalNet registry

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 database collects information about the disease Galactosemias type I, II and III. Information collected about demographics, neonatal period, general follow-up, brain follow-up, gonads and reproduction follow-up, bone health follow-up and diet.

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

No

ICU admission

Is information on intensive care unit admission available?

No

Cause of death

Not Captured

Prescriptions of medicines

Not 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

Procedures vocabulary

Orphacode

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

Data on 3 different genes: GALT, GALK and GALE. The pathogenic variants in the given are described, eg. p.Q188R or p.Gln188Arg homozygous in the GALT gene

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

The data source uses Gal-1-P in erythrocytes as biomarker: mg/dl or umol/gram Hb

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?

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?

No

Diagnostic codes

Captured

Diagnosis / medical event vocabulary

Orphacode

Medicinal product information

Captured

Medicinal product information collected

Formulation

Medicinal product vocabulary

Other

If 'other,' what vocabulary is used?

internal code

Quality of life measurements

Not Captured

Lifestyle factors

Captured

Lifestyle factors

Diet

Frequency of exercise

Sociodemographic information

Captured

Sociodemographic information collected

Education level

Ethnicity

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 (≥ 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

Rare diseases, the most common has a prevalence of 1:18.000 - 1:50.000

Median age of total unique individuals with records captured in the data source: 18 years (0-65y)

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)

World-wide

Population

Population size

672

Active population size

672

Median observation time

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

9.00

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

9.00

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).

https://www.galactosemianetwork.org/registry

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

Staff is trained on forehand to collect the data in registry. Data is collected from medical files after informed consent of patients.

Event triggering registration

Event triggering registration of a person in the data source

Disease diagnosis

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

Withdrawal from study

Event triggering creation of a record in the data source

After confirmed diagnosis of galactosemia medical data is updated in case of new findings or to add the most recent information.

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

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?

Yes

Approval for publication

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

No

Data source last refresh

20/04/2023

Common Data Model (CDM) mapping

CDM mapping

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

No