European Registry of Patients with McArdle disease or other rare form of muscle Glycogenoses

First published: 01/02/2024

Last updated: 17/10/2024

Data source | Outdated | Human | Disease registry

Administrative details

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Data source ID

18559

Data source acronym

EUROMAC

Data holder

University Hospital Vall d’Hebron (HUVH)

Data source type

Disease registry

Main financial support

Other

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.euromacregistry.eu/portal1/h_index.php

Contact details

Tomas Pinos Figueras tomas.pinos@vhir.org

Main

tomas.pinos@vhir.org

Data source regions and languages

Data source countries

Denmark

France

Germany

Greece

Italy

Netherlands

Portugal

Spain

Türkiye

United Kingdom

Data source languages

English

Data source establishment

Data source time span

First collection: 01/09/2015

The date when data started to be collected or extracted.

Publications

Data source publications

Report on the EUROMAC McArdle Exercise Testing Workshop, Madrid, Spain, 11–12 July 2014Ros Quinlivan, Alejandro Lucia, Renata S. Scalco, Alfredo Santalla, Jatin Pattni, Richard Godfrey, Ramon Martion behalf of the Workshop Participants

Development of diagnostic criteria and management strategies for McArdle Disease and related rare glyco(geno)lytic disorders to improve standards of care

Chronic Fatigue and Rhabdomyolysis p453-460In Inherited Metabolic Disease in Adults 1st Edition Oxford University Press 2016R Quinlivan, P Laforet

Skeletal Muscle Disorders of Glycogenolysis and GlycolysisNature Reviews Neurology. 12(7):393-402, 2016 Jul.Godfrey R, Quinlivan R

Creation and implementation of a European registry for patients with McArdle disease and other muscle glycogenoses (EUROMAC registry).

Studies

No information provided.

Data elements collected

The data source contains the following

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)

McArdle disease and other very rare mucle glycogenosis presenting with exercise intolerance

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

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)?

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?

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

Medicinal product information

Captured

Medicinal product vocabulary

Other

If 'other,' what vocabulary is used?

internal code

Quality of life measurements

Not Captured

Lifestyle factors

Not Captured

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)

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

313

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

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

Quarterly

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)?

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

Informed consent, other

There is a committee to evaluate requests for data access

Common Data Model (CDM) mapping

CDM mapping

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

No