# 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

(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

### **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



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).

# 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)

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.

Unique	identifier	for	persons
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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