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European registry of patients with McArdle disease and very rare muscle glycogenolytic disorders (MGD) with exercise intolerance as the major symptom (PR-MDMGD)

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General objectives

General objective: The implementation of an EU registry of patients with McArdle disease and very rare muscle glycogenolytic disorders (MGD) with exercise intolerance as the major symptom (PR-MDMGD). A web portal, with public and intranet access will support the registry and the dissemination of information on these rare diseases. The main aim of this project will be to improve health outcomes while at the same time reducing health and social care costs by disseminating knowledge. In these patients delayed diagnosis and incorrect advice leads to debilitating symptoms and increased risk of life-threatening crises necessitating admission to critical care. All of which can be prevented by correct medical care.

Strategic relevance & contribution to the programme: The project will address the lack of data available on diagnosis and treatment of McArdle disease and MGD. By registering patient data from at least 8 European countries and moving towards a wider global patient registry, access and quality of care for patients across Europe will be improved which is highly relevant to the EU.

Strategic relevance and contribution to the public health programme

EUROMAC addresses the current lack of data at EU level on treatment and care of McArdle disease and other MGD. By registering patient data, covering at least 8 European countries, and moving towards a further global patient registry on this level, it highly complies with the EU strategy on rare diseases. Bringing different key actors together to develop a key patient registry together with important and innovative dissemination action, the consortium aims at not only building a static registry, but one that could be reproduced for other diseases. The way the consortium is built, the involved groups, the geographic and societal coverage can set an example for future registries. The utility of the web portal clearly aims at a registry that can be sustainable in the future, through its appropriation by users (all key stakeholders) and the involvement of future partners and governmental bodies to keep it going.

Methods and means

EUROMAC registry will be based on the principle of "indirectly identifiable data collection". This means that all data will be anonymised for research purposes, but it will be linked to their patient sources through the use of an encrypted algorithm that will be only revealed under the specific conditions and described in the consent form. A specific Ethical Committee (EthC) for dealing with legal issues on anonymity and confidentiality, and ethical use of patient’s data will establish specific instructions to ensure that the implementation of the registry will follow the national and European legal framework. The registry will be developed according to the following methods:
i) Implementation Plan; ii) Registry Documentation; iii) Data Collection; iv) Data Access Policy and v) Data Dissemination.

Expected outcomes period

Generation of a database of patient groups and concerned professionals throughout the EU, including data available in all languages of the participating countries. This specific outcome (the core of the project) will allow us to get the following related expected outcomes: Improving access of patients to specialized care and registry entrance; enable the physicians and researchers working in the field of the glycogenoses to improve the knowledge of the natural history of McArdle Disease and other glycogenose; facilitate the involvement of local governments and regulatory agencies as well as the pharmaceutical industry; Implementation of training courses and patient support groups meetings and other impact outcomes described in Annex 1A.
COORDINATOR

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D01 - Midterm report
FUNDACIO HOSPITAL UNIVERSITARI VALL D"HEBRON- INSTITUT DE RECERCA
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D02 - Final report
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D03 - WEB Database
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D04 - Web portal of the registry with public access
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D05 - Web portal with intranet access
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D06 - Evaluation reports
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D07 - Quality assessment report
FUNDACIO HOSPITAL UNIVERSITARI VALL D'HEBRON- INSTITUT DE RECERCA
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D08 - Scientific report on MGD
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D10 - Ethical report
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