UNIVERSITY MEDICAL CENTER GRONINGEN EXPRESSION OF INTEREST
FOR APPLICATION AS A PARTNER

On behalf of Terry Derks, we would like to express interest to cooperate as a partner in the HORIZON-HLTH-2021-TOOL-06-03: More information about the researchers and the UMCG can be found below. For questions and contact initiation, please email grantsupport@umcg.nl.

Horizon Europe, Work Programme 2021-2022 (HEALTH)
Call topic: "HORIZON-HLTH-2022-DISEASE-06-04-two-stage ex-Topic 3.6 Development of new effective therapies for rare diseases
Deadline: September 21, 2021

Partner information

Country	THE NETHERLANDS
Name of the organisation	UNIVERSITY MEDICAL CENTER GRONINGEN (UMCG)
Type of organisation	Research Organisation
Short description	The University Medical Center Groningen (UMCG) is one of the largest hospitals in the Netherlands and the largest employer in the Northern Netherlands. More than 10,000 employees provide patient care, are involved in medical education and perform cutting-edge scientific research, focused on 'healthy and active ageing'. Research and education at the UMCG are funded through the University of Groningen, and the Faculty of Medical Sciences functions as an integral part of the University. UMCG is ISO9181-certified for its research and education activities. The center provides excellent facilities to work with large databases, including the LifeLines cohort that was initiated at the UMCG. These facilities include both digital infrastructures as well as methodological expertise.
Department / laboratory	Pediatrics metabolic diseases
Name(s) of the principal investigator(s)	Terry Derks

Department involved
The research group of Terry Derks are experts in the field of inherited metabolic diseases and more specifically Glycogen Storage Diseases and Fatty Acid Oxidation Disorders, all (ultra) rare diseases. Terry coordinates the UMCG Center of Expertise for Carbohydrate, Fatty Acid Oxidation and Ketone Bodies Disorders. The center of expertise is involved in fundamental, translational and clinical research, including multiple clinical trials (e.g., NCT02318966, NCT03011203, NCT03517085, NCT03970278, NCT03761693, and NCT04311307). He is a renowned international expert in this field, one of the coordinators of the Carbohydrate, fatty acid oxidation and ketone bodies disorders (C-FAO) subnetwork within the European Reference Network for Hereditary Metabolic Disorders (MetabERN). He has initiated and led the priority setting partnership for liver GSD and initiated www.emergencyprotocol.net.

The group of experts identified the following clinical, scientific and societal challenges to deliver high-quality health care to GSD patients:

A. Traditional health care does not fit, as it is focused on local delivery of care, whereas individuals with rare diseases usually do not live close to expert health care providers. Patients are relatively few and scattered across populations.

B. The number of disease experts and corresponding centers of expertise is not enough.
C. There is unwanted variation in diagnosis and treatment. Patients are underdiagnosed (due to their individual ultra-rarity and low clinical awareness), causing doctor's delay in diagnosis, unnecessary complications, and even death of patients.
D. There is large heterogeneity among patients with identical GSD subtypes and sometimes even identical genotypes.
E. Diagnostic and/or prognostic biomarkers are missing.
F. There is limited understanding of pathophysiology and treatment.
G. Exact numbers on epidemiology, natural history and mortality are not known.
H. Registered treatments are unavailable, thus leading to poor outcomes.
I. Evidence-based medicine is challenging given the small patient numbers and variable phenotypes.
J. Therapeutic effects are often challenging to measure, and the need for person centred outcomes exists.
K. Treatment access is challenging, given slow regulatory processes and reimbursement issues especially for nutritional products and high prices for enzyme and genetic treatments.
L. Partnership between relevant stakeholders is limited.
M. Expansion of population newborn screening panels is challenging, and slow and continuous evaluation is not embedded in the regular care.
N. The research and clinical expertise on GSD are very fragmented and confined to personal interest of a few experts, and interaction between patients and other stakeholders is not guaranteed in Europe (and beyond).

The group is now preparing for a COST action for the development of international research network(s) for Glycogen Storage Diseases (Rare Diseases). They are creating a strong and capable multidisciplinary stakeholder network, including health care providers, patients, scientists, and medical food and pharmaceutical companies. The network will promote (1) telehealth; (2) integrating of care and research; and (3) improving stakeholder collaborations.

The focus is on (1) defining person-centred outcomes; (2) improving interaction between health care providers and families during acute hospitalizations; (3) coordinating the research roadmap towards better clinical trials; (4) providing networking opportunities for early-stage researchers and other talented professionals and learning cycles; and (5) developing a platform for sharing best practices.

The aim of this group is to increase the quality of life of the affected individuals and their families (personal value), and create healthcare value improvements at technical, allocative and societal levels. The group of Terry Derks is also interested in furthering the research on GSD to improve high-quality health care to GSD patients.