ASSOCIATED PARTNERS

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Všeobecná fakultní nemocnice v Praze	Viktor Kozich (Dissemination)	Prague, Czech Republic
Ospedale Pediatrico Bambino Gesu	Carlo Dionisi-Vici (Evaluation)	Rome, Italy
Universitätsklinikum	Thomas Opladen (Registry) lead)	Heidelberg, Germany
Hospital Sant Joan de Deu	Angels Garcia-Cazoria (INTD)	Barcelona, Spain
Udine University Hospital	Maurizio Scarpa (MetabERN)	Udine, Italy

COLLABORATING STAKEHOLDERS

MetabERN	http://metab.ern-net.eu
ERKNet	https://www.erknet.org/index.php?id=home
E-IMD	http://www.e-imd.org/en/index.phtml
E-HOD	http://www.e-hod.org
iNTD	http://www.intd-online.org
ePAG Belgian Patient Organisation (BOKS)	http://metab.ern-net.eu/patient-association/
Gaucher Association UK	https://www.gaucher.org.uk

How to participate

U-IMD is the official registry of MetabERN but is in principle open to all European and international health care providers, treating patients with IMDs. U-IMD is designed as an observational non-interventional registry study, requiring members to obtain an ethics vote according to local standards for the U-IMD study protocol and to sign the U-IMD collaboration agreement.

Supporting information and material can be obtained by contacting U-IMD via the project or registry website.

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European Reference Network | MetabERN

Do you need more information?

Please, contact us by via the contact form on the project website www.u-imd.org

U-IMD project | www.u-imd.org

U-IMD registry | https://u-imd-registry.org

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U-IMD

Unified Registry for Inherited Metabolic Diseases

U-IMD is the acronym for the **Unified European Registry for Inherited Metabolic Disorders**. The overall aim of this project is to promote health for children, adolescents and adults affected with rare Inherited Metabolic Disorders (IMDs) using an open multiple stakeholder approach.

What are Inherited Metabolic Diseases (IMDs)?

IMDs are a class of rare genetic diseases. Defects in genes that code for enzymes interfere with the normal functioning of the metabolism of affected patients. More than 1400 IMDs have thus far been identified. Each IMD on itself is a rare condition with estimated individual prevalence ranging from 0.1 to 15 in 100,000 newborns. Taken together patients affected by IMDs are numerous with at least one in 500 newborns.

Depending on the underlying defect and its individual severity, the spectrum of clinical presentation of IMDs is wide, ranging from involvement of single organ systems to multi-systemic disease. Individuals affected by IMDs are confronted with significant and often severe health problems resulting in high morbidity, reduced life expectancy, and low quality of life.

Data Ownership

The data gathered in the U-IMD registry is pseudonymized, using local pseudonymization. Members can only access their own files and always retain full data ownership. Usage of the entire U-IMD dataset is governed by the U-IMD Consortium, granting members equal rights in initiating and deciding on mutual projects.

MetabERN

MetabERN is a European non-profit network established by the EU to facilitate access to the best available care and address the needs across the border of all patients affected by any rare inherited metabolic disease (IMDs) and their families. MetabERN is driven by the principle of patient-centred care for the provision of its services aiming at improving the quality of life of patients and families. MetabERN aims to connect the most specialised centres in the area of rare IMDs to promote prevention, accelerate diagnosis and improve standards of care across Europe for patients living with IMDs. MetabERN is entirely patient and expert-led. Through the combination of patient experience and expert knowledge from across the EU, it captures the most innovative medical advances and tailors them to patient needs.

Disease Spectrum covered by MetabERN and U-IMD

The U-IMD registry will cover all MetabERN IMD subgroups:

- Amino acid-related disorders (AOA)
- Pyruvate metabolism, mitochondrial oxidative phosphorylation disorders, Krebs cycle defects, disorders of thiamine transport and metabolism (PM-MD)
- Carbohydrate, fatty acid oxidation and ketone bodies disorders (C-FAO)
- Lysosomal storage disorders (LSD)
- Peroxisomal disorders (PD)
- Congenital disorders of glycosylation and disorders of intracellular trafficking (CDG)

WHAT ARE THE MAJOR AIMS OF U-IMD?

Establishing a patient registry for the European Reference Network for Hereditary Metabolic Disorders (MetabERN).
U-IMD has fully implemented the common data elements of the European Platform on Rare Disease Registration (EU RD Platform) and will be integrated into the European Rare Disease Registry Infrastructure (ERDRI). U-IMD is the first unified European registry that encompasses all IMDs.

Upgrading already existing IMD registries to the standard of U-IMD, starting with the registry of the International Working Group on Neurotransmitter Related Disorders (iNTD).

Developing a standard for minimal core data sets shared by the MetabERN and the European Rare Kidney Disease Reference Network (ERKNET).

User Group

U-IMD is intended to be used by physicians treating patients with rare inherited metabolic disorders. U-IMD is available for all Members of MetabERN but also for international users not associated with MetabERN.

Access

U-IMD is a web-based patient registry. U-IMD is accessible to registered users via the internet using password protected user accounts and encrypted data transfer between server and client.