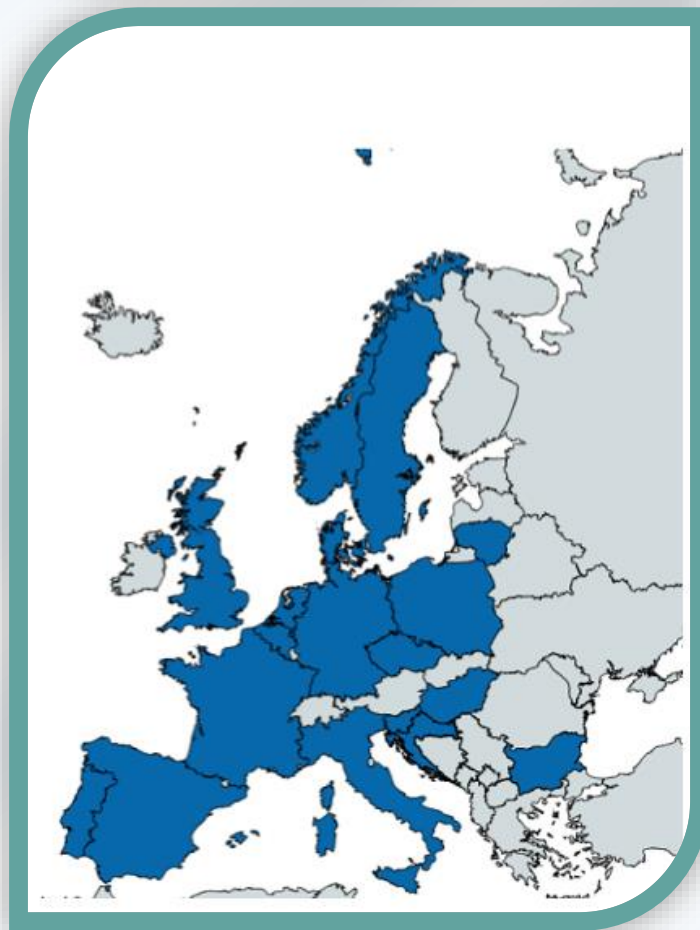


MetabERN: the European Reference Network for Rare Hereditary Metabolic Disorders

The first most comprehensive, pan-metabolic, pan-european, patient-orientated platform ever conceived



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BACKGROUND

In light of the existing Commission initiative in the area of rare diseases, the [Cross Border Directive](#) 2011/24/EU published by the EU to ensure that affected patients are given the priority they deserve and that their needs in receiving better diagnosis, treatment and management are met, an European Reference Network for Inherited metabolic diseases (MetabERN) was approved, on December 15th 2016, by the European Commission.

The major goal of this initiative is to ensure a coordinated action in creating the widest collaboration among paediatric and adult metabolic physicians and patient associations at EU level, facilitating patient access to specialists with expertise in the metabolic field and to foster research activity.

MetabERN serves as a referral hub to ensure optimal knowledge-sharing, to improve early diagnosis and treatment of inherited metabolic diseases (IMDs) at EU level, to bring expertise at patient's bed, to facilitate access to therapy and to coordinate clinical and research services to rationalize the existing resources at European level.

GOALS

MetabERN aims to

- Accommodate and interconnect expertise across Europe at the service of IMD patients,
- Harmonise data collection across participating European centres,
- Establish common approaches to optimise prevention, diagnostics, management and treatment,
- Develop and implement harmonized guidelines,
- Stimulate cross-border research and innovative treatments
- Develop training and education opportunities especially in Member States where it is lacking,
- Interact closely with patients to serve as a reliable source of information and involve them in the planning of their individual care pathways and decision-making.

FEATURES OF THE METABERN

The MetabERN's main key features:

International Coverage Today MetabERN involves **69 certified Health Care Providers endorsed by the Ministries of Health from 18 different EU countries**. MetabERN is endorsed by and partners with the Society for the Study of the Inborn Errors of Metabolism (SSIEM).

Multidisciplinary Team **MetabERN involves 1681 experts**, of which about 52% are specialized medical doctors (particularly Pediatric and Adult physicians, Geneticists, Neurologists and Metabolic Physicians).

Main Diseases Categories IMDs are a group of more than **800 genetic** and often lethal disorders affecting children and adults. All IMDs, with no exclusion, are of interest for the MetabERN, independently from their prevalence, frequency and existing previous interest for research or therapy development.

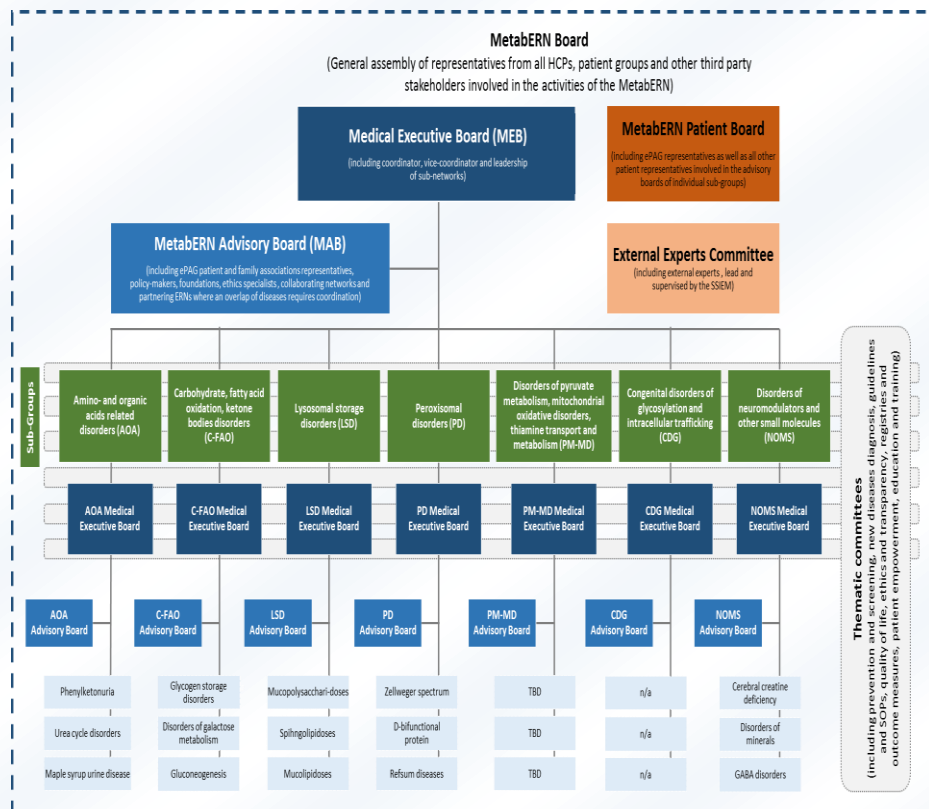
Considering the complexity of the IMDs field as a whole, additional core networks of experts interested in homogenous IMDs categories have been created.

The network is structured under the following 7 thematic groups:

1. Amino and organic acids-related disorders (AOA)
2. Pyruvate metabolism mitochondrial oxidative phosphorylation disorders, Krebs cycle defects, disorders of thiamine transport and metabolism (PM-MD)
3. Carbohydrate, fatty acid oxidation and ketone bodies disorders (C-FAO)
4. Lysosomal Storage disorders (LSD)
5. Peroxisomal disorders (PD)
6. Congenital disorders of glycosylation and disorders of intracellular trafficking (CDG)
7. Disorders of neuromodulators and other small molecules (NOMS)

The Governance

Each member of MetabERN holds a role in one or several of the identified bodies, including the MetabERN Board (all members) (MEB) as well as at least one of the disease specific sub-groups and at least one of the thematic committees spanning all disease-specific sub-groups, in which professionals and patient/family associations will collaborate within the specific disease group/disease but also to transversal programs common to all subnetworks.



The MEB is supported by the **MetabERN Patient Board (PB)** and the **MetabERN Advisory Board (MAB)**.

- The MetabERN Patient Board is the representation of all patient and family associations involved in MetabERN activities. It is an independent body which oversees and evaluates the work of MetabERN from the patients' perspective.
- The MetabERN Advisory Board consists of representatives of the European Patient Advocacy Group, Patients and Family Associations, Foundations, Policy-makers, representatives from collaborating networks and other relevant stakeholders. Members serve for a 5-year renewable term.

An **External Experts Committee (EEC)**, made up of HCPs not operating inside the MetabERN governance bodies, will also be set up to provide an external evaluation on the activities of the Network.

At its functional level, the MetabERN is composed of 7 Sub-Groups, each covering a different group of rare inherited metabolic diseases (see diagram above). Each sub-group replicates at its level the structure of the overall MetabERN.

Patients Coverage **42.427** are the patients managed by the MetabERN, 68% of which represented by paediatric patients.

MetabERN Activities Working groups and horizontal activities among all the 7 subnetworks are defined.

In the first year, the following main strategic activities of the network are planned:

- Formalisation of the set-up of the network,
- Establishment of closer collaboration between participating healthcare professionals and patients in the IMD field,
- Dissemination of information about the network to increase awareness in the relevant communities and
- Development of a comprehensive overview of the current situation in IMD diagnosis, care and management and therefore build the basis for the development of a White Book on IMDs in Europe at a later stage.

MetabERN Services The provision of the main services identified by the EC

- (Prevention (e.g. genetic screening);
- Acute care
- Ambulatory services
- Diagnostic services
- Interventional therapeutic services
- Rehabilitation services
- Social care services
- Palliative care services

Patient organisations engagement MetabERN members have developed close collaborations with 44 national and European patient organisations and will build on their strong relationships to extend the networks engagement with the wider IMD patient community and break isolate that many single patients currently experience.

- There is an elected European Patient Advocacy Group for MetabERN. Patient representatives are members of the network board and committees with the clear role to contribute to the development of the network including acting as a 'communication and information' actor between the patient and clinical community.

For more information please contact Dr. Lut de Baere, President of the Belgian Patient Organization for Rare Metabolic Diseases, responsible for the MetabERN Patients organizations management, at lut@boks.be.

CONCLUSION and FUTURE PROSPECTS

MetabERN does ensure a join-up approach to care by bringing together paediatric and adult metabolic physicians across the EU to deliver the best available care to patients affected by IMDs.

To this end, the MetabERN has already established a multi-disciplinary and multi-stakeholder platform including Patient Organizations and Scientific/Medical Organizations to foster transversal program among the subnetworks aimed to improve standards of clinical management and strengthen referral systems, seek consensus on EU guidelines and Standard Operating Procedures, share knowledge in a structured way, generate epidemiological studies and registries, facilitate diagnosis of new diseases, empower patients and help the transition process from youth to adulthood, encourage virtual counselling, design new pathways for training and education, facilitate research programs and promote innovative clinical trial design for the development of innovative therapies and evaluate efficacy and safety of drugs in independent way.

Furthermore, the MetabERN aims to help all patients to better access to therapy by helping the Member States in designing an economical strategy for the sustainability of rare diseases.

For more information please contact

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