

MetabERN: the European Reference Network for Rare Hereditary Metabolic Disorders

the first pan-European network for all metabolic diseases

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Introduction

In light of the existing Commission initiative re 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.

What are Inherited Metabolic Diseases?

Inherited Metabolic Diseases (IMDs) are a group of more than 700 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.

Aims

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 will facilitate and harmonize patients' access to diagnosis and best treatment by:

Accommodating and interconnecting expertise across EU

Harmonising data collection across participating EU centres

Establishing common approaches to optimise prevention, diagnostics, management and treatment

Developing and implementing harmonized guidelines

Stimulating cross-border research and innovative treatments

Developing training and education opportunities especially in Member States where they are lacking

Interacting closely with patients

Activities

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 HCPs and patients in the IMDs field

Dissemination of information to increase awareness in the relevant communities

Development of a comprehensive overview of the current situation in IMDs diagnosis, care and management

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).



Governance

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 & 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)

Future Prospects

MetabERN is implementing its multi-disciplinary and multi-stakeholder collaborative governmental structure that includes patients, academia, scientific organizations, politics, insurance companies and industry. It will be aimed at facilitating and harmonize patients' access to diagnosis and best treatment by fulfilling and improving the following activities:

- Prevention and Screening
- Diagnosis/New Diseases Diagnosis
- Epidemiology/Registries and Outcome
- Management/QoL, Transition Guidelines & Pathways
- Virtual Counselling
- Education and Training
- Patient Empowerment
- Clinical Trials
- Research
- Dissemination/Stakeholders Relations

Multidisciplinary Team (MDT)

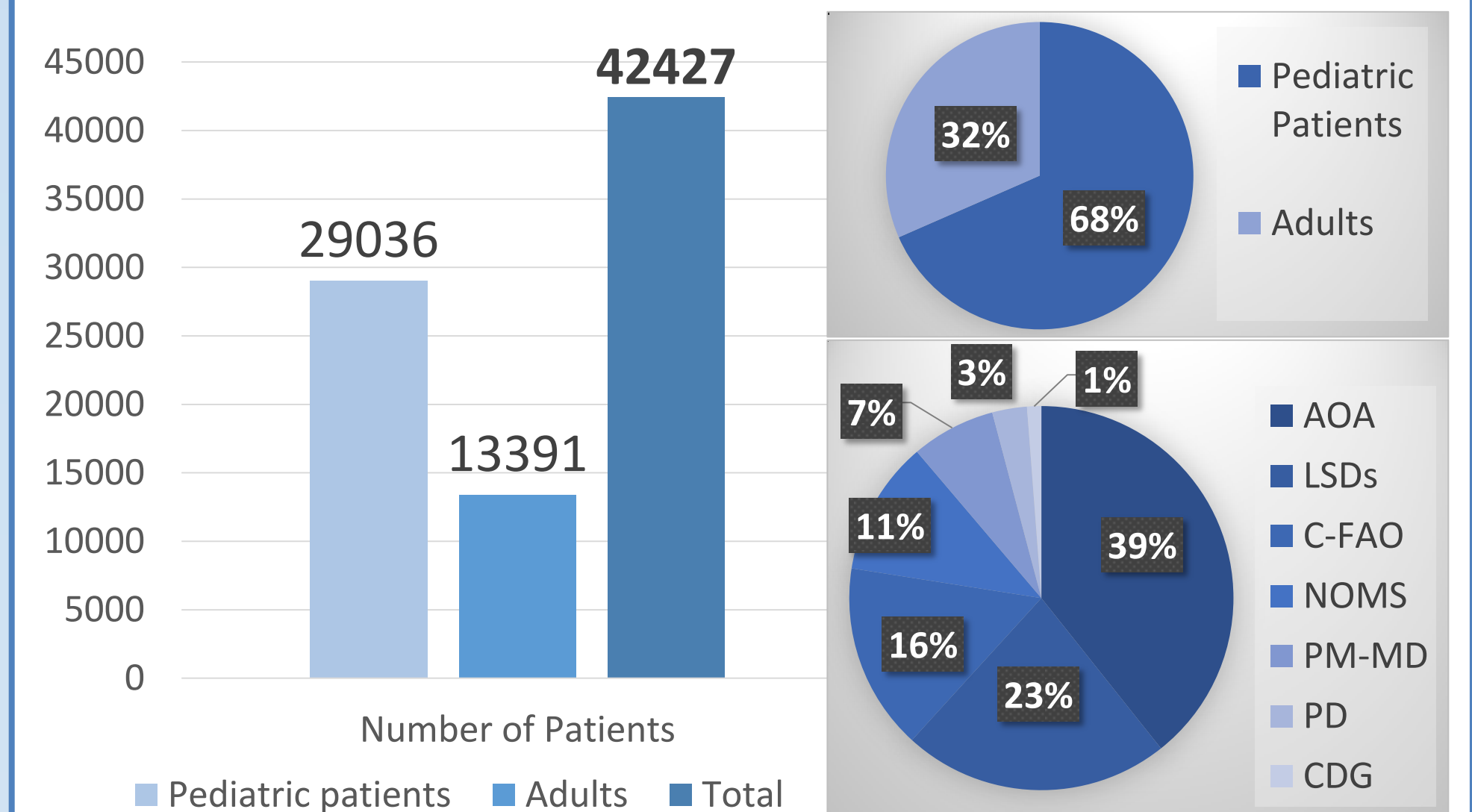
MetabERN involves 1681 experts, of which about 52% are specialized medical doctors (particularly Pediatric and Adult physicians, Geneticists, Neurologists and Metabolic Physicians)

- MDT composed of
- 871 Specialized MDs
 - 188 Biochemists/Biologists
 - 184 Nurses
 - 121 Dieticians/Nutritionists
 - 76 Physical therapists/Rehab
 - 73 Psychologists
 - 49 Social workers
 - 34 Pharmacists
 - 34 Coordinators/Medical Secretaries
 - 13 Experts in Palliative care/Pain management
 - 10 PHD/students
 - 28 Other

a Total of 1681 Experts

Patients Coverage

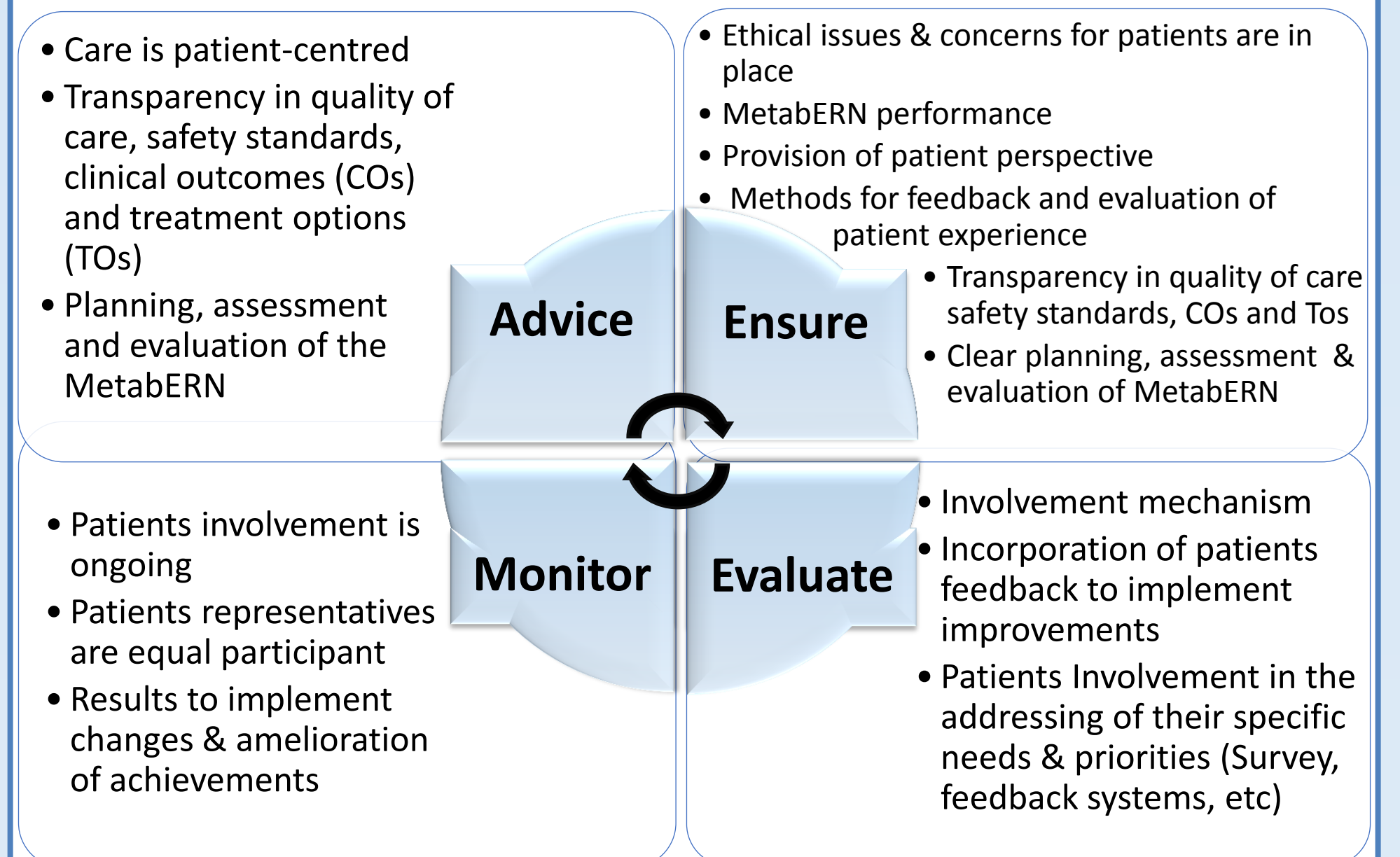
42.427 are the patients managed by the MetabERN, 68% of which represented by paediatric patients



Patient organisations engagement

Close collaborations with 44 national and European patient organisations has been established.

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.



Contact Information

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