



European Reference Network

for rare or low prevalence complex diseases



Network

Hereditary Metabolic Disorders (MetabERN)

EURORDIS
MEMBERSHIP
MEETING
2017



WHY PATIENT ASSOCIATIONS SHOULD WORK INSIDE METABERN

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VAINCRE LES MALADIES
LYSOSOMALES
RARE MAIS PAS SEUL !



My conflict of interest !





VML's Core Values *as many, many Pos....*



➤ PRECISION

Transparency, quality of the information given, coherence

➤ SOLIDARITY

Between pathologies (rare and ultra-rare),
for all our members

➤ INVOLVEMENT

Goals to find a cure for all our lysosomal diseases, strong
mobilisation for our fight :

3238 days / men in 2015



Ethical Principles

✓ Autonomy

Respect Patient Self-Determination

✓ Beneficence

Promote Patient Well-being

✓ Nonmaleficence

Do No Harm

✓ Justice

Fair Allocation of Resources

Ethical Principles

➤ The role of a Patient Organisation

➤ Information and Training

(associative « DNA », sharing knowledge between « peers »)

➤ Beneficence and non-maleficence

➤ **Autonomy:** central question of choice and freedom....

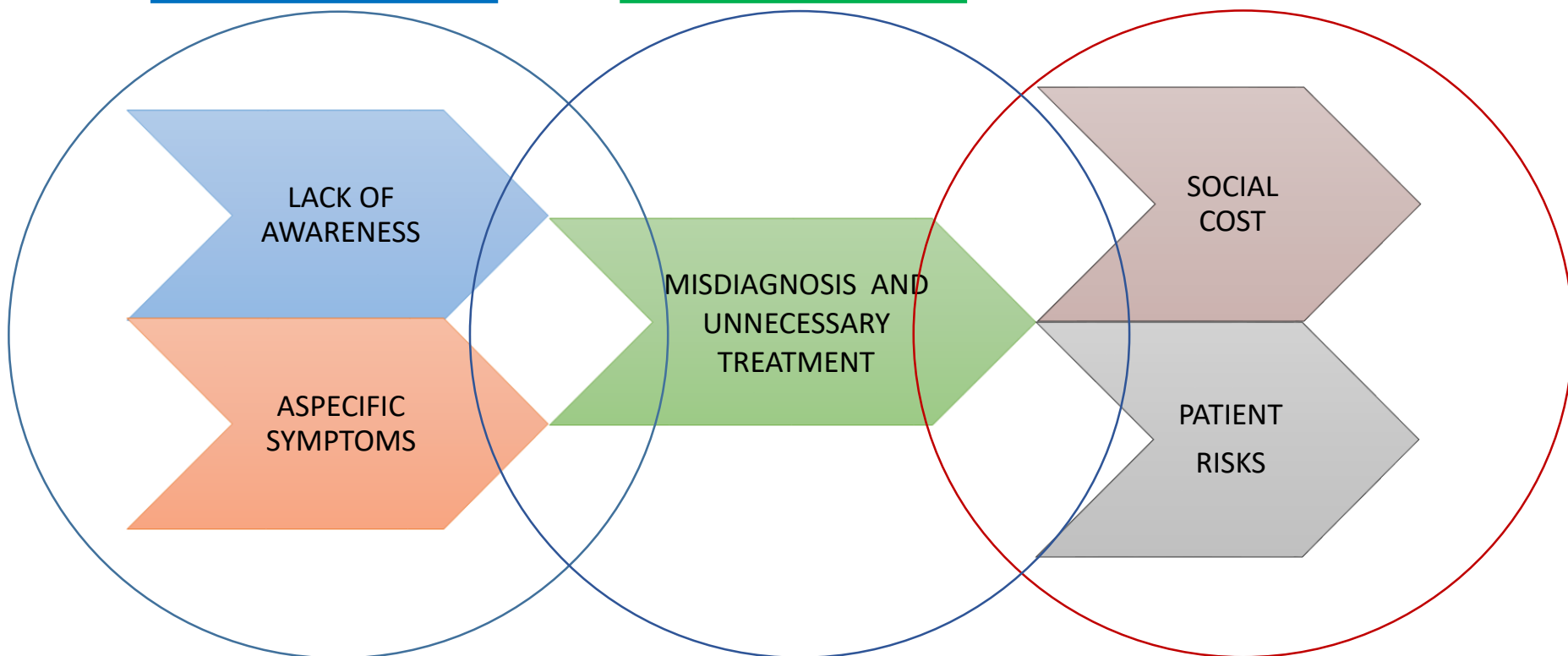
➤ **Equity and Justice:** no treatment or very few / Access for the patients in France, in EU countries and the others....

RARE DISEASES MAIN PROBLEM: Delayed Or Difficult Diagnosis

ACADEMICS

CLINICIANS

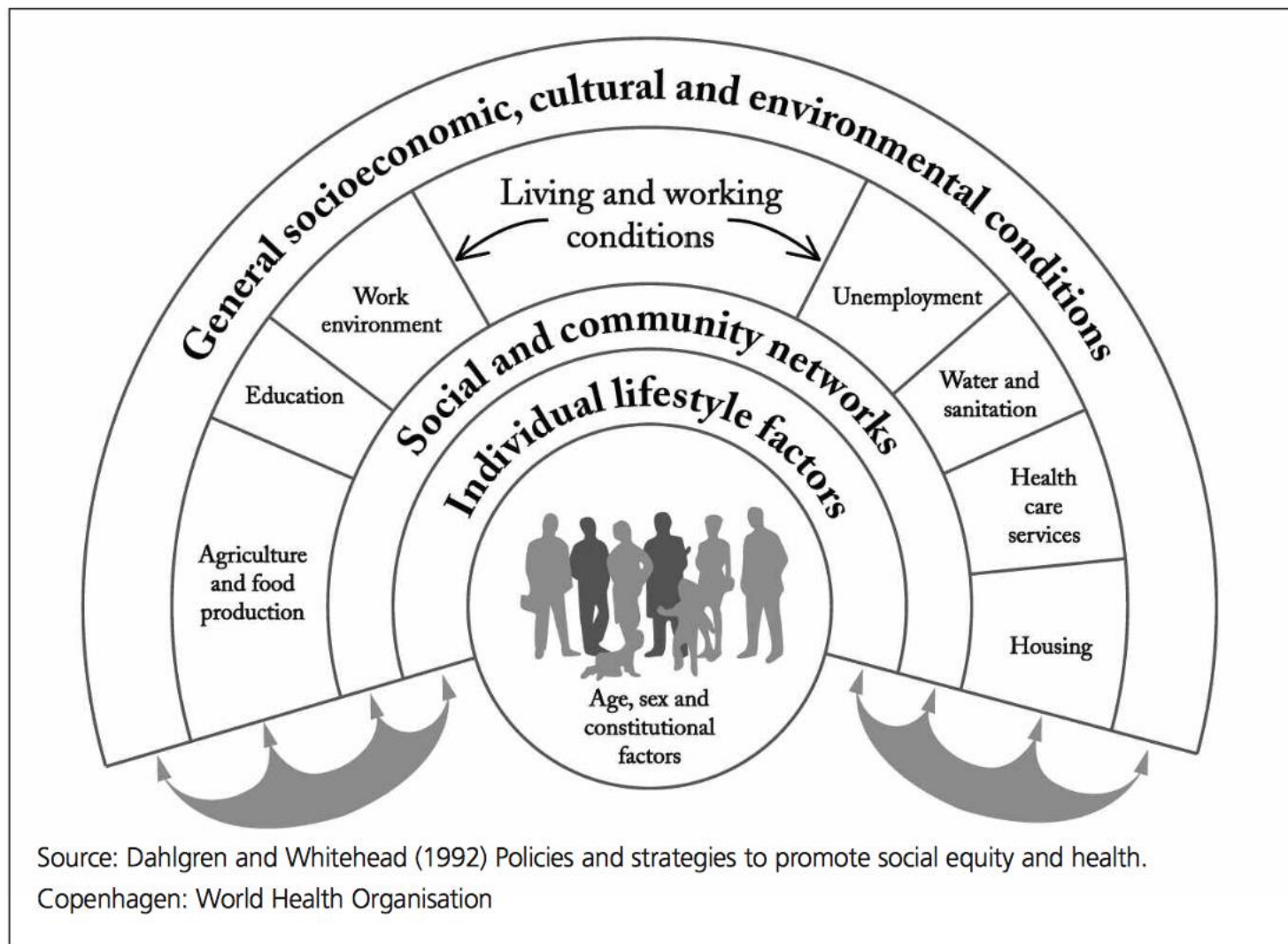
SOCIETY



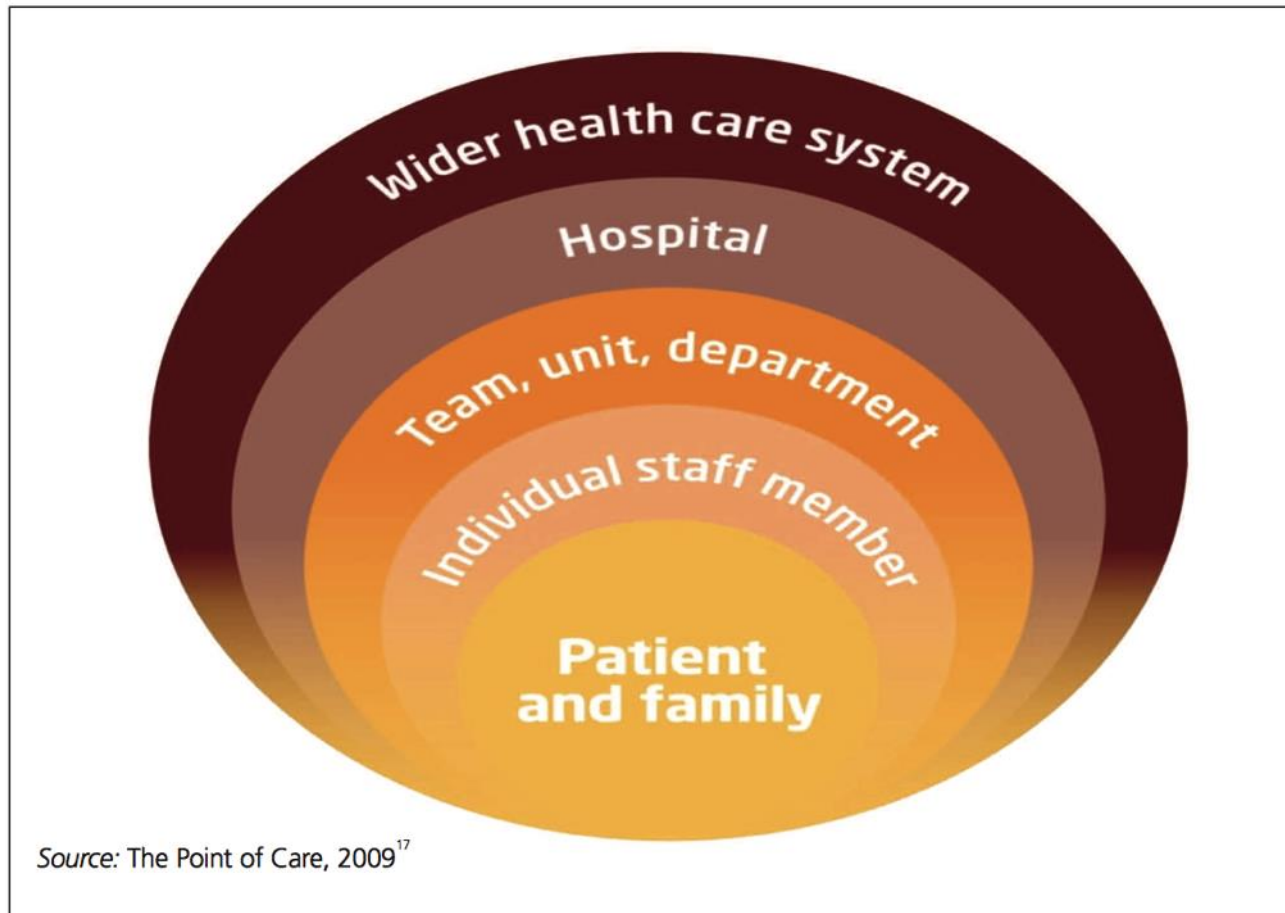
INDEX OF SUSPICION MUST BE HIGH

It's necessary to increase awareness among

FACTORS DETERMINING THE HEALTH POPULATIONS



A FRAMEWORK FOR THE ANALYSIS OF THE FACTORS INFLUENCING PATIENTS' EXPERIENCE



KEY PRIORITIES FOR PATIENTS

- Respect and support for the individual patient, their wants, preferences, values, needs and rights
- Choice and empowerment
Patient engagement in health policy
Access and Safety
- Information that is accurate, relevant and comprehensive
- Strengthen patient involvement in all processes to improve health outcomes and ensure needs are met
- The essence of patient-centred healthcare is that the healthcare system is designed and delivered so that it can answer the needs of patients

MAJOR AIMS IN COLLABORATING WITH METABERN

- **To realize active partnerships with patients' organizations,** maximizing their impact through capacity building
- **To advocate internationally** with a strong patients' voice on relevant aspects of healthcare policy, with the aim of influencing international, regional and national health agendas and policies
- **To build cross-sector alliances** and working collaboratively with like-minded medical and health professionals, policy makers, academics, researchers and industry representatives

Strengths

- ✓ Multidisciplinary work
+ Shared registries service
Natural history of the diseases
+ European Patient advocacy group
+ Patient Empowerment / Patient Involvement
=
Creating quality outcomes and clinical excellence
Improving outcomes for patients

Opportunities

- Formation / Information
- ✓ Patient centred approach
Relevant Data
 - ✓ E-health tools
- ERN Platform, multidisciplinary work between ERNs
- ✓ Research / Pilots
+ H2020

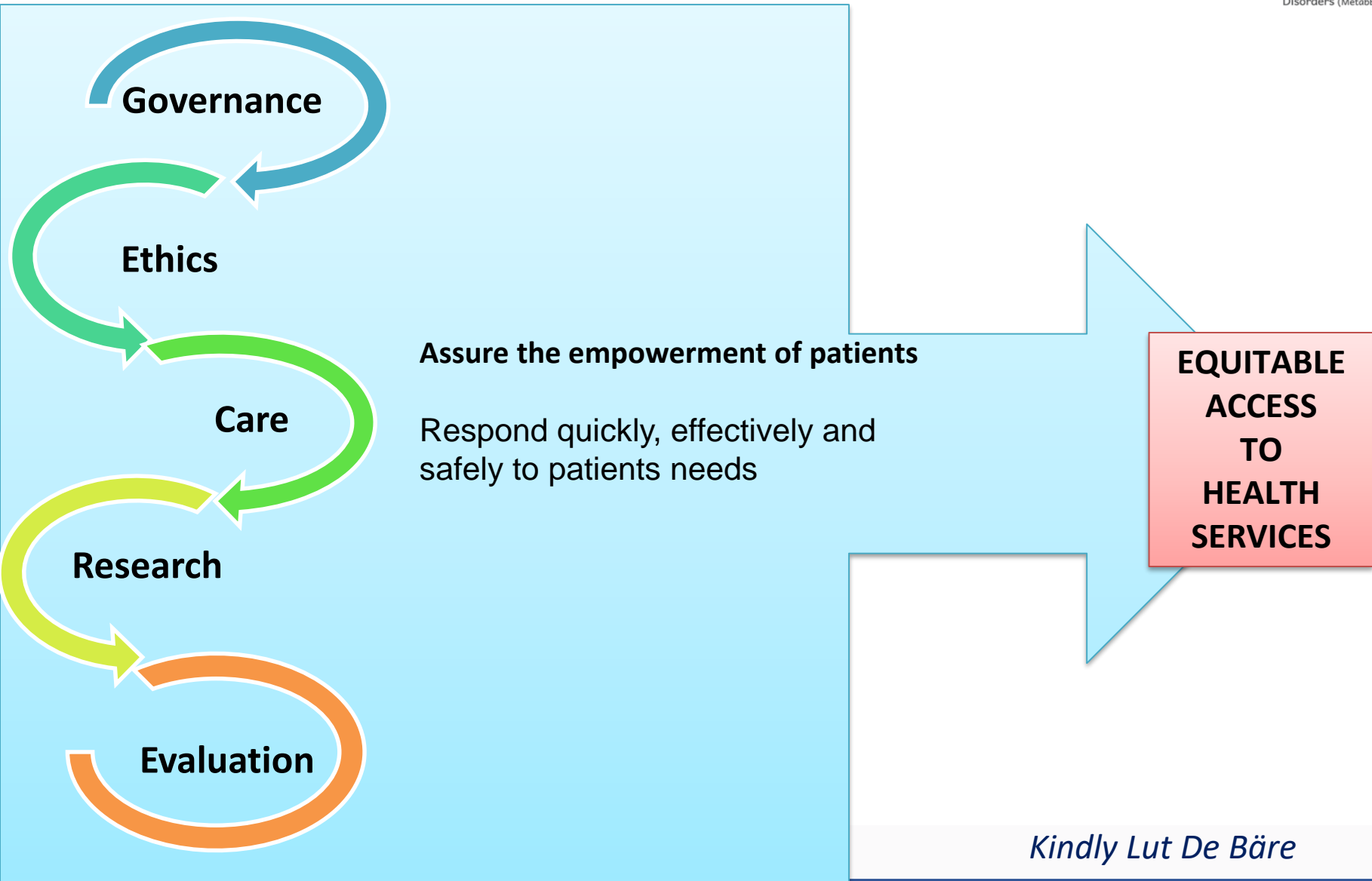
Weaknesses

- ✓ Depend of a certain amount of maturity of the network
 - ✓ E-health tools
Rarely available
- ✓ Access to treatment and diagnosis
Inequity between countries

Threats

- ✓ Complexity of rare diseases
- ✓ Lack of relevant patient data in the scope of QoL, social consequences, burden of disease
Lack of indicators to prove the effectiveness of a treatment and not only the efficacy.....
 - ✓ Transparency
POs, Investigators, Sponsors

PATIENT REPRESENTATIVES ROLE IN METABERN



Kindly Lut De Bäre

MetabERN Board

(General assembly of representatives from all HCPs, patient groups and other third party stakeholders involved in the activities of the MetabERN)

Medical Executive Board (MEB)

(including coordinator, vice-coordinator and leadership of sub-networks)

MetabERN Patient Board

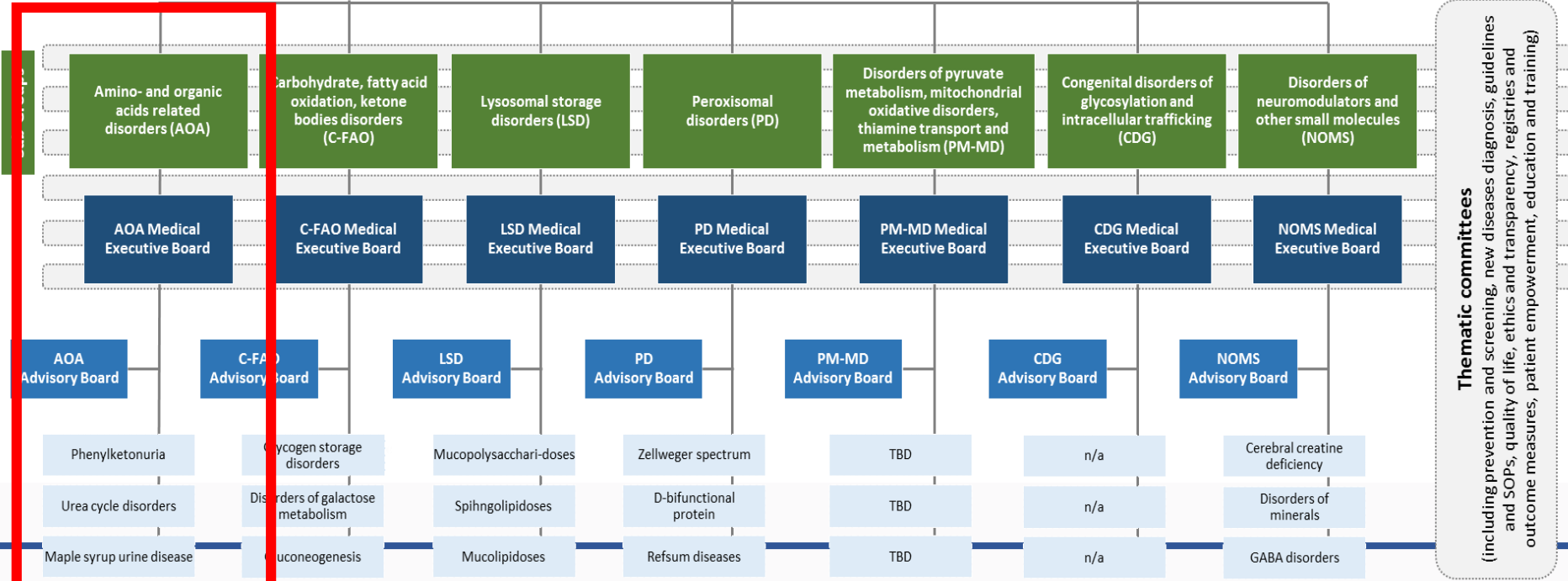
(including ePAG representatives as well as all other patient representatives involved in the advisory boards of individual sub-groups)

MetabERN Advisory Board (MAB)

(including ePAG patient and family associations representatives, policy-makers, foundations, ethics specialists, collaborating networks and partnering ERNs where an overlap of diseases requires coordination)

External Experts Committee

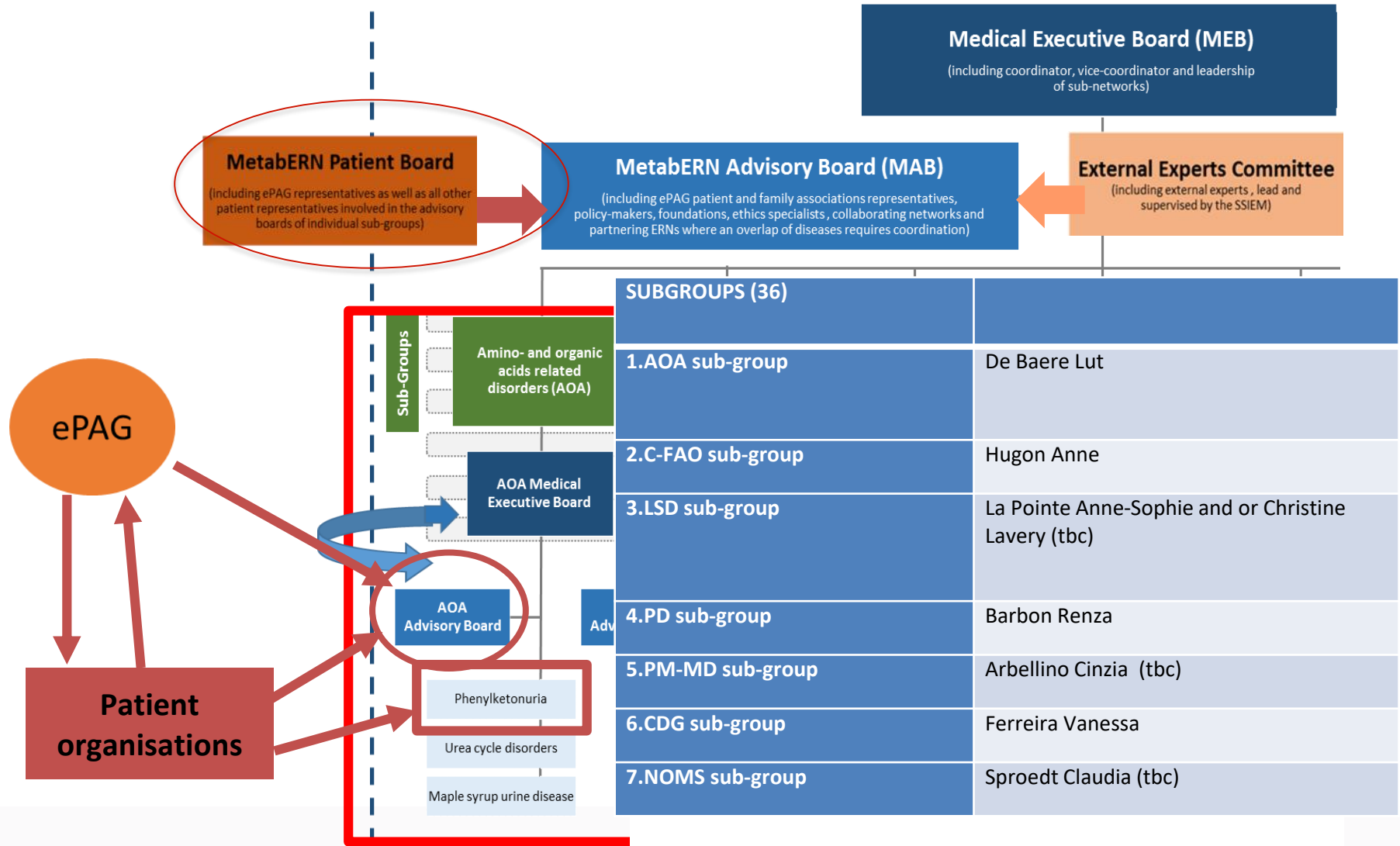
(including external experts, lead and supervised by the SSIEM)



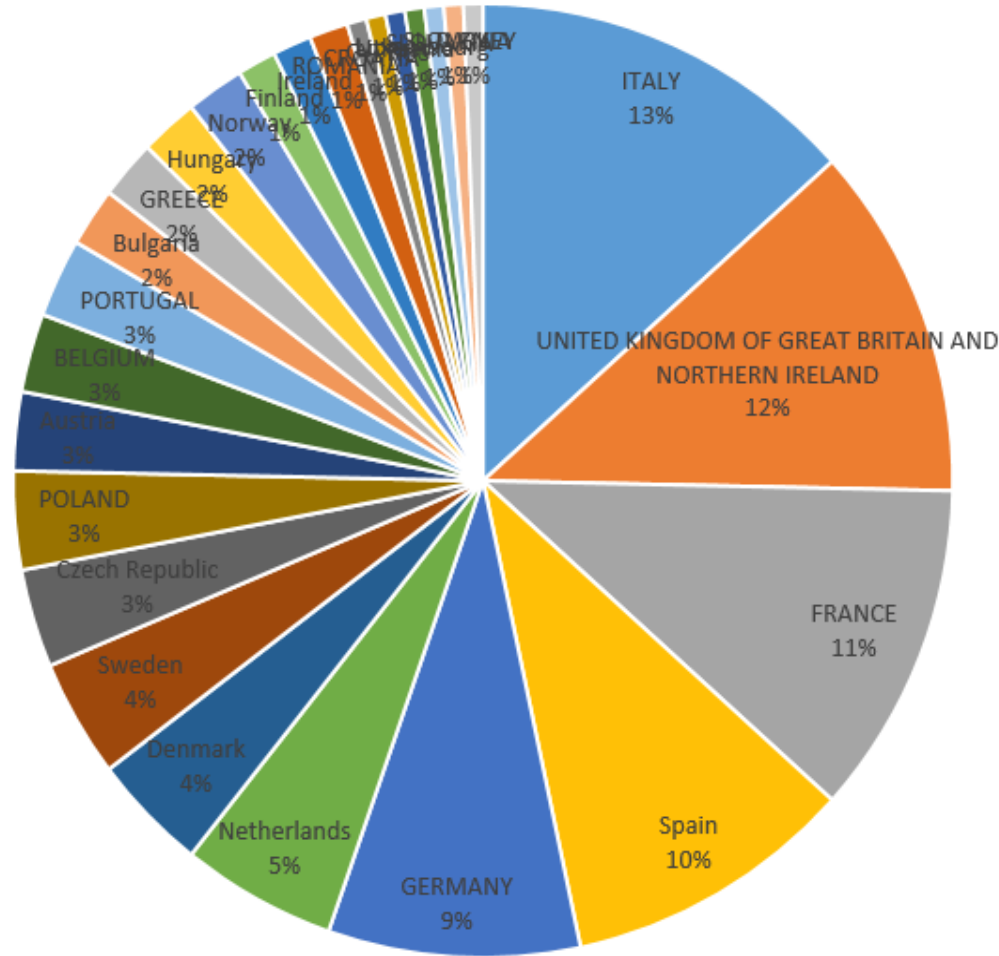
Thematic committees

(including prevention and screening, new diseases diagnosis, guidelines and SOPs, quality of life, ethics and transparency, registries and outcome measures, patient empowerment, education and training)

THE ROLE OF PATIENT ORGANISATIONS



IN TOTAL SO FAR 366 PO'S ARE MAPPED



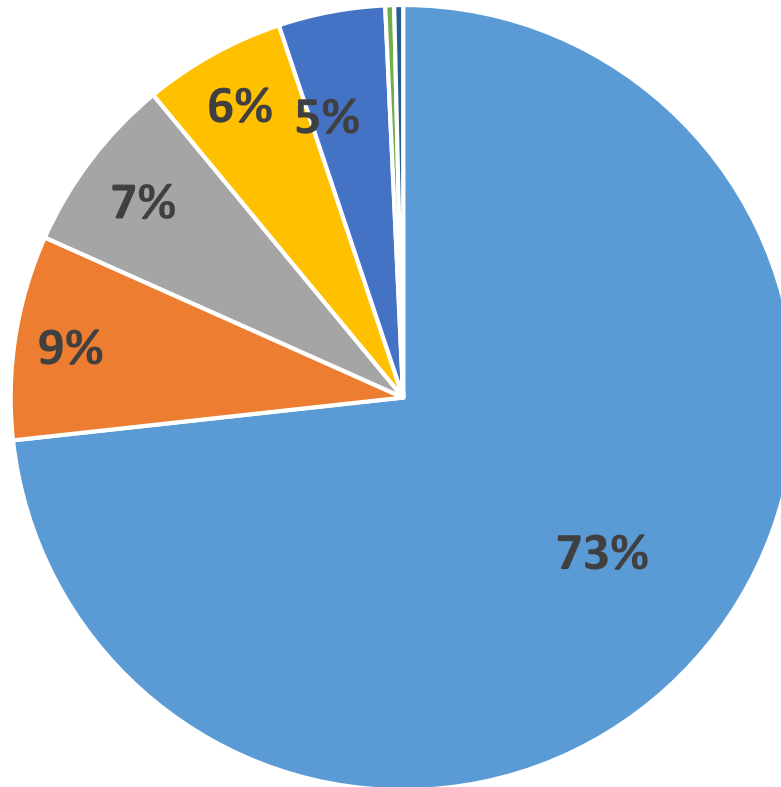
- ITALY
- FRANCE
- GERMANY
- Denmark
- Czech Republic
- Austria
- PORTUGAL

- UNITED KINGDOM OF GREAT BRITAIN AND NORTHERN IRELAND
- Spain
- Netherlands
- Sweden
- POLAND
- BELGIUM
- Bulgaria

Kindly Lut De Bäre

THE 273 PO'S HAVE BEEN GROUPED ACCORDING TO THE 7 SUBNETWORKS

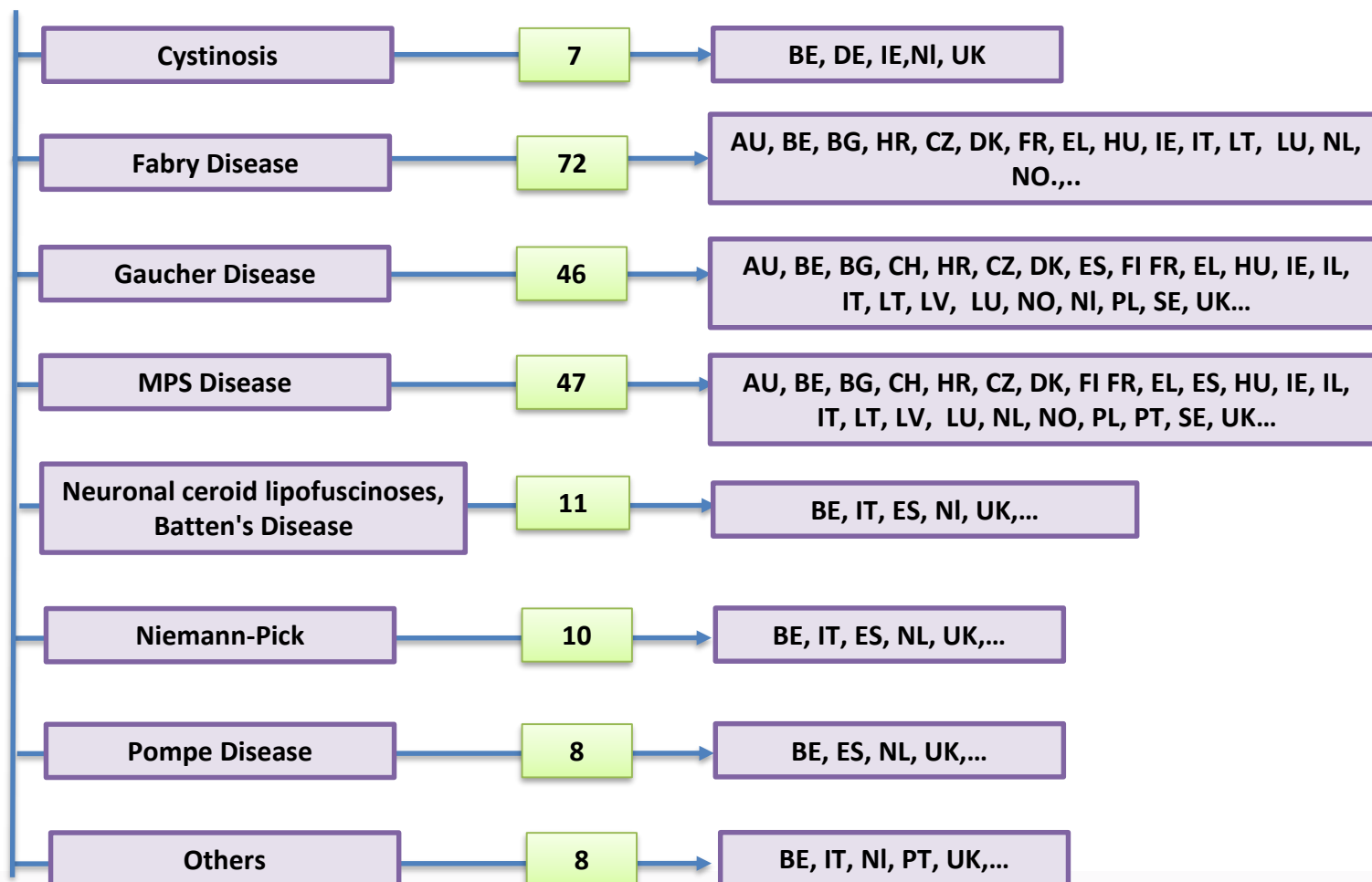
AOA	PM-MD	C-FAO	LSDs	PD	CDG	NOMS
20	16	12	200	1	23	1



■ LSDs ■ CDG ■ AOA ■ PM-MD ■ C-FAO ■ PD ■ NOMS

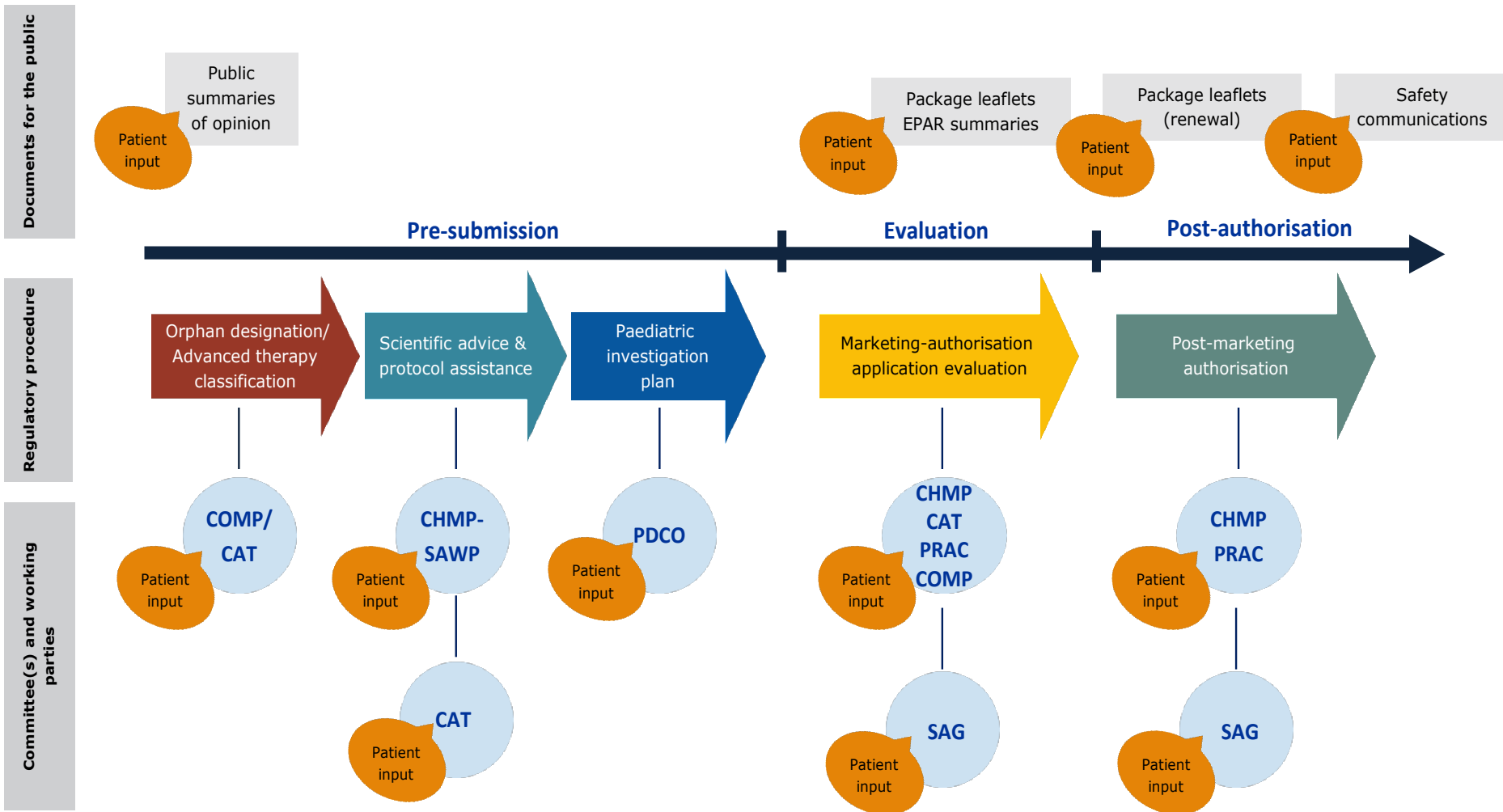
Example

Sub-Network: Lysosomal Disorders



- ✓ What are the challenges facing collaborative medical research?
- ✓ How does one recognize in this cooperation the factors driving and hampering the development of new drugs targeting rare diseases?

Overview of patient involvement along the medicines lifecycle at EMA



Quality of Life

Principles of Beneficence and Nonmaleficence and Respect for Autonomy

WP7 : Capacity-building & Training

How is evaluated the disease burden for the family environment ?

⇒ Psychologist + Social care

Which is the evaluation of patient' s quality of life ?

⇒ Dialogue with the family

⇒ Information & Formation for health care professionals :

⇒ learning and training

Care and Research

✓ How to insure quality gathering of registries?
Are the infrastructures ready and efficient in all countries?

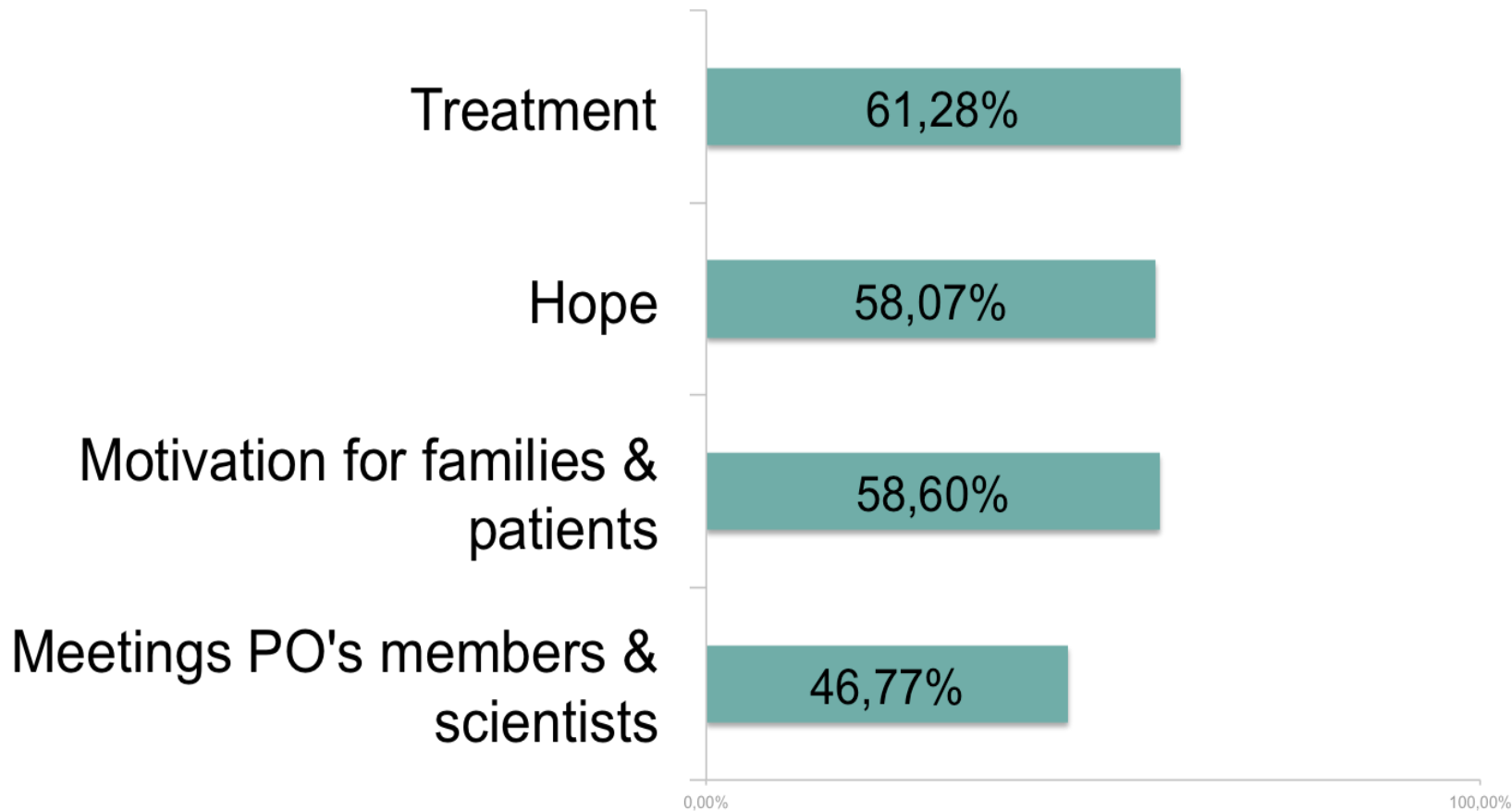
✓ New registries need to take into account the needs of the regulators, HTA and payers to collect the relevant data

✓ What will be the role of patient organisations in this process ?

✓ Need to support the patient organisations to be informed and efficient partners

Question for PO:

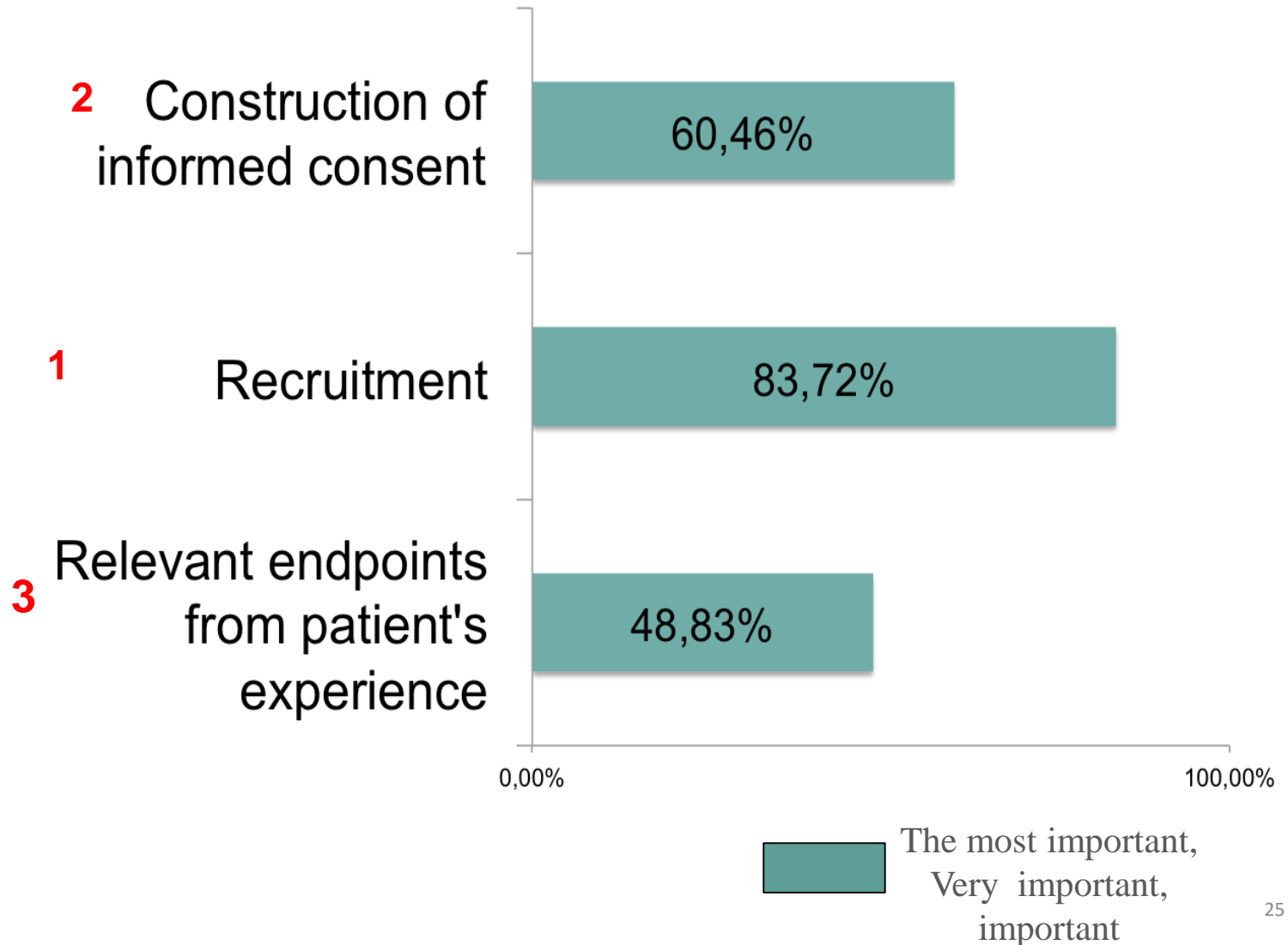
What is for you the main interest to have research collaboration ? n=62



For 61.28% of POs, it could improve access to treatment

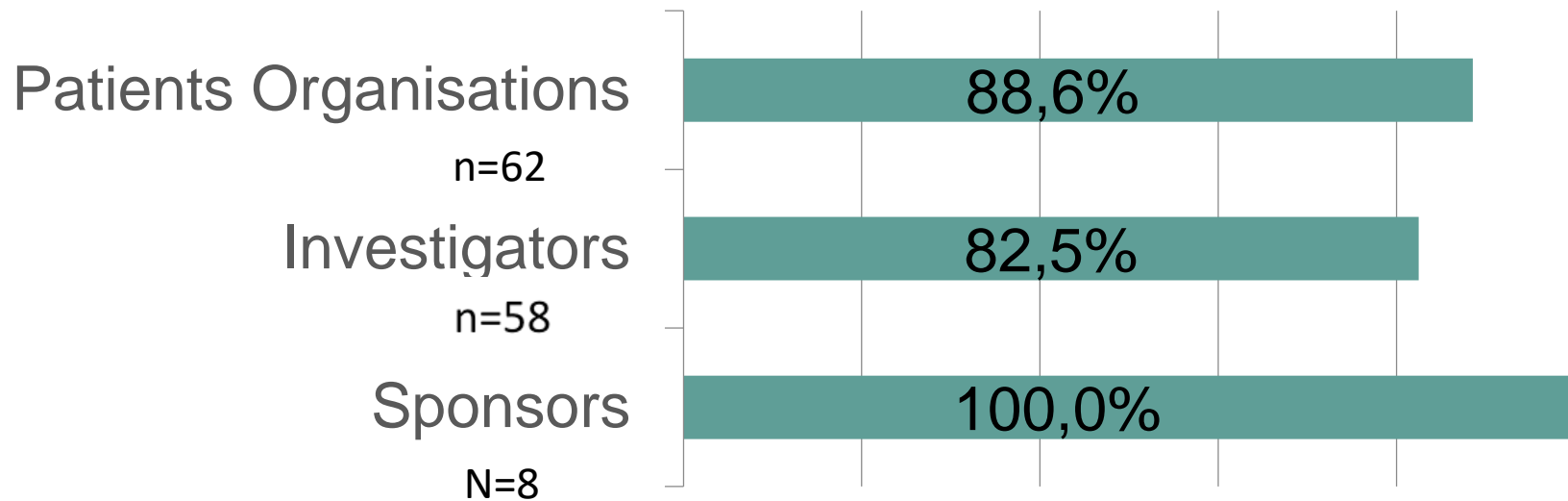
Question for investigators:

What is for you the main interest to have research collaboration with POs ? n=58



Advantage for the data incrementation by the patients Patient Reported Outcome Measures PROMSs

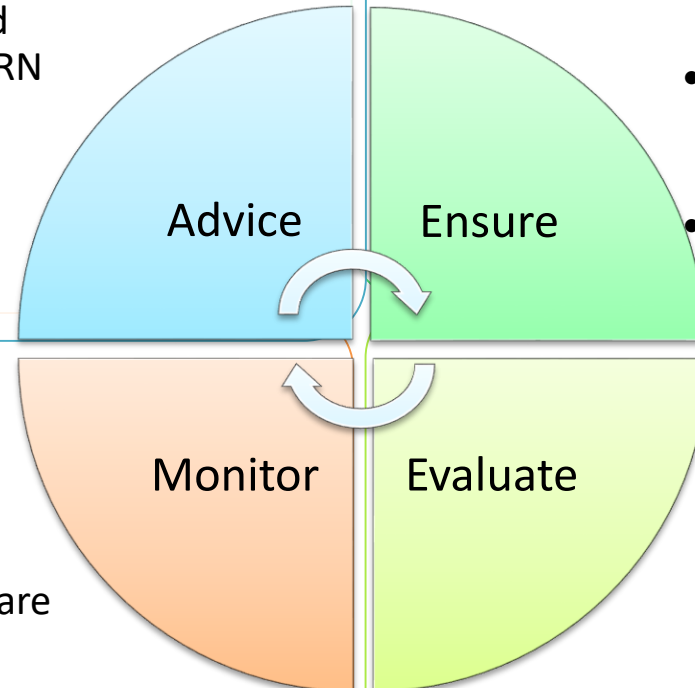
To better evaluate quality of life
=
Patient centered



SUMMARIES OF TASKS OF THE METABERN PATIENT BOARD

- care is patient-centred
- transparency in quality of care, safety standards, clinical outcomes and treatment options
- planning, assessment and evaluation of the MetabERN

- All ethical issues & concerns for patients are in place
- the performance of the MetabERN.
- Provision of patient perspective
- Methods for feedback and evaluation of patient experience,
- transparency in quality of care, safety standards, CO and treatment options
- Clear planning, assessment and evaluation of the MetabERN



- Patients involvement is ongoing
- patients representatives are equal participant
- Results and act upon them to implement changes and amelioration of the achievements

- Review involvement mechanism regularly
- Incorporate patient feedback to implement improvements
- Involve patients in the addressing of their specific needs, preferences, priorities (Survey, feedback systems, etc)

WHAT HAVE PRS DONE, TILL NOW, IN THE METABERN

Advice: - Governance & Strategy of MetabERN

Mapping of:

- The International, European, National and umbrella organisations

Engage: - POs to join the MetabERN

- e PAG members responsibility in the subgroups

- Eurordis & e PAG members

Preparing: - Annual Report, POs insights MetabERN

- MetabERN presentation for EGA (2016, June)

- MetabERN presentation for ES PKU (2016, November)

- a survey: patient needs in every country

GENERAL CONCLUSION

- **The MetabERN is an unique opportunity to impact the life of patients and the management of their diseases.**
- The patients and patients' empowerment are at the center of the interests of the MetabERN.
- We have a formal proactive collaboration with EURORDIS to facilitate the collaboration
- The PO will be involved in the different activities of the MetabERN
- The PO will participate to the governance, ethics, care, research, evaluation of the MetabERN.
- MetabERN represents the first opportunity for all the expert centres to really work together in a coordinated way.
- MetabERN : The spirit of rare diseases will make the difference.

The patients and patient's empowerment are at the center of the interests of the MetabERN.



Prevention and Screening

Diagnosis/New Diseases Diagnosis

Epidemiology/Registries and Outcome

Management/Quality of Life and Transition Guidelines and Pathways

Virtual Counselling

Education and Training

Patient Empowerment

Clinical Trials

Research

Dissemination/Stakeholders Relations

Formation

+

Training



Informations

For

families



Dialog

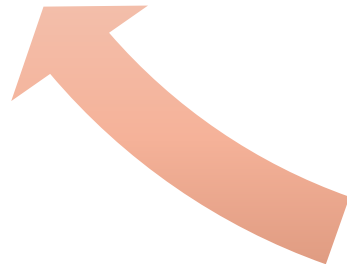


Vulnerability

For the patients

Also for the

Healthcare professionals



Mutual
attention



MERCI !
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THE BURDEN OF TREATMENT IN RARE DISEASES

***MAURIZIO SCARPA,
Coordinator MetabERN***

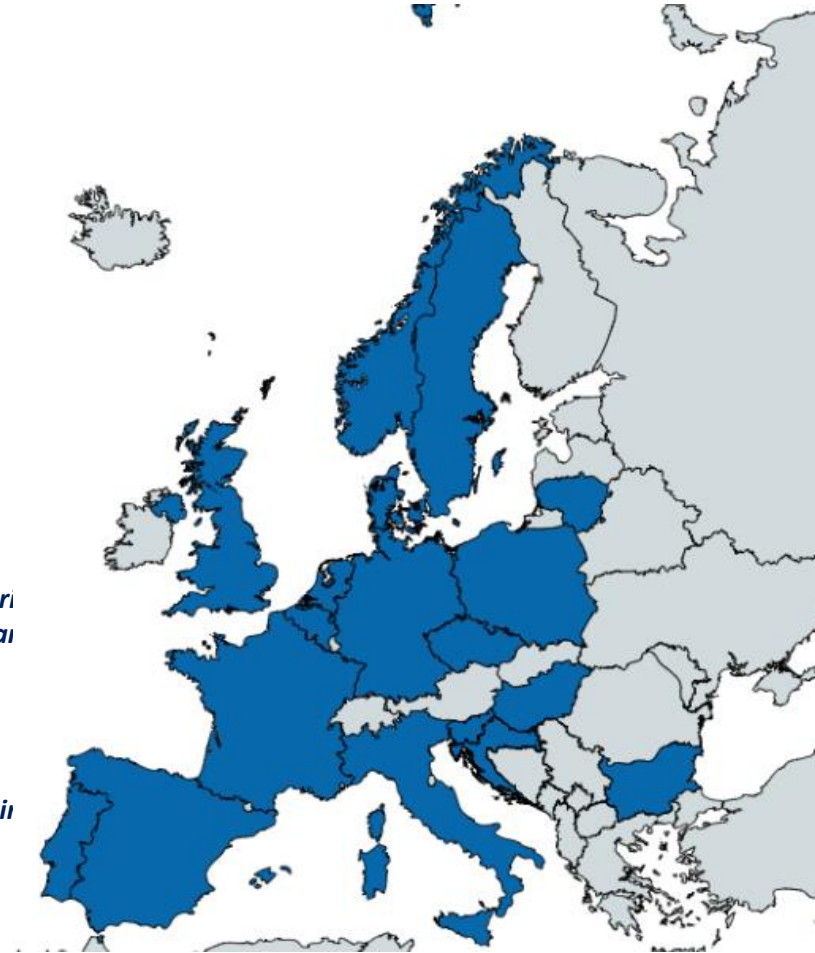
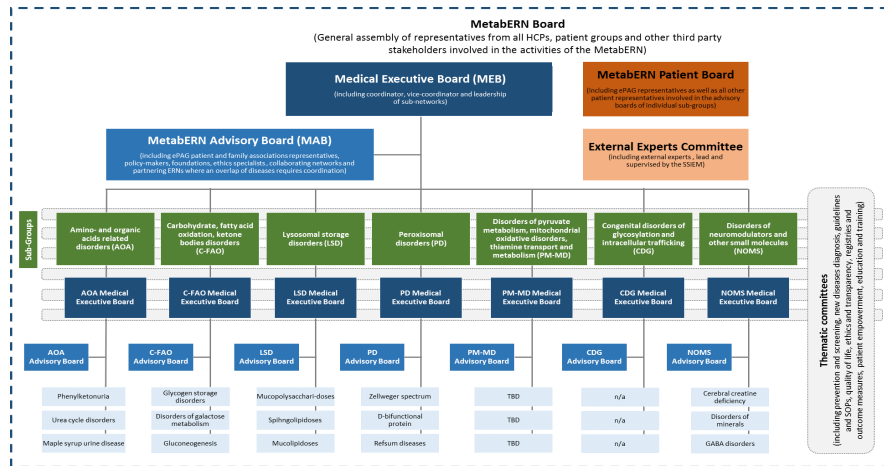


MetabERN: ERN on HEREDITARY METABOLIC DISEASES

Coordinator

Prof. Maurizio Scarpa MD PhD

Helios Dr. Horst Schmidt Kliniken Wiesbaden, Germany



- 1) **Aminoacid and organic acids related disorders**
- 2) **Disorders of pyruvate metabolism, Krebs cycle defects, mitochondri oxidative phosphorylation disorders, disorders of thiamine transport and metabolism**
- 3) **Charbohydrate, fatty acid oxidation and ketone bodies disorders**
- 4) **Lysosomal disorders**
- 5) **Peroxisomal and lipid related disorders)**
- 6) **Congenital disorders of glycosylation and disorders of intracellular traffickii**
- 7) **Disorders of Neuromodulators and Small MoleculeI**

Diseases Followed: Over 700, 80% genetically inherited;
Patients followed: 42471. Pediatrics: 29036; Adults 13391
Professionals involved in the MDT: 1681 in 69 HCPs/18 EU Countries

METABERN KICK OFF MEETING FRANKFURT APRIL 23-25, 2017

