

THE EUROPEAN REFERENCE NETWORKS: A VISION FROM INSIDE

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EUROPEAN REFERENCE NETWORK
HEREDITARY METABOLIC DISEASES, MetabERN*

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COORDINATORS GROUP*





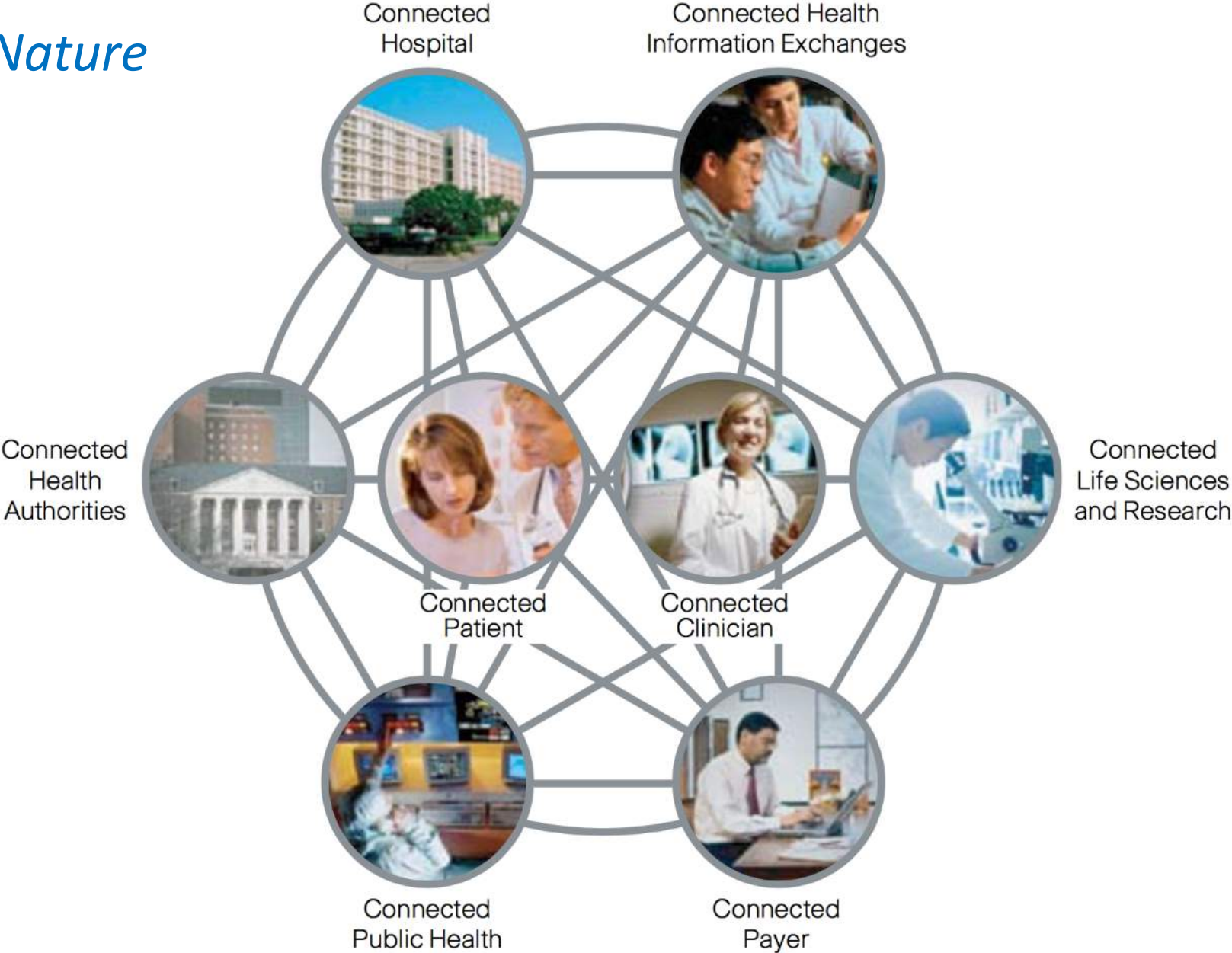
Grouping Rare Diseases In Thematic Networks

- Rare immunological and auto-inflammatory diseases
- Rare bone diseases
- Rare cancers* and tumours
- Rare cardiac diseases
- Rare connective tissue and musculoskeletal diseases
- Rare malformations and developmental anomalies and rare intellectual disabilities
- Rare endocrine diseases
- Rare eye diseases
- Rare gastrointestinal diseases
- Rare gynaecological and obstetric diseases
- Group on Cancer Control

- Rare haematological diseases
- Rare craniofacial anomalies and ENT (ear, nose and throat) disorders
- Rare hepatic diseases
- Rare hereditary metabolic disorders
- Rare multi-systemic vascular diseases
- Rare neurological diseases
- Rare neuromuscular diseases
- Rare pulmonary diseases
- Rare renal diseases
- Rare skin disorders
- Rare urogenital diseases

*Note: The networking of rare cancers is currently under discussion in EC Expert

ERNs Mimick Nature



1. ERN BOND	European Reference Network on Rare Bone Disorders
2. ERN CRANIO	European Reference Network on Rare craniofacial anomalies and ENT disorders
3. Endo-ERN	European Reference Network on Rare Endocrine Conditions
4. ERN EpiCARE	European Reference Network on Rare and Complex Epilepsies
5. ERKNet	European Rare Kidney Diseases Reference Network
6. ERN RND	European Reference Network on Rare Neurological Diseases
7. ERNICA	European Reference Network on Rare inherited and congenital anomalies
8. ERN LUNG	European Reference Network on Rare Respiratory Diseases
9. ERN Skin	European Reference Network on Rare and Undiagnosed Skin Disorders
10. ERN EURACAN	European Reference Network on Rare Adult Cancers (solid tumours)
11. ERN EuroBloodNet	European Reference Network on Rare Haematological Diseases
12. ERN EURO-NMD	European Reference Network for Rare Neuromuscular Diseases
13. ERN EYE	European Reference Network on Rare Eye Diseases
14. ERN GENTURIS	European Reference Network on Genetic Tumour Risk Syndromes
15. ERN GUARD-HEART	European Reference Network on Uncommon And Rare Diseases of the HEART
16. ERN ITHACA	European Reference Network on Rare Congenital Malformations and Rare Intellectual Disability
17. MetabERN	European Reference Network for Rare Hereditary Metabolic Disorders
18. ERN PaedCan	European Reference Network for Paediatric Cancer (haemato-oncology)
19. ERN RARE-LIVER	European Reference Network on Rare Hepatological Diseases
20. ERN ReCONNET	Rare Connective Tissue and Musculoskeletal Diseases Network
21. ERN RITA	Rare Immunodeficiency, Autoinflammatory and Autoimmune Diseases Network
22. ERN TRANSPLANT-CHILD	European Reference Network on Transplantation in Children
23. VASCERN	European Reference Network on Rare Multisystemic Vascular Diseases
24. ERN eUROGEN	European Reference Network on Rare and Complex Urogenital Diseases and Conditions

European Reference Networks Conference

& ERN Kick-off meeting

Vilnius, Lithuania, 9-10 March 2017

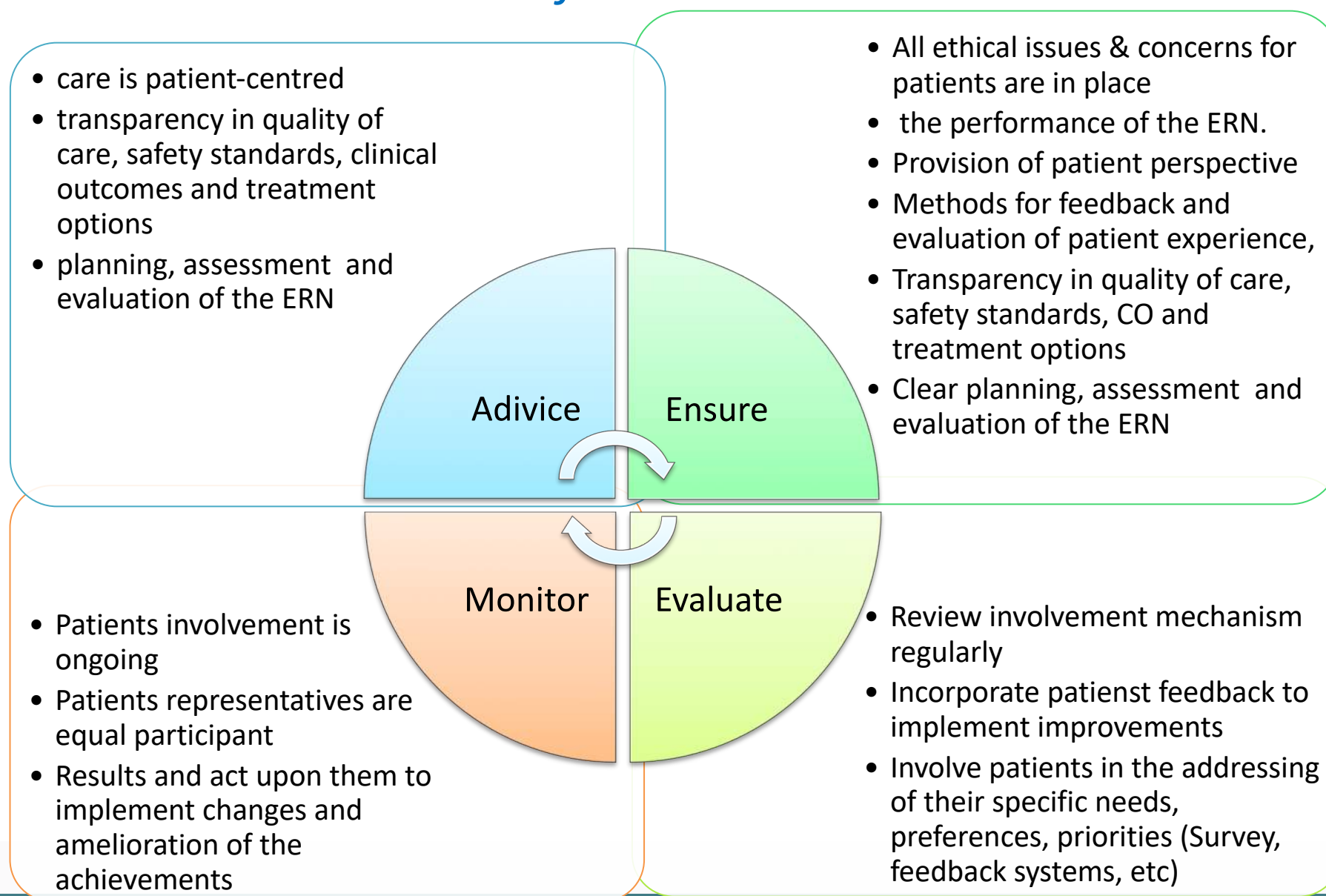




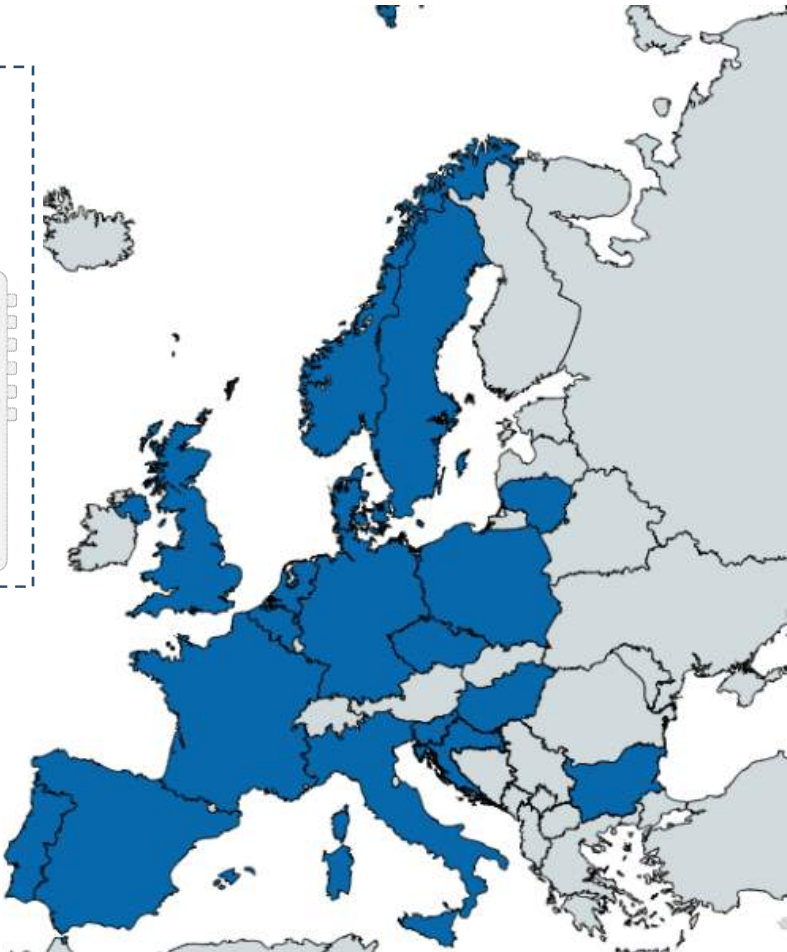
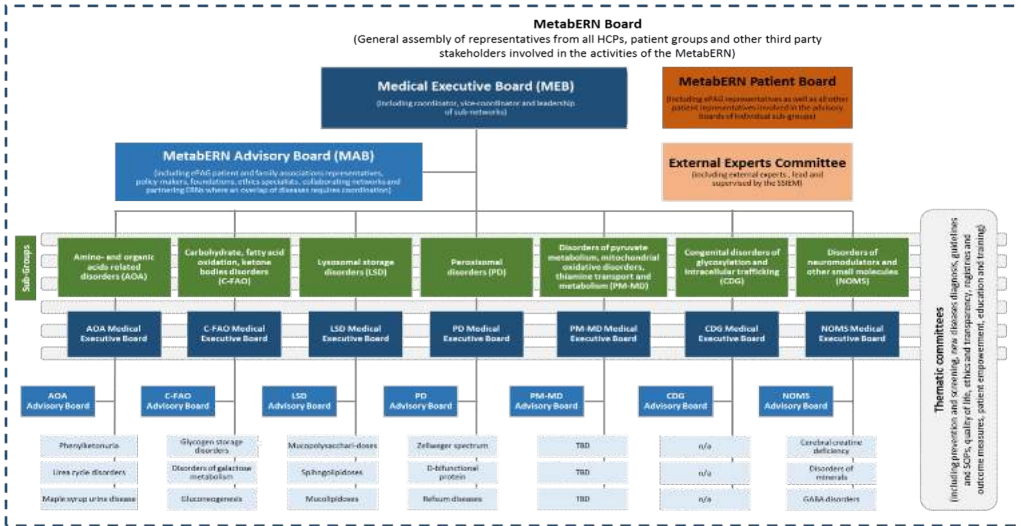
VILNIUS: EUROPEAN REFERENCE NETWORK AWARD

MEMBER STATES	No. ERNS	No. HOSPITALS	No. HCP	MEMBER STATES	No. ERNS	No. HOSPITALS	NUMBER HCP
ITALY	23	66	186	BULGARIA	6	6	7
FRANCE	ALL	38	111	ROMANIA	6	6	7
GERMANY	23	42	121	SLOVENIA	9	3	9
UK	22	35	112	ESTONIA	3	2	3
NETHERLANDS	ALL	13	90	CROATIA	2	1	2
BELGIUM	19	10	36	AUSTRIA	2	1	2
SPAIN	19	16	42	NORWAY	3	2	3
CZECH REPUBLIC	18	8	28	IRELAND	3	1	3
SWEDEN	20	6	30	LATVIA	2	1	2
PORTUGAL	16	8	29	LUXEMBOURG	1	1	1
POLAND	17	12	21	CYPRUS	2	1	2
DENMARK	11	4	20	MALTA	-	-	-
FINLAND	12	4	14	GREECE	-	-	-
HUNGARY	10	4	12	SLOVAK REP.	-	-	-
LITHUANIA	12	2	12	TOTAL		293	905

Involvement of Patients in the ERNs



MetabERN: ERN on HEREDITARY METABOLIC DISEASES
Coordinator
Prof. Maurizio Scarpa MD PhD
Helios Dr. Horst Schmidt Kliniken Wiesbaden,



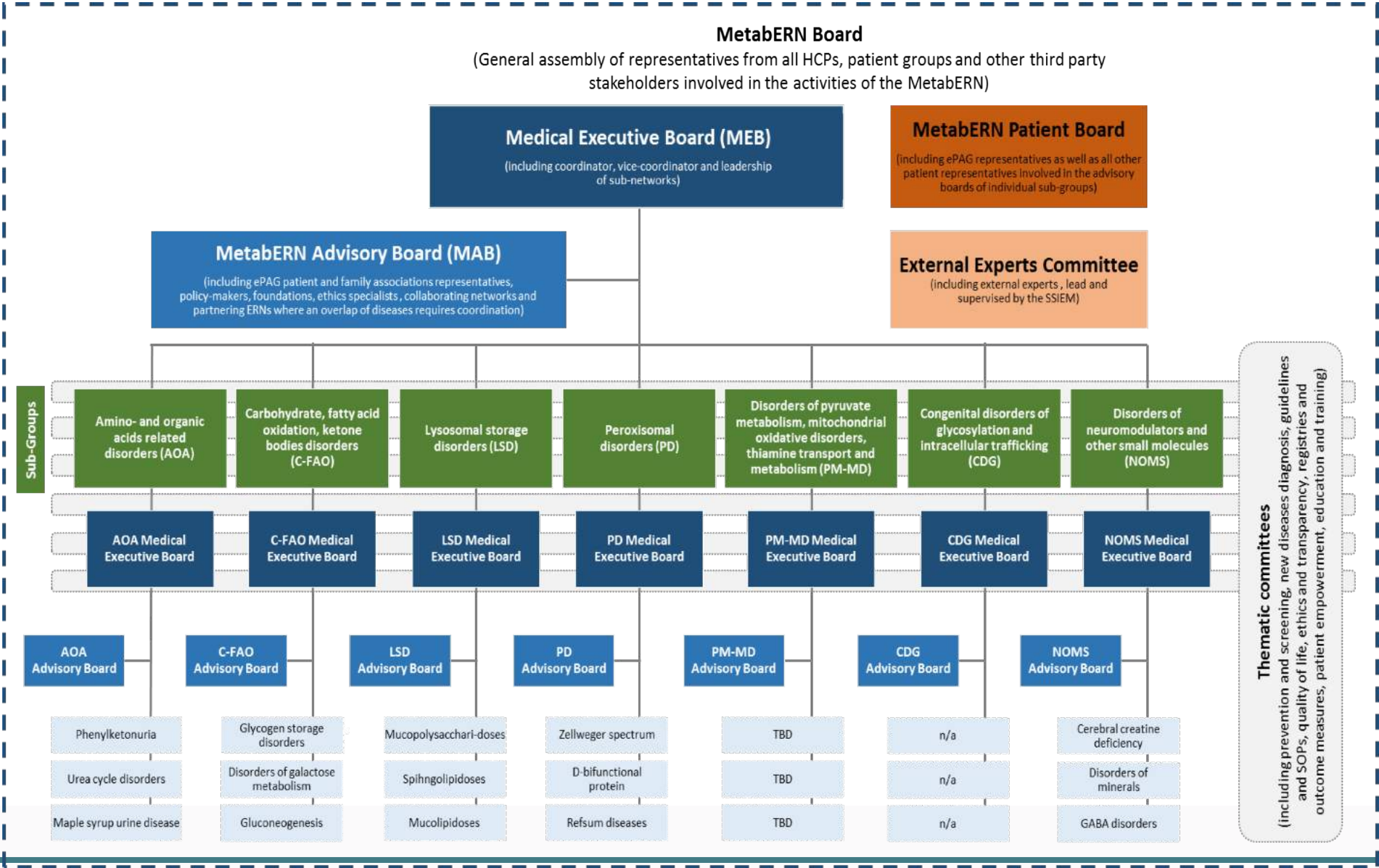
- 1) *Aminoacid and organic acids related disorders*
- 2) *Disorders of pyruvate metabolism, Krebs cycle defects, mitochondria oxidative phosphorylation disorders, disorders of thiamine transport and metabolism*
- 3) *Charbohydrate, fatty acid oxidation and ketone bodies disorders*
- 4) *Lysosomal disorders*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

BE	BG	CZ	DE	DK	ES	FR	HR	HU	IT	LT	NL	NO	PL	PT	SE	SI	UK
6	1	1	10	1	5	9	1	1	11	1	5	2	1	5	2	1	6

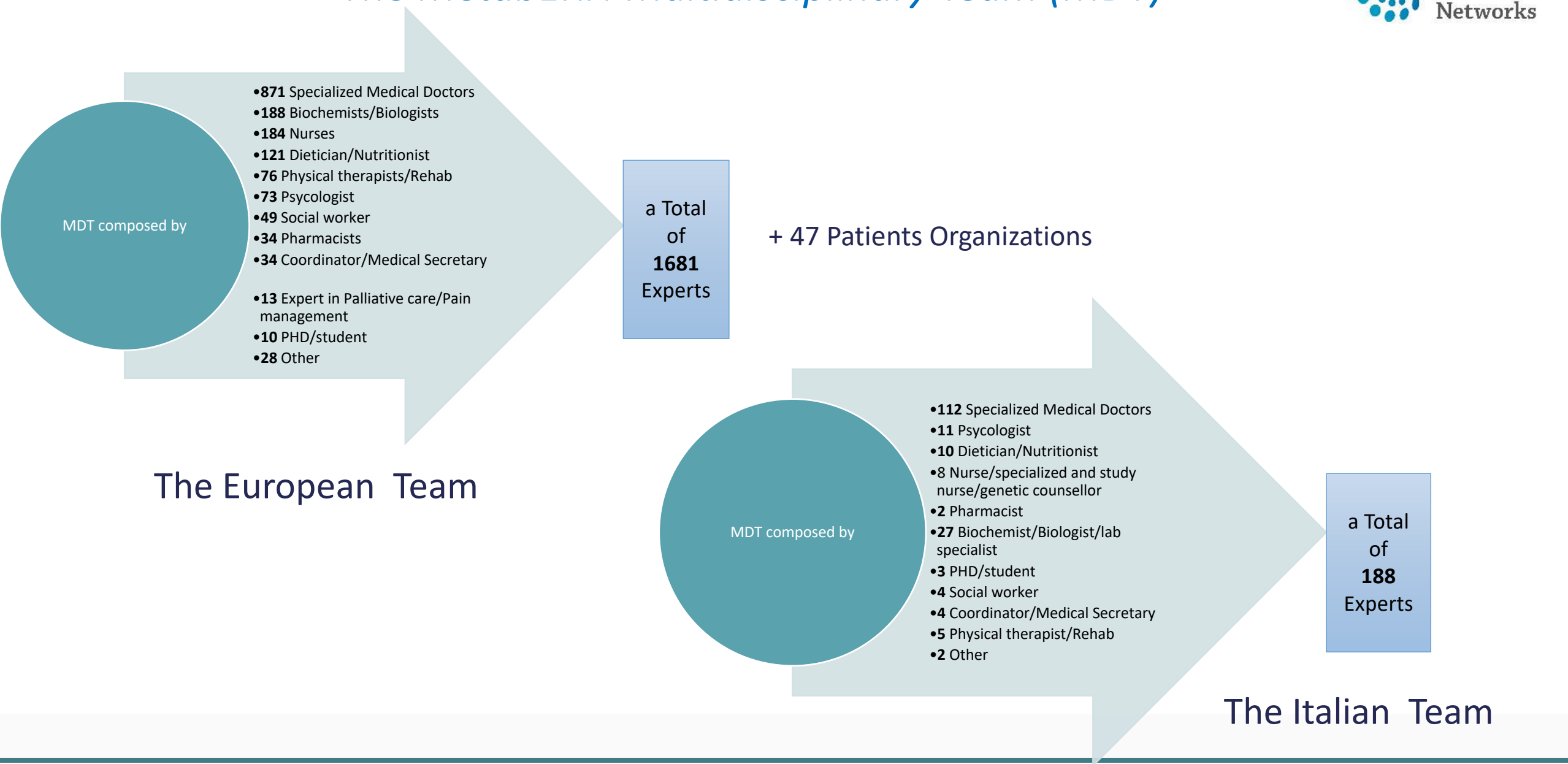
MetabERN List of the Italian HCPs

HCP	LEADER		Vice
ASST Monza San Gerardo Hospital	Serena	Gasperini	Alberto Piperno
Azienda Ospedaliera Universitaria "Federico II", Napoli	Giancarlo	Parenti	Pietro Strisciuglio
Azienda Ospedaliera Universitaria Integrata di Verona	Alessandro	Salviati	Andrea Bordugo
Azienda Ospedaliera Universitaria Senese	Antonio	Federico	Maria Teresa Dotti
Azienda Ospedaliero-Universitaria Pisana	Ferruccio	Santini	Giovanni Ceccarini
Azienda Sanitaria Universitaria Integrata di Udine	Bruno	Bembi	Andrea Dardis
Giannina Gaslini Institute (IRCCS Giannina Gaslini)	Maja	di Rocco	Roberto cerone
Meyer Children's Hospital	Maria Alice	Donati	Elisabetta Pasquini
Ospedale Pediatrico Bambino Gesù	Carlo	Dionisi Vici	Enrico Bertini
San Paolo Hospital, ASST Santi Paolo e Carlo	Graziella	Cefalo	Giuseppe Banderali
University Hospital of Padova	Alberto	Burlina	Giulia Polo

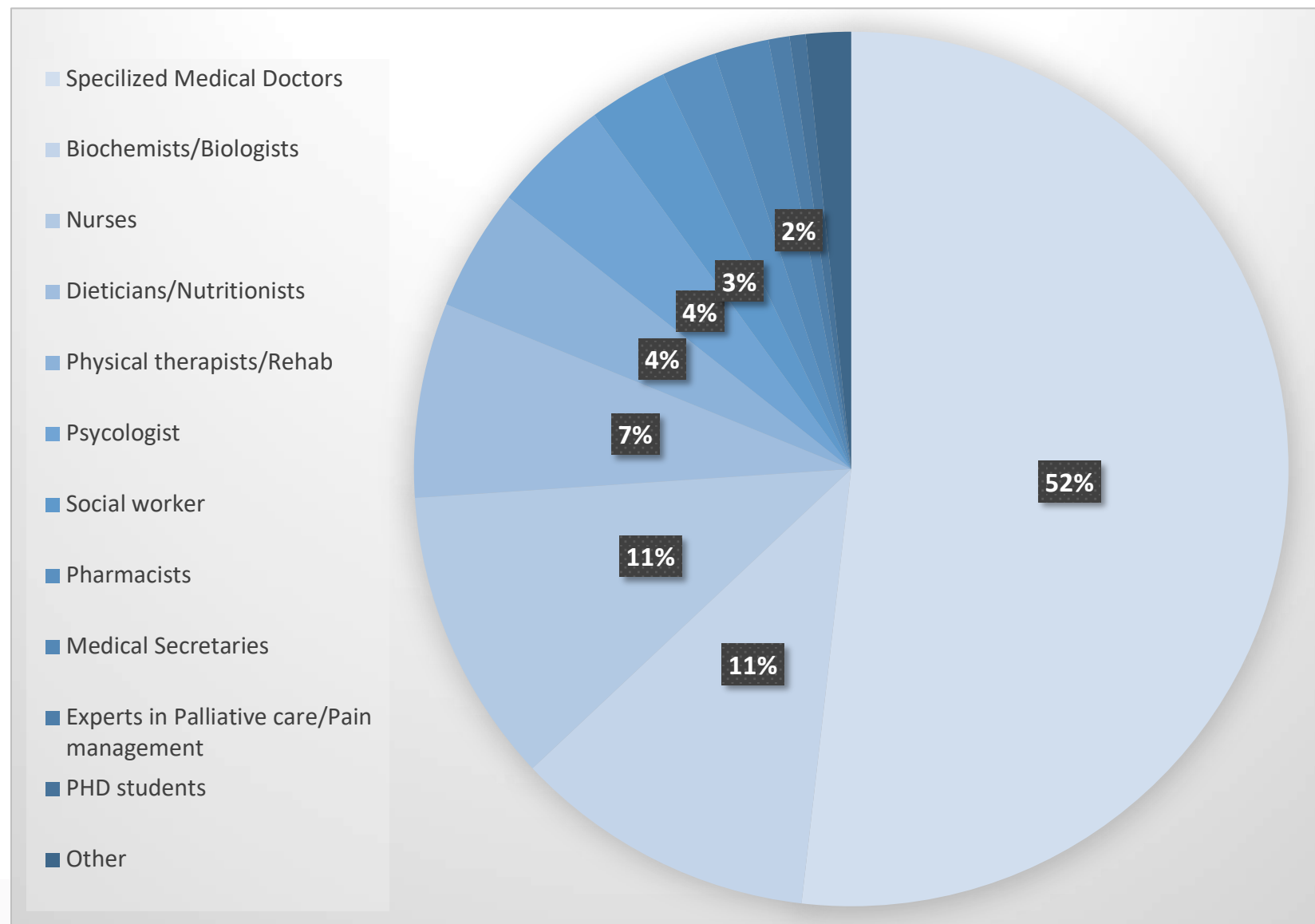
MetabERN Structure



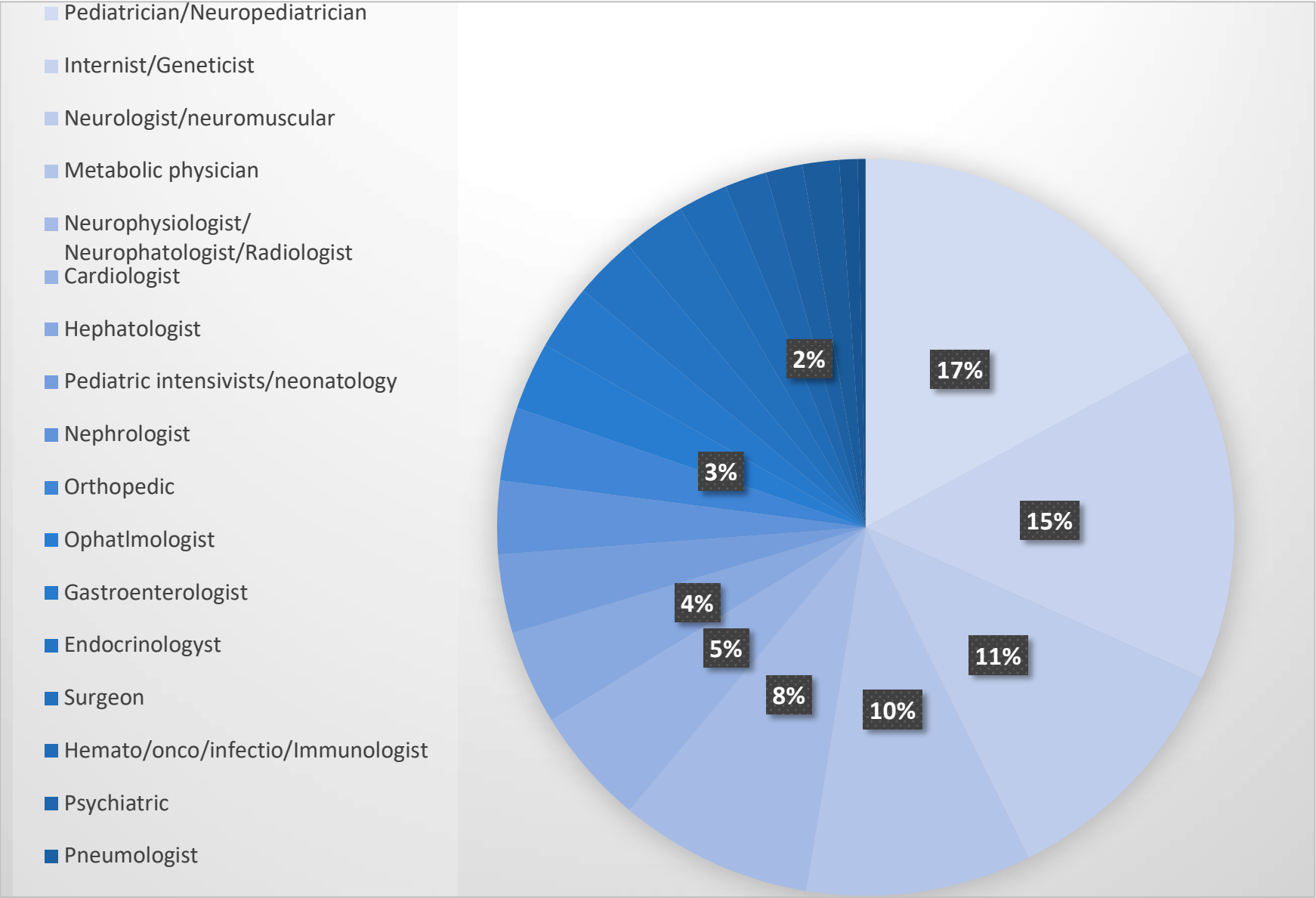
The MetabERN Multidisciplinary Team (MDT)



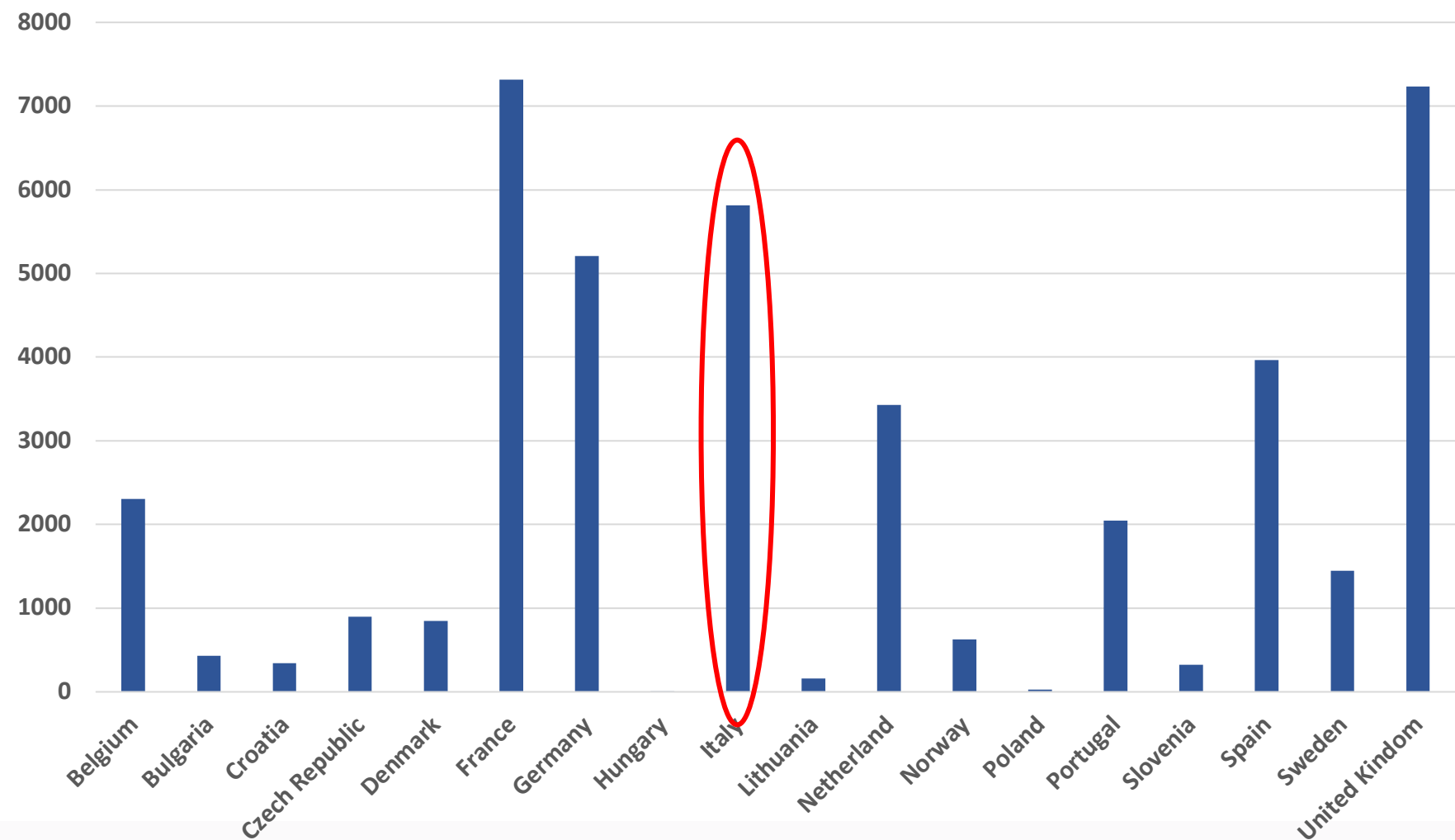
Composition of the MetabERN MDT



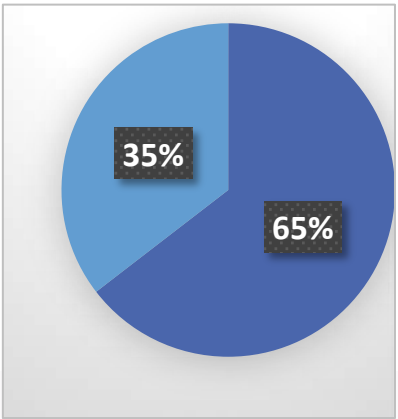
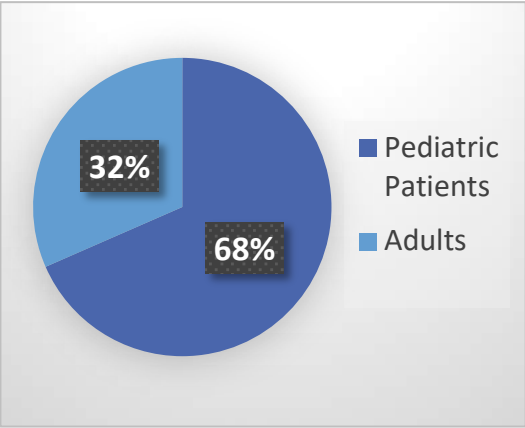
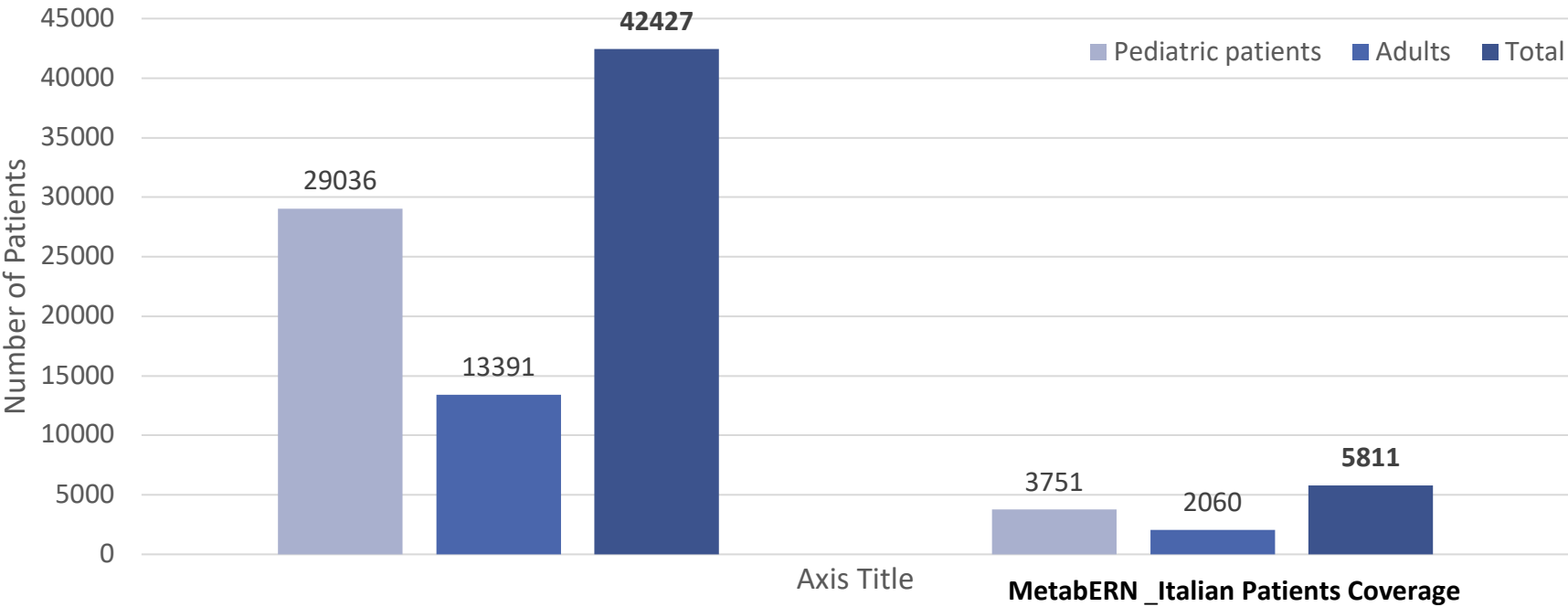
Composition of the MetabERN Specialists



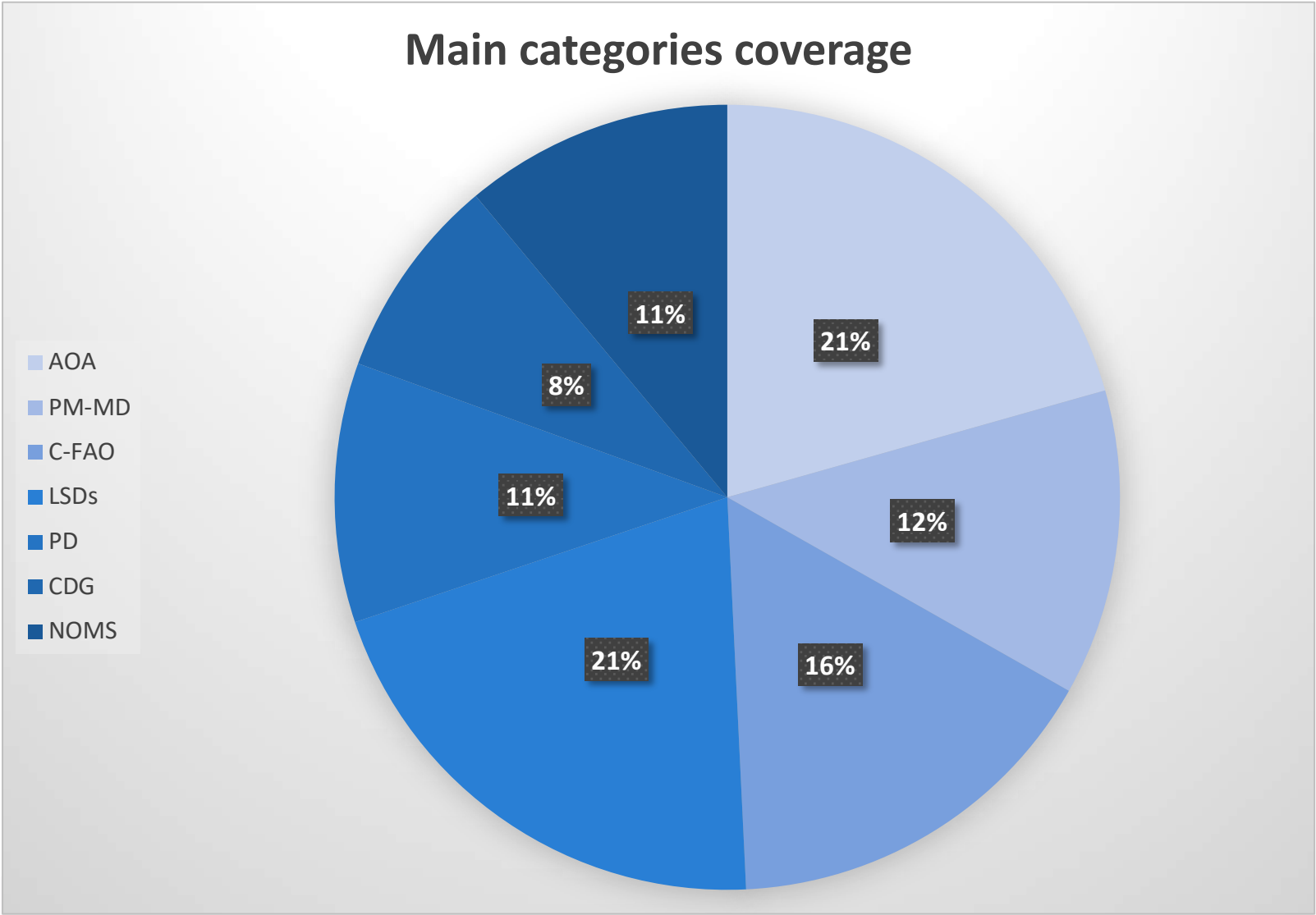
MetabERN: Number of Patients per Country



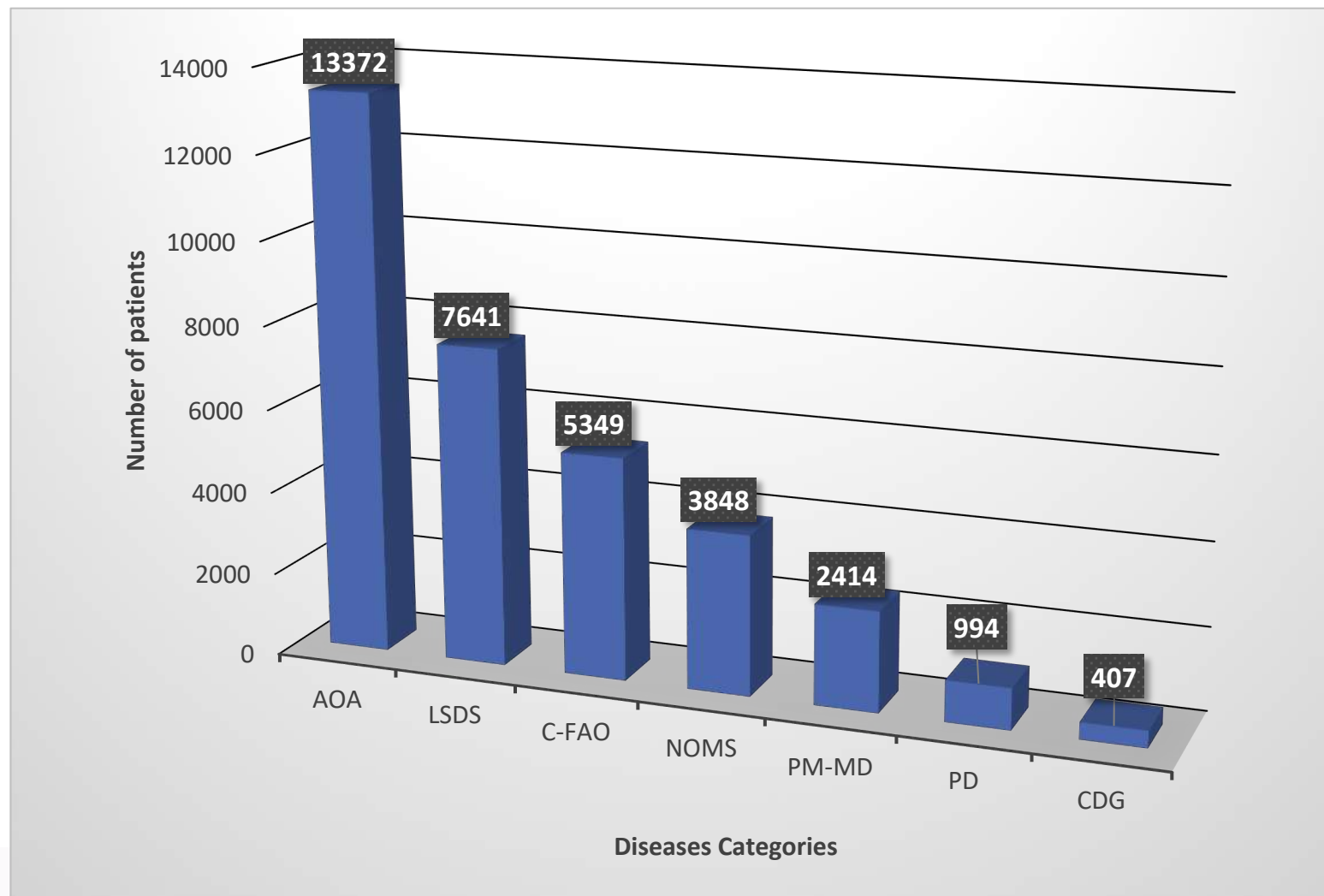
MetabERN: Patients Coverage

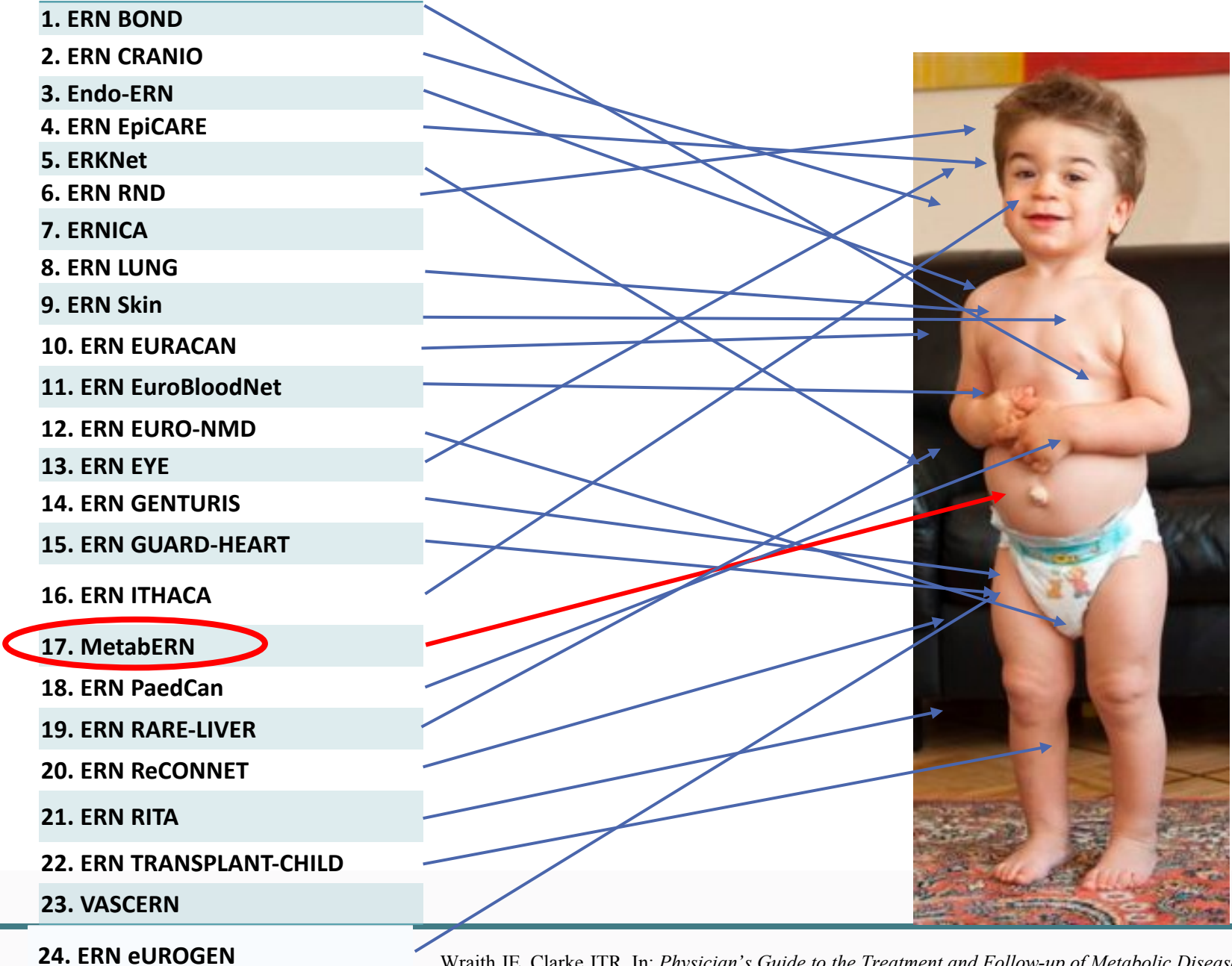


MetabERN: Main Diseases Categories



Patients Managed by MetabERN in the last year





ERN ACTIVITIES

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

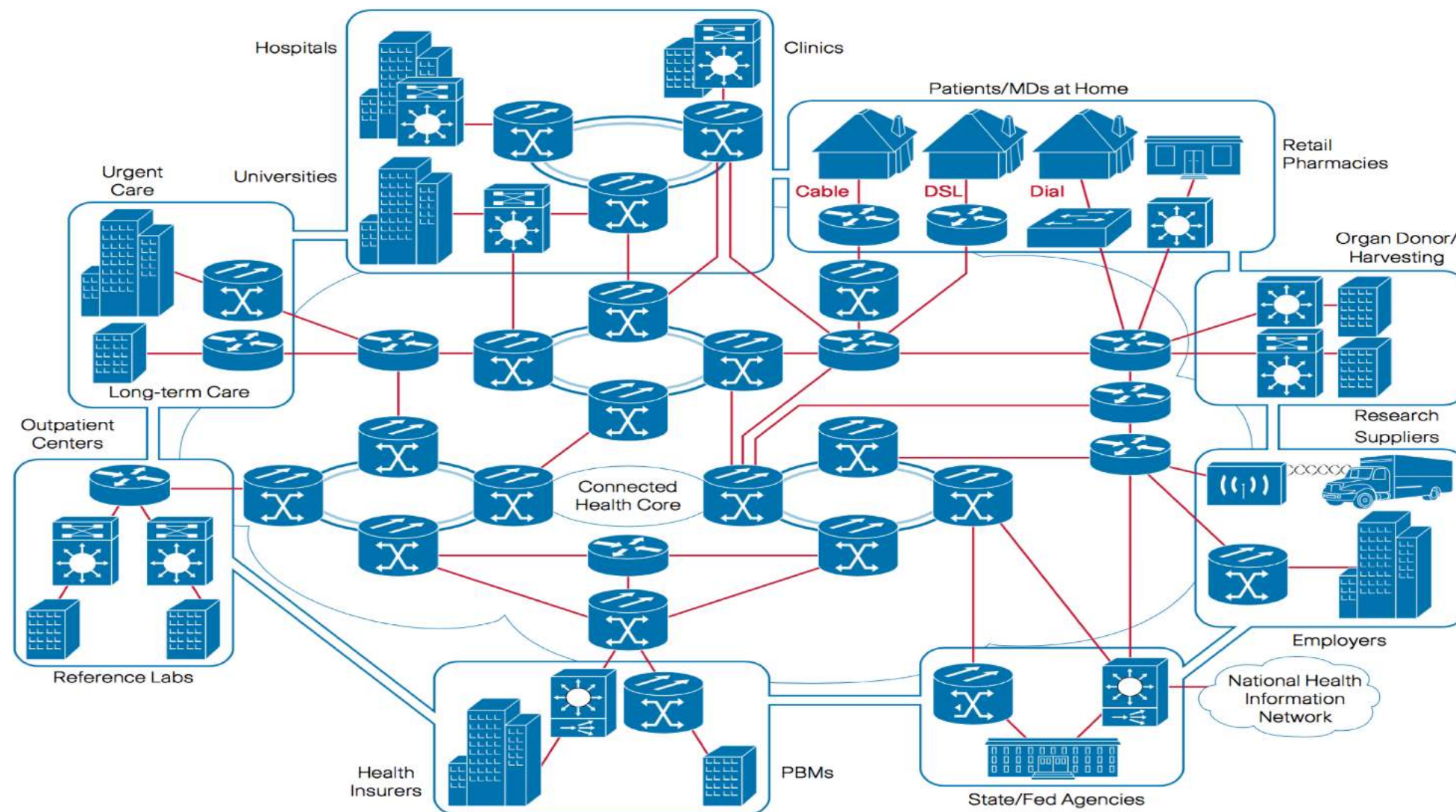
Patient Empowerment

Clinical Trials

Research

Dissemination/Stakeholders Relations

One Of The Secrets For Success Of The ERN Crosslinking : The IT Platform



Major Opportunities Offered By The ERNs Health System

- Close contact with regulatory authorities, key opinion leaders, patient advocacy organizations. and non-traditional players, like technology companies, to create a new coordinated way to manage patients affected by rare diseases and optimize access to diagnosis and therapy.
- Impact on the National Health System to spare resources, optimize services and avoid waste of expertise
- Impact on the awareness for rare diseases. Creation of an European epidemiology on rare diseases, common registries, databases, research projects.
- Impact on the awareness for cost of rare diseases. Creation of EU HTAs for the calculation of the cost of the management of patients with rare diseases and prediction of sustainability.
- Impact on the discussion on the sustainability of orphan drugs.
- Interaction with National Health Institute and Drug agency for postmarketing analysis.

Major Opportunities Offered By The ERNs Patients

- Optimization of care pathways for patients with rare disease: diagnosis, management access to therapy
- More effective clinical trial designs that consider the very special and unique circumstances of rare disease research
- Creation of databases/registries/natural histories of patients in a coordinated way
- Close interaction with family associations which may help logistical challenges associated with conducting trials for small patient population
- Close interaction among ERNs to integrate expertise needed for clinical trials
- Possibility of helping Companies to generate new clinical trial designs based on the right choice of the primary and secondary endpoints based on the well characterised patient population followed by the ERN.
- Possibility of generating studies also for ultrarare patients due to the availability of numbers of patients in a coordinated way
- Protection of patients from not well designed clinical trials
- Counselling the patients in the understanding the value of the clinical trials

The ERN Coordinators Group

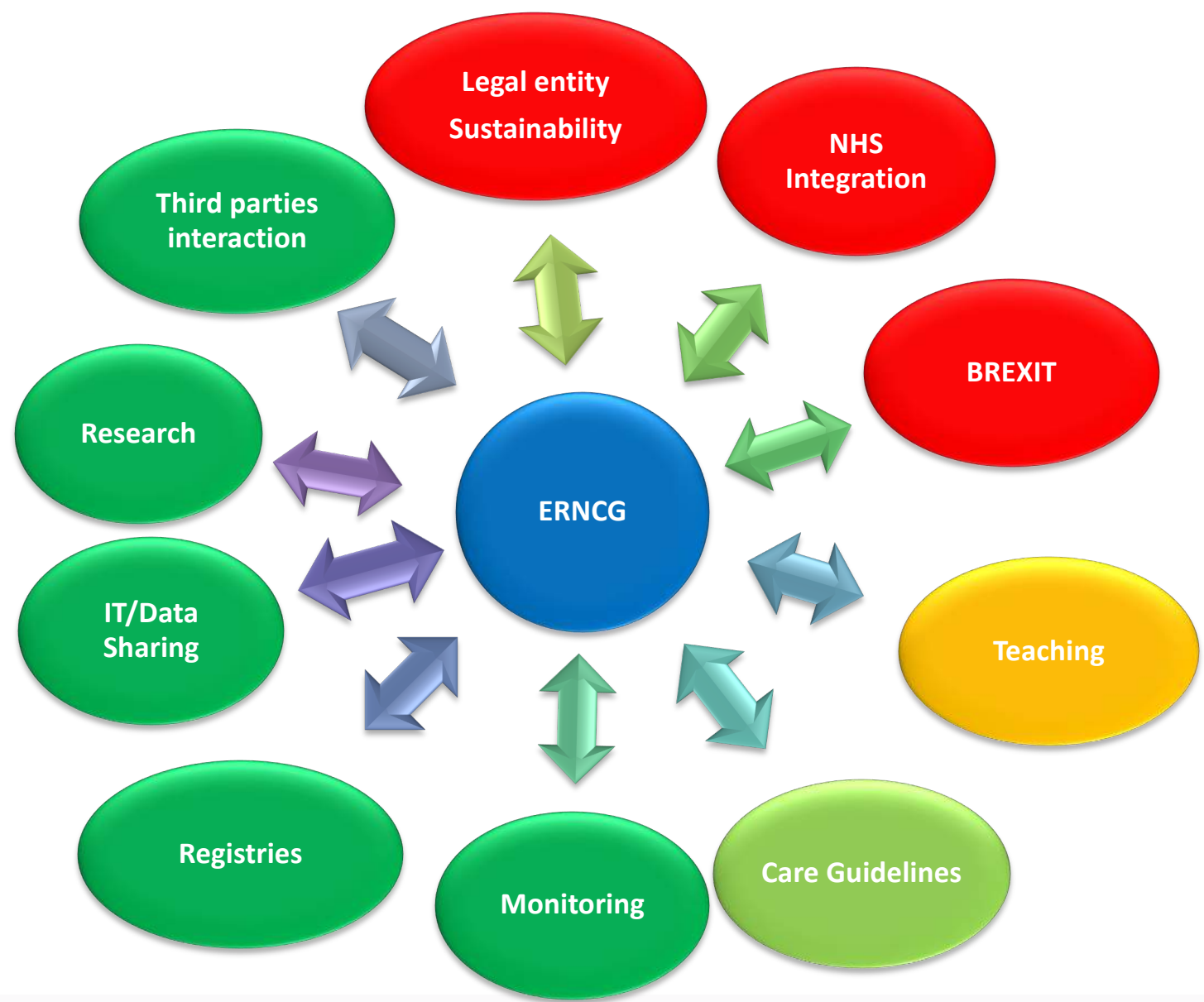
Chair: Maurizio Scarpa, MetabERN

Vice-Chair: Franz Schäfer, ERKNET

Board: WG Coordinators

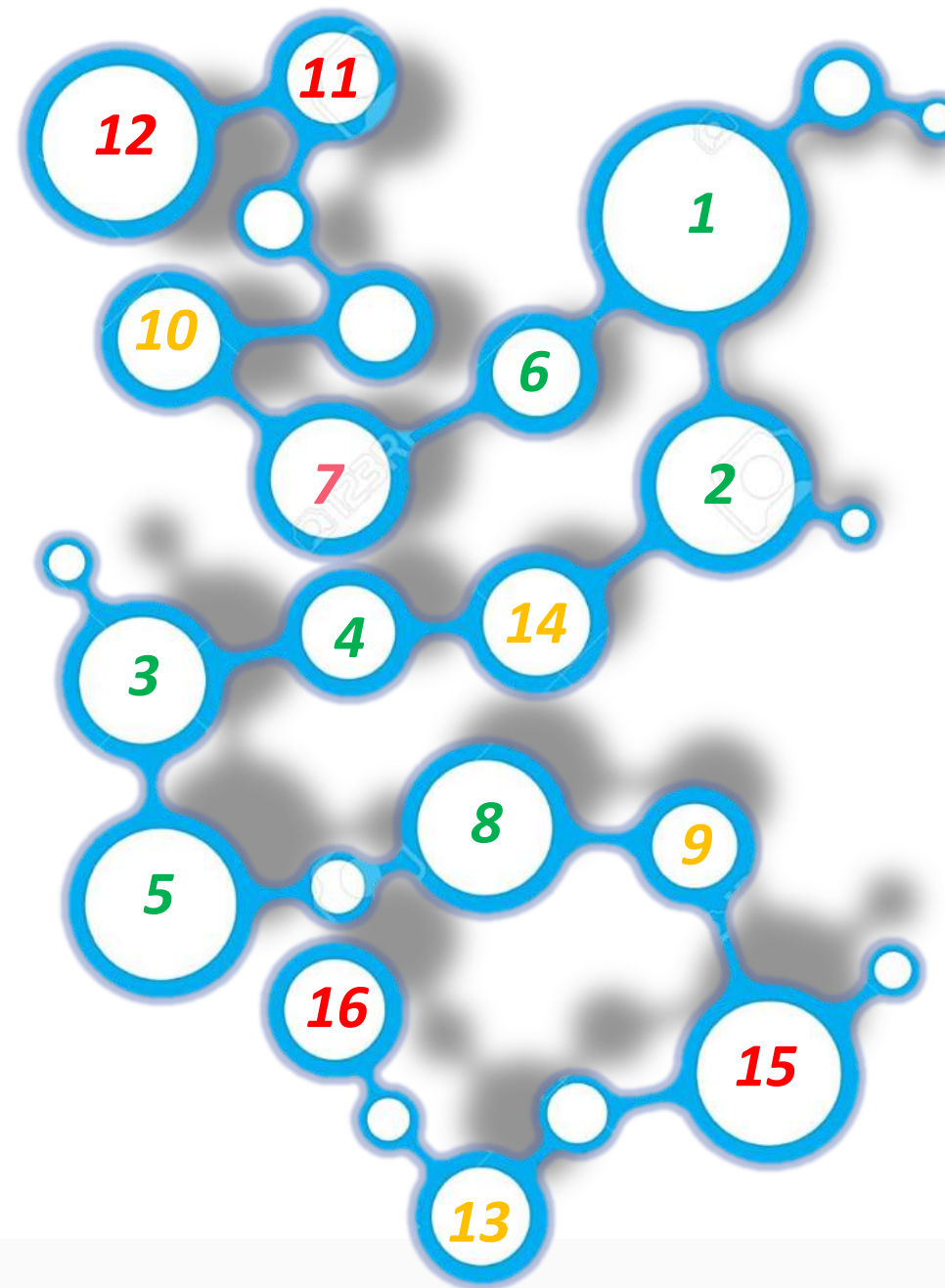
WG	LEADERSHIP
IT & DATA-SHARING	Ruth Ladenstein, ERNPaedcan
RESEARCH	Eduardo López Granados, ERN TransplantChild
LEGAL ASPECTS, DATA PROTECTION & ETHICS	Nicoline Hoogerbrugge, GENTURIS
CROSS-BORDER HEALTHCARE AND BUSINESS CONTINUITY	Holm Graessner, ERN-RND; Kate Bushby, EURO NMD
MONITORING & ASSESSMENT	Christopher Chapple, EUROGEN
GUIDELINES, EDUCATION, TRAINING	Paolo Casali, ERN EURACAN
SPECIAL PROJECTS: NHS INTEGRATION SUSTAINABILITY	Maurizio Scarpa, MetabERN

Achievements



The ERN Interactome: Coordination of the Activities

1. ERNs COORDINATION
2. PATIENT COMMITMENT
3. DISEASE IDENTIFICATION
4. REGISTRY
5. DATA SHARING
6. COMMUNICATION/AWARENESS
7. EDUCATION
8. SHARED VIRTUAL COUNSELLING
9. NETWORK OF SPECIALISTS
10. INSTITUTIONAL STAKEHOLDERS
11. PAYERS
12. NATIONAL HEALTH SYSTEMS/NPRD
13. RESEARCH COORDINATION
14. GUIDELINES/PATIENT PATHWAYS
15. STANDARD OPERATING PROCEDURES
16. QUALITY INDICATORS





25th Workshop of the EURORDIS Round Table of Companies (ERTC)

Healthcare Companies & European Reference Networks:
Expectations & Potential for Collaboration



PROGRAMME

Morning session co-chaired by:

Chairpersons: Ms. Claudia Crocione, Managing Director, HHT Europe, Italy & Pr. Josep Torrent-Farnell, Head of Medicines Division, CatSalut (Catalan Health Service), Generalitat de Catalunya, Spain

Morning Session: Exploring opportunities	
09:00 – 09:20	Welcome introduction, setting the scene & goals for the day: A reminder of why we are all here. Our shared ambition for ERNs. Mr. Yann Le Cam , Chief Executive Officer, EURORDIS, France/Belgium
09:20 – 09:35	State of the art of ERNs: What they are (core responsibilities), how they are structured and organised and their overall vision Mr. Jaroslaw Waligora , Policy Officer, European Commission
09:35 – 09:45	Board of Member States Reflections Prof. Nicoline Hoogerbrugger , Chair, EC ERN Committee on Data Protection, Ethics & Informed Consent, The Netherlands
09:45 – 09:55	From concept to reality: European Reference Network for Hereditary Metabolic Diseases, MetabERN Dr. Maurizio Scarpa , Clinical lead for Rare Metabolic Diseases, Director of the Centre for Rare Diseases Helios Dr Horst Schmidt Clinic, Germany
09:55 – 10:05	Round table Q&A: Current state of play, needs and expectations, looking ahead Moderator: Pr. Josep Torrent-Farnell, Head of Medicines Division, CatSalut (Catalan Health Service), Generalitat de Catalunya, Spain Panelists: Mr. Jaroslaw Waligora , Policy Officer, European Commission Dr. Maurizio Scarpa , ERN Chairperson, Clinical lead for Rare Metabolic Diseases, Director of the Centre for Rare Diseases Helios Dr Horst Schmidt Clinic, Germany Prof. Nicoline Hoogerbrugger , Professor in Hereditary Cancer, Radboud University Medical Center, The Netherlands & Chair, EC ERN Committee on Data Protection, Ethics & Informed Consent



20TH European Health Forum Gastein

Health in All Politics –
a better future for Europe



- 1) WORKSHOP ON PERSONALISED MEDICINE AND RD
- 2) WORKSHOP ON SUSTAINABILITY OF ORPHAN DRUGS
- 3) WORKSHOP ON BIG DATA
- 4) MEETING WITH THE COMMISSIONER:
 - LEGAL ENTITY
 - SUSTAINIBILITY

Work in Progress for the Coordinators

- Continuous collaboration with the BoMS representatives
- Sustainability of the ERNs
- Integration into National Health Systems
- Legal entity of the ERNs
- Reimbursement of the ERN activities
- Liability of Coordinators/ recognition of the Coordinator role

Conclusions

- ✓ ERNs are NOT a project or a program, they are a CONCEPT* and represent the major achievement of the European spirit of collaboration, sharing and health investment in the field of rare diseases
- ✓ ERNs group the most acknowledged centers of excellence in Europe, all sharing the same level of commitment and interest for rare diseases
- ✓ ERNs are NOT single networks, but rather a critical mass of about 300 hospitals and over 1000 specialised units, cross feeding each other to meet the needs of our patients.
- ✓ Crosslinking is in the nature of the ERNs due to multidisciplinary implications of the cared diseases.
- ✓ The ERNs' integration inside the MS National Health Systems/National Plans for Rare Diseases will represent a major achievement for the Cross Border Care
- ✓ The ERNs'/MS National Health Systems/National Plans for Rare Diseases integration is instrumental to define strategies to optimize the cost of management of patients of rare diseases
- ✓ The ERN Coordinators Group is an unvaluable instrument for the EC/ National Health Systems/National Plans for Rare Diseases to define a uniform pan-European plan for rare diseases.

* Quoted after Victoria Hedley, Newcastle, UK