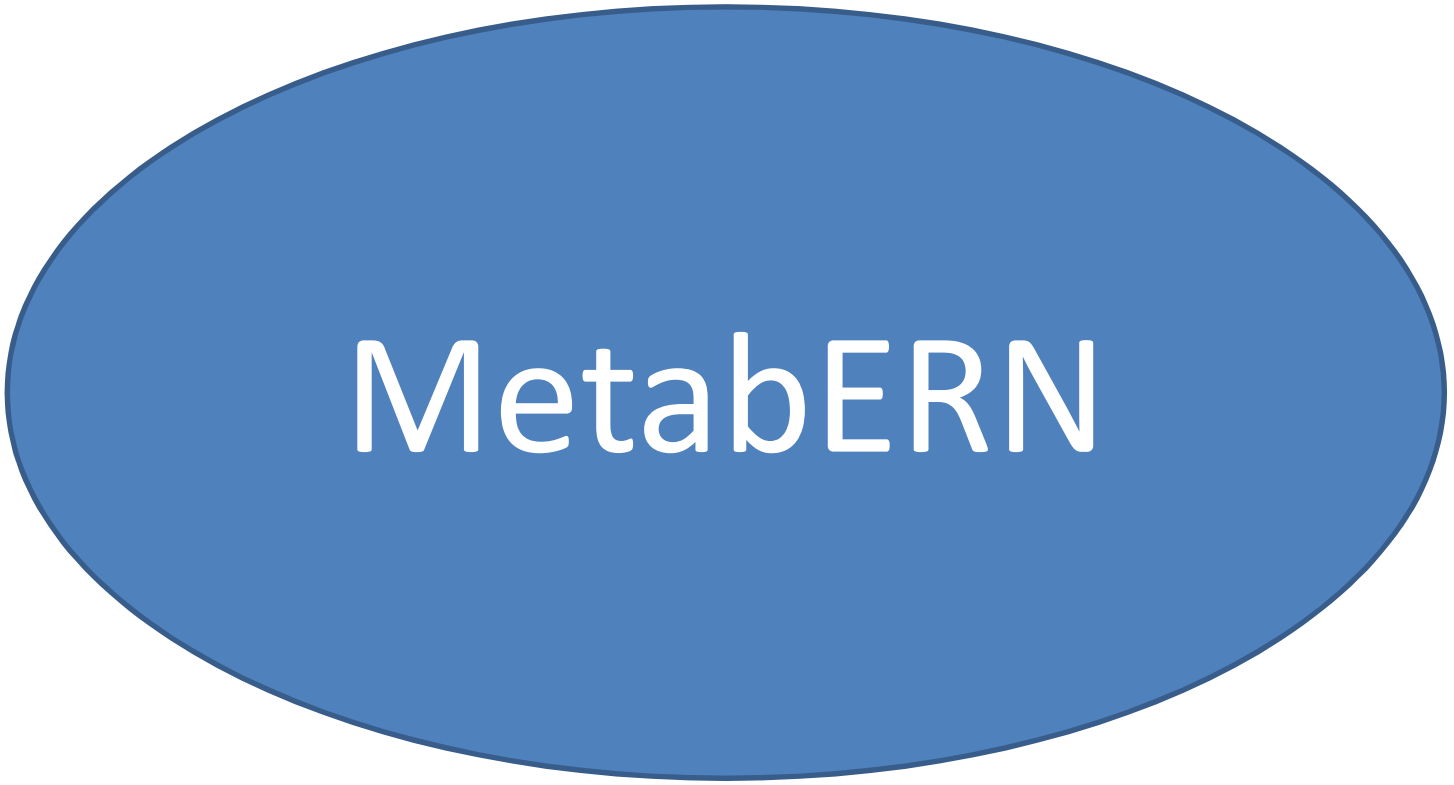
Role of ERNDIM in MetabERN

Viktor Kožich

with help of

Maurizio Scarpa


MetabERN

European Reference Network
for rare or low prevalence complex diseases
Network Hereditary Metabolic Disorders (MetabERN)

THE ORIGIN OF THE ERNS

Slide kindly provided by Enrique Terol, EU Commission

European Reference Networks

APPROVED ERNs

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 Network
for rare or low prevalence complex diseases
Network Hereditary Metabolic Disorders (MetabERN)

MetabERN: ERN on HEREDITARY METABOLIC DISEASES

Coordinator

Prof. Maurizio Scarpa MD PhD

Helios Dr. Horst Schmidt Kliniken Wiesbade

69 HCPs from 18 COUNTRIES

The MetabERN is endorsed by and partners with the Society for the Study of the Inborn Errors of Metabolism (SSIEM) and with ERNDIM

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

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METABERN MULTIDISCIPLINARY TEAM (MDT)

MDT composed of

- **871** Specialized Medical Doctors
- **188** Biochemists/Biologists
- **184** Nurses
- **121** Dieticians/Nutritionists
- **76** Physical therapists/Rehab
- **73** Psycologists
- **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

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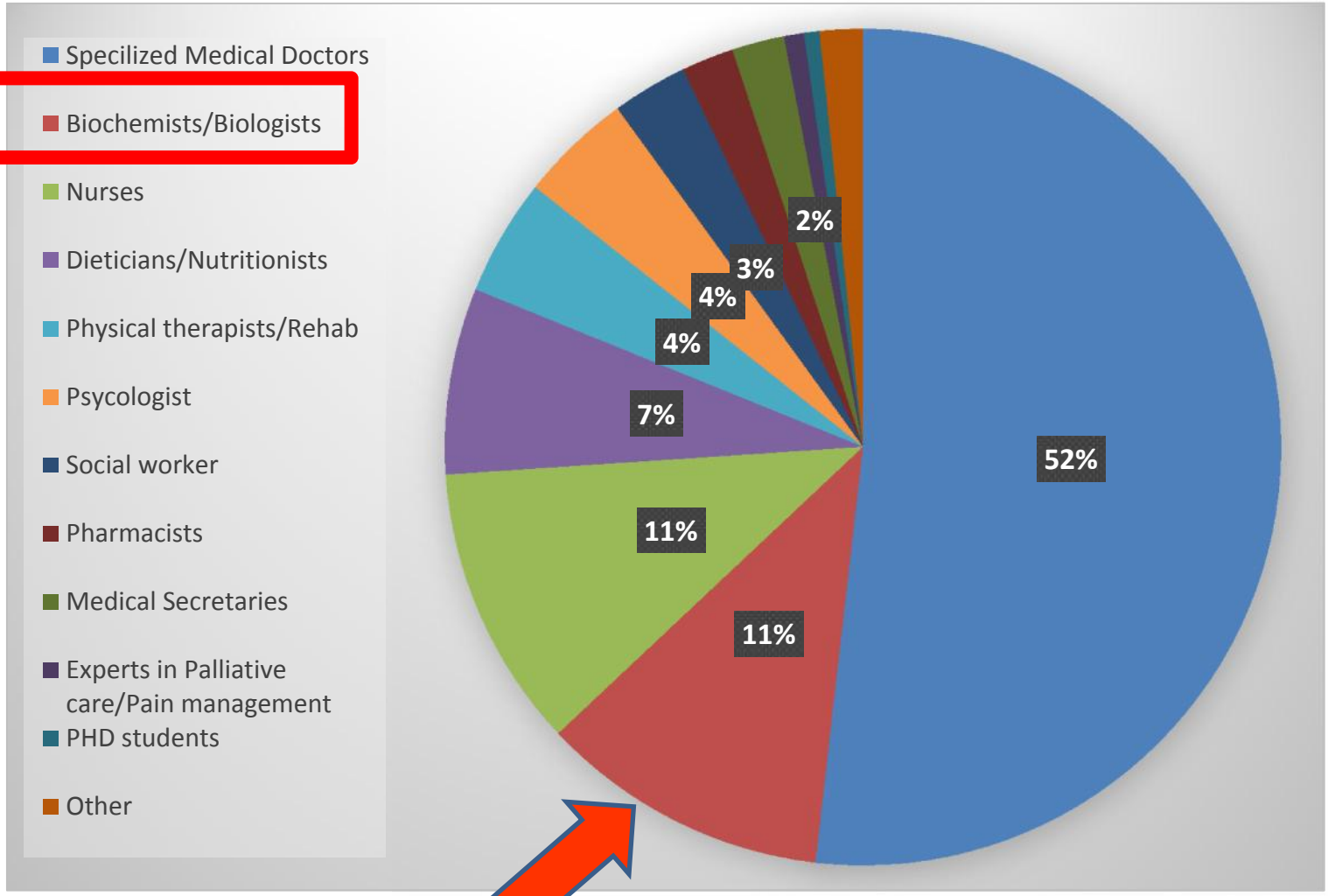
COMPOSITION OF THE METABERN MDT



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AIMS OF METABERN

- ✓ To pool knowledge and improve information exchange between network partners;
- ✓ To improve prevention, diagnosis and care in disease areas where expertise is rare;
- ✓ To support Member States with a small number of patients to provide highly specialised care;
- ✓ To advance innovation in medical science and health technologies;
- ✓ To provide cross-border medical training and research.

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Network Hereditary Metabolic Disorders (MetabERN)

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, leadership of sub-networks) + leadership of the WPs

MetabERN Advisory Board (MAB)

(including the SSIEM, ePAG patient and family associations representatives, relevant associations such as the European Metabolic Disorders Alliance, former and current policy-makers, foundations, ethics specialists , collaborating networks and partnering ERNs where an overlap of diseases requires coordination)

MetabERN Patient Board

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

Sub-

Amino- and organic acids related disorders (AOA)	Carbohydrate, fatty acid oxidation, ketone bodies disorders (C-FAO)	Lysosomal storage disorders (LSD)	Peroxisomal disorders (PD)	Disorders of pyruvate metabolism, mitochondrial oxidative disorders, thiamine transport and metabolism (PM-MD)	Congenital disorders of glycosylation and intracellular trafficking (CDG)	Disorders of neuromodulators and other small molecules (NOMS)
AOA Medical Executive Board	C-FAO Medical Executive Board	LSD Medical Executive Board	PD Medical Executive Board	PM-MD Medical Executive Board	CDG Medical Executive Board	NOMS Medical Executive Board
AOA Advisory Board	C-FAO Advisory Board	LSD Advisory Board	PD Advisory Board	PM-MD Advisory Board	CDG Advisory Board	NOMS Advisory Board
Phenylketonuria	Glycogen storage disorders	Mucopolysaccharidoses	Zellweger spectrum	TBD	n/a	Cerebral creatine deficiency
Urea cycle disorders	Disorders of galactose metabolism	Spihngolipidoses	D-bifunctional protein	TBD	n/a	Neurotransmitter Disorders
Maple syrup urine disease	Gluconeogenesis	Mucolipidoses	Refsum diseases	TBD	n/a	GABA disorders

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)

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Sub-Network Coordinators

Sub-network	Coordinator
Amino and organic acids-related disorders (AOA)	**Henk Blom** **Stefan Kölker** **Francjan van Spronsen**
Disorder of pyruvate metabolism, Krebs cycle defects, mitochondrial oxidative phosphorylation disorders, disorders of thiamine transport and metabolism (PM-MD)	**Enrico Bertini** **Shamina Rahman** **Manuel Schiff**
Carbohydrate, fatty acid oxidation and ketone bodies disorders (C-FAO)	**Carlo Dionisi** **Terry Derks** **Ute Spieterkötter**
Lysosomal storage disorders (LSD)	**Ans van der Ploeg** **Giancarlo Parenti** **Dominique Germain**
Peroxisomal disorders (PD)	**Bwee-Tien Poll The** **Linda De Meirleir** **François Eyskens**
Congenital disorders of glycosylation and disorders of intracellular trafficking (CDG)	**Eva Morava** **Pascale de Lonlay** **Thomas Honzik**
Disorders of Neuromodulators and Other Small Molecules (NOMS)	**Angela Garcia Cazorla** **Thomas Opladen** **Eliane Sardh**

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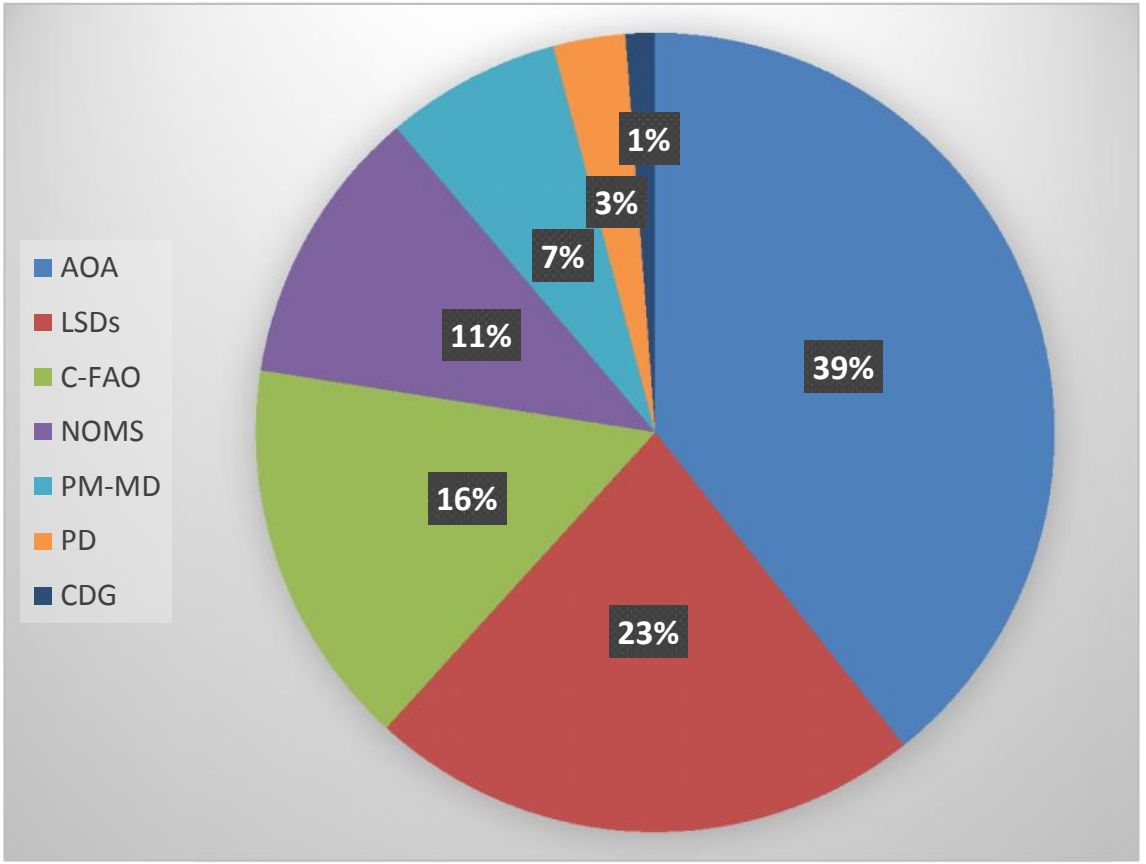
PATIENTS MANAGED BY METABERN



European Reference Network
for rare or low prevalence complex diseases

8 Work Packages

	WORK PACKAGE	MAIN OBJECTIVE	Leader
1	**Coordination and Management**	Ensuring timely execution of the Multiannual Plan	**Maurizio Scarpa (DE)**
2	**Dissemination**	Providing access to information to different target audiences and ensuring timely execution of the MAP	**Eva Morava (BE)**
3	**Evaluation**	Ensuring timely execution of the Multiannual Plan	**Viktor Kozich (CZ)**
4	**Guidelines, Care Pathways & Standardisation**	Pooling knowledge and improving information	**Ursula Plöckinger (DE)** **Carlo Dionisi Vici (IT)**
5	**Virtual Counselling Framework**	Advancing innovation in health technologies for IMDs	**Klaus Mohnike (DE)**
6	**Research, Translational Activities & CT**	Advancing innovation in medical science	**Maurizio Scarpa (DE)**
7	**Capacity-building & Training**	Increasing knowledge and skilling up competencies of target groups and MS to provide highly specialised care	**Nadia Belmatoug (FR)**
8	**Continuity of Care**	Improving prevention, diagnosis and care	**Shamima Rahman (UK)**

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COLLABORATION WITH PATIENTS ASSOCIATIONS

- The patients and patients´ empowerment are at the center of the interests of the MetabERN.
- We have identified 47 PO at national and international level
- 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.

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CONCLUSION

- The MetabERN is an unique opportunity to impact the life of patients and the management of their diseases
- MetabERN represents the first opportunity for all the expert centres to really work together in a coordinated way, in a multidisciplinary way, to share data, expertise, projects and cross feeding, to better meet the patients´need
- The MetabERN is the first opportunity to show that expertise can travel to patients and not always vice versa.

For more information, please contact:

Maurizio.scarpa@metab.ern-net.eu


ERNDIM

ERNDIM

QUALITY ASSURANCE IN LABORATORY TESTING FOR IEM

ERNDIM

European **R**esearch **N**etwork for evaluation and improvement of screening, **D**iagnosis and treatment of **I**nherited disorders of **M**etabolism

ERNDIM: Aims

ERNDIM
QUALITY ASSURANCE IN LABORATORY TESTING FOR IEM

- ERNDIM began in 1994 as an EC funded project (BIOMED-1)
- Main aim was to reach a consensus between European Biochemical Genetics Centres on reliable and standardised procedures for diagnosis, treatment and monitoring of inherited metabolic diseases

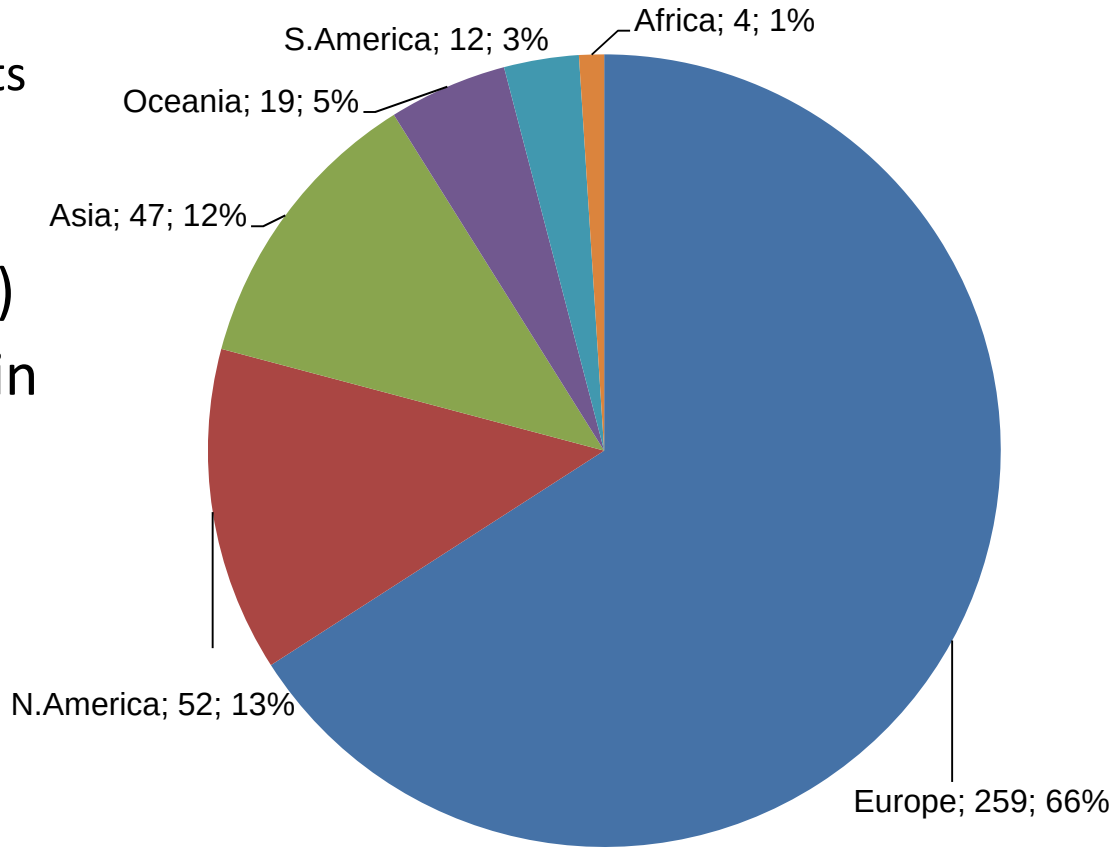
Present ERNDIM Activities

ERNDIM
QUALITY ASSURANCE IN LABORATORY TESTING FOR IEM

- **Quality control schemes** on a worldwide scale (external quality assurance, EQA)
- **Education**
  - Publication of recommended operating procedures
  - Publication of annual reports
  - Support of meetings at the national level
  - Organization of DPT meetings and workshops during the annual SSIEM symposium
  - Co-organizatin of SSIEM Academy

ERNDIM: participation

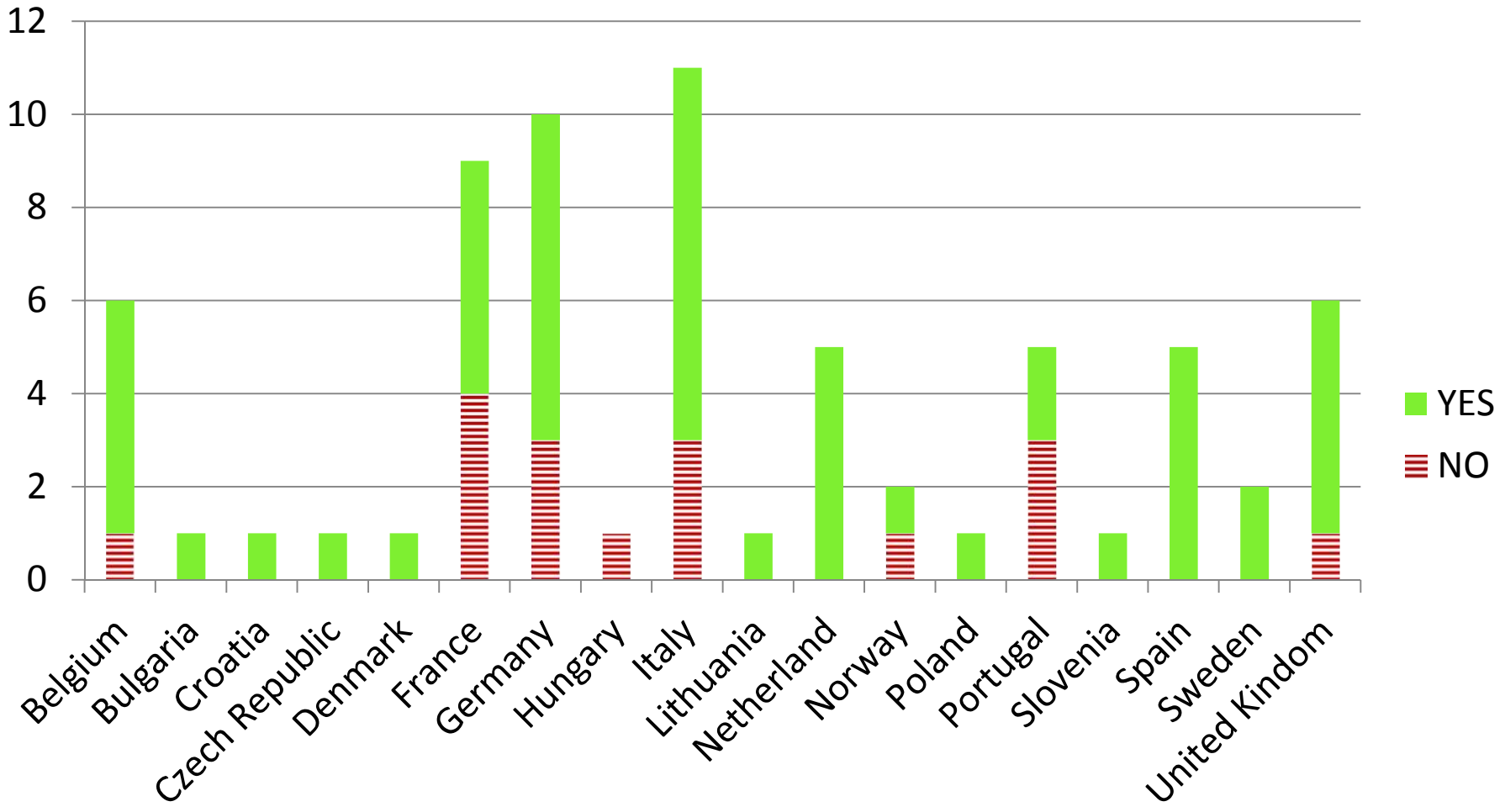
- In 1994 - an EC funded project
  - 3 DPT schemes
  - 162 European participants
- Number of QA schemes increased to **15** (in 2017)
- Number of participants in 2016 was **393** from **59** different countries
- Total number of participations in 2016 was **1604**



2016: No. of participants

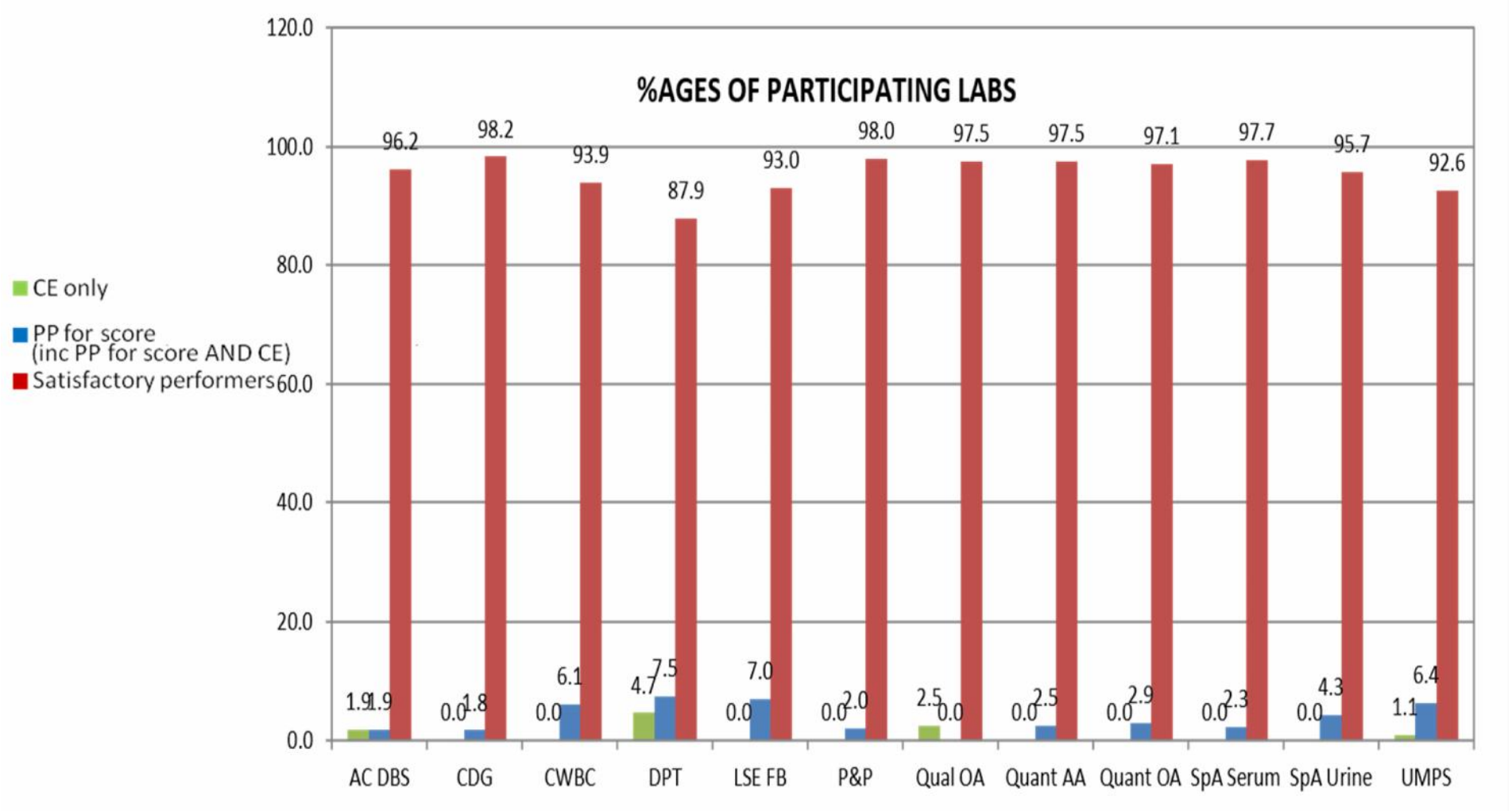
MetabERN HCPs subscribed to ERNDIM

(preliminary data based on 2017 invoices)



Scheme Results – 2015

Poor performers (PP)



Scheme Results – 2015

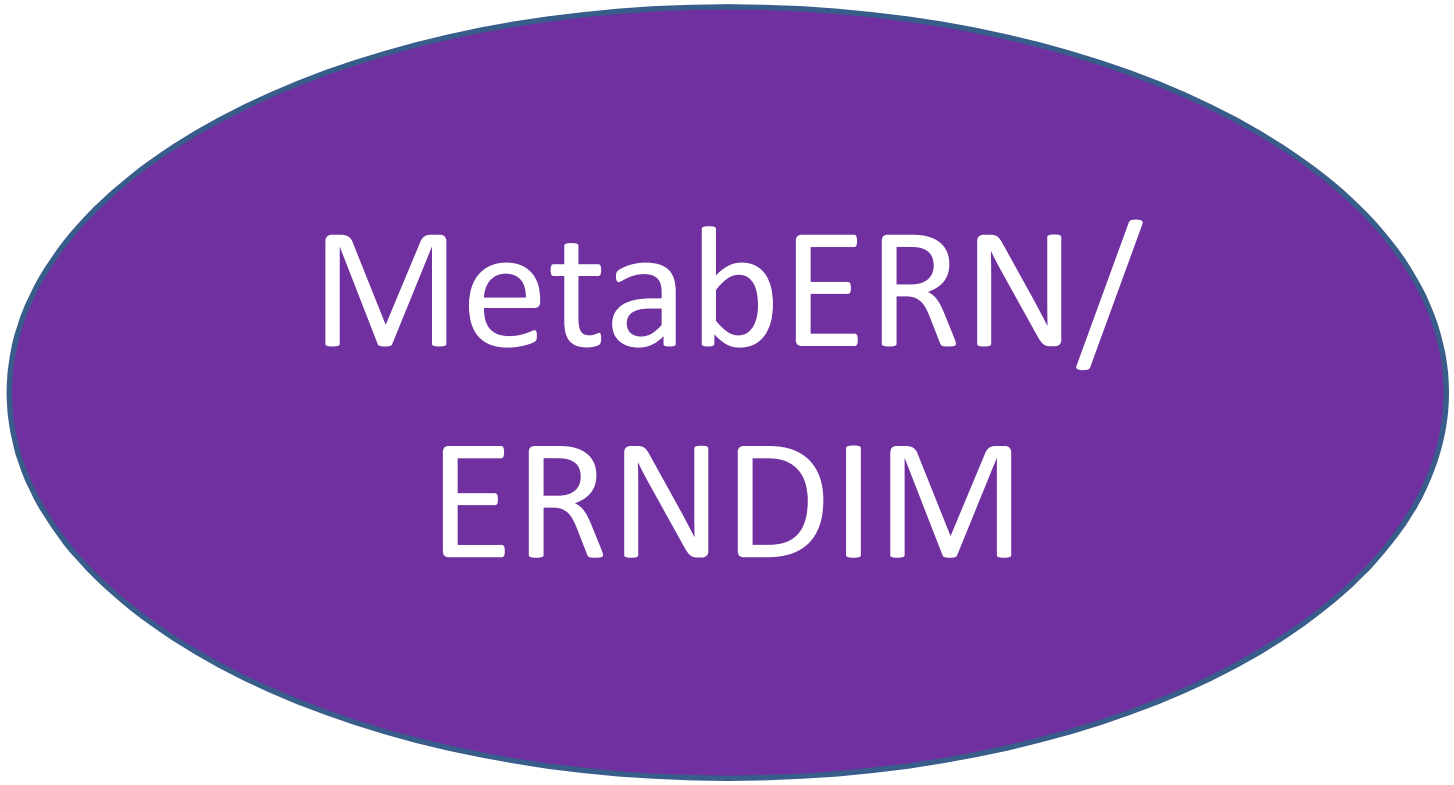
Global Poor Performance (PP)

ERNDIM
QUALITY ASSURANCE IN LABORATORY TESTING FOR IEM

- Global PP is poor performance in more than one EQA scheme in one year
- Details of labs with Global PP in 2015 are below

2015 Global PP	No of schemes with PP	AC in DBS	CDG	Cystine in WBC	DPT	LSE in Fibroblasts	Purines & Pyrimidines	Qual Organic Acids	Quant Amino Acids	Quant Organic Acids	Special Assays Serum	Special Assays Urine	Urine MPS
LAB 1	4	Y	N					CE	N	N	Y	Y	
LAB 2	3	CE			N			Y	N		Y	Y	
LAB 3	2							Y	Y		Y	N	N
LAB 4	2	CE			Y	N		Y	Y	Y	Y	Y	
LAB 5	2							D			N	N	
LAB 6	2			Y		Y	N	CE			Y	Y	
LAB 7	2							CE	Y				N

N = Poor Performance (inc PP for score AND CE); CE = Critical Error only; Y = Satisfactory Performance; D = Non-submitter; P = Partial Submitter


MetabERN/
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Potential areas for collaboration

- Awarenes of clinicians about laboratory performance and its assessment (including poor performance, PPP and global poor performance; in collaboration with QC ESHG, or e.g.uncertainty of measurement)
- Involvement of clinicians in designing new EQA schemes/review of existing schemes
- Mandating EQA participation for MetabERN members

Potential areas for collaboration

- Involvement of laboratory scientists in guidelines development/revision
- Provision of directory of tests for MetabERN members
- Involvement of laboratory scientists in research
- Patient organizations: sample donations