

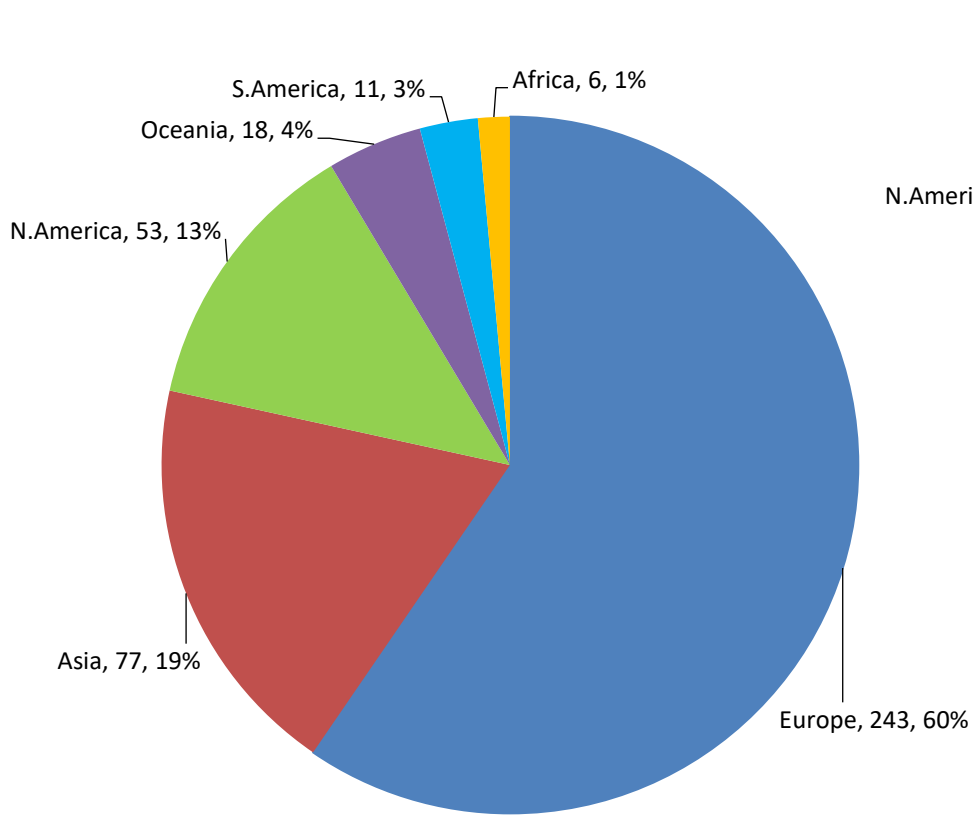


Chair's Update

Rafa Artuch

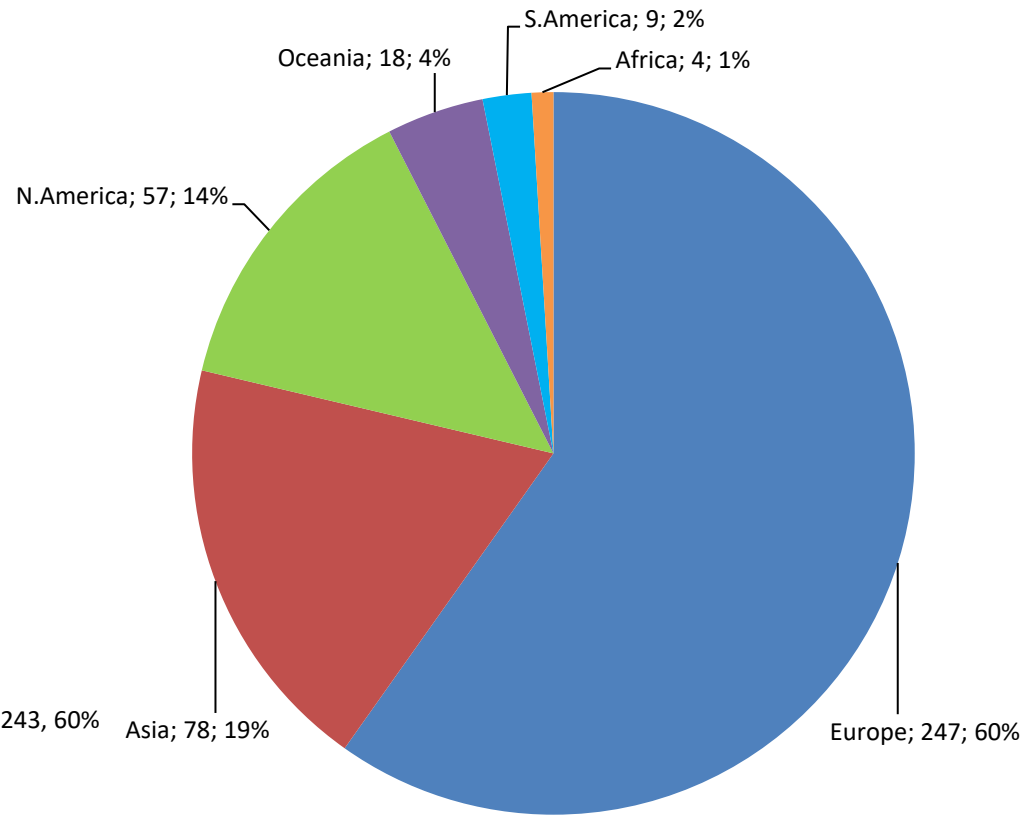
Hospital Sant Joan de Déu, Barcelona

Participants



2019: Number of participants = 408

- Number of participating countries = 63



**2024: Number of participants = 413
(1.2% ↑ on 2019)**

- Number of participating countries = 61

EQA Schemes

Quantitative schemes	Qualitative schemes
Amino acids (serum)	Amino Acids Interpretation
Acylcarnitines (serum)	Diagnostic Proficiency Testing (urine)
Organic acids (urine)	Organic acids (urine)
Purines-Pyrimidines (urine)	Acycarnitines (DBS)
Special assays serum	Mucopolysaccharides (urine)
Special assays urine	CDG (serum)
Special assays DBS	

Hybrid schemes	Interpretation scoring
Lysosomal enzymes (Fib)	Yes
Cystine (WBC)	Yes
Pterins (urine)	Yes
Neurotransmitters (CSF)	Yes

2019: 16 EQA schemes, 1953 scheme registrations

2024: 17 EQA schemes, 2188 scheme registrations (+12%)

New EQA Schemes

Pilot scheme

1. Lipids/sterols in serum

- Running as a PILOT scheme in 2024 and will continue as a pilot scheme in 2025
- SAS will continue as is in 2024 and 2025
- The pilot scheme will analyze the lipid scheme performance and participation
- Reasons for this new scheme
 - SAS scheme is currently crowded by many analytes
 - Lipids are becoming powerful biomarkers for different IEM
 - Pre-analytical and analytical issues are different from those of polar compounds included in SAS
 - Separating Lipids out of the SAS scheme would allow for other analytes to potentially be included in either the SAS or LIS schemes in the future

**Please let us know your ideas
for new EQA schemes**

Metabolite	Disorder	Group of disorders	Method	SAS/new
Lysosphingolipids				
Lyso-sphingomyelin (Lyso-SM)	Niemann Pick Disease type A/B (NPA/B)	Sphingolipidoses	LC-MS/MS	SAS
Lyso-globotriaosylceramide (Lyso-Gb3)	Fabry Disease	Sphingolipidoses	LC-MS/MS	SAS
Glucosylsphingosine	Gaucher Disease	Sphingolipidoses	LC-MS/MS	SAS
N-palmitoyl-O-phosphocholineserine (PPCS); (previously known as Lysosphingomyelin-509)	Niemann Pick Disease type C (NPC) and Niemann Pick Disease type A/B (NPA/B)	Sphingolipidoses	LC-MS/MS	New
Lyso-monosialoganglioside 1 (Lyso-GM1)	GM1 gangliosidosis	Sphingolipidoses	LC-MS/MS	New
Lyso-monosialoganglioside 2 (Lyso-GM2)	GM2 gangliosidosis (Tay Sachs and Sandhoff disease)	Sphingolipidoses	LC-MS/MS	New
Oxysterols				
Cholestane-3 β ,5 α ,6 β -triol	Niemann Pick Disease type C (NPC)	Sphingolipidoses	LC-MS/MS	SAS
7-ketocholesterol (7-KC)	Niemann Pick Disease type C (NPC)	Sphingolipidoses	LC-MS/MS	SAS
Lysophosphatidylcholines				
C26:0-lysophosphatidylcholine (C26:0-lysoPC)	X-linked adrenoleukodystrophy (X-ALD), D-bifunctional protein (DBP) deficiency, peroxisomal acyl-CoA type 1 (ACOX1) deficiency and Zellweger Spectrum Disorders (ZSD)	Peroxisomal disorders	LC-MS/MS	SAS (pilot)
Sterols				
Cholestanol	Cerebrotendinous Xanthomatosis (CTX)	Sterol biogenesis disorders	GC-MS/GC-FID	SAS
7-dehydrocholesterol (7-DHC)	Smith Lemli Opitz Syndrome (SLO)	Sterol biogenesis disorders	GC-MS/GC-FID	SAS
Desmosterol	Desmosterolosis	Sterol biogenesis disorders	GC-MS/GC-FID	New
Sitosterol	Sitosterolemia	Sterol transport disorders	GC-MS/GC-FID	New

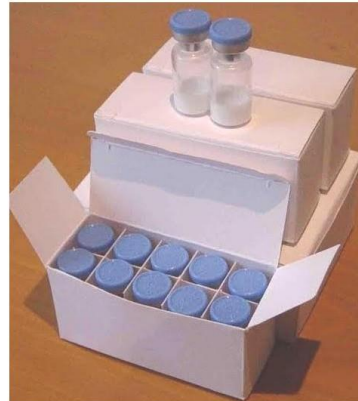
Considerations for new schemes

- Demonstrable need for new scheme
- Scientific Advisor + deputy
- Proposal → scheme design → business plan
 - Planning
 - Materials/samples
 - Metabolites (incl stability)
 - Statistical design
 - Assigned values
 - Costs
- Pilot scheme run successfully
- Approval by SAB, Executive committee & Board of Trustees
- ***UNTARGETED METABOLOMICS***

Other activities

Materials for internal QC

- Amino acids
- Organic acids
- Purines/pyrimidines
- Acylcarnitines
- SAS and SAU
- Homocysteine,
- Pterins
- Neurotransmitters



produced separate from EQA materials

Educational panels

- Oligosaccharide kit

All supplied by MCA laboratory www.erndim.org

Materials for internal QC

produced separate from EQA materials

- Cystine in WBC/protein matrix: under investigation
- Amino acids in urine
- Amino acids in CSF/CSF-like matrix
- Amino acids (and carnitine, etc) in dry blood spots

Educational panels

- Purine-pyrimidine disorders
- Bile acid disorders

ERNDIM - SSIEM Academy

SSIEM Academy 2024

- Was held on 22nd and 23rd April 2024 in Amsterdam, the Netherlands
- Topics:
Lysosomal storage disorders, peroxisomal disorders, purine and pyrimidine disorders

SSIEM Academy 2025

- Will be held on 28th and 29th April 2025 in Prague, Czech Republic
- Topics:
Mitochondrial diseases, Neurotransmitter diseases, Congenital disorders of Glycosylation and Glycogen Storage disorders

Event information will be made available on the SSIEM and ERNDIM events pages

New Meetings for 2024 and beyond

1) Online workshops for ACDB and QLOU to discuss scheme results & format similar to Diagnostic Proficiency Testing participant meetings → Done in 2024. **QLOU: 166 registrations**

2) Quantitative schemes February 2024: Virtual meetings

Amino acids (serum) /Acylcarnitines (serum)

Aims:

- Technical meeting for laboratory professionals
- Present and discuss critical laboratory issues
- Before the meeting, participants may send to us their questions of technical problems with the techniques
- Cycle of meetings over 3-4 years, focusing on 2 schemes/method areas per year
- **QTAS: 223 registrations**
- **ACS: 125 registrations**

Administration Office

Staff:

- Sara Gardner (Executive Administrator, full time)
 - Jenny Barrett (Scientific Administrator, full time, *maternity leave from late October 2024*)
 - Simon Rothwell (Scientific Administrator, full time)
 - Kate Straznikiewicz (Admin Assistant, 0.6 x full time)
 - Ismenia da Silva (Admin Assistant, 0.6 x full time)
 - Maternity cover Scientific Administrator post to be recruited (full time)
- Responsible for administration **EQA schemes**
 - Central to the day to day running of ERNDiM
 - Preparation of **ISO accreditation**
 - **Thank you to Administration office !!!!**

Path to accreditation

Progress so far:

- **ISO/IEC 17043** specifies the standards to be met for all aspects of a proficiency testing scheme and the management of the proficiency testing provider
- **Document control system**
- **Management system**
- **Formal Internal audit calendar**
- **Formal agreements with sub-contractors**
- All qualitative schemes: **centralised sample dispatch and online results submission**
- **Policies and procedures in progress**
- **Aiming to submit initial application 2024**

Laboratory directory

www.IEMBASE.ORG

- The aim of the laboratory directory is to provide a platform for physicians and labs to search laboratories that perform a certain test in the IEM field.
- We discussed with Nenad Blau to include a laboratory directory module in IEMBase. This could be a fruitful collaboration. To be able to build a laboratory directory, IEMBase must include a good list of metabolites (metabolite groups) and tests.
- You will be contacted early for further details.



Thank You!