



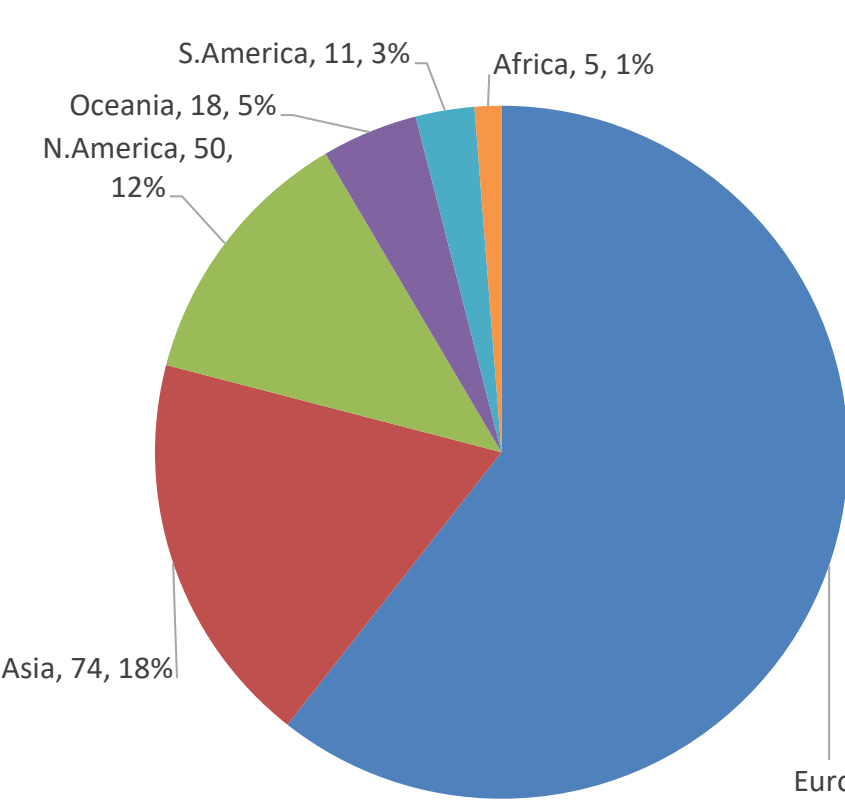
Chair's Update

**Christine Vianey Saban
(on behalf of Rafa Artuch)**

August 29th, 2023

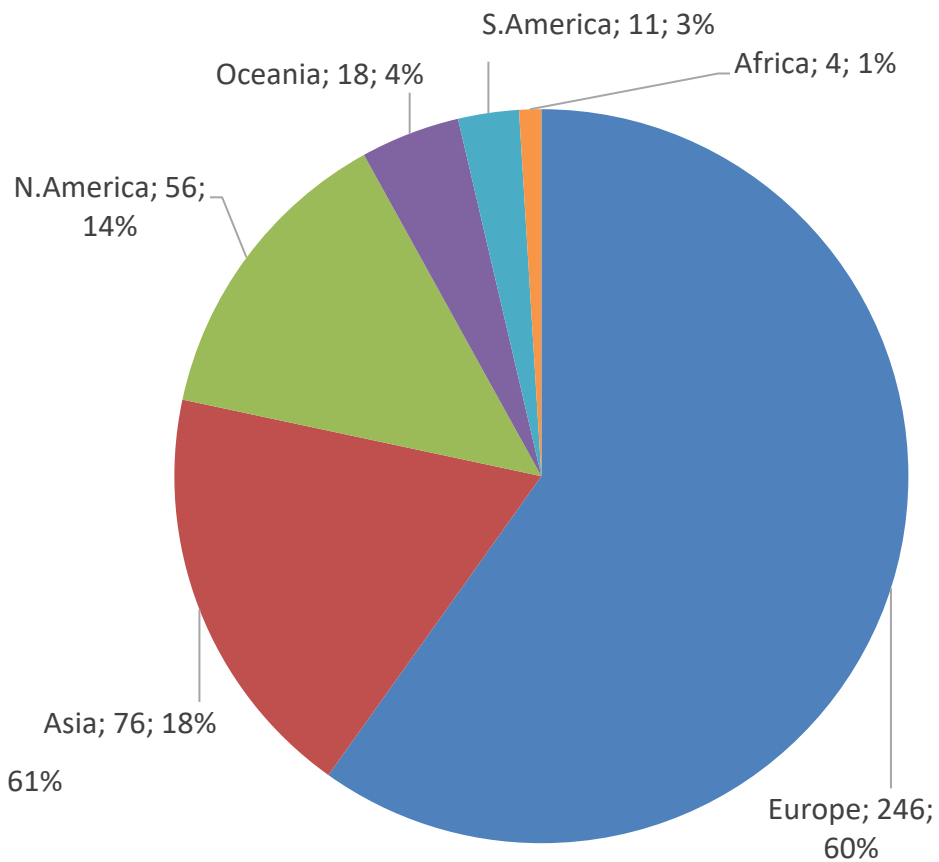
*This presentation will be uploaded to the ERNDiM
website after the event is over*

Participants



2018: Number of participants = 401

- Number of participating countries = 60



**2023: Number of participants = 411
(2.5% ↑ on 2017)**

- Number of participating countries = 60

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

2018: 15 EQA schemes, 1788 scheme registrations

2023: 17 EQA schemes, 2165 scheme registrations (+21%)

Scheme sample delivery issues:

- **Spain:** Import regulations were changed for non-EU imports into Spain between the 2021 and 2022 sample dispatches. Additional documents are now required for some ERNDiM EQA scheme packages which caused some delays. We hope the majority of these issues are now resolved. Co-operation and determination from participants in resolving this is very much appreciated.
- **Italy:** Participants in Italy experienced difficulties in 2023 with import inspections for packages entering the EU, requiring additional certificates for release of samples. ERNDiM are looking to implement a solution for this before the 2024 dispatch with the assistance of Cristiano Rizzo, Scientific Advisor for ACDB Rome and CSCQ, our scheme organiser in Switzerland.
- **Turkey:** A number of participants in Turkey experienced delays to sample delivery in 2023 due to disruption to infrastructure following the catastrophic earthquake in early 2023. Support to receive samples and submit results was provided. Our thoughts are with those affected by the earthquake, we hope that 2024 will be less difficult for those affected.

New EQA Schemes

New pilot scheme

1. Lipids/sterols in serum

- **Aiming for this as a PILOT scheme in 2024**
- **SAS will continue as is in 2024**
- **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 gangliosidoses (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

ERNDIM - SSIEM Academy

SSIEM Academy 2023

- Was held on 24th and 25th April 2023 in Manchester, UK
- Topics:
Organic acidemias, fatty acid oxidation defects and cardiomyopathy
Event information will be on the ERNDIM and SSIEM events pages

SSIEM Academy 2024

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

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

New Meetings for 2024 and beyond

1) We are planning online workshops for ACDB and QLOU to discuss scheme results & format similar to Diagnostic Proficiency Testing participant meetings. Planned to start in 2024.

2) Quantitative schemes early 2024: Virtual meeting

Proposed topics:

Amino acids (serum) → LC-MS/MS (scientific advisor)

Acylcarnitines (serum) → LC-MS/MS (scientific advisor)

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

Quantitative schemes early 2024: Virtual meeting

- **Proposed meeting format** (to present preanalytical, analytical, postanalytical aspects of the procedures). Critical steps, troubleshooting, difficult metabolites...
- General presentation of the meeting and schemes: 15 minutes: Rafa Artuch/George Ruijter
- Amino acids 30 minutes
- Open discussion: 30 minutes
- Acylcarnitines 30 minutes
- Open discussion: 30 minutes
- Concluding remarks (15 minutes)

Other activities

Materials for internal QC

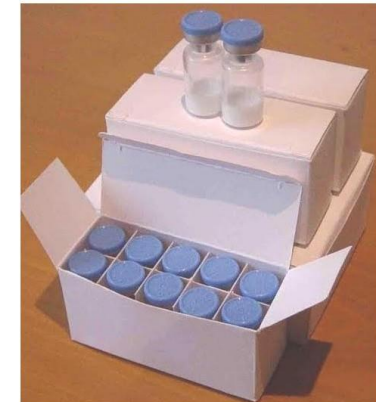
- Amino acids
- Organic acids
- Purines/pyrimidines
- Acylcarnitines
- SAS and SAU
- Homocysteine,
- Pterins
- Neurotransmitters

produced separate from EQA materials

Educational panels

- Oligosaccharide kit

All supplied by MCA laboratory www.erndim.org



Other control materials?

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

Administration Office

Staff:

- Sara Gardner (Executive Administrator, full time)
 - Jenny Barrett (Scientific Administrator, full time)
 - Kate Straznikiewicz (Admin Assistant, 0.5 x full time)
 - Ismenia da Silva (Admin Assistant, 0.5 x full time)
 - Simon Rothwell (Scientific Administrator, full time) Joined June 2023
- Responsible for administration **EQA schemes & SSIEM Academy**
 - 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**

ERNNDiM The logo for ERNDiM features the text "ERNNDiM" in a dark blue, sans-serif font. The letter "i" is lowercase and has a red dot above it. To the right of the text is a teal-colored line graph with three peaks of varying heights, resembling a heartbeat or a signal waveform. A thin teal horizontal line extends from the left side of the text to the start of the graph.

Thank You!