



SSIEM Annual Symposium
30 Aug. – 2 Sept. 2022 | Freiburg · Germany



QUALITY ASSURANCE IN LABORATORY TESTING FOR IEM



Chair's Update

George Ruijter

Rafa Artuch

September 30th, 2022. Freiburg

Report from Executive Committee

(minutes from EC meeting in Barcelona)

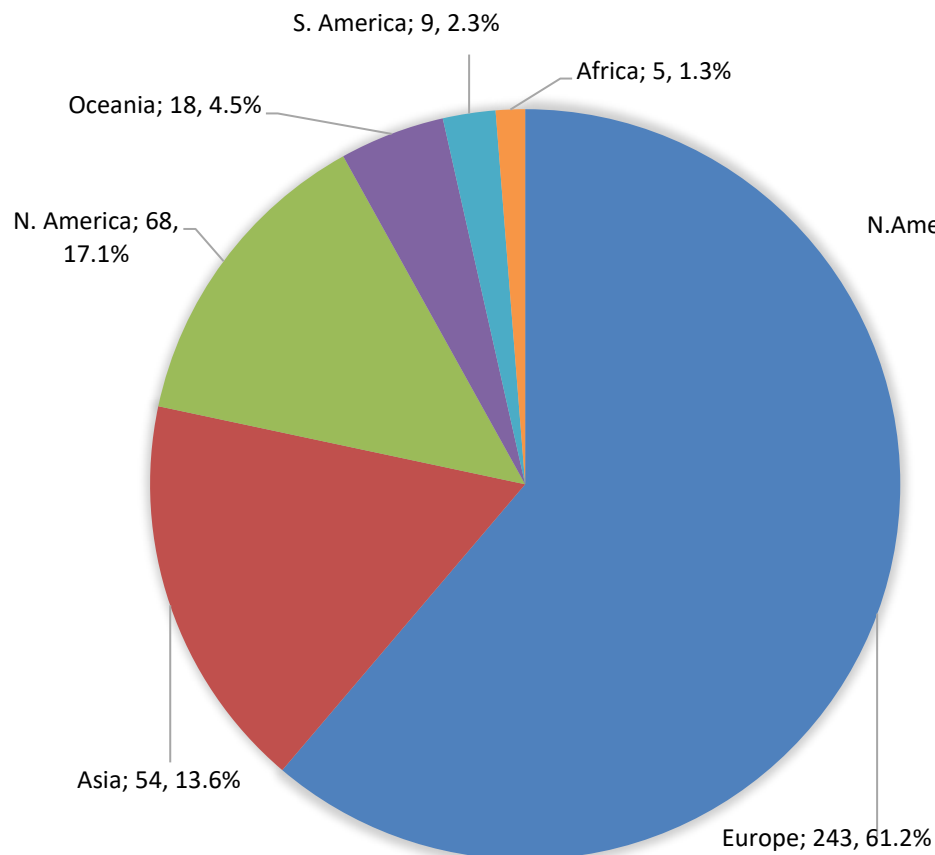
Meetings:

- EXEC (Barcelona, March 2022).
- SAB (Barcelona, March 2022).

New Executive Committee members:

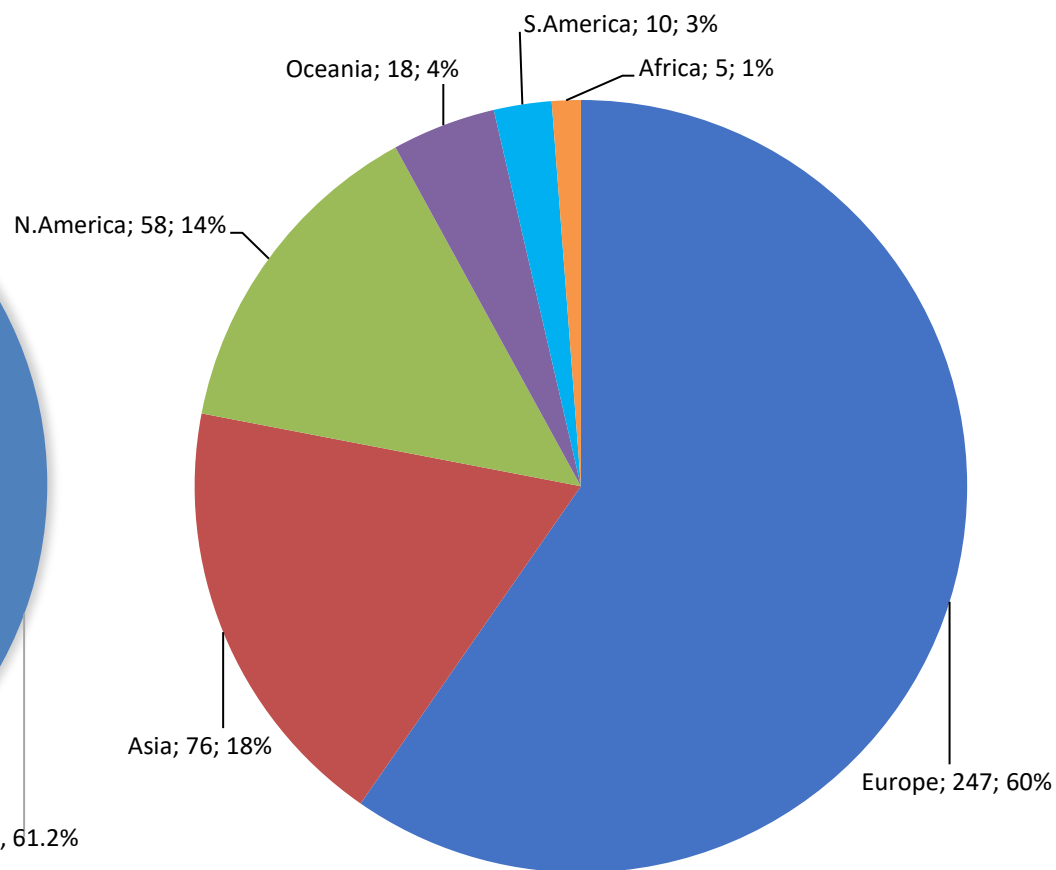
- Rafa Artuch → EC Chair.
- George Ruijter → SAB Chair
- Judit García-Villoria → Secretary
- All started on 1st January 2022.
- Daniel Herrera → Treasurer

Participants



2017: Number of participants = 397

- Number of participating countries = 60



**2022: Number of participants = 414
(4.3% ↑ on 2017)**

- Number of participating countries = 62

Participants

- EQA scheme total registrations → 2000.
- Registered countries = 62.
- Europe = 59%,
Asia = 18%
North America = 14%
Oceania = 5%
South America = 2%
Africa = 2%.
- Potential for growth in a number of geographical areas. Position paper about ERNDiM in English and Spanish.

EQA Schemes

Quantitative schemes	Qualitative schemes
Amino acids (serum)	Diagnostic Proficiency Testing (urine)
Acylcarnitines (serum)	
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



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Sample donations. Ethical issues: consent should be shifted from ERNDiM to the donating clinician (local informed consent)
Biobank activities (HSJD and CDG scheme)

EQA calendar



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[About](#) [News & Events](#) [EQA Schemes](#) [Meetings & Reports](#) [Training](#) [Newsletters](#) [Laboratory Directory](#) [Links](#)

QUALITY ASSURANCE IN LABORATORY TESTING FOR IEM

Diagnosis
and Treatment

EQA Schemes

External Quality Assurance Schemes

We will be expanding and updating the information available on our External Quality Assurance Schemes soon. In the meantime if you have any questions or would like to subscribe to one of the ERNDiM EQA schemes please [contact us](#)

Subscriptions

SUBSCRIPTIONS FOR THE ERNDiM 2021 EQA SCHEMES CLOSED ON 4TH NOVEMBER 2020.

If you are an existing ERNDiM EQA participant you will have been sent details of how to register for the 2021 EQA schemes in September 2020. If you missed the registration period but would still like to register for the 2021 EQA scheme please [contact us](#).

If you have any problems accessing the ERNDiM Registration Website please [contact us](#).

If you don't already subscribe to an ERNDiM EQA Scheme and would like to participate during 2021, subscription to a scheme may still be possible but availability will be limited.

PLEASE NOTE THE FOLLOWING 2021 EQA SCHEMES ARE NOW FULL: PTERINS IN URINE, SPECIAL ASSAYS IN DBS, SPECIAL ASSAYS IN SERUM AND SPECIAL ASSAYS IN URINE.

The ERNDiM EQA Scheme Catalogue gives information on all of the ERNDiM EQA schemes and also the prices of each scheme in Euro, GB pounds and US dollars. The 2021 catalogue can be downloaded from this page - see link on right.

Brief information on the EQA schemes offered by ERNDiM in 2021 is also below.

Quantitative Schemes

- Acylcarnitines in serum
- Cystine in white blood cells
- Lysosomal enzymes in fibroblasts
- Neurotransmitters in CSF

Participants

- ▶ [PARTICIPANT AREA](#)
Register for EQA schemes and view account details
- ▶ [QUANTITATIVE RESULTS WEBSITE](#)
Submit results for Quantitative schemes
- ▶ [QUALITATIVE RESULTS WEBSITE](#)
Submit results for DPT and Urine MPS schemes

ERNDiM EQA Scheme

- ▶ [ERNDiM EQA Scheme](#)
- ▶ [ERNDiM EQA scheme catalogue and participant guide](#)
- ▶ [EQA Scheme Annual Reports](#)
- ▶ [Support Grant Application Form](#)
- ▶ [ERNDiM EQA scheme submission calendar](#)
- ▶ [Sample Donations](#)
PDF Document download

Partnerships



Poor performance (PP)

- The percentage of poor performers in the 2021 schemes varied from 0.0% (PTU) to 13.9% (NCSF).
- Overall, there were 26 incidences of PP due to critical error only (1.5% of all participations), 70 incidences of PP for score only (4.0% of all participations) and 12 incidents of PP for score and critical error (0.7% of all participations). The total number of PP incidences was 108 (6.2% of all participations).

Path to accreditation



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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 2023**

A screenshot of a software application window displaying a list of items. The window has a standard Windows-style title bar and menu bar. The main area contains a table with several columns. The visible columns include 'Name', 'Description', 'Status', and 'Date'. The table contains numerous rows of data, with some rows highlighted in blue. The interface appears to be a database or inventory management system.

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) joined March 2020
- Responsible for administration **EQA schemes & SSIEM Academy**
- Central to the day to day running of ERNDiM
- Preparation of **ISO accreditation**
- SSIEM agreed to support the Scientific Administrator position until 2022 and software development costs **Thank you SSIEM!**
- **And Thank you to Administration office !!!!**



Meetings

- Meeting Oct 2021. Organising committee: Cristiano Rizzo, Carlo Dionisi-Vici, CVS, Nenad Blau, GR, with JBa representing the AO. Online meeting with pre-recorded presentations (Total of 472 registrations)

Continent	Symposium attendees	2021 EQA participants (number of labs)
Europe	273 (65%)	239 (59%)
Asia	71 (17%)	73 (18%)
North America	26 (6%)	57 (14%)
Oceania	20 (5%)	19 (5%)
South America	17 (4%)	10 (2%)
Africa	16 (4%)	6 (2%)

Next ERNDIM meeting → Rome, November 2022.

ERNDIM - SSIEM Academy



SSIEM Academy 2022

- June 2022, Amsterdam, The Netherlands
- First in person Academy since April 2019
- Topics were:
 - Aminoacidopathies, Hyperammonaemia and Urea Cycle Defects

SSIEM Academy 2023

- Will be 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

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

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New EQA Schemes

Pilot schemes running in 2022

1. Amino Acids Interpretation

lead organiser: Dr Sabine Scholl-Bürgi (Innsbruck, Austria)



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Ideas for new pilot schemes

1. Lipids/sterols in serum: 4th special assays scheme

Currently in SAS: C22, C24, C26, phytanic acid, pristanic acid, 7-Dehydrocholesterol, 7-Ketocholesterol, Cholestane-3b,5a,6b-triol, Cholesterol, Glucosylsphingosine, Lyso Gb3, Lysosphingomyelin, coenzyme Q10..

2. Metabolomics in plasma/serum Qualitative scheme?

3. Lysosomal enzymes in DBS Depends on sample availability

4. Qualitative bile acids?

5. Individual metabolites

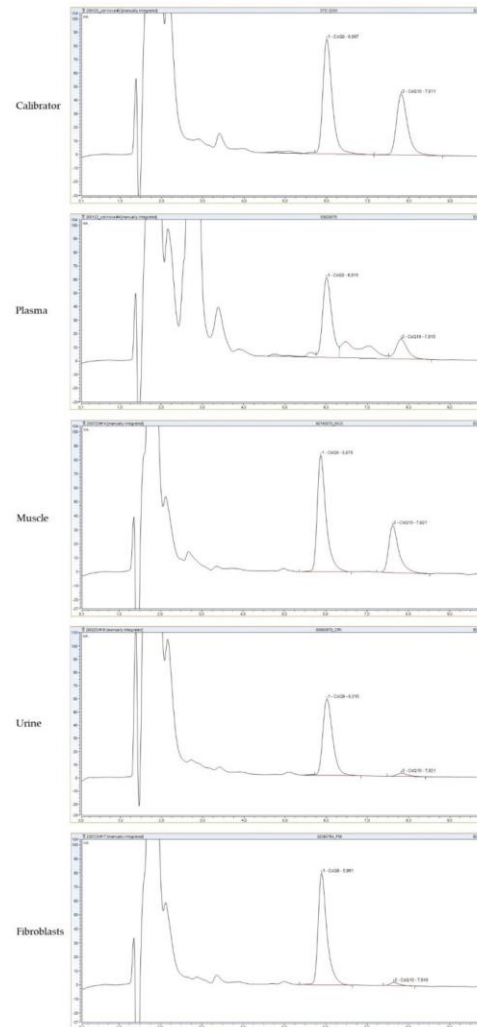


Please let us know your ideas
for new EQA schemes

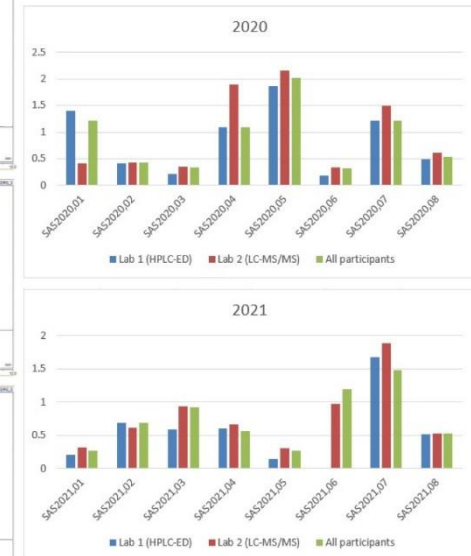
Report from Executive Committee

PLANS

- 2023 schemes – depends on workload AO. Some minor changes in individual metabolites
- New metabolites → prior to include, consider stability, price, general interest... Encouraging laboratories to do first an agreement study among 4-5 Labs, and after, plan to include in ERNDIM.
- Thanks for answering the annual survey.



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Coenzyme Q₁₀

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

Considerations for new schemes

- Demonstrable need for new scheme
- Scientific Advisor + deputy
- Proposal → scheme design → business case
 - Planning
 - Materials/samples
 - Metabolites (incl stability)
 - Statistical design
 - Assigned values
 - Costs
- Pilot scheme run successfully for 2 years
- Approval by SAB, Executive committee & Board of Trustees

ERNNDiM 

Thank You!