

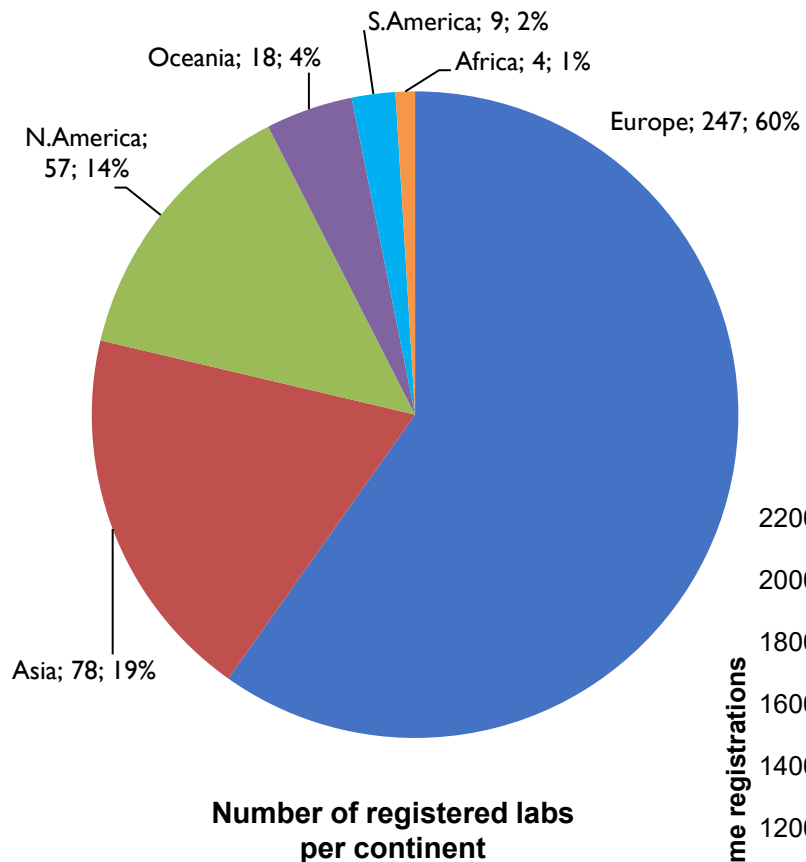


Chair's update

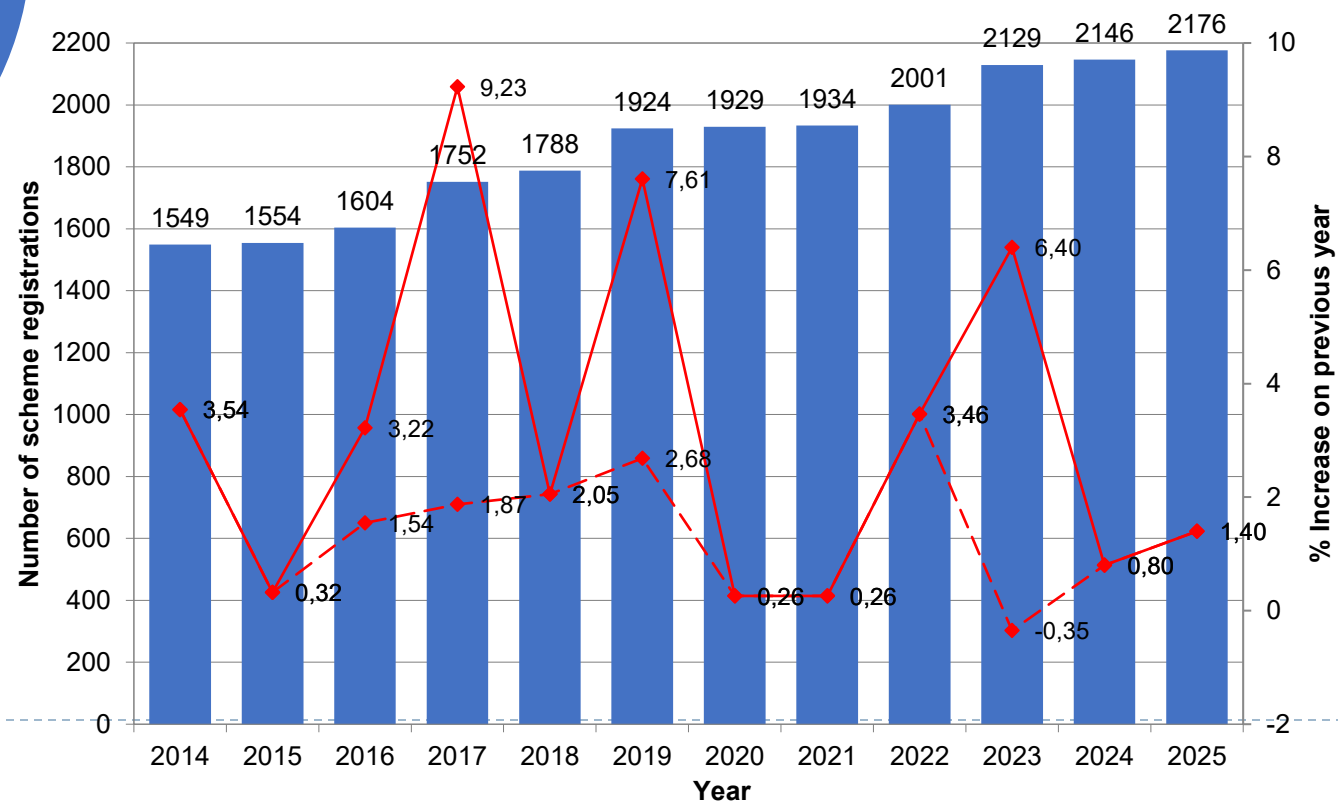
George Ruijter (Chair SAB) on behalf of Rafa Artuch (Chair EC)

Helsinki, August 25th, 2026

2025 Scheme registrations (from AMR)



	2024	2025	% difference
No. of scheme registrations	2146	2176	+1.4%
No. of labs	413	417	+1.0%
No. of countries	61	61	0.0%



EQA Schemes 2026

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

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

Lipids in Serum scheme new in 2026

- SA's: Susan Goorden (Rotterdam) and Marie van Dijk (Amsterdam)
- 2026: 69 labs registered, scheme oversubscribed
- 16 different lipids
- 6 lipids transferred from SAS to LIS
- 7DHC, cholestanol and VLCFA kept in SAS

Analyte	Your Lab	Med All Labs	n
7-dehydrocholesterol (7-DHC) (μmol/L)		54.2	32
7-ketocholesterol (7-KC) (μmol/L)		0.960	9
C26:0-lysophosphatidylcholine (C26:0-lysoPC) (μmol/L)		1.47	14
Cholestane-3β, 5α, 6β-triol (μmol/L)		0.0140	11
Cholestanol (μmol/L)		67.7	30
Cholesterol (mmol/L)		4.32	13
Coenzyme Q10 (μmol/L)		1.57	15
Desmosterol (μmol/L)		82.6	23
Glucosylsphingosine (nmol/L)		1.06	27
Lathosterol (μmol/L)		34.5	23
Lyso-globotriaosylceramide (Lyso-Gb3) (nmol/L)		0.410	31
Lyso-monosialoganglioside 1 (Lyso-GM1) (nmol/L)		96.2	9
Lyso-monosialoganglioside 2 (Lyso-GM2) (nmol/L)		14.4	9
Lyso-sphingomyelin (Lyso-SM) (nmol/L)		125	24
N-palmitoyl-O-phosphocholineserine (PPCS) (nmol/L)		899	19
Sitosterol (μmol/L)		19.8	26

Considerations for new schemes

- Demonstrable need for new scheme
- Scientific Advisor + deputy
- Proposal → scheme design → business plan
 - Materials/samples, metabolites (incl stability)
 - Statistical design, assigned values
 - Planning, costs
- Pilot scheme successfully organised 2 y
- Approval by SAB, Executive committee & Board of Trustees
- *Untargeted metabolomics (2027?)*
- *Lysosomal enzymes in Fibroblasts 2 (2028)*
- Please let us know your ideas for new EQA schemes

Websites for Quan and Qual schemes

Quantitative schemes website (erndimqa.nl)

- Small improvements based on participant suggestions
- Excel export for cycle review data
- Directional z-scores
- Standard deviations added to cycle review

Qualitative schemes website (cscq.hcuge.ch/cscq/ERNDIM/)

- New result submission website in development
- Hopefully operational in 2027
- Report availability improved

Administration Office

Staff:

- Sara Gardner (Executive Administrator)
 - Jenny Barrett (Scientific Administrator)
 - Simon Rothwell (Scientific Administrator)
 - Amelia Gamble (Scientific Administrator)
 - Ismenia da Silva (Admin Assistant)
- Responsible for administration of EQA schemes
 - Responsible for Quality Management
 - Assist Scientific Advisors
 - Central to the day-to-day running of ERNDiM
 - **Big thank you to Administration office !!!!**



Accreditation

ISO 17043 : 2023

- Specifies general requirements for the competence and impartiality of proficiency testing (PT) providers
- Accreditation sub committee set up in 2025
- Application ready; discussion with UKAS (UK) and RvA (NL) – hopefully apply autumn 2026



2025 Participant Survey

- Survey link sent to 828 contacts from **415 centres**
- Responses received from **133 centres (32%)**
- 95.3% of respondents rated the quality of services provided by ERNDiM as 'excellent' or 'good'
- Best rated: frequency of samples and annual reports
- Worst rated: sample volume CDG scheme

Online workshops 2024-2027

- Technical meetings for laboratory professionals
- Present and discuss critical laboratory issues
- Participants are invited to submit their questions in advance
- Cycle of 4 years, focusing on 2 schemes/method areas per year

2024

- QTAS (Rachel Carling)
- ACS (Pedro Ruiz-Sala)

2025

- QTOU (Clothilde Roux, Laura Steinbusch & Irene Körver-Keularts)
- PPU (Jörgen Bierau & Dr. Laura Steinbusch)

2026

- SAS and SAU (Rafa Artuch)

2027

- SADB and LIS (end of 1st cycle)

Laboratory Directory

Collaboration with IEMbase

IEMBASE.ORG



- Database of laboratories active in IMD diagnostics
- Planning to fill and maintain database from ERNDiM (and CAP?) registrations using opt-in approach
- Ultimate aim to link metabolites to labs

Other activities

Quality comparison materials (iQC)

- Batches of AA, OA, PuPy, AC, Special Assays, pterins, homocysteine, cystine in WBC (pilot sets)
- Produced separate from EQA materials
- Comprehensive data management platform
- Considering AA in urine, CSF

Educational panels

- Oligosaccharide kit

Archived scheme materials

- Residual EQA sets
- Provided for validation purposes



All supplied by **MCA laboratory** (www.erndimqa.nl)



Thank You!



Quality Assurance in Laboratory Testing for IEM

2026 Participant Workshop

Tuesday 25th August 2026

Programme	
11:00 – 11:10	Chair's update <i>George Ruijter, Netherlands (on behalf of Rafa Artuch)</i>
11:10 – 11:20	Common DPT sample <i>George Ruijter, Netherlands</i>
11:20 – 11:40	Peroxisomal Biomarkers in Blood and Urine <i>Fred Vaz, Netherlands</i>
11:40 – 12:00	Functional Testing for Peroxisomal Disorders <i>Sacha Ferdinandusse, Netherlands</i>
12:00 – 12:30	Clinical Presentation of Peroxisomal Disorders <i>Angeles García Cazorla, Spain</i>
12:30	Closing Remarks <i>George Ruijter, Netherlands (on behalf of Rafa Artuch)</i>