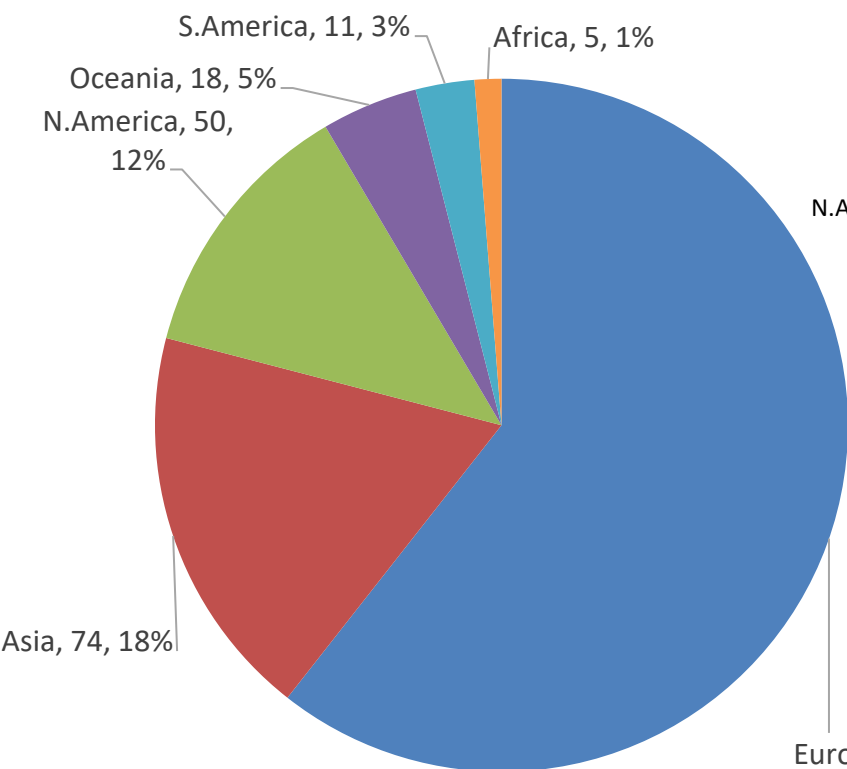




Report of the Executive Committee

Rafa Artuch
Chair EC

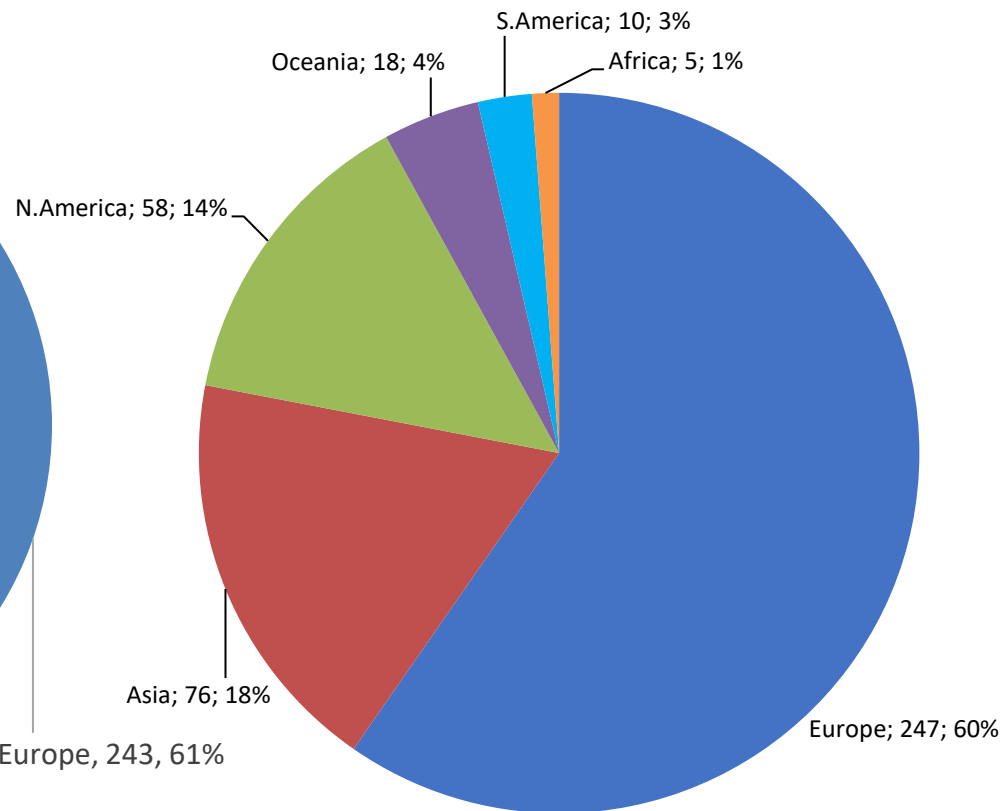
Participants



■ Europe ■ Asia ■ N.America ■ Oceania ■ S.America ■ Africa

2018: Number of participants = 401

- Number of participating countries = 60



2024: Number of participants = 417

Number of participating countries = 60

EQA Schemes

Quantitative schemes

Amino acids (serum)
Acylcarnitines (serum)
Organic acids (urine)
Purines-Pyrimidines (urine)
Special assays serum
Special assays urine (Cystine , Amino acids)
Special assays DBS

Qualitative schemes

Amino Acids Interpretation
Diagnostic Proficiency Testing (urine)
Organic acids (urine)
Acycarnitines (DBS)
Mucopolysaccharides (urine)
CDG (serum)

Hybrid schemes

Interpretation scoring

Lysosomal enzymes (Fib)	Yes
Cystine (WBC)	Yes
Pterins (urine)	Yes
Neurotransmitters (CSF)	Yes

LIS/sterols (Pilot) → New scheme in 2025

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**

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

Update on Laboratory Directory

Laboratory directory

www.IEMBASE.ORG

We have discussed with Nenad Blau to include a laboratory directory module in IEMBase, and also to review and curate the metabolite data base (HMDB, LOINC codes,). This will be a fruitful collaboration.

One young scientist as volunteer (Dieuwertje Augustijn, Rotterdam).

We have to prepare a project to be presented to IEMBASE for funding.



Disorder Information

Name

Glycerol kinase deficiency, isolated

Alternative Names

Hyperglycerolemia

Disease Abbreviation

GKD

Treatable?

yes

Prevalence

Mode of Inheritance

X-linked

Subtype

benign form

ICIMD Nosology Number

3.2.01.01

IEM Nosology Code

IEM0652

ICIMD Nosology Group

Disorders of gluconeogenesis

OMIM

307030

OrphaCode

Clinical Symptoms

Highlighted rows indicate symptoms that are characteristic of this disorder.

Symptom	Neonatal (birth -1mth)	Infancy (1-18mths)	Childhood (1.5-11yrs)	Adolescence (11-16yrs)	Adulthood (>16yrs)
Diabetes mellitus, insulin-resistant					±
Hypoglycemia	±	±	±	±	±
No clinical significance	+	+	+	+	+

Biochemical Markers

Highlighted rows indicate symptoms that are characteristic of this disorder.

Test panel	Symptom	HMDB	Neonatal (birth -1mth)	Infancy (1-18mths)	Childhood (1.5-11yrs)	Adolescence (11-16yrs)	Adulthood (>16yrs)
Routine tests	Triglyceride, pseudo (P)		↑	↑	↑	↑	↑
	Glucose (P)	HMDB0000122	↓-n	↓-n	↓-n	↓-n	↓-n
Specific tests	Glycerol (P)	HMDB00131	↑	↑	↑	↑	↑
	Glycerol (U)	HMDB00131	↑	↑	↑	↑	↑

Update from MetabERN (Yngve Blikrud)

- Yngve is the person who has replaced the position of Viktor.
- Joint educational forces during our last meeting in ERNDiM's board of trustees.





Plans for 2025/2026 Workshop

No meeting (Congress of the ICIEM in Japan)

ERNDIM 2025 Workshop 9-10 October 2025, Madrid, Spain.

<https://www.erndim.org/news-events/erndim-2025-workshop-registrations-open/>

www.geyseco.es/ERNDIM2025

2026 (August) → SSIEM meeting in Helsinki

Meetings for 2024/2027

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

2) Quantitative schemes February 2024: Virtual meetings

- QTAS (Rachel Carling) → **223 registrations**
- ACS 8Pedro Ruiz-Sala) → **125 registrations**

3) 2025

- QTOU 27th May (Clothilde Roux, Laura Steinbusch & Irene Körver-Keularts) – **95 attendees**
- PPU 6th June (Jörgen Bierau & Dr. Laura Steinbusch) – **66 attendees**

4) 2026

- SAS and SAU

5) 2027

- SADB and LIS (end of 1st cycle)

- Technical meetings 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

SSIEM meetings

SSIEM academy → Prague, 28th-29th April 2025 (MD, NT and CDG)

SSIEM anual symposium → Helsinki, August 2026

Administration Office

Staff:

- Sara Gardner (Executive Administrator)
 - Jenny Barrett (Scientific Administrator)
 - Kate Straznikiewicz (Admin Assistant)
 - Ismenia da Silva (Admin Assistant)
 - Simon Rothwell (Scientific Administrator)
 - Amelia Gamble (Last incorporation)
- 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 !!!!**



ERNDiM

Thank You!