



QUALITY ASSURANCE IN LABORATORY TESTING FOR IEM

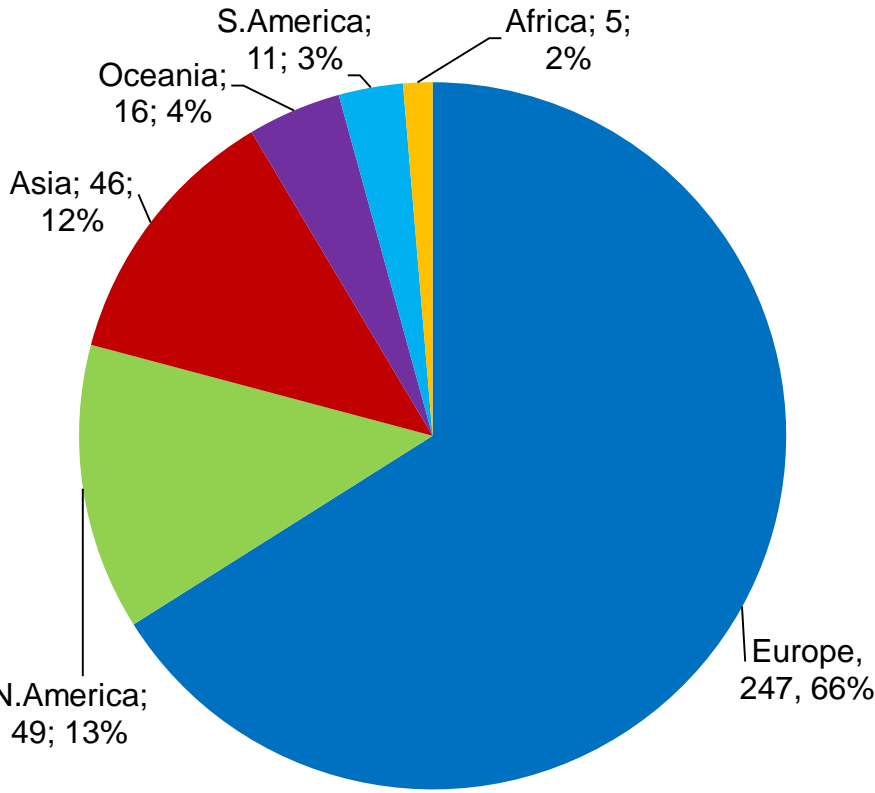
Exec Chair's Update

George Ruijter

Rotterdam

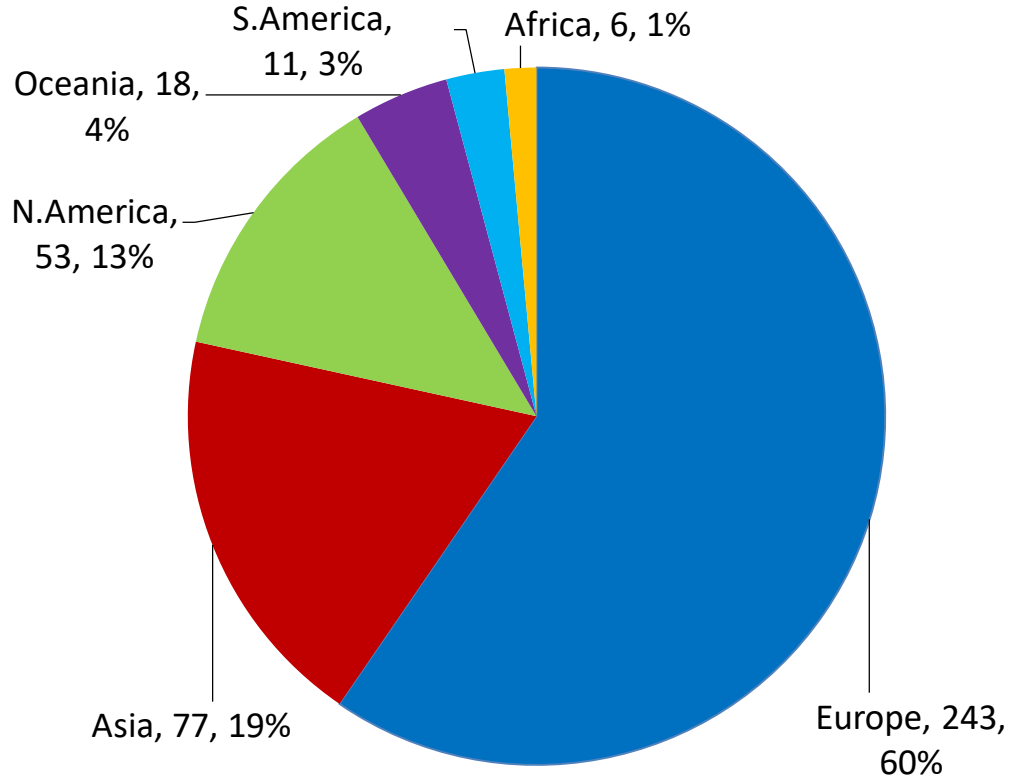
September 3rd, 2019

ERNDIM: Participants



2014: Number of participants = 374

- Number of participating countries = 59

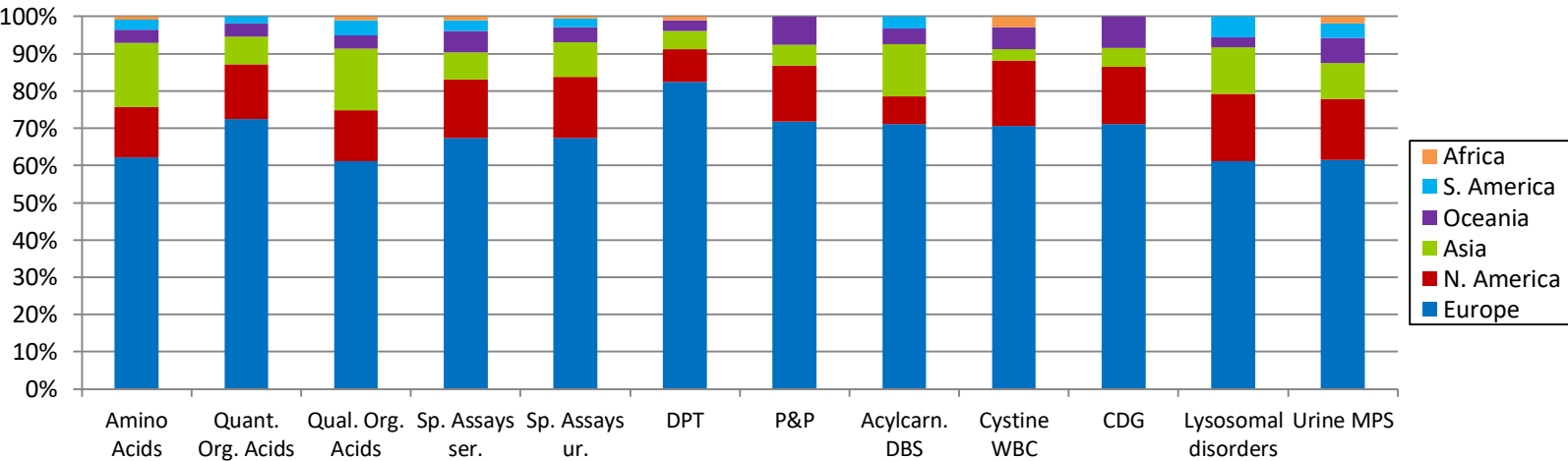


2019: Number of participants = 408
(13.6% ↑ on 2013)

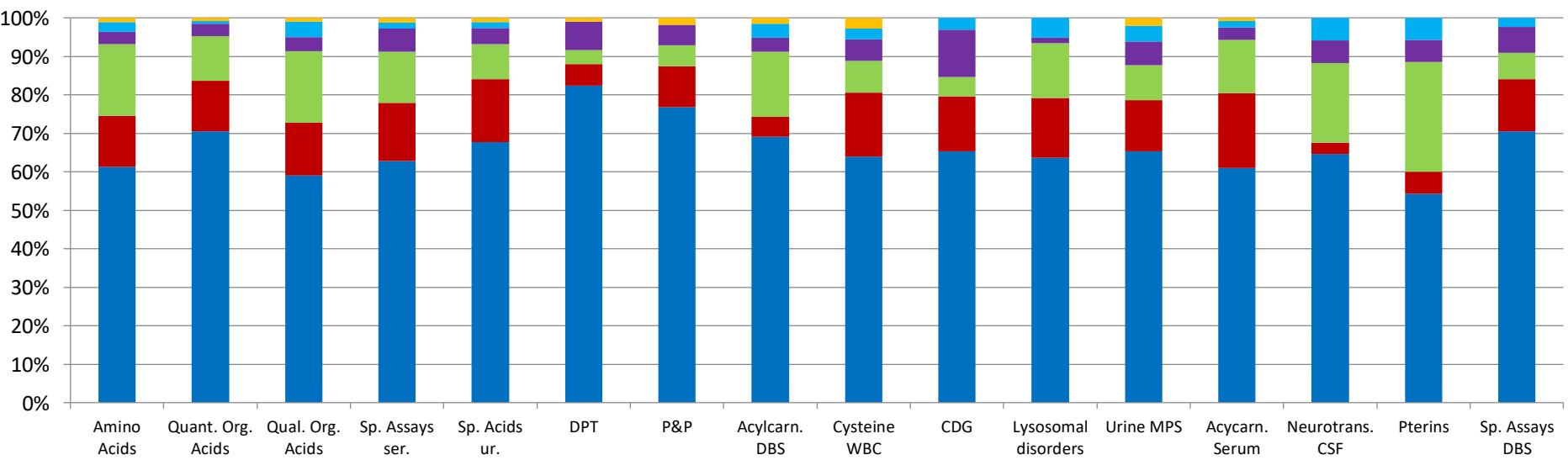
- Number of participating countries = 63

ERNDIM: EQA Schemes

2014: 12 QA schemes, 1484 scheme registrations



2019: 16 QA schemes, 1953 scheme registrations, (32% ↑ on 2014)



ERNDIM: EQA Schemes



Pilot schemes running in 2019

1. **Cognitive Amino Acids**, lead organiser is Prof Brian Fowler (Basel/Zurich, Switzerland)

Please let us know your ideas for new EQA schemes

Workshops

- To discuss results & format
- Lysosomal enzymes in Fib: September 3, 2019
- Other scheme(s) in 2020 etc

ERNDIM: EQA Schemes



Hybrid schemes vs. Quantitative/Interpretative schemes

Lysosomal enzymes in Fib

- Quantitation (enzyme activity) + interpretation (diagnosis) scored

Neurotransmitters in CSF & Pterins in Urine

- Interpretation introduced in 2018
- Pilot scoring interpretation of 2019 schemes

Cystine in WBC

- Interpretation introduced and pilot scoring 2020

ERNDIM: Changes to EQA calendar



ACCOMPLISHED

2018 schemes

- Earlier sample dispatch and submission dates
- Most 2018 annual reports finished February 2019
- Scheme year 2018 finished (issue of certificates) in **April 2019**

PLANS

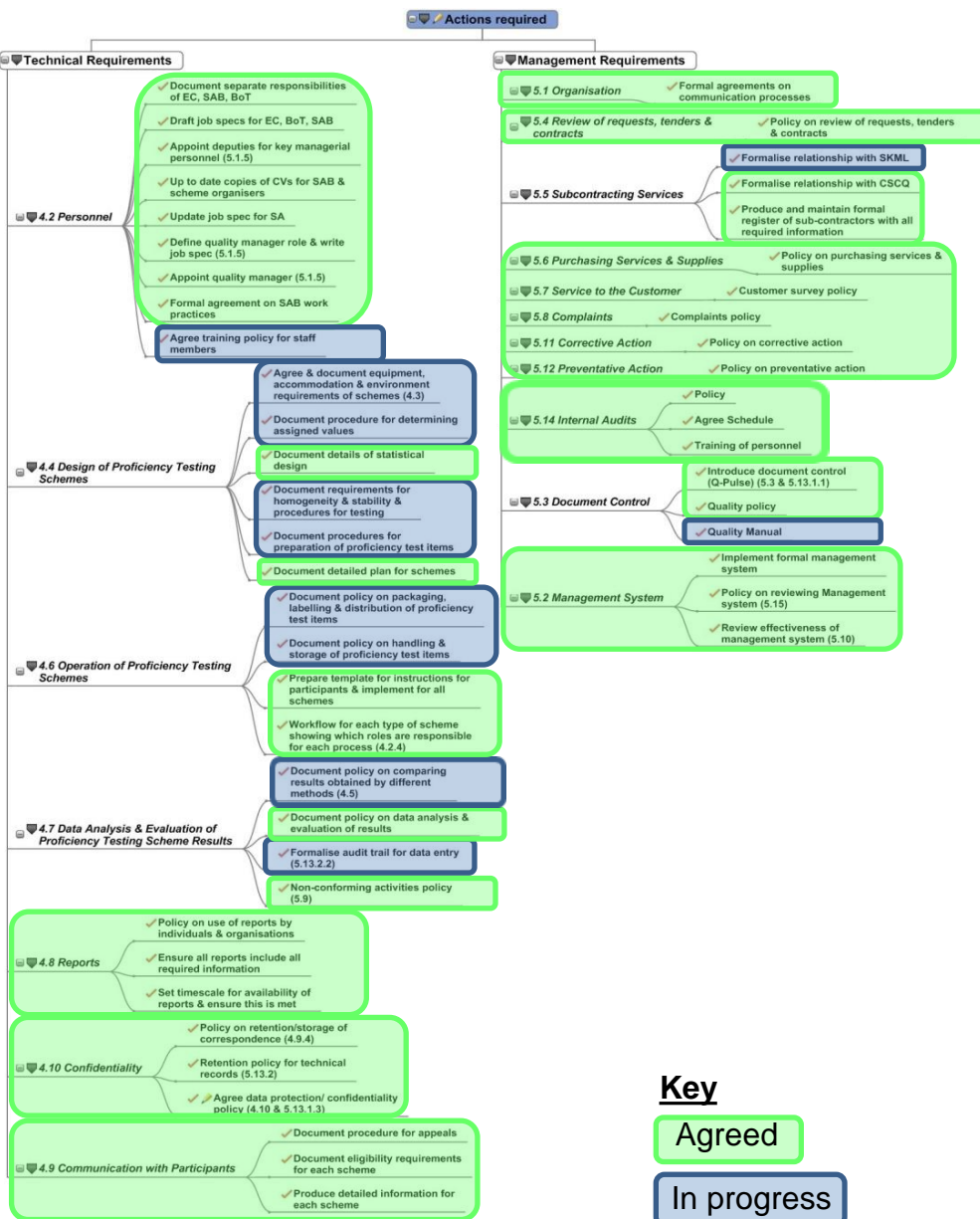
2019 & 2020 schemes

- **Finish scheme year in March of following year**

2021 schemes

- **Distribute submission dates of quantitative schemes more equally over the year: January-June, September, October (currently March-October)**

ERNDIM: Path to accreditation



Key

Agreed

In progress

- **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
- Management requirements were prioritised but now working on the Technical Requirements
- Aiming to submit initial application in early 2020

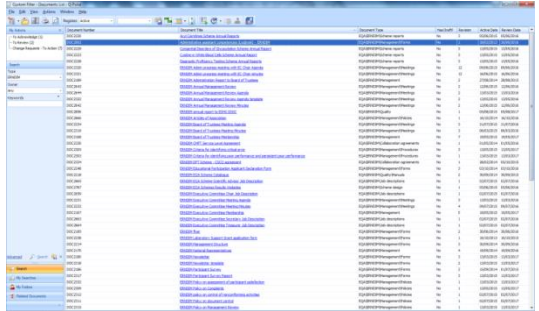
ERNDIM: Path to accreditation



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Progress so far:

- Document control system
- Management system
- Policies and procedures in progress
- Formal Internal audit calendar
- Formal agreements with sub-contractors in progress
- All qualitative schemes: **centralised sample dispatch and online results submission**



	DPT	Urine MPS	CDG	Qualitative Organic Acids	Acylcarnitines
Centralised sample dispatch	✓	✓	✓	✓	✓
Online results submission	✓	✓	✓	✓	✓

ERNDIM: Administration Office



- Staff: Sara Gardner (Executive Administrator, full time)
Jenny Barrett (Scientific Administrator, full time)
Kate Straznikiewicz (Administration Assistant, 0.5 x full time) – joined ERNDIM February 2019
- Responsible for administration **EQA schemes & SSIEM Academy**
- Central to the day to day running of ERNDIM
- Preparation of **ISO accreditation**
- In 2017, SSIEM agreed to fund a full time Scientific Administrator (deputy to Sara) for 3.5 years. Jenny Barrett was recruited in August 2017 and has made a significant impact on the activities of the Office
 - Long term aims is for ERNDIM to sustainably fund this post for the future
- SSIEM also agreed to support the software development costs which have allowed all schemes to move to on-line results submission

Thank you SSIEM!

ERNDIM: impact of BREXIT



Risks Associated with Brexit

- Increased costs, impact of exchange rates e.g. on shipping/cost of schemes for UK labs.
- Changes to customs/permits for UK labs
- Changes to VAT regulations so that VAT must be added to invoices for EU labs

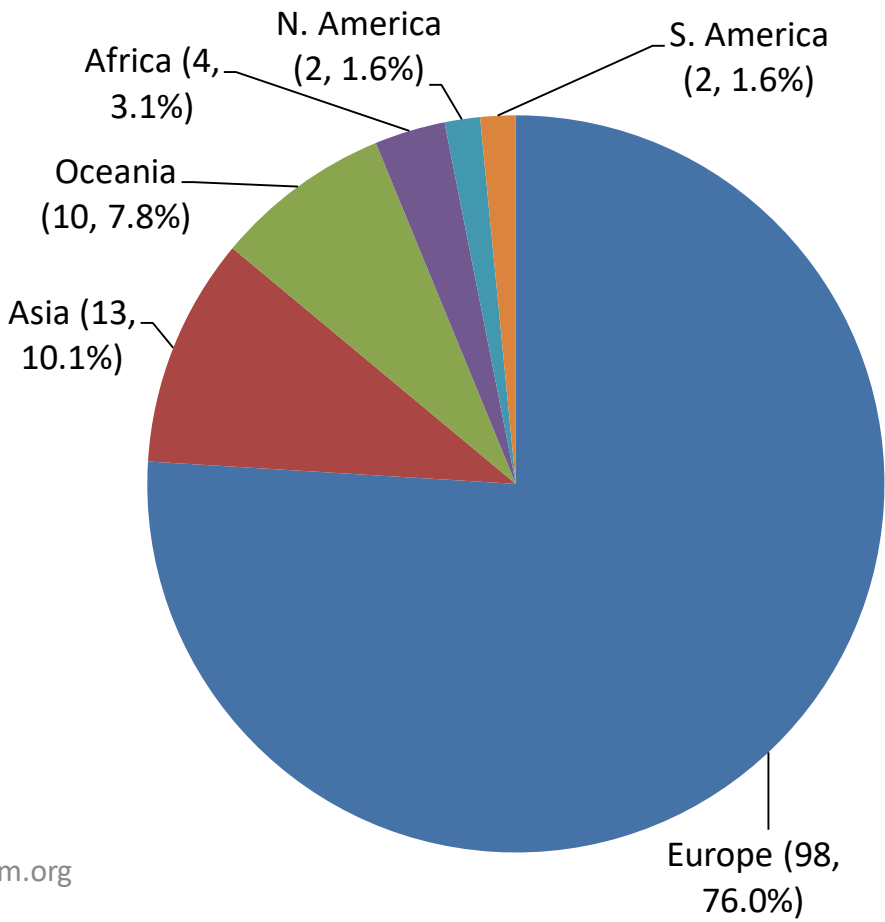
ERNDIM: SSIEM Academy



April 2019, Winterthur (near Zurich), Switzerland

- Course heavily oversubscribed (82 lab & 47 clinical applications from 45 countries)
- Accepted applications from 44 clinicians and 36 laboratory scientists from 32 countries.
- Topics: mitochondrial diseases, glycogen storage diseases, neurotransmitters and congenital disorders of glycosylation.
- The programme was a mix of joint and separate clinical/laboratory lectures and workshops.

Origin of 2019 Academy trainees



ERNDIM: SSIEM Academy



SSIEM Academy 2020

- 21st & 22nd April 2020, Amsterdam, The Netherlands
- Topics:
Aminoacidopathies, Hyperammonaemia and Urea Cycle Defects
- Event information can be found on the ERNDIM and SSIEM Events pages:
<https://erndim.org/home/news.asp> and
<http://www.ssiem.org/events/events.asp>
- Course applications will open later in September 2019 & will close on in late October 2019!



ERNDIM: other activities

Materials for internal QC

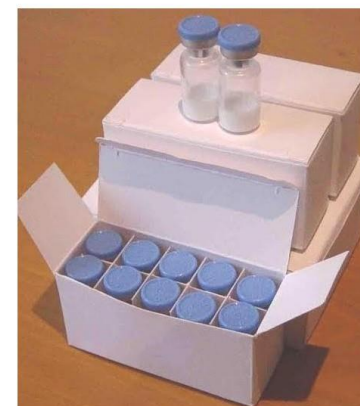
- New batches of AA, OA, PuPy, AC, SA produced separate from EQA materials

Reference standard

- Organic acids (supplied by dr Herman ten Brink)

Educational panels

- Oligosaccharide kit (supplied by MCA laboratory)



Work in progress

- Support sample exchange between labs in case EQA is not available (guidelines, sample preparation?)
- Biochemical Test directory: MetabERN-Orphanet-ERNDIM



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Thank You!