

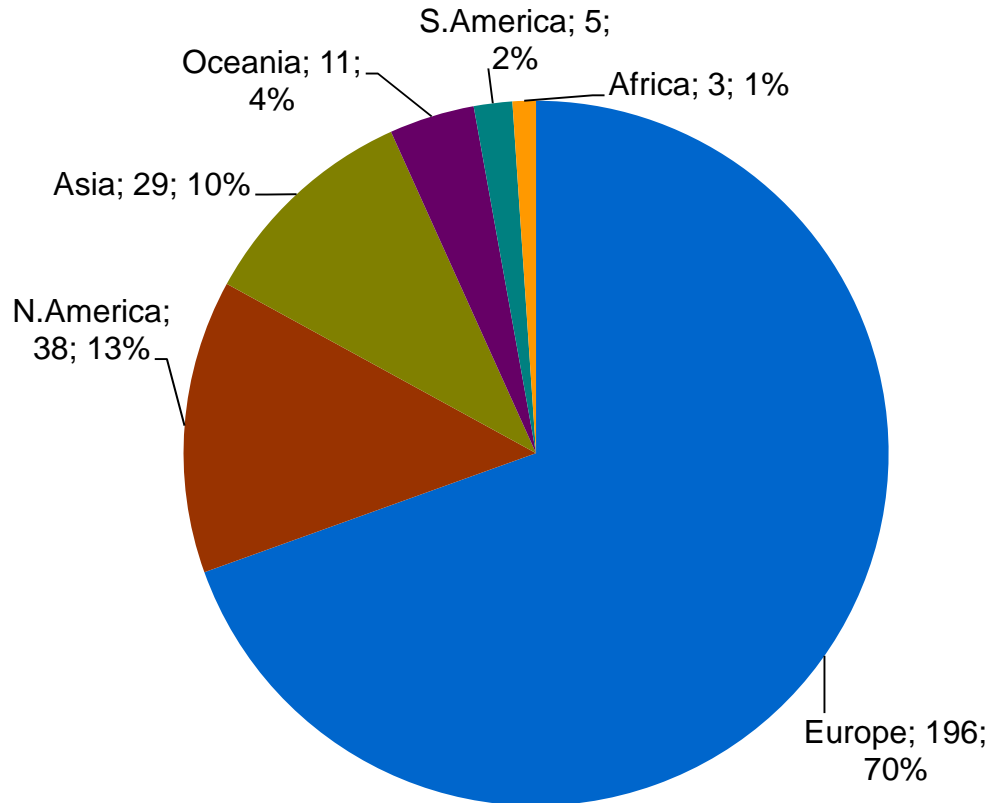


Chairman's Update

2014

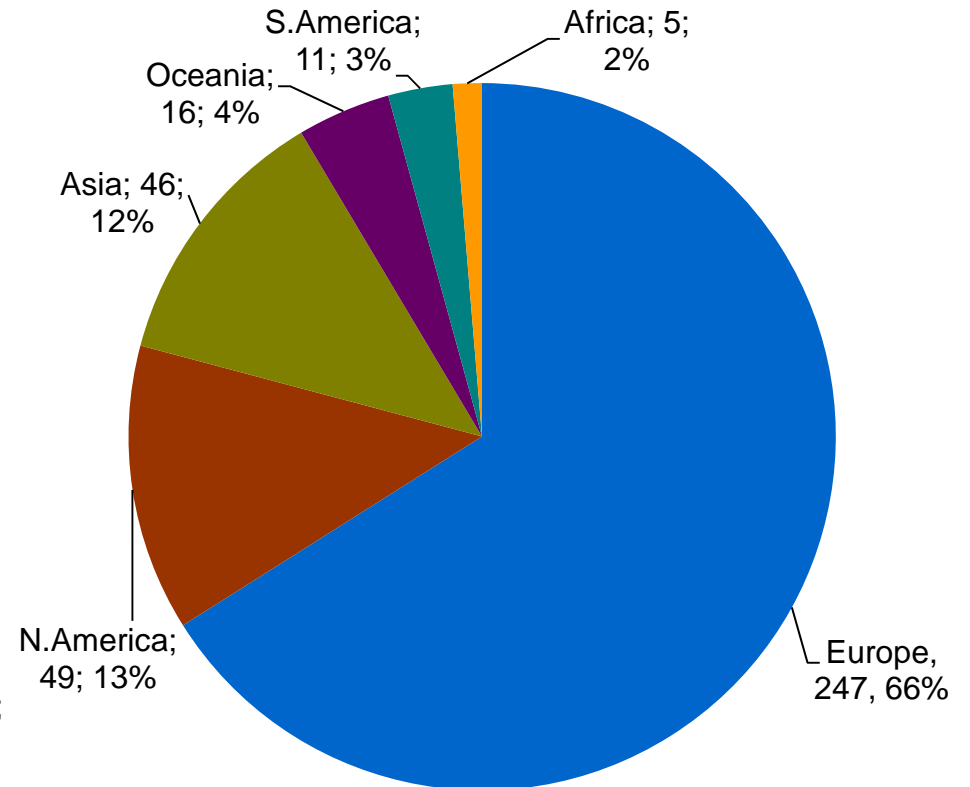
Mick Henderson

ERNDIM: Participants



2009: Number of participants = 282

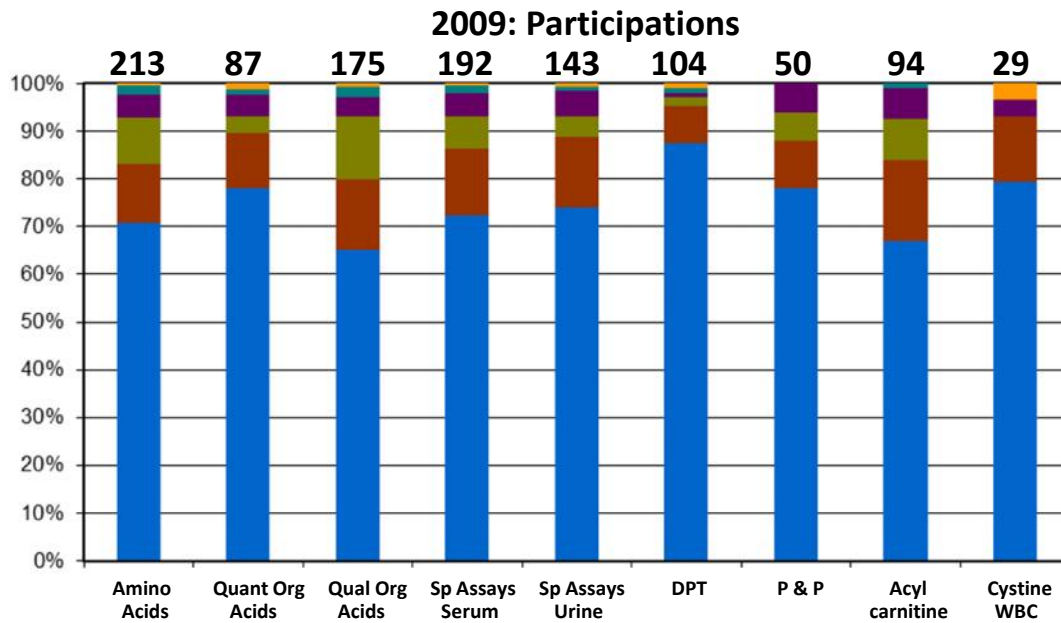
- Number of participating countries = 49



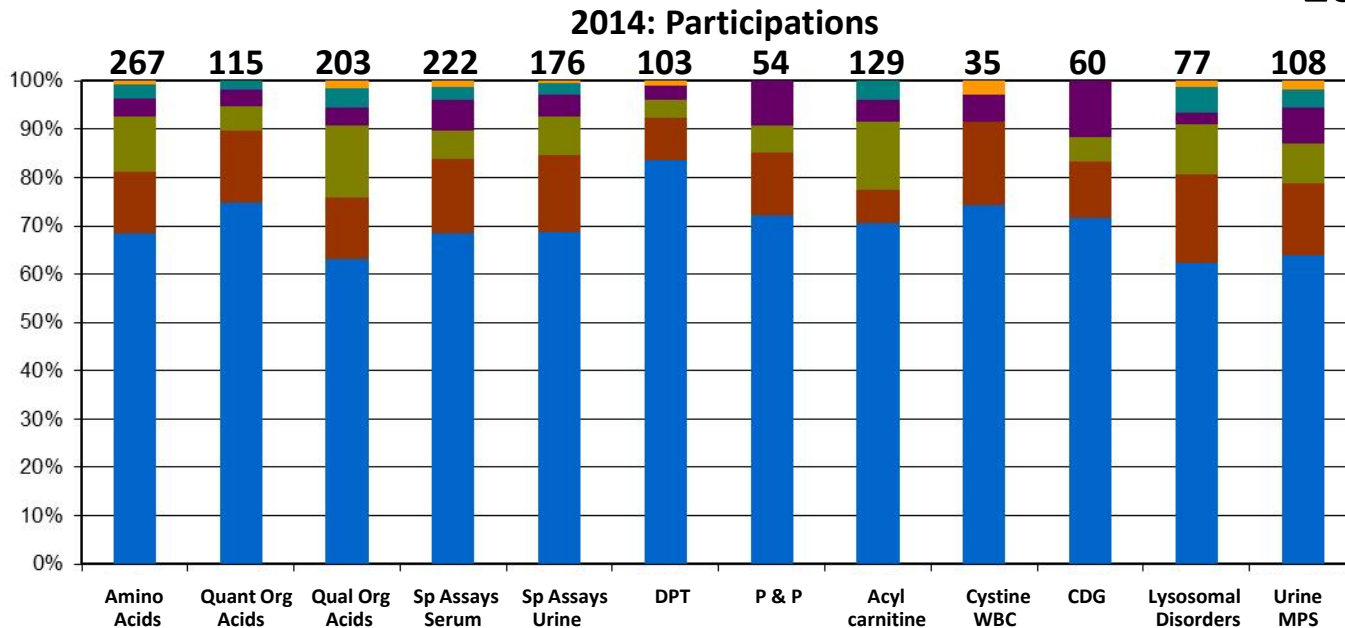
**2014: Number of participants = 374
(33% ↑ on 2009)**

- Number of participating countries = 59

ERNDIM: QA Schemes



2009: 9 QA schemes
1087 participations



2014: 12 QA schemes
1549 participations
(43% ↑ on 2009)

ERNDIM: QA Schemes



Pilot schemes

3 pilot schemes are running in 2014

1. Pterins in urine and Pterins/DHPR in dried blood spots, organised by Nenad Blau (University Children's Hospital, Zurich, Switzerland)
2. Neurotransmitters in CSF organised by Simon Heales (UCL Institute of Child Health, London, UK)
3. Lysosomal Enzymes in dried blood spots, organised by Kees Schoonderwoerd (University Medical Centre, Rotterdam, The Netherlands)

Participation in these schemes is limited but if you would like to be contacted if participation can be widened please email admin@erndim.org

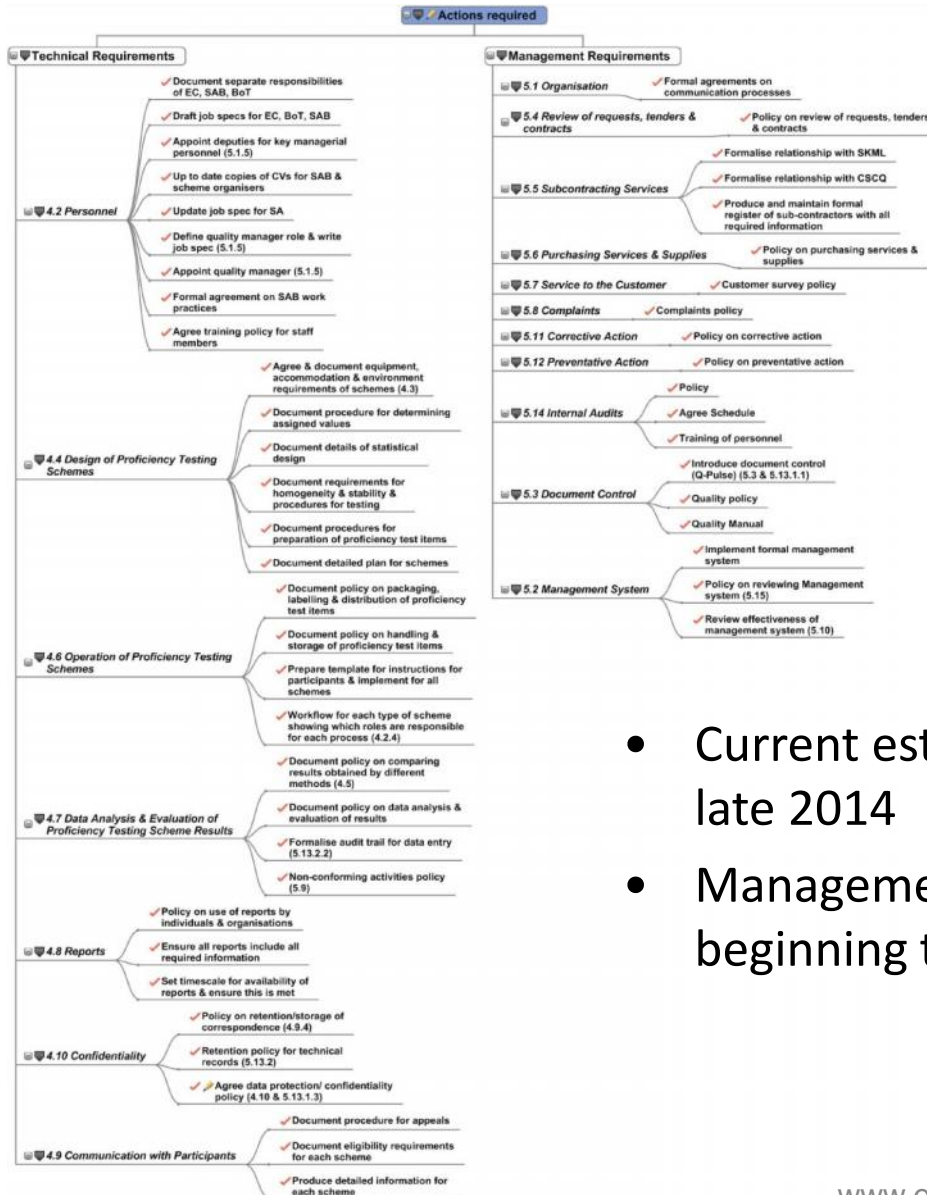
ERNDIM: Critical Error Policy



- Will be applied to Qualitative Scheme for 2014 scoring, to be ratified by SAB at March 2015 meeting
- Pressure to implement this policy from the Quality Committee ESHG
- Errors that would be unacceptable to the majority of labs and would have a serious adverse effect on patient management
- Critical Error will override a satisfactory score and will be automatic poor performance; will be shown on Certificate of Participation
- Appeals procedure in place

Full details will be on the ERNDIM website

ERNDIM: Path to accreditation



- 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
- Initial GAP analysis allowed development of list of proposed actions required to meet ISO/IEC 17043
- Current estimate for meeting these requirements is late 2014
- Management requirements were prioritised but now beginning to look at the Technical Requirements

ERNDIM: Path to accreditation



Progress so far:

- Terms of Reference for each of the ERNDIM bodies & an increased role for the Board of Trustees so that it can carry out an increased governance role.
 - Aim: half of the Trustees will be people who are not members of the Executive Committee or Scientific Advisory Board, but are experienced in the field.
 - Prof Katrin Ounap (University of Tartu, Estonia) was appointed as the Chair of the Board of Trustees in 2013.
- Job descriptions for important ERNDIM roles have been completed
- Appointment of a Quality Manager & deputies for EQA scheme Scientific Advisors
- Expansion of the Administration office (0.5 x full time Administration Assistant appointed in 2013)
- Documentation of existing ERNDIM policies and procedures is in progress
- Agreement on new policies for poor performance, critical error, appeals and educational participation for centres who are setting up a new test (currently just available for the qualitative schemes but will be expanded to include the quantitative schemes from 2015 onwards)

ERNDIM: Path to accreditation



Biography: Prof Katrin Ounap



- Prof Katrin Ounap is Head of the Department of Genetics at Tartu University Hospital, Estonia.
- She qualified as a paediatrician in 1988 and began her career in the Medical Genetic Centre at Tartu's Children's Hospital. During her early years she was instrumental in the introduction of newborn screening to Estonia in 1993. She completed her PhD thesis on phenylketonuria in 1999.
- Throughout her career Katrin has worked consistently to institute and improve clinical and laboratory services for the diagnosis and treatment of metabolic disorders.
- Katrin was invited to join the ERNDIM Board of Trustees in 2009 and from 1st January 2014 she took on the role of Chair of the Board.

ERNDIM: Path to accreditation



Online registration:

- The aim is to have online registration for the EQA schemes available for the 2015 schemes.
- All centres currently participating in ERNDIM EQA schemes will be sent information on how to register for the 2015 schemes.
- Centres not currently participating but interested in participating in 2015 should contact the ERNDIM Administration office (admin@erndim.org)

ERNDIM: SSIEM Academy



SSIEM Academy 2014

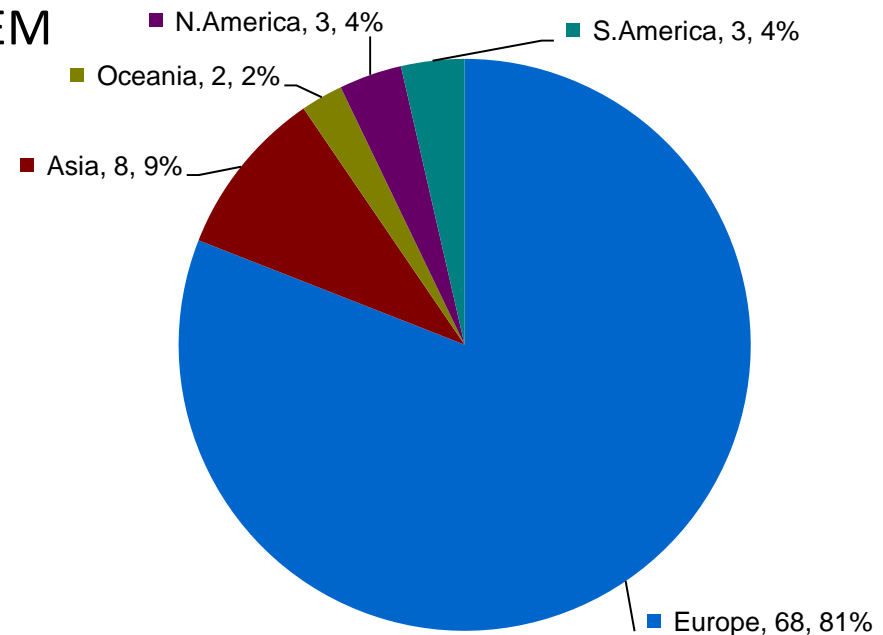
- 1st & 2nd April 2014, Paris, France
- Accommodation (2 night's) paid by SSIEM & 60 Euro non-refundable registration fee
- Course was heavily oversubscribed (55 applications for the clinical course and 53 for the laboratory course)
- Attended by 44 clinicians and 40 laboratory scientists from 35 countries
- Topics:

- **Joint Clinicians & Laboratory**

- **Scientists:** Lysosomal storage disorders, peroxisomal & other neurodegenerative disorders

- **Clinicians:** Management of LSDs, Peroxisomes, Metal disorders

- **Laboratory Scientists:** Peroxisomes, MPS, Purines & Pyrimidines



Origin of 2014 Academy delegates

ERNDIM: SSIEM Academy



SSIEM Academy 2015

- Late April/early May 2015
- Proposed venue: London, UK
- Topics:
 - **Joint Clinician & Laboratory Scientist:** CDGs, Clinicians: GSDs, Seizures, Neurotransmitters
 - **Laboratory Scientists:** GSDs, CDGs, Mitochondrial Disease
- The final programme and details of the venue will be announced at the end of September 2014
- Course applications will open in October 2014 on the SSIEM Academy page of the SSIEM website (www.ssiem.org/ssiemacademy)



Thank You!