

What is ERNDIM?



Brian Fowler, Basel / Zürich, Switzerland



What is ERNDIM?

- Origins of ERNDIM, a bit of history
- Our aims and philosophy
- Organisation
- Activities
 - External Quality Assessment Schemes
 - Teaching in SSIEM Academy
 - Training / Support
 - Meetings
 - Networking – SSIEM, EuroGentest, ESHG
- Future

What is ERNDIM? www.erndim.org

About

News & Events

EQA Schemes

Meetings & Reports

Training

Newsletters

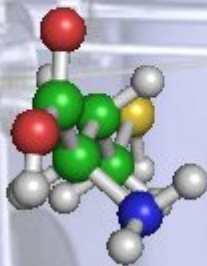
Laboratory Directory

Links



Evaluation
and Improvement

Welcome to ERNDIM



ERNDIM . . .

is synonymous with quality control of laboratory measurements within the field of the study of inborn errors of metabolism.

[read more](#)

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

Current Newsletter



Meeting of the "expert committee" of the ERNDIM,
Diepenbeek, Belgium, May 7th, 1993

REPORT

Present: C. Bachmann, J.R. Bonham, J.L. Dhondt, A.H. van
Gennip, P.P. Kamoun, J.L. Willems, L.J.M. Spaapen
Excuses from: Mrs. P. Divry, B. François, J.M.P. Trijbels.

Historical milestones:

- December 1991 establishment of ERNDIM: EUROPEAN RESEARCH NETWORK FOR EVALUATION AND IMPROVEMENT OF SCREENING, DIAGNOSIS, AND TREATMENT OF INHERITED DISORDERS OF METABOLISM.
- January, 1992: initiative of Baudouin François for a concerted action to join the expertise in the various European countries in the field of the screening, diagnostics, and therapy of inherited disorders of metabolism (IDM). This resulted in submission of the ERNDIM-I project to the Biomedical and Health Research Programme (BIOMED-I) of the European Community.
The ERNDIM-I proposal was not qualified for subsidization; too ambitious, too extensive (too many topics), and too many participants.
- End of 1992: collaboration of the two Biochemical Genetics Centres of twin universities of Belgium's and Netherlands Limburg to work out a new proposal for BIOMED. This ERNDIM-II project was restricted to the development and execution of a European Quality Assurance Program (QAP) for biochemical genetics laboratories.
- February, 1993 submission of the ERNDIM-II proposal.

FIRST MEETING OF THE ERNDIM
European Research Network for evaluation and improvement of screening,
diagnosis and treatment of inherited disorders of metabolism.

Leuven
September 12th, 1992

Dear Colleague,

On behalf of the Council of the ERNDIM, we wish to thank Prof. J. Jaeken and the **SSIEM** the opportunity offered to organize the first general meeting in Leuven.

What is ERNDIM?- History

1991 unsuccessful application for European Community grant on many aspects of IEM (**too many**)!

1992 informal ERNDIM meeting at 30th SSIEM annual symposium in Leuven

1994 BIOMED-1 grant (Maastricht and Diepenbeek) on QA in IEM

1994 September 5th ERNDIM Foundation was formally registered in Maastricht.

5 EQA Schemes

Newsletters and workshops at the SSIEM annual symposium.

1998 BIOMED-2 grant (e.g. Newborn screening, poor performance assessment, links of QA to accreditation)

2005-2010 Eurogentest One

2010-2013 Eurogentest Two

2011 --- Office, Scientific Officer (Sara Gardner)



Becoming
Professional

Baudouin François



Leo Spaapen



What is ERNDIM?

Our aims and philosophy

Quality Assurance in IEM: special problems

Tasks tested by different types of ERNDIM QC schemes

Task	What do we test
• Patient/sample ID	±
• Sample collection	no
• Transport	yes ±
• Processing /assay	±
• Technical validation	implied (int QC)
• Interpretation ❄❄	yes / no
• Reporting ❄❄	yes
• Advice ❄	yes / no
• Turn-around-time	±
• Safety (disposal)	no

AIMS of ERNDIM

The goal of ERNDIM

Consensus between European Biochemical Genetics Centres **on reliable and standardised procedures** for diagnosis, treatment and monitoring of inherited metabolic diseases.

Achieved through

Quality control schemes operated according to accepted norms

On a **European wide** scale - too few labs in a single country for sufficient data to assess performance.

Education

Meetings, Recommended Operating Procedures
Annual reports of schemes on the internet.

Aim is full value for money

Minimal administration costs and efficient subscription collection.

What is ERNDIM?

Organisation

ERNDIM: www.erndim.org



Evof inborn errors
arof metabolism

About Us

Organisation and Key Persons



[KEY PERSONS IN ERNDIM](#)

5-Oct-2016



[Structure and Organisation of ERNDIM](#)

26-Jun-2014

About Us



[Aims of the Foundation](#)



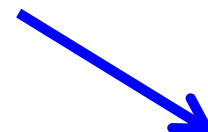
[Constitution](#)



[History of ERNDIM](#)



[Organisation and Key Persons](#)



Organisation

- **Board of Trustees**

Chair Katrin Ounap, Estonia

*6 External (Not Exec Comm or
8 SAB/ Exec Comm members)*

- **Executive Committee**

Chairs Exec Comm, SAB, Treasurer

- **Scientific Advisory board**

Scheme Advisors, *Chair Christine V*

- **National Representatives**

30 European countries

- **Administrative Office**

Manchester – 2 Scientific Officers, S

Sara Gardner



Exec. Comm Chair
George Rujter

What is ERNDIM?

Activities

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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 our schemes please [contact us](#)

Subscriptions

SUBSCRIPTIONS FOR THE ERNDIM 2017 EQA SCHEMES WILL BE OPEN FROM 12TH TO 14TH NOVEMBER 2017.

If you are an existing ERNDIM EQA participant you will be sent details of how to register for the 2018 EQA schemes on the ERNDIM Registration Website. To register for the 2018 EQA schemes you will need to log into the ERNDIM Registrations Website using the username and password which you will be sent by email. You will need to check all the information which is held on the ERNDIM Registration Website for your laboratory, update it where necessary and select the EQA schemes that you wish to participate in during 2018. 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 in one of our schemes please [contact us](#) before 14th November 2017.

Please make sure you register for the 2018 EQA schemes by **Tuesday 14th November 2017**. Subscription to a scheme may still be possible after this time but availability will be limited.

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Submit results for DPT and Urine MPS schemes

- [▶ ERNDIM EQA Scheme](#)
- [▶ EQA Scheme Catalogue 2017](#)
PDF Document download
- [▶ EQA Scheme Annual Reports](#)

Expansion of EQA schemes

	1995	2017
Numbers of schemes	5	15
Participating labs	179	397
Scheme Registrations	263	1604
Countries	36	60

Two Types of EQA schemes

Number participants

Quantitative Schemes

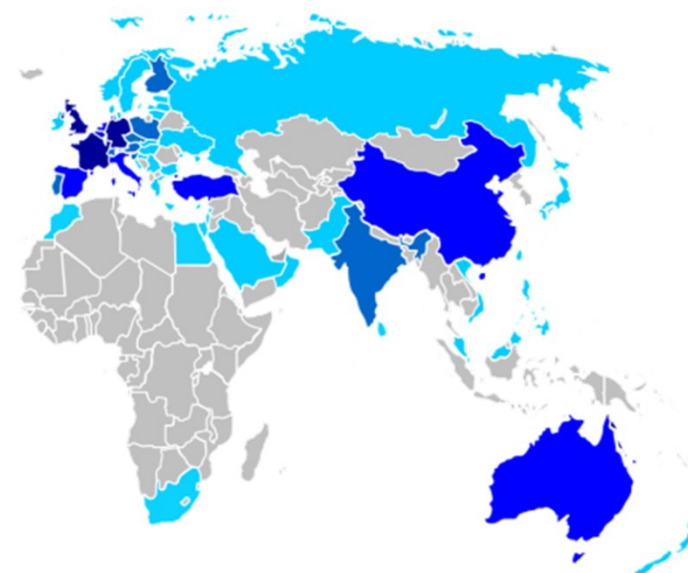
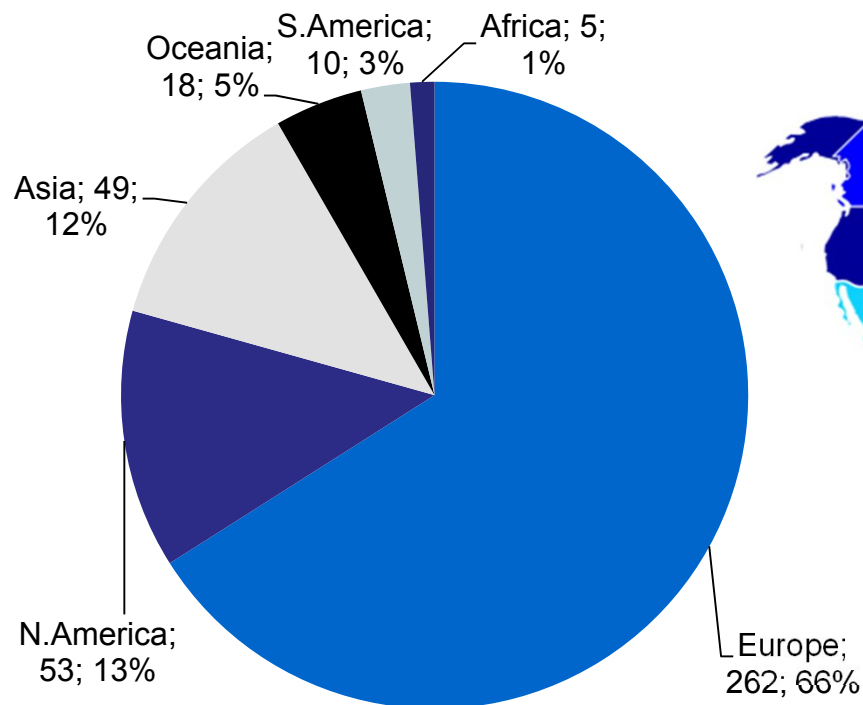
Acylcarnitines in Serum	121
Cystine in White Blood Cells	35
Lysosomal Enzymes (fibroblasts)	78
Neurotransmitters in CSF	26
Pterins in Urine	29
Purines and Pyrimidines (urine)	51
Quantitative Amino Acids (serum)	266
Quantitative Organic Acids (urine)	122
Special Assays in Serum	242
Special Assays in Urine	175

Qualitative Schemes

Acylcarnitines in dried blood spots	121
Congenital Disorders of Glycosylation	63
Diagnostic Proficiency Testing (urine)	108
Qualitative Organic Acids (urine)	214
Urine Mucopolysaccharides	100

Read all about the
Diagnostic Proficiency Scheme
Poster 899

ERNDIM: Participants



**2017: Number of participants = 397
(16% ↑ on 2012)**

- Number of participating countries = 60

No. of labs	
1-4	
5-9	
10-24	
25-45	

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Training for IEM in the EU

Training is a major issue

Interim Report on Biochemical Genetic Testing in Europe: deficits and needs and EQA

We concluded:

- Programmes are poorly developed or unavailable
- Lack of interested people for IEM
- Constitutes threat to continuance of patient care.

The SSIEM Education and Training Advisory Committee (ETAC)

Clinical	Laboratory
Andrew Morris - Chair	Simon Heales - Secretary
Johannes Häberle	Rachel Carling
Elaine Murphy	Christine Vianey-Saban
	Jorgen Bierau

Senior Advisors: Shamima Rahman, Brian Fowler

<http://www.ssiem.org/docs/ETACv6.pdf>

SSIEM ACADEMY 2017

April 2018



SOCIETY FOR THE STUDY
OF INBORN ERRORS OF METABOLISM

SSIEM ACADEMY

PRESENTATIONS ARCHIVE

SSIEM Academy 2017

SSIEM Academy 2016

SSIEM Academy 2015

SSIEM Academy 2014

SSIEM Academy 2013

SSIEM Academy 2012

SSIEM Academy 2011

SSIEM Academy 2010

SSIEM ACADEMY 2017

» [Joint Materials](#)

» [Laboratory Materials](#)

» [Clinical Materials](#)

» **JOINT LECTURES AND WORKSHOPS**

LECTURES



[Organic Acidaemias - Introduction](#)

[Organic Acidaemias - Clinical Cases](#)

Prof Henk Blom, University Medical Centre Freiburg, Freiburg, Germany

Dr Andrew Morris, Central Manchester Foundation Trust, UK

WORKSHOPS



[Organic Acidaemias \(Clinical\) \(4 parallel workshops\)](#)

ERNDIM: SSIEM Academy



SSIEM Academy 2018

- *23rd & 24th April 2018*
- Venue: London, UK
- Topics:
 - Lysosomal Storage Disorder
 - Peroxisomal Disorders
 - Purine & Pyrimidine Disorders
- The final programme and details of the venue will be announced at the end of September 2017
- Course applications will open in October 2017 on the SSIEM Academy page of the SSIEM website (www.ssiem.org/ssiemacademy)

SSIEM Academy Lisbon 2008



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Meetings and Reports

Please select an option from the menu

Meetings and Reports



[Meetings](#)



[Reports](#)



[EQA Scheme Annual Reports](#)



[Participant Survey Reports](#)

ERNDIM: Participant meeting 2017



- Meeting in Manchester separate from ICIEM in Rio
November 21-22, 2017
- Registration fee
- DPT meetings and workshops for other schemes
- Programme on ERNDIM Website
- Registration open soon

Newsletters

Newsletter Issues

General

General



Newsletter 2017

28-Jun-2017

2016 has been a special year for ERNDIM, because the first steps to establish the center were taken 25 years ago. In 1991 a group of enthusiastic Doctors and Biochemists



Newsletter 2016

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ERDIM & EuroGenTest: Achievements

- Development of harmonized performance assessment , scoring
- New participation certificate with performance
- Website submission of results
- National Representatives network established
 - Recommendations to overcome deficits in provision of BGT services in EU countries*
- Annual Scientific Workshops
 - Adjacent to SSIEM
- Training Workshops
- Directory:
- Expansion Diagnostic proficiency testing
- New schemes
- Standard methodologies and reporting procedures
- Publications

Directory of Laboratories

The ERNDIM Directory of Laboratories which perform diagnostic tests in Inherited Metabolic Diseases is intended to aid specialist workers in the field in selection of laboratories for sample referral.

[Go to the directory](#)



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Laboratories

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Links

E-mail

Eurogentest Committee

The ESHG-Eurogentest Committee focuses on quality aspects

Sub-committee members, aims and activities

The Quality subcommittee (QSC) will continue to work on issues that affect quality in genetic testing and wider aspects

Members

Ros Hastings (Chairperson, CEQA), Marta Rodriguez de Alba (Cytogenetics)

David Barton (EMQN), Els Dequecker (CF Network),

Brian Fowler (ERNDIM),, Viktor Kožich (SSIEM)

Thomy de Ravel, Conny van Ravenswaaij (Clinicians)

What is ERNDIM?

Future

What is ERNDIM? - Future

Future Requirements

- Recruit more labs
- Provide new schemes to meet changing needs
e.g scheme for Newborn –screening planned
need for schemes for metabolomics and proteomics
- Provide standard reagent kits and reference materials
- Link performance in EQA to accreditation
- Sustain efforts to improve performance
- Complete Accreditation

ERNDIM at 25 years of age



