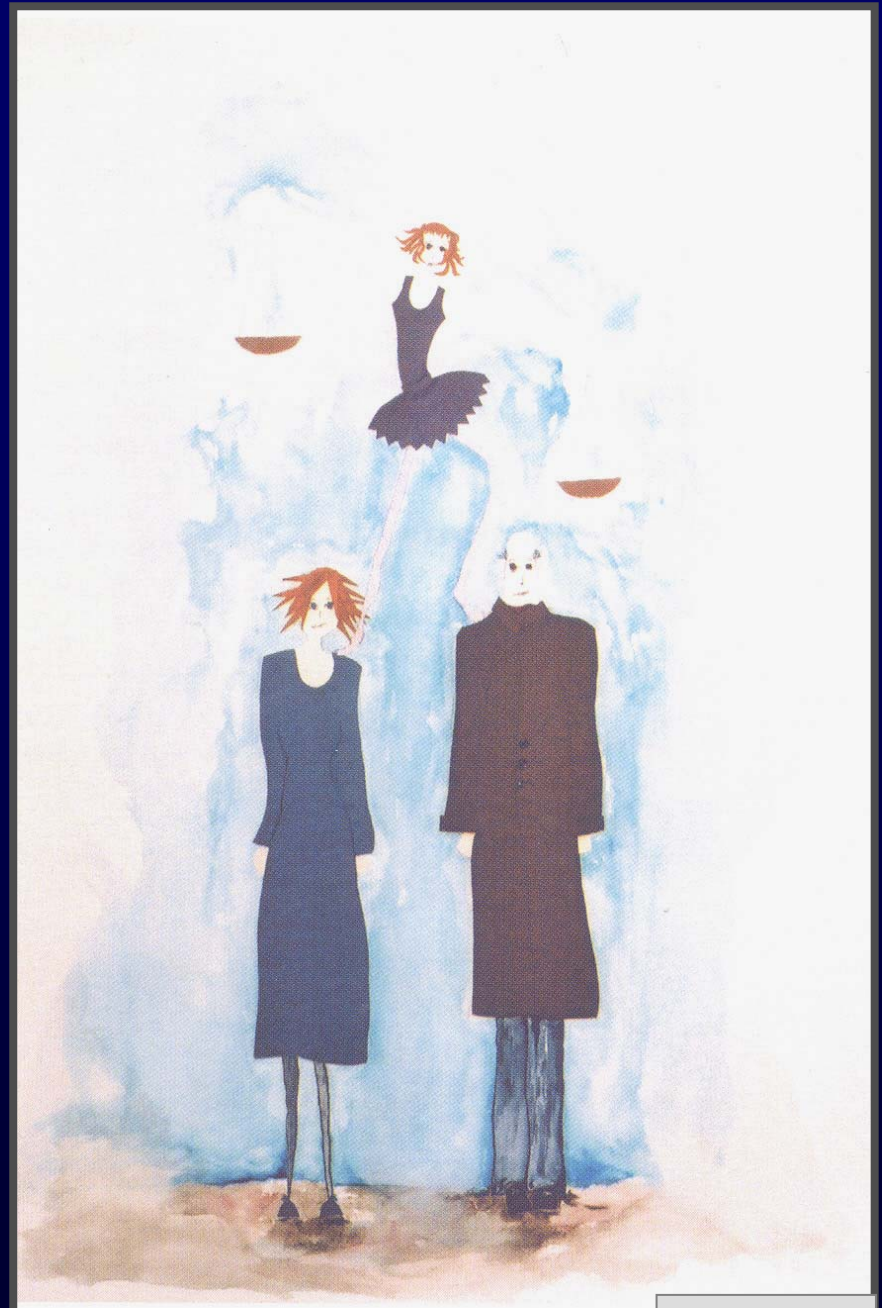




*Prof Brian Fowler, Lead Scientist for
Biochemical Genetics*

Co-ordinator: Jean-Jacques Cassiman
K.U. Leuven, Belgium



By Rebecca Kent

EU 25 Representatives meeting December 2 2005

Programme

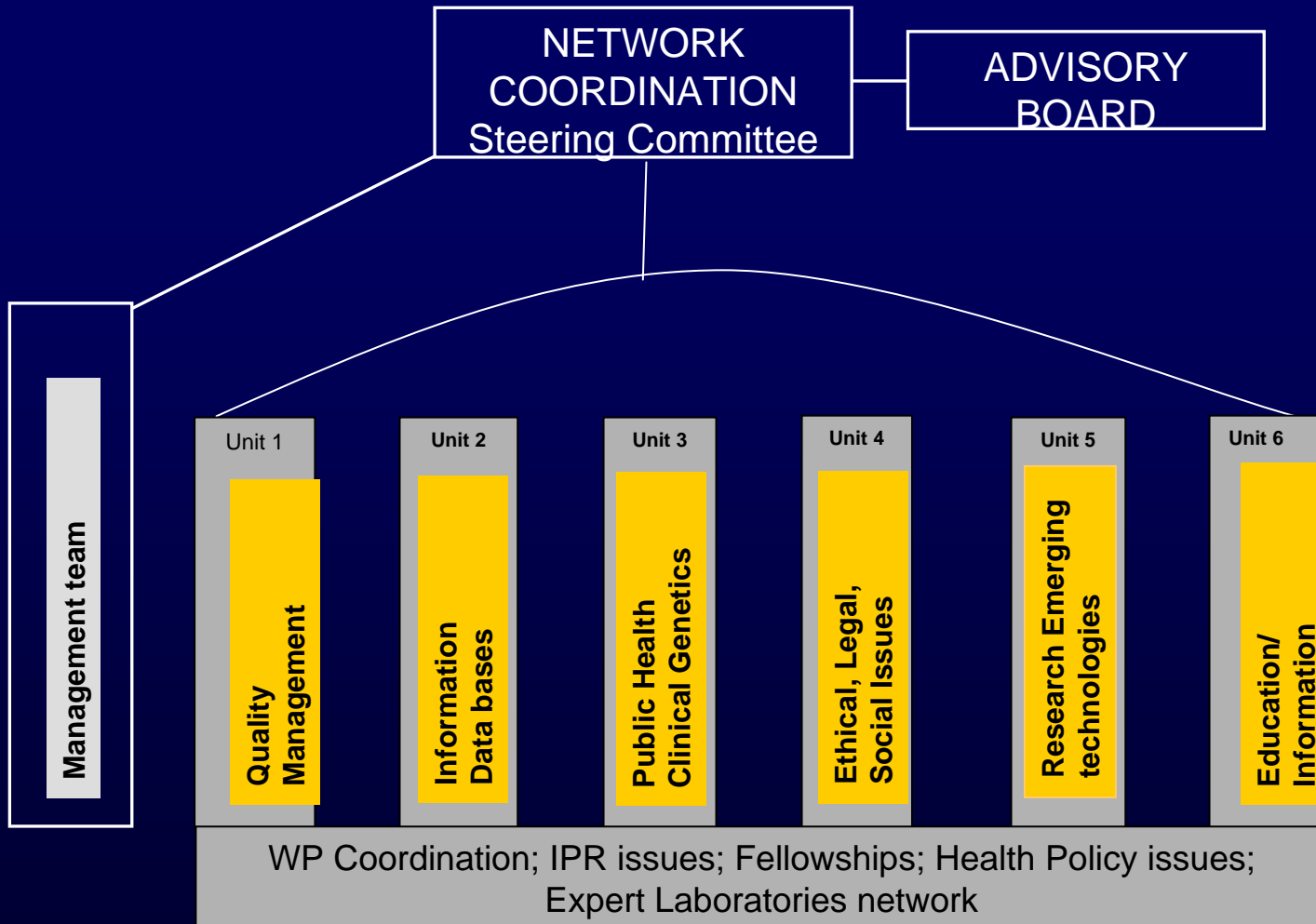
- 10:00 Introduction: ERNDIM Background, Overview of schemes and perspectives, aims of project Jim Bonham for Brian Fowler
- 10:20 Introduction of participants: strengths and weaknesses in individual countries (ca. 3 minutes each) Everyone
- 11:30 Quality control internal / external, why and how: special problems of IEM Leo Spaapen
- 11:45 Best practice / EQA experience: Amino acids / cystine Leo Spaapen for Brian Fowler
- 12:00 Best practice / experience: Organic acids Jim Bonham
- 12:15 Best practice / experience: Acyl Carnitines Charles Turner
- 12:30 Best practice / experience: Purines / pyrimidines Jorgen Bierau
- 12:45 Best practice / experience: Proficiency schemes Christine Saban
- 13:00 Lunch

EU 25 Representatives meeting December 2 2005

Programme

- 14:00 General aspects of diagnostic guidelines: UK experience
Jim Bonham
- 14:20 Needs assessment and review of IMD services in the UK
Anne Green
- 14:40 Work in five groups on needs / deficits in individual countries.
selected groups (see list for group distribution)
- 15:40 Presentation and discussion of group work
- 16:30 Summary and conclusions and future
- 17:00 Close of Meeting

Network of Excellence: "EUROGENTEST"



Relation between the units, activities and workpackages

Unit 1:
Quality Management and Accreditation / Certification of Genetic Testing
Elisabeth Dequeker (BE) / Michael Morris (CH)

Activity 1.1
Quality Procedures
Elisabeth Dequeker (BE)
Michael Morris (CH)

WP 1.1

Activity 1.2
QAu Database

Elisabeth Dequeker (BE)
Michael Morris (CH)

WP 1.2

Activity 1.3
Quality Assessment Schemes
Rob Elles (UK)
Clemmens Müller (DE)
Ros Hastings (UK)
Brian Fowler (CH)

WP 1.3
Molec

WP 1.4
Cyto

WP 1.5
Biochem.

Activity 1.4
Reference Systems and Procedures

David Barton (IE)
Christof Klein (BE)

WP 1.6

Activity 1.5
'Generic' method SOP

Gert Matthijs (BE)
Milan Macek (CZ)

WP 1.7



EUGT Work package Biochemical Genetics

Objectives of the work package

- To expand opportunities for Biochemical Genetic testing laboratories in EU25 to participate in EQA
- To link Internal Quality Control and EQA through consensus discussion on best practice

EUGT Work package Biochemical Genetics

Activities to meet objectives

- 1) Inclusion of complete data on biochemical genetics labs in the ERNDIM web site directory
- 2) Best practice meetings and development of poor performance criteria
- 3) Expansion of pediatric biochemical genetic EQA in EU25
- 4) To explore standard methodologies and standard reporting procedures
- 5) Coordination of IntQA, EQA and accreditation

EUGT Work package Biochemical Genetics

Work done Months 1-12

- 1) Directory: - Trial version completed
- all lab info available mounted
 - participant contacted to update **November**
 - open to public / new participants **December 2**
 - meeting held with A. Green
 - ? link to UK Directory **October**

ERNDIM web-site: www.erndim.ch



[HOME](#)

[About ERNDIM](#)

[ERNDIM Participants](#)

[Organisation](#)

[Educational Documents](#)

[Directory of Laboratories](#)

[E-MAIL](#)

[LINKS](#)

ERNDIM is now synonymous with quality control of laboratory measurements within the field of the study of inborn errors of metabolism. ERNDIM was founded in 1994 with its legal base in Maastricht, The Netherlands.

The full name of ERNDIM, the "European Research Network for evaluation and improvement of screening, Diagnosis and treatment of Inherited disorders of Metabolism" reflects the original ideals and vision of its founders.

Directory of Laboratories

DLD Search Results - Microsoft Internet Explorer bereitgestellt von Internet Professionell

Datei Bearbeiten Ansicht Favoriten Extras ?

Zurück Vorwärts Abbrechen Aktualisieren Startseite Suchen Favoriten Verlauf E-Mail Drucken Bearbeiten Diskussion Messenger Druckvorsch.

Adresse <http://www.golduk.net/dld/search.jsp>

ERNDIM-SSIEM
Diagnostic Laboratories Database

Search

[New Search](#)
[Last Results](#)

[Back to Options](#)

Search results for Cystathionine beta-synthase (Homocystinuria) in Switzerland:

Switzerland	Switzerland, Basel, University Childrens Hospital	Switzerland	More info
Outside Switzerland	Netherlands, Rotterdam, Erasmus University MCg,	Netherlands	More info
	UK, Belfast, Belfast Link Labs	Northern Ireland	More info
	UK, Peterborough, Peterborough District Hospital, Anglian Biochemical Genetic Unit and Neonatal Screening Unit	United Kingdom	More info
	UK, Birmingham, Birmingham Children's Hospital NHS Trust	United Kingdom	More info
	UK, Birmingham, University of Birmingham	United Kingdom	More info
	UK, Edinburgh, Royal Hospital for Sick Children, Department of Paediatric Biochemistry	United Kingdom	More info
	UK, London, Great Ormond Street Hospital, Chemical Pathology Services	United Kingdom	More info
UK, Manchester, Royal Manchester Children's Hospital, Willink Biochemical Genetics Unit	United Kingdom	More info	

ERNDIM Laboratories Directory



ERNDIM
Diagnostic Laboratories Database



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Introduction

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.

Tests, referred to as analytes, for single or groups of metabolites, enzyme activity and mutation analysis are included. In using the directory a specific analyte is chosen from the pull-down list and laboratories providing the service are listed, first from a specified country followed by other countries. Contact and other details of the listed laboratories are found under "more info".

Applications from laboratories wishing to be included in the directory or requests for inclusion of additional tests should be sent to the web site administrator (e-mail above).

Disclaimer

Information contained in this directory is provided by the individual laboratories who themselves are responsible for its accuracy.

The ERNDIM organisation takes no responsibility for the quality and reliability of the services provided by laboratories listed on this website which resides with individual laboratories.

The inclusion of a laboratory in this directory does not represent or infer endorsement by ERNDIM.

When considering referral of samples to a listed laboratory it is recommended that attention be paid to participation of the laboratory in external quality assurance schemes which can be found in the laboratory's own information sheet.

Presentation of ERNDIM web-site and
Lab directory

Piotr Litynski

EUGT Work package Biochemical Genetics

Activity 1: work planned months 13 - 30

Directory of IEM labs

- Completion / update existing participants
release for public access month 12
- Recruitment / incorporation further participants months 13-18
- Maintenance / updating /
co-ordination with other EUGT directories /
national directories (e.g. UK model) months 19-30

EUGT Work package Biochemical Genetics

Activity 2 : work planned months 13 - 30

Best Practice and Performance Assessment

- Best Practice Guidelines, months 13-18
good/poor performance criteria
developed at ERNDIM SAB / Exec. Comm.
- Standards for assessment of performance months 19- 24
in biochemical genetics EQA
(ERNDIM Board and Scientific advisors)
- Establishment of rules linking performance months 25 - 30
to certificate issue established and put
into practice (ERNDIM Exec. Comm. and SAB)

EUGT Work package Biochemical Genetics

Activity 3 : work planned months 13 - 30

Expansion in EU25

- Establishment of deficits and need in all EU countries, especially EU 16-25 months 13-18
- Establishment of additional ERNDIM Proficiency test centre in Basel months 12 -24
- 2nd Best Practice Meeting / all EU25 Reps. month 24
- On-site national or international workshops for methods and quality control months 21-30

EUGT Work package Biochemical Genetics

Activity 4 : work planned months 13 - 30

Standard methodologies

- Mucopolysaccharides (MPS), Organic Acids months 13-18
- Development of standard reporting procedures months 19-30
- Further standard methodologies months 19-30
 - groups of biochemical genetic analytes
 - Purines/Pyrimidines, cystine
 - various special assays etc

EUGT Work package Biochemical Genetics

Activity 5 : work planned months 13 - 30

Co-ordination with other Genetic disciplines

- Meetings on knowledge exchange, harmonisation months 13-24
with Cytogenetics /Molecular Genetics partners
- Instigation of harmonisation measures months 25-30
- Unit 1 meetings months 13-30

This becomes new work package WP1.9

ERNDIM National Representatives

Country	Name	Country	Name
Austria	Olaf Bodamer	Belgium	B. François
Czech	Pospisilova Eva	Cyprus	A. Drousiotou
Denmark	E. Christensen	Estonia	K. Ounap
Finland	Kari Pulkki	Hungary	Agnes Schuler
France	Christine Vaney-Saban	Ireland	Richard Walsh
Germany	Langhans Claus-Dieter	Latvia	Parsla Vevere
Greece	Helen Michelakakis	Lithuania	Jurgita Songailiene
Italy	Amelia Morrone	Luxembourg	Jean-Paul Hoffmann
Netherlands	Leo Spaapen	Malta	
Portugal	M.M. Silva	Poland	Wanda Gradowska
Spain	Antonia Ribes	Slovakia	D. Behulova
Sweden	Elisabeth Holme	Slovenia	M. Zerjav-Tansek
Switzerland	Brian Fowler		
United kingdom	James Bonham		

First meeting of EU25 national representatives

Aims

Information exchange on best practice in Biochemical Genetic Laboratory testing

Preliminary Assessment of Deficits and Needs for Biochemical Genetic Laboratory testing in all EU25 countries

Questionnaire analysis

2006 ERNDIM and EU25 national representatives meetings

No SSIEM (ICIEM in Japan)

ERNDIM meeting 2006

Day 1 DPT individual meetings

Workshop on Best Practice for group of analytes as in previous years

ERNDIM Board meetings

Day 2 Extended EU 25 best practice meeting

Free communications

Location Prague

Date 2nd. half October