

# Third meeting of ERNDIM National Representatives

Needs and deficits in Biochemical Genetic Testing in Europe

## AIM

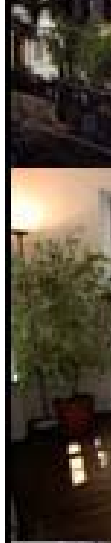
To produce recommendations for the provision of comprehensive services for biochemical genetic testing in all (small/large, old/new) EU (European) countries.

first EU25 Best Practice Meeting in Basel, December 2005

→ interim report

- meeting in Prague, October 2006 → Feedback and Comments to Interim report by National Representatives

# First EU25 Best Practice Meeting in Basel, December 2005 → interim report



**BETHESDA**  
fürs Leben gut



BETHESDA-GRUPPE



BASEL

Spital Schule Seminar Schwesterngemeinschaft



KÜSNACHT





## **The Eurogentest Project and ERNDIM**

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

- Background
- Developments in Biochemical Genetic Testing in Europe: the role of ERNDIM
- The Eurogentest Project and the role of ERNDIM
- Report of the first Eurogentest Best Practice meeting of National Representatives of EU25 held on December 2<sup>nd</sup>, 2005 in Basel.
- Conclusions and recommendations on deficits and needs
- Feedback, discussions and action based on the recommendations (second Eurogentest Best Practice meeting of National Representatives of EU25 held on October 5-6, 2006 in Prague)

# Interim Report on biochemical genetic testing in EU25

• Questionnaires from each country

• Excel table: BGT in EU25 GENERAL

Pop. Mio	in	Birth/Y	NBS No.Dis orders	NBS No. Centres	No. BGT labs	AA		Org Ac		CHO Metab		of usual dence
						Test/Y	c/Y	Test/Y	c/Y	Test/Y	c/Y	
						2700	20	2500	30			

• Excel table: BGT in EU25 Tests and C

Amino acids

Organic acids

Carbohydrate metabolism

Fatty acid oxidation defects

Respiratory chain defects

MPS defects

Sphingolipidoses

Purine/Pyrimidine disorders

Peroxisomal disorders

Creatinine synthesis disorders

Congenital glycosylation disorders

	15		12		5
350	1-2	250	1-2	30	1
6300*	23*	4000	9	2500	15
2500	10	1500	6	250	1
348	3	198	0.3	387	0.5
1500	5	1000	5	20	0
20000	?	12000	?	?	?
4250	8	840	14	30	1.6
n.a.	n.a.	n.a.	n.a.	n.a.	n.a.
n.a.	n.a.	n.a.	n.a.	n.a.	n.a.
1299	16	2556	11	20	0

# Interim Report on biochemical genetic testing in EU25

## Recommendations to overcome deficits

Progress and Feedback and discussion on recommendations by National Representatives at meeting held in Prague, October 5-6, 2006.

1. *Define basic standards / minimum core requirements / test repertoires in relation to size of country*
2. *Identification of clusters of countries/sharing where workloads are very low. Exchange visits and / or workshops*
3. *Training initiatives for implementing new tests*  
*Initiatives for training in reference labs in more developed countries*
4. *National and international reference laboratories for training should be identified*
5. *Stimulation of accreditation of laboratories by scoping present status*
6. *Accreditation of the EQA schemes themselves*

## Interim Report on biochemical genetic testing in EU25

7. *A survey of the scope of Metabolic Physician and Biochemist Training*
8. *An Initiative similar to that taking place in the UK to scope the number of patients with certain IMD disorders across Europe*
9. *Establishment of National registers of diagnosed cases*
  - *through existing national organisations*
  - *through European wide action*
10. *Expansion of EQA and improved availability of quality assessment materials including cell banks for biological material*
11. *Best Practice guidelines for methodology, minimum services and QA*
12. *The advised recommendations and issues should be a Directive of the European Commission.*

## recommendations for biochemical genetic testing services

7. *A survey of the scope of Metabolic Physician and Biochemist Training*
8. *An Initiative similar to that taking place in the UK to scope the number of patients with certain IMD disorders across Europe*
9. *Establishment of National registers of diagnosed cases*
  - *through existing national organisations*
  - *through European wide action*
10. *Expansion of EQA and improved availability of quality assessment materials including cell banks for biological material*
11. *Best Practice guidelines for methodology, minimum services and QA*
12. *The advised recommendations and issues should be a Directive of the European Commission.*

- a) Importance or relevance today
- b) Progress made, yes or no?
- c) Further measures needed

# Update from ERNDIM / EUGT

## European Human Genetics Society sub-committee

aiming to improve the quality of genetic services in Europe (EQA organisers, Clinical Geneticists, Research, patient groups, industries)

This can give authority (governance) to ERNDIM

1st meeting june 1st

Accreditation of ERNDIM as umbrella organisation



# Plan for accreditation of ERNDIM

Decision to go ahead	end of 2007
Decide which accreditation body (UKAS ?)	
GAP analysis for ERNDIM (see that for EMQN)	April 2008
Identification of necessary costs and source of funding	June 2008
Decide on physical location of activities. ERNDIM base, where are documents stored, best in centre which is accredited. Discussions with EMQN ongoing	
Documentation of all activities in SOP format	} according to generic/ SAS documents June 2009
Development of document control system guide	
Quality management handbook	
Run the system for 1-2 years	June 2010
Apply to accreditation body	End 2010

# Update from ERNDIM / EUGT

## DPT schemes: development of web-site reporting / future organisation

- pilot web-site developed with CSCQ due for results of second circulations of Basel / Prague schemes
- possible distribution of samples / administration of schemes by CSCQ

## New participants certificate

detailed for each scheme including performance

## ERNDIM Laboratory Directory

ca.200 potential new labs

new laboratory information mask including quality management details  
(integration into EUGT Quality Database)

# Certificate of Participation and Performance



This certificate is to confirm that NAME of LAB  
 participated in the following external quality assurance schemes organised by ERNDIM  
 in 2007 and has achieved performance in the individual schemes as shown. It is **important** that the laboratories'  
**detailed annual report** is referred to alongside this certificate.

[Quantitative Schemes](#)

## Certificate of Participation and Performance



	Subscribed	Submitted Results	Satisfactory performance
<b>Special assays Serum</b>	✓		
3 OH Butyric acid		✓	✓
7-Dehydrocholesterol		✓	✓
C22:0 Behenic acid			
Sodium thiosulphate		✓	✓
Succinylacetone		✓	✓
Uric acid		✓	
Creatinine		✓	✓
<a href="#">Proficiency Schemes</a>			
<b>Qualitative Organic Acids</b>	✓	✓	✓
<b>Qualitative Acyl Carnitines</b>			
<b>Diagnostic Proficiency Testing</b>	✓	✓	✓

Signed on behalf of ERNDIM, date

<b>Special assays Urine</b>	✓		
5-OH-Indolacetic acid		✓	✓

<a href="#">Proficiency Schemes</a>			
Qualitative Organic Acids	✓	✓	✓
Qualitative Acyl Carnitines			
Diagnostic Proficiency Testing	✓	✓	✓

Signed on behalf of ERNDIM, date

# ERNDIM National Representatives

Country

Name

Austria	Adolf Mühl ✨
Czech	Viktor Kozich
Denmark	Ernst Christensen
Finland	Kari Pulkki ✨
France	Christine Vaney-Saban ✨
Germany	Claus-Dieter Langhans ✨
Greece	Helen Michelakakis ✨
Italy	Ubaldo Caruso
Netherlands	Ries Duran
Portugal	Margarida Silva ✨
Spain	Begona Merinero
Sweden	Elisabeth Holme ✨
Switzerland	Brian Fowler ☹️
United Kingdom	James Bonham ✨

Country

Name

Belgium	Marie-Francoise Vincent
Cyprus	Anthi Drousiotou ✨
Estonia	Katrin Ounap ✨
Hungary	Agnes Schuler ✨
Ireland	Richard Walsh ✨
Latvia	Parsla Vevere ✨
Lithuania	Jurgita Songailiene ✨
Luxembourg	Jean-Paul Hoffmann ✨
Malta	Isabella Borg
Poland	Wanda Gradowska ✨
Slovakia	Darina Behulova ✨
Slovenia	M. Zerjav-Tansek
Bulgaria	Ivo Kremensky
Croatia	Ksenija Fumic
Rumania	?

# Role of National Representatives

Active recruitment of participants for ERNDIM EQA

list of present ERNDIM participants sent out to NRs

Inform national accreditation bodies of the existence of ERNDIM schemes

Jim Bonham will do this for the UK as prototype

Consider possible grouping of countries within the EU

do we need consortia based on size / locality etc.?

Encourage laboratory workers to attend training workshop at SSIEM  
Lisbon

see <http://www.ssiem.org/ssiemacademy.asp> (deadline 12th May)

Training Initiatives

ERNDIM web-site section

Request from Mick Henderson for National Information



HOME

About ERNDIM

ERNDIM Participants

Organisation

Training & Education

Directory of Laboratories

E-MAIL

LINKS

Meetings & Reports

## Announcement

I am very pleased to announce the introduction of two new support grants:

- [Training grants](#) to allow trainees, to spend a short visit to a European ERNDIM approved laboratory to gain experience and specialised knowledge.
- [Support grants](#) to allow laboratories from European countries suffering severe financial restrictions to participate in ERNDIM QA schemes.

Please note that two new educational documents on [White cell cystine determination and the role of EQA](#) and [Role of EQA in special assays for IEM](#) have been mounted in the [Training and Education](#) section.

Brian Fowler



HOME

About ERNDIM

ERNDIM Participants

Organisation

Training & Education

Directory of Laboratories

E-MAIL

LINKS

Meetings & Reports

### ***ERNDIM workshop at the September 2008 SSIEM meeting in Lisbon***

This year's ERNDIM meeting will take place at the SSIEM in Lisbon on Tuesday, **September 2**, 9.00 h until 12.30 h. The 5 individual DPT meetings will be followed by a joint workshop for all ERNDIM participants and will be open to all SSIEM participants. The topic of the workshop will be diagnostic aspects of creatine synthesis disorders. Please note that the PowerPoint files of the various presentations at last years workshop on peroxisomal disorders are mounted in the [Meetings and Reports](#) section of this website.

### ***Training of laboratory scientists***

A one day training workshop for laboratory workers, organised by ERNDIM under the aegis of the SSIEM academy will take place on Monday, September 1, preceeding the annual symposium of the SSIEM in Lisbon.

For full details see: <http://www.ssiem.org/ssiemacademy.asp>

A new section titled [European Training Schemes for Scientists](#) has been mounted in the [Training and Education](#) section.



## ERNDIM 2008

### National Representatives Meeting

Basel, May 9, 2008

#### Programme

9:00	Introduction	Brian Fowler	20 min
9:20	Organisation of BGT testing in 4 representative countries		
	- UK	Jim Bonham	20 min
	- Estonia	Katrin Õunap	20 min
	- Italy	Ubaldo Caruso	20 min
	- Netherlands	Marius Duran	20 min
10:40	Coffee break		20 min
11:00	Group sessions (groups 1, 2 and 3)		2 hours
13:00	Lunch time		1 hour
14:00	Report of group sessions (each group 10 min)		30 min
14:30	Develop guidelines (new groups A, B, C)		45 min
15:15	Coffee break		15 min
15:30	Final discussion and conclusion		1 hour
16:30	Close of meeting		



# recommendations for biochemical genetic testing services

## Group Work

Test criteria for categories of inherited metabolic diseases:

Table 1

Country	Tests / Disorders		
Criteria for decision	Aminoacidopathies		
	PKU	Urea cycle disorders	other (not organic acidurias)
Frequency of diseases in your country			
Presentation			
Necessity of monitoring			
Equipment, instruments			
Expertise level needed			
Organisation at present in your country			
Reason for lack of service			

# recommendations for biochemical genetic testing services

## Group Work

Test criteria for categories of inherited metabolic diseases:

- **Frequency of diseases:** high =  $< 1:20.000$ , medium =  $1: 20.000 - 1: 100.000$ , low =  $> 1: 100.000$
- **Presentation:** Is acute presentation prevailing/ important in this category?
- **Necessity of monitoring:** Is there a necessity of frequent (i.e. more than 6 times a year) and timely monitoring (i.e. 24h-service and turnaround time less than 6 hours)?
- **Equipment and instruments:** easy, medium, difficult, expensive
- **Expertise level needed:** low, medium, high, very high
- **Organisation:** regional, national, international

# Groups for ERNDIM National Representatives meeting

Chairmen: Jim Bonham, Viktor Kozich, Leo Spaapen  
Brian Fowler and Mick Henderson will move between groups.

<b>Group 1</b>	<b>Group 2</b>	<b>Group 3</b>
Jim Bonham	Viktor Kozich	Leo Spaapen
Austria Cyprus Denmark Estonia Luxembourg Poland Slovenia Spain UK	Croatia Czech Republic Finland France Germany Ireland Latvia Malta Portugal	Belgium Greece Hungary Italy Lithuania Netherlands Slovakia Sweden
Lecture hall	Seminar room	Seminar room

# Groups for ERNDIM National Representatives meeting

Chairmen: Jim Bonham, Viktor Kozich, Leo Spaapen  
Brian Fowler and Mick Henderson will move between groups.

<b>Group A</b>	<b>Group B</b>	<b>Group C</b>
Jim Bonham	Viktor Kozich	Leo Spaapen
Belgium France Germany Greece Italy Netherlands Poland Spain UK	Austria Czech Republic Denmark Finland Hungary Portugal Slovakia Sweden	Croatia Cyprus Estonia Ireland Latvia Lithuania Luxembourg Malta Slovenia
Lecture hall	Seminar room	Seminar room

