

# ERNDIM Workshop SSIEM 2007

Update from the ERNDIM Board

**Brian Fowler, Basel**

# ERNDIM Workshop SSIEM 2007

## Peroxisomal Disorders

- |               |  |                        |
|---------------|--|------------------------|
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# Albert van Gennip

- Major contributor to the establishment of ERNDIM
- "Founder Member" at inaugural meeting in Leuven 1992
- Trust Board on official grounding of ERNDIM in 1994.

Leuven Sept 12th 1992

European Research Network for evaluation and improvement of screening, Diagnosis  
and treatment of Inherited Disorders of Metabolism  
(ERNDIM)

Programme

9.30 - 9.50

Introduction **B. François**

Biomedical and health research proposal for a  
concerted action (BIOMEDI) and ERNDIM.

- Improvement of the diagnostic management of Inherited Disorders of Metabolism  
(IDM)

9.50 - 10.05

Evaluation of the efficiency of existing  
screening programs in Europe

**K. Widhalm, Vienna**

10.05 - 10.20

Development of a quality control system

**A. Van Gennip, Amsterdam**

10.20 - 10.35

Establishment of a database of IDM

# ERNDIM

*European Research Network for evaluation and improvement of screening, diagnosis and treatment of Inherited Disorders of Metabolism.*



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Meeting of the "ERNDIM-QAP panel", Manchester, UK,  
September 7<sup>th</sup>, 1993.

## REPORT

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

- The minutes of the last meeting in Diepenbeek, dated  
May 7<sup>th</sup>, 1993 are approved.

# Albert van Gennip

- Major contributor to the establishment of ERNDIM
- "Founder Member" at inaugural meeting in Leuven 1992
- Trust Board on official grounding of ERNDIM in 1994.
- From 1996 special role of liaising between ERNDIM and the Dutch based SKZL
- Executive Committee - 1997 to 2005
- Scientific Advisory Board - 2000 to 2005.
- Aimed for perfectionism of diagnostic efficiency
- Strove to improve training of our successors, an inspiring teacher and motivator of colleagues especially younger workers in our field
- His engagement and support will be greatly missed

# Albert van Gennip



# Future Perspectives of ERNDIM

- Accreditation of QC schemes
- Establishment of Performance Assessment
- New Schemes (integration of new analytes into existing schemes !!)
- Removal of Financial Risks
- Directory of Laboratories
- Eurogentest
- Training of Biochemists
- Replace Executive Committee  
(younger more energetic people needed)



# Accreditation of QC schemes

- Accreditation of individual schemes – little progress
- Accreditation of ERNDIM itself as umbrella organisation
- Feasible, agreed as aim by SAB and Exec. Comm.
- Action plan to be prepared for next SAB (November 2)

# Establishment of Performance Assessment

## System based on Web site for Scientific Advisors

- Display of annual performance for each lab
- Each analyte scored
- Ranking within all labs
- Unsatisfactory performance for each analyte flagged
- total flags = overall performance

Tested in the SAB for 2006 - all samples in AA, OA, Special Assays, P&P, WC Cystine schemes

## Decision

Apply from 2006

# Establishment of Performance Assessment

## Certificate to show

### Quantitative schemes

- No. compounds in the scheme
- No. for which results were reported
- No. with satisfactory performance
- No. with unsatisfactory performance
- No. with inadequate number of returns
- \* further detail - SEE annual report
- \* any warning letters also noted on certificate

### Special assays

- Satisfactory/unsatisfactory performance listed for each analyte
- Non-returns not be recorded

# Existing Schemes

- **Diagnostic Proficiency Schemes**

- Plans for web-site entry, processing and reporting of results (CSCQ) ? 2008
- centralised storage and shipping of samples (CSCQ)
- Sample archivist (J. Bonham)

# ERNDIM – PATIENT SAMPLE ARCHIVIST

One of the strengths of the qualitative ERNDIM schemes, such as the five regionally organised DPT schemes and the qualitative urinary organic scheme, is the opportunity for participants to encounter interesting and challenging samples. The best way of achieving this is to use **genuine patient samples** but as membership of these schemes continues to grow, supplies of suitable samples are becoming increasingly difficult to maintain.

## Proposal

While most of us are well intentioned when new diagnoses are made, it is easy to overlook the need to arrange sample collection for EQA purposes. One solution may be to identify a **single co-ordinator** to pro-actively remind all participants about the value of patient samples for EQA. In addition, this co-ordinator would act as a well advertised single point of contact ensuring that it was easy and straightforward for people to submit samples to ERNDIM that they had obtained.

## The person appointed

We are looking for a **well motivated** and enthusiastic **individual** to co-ordinate this activity, regularly advertise the need for samples by email by letter and at meetings, then receive, store and catalogue the samples sent. ERNDIM could contribute toward the cost of storage and reimburse transport costs for participants who agreed to submit samples.

This is an enormously important task, which will involve developing and maintaining contacts with laboratory staff, paediatricians and possibly patient groups throughout Europe. The person willing to undertake these tasks will have to ensure that all samples obtained are properly consented and that a record of this consent is stored.

The catalogue and sampled stored would be an extremely valuable resource for ERNDIM.

If you are **interested** in undertaking this role for a period of three to six years, please **contact Dr Jim Bonham**, [jim.bonham@sch.nhs.uk](mailto:jim.bonham@sch.nhs.uk) at Sheffield, UK

# Existing Schemes

- **Diagnostic Proficiency Schemes**

- Plans for web-site entry, processing and reporting of results (CSCQ)
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- **Amino Acids**

- Glutamine problem
- Questionnaire for participants

- **Other Schemes**

- any questions ?

# New Schemes

- **Lysosomal Enzymes** (O. van Diggelen)

Continues as pilot until 2009

- **Congenital disorders of Glycosylation** (R. Wevers)

Present scheme to be taken over as ERNDIM pilot  
2008, ? Full scheme 2009 (questionnaire ? Interest)

- **Under Discussion**

MPS

CSF analyses including neurotransmitters  
(questionnaire ? Interest)

Ceramide trihexoside in urine (Fabry)

# Removal of Financial Risks

Thanks to Melanie Downing and Malcolm Heron

Aim is Neutral Budget

	2005	2006
Total Income	140'065	162'065
Everybody must pay (and they will)		
Payment to scheme organisers		
Administration/Comm (Travel Grant)	139'917	140'931 (677)
Surplus	9'099	21'134
Reserves		121'500

No price increase  
for 2007



# Directory of Laboratories

- **Number labs** 117 with fully verified information
- **Number Analytes** 474
- **Identification remaining European labs**
  - contact with National Representatives
  - Orphanet
- **Extension of Information on labs**
  - accreditation status, harmonisation with other EUGT lab directory

# Eurogentest

- **Harmonisation with Cyto- Molecular genetics**

Continues (accreditation of labs, schemes and ERNDIM itself)

New sub-group of the European Society for Human Genetics for Quality issues with full representation of ERNDIM

- **Expansion and improvement of Biochemical Genetic Testing in Europe**

Report of present state and proposed measures for improvement (ERNDIM website – Meetings and Reports "Best Practice EUGT UPDATED")

- **Development of Guidelines for Methodology**

Organic acids, MPS, Purines / Pyrimidines  
Lysosomal Enzymes, Acyl Carnitines

# Training of Biochemists

- **Training resources on web-site** (M. Henderson)

<http://www.metbio.net>

training in various countries

- **Exploratory talks with SSIEM**

Training workshops

**EUGT workshops for BGT labs**

regional

# ERNDIM National Representatives

Country	Name	Country	Name
Austria	Olaf Bodamer	Belgium	(Baudouin François)
Czech	Viktor Kozich	Cyprus	Anthi Drousiotou
Denmark	Ernst Christensen	Estonia	Katrin Ounap
Finland	Kari Pulkki	Hungary	Agnes Schuler
France	Christine Vaney-Saban	Ireland	Philip Mayne
Germany	<b>Claus-Dieter Langhans</b>	Latvia	Parsla Vevere
Greece	Helen Michelakakis	Lithuania	Jurgita Songailiene
Italy	Ubaldo Caruso	Luxembourg	Jean-Paul Hoffmann
Netherlands	Ries Duran	Malta	Isabella Borg
Portugal	M.I. Tavares de Almeida	Poland	Ewa Pronicka
Spain	Antonia Ribes	Slovakia	Darina Behulova
Sweden	Elisabeth Holme	Slovenia	M. Zerjav-Tansek
Switzerland	Brian Fowler	Bulgaria	Ivo Kremensky
United Kingdom	James Bonham	Croatia	Ksenija Fumic
		Rumania	?

# Executive Committee Members

**Treasurer** Ann Brown 2007

**Schemes Representative, Chair of SAB**

Ries Duran (Albert van Gennip) 2006 Stands  
down

**Secretary** Christine Saban (Ernst Christensen) 2007

**Chairman** replacement in mind for 2008-9

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