

# ERNDIM Workshop SSIEM 2005

## Purine/Pyrimidine disorders

- 11.20 – 11.30 Introduction: ERNDIM Update  
**B. Fowler**
- 11.30 – 11.50 Clinical presentation of pyrimidine defects  
B. Assmann (Düsseldorf)
- 11.50 – 12.10 Clinical presentation of purine defects  
**F.G. Bowling** (Brisbane)
- 12.10 – 12.30 Analysis of purines and pyrimidines  
**J. Duley** (Brisbane)
- 12.30 – 13.00 Results of the purine/pyrimidine proficiency scheme  
**Scientific Advisory Board**

# ERNDIM Workshop SSIEM 2005

## Update on ERNDIM Activities

- Established Schemes
  - Participation / Samples
  - Level of performance
  - Performance assessment
- Pilot & New Schemes
- Directory of Laboratories
- Eurogentest Project
- Education of biochemists
- Future / Officers

# Update on ERNDIM Activities

- Established Schemes

- Participation – Slight increase in each scheme  
DPT now 92 = need for 5th. centre
- Continual review of analytes (concentrations)
- Reference materials (Consensus values NOT calibrators)

- Samples

- We are short of samples for
- Diagnostic Prof. Schemes
  - Qual. Organic acids
  - Acyl Carnitines



# Current Established Schemes

## Number of Scheme Participants

	2004	2005
Quantitative amino acids	171	177
Special Assays in Urine	119	121
Special Assays in Serum	155	158
Quant. Organic Acids in Urine	60	63
Quant. Purines and Pyrimidines	44	45
Qualitative Organic Acids	131	137
Proficiency Scheme (4 Centres)	85	92
Acylcarnitines	60	67
Cystine in WBCs	pilot	27

# Established Schemes

## Level of performance

Improvement still necessary / Link to accreditation

## Scoring and Assessment of Performance

### Aim is

- Certificate based achievement of satisfactory (good) performance
- Warning letter for unsatisfactory performance (including no return of results)
- ~~- Certificate of participation to include No warning letters~~

### Statement on all certificates

"In the event of poor performance, warning letters are issued to individual participants by the scheme organisers"

# Scoring and Assessment of Performance

## Proficiency testing schemes

- harmonised scoring
  - (good performance = 75% ???)
  - poor performance = less than 15/30 points

## Quantitative Schemes

### Web site for Scientific Advisors

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

### System under evaluation

Warning letters for 2005 performance after discussion in SAB

# New Schemes

## Neurotransmitters etc. in CSF (N. Abeling)

- Evaluation of potential participant number and metabolites

## Lysosomal Enzymes (O. van Diggelen)

- Evaluation of matrix, which enzymes, preliminary study with few labs. Etc.

Steroids ?? NO

# 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> Wechseln zu Links >>

**ERNDIM-SSIEM**  
Diagnostic Laboratories Database

Search

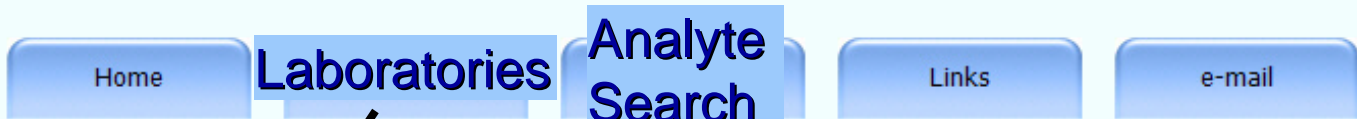
[New Search](#)  
[Last Results](#)

[Back to Options](#)

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

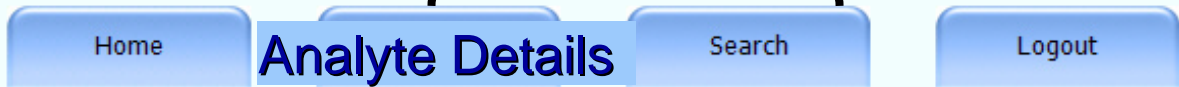
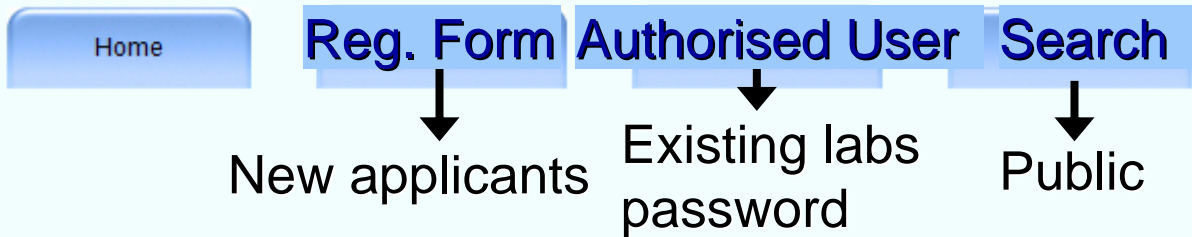
Switzerland	Switzerland, Basel, University Childrens Hospital	Switzerland	<a href="#">More info</a>
Outside Switzerland	Netherlands, Rotterdam, Erasmus University MCg,	Netherlands	<a href="#">More info</a>
	UK, Belfast, Belfast Link Labs	Northern Ireland	<a href="#">More info</a>
	UK, Peterborough, Peterborough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





List by country

Automatic updating online with verification



Update by labs + vetting

# EU Project: Eurogentest

**Title:** Genetic testing in Europe - Network for test development harmonization, validation and standardization of services

[www.eurogentest.org](http://www.eurogentest.org)

## Main activities

ERNDIM directory of IEM laboratories

Meetings with partners from other Genetic disciplines

Selection of National representatives from each EU25 country

Workshop for all in Basel December 2, 2005

Best practice meetings: eg. performance assessment, recommended methods, linking internal QA / external QA & accreditation (mainly SAB)

**MANY REPORTS TO WRITE**

# Future Perspectives

- 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)

# Future Perspectives

- Accreditation of QC schemes
- Establishment of Performance Assessment
- New Schemes (integration of new analytes into existing schemes !!)
- Removal of Financial Risks

- From Sept 2005

## Treasurer

Jim Bonham - stands down

Melanie Downing - takes over

## Executive Committee Representative of SKZ

Albert van Gennip - Stands down

Ries Duran – takes over



# Eurogentest Project: ERNDIM Activity

ERNDIM responsible for Work package “Organize and harmonize EQA for biochemical Testing”

**Objectives** 1) To expand opportunities for Biochemical Genetic testing laboratories in EU25 to participate in EQA.

(2) To link Internal Quality Control and EQA through consensus discussions on best practice.

## Description of work

(1) To research the current situation in EU25 including needs and deficits.

(2) A program of meetings to discuss best practice and agree consensus guidelines in Biochemical Genetic practice.

(3) To instigate the links between internal quality control, EQA and accreditation.

(4) To explore standard methodologies and standard reporting procedures.

(5) Co-ordination with other work packages through correspondence and meetings.