

ERNDIM /EUGT meeting October 5 and 6, 2006,Prague

The Role of QA in the diagnosis and dietary management of IEM The European perspective

Programme

Thursday October 5

9:00 – 12:30 Session 1

- 9:00 Opening
Brian Fowler (Basel)
Prof. Tomas Zima (Dean, First Faculty of Med.-Charles Univ.)
- 9:10 Special Guest Lecture: Diagnosis of Lysosomal storage diseases
Milan Elleder (Prague)
- 9:55 Results obtained with samples from Lysosomal storage diseases in Diagnostic Proficiency Testing
DPT scheme organisers
- 10:10 Lysosomal enzymes pilot EQA scheme
Otto van Diggelen (Rotterdam)

10:30 Coffee

- 11:00 Eurogentest: activities: Overview
Brian Fowler (Basel)
- 11:15 Perspective of Cytogenetics
Rod Howell (Oxford)
- 11:35 Eurogentest NoE: towards analytical and diagnostic validation
Milan Macek (Prague)

12:00 Lunch

ERNDIM Board meeting

14:15 – 18:00 Session 2

Separate meetings of participants of the 5 DPT schemes.

Chairs: Marius Duran, DPT-Amsterdam
Brian Fowler, DPT-Basel
Christine Vianey-Saban, DPT-Lyon
Viktor Kozich, DPT- Prague
Jim Bonham, DPT-Sheffield

14:15 **Discussions and reports on 2006 samples.**

16:00 Coffee

- 16:30 **Separate meetings contd.:** needs and deficits in Biochemical Genetic Testing in Europe (including issues arising from last years EU25 best practice meeting)
These meetings will be joined by none-DPT participants according to allocation
- 18:00 Close

Friday October 6

8:30 – 12:00 Session 3

8:30 Presentation and discussion of the DPT common samples for 2005, 2006 **in plenum**

9:00 ERNDIM EQA Schemes: Issues and Questions

- Urinary Qualitative Organic acid analyses: Analytical approaches and Performance
Jim Bonham (Sheffield)

- Purines and Pyrimidine EQA: specific issues and lessons for other schemes
Jörgen Bierau (Maastricht)

- Approaches to Reporting

Jim Bonham (Sheffield)

- ERNDIM EQA Schemes: Questions and Answers

Scientific Advisors

10:00 Coffee

10:30 Explanation for scoring in assessment of performance of quantitative schemes
Cas Weykamp (Winterswijk)

11:00 Debate on "critical aminoacids" in relation to diagnosis and management of treatment

Chair: Jim Bonham (Sheffield)

12:00 Lunch

13:30 – 15:00 Session 4

13:30 **Short communications – novel disorders and potential pitfalls in diagnosis**

1. Aminoacylase 1 deficiency

J.O. Sass (Freiburg)

2. MMA epimerase deficiency

M. Duran (Amsterdam)

3. alpha-amino adipic semialdehyde and pyridoxine responsive fits

E. Struys (Amsterdam)

4. SCHAD-deficiency + 3-OH-isobutyryl-CoA hydrolase deficiency and the value of OH-C4-carnitine

S. Krywawych (London)

14:30 Summary of the DPT participant meetings on needs and deficits in Biochemical Genetic Testing in Europe
Future Challenges (Eurogentest, Best Practice)

Brian Fowler (Basel)

15:00 **Close of meeting**

Organized under the auspices of the Dean of the First Faculty of Medicine of the Charles University, Prague

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