

## **ERNDIM /EUGT meeting October 5 and 6, 2006 in Prague**

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

**Thursday October 5**

#### **9:00 – 12:30 Session 1**

- 9:00 Special Guest Lecture: Diagnosis of Lysosomal storage diseases  
*Milan Elleder (Prague)*
- 9:45 Results obtained with samples from Lysosomal storage diseases in Diagnostic Proficiency Testing  
*DPT scheme organisers*
- 10:00 Lysosomal enzymes pilot EQA scheme  
*Otto van Diggelen (Rotterdam)*
- 10:15 Discussion

#### **10:30 Coffee**

- 11:00 Eurogentest: activities  
Perspective of Molecular Genetics and Cytogenetics

#### **12:30 Lunch**

#### **ERNDIM Board meeting**

#### **14:30 – 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

14:30 **Discussions and reports on 2006 samples.**

#### **16:00 Coffee**

16:30 **Discussions on needs and deficits in Biochemical Genetic Testing in Europe**  
(including issues arising from last years EU25 best practice meeting)

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 4 case presentations of nearly or actually missed diagnoses with recommendations by Scientific Advisors of the relevant EQA schemes

**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  
clinician  
Chair: *Jim Bonham (Sheffield)*

**12:00 Lunch**

**13:30 – 15:00 Session 4**

13:30 **Short communications – novel disorders and the 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

15:00 **Close of meeting**