

Welcome to Basel

ERDIM Scientific and DPT  
meetings

October 22-23, 2009



San Diego





# **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)*

## ERNDIM Meeting, October 22 – 23, 2009

### Programme

#### DAY 1: Thursday, 22.10.2009

Session 1: Diagnostic approaches		
09.00 – 09.10	Opening	Brian Fowler, Basel
09.10 – 09.50	Clinical approach to Diagnosis of Lysosomal Storage Diseases	Marianne Rohrbach, Zürich
09.50 – 10.10	ERNDIM EQA scheme for Lysosomal Enzymes	O. van Diggelen, Rotterdam
10.10 – 10.30	Coffee	
10.30 – 11.30	Diagnosis of Congenital disorders of Glycosylation ERNDIM EQA scheme for CDG	Dirk Lefeber, Nijmegen
11:30 – 12.20	Clinical presentation and diagnostic difficulties of Glycogen storage diseases	Matthias Baumgartner, Zürich

### Session 2: Future Developments in Genetic Testing / EQA

14.15 – 14.45	ERNDIM and the Eurogentest Project	Brian Fowler Basel
14.45 – 15.15	EMQN and EQA in molecular genetic testing	David Barton, Dublin
15.15 – 15.45	Homozygosity mapping as a primary diagnostic tool: LIPN1-mutations as the major cause of recurrent rhabdomyolysis	Orly Elpeleg, Jerusalem

15.45 – 16.30	<b>Coffee and Poster session</b>
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### Session 3: Novel Disorders and Biochemical Diagnosis

16.30 – 17.00	State of art of intracellular cobalamin defects: CbID, CbIF	Brian Fowler Basel
17.00– 17.30	A final solution to the ethylmalonic acid syndrome	Valeria Tiranti, Milan
17.30 – 18.00	The application of UHPLC-separation of acylcarnitines prior to MS/MS detection	Wim Kulik, AMC, Amsterdam

Evening	<b>Dinner restaurant Rialto</b>
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On leaving  
this building  
turn left and  
you see  
"Rialto"

**DAY 2: Friday, 23.10.2009**

09.00 – 10.30	<b>Separate DPT meetings</b> DPT Sheffield DPT Basel DPT Lyon DPT Prague DPT Amsterdam	first floor: 1A 232 second floor: 2B 241 second floor: 2B 242 second floor: 2B 233 second floor: 2B 105	<b>Chairs:</b> Jim Bonham Brian Fowler Christine Vianey-Saban Viktor Kozich Ries Duran
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10.30 – 11.00	Coffee and Poster session
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<b>Common ERNDIM meeting</b>		
11.00 – 11.15	Chairman's update	Mick Henderson, Leeds
11.15 – 11.30	Common DPT sample	Brian Fowler Basel
11.30 – 12.00	Neurotransmitter disorders: clinical presentation, when to test	Nenad Blau, Zürich
12.30 – 13.00	Analysis of neurotransmitters	Simon Heales, London

# Sponsors

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## Name Badges

Please write your  
name yourself

## ERNDIM meeting Registration

Please do not forget to pay any outstanding fees on  
Wednesday (exact amount, Katharina is not a  
banker)

## Participation Certificates

## Dinner