ERNDIM 2025 SYMPOSIUM

ERNDiM

Thursday 9th
Friday 10th
October 2025
Madrid SPAIN
Crowne Plaza Madrid
Centre Retiro

THURSDAY, 9TH OCTOBER

12:00 – 13:25 **Registration, and Lunch**

13:25 – 13:30 **Welcome**

Rafa Artuch, Spain

13:30 – 15:30 **Session 1, Lysosomal metabolism & Lipids**

Session chair: George Ruijter

13:30-14:00 **Cell trafficking and autophagy in neurometabolic disorders**
Alfonso de Oyarzabal, Spain

14:00-14:30 **Application of lipidomics to enhance targeted diagnostic lipid panels**
Marie van Dijk, The Netherlands

14:30-15:00 **History and updates on the use of Biomarkers in lysosomal diseases: FABRY, GAUCHER, NPC, ASMD**
Sara Boenzi, Italy

15:00-15:30 **Analysis of Endogenous-NRE GAG Biomarkers Provides the Most Reliable Method for Reducing False Positives in Newborn Screening for MPS Disorders and for Callibration of Values Across Multiple Reference Laboratories**
Michael Gelb, USA

15:30 – 16:00 **Coffee Break**

16:00 – 17:20 **Oral abstracts. New diagnostic methods/new metabolites**

Session chairs - Déborah Mathis and Petr Chrastina

***Feasibility of -omics technologies on platelets as rapid high-throughput second-tier diagnostic test for OXPHOS diseases**. Bram Decru, Belgium.*

***Determining cut-off values to increase selectivity and specificity in X-ALD testing within at-risk populations**. Ewa Wielogórska, Poland.*

***KS-462-282: An alternative Krabbe Disease biomarker with excellent diagnostic potential**. James Cooper, UK.*

***Adaptation of a UPLC-MS/MS Method for the Analysis of Glycosaminoglycan Fragments for the Diagnosis of Mucopolysaccharidosis in a UK Clinical Laboratory**. Kate Neal, UK.*

***The effect of maternal methyl-DOPA and other exo- and endogenous interfering substances on the results of 3-O-methyl-DOPA determinations and therefore, the screening for AADC deficiency**. Malgorzata Rogozinska, Poland.*

***Urinary GAGs quantification by a simplified UPLC-MS/MS assay for mucopolysaccharidoses diagnosis and monitoring**. Marianna Caterino, Italy.*

17:20 – 17:30 **Break**

17:30 – 19:00 **Diagnostic Proficiency Testing Workshops (Parallel sessions)**

DPT Czech Republic
Petr Chrastina, Czech Republic

DPT France
Christine Vianey-Saban, France

DPT Switzerland
Déborah Mathis, Switzerland

DPT The Netherlands
George Ruijter, The Netherlands

DPT United Kingdom
Joanne Croft, United Kingdom

20:00 **Networking Dinner**

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DAY 2 – FRIDAY, 10TH OCTOBER

09:00 – 09:55 **The role of ERNDIM in current and future EQAs.** Session chair - Rafa Artuch

09:00-09:10 **Chair's update -** Rafa Artuch, Spain

09:10-09:25 **Results of the common sample 2025 (DPT-F) -** Christine Vianey Saban, France

09:25-09:40 **Introduction to the Lipids in Serum (LIS) Scheme -** Susan Goorden, The Netherlands

09:40-09:55 **Overview of critical errors, including definitions and example cases -** George Ruijter, The Netherlands

10:00 – 11:00 **ERNDIM Workshops (Parallel sessions)**

Qualitative Organic Acids
Camilla Scott, United Kingdom and Judit García Villoria, Spain

Lysosomal enzymes and Urine Mucopolysaccharides
Chair: Berthil Prinsen, The Netherlands
New insights into diagnosing lysosomal storage disorders using dried blood spot MS/MS testing
Magali Pettazoni, France
Lysosomal enzymes in fibroblasts (LEFB) and in dried blot spots (LEDB) schemes: present and future
Ed Jacobs, The Netherlands
Improving detection of oligosaccharidoses: advanced diagnostics with mass spectrometry
Marne Hagemeijer, The Netherlands

Amino Acids Interpretations
Sabine Scholl-Bürgi, Austria and Apolline Imbard, France

11:00 – 11:30 **Coffee Break**

11:30 – 13:15 **Session 2 Rare and Interesting cases**
Session chairs - Camilla Scott and Pedro Ruiz Sala

11:30-12:05 **Rare/interesting cases -** Cristiano Rizzo, Italy

12:05-13:15 **Oral abstracts Challenging cases**

Dihydrolipoamide Dehydrogenase (E3 Enzyme) Deficiency Manifesting as Acute Liver Failure: Case Series of Two Patients*. Elena Dumin, Israel.*

Maternal glutaric aciduria type-1 detected by newborn screening. Ewa Glab- Jablonska, Poland.

Diagnostic Challenges in Non-Classical Forms of Maple Syrup Urine Disease in newborn screening*. Mariola Sasin-Rokita, Poland.*

Study of a Patient with Symptoms Compatible with Niemann-Pick Disease Type C but with Intermediate Levels of Lysosphingomyelin-509 and a Genetic Carrier of NPC1*. Mercedes del Valle Sanz, Spain.*

Laboratory diagnosis of a case with sodium-dependent citrate transporter deficiency*. Noelia Muñoz-Puga, Spain.*

Implication of neurotransmitter analysis on the diagnosis of SCN2A-Related disorder: Diagnostic and therapeutic considerations*. Pablo Martínez, Spain.*

13:15 – 13:30 **Closing remarks**

13:30 **Lunch**

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