



QUALITY ASSURANCE IN LABORATORY TESTING FOR IEM

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ERNDIM 2021 SYMPOSIUM PROVISIONAL SCIENTIFIC PROGRAMME

The 2021 ERNDIM SYMPOSIUM will take place on:

Dates: Thursday 21st and Friday 22nd October 2021

Time: see programme below for details, all times in CET

Venue: Online

Organising committee:

Nenad Blau – Zurich
Carlo Dionisi-Vici – Rome
Cristiano Rizzo – Rome
George Ruijter – Rotterdam
Christine Vianey-Saban – Lyon

Please note that titles of presentations in this provisional programme may be subject to change, however the topics will remain as listed below.

Day 1:

- 10:50-11:00** **Welcome 1st day – George Ruijter**
- 11:00-12:50** **Vitamin B disorders**
- Overview on vitamin B disorders: Carlo Dionisi-Vici (Rome)
 - Inborn errors of thiamine metabolism: Rafael Artuch (Barcelona)
 - How to diagnose inborn errors of riboflavin metabolism?: Christine Vianey-Saban (Lyon)
 - LC-MS/MS for the differential diagnosis of treatable early onset epilepsy: Déborah Mathis (Bern)
 - Diagnosis of inherited disorders of folate metabolism: Nenad Blau (Zurich)
 - New insights in the metabolism of vitamin B12: Luciana Hannibal (Freiburg)
 - Discussion
- 12:50-13:20** **Sponsored symposium – Sciex title TBC**
- 13:20-14:20** **Break**
- 14:20-14:50** **Sponsored symposium – Shimadzu title TBC**
- 14:50-15:40** **ERNDIM schemes - present and future**
- Chair's update including potential new schemes: George Ruijter (Rotterdam)
 - Results common DPT sample 2021: George Ruijter
 - Limitation of metabolites measurement in DBS & potential alternatives: Rachel Carling (London)
 - Discussion
- End session**

Important note: The complete history of this document including its author, authoriser(s) and revision date, can be found on Q-Pulse

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ERNDIM (European Research Network for evaluation and improvement of screening,
Diagnosis and treatment of Inherited disorders of Metabolism

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Email: admin@erndim.orgWeb: www.erndim.org**Day 2:**

- 10:45-11:45 Workshops [parallel sessions]**
- Pterins: Nenad Blau (Zurich), Claudia Carducci (Rome), Alessio Cremonesi (Zurich)
 - Acylcarnitines: Cristiano Rizzo (Rome), Pedro Ruiz Sala (Madrid), Charles Turner (London)
 - Organic acids: Judit Garcia Villoria (Barcelona), Clothilde Roux (Lausanne), Camilla Scot (Sheffield)
 - Amino acids: Rachel Carling (London), Brian Fowler (Basel), Sabine Scholl-Burgi (Innsbruck)
- 11:45-11:50 Break for connection to plenary session**
- 11:50-12:40 New biomarkers detected by mass spectrometry**
- Application to newborn screening and laboratory medicine: Giancarlo La Marca (Florence)
 - Amadori rearrangement products as novel biomarkers for inborn errors of amino acid metabolism: Karlien Coene (Nijmegen)
 - Discussion
- 12:40-13:10 Sponsored symposium – PTC title TBC**
- 13:10-13:55 Break**
- 13:55-16:00 Omics, Big data and IEM**
- Targeted and untargeted metabolomics and lipidomics, including lysosphingolipids
 - Positioning untargeted metabolomics in the diagnostic workup of inborn metabolic disorders: Judith Jans (Utrecht)
 - Targeted lipidomics, experience from an IEM diagnostic lab: Giulio Catesini (Rome)
 - Untargeted lipidomics: Fred Vaz (Amsterdam)
 - Discussion
- Data processing for untargeted metabolomics: Pietro Franceschi (San Michele all'Adige)
- Bridging the gap between a negative exome and diagnosis for personalized medicine: Clara van Karnebeek (Nijmegen)
 - Discussion
- 16.00-16:05 Closing remarks – George Ruijter (Rotterdam)**

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