

ERNDIM Administration Office

c/o EMQN CIC Office, Unit 4, Enterprise House, Manchester Science Park,
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ERNDIM 2021 SYMPOSIUM 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 CEST

Venue: Online

Organising committee:

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

Day 1:

- 10:50-11:00** **Welcome 1st day – George Ruijter**
- 11:00-12:50** **Vitamin B disorders** *Chaired by Alessio Cremonesi & Brian Fowler*
- Overview on vitamin B disorders: Carlo Dionisi-Vici (Rome)
 - Acquired and genetic defects related to thiamine transport and metabolism: Rafa Artuch (Barcelona)
 - How to diagnose inborn errors of riboflavin metabolism?: Christine Vianey-Saban (Lyon)
 - LC-MS/MS method for the differential diagnosis of treatable early onset epilepsy: Déborah Mathis (Bern)
 - Diagnosis of inherited disorders of folate metabolism: Nenad Blau (Zürich)
 - New insights in the metabolism of vitamin B₁₂: Luciana Hannibal (Freiburg)
 - Discussion
- 12:50-13:20** **Sponsored symposium – Sciex** Advancements in metabolomics and lipidomics with the ZenoTOF 7600 system: Mário Armelão
- 13:20-14:20** **Break**
- 14:20-14:50** **Sponsored symposium – Shimadzu** Hemoglobinopathies Newborn Screening by LCMS-8050 using ZenTech Targeted MS/MS Hemo device: Madeleine Boulanger (Liege)
- 14:50-15:40** **ERNDIM schemes - present & future** *Chaired by Judit Garcia-Villoria & Christine Vianey Saban*
- Chair's update including potential new schemes: George Ruijter (Rotterdam)
 - Results common DPT sample 2021: George Ruijter
 - Limitation of metabolite measurement in DBS: Rachel Carling (London)
 - Discussion
- End session**

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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 Scott (Sheffield)
- Amino acids: Rachel Carling (London), Brian Fowler (Basel), Sabine Scholl-Burgi (Innsbruck), Maryanne Preece (Birmingham)

11:45-11:50 Break for connection to plenary session**11:50-12:40 New biomarkers detected by mass spectrometry Chaired by Rafael Artuch & Déborah Mathis**

- 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 Understanding the diagnostic tests to identify aromatic L-amino acid decarboxylase (AADC) deficiency: Simon Heales (London) & Alberto Burlina (Padua)**13:10-13:55 Break****13:55-16:00 Omics, Big data and IEM Chaired by Nenad Blau & Cristiano Rizzo**

- 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)
- Disorders of complex lipid metabolism, application of 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, towards personalized medicine: Clara van Karnebeek (Nijmegen)
- Discussion

16:00-16:05 Closing remarks – George Ruijter (Rotterdam)

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Faculty:

<u>Name</u>	<u>Affiliation</u>
Dr Rafa Artuch	Hospital Sant Joan de Déu, Barcelona, Spain
Prof Nenad Blau	ERNDIM Scientific Advisor Children's Hospital Zürich, Switzerland
Dr Claudia Carducci	ERNDIM deputy Scientific Advisor Università di Roma Sapienza, Rome, Italy
Dr Rachel Carling	Guy's & St Thomas' Hospital, London, United Kingdom
Giulio Catesini	ERNDIM Scientific Advisor Bambino Gesù Children Hospital, Rome, Italy
Dr Karlien Coene	Radboud UMC Nijmegen, The Netherlands
Dr Alessio Cremonesi	Children's Hospital Zürich, Switzerland
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Prof Brian Fowler	Children's Hospital Basel and Zürich, Switzerland
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Dr Deborah Mathis	Inselspital, University Hospital Bern, Switzerland
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Mme Chlothilde Roux	ERNDIM Scientific Advisor CHUV Lausanne, Switzerland
Dr George Ruijter	ERNDIM Scientific Advisor Erasmus MC, Rotterdam, The Netherlands
Dr Pedro Ruiz Sala	Chair ERNDIM Executive Committee and Scientific Advisor Universidad Autónoma de Madrid, Spain
Dr Sabine Scholl-Burgi	ERNDIM Scientific Advisor Medical University of Innsbruck, Austria
Mrs Camilla Scott	ERNDIM Scientific Advisor Sheffield Children's Hospital, United Kingdom
Dr Charles Turner	ERNDIM Scientific Advisor Evelina Children's Hospital, London, United Kingdom
Prof Clara van Karnebeek	ERNDIM Scientific Advisor Radboud UMC, Nijmegen & Amsterdam UMC, The Netherlands
Fred Vaz	Amsterdam UMC, The Netherlands
Dr Christine Vianey Saban	CHU de Lyon, France
	Chair ERNDIM Scientific Advisory Board and Scientific Advisor

Sponsored symposium:

<u>Sponsor</u>	<u>Name</u>	<u>Affiliation</u>
Sciex	Mário Armelão	Advanced workflow specialist, SCIEX
Shimadzu	Dr Madeleine Boulanger	R&D Scientist, ZENTECH
PTC	Prof Alberto Burlina	Great Ormond St Hospital, London, United Kingdom
	Prof Simon Heales	University of Padua, Padua, Italy

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ERNDIM (European Research Network for evaluation and improvement of screening,
Diagnosis and treatment of Inherited disorders of Metabolism
Version 3

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