SSIEM Virtual Symposium Organizer

Society for the Study of Inborn Errors of Metabolism (SSIEM)

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For registration and more information please visit: ssiemvirtual.org
Welcome

Dear SSIEM Members, Colleagues and Friends,

Welcome to the first SSIEM 2020 Virtual Symposium Day, December 3, 2020! This is an adventure to all of us after having practiced more than half a century of ‘live’ SSIEM annual meetings.

The evolving SARS-CoV-2 (COVID-19) pandemic has not only changed the world tremendously, but also forced us to make the difficult decision to postpone our 53rd Annual Meeting to 2022 in Freiburg. Instead for the current year, SSIEM council members and the editors of our journals (Journal of Inherited Metabolic Disease and JIMD Reports) organised a challenging one day online symposium, to which we welcome you all very much. In addition, we organised our first online annual general meeting to be held in conjunction at 7 pm, GMT to which all SSIEM members are invited.

The aims of this symposium day are to bring together our global community, to foster the study of inherited metabolic disorders, and to showcase research in different metabolic disciplines from all over the world.

We have put together an exciting scientific programme for our online symposium day, including invited lectures, and the Komrower and Garrod lectures. In the late breaking news section, four selected abstracts will be presented as oral presentations. The recorded lectures will remain available for 3 months.

Finally, we would like to encourage attending the ICIEM 2021 in Sydney as well as to announce the upcoming SSIEM Annual Symposia in Freiburg (2022), Jerusalem (2023) and Porto (2024).

Please participate/join, enjoy and profit from the virtual day, and stay healthy!

Thank you very much for joining us “digitally“.

With our best wishes,

On behalf of the Organisation Team,

Gajja Salomons, Chair of SSIEM

Eva Morava, Editor in Chief JIMD / JIMD Reports
SSIEM Scientific Committee

Matthias Baumgartner
Zurich, Switzerland

Anny Brown
Nottingham, United Kingdom

David Cassiman
Leuven, Belgium

Helen Michelakakis
Athens, Greece

Phillipa Mills
London, United Kingdom

Fanny Mochel
Paris, France

Eva Morava
Rochester, United States

Marc Patterson
Rochester, United States

Shamima Rahman
London, United Kingdom

Verena Peters
Heidelberg, Germany

Gajja Salomons
Amsterdam, The Netherlands

Manuel Schiff
Paris, France

Ute Spiekerkötter
Freiburg, Germany

Johannes Zschocke
Innsbruck, Austria
All indicated times are based on Greenwich Mean Time (GMT, London).

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<td>Gaija Salomons (Amsterdam, The Netherlands)</td>
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<td>Sandra Sirrs (Vancouver, Canada), Annalisa Secchi (Udine, Italy)</td>
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<td>Update on complex lipid disorders</td>
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<td>Clinical, radiological, and genetic characteristics in patients with ACO2 gene defects: an emerging neurometabolic syndrome</td>
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<td>Ronen Spiegel (Afula, Israel)</td>
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<td>15.00-15.15</td>
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15.15-17.00 Invited lectures
Chairs: Ute Spiekerkötter (Freiburg, Germany), Fanny Mochel (Paris, France)

Metabolic cutis laxa and The Golgi
Björn Fischer (Berlin, Germany)

The International Classification of Inherited Metabolic Disorders, ICIMD
Carlos Ferreira (Bethesda, United States)
Shamima Rahman (London, United Kingdom)
Johannes Zschocke (Innsbruck, Austria)

News from the FDA: Orphan drug development
Patroula Smpokou (Maryland, United States)

In vivo gene therapy for Metabolic Diseases: state of the art and future perspectives
Nicola Brunetti Pierri (Naples, Italy)

Developing therapy for ultra-rare inherited metabolic disease: lessons from Niemann-Pick disease type C
Marc Patterson (Rochester, United States)

16.45 Q&A

17.00-17.15 Break

17.15-18.00 Komrower Lecture
Introduction: Gajja Salomons (Amsterdam, The Netherlands)

Elucidating the causes of metabolic disorders:
the new veins to explore
Emile van Schaftingen (Leuven, Belgium)
18.00-19.00  Late breaking news

Chair: Manuel Schiff (Paris, France)

An autosomal dominant neurological disorder caused by de novo variants in FAR1 resulting in uncontrolled synthesis of ether lipids
Sacha Ferdinandusse (Amsterdam, The Netherlands)

A novel brain and heart developmental syndrome caused by impairment of the mitochondrial one-carbon metabolism enzyme SHMT2
Aurora Pujol (Barcelona, Spain)

A new neurological disease due to PGM2L1 deficiency leads to defective production of glucose-1,6-bisphosphate, but not to a glycosylation defect
Maria Veiga-da-Cunha (Brussels, Belgium)

GFUS-CDG, a new glycosylation disorder treatable with L-fucose
Saskia Wortmann (Salzburg, Austria)

Q&A

19.00-19.50  SSIEM Annual General Meeting
(all SSIEM Members invited but RSVP)

19.50-20.00  A message from the ICIEM 2021
Closing remarks

20.00-20.30  Industry Sponsored Symposia (p. 13)
Our new virtual home
Nutricia Metabolics Campus

Even when we are miles apart, our commitment to support you with tools and educational content knows no distance. That is why we are excited to welcome you to the Nutricia Metabolics Virtual Campus.

Our campus is an interactive experience, bringing together a range of educational content, patient resources and networking opportunities.

Visit Nutricia Metabolics Campus to find downloadable educational content including podcasts and round table discussions.

Webinar: The Power of Personalized Education - Improving Adherence to Diet and Patient Outcomes in IEMs

Request your Copy

An exclusive printed copy of the updated 5th edition of Zschocke/Hoffmann’s Vademecum Metabolicum providing a systematic and practical approach to the diagnosis and treatment of metabolic disease using promotional code: SSIEM2020*

*Limited copies available, 1 per person

https://metabolics.nutricia-campus.com/
All indicated times are based on Greenwich Mean Time (GMT, London).

12.00-12.30  Sponsored Satellite Symposium 1:
Glycomacropeptide (GMP) and Bone Health
Chair: Peter Freisinger (Reutlingen, Germany)

Speaker: Anne Daly (Birmingham, United Kingdom)

12.00-12.30  Sponsored Satellite Symposium 2:
Long-Chain Fatty Acid Oxidation Disorders: Evolving Best Practices

Welcome and Introduction
Jerry Vockley (Pittsburgh, United States)

LC-FAODs: Is Guideline-Based Management Enough?
Jerry Vockley (Pittsburgh, United States)
Barbara K. Burton (Chicago, United States)
Ute Spiekerkötter (Freiburg, Germany)

What Will Emerging Treatments Mean for LC-FAOD Management?
Jerry Vockley (Pittsburgh, United States)
Barbara K. Burton (Chicago, United States)
Ute Spiekerkötter (Freiburg, Germany)

Concluding Remarks
Jerry Vockley (Pittsburgh, United States)

Patients with long-chain fatty acid oxidation disorders (LC-FAODs) often experience significant metabolic events leading to emergency room visits, hospitalizations and even mortality, all despite following guideline-based dietary interventions. During this interactive roundtable discussion, expert faculty will discuss this challenge, how they address it, as well as how the treatment landscape for LC-FAODs is evolving to enable better outcomes for patients.

A Medscape LIVE symposium supported by an educational grant from Ultragenyx.
PKU sphere™ is Vitaflor’s first GMP-based protein substitute, specifically designed to optimise adherence. PKU sphere is:

- The only GMP-based protein substitute to be evaluated long term in children and teenagers.¹
- Suitable from 4 years of age.
- Available in three flavours - Vanilla, Red Berry and Chocolate.
- Available in two pack sizes - 15g and 20g PE pre-measured sachets.
- Low in volume and designed to support an overall healthy dietary intake by avoiding excess calories and sugar.

Join us for our symposium… on Glycomacropeptide (GMP) and Bone Health at 12-12:30pm (GMT). Presented by guest speaker Anne Daly, Metabolic Dietitian, Birmingham Children’s Hospital, UK and chaired by Prof. Dr. Freisinger, Head of the paediatrics department and the metabolic centre, Klinikum Reutlingen, Germany.

Visit our virtual stand… to find out more about PKU sphere and our comprehensive range of products for Inborn Errors of Metabolism.

PKU (Phenylketonuria), GMP (Glycomacropeptide), PE (Protein Equivalent), Phe (Phenylalanine). PKU sphere™ is a food for special medical purposes and must be used under medical supervision. For the dietary management of PKU. Refer to product labels for allergen and other important information. PKU sphere chocolate contains 2.8g/ 20g PE and 2.2g/ 15g PE. Reference available upon request.

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12.00-12.30
Sponsored Satellite Symposium 3
One year of treatment experience with PALYNZIQ® (pegvaliase) in Europe

Welcome
Ania C. Muntau (Hamburg, Germany)

One year of treatment experience with PALYNZIQ in Europe
Ania C. Muntau (Hamburg, Germany)

Live Q&A
Faculty & audience

12.30-12.45
Break

12.45-13.15
Sponsored Satellite Symposium 4:
Re-defining management of alkaptonuria: Transforming the outlook for patients with the iconic Mendelian disease - a tribute to Archibald Garrod

DevelopAKUre: Disease-modifying pharmacological treatment of AKU with nitisinone – results from an international, randomised controlled Phase 3 study (SONIA 2)
Lakshminarayan Ranganath (Liverpool, United Kingdom)

AKU from the patients’ perspective – Impact of AKU symptoms as perceived by the patients
Nicolas Sireau (Cambridge, United Kingdom)

12.45-13.15
Sponsored Satellite Symposium 5:
Shortening the Journey to Fabry Disease Diagnosis: Perspectives from a Patient and Nephrologist

David G. Warnock (Birmingham, United States)

Paul R.
Fabry Patient (United States)

12.45-13.15
Sponsored Satellite Symposium 6:
Optimizing IEM Care During a Pandemic: A Case-Based Look at UCD Management

A case-based program that reviews key aspects of urea cycle disorders (UCD) and illustrates real-world challenges and strategies for successful management of inborn errors of metabolism during the pandemic.

Speaker:
Gregory Enns (Stanford - Palo Alto, United States)
Lucile Salter (Palo Alto, United States)
YOUR HEALTH, OUR COMMITMENT

We are an Italian pharmaceutical company with over one hundred years of experience in several specialist therapeutic areas where we continue to be active today. We also strive to bring to market treatments for rare and niche diseases, thereby ensuring that patients have access to the pharmaceutical and medical food options they need.

We work closely together with the medical and scientific community, liaising with international partners with high technological expertise so that the best therapeutic solutions may be identified and delivered more quickly.

We promote excellence that benefits patients by standing as a reference point for medical specialists and through presenting state-of-the-art solutions that help foster therapeutic compliance.

Our heart is in Italy, is our head in the world and our eyes are constantly oriented towards the future.
Sponsored Satellite Symposium 7: Distinguishing Pediatric Movement Disorders - Uncovering AADC Deficiency

Welcome and introductions
Sunay Ozdas

Distinguishing AADC deficiency: Clinical experience with differential diagnosis
Berrin Monteleone

Current management strategies and emerging therapies for AADC deficiency
Wuh-Liang Hwu

Q&A panel discussion
Sunay Ozdas
Berrin Monteleone

Symposium Objectives:
- Review pediatric disorders that may clinically resemble AADC deficiency
- Share clinical experience with differential diagnosis of AADC deficiency
- Discuss current management options and treatments in development for AADC deficiency

Sponsored Satellite Symposium 8: Fabry phenotypes: GLA variants in the real world

Welcome & introduction
Jörn Schenk (Zurich, Switzerland)

What can GLA variants tell us and what can’t they?
Gheona Altarescu (Jerusalem, Israel)

Working with phenotypic variability
Derralynn Hughes (London, United Kingdom)

Live Q&A
Aromatic-L-amino acid decarboxylase (AADC) deficiency is an inborn error of neurotransmitter biosynthesis. It is an autosomal recessive disorder caused by pathogenic variants in the dopa decarboxylase gene, DDC, encoding for the AADC enzyme. In patients with AADC deficiency, lack of AADC enzyme leads to accumulation of its substrate L-DOPA.

What is AADC deficiency? What is 3-OMD?

3-O-methyldopa (3-OMD) is a more stable, catabolic product of L-DOPA. Accumulation of L-DOPA leads to increased concentrations of 3-OMD through conversion by catechol-O-methyltransferase. 3-OMD is detectable and stable in blood.

How does 3-OMD screening help in the diagnosis of AADC deficiency?

Elevated levels of 3-OMD can be detected in blood.

To diagnose AADC deficiency, genetic testing should be performed and 2 of the 3 core diagnostic tests should be positive.

3-OMD can be used as an initial screen when AADC deficiency is suspected.
Elevated levels of 3-OMD in the blood should prompt further testing to confirm the diagnosis of AADC deficiency.
Dried blood spot testing for 3-OMD is an option for screening individuals at high risk for AADC deficiency and a potential option for newborn screening.
This test may help to bridge the delay (typically months to years) in the diagnosis of the disease.

Screening for elevated 3-OMD in the blood of patients with suspected AADC deficiency may aid in early diagnosis.

PTC has partnered with CENTOGENE to offer 3-OMD testing via dried blood spot for initial screening of patients in whom AADC deficiency is clinically suspected and/or in at-risk patient populations.

For additional information, contact aadc.deficiency.info@ptcbio.com.

Abbreviations: L-DOPA, L-3,4 dihydroxyphenylalanine; CSF, cerebrospinal fluid; DDC, dopa decarboxylase; 3-OMD, 3-O-methyldopa.

Sponsored Satellite Symposium 9:
Long-Term Management of Organic Acidemia Patients With CAR-BAGLU®: The PROTECT Trial Overview & Interim Analysis.
Chair: Johannes Häberle (Zurich, Switzerland)

Introduction & Unmet needs in the long-term management of organic acidurias
Johannes Häberle (Zurich, Switzerland)

PROTECT Trial - Overview & enrolment status
Sufin Yap (Sheffield, United Kingdom)

PROTECT Trial - Interim analysis results
Sufin Yap (Sheffield, United Kingdom)

Open discussion & Q&A
but if we could find a way to alter the genes the body might start to heal itself

LET’S RECODE WHAT’S POSSIBLE

We don’t accept that the way things are is the way they need to be. If we can use gene therapy to recode the very building blocks of life, we should be able to recode everything about healthcare: the science, the system, even the status quo. We’re working to help create a world where lives can be lived more fully.
We would like to thank our corporate partners for supporting the SSIEM Virtual Symposium Day.

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Long-Chain Fatty Acid Oxidation Disorders
Evolving Best Practices

THURSDAY, 3 DECEMBER 2020 | 12:00 – 12:30 GMT

CHAIR
Jerry Vockley, MD, PhD
Cleveland Family Endowed Chair in Pediatric Research
Professor of Human Genetics
University of Pittsburgh
Chief of Medical Genetics
Director of the Center for Rare Disease Therapy
UPMC Children’s Hospital of Pittsburgh
Pittsburgh, Pennsylvania
United States

PANELISTS
Barbara K. Burton, MD
Professor of Pediatrics
Northwestern University Feinberg School of Medicine
Clinical Practice Director
Genetics and Metabolism
Ann & Robert H. Lurie Children’s Hospital of Chicago
Chicago, Illinois
United States

Ute Spiekerkötter, MD
Professor of Pediatric Metabolic Diseases
Medical Director
University Children’s Hospital Freiburg, Germany

TARGET AUDIENCE
This activity is intended for an international audience of non-US metabolic geneticists, genetic counsellors, metabolic dieticians, endocrinologists, and pediatricians.

LEARNING OBJECTIVES
Upon completion of this activity, participants will:
- Have increased knowledge regarding the
  • Morbidity associated with LC-FAODs despite guideline-based management
  • Role of emerging therapies for LC-FAODs
- Have greater competence related to
  • Identifying patients who may benefit from emerging therapies for LC-FAODs

AGENDA
Introduction
Jerry Vockley, MD, PhD

LC-FAODs: Is Guideline-Based Management Enough?
Jerry Vockley, MD, PhD, Barbara K. Burton, MD
Ute Spiekerkötter, MD

What Will Emerging Treatments Mean for LC-FAOD Management?
Jerry Vockley, MD, PhD, Barbara K. Burton, MD
Ute Spiekerkötter, MD

Concluding Remarks
Jerry Vockley, MD, PhD
Join us on December 8, 2020 for a 24-hour, live Newborn Screening Virtual Summit, a global event featuring sessions from over 40 international thought leaders on the latest trends and advances in Newborn Screening. This is a free event open to anyone interested in Newborn Screening.

Topics being discussed include:
- Newborn Screening workflow
- New Disorders
- Panel Expansion
- Duchenne Muscular Dystrophy

December 8, 2020

CLICK HERE TO REGISTER