SSIEM

VIRTUAL SYMPOSIUM DAY 2020

December
03

www.ssiemvirtual.org

SSIEM Virtual Symposium Organizer

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

Registered Office:
130-132 Tooley Street
London SE1 2TU

admin@ssiem.org

SSIEM Virtual Symposium Secretariat

S12! studio12 gmbh

Kaiser-Josef-Straße 9
6020 Innsbruck
AUSTRIA

office@studio12.co.at

studio12.co.at

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 Virtual Symposium Day 2020
December 03, 2020

All indicated times are based on Greenwich Mean Time (GMT, London).

12.00	**Start of the SSIEM Virtual Symposium Day**
12.00-12.30	**Industry Sponsored Symposia (p. 8)**
12.30-12.45	**Break**
12.45-13.15	**Industry Sponsored Symposia (p. 10)**
13.15-13.30	**Break**
13.30-13.35	**Introduction from the SSIEM President** *Gajja Salomons (Amsterdam, The Netherlands)*
13.35-14.30	**Adult Group Session** Chair: *David Cassiman (Leuven, Belgium)*
13.35	Educational programme for Adult Metabolic Medicine *Sandra Sirrs (Vancouver, Canada), Annalisa Secchi (Udine, Italy)*
14.00	Update on complex lipid disorders *Fanny Mochel (Paris, France)*
14.25	Q&A
14.30-15.00	**Archibald Garrod Award Lecture**
14.30	Introduction *Eva Morava (Rochester, United States)*
14.35	Archibald Garrod Award Lecture Clinical, radiological, and genetic characteristics in patients with ACO2 gene defects: an emerging neurometabolic syndrome *Ronen Spiegel (Afula, Israel)*
15.00-15.15	**Break**

15.15-17.00 **Invited lectures**
Chair: Ute Spiekerkötter (Freiburg, Germany)

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. 12)**

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

Register today

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*

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*Limited copies available, 1 per person

https://metabolics.nutricia-campus.com/

NUTRICIA METABOLICS
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SSIEM Virtual Symposium Day 2020
Industry Sponsored Symposia

All indicated times are based on Greenwich Mean Time (GMT, London).

12.00-12.30
sponsored by:

Sponsored Satellite Symposium 1

TBA

12.00-12.30
sponsored by:

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 round-table 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.

A Medscape LIVE! EVENT

Long-Chain Fatty Acid Oxidation Disorders

Evolving Best Practices

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

VIRTUAL EVENT

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

Supported by an independent educational grant from ultragenyx pharmaceutical

Medscape EDUCATION GLOBAL

12.00-12.30
sponsored by:
BIOMARIN

Sponsored Satellite Symposium 3

TBA

12.30-12.45 **Break**

12.45-13.15
sponsored by:
sobi rare strength

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 by:
Amicus Therapeutics

Sponsored Satellite Symposium 5:

TBA

12.45-13.15
sponsored by:
HORIZON

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

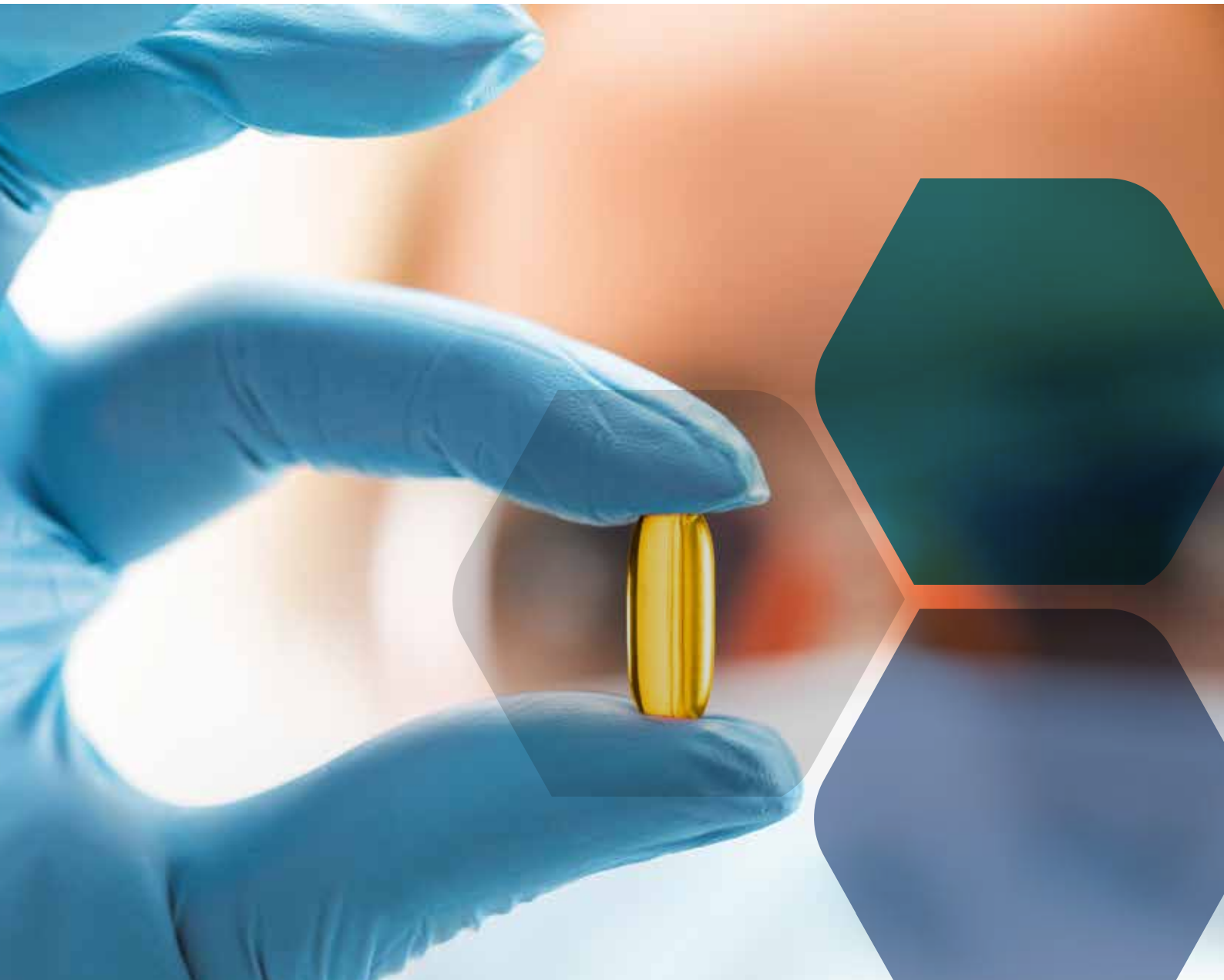
We are an italian pharmaceutical company with over **one hundred years of experience in several specialist therapeutic areas** where we continue to be active today.
We also strive to bring to the market treatments for **rare and niche diseases**, thereby ensuring that patients have access to the pharmaceutical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

PIAM
Pharma &
Integrative Care



20.00-21.00
sponsored by:

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

20.00-21.00
sponsored by:

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

NEWBORN SCREENING
VIRTUAL SUMMIT 2020
Learn. Share. Develop.

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

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20.00-20.30
sponsored by:

RECORDATI RARE DISEASES GROUP

Sponsored Satellite Symposium 9:
Long-Term Management of Organic Acidemia Patients With CARBAGLU®: 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.

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recode for life™