SSIEM

2026 ANNUAL SYMPOSIUM • HELSINKI FINLAND

SSIEM 2026 ANNUAL SYMPOSIUM:

PROGRAMME

25 - 28 AUGUST 2026 • HELSINKI FINLAND

SSIEM Symposium 2027

Society for the Study of Inborn Errors of Metabolism

31 AUG - 3 SEP 2027, DUBLIN, IRELAND

complexity | diversity | inclusion

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See you in Dublin!

- Vibrant European capital city
- Strong clinical, research, academic and patient communities
- Dublin 'buzz' - endlessly exciting
- Easy international access
- Rich culture, history and hospitality

ISIMD Irish Society of Inherited Metabolic Disorders

SSIEM

www.ssiem2027.org

#SSIEM2027

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Welcome to the 2026 SSIEM Symposium!

Let's work together to secure a sustainable future for both research and clinical services in inborn errors of metabolism. I want to extend a huge thank you to everyone who submitted or evaluated abstracts, agreed to present or chair a session, and supported us in putting this meeting together. No matter your age, nationality, or profession, this is a wonderful opportunity to learn from one another and share new insights.

Our theme, **Next Generation Metabolic Medicine**, is about more than just parents and children. It encompasses the education of future healthcare professionals and scientists. Today's research is tomorrow's clinical practice—and we will also catch an exciting glimpse into the research of the future. Our patients remind us every day that many questions remain unsolved in our field. Let's tackle them together!

This booklet gives you a handy summary of the Symposium programme. As mentioned before, we have introduced some exciting new features and unique highlights. You can find the full details, latest updates, and any program changes on our website (ssiem2026.org) or via the official app.

Thank you so much for making the journey to the northernmost SSIEM Symposium! Helsinki is one of the greenest capitals in the world, famous for its beautiful parks and stunning coastline. I highly encourage you to head outdoors and recharge with a walk, run, or bike ride. In our effort to minimize waste, we kindly ask you to consider bringing your own reusable water bottle.

We are also deeply grateful to our industrial sponsors. Their generous contributions not only make this Symposium possible but keep the SSIEM active and thriving in its mission.

We are truly looking forward to welcoming you to Helsinki for what promises to be an inspiring, productive, and memorable event. I am confident that our time together will ultimately lead to even better care for patients with an IEM.

Warm regards,

Risto Lapatto

Symposium President

ORGANISATION & COMMITTEES

Organisation

SSIEM

SUOMEN METABOLINEN KLUBI RY

With the support of City of Helsinki

Helsinki

Symposium President

- **Risto Lapatto** (Helsinki, Finland)

Committees

Local Organising Committee

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- **Mervi Hyvönen** (Helsinki, Finland)
- **Risto Lapatto** (Helsinki, Finland)
- **Päivi Miettinen** (Helsinki, Finland)
- **Irina Nagy** (Oulu, Finland)
- **Pasi Nevalainen** (Tampere, Finland)
- **Harri Niinikoski** (Turku, Finland)
- **Katrin Strengell** (Helsinki, Finland)
- **Jetta Tuokkola** (Helsinki, Finland)
- **Henna Tyynismaa** (Helsinki, Finland)

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- **Alice Dianin** (Verona, Italy)
- **Angeles Garcia Cazorla** (Barcelona, Spain)
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- **Helen Michelakakis** (Athens, Greece)
- **Philippa Mills** (London, United Kingdom)
- **Elaine Murphy** (London, United Kingdom)
- **Shamima Rahman** (London, United Kingdom)
- **Manuel Schiff** (Paris, France)
- **Trine Tangeraas** (Oslo, Norway)
- **Jerry Vockley** (Pittsburgh, United States)

SCIENTIFIC PROGRAMME

DAILY OVERVIEW

DAILY OVERVIEW

SUNDAY, AUGUST 23, 2026

15:00-21:30 INFORM Meeting Part I (registration at INFORM – limited places, venue Hilton Helsinki Kalastajatorppa, NOT Messukeskus)

MONDAY, AUGUST 24, 2026

8:00-17:30 INFORM Meeting Part II (registration at INFORM – limited places, venue Hilton Helsinki Kalastajatorppa, NOT Messukeskus)

	101ab	101c	101d	102	203a	208	209	210	211	212	213	214	215	216
8:00								8:00-18:00 UCD Guideline group meeting (*)						
8:30														
9:00														
9:30														
10:00										10:00-12:00 eHOD (*)				
10:30														
11:00														
11:30														
12:00				12:00-14:00 Recon4IMD (*)										
12:30														
13:00	13:00-16:00 SSIEM		13:00-15:00 eHOD (*)											
13:30														
14:00	Dietitians'	14:00-16:00 MetabERN												
14:30	Group													
15:00	Academy	plenary												
15:30	Review	meeting (*)												
16:00					16:00-18:00 MetabERN/ C-FAO (*)	16:00-18:00 MetabERN/ LSD SNW (*)			16:00-18:00 MetabERN/ PD SNW (*)		16:00-18:00 MetabERN/ PM-MD (*)	16:00-18:00 MetabERN/ NOMS (*)	16:00-18:00 SSIEM Council meeting (*)	16:00-18:00 MetabERN/ AOA SNW (*)
16:30							16:30-20:30 ERNDIM Board meeting (*)							
17:00														
17:30														
18:00														
18:30														
19:00														
19:30														
20:00														
20:30														

(*) = by invitation only

TUESDAY, AUGUST 25, 2026

	HALL 1	HALL 5A	HALL 5B	HALL 5C	101ab	101c	101d	102	208	209	210	214
8:00			8:15-9:15 Introductory session by the local organizing committee								8:00-12:00 UCD Guideline group meeting (*)	
8:30	8:30-10:35 SSIEM Dietitians' Group (DG): Nutrition and Dietetics session	8:30-10:35 SSIEM Adult Metabolic Physicians' Group: Tailoring and discontinuation of therapy in adulthood										
9:00						9:00-10:30 ERNDIM Workshop (*)	9:00-10:30 ERNDIM Workshop (*)	9:00-10:30 ERNDIM Workshop (*)	9:00-10:30 ERNDIM Workshop (*)	9:00-10:30 ERNDIM Workshop (*)		9:00- 13:00 JIMD Editorial Board Meeting (*)
9:30												
10:00												
10:30					10:30-11:00 COFFEE							
11:00	11:00- 12:00 SSIEM DG & AMPG: Relaxation of PKU treatment in adulthood					11:00-12:45 ERNDIM Open meeting						
11:30												
12:00												
12:30			12:30-14:00 LUNCH				12:45-13:45 SSIEM Adult Metabolic Physicians' Group business meeting (AMPG members only)		12:30-14:00 LUNCH			
13:00		12:45-13:45 Satellite meetings by the sponsors										
13:30												
14:00	14:00-14:10 Opening of the Symposium 14:10-14:40 Opening Lecture 14:40-15:40 Plenary 1 The Foeto-Maternal Unit and Inborn Errors of Metabolism (IEM)											
14:30												
15:00												
15:30				15:40-16:15 COFFEE								
16:00	16:15-17:45 Parallel session A1: Dietetics and nutrition	16:15 – 17:45 Parallel session A4: Gene and innovative therapies	16:15-17:45 Parallel session A3: Mitochondrial disorders	16:15-17:45 Parallel session A2: Protein modification disorders including CDG		16:00-17:45 SSIEM, MetabERN and ISNS session on Newborn screening						
16:30												
17:00												
17:30												
18:00	17:50-18:30 Garrod Lecture											
18:30	18:30-20:30 Welcome Reception, Posters											
19:00												
19:30		19:30-20:30 YSSIEM Inaugural Meeting										
20:00												
20:30												
21:00												

(*) = by invitation only

= Plenary Session
= Side & Administrative Meetings
= Parallel Session
= Special Sessions
= Posters
= Satellite Symposia by sponsors
= Networking

WEDNESDAY, AUGUST 26, 2026

	HALL 1	HALL 5A	HALL 5B	HALL 5C	101ab	101c	101d	102	104	203b	208
7:30		7:30-8:30 Satellite meetings by the sponsors 2				7:30-8:30 Speed Mentoring					
8:00											8:00-8:30 Lab Talk
8:30											
9:00	8:45-10:15 "From presentation to diagnosis: a case-based education session with the SSIEM Educational and Training (ETAC) Academy"	8:45-10:15 Parallel session B1: Fatty acid oxidation and related disorders	8:45-10:15 Parallel session B2: Clinical studies and real world data	8:45-10:15 Parallel session B3: Newborn screening							
9:30											
10:00				10:00-10:40 COFFEE + COFFEE WITH JIMD EDITORS							
10:30							10:30-14:00 Patient advocacy group (PAG) session: Making PAG work sustainable				
11:00	10:40-12:15 Plenary 2 - New Diseases and Mechanisms										
11:30											
12:00				12:20-12:50 SSIEM Advisory Council & Council meeting (*)	12:15-14:15 LUNCH					12:00-14:00 Drug Repurposing consortium meeting (*)	
12:30											
13:00				13:00-14:00 SSIEM Annual General Meeting (AGM) (SSIEM members only)							
13:30											
14:00	14:00-16:00 Ensuring Patient Access to Innovation in Rare Diseases (details to follow)	14:15-15:45 Parallel session C1: Disorders of vitamins and cofactors	14:15-15:45 Parallel session C2: New methods	14:00-16:00 SSIEM Dietitians' Group Business Meeting (DG members only)							
14:30											
15:00					15:00-16:00 COFFEE						
15:30											
16:00	16:00-17:30 Plenary 3 - Physics and Mathematics Enter the World of IEM										
16:30											
17:00											
17:30					17:30-18:30 Poster Walk						
18:00											
18:30		18:30-19:30 Satellite meetings by the sponsors 3	18:30-19:30 Satellite meetings by the sponsors 4	18:30-19:30 Satellite meetings by the sponsors 5						18:00-20:00 GalNet meeting(*)	
19:00											
19:30											
20:00	20:00-22:00 Young Investigators Evening (YSSIEM)										
20:30											
21:00											

(*) = by invitation only

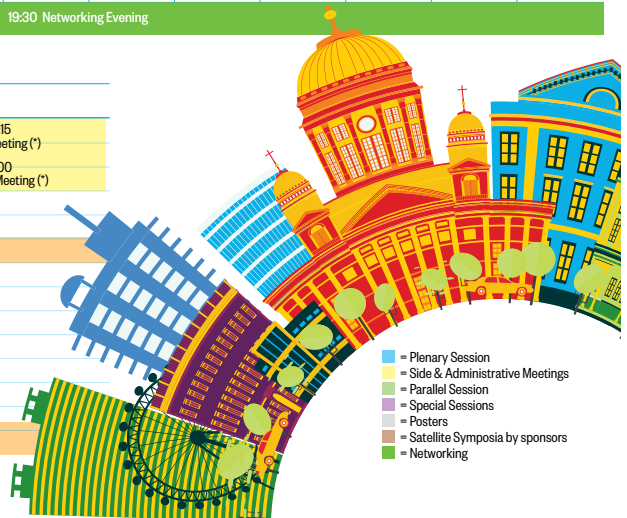
= Plenary Session
= Side & Administrative Meetings
= Parallel Session
= Special Sessions
= Posters
= Satellite Symposia by sponsors
= Networking

THURSDAY, AUGUST 27, 2026

	HALL 1	HALL 5A	HALL 5B	HALL 5C	101ab	101c	101d	102	104	203B	208	210
7:00									7:00-8:00 eHOD CBS guidelines group (*)			
7:30		7:30-8:30 Satellite meetings by the sponsors	7:30-8:30 Satellite meetings by the sponsors	7:30-8:30 Satellite meetings by the sponsors	7:30-8:30 Satellite meetings by the sponsors	7:30-8:30 Speed Mentoring 2						
8:00												
8:30											8:30-9:00 LAB Talk	
9:00	9:00-10:30 Plenary 4 - OMICS data help us understand pathology in metabolic pathways											
9:30												
10:00												
10:30					10:30-11:00 COFFEE							
11:00	11:00-12:30 Parallel session D1: Lysosomal disorders	11:00-12:30 Parallel session D4: Organic acidurias	11:00-12:30 Parallel session D3: Neurometa-bolic disorders	11:00-12:30 Parallel session D2: Nurses' and allied healthcare professionals' meeting						11:15-12:15 JIMD Editors meeting (*)		
11:30												
12:00												
12:30					12:30-13:30 LUNCH				12:30-13:30 Gyrate Atrophy Meeting			
13:00												
13:30	13:30-15:00 Plenary 5 - Mitochondria Now and Forever											
14:00												
14:30												
15:00					15:00-15:30 COFFEE							
15:30	15:30-17:00 Parallel session E1: Phenylke-tonuria	15:30-17:00 Parallel session E3: Carbohydrate disorders	15:30-17:00 Parallel session E2: Urea cycle disorders	15:30-17:00 Training in IEM: A roundtable discussion (paed/adult clin, clin lab, diet, nursing)								
16:00												
16:30												
17:00					17:00-18:00 Poster viewing							
17:30												
18:00												
18:30												
19:00												
19:30	19:30 Networking Evening											

FRIDAY, AUGUST 28, 2026

	HALL 1	104
7:30		7:30-8:15 ICIEM IOC Meeting (*)
8:00		
8:30		8:15-9:00 ICIEM SciOrg Meeting (*)
9:00	9:00-10:15 Late-Breaking News: 5 talks	
9:30		
10:00	10:15-10:45 COFFEE	
10:30	10:45-12:15 Plenary 6 - Looking Forward	
11:00		
11:30	12:15-13:00 Komrower lecture	
12:00		
12:30	13:00-13:20 Closing session Awards Ceremonie (LBN, posters) SSIEM 2027 Dublin presentation	
13:00		
13:30		
14:00	13:20-14:00 LUNCH	



SCIENTIFIC PROGRAMME

SUNDAY 23 AUGUST

15:00 - 21:30 **INFORM Meeting Part I**
Hilton Helsinki Kalastajatorppa
(registration at INFORM – limited places)

MONDAY 24 AUGUST

08:00 - 17:30 **INFORM Meeting Part II**
Hilton Helsinki Kalastajatorppa
(registration at INFORM – limited places)

08:00 - 18:00 **UCD Guideline group meeting**
Meeting room 210
(by invitation only)

10:00 - 12:00 **eHOD**
Meeting room 212
(by invitation only)

12:00 - 14:00 **Recon4IMD**
Meeting room 102
(by invitation only)

13:00 - 15:00 **eHOD**
Meeting room 101D
(by invitation only)

13:00 - 16:00 **SSIEM Dietitians' Group Academy Review: addressing dietetic challenges**
Meeting room 101AB
(open to all)

14:00 - 16:00 **MetabERN plenary meeting**
Meeting room 101C
(by invitation only)

16:00 - 18:00 **SSIEM Council meeting**
Meeting room 215
(by invitation only)

16:00 - 18:00 **MetabERN meetings**
(by invitation only)

- MetabERN LSD SNW, Meeting room 208
- MetabERN AOA SNW, Meeting room 216
- MetabERN NOMS, Meeting room 214
- MetabERN PM-MD, Meeting room 213
- MetabERN PD SNW, Meeting room 211
- MetabERN C-FAO SNW, Meeting room 203A

16:30 - 20:30 **ERNDIM Board meeting**
Meeting room 209
(by invitation only)

TUESDAY 25 AUGUST

08:00 - 12:00 **UCD Guideline group meeting**
Meeting room 210
(by invitation only)

08:15 - 09:15 **Introduction for those who are new to the field: multidisciplinary collaboration for diagnosing and treating inborn errors of metabolism:**
Hall 5B
CHAIR: *Pasi Nevalainen (Tampere, Finland)*

08:15 - 08:30 Introduction to the SSIEM organization and the SSIEM 2026 meeting
Pasi Nevalainen (Tampere, Finland)

08:30 - 08:45 Pediatrician's view
Mervi Hyvönen (Helsinki, Finland)

08:45 - 08:55 Clinical laboratory view
Irina Nagy (Oulu, Finland)

08:55 - 09:05 Dietician's view
Jetta Tuokkola (Helsinki, Finland)

09:05 - 09:15 Adult metabolist's view
Pasi Nevalainen (Tampere, Finland)

08:30 - 10:35 **SSIEM Dietitians' Group (DG): Nutrition and Dietetics session**
Hall 1
(open to all)
CHAIRS: *Camilla Caroee (Copenhagen, Denmark); Alexander Hoeller (Innsbruck , Austria)*

08:30 - 08:55 Dietary management of Urea Cycle Disorders across the lifespan: Consensus and Controversies in practice
Krista Viau (Alexandria, United States)

08:55 - 09:20 Lysinuric Protein Intolerance: metabolic challenges and dietary management
Harri Niinikoski (Turku, Finland) and Katrin Strengell (Helsinki, Finland)

09:20 - 09:45 Dietary management of Pyruvate Dehydrogenase Deficiency
Rachel Skeath (London, United Kingdom)

09:45 - 10:10 Point-of-Care testing for blood phenylalanine: Impact on clinical decision--making in PKU
Alex Pinto (Birmingham, United Kingdom)

10:10 - 10:35 Protein substitutes used in the treatment of Phenylketonuria distinctly modulate gut nutrient absorption and bacterial growth: an in vitro study
Catarina Rodrigues (Lisbon, Portugal)

08:30 - 10:35 **SSIEM Adult Metabolic Physicians' Group (AMPG):**
Tailoring and discontinuation of therapy in adulthood
(open to all)
Hall 5A
CHAIRS: *Sandra Sirrs (Vancouver, Canada) and Risto Lapatto (Helsinki, Finland)*

08:30 - 08:35 Opening of the session
(Mirjam Langeveld, Amsterdam, Netherlands)

08:35 - 10:00 Discontinuation of ERT for lysosomal storage disorders

Introduction

Talks

1. Ethical considerations regarding discontinuation of enzyme replacement therapy
Hans-Jürgen Christen (Hanover, Germany)

2. Progression of cardiac manifestations of Fabry disease despite enzyme replacement therapy
Bram Veldman (Amsterdam, Netherlands)

3. What happens if we discontinue enzyme replacement therapy in Fabry disease?
Maud Janssens (Amsterdam, Netherlands)

4. Discontinuation of ERT in Pompe disease
Lianne Potters (Netherlands)

10:00 - 10:35 Debate: "ERT should be discontinued in patients with Fabry disease and advanced fibrosis"
For / against: *Mirjam Langeveld (Amsterdam, Netherlands)/ Rick Steeds (Birmingham, United Kingdom)*

Closing of the session: *Sandra Sirrs (Vancouver, Canada)*

09:00 - 10:30 **ERNDIM Workshops**
Meeting rooms 101C, 101D, 102, 208 and 209
(by invitation only)

09:00 - 13:00 **JIMD Editorial Board Meeting**
Meeting room 214
(by invitation only)

10:35 - 11:00 **COFFEE** Halls 4&5

11:00 - 12:00 **SSIEM Dietitians' Group (DG) and Adult Metabolic Physicians' Group (AMPG) joint meeting:**
Relaxation of PKU treatment in adulthood
Hall 1
Collaborative roundtable with the Adult Metabolic Physicians and Dieticians Group
LEAD: *Elaine Murphy (London, United Kingdom) and Alice Dianin (Verona, Italy)*
PANELLISTS: *Peter Burgard (Heidelberg, Germany), Anita MacDonald (Birmingham, United Kingdom), Robin Lachmann (London, United Kingdom), Francjan van Spronsen (Groningen, Netherlands), Kirsten Ahring (Copenhagen, Denmark)*

11:00 - 12:45 **ERNDIM Open meeting**
Meeting room 101C

12:30 - 14:00 **LUNCH** Halls 4&5

12:45 - 13:45 **SSIEM Adult Metabolic Physicians' Group business meeting**
Meeting room 101D
(SSIEM AMPG members only)

12:45 - 13:45 **Satellite meetings by the sponsors, please see from page 46**
Halls 5A, 5B, 5C and 101AB

14:00 - 14:10 **Opening of the Symposium**
Hall 1

14:10 - 14:40 **Opening Plenary Lecture**
Hall 1
CHAIRS: *Manuel Schiff (Paris, France) and Risto Lapatto (Helsinki, Finland)*

From the zygote to a person: genes, epigenetics, environment, and chance
Juha Kere (Stockholm, Sweden)

14:40 - 15:40 **Plenary 1 - The Foeto-Maternal Unit and Inborn Errors of Metabolism (IEM)**
Hall 1
CHAIRS: *Helen Michelakakis (Athens, Greece) and Alice Dianin (Verona, Italy)*

- Prenatal Therapies for IEM
  Tippi McKenzie (San Francisco, United States)
- Managing Pregnancy in Inherited Metabolic Disorders: A Dietary Approach
  Charlotte Ellerton (London, United Kingdom)

15:40 - 16:15 **COFFEE** Halls 4&5

16:00 - 17:45 **SSIEM, MetabERN and ISNS session on Newborn Screening on Lysosomal Diseases**
Hall 101C
CHAIR: *Rolf Zetterström (Stockholm, Sweden)*
TALKS BY: *Stefan Kölker (Heidelberg, Germany), Giancarlo la Marca (Florence, Italy), Mirjam Langeveld (Amsterdam, The Netherlands), Maria Jose De Castro Lopez (Manchester, United Kingdom)*
Discussion

Parallel sessions A

16:15 - 17:45 **A1: Dietetics and nutrition**
Hall 1
CHAIRS: *Julio Rocha (São José, Portugal) and Jetta Tuokkola (Helsinki, Finland)*

O-001 Safety of overnight dietary treatment in LCHAD deficiency patients
Katrin Strengell (Helsinki, Finland)

O-002 Variability in dietary management in Isovaleric Acidaemia (IVA): A retrospective longitudinal cohort study
Sarah Cawtherley (London, United Kingdom)

O-003 Single-centre experience of parenteral nutrition in acutely managing methylmalonic acidaemia and propionic acidaemia
Georgina Wood (Manchester, United Kingdom)

O-004 Dietary Protein Modulation, Gut Microbiota, and Metabolic Control in Methylmalonic Acidemia: A Prospective Longitudinal Study
Engin Kose (Ankara, Türkiye)

O-005 Breastfeeding in inborn errors of metabolism: a retrospective study in cases detected by neonatal screening
Sinziana Stanescu (Madrid, Spain)

O-006 Twenty Four Years of Liver Transplantation for Inherited Metabolic Disorders: Have We Improved Nutritional Outcomes?
Anne Daly (Birmingham, United Kingdom)

O-007 Increased Adiposity Despite Protein and Energy Restriction in Adults with Inborn Errors of Protein Metabolism
Giorgia Gugelmo (Padova, Italy)

O-008 Large Language Models' Responses to Patient-Like Questions in Metabolic Dietetics: A Comparative Evaluation
Martina Tosi, Department of Dietetics (Birmingham, United Kingdom)

O-009 Developing a paediatric dietetic complexity tool to inform workforce requirements using a best practice model
Fiona White (Manchester, United Kingdom)

16:15 - 17:45 **A2: Protein modification disorders including CDG**
Hall 5C
CHAIRS: *Dulce Quelhas (Porto, Portugal) and Kimitoshi Nakamura (Kumamoto, Japan)*

O-010 Scaling phenotypes from clinical notes to identify hidden inborn errors of metabolism using machine learning
Mohammad Ghouse Syed (New York, United States)

O-011 A Discovery-driven Glycoproteomic Workflow Reveals Candidate Plasma Biomarkers for Congenital Disorders of Glycosylation
Maria Blomqvist (Gothenburg, Sweden)

O-012 SLC39A8-related Manganese Deficiency: Phenotypic Spectrum and Treatment Response in an International Cohort of 31 Patients
Julien Park (Münster, Germany)

O-013 Development of a splice-switching antisense oligonucleotide for a deep-intronic ATP6AP1-CDG variant
Ludvík Hejl (Radboud, Netherlands)

O-014 Preliminary results from a PMM2-CDG natural history study: the international cooperative assessment of ataxia scale
Fabio Pettinato (Catania, Italy)

O-015 Complex Ganglioside Deficiency as a Hallmark of Golgi-Related CDG
Peter Witters (Leuven, Belgium)

O-016 AAV9-PMM2 therapy rescues neuronal phenotypes in a human cortical organoid model of PMM2-CDG
Tamas Kozicz (New York, United States)

O-017 COG5-CDG organoids reveal a novel role for Golgi organization in human brain development
Elena Taverna (Milan, Italy)

16:15 - 17:45 **A3: Mitochondrial disorders**
Hall 5B
CHAIRS: *Johan van Hove (Aurora, United States) and Pirjo Isohanni (Helsinki, Finland)*

O-019 Novel ACAA2 variant causes autosomal dominant metabolic disorder with infantile hypoglycemia, steatohepatitis, lipodystrophy, lipomatosis
Mary Kate LoPiccolo (New York, United States)

O-020 Modulation of redox homeostasis in malate-aspartate shuttle deficiencies: mechanistic and therapeutic insights
Hannah German (Utrecht, Netherlands)

O-021 PDH Deficiency: Outcome and Therapeutic Management in a Large French Cohort of Patients Born before 2010
Cecilia Marelli (Montpellier, France)

O-022 Pleuroparenchymal Fibroelastosis in patients with DGUOK-deficiency – a case series
Anibh Das (Hanover, Germany)

O-023 Sodium valproate in non-POLG primary mitochondrial disease: safety and effectiveness from an international survey
Marcello Bellusci (Madrid, Spain)

O-024 Evolutionarily conserved temperature dependency leads to loss of protein O-GlcNAc in complex III deficient mice
Christa Kietz (Helsinki, Finland)

O-025 When silence is noise: a silent start codon single nucleotide variant impairs mitochondrial RNA processing
Saskia Wortmann (Salzburg, Austria)

O-026 MECP2-Dependent LYRM4 Deficiency Leads to Multifaceted Mitochondrial Dysfunction and Derailed Neuronal Maturation
Gerarda Cappuccio (Houston, United States)

O-027 Tetrahydrobiopterin metabolism is a shared vulnerability in acquired and primary mitochondrial deficiencies
Luisa Cruz (Florianópolis, Brazil)

O-029 Consensus statements for diagnostic criteria and management of MELAS and stroke-like episodes
Shamima Rahman (London, United Kingdom)

16:15 - 17:45 **A4: Gene and innovative therapies**
Hall 5A
CHAIRS: *Nicola Brunetti-Pierri (Pozzuoli, Italy) and Mervi Hyvönen (Helsinki, Finland)*

O-030 Development of mRNA therapy in clinically relevant mouse models of Glycogen Storage Disease type 1b
Lucia De Stefano (Pozzuoli, Italy)

O-031 Efficacy and nutritional changes from a phase 3 trial of DTX401 gene therapy for GSDIa
Andrea Haijer-Schreuder (Groningen, Netherlands)

O-032 Liver-Directed AAV-mediated Gene Therapy for Wolman Disease
Iolanda Boffa (Pozzuoli, Italy)

O-033 AMT-191 gene therapy in males with Fabry disease; phase 1/2 initial safety and biomarker results
Maryam Banikazemi (New York, United States)

O-034 AAV and mRNA therapies: A sequential and mirror therapeutic approach in MSUD
Clément Pontoizeau (Paris, France)

O-035 Data from 50+ cumulative patient years exposure to mRNA-3927, an investigational treatment for propionic acidemia
BC Schwahn (Manchester, United Kingdom)

O-036 In vivo gene editing in a novel mouse model of argininosuccinate lyase deficiency
Timo Keskinen (Helsinki, Finland)

O-037 Hepatocyte-specific gene replacement improves energy metabolism and survival in mouse model of mitochondrial CIII Deficiency
Rishi Banerjee (Helsinki, Finland)

O-038 Dual mRNA lipid nanoparticles rescue metabolic performance in a mouse model of trifunctional protein deficiency
Troy von Beck (Durham, United States)

O-039 Single-vector AAV8 genome editing overcomes the dominant-negative effect in mitochondrial trifunctional protein deficiency mouse
Tatiana Terranova (Durham, United States)

O-040 CRISPR base editing as a one-fits-many gene editing therapy for OTC deficiency
Sven Klassa (Zurich, Switzerland)

17:50 - 18:30 **Garrod Award Lecture: Cystathionine β-synthase deficiency in the E-HOD registry**
Hall 1
CHAIR: *Matthias Baumgartner (Zurich, Switzerland)*
Andrew Morris (Manchester, United Kingdom)

18:30 - 21:00 **Welcome Reception, Posters**
Halls 4 & 5 (Exhibition and Poster area)

19:30 - 20:30 **YSSIEM (Young SSIEM) Inaugural Meeting**
Hall 5A
(open to all)

WEDNESDAY 26 AUGUST

07:30 - 08:30 **Speed Mentoring**
Meeting room 101C
(details to follow)

07:30 - 08:30 **Satellite meetings by the sponsors, please see from page 46**

08:00 - 08:30 **Lab Talk by *Philippa Mills* (London, United Kingdom)**
Meeting room 208

08:45 - 10:15 **From presentation to diagnosis: a case-based education session with the SSIEM Educational and Training (ETAC) Academy**
Hall 1
CHAIR: *Elaine Murphy (London, United Kingdom)*

Parallel sessions B

08:45 - 10:15 **B1: Fatty acid oxidation and related disorders**
Hall 5A
CHAIRS: *Ute Spiekerkoetter (Freiburg Im Breisgau, Germany) and Jerry Vockley (Pittsburgh, United States)*

O-041 Mortality in fatty acid oxidation disorders despite newborn screening: insights from a European multicentre survey
Marie-Cecile Nassogne (Brussels, Belgium)

O-042 Validation of carnitine transport (OCTN2) activity assay in cultured urothelial cells for primary carnitine deficiency
Sacha Ferdinandusse (Amsterdam, Netherlands)

O-043 N-Adipyl-D-Ala-D-His as a Superior Anaplerotic Therapy for VLCAD Deficiency in a Knockout Mouse Model
Al-Walid Mohsen (Pittsburgh, United States)

O-044 Long-term clinical outcomes of patients with VLCAD deficiency identified by newborn screening
Luisa Burgenmeister (Heidelberg, Germany)

O-045 Metabolic rerouting drives odd-chain lipid enrichment in mitochondrial **β**-oxidation disorders
Viktorija Juric (Innsbruck, Austria)

O-046 The Circulating Non-Coding Genome Reveals a Distinct Interferon Signature in X-linked Adrenoleukodystrophy
Lara M Marten (Göttingen, Germany)

O-047 Hydroxylated long-chain acylcarnitines cause Schwann cell bioenergetic failure: implications for LCHADD/MTPD peripheral Neuropathy
Chen Zhang (Aarhus, Denmark)

08:45 - 10:15 **B2: Clinical studies and real world data**
Hall 5B
CHAIRS: *David Cassiman (Leuven, Netherlands) and Allan Lund (Copenhagen, Denmark)*

O-048 The Online Inherited Metabolic Diseases (OIMD) knowledgebase
Johannes Zschocke (Innsbruck, Austria)

O-049 Genetic diagnosis network and sustainable registry for real-world evidence in inborn errors of metabolism
Takashi Hamazaki (Osaka, Japan)

O-050 A Multinational Study of Patient Characteristics, Management and Outcomes in Lesch-Nyhan Syndrome
Alwyn Charles (Crumlin, Ireland)

O-051 International Recommendations on Use of Brain MRI for Screening, Monitoring, and Research in Cerebral Adrenoleukodystrophy
Hemmo Yska (Paris, France)

O-052 Results from a phase 2 study evaluating the effects of GLM101 in patients with PMM2-CDG
Mel McSweeney (London, United Kingdom)

O-053 Cognition and Ataxia Benefits of Venglustat in GD3: Prespecified Analyses of Neurologic Endpoints in LEAP2MONO
Pramod Mistry (New Haven, United States)

O-054 Therapeutic Strategies in Adult Cerebral Adrenoleukodystrophy: Balancing Hematopoietic Stem Cell Transplantation and Leriglitazone
Fanny Mochel (Paris, France)

O-055 Bridging the Treatment Gap: A Systematic 10-Principle Framework for Drug Repurposing in Inherited Metabolic Diseases
Clara van Karnebeek (Amsterdam, Netherlands)

08:45 - 10:15 **B3: Newborn screening**
Hall 5C
CHAIRS: *Trine Tangeraas (Oslo, Norway) and Giancarlo la Marca (Florence, Italy)*

O-056 Newborn screening for alpha-mannosidosis: quantitative assessment of a novel biomarker in dried blood spots
Simona Murko (Hamburg, Germany)

O-057 First prospective newborn screening for metachromatic leukodystrophy enables presymptomatic intervention
Thomas Neisse (Hanover, Germany)

O-058 Comprehensive newborn screening of lysosomal diseases: insights from the LysoNeo study
Abdellah Tebani (Rouen, France)

O-059 smMIP-based genomic newborn screening in India: prospective validation against tandem mass spectrometry and whole-exome sequencing
Harsh Sheth (Ahmedabad, India)

O-060 Expanded Newborn Screening by Tandem Mass Spectrometry: 25-year experience in Tuscany and Umbria regions (Italy)
Giancarlo la Marca (Florence, Italy)

O-061 A Novel Predictive Metabolomic Algorithm for Genotype-Phenotype Stratification in Newborn Screening of β-Oxidation Defects
Michela Perrone Donnorso (Genova, Italy)

O-062 CRINGENES: A National Pilot Integrating Genomics, Metabolomics, and Functional Testing to Enhance Newborn Screening
Judit Garcia-Villoria (Barcelona, Spain)

10:00 - 10:40 **COFFEE** Halls 4&5

10:30 - 14:00 **Patient advocacy group (PAG) session: Making PAG work sustainable**
Meeting room 101D
CHAIRS: *Päivi Miettinen (Helsinki, Finland) and Anita Ingwood (Brisbane, Australia)*

10:40 - 12:15 **Plenary 2 - New Diseases and Mechanisms**
Hall 1
CHAIRS: *Mirjam Langeveld (Amsterdam, Netherlands) and Henna Tyynismaa (Helsinki, Finland)*

What have we learned from the undiagnosed disease program?
William Gahl (Bethesda, United States)
Niemann-Pick Disease Type C: From Molecular Pathways to Therapeutic Impact
Frances Platt (Oxford, United Kingdom)
From Curiosity to Mechanism: Discovering a Ribosomopathy in Cobalamin Deficiency
Annita Achilleos (Nicosia, Cyprus)

12:00 - 14:00 **Drug Repurposing consortium meeting**
Meeting room 203B

12:15 - 14:15 **LUNCH** Halls 4&5

12:20 - 12:50 **SSIEM Advisory Council & Council meeting**
Hall 5C
(by invitation only)

13:00 - 14:00 **SSIEM Annual General Meeting (AGM)**
Hall 5C
(SSIEM members only)

14:00 - 16:00 **SSIEM Dietitians' Group business meeting**
(SSIEM DG members only)
Hall 5C
CHAIR: *Alice Dianin*

14:00 - 14:15 Welcome and report from the SSIEM DG Committee
Alice Dianin (Verona, Italy) and Charlotte Ellerton (London, United Kindgom)

14:15 - 14:45 Metabolic dietitians' education and workforce project
Martina Tosi (Birmingham, United Kindgom) and Fiona White (Manchester, United Kindgom)

14:45 - 15:25 short oral presentations and discussion:
- Use of Glycomacropeptide (GMP) in Maternal PKU; a multicentre prospective observational study
Charlotte Ellerton (London, United Kingdom)

- Impact of Low Phenylalanine Free Diet on Compliance and Metabolic Control in Tyrosinemia Type 1
Esma Uygur (Istanbul, Turkey)

- Infantile-onset lysosomal acid lipase deficiency – impact of fat intake on growth and duodenal histology
Fiona White (Manchester, United Kingdom)

15:25 - 15:55 Celebration of 30 years of European Metabolic Dietitians and SSIEM Dietitians Group

15:55 - 16:00 Closing of the session

14:00 - 16:00 **Ensuring Patient Access to Innovation in Rare Diseases**
Hall 1
CHAIRS: *Cary Harding (Portland, United States), Kirmo Wartiovaara (Helsinki, Finland)*

Accelerating access to orphan drugs through regulatory innovation
Janet Woodcock (Washington, United States)

Recent EU regulatory developments and useful tools for orphan drug development
Kristina Larsson (Amsterdam, Netherlands)

Economic realities and the case for patient-centered orphan drug access models
Barbara Yu (London, United Kingdom)

From research to approval: a non-profit development and distribution model for rare disease gene therapies
Ilaria Villa (Rome, Italy)

Panel and Audience Discussion
PANELISTS: *Speakers and Julien Baruteau (London, United Kingdom)*

Parallel abstract sessions C

14:15 - 15:45 **C1: Disorders of vitamins and cofactors**
Hall 5A
CHAIRS: *Bernd Schwann (Manchester, United Kingdom) and Barbara Plecko (Graz, Austria)*

O-063 Preclinical Evaluation of Iron Nanoclusters for IRIDA Therapy: Efficacy and Safety Assessment
François Feillet (Nancy, France)

O-064 Towards a new definition of cobalamin C and epi-cobalamin deficiency in the newborn screening era
Giorgia Olivieri (Rome, Italy)

O-065 Descriptive analysis of clinical and genetic characteristics in 114 patients with cblE-MTRR or cblG-MTR disease
Anna Huemer (Graz, Austria)

O-066 Dried blood spot vitamin B_6 vitamer profiling reveals altered B_6 homeostasis in ALDH7A1-related pyridoxine-dependent Epilepsy
Fatimah Almousawi (London, United Kingdom)

O-067 Copper Nanoclusters Delivered Prenatally Mitigate Neurological, Mitochondrial, and Vascular Deficits in Menkes Disease
François Feillet (Nancy, France)

O-068 Essential single nutritional therapy products for IMDs: Evidence and consensus assessment using modified Delphi method
Nina Stolwijk (Amsterdam, Netherlands)

O-069 Perinatal Lysine Drives Neurodevelopmental, Metabolic, and Neuropathological Outcomes in a Mouse Model of Pyridoxine-Dependent Epilepsy
Hilal Al-Shekaili (Seeb, Oman)

O-070 Vitamin B6-dependent and independent alterations in pyridoxal phosphate-binding protein deficiency identified by large-scale metabolomics profiling
Jolita Ciapaite (Utrecht, Netherlands)

O-071 NAD(P)HX Repair Deficiency Triggers Glial Loss, Excitatory Shift, and Metabolic Collapse in Human Brain Organoids
Yehoshua (Josh) Manor (Ramat Gan, Israel)

O-072 NADHX Repair Deficiency: From Case Reports to Multi-Omics Characterization of Systemic Metabolic and Cellular Perturbations
Najmesadat Seyedkatouli (Luxembourg)

O-073 Clinical spectrum and treatment outcomes in Cobalamin D deficiency: A retrospective multicentre cohort study
Julia Neugebauer (Berlin, Germany)

14:15 - 15:45 **C2: New methods**
Hall 5B
CHAIRS: *Fred Vaz (Amsterdam, Netherlands) and Andrea Dardis (Udine, Italy)*

O-074 Proteome-wide mapping of S-adenosylmethionine interactions reveals novel regulatory targets
Evgeniya Warmer (Zurich, Switzerland)

O-075 UHPLC/HRAM-MS profiling of endogenous urinary GAG-oligosaccharides for rapid mucopolysaccharidoses screening
Marne Hagemeijer (Rotterdam, Netherlands)

O-076 Implementing user-engaged IEM research for accelerating positive societal impact
Cyrille Thinnes (Galway, Ireland)

O-077 An AI-Driven Multilayer Model of Lung Involvement in NeuronopathicGaucher Disease (nGD) and Management Strategies
Ozlem Goker-Alpan (Fairfax, United States)

O-078 Whole-body modelling captures biomarker changes in TYMP deficiency (MNGIE)
Martina Messina (London, United Kingdom)

O-079 An integrative orthology and model organism-based predictive framework for IMD gene discovery
Travis Johnson (Melbourne, Australia)

O-080 Hexose-phosphate remodeling in congenital disorders of glycosylation revealed by tracer metabolomics
Bart Ghesquiere (Leuven, Belgium)

O-081 Structural lipidomics reveals dolichyl phytanates and a compensatory PUFA redistribution in Refsum disease
Frederic Vaz (Amsterdam, Netherlands)

O-082 RAINDROP: leveraging a large fibroblast cohort to establish multi-omics diagnostics of IEM
Vito Zanotelli (Zurich, Switzerland)

15:00 - 16:00 **COFFEE** Halls 4&5

16:00 - 17:30 **Plenary 3 - Physics and Mathematics Enter the World of IEM**
Hall 1
CHAIRS: *Angeles Garcia Cazorla (Barcelona, Spain) and Fanny Mochel (Paris, France)*

- Metabolism Through the Lens of Physics: Transduction Gears, Regulation, and Metabolic Resilience
  Massimiliano Esposito (Luxembourg)
- From basic research to disease mechanisms in lipid cell biology
  Andre Nadler (Dresden, Germany)
- Digital Metabolic Twins and Inherited Metabolic Diseases
  Ines Thiele (Galway, Ireland)

17:30 - 18:30 **Poster Walk**

Posters

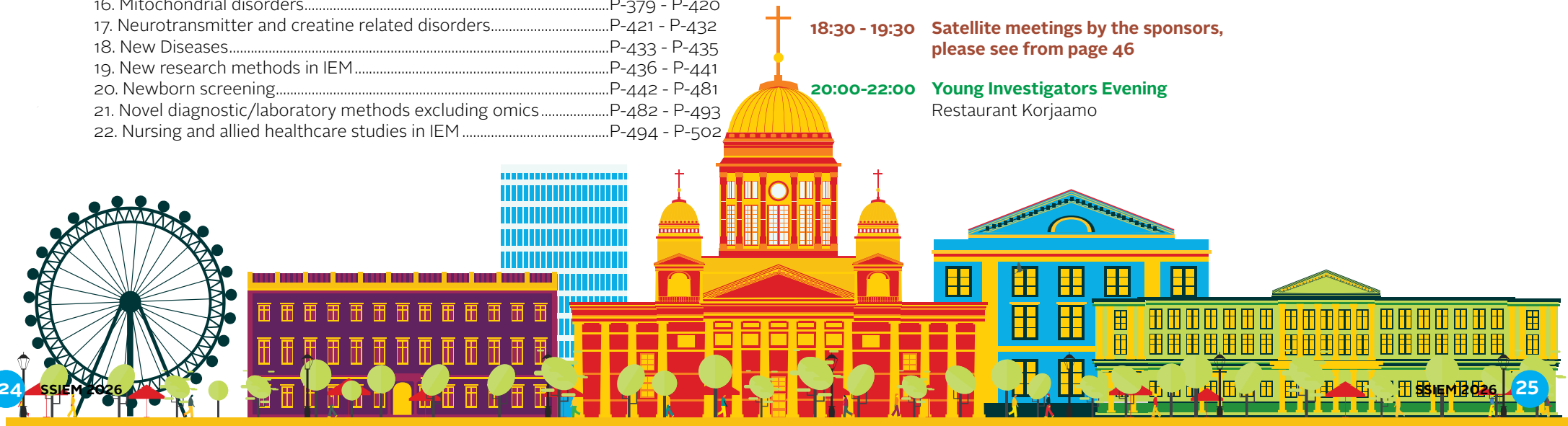
Oral presenter posters O-001-O-132

ePosters

18:00 - 20:00 **GalNet meeting**
Meeting room 203B
(by invitation only)

18:30 - 19:30 **Satellite meetings by the sponsors, please see from page 46**

20:00-22:00 **Young Investigators Evening**
Restaurant Korjaamo



THURSDAY 27 AUGUST

07:00 - 08:00 **EHOD – CBS guidelines group**
Meeting room 104
(by invitation only)

07:30 - 08:30 **Speed Mentoring**
Meeting room 101C
(details to follow)

07:30 - 08:30 **Satellite meetings by the sponsors, please see from page 46**

08:30 - 09:00 **LAB Talk by *Sean Froese (Zurich, Switzerland)***
Meeting room 208

09:00 - 10:30 **Plenary 4 - OMICS data help us understand pathology in metabolic pathways**
Hall 1
CHAIRS: *Philippa Mills (London, United Kingdom) and Pasi Nevalainen (Tampere, Finland)*

- Metabolomics and glycoproteomics to understand sugar metabolism
  Dirk Lefeber (Nijmegen, Netherlands)
- Bileomics – Uncovering New Pathways in Health and Disease
  William Griffiths (Swansea, United Kingdom)
- Global metabolomics and lipidomics implemented in routine diagnostics - examples of added value for unsolved cases
  Katja Elgstoen (Oslo, Norway)

10:30 - 11:00 **COFFEE** Halls 4&5

Parallel abstract sessions D

11:00 - 12:30 **D1: Lysosomal disorders**
Hall 1
CHAIRS: *Roberto Giugliani (Porto Alegre, Brazil) and Kaustuv Bhattacharaya (Dublin, Ireland)*

O-083 Presymptomatic and symptomatic disease stage-associated outcomes of cerliponase alfa treatment in siblings with CLN2 disease
Lena Marie Westermann (Hamburg, Germany)

O-084 Cerliponase alfa for the treatment of CLN2 disease: Combined results from two ongoing observational studies
Angela Schulz (Hamburg, Germany)

O-085 Screening of Mucopolysaccharidoses in High-Risk Patients: Comparison Among Three Different Strategies in Urine
Roberto Giugliani (Porto Alegre, Brazil)

O-086 Long-term real-world outcomes of enzyme replacement therapy in MPS I: a 10-year single-centre cohort study
Karla Cifuentes Uribe (Lyon, France)

O-087 CSF heparan sulfate disaccharides across the phenotypic spectrum of MPS IIIA
Marion Brands (Amsterdam, Netherlands)

O-088 UX111 reduced cerebrospinal fluid heparan sulfate exposure and stabilized or improved functioning in MPS IIIA
Maria Jose De Castro Lopez (Manchester, United Kingdom)

O-089 Integrated Clinical, Biochemical, and Genetic Characterization of α-Mannosidosis in a Spanish Cohort: The α-REVEAL Study
Bélen Peréz (Madrid, Spain)

O-090 Secondary Mitochondrial Dysfunction in Alpha-Mannosidosis: Implications for Disease Pathophysiology and Therapeutic Targeting
Mollie Dewsbury (Liverpool, United Kingdom)

O-091 Oxysterols and PPCS as biomarkers for Niemann-Pick disease type C: differential diagnosis and pitfalls
Cecile Pagan (Lyon, France)

O-092 Long-term assessment of motor function, muscle strength and respiratory function in classic-infantile Pompe disease
Julia Holdorp (Rotterdam, Netherlands)

11:00 - 12:30 **D2: Nurses' and allied healthcare professionals' meeting**
Hall 5C
CHAIRS: *Maureen Evans (Melbourne, Australia) and Mel McSweeney (London, United Kingdom)*

O-093 A single centre functional evaluation and exercise trial in Glycogen Storage Disease Type III patients
Amy Young (London, United Kingdom)

O-094 Divergent Long-Term Outcomes in Two Siblings with Mucopolysaccharidosis Type VI Despite Early Enzyme Replacement Therapy
Anita Inwood (Brisbane, Australia)

O-095 Rehabilitation healthcare follow-up in patients with Cystinosis – outcomes and experiences
Annemarie de Vreugd (Nijmegen, Netherlands)

O-096 Beyond Pediatrics: 40% of Metabolic Patients Are Now Adults
Gustavo Spolador (Sao Paulo, Brazil)

O-097 Metabolic Pharmacist Roles Across the United Kingdom: Current Practice and Service Provision
Antonio Ochoa-Ferraro (Birmingham, United Kingdom)

11:00 - 12:30 **D3: Neurometabolic disorders**
Hall 5B
CHAIRS: *Gajja Salomons (Amsterdam, Netherlands) and Andreas Schulze (Toronto, Canada)*

O-098 Exploring Disease Mechanisms in AADC Deficiency Using 3D Patient-Derived Midbrain Organoids and Metabolomic Profiling
Mari Oppebøen (Oslo, Norway)

O-099 A tool for predict AADC deficiency from genotype: the impact of the characterized AADC variants
Mariarita Bertoldi (Verona, Italy)

O-100 Creatine Homeostasis is Controlled via Multilayered Negative Creatine Feedback Loops
Michael Tropak (Toronto, Canada)

O-101 Altered Creatine Metabolism in Spinal Muscular Atrophy, Jokela Type
Sandra Harjuhaahto (Helsinki, Finland)

O-102 Adult Phenotype and Clinical Outcomes in Grin-Related Developmental and Epileptic Encephalopathies
Natalia Alexandra Julia Palacios (Madrid, Spain)

O-103 Levodopa responsiveness in IEM: a systematic review and case series, beyond neurotransmitter defects
Paula Juliana Rodriguez Soler (Madrid, Spain)

11:00 - 12:30 **D4: Organic acidurias**
Hall 5A
CHAIRS: *Harri Niinikoski (Turku, Finland) and Matthias Baumgartner (Zurich, Switzerland)*

O-104 25 years of newborn screening for glutaric aciduria type 1: an Australian centre's experience
Arthavan Selvanathan (Sydney, Australia)

O-105 Early liver transplant in propionic and methylmalonic aciduria: differences and similarities at follow-up
Benedetta Greco (Rome, Italy)

O-106 Cryo-electron microscopy tomography reveals N-adipyl-D-Ala-D-His ameliorates mitochondrial structural abnormalities in propionic acidemia patient cells
Zachary Freyberg (Pittsburgh, United States)

O-107 Residual propionyl-CoA carboxylase enzymatic activity predicts responsiveness to pseudoexon-skipping antisense oligonucleotide therapy in propionic acidemia
Eriko Totsune (Sendai, Japan)

O-108 Propionic acidemia impairs TCA cycle anaplerosis and cataplerosis in hiPSC-derived brain organoids
Irene González Garnacho (Madrid, Spain)

O-109 Long term Follow-up After Transplantation in Propionic Acidemia: A Retrospective French Cohort Study
Tristan Mekdade (Lausanne, Switzerland)

O-110 Malonyl-CoA decarboxylase catalyzes a new pathway for propionate catabolism in methylmalonic aciduria
Caroline T. Glatthard-Frei (Zurich, Switzerland)

O-111 Energy metabolism is rewired in methylmalonic aciduria
Lisa Tidecks (Zurich, Switzerland)

11:15 - 12:15 **JIMD Editors meeting**
Meeting room 203B
(by invitation only)

12:30 - 13:30 **LUNCH** Halls 4&5

12:30-13:30 **Gyrate Atrophy Meeting (*)**

13:30 - 15:00 **Plenary 5 - Mitochondria Now and Forever**
Hall 1
CHAIRS: *Shamima Rahman (London, United Kingdom) and Peter Freisinger (Tübingen, Germany)*

- Integrating Multi-Omics to Define Mitochondrial Homeostasis
  Patrick Forny (Zurich, Switzerland)
- Mitochondrial Insights into Fatty Acid Oxidation Disorders
  Rikke Olsen (Aarhus, Denmark)
- Metabolic keys to unlock mechanisms in mitochondrial diseases
  Anu Suomalainen (Helsinki, Finland)

15:00 - 15:30 **COFFEE** Halls 4&5

15:30 - 17:00 **Training in IEM: A roundtable discussion**
(paed/adult clin, clin lab, diet, nursing)
Hall 5C
CHAIRS: *Sandra Sirrs (Vancouver, Canada), Risto Lapatto (Helsinki, Finland)*
TALKS BY: *Sandra Sirrs (Vancouver, Canada), Dulce Quelhas (Porto, Portugal), Ann Bowron (Newcastle, United Kingdom), Alice Dianin (Verona, Italy), Anita Inwood (Brisbane, Australia), Risto Lapatto (Helsinki, Finland)*
Discussion

15:30 - 17:00 **Parallel abstract sessions E**

E1: Phenylketonuria
Hall 1
CHAIRS: *Anita MacDonald (Birmingham, United Kingdom) and Francjan van Spronsen (Groningen, Netherlands)*

O-113 Which Phe is it?: Visualising the effect of lifetime Phe on neurocognition in adult PKU-patients
Ellis van Steenis (Groningen, Netherlands)

O-114 MyGut4Phe: An Italian Multicentre Pilot Study on Gut Microbiota Composition in Infants with Hyperphenylalaninaemia
Elvira Verduci (Milan, Italy)

O-115 Redefining the number of true null PAH variants in phenylketonuria and sepiapterin response
Nicola Longo (Los Angeles, United States)

O-116 Sepiapterin Enables Liberalisation of Natural Protein Intake While Maintaining Metabolic Control in Adolescents with Phenylketonuria
Alex Pinto (Birmingham, United Kingdom)

O-117 Re-evaluating Sapropterin Responsiveness in PKU Using Dose Escalation Beyond 20 mg/kg/day
Christine Horne (Chicago, United States)

O-112 Evaluation of CDX-6114, an Acid-Stable Oral Phenylalanine Ammonia Lyase, in a Porcine Model of PKU
Jerry Vockley (Pittsburgh, United States)

O-118 Improved efficacy with sepiapterin in participants with phenylketonuria receiving sapropterin at study screening in AMPLIPHY
Maria Giżewska (Szczecin, Poland)

O-119 Intra-individual variation of in vivo Phenylalanine oxidation using the 13C-Phenylalanine breath test in Phenylketonuria
Sietske Haitjema (Groningen, Netherlands)

O-120 Long-term metabolic control, renal, bone and neuropsychiatric features in early-treated phenylketonuria adults: 5-year study
Yannick Moutapam-Ngamby-Adriaansen (Tours, France)

15:30 - 17:00 **E2: Urea cycle disorders**
Hall 5B
CHAIRS: *Yair Anikster (Ramat Gan, Israel) and Johannes Häberle (Zurich, Switzerland)*

O-121 Newly established hepatic cell model of citrin deficiency uncovers defects in mitochondrial function and ureagenesis
Toni Vukovic (Zurich, Switzerland)

O-122 The experimental structure of yeast N-acetylglutamate synthase (NAGS) sheds light on human NAGS deficiency (NAGSD)
Vicente Rubio (Valencia, Spain)

O-123 Extended Follow-up of DTX301: Safety and Efficacy in Adults with Late-onset Ornithine Transcarbamylase Deficiency (OTCD)
Nathalie Guffon-Fouilhoux (Bron, France)

O-124 Decreased rate of hyperammonemic crises in infants with neonatal-onset OTC deficiency post ECUR-506 administration
Julien Baruteau (London, United Kingdom)

O-125 Preclinical validation of anaplerotic therapies to promote ureagenesis in urea cycle defects
Nathan Breuillard (Zurich, Switzerland)

O-126 Metabolic dysfunction–associated steatotic liver disease and fibrosis in adults with UCDs: a cross-sectional study
Nicola Vitturi (Padova, Italy)

15:30 - 17:00 **E3: Carbohydrate disorders**
Hall 5A
CHAIRS: *Terry Derks (Groningen, Netherlands) and Alessandro Rossi (Naples, Italy)*

O-127 Developmental and transcriptional consequences of Classic Galactosemia
E Naomi Vos (Maastricht, Netherlands)

O-128 Longitudinal multimodal neuroimaging analyses in GLUT1 deficiency syndrome
Agata Maria Capodiferro (Paris, France)

O-129 Liver-specific phospho-proteomic profiling reveals remodeling of insulin signaling in a mouse model of GSD Ia
Margherita Ruoppolo (Napoli, Italy)

O-130 Clinical Impact of Biotin Supplementation on Glucose Homeostasis in Glycogen Storage Disease Type Ia
Sema Kalkan Uçar (Bornova-İzmir, Turkey)

O-131 Impact of early liver transplantation in four young patients with glycogen storage disease Ia
Maria Caprella (Catanzaro, Italy)

O-132 Are the different SGLT2 inhibitors equally effective in treating neutropenia in GSDIb and G6PC3 deficiency?
Maria Veiga-da-Cunha (Louvain, Belgium)

17:00 - 18:00 **Poster viewing**

19:30 - **Networking Evening**
Finlandia Hall

FRIDAY 28 AUGUST

07:30-08:15 **ICIEM IOC Meeting**
Meeting room 104
(by invitation only)

08:15-09:00 **ICIEM SciOrg Meeting**
Meeting room 104
(by invitation only)

09:00 - 10:15 **Late-Breaking News: 5 talks**
Hall 1
CHAIRS: *Ivo Baric (Zagreb, Croatia) and Tomas Honzik (Prague, Czech Republic)*

ANGEL2 deficiency as possible cause of mitochondriopathy – clinical and biochemical findings
Anibh M. Das (Hanover, Germany)

Failure of inflammation resolution defines cerebral X-linked adrenoleuko-dystrophy and is reversed by hematopoietic stem cell transplantation
Aurora Pujol (Barcelona, Spain)

Defining Infantile Niemann-Pick Disease Type C: Plasma Neurofilament Light, Genotype-Phenotype Correlations, and Severity Scoring
Berna Seker Yilmaz (Guildford, United Kingdom)

Ivosidenib Reverses the Neurological Phenotype in Ollier Disease with D-2-Hydroxyglutaric Aciduria: Proof of Concept for Precision Therapy
Diego Martinelli (Rome, Italy)

Structure-based drug discovery of aminoadipate semialdehyde synthase (AASS), a therapeutic target for lysine metabolic disorders
Wyatt W. Yue (Newcastle Upon Tyne, United Kingdom)

10:00 - 10:30 **COFFEE** Halls 4&5

10:45 - 12:15 **Plenary 6 - Looking Forward**
Hall 1
CHAIRS: *Ina Knerr (Dublin, Ireland) and Robin Lachmann (London, United Kingdom)*

Metabolic gene regulation - from molecular mechanism to physiological outcomes
Ville Hietakangas (Helsinki, Finland)

Through the looking-glass; Congenital Disorders of Glycocylation
Eva Morava (New York, United States)

Nutrition for Precision Health: Integrating Genetics into the Future of Personalized Nutrition
Holly Nicastro (Bethesda, United States)

12:15 - 13:00 **Komrower Lecture**
Hall 1
CHAIR: *Manuel Schiff (Paris, France)*

Protein, Pathways and Personalised Nutrition, *Marjorie Dixon (London, United Kingdom)*

13:00 - 13:20 **Closing session**
Hall 1
CHAIRS: *Manuel Schiff (London, United Kingdom) and Risto Lapatto (Helsinki, FInland)*

LBN Awards, Posters

Summary

SSIEM 2027 Dublin presentation

13:20 - 14:00 **LUNCH** (Main Venue lobby area)

PLENARY SPEAKERS


Annita Achilleos, Nicosia, Cyprus


Charlotte Ellerton, London, United Kingdom


Massimiliano Esposito, Luxembourg


Patrick Forny, St Louis, United States


William Gahl, Bethesda, United States


William Griffiths, Swansey, United Kingdom


Ville Hietakangas, Helsinki, Finland


Juha Kere, Stockholm, Sweden


Dirk Lefeber, Nijmegen, Netherlands


Tippi MacKenzie, San Francisco, United States


Eva Morava, New York, United States


Andre Nadler, Dresden, Germany


Holly Nicastro, Bethesda, United States


Rikke Olsen, Aarhus, Denmark


Frances Platt, Oxford, United Kingdom


Katja Elgstøen, Oslo, Norway


Anu Suomalainen, Helsinki, Finland


Ines Thiele, Galway, Ireland

SPECIAL SESSIONS

POSTER WALK
Wednesday, 26 August at 17:30-18:30 (Hall 4&5)

Posters will be available for viewing throughout the Symposium. We are asking the presenters to be near their poster during the Poster Walk to answer your questions. This offers an unrivalled platform for the exchange of cutting-edge research and ideas. By interacting with the diverse range of posters, you will have the opportunity to explore emerging trends, new methodologies and ground-breaking discoveries, broadening your intellectual horizons.

The Poster Walk provides a unique opportunity to engage with innovative research and fresh perspectives. As you interact with the poster presenters, you will discover emerging trends, explore advanced methodologies, and gain insight into significant new discoveries. This experience is designed to expand your knowledge and inspire meaningful intellectual growth.

The presenter for odd-numbered posters will be present from 17:30 to 18:00 and for even-numbered posters from 18:00 to 18:30.

YSSIEM INAUGURAL MEETING
Tuesday 25 August at 19:30-20:30 (Hall 5A)

We are super excited to host the very first meeting of the Young SSIEM (YSSIEM)! Inspired by our theme, we want to build a supportive community for our younger members—one that will grow to be just as impactful and helpful to our society as the Dietitians' and Adult Metabolic Physicians' groups. You don't need to be an SSIEM member yet to attend, so if you are currently in training, we highly encourage you to jump in. Stay tuned, more details are coming soon!

SSIEM, METABERN AND ISNS SESSION ON NEWBORN SCREENING
Tuesday 25 August at 16:00-17:45 (Hall 101C)

Newborn screening is a true success story! This year, we are diving into screening for lysosomal diseases. Top experts will cover everything from clinical and ethical aspects to technical details, followed by an open discussion session. Come share your thoughts!

ENSURING PATIENT ACCESS TO INNOVATION IN RARE DISEASES
Wednesday 26 August at 14:00-16:00 (Hall 5B)

Turning scientific innovation into sustainable treatments remains a critical challenge for the rare disease community. Join senior regulators, patient advocates, researchers and funders for an interactive session exploring emerging regulatory pathways, innovative economic models, patient-centered and real-world solutions to enabling treatment access.

TRAINING OF HEALTHCARE PROFESSIONALS IN IEM
Thursday 27 August at 15:30-17:00 (Hall 5C)

What does training look like across the globe? In this session, we will hear diverse perspectives from various healthcare professionals, including nurses, dietitians, lab scientists, geneticists, and clinicians. After a few quick intro presentations, we will open the floor for a discussion on training requirements and future steps.

SPEED MENTORING

Speed Mentoring Sessions provide young professionals in IEM a unique chance to have one-on-one discussions with experienced researchers, clinicians and nutritionists from various specialties. In these 7-minute individual interviews with different mentors, participants can gain valuable advice on career planning, navigating career transitions and promotions, handling challenging work situations, and more. This event offers a supportive professional environment for attendees to interact with mentors who have diverse scientific backgrounds.

WHERE	Messukeskus
WHEN	Wednesday, August 26, 2026 , from 7:30 AM to 8:30 AM Thursday, August 27, 2026, from 7:30 AM to 8:30 AM
HOW	Rolling system with different mentors
WHO	Clinicians, scientists and laboratory specialists in training
REGISTRATION	Online

COFFEE WITH THE EDITORS

Wednesday 26 August at 10:00-10:40 (Members' Lounge)

In Finland we say: "Let's go for a coffee!" which means Let's talk! This is your possibility to interact directly with the JIMD Editorial team. This will be an informal coffee moment, designed to foster a more personal and collaborative atmosphere, encouraging open dialogue and the exchange of ideas. And you get your coffee as well.

All participants are welcome.

NETWORKING PROGRAMME

WELCOME RECEPTION

Tuesday 25 August at 18:30 - 21:00
VENUE: Messukeskus Helsinki, Halls 4&5

The Welcome Reception offers a unique opportunity to connect with fellow attendees, speakers, and sponsors. This opening gathering serves as a catalyst for meaningful conversations, fostering new relationships and paving the way for potential collaborations. The exchange of ideas during this time can enrich your event experience and create lasting connections that continue well beyond the event itself.

We are delighted to invite you to join us for a welcome cocktail, a gift from the City of Helsinki, designed to set a warm and inviting tone for the days ahead. This special evening promises an atmosphere of camaraderie and inspiration, marking the perfect start to an engaging event.

MEMBERS' LOUNGE

This is new to the SSIEM. We offer our members a peaceful area for relaxing and networking with coffee and tea. The lounge is located in the Main venue lobby (see the Exhibition and Poster area floor plan on page 44-45). It will be open during the Symposium hours. If unsure, you can check your membership status across the corridor by the SSIEM lounge.

YOUNG INVESTIGATORS EVENING

Wednesday 26 August at 20:00 - 22:00
VENUE: Restaurant Korjaamo, Töölönkatu 51, Helsinki

Continuing from Tuesday's meeting this networking event gives you a chance to meet your peers in a relaxed atmosphere. Young scientists and professionals from every discipline within SSIEM, as well as those who appreciate engaging with emerging members, are warmly invited to attend the Young Investigator Evening.

This special event is designed to encourage meaningful connections and promote the exchange of innovative ideas among participants. While there is no age restriction and all are welcome, the central purpose of this unique networking opportunity is to empower and support the next generation of SSIEM members, fostering their growth within the community.

Complimentary drinks and snacks will be provided for the first 150 guests.

NETWORKING EVENING

Thursday 27 August starting at 19:30
VENUE: Finlandia Hall, Mannerheimintie 13/Karamzininkatu , Helsinki

As the event draws to a close, we warmly invite you to join us for a memorable closing dinner. This special occasion is designed to celebrate the accomplishments, partnerships, and meaningful discoveries that have emerged throughout our time together. Your participation enhances the sense of collective pride and unity, making this evening a fitting tribute to all that we have achieved as a community.

The event takes place in Finlandia Hall designed by the internationally best known Finnish architect Alvar Aalto. The venue has been recently renovated and was just added to the list of UNESCO World Heritage Sites.

In addition to networking, enjoying Finnish food and dancing the night away, you can also visit the special exhibition on Finnish culture, nature and design, "Visions of Alvar Aalto", at any time you like during the evening.

TICKETS: 45 € for delegates, 90 € for industry and accompanying persons

GENERAL INFORMATION

SYMPOSIUM DATES
24 – 28 August 2026

WEBSITE
www.ssiem2026.org

SYMPOSIUM VENUE
Messukeskus
Messuaukio 1 (Main Entrance)
00520 Helsinki, Finland

SYMPOSIUM PRESIDENT
Dr. Risto Lapatto
Helsinki University Hospital – HUS

PROFESSIONAL CONGRESS ORGANISER
Confedent International Ltd
+358 50 4644757
ssiem2026@confedent.fi
www.confedent.fi

EXHIBITION MANAGEMENT AND SPONSORING
Society for the Study of Inborn Errors of Metabolism (SSIEM)
Ralph Kerschbaumer – Corporate Liaison Officer
PO Box 3375, South Croydon, CR2 1PN
United Kingdom
phone +43 512 890438
corporateliaison@ssiem.org
www.ssiem.org

ACCREDITATION
An application has been made to the UEMS EACCME® for CME/CPD accreditation of this event.

ARRIVAL
Arrive early to the Symposium venue to pick up your badge and Symposium material. Our registration desk will open on Monday 24 August.

GENERAL INFORMATION

BADGE
All participants must collect a name badge from the registration desk at the Symposium venue's main entrance.
To ensure security, wearing the badge is mandatory throughout the event, including at Networking events.

CLOAKROOM
There is a cloakroom available during the Symposium.

WIFI
WiFi service will be available during the Symposium.
Access the SSIEM 2026 network. Supported by SENTYNL THERAPEUTICS, INC.

REGISTRATION DESK OPENING HOURS

Monday	12:00 - 19:00
Tuesday	08:00 - 19:00
Wednesday	08:00 - 19:00
Thursday	08:00 - 17:00
Friday	08:00 - 14:00

EXHIBITION HOURS

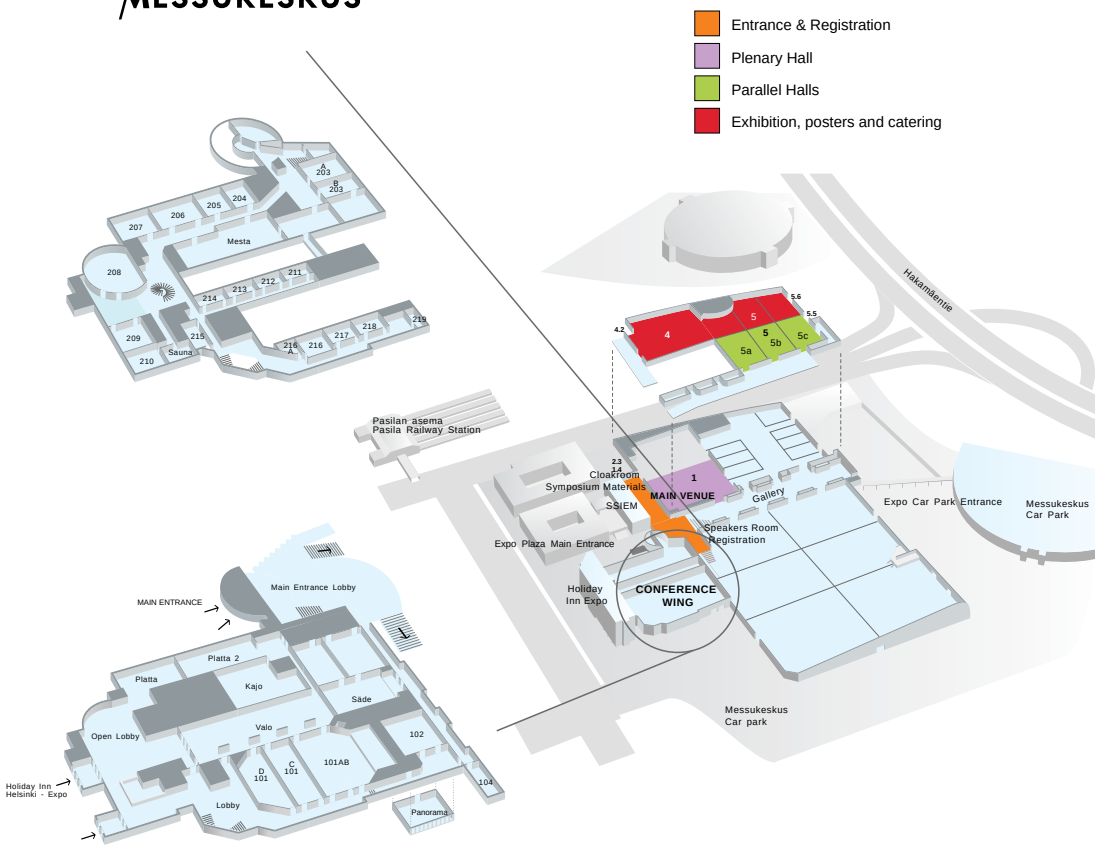
Tuesday	12:00 - 18:00
Wednesday	08:30 - 17:30
Thursday	08:30 - 15:30
Friday	09:00 - 13:00

SPEAKERS ROOM
Presentations held in Halls 1, 5A, 5B, 5C and Meeting Room 101AB as well as LAB talk presentations in Meeting Room 208 and presentations for Tuesday session on Newborn screening in Meeting Room 101C should be brought to the Speakers Room (located next to our Registration Desk) the day before the presentation time. If you arrive to the Symposium on the day of your presentation, please bring your slides to the Speakers Room technicians latest one hour before the presentation.

The Speakers Room will be open from 16:00 to 19:00 on Monday, 24 Aug and from 7:30 to 16:30 on Tue, Wed and Thu.

Other presentations should be taken directly to the meeting room on a USB.

MESSUKESKUS



SYMPOSIUM REGISTRATION FEES

Registration Category	Early Bird until 23 June	Full Price from 24 June
SSIEM Member	250 €	400 €
Non-Member	650 €	800 €
SSIEM Member Student/Trainee**	100 €	250 €
Non-Member Student/Trainee**	350 €	500 €
SSIEM Member Dietitian/Nurse/Patient Group (Charities Employee)	100 €	250 €
Non-Member Dietitian/Nurse/Patient Group (Charities Employee)	350 €	500 €
SSIEM Member Single Day Ticket	75 €	155 €
Non-Members Single Day Ticket	200 €	300 €
Exhibitor registration only	250 €	250 €
Group registration fee (per person)	550 €	700 €

All registration fees are in Euro and include VAT 25,5%.

**Proof of being a Student or Trainee will be requested. Please note that working doctors with an MD are not eligible and research scientists with a PhD are not eligible for this category.

WHAT'S INCLUDED?

Full Delegate* fees

- Admission to all oral and poster sessions and exhibition area
- Scientific Programme and Symposium bag
- Access to the online abstract book
- Coffee, tea and 4 lunches during Symposium breaks
- Welcome Reception
- 5 day free travel pass for all public transportation (local trains, buses, trams and ferry to Suomenlinna Fortress Island) in Helsinki metropolitan area including the Helsinki-Vantaa Airport
- Official Attendance certificate.

*SSIEM Members, SSIEM Member Dietitians/Nurses/Patient Group (Charities Employee), SSIEM Member Students/Trainees, Non-Members, Non-Member Dietitians/Nurses/Patient Group (Charities Employee and Non-Member Students/Trainees, Group registrations.

Single-day Tickets

- Admission to all oral and poster sessions and exhibition area on the day of the ticket
- Scientific programme on the chosen day and Symposium bag
- Access to the online abstract book
- Coffee, tea and lunch during Symposium breaks of the day of the ticket.

Single day tickets do not allow access to social events. No more than two consecutive single day tickets can be purchased by one delegate.
NOTE! Students/Trainees will need to upload a letter from the Head of Department or a senior authority of their institution (on an official letter-headed paper) confirming their Student/Trainee status.

Exhibitor Registration Only

- Admission to the exhibition area
- Admission to industry symposia
- Coffee, tea and lunch during Symposium breaks
- Welcome Reception

Networking Evening and Social Programme are to be paid for additionally.

Participation in Networking Events

Activity	Participant	Accompanying person	Industry
Welcome Reception	Complimentary	Complimentary	Complimentary
Young Investigators Evening	Complimentary	–	–
Networking Evening	45 €	90 €	90 €

Travel

Registered delegates get a free 5-day eTicket for all public transportation in the Helsinki area (including the Airport).
A personal activation code with instructions has been sent to each participant by email in the beginning of August. The eTicket can only be activated in Finland and it will be valid during the conference days, from Monday 24 August to Friday 28 August.

SSIEM 2026 APP

During the Symposium days, you can access features like:

- Scientific programme
- Networking events
- Satellite symposia
- Faculty list
- Abstract texts
- Create your own daily schedule
- Explore partner and exhibitor profiles
- Stay updated with the latest news
- Chat with other attendees

After downloading the app, do not forget to enable push notifications to stay up-to-date on the latest news!

DOWNLOAD THE APP

SPONSOR SECTION

SPONSORS LIST

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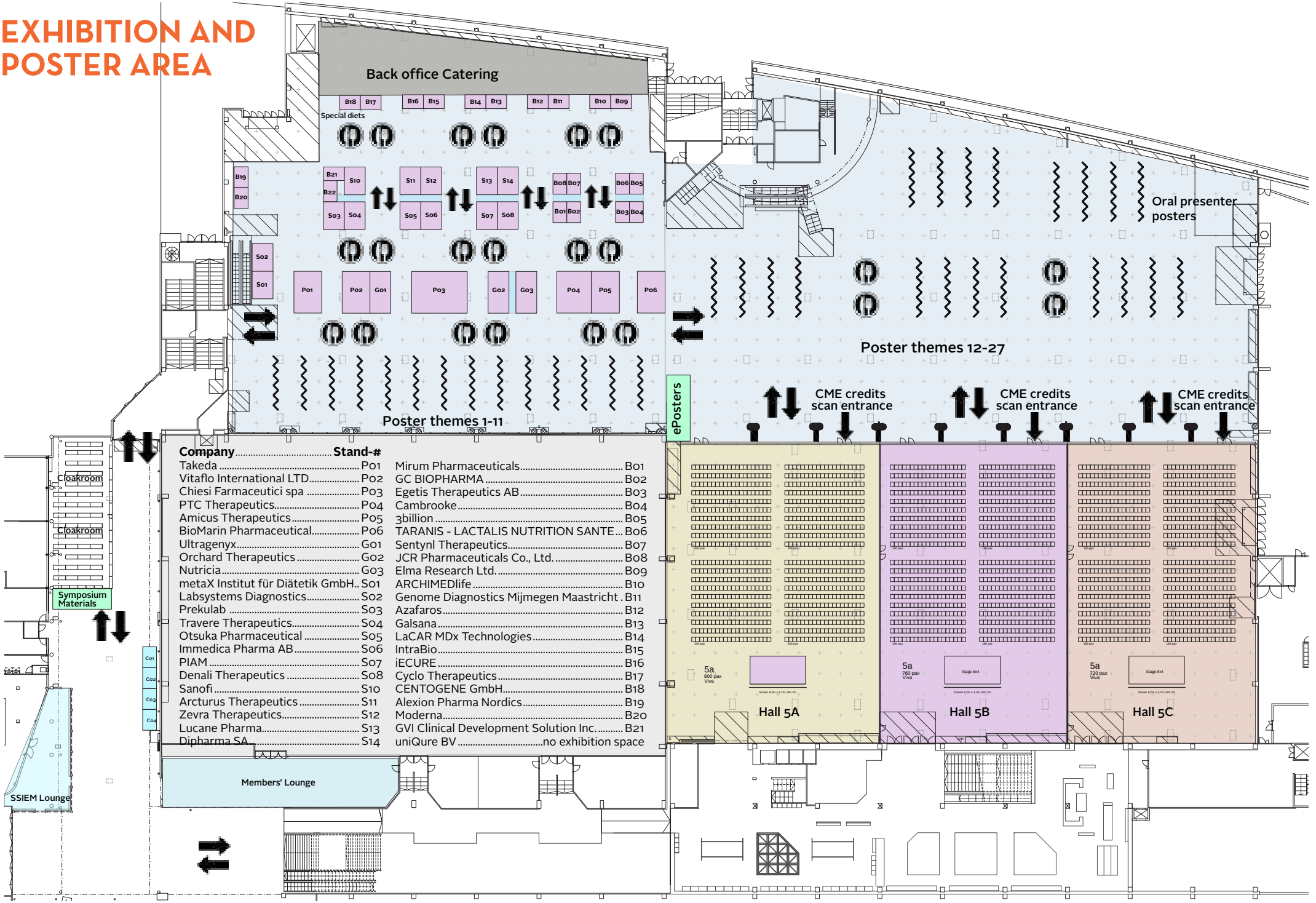
EXHIBITION AND POSTER AREA



Company	Stand-#
Takeda	P01
Vitaflo International LTD	P02
Chiesi Farmaceutici spa	P03
PTC Therapeutics	P04
Amicus Therapeutics	P05
BioMarin Pharmaceutical	P06
Ultragenyx	G01
Orchard Therapeutics	G02
Nutricia	G03
metaX Institut für Diätetik GmbH	S01
Labsystems Diagnostics	S02
Prekulab	S03
Travere Therapeutics	S04
Otsuka Pharmaceutical	S05
Immedica Pharma AB	S06
PIAM	S07
Denali Therapeutics	S08
Sanofi	S10
Arcturus Therapeutics	S11
Zevra Therapeutics	S12
Lucane Pharma	S13
Dipharma SA	S14
Mirum Pharmaceuticals	B01
GC BIOPHARMA	B02
Egetis Therapeutics AB	B03
Cambrooke	B04
3billion	B05
TARANIS - LACTALIS NUTRITION SANTE	B06
Sentynl Therapeutics	B07
JCR Pharmaceuticals Co., Ltd.	B08
Elma Research Ltd.	B09
ARCHIMEDlife	B10
Genome Diagnostics Mijmegen Maastricht	B11
Azafaros	B12
Galsana	B13
LaCAR MDx Technologies	B14
IntraBio	B15
iECURE	B16
Cyclo Therapeutics	B17
CENTOGENE GmbH	B18
Alexion Pharma Nordics	B19
Moderna	B20
GVI Clinical Development Solution Inc.	B21
uniQure BV	no exhibition space

SATELLITE SYMPOSIA

SSIEM 2026 gratefully acknowledges the following companies for hosting symposia during the meeting.

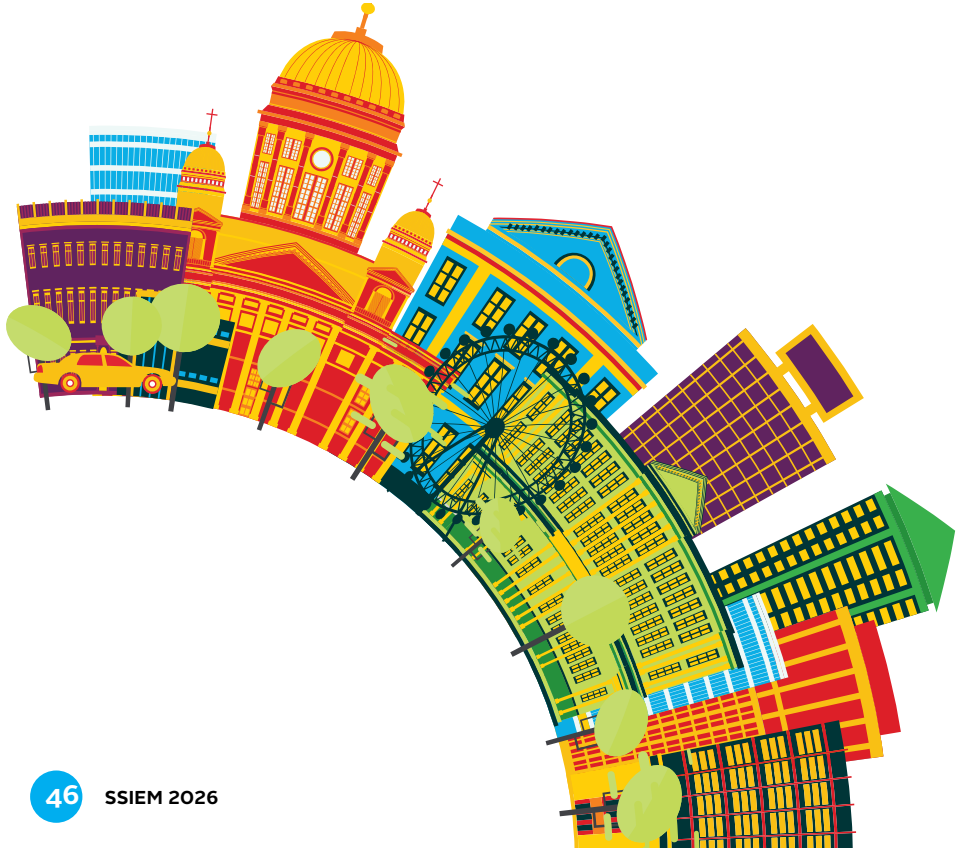
TUESDAY, AUGUST 25TH, 2026
12.45-13.45

Ultragenyx **Hall 5A**

Challenges of current treatment in LC-FAOD and GSDIa

Case series to illustrate and highlight the challenges and opportunities in managing LC-FAOD and GSDIa

Ute Spiekerkötter (Freiburg, Germany)
Sara Guillén (Mexico City, Mexico)
Terry Derks (Groningen, Netherlands)
Malaya Mount (West Hartford, United States)



JOIN US
Tuesday
12:45

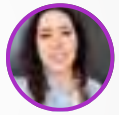
ultragenyx

SATELLITE SYMPOSIUM
Tuesday, 25th August / 12:45 - 13:45 / Hall 5A

Challenges of Current Treatment in LC-FAOD and GSDIa

Symposium will use cases to illustrate and highlight the challenges and opportunities in managing LC-FAOD and GSDIa

Prof Dr. Ute Spiekerkötter
Medical Dir., Dept. General Pediatrics, Adolescent Medicine and Neonatology
Universitätsklinikum Freiburg, Germany



Sara Guillén, MS, RD, CSP
Metabolic Dietitian, Int'l Committee Chair Member of GMDI
Instituto Nacional de Pediatría, Mexico City, Mexico

Malaya Mount, MS, RD, CND
Metabolic Dietitian
Connecticut Children's Hospital, USA

Prof Dr. Terry Derks
Pediatrician and Consultant in Metabolic Medicine
University Medical Center Groningen (UMCG)
University of Groningen, The Netherlands

Visit Our Booth #G1
Learn about our programs and clinical trials

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MRCNP-DTX401-00028, 6/2026

TUESDAY, AUGUST 25TH, 2026
12.45-13.45

Orchard Therapeutics **Hall 5B**

Realising the vision for MLD

CHAIR: *Erik Eklund (Lund, Sweden)*
MODERATOR: *Pascale Vincendon (Orchard Therapeutics France)*

Ex-vivo HSC-GT for MLD: the sooner the better (long term efficacy data and sibling analysis)
Valeria Calbi (Milan, Italy)

Global MLD NBS status & Nordic MLD NBS to-date
Erik Eklund (Lund, Sweden)

Patient association voice on NBS and support needed for MLD patients and families
Michael Scholz (Neustadt, Germany)

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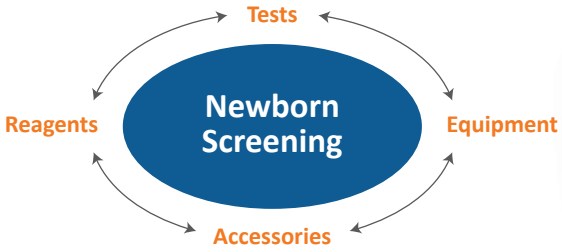

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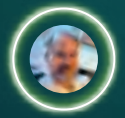
Orchard Therapeutics

SATELLITE SYMPOSIUM

REALISING THE VISION FOR MLD

CHAIR & SPEAKER



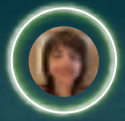
Erik **Eklund** - *Skåne University Hospital*
Global MLD NBS status & Nordic MLD NBS to date

FEATURED SPEAKERS

Valeria **Calbi** - *IRCCS San Raffaele Hospital*
Ex-vivo HSC-GT for MLD: the sooner the better (long-term efficacy data and sibling analysis)

Michael **Scholz** - *ELA Germany e.V.*
Patient association voice on NBS and support needed for MLD patients and families

MODERATOR



Pascale **Vincendon** - *Orchard Therapeutics France*

TUESDAY, AUGUST 25, 2026 12.45-13.45 **HALL 5**

EM-NoP-2600024 – date of preparation July 2026

AUTOLOGOUS HSC GENE THERAPY

Orchard therapeutics
A Kyowa Kirin Company

TUESDAY, AUGUST 25TH, 2026
12.45-13.45

BIOMARIN

BioMarin Pharmaceutical Hall 5C

Unlocking new possibilities in adolescents with PKU

MODERATOR: *Ania C. Muntau (Hamburg, Germany)*

Why adolescents are at critical window of intervention
Erika Vucko (Chicago, United States)

Phase 3 PEGASUS outcomes: Impact on Phe levels and diet
Julia B. Hennermann (Mainz, Germany)

Transforming care for adolescents: Real-world strategies
Iris Scala (Naples, Italy)

A fireside chat leaning into lived experience of PKU
An individual with PKU & Ania C. Muntau (Hamburg, Germany)

PREKULAB

Freer daily PKU diet

LNAA

A freer daily PKU diet?

This is what LNAA NeoPhe and PreKUnil can contribute to.

Visit us at booth S03

SATELLITE SYMPOSIUM

Unlocking new possibilities in adolescents with PKU

Why early intervention and normal Phe (30–120 µmol/L) matter, PEGASUS and beyond

Tuesday, 25 August 2026
12:45-13:45 hrs | Hall 5-C (floor 0)

Complimentary lunch boxes will be provided by SSIEM

Join this symposium moderated by Dr. Muntau for insights into:

- **Why adolescents are at critical window of intervention** - Erika Vucko
- **Phase 3 PEGASUS outcomes: Impact on Phe levels and diet** - Julia B. Hennermann
- **Transforming care for adolescents: Real-world strategies** - Iris Scala
- **A fireside chat leaning into lived experience of PKU** - An individual with PKU & Ania C. Muntau

Faculty

Ania C. Muntau, MD
Hamburg, Germany

Erika Vucko, APP
Chicago, IL, USA

Julia B. Hennermann, MD
Mainz, Germany

Iris Scala, MD
Naples, Italy

An individual with PKU

This symposium is intended for Healthcare Professionals only and will include information on BioMarin medicines.
This symposium is organised and funded by

BIOMARIN

COM-ET-2771 – July 2026

Scan here to see the PALYNZIQ® (pegvaliase) Abbreviated Prescribing Information

TUESDAY, AUGUST 25TH, 2026
12.45-13.45

Takeda **101ab**

Challenges in Gaucher disease: More than meets the bone

CHAIR: *Timothy Cox (Cambridge, United Kingdom)*

The bone paradox in Gaucher disease
Timothy Cox (Cambridge, United Kingdom)

Bone outcomes in Gaucher disease – What's the verdict?
Timothy Cox (Cambridge, United Kingdom)

The invisible patient: Bone disease in women with Gaucher disease
Ozlem Goker-Alpan (Fairfax, United States)

Long-term bone outcomes in Gaucher disease: Evidence from real-world practice
Maurizio Scarpa (Udine, Italy)

Panel discussion – What must change and who is responsible?
Timothy Cox (Cambridge, United Kingdom),
Ozlem Goker-Alpan (Fairfax, United States),
Maurizio Scarpa (Udine, Italy)

Symposium closing remarks
Timothy Cox (Cambridge, United Kingdom)

We invite you to a satellite symposium at SSIEM 2026

NOT ALL EVIDENCE IS EQUAL:
INFORMING FABRY TREATMENT CHOICES

August 27, 2026
07:30-08:30 am
EEST

Room 101 AB
Conference Centre (Kokoussiipi)
Helsinki, Finland

Sandro Feriozzi, MD
Nephrology Unit
University Campus Bio-Medico
Rome, Italy

Patrício Aguiar, MD, PhD
School of Medicine, University of Lisbon, Reference Center for Inherited Metabolic Disorders, ULS Santa Maria, Portugal

Guillem Pintos-Morell, MD, PhD
Vall d'Hebron Research Institute
Barcelona, Spain

JOIN US at this symposium where our experts will explore how real-world evidence and long-term registry data can inform Fabry disease management, including

- long-term outcomes with agalsidase alfa
- key considerations in therapy selection
- meaningful endpoints

PROTECT EACH STEP,
FOR THE LONG-TERM

Takeda

WEDNESDAY, AUGUST 26TH, 2026
07.30-08.30

Chiesi Farmaceutici spa Hall 5A

Flexible by design: redefining treatment possibilities with Pegunigalsidase alfa

MODERATOR: *Christina Lampe (Giessen, Germany)*

PEGylation at the core: how molecular engineering shapes protein properties
Diego Fornasari (Milan, Italy)

Long term clinical evidence with pegunigalsidase alfa
Patrício Aguiar (Lisbon, Portugal)

Flexible care in real life: experience with dosage regimens of pegunigalsidase alfa
Christina Lampe (Giessen, Germany)

Mirum Pharmaceuticals Hall 5B

Voices that Matter
Lived Experiences With Cerebrotendinous Xanthomatosis

Learn about CTX from a leading expert in lipid metabolism and rare disease
P. Barton Duell (Portland, United States)

Hear first-hand perspectives from those affected by CTX
Evalyn shares her personal journey to diagnosis and what living with CTX means to her
Bobbi shares her experiences through diagnosis and management as a parent and caregiver to her two children living with CTX

SCIENTIFIC SYMPOSIUM

POWERING NEXT-GENERATION LYSOSOMAL STORAGE DISORDERS'

CARE, RESEARCH AND INNOVATION LEVERAGING DATA, DIGITAL TECHNOLOGIES AND AI

Thursday, August 27, 2026
07.30 - 08.30 | Room: Hall 5B

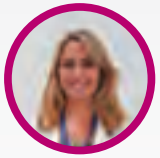
Bringing together clinical, patient, innovation and policy experts this session will explore how data, digital health and AI can enhance LSDs' care.
Come meet our panel of leading voices in the field:

Maurizio Scarpa

Coordinator of metabERN; Coordinator of the Regional Coordinating Centre for Rare Diseases at the University Hospital of Udine.

Kim Angel

Executive Director of International MPS Network



María Del Mar Mañú Pereira

Head of Research Lab in rare anemia disorders at the Vall d'Hebron Research Institute in Barcelona, Senior researcher Scientific Director of EuroBloodNet & Coordinator of the European platforms for ENROL and RADeep

Alberta Spreafico

Digital Health and Health Policy Expert; Senior Vice President of Health Innovation at EVERSANA; Adjunct Professor at the University of Pavia; Founding President Innovation for Global Health Institute

in collaboration with

This symposium is intended exclusively for healthcare professionals.
ALL_26_1296 | Date of preparation: June 2026
22236-21.05.2026

WEDNESDAY, AUGUST 26TH, 2026
07.30-08.30

Otsuka Pharmaceutical Hall 5C

PKU & the Brain - Exploring Neurodevelopment, Symptoms, and Dietary Impact

CHAIR: *Brighid Imperiale (Otsuka Pharmaceuticals Co., Ltd., United States)*

Brain Development and PKU
Deborah Bilder (Salt Lake City, United States)

Review of Neuropsychiatric Symptoms in PKU
Shawn E. Christ (Columbia, United States)

Q&A Fireside Chat
Brighid Imperiale (Otsuka Pharmaceuticals Co., Ltd., United States), Deborah Bilder (Salt Lake City, United States), Shawn E. Christ (Columbia, United States)

Sanofi 101ab

The Diagnostic Revolution in Inherited Metabolic Diseases: From Suspicion to Precision

CHAIR: *Dawn Laney (Atlanta, United States)*

The Changing Landscape of Inherited Metabolic Disease Diagnosis
Dawn Laney (Atlanta, United States)

From Clinical Suspicion to Molecular Diagnosis
Nancy Chien (Taipei, Taiwan)

Closing the Diagnostic Gap in Lysosomal Storage Disorders
Mehdi Namdar (Zurich, Switzerland)

PKU & the Brain

Exploring Neurodevelopment, Symptoms, and Dietary Impact

OUR SPEAKERS

Please join us:

Wed, Aug 26, 2026
7:30-8:30AM

Messukeskus, Helsinki, Hall 5C

Dr. Deborah Bilder

Dr. Shawn Christ

Otsuka

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OPDC-MED-SSIEM - 2026-JUNE-04

sanofi

We are committed to *closing the gaps in rare care*

Come visit us at *booth S10* to learn more

We strive to:

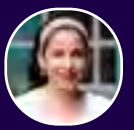
- Break down the barriers to timely and accurate rare disease diagnoses
- Discover and develop new, innovative treatments that improve real-world outcomes
- Advocate for equitable access to medicines
- Elevate the voices of people living with rare diseases and support them across their lifelong journey

Nick
MPS I, US

George
Fabry Disease, India

Shu
Pompe Disease, Japan

JJ
ASMD, US



Ailin
Gaucher Disease, Cuba

Sanofi
MAT-GLB-2604016-1.0 - 07/2026

WEDNESDAY, AUGUST 26TH, 2026
18.30-19.30

PTC Therapeutics **Hall 5A**

Evolving the Standard of Care for PKU

CHAIR: *Ania C. Muntau (Hamburg, Germany) - Alberto B. Burlina (Padua, Italy)*

Defining the New Standard: Current Evidence and Evolving Practice
Ania C. Muntau (Hamburg, Germany) - Alberto B. Burlina (Padua, Italy)

A Dietitian's Perspective: Redefining What's Possible
Annemiek Van Wegberg (Groningen, The Netherlands)

Translating Evidence Into Clinical Practice Part 1
Ania C. Muntau (Hamburg, Germany)

Translating Evidence Into Clinical Practice Part 2
Erika R. Vucko (Chicago, United States)

Looking forward to meeting you at booth S01

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Sephience is indicated for the treatment of hyperphenylalaninaemia (HPA) in adult and paediatric patients with phenylketonuria (PKU).

In compliance with Finnish regulations, this symposium is intended exclusively for healthcare professionals with prescribing rights.

Sephience™: Evolving the Standard of Care for PKU[1]

Lower Phe levels. Better outcomes. It's time to rethink what's possible in PKU care.[1,2]*

Attend the Sephience symposium to hear leading PKU experts discuss the impact of Sephience in their clinical practice. Through real-world case studies and the latest data, the symposium will explore how achieving lower Phe levels may help support meaningful improvements in overall quality of life.[2]

Wednesday, 26 August 2026
18:30-19:30 EEST
Hall 5A

DISCOVER MORE.
Visit our booth to discover what's possible for PKU patients in your clinic.[1]

Sephience™ sepiapterin

Actor portrayals.

*In Part 2 of the AMPLIPHY study, blood Phe reduction from baseline to Weeks 3–4 was significantly greater with sepiapterin than with sapropterin in the primary analysis set (LS mean difference, −180.4 µmol/L), corresponding to a 70% greater reduction.

LS=least squares; Phe=phenylalanine; PKU=phenylketonuria.

REFERENCES: 1. Gizewska M, Inwood A, Tyčová R, et al. Efficacy and safety of sepiapterin versus sapropterin in patients with phenylketonuria: results from the phase 3, randomized, crossover, open-label, active-controlled AMPLIPHY trial. *Metabolism*. 2026;178:156513. **2.** van Spronsen FJ, Peters H, Margvelashvili L, et al. Effect of long-term sepiapterin treatment on dietary phenylalanine tolerance in patients with phenylketonuria: interim results from the phase 3 APHENITY Extension Study. *Genet Med*. 2026;28(4):101683.

Abbreviated Prescribing Information

Sephience 250 mg and 1000 mg oral powder in sachet. INDICATION: Sephience™ (active ingredient: sepiapterin) is indicated for the treatment of hyperphenylalaninaemia (HPA) in adult and paediatric patients with phenylketonuria (PKU). **POSOLOGY AND ADMINISTRATION:** Treatment with Sephience must be initiated and supervised by a physician experienced in the treatment of PKU. Sephience should be administered orally once daily. The maximum recommended dose is 60 mg/kg/day. The recommended dose for patients aged 0 to < 6 months is 7.5 mg/kg/day, 6 to < 12 months is 15 mg/kg/day, 12 months to < 2 years is 30 mg/kg/day, and ≥ 2 years is 60 mg/kg/day. Sephience should be administered once daily with a meal, using mg/kg dosing. Sachets should be mixed in water, apple juice, or a small amount of soft food such as apple sauce and jams. Once mixed, the dose should be administered immediately. If not administered immediately, the liquid and soft food mixtures can be administered within 6 hours or 24 hours, when stored at room temperature (below 25 °C) or in a refrigerator (2 °C - 8 °C), respectively. Sephience oral powder may be administered via an enteral feeding tube 6 Fr or 8 Fr after mixing with water. For more details see full prescribing information. **CONTRAINDICATIONS:** Hypersensitivity to the active substance or to any of the excipients. **SPECIAL WARNINGS AND PRECAUTIONS FOR USE:** Patients treated with Sephience should undergo regular clinical assessments to align with their health care provider on appropriate dietary Phe intake (such as monitoring of blood Phe and tyrosine levels and nutritional intake). Co-administration of sepiapterin with DHFR inhibitors (e.g. trimethoprim, methotrexate, pemetrexed, pralatrexate, and trimetrexate) may require more frequent monitoring of blood Phe levels. Sephience 250 mg and Sephience 1000 mg oral powder, each sachet contains 400 mg and 1600 mg of isomalt, respectively. Patients with rare hereditary problems of fructose intolerance should not take this medicinal product. **INTERACTIONS:** Caution and more frequent monitoring of blood Phe are recommended when Sephience is co-administered with SR inhibitors, such as sulphasalazine or sulphamethoxazole. Inhibition of DHFR could potentially result in lower BH_4 concentration. Caution and more frequent monitoring of blood Phe are required in patients when sepiapterin is co-administered with a DHFR inhibitor, such as trimethoprim, methotrexate, pemetrexed, pralatrexate, and trimetrexate. Caution is recommended during concomitant use of Sephience with medicinal products that cause vasodilation by affecting nitric oxide (NO) metabolism or action, including classical NO donors (e.g. glyceryl trinitrate [GTN], isosorbide dinitrate [ISDN], sodium nitroprusside [SNP], and molsidomin), phosphodiesterase type 5 (PDE 5) inhibitors (e.g. sildenafil, vardenafil, or tadalafil), and minoxidil. Caution should be exercised when prescribing Sephience to patients receiving treatment with levodopa to monitor neurological disorders such as exacerbation of convulsion, increased excitability and irritability, seizures, and exacerbation of seizures. **FERTILITY, PREGNANCY AND LACTATION:** No clinical studies on the effect on human fertility have been conducted for sepiapterin. As the data on pregnant women is limited, it is preferable to avoid the use of Sephience during pregnancy. It is unknown whether sepiapterin/metabolites are excreted in human milk. A risk to the newborns/infants cannot be excluded. A decision must be made whether to discontinue breast-feeding or to discontinue/abstain from Sephience therapy taking into account the benefit of breast-feeding for the child and the benefit of therapy for the woman. **EFFECTS ON ABILITY TO DRIVE AND USE MACHINES:** No or negligible influence on the ability to drive and use machines. **ADVERSE REACTIONS:** The frequency of adverse reactions was calculated based on pooled data from 2 pivotal clinical studies in patients with PKU (n=222): Very common (≥1/10): upper respiratory tract infection (19.8%), headache (15.3%), diarrhoea (14.9%), and abdominal pain (12.2%). Common (≥1/100 to <1/10): faeces discoloured (4.5%), and hypophenylalaninaemia (2.7%). Long-term safety data are limited. **MARKETING AUTHORISATION HOLDER:** PTC Therapeutics International Limited. **PACKAGES AND PRICING:** Not available in Finland: **CLASSIFICATION FOR SALE OR SUPPLY:** Prescription-only medicine. Please, always consult the SmPC before prescribing. **DATE OF PREPARATION:** June 2026.

▼ This medicine is subject to additional monitoring. Adverse events should be reported to PTC at pharmacovigilance@ptcbio.com.

Registration conditions differ internationally. Always consult local prescribing information and/or Summary of Product Characteristics before prescribing any product.

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July 2026. GL-SEP-0268

WEDNESDAY, AUGUST 26TH, 2026
18.30-19.30

Denali Therapeutics **Hall 5B**

The Blood–Brain Barrier and Therapeutic Strategies for Neurodegenerative Lysosomal Diseases

CHAIR: *Simon Jones (Denali Therapeutics Inc., United States)*

Engineering Efficient Delivery of Antibodies and Enzymes to the Brain and Other Body Tissues Through Transferrin Receptor Targeting: Transporting Biotherapeutics to the Final Frontier
Robert Thorne (Denali Therapeutics Inc., United States)

The TransportVehicle™ Platform for Diseases Impacting the Brain and Body
Simon Jones (Denali Therapeutics Inc., United States)

Treatment of MPS II with Intravenous Tividenofusp Alfa
Joseph Muenzer (Chapel Hill, United States)

WEDNESDAY, AUGUST 26TH, 2026
18.30-19.30

Amicus Therapeutics **Hall 5C**

Reflecting on the Fabry journey to write the future of care

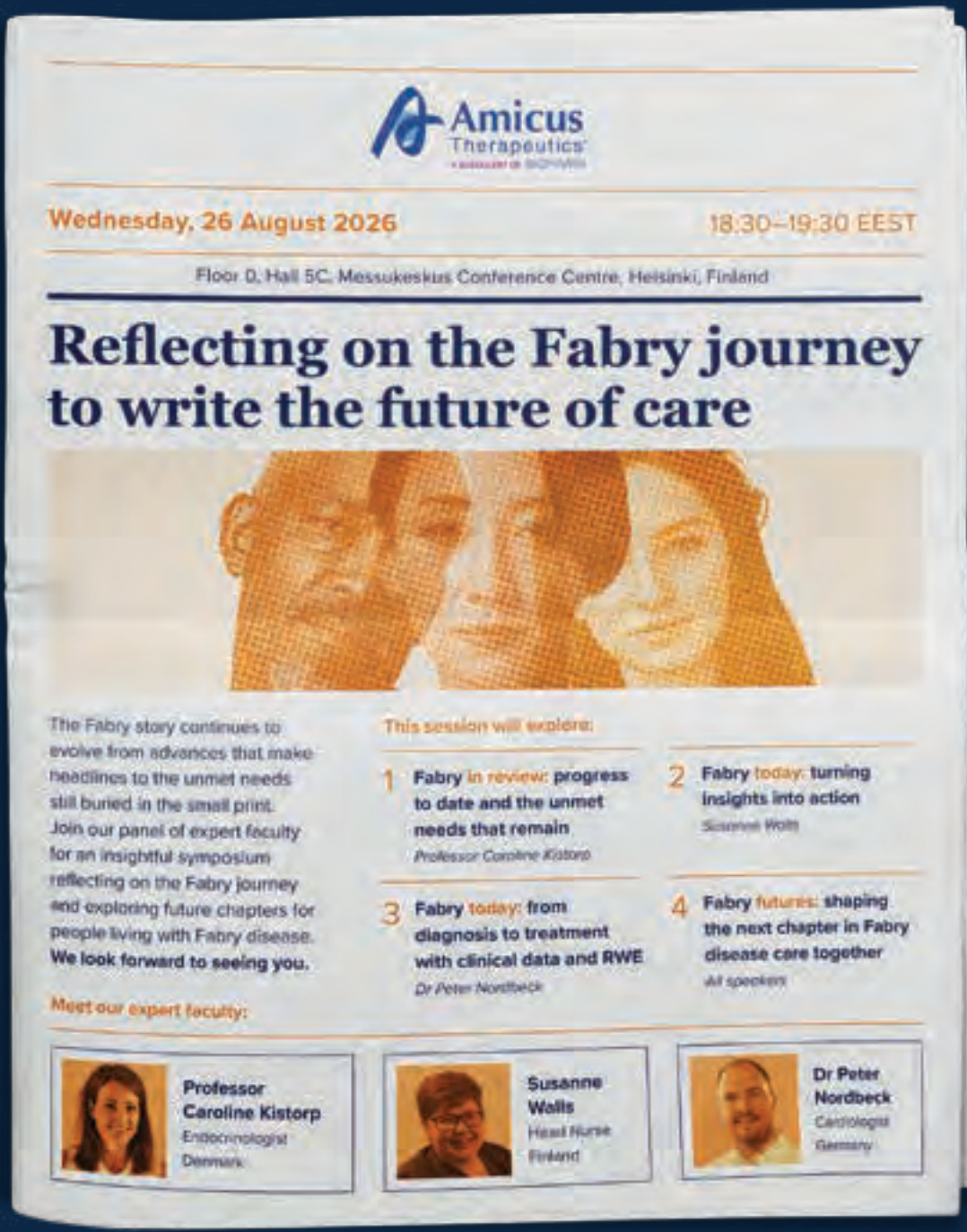
CHAIR: *Caroline Kistorp (Kopenhagen, Denmark)*

Progress to date and the unmet needs that remain
Caroline Kistorp (Copenhagen, Denmark)

Turning insights into action
Susanne Walls (Turku, Finland)

From diagnosis to treatment with clinical data and real-world evidence
Peter Nordbeck (Würzburg, Germany)

Shaping the next chapter in Fabry disease care together
All speakers



RWE, real-world evidence

This satellite symposium has been funded and initiated by Amicus Therapeutics and is intended for healthcare professionals only. This is a promotional symposium, and product information will be discussed. A light meal will be available.

www.amicusrx.com

Com-NN-IN-26-00028-1 I June 2026

THURSDAY, AUGUST 27TH, 2026
07.30-08.30

Immedica Pharma AB Hall 5A

Beyond Survival: Redefining Quality of Life in Urea Cycle Disorders

CHAIR: *Spyros Batzios (Athens, Greece)*

Defining Treatment Goals in UCDs: Are Our Current Targets Fit for Purpose?
Spyros Batzios (Athens, Greece)

Balancing Nutritional and Pharmacological Strategies: Optimizing Care Without Compromise
Erin MacLeod (Washington DC, United States)

Living in "Slow Mode": Quality of Life Challenges in Female OTC-D Patients
Soledad Kleppe (Buenos Aires, Argentina)

Where is the Limit? Redefining Motor Outcomes in ARG1-D Treatment
Marcello Bellusci (Madrid, Spain)

THURSDAY, AUGUST 27TH, 2026
07.30-08.30

Chiesi Farmaceutici spa Hall 5B

Powering Next-Generation Lysosomal Storage Disorders´
Bringing together clinical, patient, innovation and policy experts this session will explore how data, digital health and AI can enhance LSDS` care.

Come meet our panel of leading voices in the filed:
Maurizio Scarpa (Udine, Italy)
Kim Angel (International MPS Network, Canada)
Maria del Mar Manu Pereira (Barcelona, Spain)
Alberta Spreafico (Pavia, Italy)

Immedica Pharma
Dedicated to patients with UCDs

Immedica is a rare disease company focused on improving the lives of patients with urea cycle disorders (UCDs) including arginase 1 deficiency (ARG1-D). By working closely with healthcare professionals, patient communities and caregivers, we aim to improve diagnosis, expand access to treatment and help patients achieve better long-term outcomes.

Together, we can help more patients receive the care they deserve.

Visit immedica.com to learn more about our commitment to rare metabolic diseases.

Immedica pharma

COM-007381, Date of preparation: June 2026

We remain committed to bringing innovative treatments for people living with rare diseases

We believe that mRNA therapeutics will target the root cause of many rare diseases and change the future course of treatments

Our rare disease pipeline includes clinical programs for ornithine transcarbamylase (OTC) deficiency and cystic fibrosis.

ARCTURUS® therapeutics

For more information visit: **ArcturusRx.com**

THURSDAY, AUGUST 27TH, 2026
07.30-08.30

Travere Therapeutics **Hall 5C**

Pegtibatinase: An Investigational Enzyme Replacement Therapy in Development for Treatment of Classical Homocystinuria

SPEAKERS: *François Maillot (Tours, France)*
Can Ficicioglu (Philadelphia, United States)

THURSDAY, AUGUST 27TH, 2026
07.30-08.30

Takeda **101ab**

Not all evidence is equal: Informing Fabry treatment choices

CHAIR: *Sandro Feriozzi (Rome, Italy)*

Trials, studies and registries: framing the evidence in Fabry disease
Sandro Feriozzi (Rome, Italy)

Interpreting the evidence in Fabry disease: a hierarchy of endpoints
Patrício Aguiar (Lisbon, Portugal)

Twenty years of agalsidase alfa in FOS: long-term outcomes in Fabry disease
Guillem Pintos-Morell (Barcelona, Spain)

Positioning agalsidase alfa: personalizing therapy in Fabry disease
Sandro Feriozzi (Rome, Italy)

Expert panel: your questions, our perspectives
All faculty

Closing thoughts: Guiding your next Fabry decision
Sandro Feriozzi (Rome, Italy)

VISIT US AT BOOTH #S04

GROWING AWARENESS. LEARN MORE ABOUT
Classical Homocystinuria (HCU)

NOW ENROLLING

Global Phase 3 Study
Harmony

TRAVERE THERAPEUTICS
Global Medical Affairs

For more information, visit this US website – clinicaltrials.gov: HARMONY NCT06247085.
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MA-PE-26-0044 July 2026

NUTRICIA
LIFE-TRANSFORMING NUTRITION | METABOLICS

EVERY JOURNEY IS UNIQUE

BUT NO ONE WALKS IT ALONE

Celebrating 70 Years of Nutricia Metabolics, supporting lives with care, experience, and impact

Just as a real passport marks journeys and milestones, the Nutricia Metabolics passport represents **70 years of expertise, innovation, and lifelong patient support**

VISIT AND COLLECT ALL STAMPS FOR A SPECIAL SURPRISE

VISIT OUR STAND **G03**

PKU sphere™

The most extensively researched GMP-based protein substitute available. [1–10]

We are expecting something significant.

New evidence coming soon.

References can be found here

Vitaflo
Enhancing Lives Together

Find out more at stand P02

Swiss quality For life

Dipharma

Better Together

Improved Solutions for Rare Diseases

Specialized in Rare Diseases

high Quality Generics

Going Beyond.
Bringing Greater Ease to Everyday Therapy.

Manufacturing in Europe

Reliable Supply

Improved Solutions

Scan here to learn more or visit us at our booth (No. S14)

- Worldwide distribution through a network of highly selected partners
- Marketing in Germany through the affiliate Dipharma Arzneimittel

ZEVRA THERAPEUTICS

We are a purpose-driven, commercial-stage company focused on bringing life-changing therapeutics to people living with rare diseases.

Adult living with Niemann-Pick disease type C

To learn more, visit **Zevra.com**

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Voices That Matter

Lived Experiences With Cerebrotendinous Xanthomatosis

26 August 2026
07:30 AM – 08:30
(Breakfast will be provided)

Hall 5B

Learn about CTX from a leading expert in lipid metabolism and rare disease

P. Barton Duell, MD
Oregon Health and Science University

Hear first-hand perspectives from those affected by CTX

Evalyn
Evalyn shares her personal journey to diagnosis and what living with CTX means to her

Bobbi
Bobbi shares her experiences through diagnosis and management as a parent and caregiver to her two children living with CTX

CTX, cerebrotendinous xanthomatosis.

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FI-NON-2600002; June 2026.

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SSIEM 2026 | 25-28 AUGUST 2026 | HELSINKI, FINLAND

SYMPOSIUM – Wednesday, August 26, 2026
07.30 – 08.30 AM | Room: Hall 5A

FLEXIBLE BY DESIGN:

REDEFINING TREATMENT POSSIBILITIES WITH PEGUNIGALSIDASE ALFA

MODERATOR:
Christina Lampe *(Germany)*

SPEAKERS:
Diego Fornasari *(Italy)*
Patrício Aguiar *(Portugal)*
Christina Lampe *(Germany)*

This symposium is intended exclusively for healthcare professionals.
ALL_26_1415. Date of preparation July 2026.
21585-27.04.2026

Chiesi
global rare diseases