where EAST meets WEST

SSIEM

SSIEM Annual Symposium 2023

29 August - 1 September 2023 | Jerusalem, Israel

Pre-Program

החברה הישראלית
למחלות מטבוליות
Israel Society for
Metabolic Diseases

Table of Contents

WELCOME ADDRESS

Yair Anikster
SSIEM 2023 president

Welcome to the SSIEM 2023 in Jerusalem! This year's theme, "**East Meets West**", will explore what different regions of the world can learn from each other. We have a plateful of topics to discuss, including:

The differential expression of the same disease across the globe. We will discuss the vastly different phenotypes of citrin deficiency, dihydrolipoamide dehydrogenase deficiency, and neuronopathic Gaucher disease, and discuss the lesser familiar phenotypes of diseases you are familiar with.

New disease pathways in the mitochondria. We will explore new roles of vitamin processing within the mitochondria via newly described inborn errors of metabolism, as shed light on the role of the mitochondria outside of the oxidative phosphorylation realm.

What we can learn from our neighbors. We will discuss how pediatricians, adult doctors, clinicians, laboratory experts, and nutritionists can use each other's experience to better personalize care. We will also discuss the latest on diagnostic metabolomics, nutrition-based deep phenotyping, and hindsight wisdom from adults.

The complexity of brain traffic. We will discuss the molecular mechanisms underlying disorders of cellular trafficking and RNA metabolism, as well as biomarkers and potential therapeutic targets.

Where big data meets small practices. In the age of big data, are we screening more that we can handle? Are we ready to make an in silico diagnosis? We will discuss the challenges and opportunities of using big data in small practices and the potential impact of big data on the cost of care.

Therapeutic updates. We will discuss the latest advances in the treatment of inherited metabolic disorders, including antisense oligonucleotide therapy, gene therapy, and mitochondrial augmentation therapy.

In addition, join us for a variety of other events, including an open-for-all nutrition and dietary management session, dedicated educational and adult IEM sessions, speed mentoring opportunities, a highest ranked posters competition, breakthrough works, and many networking opportunities.

Last but not least – Do not forget to check our satellite symposia – the CTX, the INFORM, and the brand-new Leukodystrophy meeting.

All taking place where East meets West - see you all in Jerusalem!

Kind regards,
Yair Anikster

SCIENTIFIC PROGRAM

COMMITTEES

Local Scientific Committee

Yair Anikster, Chair
Sheba Medical Center | Ramat Gan, Israel

Shlomo Amashanu
Ministry of Health, Israel

Stanley Korman
Shaare Zedek Medical Center | Jerusalem, Israel

Hanna Mandel
Ziv Medical Center | Safed, Israel

Yehoshua (Josh) Manor
Sheba Medical Center | Ramat Gan, Israel

Ann Saada
Hadassah Medical Center | Jerusalem, Israel

Ronen Spiegel
Emek Medical Center | Afula, Israel

Galit Tal
Rambam Medical Center | Haifa, Israel

INTERNATIONAL SCIENTIFIC COMMITTEE

Nenad Blau | University Children's Hospital, Zurich, Switzerland

Avihu Boneh | University of Melbourne, Australia

Carlo Dionisi Vici | Bambino Gesù Children's Hospital, Rome, Italy

Maria Dulce Silva Quelhas | Porto, Portugal

Andrea Gropman | Children's National Medical Center, Washington. D.C., USA

Meral Gunay-Aygun | John Hopkins University School of Medicine, St. Petersburg, USA

Helen Michelakakis | Athens, Greece

Philippa Mills | London, United Kingdom

Eva Morava | Mayo Clinic, USA

Shamima Rahman | UCL Great Ormond Street Institute of Child Health, London, UK

Agnes Rotig | Institute Imagine, Hôpital Necker-Enfants Malades and Université Paris Descartes, Paris, France

Manuel Schiff | Paris, France

Ellen Sidransky | National Human Genome Research Institute, National Institute of Health, Maryland, USA

Priya S. Kishnani | Duke University Medical Center , Durham, NC, USA

SUNDAY, AUGUST 27, 2023
Ramada Hotel

13:00-18:30
2023 CTX International Scientific Meeting

15:00-21:30
INFORM Meeting
(Registration at INFORM - limited places)
International Network for Fatty Acid Oxidation Research and Management

MONDAY, AUGUST 28, 2023
Ramada Hotel

07:30-18:30
INFORM Meeting
(Registration at INFORM - limited places)
International Network for Fatty Acid Oxidation Research and Management

08:00-17:15
2023 CTX International Scientific Meeting

MONDAY AFTERNOON
Location: Neve Shalom

16:30-20:00
White Matter Diseases Meetinfg
Local Committee Endorsed

Register Here

MONDAY 28 AUGUST 2023

	Oranim 2	Oranim 3	Oranim 4	Seminar Room 310	Seminar Room 311	Seminar Room 312	Seminar Room 313	Seminar Room 314
11:00	Registration							
12:00	12:00-14:00 NBS Collaboration Meeting MetabERN/ISNS/ SSIEM (By invitation only)							
12:30								
13:00								
13:30								
14:00		14:00-16:30 MetabERN Meeting (By invitation only)		14:00-16:00 EHOD Executive Board (By invitation only)			14.00-16.00 SSIEM Pre-Council Meeting (SSIEM Honorary Officers & Staff Only)	
14:30								
15:00								
15:30								
16:00							16.00-18.00 SSIEM Council Meeting (Council Members Only)	
16:30	16:30-19:30 MetabERN AOA SNW (By invitation only)	16:30-19:30 MetabERN C-FAO SNW (By invitation only)	16:30-19:30 MetabERN LSD SNW (By invitation only)	16:30-19:30 MetabERN CDG SNW (By invitation only)	16:30-19:30 MetabERN PM-MD (By invitation only)	16:30-19:30 MetabERN PD SNW (By invitation only)		16:30-19:30 MetabERN NOMS SNW (By invitation only)
17:00								
17:30								
18:00								
18:30								
19:00								
19:30								

18:30-21:30 SSIEM Council & JIMD Editors Dinner
(By invitation only)

Networking
Parallel Session
Poster
Satellite Symposia
Speed Mentoring
Plenary Session
Poster Walk
Side & Administraive Meetings

TUESDAY 29 AUGUST 2023

	Ussishkin Hall - Plenary/Paralel	Pincus Parallel	Dulchin 1 paralell	Oranim 1	Oranim 2	Oranim 3	Oranim 4	Seminar Room 310	Seminar Room 311	Seminar Room 312	Seminar Room 313	Seminar Room 314	Posters in Exhibition hall
7:30	07:30 - 09:00 Registration												
9:00				09:00-12:00 SSIEM Adult Meeting (Open to all participants)	09:00-12:30 SSIEM Nutrition & Dietetics session (Open to all participants)	09:00-12:00 GalNet galactosemia symposium (By invitation only)	09:00-12:00 Patient Representative Meeting: IMD Patient advocacy session	09:00-12.30 SSIEM JIMD & JIMD Reports Editorial Board Meeting (By invitation only)	09.00-10.30 ERNDIM Workshop (By invitation only)	09.00-10.30 ERNDIM Workshop (By invitation only)	09.00-10.30 ERNDIM Workshop (By invitation only)	09:00-11:00 EHOD "Remethylation Guidelines Group" (By invitation only)	
9:30													
10:00													
10:30													
11:00											11.00-12.00 ERNDIM Participant Meeting (Open to all ERNDIM participants)		
11:30													
12:00													
12:30		12:45-13:45 Satellite Symposia BioMarin	12:45-13:45 Satellite Symposia PTC			12:45-13:45 Satellite Symposia Chiesi							
13:00													
13:30													
14:00	14:00-14:50 Opening Ceremony – Epitranscritome												
14:30													
15:00	14:50-16:20 Where East Meets West												
15:30													15:30-19:30 Posters
16:00													
16:30	16:50-18:20 The mitochondrial Bs – old players, new roles												
17:00													
17:30													
18:00	18.20-18.50 Archibald Garrod Lecture												
18:30													
19:00	18:50-19:00 The case of Citrin deficiency: A new and holistic approach to inborn error of metabolism												

19.00-20.30 Welcome Reception

Networking | Parallel Session | Poster | Satellite Symposia

Speed Mentoring | Plenary Session | Poster Walk | Side & Administraive Meetings

WEDNESDAY 30 AUGUST 2023

	Ussishkin Hall - Plenary/Paralel	Pincus Parallel	Dulchin 1 paralell	Oranim 1	Oranim 2	Oranim 3	Oranim 4	Seminar Room 310	Seminar Room 311	Seminar Room 312	Seminar Room 313	Seminar Room 314	Posters in Exhibition hall
6:30	Registration												
7:00													
7:30			07:30-08:30 Satellite Symposia Recordati		07:30-08:30 Speed Mentoring			07:30-08:30 IOC Meeting					07:30-20:15 Posters
8:00									08:15-10:15 SSIEM Dietitians Group Committee Meeting				
8:30	08:30-10:00 Parallel Session - Educational	08:30-10:00 Parallel Session	08:30-10:00 Parallel Session		08:30-10:00 Parallel Session	08:30-10:00 SSIEM Nurses Meeting (Open to all participants)	08:30-10:00 Parallel Session						
9:00													
9:30													
10:00													
10:30	10:30-12:00 Learning from the Neighbours												
11:00													
11:30													
12:00													
12:30							12:30-13:00 SSIEM Council & Advisory Council Meeting (By invitation only)						
13:00							13.00-14.00 SSIEM Annual General Meeting (SSIEM Members Only)						
13:30													
14:00	14:00-15:30 Parallel Session	14:00-15:30 Parallel Session	14:00-15:30 Parallel Session		14:00-15:30 Parallel Session	14:00-15:30 Nutritional Management of IEM Session							
14:30													
15:00													
16:00	16:00-17:30 The Complexity of brain traffic: new insights from neurometabolism												
16:30													
17:00													
17:30		17:30-18:30 Satellite Symposia Takeda				17:30-18:30 Satellite Symposia Travere							18:35-20:30 Poster Walk + Highest Ranked Posters
18:00													
18:30													
19:00													
19:30													
20:00													

20:30 SSIEM Symposium President´s Dinner (By invitation only)

Networking · Parallel Session · Poster · Satellite Symposia
Speed Mentoring · Plenary Session · Poster Walk · Side & Administraive Meetings

THURSDAY 31 AUGUST 2023

	Ussishkin Hall - Plenary/Paralel	Pincus Parallel	Dulchin 1 paralell	Oranim 1	Oranim 2	Oranim 3	Oranim 4	Posters in Exhibition hall
6:30								
7:00	06:45 Registration							
7:30					07:45-08:45 Speed Mentoring			07:45-15:30 Posters
8:00								
8:30	08:45-10:15 Where big data meets small practices							
9:00								
9:30								
10:00					10:15-10:45 Coffee with the SSIEM JIMD Editors			
10:30	10:45-12:15 Parallel Session	10:45-12:15 Parallel Session	10:45-12:15 Parallel Session			10:45-12:15 Parallel Session		
11:00								
11:30								
12:00					12:00-13:30 SSIEM JIMD Editors, JIMD Communicating Editors and JIMD Advisory Members Board Meeting			
12:30		12:30-13:30 Satellite Symposia BioMarin	12:30-13:30 Satellite Symposia PTC			12:30-13:30 Satellite Symposia Sanofi		
13:00								
13:30	13:30-14:15 Komrower Lecture							
14:00	14:15-15:30 Parallel Session	14:15-15:30 Parallel Session	14:15-15:30 Parallel Session		14:15-15:30 Parallel Session	14:15-15:30 Parallel Session		
14:30								
15:00								
15:30								

15:30-18:30 SSIEM Networking Tours

20:00-23:00 SSIEM Networking Evening & Dinner Speech

Networking | Parallel Session | Poster | Satellite Symposia

Speed Mentoring | Plenary Session | Poster Walk | Side & Administraive Meetings

FRIDAY 1 SEPTEMBER 2023

	Ussishkin Hall - Plenary/ Paralel	Posters in Exhibition hall
7:30	07:45 Registration	
8:00	08:15-09:45 Therapeutic updates - Advances in therapy modalities	08:15-12:30 Posters
8:30		
9:00		
9:30		
10:00	10:15-11:45 Late Breaking News	
10:30		
11:00		
11:30	11:45-12:15 Awards and Introduction to SSIEM 2024 at Porto	
12:00		
12:30	12:15-12:30 Closing Remarks	

* Program outline is subject to changes.

Please note: This is a preliminary programme, which is subject to change without prior notice.
For updated information please visit: www.ssiem2023.org

SUNDAY, AUGUST 27, 2023

2023 CTX International Scientific Meeting
13:00-18:30, Ramada Hotel

INFORM Meeting Part I *(registration at INFORM - limited places)*
15:00-21:30, Ramada Hotel
International Network for Fatty Acid Oxidation Research and Management

MONDAY, AUGUST 28, 2023

INFORM Meeting Part II *(registration at INFORM - limited places)*
07:00-18:30, Ramada Hotel
International Network for Fatty Acid Oxidation Research and Management

2023 CTX International Scientific Meeting
08:00-17:15, Ramada Hotel

Registration
11:00-12:00, Exhibition Hall (ICC)

NBS Collaboration Meeting MetabERN/ISNS/SSIEM
12:00-14:00, Oranim 2 (ICC) *(By invitation only)*

EHOD Executive Board
14:00-16:00, Seminar Room 310 (ICC) *(By invitation only)*

MetabERN Meeting
14:00-16:30, Oranim 3 (ICC) *(By invitation only)*

SSIEM Pre-Council Meeting
14:00-16:00, Seminar Room 313 (ICC) *(SSIEM Honorary Officers & Staff Only)*

SSIEM Council Meeting
16:00-18:00, Seminar Room 313 (ICC) *(Council Members Only)*

MetabERN AOA SNW
16:30-19:30, Oranim 2 (ICC) *(By invitation only)*

MetabERN C-FAO SNW
16:30-19:30, Oranim 3 (ICC) *(By invitation only)*

MetabERN LSD SNW
16:30-19:30, Oranim 4 (ICC) *(By invitation only)*

MetabERN CDG SNW
16:30-19:30, Seminar Room 310 (ICC) *(By invitation only)*

MetabERN PM-MD
16:30-19:30, Seminar Room 311 (ICC) *(By invitation only)*

MetabERN PD SNW
16:30-19:30, Seminar Room 312 (ICC) *(By invitation only)*

MetabERN NOMS SNW
16:30-19:30, Seminar Room 314 (ICC) *(By invitation only)*

SSIEM Council & JIMD Editors Dinner
18:30-21:30 *(By invitation only)*

TUESDAY, AUGUST 29, 2023

Registration
07:30-09:00, Exhibition Hall (ICC)

ERNDIM Workshop
09:00-10:30, Seminar Room 311 *(By invitation only)*

SSIEM Adult Meeting
09:00-12:00, Oranim 1 (ICC) *(Open to all participants)*
Session details to be announced.

SSIEM Nutrition & Dietetics session *(Open to all participants)*
09:00-12:30, Oranim 2 (ICC)
Chairs: *Rani Singh, USA; Bénédicte Samba, France*

09:00 **Revised Glutaric Aciduria Type 1 Guidelines: new insights for dietary treatment?**
Nikolas Boy, Heidelberg, Germany

09:30 **Metabolic control in PKU in Europe: what's the picture?**
Alex Pinto, Birmingham, UK

10:00 **Ketogenic Dietary Therapy in McArdle Disease (GSD V)**
Ros Quinlivan, London, UK

10:30 ***Refreshment Break***

11:00 **Personalised nutrition approaches for metabolic disorders – what can we learn from nutrigenomics research?** | *Baukje de Roos, Aberdeen, UK*

11:30 **Clinical and nutritional outcomes in GSD Type 1 patients using Continuous Glucose Monitoring (CGM)** | *Alessandro Rossi, Naples, Italy*

12:00 **Branched-chain amino acids in propionic academia: what is ideal?**
Rajavel Elango, Vancouver, Canada

GalNet galactosemia symposium
09:00-12:00, Oranim 3 (ICC) *(By invitation only)*

Patient Representative Meeting: IMD Patient advocacy session
09:00-12:00, Oranim 4 (ICC)

SSIEM JIMD & JIMD Reports Editorial Board Meeting
09:00-12:30, Seminar Room 310 (ICC) *(By invitation only)*

ERNDIM Workshop
09:00-10:30, Seminar Room 312 (ICC) *(By invitation only)*

ERNDIM Workshop
09:00-10:30, Seminar Room 313 (ICC) *(By invitation only)*

EHOD "Remethylation Guidelines Group"
09:00-11:00, Seminar Room 314 (ICC) *(By invitation only)*

ERNDIM Participant Meeting
11:00-12:00, Seminar Room 313 (ICC) *(Open to all ERNDIM participants)*

Satellite Symposia: BioMarin
12:45-13:45, Pincus (ICC)

Satellite Symposia: PTC
12:45-13:45, Dulchin 1 (ICC)

Satellite Symposia: Chiesi
12:45-13:45, Oranim 3 (ICC), Oranim 4 (ICC)

SSIEM 2023 Symposium Opening
14:00-14:50, Ussishkin (ICC)

14:00 **Opening Ceremony**

14:20 **Opening Lecture – Epitranscritome** | *Gideon Rechavi, Sheba Medical Center, Israel*

PL1: Where East meets West – differential expression of the same disease in different regions in the world
14:50-16:20, Ussishkin (ICC)

14:50 **Neuronopathic GD – rare in the West, common in the East**
Ozlem Goker-Alpan, Lysosomal & Rare Disorders Research & Treatment Center, USA

15:20 **Dihydrolipamide dehydrogenase deficiency – An Israeli heterogeneity: mild in the West, severe in the South**
Orna Staretz-Chacham, Ben Gurion University, Soroka Medical Center, Israel

15:50 **Citrin deficiency – The East-side story**
Johannes Häberle, University Children's Hospital, Switzerland

Posters
15:30-19:30, Exhibition Hall (ICC)

Coffee Break
16:20-16:50, Exhibition Hall (ICC)

PL2: The State of the Mitochondria – old players, new roles
16:50-18:20, Ussishkin (ICC)

16:50 **Mitochondrial DNA in health and disease**
Douglas C. Wallace, Children's Hospital of Philadelphia, USA

17:20 **B3 pathway – A newly described fatal encephalopathy triggered by fever**
Carole Linster, University of Luxembourg, Luxembourg

17:50 **B5 pathway – More than NBIA: new findings in the CoA synthesis pathways**
Arcangela Iuso, Helmholtz Zentrum of München, Germany

Archibald Garrod Lecture
18:20-18:50, Ussishkin (ICC)

18:20 **Clinical manifestation and long-term outcome of citrin deficiency: Report from a nationwide study in Japan** | *Jun Kido, Kumamoto University, Japan*

Short Break
18:50-19:00, Exhibition Hall (ICC)

The case of Citrin deficiency: A new and holistic approach to inborn error of metabolism
19:00-19:15, Ussishkin (ICC)

19:00 **The case of Citrin deficiency: A new and holistic approach to inborn error of metabolism**
Barbara Yu, Citrin Foundation, Singapore (Local Committee Endorsed)

WELCOME RECEPTION
19:00-20:30, Exhibition Hall (ICC)

SSIEM JIMD EDITORS & JIMD COMMUNICATING EDITORS' DINNER
(By invitation only)

WEDNESDAY, AUGUST 30, 2023

Registration
06:30-07:30, Exhibition Hall (ICC)

Satellite Symposia: Recordati
07:30-08:30, Dulchin 1 (ICC)

Speed Mentoring
07:30-08:30, Oranim 2 (ICC)

IOC Meeting
07:30-08:30, Seminar Room 310 (ICC)

POSTERS
07:30-20:15, Exhibition Hall (ICC)

SSIEM Dietitians Group Committee Meeting
08:15-10:15, Seminar Room 311 (ICC)

SSIEM Nurses Meeting
08:30-10:00, Oranim 3 (ICC) *(Open to all participants)*

Parallel Session - Educational: Pitfalls to avoid in the management of IEM
08:30-10:00, Ussishkin (ICC)

08:30 **Common and uncommon pitfalls in pediatric IEM**
Andrew Morris, Willink Metabolic Unit, UK

08:50 **Cases with challenging biochemical signatures**
Jean-Marc Nuoffer, University of Bern, Switzerland

09:10 **Adult IEM: What's different and what stays the same**
Robin Lachmann, University College London Hospitals, UK

09:30 **Common mistakes with diet in IEMs**
Rani H. Singh, Emory University of Atlanta, USA

Parallel Session: Innovative Therapies I + New Diseases
08:30-10:00, Pincus (ICC)

08:30 **Fortunate discovery and fast treatment – Bachmann-Bupp syndrome is a new potentially treatable inborn error of polyamine metabolism**
Caleb Bupp, Corewell Health & Helen Devos Children's Hospital, USA

08:45 **Atidarsagene autotemcel (autologous hematopoietic stem cell gene therapy) preserves cognitive and motor development in early-onset metachromatic leukodystrophy with up to 12 years follow-up**
Valeria Calbi, San Raffaele Telethon Institute for Gene Therapy (SR-TIGET), IRCCS San Raffaele, Italy

09:00 **Early skeletal outcome after Hematopoietic Stem & Progenitor Cell Gene Therapy for Hurler Syndrome**
Maria Ester Bernardo, IRCCS San Raffaele Scientific Institute, Italy

09:15 **RGX-111: an investigational gene therapy for the treatment of severe mucopolysaccharidosis type i (mps i): interim analysis of data from the first in human study**
Raymond Wang, Choc Children's Hospital, USA

09:30 **Single AAV gene therapy with mini-GDE for glycogen storage disease type III**
Antoine Gardin, Genethon, France

09:45 **AXO-AAV-GM2 Gene Therapy for the Treatment of GM2 Gangliosidosis: Interim Results from an Ongoing Phase 1/2 Trial**
Florian Eichler, Massachusetts General Hospital, USA

Parallel Session: Mitochondrial Disorders I
08:30-10:00, Dulchin 1 (ICC)

08:30 **A large-scale drug screen for compounds that improve reductive stress in melas cardiomyocytes** | *Tamas Kozicz, Mayo Clinic, USA*

08:45 **Shedding light on the cellular consequences of Friedreich's ataxia**
Tslil Ast, The Weizmann Institute of Science, Israel

09:00 **A novel mitochondrial assembly factor RTN4IP1 has an essential role in the final stages of Complex I assembly** | *Rob Taylor, Newcastle University, UK*

09:15 **PALFES study: exome sequencing reveals a genetic cause in 40% of pediatric acute liver failure cases of indeterminate origin**
Lea Dewi Schlieben, Klinikum Rechts Der Isar, Technical University Munich, Germany

09:30 **Mavodelpar clinical development program in adult patients with primary mitochondrial myopathy: results from a Phase 1b study and design of ongoing pivotal study (STRIDE)**
Robert D. S. Pitceathly, UCL Queen Square Institute of Neurology, UK

09:45 **Mitochondrial ATP synthase deficiency and its relationship with the urea cycle**
Barbara Siri, Bambino Ges Children Hospital, IRCCS, Italy

Parallel Session: Clinical Studies & Outcomes I
08:30-10:00, Oranim 2 (ICC)

08:30 **Pegzilarginase demonstrates long-term, clinically meaningful improvements in functional mobility in ARG1-D: patient-level analysis from the Phase 3 PEACE trial**
Serena Gasperini, Fondazione IRCCS San Gerardo dei Tintori, Italy

08:45 **Switching treatment from alglucosidase alfa to cipaglucosidase alfa plus miglustat positively affects motor function and quality of life in patients with late-onset Pompe disease** | *Priya Kishnani, Duke University Medical Center, USA*

09:00 **Liver Transplant for the Treatment of Inborn Errors of Metabolism - the Israeli Experience** | *Adi Efron Srour, Schneider's Children Medical Center, Israel*

09:15 **Standardized protocols to optimize the emergency management of patients with inherited metabolic diseases in France**
Camille Wicker, Universitary Hospital Strasbourg, France

09:30 **The ZOEMBA-Amsterdam study for unsolved metabolic phenotypes: The diagnostic yield of untargeted metabolomics and exome reanalysis**
Elise Ferreira, Amsterdam UMC, The Netherlands

09:45 **Pegtibatinase, an Investigational Enzyme Replacement Therapy for the Treatment of Classical Homocystinuria (HCU): Latest Findings from the COMPOSE Phase 1/2 Trial**
Can Ficicioglu, University of Pennsylvania, Perelman School of Medicine, USA

Parallel Session: Disorders of FAO and Ketones
08:30-10:00, Oranim 4 (ICC)

08:30 **Elamipretide restores mitochondrial function in trifunctional protein deficiency mice and human fibroblasts** | *Eduardo Vieira Neto, University of Pittsburgh, USA*

08:45 **Medium-chain acylcarnitines target muscular phenotypes in long-chain fatty acid oxidation disorders**
Keaton Solo, University of Pittsburgh/University of Pittsburgh Medical Center, USA

09:00 **Correlation between cardiopulmonary exercise tests and biochemical parameters in patients affected by fatty acid oxidation disorders**
Apolline IMBARD, Hopital Necker-Enfants Malades, APHP, France

09:15 **Fasting metabolism in children with fatty acid oxidation disorders: Earlier lipolysis in VLCAD compared to MCAD patients**
David Olsson, Karolinska Institutet/Karolinska University Hospital, Sweden

09:30 **Cardiac-specific deficiency of 3-hydroxy-3-methylglutaryl coenzyme a lyase in mice causes cardiomyopathy and a distinct pattern of acyl-coenzyme A-related biomarkers**
Grant Mitchell, CHU Sainte-Justine, Canada

09:45 **A new old player in MCADD: reduced coenzyme A availability in medium-chain acyl-CoA dehydrogenase deficiency**
Ligia A. Kiyuna, University Medical Center Groningen, University of Groningen, The Netherlands

Coffee Break
10:00-10:30, Exhibition Hall (ICC)

PL3: Learning from the neighbors
10:30-12:00, Ussishkin (ICC)

10:30 **What can pediatricians learn from adult IEM**
Fanny Mochel, Sorbonne University of Paris, France

11:00 **Personalized medicine and nutrition based on deep human phenotyping**
Eran Segal, Weizmann Institute of Science, Israel

11:30 **What clinicians can learn from laboratory metabolomics**
Sarah H. Elsea, Baylor College of Medicine, USA

Lunch
12:00-13:00, Exhibition Hall (ICC)

SSIEM Council & Advisory Council Meeting
12:30-13:00, Oranim 4 (ICC) *(By invitation only)*

SSIEM Annual General Meeting
13:00-14:00, Oranim 4 (ICC) *(SSIEM Members Only)*

Parallel Session: Lysosomal disorders
14:00-15:30, Ussishkin (ICC)

14:00 **RGX-121: An investigational gene therapy for the treatment of neuronopathic mucopolysaccharidosis type II (MPS II), an interim analysis of data from the first-in-human study** | *Paul Harmatz, UCSF Benioff Children's Hospital, USA*

14:15 **RNA based therapies for Lysosomal Storage disorders (LSDs)**
Andrea Dardis, University Hospital Udine, Italy

14:30 **Glucosylsphingosine causes mitochondrial dysfunction in a neuronal cell model**
Valeria Nikolaenko, UCL Institute of Child Health, UK

14:45 **CLN3 deficiency leads to neurometabolic perturbations during early development**
Ursula Heins Marroquin, Luxembourg Centre for Systems Biomedicine, Luxembourg

15:00 **Innovation in the diagnostic field: A new accurate and robust screening method for the most common lysosomal storage diseases**
Amber Van Baelen, University Hospital of Antwerp, Belgium

15:15 **Interim, 24-month results of a phase 1/2 study of weekly intravenous DNL310 (brain-penetrant enzyme replacement therapy) in MPS II**
Barbara Burton, Lurie Childrens Hospital, USA

Parallel Session: CDG / Protein Modifications
14:00-15:30, Pincus (ICC)

14:00 **A iCardiomyocyte model of PGM1-CDG reveals defective energy metabolism with implications for therapy** | *Silvia Radenkovic, Mayo Clinic, USA*

14:15 **Novel fractionated plasma N-glycan test identifies sensitive hepatic and extra-hepatic biomarkers for Congenital Disorders of Glycosylation (CDG)**
Earnest James Paul Daniel, Childrens Hospital of Philadelphia (CHOP), USA

14:30 **NGLY1 deficiency zebrafish model manifests abnormalities of the nervous system**
Aviv Mesika, Bar Ilan University, Israel

14:45 **Ongoing Natural History study in Phosphomannomutase 2 Congenital Disorder of Glycosylation (PMM2-CDG): Clinical and Basic Investigations**
Eva Morava, Mayo Clinic, USA

15:00 **Acute neurological symptoms in patients with PMM2-CDG: a link with perturbed hemostasis?** | *Camille Wicker, Universitary Hospital Strasbourg, France*

15:15 **Cellular oxidative damage in congenital disorders of glycosylation**
Belkis Ak, Istanbul University Medical Faculty, Turkiye

Parallel Session: Urea cycle disorders
14:00-15:30, Dulchin 1 (ICC)

14:00 **Impaired nuclear glycogen metabolism affects liver homeostasis in Argininosuccinic aciduria** | *Alfonso Manuel D'Alessio, Telethon Institute of Genetics and Medicine, Italy*

14:15 **Small molecules for targeting moonlighting functions of urea cycle proteins**
Georgios Makris, University Children's Hospital Zurich, Switzerland

14:30 **Screening of stabilizing ornithine transcarbamylase variants for use in a one-fits-many gene editing approach for OTC deficiency**
Sven Klassa, University Children's Hospital Zurich, Switzerland

14:45 **Long-term Safety and Efficacy of DTX301 in Adults with Late-Onset Ornithine Transcarbamylase (OTC) Deficiency: A Phase 1/2 Trial**
Tarekegn Hiwot, University Hospital of Birmingham, UK

15:00 **The markers distinguishing healthy and citrin deficiency newborns in the newborn screening** | *Jun Kido, Kumamoto University, Japan*

15:15 **Disruption of a possible compensatory mechanism by increasing ureagenesis can be the cause of the development of hyperammonemia in citrin deficiency**
Kimihiko Oishi, Jikei University School of Medicine, Japan

Parallel Session: Vitamins + Neurotransmitters
14:00-15:30, Oranim 2 (ICC)

14:00 **Dose-ranging pre-clinical studies of systemic AAV9 with codon-optimized reduced size ATP7A (cors-ATP7A) plus subcutaneous Cu-Histidinate in a Menkes disease model**
Stephen G. Kaler, Nationwide Children's Hospital, USA

14:15 **Increased survival and improved clinical outcomes in patients with MoCD Type A treated with cyclic pyranopterin monophosphate (cPMP)/fosdenopterin**
Guenter Schwarz, University of Cologne, Germany

14:30 **MEDNIK- like syndrome: delineation of the copper metabolism phenotype and response to zinc therapy** | *Diego Martinelli, Bambino Ges Childrens Hospital, IRCCS, Italy*

14:45 **An efficient workflow for diagnosis of AADC deficiency – A template for reducing health disparities in pediatric care** | *Sarah Elsea, Baylor College of Medicine, USA*

15:00 **Tolerability and efficacy of L-serine in patients with GRIN-related encephalopathy**
Natalia Julia-Palacios, Hospital Sant Joan de DuCIBERER, Spain

15:15 **AADC deficiency severity/mildness predictions can be suggested by unraveling the structural dynamics of the AADC protein**
Mariarita Bertoldi, University of Verona, Italy

Parallel Session: Nutritional Management of IEM Session
14:00-15:30, Oranim 3 (ICC)

14:00 **Nutrition, exercise and growth in 17 young Greek patients with carnitine palmitoyltransferase II (CPT II) deficiency**
Eleana Petropoulou, Institute of Child Health, Greece

14:15 **When patients' metabolism is challenged twice - managing autoimmune diabetes mellitus with a coexisting inherited metabolic disorder**
Christina Spyridoula Sidira, Great Ormond Street Hospital for Children NHS Foundation Trust, UK

14:30 **Free Use of fruit and vegetables containing 76-100mg of Phenylalanine per 100g in children with phenylketonuria: a 6 months follow-up**
Alex Pinto, Birmingham Women's and Children's Hospital, UK

14:45 **Natural protein intake in children with Phenylketonuria: prescription vs. intake**
Alex Pinto, Birmingham Women's and Children's Hospital, UK

15:00 **An assessment of swallowing function, nutrition and growth in young children with late-onset Pompe disease diagnosed via newborn screening**
Surekha Pendyal, Duke University, USA

15:15 **Dietary management of Hereditary Fructose Intolerance (HFI) in eleven Italian meta bolic centres: current practices and new challenges**
Alice Dianin, IRCCS University Hospital of Bologna, Ital

Coffee Break
15:30-16:00, Exhibition Hall (ICC)

PL4: The Complexity of brain traffic: new insights from neurometabolism
16:00-17:30, Ussishkin (ICC)

16:00 **Disorders of cellular trafficking causing neurological phenotypes**
Angeles García-Cazorla, Sant Joan de Déu Hospital of Barcelona, Spain

16:30 **RNA metabolism, leukodystrophies, and CNS pathology**
Laura Adang, Children's Hospital of Philadelphia, USA

17:00 **Brain Pathology Reflected in Blood Biomarkers: A Lesson From X-Linked Adrenoleukodystrophy**
Johannes Berger, Medical University of Vienna, Austria

Satellite Symposia: Takeda
17:30-18:30, Pincus (ICC)

Satellite Symposia: Travere
17:30-18:30, Oranim 3 (ICC),Oranim 4 (ICC)

Poster Walk + Highest Ranked Posters
18:35-20:30, Exhibition Hall (ICC)

Failure to thrive, ichthyosis, deafness, and endocrinopathies in an infant with a novel biallelic AP1B1 mutation causing abnormal intracellular ATP7A trafficking
Raz Rotman, Edmond and Lily Safra Childrens Hospital, Sheba Medical Center, Israel

Survival, Cardiac, and Pulmonary Outcomes In Individuals with Attenuated MPS I Receiving Laronidase Enzyme Replacement Therapy: Data from The MPS I Registry
Roberto Giugliani, Universidade Federal do Rio Grande do Sul, Brazil

Newborn Screening Programs for Mucopolysaccharidoses Types I, II, IVA, and VI in Taiwan and the Application of Gene Variants | *Hsiang-Yu Lin, MacKay Memorial Hospital, Taiwan*

Identification of a novel deep intronic variant causing pseudo-exon inclusion in citrin deficiency and the development of a new drug for the variant by using splice-switching oligonucleotides
Eri Imagawa, The Jikei University School of Medicine, Japan

Daily Living Skills on the Vineland Adaptive Behavioral Scale Version 2 (VABS-II) in Neuronopathic Mucopolysaccharidosis Type II (MPS II)
Michelle Wood, Greater Ormond Street NHS Foundation Trust, UK

Role of Osteoclast biomarker (OSCAR) and pro-inflammatory cytokines in Gaucher disease
Margarita Ivanova, Lysosomal and Rare Disorders Research and Treatment Center, USA

Lysine hyposuccinylation in human MCAD deficient fibroblast cells alleviated with heptanoic and medium branched-chain fatty acids and in Acadm-/- mice with triheptanoin
Anuradha Karunanidhi, University of Pittsburgh, USA

Evaluation of Repeated Biotinidase Enzyme Activity and The Effect of BTD Gene p.Asp444His Variant on Enzyme Activity and Clinical Findings | *Fatma Tuba Eminoglu, Ankara University, Turkiye*

Therapeutic potential of intracerebroventricular recombinant human Heparan-N-Sulfatase enzyme replacement therapy in MPS IIIA mice
Aram Yang, Kangbuk Samsung Hospital, Sungkyunkwan University, School of Medicine, South Korea

Long term clinical outcome of patients diagnosed by newborn screening for Gaucher Disease in Italy
Vincenza Gragnaniello, Padua University Hospital, Italy

Clinical burden of classical homocystinuria in the United States: a retrospective analysis of Optum Market Clarity | *MAHIM JAIN, Johns Hopkins Medicine, USA*

The glycine N-acyltransferases, GLYAT and GLYATL1, contribute to the detoxification of isovaleryl-CoA: an in-silico and in vitro validation | *Stefan Kuhn, North-West University, South Africa*

Investigation of mucopolysaccharidoses by measuring disease-specific oligosaccharides by LC-MS/MS: A study on multiples matrices | *Gabrielle Dineck Iop, Hospital de Clnicas de Porto Alegre, Brazil*

Validation of a tandem mass spectrometry methodology for the analysis of urinary oligosaccharides and free sialic acid for the screening of lysosomal storage disorders
Blai Morales Romero, Hospital Clinic of Barcelona, Spain

Generation of iPSC-derived human neuronal progenitors for the study of GM1 gangliosidosis
Rodolfo Tonin, A.O.U. MEYER, Italy

Predicting correct IMD diagnosis using HPO phenotype association algorithms
Judith Jans, UMC Utrecht, The Netherlands

First in-human intracisternal dosing of RGX-181 (adeno-associated virus 9 / human tripeptidyl peptidase 1) for a 5-year-old child with late infantile neuronal ceroid lipofuscinosis type 2 (CLN2): 6 month follow-up | *Carolina Fischinger de Souza, HCPA, Brazil*

Newborn screening for Fabry disease in Japan: 16 years of experience
Takaaki Sawada, Kumamoto University, Japan

AI-based OMICs integration facilitates genetic diagnostics and provides a framework for automated analysis of clinical NGS data | *Dmitrii Smirnov, Technical University of Munich, Germany*

Dysmyelination or Demyelination: Investigating the link between lysosomal function and myelination in a murine model of Free Sialic Acid Storage Disorder
May Christine Malicdan, NHGRI, NIH, USA

Utility of genetic testing in an Israeli cohort of children with leukodystrophy
Ayelet Zerem, Dana-Dwek Children's Hospital, Tel Aviv Sourasky Medical Center, Israel

Trio genome sequencing and detection of metabolic disorders in an Israeli cohort of critically ill neonates, the The Israeli NICU-Genomics consortium
Daphna Marom, Tel Aviv Sourasky Medical Center, Israel

Cerebral folate deficiency due to DHFR mutation
Nasrin Hamed, Edmond and Lily Safra Children's Hospital, Sheba Medical Center, Tel Hashomer, Israel

Differential diagnosis of inherited metabolic disorders according to organ system involvement: a lesson from the knowledgebase | *Nenad Blau, University Children's Hospital, Switzerland*

AI-Powered Genomic Analysis: A New Frontier in Diagnosing Rare Diseases
Jaime Lopes, Cincinnati Children's Hospital, USA

De novo DNM1L mutations – an emerging mitochondrial cause of ultra-refractory status epilepticus in children | *Leo Arkush, Safra Children's Hospital, Sheba Medical Center, Israel*

A new animal model of Mucopolysaccharidosis IVA unravels potential new mechanisms involved in the disease pathogenesis | *Lucia De Stefano, Telethon Institute of Genetics and Medicine, Italy*

Novel homozygous variant in TOP3A in a pediatric patient resulting in ataxia, sensorimotor neuropathy and cardiomyopathy due to mitochondrial dysfunction that is partially rescued by ketogenic conditions in skin fibroblasts
Jaya Ganesh, Icahn School of Medicine at Mount Sinai, USA

The French Gaucher disease registry: clinical features, complications, and treatment trends of 688 patients

Nadia Belmatoug, Hôpital Beaujon, AP-HP.Nord, Université Paris Cité, France

Allow Natural Death (AND) in Inborn Errors of Metabolism

Carolina Fraga, Centro Hospitalar Universitario Santo Antonio, Portugal

A 12-month, longitudinal, intervention study examining a tablet protein substitute preparation in the management of tyrosinemia | *Anne Daly, Birmingham Children's Hospital, UK*

Assessment of the level of indepencence and knowledge of 13 young people with an inborn error of metabolism: Twelve months post transition to an adult clinic from a paediatric service

Anita Inwood, Queensland Lifespan Metabolic Medicine Service, Australia

SSIEM Symposium President´s Dinner

20:30-22:30 *(By invitation only)*

THURSDAY, AUGUST 31, 2023

Speed Mentoring
07:45-08:45, Oranim 2 (ICC)

Posters
07:45-15:30, Exhibition Hall (ICC)

PL5: Where big data meets small practices
08:45-10:15, Ussishkin (ICC)

08:45 **Newborn screening – finding more than we want to know?**
Rachel Rock, Sheba Medical Center, Israel

09:15 Use of the knowledge base IEMBase for the in silico diagnosis of inherited metabolic diseases
Francis Rossignol, National Human Genome Research Institute, USA

09:45 Big data meets small budget: Inborn errors of metabolism in developing countries
Anil B. Jalan, Nirman Metabolic Clinic of Mombai, India

Coffee Break
10:15-10:45, Exhibition Hall (ICC)

Coffee with the SSIEM JIMD Editors
10:15-10:45, Exhibition Hall (ICC)

Parallel Session: Phenylketonuria
10:45-12:15, Ussishkin (ICC)

10:45 Phase 3 APHENITY study: Oral sepiapterin for treatment of phenylketonuria
Nicola Longo, University of Utah, USA

11:00 Exploring the therapeutic potential of proteostasis regulators in phenylketonuria: Insights from the newly established phenylalanine hydroxylase proteostasis network
Luka Janina Haupt, University Medical Center Hamburg-Eppendorf, Germany

11:15 Iron supplementation links to reduced b-Phe levels in PKU patients: new evidences after a 3-year longitudinal randomized study
Raed Selmi, University of Milan, San Paolo Hospital, Italy

11:30 Safety, Tolerability and Proof-Of-Mechanism in Healthy Volunteers for JNT-517, a First-In-Class SLC6A19 Inhibitor for the Treatment of Phenylketonuria
Cary Harding, Oregon Health & Science University, USA

11:45 Brain hyperphenylalaninemia induces glucose hypometabolism: Metabolic findings in early treated adult PKU patients compared to controls
Friedrich Trefz, Metabolic Consulting, Germany

12:00 Metabolic and Biochemical Changes Associated with Long-Term Pegvaliase Therapy in Adults with Phenylketonuria
Rani Singh, Emory University School of Medicine, USA

Parallel Session: Novel diagnostic/laboratory methods including omics
10:45-12:15, Pincus (ICC)

10:45 Transcriptome profiles at different developmental stages in a zebrafish model of Classic Galactosemia

M. Estela Rubio-Gozalbo, Maastricht University Medical Center, The Netherlands

11:00 A multiplexed mass spectrometry assay to identify multiple inborn errors of cholesterol synthesis, metabolism and transport

Yuqin Wang, Swansea University, UK

11:15 A glycomic workflow for LC-MS/MS analysis of urine glycosaminoglycan biomarkers in mucopolysaccharidoses

Maria Blomqvist, Sahlgrenska University Hospital, Sweden

11:30 Diagnostic proficiency testing of untargeted metabolomics: pilot experience

Judith Jans, University Medical Center Utrecht, The Netherlands

11:45 Glycoproteomics unlocks novel and improved functional diagnostics for congenital disorders of glycosylation (CDG)

Purva Kulkarni, Radboud University Medical Center, The Netherlands

12:00 Untargeted metabolomics using direct-infusion mass spectrometry as a diagnostic modality for patients with inborn metabolic disorders

Anke Willems, University Medical Centre Utrecht, Netherlands

Parallel Session: Newborn Screening
10:45-12:15, Dulchin 1 (ICC)

10:45 A Pilot Study of Multiplexed Proteomic Newborn Screening for Wilson Disease and Inborn Errors of Immunity in WA State

Sihoun Hahn, University of Washington Seattle Children's Hospital, USA

11:00 Classical Galactosaemia Outcomes in a Screened Paediatric Population in the Republic of Ireland: A Fifty-Year Retrospective Study

Claire E Thompson, National Centre for Inherited Metabolic Disorders, Ireland

11:15 Towards recommendations for Newborn Screening for Classical Galactosemia – a world wide survey

Matthias Gautschi, Inselspital, University Hospital Bern, Switzerland

11:30 A digital-tier strategy based on machine learning methods improves specificity in newborn screening for isovaleric aciduria

Elaine Zaunseder, University Heidelberg, Germany

11:45 Universal Neonatal Screening for X-linked Adrenoleukodystrophy: Preliminary results of the pilot study carried out in Eastern Andalusia

Raquel Yayhaoui-Macias, Regional University Hospital of Malaga, Spain

12:00 Multiplex, biochemical newborn screening for an ever-increasing collection of treatable, neonatal diseases

Michael Gelb, University of Washington, USA

Parallel Session: Peroxisomal and Purines
10:45-12:15, Oranim 3 (ICC),Oranim 4 (ICC)

10:45 Suppression of de novo purine biosynthesis by exogenous purine supplementation as a treatment for AICA ribosiduria | *Julien H. Park, University of Münster, Germany*

11:00 Two girls with fast progressive leukoencephalopathy due to bi-allelic HMBS variants
Arnaud Vanlander, Ghent University Hospital, Belgium

11:15 Peroxisomes – from systematic screens in yeast to human diseases
Einat Zalckvar, Weizmann Institute of Science, Israel

11:30 Interim results from the NEXUS open-label registration study on the efficacy of leriglitazone in the treatment of childhood cerebral adrenoleukodystrophy
Angeles Garcia-Cazorla, Hospital Sant Joan De Deu, Spain

11:45 Investigating the role of Miglustat in the management of a patient with Tangier's Disease: An n-of-1 study with alternating periods of intervention and contro
Tarekegn Hiwot, University Hospital of Birmingham, UK

12:00 Symptomatic management in cerebrotendinous xanthomatosis: a single center case series and literature review
Brian Wishart, Massachusetts General Hospital / Spaulding Rehabilitation Hospital, USA

SSIEM JIMD Editors, JIMD Communicating Editors and JIMD Advisory Members Board Meeting
12:00-13:30, Oranim 2 (ICC) *(By invitation only)*

Lunch & Posters
12:15-13:30, Exhibition Hall (ICC)

Satellite Symposia: BioMarin
12:30-13:30, Pincus (ICC)

Satellite Symposia: PTC
12:30-13:30, Dulchin 1 (ICC)

Satellite Symposia: Sanofi
12:30-13:30, Oranim 3 (ICC),Oranim 4 (ICC)

Satellite Symposia: Sanofi
12:30-13:30, Oranim 3 (ICC),Oranim 4 (ICC)

Komrower Lecture
13:30-14:15, Ussishkin (ICC)

13:30 Molecular basis of phenotype expression in homocystinuria: where are we thirty years later?
Viktor Kožich, General University Hospital in Prague, Czech Republic

Parallel Session: Mitochondrial disorders II
14:15-15:30, Ussishkin (ICC)

14:15 Abnormal autophagy is a critical mechanism in TANGO2-related rhabdomyolysis
Hortense de Calbiac, Necker Institute, France

14:30 Association of 37 mitochondrial DNA genes with primary mitochondrial disease: standardized assessment using the ClinGen clinical validity framework
Shamima Rahman, UCL Great Ormond Street Institute of Child Health, UK

14:45 Pathological variants in TOP3A cause distinct disorders of mitochondrial and nuclear genome stability
Rob Taylor, Newcastle University, UK

15:00 Novel homozygous variants in PRORP expand the genotypic spectrum of combined oxidative phosphorylation deficiency 54
Nava Shaul Lotan, Hadassah University Hospital, Israel

15:15 Ketogenic diet as a potential new treatment for SPATA5-encephalopathy: in vitro and clinical assessment | *Alfonso Oyarzabal, Hospital Sant Joan De Deu, Spain*

Parallel Session: Organic Acidurias
14:15-15:30, Pincus (ICC)

14:15 Development of second-tier LC-MS/MS DBS method for the differential diagnosis of organic acidurias | *Malgorzata Rogozinska, University of Warsaw, Poland*

14:30 Methylmalonic acidemia, propionic acidemia, and cblC defect: comparing untargeted urine metabolomic profiles allows better disease understanding
Anna Sidorina, Bambino Gesu Children's Hospital, IRCCS, Italy

14:45 Pre-symptomatic diagnosis and optimized treatment lead to improved outcomes in early onset cobalamin C deficiency | *Irini Manoli, National Institutes of Health, USA*

15:00 The impact of liver trasplantation on targeted metabolomic profile in propionic acidemia: in vivo and ex vivo studies | *Carlo Dionisi-Vici, Bambino Gesù Children's Hospital IRCCS, Italy*

15:15 Deficiency of methylmalonyl-CoA mutase correlates with higher autophagy in HEK cells
Miriam Guera, University Children's Hosptial Zurich, Switzerland

Parallel Session: Clinical Studies & Outcomes II
14:15-15:30, Dulchin 1 (ICC)

14:15 Continued improvement in liver and lipid outcomes in clinical trials of olipudase alfa in children and adults with chronic acid sphingomyelinase deficiency treated for 2 to 6.5 years | *Robin Lachmann, National Hospital for Neurology and Neurosurgery, UK*

14:30 Transcranial Alternating Current Stimulation (tACS) as a novel treatment option: a proof of concept study in Adult Classic Galactosemia
M. Estela Rubio-Gozalbo, Maastricht University Medical Center, Italy

14:45 Validation of Wearable Devices to Measure Balance and Gait in Patients with X-Linked Adrenoleukodystrophy
Hemmo Yska, Amsterdam Medical Centers, The Netherlands

15:00 Prospective, multicenter validation of a simple blood test for the diagnosis of Glut1 deficiency syndrome | *Fanny Mochel, University Hospital Pitie-Salpetriere, France*

15:15 The AMETHIST phase 3 trial of venglustat in patients with GM2 gangliosidoses and related diseases: baseline characteristics | *Cynthia Tifft, National Institutes of Health, USA*

Parallel Session: Sulphur related and other amino acid disorders
14:15-15:30, Oranim 2 (ICC)

14:15 ACMSD deficiency a new disorder of tryptophan catabolism responsive to protein restriction
Saskia Wortmann, Paracelsus Medical University, The Netherlands

14:30 Pubertal origin of growth retardation in Inborn Errors of Protein Metabolism: A longitudinal cohort study | *Kanetee Busiah, Lausanne University Hospital, France*

14:45 Sulfide:quinone oxidoreductase deficiency presenting as acute hemorrhagic necrotizing encephalitis with cardiorespiratory failure
Tamara Zigman, University Hospital Center Zagreb, Croatia

15:00 Structural understanding of delta1-pyrroline-5-carboxylate synthetase (P5CS) deficiency
Vicente Rubio, Instituto de Biomedicina de Valencia, IBV-CSIC, Spain

15:15 Development of a robust high-throughput screenings assay for the evaluation of bacterial tyrosine ammonia lyases in the context of tyrosine-inherited metabolic disorders
Ine Nulmans, Vrije Universiteit Brussel, Belgium

Parallel Session: Innovative Therapies II
14:15-15:30, Oranim 3 (ICC),Oranim 4 (ICC)

14:15 Consistent long-term clinical benefit with govorestat treatment: results of the ACTION-Galactosemia Kids trial | *Jerry Vockley, University of Pittsburgh, USA*

14:30 Repurposing bempedoic acid as a therapeutic option in GSD type 1: from biochemical principles to first clinical data | *Anibh Das, Hannover Medical School, Germany*

14:45 Alleviation of a polyglucosan storage disorder by enhancement of autophagic glycogen catabolism | *Or Kakhlon, Hadassah Medical Center, Israel*

15:00 Limb girdle muscular disease caused by HMGCR mutation and statin myopathy treatable with mevalonolactone | *Yuval Yogev, Ben-Gurion University of the Negev, Israel*

15:15 Seeking personalised therapy for Niemann Pick Type C disease with a rare deep intronic mutation: a role for ASO-induced exon skipping?
Siyamini Sivananthan, Great Ormond Street Hospital, UK

SSIEM Networking Tours
15:30-18:30

SSIEM Networking Evening and Dinner Speech
20:00-23:00

FRIDAY, SEPTEMBER 1, 2023

Registration
07:45-08:15, Exhibition Hall (ICC)

PL6: Therapeutic updates – Advances in therapy modalities
08:15-09:45, Ussishkin (ICC)

08:15 Individualized antisense oligonucleotide therapies for genetic brain and eye diseases
Annemieke Aartsma-Rus, Medical Center of Leiden, The Netherlands

08:45 Development of gene therapies for inborn errors of metabolism
Simon Waddington, University College London, UK

09:15 Mitochondrial Augmentation Therapy (MAT)
Elad Jacoby, Sheba Medical Center, Israel

Posters
08:15-12:30, Exhibition Hall (ICC)

Coffee Break
09:45-10:15, Exhibition Hall (ICC)

Closing Session
10:15-12:30, Ussishkin (ICC)

10:15 Late Breaking News

11:45 Awards and Introduction to SSIEM 2024 at Porto

12:15 Closing Remarks

POSTERS

AMINO ACID DISORDERS

B Pitfall in Non-Ketotic Hyperglycinemia (NKH) Diagnosis
Rima Abu-Asaad, Rambam Medical Center, Israel

B Factors associated with poor outcomes in patients with maple syrup urine disease in a tertiary government hospital: a retrospective cohort study
CHRISTINE Mae AVILA, National Institutes of Health, Philippines

B Mitochondrial dysfunction in a disorder of transsulphuration: Cystathionine β-synthase deficiency
Mehmet Cihan Balci, Istanbul Medical Faculty Children's Hospital, Turkiye

B Disturbance of mitochondrial functions caused by N-acetylglutamate and N-acetylmethionine in brain of adolescent rats as a contributing cause of neurodegeneration in aminoacylase 1 deficiency
Vanessa Trindade Bortoluzzi, Universidade Federal Do Rio Grande Do Sul, Brazil

B N-Acetylglutamate and N-Acetylmethionine inhibit citric acid cycle enzyme and respiratory chain activities in brain of young rats: potential relevance for the neurological dysfunction in aminoacylase 1 deficiency
Vanessa Trindade Bortoluzzi, Universidade Federal Do Rio Grande Do Sul, Brazil

B Amino Acid Analyses of Plant Foods used in the Dietary Management of Inherited Amino Acid Disorders
Anita McDonald, Birmingham Children's Hospital, Birmingham, UK

B Neonatal ethylmalonic encephalopathy with neuroradiological lesions at birth: is it an in utero disease?
Alberto Burlina, University Hospital of Padua, Italy

B Bezafibrate prevents myelin alterations, neuroinflammation, and oxidative stress induced by sulfite intrastriatal administration in rats
Moacir Wajner, UFRGS, Brazil

B Succinylacetone reduces the antioxidant defenses and induces reactive nitrogen species generation in liver and kidney of developing rats
Moacir Wajner, UFRGS, Brazil

B Hydrogen sulfide impairs redox homeostasis and mitochondrial bioenergetics in the striatum of rats
Moacir Wajner, UFRGS, Brazil

B Hyperhomocysteinemia alters growth factors and neurotrophins in rat hippocampus: Neuroprotective role of ibuprofen and rivastigmine
Angela Wyse, Universidade Federal do Rio Grande do Sul, Brazil

CLINICAL STUDIES, PATIENT REPORTED OUTCOME MEASURES

B Exploring the Experiences of Females Living with Fabry Disease in Canada
Julia Alton, Canadian Fabry Association, Canada

B RESTORE, a phase 3 study to evaluate the effects of chenodeoxycholic acid adult and pediatric patients with cerebrotendinous xanthomatosis
John Bernat, University of Iowa, USA

B Management of Inherited Metabolic Diseases in France
Pascale De Lonlay, Filiere de sante maladies rares G2M, France

B Breaking bad news in paediatric metabolic disorders; Lessons learnt
Unai Diaz-Moreno Elorz, Great Ormond Street Hospital, UK

B Long-term follow-up of three pediatric patients with glycogen storage disease type 1b treated with SGLT2 inhibitor
Ana Drole Torkar, University Children's Hospital, University Medical Centre Ljubljana, Slovenia

B Long-term clinical evaluation of patients with alpha-mannosidosis – A multicenter study
Fatma Tuba Eminoglu, Ankara University, Turkiye

B IGAm index predicts long-term survival in patients with early-diagnosed inherited metabolic disorders
Fatma Tuba Eminoglu, Ankara University, Turkiye

B Clinical Characterization of Classical Homocystinuria due to Cystathionine β-Synthase Deficiency: Results from the ACAPPELLA Study
Can Ficicioglu, University of Pennsylvania, Perelman School of Medicine, USA

B Plasma Lyso-Sphingomyelin Levels Correlate with Baseline Disease and Decline with Olipudase Alfa Treatment in Clinical Trials of Adults and Children with Acid Sphingomyelinase Deficiency
Roberto Giugliani, Universidade Federal do Rio Grande do Sul, Brazil

B Safety of home administration of cipaglucosidase alfa + miglustat in late-onset Pompe disease: results from multiple clinical trials
Ozlem Goker-Alpan, Lysosomal and Rare Disorders Research and Treatment Center, USA

B Evaluation of Clinical and Potential Disease Modifying Effects of Pentosan Polysulfate Sodium in Subjects with Mucopolysaccharidosis I
Drago Bratkovic, Women's and Children's Hospital, South Australia, Australia

B Characteristics of patients with inborn errors of metabolism and incidence of these disorders in Slovenia - a nation-wide cross-sectional study
Urh Groselj, UMC - University Children's Hospital Ljubljana, Slovenia

B Avalglucosidase alfa safety and immunogenicity profile in alglucosidase alfa-experienced participants with Pompe disease: Pooled analysis of clinical trial data
Andreas Hahn, University Hospital Giessen, Germany

B Validation of a patient identification algorithm to estimate the prevalence of classical homocystinuria (HCU) in the United States (US)
Mahim Jain, Johns Hopkins Medicine, USA

B Breakthrough in the treatment of glycogen storage disease type 1b (GSD1b)
Magdalena kaczor, The Children's Memorial Health Institute, Poland

B Mini-COMET study: Safety and efficacy data after avalglucosidase alfa dosing for ≥145 weeks in patients with infantile-onset Pompe disease (IOPD) who had demonstrated clinical decline or sub-optimal response whilst receiving alglucosidase alfa
Priya Kishnani, Duke University Medical Center, USA

B A rare partnership: community and industry collaboration to shape the impact of real-world evidence on the rare disease ecosystem
Terri Klein, National MPS Society, USA

B Designing patient-oriented longitudinal disease registries for children with rare metabolic diseases in Canada
Michal Inbar-Feigenberg, The Hospital for Sick Children, Canada

B wrong diagnoses prior to the ultimate diagnosis of late-onset pompe disease: a multicenter experience
Tahseen Mozaffar, The University of California, USA

B Endogenous glucose production in glycogen storage disease type Ia estimated by a single oral dose of D-[6,6-2H2]-glucose: an investigator-initiated human pilot study
Alessandro Rossi, University Medical Center Groningen, The Netherlands

B Health-related quality of life and fatigue in children with Pompe disease
Hannerieke van Houten, Erasmus MC - Sophia Children's Hospital, Netherlands

B Insights into liver disease progression in glycogen storage disease IX γ2: A review of histology
Maheen Sheikh, Duke University, USA

B Brain-Type Creatine Kinase Release from Cultured Osteoclasts Exposed to Neridronate in Children Affected by Osteogenesis Imperfecta Type 1
Albina Tummolo, Children Hospital Giovanni XXIII Azienda Ospedaliero-Universitaria Consorziale, Italy

B First in-human intracisternal dosing of RGX-181 (adeno-associated virus 9 / human tripeptidyl peptidase 1) for a 5-year-old child with late infantile neuronal ceroid lipofuscinosis type 2 (CLN2): 6 month follow-up
Carolina Fischinger de Souza, Hospital de Clinicas de Porto Alegre, Brazil

B Evaluation of Repeated Biotinidase Enzyme Activity and The Effect of BTD Gene p.Asp444His Variant on Enzyme Activity and Clinical Findings
Fatma Tuba Eminoglu, Ankara University, Turkiye

B Clinical burden of classical homocystinuria in the United States:a retrospective analysis of Optum Market Clarity
Mahim Jain, Johns Hopkins Medicine, USA

B Daily Living Skills on the Vineland Adaptive Behavioral Scale Version 2 (VABS-II) in Neurono pathic Mucopolysaccharidosis Type II (MPS II)
Michelle Wood, Great Ormond Street NHS Foundation Trust, UK

B Utility of genetic testing in an Israeli cohort of children with leukodystrophy
Ayelet Zerem, Dana-Dwek Children's Hospital, Tel Aviv Sourasky Medical Center, Israel

DIETETICS AND NUTRITION

B A Dietician Experience with the Ketogenic Diet in Dihydrolipoamide Dehydrogenase Deficiency
Smadar - Yaala Abraham, Safra Children's Hospital, Sheba Medical Center, Tel Hashomer, Israel

B Management of lipoprotein lipase deficiency with medium-chain triglycerides: a retrospective chart review
Andreas Schulze, University of Toronto, Canada

B evaluation of the risk factors for noncommunicable diseases in patients with inborn errors of amino acid metabolism receiving nutrition therapy
Mehmet Cihan Balci, Istanbul Medical Faculty Children's Hospital, Turkiye

B Integrating metabolic dietetics in newborn screening continuity clinics: challenges and opportunities in a developing country
Jeanne Ruth Basas, Institutes of Human Genetics, National Institutes of Health, University of the Philippines Manila, Philippines

B Triheptanoin use in TANGO2-Related Disease (RD)
Heather Bausell, Ann & Robert H. Lurie Children's Hospital of Chicago, USA

B Glycogen storage disease Type IIIa with hypertrophic cardiomyopathy: Efficacy and safety of long- term ketogenic diet
Charlotte Mindermann, Hannover Medical School, Germany

B Determination of iron deficiency in patients with gluten-related diseases on an agliadin diet
Oleg Denesiuk, Bogomolets National Medical University, Ukraine

B Dietary management of fatty acid oxidation defects: easy biomarkers are needed to personalize treatment in the new era of expanded newborn screening
Alice Dianin, Azienda Ospedaliera Universitaria Integrata Verona, Italy

B A study on the understanding of classical homocystinuria by patients and caregivers
Ida Vanessa Doederlein Schwartz, Hospital de Clinicas de Porto Alegre, Brazil

B Biochemical and dietary evaluation of B vitamins deficiencies in patients with hepatic Glycogen Storage Diseases
Ida Vanessa Doederlein Schwartz, Hospital de Clinicas de Porto Alegre, Brazil

B Audit of maternal iodine intakes during pregnancy in PKU
Charlotte Ellerton, University College London Hospitals, UK

B Opinion of parents regarding early support and information in PKU
Anita McDonald, Birmingham Women's and Children's NHS Foundation Trust, UK

B Change in quality of diet and burden of care in children with PKU treated with sapropterin dihydrochloride: a longitudinal study
Maria Ines Gama, Birmingham Children's Hospital, UK

B Intestinal microbiota composition of children with glycogen storage type 1 patients
Sabire Gokalp, Gazi University Faculty of Medicine, Turkiye

B Classic galactosemia: relationship between calcium, vitamin D, and bone mineral density status
Esmeralda Martins, Centro Hospitalar Universitario De Santo Antonio, Portugal

B Phenylalanine Tolerance over Time in Phenylketonuria: A systematic review and meta-analysis
Alex Pinto, Birmingham Women's and Children's Hospital, UK

B Ketogenic Diet: make it possible for patients with metabolic disorders
Keren Porper, Safra Pediatric Hospital, Chaim Sheba Medical Center, Israel

B Effective Management of Severe Necrotising Dermatosis as a Side Effect of dietary restriction of protein in Glutaric acidemia-I(GA-1) & Homocystinuria
Ketki Kudalkar, Nirman, India

B Clinical case: early nutritional approach in two cases of type i glutaric acidury
Marta Suarez Gonzalez, Central University Hospital of Asturias, Spain

B Effects of Combine therapy Ketogenic diet and alglucosidase alfa on Creatine Kinase Levels and motor outcome in Infantile Pompe Disease: Case Series
Pelin Teke Kisa, Dr. Behçet Uz Children's Education and Research Hospital, Turkiye

B Optimized diet in a patient with familial chylomicronemia syndrome (FCS) with increased energy requirements receiving Volanesorsen
Alexandra Thajer, Medical University of Vienna, Austria

B Early manifestation of GLUT1 deficiency syndrome
Sara Via Dorembus, Edmond and Lily Safra Children's Hospital, Sheba Medical Center, Israel

B High-protein diet during gestation impairs behavior in rats offspring:
Could neuroinflammation and homocysteine levels be involved in this impairment?
Angela Wyse, Universidade Federal do Rio Grande do Sul, Brazil

B Clinical experiences in transitioning PKU patients from second to third stage protein substitutes: A global survey
Ozlem Yilmaz, Ankara Yildirim Beyazit University, Turkiye

B Phenylalanine free infant formula in patients with phenylketonuria: A retro spective study
Ozlem Yilmaz, Ankara Yildirim Beyazit University, Turkiye

B Phenylalanine-free infant protein substitute in the dietary management of phenylketonuria
Ozlem Yilmaz, Ankara Yildirim Beyazit University, Turkiye

B A 12-month, longitudinal, intervention study examining a tablet protein substitute preparation in the management of tyrosinemia
Anne Daly, Birmingham Children's Hospital, UK

DISORDERS OF FATTY ACID OXIDATION AND KETONE BODY METABOLISM

B Long-chain fatty acid oxidation disorder genotypes, clinical signs and symptoms from a gene panel sponsored program
Ida Vanessa Doederlein Schwartz, Hospital de Clinicas de Porto Alegre, Brazil

B Controlled Fasting Test in Pediatric Patients in the Era of Gene Panels
Carolina Fraga, Centro Materno-Infantil do Norte, Centro Hospitalar Universitario Santo Antonio, Portugal

B Early diagnosis by newborn screening (NBS) or prior family history is associated with improved visual outcomes of Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHADD) chorioretinopathy
Melanie Gillingham, Oregon Health & Science University, USA

B Clinical, Biochemical and Molecular Characteristics of Filipino patients with Very Long-Chain Acyl-CoA Dehydrogenase Deficiency (VLCADD) and Medium-Chain Acyl-CoA Dehydrogenase Deficiency (MCADD)
Ebner Bon Maceda, National Institutes of Health, University of the Philippines Manila, Philippines

B Carnitine-acylcarnitine translocate deficiency (CACTD) in the Vietnam referal center: genotype, phenotype and outcome
Khanh Nguyen Ngoc, Vietnam National Children's Hospital, Vietnam

B Cardiac Transplantation in Long-chain 3-hydroxyacyl-CoA dehydrogenase (LCHAD) deficiency with severe cardiomyopathy: a case report
Paulo Castro Chaves, São João University Hospital Centre, Portugal

B Development and characterization of a VLCAD/LCAD muscle double knock out mouse model
Bianca Seminotti, University of Pittsburgh School of Medicine, USA

B Comprehensive multi-parametric cardiac MRI (CMR) in mice with a mutation in the mitochondrial trifunctional protein β-subunit - a model of cardiac fibrosis
Eduardo Vieira Neto, University of Pittsburgh, USA

B Characterizing pathogenicity of exon and intron variants in very long-chain acyl-CoA dehydrogenase deficiency
Jerry Vockley, University of Pittsburgh School of Medicine, USA

B Genetic landscape findings from a comprehensive database of long-chain fatty acid oxidation disorder gene variants
Jerry Vockley, University of Pittsburgh School of Medicine, USA

B Genotype, phenotype and outcome of ketone synthesis defects due to mHS deficiency
Dung Vu Chi, Vietnam National Children's Hospital, Vietnam

B Pyruvate dehydrogenase interacts with electron transfer chain supercomplexes

Yudong Wang, University of Pittsburgh School of Medicine, USA

B Lysine hyposuccinylation in human MCAD deficient fibroblast cells alleviated with heptanoic and medium branched-chain fatty acids and in Acadm-/- mice with triheptanoin
Anuradha Karunanidhi, University of Pittsburgh, USA

DISORDERS OF PURINES, PYRIMIDINES, NUCLEIC ACIDS AND PORPHYRIAS

B Effect of Methione restricted diet in Molybdenum Cofactor Deficiency in a resource limited setting
Ketki Kudalkar, Navi Mumbai Institute of Research in Mental and Neurological Handicap (NIRMAN), India

B Biochemical Spectrum and Outcome of Acute Hepatic Porphyrias- a single center experience
Ketki Kudalkar, Navi Mumbai Institute of Research in Mental and Neurological Handicap (NIRMAN), India

B Dihydropyrimidine dehydrogenase deficiency caused by a novel intragenic large deletion in the DPYD gene
Anna Malekkou, The Cyprus Institute of Neurology and Genetics, Cyprus

B Clinical and genetic characteristics of a patient with phosphoribosylpyrophosphate synthetase 1 deficiency and a systematic literature review
Jaka Sikonja, University Children's Hospital, University Medical Centre Ljubljana, Slovenia

B Mild phenotype and late onset in molybdenum cofactor deficiency: a novel variant in MOCS2 gene in Roma Population
Blanka Stiburkova, Institute of Rheumatology, Czech Republic

DISORDERS OF VITAMINS, COFACTORS AND TRACE ELEMENTS

B A Southern African MADD cohort: clinical, biochemical, and genetic spectrum
Michelle Bisschoff, North-West University, South Africa

B Activity of NF-kB in lymphocyte populations in children with Wilson`s disease
Tatiana Bushueva, National Medical Research Center of Children's Health, Russia

B Pyridoxine-dependent epilepsy with neonatal onset: A Report of Two Cases
Martha Caterina Faraguna, Fondazione IRCCS San Gerardo Dei Tintori, Italy

B First late onset cerebral folate transporter deficiency with a novel mutation mimicking sspe and wilson's disease
Recep Kamil Kilic, Gazi University School of Medicine, Turkiye

B Biotinidase deficiency: Is redefinition for treatment necessary?
Huseyin Kutay Korbeyli, Istanbul University, Turkiye

B Clinical, biochemical and molecular features of a cohort of 8 patients with inherited disorders of vitamin B12 metabolism in a metabolic reference center
Goncalo Padeira, Hospital Dona Estefania, Portugal

B The importance of high doses of hydroxocobalamin in Brazilian cblC patients
Flavia Piazzon, University of Liege, Belgium

B Progressive myoclonus ataxia and resting tremor in hypoceruloplasminemia
Luca Pollini, Sapienza, University of Rome, Italy

B RNA analysis in the diagnosis of Wilson`s disease
Mikhail Skoblov, Research Centre for Medical Genetics, Russia

B Failure to thrive, ichthyosis, deafness, and endocrinopathies in an infant with a novel biallelic AP1B1 mutation causing abnormal intracellular ATP7A traf ficking
Raz Rotman, Edmond and Lily Safra Children's Hospital, Sheba Medical Center, Israel

GLYCOSYLATION DISORDERS/CDG, PROTEIN MODIFICATION DISORDERS

B Relationship between the metabolic state of copper and oxidative status in patients with and without GLUT1 deficiency in treatment with ketogenic diet
Veronica Cornejo, Inta, University of Chile, Chile

B Movement difficulties in children with Glucose Transporter 1 Deficiency (GLUT-1D) both off and on ketogenic diet
Sian Waller, Great Ormond Street Children's Hospital, UK

B SRD5A3-CDG restricted to inherited retinal disease phenotype
Hana Hansikova, Charles University and General University Hospital Prague, Czech Republic

B Congenital disorders of glycosylation (cdg) induce reduced biotinidase activity: supplementation of biotin as a new therapeutic option?
Nastassja Himmelreich, University Children's Hospital Heidelberg, Germany

B Retrospective Analysis Of Carbohydrate-Deficient Transferrin For CDG Screening: A Single Center Study
Ozge Ozgen, Istanbul University, Turkiye

B Phenotypic profile of inherited GPI deficiency disorders in Polish group of patients
Michal Patalan, Michal Patalan, Pomeranian Medical University, Poland

B A negative exome is not the end of the story
Batel Terespolskye, Hebrew University of Jerusalem, Israel

B Four year follow-up of mannose therapy in the first Belgian patient with MPI-CDG
Patrick Verloo, University Hospital Ghent, Belgium

B Two sibling cases of GPI deficiency presenting with a complex febrile seizure and intellectual disability
Yoriko Watanabe, Kurume University School of Medicine, Japan

INBORN ERRORS OF METABOLISM IN ADULTS

B Clinical experience of three cases of late-onset Krabbe disease
Gabriella Horvath, University of British Columbia, Canada

B Understanding the burden of classical homocystinuria (HCU) from the patient's perspective: A qualitative study
Danae Bartke, HCU Network America, USA

B Characterization of Gbe1ys/ys mouse model representing a phenotype similar to Adult Polyglucosan Body Disease (APBD)
Priya Kishani, Duke University, USA

B Activating mutation of the Glucokinase gene in Hyperinsulinemic Hypoglycemia: phenotype and genotype in 9 adult patients
Claire Douillard, Lille University Hospital, France

B A Large retrospective case series of managmnent of mothers with hyperphenylalaninaemia during pregnancy
Tarekegn Hiwot, University Hospital of Birmingham, UK

B Late diagnosis of childhood-onset hypophosphatasia in an adult with recurrent fractures: the impact of enzyme replacement therapy
Michel Hochuli, Inselspital, Bern University Hospital, University of Bern, Switzerland

B The landscape of long-term problems in adult patients with glycogen storage disorder type I
Michel Hochuli, Inselspital, Bern University Hospital, University of Bern, Switzerland

B Late-onset symptomatic hyperprolactinemia in 6-pyruvoyl-tetrahydropterin synthase deficiency
Wuh-Liang Hwu, National Taiwan University Hospital, Taiwan

B Pregnancy, maternal and child health in women with inherited metabolic disorders
Meryem Karaca, İstanbul Medical Faculty, Turkiye

B Cardiometabolic risk factors and muscle quality in adult patints with inherited metabolic diseases
Luis M. Luengo-Perez, University of Extremadura / Badajoz University Hospital, Portugal

B Clinical features of glucose transporter type 1 deficiency syndrome in a Portuguese Reference Centre
Maria Carmo Macario, Centro Hospitalar e Universitario de Coimbra, Portugal

B Hyperammonemia in adults: try to think outside the box
Francesca Maria Menni, Fondazione IRCCS Ca' Granda Ospedale Maggiore, Italy

B Dilated cardiomyopathy in adult propionic acidemia improved by adjustment of nutrition and heart failure treatment drugs: A case report
Yoko Nakajima, Fujita Health University School of Medicine, Japan

B Development of tools to facilitate the diagnosis of hereditary fructose intolerance
Bianca Panis, Maastricht University Medical Center, The Netherlands

B Imprinted cell memory in glycogen storage disorder 1a patients`fibroblasts
Uri Sprecher, Tel Aviv University, Israel

B The cognitive and mental health support of adults with Inherited Metabolic Diseases-a review of the newly developed neuropsychology service in one tertiary Metabolic Centre
Adrian Heald , Salford Royal NHS Foundation Trust, UK

B The relationship between metabolic control and cardiovascular risk factors in adult early-treated classic phenylketonuria patients
Kata Rebeka Utassy, Semmelweis University, Hungary

INNOVATIVE THERAPIES SUCH AS RNA-BASED THERAPY, GENE THERAPY AND REGENERATIVE MEDICINE

B Updates on the FBX-101 RESKUE Phase I/II Gene Therapy Clinical Trials for Patients with Infantile Krabbe Disease
Maria Escolar, University of Pittsburgh, USA

B Updates on the FBX-101 REKLAIM Phase Ib Gene Therapy Clinical Trials for Patients with Infantile and Late-Infantile Krabbe Disease
Maria Escolar, University of Pittsburgh, USA

B AT845 gene replacement therapy for Late Onset Pompe disease: preliminary muscle biomarkers and histopathology results from FORTIS, a phase1/2 open-label clinical study
Michael Lawlor, Medical College of Wisconsin, USA

B An AAV-mediated liver-directed gene therapy metabolically corrects alkaptonuria in an Hgd deficient mouse model
Sien Lequeue, Vrije Universiteit Brussels, Belgium

B Natural history of disease progression in GSD IX γ2 mouse model and a long-term follow-up study of AAV gene therapy
Priya Kishani, Duke University School of Medicine, USA

B What are the present obstacles to the incorporation of authorized cell & gene therapies into clinical practice?
Cecilia Marinova, Medasol Outpatient Clinic, Czech Republic

B Neonatal AAV8 gene therapy successfully treats severe MSUD in Bckdhb-/- mice
Clement Pontoizeau, Necker-Enfants Malades Hospital, France

B A Novel Muscle VLCAD/LCAD Double Knock Out Mouse Model Explores Very Long Chain Acyl-CoA Dehydrogenase (VLCAD) Deficiency Induced Skeletal Muscle Myopathy and Its Therapies
Jerry Vockley, UPMC Children's Hospital of Pittsburgh, USA

LYSOSOMAL DISORDERS

B Evaluating glycosphingolipids on dried blood spots samples of Gaucher disease patients by LC/MS-MS
Pilar Giraldo, Fundación para el Estudio y la Terapéutica de la Enfermedad de Gaucher y Otras Lisosomales (FEETEG), Spain

B A Novel Multiplex approach for determining anti-drug antibodies in Fabry Disease
Tomas Baldwin, University College London, UK

B A retrospective and prospective multicenter observational study of bone MRI changes in patients with type 1 Gaucher disease treated with velaglucerase alfa: the EIROS study
Nadia Belmatoug, Beaujon Hospital, France

B Impact of PEGylated COVID-19 vaccination on patient tolerability to peguni galsidase alfa – a new PEGylated enzyme replacement therapy for Fabry disease
John Bernat, University of Iowa, USA

B Pooled safety profile of pegunigalsidase alfa: an analysis of data from 142 patients in the pegunigalsidase alfa clinical program
John Bernat, University of Iowa, USA

B The importance of examining skeletal x-rays for dysostosis multiplex in the early diagnosis of mucopolysaccharidosis
Huseyin Bilgin, Diyarbakir Children's Hospital, Turkiye

B Pseudo-polymorphic variant c.121-210CT in the GALNS gene causes cryptic exon activation and is associated with Mucopolysaccharidosis type IVA
Igor Bychkov, Federal State Budgetary Institution, Russia

B The multiomic landscape of Mucopolysaccharidosis IIIB models
Marianna Caterino, University of Naples Federico II, Italy

B Underlying neuropathophysiology of mucopolysaccharidosis type II
Chong Kun Cheon, Pusan National University Children's Hospital, South Korea

B Towards personalized medicine in cystinosis: measuring cystine levels in leucocytes and cysteamine concentration in blood
Anibh Das, Hannover Medical School, Germany

B Atypical parkinsonism and the relationship of lysosomal disease variants
Ida Vanessa Doederlein Schwartz, Hospital de Clinicas de Porto Alegre, Brazil

B Correlation between cognitive function and brain metabolites in late-infantile metachromatic leukodystrophy
Mireia del Toro, Vall d'Hebron University Hospital, Spain

B Natural history and clinical characteristics of the patients with acid sphingomyelinase deficiency in the era of enzyme replacement therapy: Single Center Experience
Asli Durmus, İstanbul University, Turkiye

B ELIKIDS: baseline characteristics from the eliglustat substrate reduction therapy trial in children with Gaucher disease type 1 or type 3
Pilar Giraldo, Hopital Quirnsalud Zaragoza and FEETEG, Spain

B A prospective, longitudinal study of neurological disease trajectory in chil dren with late-infantile and juvenile-onset GM1 or GM2 gangliosidoses (PRONTO): Interim results
Roberto Giugliani, Universidade Federal do Rio Grande do Sul, Brazil

B An integrated analysis to evaluate neurocognitive function in individuals with mucopolysaccharidosis II (MPS II) following long-term treatment with pabinafusp alfa
Roberto Giugliani, Universidade Federal do Rio Grande do Sul, Brazil

B Vestronidase Alfa for the treatment of mucopolysaccharidosis VII (MPS VII): Updated results from a novel, longitudinal, multicenter disease monitoring program (DMP)
Roberto Giugliani, Universidade Federal do Rio Grande do Sul, Brazil

B Global Variations in Diagnostic Methods and Epidemiological Estimates in Pompe Disease: Findings from a Systematic Review
Roberto Giugliani, Universidade Federal do Rio Grande do Sul, Brazil

B An Alternative for Early Detection of Cardiac Involvement in Gaucher Type 1 Disease: Speckle Tracking Echocardiograpy
Sabire Gokalp, Gazi University Faculty of Medicine, Turkiye

B Safety and tolerability of pegunigalsidase alfa: Insights from a single site experience from the Expanded Access Program in the United States
Ozlem Goker-Alpan, Lysosomal and Rare Disorders Research and Treatment Center, USA

B Treatment of infants and very young children with Gaucher disease with velaglucerase alfa: a single-center experience
Ozlem Goker-Alpan, Lysosomal and Rare Disorders Research and Treatment Center, USA

B Oxidative and inflammatory stress parameters in patients with lysosomal acid lipase deficiency
Roberto Giugliani, Universidade Federal do Rio Grande do Sul, Brazil

B Qualitative study in adult and caregiver MPSI, II and VI patients : Understanding their challenges, needs and expectations
Nathalie Guffon, HFME Hospital, HCL, France

B Peripheric neurodegeneration biomarkers in Niemann-Pick type C1 disease
Roberto Giugliani, Universidade Federal do Rio Grande do Sul, Brazil

B Nanoparticulated β-Cyclodextrin improves cholesterol accumulation and mitochondrial function in Niemann-Pick type C disease
Roberto Giugliani, Universidade Federal do Rio Grande do Sul, Brazil

B Early detection and follow-up of patients with Fabry disease. Approaching new biomarkers
Alvaro Hermida, University of Santiago de Compostela, Spain

B Long-term efficacy of velmanase alfa treatment in patients with alpha mannosidosis: updated integrated analysis of data from phase I/II, III, and follow-up clinical trials
Benedicte Heron, Armand Trousseau-La Roche Guyon University Hospital, Sorbonne University, France

B Hematopoietic stem cell transplantation in three infants with Hunter Syndrome: a good treatment option for the neuronopathic form
Dafne Horovitz, Instituto Fernandes Figueira - Fiocruz, Brazil

B Identifying outcomes that matter: developing a core outcome set for pediatric mucopolysaccharidoses
Michal Inbar-Feigenberg, Hospital for Sick Children, Canada

B A 10-year experience with high-dose ambroxol in combination with enzyme replacement therapy for neuropathic Gaucher disease
Hyunwoo Bae, University of Ulsan College of Medicine, South Korea

B Incidence and management of Adverse events of Enzyme replacement therapy in Hunter syndrome
JiHoon Hwang, Sungkyunkwan University School of Medicine, South Korea

B MicroRNA expression profiling in Fabry mice to identify therapeutic target for Fabry disease-related fibrosis
Hae Soon Kim, College of Medicine, Ewha Womans University, South Korea

B Genetic characteristics of Fabry disease in a Korean population: A systematic review
Ji-Hee Kim, St. Vincent's Hospital, College of Medicine, The Catholic University of Korea, South Korea

B The benefit of lyso-Gb3Cer biomarker for screening and therapy monitoring of Fabry disease: Study in a group of Czech patients
Ladislav Kuchar, Charles University Prague, Czech Republic

B Correlation of lyso-Gb3Cer biomarker to X-chromosome inactivation in females with Fabry disease and carriers of GLA variants of uncertain significance
Ladislav Kuchar, Charles University Prague, Czech Republic

B Endothelial to Mesenchymal Transition in Gaucher Disease and Role of High-Dose Ambroxol
Beom Hee Lee, Asan Medical Center Children's Hospital, University of Ulsan College of Medicine, South Korea

B Phenotypic characterization of Alpha-mannosidosis patients in Portugal
Maria Carmo Macario, Centro Hospitalar e Universitario de Coimbra, Portugal

B Measurement of Galactocerebrosidase (GALC) activity in serum and cerebrospinal fluid (CSF) in support of clinical gene therapy trials for Krabbe disease
Dietrich Matern, Mayo Clinic, USA

B Psychosine analysis in dried blood spots and red blood cells from patients with Krabbe disease
Dietrich Matern, Mayo Clinic, USA

B IgG glycosylation in patients with alpha-mannosidosis and the impact of enzyme replacement therapy
Angela Messina, Istituto per i Polimeri, Compositi e Biomateriali, (IPCB), Italy

B Anesthesia for patients with mucopolysaccharidosis – Analysis of over 600 cases and its learnings in comparison with published data
Florian B Lagler, Paracelsus Medical University, Austria

B 104-week efficacy and safety of cipaglucosidase alfa+miglustat in patients with late-onset Pompe disease previously treated with alglucosidase alfa
Tahseen Mozaffar, University of California, USA

B Effect size analysis of cipaglucosidase alfa + miglustat versus alglucosidase alfa in ERT-expe rienced adults with late-onset Pompe disease in PROPEL
Tahseen Mozaffar, University of California, USA

B Whole blood transcriptomic profiling in the interpretation of variable pheno type presentation in Fabry Disease
Omer Murik, Shaare Zedek Medical Center, Israel

B Multiorgan involvement in females with Fabry disease: results from 2 phase III trials and the followME registry
Peter Nordbeck, University Hospital Würzburg, Germany

B Long-term monitoring of cardiac involvement under migalastat treatment using magnetic resonance tomography in Fabry disease
Albina Nowak, University Hospital Zurich, Switzerland

B Facial dysmorphologies in Mucopolysaccharidosis type IVA: exploring the morphometry and ancestry component
Harry Pachajoa, Universidad Icesi, Fundacion Valle del Lili, Colombia

B Association between heparan sulfate non-reducing ends and clinical symptom score in patients with severe mucopolysaccharidosis type 1(MPS1)
Marzia Pasquali, University of Utah, USA

B LYSO-GB3 normalization in fabry disease treated patients
Fernando Javier Perretta, Fresenius Medical Care Escobar, Argentina

B Measurement of lysosphingolipids in dried blood spots by LC-MSMS: a useful tool for the diagnosis of sphingolipidoses
Magali PETTAZZONI, Hospices Civils de Lyon, France

B Using machine learning to develop an algorithm for early diagnosis of Gaucher disease
Shoshana Revel-Vilk, Shaare Zedek Medical Center, Israel

B Pregnancy outcomes in imiglucerase-treated patients with Gaucher disease: real-world data from the International Collaborative Gaucher Group (ICGG) Gaucher Registry Pregnancy Sub-Registry
Shoshana Revel-Vilk, Shaare Zedek Medical Center, Israel

B Reduced renal function and other factors influence the measured level of glycosaminoglycans (GAGs) and could lead to diagnostic misinterpretation
Cathrin Lytomt Salvador, Oslo University Hospital, Norway

B Genotype/phenotype correlations in the patients with alpha-mannosidosis in Ukraine
Nataliia Samonenko, National Children's Specialized Hospital "Okhmatdyt", Ukraine

B Analysis of gut microbiota in patients with late-onset Pompe disease
Annalisa Sechi, University Hospital of Udine, Italy

B Study of the development and involvement of Neutrophil Extracellular Traps (NETs) in vascular complications in Lysosomal disorders
Pilar Giraldo, Fundación Española para el Estudio y Terapéutica de la enfermedad de Gaucher y otras lisosomales (FEETEG), Spain

B Leukocyte Chondroitin Sulfate (CS) and Dermatan Sulfate (DS) as Biomarkers of Intracellular Glycosaminoglycans (GAG) Accumulation in Patients with Mucopolysaccharidosis Type VI
Young Bae Sohn, Ajou University Hospital, South Korea

B Rapid, accurate and comprehensive diagnostic method for the detection of Neuronal Ceroid Lipofuscinosis Type 2 (CLN2) Disease using long-read third-generation sequencing technology
Betul Teker, Istanbul University, Turkiye

B Living with Pompe disease: results from a qualitative interview study with pediatric patients and their caregivers
Moritz Ilan Truninger, University Children's Hospital Zurich, University of Zurich, Switzerland

B Association between lysosomal hydrolase activity and LRRK2 kinase activity in induced pluripotent stem cell-derived dopaminergic neurons of patients with GBA-associated Parkinson's disease
Tatiana Usenko, Petersburg Nuclear Physics Institute named by B.P.Konstantinov of NRC Kurchatov, Russia

B Mitochondrial biogenesis and autophagy induction in the cerebral cortex of mucopolysaccharidosis type II mice
Moacir Wajner, UFRGS, Brazil

B Pooled analysis of the effect of pegunigalsidase alfa on renal function: Data from 113 patients in the pegunigalsidase alfa clinical trial program
David Warnock, The University of Alabama at Birmingham, USA

B Evidence-based Individual Treatment Trials with Immunomodulatory Drugs in Mucopolysaccharidosis
Anna-Maria Wiesinger, Paracelsus Medical University, Austria

B Development of Suspicion Index Tool to Aid Diagnosis of ASMD Disease
Anna-Maria Wiesinger, Paracelsus Medical University, Austria

B Individual Treatment Trials – Do Experts Know and Use this Option to Improve the Treatability of Mucopolysaccharidosis?
Anna-Maria Wiesinger, Paracelsus Medical University, Austria

B Brain MRI pattern in VPS11 hypomyelinating leukodystrophy
Ayelet Zerem, Dana-Dwek Children's Hospital, Tel Aviv Sourasky Medical Center, Israel

B A new animal model of Mucopolysaccharidosis IVA unravels potential new mechanisms involved in the disease pathogenesis
Lucia De Stefano, Telethon Institute of Genetics and Medicine, Italy

B Survival, Cardiac, and Pulmonary Outcomes In Individuals with Attenuated MPS I Receiving Laronidase Enzyme Replacement Therapy: Data from The MPS I Registry
Roberto Giugliani, Universidade Federal do Rio Grande do Sul, Brazil

B Long term clinical outcome of patients diagnosed by newborn screening for Gaucher Disease in Italy
Vincenza Gragnaniello, University Hospital of Padua, Italy

B Role of Osteoclast biomarker (OSCAR) and pro-inflammatory cytokines in Gaucher disease
Margarita Ivanova, Lysosomal and Rare Disorders Research and Treatment Center, USA

B Dysmyelination or Demyelination: Investigating the link between lysosomal function and myelination in a murine model of Free Sialic Acid Storage Disorder
May Christine Malicdan, NIH Undiagnosed Diseases Program, National Human Genome Research Institute, National Institutes of Health, USA

B The French Gaucher disease registry: clinical features, complications, and treatment trends of 688 patients
Nadia Belmatoug, Beaujon Hospital, France

B Generation of iPSC-derived human neuronal progenitors for the study of GM1 gangliosidosis
Rodolfo Tonin, Meyer Children's Hospital IRCCS, Italy

B Therapeutic potential of intracerebroventricular recombinant human Heparan-N-Sulfatase enzyme replacement therapy in MPS IIIA mice
Aram Yang, Sungkyunkwan University, School of Medicine, South Korea

METABOLIC MYOPATHIES

B Deoxynucleoside therapy for thymidine kinase 2-deficient myopathy: clinical case
Irina Artamonova, Almazov National Medical Research Centre, Russia

B Neutral lipid storage disease with myopathy (NLSDM), with cardiac and hepatic involvement: A 23 year evolution and the identification of a new PNPLA2 variant: case report
M Teresa Cardoso, University Hospital São João, Portugal

B Favorable response in muscle strength and endurance with chronic exercise in patients with metabolic myopathy
Tanyel Zubarioglu, Istanbul University-Cerrahpasa, Turkiye

B Defining the atlas of mitochondrial haplotypes and their regulation on T cell activation
Angi Zenab, Sheba Medical Center, Israel

B Mitochondrial aminoacyl- tRNA synthetase deficiency as a new cause of primary Pediatric Intestinal Pseudo-Obstruction
Barbara Siri, Bambino Gesu Children Hospital IRCCS, Italy

B Pathogenic variants of the coenzyme A biosynthesis-associated enzyme phosphopantothenoylcysteine decarboxylase cause autosomal-recessive dilated cardiomyopathy
Beln Perez, Centro de Biología Molecular Severo Ochoa, Universidad Autónoma de Madrid, Spain

B Comprehensive Intra- and Extracellular Metabolic Investigation of Perfused Fibroblasts by Real-time NMR Allows for Discrimination of Mitochondrial Defects
Christian Urzi, University of Bern, Switzerland

B Investigation of bioenergetic and metabolic adaptations under galactose stress in CI, CV and MDH2 deficient fibroblasts using metabolic flux and HR-MAS NMR analysis
Christoph Meyer, University of Bern, Switzerland

B Novel variant in COQ4 causes developmental delay, regression, epilepsy and cardiomyopathy associated with CoQ10 deficiency
Elene Kirtadze, Galil Medical Center, Israel

B King Denborough syndrome in South African congenital myopathy
patients is associated with an autosomal recessive STAC3 variant and secondary mitochondrial dysfunction
Francois van der Westhuizen, North-West University, South Africa

B A homozygous NDUFA6 variant associated with alopecia, neutropenia, growth retardation and encephalopathy and isolated complex I deficiency
Yoav Zehavi, Emek Medical Center, Israel

B What is a diagnosis worth? The MitoCope-study on the psychosocial experience of parents of children with a mitochondrial disease
Saskia B. Wortmann, University Children's Hospital, Austria

B Towards a Novel Target-Based Small Molecule Therapeutics for Pyruvate Dehydrogenase Complex Deficiency due to Common Recurrent E1α Amino Acid Replacements
Jirair K. Bedoyan, University of Pittsburgh School of Medicine, USA

B Novel bi-allelic variants in COA7 associated with isolated cytochrome c oxidase deficiency and mitochondrial cardiomyopathy
Lucie Taylor, Newcastle University, UK

B Mitochondrial dysfunction and thiamine response in a child with TELO2 mutation
Maria Novelli, Sapienza University, Italy

B Machine Learning can Identify Newborns with Energy Metabolism Conditions
Rebecca Ganetzky, Children's Hospital of Philadelphia, USA

B Genotype phenotype correlation in DLD deficiency based on mitochondrial function
Yarden Haham, Sheba Medical Center, Israel

B Anastrozole modulates mitochondrial activity via Inhibitory action on mitochondrial permeability transition pore opening : An Initial Perspective on repurposive drug therapeutic
Somesh Kumar, Maulana Azad Medical College, India

B Barth syndrome: lessons in metabolism from cellular models
Yana Sandlers, Cleveland State University, USA

B Interrogating the role of COA5 in mitochondrial cytochrome c oxidase biogenesis
Rob Taylor, Newcastle University, UK

B De novo DNM1L mutations - an emerging mitochondrial cause of ultra-refractory status epilepticus in children
Leo Arkush, Safra Children's Hospital, Sheba Medical Center, Israel

B Novel homozygous variant in TOP3A in a pediatric patient resulting in ataxia, sensorimotor neuropathy and cardiomyopathy due to mitochondrial dysfunction that is partially rescued by ketogenic conditions in skin fibroblasts
Jaya Ganesh, Icahn School of Medicine at Mount Sinai, USA Neurotransmitter and creatine related disorders

B Autosomal recessive GTP-cyclohydrolase I deficiency spectrum: a severe case without hyperphenylalaninemia
Filippo Manti, University of Rome La Sapienza, Rome, Italy

B Improved outcomes in early treated GAMT deficiency – a sibling study
Liora Caspi, Hospital for Sick Children, Canada

B Unexplained cerebrospinal fluid findings in GABA transaminase deficiency
Unai Diaz-Moreno Elorz, Great Ormond Street Hospital, UK

B "Standing in the Shoulder of Giants": Integrating Biochemical and Genomic Approaches in the Investigation of Neurotransmitters Disorders
Charles Lourenco, Faculdade De Medicina De Sao Jose Do Rio Preto (Famerp), Brazil

B DNAJC12 defect a new neurodevelopmental disorder associated with parkinsonism-dystonia
Filippo Manti, University of Rome La Sapienza, Rome, Italy

B Metabolic testing in the differential diagnosis of childhood onset movement disorders: when GLUT1 deficiency meets Segawa disease
Giacomina Ricciardi, Umberto I Hospital, Italy

B Functional characterization of DOPA Decarboxylase variants found in Polish patients with L-amino acid decarboxylase deficiency
Agnieszka Magdalena Rygiel, Institute of Mother and Child, Poland

B Monitoring changes in intracellular creatine using an AGAT-luciferase reporter provides clues about regulatory mechanism in creatine homeostasis
Andreas Schulze , Hospital for Sick Children, Canada

B Cerebral folate deficiency due to DHFR mutation
Nasrin Hamed, Edmond and Lily Safra Children's Hospital, Sheba Medical Center, Tel Hashomer, Israel

NEW DISEASES

B A rare disease with neurodevelopmental delay and recurrent rhabdomyolysis: a case report of trappc2l-related disorder
Niccolo' Campagna, University of Florence, Italy

B Sodium-dependent multivitamin transporter defect, a new case characterized by an unusual cardiac presentation
Dominique Roland, Institute of Pathology and Genetics, Belgium

NEWBORN SCREENING

B Newborn detection of two Spanish cases of D- bifunctional protein deficiency: the importance of X-ALD Newborn Screening
Maria Isabel Cabrera Gonzalez, Malaga Regional University Hospital, Spain

B Portuguese Neonatal Screening Programme: a retrospective cohort study of 18 years of MS/MS
Maria Miguel Goncalves, National Institute of Health Doutor Ricardo Jorge, Portugal

B Expanded metabolic newborn screening in catalonia: 10 years of experience
Jose Manuel Gonzalez De Aledo Castillo, Hospital Clinic De Barcelona, Spain

B Predicting phenotypes in metachromatic leukodystrophy: implications for newborn screening
Samuel Groeschel, University Children's Hospital of Tübingen, Germany

B A New biochemical marker as a game changer in Hyperphenylalaninemia Newborn Screening
Nitsan Haham, Sheba Tel Hashomer Hospital, Israel

B 22-year Follow-up of Extended Newborn Screening for Metabolic and Endocrine Disorders
Sook Za Kim, Korea Genetics Research Center, South Korea

B Developing a Novel Method for Glycosaminoglycan Analysis as a Second-Tier Test in Newborn Screening for Mucopolysaccharidosis
Hironori Kobayashi, Shimane University Hospital, Japan

B Retrospective Audit of Inherited Metabolic Disease Diagnoses: Implications for Newborn Screening in South Africa
Sarah Lampert, University of Cape Town, South Africa

B A case of 3-methylglutaconic aciduria type I detected by expanded neonatal screening in Ukraine
Nataliia Olkhovych, National Children Hospital Ohmatdyt, Ukraine

B Uniform gene list for genomic newborn screening: the time is now
Flavia Piazzon, University of Liege, Belgium

B Diagnosis of inborn errors of metabolism within the expanded newborn screening in the Balearic Islands (Spain)
Montse Pons, Hospital Universitari Son Espases, Spain

B A Pilot Project for Newborn Screening of four Lysosomal Storage Diseases in Campania region (Italy) using digital microfluidics
Margherita Ruoppolo, Università Degli Studi Di Napoli Federico II, Italy

B Inherited Metabolic Disorders and Primary Immunodeficiencies in Indigenous Populations of Southern Brazil
Leonardo Simao Medeiros, Hospital De Clinicas De Porto Alegre (HCPA), Brazil

B Elevated orotic acid with normal citrulline on NBS – consider hyperornithinemia-hyperammonemia-homocitrullinuria (HHH)
Galit Tal, Ruth Rappaport Children's Hospital, Rambam Medical Center, Israel

B Possible pitfalls in the diagnosis of Multiple Acyl-CoA Dehydrogenase Deficiency at Expanded Newborn Screening Program. The importance of the molecular characterization
Margherita Ruoppolo, University "Federico II", Italy

B Newborn Screening Programs for Mucopolysaccharidoses Types I, II, IVA, and VI in Taiwan and the Application of Gene Variants
Hsiang-Yu Lin, Mackay Memorial Hospital, Taiwan

B Trio genome sequencing and detection of metabolic disorders in an Israeli cohort of critically ill neonates, the The Israeli NICU-Genomics consortium
Daphna Marom, Tel Aviv Sourasky Medical Center, Israel

B Newborn screening for Fabry disease in Japan: 16 years of experience
Takaaki Sawada, Kumamoto University, Japan

NOVEL DIAGNOSTIC/LABORATORY METHODS INCLUDING OMICS

B Pterin profiling in serum, dry blood spot and urine using LC-MS/MS in patients with hyperphenylalaninemia
Harun Bayrak, Gazi University, Turkiye

B Multi-OMICs approach to improving diagnosis in metabolic disorders
Peter Bauer, Universitätsmedizin Rostock, Germany

B Genetic variants in the 5` untranslated regions cause hereditary diseases
Alexandra Filatova, Research Centre for Medical Genetics, Russia

B The diagnostic yield of critical sample and elective fasting test in children after hypoglycemic event
Ran Hazan, Soroka Medical Center, Israel

B Topology and machine learning based analysis of untargeted metabolomics profiles of patients with Propionic Aciduria reveal novel insights into the disease progression
Purva Kulkarni, Radboud University Medical Center, The Netherlands

B Modelling inborn errors of redox metabolism using patient-derived lymphoblastoid cell lines
Julien H. Park, University of Münster, Germany

B Comprehensive diagnostic approach for patients with inborn phosphate metabolism damage
Margarita Sharova, Research Centre for Medical Genetics, Russia

B Characterizing the Metabolomic Profile of Aicardi Goutières syndrome
Brian Shayota, University of Utah, USA

B Differential diagnosis of inherited metabolic disorders according to organ system involvement: a lesson from the knowledgebase
Nenad Blau, University Children's Hospital, Switzerland

B Investigation of mucopolysaccharidoses by measuring disease-specific oligosaccharides by LC-MS/MS: A study on multiples matrices
Gabrielle Dineck Iop, Hospital de Clinicas de Porto Alegre, Brazil

B Predicting correct IMD diagnosis using HPO phenotype association algorithms
Judith Jans, University Medical Center Utrecht, The Netherlands

B AI-Powered Genomic Analysis: A New Frontier in Diagnosing Rare Diseases
Jaime Lopes, Cincinnati Children's Hospital, USA

B Validation of a tandem mass spectrometry methodology for the analysis of urinary oligosaccharides and free sialic acid for the screening of lysosomal storage disorders
Blai Morales Romero, Hospital Clínic of Barcelona, Spain

B AI-based OMICs integration facilitates genetic diagnostics and provides a framework for automated analysis of clinical NGS data
Dmitrii Smirnov, Technical University of Munich, Germany

NURSING IN METABOLIC DISORDERS

B Nursing experiences caring for Metabolic patients in an out patient clinic of a tertiary medical Center
Yifat Ratzabi, Safra Children's Hospital, Sheba Medical Center, Tel Hashomer, Israel

B Outlining the care situation in Canavan patients
Jama Wahid, University Medical Center Hamburg-Eppendorf, Germany

B Allow Natural Death (AND) in Inborn Errors of Metabolism
Carolina Fraga, Centro Materno-Infantil Do Norte, Centro Hospitalar Universitario Santo Antonio, Portugal

B Assessment of the level of indepencence and knowledge of 13 young people with an inborn error of metabolism: Twelve months post transition to an adult clinic from a paediatric service
Anita Inwood, Queensland Lifespan Metabolic Medicine Service, Australia

ORGANIC ACIDURIAS

B Factors associated with the long-term prognosis in patients with methylmalonic acidemia and propionic acidemia
Hyunwoo Bae, Asan Medical Center, University of Ulsan College of Medicine, South Korea

B Citraconate isomers in methylmalonic acidemia revisited: possible role in pathophysiology and as biomarkers
Anibh Das, Hannover Medical School, Germany

B Investigating gene essentiality and gene:gene interactions in MMUT deficiency
Sean Froese, University of Zurich, Switzerland

B Neurodevelopmental delay and cognitive disabilities caused by intracerebroventricular administration of L-2-hydroxyglutaric acid to neonatal rats
Moacir Wajner, Universidade Federal do Rio Grande do Sul, Brazil

B Experimental evidence that intracerebral administration of L-2- hydroxyglutaric acid to neonatal rats causes oxidative stress, neuronal death, astrogliosis, microglia activation associated with neuroinflammation and delayed myelination
Moacir Wajner, Universidade Federal do Rio Grande do Sul, Brazil

B The glycine N-acyltransferases, GLYAT and GLYATL1, contribute to the detoxification of isovaleryl-CoA: an in-silico and in vitro validation
Stefan Kuhn, North-West University, South Africa

B Three cases of glycerol-3-phosphate dehydrogenase 1 deficiency followed at a tertiary care hospital: the importance of molecular genetic testing
Niccolo' Campagna, University of Florence, Italy

B Clinical, biochemical, genetic and neuroradiological data in a single cohort of DBP deficiency patients
Unai Diaz-Moreno Elorz, Great Ormond Street Hospital, UK

B A girl with strawberry milk-like blood – acute stabilisation in familial chylomicronemia syndrome
Oliver Heath, Royal Children's Hospital, Australia

B Phosphatidylserine flippase deficiency diagnosed by whole exome sequencing – a case report
Ariana Mendes, Centro Hospitalar e Universitário de Coimbra, Portugal

B Untargeted metabolomics profiling in three cohorts indicates lipid imbalance in individuals with epilepsy
Kaisa Teele Oja, University of Tartu, Estonia

B Cholic acid increases serum cholesterol in Smith-Lemli-Opitz syndrome: a pilot study
William Rizzo, University of Nebraska Medical Center, USA

B Persistent hypertriglyceridemia and liver disease in Roma children with transient infantile hypertriglyceridemia caused by the homozygous mutation c.895GA in GPD1 gene.
Jana Saligova, Children's Faculty Hospital Kosice, Slovakia

B Characterization of Alagille Syndrome in Patients with Cholestatic Liver Disease: Clinical Features and Genetic Analysis
Natalia Semenova, Research Centre for Medical Genetics, Russia

B Successful treatment of child with congenital bile acid synthesis defect type 3 with oral chenodeoxycholic acid
Natalia Semenova, Research Centre for Medical Genetics, Russia

B Cell imaging-based screening for small molecules that rescue peroxisome function in mild disorders of the Zellweger spectrum
Beatriz Silva, University of Luxembourg, Luxembourg

B Primary dyslipidemias in Russian population: novel genetic variants
Peter Vasiluev, Research Centre for Medical Genetics, Russia

B Disturbances of mitochondrial functions involving permeability transition pore opening are caused by phytanic acid in rat heart
Moacir Wajner, Universidade Federal Do Rio Grande Do Sul, Brazil

B Phytanic acid impairs mitochondrial respiration in rat heart and decreases cardiomyocyte viability
Moacir Wajner, Universidade Federal Do Rio Grande Do Sul, Brazil

PHENYLKETONURIA

B The effect of Large Neutral Amino Acids on protein- and diet intake for PKU patients
Kirsten Ahring, Copenhagen University Hospital, Denmark

B PHEFREE (Phenylalanine Families and Researchers Exploring Evidence): The rare disease research consortium for hyperphenylalaninemia
Georgianne Arnold, University of Pittsburgh, USA

B Phenylketonuria (PKU) in Iraq: urgent need for education of health care providers to establish medical support for PKU patients
Joshua Baker, Ann & Robert H. Lurie Children's Hospital of Chicago, USA

B Relating parental personality and stress with eating styles and metabolic control in early treated PKU patients
Filippo Manti, University of Rome La Sapienza, Italy

B Phase 3 APHENITY long-term study: Sepiapterin for treatment of phenylketonuria
Drago Bratkovic, Royal Adelaide Hospital, Australia

B Genetic profile of phenylketonuria in Brazil
Ida Vanessa Doederlein Schwartz, Hospital de Clínicas de Porto Alegre, Brazil

B Exploring the Prevalence of Inborn Errors of Metabolism in Brazil: Insights from the Brazilian Rare Diseases Network
Ida Vanessa Doederlein Schwartz, Hospital de Clínicas de Porto Alegre, Brazil

B Preclinical Evaluation of a Prolonged-Release Protein Substitute on Blood Phenylalanine Levels when Combined with Dietary Protein
Luciana Giardino, Bologna University, Italy

B The role of mitogen-activated protein kinase (MAPK) in Phenylketonuria: new markers of oxidative stress
Vincenza Gragnaniello, University Hospital of Padua, Italy

B Rate of fall of phenylalanine (phe) in classical phenylketonuria (PKU) patients after commencement of phe free supplement from a single UK centre
Rebecca Halligan, Guys & St. Thomas NHS Foundation Trust, UK

B Oxidant status of 26 phenylketonuria patients in follow-up: Determination of superoxide dismutase-2 activity and Malondialdehyde estimation
Verónica Cornejo, University of Chile, Chile

B Cardiovascular risk and cardiac disease in adult patients with phenylketonuria: a review
Francois Maillot, University Hospital of Tours, France

B Study of peripheral biogenic amines metabolites in phenylketonuric patients
Filippo Manti, University of Rome La Sapienza, Italy

B A National Referral Clinic for Phenylketonuria (PKU) patients- treatment from a dietician`s point of view.
Lior Marinescu, Edmond and Lily Safra Children's Hospital, Sheba Medical Center, Tel-Hashomer, Israel

B Evaluating trends in self-rated historic metabolic control and treatment history among PRISM participants
Markey McNutt, University of Texas Southwestern Medical Center, USA

B Diet compliance and body mass index at Slovenian adult patients with phenylketonuria
Urh Groselj, University Children's Hospital, UMC Ljubljana, Slovenia

B An audit of DEXA results in adult patients with PKU; examination of metabolic control and dietary factors
Robert O'Byrne, Mater Misericordiae University Hospital, Ireland

B Trimethylamine increases intestinal fatty acid absorption
Júlio César Rocha, Universidade NOVA De Lisboa, Portugal

B Protein intake and prevalence of overweight and obesity in patients with phenylketonuria: a 10 year-longitudinal TNSPKU study
Júlio César Rocha, Universidade NOVA De Lisboa, Portugal

B OPAL: A multicenter, observational study to evaluate the real-world outcomes of pegvaliase in adults with phenylketonuria
Frank Rutsch, Muenster University Children's Hospital, Germany

B The challenge of adults with Phenylketonuria who have been lost to care: A single center's attempt to reach those diagnosed with PKU over 60 years of Newborn Screening
Stephanie Sacharow, Boston Children's Hospital, USA

B Menstrual cycle characteristics, premenstrual syndrome and blood phenylalanine level relationship in women with PKU
Arzu Selamioglu, Istanbul University, Turkiye

B Total Choline Intake and Working Memory Performance in Adults with Phenylketonuria
Meriah Schoen, Emory University, USA

B Intake modalities of amino acid mixtures: a real-world data collection from PKU patients
Albina Tummolo, Children Hospital Giovanni XXIII Azienda Ospedaliero-Universitaria Consorziale, Italy

B A feasible point-of-care testing method for monitoring phenylalanine levels using phenylalanine ammonia lyase and a portable ammonia detection system
Yoichi Wada, Tohoku University School of Medicine, Japan

UREA CYCLE DISORDERS

B A rapid differential diagnosis for congenital neonatal hyperammonemia by NGS targeted-resequencing gene panel improves therapy and patient management
Lorenzo Ferri, Meyer Children's Hospital IRCCS, Italy

B Variant in the allosteric domain of CPS1 protein associated with effectiveness of N-carbamoyl glutamate therapy in CPS1 deficiency
Vincenza Gragnaniello, University Hospital of Padua, Italy

B Urea cycle disorders in Argentina, analysis from a cohort of 135 patients diagnosed in the last 20 years
Soledad Kleppe, Hospital Italiano De Bsas, Argentina

B Metabolic parameters and development in patients with argininosuccinic acid synthase 1 and lyase deficiency
Nicola Longo, University of Utah, USA

B Evaluation of oxidative damage to biomolecules in patients with ornithine transcarbamilase deficiency
Moacir Wajner, Universidade Federal Do Rio Grande do Sul, Brazil

B Acute management and outcome of inaugural hyperammonemia in urea cycle defects in a tertiary UK metabolic centre
Christina Spyridoula Sidira, Great Ormond Street Hospital for Children NHS Foundation Trust, UK

B Towards an Algorithm-Based Tailored Treatment of Acute Neonatal Hyperammonemia
Patrick Verloo, University Hospital Ghent, Belgium

B Identification of a novel deep intronic variant causing pseudo-exon inclusion in citrin deficiency and the development of a new drug for the variant by using splice-switching oligonucleotides
Eri Imagawa, The Jikei University School of Medicine, Japan

AMINO ACID DISORDERS

Newborn screening in Emilia-Romagna (Italy) for cystathionine beta-synthase deficiency: never give up
Andrea Pession, IRCCS Azienda Ospedaliero-Universitaria di Bologna, Italy

Prolidase deficiency, natural history and identification of a rare variant c.1409G>A (p.(Arg470His)) in PEPD gene. Case report
M Teresa Cardoso, University Hospital Center S. Joao, Portugal

Rare complication of hereditary tyrosinemia type 1: neurogenic crisis
Fehime Erdem, Ege University Faculty of Medicine, Turkiye

Intermittent maple syrup urine disease (MSUD) uncovered by screening
Olga Grafakou, OKYPY, Archibishop Makarios III Hospital, Cyprus

Specific biosensor of amino acid
Hen Hadad, Ben-Gurion University of the Negev, Israel

Two siblings with Lisinuric Protein Intolerance (LPI) misdiagnosed as Glycogen Storage Disease type 1 (GSD 1) in resource-limited settings; a case report from Sri Lanka
Chamal Palingu Imalke Kankanan Arachchige, University of Ruhuna, Sri Lanka

Genotype and Clinical Characteristics of Methioninemia Patients: A Comprehensive Study in a Single Institution
Min-Ji Kim, Pusan National University Children's Hospital, South Korea

Maple syrup urine disease type Ib: 3-years outcome of the case of early liver transplantation in Russia
Natalia Pechatnikova, GBIH Morozov's Children Clinical Hospital, Russia

Specific biosensor of amino acid
Noam Shlush, Ben Gurion University of the Negev, Israel

Disruption of brain redox homeostasis by acute intracerebral accumulation of α-ketoisocaproic acid in neonatal rats
Moacir Wajner, Universidade Federal Do Rio Grande Do Sul, Brazil

Oculocutaneous tyrosinemia in a Colombian patient: case report and literature review
Ana Maria Zarante-Bahamon, Hospital Universitario San Ignacio, Colombia

CLINICAL STUDIES, PATIENT REPORTED OUTCOME MEASURES, OUTCOME

Individual therapeutic trial of a rocking bed for a sleep disorder in a patient with a severe mitochondrial disease
Alexander Breuss, ETH Zurich, Switzerland

The International Niemann-Pick disease registry-A resource for researchers worldwide
Jacqueline Imrie, INPDR, UK

Cognitive functioning in pre-school children with classical galactosemia
Amanda Krzywdzinska, Institute of Mother and Child, Poland

The Impact of Gender Identity and Transgender Related Issues on the Management of Inborn Errors of Metabolism
Stephanie Newman, Children's Hospital London Health Sciences Centre, Canada

Metabolomic profiles of Down syndrome patients by Liquid Chromatography Mass Spectrometry (LC-MS)
Sunil Kumar Polipalli, Maulana Azad Medical College & Associated Lok Nayak Hospital, India

2-year experience of a interdisciplinary IEM group in a tertiary care center in Bogotá, Colombia: an integral view
Jorge Luis Ramon-Gomez, Instituto Roosevelt, Colombia

The interface of pediatric palliative care and metabolic diseases - A 20-year epidemiological survey of outpatients at a quaternary hospital
Gustavo Spolador, Hospital Das Clinicas USP, Brazil

Alpha-mannosidosis: first symptoms in ten cases
Nato Vashakmadze, Pirogov Russian National Research Medical University, Russia

Role of gene interactions in the Pathophysiology of skeletal dysplasias
Lina Moreno, Universidad Libre, Colombia

Two cases of diazoxide-responsive congenital hyperinsulinism with diffuse uptake of pancreas by 18F-DOPA PET-CT
Jeesuk Yu, Dankook University Hospital, South Korea

DIETETICS AND NUTRITION

Neonatal MSUD Metabolic Crisis - to Dialyse or Not to Dialyse?
Danit Javasky, Schneider Children's Medical Center, Israel

DISORDERS OF FATTY ACID OXIDATION AND KETONE BODY METABOLISM

A Case of Glutaric Aciduria Type II Initially Detected through Newborn Screening in the Philippines
Michelle Abadingo, University of the Philippines Manila, Philippines

The Challenges of Introducing Triheptanoin in a Child with Carnitine Acylcarnitine Translocase Deficiency (CACT)
Drago Bratkovic, Women's and Children's Hospital, Australia

First results from a long-term disease monitoring program in patients with long-chain fatty acid oxidation disorders
Barbara Burton, Ann & Robert H. Lurie Children's Hospital, USA

Medium Chain Acyl CoA Dehydrogenase (MCAD) Deficiency Due To An Exon 8 Duplication In ACADM
Aviva Eliyahu, Edmond and Lily Safra Children's Hospital, Sheba Medical Center, Israel

Challenging diagnosis of type 2 diabetes mellitus in patient with multiple acyl-CoA dehydrogenase deficiency
Nodoka Ikeda, Tohoku University School of Medicine, Japan

Acute renal failure due to severe rhabdomyolysis provoked by a mild covid-19 infection in a patient with LCHAD deficiency
Dunja Leskovar, University Hospital Centre Zagreb, Croatia

The synergy of different methods for the final diagnosis of MCADD in Slovenian patient
Barbka Repic Lampert, University Children's Hospital, University Medical Centre Ljubljana, Slovenia

Case report: A newborn infant with cardiac arrest
Ana Morais López, University Hospital La Paz, Spain

A previously undescribed mutation in CPT-II deficiency. Case report
Monserrat Pons Rodríguez, Hospital Universitario Son Espases, Spain

Importance of enzyme activity measurement for MCAD deffiency classification in patients with inconclusive results. Case report
Monserrat Pons Rodríguez, Hospital Universitario Son Espases, Spain

Congenital Hyperinsulinemia Hypoglycemia,pregnancy in a case affected of Hyperinsulinemia Hypoglycemia Familial 4
Talieh Zaman, Iranian National Society of SSIEM, Iran

DISORDERS OF PURINES, PYRIMIDINES, NUCLEIC ACIDS AND PORPHYRIAS

Late onset isolated sulfite oxidase deficiency: a case report
Aynur Kucukcongar Yavas, Ankara Bilkent City Hospital, Turkiye

DISORDERS OF VITAMINS, COFACTORS AND TRACE ELEMENTS

Family case of biotin-thiamine-responsive basal ganglia disease
Tatiana Bushueva, National Medical Research Center for Children's Health, Russia

GLYCOSYLATION DISORDERS/CDG, PROTEIN MODIFICATION DISORDERS

A novel variant in Lafora`s disease: case report in Afro-Colombian adolescent
Oscar Mauricio Espitia Segura, HOMI Fundación Hospital Pediatrico la Misericordia, Colombia

Glycogen storage disorders type i in vietnam: genotype, phenotype, and outcome
Hang Nguyen Thi, Vietnam National Children's Hospital, Vietnam

Homozygous PGAP2 mutation cause hyperphosphatasia with mental retardation syndrome-3 (HP-MRS3): Genetic and clinical evaluation of the ultra rare inherited glycosylphosphatidylinositol (GPI) biosynthesis defect
Aynur Kucukcongar Yavas, Ankara Bilkent City Hospital, Turkiye

First Korean case of NANS gene mediated deficiency of N-acetylneuraminic acid synthase causing late onset progressive skeletal dysplasia
Sukdong Yoo, Pusan National University School of Medicine, South Korea

INBORN ERRORS OF METABOLISM IN ADULTS

Neonatal Screening In Portugal: The Results of a Retrospective Cohort Study with 113 Adult PKU Patients
Esmeralda Martins, Centro Hospitalar Universitário de Santo António, Portugal

Late presentation of McArdle Disease: a special focus on symptomatology
Paulo Castro Chaves, Centro Hospitalar Universitario Sao Joao, Portugal

VARS2-related mitochondrial disease: a glimpse into mitochondrial complexity
Paulo Castro Chaves, Centro Hospitalar Universitario Sao Joao, Portugal

High vitamin B12 levels
Maria Francesca Font Pico, Hospital Universitari Sant Joan De Reus, Spain

Impact of the COVID-19 pandemic on the care of adult PKU patients in Germany: a retrospective analysis of 115 patient data over a four years period
Jan Philipp Koehler, Medical Faculty and University Hospital Duesseldorf, Germany

Low back pain revealing an inborn metabolic disease: a case report
Francois Maillot, University Hospital of Tours, France

Classical homocystinuria in adult patients - one centre experience
Drazen Perica, University Hospital Centre Zagreb, Croatia

Prevalence of Comorbid Conditions in 222 Hungarian Patients with Phenylketonuria
Csaba Sumanszki, Semmelweis University, Hungary

Genetic variants associated with inborn errors of carbohydrate metabolism in southwestern Colombia
Lina Johanna Moreno Giraldo, Universidad del Valle, Colombia

Diagnosis of Ornithine Transcarbamylase Deficiency during Pregnancy: Case Report
Ana Maria Zarante-Bahamon, Hospital Universitario San Ignacio, Colombia

LYSOSOMAL DISORDERS

A CRIM-negative Pompe Case Receiving Enzyme Replacement Therapy with a Successful Immunotolerance Regimen
Halil Tuna Akar, Hacettepe University, Turkiye

C.239 G>A: from uncertain significance to a pathogenic variant in a Colombian family cluster
Alejandra Bello, University Foundation of Health Sciences, Colombia

Application of RNA analysis and whole genome sequencing for confirmation of diagnosis in Mucopolysaccharidosis type VI patients
Igor Bychkov, Research Centre for Medical Genetics, Russia

Phenotype-Genotype in Colombian women with Anderson-Fabry disease
Claudia C Colmenares-Mejia, Fundación Cardiovascular De Colombia, Colombia

GALNS c.304G>A (p.Ala102Thr) variant of uncertain significance (VUS) as cause of Morquio A disease
Cristobal Colon, Complexo Hospitalario Universitario de Santiago de Compostela, Spain

Experience of miglustat therapy in pediatric patients with niemann-pick type c disease
Esmeralda Martins, Centro Hospitalar Universitário De Santo António, Portugal

Mucolipidosis Type 3 Gamma: Case Report
Serpil Dincer, Ankara City Hospital, Turkiye

Cystinosis in Latin America: insights from local experience on biochemical diagnosis
Olga Echeverri, Pontificia Universidad Javeriana, Colombia

Evaluation of experienced events in pompe disease based on real-life data
Fehime Erdem, Ege University Faculty of Medicine, Turkiye

Case Report: Niemann-Pick Disease Type C (NPC) due to E20X variant in NPC2: First case report in Latin America
Oscar Mauricio Espitia Segura, HOMI Fundacion Hospital Pediátrico la Misericordia, Colombia

Cerliponase alfa treatment in a pre symptomatic patient with Neuronal Ceroid Lipofuscinosis type 2 CLN2 and atypical phenotype: Case report
Oscar Mauricio Espitia Segura, HOMI Fundacion Hospital Pediátrico la Misericordia, Colombia

Prenatal diagnosis of Mucopolysaccharidosis I by LC-MS/MS determination of disease-specific oligosaccharides in amniotic fluid supernatant
Larissa Faqueti, Hospital de Clinicas de Porto Alegre, Brazil

Alpha-mannosidosis: positive effects of Enzyme Replacement Treatment on cognitive function
Martha Caterina Faraguna, Fondazione IRCCS San Gerardo Dei Tintori, Italy

Burden of illness of acid sphingomyelinase deficiency (ASMD) in Brazil: A retrospective chart review study
Roberto Giugliani, Federal University of Rio Grande do Sul, Brazil

Evaluation of inflammatory process in mucopolysaccharidosis type IV A patients under long-term enzyme replacement therapy
Roberto Giugliani, Federal University of Rio Grande do Sul, Brazil

Bone turnover in patients with lysosomal storage disorders
Sabire Gokalp, Gazi University Faculty of Medicine, Turkiye

Juvenile Canavan Disease presenting with intention tremor: a second case report
Rebecca Halligan, Evelina London Children's Hospital, UK

In vitro beneficial effects of N-acetylcysteine and Coenzyme Q10 in Niemann-Pick type C patient-derived fibroblasts
Roberto Giugliani, Federal University of Rio Grande do Sul, Brazil

Association of Elosulfase Alpha for quality of life in Morquio IV-A syndrome: a case series
Gloria Liliana Porras, Comfamiliar Risaralda, Colombia

Hematopoietic stem cell transplantation in Mucopolysaccharidosis VI: case report and 4 year follow up
Dafne Horovitz, Children and Adolescents Health Fernandes Figueira - Fiocruz, Brazil

A case of avalglucosidase treatment in a patient with Infantile-onset pompe disease(IOPD) in Korea
JiHoon Hwang, Sungkyunkwan University School of Medicine, South Korea

Evaluation of Lysosphingolipid analysis in the diagnosis of Lysosomal Storage Disorders
Aynur Kucukcongar Yavas, Ankara Bilkent City Hospital, Turkiye

Single institutional experience with metachromatic leukodystrophy: demographic, biochemical and genetic characteristics of 7 patients
Aynur Kucukcongar Yavas, Ankara Bilkent City Hospital, Turkiye

Event based treatment outcomes of patients with gaucher disease: a different perspective
Ayse Kilic, Gazi University Faculty of Medicine, Turkiye

Status epilepticus with exitus in symptomatic leukodystrophy-related epilepsy of infantile-onset Pompe disease (IOPD)
Francesca Maria Menni, Fondazione IRCCS Ca' Granda Ospedale Maggiore Policlinico, Italy

Characterization and treatment of Fabry's disease in female pediatric patients
Lina Moreno Giraldo, Universidad Santiago de Cali, Colombia

Impact of early diagnosis and timely treatment on the natural history of type I Mucopolysaccharidosis - Scheie Phenotype
Lina Moreno Giraldo, Universidad Santiago de Cali, Colombia

Real-world clinical experience of unexpected thrombocytopenia in patients with Gaucher disease as an initial diagnostic clue
Jiyoung Oh, Yonsei University, College of Medicine, Severance Children's Hospital, South Korea

Importance of the pedigree in the neonatal diagnosis of orphan diseases: MPS VI case report
Lina Johanna Moreno Giraldo, Universidad Santiago de Cali, Colombia

Expert opinion on clinical presentation, diagnosis and treatment of infantile onset pompe disease: a delphi study in Turkey
Ekin Ozsaydi Aktasoglu, Gazi University, Turkiye

Clinical and laboratory findings in 3 rare cases from gangliosidosis family
Vasilica Plaiasu, INSMC Alessandrescu-Rusescu Regional Center of Medical Genetics, Romania

Advances in the sociodemographic, clinical, enzymatic and molecular characterization of patients with Gaucher disease in southwestern Colombia
Lina Johanna Moreno Giraldo, Universidad Santiago de Cali, Colombia

Patient Characterization before and after enzyme therapy.The GALNS p.Gly301Cys variant is a probable founder mutation from the coffee-growing area of Colombia that causes mucopolysaccharidosis IVA syndrome
Liliana Porras, Comfamiliar Risaralda, Colombia

Impact of COVID-19 Pandemic on quality of life of patients with lysosomal storage disorders receiving treatment at a tertiary care public health Institute in Mumbai, India
Sakshi Rajoria, Seth G S Medical College & KEM Hospital, India

Heterogenic presentations of Sialidosis in two Sri Lankan children
Kishanjalee Rammuthupura, Lady Ridgeway Hospital, Sri Lanka

Ambroxol as adjunctive therapy for neurologic symptoms in Gaucher disease type 3: Case report
Jorge Luis Ramon-Gomez, Instituto Roosevelt, Colombia

Micro learning-based education in Fabry disease significantly improves physician confidence
Derralynn Hughes, University College London, Royal Free London NHS Foundation Trust, UK

Heart-transplantation in a patient with pathogenic saromere and α galactosidase A mutation: a case report
Karolina Schnabel, Semmelweis University, Hungary

Unsuspected Gaucher disease in a splenectomized young adult with acute onset of back pain
Mariana Serres Gomez, La Paz University Hospital, Spain

METABOLIC MYOPATHIES

Nosological, therapeutic and prognostic implications of genomics in juvenile refractory dermatomyositis, a case report
Lina Johanna Moreno Giraldo, Universidad Santiago de Cali, Colombia

A rare case of GABA transaminases deficiency diagnosed by trio-based whole exome sequencing analysis
Jieun Lee, Inha University Hospital, South Korea

Impact of genomic characterization in patients with non-5q spinal muscular atrophy
Lina Johanna Moreno Giraldo, Universidad Santiago de Cali, Colombia

A case of ECTH1 deficiency with elective liver transplantation
Tomohiro Ebihara, Chiba Children's Hospital, Japan

Investigation of mitochondrial DNA depletion syndromes in children under 5 years old with acute liver manifestations of unknown etiology
Fatma Tuba Eminoglu, Ankara University School of Medicine, Turkiye

Novel mitochondrial phenotype in a patient with SYNGAP1 encephalopathy
Andrea Gropman, Children's National Hospital, USA

Mimics and chameleons in mitochondrial pathology
Yulia Itkis, Research Centre for Medical Genetics, Russia

Familial presentation of heteroplasmic mutation m.3291T>C in MT-TL1 gene
Kairit Joost, East-Tallinn Central Hospital, Estonia

Variants Detected in Mitochondrial DNA D-loop in Patients with Non-alcoholic Fatty Liver Disease
Sharareh Kamfar, Shahid Beheshti University of Medical Sciences, Iran

Hereditary spastic paraplegia type 35 in a Turkish girl with fatty acid hydroxylase associated neurodegeneration
Aynur Kucukcongar Yavas, Ankara Bilkent City Hospital, Turkiye

A case diagnosed with mitochondrial disease caused by surf1 mutation by candidate gene method
Aynur Kucukcongar Yavas, Ankara Bilkent City Hospital, Turkiye

Personalized diagnostics is the basis for individual effective therapy of mitochondrial diseases
Grechanina Ye.Ya, Interregional Specialized Medical and Genetic Center Center for Rare (Orphan) Diseases, Ukraine

A case of NDUFAF6-associated mitochondrial respiratory chain complex I deficiency in two siblings
Denis Kistol, Research Centre for Medical Genetics, Russia

Successful long-term high-dose N-Acetylcysteine therapy in a patient with transient infantile liver failure due to TRMU gene mutations
Nina Krasnoshchekova, Gbih Morozov's Children Clinical Hospital, Russia

The heterogeneity of m.13513G>A variant - related phenotypes depending on heteroplasmy level
E.Yu. Zakharova, Research Centre for Medical Genetics, Russia

Heteroplasmic Mutant Load Differences in Mitochondrial Disease
Young-Mock Lee, Yonsei University College of Medicine, South Korea

Expanding phenotype of FDXR-related mitochondrial disorder: the role of ferredoxin reductase in steroidogenesis
Nicola Longo, University of Utah, USA

Insight into the effect of mitochondrial function on immune function in the context of tumor infiltrating lymphocyte therapy
Shani Kassia Lyskov, Sheba Medical Center, Israel

Phenotypic characterization of the ECHS1 pathogenic variant c.476A>G (p.Q159R): a review of 9 cases
Alejandro Iglesias, Columbia University Vagelos College of Physicians and Surgeons and New York-Presbyterian Morgan Stanley's Children's Hospital, USA

High prevalence of mitochondrial membrane protein-associated neurodegeneration (MPAN) in Estonia
Katrin Ounap, University of Tartu, Estonia

A novel pathogenic variant m.9122T>G in MT-ATP6 presenting with neonatal hypertrophic cardiomyopathy, hyperammonemia and anaemia
Barbara Siri, Bambino Ges Children Hospital, IRCCS, Italy

Neuropsychological and neuropsychiatric disease spectrum in Single Large Scale Mitochondrial Deletions
Barbara Siri, Bambino Ges Children Hospital, IRCCS, Italy

Lack Of mitochondrial complex I assembly factor NDUFAF2 results in a distinctive infantile onset brainstem neurodegenerative disease with early lethality
Ronen Spiegel, Emek Medical Center, Israel

A patient with a RARS2 mutation exhibiting anemia as a possible new clinical feature
Rachel Rock, Edmond and Lily Safra Children's Hospital, Sheba Medical Center, Tel Hashomer, Israel

Mitochondrial DNA testing increases the diagnostic efficacy of mitochondrial disorders by 8.3%
Elis Tiivoja, University of Tartu, Estonia

Mitochondrial diseases masks revealed on WES/WGS analysis
Polina Tsygankova, Research Centre for Medical Genetics, Russia

Personalized Medicine in Mitochondrial Health and Disease: Molecular Basis of Therapeutic Approaches Based on Nutritional Supplements and Their Analogs
Albina Tummolo, Children Hospital Giovanni XXIII Azienda Ospedaliero-Universitaria Consorziale, Italy

Successful implementation of ketogenic diet in a TRIT1 deficient patient with diabetes mellitus leads to clinical improvement
Arnaud Vanlander, Ghent University Hospital, Belgium

Another case of UQCRC2-related mitochondrial disease: a novel variant with atypical onset
Jacopo Maria Venanzi, Università degli Studi di Firenze, Italy

NEUROTRANSMITTER AND CREATINE RELATED DISORDERS

Laboratory LC-MS/MS algorithm for detecting primary creatine disorders.
Josef Bartl, General University Hospital and Charles University, Czech Republic

Baseline clinical characteristics and disease burden of patients with aromatic L-amino acid decarboxylase deficiency (AADCd) enrolled in the AADCAware registry
Bruria Ben Zeev, Sheba Medical Center, Israel

Clinical presentation and follow up of two AADC deficiency cases in Brazil prior to gene therapy
Matheus Guerra-Peixe, Hospital Santa Helena, Brazil

Mild form of aromatic L-amino acid decarboxylase deficiency
Sabine Laktina, Childrens Clinical University Hospital, Latvia

Optimization of a stable isotope-labelled substrate assay for measuring AGAT activity
Andreas Schulze, University of Toronto, Canada

Creatine transporter deficiency in a family with unexplained intellectual disability
Maria Carmo Macario, Centro Hospitalar e Universitário de Coimbra, Portugal

Case Series. Nonketotic hyperglycinemia patients with and without a ketogenic diet treatment
Liliana Porras, Comfamiliar Risaralda, Colombia

LC-MS/MS quantification of 3-O-methyldopa in DBS for the diagnosis of AADC Deficiency using a derivatization method to improve sensitivity
Dana Velasquez Rivas, Laboratorio de Neuroquímica Dr. Chamoles, Argentina

NEW DISEASES

Variants in the ERCC4 gene as a rare cause of cerebellar ataxia with dystonia: The First Case in Korea
In Kim, Pusan National University Children's Hospital, South Korea

The minacious dance of TANGO2 deficiency in Brazilian patients
Caroline Olivati, Rare Rosy Clinic, Brazil

Claudin-1 mutation in a case affected of cholestatic liver disease,ichthyosis and scalp hypotrichosis
Talieh Zaman, Iranian National Society of SSIEM, Iran

NEWBORN SCREENING

Newborn screening for acid sphingomyelinase deficiency in Illinois
Joshua Baker, Ann & Robert H. Lurie Children's Hospital of Chicago, USA

Geographical Variations in MCADD Frequency: Findings from Expanded Newborn Screening in Russia
Galina Baydakova, Research Center for Medical Genetics, Russia

Case report: Spanish male newborn with Zellweger Syndrome detected by X-ALD newborn screening
Maria Isabel Cabrera Gonzalez, Malaga Regional University Hospital, Spain

Newborn screening for biotinidase deficiency: A 6-year single center experience
Petr Chrastina, General University Hospital and 1st Faculty of Medicine, Charles University, Czech Republic

Survey of 8,799 cases included in a pilot program of phase 2 to expand neonatal screening in Porto Alegre, RS, Brazil
Ida Vanessa Doederlein Schwartz, Hospital de Clínicas de Porto Alegre, Brazil

Mild/Variant cases of argininosuccinic acid lyase deficiency by newborn screening using argininosuccinic acid as a primary marker
Xiaowei Fu, University of Tennessee Health Science Center, USA

Frequency of iduronate-2-sulfatase gene variants detected in newborn screening for mucopolysaccharidoses type II in Japan
Yusuke Hattori, Kumamoto University, Japan

Citrin deficiency: a metabolic disorder misdiagnosed as galactosemia - a case report
Nina Krasnoshchekova, Gbih Morozov's Children Clinical Hospital, Russia

New Born Screening and mutational spectrum for Hemoglobinopathies & Thalassemia in Delhi, India
Sunil Kumar Polipalli, Maulana Azad Medical College & Associated Lok Nayak Hospital, India

Psychometric parameters in patients with phenylketonuria and galactosemia of the National Newborn Screening in Greece
Triantafyllia Sdogou, Institute of Child Health, Greece

Psychosocial Issues of Filipino Parents with a Child with Maple Syrup Urine Disease
Ma-Am Joy Tumulak, University of the Philippines, Philippines

NOVEL DIAGNOSTIC/LABORATORY METHODS INCLUDING OMICS

Using urine NMR spectroscopy in the diagnosis of intoxication-type inborn errors of metabolism
Daniela Blanita, Institute of Mother and Child, Moldova

Metab-Latam: Sharing of scientific knowledge in inborn errors of metabolism
Ida Vanessa Doederlein Schwartz, Universidade Federal do Rio Grande do Sul, Brazil

Comparison of two branched-chain amino acids (BCAAs) measurement methods for monitoring the treatment in inborn errors of metabolism
Ewa Glab- Jablonska, Institute of Mother and Child, Poland

Personalised System Practice - eXamination Transformation
James Henry, Children's Hospital, Ireland

Landscape of genetic testing in MCT8-deficiency
Charlotte Hoffman, Independent, Netherlands

NSAID-induced intestinal lesions in patients with osteoarthritis: the diagnostic capabilities of noninvasive fecal biomarkers
Olena Gubska, Bogomolets National Medical University, Ukraine

Hypotonic syndrome as a manifestation of an ultra-rare disease of genetic origin
Lina Moreno Giraldo, Universidad Santiago de Cali, Colombia

Clinical Applications of a Rapid Real-Time Analysis System for Whole Genome/Exome Sequencing
Dau-Ming Niu, Taipei Veterans General Hospital, Taiwan

Current challenges of genomic diagnostics in hemophagocytic syndromes in pediatrics: a case report
Lina Johanna Moreno Giraldo, Universidad Libre, Colombia

Characterization of genomic variants of the PTPN11 gene associated with congenital heart disease in a population of southwestern Colombia
Jose Maria Satizabal Soto, Universidad Del Valle, Colombia

ORGANIC ACIDURIAS

Multi-omics to the rescue – genome sequencing and RNA analysis decipher the cause of previously unresolved propionic acidemia
Hagit Baris Feldman, Tel Aviv Sourasky Medical Center, Israel

Transient bilateral vision loss during acute metabolic decompensation in a patient with methylmalonic acidemia
Andrea Pession, IRCCS Azienda Ospedaliero-Universitaria di Bologna, Bologna, Italy

Diagnosis and follow-up of four organic acidurias in three regions of central Italy (Emilia-Romagna, Tuscany and Umbria): what has been learnt, what can be improved?
Elena Procopio, Meyer Children's Hospital IRCCS, University of Florence, Italy

Non-Hodgkin Lymphoma after kidney trasplantation in a Cobalamin B deficiency patient: an incidental finding or increased risk of cancer?
Alberto B Burlina, University Hospital of Padua, Italy

Osteosarcoma and Propionic Acidemia: Chronicle of a Crisis Foretold
Paola Andrea Cubides Villamil, Universidad Militar Nueva Granada, Colombia

Unraveling the Genetic Basis of Propionic Acidemia using Advanced Molecular Techniques
Artur Galushkin, Research Centre for Medical Genetics, Russia

Glutaric aciduria type 1 in Poland detected through newborn screening – incidence, initial management and outcome
Jolanta Sykut-Cegielska, Institute of Mother and Child, Poland

Patient with Glutaric aciduria type 1 and a familial ACAN gene variant
Pedro Louro, University Hospital Center of Sao Joao, Portugal

Aminoacylase 1 deficiency: the first Czech patient
Dagmar Prochazkova, Masaryk University and University Hospital Brno, Czech Republic

D-2-hydroxyglutaric acid disrupts heart mitochondrial calcium retention capacity and causes cardiomyocyte death
Moacir Wajner, Universidade Federal Do Rio Grande Do Sul, Brazil

Hyperammonemia, Hypocarnitinemia, Rhabdomyolysis, and Pancreatitis in a Patient with Undiagnosed 3-Methylcrotonyl-CoA Carboxylase Deficiency
Shoji Yano, University of Southern California, USA

PEROXISOMAL, STEROL, BILE ACID, LIPID AND LIPOPROTEIN METABOLISM

Metabolic responses to in-vitro biomass burning aerosol exposure in liver disease model
Dror Bittner, Sheba Medical Center, Israel

Cerebrotendinous xanthomatosis - clinical manifestation in two Slovak children
Katarina Brennerova, National institute of Children´s Diseases, Slovakia

Effectiveness of Cholic acid treatment in Cerebrotendinous Xanthomatosis
Ketki Kudalkar, Navi Mumbai Institute of Research in Mental and Neurological handicap (NIRMAN), India

Genomic variants associated with inborn errors of lipid metabolism in southwestern Colombia
Lina Johanna Moreno Giraldo, Universidad Libre, Colombia

PEX7 related nonclassic (mild) rhizomelic chondrodysplasia punctate type 1 patient with global developmental delay without skeletal dysplasia and cataract
Young-Lim Shin, Soonchunhyang University Bucheon Hospital, South Korea

Cerebellar ataxia with normal intelligence in an adult patient with PEX10 mutation
Tinatin Tkemaladze, Tbilisi State Medical University, Georgia

Zellweger Spectrum Disorder with Nystagmus and Acquired Motor Skill Loss and Short-Term Benefit from IVIG Treatment: A Case Report
Harun Yildiz, Ankara Etlik City Hospital, Turkiye

PHENYLKETONURIA

The nature of metabolic disorders in patients with phenylketonuria
Olga Khmil, Interregional Specialized Medical and Genetic Center Center for Rare (Orphan) Diseases, Ukraine

Our experience with Long Neutral Amino acid supplementation in paediatric PKU patients in India
Ketki Kudalkar, Navi Mumbai Institute of Research in Mental and Neurological handicap (NIRMAN), India

Design of a global, multicenter study to assess maternal, fetal, and infant outcomes of pegvaliase exposure during pregnancy and breastfeeding
Nicola Longo, University of Utah, USA

Is there an intrauterine growth retardation in Bulgarian patients with phenylketonuria and hyperphenylalaninemia?
Maria Sredkova-Ruskova, University Pediatric Hospital, Bulgaria

Phenylketonuria during pregnancy: how we managed four women
Albina Tummolo, Children Hospital Giovanni XXIII Azienda Ospedaliero-Universitaria Consorziale, Italy

Dynamics of Newborn Screening for Phenylketonuria in Moldova
Dan-Cristian Usurelu, Institute of Mother and Child, Moldova

Managing the uncontrolled PKU patient who cannot feed
Greg Woodhead, The Royal Children's Hospital, Australia

UREA CYCLE DISORDERS

Design of a phase 3 study of AAV-mediated gene transfer of ornithine transcarbamylase (OTC) in patients with late-onset OTC deficiency
Jean-Baptiste Arnoux, Necker-Enfants Malades University Hospital, France

Retrospective diagnosis of OTC deficiency in a deceased neonate following presentation in a subsequent sibling - A case for postmortem genomic screening in all neonatal deaths
Drago Bratkovic, Women's and Children's Hospital, Australia

First two cases of arginase 1 deficiency in Slovakia
Vladimir Bzduch, National Institute of Children's Diseases, Slovakia

Biochemical profile of argininosuccinate synthase 1 deficiency patients from a referral hospital in Brazil
Moacir Wajner, Universidade Federal Do Rio Grande Do Sul, Brazil

Clinical trial of N-carbamoyl-L-Glntamic acid in two patients with late-onset ornitine transcarbamirase deficiency (OTCD)
Yuta Sudo, Fujita Health University School of Medicine, Japan

INFORM 2023 to Bring Latest in Fatty Acid Oxidation Disorders Research to Israel

10th gathering of International Network for Fatty Acid Oxidation Research and Management to showcase global advancements.

Global experts, practitioners and researchers in fatty acid oxidation disorders will convene in Jerusalem, Israel, August 27th and 28th for the 10th Anniversary meeting of INFORM, the International Network for Fatty Acid Oxidation Research and Management. The yearly gathering is recognized as the premier international meeting on fatty acid oxidation disorders.

“INFORM was first envisioned more than a decade ago as a global event to encourage and foster collaboration by participating centers and practitioners, and to feature breakthrough research and best practices for treatment of fatty acid oxidation defects,” said Dr. Jerry Vockley, MD, PhD. INFORM’s founding co-chair and scientific organizer. “This year promises to elevate our mission.”

The event will precede the Society for the Study of Inborn Errors of Metabolism (SSIEM); Prof. Yair Anikster MD, PhD, the Director of the Metabolic Disease Unit at Edmond and Lily Safra Children's Hospital and Chairman of the Israel Society for Metabolic Diseases, serves as president of SSIEM 2023.

INFORM presenters include keynote speaker Gerard Vockley, M.D., Ph.D., University of Pittsburgh, Cleveland Family Professor of Pediatric Research, Professor of Human Genetics , Chief of Medical Genetic and Genomic Medicine, Director of the Center for Rare Disease Therapy Children's Hospital of Pittsburgh of UPMC, whose presentation is entitled, ***“Gene therapy meets fatty acid oxidation disorders (finally!)”***. Israeli presenters will include Dr. Shlomo Almashanu, the head of the Israeli national newborn screening program, who will speak on: *"Fatty acid oxidation disorders in Israel"*; and Dr. Orna Staretch-Chacham, who will report on: *"Unique MCAD exon 8 duplication with a founder effect among Northern African Jews".*

INFORM began more than a decade ago as part of a National Institutes of Health grant application to establish a Rare Disease Research Network on fatty acid oxidation disorders. While the application was ultimately unsuccessful, a suggestion from one reviewer to begin demonstrating the ability to work together led the founders to seek funding to meet the goals of INFORM through industry sponsorships and unrestricted educational grants.

Now, more than a decade later, INFORM provides a platform for collaborating centers to interact face-to-face, present state of the art research, and share best practices for treatment of fatty acid oxidation defects. This year, the steering committee of international experts on fatty acid oxidation disorders is planning the agenda, from evaluating abstracts to vetting potential speakers. The resulting broad-based program will bridge clinical and research issues, and includes patients and patients advocates.

The inaugural INFORM event was held in 2014 in Innsbruck, Austria. Subsequent meetings have been held in Lyons, France; Boston, Massachusetts; Rio De Janeiro Brazil; Athens, Greece, Amsterdam, The Netherlands; and Freiburg, Germany. Two were held virtually due to COVID. As with all previous events, the upcoming one will invite families and patients from Israel to participate.

Funding to support the program continues to be provided by industry sponsorships and unrestricted educational grants as well as individual donors. Last year's donors included Acer Therapeutics; American Gene Technologies; Arcturus Therapeutics; CoA a BioBridge Company; Homology Medicines; Kriya; Moderna; Nestle Health Science; PerkinElmer Genetics; Reneo; Solace Nutrition; Stealth; Ultragenyx Pharmaceuticals; and Vitaflo. All funds are collected and managed by the Children's Hospital of Pittsburgh Foundation.

"This meeting showcases the role that the Center for Rare Disease Therapy at the UPMC Children's Hospital of Pittsburgh plays in training national and international clinicians and its reputation as a destination for the development and pursuit of clinical trials for novel therapeutics for these disorders", said Dr. Vockley.

Future events are scheduled for Porto, Portugal, in 2024, Japan in 2025 during preceding the ICIEM, Helsinki, Finland, in 2026 and Dublin, Ireland in 2027.

INFORM is committed to discovering new FAOD treatments and cures to improve the lives of patients and their families. Learn more at https://informnetwork.org/

Past and present members of the INFORM Scientific and Organizing Committee:

Jerry Vockley, MD, PhD Co-Chairman University of Pittsburgh School of Medicine

Ute Spiekerköetter, MD Co-Chairman Department of Pediatrics and Adolescent Medicine, University Children's Hospital, Freiburg

Dr. Nicola Longo, M.D. Ph.D., University of Utah, Division of Medical Genetics

Michael Bennett, PhD Michael J. Palmieri Metabolic Laboratory, University of Pennsylvania School of Medicine

*Jean Bastin, PhD INSERM-University Hôpital Necker-Enfants Malades

*Niels Gregersen, PhD Research Unit for Molecular medicine, MMF Aarhus University Hospital, Skejby Brend-strupgaardsvej

Daniela Karall, MD Department for Child and Adolescent Medicine, Medical University of Innsbruck

Melanie Gillingham, PhD, RD Molecular and Medical Genetics Department, Oregon Health & Science University

Rikke Katrine Jentoft Olsen, MSc, PhD, Associate Professor Research Unit for Molecular Medicine, Aarhus, Denmark

Dr. Kimihiko Oishi, M.D., Chair and Professor of the Department of Pediatrics at the Jikei University School of Medicine, Tokyo, Japan, and Vice Chair of the Japanese Society for Inherited Metabolic Diseases

Keith McIntire, INFORM Program Manager

*Past Committee Members

INFORM

Celebrating 10 YEARS

2023 AGENDA

SUNDAY, August 27th

Session 1 | KEYNOTE | POSTER PRESENTATIONS

3:00 - 4:00 PM	Registration Opens
4:00 Keynote	Keynote: "Gene therapy meets fatty acid oxidation disorders (finally!)": **Jerry Vockley**
5:00 - 5:30	Questions and Open Discussion
6:00 - 7:30	Casual Dinner & Reception
7:30 - 9:30	Poster Presentations

MONDAY August 28th

Session 2 | New Therapies for FAODs

7:00 - 8:00 AM	Registration Opens
8:00 - 8:15	New therapies for MCADD / C7 (Dojolvi): **Georgianne Arnold**
8:15 - 8:30	Sodium Phenylbutyrate as Therapeutics: **Jerry Bedoyan**
8:30 - 9:00	Return to activity following rhabdomyolysis: toward developing a clinical guideline: **Pamela Tucker**
9:00 - 9:15	Medium-chain acylcarnitines target muscular phenotypes in long-chain fatty acid oxidation Disorders: **Keaton Solo**
9:15 - 9:30	Elamipretide restores mitochondrial function in trifunctional protein deficiency mice and human fibroblasts: **Eduardo Vieira Neto**
9:30 -10:00	Break

Session 3 | Ketone Use in FAODs

10:00 - 10:30	Animal Models: **Emmalie Jager**
10:30 - 11:00	Human Studies: **Melanie Gillingham**
11:00 - 11:30	Sports Medicine and Ketones: **Daniele Zaccaria**
11:30 - 11:45	Fasting metabolism in children with fatty acid oxidation disorders: Earlier lipolysis in VLCADD compared to MCADD patient: **David Olsson**
12:30 - 2:00PM	Lunch

AGENDA

MONDAY August 28th (continued)

Session 4 | FAODs in Israel and abroad

2:00 - 2:30	FAOD in Israel's NBS Program: **Shlomo Almashanu**
2:30 - 3:00	Unique MCAD exon 8 duplication with a founder effect among Northern African Jews: **Staretch-Chacham**
3:00 - 3:15	Comprehensive multi-parametric cardiac MRI (CMR) in mice with a mutation in the mitochondrial trifunctional protein β-subunit – a model of cardiac fibrosis: **Eduardo Viera Neto**
3:15 - 3:30	Early diagnosis by newborn screening (NBS) or prior family history is associated with improved visual outcomes of Long-chain 3-hydroxyacyl-CoA dehydrogenase deficiency (LCHADD) chorioretinopathy: **Melanie Gillingham**
3:30 - 3:45	Break

Session 5 | Basic Science

3:45 - 4:15	A high-throughput screening platform to probe protein/metabolite interactions enabled rapid discovery of a functional allosteric binding site on MCAD: **Shomit Sengupta**
4:15 - 4:45	Immunology and FAODs: **Abbe De Vallejo**
4:45 - 5:00	Lysine hyposuccinylation in human MCAD deficient fibroblast cells alleviated with heptanoic and medium branched-chain fatty acids and in Acadm-/- mice with triheptanoin: **Anuradha Karunanidhi**
5:00 - 5:15	Muscle contraction-related proteome and calcium homeostasis are disrupted in CPT2-deficient mice: **Andrea S. Pereyra**
5:15 - 5:30	Adjournment
5:30 - 6:30	INFORM business meeting

INFORM

www.informnetwork.org

FOR MORE INFORMATION, PLEASE CONTACT:
Keith W. McIntire, INFORM Program Manager
UPMC / Children's Hospital of Pittsburgh
Division of Genetics and Genomics
Phone: **412.692.5099 | keith.mcintire@chp.edu**

UPMC | CHILDREN'S HOSPITAL OF PITTSBURGH

NETWORKING

Welcome Reception

Tuesday, 29 August from 19:00-20:30

ICC Exhibition area

Join us for an extraordinary scientific exchange at the highly anticipated SSIEM 2023 Welcome Reception held at the prestigious ICC in the exhibition area. This exclusive event marks the beginning of the symposium, igniting new research ideas and fostering invaluable networking opportunities. Be part of this memorable occasion as we gather in Jerusalem to celebrate the opening of SSIEM 2023!

Poster Walk + Highest Ranked Posters

Wednesday, 30 August from 18:35-20:30

ICC Exhibition Hall

Engage in thought-provoking discussions with abstract authors and gain valuable insights into their research findings. Additionally, talented young investigators will present their outstanding posters in 4-minute talks, followed by an open discussion. This engaging event offers a platform for meaningful exchanges and opportunities to explore emerging ideas. Take advantage of this valuable opportunity to broaden your knowledge and connect with fellow researchers.

Networking Activities

Thursday, 31 August from 15:30 – 18:30

Jerusalem city

Embark on an immersive journey in the company of friends and colleagues to experience the wonders of magical Jerusalem. This extraordinary city intertwines tradition, religion, history, and modern culture, offering an abundance of treasures to explore. From guided tours of historic sites to culinary adventures in the vibrant Machane Yehuda Market, diverse networking activities suit every interest.

Participation in these activities is optional and requires pre-registration. Don't miss the opportunity to enhance your conference experience and connect with fellow attendees.

Networking Evening

Thursday, 31 August from 20:00-23:00

Jerusalem Botanical Gardens

Experience an enchanting evening networking event at the Jerusalem Botanical Gardens, exclusively for the SSIEM 2023 attendees.

Enjoy the serene beauty of the gardens, featuring a diverse collection of plants from around the world. Connect with fellow participants, indulge in delectable refreshments, and create lasting memories while networking in this picturesque setting.

The evening will be filled with live entertainment, and transportation from the ICC to the Botanical Gardens will be available for symposium participants via shuttle bus. Join us for an unforgettable evening of networking, entertainment, and celebration during SSIEM 2023!

GENERAL INFORMATION

Venue

The conference will be held at The Jerusalem International Convention Centre (ICC) located at the entrance of Jerusalem. It has hosted a variety of events from conferences and business meetings, exhibitions, shows, cultured events, product launches and banquets.
The ICC has easy access to the Tel Aviv and Ben Gurion International Airport and the surrounding areas.

ICC Jerusalem – International Convention Center
PO 6001 Jerusalem 9106001
Tel: +972-2-655-8558

Language

Israel's official languages are Hebrew and Arabic. Most Israelis speak English.
The official language of the conference is English.

Weather and Clothing

Israel's northern and coastal regions are classified as Mediterranean climate characterized by hot and dry summers and cool rainy winters; whereas the southern and eastern areas of Israel are characterized by an arid climate.
Israel enjoys temperate weather throughout the year. No rain is expected from early spring through late autumn.
Along with avoiding excessive exposure to the sun, visitors are recommended to wear protective head coverings and use sun blocks.
Jerusalem and the Galilee are colder than Tel Aviv, especially in the evening. Jacket is recommended.

Currency

The unit of currency in Israel is the New Israeli Shekel (NIS), commonly known as the Shekel.

Electricity

Electricity in Israel is supplied at 220V, 50Hz and has a three-pin connecting plug.

Value Added Tax Reimbursement

According to the Value Added Tax (VAT) Law, a foreign tourist is able to benefit from a zero rate of VAT on a vast array of services obtained during a stay in Israel. In addition, a special arrangement exists whereby a tourist may also request a VAT refund on purchases made during a stay.

SHABBAT IN ISRAEL

Learn more about Shabbat

Shabbat in Jerusalem is a unique and special time. However, it raises many practical questions about what to do, where to eat, and how to get around.

Shabbat (Saturday) begins at sundown on Friday and ends at sundown on Saturday (when a new week begins).

Starting from early Friday afternoon, businesses, shops, and most restaurants begin to shut; however, some non-Kosher restaurants remain open during Shabbat, as well as a limited number of businesses. Public transportation (buses and railways) do not run at all in Jerusalem during Shabbat, and all these services pause in the hours leading up to sunset. The final train from Jerusalem to Tel Aviv/ airport departs at 14:39 on Friday afternoon, while shared taxis and private taxis continue to operate .

When considering your travel plans home, please be aware that it takes approximately 25 minutes to get to the airport from Jerusalem by train, or approximately 30 minutes by taxi and that the advised time to arrive for check-in at the airport is 3 hours before departure.

Please keep in mind the timing described above when planning your travels on Friday afternoon/evening.

Since sunset hours vary throughout the year, the time that Shabbat begins and ends also varies. Please keep in mind that each business has its own rules regarding hours of operation, but it is safe to say that in the summer, most businesses will close on Friday between 2 pm to 4 pm.

Regarding hotels on Shabbat – hotels have check-ins/checks out regularly. Before booking, check the hotel website for their hours of operation during Shabbat.

Saturday evenings after Shabbat ('Motzei Shabbat' in Hebrew) is when most businesses, restaurants, and public transportation begin again, usually around one hour after the end of Shabbat. Bus services resume almost immediately after the end of Shabbat, and the first train from Jerusalem (after Shabbat) to Tel Aviv/ airport departs at 21:39.

Shabbat is a wonderful time to explore Jerusalem; since most businesses and services are closed, the city is much quieter than usual, with minimal traffic.

Departing Israel

Security Check

As from 2014, the Israel Airports Authority has implemented a modern advanced security system for baggage inspection for flights departing Ben Gurion Airport – Hold Baggage Screening (HBS).
The HBS was developed by the Israel Airports Authority for totally automatic passenger baggage security inspection, and to improve the level of service. The system complies with the strictest of requirements and is based on the highest level of technology.

The new security inspection process includes a few questions, after which passengers will proceed directly to the counter of their respective airlines in order to check in for their flight and hand over their baggage.

After tagging your baggage at the airline counter, the baggage will be sent for the automatic inspection by the new HBS system. In certain cases the need may arise to open the baggage for manual inspection. This manual inspection is carried out under total electronic surveillance and documented.

NOTE:
Bags should be unlocked.
Security leaves the 3.5 hrs. time which is published for the whole security/check-in procedure as unchanged. This means that the security lines will open at D-3.5 hrs and check-in opens at D-3hrs.

Late passengers' acceptance – will be coordinated with the airlines as done before. It should be clear that 1 hour time is still needed for the baggage security process in the HBS. Late passengers will therefore be advised that there is a chance that their luggage will not make it to the flight on time. Therefore counters will be closed 60 min before departure.

Oversized baggage will be dealt in front of the elevator dedicated for OOG luggage.
No oversize baggage and/or unsuitable luggage will be accepted at check-in and to be sent to the HBS system and the baggage sorting system accordingly.
The Maximum dimensions allowed to be accepted at check-in – Length: 115 cm, Width: 80 cm, Height: 70 cm, Weight: 40 kg

Check-In

After the security check, each passenger must check in at the counter of the airline they are flying with. They will then check in their luggage and receive a boarding pass and seat number.

Early Check-In Service – Some of the airline companies have early check-in service. Passengers can clarify this with the airline. Information about early check-in for airlines other than El Al is available from the airport information service: +972-3-9755555 or on the Israel Airports Authority website . For early check-in services on El Al flights please see El Al Website.

Passport control

After check-in passengers continue to passport control. They must present their passport, airline ticket, and the form that they filled out upon arrival in Israel.

Value Added Tax Reimbursement

According to the Value Added Tax (VAT) Law, a foreign tourist is able to benefit from a zero rate of VAT on a vast array of services obtained during a stay in Israel. In addition, a special arrangement exists whereby a tourist may also request a VAT refund on purchases made during a stay. For more details please visit the official website of the Ministry of Finance, Israeli Department of Customs and VAT.

SPONSORS SECTION

SPONSOR ACKNOWLEDGEMENT

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New perspectives in PKU management

The change we can see

Tuesday, 29 August 2023

12:45-13:45 hrs, The Pincus Hall (ground floor)

Faculty

Cary Harding
Oregon Health & Science University, Portland, OR, USA

Johannes Krämer
Ulm University Medical School, Ulm, Germany

Frank Rutsch
Münster University Children's Hospital, Münster, Germany

Treating adult MPS IVA patients: Recent findings from the largest global Morquio A Registry Study

Thursday, 31 August 2023

12:30–13:30 hrs, The Pincus Hall (ground floor)

Nathalie Guffon, MD
Reference Centre for Inherited Metabolic Diseases, Femme Mère Enfant Hospital, Hospices Civils of Lyon, Lyon, France

Christina Lampe, MD
Centre for Rare Diseases, University of Giessen, Giessen, Germany

These symposia have been initiated, organised and funded by BioMarin. These symposia are intended for healthcare professionals registered for SSIEM 2023.

PALYNZIQ®▼ Abbreviated Prescribing Information

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▼This medicinal product is subject to additional monitoring. This will allow quick identification of new safety information. Healthcare professionals are asked to report any suspected adverse reactions.

EU-MPRL-00070 - June 2023

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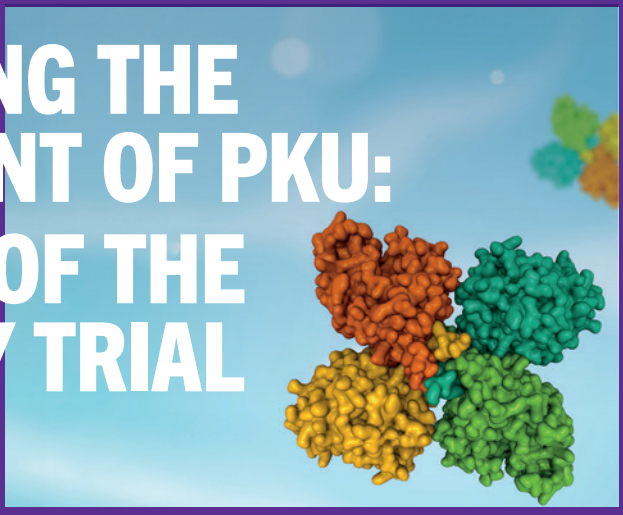
Tuesday, 29 August
12:45–13:45
Dulcin Hall

AADC DEFICIENCY: ADVANCES IN DIAGNOSIS AND TREATMENT

ADVANCING THE TREATMENT OF PKU: RESULTS OF THE APHENITY TRIAL



Thursday, 31 August
12:30–13:30
Dulcin Hall

AADC, aromatic L-amino acid decarboxylase; PKU, phenylketonuria.
These industry-sponsored symposia are organised and funded by PTC Therapeutics and are intended for healthcare professionals only.

MED-ALL-CORP-2300013 | July 2023

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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

Come visit us at our booth

Sanofi aventis Israel ltd.
Approval Date: July 2023
MAT-GLB-2203149 v1.0

SSIEM Annual Symposium

Neurocognitive aspects of MPS II

from early diagnosis to developmental assessment

Wednesday 30 August 2023, 17:30–18:30 IDT
International Convention Center – ICC Jerusalem – Pincus Hall

Come and join us to learn more about Mucopolysaccharidosis (MPS) II

Join our international expert faculty for a discussion of the inherited lysosomal storage disease MPS II in an informative satellite symposium. The discussions will focus on disease burden for patients and caregivers; diagnosis and natural history of cognitive development in patients; and the psychological and neuropsychological testing and assessment process.

A Takeda-sponsored satellite symposium at the annual Society for the Study of Inborn Errors of Metabolism (SSIEM) 2023 congress

This meeting is intended for healthcare professionals registered for SSIEM 2023 congress only and is initiated, organized, and funded by Takeda.

VV-MEDMAT-88754 Date of preparation: June 2023

As the first Italian Company to commit to researching more effective and practical formulations in the field of inherited, inborn metabolic diseases, **PIAM** strive to introduce innovative and technologically advanced products that aim to improve patients' adherence to medication.

Our Vision

We offer effective solutions and enable the dissemination of a culture of health where people and their needs come first. Our approach to care stems not only from over a century of experience working alongside physicians and within the scientific community of the pharmaceutical industry, but also **from 30 years of research in nutrition care for patients with rare diseases.**

Our unique past and expertise mean that we have built our offer on and around the real needs of physicians, patients and caregivers, to whom we offer all the pharmacological and nutritional treatment options and services needed for the management of specific needs.

Our Mission

For over a century, our one mission has been to provide the best possible and most cutting-edge responses to a growing need for better health.

Through harmonizing nutrition and pharmacological therapy options, we are able to offer the specialists who consistently choose us, a comprehensive platform of integrated therapeutic solutions and personalised services that cater to individual needs.

A strong, long-standing network that brings together high-profile international partners and focuses on clinical research and development implies that, at **PIAM**, we have the know-how to seize and offer the best and most innovative, effective and safe therapeutic opportunities on the global market.Our lean, yet ethically and scientifically rigorous business model means that we are in the position to address unmet needs through releasing the top-most pharmacotherapy solutions.

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