where EAST meets WEST

SSIEM

SSIEM Annual Symposium 2023

29 August - 1 September 2023 | Jerusalem, Israel

PROGRAM

החברה הישראלית למחלות מטבוליות
Israel Society for Metabolic Diseases

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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 Symposium – 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

SSIEM 2023 Symposium President

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

Local Scientific Committee

Shlomo Almashanu
Ministry of Health, Jerusalem, 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

Dulce Quelhas | Centro De Genetica Medica Centro Hospitalar Universitario Santo Antonio, 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 | SSIEM Council, Athens, Greece

Philippa Mills | University College London, 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 | Hôpital Necker-Enfants Malades, 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 Jerusalem Hotel

13:00-18:30	**2023 CTX InternationalScientific Meeting**
15:00-21:30	**INFORM Meeting** International Network for Fatty Acid Oxidation Research and Management

MONDAY 28 AUGUST 2023
ICC Jerusalem

	Oren 2	Oren 3	Oren 4	Seminar Room 310	Seminar Room 311	Seminar Room 312	Seminar Room 313
13:00	13:00-21:00 **Registration** (ICC Main Entrance)						
13:30							
14:00				14:00-16:00 **EHOD Executive Board** (By invitation only)			
14:30							
15:00							
15:30							
16:00							
16:30							
17:00							
17:30							
18:00							
18:30							
19:00							
19:30							

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

	MONDAY AUGUST 28 2023 Ramada Jerusalem Hotel		MONDAY AUGUST 28 2023 Location: Neve Shalom	
Seminar Room 314	07:30-18:30	**INFORM Meeting** International Network for Fatty Acid Oxidation Research and Management	16:30-20:00	**White Matter Diseases Meeting** *Local Committee Endorsed*
14.00-16.00 **SSIEM Pre-Council Meeting** (SSIEM Honorary Officers & Staff Only)	08:00-17:15	**2023 CTX International Scientific Meeting**		
16.00-18.00 **SSIEM Council Meeting** (Council Members Only)				

TUESDAY 29 AUGUST 2023 | ICC Jerusalem

	Ussishkin Hall	Pincus	Dulzin	Oren 1	Oren 2	Oren 3
7:30	07:30 - 20:30 **Registration** (ICC Main Entrance)					
9:00				09:00-12:00 **Galnet Galactosemia Symposium** (By invitation only)	09:00-12:30 **SSIEM Nutrition & Dietetics Session** (Open to all participants)	09:00-12:00 **SSIEM Adult Session** (Open to all participants)
9:30						
10:00						
10:30						
11:00						
11:30						
12:00						
12:30				12:30-13:30 **SSIEM Adult Business Meeting** (Open to all participants)		
13:00		12:45-13:45 **Satellite Symposium**	12:45-13:45 **Satellite Symposium**			12:45-13:45 **Satellite Symposium**
13:30						
14:00	14:00-14:50 **Opening Ceremony – Epitranscriptome**					
14:30						
15:00	14:50-16:20 **Where East Meets West**					
15:30						
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						

19:00-20:30 Welcome Reception (Open to all participants)

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

	Oren 4	Seminar Room 310	Seminar Room 311	Seminar Room 312	Seminar Room 313	Seminar Room 314	Exhibitions Hall | Hadarim
	09:00-12:00 **SSIEM Patient Advocacy Session: The Psychosocial Impact of Living with an Inborn Error of Metabolism** (Open to all participants)	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)		09:00-11:00 **EHOD "Remethylation Guidelines Group"** (By invitation only)	
					11.00-14.00 **ERNDIM Participant Meeting** (Open to all ERNDIM participants)		
							Exhibitions Hall 15:30-19:30 **Posters**

WEDNESDAY 30 AUGUST 2023 | ICC Jerusalem

	Ussishkin Hall	Pincus	Dulzin	Oren 1	Oren 2	Oren 3
7:00	07:00-20:00 **Registration** (ICC main entrance)					
7:30			07:30-08:30 Satellite Symposium			
8:00						
8:30	08:30-10:00 Educational: Pitfalls to avoid in the management of IEM	08:30-10:00 Innovative Therapies I + New Diseases	08:30-10:00 Mitochondrial Disorders I		08:30-10:00 Clinical Studies & Outcomes I	08:30-10:00 SSIEM Nurses Meeting (Open to all participants)
9:00						
9:30						
10:00						
10:30	10:30-12:00 Learning from the Neighbours					
11:00						
11:30						
12:00						
12:30						
13:00						
13:30						
14:00	14:00-15:30 Lysosomal disorders	14:00-15:30 CDG / Protein Modifications	14:00-15:30 Urea cycle disorders		14:00-15:30 Vitamins + Neuro-transmitters	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 Symposium				17:30-18:30 Satellite Symposium
18:00						
18:30						
19:00						
19:30						
20:00						

Networking | Parallel Session | Poster | Satellite Symposium

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

Oren 4	Seminar Room 310	Seminar Room 311	Seminar Room 312	Seminar Room 313	Seminar Room 314	Exhibitions Hall | Hadarim
	07:30-08:30 **IOC Meeting** (By invitation only)					**Hadarim Hall** 07:30-08:30 Speed Mentoring
08:30-10:00 **Disorders of FAO and Ketones**				08:15-10:15 **SSIEM Dietitians Group Committee Meeting** (Open to all participants)		**Exhibitions Hall** 07:30-20:15 Posters
12:30-13:00 **SSIEM Council & Advisory Council Meeting** (By invitation only)						
13.00-14.00 **SSIEM Annual General Meeting** (SSIEM Members Only)						
						Exhibitions Hall 18:35-20:35 Poster Walk + Highest Ranked Posters

THURSDAY 31 AUGUST 2023 | ICC Jerusalem

	Ussishkin Hall	Pincus	Dulzin	Oren 1	Oren 2	Oren 3
7:00	07:00-15:00 **Registration** (ICC main entrance)					
7:30						
8:00						
8:30	08:45-10:15 **Where big data meets small practices**					
9:00						
9:30						
10:00						
10:30	10:45-12:15 **Phenylketonuria**	10:45-12:15 **Novel diagnostic/ laboratory methods including omics**	10:45-12:15 **Newborn Screening**			10:45-12:15 **Peroxisomal and Purines**
11:00						
11:30						
12:00				12:00-13:30 **SSIEM JIMD Editors, JIMD Communicating Editors and JIMD Advisory Members Board Meeting** (By invitation only)		
12:30		12:30-13:30 **Satellite Symposium**	12:30-13:30 **Satellite Symposium**			12:30-13:30 **Satellite Symposium**
13:00						
13:30	13:30-14:15 **Komrower Lecture**					
14:00	14:15-15:30 **Mitochondrial disorders II**	14:15-15:30 **Organic Acidurias**	14:15-15:30 **Clinical Studies & Outcomes II**		14:15-15:30 **Sulphur related and other amino acid disorders**	14:15-15:30 **Innovative Therapies II**
14:30						
15:00						
15:30						

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

FRIDAY 1 SEPTEMBER 2023 | ICC Jerusalem

	Oren 4	Exhibitions Hall | Hadarim
7:30		
8:00		Hadarim Hall 07:45-08:45 Speed Mentoring
8:30		
9:00		
9:30		Exhibitions Hall 07:45-15:30 Posters
10:00		Hadarim Hall 10:15-10:45 Coffee with the SSIEM JIMD Editors
10:30		
11:00		
11:30		
12:00		
12:30		

	Ussishkin Hall	Exhibitions hall | Hadarim
7:30	**07:45-12:45 Registration** (ICC main entrance)	
8:00		
8:30	08:15-09:45 Therapeutic updates - Advances in therapy modalities	Exhibitions Hall 08:15-12:30 Posters
9:00		
9:30		
10:00	10:15-11:15 Late Breaking News	
10:30		
11:00	11:15-12:00 Latest innovations in Research and Therapy – LOC Endorsed	
11:30		
12:00	12:00-12:20 SSIEM 2023 Best Poster Awards	
12:30	12:20-12:30 Introduction to SSIEM 2024 Porto and Closing Remarks	

Please note: This program is subject to change without prior notice.
For updated information please visit: www.ssiem2023.org

SUNDAY | 27 AUGUST 2023

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

INFORM Meeting
15:00-21:30, Ramada Jerusalem Hotel
International Network for Fatty Acid Oxidation Research and Management

MONDAY, AUGUST 28, 2023

INFORM Meeting
07:00-18:30, Ramada Jerusalem Hotel
International Network for Fatty Acid Oxidation Research and Management

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

Registration
13:00-21:00, ICC Main Entrance

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

SSIEM Pre-Council Meeting
14:00-16:00, Seminar Room 314 (ICC) *(SSIEM Honorary Officers & Staff Only)*

SSIEM Council Meeting
16:00-18:00, Seminar Room 314 (ICC) *(Council Members Only)*

TUESDAY | 29 AUGUST 2023

Registration
07:30-20:30, ICC Main Entrance

GalNet Galactosemia Symposium
09:00-12:00, Oren 1 (ICC) *(By invitation only)*

SSIEM Nutrition & Dietetics Session *(Open to all participants)*
09:00-12:30, Oren 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)**
Richard Godfrey, London, UK

10:30 ***Refreshment Break*** | *Exhibition Area*

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 acidemia: what is ideal?**
Rajavel Elango, Vancouver, Canada

SSIEM Adult Session
09:00-12:00, Oren 3 (ICC) *(Open to all participants)*

09:00-10:30 | Update on therapeutic options for adult patients with IMDs

09:00 **SGLT2 inhibitors in adult patients with glycogenosis 1B**
Saskia Wortmann, Austria

09:30 **A large retrospective case series of management of mothers with hyperphenylalaninaemia during pregnancy**
Tarekegn Hiwot, United Kingdom

09:50 **Hematopoietic stem cell transplantation in adult-onset Krabbe disease**
Gabriella Horvath, Canada

10:10 **Leriglitazone in adult patients with cerebral forms of X-linked adrenoleukodystrophy**
Fanny Mochel, France

10:30 ***Refreshment Break*** | *Exhibition Area*

11:00-12:00 | Take-home messages from case series in adult patients with IMDs10:10

11:00 **The cognitive and mental health support of adults with IMDs – a review of the newly developed neuropsychology service in one tertiary metabolic centre**
Adrian Heald, United Kingdom

11:15 **The landscape of long-term problems in 20 adult patients with glycogen storage disorder type 1**
Michel Hochuli, Switzerland

11:30 **Activating mutation of the glucokinase gene in hyperinsulinemic hypoglycemia: phenotype and genotype in 9 adult patients**
Claire Douillard, France

11:45 **Evaluation of novel tools to facilitate the diagnosis of hereditary fructose intolerance in 15 adult patients**
Bianca Panis, the Netherlands

SSIEM Patient Advocacy Session: The Psychosocial Impact of Living with an Inborn Error of Metabolism

09:00-12:00, Oren 4 (ICC)

Presenters:
Kirsty Hoyle – *CEO at Metabolic Support UK*
Laura Smith van Carroll – *Head of Insight & Advocacy at Metabolic Support UK*
Karen Dolins – *Research Lead at Maple Syrup Urine Disease (MSUD) Family Support Group*
Gulcin Gumus – *Research and Policy Project Senior Manager at EURORDIS*
Danaé Bartke – *Executive Director at HCU Network America*
Audience: These sessions are intended for everyone with an interest in the lived experience of people living with inborn errors of metabolism. They are an opportunity to hear stories that give cultural context to the experiences of patients and understand more about the psychosocial impact of living with a rare disease.

09:00 **Welcome** Chair: ***Kirsty Hoyle***

09:05 **What do patients want? MSUK present early findings from 'Thoughts into Action', a research project looking at Quality of Life as defined by patients** | *Metabolic Support UK*

09:20 **MSUD Family Support Group present their story of change through community and cross-cultural collaboration** | *MSUD Family Support Group*

09:35 **The wider context: psychosocial impact of living with a rare disease** | *EURORDIS*

09:50 **Understanding the burden of classical homocystinuria (HCU) from the patient's perspective: A qualiative study** | *HCU Network America*

10:00 **Panel discussion | Q&A /** *All*

10:30 ***Refreshment Break*** | *Exhibition Area*

11:00 **Using the concept of 'Open Space' this free-flowing session will be an opportunity for all stakeholders with an interest in Inborn Errors of Metabolism to raise issues and fund solutions.**
A unique opportunity for patients, industry and healthcare professionals to listen and to speak in a shared space. | *Patient Community Meet up*

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 311 (ICC) *(By invitation only)*

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

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

ERNDIM Participant Meeting
11:00-14:00, Seminar Room 313 (ICC) *(Open to all ERNDIM participants)*

10:30-11:00 ***Coffee Break*** | *Exhibition Area (ICC)*

12:30-13:30 ***Lunch*** | *Exhibition Area (ICC)*

SSIEM Adult Business Meeting
12:30-13:30, Oren 1 (ICC) *(Open to all participants)*

Satellite Symposium
12:45-13:45, Pincus (ICC)

Satellite Symposium
12:45-13:45, Dulzin (ICC)

Satellite Symposium
12:45-13:45, Oren 3 (ICC), Oren 4 (ICC)

SSIEM 2023 Symposium Opening | *Chairs: Manuel Schiff, France; Yair Anikster, Israel*
14:00-14:50, Ussishkin (ICC)

14:00 **Opening Ceremony**

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

PL1: Where East meets West – differential expression of the same disease in different regions in the world | *Chairs: Ellen Sidransky, USA; Stanley Korman, Israel*
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, Exhibitions Hall (ICC)

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

PL2: The State of the Mitochondria – old players, new roles
Chairs: Shamima Rahman, UK; Ann Saada, Israel
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 | *Chair: Shamima Rahman, UK*
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*

WELCOME RECEPTION
19:00-20:30, Exhibition Area (ICC) *(Open to all participants)*

WEDNESDAY | 30 AUGUST 2023

Registration
07:00-20:00, ICC Main Entrance

Satellite Symposium
07:30-08:30, Dulzin (ICC)

Speed Mentoring
07:30-08:30, Hadarim Hall (ICC)

IOC Meeting
07:30-08:30, Seminar Room 310 (ICC) *(By invitation only)*

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

SSIEM Dietitians Group Committee Meeting
08:15-10:15, Seminar Room 313 (ICC)

08:15 **Welcome and program outline**
Charlotte Ellerton, UK

08:20 **Intestinal microbiota composition of children with glycogen storage type 1 patients**
Sabire Gokalp, TK | Discussion (5 minutes)

08:35 **Phenylalanine Tolerance over Time in Phenylketonuria: A systematic review and meta-analysis** *| Alex Pinto, UK | Discussion (5 minutes)*

08:50 **Effects of combined therapy Ketogenic diet and alglucosidase alfa on Creatine Kinase Levels and motor outcome in Infantile Pompe Disease: Case Series**
Pelin Teke Kisa, TK | Discussion (5 minutes)

09:05 **SSIEM-DG report and activities**
Júlio César Rocha, PT

09:30 **Anita MacDonald Lecture** *| Anne Daly, UK*
Advances and future directions in the nutrition management of long-chain fatty acid oxidation disorders *| Speaker: Melanie Gillingham, USA*

10:10 **Conclusion**
Júlio César Rocha, PT

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

Parallel Session - Educational: Pitfalls in the diagnosis & management of inherited metabolic diseases

Chairs: Stephanie Grunewald, UK; Cristiano Rizzo, Italy

08:30-10:00, Ussishkin (ICC)

08:30 *Andrew Morris, Willink Metabolic Unit, UK*

08:50 *Jean-Marc Nuoffer, University of Bern, Switzerland*

09:10 *Robin Lachmann, University College London Hospitals, UK*

09:30 *Rani H. Singh, Emory University of Atlanta, USA*

Parallel Session: Innovative Therapies I + New Diseases

Chairs: David Weinstein, USA; Carolina Fischinger de Souza, Brazil

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
Chairs: Peter Freisinger, Germany; May Christine Malicdan, USA
08:30-10:00, Dulzin (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
Chairs: Matthias Baumgartner, Switzerland; Raphael Schiffmann, USA
08:30-10:00, Oren 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
Chairs: Ute Spiekerkoetter, Germany; Jerry Vockley, USA
08:30-10:00, Oren 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 Area (ICC)

PL3: Learning from the neighbors
Chairs: Helen Michelakakis, Greece; Risto Lapatto, Finland
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 Area (ICC)

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

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

Parallel Session: Lysosomal disorders | *Chairs: Roberto Giugliani, Brazil; Marc Patterson, USA*
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 | *Chairs: Pascale de Lonlay, France; Katrin Ounap, Estonia*
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 | *Chairs: Carlo Dionisi-Vici, Italy; Galit Tal, Israel*
14:00-15:30, Dulzin (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 during newborn screening**
Jun Kido, Kumamoto University, Japan

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

Parallel Session: Vitamins + Neurotransmitters
Chairs: Sean Froese, Switzerland; Gajja Salomons, The Netherlands
14:00-15:30, Oren 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
Chairs: Anita MacDonald, UK; Smadar Avraham, Israel
14:00-15:30, Oren 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 **A 12-month, longitudinal, intervention study examining a tablet protein substitute preparation in the management of tyrosinemia**
Anne Daly, Birmingham Children's Hospital, UK

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

PL4: The Complexity of brain traffic: new insights from neurometabolism
Chairs: Andrea Gropman, USA; Nicole Wolf, The Netherlands
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 Symposium
17:30-18:30, Pincus (ICC)

Satellite Symposium
17:30-18:30, Oren 3 (ICC),Oren 4 (ICC)

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

B91 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

B191 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

B242 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*

B310 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

B37 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

B192 Role of Osteoclast biomarker (OSCAR) and pro-inflammatory cytokines in Gaucher disease
Margarita Ivanova, Lysosomal and Rare Disorders Research and Treatment Center, USA

B78 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

B35 Evaluation of Repeated Biotinidase Enzyme Activity and The Effect of BTD Gene p.As p444His Variant on Enzyme Activity and Clinical Findings
Fatma Tuba Eminoglu, Ankara University, Turkiye

B196 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

B36 Clinical burden of classical homocystinuria in the United States: a retrospective analysis of Optum Market Clarity | *MAHIM JAIN, Johns Hopkins Medicine, USA*

B267 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*

B254 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

B257 Validation of a tandem mass spectrometry methodology for the analysis of urinary oligo saccharides and free sialic acid for the screening of lysosomal storage disorders
Blai Morales Romero, Hospital Clinic of Barcelona, Spain

B195 Generation of iPSC-derived human neuronal progenitors for the study of GM1 gangliosidosis */ Rodolfo Tonin, A.O.U. MEYER, Italy*

B255 Predicting correct IMD diagnosis using HPO phenotype association algorithms
Judith Jans, UMC Utrecht, The Netherlands

B34 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*

B244 Newborn screening for Fabry disease in Japan: 16 years of experience
Takaaki Sawada, Kumamoto University, Japan

B258 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*

B193 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

B38 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

B243 Trio genome sequencing and detection of metabolic disorders in an Israeli cohort of criti cally ill neonates, the The Israeli NICU-Genomics consortium
Daphna Marom, Tel Aviv Sourasky Medical Center, Israel

B224 Cerebral folate deficiency due to DHFR mutation
Nasrin Hamed, Edmond and Lily Safra Children's Hospital, Sheba Medical Center, Tel Hashomer, Israel

B253 Differential diagnosis of inherited metabolic disorders according to organ system involve ment: a lesson from the knowledgebase | *Nenad Blau, University Children's Hospital, Switzerland*

B256 AI-Powered Genomic Analysis: A New Frontier in Diagnosing Rare Diseases
Jaime Lopes, Cincinnati Children's Hospital, USA

B217 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*

B190 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*

B218 A Novel Homozygous TOP3A Variant in a Pediatric Patient Resulting in Ataxia, Sensorimotor Neuropathy and Cardiomyopathy Due to Mitochondrial Dysfunction Partially Rescued by Ketogenic Conditions in Skin Fibroblasts
Jaya Ganesh, Icahn School of Medicine at Mount Sinai, USA

B194 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

B260 Allow Natural Death (AND) in Inborn Errors of Metabolism
Carolina Fraga, Centro Hospitalar Universitario Santo Antonio, Portugal

B261 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

B197 Long-Term Clinical Outcome of Patients Diagnosed via Newborn Screening for Gaucher Disease in Italy*/Alberto Burlina, Padua University Hospital, Italy*

THURSDAY | 31 AUGUST 2023

Registration
07:00-15:00, ICC Main Entrance

Speed Mentoring
07:45-08:45, Hadarim Hall (ICC)

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

PL5: Where big data meets small practices | *Chairs: Nenad Blau, Switzerland; Shlomo Almashanu, Israel*
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 Area (ICC)

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

Parallel Session: Phenylketonuria | *Chairs: Stephanie Sacharow, USA; Georgianne Arnold, USA*
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

Chairs: Dulce Quelhas, Portugal; Elena Dumin, Israel
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 | *Chairs: Andreas Schulze, Canada; Giancarlo la Marca, Italy*

10:45-12:15, Dulzin (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 | *Chairs: Peter Clayton, UK; Josh Manor, Israel*
10:45-12:15, Oren 3 (ICC),Oren 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 control
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, Oren 1 (ICC) *(By invitation only)*

Lunch
12:15-13:30, Exhibition Area (ICC)

Satellite Symposium
12:30-13:30, Pincus (ICC)

Satellite Symposium
12:30-13:30, Dulzin (ICC)

Satellite Symposium
12:30-13:30, Oren 3 (ICC),Oren 4 (ICC)

Komrower Lecture | *Chair: Manuel Schiff, France*
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 | *Chairs: Rebecca Ganetsky, USA; Hanna Mandel, Israel*
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
Chairs: Stefan Kuhn, South Africa; Yuval Landau, Israel
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
Chairs: David Cassiman, Belgium; Ida Vanessa Doederlein Schwartz, Brazil
14:15-15:30, Dulzin (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
Chairs: Ronen Spiegel, Israel; Ina Knerr, Ireland
14:15-15:30, Oren 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

Chairs: Gerry Berry, USA; Mireia del Toro Riera, Spain

14:15-15:30, Oren 3 (ICC), Oren 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 Activities / **Pre-registration required*

15:30-18:30

SSIEM Networking Evening / **Pre-registration required*

20:30-23:00

FRIDAY | 1 SEPTEMBER 2023

Registration
07:45-12:45, ICC Main Entrance

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

PL6: Therapeutic updates – Advances in therapy modalities
Chairs: Irini Manoli, USA; Eva Morava, USA
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

Coffee Break
09:45-10:15, Agam Foyer (ICC)

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

10:15-11:15 Late Breaking News
Chairs: Stephanie Grunwald, UK; Philippa Mills, UK

10:15 Enasidenib treatment in two individuals with D-2-hydroxyglutaric aciduria carrying a germline IDH2 mutation
Manuel Schiff, Necker University Hospital, APHP and University of Paris Cité, France

10:30 Free cytosolic-mitochondrial DNA triggers a potent type-I Interferon response in Kearns–Sayre patients treatable by mycophenolate mofetil therapy
Martinelli Diego, Bambino Gesu' Children's Hospital, IRCCS, Italy

10:45 Ataxia-telangiectasia: Treating Mitochondrial Dysfunction With a Novel Form of Anaplerosis (A-TC7)
David Coman, Queensland Children's Hospital, Australia

11:00 Glycine amyloid-like Structures in Nonketotic Hyperglycinemia: Immunogenicity and Therapeutic Leads
Dana Laor Bar-Yosef, Tel Aviv University, Israel

11:15-12:00 Latest Innovations in Research and Therapy – LOC Endorsed

Chairs: Dulce Quelhas, Portugal; Yair Anikster, Israel

11:15 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, University of Pittsburgh, USA*

11:24 Results of a Phase III, randomized, placebo-controlled crossover trial with N acetyl-L-leucine for Niemann-Pick disease type C
Julien Park, University of Munster, Germany

11:33 Biallelic variants in RCC1 result in fever associated axonal neuropathy with encephalopathy
Robert Harkness, Manchester University NHS Foundation Trust, Health Innovation Manchester, UK

11:40 PMM2 deficient brain organoids show GLUT1 defect-like metabolic phenotype
Silvia Radenkovic, Mayo Clinic, USA

11:47 Efficacy of oral Manganese and D-Galactose therapy for a novel TMEM165-CDG patient
Arnaud Bruneel, Hospital Bichat-Claude Bernard, France

11:54 In utero enzyme replacement therapy for lysosomal storage disorders - interim results
Billie Lianoglou, San Francisco (UCSF) Benioff Children's Hospital and the UCSF Center for Maternal–Fetal Precision Medicine, USA

12:00-12:20 SSIEM 2023 Best Posters Awards

Chairs: Dulce Quelhas, Portugal; Yair Anikster, Israel

12:00 Highest ranked poster – Runner up TBD

12:06 Highest ranked poster – Winner TBD

12:20-12:30 Introduction to SSIEM 2024 at Porto and Closing Remarks

Lunch

12:30-13:30, Agam Foyer (ICC)

POSTERS

AMINO ACID DISORDERS

B1 Pitfall in Non-Ketotic Hyperglycinemia (NKH) Diagnosis
Rima Abu-Asaad, Rambam Medical Center, Israel

B2 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

B3 Mitochondrial dysfunction in a disorder of transsulphuration: Cystathionine β-synthase deficiency
Mehmet Cihan Balci, Istanbul Medical Faculty Children's Hospital, Turkiye

B4 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

B5 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

B6 Amino Acid Analyses of Plant Foods used in the Dietary Management of Inherited Amino Acid Disorders
Anita McDonald, Birmingham Children's Hospital, Birmingham, UK

B7 Neonatal ethylmalonic encephalopathy with neuroradiological lesions at birth: is it an in utero disease?
Alberto Burlina, University Hospital of Padua, Italy

B8 Bezafibrate prevents myelin alterations, neuroinflammation, and oxidative stress induced by sulfite intrastriatal administration in rats
Moacir Wajner, UFRGS, Brazil

B9 Succinylacetone reduces the antioxidant defenses and induces reactive nitrogen species generation in liver and kidney of developing rats
Moacir Wajner, UFRGS, Brazil

B10 Hydrogen sulfide impairs redox homeostasis and mitochondrial bioenergetics in the striatum of rats
Moacir Wajner, UFRGS, Brazil

B11 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

B12 Exploring the Experiences of Females Living with Fabry Disease in Canada
Julia Alton, Canadian Fabry Association, Canada

B13 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

B14 Breaking bad news in paediatric metabolic disorders; Lessons learnt
Unai Diaz-Moreno Elorz, Great Ormond Street Hospital, UK

B15 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

B16 Long-term clinical evaluation of patients with alpha-mannosidosis – A multicenter study
Fatma Tuba Eminoglu, Ankara University, Turkiye

B17 IGAm index predicts long-term survival in patients with early-diagnosed inherited metabolic disorders
Fatma Tuba Eminoglu, Ankara University, Turkiye

B18 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

B19 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

B20 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

B21 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

B22 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

B23 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

B24 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

B25 Breakthrough in the treatment of glycogen storage disease type 1b (GSD1b)
Magdalena kaczor, The Children's Memorial Health Institute, Poland

B26 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

B27 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

B28 Designing patient-oriented longitudinal disease registries for children with rare metabolic diseases in Canada
Michal Inbar-Feigenberg, The Hospital for Sick Children, Canada

B29 wrong diagnoses prior to the ultimate diagnosis of late-onset pompe disease: a multicenter experience
Tahseen Mozaffar, The University of California, USA

B30 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

B31 Health-related quality of life and fatigue in children with Pompe disease
Hannerieke van Houten, Erasmus MC - Sophia Children's Hospital, Netherlands

B32 Insights into liver disease progression in glycogen storage disease IX γ2: A review of histology
Maheen Sheikh, Duke University, USA

B33 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

B34 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

B35 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

B36 Clinical burden of classical homocystinuria in the United States:a retrospective analysis of Optum Market Clarity
Mahim Jain, Johns Hopkins Medicine, USA

B37 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

B38 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

B39 Management of Inherited Metabolic Diseases in France
Pascale De Lonlay, Filiere de sante maladies rares G2M, France

DIETETICS AND NUTRITION

B40 A Dietician's Experience with the Ketogenic Diet in Dihydrolipoamide Dehydrogenase Deficiency
Smadar - Yaala Abraham, Safra Children's Hospital, Sheba Medical Center, Tel Hashomer, Israel

B41 Management of lipoprotein lipase deficiency with medium-chain triglycerides: a retrospective chart review
Andreas Schulze, University of Toronto, Canada

B42 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

B43 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

B44 Glycogen storage disease Type IIIa with hypertrophic cardiomyopathy: Efficacy and safety of long- term ketogenic diet
Charlotte Mindermann, Hannover Medical School, Germany

B45 Determination of iron deficiency in patients with gluten-related diseases on an agliadin diet
Oleg Denesiuk, Bogomolets National Medical University, Ukraine

B46 A study on the understanding of classical homocystinuria by patients and caregivers
Ida Vanessa Doederlein Schwartz, Hospital de Clinicas de Porto Alegre, Brazil

B47 Biochemical and dietary evaluation of B vitamin deficiencies in patients with hepatic Glycogen Storage Diseases
Ida Vanessa Doederlein Schwartz, Hospital de Clinicas de Porto Alegre, Brazil

B48 Audit of maternal iodine intakes during pregnancy in PKU
Charlotte Ellerton, University College London Hospitals, UK

B49 Opinion of parents regarding early support and information in PKU
Anita McDonald, Birmingham Women's and Children's NHS Foundation Trust, UK

B50 Change in quality of diet and burden of care in children with PKU treated with sapropterin dihydrochloride: a longitudinal study
Alex Pinto, Birmingham Children's Hospital, UK

B51 Intestinal microbiota composition of children with glycogen storage type 1
Sabire Gokalp, Gazi University Faculty of Medicine, Turkiye

B52 Classic galactosemia: the relationship between calcium, vitamin D, and bone mineral density status
Esmeralda Martins, Centro Hospitalar Universitario De Santo Antonio, Portugal

B53 Phenylalanine Tolerance over Time in Phenylketonuria: A systematic review and meta-analysis
Alex Pinto, Birmingham Women's and Children's Hospital, UK

B54 Ketogenic Diet: make it possible for patients with metabolic disorders
Keren Porper, Safra Pediatric Hospital, Chaim Sheba Medical Center, Israel

B55 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

B56 Clinical case: early nutritional approach in two cases of type i glutaric acidury
Marta Suarez Gonzalez, Central University Hospital of Asturias, Spain

B57 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

B58 Optimized diet in a patient with familial chylomicronemia syndrome (FCS) with increased energy requirements receiving Volanesorsen
Alexandra Thajer, Medical University of Vienna, Austria

B59 Early manifestation of GLUT1 deficiency syndrome
Sara Via Dorembus, Edmond and Lily Safra Children's Hospital, Sheba Medical Center, Israel

B60 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

B61 Clinical experiences in transitioning PKU patients from second to third stage protein substitutes: A global survey
Ozlem Yilmaz, Ankara Yildirim Beyazit University, Turkiye

B62 Phenylalanine free infant formula in patients with phenylketonuria: A retro spective study
Ozlem Yilmaz, Ankara Yildirim Beyazit University, Turkiye

B63 Phenylalanine-free infant protein substitute in the dietary management of phenylketonuria
Ozlem Yilmaz, Ankara Yildirim Beyazit University, Turkiye

B64 A 12-month, longitudinal, intervention study examining a tablet protein substitute preparation in the management of tyrosinemia
Anne Daly, Birmingham Children's Hospital, UK

B65 Dietary management of Hereditary Fructose Intolerance (HFI) in eleven Italian metabolic centres: current practices and new challenges
Christian Loro, University of Padova, Italy

DISORDERS OF FATTY ACID OXIDATION AND KETONE BODY METABOLISM

B66 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

B67 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

B68 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

B69 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

B70 Carnitine-acylcarnitine translocate deficiency (CACTD) in the Vietnam referral center: genotype, phenotype and outcome
Khanh Nguyen Ngoc, Vietnam National Children's Hospital, Vietnam

B71 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

B72 Development and characterization of a VLCAD/LCAD muscle double knock out mouse model
Bianca Seminotti, University of Pittsburgh School of Medicine, USA

B73 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

B74 Characterizing pathogenicity of exon and intron variants in very long-chain acyl-CoA dehydrogenase deficiency
Jerry Vockley, University of Pittsburgh School of Medicine, USA

B75 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

B76 Genotype, phenotype and outcome of ketone synthesis defects due to mHS deficiency
Dung Vu Chi, Vietnam National Children's Hospital, Vietnam

B77 Pyruvate dehydrogenase interacts with electron transfer chain supercomplexes
Yudong Wang, University of Pittsburgh School of Medicine, USA

B78 Lysine hyposuccinylation in human MCAD deficient fibroblast cells alleviated with heptanoic and medium branched-chain fatty acids and in Acadm-/- mice wit triheptanoin
Anuradha Karunanidhi, University of Pittsburgh, USA

DISORDERS OF PURINES, PYRIMIDINES, NUCLEIC ACIDS AND PORPHYRIAS

B79 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

B80 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

B81 Dihydropyrimidine dehydrogenase deficiency caused by a novel intragenic large deletion in the DPYD gene
Anna Malekkou, The Cyprus Institute of Neurology and Genetics, Cyprus

B82 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

B83 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

B84 A Southern African MADD cohort: clinical, biochemical, and genetic spectrum
Michelle Bisschoff, North-West University, South Africa

B85 NF-Kb Activity in Lymphocyte Populations in Children with Wilson's Disease
Tatiana Bushueva, National Medical Research Center of Children's Health, Russia

B86 Pyridoxine-dependent epilepsy with neonatal onset: A Report of Two Cases
Martha Caterina Faraguna, Fondazione IRCCS San Gerardo Dei Tintori, Italy

B87 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

B88 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

B89 The importance of high doses of hydroxocobalamin in Brazilian cblC patients
Flavia Piazzon, University of Liege, Belgium

B90 RNA analysis in the diagnosis of Wilson`s disease
Mikhail Skoblov, Research Centre for Medical Genetics, Russia

B91 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

B92 Relationship between the metabolic state of copper and oxidative status in patients with and without GLUT1 deficiency in treatment with a ketogenic diet
Veronica Cornejo, Inta, University of Chile, Chile

B93 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

B94 SRD5A3-CDG restricted to inherited retinal disease phenotype
Hana Hansikova, Charles University and General University Hospital Prague, Czech Republic

B95 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

B96 Retrospective Analysis Of Carbohydrate-Deficient Transferrin For CDG Screening: A Single Center Study
Ozge Ozgen, Istanbul University, Turkiye

B97 Phenotypic profile of inherited GPI deficiency disorders in Polish group of patients
Michal Patalan, Michal Patalan, Pomeranian Medical University, Poland

B98 A negative exome is not the end of the story
Batel Terespolskye, Hebrew University of Jerusalem, Israel

B99 Four year follow-up of mannose therapy in the first Belgian patient with MPI-CDG
Patrick Verloo, University Hospital Ghent, Belgium

B100 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

B101 Clinical experience of three cases of late-onset Krabbe disease
Gabriella Horvath, University of British Columbia, Canada

B102 Understanding the burden of classical homocystinuria (HCU) from the patient's perspective: A qualitative study
Danae Bartke, HCU Network America, USA

B103 Characterization of Gbe1ys/ys mouse model representing a phenotype similar to Adult Polyglucosan Body Disease (APBD)
Priya Kishani, Duke University, USA

B104 Activating mutation of the Glucokinase gene in Hyperinsulinemic Hypoglycemia: phenotype and genotype in 9 adult patients
Claire Douillard, Lille University Hospital, France

B105 A Large retrospective case series of managmnent of mothers with hyperphenylalaninaemia during pregnancy
Tarekegn Hiwot, University Hospital of Birmingham, UK

B106 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

B107 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

B108 Late-onset symptomatic hyperprolactinemia in 6-pyruvoyl-tetrahydropterin synthase deficiency
Wuh-Liang Hwu, National Taiwan University Hospital, Taiwan

B109 Pregnancy, maternal and child health in women with inherited metabolic disorders
Meryem Karaca, İstanbul Medical Faculty, Turkiye

B110 Cardiometabolic risk factors and muscle quality in adult patints with inherited metabolic diseases
Luis M. Luengo-Perez, University of Extremadura / Badajoz University Hospital, Portugal

B111 Clinical features of glucose transporter type 1 deficiency syndrome in a Portuguese Reference Centre
Maria Carmo Macario, Centro Hospitalar e Universitario de Coimbra, Portugal

B112 Hyperammonemia in adults: try to think outside the box
Francesca Maria Menni, Fondazione IRCCS Ca' Granda Ospedale Maggiore, Italy

B113 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

B114 Development of tools to facilitate the diagnosis of hereditary fructose intolerance
Bianca Panis, Maastricht University Medical Center, The Netherlands

B115 Imprinted cell memory in glycogen storage disorder 1a patients`fibroblasts
Uri Sprecher, Tel Aviv University, Israel

B116 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

B117 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

B118 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

B119 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

B120 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

B121 An AAV-mediated liver-directed gene therapy metabolically corrects alkaptonuria in an Hgd deficient mouse model
Sien Lequeue, Vrije Universiteit Brussels, Belgium

B122 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

B123 What are the present obstacles to the incorporation of authorized cell and gene therapies into clinical practice?
Cecilia Marinova, Medasol Outpatient Clinic, Czech Republic

B124 Neonatal AAV8 gene therapy successfully treats severe MSUD in Bckdhb-/- mice
Manuel Schiff, University Paris Cité, France

B125 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

B126 Evaluation of Glycosphingolipids on Dried Blood Spot 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

B127 A Novel Multiplex approach for determining anti-drug antibodies in Fabry Disease
Tomas Baldwin, University College London, UK

B128 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

B129 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

B130 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

B131 The importance of examining skeletal x-rays for dysostosis multiplex in the early diagnosis of mucopolysaccharidosis
Huseyin Bilgin, Diyarbakir Children's Hospital, Turkiye

B132 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

B133 The multiomic landscape of Mucopolysaccharidosis IIIB models
Marianna Caterino, University of Naples Federico II, Italy

B134 Underlying neuropathophysiology of mucopolysaccharidosis type II
Chong Kun Cheon, Pusan National University Children's Hospital, South Korea

B135 Towards personalized medicine in cystinosis: measuring cystine levels in leucocytes and cysteamine concentration in blood
Anibh Das, Hannover Medical School, Germany

B136 Atypical parkinsonism and the relationship of lysosomal disease variants
Ida Vanessa Doederlein Schwartz, Hospital de Clinicas de Porto Alegre, Brazil

B137 Correlation between cognitive function and brain metabolites in late-infantile metachromatic leukodystrophy
Mireia del Toro, Vall d'Hebron University Hospital, Spain

B138 Natural history and clinical characteristics of patients with acid sphingomyelinase deficiency in the era of enzyme replacement therapy: Single Center Experience
Asli Durmus, İstanbul University, Turkiye

B139 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

B140 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

B141 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

B142 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

B143 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

B144 An Alternative for Early Detection of Cardiac Involvement in Gaucher Type 1 Disease: Speckle Tracking Echocardiograpy
Sabire Gokalp, Gazi University Faculty of Medicine, Turkiye

B145 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

B146 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

B147 Oxidative and inflammatory stress parameters in patients with lysosomal acid lipase deficiency
Roberto Giugliani, Universidade Federal do Rio Grande do Sul, Brazil

B148 Qualitative study in adult and caregiver MPSI, II and VI patients : Understanding their challenges, needs and expectations
Nathalie Guffon, HFME Hospital, HCL, France

B149 Peripheric neurodegeneration biomarkers in Niemann-Pick type C1 disease
Roberto Giugliani, Universidade Federal do Rio Grande do Sul, Brazil

B150 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

B151 Early detection and follow-up of patients with Fabry disease. Approaching new biomarkers
Alvaro Hermida, University of Santiago de Compostela, Spain

B152 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

B153 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

B154 Identifying outcomes that matter: developing a core outcome set for pediatric mucopolysaccharidoses
Michal Inbar-Feigenberg, Hospital for Sick Children, Canada

B155 Ten-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

B156 Incidence and Management of Adverse Events Associated with Enzyme Replacement Therapy in Hunter Syndrome
JiHoon Hwang, Sungkyunkwan University School of Medicine, South Korea

B157 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

B158 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

B159 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

B160 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

B161 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

B162 Phenotypic characterization of Alpha-mannosidosis patients in Portugal
Maria Carmo Macario, Centro Hospitalar e Universitario de Coimbra, Portugal

B163 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

B164 Psychosine analysis in dried blood spots and red blood cells from patients with Krabbe disease
Dietrich Matern, Mayo Clinic, USA

B165 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

B166 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

B167 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

B168 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

B169 Multiorgan involvement in females with Fabry disease: results from 2 phase III trials and the followME registry
Peter Nordbeck, University Hospital Würzburg, Germany

B170 Long-term monitoring of cardiac involvement under migalastat treatment using magnetic resonance tomography in Fabry disease
Albina Nowak, University Hospital Zurich, Switzerland

B171 Facial dysmorphologies in Mucopolysaccharidosis type IVA: exploring the morphometry and ancestry components
Harry Pachajoa, Universidad Icesi, Fundacion Valle del Lili, Colombia

B172 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

B173 LYSO-GB3 normalization in fabry disease-treated patients
Fernando Javier Perretta, Fresenius Medical Care Escobar, Argentina

B174 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

B175 Using machine learning to develop an algorithm for early diagnosis of Gaucher disease
Shoshana Revel-Vilk, Shaare Zedek Medical Center, Israel

B176 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

B177 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

B178 Genotype/phenotype correlations in the patients with alpha-mannosidosis in Ukraine
Nataliia Samonenko, National Children's Specialized Hospital "Okhmatdyt", Ukraine

B179 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

B180 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

B181 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

B182 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

B183 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

B184 Mitochondrial biogenesis and autophagy induction in the cerebral cortex of mucopolysaccharidosis type II mice
Moacir Wajner, UFRGS, Brazil

B185 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

B186 Evidence-based Individual Treatment Trials with Immunomodulatory Drugs in Mucopolysaccharidosis
Anna-Maria Wiesinger, Paracelsus Medical University, Austria

B187 Development of Suspicion Index Tool to Aid Diagnosis of ASMD Disease
Anna-Maria Wiesinger, Paracelsus Medical University, Austria

B188 Individual Treatment Trials – Do Experts Know and Use this Option to Improve the Treatability of Mucopolysaccharidosis?
Anna-Maria Wiesinger, Paracelsus Medical University, Austria

B189 Brain MRI pattern in VPS11 hypomyelinating leukodystrophy
Ayelet Zerem, Dana-Dwek Children's Hospital, Tel Aviv Sourasky Medical Center, Israel

B190 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

B191 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

B192 Role of Osteoclast biomarker (OSCAR) and pro-inflammatory cytokines in Gaucher disease
Margarita Ivanova, Lysosomal and Rare Disorders Research and Treatment Center, USA

B193 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

B194 The French Gaucher disease registry: clinical features, complications, and treatment trends of 688 patients
Nadia Belmatoug, Beaujon Hospital, France

B195 Generation of iPSC-derived human neuronal progenitors for the study of GM1 gangliosidosis
Rodolfo Tonin, Meyer Children's Hospital IRCCS, Italy

B196 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

B197 Long-Term Clinical Outcome of Patients Diagnosed via Newborn Screening for Gaucher Disease in Italy
Alberto Burlina, University Hospital of Padua, Italy

METABOLIC MYOPATHIES

B198 Deoxynucleoside therapy for thymidine kinase 2-deficient myopathy: clinical case
Irina Artamonova, Almazov National Medical Research Centre, Russia

B199 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

B200 Favorable response in muscle strength and endurance with chronic exercise in patients with metabolic myopathy
Tanyel Zubarioglu, Istanbul University-Cerrahpasa, Turkiye

MITOCHONDRIAL DISORDERS

B201 Defining the atlas of mitochondrial haplotypes and their regulation on T cell activation
Angi Zenab, Sheba Medical Center, Israel

B202 Mitochondrial aminoacyl- tRNA synthetase deficiency as a new cause of primary Pediatric Intestinal Pseudo-Obstruction
Barbara Siri, Bambino Gesu Children Hospital IRCCS, Italy

B203 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

B204 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

B205 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

B206 A Novel COQ4 Variant Causes Developmental Delay, Regression, Epilepsy and Cardiomyopathy Associated with CoQ10 Deficiency
Elene Kirtadze, Galil Medical Center, Israel

B207 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

B208 A homozygous NDUFA6 variant associated with alopecia, neutropenia, growth retardation and encephalopathy and isolated complex I deficiency
Yoav Zehavi, Emek Medical Center, Israel

B209 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

B210 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

B211 Novel bi-allelic variants in COA7 associated with isolated cytochrome c oxidase deficiency and mitochondrial cardiomyopathy
Lucie Taylor, Newcastle University, UK

B212 Machine Learning can Identify Newborns with Energy Metabolism Conditions
Rebecca Ganetzky, Children's Hospital of Philadelphia, USA

B213 Genotype phenotype correlation in DLD deficiency based on mitochondrial function
Yarden Haham, Sheba Medical Center, Israel

B214 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

B215 Barth syndrome: lessons in metabolism from cellular models
Yana Sandlers, Cleveland State University, USA

B216 Interrogating the role of COA5 in mitochondrial cytochrome c oxidase biogenesis
Rob Taylor, Newcastle University, UK

B217 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

B218 A Novel Homozygous TOP3A Variant in a Pediatric Patient Resulting in Ataxia, Sensorimotor Neuropathy and Cardiomyopathy Due to Mitochondrial Dysfunction Partially Rescued by Ketogenic Conditions in Skin Fibroblasts
Jaya Ganesh, Icahn School of Medicine at Mount Sinai, USA

NEUROTRANSMITTER AND CREATINE RELATED DISORDERS

B219 Improved outcomes in early treated GAMT deficiency – a sibling study
Liora Caspi, Hospital for Sick Children, Canada

B220 Unexplained cerebrospinal fluid findings in GABA transaminase deficiency
Unai Diaz-Moreno Elorz, Great Ormond Street Hospital, UK

B221 "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

B222 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

B223 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

B224 Cerebral folate deficiency due to DHFR mutation
Nasrin Hamed, Edmond and Lily Safra Children's Hospital, Sheba Medical Center, Tel Hashomer, Israel

NEW DISEASES

B225 A rare disease with neurodevelopmental delay and recurrent rhabdomyolysis: a case report of trappc2l-related disorder
Niccolo' Campagna, University of Florence, Italy

B226 Sodium-dependent multivitamin transporter defect, a new case characterized by an unusual cardiac presentation
Dominique Roland, Institute of Pathology and Genetics, Belgium

NEWBORN SCREENING

B227 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

B228 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

B229 Expanded metabolic newborn screening in catalonia: 10 years of experience
Jose Manuel Gonzalez De Aledo Castillo, Hospital Clinic De Barcelona, Spain

B230 Predicting phenotypes in metachromatic leukodystrophy: implications for newborn screening
Samuel Groeschel, University Children's Hospital of Tübingen, Germany

B231 A New biochemical marker as a game changer in Hyperphenylalaninemia Newborn Screening
Nitsan Haham, Sheba Tel Hashomer Hospital, Israel

B232 22-year Follow-up of Extended Newborn Screening for Metabolic and Endocrine Disorders
Sook Za Kim, Korea Genetics Research Center, South Korea

B233 Development of a Novel Method for Glycosaminoglycan Analysis as a Second-Tier Test in Newborn Screening for Mucopolysaccharidosis
Hironori Kobayashi, Shimane University Hospital, Japan

B234 Retrospective Audit of Inherited Metabolic Disease Diagnoses: Implications for Newborn Screening in South Africa
Sarah Lampert, University of Cape Town, South Africa

B235 A case of 3-methylglutaconic aciduria type I detected by expanded neonatal screening in Ukraine
Nataliia Olkhovych, National Children Hospital Ohmatdyt, Ukraine

B236 Uniform gene list for genomic newborn screening: the time is now
Flavia Piazzon, University of Liege, Belgium

B237 Diagnosis of inborn errors of metabolism within the expanded newborn screening in the Balearic Islands (Spain)
Montse Pons, Hospital Universitari Son Espases, Spain

B238 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

B239 Inherited Metabolic Disorders and Primary Immunodeficiencies in Indigenous Populations of Southern Brazil
Leonardo Simao Medeiros, Hospital De Clinicas De Porto Alegre (HCPA), Brazil

B240 Elevated orotic acid with normal citrulline on NBS – consider hyperornithinemia-hyperammonemia-homocitrullinuria (HHH)
Galit Tal, Ruth Rappaport Children's Hospital, Rambam Medical Center, Israel

B241 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

B242 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

B243 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

B244 Newborn screening for Fabry disease in Japan: 16 years of experience
Takaaki Sawada, Kumamoto University, Japan

NOVEL DIAGNOSTIC/LABORATORY METHODS INCLUDING OMICS

B245 Pterin profiling in serum, dry blood spot and urine using LC-MS/MS in patients with hyperphenylalaninemia
Harun Bayrak, Gazi University, Turkiye

B246 Multi-OMICs approach to improving diagnosis in metabolic disorders
Peter Bauer, Universitätsmedizin Rostock, Germany

B247 Genetic variants in the 5` untranslated regions cause hereditary diseases
Alexandra Filatova, Research Centre for Medical Genetics, Russia

B248 The diagnostic yield of critical sample and elective fasting test in children after hypoglycemic event
Ran Hazan, Soroka Medical Center, Israel

B249 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

B250 Modelling inborn errors of redox metabolism using patient-derived lymphoblastoid cell lines
Julien H. Park, University of Münster, Germany

B251 Comprehensive diagnostic approach for patients with inborn phosphate metabolism damage
Margarita Sharova, Research Centre for Medical Genetics, Russia

B252 Characterizing the Metabolomic Profile of Aicardi Goutières syndrome
Brian Shayota, University of Utah, USA

B253 Differential diagnosis of inherited metabolic disorders according to organ system involvement: a lesson from the knowledgebase
Nenad Blau, University Children's Hospital, Switzerland

B254 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

B255 Predicting correct IMD diagnosis using HPO phenotype association algorithms
Judith Jans, University Medical Center Utrecht, The Netherlands

B256 AI-Powered Genomic Analysis: A New Frontier in Diagnosing Rare Diseases
Jaime Lopes, Cincinnati Children's Hospital, USA

B257 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

B258 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

B259 Outlining the care situation in Canavan patients
Jama Wahid, University Medical Center Hamburg-Eppendorf, Germany

B260 Allow Natural Death (AND) in Inborn Errors of Metabolism
Carolina Fraga, Centro Materno-Infantil Do Norte, Centro Hospitalar Universitario Santo Antonio, Portugal

B261 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

B262 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

B263 Citraconate isomers in methylmalonic acidemia revisited: possible role in pathophysiology and as biomarkers
Anibh Das, Hannover Medical School, Germany

B264 Investigating gene essentiality and gene:gene interactions in MMUT deficiency
Sean Froese, University of Zurich, Switzerland

B265 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

B266 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

B267 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

B268 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

B269 Clinical, biochemical, genetic and neuroradiological data in a single cohort of DBP deficiency patients
Unai Diaz-Moreno Elorz, Great Ormond Street Hospital, UK

B270 A girl with strawberry milk-like blood – acute stabilisation in familial chylomicronemia syndrome
Oliver Heath, Royal Children's Hospital, Australia

B271 Phosphatidylserine flippase deficiency diagnosed by whole exome sequencing – a case report
Ariana Mendes, Centro Hospitalar e Universitário de Coimbra, Portugal

B272 Untargeted metabolomics profiling in three cohorts indicates lipid imbalance in individuals with epilepsy
Kaisa Teele Oja, University of Tartu, Estonia

B273 Cholic acid increases serum cholesterol in Smith-Lemli-Opitz syndrome: a pilot study
William Rizzo, University of Nebraska Medical Center, USA

B274 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

B275 Characterization of Alagille Syndrome in Patients with Cholestatic Liver Disease: Clinical Features and Genetic Analysis
Natalia Semenova, Research Centre for Medical Genetics, Russia

B276 Successful treatment of child with congenital bile acid synthesis defect type 3 with oral chenodeoxycholic acid
Natalia Semenova, Research Centre for Medical Genetics, Russia

B277 Cell imaging-based screening for small molecules that rescue peroxisome function in mild disorders of the Zellweger spectrum
Beatriz Silva, University of Luxembourg, Luxembourg

B278 Primary dyslipidemias in Russian population: novel genetic variants
Peter Vasiluev, Research Centre for Medical Genetics, Russia

B279 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

B280 Phytanic acid impairs mitochondrial respiration in rat heart and decreases cardiomyocyte viability
Moacir Wajner, Universidade Federal Do Rio Grande Do Sul, Brazil

PHENYLKETONURIA

B281 The effect of Large Neutral Amino Acids on protein- and diet intake for PKU patients
Kirsten Ahring, Copenhagen University Hospital, Denmark

B282 PHEFREE (Phenylalanine Families and Researchers Exploring Evidence): The rare disease research consortium for hyperphenylalaninemia
Georgianne Arnold, University of Pittsburgh, USA

B283 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

B284 Phase 3 APHENITY long-term study: Sepiapterin for treatment of phenylketonuria
Drago Bratkovic, Royal Adelaide Hospital, Australia

B285 Genetic profile of phenylketonuria in Brazil
Ida Vanessa Doederlein Schwartz, Hospital de Clínicas de Porto Alegre, Brazil

B286 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

B287 Preclinical Evaluation of a Prolonged-Release Protein Substitute on Blood Phenylalanine Levels when Combined with Dietary Protein
Luciana Giardino, Bologna University, Italy

B288 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

B289 Oxidant status of 26 phenylketonuria patients in follow-up: Determination of superoxide dismutase-2 activity and Malondialdehyde estimation
Verónica Cornejo, University of Chile, Chile

B290 Cardiovascular risk and cardiac disease in adult patients with phenylketonuria: a review
Francois Maillot, University Hospital of Tours, France

B291 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

B292 Evaluating trends in self-rated historic metabolic control and treatment history among PRISM participants
Markey McNutt, University of Texas Southwestern Medical Center, USA

B293 Diet compliance and body mass index of Slovenian adult patients with phenylketonuria
Urh Groselj, University Children's Hospital, UMC Ljubljana, Slovenia

B294 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

B295 Trimethylamine increases intestinal fatty acid absorption
Júlio César Rocha, Universidade NOVA De Lisboa, Portugal

B296 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

B297 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

B298 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

B299 Menstrual cycle characteristics, premenstrual syndrome and blood phenylalanine level relationship in women with PKU
Arzu Selamioglu, Istanbul University, Turkiye

B300 Total Choline Intake and Working Memory Performance in Adults with Phenylketonuria
Meriah Schoen, Emory University, USA

B301 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

B302 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

B303 The role of mitogen-activated protein kinase (MAPK) in Phenylketonuria: new markers of oxidative stress
Alberto Burlina, University Hospital of Padua, Italy

UREA CYCLE DISORDERS

B304 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

B305 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

B306 Metabolic parameters and development in patients with argininosuccinic acid synthase 1 and lyase deficiency
Nicola Longo, University of Utah, USA

B307 Evaluation of oxidative damage to biomolecules in patients with ornithine transcarbamilase deficiency
Moacir Wajner, Universidade Federal Do Rio Grande do Sul, Brazil

B308 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

B309 Towards an Algorithm-Based Tailored Treatment of Acute Neonatal Hyperammonemia
Patrick Verloo, University Hospital Ghent, Belgium

B310 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

B311 A Variant in the Allosteric Domain of CPS1 Protein Associated With Effective N-carbamoyl Glutamate Therapy in CPS1 Deficiency
Alberto Burlina, University Hospital of Padua, Italy

E-Posters

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

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 and **Hen Hadad**, 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

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
Khanh Nguyen Ngoc , Vietnam National Children's Hospital, Vietnam

Homozygous PGAP2 mutation cause hyperphosphatasia with mental retardation syndrome-3 (HPMRS3): 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
Montserrat Pons, 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 GA: 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.304GA (p.Ala102Thr) variant of uncertain significance (VUS) as a 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

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
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 the 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-de–rived 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

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

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

MITOCHONDRIAL DISORDERS

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

A novel mitochondrial phenotype in a patient with SYNGAP1 encephalopathyy
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.3291TC in the MT-TL1 gene
Kairit Joost, East-Tallinn Central Hospital, Estonia

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

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

A 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

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

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

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

INVITED SPEAKERS

INVITED SPEAKERS

Annemieke Aartsma-Rus
Medical Center of Leiden
Netherlands

Laura Adang
Children's Hospital of Philadelphia
USA

Johannes Berger
Center of Brain research,
Medical University of Vienna
Austria

Sarah H. Elsea
Baylor College of Medicine
USA

Angeles Garcia Carzola
Sant Joan de Déu Hospital of Barcelona
Spain

Ozlem Goker-Alpan
Lysosomal & Rare Disorders
Research & Treatment Center, Inc
Virgnia, USA

Johannes Häberle
University Children's Hospital, Zurich
Switzerland

Arcangela Iuso
Helmholtz Zentrum of München
Germany

Elad Jacoby
Sheba Medical Center
Israel

Anil B. Jalan
Nirman Metabolic Clinic of Mombai
India

Viktor Kožich
Charles University-First Faculty of Medicine
Czech Republic

Robin Lachmann
University College London Hospitals
UK

INVITED SPEAKERS

Carole Linster
University of Luxembourg, Centre for Systems Biomedicine
Luxembourg

Fanny Mochel
Sorbonne University of Paris
France

Andrew Morris
Willink Metabolic Unit
UK

Jean-Marc Nuoffer
University of Bern
Switzerland

Gideon Rechavi
Sheba Medical Center
Israel

Rachel Rock
Sheba Medical Center, Ramat Gan
Israel

Francis Rossignol
National Human Genome Research Institute
USA

Eran Segal
Weizmann Institute of Science
Israel

Rani H. Singh
Emory University of Atlanta
USA

Orna Staretz-Chacham
Ben Gurion University,
Soroka Medical Center
Israel

Simon Waddington
University College London , Institute for Women's Health
UK

Douglas C. Wallace
Perelman School of Medicine,
University of Pennsylvania
USA

SPECIALS

Meet Shalva

Shalva, the Israel Association for the Care and Inclusion of Persons with Disabilities, is a transformative force in providing comprehensive care for individuals with disabilities and empowering their families while fostering social inclusion.

Their cutting-edge services encompass all stages of life, offering innovative therapies, inclusive education, recreational activities, vocational training, and more, making a lasting impact on the lives of thousands. With advanced programs and facilities, Shalva sets new standards in disability rehabilitation and research, positively influencing the broader community and advocation for equal access and opportunity for all, irrespective of background or financial means.

The Shalva Band

The Shalva Band, an extraordinary ensemble of eight talented musicians with disabilities, captivates audiences worldwide with their exceptional musicality and infectious charm.

As one of Shalva's renowned inclusion programs, they perform at prestigious venues, cultural events, and community gatherings across the globe. Their performance at the SSIEM Opening Ceremony on August 29th at 14:00 promises to be a journey of unity, showcasing the power of music to connect people of all abilities and cultures. This concert celebrates the beauty of inclusion, inspiring us to embrace challenges, recognize human potential, and unite in a world where differences are celebrated, and all voices are heard.

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NETWORKING PROGRAM

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

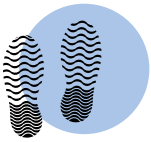
Wednesday, 30 August from 18:35-20:35

ICC Poster Area (Exhibitions 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.

Departure point: ICC main entrance, Return to the ICC

Networking Evening

Thursday, 31 August from 20:30-22:30

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!

Pick-up and return from the Symposium hotel collection points. See route details on the symposium website and APP.

GENERAL INFORMATION

SYMPOSIUM VENUE

The symposium 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

SYMPOSIUM WEBSITE

www.ssiem2023.org

SYMPOSIUM PRESIDENT

Professor Yair Anikster
Sheba Medical Center
Ramat Gan, Israel

SYMPOSIUM SECRETARIAT

ORTRA LTD. (Professional Conference Organizer)
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EXHIBITION MANAGEMENT AND SPONSORING

Society for the Study of Inborn Errors of Metabolism (SSIEM)
Ralph Kerschbaumer – Corporate Liaison Officer
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Phone +43 512 890438
www.ssiem.org

SYMPOSIUM REGISTRATION DESK

Monday, 28 August | 13:00-21:00
Tuesday, 29 August | 07:30-20:30
Wednesday, 30 August | 07:00-20:00
Thursday, 31 August | 07:00-15:00
Friday, 1 September | 07:45-12:45

PRESENTATION UPLOAD FOR SPEAKERS

Speakers' Preview Room is located in the upper level (ICC) (follow the signs)
Tuesday, 29 August | 08:00-19:30
Wednesday, 30 August | 07:00-18:00
Thursday, 31 August | 07:30-15:30
Friday, 1 September | 07:30-12:00

EXHIBITION OPENING HOURS

Tuesday, 29 August | 10:00 - 19:30
Wednesday, 30 August | 08:30-17:30
Thursday, 31 August | 08:30-15:30

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CME CERTIFICATION

"The SSIEM Annual Symposium 2023, Jerusalem, Israel, 29/08/2023-01/09/2023 has been accredited by the European Accreditation Council for Continuing Medical Education (EACCME®) with 17 European CME credits (ECMEC®s). Each medical specialist should claim only those hours of credit that he/she actually spent in the educational activity."

"Through an agreement between the Union Européenne des Médecins Spécialistes and the American Medical Association, physicians may convert EACCME® credits to an equivalent number of AMA PRA Category 1 CreditsTM. Information on the process to convert EACCME® credit to AMA credit can be found at www.ama-assn.org/education/earn-credit-participation-international-activities.

"Live educational activities, occurring outside of Canada, recognised by the UEMS-EACCME® for ECMEC®s are deemed to be Accredited Group Learning Activities (Section 1) as defined by the Maintenance of Certification Program of the Royal College of Physicians and Surgeons of Canada."

TOURISM INFORMATION DESK

Tuesday, 29 August | 10:00-18:00
Wednesday, 30 August | 10:00-18:00
Thursday, 31 August | 09:00-15:00

Pre and Post-Conference tours in Israel will be arranged for interested participants. If you wish to enjoy the beautiful sights of Israel and for more information, please contact Ortra's representative at the Information & Tours desk during operating hours.

REGISTRATION

TO REGISTER FOR THE SSIEM 2023 ANNUAL SYMPOSIUM VISIT THE LINK:

ssiem2023.org/registration-2

We are pleased to offer **discounted registration rates** for **SSIEM members, students** (with student card), **medical residents, nurses** and **technicians** as detailed below.

Registration Category	Late Registration
SSIEM Member	2050 NIS
Non-Member	2985 NIS
SSIEM Member Students/Residents-in-Training (<35 yrs)	1495 NIS
Non-Member Students/Residents-in-Training (<35 yrs)	1830 NIS
SSIEM Member Dietitians/Nurses	1495 NIS
Non-Member Dietitians/Nurses	1830 NIS
SSIEM Members Single Day Ticket	750 NIS
Non-Members Single Day Ticket	1680 NIS

Registration Fees Include:

Full Delegate

(SSIEM Members, SSIEM Member Dietitians/Nurses, SSIEM Member students & residents-in-training, Non-Members, Non-Members Dietitians/Nurses and Non-Member students & residents-in-training):

- Admission to all oral and poster sessions and exhibition area
- Scientific program and Symposium bag
- Access to the online abstract book
- Coffee, tea and 4 lunches during Symposium breaks
- Welcome Reception

Single-day Ticket

Single day tickets do not allow access to social events. No more than two consecutive single day tickets can be purchased by one delegate.

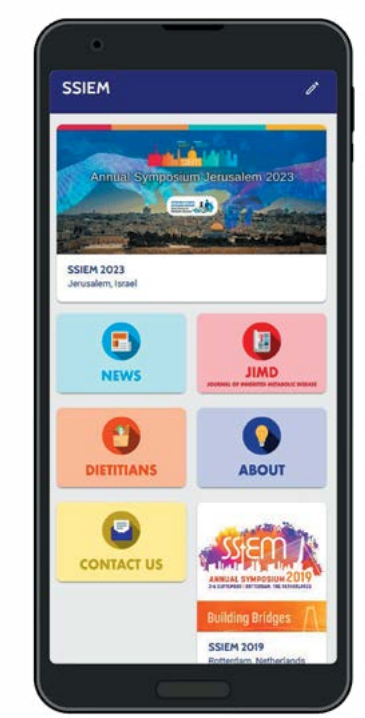
- Admission to all oral and poster sessions and exhibition area
- Scientific program and Symposium bag
- Access to the online abstract book
- Coffee, tea and lunch during Symposium breaks of the day of the ticket

Networking

The Symposium participants are welcome to enjoy the magic of the city with friends and colleagues. For additional information about networking visit the link: **ssiem2023.org/networking-activities**

	Participant	Accompanying Person	Industry
Welcome Reception Tuesday, 29 August	Included	120 NIS	120 NIS
Networking Activities Thursday, 31 August	60 NIS	160 NIS	160 NIS
Networking Evening Thursday, 31 August	125 NIS	315 NIS	315 NIS

APP



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TRANSPORTATION WITHIN JERUSALEM

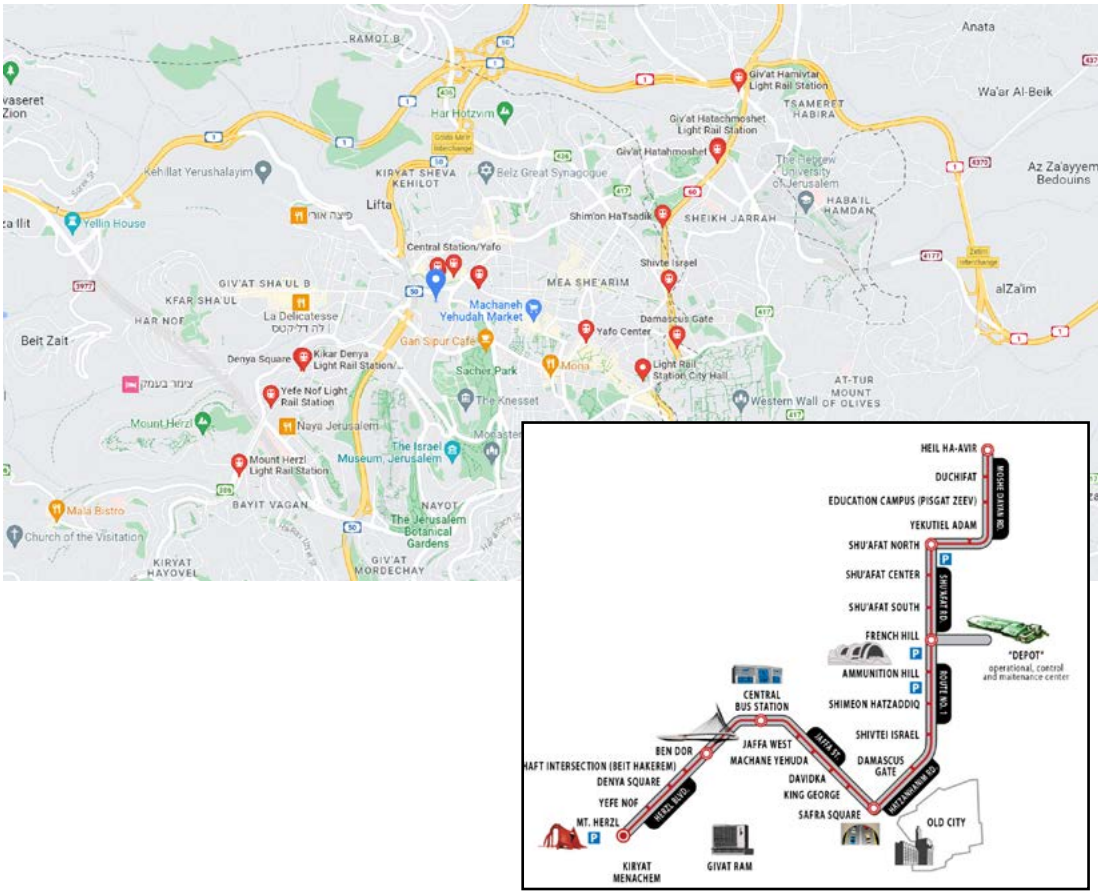
InterCity – The Jerusalem Light Rail (Harakevet Hakala) begins at Mt. Herzl, runs along Herzl Blvd., then along the suspension bridge towards the Central Bus Station, it then continues along the Jaffa Rd. pedestrian mall to Safra Square. From there it travels by the Old City walls, passing closely by Damascus Gate, it then continues through Road No. 1 towards the French Hill Junction, across Shu'afat and terminates in the Pisgat Ze'ev neighborhood.

The closest station to the Sympsoium venue is the Central Station.

The train's arrival time is continuously updated on the electronic boards at the stations. Know the right time for you!

Use the "**plan your journey**" option on the rail's official website to stay updated with the schedule: www.cfir.co.il/en

APP for travelling in Jerusalem on buses and the light rail here: pti.org.il/datasites/lightrail/eng



SHABBAT IN ISRAEL

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.

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.

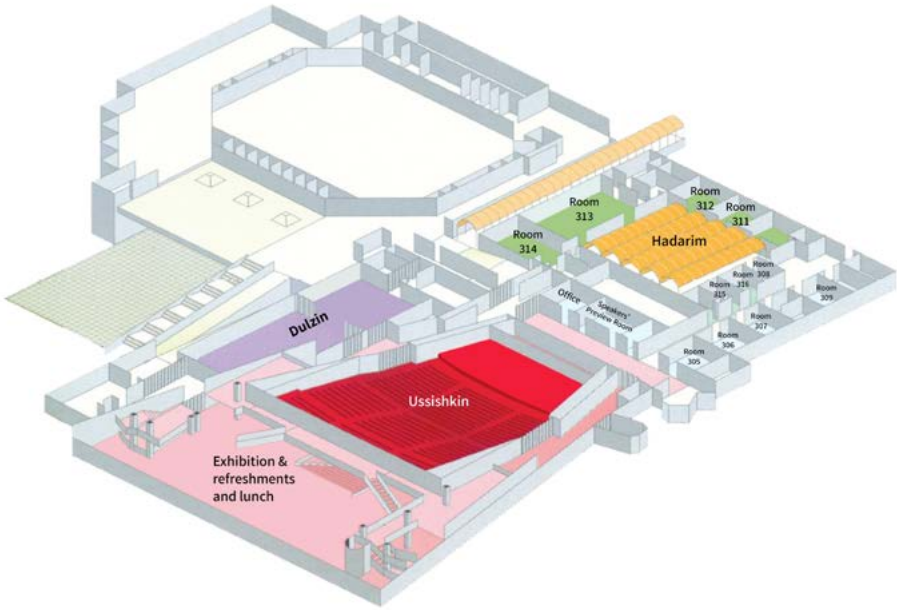
Passport control

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.

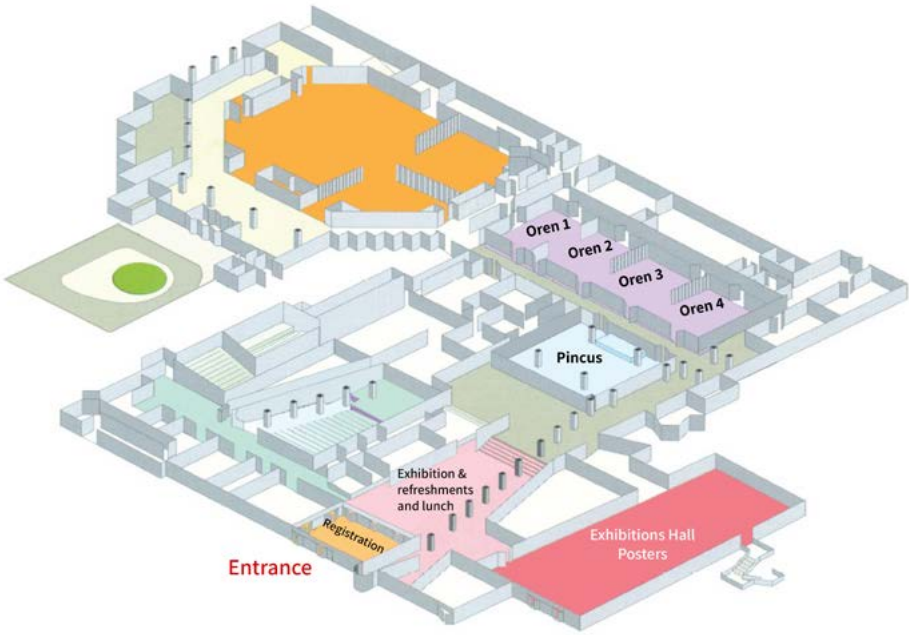
Value Added Tax Reimbursement

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.

ICC Upper Level Map



ICC Lower Level Map



JERUSALEM

Jerusalem, City of Gold

Jerusalem, a city where the past intertwines seamlessly with the present, steeped in history and spirituality, beckons visitors with its captivating allure. Its ancient walls hold stories of civilizations past, and the echoes of its diverse cultural heritage reverberate through its winding streets.

Whether you seek moments of reflection at ancient landmarks or wish to savor the vibrant tapestry of modern life, Jerusalem promises an experience that transcends mere sightseeing. Discover the enchanting blend of tradition and innovation, explore its rich cultural tapestry, indulge in the warmth of its people, and embark on a journey that will leave an indelible mark on your soul. Prepare to be captivated by Jerusalem's timeless charm and profound essence, and to leave with a piece of its magic forever etched in your heart.

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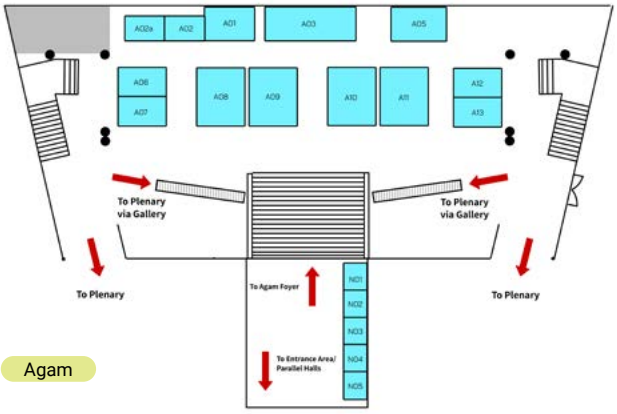
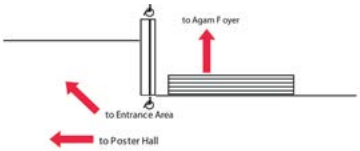
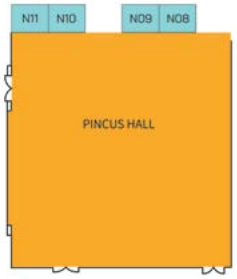
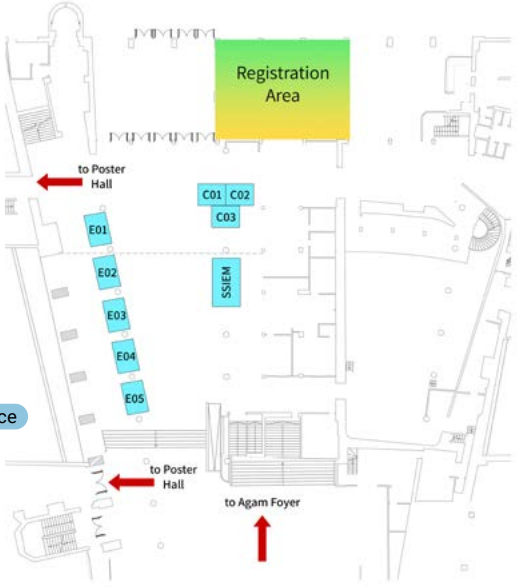


Agam





Nesher



Entrance

Stand-#	Company Name
N01	Ajinomoto Cambrooke
A03	Amicus Therapeutics
E03	Arcturus
E01	Azafaros
N03	Baebies
A10	BioMarin
A08	Chiesi Global Rare Diseases
N04	Denali Therapeutics
N11	Dipharma SA
E05	Egetis Therapeutics AB
A02	GHF - Golden Heart Flower
A12	Immedica
N08	JCR Pharmaceuticals
N02	Lactalis Nutrition Sante
A01	metaX Institut für Diätetik GmbH
N09	Orchard Therapeutics
A02a	Protalix BioTherapeutics
A09	PTC Therapeutics
A13	Recordati Rare Diseases
E04	Revvity
A11	Sanofi
E02	Sentynl Therapeutics, Inc.
A05	Takeda
A07	Travere Therapeutics
N05	TrueMed LTD.
A06	UCB
N10	Ultragenyx

SATELLITE SYMPOSIA & SPONSORING

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

TUESDAY | 29 AUGUST 2023 | 12.45-13.45 | PINCUS HALL

New perspectives in PKU management - The change we can see
Chair: Cary Harding (Portland, OR, United States)

Welcome & introduction
Cary Harding (Portland, OR, United States)

Behind the science: My PALYNZIQ® ▼ experience
Cary Harding (Portland, OR, United States)

Tackling barriers: Journey of a centre with a single specialist to deliver PALYNZIQ®
Johannes Krämer (Ulm, Germany)

From practice to evidence: The real-world impact of PALYNZIQ®
Frank Rutsch (Münster, Germany)

Q&A
Faculty & audience

▼ 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.

Healthcare professionals should report adverse events in accordance with their local requirements. Adverse events should also be reported to BioMarin on + 1 415 506 6179 or drugsafety@bmrn.com.

PALYNZIQ® (pegvaliase) is indicated for the treatment of patients with PKU aged 16 years and older who have inadequate blood phenylalanine control (blood phenylalanine levels greater than 600 μmol/L) despite prior management with available treatment options.

This will contain information on PALYNZIQ® which is not licensed in Israel, but is licensed in the European Union. Refer to your local Prescribing Information prior to using PALYNZIQ®.
This symposium is intended for Healthcare Professionals registered for SSIEM 2023.

This symposium is sponsored and funded by

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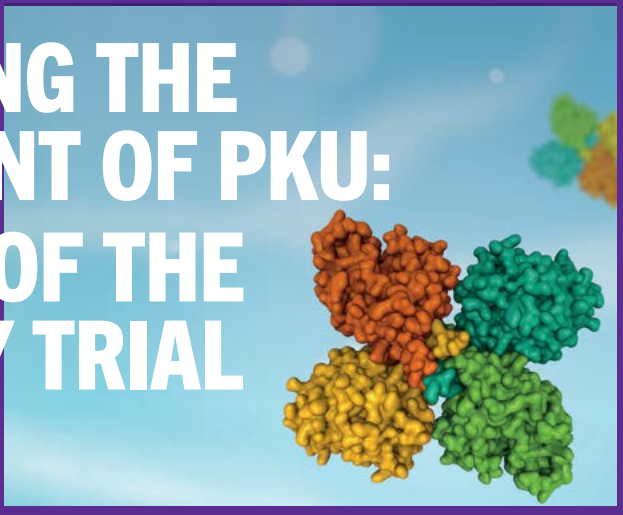
Tuesday, 29 August
12:45–13:45
Dulcin Hall

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

PTC THERAPEUTICS™

AADC Deficiency: Advances in Diagnosis and Treatment

Chair: Bruria Ben-Zeev (Tel Aviv, Israel)

Welcome, introductions and objectives

Bruria Ben-Zeev (Ramat Gan, Israel)

The importance of early recognition in the diagnosis of patients with AADC deficiency

Ángeles García Cazorla (Barcelona, Spain)

Addressing neurometabolic dysregulation through gene therapy: Intraputaminal gene therapy for the treatment of AADC deficiency

Agathe Roubertie (Montpellier, France)

In conversation: Finding and fighting a rare neurometabolic disease

All faculty

Summary and close

Bruria Ben-Zeev (Tel Aviv, Israel)

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Creating A New Roadmap: Advances in Fabry Disease Monitoring and Management

In this symposium, the presenters will identify gaps in current monitoring practices in Fabry disease and provide insights on the use of imaging, integrating testing for biomarkers and antidrug antibodies (ADAs), and patient reported outcome measures in patient care.

The presenters will then examine ways to use the patient experience to guide monitoring and management and address realistic treatment goals that consider patient-reported outcomes.

The presentation will include case studies and conclude with a Q&A *session.*

Presenters

Dominique Germain (Garches, France)
Dawn Laney (Atlanta, GA, United States)
Derralynn Hughes (London, United Kingdom)

Sponsored by

WEDNESDAY | 30 AUGUST 2023 | 07.30-08.30 | DULCIN HALL

Pharmacological optimization of propionic and methylmalonic acidurias long-term management: the latest evidence from the bench to the bed side

Introduction

Sufin Yap (Sheffield, United Kingdom)

The anaplerotic role of Carbaglu: Interplay between urea and Krebs cycles Preclinical and clinical evidence

Francois Feillet (Nancy, France)

Carbaglu mitochondrial protection in PA & MMA and improved ATP production: ex-vivo and long-term clinical outcomes. Is Carbaglu better than liver transplant? A comparative LT prospective trial

Shirou Matsumoto (Kumamoto, Japan)

The very long-term European PA & MMA PROTECT prospective trial, the interim analysis update and paradigmatic cases presentation

Sufin Yap (Sheffield, United Kingdom)

Open Discussion & Q&A - *All*

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RECORDATI RARE DISEASES
Focused on the Few

SSIEM 2023 Annual Symposium

Neurocognitive aspects of MPS II

from early diagnosis to developmental assessment

Wednesday 30 August 2023, 17:30–18:30 IDT

Chaired by Prof. Barbara K. Burton, this highly informative symposium on Mucopolysaccharidosis (MPS) II, a rare inborn error of metabolism, will cover a range of topics. During three presentations our expert faculty will provide insights from their professional experience on the burden of MPS II for patients and caregivers; diagnosis and natural history of cognitive development in children with MPS II; and psychological and neuropsychological testing and assessment.

A Takeda-sponsored satellite symposium at the Society for the Study of Inborn Errors of Metabolism (SSIEM) 2023 Annual Symposium.

Learning objectives

After completing this activity, attendees will be able to:

- Appreciate the burden of disease of neuronopathic Mucopolysaccharidosis II in patients, and their family and caregivers
- Describe the natural history of cognitive development in patients with Mucopolysaccharidosis II
- Understand practical considerations when performing routine cognitive testing in children with neuronopathic Mucopolysaccharidosis II

Prof. Barbara K. Burton
Northwestern University Feinberg School of Medicine, Chicago, IL, USA

Natural history of cognitive development in children with Mucopolysaccharidosis II

Prof. Ronen Spiegel
Emek Medical Center, Afula, Israel

Burden of disease of neuronopathic Mucopolysaccharidosis II and the effects on family and caregivers

Dr Hannerieke van den Hout
Erasmus Medical Center, Rotterdam, Netherlands

(Neuro)psychological testing and assessment in children with Mucopolysaccharidosis II

This meeting is intended for healthcare professionals registered for the SSIEM congress only and is initiated, organized, and funded by Takeda.

VV-MEDMAT-89513 Date of preparation: July 2023

Neurocognitive aspects of Mucopolysaccharidosis II: from early diagnosis to developmental assessment

Chair: Barbara K. Burton (Chicago, IL, United States)

Welcome and introduction

Barbara K. Burton (Chicago, IL, United States)

Burden of disease of neuronopathic Mucopolysaccharidosis II and the effects on family and caregivers

Ronen Spiegel (Haifa, Israel)

Natural history of cognitive development in children with Mucopolysaccharidosis II

Barbara K. Burton (Chicago, IL, United States)

(Neuro)psychological testing and assessment in children with Mucopolysaccharidosis II

Hannerieke van den Hout (Rotterdam, Netherlands)

Panel discussion and Q&A

All faculty, moderated by Barbara K. Burton

Closing remarks

Barbara K. Burton (Chicago, IL, United States)

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Society for the Study of Inborn Errors of Metabolism (SSIEM) 2023 Satellite Symposium

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

Wednesday, August 30, 2023 | 5:30-6:20pm IDT

ICC | Ground Floor, Oranim Rooms 3 & 4 | Jerusalem, Israel

Welcome and Introduction
Can Ficicioglu, MD, PhD

Overview of Classical Homocystinuria
Jaya Ganesh, MD

COMPOSE Phase 1/2 Clinical Trial: Pegtibatinase Data Update
Can Ficicioglu, MD, PhD

Your Questions Answered: Panel Discussion

This meeting is hosted by Travere Therapeutics, Inc. and is open to all registered delegates at the SSIEM 2023 congress. Food and refreshments will be provided.

TRAVERE™ THERAPEUTICS

WEDNESDAY | 30 AUGUST 2023 | 17.30-18.30 | OREN HALL

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

Welcome and Introduction
Can Ficicioglu (Philadelphia, United States)

Overview of Classical Homocystinuria
Jaya Ganesh (New York, United States)

COMPOSE Phase 1/2 Clinical Trial: Pegtibatinase Data Update
Can Ficicioglu (Philadelphia, United States)

Your Questions Answered: Panel Discussion

Sponsored by

TRAVERE™ THERAPEUTICS

Treating adult MPS IVA patients: Recent findings from the largest global Morquio A Registry Study

Chair: Nathalie Guffon (Lyon, France)

Welcome & introduction
Nathalie Guffon (Lyon, France)

Evolving patient needs and management strategies for adults with Morquio A
Christina Lampe (Giessen, Germany)

Long-term treatment of adults in real-world clinical practice: Data from the Morquio A Registry Study
Nathalie Guffon (Lyon, France)

Starting treatment in adulthood – a Morquio A case report
Christina Lampe (Giessen, Germany)

Q&A
Faculty & audience

This symposium is sponsored and funded by

BIOMARIN®

Advancing the Treatment of PKU: Results of the APHENITY Trial

Welcome and Introduction
Nicola Longo (Salt Lake City, Utah, United States)

An Introduction to Sepiapterin and the APHENITY Trial
Nicola Longo (Salt Lake City, Utah, United States)

The APHENITY Results and Implications for Clinical Practice
Ida Schwartz (Porto Alegre, Brazil)

APHENITY Open-label Extension Study
Anita Mac Donald (Birmingham, United Kingdom)

Conclusion, Closing Remarks, and a Brief Q&A
Nicola Longo (Salt Lake City, Utah, United States)

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We chase the *miracles of science*, with an enduring commitment to *better care for rare*.

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

Addressing the needs of patients with Pompe disease – translating clinical trial data into real-world practice

Chair: Priya Kishnani (Durham, NC, United States)

1. To share the current evidence related to avalglucosidase alfa as an alternative therapy option for patients with Pompe disease
2. To discuss how patients, in real-world settings, are responding to avalglucosidase alfa

Introduction by Chair
Priya Kishnani (Durham, NC, United States)

Efficacy and safety of avalglucosidase alfa in participants with late-onset Pompe disease after 145 weeks' treatment during the COMET trial
Priya Kishnani (Durham, NC, United States)

A cohort summary: switching to avalglucosidase alfa in patients with infantile-onset and late-onset disease
Galit Tal (Haifa, Israel)

Real-world data: switching to avalglucosidase alfa and exploring the impact of home-infusion on the quality of life of patients with late-onset Pompe disease
Derralynn Hughes (London, United Kingdom)

Closing remarks & Q&A
Chair & All speakers

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FLEXIBILITY

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* Focus groups were organised in Italy, Germany and UK, involving PKU healthcare professionals and some of their patients.

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Rethink potential.

Long term metabolic control for UCD patients of all ages[1,2]

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References: 1. Ravicti SPC 10/2022. **2.** Diaz GA et al. Hepatology. 2013;57:2171–2179.

RAVICTI® 1.1 g/ml oral liquid. Active ingredient: Glycerol phenylbutyrate. **Composition:** Each ml of the liquid contains 1.1 g glycerol phenylbutyrate (equivalent to a density of 1.1 g/ml). **Indications:** adjunctive therapy in patients with urea cycle disorders (UCDs) including carbamoyl phosphate synthetase 1 (CPS), ornithine transcarbamylase (OTC) deficiency, argininosuccinate synthetase (ASA), argininosuccinate lyase (ASL), arginase 1 (ARG) and ornithine translocase (hyperammonemia-hyperornithinemia-homocitrullinuria syndrome, HHH), which cannot be treated by dietary protein restriction and/or amino acid substitution alone. RAVICTI® must be used with dietary protein restriction and, in some cases, with nutritional supplements (e.g., essential amino acids, arginine, citrulline, protein-free calorie supplements). **Contraindications:** Hypersensitivity to the active substance, treatment of acute hyperammonemia. **Side effects: Common:** Decreased appetite, increased appetite, aversion to food, dizziness, headache, tremor, flatulence, diarrhea, vomiting, nausea, abdominal pain, dyspepsia, abdominal distension, constipation, oral discomfort, retching, skin odour abnormal, acne, metrorrhagia, fatigue, oedema peripheral, aspartate aminotransferase increased, alanine aminotransferase increased, anion gap increased, lymphocyte count decreased, vitamin D decreased. **Uncommon:** gastrointestinal viral infection, hypothyroidism, hypoalbuminaemia, hypokalaemia, dysgeusia, lethargy, paraesthesia, psychomotor hyperactivity, somnolence, speech disorder, confusional state, depressed mood, ventricular arrhythmia, hot flushes, dysphonia, epistaxis, nasal congestion, oropharyngeal pain, throat irritation, abdominal discomfort, abnormal stool, dry mouth, belching, urgency to defecate, abdominal pain upper, abdominal pain lower, painful stool, steatorrhea, stomatitis, gallbladder pain, alopecia, hyperhidrosis, itchy rash, back pain, joint swelling, muscle spasms, pain in extremity, plantar fasciitis, bladder pain, amenorrhea, menstrual irregularity, hunger, pyrexia, increase in blood potassium, increase in blood triglycerides, abnormal electrocardiogram, increase in low density lipoproteins, increase in prothrombin time, increase in white blood cell count, weight increase, weight decrease. Side effects that occur during long-term treatment with glycerol phenylbutyrate in pediatric and adult patients included upper abdominal pain (3 v. 49 pediatric [6.1%] vs. 1 v. 51 adult patients [2.0%]) and increased anion gap (2 v. 49 pediatric [4.1%] versus 0 v. 51 adult patients [0%]). **Permitted Owner:** Immedica Pharma AB · 113 63 Stockholm · Sweden. **Prescription only.** Email: info@immedica.com Internet: www.immedica.com.

COM-003023. Date of preparation: July 2023

Immedica Pharma AB
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Sweden

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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.

SPECIALIZED CLINICAL NUTRITION
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A DIVISION OF PIAM

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LIFE-TRANSFORMING NUTRITION | METABOLICS

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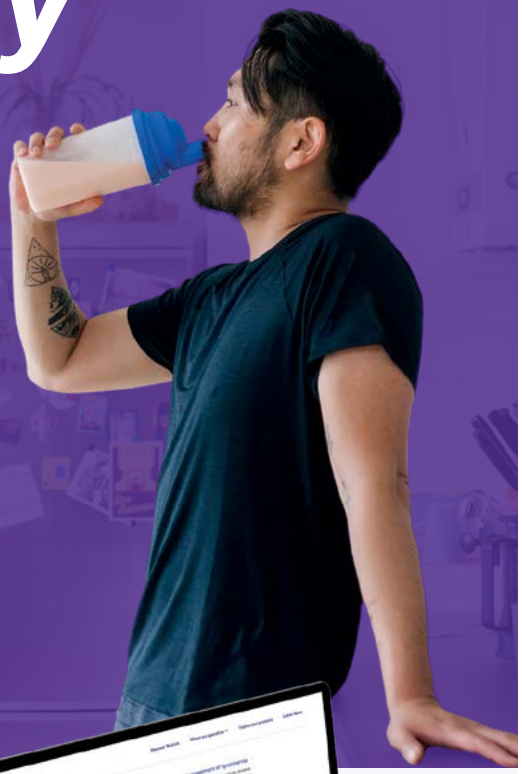

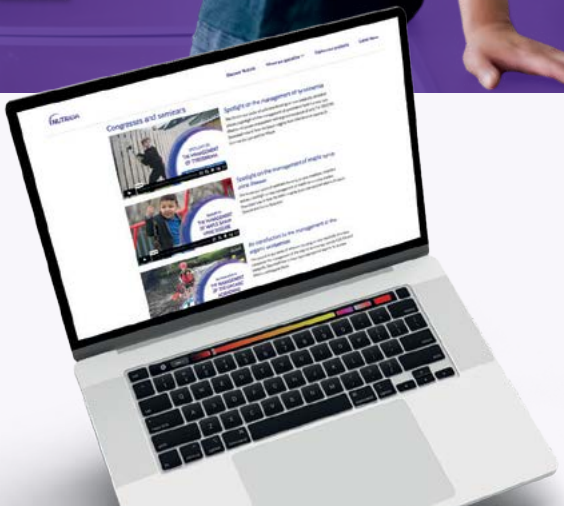

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