

Annual Conference 2025

Friday, 14th November 2025, The Gibson Hotel, Dublin

'New Insights and Personalised Treatments in Inborn Metabolic Disorders'

Hybrid Event - in-person event & live streaming

9.00 Registration & welcome Coffee/Tea

Session 1, Chairpersons: Ina Knerr / Siobhan O'Sullivan

9.30 'Neonatal Dyslipidaemia and Phytosterolaemia', *Kevin McCarthy, Dublin*

10.00 'Clinical Spectrum and Management of Hyperprolinaemia Type 1 & 2 in a Cohort of Irish Children', *Alwyn Charles, Annemarie Hayes, Ritma Boruah, Ahmad A. Monavari, Sabah Abdelfadil, Mohamed Elsammak, Ina Knerr, Dublin*

10.20 'Navigating the Complexities of Personalized Treatments' *Caroline Hart, Siobhan O'Sullivan, Belfast*

10.40 'The Clinical Phenotype of Paediatric Patients with Mucopolysaccharidosis Type VI', *Cherian D. Mathew, Eithne Losty, Ellen Crushell, Ahmad A. Monavari, Mohamed Elsammak, Ina Knerr, Dublin*

11.00-11.30 Coffee/Tea Break – Poster Presentations* – Industry Exhibition

Session 2, Chairpersons: Ahmad A. Monavari / Jenny McNulty

11.30 'A Retrospective Review of Patients with Idiopathic Ketotic Hypoglycaemia', *Fiona Boyle, Christine Merrigan, Emma Crean, Kevin McCarthy, Mohamed Elsammak, Dublin*

11.50 'Late-onset Argininosuccinic Aciduria unmasked by Sodium Valproate', *Robert H Field, Hongying Chen, Aine Redmond, Alex Dudley, Sabah Abdelfadil, Mohamed Elsammak, Eavan McGovern, Stephen Ryan, Loai Shakerdi, James J. O'Byrne, Dublin*

12.10 'Inherited Metabolic Conditions in the Republic of Ireland: Estimation of Prevalence and Carrier Frequency in Adulthood', *Ahsan Rasool, Cliona Carroll, Aya Ibrahim, Mehwish Yunas, Loai Shakerdi, Karl Kavanagh, Ritma Boruah, James J. O'Byrne, Dublin*

12.30 'Exploring Transition to Adult Services for Phenylketonuria Patients in Ireland: Experiences and Outcomes' *Yuxin Woon, Jessica Ivory, Alison Sheerin, Barbara Gillman, Aya Ibrahim, James J. O'Byrne Dublin*

13.00-14.00 Lunch – Poster Displays – Industry Exhibition

Session 3, Chairpersons: Ina Knerr / James J. O'Byrne

14.00 'Patient-Specific In Vivo Genome Editing to Treat Rare Metabolic Disorders' (Keynote Lecture)
Kiran Musunuru, MD, PhD; Director of the Penn Cardiovascular Institute's Genetic and Epigenetic Origins of Disease Program; Children's Hospital of Philadelphia, Perelman School of Medicine at the University of Pennsylvania, Philadelphia, PA.
Rebecca Ahrens-Nicklas, MD, PhD; Director, Gene Therapy for Inherited Metabolic Disorders Frontier Program, Children's Hospital of Philadelphia, Perelman School of Medicine at the University of Pennsylvania, Philadelphia, PA.

- 15.00 'CPS1 Deficiency: Case Report and the Lived Experience', *Louise Perris, Christine Merrigan, Maria O'Regan, Katie Lynam (Dublin), Roshni Vara, Nedim Hadzic (London), Emer Fitzpatrick, Ina Knerr, Dublin*
- 15.20 'Liver Transplantation in Children with Inborn Errors of Metabolism: Over 30 Years' Experience in NSW, Australia' *Kaustuv Bhattacharya, Sydney & Dublin*
- 15.50 'Save the Date - 2027 Symposium of the SSIEM/Society for the Study of Inborn Errors of Metabolism', *Ina Knerr, Dublin (on behalf of LOC)*
- 16.00 Conference Close, *Ahmad A. Monavari, Dublin (followed by AGM, for ISIMD Members only)*

***Poster Presentations (onsite Poster Area & Virtual)**

Chairpersons: Sabah Abdelfadil / James O'Byrne

'A Review of Dietary Intake and Physical Activity in patients with Familial Hypercholesterolaemia attending the National Centre for Inherited Metabolic Disorders', *Emma Lennon, Breige McNulty, Fiona Boyle, Dublin*

'Evaluating Patient Outcomes with GLP-1 Agonist Treatment: Experience to date from the Adult National Centre for Inherited Metabolic Disorders', *Orla Torrallardona-Murphy, Hollie Corrigan, James J. O'Byrne, Dublin*

'A Case of mild PTPS Deficiency- An unexpected Finding from Newborn Screening', *Caroline Hart, Siobhan O'Sullivan, Belfast*

'Sodium-Dependent Multivitamin Transporter Deficiency: A Case Series', *Kevin Gaughan, Helen Mundy, London*

'Vitamin B12 Deficiencies identified incidentally by the detection of urinary Methylmalonic Acid', *Kevin Gaughan, Dinusha Pandithan, Berna Yilmaz, Roshni Vara, Hugh LeMonde, Helen Mundy, London*

'Patient Outcome from Laboratory Alerts for elevated Glutamine on Plasma Amino Acids', *Kevin Gaughan, Dinusha Pandithan, Berna Yilmaz, Roshni Vara, Hugh LeMonde, Helen Mundy, London*

'Review of Sapropterin (Kuvan) Trial Preparation and Outcome for Phenylketonuria (PKU) – Impact on Natural Protein Exchanges and Response Rate' *Kevin Gaughan, Berna Yilmaz, Helen Mundy, London*

+ More!

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