



Irish Society for Inherited Metabolic Disorders *Spring Conference*

'Inborn Metabolic Disorders and the Brain'

Friday, 8th April 2022, Dublin:
VIRTUAL (WEBINAR), 14.00-16.30

Sponsor Displays

14.00 – 14.05 Welcome address (Prof. Monavari)

Session 1 (Chair Prof. Monavari)

14.05 – 14.25 Dr L. Healy/M.O'Shea - Glutaric Aciduria Type 1: Long term outcome in 34 Irish patients with events above 6 years of age

14.25 – 14.45 Dr D. O'Reilly - Point of care biochemical testing for inborn errors of metabolism: Experience from a large tertiary neonatal centre

14.45 – 15.05 C. Merrigan/Senior Metabolic Dietitian - The use of a valine-restricted diet in short-chain enoyl-CoA hydratase (ECHS1) deficiency

15.05 – 15.25 Dr J. Brady - OTC deficiency: A laboratory perspective

15.25 – 15.30 *Live Questions and Answers*

15.30-15.40 *Comfort Break & Sponsor Displays*

Session 2 (Chair Prof. Ina Knerr)

15.40 – 15.55 Prof. I Knerr - Reflections on 50 years of Newborn Screening for classical Galactosaemia in Ireland: a success story with challenges

15.55 – 16.30 Prof. Phillip L. Pearl, William G. Lennox Chair, Boston Children's Hospital, Harvard Medical School, Boston, MA - New Paradigms for Metabolic Epilepsies (*Keynote Lecture*)

16.30 *Summary and Close (Prof. Monavari)*

Sponsor Acknowledgement