



Irish Society for Inherited Metabolic Disorders

Developmental Delay in Inherited Metabolic Disorders

Friday 21st April 2017, The Pillar Room, Rotunda Hospital

RCPI/ACBI/ACSLM 5 points. Applicable for CORU & NMBI CPD points

9.30 **Registration & Coffee**

9.45 **Opening Speech:** *Professor Philip Mayne*

Session 1: Investigations for developmental delay

10.00 Diagnostic approaches in infants and children with suspected mitochondrial disorder presenting with developmental delay

Dr Saskia Wortmann-Hagemann, Consultant in Inherited Metabolic Disorders, Children's University Hospital, Salzburg

10.45 Sequence first, ask questions later: genetic diagnostics in the genome age

Dr Shane McKee, Consultant in Clinical Genetics, Belfast HSC Trust

11.30 **Coffee**

Session 2: Lysosomal storage disorders

11.45 Lysosomal storage disorders presenting with developmental delay

Prof. Gregory M. Pastores, Consultant in Inherited Metabolic Disorders, Mater Misericordiae University Hospital Dublin

12.15 When to start and stop enzyme replacement therapy

Dr Uma Ramaswami, Consultant in Inherited Metabolic Disorders, Royal Free London

13.00 **Lunch & Sponsor Displays**

Session 3: Challenges, advances and management of paediatric patients (PKU)

14.00 Clinical outcomes following hospitalisation to stabilise blood phenylalanine level

Ms. Christine Merrigan, Senior Metabolic Dietitian, Temple Street Children's University Hospital, Dublin

14.20 Overweight & obesity in patients with PKU in comparison with the general population.

Ms. Aoife Fitzgerald, Senior Metabolic Dietitian, Temple Street Children's University Hospital, Dublin

14.40 Embracing technology to advance medical communication

Ms. Anne Clark, Metabolic Dietitian Manager, Temple Street Children's University Hospital, Dublin
Ms. Deirdre Deverell, Principal Biochemist, Temple Street Children's University Hospital, Dublin

15.00 **Coffee**

Session 4: Case studies of critically ill neonates (Inherited Metabolic Disorders)

15.15 Three case studies presented by the junior doctors,

Temple Street Children's University Hospital, Dublin

16.00 Aspects of investigating the critically ill child

Dr Ingrid Borovickova, Consultant Chemical Pathologist, Temple Street Children's University Hospital, Dublin

16.15 Close – *Dr. A.A. Monavari, Consultant in Inherited Metabolic Disorders,*

Temple Street Children's University Hospital, Dublin