

Irish Society of Inherited Metabolic Disorders

Friday 12th April 2019, The Pillar Room, Rotunda Hospital

Multi-system presentation of Metabolic Disorders

9.30 Registration & Coffee

9.45 Welcome address - Prof. Philip Mayne

Session 1: Glycosylation Disorders

Chair: Prof. Ina Knerr

10.00 Introduction: diagnostic dilemmas, patient numbers, MDT involvement, international testing

Prof. Monavari, Metabolic Consultant, National Centre for Inherited Metabolic Disorders (NCIMD), Temple Street Children's University Hospital, Dublin

10.15 The diagnosis and management of complex disorders involving Glycosylation of proteins and lipids

Prof. Dirk Lefeber, Radboud University Medical Centre, Nijmegen, The Netherlands

11.00

Coffee

Session 2: Looking at Neurometabolic Disorders

Chair: Dr. Siobhán O'Sullivan

11.15 Diagnosis of IEM through an Irish Neurology Clinic

11.45 *Dr. Bryan Lynch, Consultant Paediatric Neurologist, Temple Street Children's University Hospital, Dublin*

12.15 Radiological abnormalities in neuro-metabolic patients

Dr. Stephanie Ryan, Consultant Paediatric Radiologist, Temple Street Children's University Hospital, Dublin

12.45

Metabolic disorders through the eyes

Prof. Flitcroft, Consultant Ophthalmologist, Temple Street Children's University Hospital, Dublin

1.30

Lunch

Session 3: Disorders affecting the Liver

Chair: Prof. Philip Mayne

2.15

What can we learn from Bile Acid analyses?

Prof. Peter Clayton, Genetics & Genomic Medicine Programme, Institute of Child Health, Great Ormond Street, London, UK

2.45

Infantile liver failure syndromes

Prof. Ellen Crushell, Metabolic Consultant, National Centre for Inherited Metabolic Disorders (NCIMD), Temple Street Children's University Hospital, Dublin

3.00

Coffee

4.15

Session 4: Case Studies

Chair: Dr. Joanne Hughes

Case Studies

Close - Prof. A.A. Monavari