



Irish Society for Inherited Metabolic Disorders

Celebrating 50 years of Newborn Bloodspot Screening in Ireland

Friday 15th April 2016
Pillar Room, Rotunda Hospital

Programme

09.00 Registration and refreshments

Session 1 Chair: Prof Philip Mayne

10.00 Introduction: 50 years of Newborn Bloodspot Screening in Ireland
(Prof Philip Mayne)

10.30 Ancient Genomes and Irish prehistory
(Prof Dan Bradley, Smurfit Institute of Genetics, Trinity College Dublin)

11.00 Effects of population migration on PKU and Classical Galactosaemia
(Dr Orna Tighe, Molecular and Cellular Therapeutics, RCSI)

11.30 Harmonisation of newborn bloodspot screening across Europe and its impact on the Irish screening programme
(Dr Gerard Loeber, Past President, International Society of Neonatal Screening; Bilthoven, The Netherlands)

12.15 Should screening for Lysosomal Storage Disorders be included in newborn bloodspot screening programmes
(Prof Greg Pastores, MMUH and TSCUH, Dublin)

12.45-13.45 Lunch

Session 2 Chair: Dr Ardeshir Monavari

13.45 Outcome of newborn bloodspot screening for metabolic disorders in Ireland
Introduction (Dr Ardeshir Monavari)
Outcome for Homocystinuria (Dr Siobhan O'Sullivan)
Outcome for MSUD (Prof Ina Knerr)

14.45 Outcome of pregnancy in women diagnosed with an IMD through newborn screening
(Dr Joanne Hughes)

15.15 Living with an inborn error of metabolism detected by newborn screening- parents' perspectives on child development and impact on family life
(Dr Peter Burgard – Child Psychologist, University Children's Hospital, Heidelberg)

16.00 Conclusion
(President of the ISIMD Dr Ardeshir Monavari)