Programme overview

9.30-10.00 Registration, tea & coffee

Morning chair
Professor John Newton, Chief Knowledge Officer, Public Health England

10.00-10.05 Introduction by Chair

10.05-10.25 30 years of the BPSU: Past, present and the future
Dr Richard Reading, Consultant Paediatrician and BPSU Chair, Norfolk and Norwich University Hospitals

Session 1 Improving and speeding-up diagnosis

Early diagnosis of rare disease can frequently be made prior to delivery giving families the opportunity to make choices and prepare for the delivery of their child.

Advances in genomics will facilitate this early rare disease diagnosis prenatally, antenatally and soon after birth.

10.25-10.45 100,000 genome project and its potential for rare disease research
Professor Eamonn Sheridan, Professor of Clinical Genetics, University of Leeds

10.45-11.00 Prenatal/antenatal diagnosis of rare disease
Dr Kelly Cohen, Consultant Obstetrician, Leeds Teaching Hospitals

11.00-11.15 Neonatal screening – progress, controversies and the impact of the BPSU
Dr Rachel Knowles, Consultant Public Health Medicine, UCL Institute of Child Health

11.15-11.30 Panel Q&A

11.30-11.55 Coffee break

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Session 2
Research into practice
The findings of clinical research must be translated into real practice in order to benefit the lives of children and families living with rare disease

11.55-12.10 Rare disease registries
Dr Martin Ward Platt, Consultant Paediatrician, Newcastle upon Tyne Hospitals

12.10-12.25 Sudden and Unexpected Postnatal Collapse
Dr Julie-Clare Becher, Consultant Neonatologist, Royal Infirmary of Edinburgh

12.25-12.45 Severe Combined Immunodeficiency: from 100% mortality to 100% survival in 50 years?
Professor Bobby Gaspar, Professor of Paediatrics and Immunology, Great Ormond Street Hospital

12.45-13.00 Panel Q&A

13.00-14.00 Lunch

Afternoon chair
Professor Tim Barrett, Consultant paediatrician, Program Director for the Wellcome Trust Clinical Research Facility

14.00-14.20 Infectious disease monitoring: predicting what the future holds and it’s impact on clinical practice
Professor Richard Pebody, Consultant in Infectious Disease, Public Health England

14.20-14.40 Transition: how can paediatricians make it work?
Dr Larissa Kerecuk, Consultant Nephrologist, Rare Diseases Lead, Birmingham Children’s Hospital
Dr Graham Lipkin, Consultant Nephrologist, Rare Diseases Centre at the Queen Elizabeth Hospital Birmingham

14.40-14.55 Life limiting care – the effect of changing disease profiles
Dr Heather McCluggage, Associate Specialist, Paediatric Palliative Care, Western Health and Social Care Trust, Northern Ireland

14.55-15.15 Real lives: growing up with rare disease, the impact on the child and family
Dan Lewi, Charity Director, The CATS Foundation

15.15-15.30 Panel Q&A

15.30 Close

15.30-16.30 Rare disease tea party & networking event introduced by Professor Tim Barrett