RSM Joint meeting RCPCH and RSM Paediatric & Child Health Section and in association with the Clinical Genetics Society

Genomics of paediatric disease

Tuesday 21 October 2014

Chairs: Dr Andrew Long, Dr Mick Parker & Dr Louise Fleming

9.30am  Registration, tea and coffee

10.00am  Impact of genomics on paediatric practice –
Professor Jill Clayton-Smith
Consultant Clinical Geneticist, North Western Regional Genetic Service

10.35am  Understanding the jargon – genes, panels, exomes, genomes and arrays
Dr Serena Nik-Zainal, Clinical Research Fellow, Wellcome Trust Sanger Institute, Cambridge

11.10am  Coffee break

11.40am  The deciphering developmental disorders study
Dr Helen V Firth, Consultant Clinical Geneticist, Cambridge University Hospitals Trust

12.15am  Translational genomics – identifying clinically important variants –
Dr Caroline Wright, Programme Associate, PHG Foundation,
Wellcome Trust Sanger Institute, Cambridge

12.50pm  Lunch

1.50pm  Targeting treatment based on accurate genetic diagnosis
Dr Simon Jones, Manchester, Consultant in Paediatric Inherited Metabolic Disease, Manchester Centre for Genomic Medicine

2.25pm  Ethics of genomics in paediatric practice
Professor Michael Parker
Professor of Bioethics and Director of the Ethox Centre, University of Oxford

3.00pm  Tea

3.20pm  Attitudes of young people to receiving data from sequencing technologies
Dr Anna Middleton, Registered Genetic Counsellor and Ethics Researcher, Wellcome Trust Sanger Institute, Cambridge

3.55 pm  Unique - supporting families and professionals with a genomic diagnosis
Dr Beverly Searle, Chief Executive, Unique the Rare Chromosome Disorder Support Group

4.30pm  Concluding remarks

4.40pm  Close of meeting