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