CMGS
CLINICAL MOLECULAR GENETICS SOCIETY

CMGS Spring Scientific Conference 2009

Thursday 26th and Friday 27th March
University of Exeter

FINAL PROGRAM AND ABSTRACT BOOK

Welcome to the CMGS Spring Scientific Conference 2009

This year's conference is being jointly hosted by the Exeter and Bristol laboratories and is being held in the Peter Chalk Centre, Streatham Campus, University of Exeter.

Due to the excellent quality of the abstracts submitted, we have tried to allocate a spoken presentation to as many people as possible and so have been unable to include spoken presentations on posters. The posters are easily accessible near the reception desk and we hope that people will view them when taking their refreshments.

We would like to thank the trade exhibitors that have contributed to the conference this year. The trade exhibits are situated en route to the main lecture room so please make sure that you take the time to visit their stands.

The evening entertainment on Thursday will be held in the restaurant in Holland Hall, which has panoramic views of the Exe estuary.

Thank you for coming to Exeter for this year's conference, particularly those who are contributing. Our special thanks go to our invited speakers, Dr. Maggie Shepherd and Professor Tim Frayling. For those of you who have decided to take the opportunity of staying for the weekend and exploring the area, there is a lot to do! (see http://www.exeterandessentialdevon.com for some ideas).

We hope you enjoy the conference and we look forward to seeing you in Exeter.

2009 CMGS Organising Committee
Exeter and Bristol Molecular Genetics Laboratories

The trainee sessions are dedicated in memory of Danny Routledge. Ann Curtis will speak a few words before the afternoon trainee session on Thursday.

Danny Routledge 1977-2009

Danny was awarded the CMGS Certificate Competence in 2006 after completing his A grade training in the Newcastle lab. For the past 2 years he has bravely fought a battle with cancer but very sadly died, peacefully, at his home on 14th February. Everyone who came into contact with Danny appreciated what a wonderful person he was. Throughout his illness he was determined, optimistic and uncomplaining. He worked in the lab until just 3 weeks before he died. Danny will be greatly missed by all of us in Newcastle, and he is a great loss to our profession. Our thoughts are with his family.

Programme CMGS 2009

Thursday 26 March 2009

10:30 **Coffee and registration**

11:00 **Welcome**

11:05-12:20		**Trainee Presentations Session 1**	**Chair:** Mrs Maggie Williams
11:05	SP1	Could distal *MSH2* upstream deletions cause HNPCC?	**John Taylor**, Jennie Bell, Fiona Macdonald, Philippe Taniere Oxford Medical Genetics Laboratories
11:20	SP2	Epidermal Growth Factor Receptor and K-Ras mutations in patients with Non Small Cell Lung Cancer	**H. Mugalaasi**, J. Davies, L Medley, D Talbot, R. Butler, R. Britto All Wales Molecular Genetics Laboratory, Cardiff
11:35	SP3	Identification and RQ-PCR monitoring of CML patients with rare variant BCR-ABL transcripts	**Christopher Bowles**, Joanne Mason, Susanna Akiki and Mike Griffiths West Midlands Regional Genetics Laboratory, Birmingham
11:50	SP4	Fumarate Hydratase; one gene, two inheritance patterns and two or three diseases	**Oliver Ridgway** West Midlands Regional Genetics Laboratory, Birmingham
12:05	SP5	Validation of *BRCA2* mutation scanning using the Light-Scanner system for high resolution melt analysis	**Lewis Darnell**, Rachael Tredwell and Gareth Cross Nottingham Regional Molecular Genetics Service

12:20-13:15		**Guest Speakers**	**Chair:** Prof Sian Ellard
12:20	SP6	The impact of a genetic diagnosis of neonatal diabetes	**Dr Maggie Shepherd** Peninsula Medical School, Exeter
12:40	SP7	Polygenic traits reach new heights	**Professor Tim Frayling** Peninsula Medical School, Exeter

13:15-14:15 **Lunch, trade exhibition, posters**

14:15-16:00		**Trainee Presentations Session 2** **Tribute to Danny Routledge by Ann Curtis**	**Chair:** Dr Sarah Warburton
14:15	SP8	Development of a mutation screening service for ARPKD	**Wendy Lewis,** David Goudie and David Baty Molecular Genetics, Dundee
14:30	SP9	Development of a molecular genetic diagnostic service for X-linked ichthyosis, with emphasis on carrier detection	**Eleanor Reavey**, Dr A Cooke, Dr G Graham, Su Stenhouse West of Scotland Regional Genetics Service, Glasgow
14:45	SP10	Development of a New Method to Prioritise Gene Analysis in Familial Hypertrophic Cardiomyopathy	**Jayne Duncan**, V Murday, S Stenhouse, D Ellis West of Scotland Regional Molecular Genetics Service, Glasgow
15:00	SP11	Sequencing of the Hypertrophic Cardiomyopathy genes using an automated high throughput strategy	**Aisha Ansari**, Stuart MacKay, Paul Westwood, Judith Pagan, Austin Diamond, Nicola Williams & Jon Warner South East Scotland Clinical Genetic Services, Edinburgh
15:15	SP12	Development and validation of a diagnostic service for epimutations that cause Pseudohypoparathyroidism type 1b	**Jennifer Greatwood**, Deborah Mackay, David Robinson Wessex Regional Genetics Laboratory
15:30	SP13	Affymetrix Resequencing Arrays	**Matthew Smith**, Paul Gissen, Chris Bruce, Fatima Rahman, Fiona Macdonald West Midlands Regional Genetics Laboratory, Birmingham
15:45	SP14	Early experiences in amplicon sequencing using the Roche GS-FLX massively parallel DNA sequencer and its application within a diagnostic laboratory	**Louise Stanley**, Jonathan Coxhead, John Burn and Ann Curtis Northern Genetics Service, Newcastle

16:00-16:30 **Tea**

16:30-17:15		Service reviews and interesting case reports	**Chair:** Dr Anneke Seller
16:30	SP15	An assessment of the clinical sensitivity of molecular genetic testing in Hypertrophic Cardiomyopathy (HCM).	**Kate Thomson**, J. Thistleton, M. Wilson, S. Adams, K. McGuire, P. Clouston, E. Blair, A. Seller Churchill Hospital, Oxford
16:45	SP16	Mutation Screening in patient affected with autosomal dominant hypercholesterolemia – results from the Department of Health (DH) Pilot Project	**Alison Taylor**, K Patel, D Wang, J Tsedeke, B Martin, D Neely, D Nair, M Barbir, S Egan, Y Lolin, R Whittall, G Hadfield, S Humphries, G Norbury Great Ormond Street Hospital
17:00	SP17	Suspected confined blood chimerism in monochorionic dizygotic (MCDZ) twins	**David Gokhale**, Julie Sibbring, Frances White, Carly Broadhurst, Aram Buchanan, Dan Hawcutt, Nim Subhedar, Leanne Bricker Liverpool Women's Hospital

17:15-17:45 **AGM**

19:00 **Conference Dinner**

Friday 27 March 2009

9:00-10:15		Trainee **Presentations Session 3**	**Chair: Dr** David Bourn
9:00	SP18	Developing a diagnostic service for Stargardt disease – a feasibility study	**Emily Packham**, Treena Cranston, Susan Downes, Andrea Nemeth and Anneke Seller Oxford Medical Genetics Laboratories
9:15	SP19	Developing a molecular genetic diagnostic service for arrhythmogenic right ventricular dysplasia/cardiomyopathy (ARVD/C) in Scotland	**Silvia Borras**, C. Clark, D. Walker, K.Kelly and J.Dean Medical Genetics, Aberdeen
9:30	SP20	Non-invasive diagnosis of fetal sex using free fetal DNA: our experiences so far	**Rebecca Woodward**, Joanne Dunlop, Stephanie Allen and Fiona MacDonald West Midlands Regional Genetics Laboratory, Birmingham
9:45	SP21	A false negative result for Myotonic Dystrophy type 2 using quadruplet primed PCR	**Nick Parkin**, Oliver Wood, David Robinson, John Harvey Wessex Regional Genetics Laboratory
10:00	SP22	Development of Methylation Specific High Resolution Melt analysis for detection of 11p15 methylation abnormalities and comparison to MS-MLPA.	**Catherine Willoughby**, Elizabeth Ormshaw, Alice Johnson-Marshall, Linda Baskcomb, Katrina Tatton Brown and Rohan Taylor SW Thames Molecular Genetics Diagnostic Laboratory, St George's Hospital, London
10:15-10:45		**Coffee**	
10:45		**Trainee prize**	
10:50-11:35		**Haematological Malignancies**	**Chair: Dr** Joanna Farrugia
10:50	SP23	The measurement of minimal residual disease for UKALL2003	**Jeremy Hancock**, on behalf of the UKMRD Laboratory Network Bristol Genetics Laboratory
11:05	SP24	Acute Promyelocytic Leukemia with t(15;17) (q22;q21) with inv16 (p13q22) secondary AML	**Susanna Akiki**, Jane Bryon, Joanne Mason, Charles Craddock, Mike Griffiths Birmingham Women's NHS Foundation Trust
11:20	SP25	Acquired isodisomy (uniparental disomy) of chromosome 21 in an acute myeloid leukaemia (AML) patient as an incidental finding during routine chimaerism analysis, and the introduction of a new RUNX1 screening service	**Joanne Mason** and Mike Griffiths Birmingham Women's NHS Foundation Trust
11:35-12:35		**Molecular Pathology**	**Chair:** Dr Ian Frayling
11:35	SP26	Rapid KRAS mutation screening by pyrosequencing	**Philip Chambers**, Susan Richman, Chris Booth, Sophie Grant, Graham Taylor and Philip Quirke St James Hospital, Leeds
11:50	SP27	The Bristol Experience of Molecular Genetic Analysis of Gliomas (LOH and MGMT) for Optimisation of Treatment	**Hilary Sawyer**, Sarah Burton-Jones, Mark Greenslade, Paula Waits, Kirsten Hopkins, Hugh Newman, Seth Love, Maggie Williams Bristol Genetics Laboratory
12:05	SP28	Detection of BRAF Mutations in Tumour and Serum of Patients with Advanced Melanoma.	**Ruth E Board**, Gillian Ellison, Maria C M Orr, Karin R Kemsley, Gael McWalter, Laura Y Blockley, Simon P Dearden, Caroline Dive, Clive Morris, Mireille V Cantarini AstraZeneca Pharmaceuticals
12:20	SP29	A survey of current pharmacogenetic testing in the UK and Ireland	**Simon Ramsden**, Nikki Gambhir, Jennifer Higgs, Kay Poulton, William Newman St Mary's Hospital, Manchester
12:35-14:00		**Lunch, trade exhibition, posters**	
13:15-14:00		*Next Generation Sequencing Group meeting*	
14:00-14:30		UK NEQAS for Molecular Genetics participants meeting	Chair: Dr David Robinson
14:30	SP30	Initial experiences of the UK NEQAS Immunocytochemistry scheme for DNA mismatch repair proteins	**Ian Frayling**, Lisa Happerfield, Merdol Ibrahim, Keith Miller and Mark J Arends University Hospital of Wales, Cardiff

14:45-15:45		Into the Future	**Chair:** Mrs Rachel Butler
14:45	SP31	TP-PCR revisited	**Jon Warner**, Aisha Ansari, Tara Azam Western General Hospital, Edinburgh
15:00	SP32	From kit car to Toyota production line; process redesign in the Exeter laboratory	**Sian Ellard**, Carolyn Tysoe, Martina Owens, Melissa Sloman, Kevin Colclough, Andrew Parrish, Neil Goodman, Karen Stals, Katie Guegan, Ann-Marie Patch and Beverley Shields Genetics Laboratory, Royal Devon & Exeter Hospital
15:15	SP33	STARLIMS goes live in 2009	**Stuart Bayliss**, Richard Renshaw, Gary Dawson, Steve Evans, Carolyn Tysoe, Kirsty Manger and Yvonne Wallis St Mary's Hospital, Manchester
15:30	SP34	Illumin8er: Novel Analytical Software for Diagnostic next-generation sequencing using the Illumina GA-II	Ian M. Carr, Philip Chambers, Joanne Morgan, Alexander F. Markham, David T. Bonthron, **Graham R. Taylor** St James Hospital, Leeds
15:45		**Close and cream tea**	