British Human Genetics Conference 2006

Monday-Wednesday, 18-20 September

University of York

PROGRAMME AND REGISTRATION BOOKLET

Closing date for registration: 14th August 2006

There will be plenary sessions of contributed papers and symposia on:

- Dvelopmental and Prenatal Genetics
- Service Developments in Clinical Genetics
- Mechanisms of Genetic Disease
- Complex Genetic Disorders
- Protecting the Public, Protecting the Professional
- Genetics and Therapy
- Structural Chromosome rearrangements – breakpoints, expression and identity by descent
- Genomics and Medicine
- Microarray Implementation Workshop
- Genomic Tools to target diagnosis, prognosis and therapy of common disease
- Genomic Disorders
- *Carter Lecture*
  Professor Patricia Jacobs on "A new look at old Chromosomes"
- *Great Debate:*
  "This house believes that genetic services have a duty to attempt to contact at-risk relatives directly, rather than rely on communication by patients"
- *Researchers Forum*

LATE BREAKING ABSTRACTS

Deadline 1st September 2006

Details on website www.bshg.org.uk

SCIENTIFIC PROGRAMME

The BSHG Scientific Programme Committee welcomes you to the 2006 meeting of the British Society for Human Genetics. The SPC has compiled a wide-ranging programme comprising many diverse symposia and outstanding speakers and, 50 years after the discovery of the correct number of human chromosomes, Professor Patricia Jacobs will deliver the Clinical Genetics Society Carter Lecture on "A new look at old chromosomes".
Whilst the programme format is generally similar to that of previous years (including the well received "Late Breaking Session" and Great Debate), this year, in response to members requests for more symposia options, two additional parallel sessions have been added. Furthermore, a "Researchers Forum" meeting has been scheduled to provide younger members of the Society, who are interested in a research career, with the opportunity to meet informally with more experienced members.
The combination of excellent invited speakers and submitted abstracts should ensure another high quality meeting, but your feedback is most important – please do take the time to provide comments and suggestions for future meetings.

Eamonn Maher
Chairman, Scientific Programme Committee 2006

There is a varied programme of symposia, workshops and submitted spoken and poster presentations.

Posters are in the Exhibition Centre and Goodricke Function Room. Poster presenters will be asked to stand next to their posters:

Odd numbers (i.e. 1.01, 1.03 etc) on TUESDAY 13:30 - 14:15

Even numbers (i.e. 1.02, 1.04 etc) on TUESDAY 14:30 - 15:15

Carter Lecture - Professor Patricia Jacobs on "A new look at old chromosomes"

The Great Debate is being held again on Monday evening (19:00-20:00 in Room PX001, Exhibition Centre). The motion for the debate is "This house believes that genetic services have a duty to attempt to contact at-risk relatives directly, rather than rely on communication by patients"

Debators are Professor Steve Humphries, Dr Ainsley Newson, Professor Graeme Laurie, Mrs Lauren Kerzin-Storrar

Come and join in what promises to be a very lively debate.

Researchers Forum - A "Researchers Forum" meeting is scheduled for Tuesday lunchtime. This is a development of last year's "Meet the Professors" initiative. The purpose of these sessions is to provide trainees and junior faculty the opportunity to meet with senior academics, who can provide insights in their field and career guidance in a small interactive group. Attendance at each session is limited and available on a first-come, first-served basis to maintain an interactive format. In order to reserve a place (and lunch!) please notify Ruth Cole (bshg@bshg.org.uk) if you intend to attend. In addition, please feel free to email suggestions for topics for discussion that you think would be of general interest.

ABSTRACT BOOKLET

The Abstract Booklet is a supplement to the *Journal of Medical Genetics.* Members unable to attend can order Abstract Booklets from the Conference Organiser *before the meeting* price £6.00.

CONFERENCE VENUE

The plenary sessions, symposia, workshops and posters are being held in Central Hall and the Exhibition Centre on the campus of the University of York. The venues overlook the lake and are linked together with a covered walkway.

ACCOMMODATION

Accommodation has been arranged on campus close to Central Hall and the Exhibition Centre. The accommodation is situated in James and Langwith Colleges. Langwith College comprises single study bedrooms with approximately 4 people sharing a bathroom. James College comprises mainly single study bedrooms with en-suite facilities but also has single study bedrooms with shared bathroom facilities. Full details of the location of your accommodation will be given in the conference pack, which will be sent to you at the end of August.

TRAVEL

The University is sited in the village of Heslington to the south-east of York city centre. A map with more detailed travel information will be sent on receipt of the Registration Form.

By Road The University is extremely well signposted from the junction of the A64 and A19.

Car Parking Please note that the University makes a charge for car parking and have introduced Pay and Display machines. Car Parking will only be available in Car Parks—Campus Central, Campus West and Campus South (a map of the University will be included in your conference pack).

We can provide a 3-day parking permit if delegates wish, but unfortunately there is no longer a special rate for conference delegates and, in fact, the cost of the 3-day permit is considerably higher (£11) than buying a daily ticket. The Pay and Display charges are £1 for 5 hours, £2 for 8 hours and £3 for 24 hours (Monday-Friday). We have tried to negotiate the price of the 3-day permit down to £9 (the charge for 3 separate 24hr rates), but the University of York Conference Services are unable to comply, as they have to "buy in" the service from another department. If you pay for your parking daily, you must remember to put a new ticket on in time or you will be clamped - the release fee is £25.00.

If you wish to purchase a permit it will be included in your delegates pack and is designed to be hung on the rear view mirror. Don't forget to complete the permit with your details or display it – failure to do so will result in your car being clamped.

By Rail Regular trains serve York from stations throughout the UK. Please enquire at your main station or telephone the National Rail helpline (Tel: 08457 484950 website: www.thetrainline.com) for special fares—such as Apex fares or Saver tickets. Taxis are available at the station and buses are close by. The taxi journey from the railway station to the University takes approximately 15 minutes and costs about £6.00.

By Air The nearest airports for York are Leeds and Manchester. There is a good rail service between Manchester airport and York.

SOCIAL PROGRAMME

Wine Tasting Reception - Start Monday evening with the traditional wine tasting reception (18:00-19:00 Exhibition Centre). View the posters and visit the trade exhibitors whilst enjoying a wine tasting. As usual there will be a prize for the competition winner.

5-a-side Soccer - As usual you can kick a football around at 19:00 on Monday evening. The soccer will be inside the University Sports Hall. Bring a team from your department or turn up and make a scratch team. Age/sex unimportant. Nick Lench has again kindly agreed to take on the organisation of this event. Please contact him on lenchnj@cardiff.ac.uk

Conference Party - This year, the Conference Dinner will be on campus. Often our on-campus dinners have had exotic themes, but this year we are celebrating the British heritage. The theme is local – there will be a Yorkshire-themed meal, with the finest quality locally-sourced ingredients and a selection of real ales pulled direct from the barrel. Wine (not British!) will also be available. To complete the evening, there will be traditional dancing from the Ebor Morris Men followed by the opportunity to "strip the willow", etc. in a barn dance - just like the old days...

ON-LINE REGISTRATION

This year registration is being processed on-line and no other method of registration will be accepted. Details are given at the end of this booklet. Deadline for registration is 14th August.

A full-time student, who is not a member of the Society, may pay the lower registration fee as a Society member. Would you please note that we are unable to supply invoices in respect of registration fees.

CONFIRMATION OF REGISTRATION

UK Delegates - your Delegates pack will be sent to you at the end of August.
Overseas Delegates – general information, including details of accommodation, will be sent to you at the end of August – your Delegates pack will be available at the conference from Sunday 17 September at 4.00p.m.

CPD APPROVAL

Like previous conferences it is expected to be accredited by the Royal Colleges of Physicians and Pathologists.

EMERGENCY TELEPHONE NUMBERS

Information Desk during Conference hours: 01904 432475
Goodricke Porters' Lodge: 01904 433100 (for delegates staying in James College)
Langwith Porters' Lodge: 01904 433400

COUNCIL AND SOCIETY MEETINGS

BRITISH SOCIETY FOR HUMAN GENETICS
Council Meeting - 08:30 Monday 18th September - *Room G/020 Goodrick College*
Annual General Meeting -17:30 Monday 18th September - *Central Hall*

ASSOCIATION OF GENETIC NURSES AND COUNSELLORS
Business Meeting - 13:30 Tuesday 19th September - *Room PX001*

ASSOCIATION OF CLINICAL CYTOGENETICISTS
Annual General Meeting - 14:30 Tuesday 19th September - *Central Hall*

OTHER MEETINGS

A Rendezvous with the Junior Liaison Committee – 13:45 Tuesday 19 September – Central Hall
This session is designed for those employed at Band 7 and below (including A-grade trainees). Come along and find out about the ACC, what we do and how we can help you. There will also be a platform for discussing any issues, which in particular, concern the junior members of the profession. If you want to raise anything in advance or just to let us know you are coming drop us a line at h_harvey_smith@hotmail.com

FUTURE MEETINGS

BRITISH SOCIETY FOR HUMAN GENETICS - UNIVERSITY OF YORK
17-19 September 2007
15-17 September 2008
14-16 September 2009

CONFERENCE OFFICE

British Society for Human Genetics
Clinical Genetics Unit, Birmingham Women's Hospital,
Edgbaston, Birmingham. B15 2TG
Tel: 0121 627 2634 Fax: 0121 623 6971
Email: york2006@bshg.org.uk Website: www.bshg.org.uk

PROGRAMME

Sunday 17 September 2006

19:30-20:15 **DINNER** - (Roger Kirk Centre)

Monday 18 September 2006

08:30-10:30 **BSHG - Council Meeting** - Room G/002 (Goodricke College)

11:25 **CHAIRMAN'S WELCOME** (Central Hall)

11:30-13:00 *Symposium – Developmental and Prenatal Genetics*

Chair: Prof Peter Scambler, Dr Charles Shaw-Smith
Central Hall

11:30	(SP01)	The "other" trinucleotide repeat: polyalanine expansions disorders *- Dr Stefan Mundlos (Berlin, Germany)*
12:00	(SP02)	Long-range gene regulation in development and disease *– Prof Veronica van Heyningen (Edinburgh)*
12:30	(SP03)	Fetal cell-free nucleic acids as novel biomarkers for prenatal diagnosis *– Prof Diana Bianchi (Boston, USA)*

13:00-14:15 **LUNCH**

14.15-15:15 *Concurrent Sessions*

Mechanisms of Disease 1

Chair: Dr Joanne Whittaker, Dr Emma Roper
Central Hall

14:15	(SP04)	Mutations in the gene encoding the 3->5 prime DNA exonuclease TREX1 cause Aicardi-Goutieres syndrome at the AGS1 locus *- Dr Yanick Crow, BE Hayward, R Parmar, P Robins, A Leitch, P Lebon, DT Bonthron, AP Jackson, DE Barnes, T Lindahl*
14:30	(SP05)	Mutations in the embryonal subunit of the Acetylcholine receptor (CHRNG) cause lethal and Escobar variants of Multiple Pterygium Syndrome *- Dr Neil Morgan, LA Brueton, P Cox, MT Greally, J Tolmie, S Pasha, IA Aligianis, H van Bokhoven, T Marton, L Al Gazali, J Morton, C Oley, CA Johnson, RC Trembath, HG Brunner, ER Maher*
14:45	(SP06)	The transmembrane protein meckelin (MKS3) is mutated in Meckel-Gruber syndrome *- Dr Colin Johnson, UM Smith, S Pasha, S Malik Sharif, PA Batman, CP Bennett, CG Woods, C McKeown, P Cox, T Attie-Bitach*
15:00	(SP07)	A maternal hypomethylation syndrome presenting as transient neonatal diabetes mellitus *- Dr Deborah Mackay, DO Robinson, SE Boonen, J Clayton-Smith, J Goodship, JMD Hahnemann, SG Kant, PAL Njolstad, NH Robin, R Siebert, JPH Shield, HE White, IK Temple*

Complex Disease

Chair: Dr Philip Beales, Dr Richard Sandford
Exhibition Centre Room PX001

14:15	(SP08)	Prevalent mutations in the filaggrin gene are a major genetic factor in atopic disease. *- Prof Irwin McLean, A Sandilands, FJD Smith, A Terron-Kwiatkowski, SP Lee, Y Zhao, H Liao, G O'Regan, CNA Palmer, AD Irvine*
14:30	(SP09)	Filaggrin mutations are a major predisposing factor in childhood-onset adult-persistent atopic dermatitis *- Prof Richard Trembath, CNA Palmer, SP Lee, MH Allen, Y Zhao, H Liao, SJ Meggit, NJ Reynolds, WHI McLean, JN Barker*
14:45	(SP10)	The genomic organisation of the RCA cluster, including CFH, predisposes to gene conversion and non-homologous recombination as a cause of atypical haemolytic uraemic syndrome *- Prof Judith Goodship, L Strain, J Venables, DM Routledge, H Powell, K Marchbank, THJ Goodship*
15:00	(SP11)	Deletion of CFHR1 and CFHR3 is common and strongly protective against development of age-related macular degeneration. *- Prof Anne Hughes, N Orr, H Esfandiary, U Chakravarthy*

Cytogenetics 1

Chair: Dr Nick Bown, Mr Neil Atkey
Exhibition Centre Room PL001

14:15	(SP12)	Comprehensive DNA copy number profiling of malignant peripheral nerve sheath tumours (MPNSTs) using array based comparative genomic hybridisation (array-CGH) *- Dr Kiran K Mantripragada, G Spurlock, L Kluwe, A Pandita, A Guha, G Evans, R E Ferner, V Mautner, I Frayling, JP Dumanski, M Upadhyaya*
14:30	(SP13)	DNA microarray for detection of copy number changes at the NF1 locus using array comparative genomic hybridisation (CGH) analysis *- Dr Ming Hong Shen, K Mantripragada, J Dumanski, I Frayling, M Upadhyaya*
14:45	(SP14)	Two novel PDGFRA fusions in imatinib-responsive chronic eosinophilic leukaemia *- Ms Claire Curtis, P Musto, G Perla, M Minervini, A Clark, J Stewart, FH Grand, NCP Cross*
15:00	(SP15)	PAX5-ETV6 fusion in acute lymphoblastic leukaemia des not define the dic(9;12) *- Ms Zoe Broadfield, M Martineau, AV Moorman, SL Wright, N Bown, CJ Harrison*

Counselling		Chair: Mrs Gilly Bromilow, Ms Rhona MacLeod Exhibition Centre Room PL002
14:15	(SP16)	Involvement of Clinical Genetics Unit in Establishing Neonatal Sickle Cell Screening in Essex *- Ms Bernadette Farren,*
14:30	(SP17)	Genetics in Health- a service development initiative to explore the South Asian community's attitudes and beliefs regarding genetics and the genetic service *- Ms Heena Patel,*
14:45	(SP18)	Effective risk communication in clinical genetics: a systematic review *- Ms Stephanie Sivell, J Dundon, R Evans, C Gaff, R Iredale, C Shaw, A Edwards*
15:00	(SP19)	Information and support needs of individuals concerned about familial breast cancer *- Ms Stephanie Sivell, R Iredale, A Edwards, J Gray, W Jones, F Rapport, G Elwyn*

15:15-16:00 **TEA**

16:00-17:30 Concurrent Symposia

Mechanisms of Genetic Disease		Chair: Dr Willie Reardon, Dr Sahar Mansour Central Hall
16:00	(SP20)	The VHL tumor suppressor gene and oxygen sensing *– Prof Patrick Maxwell (London)*
16:30	(SP21)	One gene – many diseases. The Lamins and the functional Architecture of the Nucleus *– Dr Colin Stewart (Frederick, USA)*
17:00	(SP22)	To be confirmed

Service Developments in Clinical Genetics		Chair: Ms Christine Patch, Miss Esther Williams Exhibition Centre Room PX001
16:00	(SP23)	Laboratory Service Developments: How have we invested in the future? *- Dr Jo Whittaker*
16:15	(SP24)	Review and outcomes of the initial DoH pilot year as a GPwSI in Clinical Genetics *- Dr Ian Robinson*
16:30	(SP25)	'Cancer - a family affair? *- Ms Chris Jacobs*
16:45	(SP26)	Implementing Service Developments in Genetics – The Manchester Experience *- Mrs Pamela Griffiths*
17:00	(SP27)	Renal genetics: old diseases, new horizons *– Prof Fiona Karet (Cambridge)*
17:15	(SP28)	Demystifying genetics in clinical practice: Haemochromatosis *– Ms Samantha Gamble (Nottingham)*

17:30 **BRITISH SOCIETY FOR HUMAN GENETICS – Annual General Meeting** (Central Hall)

18:00-19:00 **WINE TASTING around TRADE EXHIBITION** (Exhibition Centre)

19:00-20:00 The Great Debate

Chair: Prof Michael Parker
Exhibition Centre Room PX001

This house believes that genetic services have a duty to attempt to contact at-risk relatives directly, rather than rely on communication by patients.

For: Professor Steve Humphries and Dr Ainsley Newson

Against: Professor Graeme Laurie and Ms Lauren Kerzin-Storrar

19:00 **FIVE-A-SIDE FOOTBALL** (Main Sports Hall)

19:30-20:45 **DINNER** (Roger Kirk Centre)

PROGRAMME

PROGRAMME

Tuesday 19 September 2006

09:00-10:30 Symposium – Complex Genetic Disorders

Chair: Prof Dorothy Trump, Dr William Newman
Central Hall

09:00	(SP29)	The genetics of inflammatory bowel disease *– Prof Jack Satsangi (Edinburgh)*
09:30	(SP30)	Aberrant complement activation and regulation in Age-related Macular Degeneration *– Prof Gregory Hageman (Iowa, USA)*
10:00	(SP31)	Hearing impairment: from extreme genetic heterogenity for monogenic forms to complex forms *– Prof Guy van Camp (Antwerp, Belgium)*

10:30-11:15 **COFFEE**

11:15-12:45 Concurrent Sessions

Mechanisms of Disease 2

Chair: Prof David Wilson, Dr Sian Ellard
Central Hall

11:15	(SP32)	The calcium-independent phospholipase A2 gene, PLA2G6, is mutated in a spectrum of childhood neurodegenerative disorders with high brain iron *- Dr Neil Morgan, SK Westaway, JEV Morton, A Gregory, P Gissen, S Sonek, H Cangul, J Coryell, N Canham, N Nardocci, G Zorzi, S Pasha, D Rodriguez, I Desguerre, A Mubaidin, E Bertini, RC Trembath, A Simonati, C Schanen, CA Johnson, B Levinson, CG Woods, B Wilmot, P Kramer, J Gitschier, SJ Hayflick, ER Maher*
11:30	(SP33)	Multiple APC missense variants predispose to colorectal adenomas *- Ms Natalie Jones, K Eliason, JR Sampson, T Scholl, JP Cheadle*
11:45	(SP34)	Exon sequencing: The search for sequence variation in the human genome. *- Dr Alison Coffey*
12:00	(SP35)	RNA processing defects in genetic disease *- Dr Diana Baralle, M Rapon, M Upadhyaya*
12:15	(SP36)	The Dyggve Melchior Clausen syndrome protein, dymeclin interacts with vacuolar protein sorting protein 25 supporting its role in endoplasmic reticulum trafficking *- Dr Esther Kinning, S Shackleton, RC Trembath*
12:30	(SP37)	Tsc1-haploinsufficiency without mTOR activation is sufficient for renal cyst formation in Tsc1+/- mice *- Ms Catherine Wilson, C Bonnet, C Guy, S Idziaszczyk, J Colley, V Humphreys, J Maynard, J Sampson, J Cheadle*

Protecting the Public, Protecting the Professional

Chair: Ms Alison Lashwood, Ms Vicki Wiles
Exhibition Centre, Room PX001

11:15	(SP38)	Offering a good, but incomplete, Prenatal diagnosis option *– Dr Geoff Woods (Cambridge)*
11:30	(SP39)	Taking responsibility *– Ms Nicola Crawford (Sheffield)*
11:45	(SP40)	Medical legal aspects of clinical work *– Mr James Lawford Davies (London)*
12:00	(SP41)	The HPC and Accountability: A Partnership, but with whom? *– Prof Tony Hazell (Cardiff)*
12:15	(SP42)	The principles of root cause analysis *– Mr Mike Surkitt-Parr (Market Harborough)*
12:30		Open Panel Discussion

Diagnosis and Management of Genetic Disease

Chair: Dr Ed Blair
Exhibition Centre, Room PL001

11:15	(SP43)	Effectiveness of non-invasive prenatal diagnosis using free fetal DNA in the maternal circulation. *- Dr Lyn Chitty, J Hogg, P.G Martin, C Meaney, G Norbury*
11:30	(SP44)	Is DNA testing for hypertrophic cardiomyopathy cost-effective? *- Dr Sarah Wordsworth, J Leal, R Legood, J Taylor, K Thomson, A Seller, H Watkins, E Blair*
11:45	(SP45)	Molecularly targeted therapy for tuberous sclerosis complex (TSC) and lymphangioleiomyomatosis (LAM): the TESSTAL trial *- Dr Mark Davies, P Batsford, J A Cox, J Cross, PJ de Vries, T Doyle, F Elmslie, S Johnson, J C Kingswood, D McCartney, K Poynton, A Saggar, A Tattersfield, JR Sampson*
12:00	(SP46)	Developments in DECIPHER - A clinical and research tool for the genomic array era linking phenotype to genomic copy number changes and breakpoints within the Ensembl genome browser *- Dr Helen Firth, RM Pettett, AP Bevan, S Richards, D Kalaitzopoulos*
12:15	(SP47)	'DiaLGEN' - A Liaison Service for the Genetic Assessment of Patients at Risk of an Inherited Arrhythmia *- Dr Ian Ellis, C Benjamin, S Morgan, T Dutton, D Todd*
12:30	(SP48)	ORPHANET – Data Collection in the United Kingdom: Experience and Progress *- Dr Emma Gillaspy, K Strong, J Taylor, S Ayme, D Donnai*

Cytogenetics 2		Chair: Ms Magda Ainscough, Mr Steve Williams Exhibition Centre, Room PL002
11:15	(SP49)	Aneuploidy detection by comparative genomic hybridisation analysis of oocytes and first polar bodies *- Prof Joy D A Delhanty, D Wells, E Fragouli, MJW Faed*
11:30	(SP50)	Evaluation of array CGH platforms for clinical diagnostic purposes using idiopathic learning disability as a model condition. *- Dr Regina Regan, K Smith, M Crocker, C Guiver, J Taylor, SJL Knight*
11:45	(SP51)	Application of a sex chromosome tiling path BAC array for the CGH analysis of X and Y abnormalities *- Ms Alexandra Karcanias, NS Thomas, PA Jacobs, MJ Mitchell, NA Affara*
12:00	(SP52)	A novel 17q21.31 microdeletion syndrome and other cryptic genome imbalances causing idiopathic learning disability discovered using targeted segmental duplication arrays *- S Knight, R Regan, JA Hurst, E Blair, S Price, C Guiver, RC Hennekam, AJ Sharp, S Hansen, R Selzer, Z Cheng, CA Fitzpatrick, S Schwartz, DG Albertson, D Pinkel, EE Eichler*
12:15	(SP53)	FISH studies in 194 patients with aniridia and/or WAGR syndrome *- Dr John Crolla, V van Heyningen, D Robinson*
12:30	(SP54)	Incidence of submicroscopic imbalance at subtelomeres and microdeletion loci in patients referred for Fragile X syndrome *- Dr Joo Wook Ahn, K Mann, H Thomas, A Hallam, C Mackie Ogilvie*

12:45-16:00 **LUNCH AND POSTER VIEWING** (including Tea at 15:15)

13:30 **ASSOCIATION OF GENETIC NURSES AND COUNSELLORS – Business Meeting** (Exhibition Centre, Room PX001)

14:30 **ASSOCIATION OF CLINICAL CYTOGENETICISTS – Annual General Meeting** (Central Hall)

16:00-17:15 Plenary Session - to include Late Breaking Research

		Chair: Prof Richard Trembath, Prof Karen Temple Central Hall
16:00	(SP55)	Mutations in three Ribonuclease H2 subunits cause Aicardi-Goutieres syndrome *- Dr Andrew Jackson, A Leitch, BE Hayward, A Garner, R Parmar, E Griffith, M Ali, The AGS consortium, P Lebon, DT Bonthron, C Ponting, YJ Crow*
16:15	(SP56)	The Role of Wnt7A in human limb development *- Dr Eamonn Sheridan, S Stricker, P Seeman, R Stern, J Cox, E Roberts, K Springell, S Scott, G Karbani, S Malik, J Bond, D Kumar, L Al-Gazali, S Mundlos*
16:30-17:15	**Late Breaking Research**	

17:15-18:15 THE CARTER LECTURE

	Chair: Prof Karen Temple Central Hall
(SP57)	A new look at old Chromosomes – *Professor Patricia Jacobs*

19:30 **CONFERENCE PARTY** – Roger Kirk Centre

PROGRAMME

PROGRAMME

Wednesday 20 September 2006

09:00-10:30 Concurrent Symposium

Genetics and Therapy

Chair: Dr David Rubinzstein, Dr David Barton
Central Hall

09:00	(SP58)	Advances in human genetics: what are the benefits for patients? *– Dr Arnold Munnich (Paris, France)*
09:30	(SP59)	Drug development for neurodegenerative diseases *– Dr Mene Pangalos (Princetown, USA)*
10:00	(SP60)	Novel Therapeutic approaches for BRCA1 and BRCA2 associated cancers *– Dr Andrew Tutt (London)*

Structural chromosome rearrangements - breakpoints, expression and identity by descent

Chair: Dr John Barber, Mr Richard Hall
Exhibition Centre, Room PX001

09:00	(SP61)	Molecular cytogenetic analysis of apparently balanced chromosome rearrangements *– Dr Judy Fantes (Edinburgh)*
09:30	(SP62)	The origins of chromosome inversions and balanced translocations *– Dr Simon Thomas (Salisbury)*
10:00	(SP63)	Large genomic deletions influence expression levels of the non-hemizygous flanking genes *– Prof Alexandre Reymond (Lausanne, Switzerland)*

10:30-11:15 **COFFEE**

11:15-12:45 Symposium – Genomics and Medicine

Chair: Dr David Fitzpatrick, Dr Diana Johnson
Central Hall

11:15	(SP64)	Properties of high- and low-penetrance RET mutations in Hirschsprung aganglionosis: Generalizations *– Dr Aravinda Chakravarti (Baltimore, USA)*
11:45	(SP65)	Patterns of somatic mutation in human cancer genomes *– Prof Mike Stratton (Cambridge)*
12:15	(SP66)	Genetics of natural variation in human gene expression *– Dr Vivien Cheung (Philadelphia, USA)*

12:45-14:00 **LUNCH**

14:00-16:00 Workshops

Genomic Tools to target diagnosis, prognosis and therapy of common disease

Central Hall
Chair: Dr Graham Taylor, Dr Fiona Macdonald

14:00	(SP67) A genome wide scan to find common, low-penetrance breast cancer susceptibility loci *–- Dr Alison Dunning (Cambridge)*
14:20	(SP68) EGFR mutations and treatment response in cancer *– Ms Sally Cottrell (London)*
14:40	(SP69) Detection of ABL Kinase Domain Mutations in Chronic Myeloid Leukaemia patients resistant to Imatinib *– Dr Elizabeth Ormshaw (Birmingham)*
15:00	(SP70) Treating monogenic diabetes according to genotype *– Dr Sian Ellard (Exeter)*
15:20	(SP71) TARGET – predicting adverse reaction to azathioprine prescription *– Dr Simon Ramsden (Manchester)*
15:40	5 minute comments / contributions from the floor

Genomic Disorders

Exhibition Centre Room PX001
Chair: Dr Helen Firth, Dr Jane Hurst

14:00	(SP72) Mechanisms of genomic rearrangements *– Prof Andrew Wilkie (Oxford)*
14:15	(SP73) 7q– Williams syndrome and the WBS reciprocal duplication syndrome *– Prof Dian Donnai (Manchester)*
14:35	(SP74) Genetics of Silver-Russell syndrome *– Dr Thomas Eggermann (Aachen, Germany)*
14:55	(SP75) 15q11-13 phenotypes: Angelman and Prader-Willi syndromes *– Dr Jill Clayton-Smith (Manchester)*
15:15	(SP76) A novel and clinically distinct 17q21.31 microdeletion syndrome *– Dr Jane Hurst, Dr Helen Firth (Oxford)*
15:35	(SP77) Genotype-Phenotype correlation in 22q11.2 associated syndromes *– Dr Anita Rauch (Erlangen, Germany)*

Microarray Implementation

Exhibition Centre Room PL001
Chair: Prof Nick Cross, Dr Carol Delaney

14:00 (SP78) Bringing array CGH from the scientific into the diagnostic laboratory
– Dr Joris Vermeesch (Leuven, Belgium)

The aim of the Microarry CGH Implementation Group is to facilitate the introduction of microarray CGH into NHS genetic services by sharing experiences and planning common ways forward. Presentation from both research and service laboratories.

16:00 **TEA**

Poster presentations

1. Clinical genetics

(1.01) Does genotype predict tumour risk in Costello syndrome - *Mr Ben Tang, B Kerr, GCM Black*

(1.02) Surveillance for Malignancy in Bloom Syndrome: A Case Report - *Dr Lisa Walker, E Thomas, S Shanley, R Eeles*

(1.03) Aminoglycoside-induced Deafness: prevention by genetic testing is cost-effective - *Dr Maria Bitner-Glindzicz, V Osei-Lah, I Colvin, D Lucas, B MacArdle, T Sirimanna, D Webb, A Shankar, J Kingston, L Jenkins, S Rahman*

(1.04) Aetiology of sensorineural hearing loss in British Bangladeshi children in East London - *Dr Maria Bitner-Glindzicz, Y Bajaj, T Sirimanna, P Quader, L Jenkins, S Loughlin, M Cortina-Borja, D Albert*

(1.05) Molecular analysis and genetic counselling in Deaf-Deaf marriages - *Dr Fiona Connell, D Ruddy, L Jenkins, M Bitner-Glindzicz*

(1.06) Hypertrophic Cardiomyopathy in Fabry disease presenting with sudden cardiac death - *Dr Adila AlKindy, D Kumar, S Davies*

(1.07) Lipoedema associated with familial growth hormone deficiency - *Dr Sahar Mansour, G Brice, S Jeffery, P Mortimer*

(1.08) Exon resequencing and mutation detection in syndromic oesophageal atresia - *Dr Charles Shaw-Smith, E Howard, K McLay, A Dunham, J Burton, S Hunt, S Leonard, J Rogers, A Coffey*

(1.09) Congenital Synspondylism: 3 further cases of this rare skeletal dysplasia - *Dr Audrey Smith, K Metcalfe*

(1.10) Prevention Of Neural Tube defects by Inositol (PONTI). - *Dr Lyn Chitty, NDE Greene, K Burren, T Hesketh, A Copp*

(1.11) A further case of Bartsocas-Papas syndrome - *Dr Duncan Rourke*

(1.12) Osteocraniostenosis: a further case report documenting the antenatal findings - *Dr Audrey Smith, P Bullen, J Clayton-Smith*

(1.13) Bruck Syndrome: a case of osteogenesis imperfecta with joint contractures and minimal fractures - *Dr Ajoy Sarkar, RA Bank, CM Hall, AF Brady*

(1.14) A manifesting carrier with X-linked lymphoproliferative disease? - *Dr Rachel Cole, K Gilmour, C Meaney, C Cale, F Lalloo*

(1.15) Familial mild Jeune syndrome with a manifesting heterozygote - *Dr Diana Johnson, VA Murday, R Thomson*

(1.16) A case of Loeys Dietz Syndrome - *Dr Dorothy Halliday*

(1.17) Cystic Fibrosis in Echogenic Bowel Referrals in the Northwest UK - *Mr David Jones, D Trump, M Schwarz*

(1.18) Lowe (Oculocerebrorenal) syndrome: Lessons learned from three new cases - *Dr Kathryn Leask, A Smith, C Lloyd, G Black, A Wallace, J Clayton-Smith*

(1.19) Familial Multiple Biliary Hamartomas-a new clinical syndrome? - *Dr Richard Sandford, W Gelson, DJ Lomas, SE Davies, AC Douds, M Hoare, P Charoen, GJM Alexander*

(1.20) X-linked epilepsy affecting females and sparing males: report of a second family - *Dr Shane McKee*

(1.21) Metachromatic Leucodystrophy: A family with uncertain genotype-phenotype correlation - *Dr David Halsall, JCP Roos, TS Elsey, P Stein, J Calvin*

(1.22) The first reported case of Ulnar Mammary Syndrome with heart abnormalities. - *Dr Katrina Prescott, R Williams, J King, E Blair, U Kini*

(1.23) Familial multiple ventricular extrasystoles and skeletal features - a further family with Stoll syndrome? - *Dr Catherine Mercer*

(1.24) Clinical and molecular details on a family with LADD syndrome - *Dr Angus Dobbie, B Wollnik*

(1.25) A case series of aniridia patients referred to the Northern Ireland Genetics Service during 1970-2005 - *Dr Vivienne McConnell, F Stewart*

(1.26) A novel presentation of thyroid agenesis in a child with the microdeletion Del (1) (q44) - *Dr Asma Shah, E Paul, S Al Wahab, A Saggar*

(1.27) Subtelomeric 2p microdeletions: Another cause of a Prader-Willi like phenotype? - *Dr Joanna Prothero, R Palmer, R Hennekam, E Rosser*

(1.28) Case Report: Deletion 3q22.1-q23 with BPES and an AHO-like brachydactyly phenotype - *Dr Marion Croft, J Crolla, A M Differ, L Burville-Holmes, V Maloney, P D Turnpenny*

(1.29) Description of a subtle de novo deletion/duplication involving chromosome 5p15 and the associated phenotype - *Dr Diana Johnson, A Kay, M Callaghan, E Ryan, J Boyce, N Morrison*

(1.30) Novel duplication: ish dup(11)(p12p15.2). Report of a case and review of the dup(11p) phenotype - *Dr Diana Johnson, M Callaghan, J Simpson, F Glencross, G Bakirtzis, N Morrison, R Morrison*

(1.31) Inversion 13q vs Insertion 13q – clinical phenotype and consequences? - *Dr Marie Leema Patricia Robert, CM Brewer, E Roberts, L Burvill-Holmes, L Morgan, T Davies*

(1.32) Dicentric Chromosome 15 Syndrome - *Dr Cheng Hiang Lee, P Woods, AC Magee*

POSTER PRESENTATIONS

(1.33) A family with overgrowth associated with partial trisomy 15q - *Dr Katrina Tatton Brown, J Barber, C Donaghue, VK Maloney, K Marks, S Tomkins, P Waits, N Rahman, M McEntagart*

(1.34) Microdeletion encompassing the MAPT gene at chromosome 17q21.3 is associated with developmental delay and learning disability - *Dr Charles Shaw-Smith, A Pittman, L Willatt, H Martin, L Rickman, S Gribble, C Rosenberg, HV Firth, R de Silva, NP Carter*

(1.35) Familial 22q11.2 duplication: the mild and the severe - *Dr Dragana Josifova, DJ Josifova, D Ruddy, P Warburton, S Bint*

(1.36) Fronto-Ocular Syndrome: A further report in a male with a cryptic telomere imbalance - *Dr Louise C Wilson, RCM Hennekam, S NIsbet, D Morrogh, R Palmer*

(1.37) A large patient study confirming that facioscapulohumeral muscular dystrophy (FSHD) disease expression is almost exclusively associated with an FSHD locus located on a 4qA-defined 4qter subtelomere - *Dr Meena Upadhyaya, NST Thomas, K Wiseman, G Spurlock, M MacDonald, D Üstek, M Upadhyaya*

(1.38) X-linked Alport syndrome and severe constipation associated with a contiguous COL4A5 and COL4A6 gene deletion - *Dr Jenny Thomson, J Pagan, CE Chu, F Flinter, P Renwick*

(1.39) A novel autosomal dominant progressive limb-girdle myopathy with bone fragility maps to chromosome 9p21-p22 - *Dr Sarju Mehta, GDJ Watts, C Zhao, S Mumm, D Novack, S Ramdeen, SJ Hamilton, C Briggs, MP Whyte, B McGillivray, VE Kimonis*

(1.40) Autozygosity mapping of Bardet-Biedl syndrome to 12q21.2 and mutation analysis - *Mr Dominic White, D Nishimura, E Maher*

(1.41) Establishing a high throughput sequencing service for mutation detection in several syndromic X-linked mental retardation genes - *Dr James Drummond, H Martin, L Raymond, J Whittaker*

(1.42) Esco1, a candidate gene for TAR syndrome - *Ms Stephanie Barton, T Singh-Dang, K Greenhalgh, R Newbury-Ecob, M Crosier, AH Trainer*

(1.43) The investigation of quantitative and qualitative effects of CFTR variants on cystic fibrosis phenotype - *Mr Shadi Al-Baba, HN Hughes, MJ Schwarz*

(1.44) Familial isolated hyperparathyroidism, arising from somatic mosaicism for a missense mutation in the MENIN gene - *Dr Louise Izatt, C Jacobs, C Fratter, T Cranston, F Hannan, R Thakker, S Aylwin*

(1.45) Bloom Syndrome in the Midlands of England - a founder mutation? - *Dr Emma R Woodward, S Patel, N Canham, M Barrow, L Brueton*

(1.46) A Northern Irish Founder Mutation in SDHD causing Familial Phaeochromocytoma/Paraganglioma Syndrome - *Dr Lisa Walker, P Morrison, J Paterson, V McConnell*

(1.47) Prenatal Diagnosis of Autosomal Recessive Antley Bixler Syndrome caused by P450 Oxidoreductase Deficiency - *Dr Katrina Prescott, W Arlt, J Hurst, R Black, S Gould, H Stewart, E Blair*

(1.48) Preimplantation Genetic Diagnosis for Non-syndromic deafness - *Ms Georgia Kakourou, S Dhanjal, R Sud, T Mamas, A Doshi, S Nuttall, S Gotts, P Serhal, JD Delhanty, JC Harper, S SenGupta*

(1.49) Preimplantation Genetic Diagnosis for Retinoblastoma Predisposition - *Ms Seema Dhanjal, G Kakourou, N Saleh, T Mamas, A Doshi, S Gotts, S Nuttall, K Fordham, P Serhal, J Delhanty, J Harper, S SenGupta*

(1.50) The frequency of inherited metabolic disorders in the West Midlands, United Kingdom - *Dr Simon Sanderson, A Gree, MA Preece, H Burton*

(1.51) Polycystic Kidney Disease - At strategy for the identification of at risk individuals - *Ms Lucy Burgess, F McGlynn, L Foggensteiner, T Cole, V Sleightholme, P Farndon*

(1.52) Bringing Genetics into Mainstream Medicine: Development of a Multidisciplinary Renal Genetic and Tubular Disorders Clinic - *Dr Richard Sandford, DA Spencer, AGW Norden, NC Shah, S Waller, J Whittaker, FE Karet*

(1.53) 2006 national survey of cardiac genetic services in the UK: a gathering storm? - *Dr Paul Brennan*

(1.54) Genetics and mainstream medicine- the importance of two way traffic. Lessons from management review of Osteogenesis Imperfecta (OI) and Congenital Adrenal Hyperplasia (CAH) - *Dr Trevor Cole, V Sleightholme, L Burgess, S Stewart, R Glennis, W Arlt, N Gittoes*

(1.55) Cervical spine neurofibromas in NF1 microdeletion patients - *Dr Shincy John, B Castle, M Upadhyaya, J Fairbank, RSC Kerr, SM Huson, LE Side*

(1.56) Mainstreaming Genetics: Inherited cardiovascular disease as a model - *Ms Christina Honeywell, H Watkins, C Fraser-Jones, E Blair*

(1.57) Genetics for Learning Disability Psychiatrists - *Ms Jo Haydon, C Bennett, P Farndon*

(1.58) Towards outcome measures for clinical genetics services: a comparison of genetics healthcare professionals and patients' views - *Dr Katherine Payne, SG Nicholls, M McAllister, R MacLeod, D Donnai, LM Davies*

(1.59) Process and outcome in clinical genetics services: a new model developed from qualitative research - *Dr Marion McAllister, S Nicholls, K Payne, R MacLeod, D Donnai, L Davies*

(1.60) Outcome measurement in clinical genetics services: a systematic review of validated measures - *Dr Katherine Payne, SG Nicholls, M McAllister, R MacLeod, D Donnai, LM Davies*

(1.61) A pilot study evaluating the introduction of patient-clinician email communication in a genetics service - *Dr Emma McCann, A Fryer, P Bundred*

(1.62) Identification and appropriate referral of patients at risk of genetic disorders; the role of primary care - *Ms Deborah McCahon, S Wilson, A Metcalfe, P Gill, T Cole, HV Sleightholme*

(1.63) The Development of Personal Health Records for Genetic Disorders - *Mrs Pamela Griffiths, R Abbott, S Huson, E Howard, J Clayton-Smith, K Metcalfe, G Evans, S Harrison, M Kenney, S Collitt, B Kerr*

(1.64) Reciprocal Observation in Clinical Genetics Departments: A useful training exercise? - *Ms Caroline Philp, Dr Roberts*

(1.65) To test or not to test.... - *Dr Peter Lunt, Maggie Williams*

(1.66) Audit to establish whether BRCA carriers undergoing bilateral prophylactic salpingoophorectomy (BPSO) have evidence of fallopian tube removal in the Genetics notes - *Dr Emily Craft, L Izatt, S Rose, G Pichert*

(1.67) Audit: Clinical Management of Familial Breast Cancer Patients in UK tertiary care - *Ms Sarah Harris*

(1.68) Audit of referrals to the Cheshire & Merseyside Clinical Genetics Service from the regional Multiple Endocrine Neoplasia clinic - *Dr Ruth Day, KL Greenhalgh, K Gamet*

(1.69) Re-Audit of referral practice to the cancer genetics service at Guy's Hospital -appropriateness and outcome - *Dr Louise Izatt*

2. Molecular Genetics

(2.01) In ovo knock-down of Tbx5 and Mef2c expression - *Ms Feifei Song, T E Robinson, J D Brook*

(2.02) Elevated maternal expression of the imprinted PHLDA2 gene is associated with low birth weight - *Dr Sophia Apostolidou, S Abu-Amero, K O'Donoghue, O Olafsdottir, KM Chavele, J Frost, JC Whittaker, P Loughna, P Stanier, GE Moore*

(2.03) Fetal cell-free DNA in maternal circulation: a potential prognostic marker for chromosomal abnormalities? - *Ms Ageliki Gerovassili, CP Garner, KH Nicolaides, SL Tein, D Rees*

(2.04) Effects of Blood Contamination in Amniotic Fluid for Amnio PCR Testing (QF-PCR) - *Mr Duncan Black, TP Bradley, LJ Levett, S Liddle*

(2.05) A potential false positive result for Prader-Willi syndrome using a PCR-based assay on bisulphite modified DNA. - *Ms Ruth Elaine Sutton, D Bourn, HM Powell, E Selkirk, A Curtis*

(2.06) Analysis of a cohort of Silver Russell Syndrome patients for epigenetic changes at the H19DMR and KvDMR loci at 11p15 - *Ms Anne-Marie Coupe, S Thomas, D Mackay, H Bullman, J Harvey*

(2.07) Establishment of a Diagnostic Service for Sotos - *Mr Sam McCall, S Ramos, M Fidanis, C Bunn, R Taylor*

(2.08) Clinical phenotype and autozygosity mapping of phenotypic diarrhoea of infancy - *Dr Jane Hartley, P Gissen, B Dawood, S Watson, R Pollitt, W Kahr, D Chitayat, D Kelly, C Johnson*

(2.09) Towards more Comprehensive Diagnostic Screening of SHOX Mutations. - *Mr Kevin Baker, S Thomas, D Bunyan, J Harvey*

(2.10) The CFTR mutation I148T: time for demotion? - *Dr Lampros Mavrogiannis, CP Bennett*

(2.11) Validation of a Luminex-Based Multiplex Assay for 25 Cystic Fibrosis Mutations - *Dr Trudi McDevitt, C King, D E Barton*

(2.12) An Audit of Genotypes and Borderline Sweat Tests in Irish CF Patients - *Dr Caitriona King, T McDevitt, D Lambert, DE Barton, SA Lynch*

(2.13) Current and future molecular testing of cancers: A survey of UK laboratories - *Ms Irene Papanicolas, S Wordsworth, J Buchanan, J Taylor, I Frayling, I Tomlinson*

(2.14) The Role of CASP8 D302H and Other Apoptosis Gene Variants in Breast Cancer - *Dr Sushila Mistry, S Rafii, A L Shippen, G MacPherson, S Balasubramanian, M W Reed, A Cox*

(2.15) Implementation of a new BRCA1 and BRCA2 screening service that meets Genetics White Paper 8 week turnaround time - *Dr Yvonne Wallis, N Motton, C Morgan, E Ormshaw, N Morrell, JA Bell, F Macdonald*

(2.16) Detection of large deletions and duplications in BRCA1 by WAVE analysis: - *Mr Navaratnam Elanko, W King, C Bunn, A Sinclair, R Taylor*

(2.17) Application of sequencing and high-resolution melting in a high-throughput BRCA1 screening service - *Dr Rachel Robinson, R Charlton, IM Carr, S Bibi, K Bransfield, C Taylor, GR Taylor*

(2.18) Screening of BRCA1 & 2 by DNA sequencing and MLPA – increased throughput and coverage equals more unclassified variants - *Dr Caroline Bunn, E Fidanis, W King, S Ramos, J Short, S Cottrell, R Taylor*

(2.19) Design and optimisation of a validated primer set for automated mutation scanning of the BRCA1 and BRCA2 genes. - *Mr Chris Mattocks, D Ward, JF Harvey, NCP Cross*

(2.20) Investigation of an HNPCC mutation of unknown significance - a clinical and laboratory perspective. - *Ms Lisa Hughes, S Palmer-Smith, M T Rogers, I Frayling, M Stephens, L Rosser, B Jasani, G Williams, B Jasani*

(2.21) A diagnostic service for PMS2 mutation screening - *Ms Jenny Bentley, R Charlton, G Taylor*

(2.22) Evaluation of an amplicon array-CGH technique for gene dosage analysis in hereditary cancers - *Mrs Sarah Shepherd, I Berry, CA Delaney, GR Taylor*

(2.23) Development of a genetic diagnostic service for Birt Hogg Dube syndrome - *Ms Louise Carr, J Bell, E Woodward, T Cole, F Macdonald, ER Maher*

(2.24) A Molecular Diagnostic Screening Service for Gorlin Syndrome. - *Dr Joan Forsyth, M Allen, JA Bell, V Lindley, K O' Leary, C Hardy, F Macdonald, P Farndon*

(2.25) Comparison of germline and somatic mutational spectra in NF1-associated peripheral nerve sheath tumours (MPNSTs) - *Dr Meena Upadhyaya, G Spurlock, Lan Kluwe, K Mantripragada, E Majounie,A Pandit, G Evans, A Guha, J Dumanski, R Ferner, V Mautner, M Upadhyaya*

(2.26) An absence of cutaneous neurofibromas associated with a 3-bp in-frame deletion in exon 17 of the NF1 gene (c.2970_2972 delAAT - *Dr Meena upadhyaya, M Upadhyaya, SM Huson, M Davies, S Giovannini, S Griffiths, C Consoli, L Side, D Adams, M Pierpoint, R Hachen, D Stevenson, D Viskochil, P Wallace, A Barnicoat, N Chuzhanova, D Baralle, C Lazaro, E Haan, P Turnpenny, L Messiaen*

(2.27) PTPN11 mutations in a group of patients with indolent JMML. - *Ms Jane Chalker, A Rao, R Gale, D Webb, H Kempski*

(2.28) Incidence and molecular anatomy of the FIP1L1-PDGFRA fusion gene - *Ms Joannah Score, C Walz, A Reiter, NCP Cross*

(2.29) Identifying the V617F JAK2 mutation: a cost effective method of diagnosing clonal polycythemia vera (PV) and other chronic myeloproliferative disorders. - *Ms Stacey Sandell, O Wood, AV Jones, S Burgstaller, JF Harvey, NCP Cross*

(2.30) Oxford Molecular Genetics Laboratory Cardiomyopathy Service - *Ms Jessica Thistleton, K Thomson, E Blair, H Watkins, A Seller*

(2.31) Screening for mutations in the cardiac Troponin T (TNNT2) gene in Scottish Grampian patients with Hypertrophic cardiomyopathy - *Ms Shalaka Samant, KF Kelly, C Clark, J Dean*

(2.32) Oxford Molecular Genetics Laboratory service for LongQT and Brugada Syndromes. - *Ms Jessica Thistleton, K Livesey, K Thomson, E Blair, A Seller*

(2.33) Long QT Syndrome: Does clinical information predict genotype? - *Ms Dawn Walker, S Borras, G Rettie, C Clark, K Kelly, J Dean*

(2.34) Expression of HIF-1a, HIF-2a (EPAS1), and their target genes in paraganglioma and phaeochromocytoma with VHL and SDH mutations - *Dr Patrick Pollard, M El-Bahrawy, R Poulsom, P Killick, F Latif, S Hodgson, I Tomlinson, E Maher*

(2.35) A Molecular Diagnostic Screening Service for Familial Paraganglioma and Phaeochromocytoma - *Mrs Claire Morgan, JA Bell, F Macdonald, ER Maher*

(2.36) Lost for Words: The Search for a Candidate Gene - *Ms Uma Mallya, J Moon, I Waterson, H Cordell, FL Raymond*

(2.37) Absence of Mitochondrial DNA Abnormalities in two Saudi Families with Gaucher Disease - *Dr Khaled Abu-Amero*

(2.38) Hi-Res MeltingTM as a mutation scanning tool in malignant hyperthermia - *Ms Jennifer Munkley, C Taylor, PJ Halsall, PM Hopkins, RL Robinson*

(2.39) Hepatocyte nuclear factor-1beta gene deletions are common in subjects with unexplained renal disease and diabetes - *Ms Martina Owens, EL Edghill, LW Harries, C Bingham, R Oram, AT Hattersley, S Ellard*

(2.40) Molecular genetic diagnosis of mitochondrial DNA depletion syndrome and Alpers syndrome - *Mr Carl Fratter, A O'Rourke, D Marchington, K Morten, A Seller, J Poulton*

(2.41) Mental retardation and epilepsy: A study of six cases with the common polyalanine expansions in Exon 2 of the Aristaless-related homeobox gene (ARX) at Xp22.13. - *Mrs Moira J MacDonald, L P Lazarou, L Wilson, SJ Davies, J Hurst, T McShane, A Green, DT Pilz*

(2.42) MLPA analysis of patients with classical lissencephaly reveals a high frequency of deletions - *Dr Thalia Antoniadi, LP Lazarou, E Robson, H Duarte, IM Frayling, DT Pilz*

(2.43) Detection and Accurate Sizing of Expanded Fragile X alleles using a Multiplex Fluorescent PCR Assay - *Dr Michael Sweeney*

(2.44) Partial rescue of truncating mutations in the ATRX gene through unusual intra-exonic splicing - *Mrs Nicola Malik, C Fisher, A Fryer, DR Goudie, ID Krantz, J Traeger-Synodinos, RJ Gibbons*

(2.45) Diagnostic testing of the Tau gene for patients with Frontotemporal dementia with Parkinsonism (FTDP-17) - *Ms Rebecca Treacy, MG Spillantini, J Hodges, J Whittaker*

(2.46) Identification of mutations within the spastin gene (SPG4) by Temperature Gradient Capillary Electrophoresis. - *Mr David Moore, C Shaw, S Abbs*

(2.47) Further refinement of the SPG11 critical interval in a consanguineous family with autosomal recessive hereditary spastic paraplegia and thin corpus callosum - *Ms Laura Baumber, RC Trembath*

(2.48) Mutation Analysis of CCM1 and CCM2 in Patients Affected with Hereditary Cerebral Cavernous Malformations - *Mr Thomas Carrick, A Stewart, D Baty, J Berg*

(2.49) Mutations in mitofusin 2 are a common cause of CMT2 - *Dr Vaneesha Gibbons, MG Sweeney, MM Reilly, MB Davis*

(2.50) Coincidental finding of 2 different CMT mutations segregating in a family - *Ms Davina Clavering, K O'Leary, F Macdonald, R Barber*

(2.51) Mutation scanning for nearly all mutation types in 109 individuals diagnosed with Xp21 muscular dystrophy - *Dr Emma Ashton, SC Yau, ZC Deans, SJ Abbs*

(2.52) Mutation detection in a large cohort of muscular dystrophy patients widens the clinical spectrum associated with dystroglycanopathy genes. - *Ms Caroline Godfrey, R Mein, C Jimenez-Mallebrera, L Feng, M Brockington, F Muntoni, S Abbs*

(2.53) Clinical and Genetic Testing for Inherited Renal Disorders at Addenbrooke's Hospital - *Dr Sarah Waller, E Barlow, V Thomas, J L Whittaker, R Sandford, F Karet*

(2.54) Application of Real-Time PCR Allelic Discrimination to Mutation Analysis in Autosomal Recessive Polycystic Kidney Disease (ARPKD) - *Dr Rhianedd Ellwood-Thompson, AL Meredith*

(2.55) A genome-wide scan for genes involved in vesicoureteral reflux - *Dr David E Barton, H Kelly, A Yoneda, JM Darlow, D Shields, C Molony, P Puri*

(2.56) NGRL (Wessex) evaluation of high resolution melt curve analysis for mutation scanning - *Dr Helen White, GL Potts, NCP Cross*

(2.57) Reference Reagents for Mutation Scanning: Development of Plasmid Based Reagents by NGRL (Wessex) - *Dr Helen White, GL Potts, VJ Hall, N Owen, CJ Mattocks, NCP Cross*

(2.58) Development of a rapid, accurate and cost effective service using MALDI-Tof Mass spectrometry for the genetic screening of mismatch repair genes MLH1, MSH2 and MSH6 in HNPCC patients. - *Ms Jeanne Buys, JA Thomson, H Field, C Lucas, S Cowen, R Flora, PG Debenham*

(2.59) Mutation scanning by high-resolution melting curve analysis – one year's experience of routine service use. - *Dr Claire Taylor, C Booth, P Chambers, J Lowery, T Stanton*

(2.60) Implementation of the AmpliChip CYP450 Test in the Diagnostic Laboratory - *Dr Nathalie Dhomen, R Lucey, S Liddle, LJ Levett*

(2.61) Developments leading to a "one patient per plate" high-throughput sequencing strategy - *Dr Howard Martin, I Johnson, L Daboo, V Burkitt, E Barlow, J Whittaker,*

(2.62) Functional characterisation of genome sequence variation - *Dr Talat Nasim, AG Gouri), B Patel), RC Trembath*

(2.63) Development of Affymetrix SNP arrays for the diagnostic assessment of SNP genotype in genetically heterogeneous disorders - *Ms Eleanor Rattenberry, EJ Prebble, F Macdonald, JA Bell, DR White, C Johnson*

(2.64) Keeping track: increased data management in diagnostic molecular genetics - *Mr Will King, I Turner, A Marsh, C Bunn, E Fidanis, R Taylor*

(2.65) Mutation Searching in SCN1A: Implications for management of SMEI patients - *Ms Rachael Birch, SM Zuberi, S Stenhouse, JG Kelly, L Conlin*

(2.66) Characterisation of wild type fibulin 5 and mutants associated with age-related macular degeneration - *Dr Richard PO Jones, C Ridley, AC Lomas, T Wang, M Howard, T Jowitt, C Baldock, T Nakamura, A Lotery, PN Bishop, CM Kielty, D Trump*

(2.67) Chromatin packaging in human spermatozoa - *Dr David Miller, DE Iles, A Arpanahi, MH Brinkworth*

(2.68) Y microdeletions in Male Infertility- 8 years service experience - *Ms Eleni Mavraki, H Sawyer, A Gardner, P Lunt, P Wardle, D Siassakos, PJ Stannard, K Bigwood, N Sharp, U Acharya, H Mekhael, M Williams*

(2.69) Methylation status of the MGMT gene promoter: detection by methylation-specific MLPA - *Dr Lampros Mavrogiannis, C A Delaney, A Chakrabarty, P Chumas, J P Schouten, P Roberts*

(2.70) Implementation of the SCOBEC High Throughput Screening Facility - *Mr Chris Mattocks, D Ward, J Sillibourne, T Herbert, N Owen, M Wall, P Goddard, JF Harvey, NCP Cross*

3. Cytogenetics and Molecular Cytogenetics

(3.01) NGRL (Wessex) evaluation of CE marked in vitro diagnostic test kits for prenatal diagnosis of aneuploidy - *Dr Helen White, VJ Hall, T Boyle, J Warner, J McLuskey, J Sibbring, S Hamilton, J Diack, S Allen, M Jones, NCP Cross*

(3.02) Telomere length analysis and Aneuploidy screening in human pre-implantation embryos using a quantitative FISH approach - *Ms Anastasia Mania, A Mantzouratou, J Delhanty*

(3.03) Discrepant Uncultured Amniocyte FISH and Long Term Culture Results, as a Result of Maternal Cell Contamination, Indicate a Need for Professional Guidelines for Interpreting Amniocyte FISH - *Mrs Karen Thompson, AL Hammersley, J Evans*

(3.04) Balanced chromosomal abnormalities and normal karyotypes in amniocyte cultures of fetuses with ultrasound scan abnormalities. - *Mr Terry Ballard, S Joseph*

(3.05) A study of how the enzymatic disaggregation of villi versus the sampling of individual fronds of villi can alter the result of both QF -PCR and FISH in a mosaic placenta. - *Ms Charlotte Noakes, M Crocker,*

(3.06) A Case of a Jumping Translocation in a Prenatal Case of Down Syndrome - *Ms Jennifer Corfield, D Delemge, K Donaldson, E Bagley, M Jones, E Clements, P Brady, E Roberts, S Gran), T Davies,*

(3.07) A false negative trisomy 18 QF-PCR result due to CVS mosaicism: the West Midlands Regional Genetics Laboratory Experience. - *Mrs Anita Luharia, S Allen, N Bibb, A Marshall, C Gould, S Larkins, G Fews, F MacDonald, EV Davison*

(3.08) A familial X centromere alpha satellite variant resulting in a false positive prenatal diagnosis of monosomy X by interphase FISH - *Mr Richard Nash, M Batema, A Clarkson, K Morgan, C Lees, I Simonic*

(3.09) Analysis of whole genome amplification methods prior to genome wide detection of chromosomal imbalances by array CGH - *Dr Carol A Delaney, M Mohammed, GR Taylor*

(3.10) Challenges for evaluating emerging genetic tests: Lessons learned from an evaluation of array-based comparative genomic hybridisation for investigating chromosomal rearrangements in patients with learning disability. - *Dr Simon Sanderson, RL Zimmern, JPT Higgins, H Burton, M Kroese, S Subramonia-Iyer, P Brice*

(3.11) Array-CGH Reveals A Duplication Of The Williams-Beuren Region - *Mrs Lucy Eastoe, E Prebble, G Fews, D McMullan, M Griffiths, L Brueton, V Davison*

(3.12) Introduction of Array CGH into the clinical setting: The Oxford laboratory experience - *Dr Mark Crocker, R Candlin, S Knight, R Regean, K Smith*

(3.13) The Beginning of the Ends, Sheffield's experience of subtelomeric FISH testing. - *Mrs Sarah Willis, M Dyson, S Williams, E Maltby*

(3.14) MLPA analysis in neuroblastoma: a pilot study using six cell lines. - *Dr Katrina Gill, C Knox, D Tweddle, A Curtis, N Bown*

(3.15) A variant translocation of t(10;11)(p12;q23) - *Dr George Bakirtzis, W Baird, J Stewart, G Lowther*

(3.16) Follow up of potential constitutional chromosome abnormalities ascertained through cancer cytogenetics - *Ms Anna Benson, J Emslie, N Bown*

(3.17) A de novo microdeletion of 5p15.1 in a boy with autism spectrum disorder - *Dr Lionel Willatt, S Kenwrick, G Parkin, J Whittaker, D Baralle*

(3.18) Duplication of 15q11-q13 associated with autistic spectrum disorder - *Ms Layla Nadine Al-Bustani, K. Martin, M. Suri*

(3.19) FISH fine-mapping detects an inversion and two 2Mb deletions in a familial 4;11 translocation associated with autosomal dominant polycystic kidney disease and macular dystrophy - *Dr Lionel Willatt, A C Clarkson, AT Moore, FE Karet, G Parkin, J Burbridge, I Simoniz, R Sandford*

(3.20) Introduction of the Affymetrix 10K SNP oligonucleotide array into the West Midlands Regional Genetics Laboratory – Its uses in AML karyotype characterisation - *Ms Judith Walker, DJ McMullan, MJ Griffiths, EV Davison*

(3.21) Evaluation of the use of array CGH on bone marrow from AML patients with a normal karyotype - *Ms Maria Sedlmeier, S Hewitt, H Dickinson, P Roberts, C A Delaney*

(3.22) Molecular assessment of post transplant BCR/ABL transcript levels in CML patients - *Dr Susanna Akiki, J Mason, E Rattenberry, M J Griffiths*

(3.23) Band-specific distribution of 5-methylcytosine in metaphase chromosomes from adult and fetal human lymphocytes - *Ms Olga Efimova, A Pendina, T. Kuznetzova, V Baranov*

(3.24) Novel deletion variants of 9q21.11-q21.12 and classical euchromatic variants of 9q12/qh involve large scale copy number variation of segmentally duplicated pericentromeric euchromatin - *Dr Lionel Willatt, JCK Barber, AJ Clarkson, I Simonic, FL Raymond, Z Docherty, CM Ogilvie*

(3.25) Apparently balanced 1;9 translocation provides further evidence for the role of euchromatin histone methyltransferase 1 (EuHMTase1) in the 9q34 subtelomeric deletion syndrome - *Dr Lionel Willatt, MS Bateman, PE Jenks, I Simoni, E Kerr, E Roper*

(3.26) The use of pyrosequencing to identify copy number variation of 16p11.2 in euchromatic variant carriers and the normal population - *Mrs Victoria Hall, VK Maloney, H White, T Liehr, M Volleth, JCK Barber*

(3.27) A case of segmental paternal uniparental disomy 14 which narrows the critical region to 14q32.33&-8594;qter - *Dr Celia Donaghue, K Mann, M Irving, P Amess, C Mackie Ogilvie, S Mohammed*

(3.28) An unusual finding of both a high-hyperdiploid and selective near-haploid clone in a patient with acute lymphoblastic leukaemia (ALL) - *Ms Amy Roe, NA Austin, SC Chatters, BDO De Oliveira, HK Kempski, HK Kempski*

(3.29) A retrospective audit of the CytoCell multiprobe CLL panel - *Ms Trisevgeni Kontou, B Czepulkowski*

(3.30) A patient with exostoses diagnosed as having Potocki-Shaffer syndrome. - *Ms Louise Monkman, E Ryan, R Morrison, M Whiteford, N Morriso), J M Colgan, G W Lowther*

(3.31) Insertion or inversion: that is the question - *Ms Kathryn Watts, B Pearce, V Maloney, T Boyle, M Collinson), J Barber*

(3.32) Microscope Vs Monitor: A comparison of chromosome analysis - *Dr Crista Illingworth, J Steer, E Maltby*

(3.33) Breakpoint mapping in apparently balanced translocations: comparing phenotypically abnormal individuals with controls - *Ms Julia Baptista, E Prigmore, SM Gribble, PA Jacobs, NP Carter, JA Crolla*

(3.34) A case of an Inherited Interstitial Deletion of 2p - *Ms Jennifer Corfield, D Delemege, C Delemege, E Roberts, J Crolla, J Barber, S Buston, P Lunt, T Davies*

(3.35) FISH for the Detection of 1p and 19q Deletions in Oligodendrogioma Paraffin Wax Sections - A Retrospective Study of 55 Cases. - *Mr Neil Atkey, SB Wharton, D Jellinek, EL Maltby*

(3.36) Testing for subtelomeric rearrangements using MLPA in children under five with developmental delay in the Grampian Region. - *Ms Caroline Clark, D Massie, J Brady, KF Kelly, JCS Dean*

(3.37) ECARUCA – an expanding resource for genotype-phenotype correlations of subtle cytogenetic and sub-microscopic chromosome abnormalities - *Dr John Barber, I Feenstra, D Koolen, A Rauch, S Ayme, A Schinzel, B de Vries, C van Ravenswaaij*

(3.38) Phenotypic Variation in a Family with a 14q31 Duplication and a Balanced Translocation - *Ms Abigail Palmer, L Morgan, E Smith, R Newbury-Ecob, L Burvill-Holmes, E Roberts, T Davies*

(3.39) Validation of ELUCIGENE QST*R, a QF-PCR Assay - *Mr Scott Higgins, C Stay, N Duxbury, C Smit*

(3.40) Microarray technology in healthcare: A cost analysis in idiopathic learning disability - *Dr Sarah Wordsworth, J Buchanan, R Regan, K Smith, V Davison, S Dyer, C Campbell, E Maher, SJL Knight, J Taylor*

(3.41) Translation of Genetics from Research into Clinical Practice; the Oxford Genetics Knowledge Park Model - *Dr Jenny Taylor, AOM Wilkie*

(3.42) Congenital fibrosis of the extraocular muscles associated with a de-novo interstitial deletion of chromosome 6 - *Dr Deborah Ruddy, AT Moore, R Palmer, R Ekong, E Spandoni, M Bitner-Glindzicz*

(3.43) Developmental delay and hyperactivity in a patient with the rare dup(16)(q11.2q13) anomaly characterised by array-CGH - *Mrs Sally Spillane, SM Morgan, DT Pilz, N Barrett, PW Thompson, SH Roberts*

(3.44) A method of checking for complete digestion of DNA samples to undergo microarray analysis using QF-PCR. - *Ms Rebecca Candlin, K Smith, M Crocker*

(3.45) Premature Chromosome Condensation Associated with Microcephaly - *Ms Louisa Wilson, D Stevenson, B Goldie, J Dean*

4. Counselling, Social, Policy and Education

(4.01) Needs assessment and review of services for people with inherited metabolic disease in the United Kingdom - *Dr Hilary Burton, S Sanderson, G Shortland, P Lee*

(4.02) Quality assuring the UK MCADD newborn screening pilot project - *Dr Sarah Ball, R Raynor), P Goddard, T Hutchin, N Dalton, J Bonham, G Besley, M Henderson, Y Foo, J Oerton, C Dezateux, A Green*

(4.03) Familial Breast Cancer Moderate risk Families: A Comparison of the Cancer Genetics Service for Wales (CGSW) and National Institute of Clinical Excellence (NICE) Guidelines: Screening and Cost Implications. - *Ms Liwsi Kim Protheroe, FMF Wadrup, Dr MTRogers, Dr AJ Murray*

(4.04) Creation of a DVD to assist patients in filling in cancer questionnaires - *Mr Gui Tran, B Shather, RG Jones, CE Chu*

(4.05) Men diagnosed with breast cancer: reflections on genetics - *Dr Rachel Iredale, J Hafner, S Sivell, K Brain, L France, B Williams, J Gray*

(4.06) Disseminating BRCA2 test results identified in the research context to relatives of deceased prostate cancer patients: a qualitative study of relatives' experiences. - *Dr Liz Ormondroyd, C Moynihan, A Ardern-Jones, S Davolls, C Foster, R Eeles, M Watson*

(4.07) Familial Cancer and Primary Care: An assessment of practice nurses' confidence and knowledge - *Ms Jennifer Wiggins, M Turnbull, MM Lees, LS Chitty*

(4.08) Genetic Counselling in Primary Care - *Ms Jan Moore, C Bennett, P Farndon, A Norman*

(4.09) Improving access to cancer genetics services in primary care - socioeconomic data from North Kirklees - *Dr Carol Chu, E Rowett, N Dharni, J Srinivasa, H Bhatt, M Day*

(4.10) How to survive genetic counselling and testing in cardiogenetics? - *Dr Irene M. van Langen, E Birnie, G.J. Bonsel, JP Van Tintelen, AAM. Wilde*

(4.11) Telling Stories: Understanding Real Life Genetics - *Mr Kevin McDonald, M Kirk, H Skirton, B Williams, K Kaur-Mann, E Tonkin*

(4.12) The Experiences of Predictive Testing for Myotonic Dystrophy - *Ms Anita Bruce,*

(4.13) What educators want: genetics and the nursing professions - *Dr Emma Tonkin, M Kirk*

(4.14) Who will be responsible for pharmacogenetics in the NHS? - *Ms Emily A Fargher, K Payne, C Eddy, K Tricker, RA Elliott, F Qasim, I Bruce, C Griffiths, K Moriarty, C Pearson, W Newman*

(4.15) Psychiatric Genetics: Challenging the Kraepelinian dichotomy - *Ms Flo Ticehurst, K Featherstone, P Atkinson, N Craddock*

(4.16) Exploring the role of patients' spiritual/religious beliefs around predictive genetic testing - *Ms Anna Clee, J Clayton-Smith, T Clancy*

(4.17) PEGASUS: Genetic education for front line health professionals - *Mrs Olive Downer-Robinson, PA Farndon*

(4.18) MAGNet: Medicine and Genomics Network, Research Capacity Building for Social Scientists - *Ms Flo Ticehurst, K Featherstone, M Gregory, R Gertz, G Haddow, N Kanellopoulou, P Saukko*

5. Other

(5.01) Outcome of preimplantation aneuploidy screening for high risk couples. - *Prof Joy D A Delhanty, A Mantzouratou, A Mania, K Fordham, A Doshi, P Serhal*

(5.02) Characterization of novel lysine-based dendrimers as gene delivery vehicles - *Ms Polina Ilina, A Egorova, A Kiselev, A Baranov, I Gur'janov, I Tarasenko, G Vlasov, V Baranov*

(5.03) Barriers to the integration of genetics into mainstream medical services - Ms H V Sleightholme, L Burges, D McCahon, D Cross, *F McGlynn, S Stewart, T Cole*

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