British Human Genetics Conference 2009

Monday 31 August (welcome reception at 19:00) – Wednesday 2 September

University of Warwick

Programme and Registration Booklet
Closing date for registration: 12 August 2009

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

- Complex Genetics
- Trisomy 21 Down Syndrome: Fifty years on – where are we now?
- Gene Patents: What have they got to do with us?
- Neurogenetics
- Chronic Leukaemia and Lymphoma
- Genomics and Phenomics
- Cross Boundary Working
- Cardiovascular Genetics
- Circulating nucleic acids; their use in non-invasive prenatal diagnosis and as markers for cancer
- Communication about genetic risk between children, young people and their parents
- Update of Cancer Genomics
- *Carter Lecture:*
  Professor Leena Peltonen on "Human Disease Genome: Lessons Learned from Special Populations"
- *BSHG Lecture:*
  Professor Stephen O'Rahilly on "Genetics of obesity and insulin resistance"
- *Debate:*
  "Personalised Genomics"
- *Young Researchers Forum*

LATE BREAKING ABSTRACTS
Deadline 7 August 2009
Details on website www.bshg.org.uk

SCIENTIFIC PROGRAMME

The BSHG Scientific Programme Committee welcomes you to the 2009 meeting of the British Society for Human Genetics at the **University of Warwick**. The SPC has compiled a wide-ranging programme comprising diverse symposia and outstanding UK and international speakers, including the Carter Lecture by Professor Leena Peltonen and the second BSHG Lecture by Professor Stephen O'Rahilly. Multiple parallel sessions ensure relevance to all constituent groups. The "Late Breaking Session" traditionally provides the hottest developments in human genetics research. The combination of excellent invited speakers and submitted abstracts should ensure another high quality meeting. Nevertheless your feedback is most important in shaping future meetings – please do take the time to complete the questionnaire and provide comments and suggestions for the 2010 meeting!

Eamonn Maher
Chairman, Scientific Programme Committee 2009

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

Posters are in the "Big Space". Poster presenters will be asked to stand next to their posters:

Odd numbers (i.e. 1.01, 1.03 etc) on TUESDAY 15:15 - 16:00

Even numbers (i.e. 1.02, 1.04 etc) on WEDNESDAY 10:00 - 10:45

Carter Lecture – Professor Leena Peltonen on "Human Disease Genome: Lessons Learned from Special Populations"

Second BSHG Lecture – Professor Stephen O'Rahilly on "Genetics of obesity and insulin resistance"

Debate: Personalised Genomics – is being held on Monday evening (20:00-21:00 in Conference Room, Arts Centre). Speakers are Dr Paul Pharoah and Dr Brent Zanke. This follows the "Welcome Reception" at 19:00.

YOUNG RESEARCHERS FORUM

A "Young Researchers Forum" meeting is scheduled for Wednesday lunchtime. This session provides an opportunity for trainees, PhD students and junior faculty to meet with senior academics and discuss issues relevant to junior academic researchers. Places are limited. In order to reserve a place (and lunch!) please notify Ruth Cole (bhgc2009@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 published as a supplement to the ***Journal of Medical Genetics***. Members unable to attend can order Abstract Booklets from the Conference Organiser ***before the meeting*** price £10.00.

CONFERENCE VENUE

This year sees us moving to a new and exciting venue – The University of Warwick (just outside Coventry). The plenary sessions, symposia and workshops are being held in the Arts Centre with the Registration Desks, Trade Exhibition, posters, internet café and catering in the "Big Space".

ACCOMMODATION

Accommodation has been arranged on campus close to the Arts Centre, Rootes Building and the "Big Space". The accommodation comprises single study bedrooms with en-suite facilities and single study bedrooms with shared bathroom facilities.

TRAVEL

The University of Warwick is located on the southern outskirts of Coventry. A map with more detailed travel information will be sent prior to the conference via email. Information is available on the University of Warwick website – http://www2.warwick.ac.uk/conferences/aboutus/gettinghere/77256_new_wc_campus_map.pdf

By Road Detailed information available at the above website

Car Parking Car Parking is FREE but please ensure that you park in Car Parks 7,8,15 otherwise a charge will be levied. To exit the car parks you will need a code. For delegates staying on campus – please ask for this when you get your room key and for Day Delegates – the code can be obtained from the Registration Desk.

By Rail The nearest railway station is Coventry. It is approximately 12 minutes by taxi from the station to the University. Alternatively, take the Travel West Midlands number 12 bus from the Warwick Road Bus Stop – again details on the above website.

By Air The nearest airport is Birmingham International, which has an excellent train link to Coventry, or approximately 30 minutes by road. Coventry airport is 15 minutes away, Nottingham East Midlands Airport is 1 hour away and Luton Airport is 2 hours away.

SOCIAL PROGRAMME

Welcome Reception – The meeting kicks off with a splendid Welcome Reception at 19:00 on Monday evening – making the most of our new venue this will be held in the Gallery of the Arts Centre. There will be wine, nibbles and, by popular request, a chocolate fountain!
You will also have chance to look at the "Our Kid" exhibition – "Our Kid" celebrates biomedical research – tracking the key achievements over the last 60 years and looking to the future of healthcare. Its centrepiece is a multimedia sculpture that displays over 3 hours of short films, including futuristic films made by teenagers, historic public health films and interview with experts.

Conference Party – We are very excited to be at the University of Warwick this year – voted best conference venue 2008 (Godiva Award) and so for the conference dinner, we will stay on campus to sample the delights of the University's award-winning banqueting chefs, who are renowned for creating imaginative food using the freshest seasonal ingredients. After a tiring day of engaging with world-leading science, visiting the trade stands in the "Big Space", we will repair to the Panorama Suite (with drinks reception in the Chancellor's Suite), Rootes Building. This is at the centre of the conference village and just a short meander from our bedrooms. Before dinner we will have a glass of Bucks Fizz to get the party off with a swing, and then will enjoy a delightful meal – featuring a choice of tranche of sesame-crusted salmon served with tomato, capsicum and white bean ragout or lentil and mushroom Wellington with peppercorn and capsicum sauce, both served with potatoes and seasonal vegetables, as well as starter, dessert, coffee and chocolates. Yum!

Sports Facilites
Available free to all delegates staying on campus – you can grab a swim, work out in the fitness suite or unwind in the sauna.

Cinema
Films are shown each evening (no details available yet) but they will be advertised in the Arts Centre. Tickets can be purchased from Box Office in Arts Centre.

REGISTRATION

Registration will be via the website – no other method of registration will be accepted. Once you have completed your on-line registration and pressed the submit button, this will then generate an acknowledgement sheet and give details of methods of payment. Please print off the acknowledgement sheet and send it **(together with payment)** to the administrative office in ***Birmingham before 12 August***. Do **NOT** resubmit, if you have any amendments please email the administrative office. Please ensure that you give your full contact details - postal address (including post code), telephone number and most importantly your ***email*** address.
A full-time student, who is not a member of the Society, may pay the lower registration fee as a Society member.

Please note:

- completing the on-line registration form commits the registrant to payment, whether or not they attend the conference. Any cancellations must be made (in writing or by email) by 12 August.
- we regret that fees cannot be reimbursed if you have to cancel your registration after 12 August.
- we **CANNOT** supply invoices in respect of registration fees

CONFIRMATION OF REGISTRATION

This year we are asking all delegates to collect their Conference Packs from the Registration Desks in the foyer to the "Big Space". The Registration desks will be open from 15:00-19:00 on Monday 31 August and from 07:30 on Tuesday 1 September. General information, including details of accommodation, map of campus and travel information, will be sent to you after the closing date of 12 August by **EMAIL** (please double check your email address when entering it in the registration details).
For those staying on campus, room keys need to be collected from the "Rootes Building" (situated near to the Arts Centre and the "Big Space")

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:	tbc
Rootes Reception (accommodation)	02476 5 22280 (07:00-23:00)
Security	02476 5 22083 (out of hours)

COUNCIL AND SOCIETY MEETINGS

BRITISH SOCIETY FOR HUMAN GENETICS
Council Meeting – 17:00 Monday 31 August - *Room SS0.10 - (Social Studies Building)*
Annual General Meeting -18:00 Tuesday 1 September – *Butterworth Hall, Arts Centre*

ASSOCIATION OF GENETIC NURSES AND COUNSELLORS
Members Meeting – 15:15 Tuesday 1 September – *Conference Room, Arts Centre*

FUTURE MEETINGS

BRITISH SOCIETY FOR HUMAN GENETICS

UNIVERSITY OF WARWICK	2010 - 30 August - 1 September
	2011 – dates to be confirmed

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: bhgc@bshg.org.uk Website: www.bshg.org.uk

PROGRAMME

Monday 31 August 2009

15:00	**REGISTRATION**
17:00-19:00	**BSHG – Council Meeting** – Room: SSO.10, Social Studies Building
19:00-20:00	**Welcome Reception** – Gallery, Arts Centre
20:00	**CHAIRMAN'S WELCOME** – Conference Room, Arts Centre

20:00-21:00 Debate – Personalised Genomics

Chair: Prof John Burn	**Conference Room, Arts Centre**
Debators are Dr Paul Pharoah and Dr Brent Zanke.	

Tuesday 1 September 2009

07:30 **REGISTRATION**

09:00-10:30 Concurrent Sessions

Clinical/Molecular		**Chair: Dr Julia Rankin / Mrs Rachel Butler**	**Butterworth Hall, Arts Centre**
09:00	(SP01)	A case of familial imprinting disorder (Beckwith-Wiedemann Syndrome) with germline mutation in NLRP2 (NALP2) - *Dr Esther Meyer, D Lim, S Pasha, LJ Tee, F Rahman, JRW Yates, CG Woods, W Reik, ER Maher*	
09:15	(SP02)	Prohormone convertase-2 haploinsufficiency causing obesity, insulin resistance and hypogonadotrophic hypogonadism - *Dr Rob Hastings, J Shield, J Davies, J Crolla, R Newbury-Ecob*	
09:30	(SP03)	A Novel Human Immunodeficiency Disorder associated with a Mutation in TRAC at the Fourth base of the Translation Termination Signal (*+1) - *Dr Neil Morgan, S Goddard, TS Cardno, F Rahman, A Ciupek, A Straatman-Iwanowska, S Pasha, G Anderson, A Huissoon, WP Tate, ER Maher*	
09:45	(SP04)	Loss of the metalloprotease ADAM9 leads to a novel form of cone-rod dystrophy in humans and retinal degeneration in mice - *Dr Carmel Toomes, DA Parry, L Bida, KV Towns, M McKibbin, CV Logan, M Ali, J Bond, S Saveliev, R Chance, SG Jacobson, K Springell, M Adams, E Banin, CA Johnson, D Sharon, CP Blobel, EN Pugh, EA Pierce, CF Inglehearn*	
10:00	(SP05)	Homozygous Loss-of-Function Mutations in the Dopamine Transporter (DAT), SLC6A3 Cause Infantile Parkinsonism-Dystonia (IPD) - *Dr Manju Kurian, J Zhen, S-Y Cheng, Y Li, S Mordekar, P Jardine, NV Morgan, E Meyer, L Tee, S Pasha, E Wassmer, B Assmann, SJR Heales, P Gissen, MEA Reith, ER Maher*	
10:15	(SP06)	A new syndrome presenting with epilepsy, ataxia, sensorineural deafness, and tubulopathy (EAST syndrome) - *Dr Angus Dobbie, S Feather, D Bockenhauer, H Stanescu, E Sheridan*	

Cross Boundary Working		**Chair: Ms Cath King / Ms Laura Boyes**	**Theatre, Arts Centre**
09:00	(SP07)	The role of the Neurofibromatosis Specialist Advisor - *Ms Carolyn Smyth*	
09:18	(SP08)	The role of the Huntington's Disease Association in supporting families – *Ms Cath Stanley*	
09:36	(SP09)	Inherited Prion Disease - *Miss Michele Gorham, S Mead, I Eastwood, L Ford*	
09:54	(SP10)	Supporting families affected by a single gene condition - *Mrs Grace MacLeod*	
10:12	(SP11)	Epidermolysis Bullosa – *Ms Jackie Denyer*	

Cytogenetics	Chair: Mr Rodger Palmer / Mrs Katie Waters	Conference Room, Arts Centre
09:00	(SP12)	Resolution at Ring Breakpoint: Role in phenotypic correlation and predicting natural history - *Dr Serena Nik-Zainal, PE Cotter, L Willatt, EW O'Brien*
09:15	(SP13)	Detection of chromosomal abnormalities in single cells from epithelial cell lines by array comparative genomic hybridisation - *Miss Thalia Mamas, S SenGupta, H Sultan, D Dafou, T Gordon, J. C. Harper*
09:30	(SP14)	Low level aneuploidy mosaicism detected by array CGH - *Mrs Mary Glancy, R Wang, S Edwards, P Stubbs, C Vince, M Bitner-Glindzicz, V Puthi, R Palmer*
09:45	(SP15)	XLMR patients screened using exon resolution X chromosome array CGH - *Mr Lee Silcock, D McMullan, L Brueton, D Lim, V Davison*
10:00	(SP16)	Copy Number Alterations of Xp11.22-p11.23 in Females Associated with Developmental Delay and Other Anomalies - *Dr Jill Urquhart, FH Sharkey, S Beri, R Giorda, L Gaunt, S Lynch, GCM Black, J Clayton-Smith*
10:15	(SP17)	Copy Number Variant (CNV) analysis: fishing for the truth amid red herrings - *Ms Elaine Doherty, K Claxton, J Love, D Love*

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

11:15-12:45 Concurrent Symposia

Complex Genetics	Chair: Dr Alexandra Blakemore / Dr Christine Patch	Theatre, Arts Centre
11:15	(SP18)	Genetics of Autism – *Dr Jeremy Parr (Newcastle)*
11:45	(SP19)	Parkinson's disease: simple answers to complex problems? - *Prof Nicholas Wood*
12:15	(SP20)	Genetic predisposition to prostate cancer: rare and common variants. Implications for targeted prostate cancer screening - *Dr Ros Eeles*

Trisomy 21 Down Syndrome: Fifty years on – where are we now?		Chair: Prof Maj Hulten / Dr Jonathan Waters Conference Room, Arts Centre
11:15	(SP21)	Introduction – *Prof Maj Hulten (Warwick)*
11:30	(SP22)	Folic acid metabolism and Down syndrome - *Prof Lucia Migliore*
11:45	(SP23)	A system biology approach to Down Syndrome: identification of Notch/Wnt dysregulation in stem cells with age - *Dr Ilaria Bellantuono, C Cairney, G Sanguinetti, E Ranghini, MC Nostro, A Bhattacharya, CN Svendsen, WN Keith*
12:00	(SP24)	Trisomy 21, Down Syndrome and the origin of Alzheimer's disease - *Dr Kevin Moffat, S Patel, M Hulten*
12:15	(SP25)	CNV in Down Syndrome heart conditions – *Dr Alex Blakemore (London)*
12:30	(SP26)	Advances in quality of life for individuals with Down syndrome and their families – *Prof Sue Buckley (Southsea)*

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12:45-14:15 **LUNCH and POSTER VIEWING**

13:00-14:00 Gene patents: What have they got to do with us?

Chair: Mr Stuart Hogarth — **Conference Room, Arts Centre**

Join *Trevor Cook* (Biotech expert in Patent law, at Bird and Bird) *Michael Hopkins* (Researcher in Science and Technology Policy at the University of Sussex) and *Stuart Hogarth* (Researcher from the King's College London) for an overview of recent events in the field followed by an open discussion.

14:15-15:15 Concurrent Symposia

Neurogenetics (sponsored by the NF Association) — **Chair: Prof Dorothy Trump / Dr Susan Huson, Theatre, Arts Centre**

14:15	(SP27)	Rare disorders of the RAS/MAPK pathway - *Dr Bronwyn Kerr (Manchester)*
14:45	(SP28)	Neurofibromatosis Type 1: from gene, to animal model, to therapy – *Prof Luis Parada (Dallas)*

Chronic Leukaemia and Lymphoma — **Chair: Dr Jon Strefford / Mr Mike Griffiths, Conference Room, Arts Centre**

14:15	(SP29)	The Genetics of Chronic Lymphocytic Leukaemia – *Dr Stephan Stilgenbauer (Ulm, Germany)*
14:45	(SP30)	The Molecular Pathogenesis of Follicular Lymphoma – *Dr Jude Fitzgibbon (London)*

15:15-16:00 **TEA AND POSTER SESSION** – "Odd Numbers"

15:15 **Association of Genetic Nurses and Counsellors** – Members Meeting – Conference Room, Arts Centre

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

Chair: Dr Frances Flinter / Dr David Barton — **Butterworth Hall, Arts Centre**

16:00	(SP31)	The Primordial Growth Disorder 3-M Syndrome Connects Ubiquitination to the Cytoskeletal Adaptor Obscurin-Like 1 - *Mr Daniel Hanson, PG Murray, A Sud, SA Temtamy, M Aglan, A Superti-Furga, SE Holder, J Urquhart, EN Hilton, FDC Manson, P Scambler, PE Clayton, GCM Black*
16:15-17:00		**Late Breaking Research**

17:00-18:00 The Carter Lecture

Chair: Dr Frances Flinter — **Butterworth Hall, Arts Centre**

(SP32)	Human Disease Genome: Lessons Learned from Special Populations – *Dr Leena Peltonen (Cambridge)*

18:00 **BRITISH SOCIETY FOR HUMAN GENETICS – Annual General Meeting** – Butterworth Hall, Arts Centre

19:30 **Conference Party** – University of Warwick (Panorama Suite with Drinks Reception in Chancellor's Suite - Rootes Building)

Wednesday 2 September 2009

08:00 **REGISTRATION**

09:00-10:00 Concurrent Sessions

Clinical/Molecular	**Chair: Dr William Newman / Dr Ian Frayling**	**Butterworth Hall, Arts Centre**
09:00	(SP33)	Oligogenic inheritance in long QT syndrome and hypertrophic cardiomyopathy - *Dr John Dean, C Brown, D Walker, C Clark, K Kelly, P Broadhurst, A-M Choy, W Lam, V Murday, D Goudie, J Grieve*
09:15	(SP34)	Polygenic risk variants for type 2 diabetes susceptibility modify age at diagnosis in monogenic HNF1A diabetes - *Dr Sian Ellard, H Lango Allen, S Johansson, JK Hertel, B Shields, H Ræder, K Colclough, A Molven, TM Frayling, PR Njølstad, AT Hattersley, MN Weedon*
09:30	(SP35)	Null Mutations in LTBP2 cause Primary Congenital Glaucoma - *Dr Manir Ali, M McKibbin, A Booth, DA Parry, SA Riazuddin, JF Hejtmancik, SN Khan, S Firasat, M Shires, DF Gilmour, K Towns, D Azmanov, I Tournev, S Cherninkova, H Jafri, Y Raashid, C Toomes, L Kalaydjieva, S Riazuddin, CF Inglehearn*
09:45	(SP36)	Aspirin prevents cancer in Lynch syndrome - *Prof John Burn, A-M Gerdes, J Mathers, J-P Mecklin, F Macrae, G Moeslein, M-L Bisgaard, R Ramesar, D Eccles, E Maher, G Evans, P Morrison, DT Bishop*

Cytogenetics	**Chair: Ms Una Maye / Mr Dominic McMullan**	**Conference Room, Arts Centre**
09:00	(SP37)	Evaluation of technologies for detection and quantification of foetal DNA in maternal plasma in a clinical setting - *Dr Kalliroi Stergianou, K Carpenter, K Ocraft, G Cross, M Hultén, T Parkin*
09:15	(SP38)	International Standardised Cytogenomic Array (ISCA) Consortium: an approach to the design, implementation and reporting of constitutional oligo array-cgh - *Dr John Crolla, DH Ledbetter, CL Martin, S Aradhya, SJL Knight, K Smith, K Kok, J Vermeesch*
09:30	(SP39)	The use of large control datasets: Assessing CNVs from high resolution microarray analysis in a diagnostic setting. - *Dr Kristin Abbott, I Simonic*
09:45	(SP40)	Array CGH detection of imbalance for cancer predisposition genes or late-onset conditions – how should we proceed? - *Dr Joo Wook Ahn, SN Mohammed, S Holden, C Mackie Ogilvie*

Counselling	**Chair: Ms Gillian Bromilow / Ms Amanda Barry**	**Theatre, Arts Centre**
09:00	(SP41)	Identification of a de novo BRCA1 mutation in a woman with young onset bilateral breast cancer - *Mrs Emma Edwards, C Yearwood, J Sillibourne, D Baralle, D Eccles*
09:15	(SP42)	Exploring family communication after receiving BRCA1/2 results - *Dr Caroline Dancyger, J Smith, C Jacobs, S Michie*
09:30	(SP43)	Whose test is it anyway? Factors influencing the decision to carry out genetic testing in childhood in a series of cases - *Dr Ruth Newbury-Ecob, A Kendall, M Williams, P Rudd, R Lumsden, C King*
09:45	(SP44)	21 years, 260 predictive tests for Huntington's disease: was it worth it? - *Dr Sheila Simpson*

10:00-10:45 **COFFEE AND POSTER SESSION** – "Even Numbers"

10:45-12:45 Symposium: Genomics and Phenomics (including the BSHG Lecture)

Chair: Dr Rob Elles / Prof Maj Hulten		**Butterworth Hall, Arts Centre**
10:45	(SP45)	Microfluidics and next-generation sequencing for non-invasive prenatal diagnosis - *Prof Yuk Ming Dennis Lo (Hong Kong)*
11:15	(SP46)	Reverse Phenotyping: Towards an integrated (epi)genomic approach to complex phenotypes and common disease - *Prof Stephan Beck (London)*

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11:45-12:45 BSHG Lecture

Chair: Dr Rob Elles / Prof Maj Hulten		Butterworth Hall, Arts Centre
11:45	(SP47)	Genetics of obesity and insulin resistance - *Prof Stephen O'Rahilly (Cambridge)*

12:45-14:00 **LUNCH and POSTER VIEWING**

13:00-14:00 **Young Researchers Forum**
Room: SSO.10 – Social Studies Building

14:00-16:00 Concurrent Symposia

Cardiovascular Genetics	Chair: Dr Edward Blair / Prof David Wilson	Theatre, Arts Centre
14:00	(SP48)	The genetic basis of TGFß cell signalling and vascular disease – *Prof Richard Trembath (London)*
14:30	(SP49)	Genetic Causes of Vascular Malformations - *Prof Miikka Vikkula (Brussels)*
15:00	(SP50)	Clinical and Molecular Genetics of Arrhythmogenic Right Ventricular Cardiomyopathy – *Prof William McKenna (London)*
15:30	(SP51)	Monogenetic causes of hypertension – *Prof Fiona Karet (Cambridge)*

Circulating Nucleic Acids; their use in non-invasive prenatal diagnosis and as markers for cancer		Chair: Prof Sian Ellard Conference Room, Arts Centre
14:00	(SP52)	New, less-invasive genetic diagnostic technologies from a potential user's standpoint – *Dr Ian Frayling (Cardiff)*
14:30	(SP53)	Non-invasive pre-natal detection of Down syndrome: an update and overview of the use of new genetic technologies - *Dr Helen White (Salisbury)*
15:00	(SP54)	Non-invasive pre-natal diagnosis using cell free fetal nucleic acids in maternal plasma: current uses and implications for wider implementation – *Dr Lyn Chitty (London)*
15:30	(SP55)	Potential role of cell free DNA for mutation detection in patients with cancer – *Dr Ruth Board (Manchester)*

Communication about genetic risk between children, young people and their parents		Chair: Ms Jennifer Wiggins / Mr Mark Longmuir Cinema, Arts Centre
14:00	(SP56)	What do children know about genetics and medical terminology generally – *Dr Fiona Ulph (Manchester)*
14:30		What do children from families affected by genetic conditions know – *Dr Jane Coad (Bristol)*
15:00		Parents experiences and views on talking to children about genetic conditions – *Ms Gill Plumridge (Birmingham)*
15:30		Families' coping with genetic risk communication: implications for clinical practice – *Dr Alison Metcalfe (Birmingham)*

Update of Cancer Genomics – UK Cancer Cytogenetics Group		Chair: Prof Christine Harrison Studio, Arts Centre
14:00	(SP57)	Characterisation of structural variation in 24 breast cancer genomes using paired-end sequencing on the Illumina Genome Analyser – *Dr Phil Stephens (Cambridge)*
14:40	(SP58)	Integrative genomic, transcriptomic and functional genomic analysis identifies novel therapeutic targets for breast cancers - *Dr Jorge Reis-Filho (Sutton,Surrey)*
15:20	(SP59)	Genome-wide analysis of chromosomal alterations in Chronic Lymphocytic leukemia - *Dr Jonathan Strefford (Southampton)*

16:00 **End of Conference**

Poster presentations

1. Clinical genetics

(1.01) The Role of Fibrillin-1 Mutations in Families with Dominantly Inherited Ectopia Lentis. - *Dr Anne Child, D Ahnood, P Comeglio, L Ocaka, D Charteris, G Arno*

(1.02) MELAS masquerading as Alport Syndrome: A family with renal impairment and deafness in the presence of a MELAS (m.3243A>G) mutation - *Dr Chirag Patel, L Foggensteiner, J Prothero*

(1.03) South West of Britain Audit of the Investigation and Management of Patients with 22q11 Deletion - *Dr Chirag Patel, C Mercer, N Harper, H Cox*

(1.04) The lower limb radiology of distal myopathy due to the S60F myotilin mutation - *Dr Alisdair McNeill, D Birchall, V Straub, L Goldfarb, P Reilich, M Walter, N Schramm, H Lochmuller, P Chinnery*

(1.05) SNAP29 mutations in two siblings: a report of the third family described with cerebral dysgenesis, neuropathy, ichthyosis and keratoderma (CEDNIK) syndrome - *Dr Helen Stewart, E Sprecher*

(1.06) Genotype-phenotype correlations in VHL exon deletions - *Dr Alisdair McNeill, E Rattenberry, R Barber, F MacDonald, P Killick, ER Maher*

(1.07) Incidence and combinations of Niemann-Pick Type C mutations with clinical data - *Mrs Jackie Imrie, S Knight, L Heptinstall, G Besley, M Vanier, E Wraith, E Jacklin*

(1.08) Trisomy 18 mosaicism: 2 more cases and important learning points - *Dr Siddharth Banka, K Metcalfe, J Clayton-Smith*

(1.09) evaluation of a cascade screening method for adpkd - *Miss Kathryn Banner, L Foggensteiner, F McGlynn*

(1.10) Phenotypic Spectrum of PLA2G6-related disorders: Infantile, Childhood and Adult Onset Presentation of Phospholiapse Associated Neurodegeneration (PLAN) - *Dr Manju Kurian, P Rehal, NV Morgan, P Gissen, F MacDonald, ER Maher*

(1.11) Developing a research register for patients with 6q24-related transient neonatal diabetes mellitus - *Dr Anna Lehmann, JP Shield, D Robinson, DJG Mackay, IK Temple*

(1.12) Rhombencephalosynapsis in the West Midlands - *Dr Alisdair McNeill, PM Cox, L Brueton, D Williams*

(1.13) Anterior versus posterior tooth agenesis - report of new cases - *Prof Emilia Severin, C. Albu, D. Albu, D. Stanciu*

(1.14) The clinical, therapeutic and molecular genetic investigation of Birt-Hogg-Dube syndrome - *Dr Derek Lim, PK Rehal, GA Kirby, F MacDonald, ER Maher*

(1.15) Rhombencephalosynapsis: review of case reports - *Dr Alisdair McNeill, C Ellison*

(1.16) Genetics in the Ancient World – was Bes the god of Down syndrome in Pharaonic Egypt? - *Dr Shane McKee*

(1.17) Beckwith Wiedemann syndrome in an infant with a mosaic chromosome 11p11.2 inversion found at prenatal diagnosis - *Dr Michael Gattas, E Algar, N Martin*

(1.18) Are mutations in the X-linked UBE1 gene present in diagnostic referrals for spinal muscular atrophy? - *Mrs Jo McCauley, Y Hughes, N Masand, S Abbs*

(1.19) The outcomes of antenatally detected isolated cardiac anomalies - *Dr Moira Blyth, C Rounding, PA Boyd, D Wellesley*

(1.20) A Review of DMD families: Is our approach to carrier testing effective - *Dr Ruth McGowan, CM Longman, DW Wilcox, SJ Joss*

(1.21) Identification of 5 novel mutations causing autosomal recessive bestrophinopathy (ARB) - *Miss Yusrah Shweikh, AE Davidson, I Miller, P Brown, AR Webster, FDC Manson, GCM Black*

(1.22) A Third Case of SAMS Syndrome - *Dr Angus Dobbie, C Harrison*

(1.23) Duplication of Chromosome 3p23.3p25.1 results in a Prader willi-like phenotype. - *Dr Shincy John, D Shears, K Smith, J Hurst, U Kini*

(1.24) Inherited Cardiac Conditions – current issues and future directions - *Dr Liz Ormondroyd, S Oates, K Thomson, E Blair, H Watkins*

(1.25) DYSCERNE: Results from a pilot of the electronic Dysmorphology Diagnostic System (DDS) - *Dr Ruth Day, S Gardner, P Griffiths, K Strong, C Harrison, D Donnai, B Kerr, K Metcalfe, H Brunner, B Dallapiccola, K Devriendt, M Krajewska-Walasek, N Philip, J Clayton-Smith*

(1.26) Numerous naevi in 17q21.31 deletion make it an important differential diagnosis for cardiofaciocutaneous syndrome - *Dr Emma Burkitt Wright, D Donnai, B Kerr, J Clayton-Smith*

(1.27) Study into Pallister-Killian syndrome in Great Britain - *Dr Moira Blyth, D Baralle, A Lucassen, IK Temple*

(1.28) Lung agenesis, congenital heart abnormalities and thumb anomalies: Four new cases and review – is there a distinct recessive syndrome? - *Dr Rob Hastings, A Donaldson, D Harding, R Liebling, A Hayes, A Kraus, S Joss, P Turnpenny, S Smithson*

(1.29) Pierpont syndrome: delineation of the phenotype - *Dr Emma Burkitt Wright, D Donnai*

(1.30) An audit of BRCA mutation detection rate in relation to Manchester score and additional risk factors, including ER negativity - *Dr David Wilkinson, K Oakhill, H Martin, S Waller, V Burkitt, P Seymour, J Whittaker, J Paterson*

(1.31) BRCA testing in Liverpool: an audit. - *Dr Caroline Pottinger, E. Lyulcheva, D. Cairns, C. Kightly, K.L. Greenhalgh*

POSTER PRESENTATIONS

(1.32) Aneuploidy in fetus' with anomaly on prenatal ultrasound in the Welsh population - *Dr Caroline Pottinger, D Robinson*

(1.33) Brothers with mental retardation, premature balding, small genitalia, small acra and small patellae: an X-linked disorder? - *Dr Anthony Vandersteen, RC Hennekam*

(1.34) A focus group study of health care professionals involved in the care of families in Wales affected by sudden arrhythmic death syndrome (SADS) - *Dr Bethan Henderson, D Kumar, M Nicol, K Brain, R Iredale, A Clarke*

(1.35) Congenital non-progressive cerbellar atrophy associated with CACNA1A gene mutation - *Dr Swati Naik, K Pohl, D Josifova*

(1.36) Effect of mutation type on myocardial function in adult patients with Marfan syndrome - *Dr Gavin Arno, A Kiotsekoglou, P Comeglio, MJ Mullen, N Bunce, DK Nassiri, AJ Camm, GR Sutherland, A Younis, AH Child*

(1.37) A Case of an Interstitial del(3)(p21.1p14.3) detected by ArrayCGH - *Miss Kath Smith, D S Johnson, E L Maltby*

(1.38) Methylation analysis of 79 patients with growth restriction reveals novel patterns of methylation change at imprinted loci - *Dr Claire Turner, DJG Mackay, JLA Callaway, L Docherty, RL Poole, H Bullman, M Lever, B Castle, EC Kivuva, PD Turnpenny, SG Mehta, S Mansour, JH Davies, IK Temple*

(1.39) Endocrine Genes- have we improved the fit? - *Mrs Susan Stewart, N Gittoes, PM Stewart, W Arlt, T Cole*

(1.40) Genetics in mainstream medicine- best practice review - *Mrs Susan Stewart, J Watkinson, J Franklin, E Woodward, T Cole*

(1.41) Developing a National Specialist Ehlers Danlos Syndrome Service. - *Miss Jessica Bowen, J.H Tocher, N.E Crawford, G.J Sobey, A. Dalton*

(1.42) Effects of stress during pregnancy on growth and adult glucose tolerance: are there implications for prenatal screening? - *Prof Joanna Poulton, S Malik, CAR Boyd, KJ Morten*

(1.43) Moebius syndrome, intestinal atresia, thumb abnormalities, hearing loss and learning difficulties: a new autosomal dominant syndrome? - *Dr Victoria Harrison, D Rourke, R Williams, U Kini*

(1.44) Vertical Transmission of Ring Chromosome 18 - *Dr Victoria Harrison, M Fraser, E Stevenson, M Crocker, E Blair*

(1.45) SDHB: further evidence of the pathogenicity of the Ser163Pro variant associated with phaeochromocytoma and a Cowden-like phenotype - *Dr Nicola Cooper, T Cole, E Rattenberry, P Rehal, PM Stewart, S Stewart, J Benn*

(1.46) Monozygotic twins discordant for Pseudohypoparathyroidism type 1b - *Dr Emma Baple, A Albanese, IK Temple, DJ Mackay*

(1.47) The clinical phenotype of FG syndrome: An algorithm for diagnostic testing - *Dr Robin Clark, J M Graham, M J Friez, J J Hoo, K L Jones, C McKeown, J B Moeschler, F L Raymond, R C Rogers, C E Schwartz, A Battaglia, M J Lyons, R E Stevenson*

(1.48) Demonstration project- GP Geneticists and a model of care for patients with 'common' genetic conditions in Scotland. - *Dr Helen Gregory, A-M Taylor, Z Miedzybrodzka, J Berg*

(1.49) The genetic investigations of neuronal migration disorder and cortical dysplasia - *Dr Neeti Ghali, S Naik, D Josifova*

(1.50) Should we use the term "selective inactivation of an X chromosome"? - *Dr Angus Dobbie*

(1.51) Detailed Neuropathological findings in a case of Galloway-Mowat Syndrome - *Dr Lisa Robertson, M Parker, M Al-Adnani, TS Jacques, TS Jacques*

(1.52) Impact of expert radilogical opinion on clinical management in suspected skeletal dysplasia - *Dr Angharad Roberts, A Offiah, E Wakeling*

(1.53) Mutations in CNNM4 Cause Jalili Syndrome, Consisting of Autosomal Recessive Cone-Rod Dystrophy and Amelogenesis Imperfecta - *Dr David Parry, AJ Mighell, W El-Sayed, RC Shore, IK Jalili, H Dollfus, A Bloch-Zupan, R Carlos, IM Carr, LM Downey, KM Blain, DC Mansfield, M Shahrabi, M Heidari, P Aref, M Abbasi, M Michaelides, AT Moore, J Kirkham, CF Inglehearn*

(1.54) Audit of Marfan Syndrome Families in The West of Scotland Clinical Genetics Service - *Dr Catherine McWilliam, V Murday*

(1.55) Lowry-Wood Syndrome: An uncommon or less recognized condition - *Dr Dragana Josifova, K Becker, M Bober*

(1.56) Clouston Syndrome due to de novo GJB6 mutations in two unrelated British patients - *Dr Laura Yates, C Healey, A Taylor, S Natarajan*

(1.57) Cardiomyopathy: To test or not to test? - *Miss Elizabeth Hardman, Ramdeep Bajwa, M Frenneaux, H Cox*

(1.58) Association of Neurofibrosarcoma, Dermatofibrosarcoma protruberans and Pilomatricoma in a family suggests a rare tumour predisposition syndrome - *Dr Sarju Mehta, RM Graham, AM Logan, H Baillie-Johnson, E Reid*

(1.59) A 3q29 microdeletion syndrome consisting of a Diamond-Blackfan Anaemia phenotype, mild dysmorphisms and developmental delay - *Dr Virginia Clowes, A Clarkson, L Willatt, M Murray, M Gattens, S-M Park*

(1.60) Audit on Genetics Ward Round- Wessex Experience - *Dr Meena Balasubramanian, A L Collins*

(1.61) Risk-Reducing Operations in Healthy BRCA Mutation Carriers - *Dr Anne-Bine Skytte, A Gerdes, M.K Andersen, L Sunde, K Brøndum-Nielsen, M Waldstrøm, S Koelvraa, D Crüger*

(1.62) What is Coffin-Siris syndrome? - *Dr Caroline Pottinger, M Tassabehji, E Sweeney, J Clayton-Smith*

(1.63) A clinical and molecular study of 20 females with Xq deletions or rearrangements - *Dr Catherine Mercer, K Lachlan, S Thomas, P Jacobs*

(1.64) Previously unreported feature in IMAGe Syndrome: Case discussion and review of published literature - *Dr Meena Balasubramanian, D S Johnson*

(1.65) Blue rubber bleb naevus syndrome- severe presentation in a neonate - *Dr Meena Balasubramanian, D S Johnson, G J Sobey*

(1.66) The National Down Syndrome Cytogenetic Register: 20 Years on – where are we now? - *Prof Joan Morris*

(1.67) Familial Chilblain Lupus due to a heterozygous mutation in the SAMHD1 gene - *Dr Mohnish Suri, J Ravenscroft, Y Crow*

(1.68) Regional variations in prenatal diagnosis of Down syndrome in England and Wales - *Prof Joan Morris*

(1.69) Expanding the Alport Syndrome service at Guy's - *Dr Helen Storey, J.C. Pagan, J. Prescott, F. Flinter, S. Abbs*

(1.70) The National Down Syndrome Cytogenetic Register: Research findings from 20 years of data - *Prof Joan Morris, D Mutton*

(1.71) The genetic basis of congenital talipes equinovarus: a genome wide association study in the population of Vanuatu - *Dr Guoqing Liu, P Giblin, J Milburn, J Inglis, D Shaw, A Cardy, L Sharp, Z Miedzybrodzka*

(1.72) Insights into the developmental basis of congenital talipes equinovarus from pma mice - *Dr Zosia Miedzybrodzka, N Lindstrom, R McIntosh, S Duce, H Sapsford, JA Chudek, C Tickle, N Vargesson, JM Collinson*

2. Molecular Genetics

(2.01) A Novel Homozygous 62bp Insertion In ECM1 Gene Causes Lipoid Proteinosis In A Multigenerational Pakistani Family - *Dr Abdul Hameed, M Nasir, M Ajmal, A Latif*

(2.02) From primer design to sequence analysis- a pipeline tool for use in the molecular genetic diagnostic laboratory - *Miss Elaine Doherty, A Vaughan, D Prosser, J Love, D Love*

(2.03) A Novel Genomic Region Upstream of the Human Caveolin 1 Gene is Involved in the Pathophysiology of Late-Onset Alzheimer's Disease. - *Dr Mina Ohadi, A Mirabzadeh, M Zarif Yeganeh, Y Heshmati*

(2.04) Dysregulation of Genomic Imprinting in IUGR Placentas - *Mr Andreas Diplas, L Lambertini, MJ Lee, R Sperling, YL Lee, J Wetmur, J Chen*

(2.05) Diagnostic analysis of Norwegian BRCA mutations using Sequenom MALDI TOF mass spectrometry - *Dr Helen Griffin, J Eden, J Burn, A Curtis*

(2.06) Calreticulin as a Promising Candidate in the Etiopathophysiology of Schizoaffective Disorder. - *Dr Mina Ohadi, A Mirabzadeh, M Olaad Nabi*

(2.07) Association of variants in the Sirtuin 1 (SIRT1) gene with severe obesity - *Mr Stephen Clark, M Falchi, B Olsson, D Meyre, J Andersson, B Balkau, M Marre, J Tichet, H Y Wong, L Carlsson, P Froguel, A J Walley*

(2.08) Development of a simplified and sensitive microarray based mutation detection method with optimised probes for Cystic Fibrosis - *Ms Marta Paolucci, J Ragoussis, L Lonie, M Macek, A Seller*

(2.09) A screening assay for BRCA1 and BRCA2 mutations based on nonsense mediated decay - *Dr Helen Griffin, R Brown, M Santibanez-Koref, J Burn, A Curtis*

(2.10) Endogenous spartin (SPG20) is recruited to endosomes and lipid droplets and interacts with the ubiquitin E3 ligases AIP4 and AIP5 - *Dr Virginia Clowes, TL Edwards, HTH Tsang, JW Connell, CM Sanderson, JP Luzio, E Reid*

(2.11) Molecular Genetic Investigation of Autosomal Recessive Cryptogenic Infantile Spasms (ARCIS): Evidence of Clinical and Genetic Locus Heterogeneity - *Dr Manju Kurian, NV Morgan, S Pasha, E Meyer, F Rahman, G Vassallo, P Gissen, E Wassmer, ER Maher*

(2.12) Prognosis for splicing factor PRPF8 Retinitis pigmentosa and correlation between human and yeast phenotypes - *Prof Chris Inglehearn, KV Towns, T Kipioti, V Long, M McKibbin, P Ehsani, K Springell, M Kamal, RS Ramesar, DA Mackey, AT Moore, R Mukhopadhyay, AR Webster, GCM Black, J O'Sullivan, EA Pierce, J Beggs*

(2.13) Diagnostic Tests for Costello Syndrome and Cardio-Facio-Cutaneous Syndrome – Three Years Service Experience - *Dr Jenny Shorto, B Kerr, B Tang, R Elles*

(2.14) Mutations of the SBDS gene in Shwachman-Diamond syndrome - *Dr Joanna Brock, J Shorto, S McCormack, RG Elles, MJ Schwarz*

(2.15) Myotonic Dystrophy Type 1 TP-PCR: A strategy to avoid false negatives. - *Dr Natalie Trump, A.L Sharif, G.S Cross*

(2.16) Urine epithelial cells as an alternative to muscle biopsies in the detection of mitochondrial DNA mutations - *Miss Mary Sweeney, CE Woodward, EE Mudanohwo, S Rahman, MG Hanna, MB Davis*

(2.17) Probe and high resolution melt analysis for Cystic Fibrosis and Hereditary Haemochromatosis using the Corbett Rotor-Gene 6000 - *Dr James Drummond, E Thompson, T Legerton, T Elsey, J Whittaker*

(2.18) An update on the BCR/ABL kinase domain mutation analysis service in CML patients at the West Midlands Regional Genetics Laboratory - *Miss Charlene Crosby, E Perrott, D Clavering, E Ormshaw, J Borrow, J Mason, S Akiki, M Griffiths*

(2.19) Sensitive and quantitative detection of KIT D816V in patients with systemic mastocytosis using allele specific real time PCR and digital PCR - *Dr Helen White, D Scott, VJ Hall, K Waghorn, R Sanders, CA Foy, NCP Cross*

(2.20) The Locus Reference Genomic (LRG) DNA sequence format for LSDBs - *Dr Raymond Dalgleish*

(2.21) Screening for mutations causing episodic ataxia type 1 (EA1) and 2 (EA2) - *Dr Robyn Labrum, S Rajakulendran, MG Sweeney, R Bevan, MG Hanna, MB Davis*

(2.22) Identification of a CYP27A1 splice mutation in a family presenting with pulverulent cataracts and developmental delay - *Ms Sarah Joyce, R Bourkiza, H Patel, M Chan, E Meyer, MA Reddy, ER Maher*

(2.23) Identification of large deletions and duplications in the CFTR gene using Multiple Ligation-dependent Probe Assay (MLPA), including one novel duplication and two novel deletions. - *Dr Mike Bulman, J Brock, E Brownsell, S Smith, S McCormack, M Adaway, C Charlton, M Schwarz*

(2.24) Application of Multiple Displacement Amplification in the Diagnostic Laboratory - *Mr Thomas Cullup, S Ganjavian, P Renwick, S Abbs*

(2.25) Initiation codon mutation in CRYBB1 associated with autosomal recessive nuclear pulverulent cataract - *Dr Esther Meyer, F Rahman, J Andorf, S Pasha, NV Morgan, RC Trembath, EM Stone, AT Moore, ER Maher*

(2.26) Single Exon Rearrangements of GTP Cyclohydrolase I in Two Families with Dopa-Responsive Dystonia - *Dr James Polke, H Ling, MG Sweeney, A Haworth, H Houlden, P Foskett, N Wood, A Lees, M Davis*

(2.27) A strategy for identification of trisomy 21 in embryos tested for monogenic disorders - *Dr Heema Patel, P Renwick, A Lashwood, C Ogilvie*

(2.28) Preimplantation Genetic Haplotyping for monogenic disorders: 141 clinical diagnostic cycles demonstrating a robust and efficient alternative to direct mutation testing on single cells. - *Dr Pamela Renwick, J Trussler, E Ronaldson, A Lashwood, P Braude, C Ogilvie*

(2.29) Genetic heterogeneity in congenital hypothyroidism - *Dr Hakan Cangul, NV Morgan, S Pasha, GA Kirby, M Karkucak, E Eren, T Yakut, H Saglam, W Hogler, O Tarim, TG Barrett, ER Maher*

(2.30) BRAF testing as a low-cost means of pre-screening before MLH1 and MSH2 sequencing - *Mrs Jenny Myring, H Mugalassi, R Hussein, S Palmer-Smith, R Butler*

(2.31) An intronic polymorphic deletion in the PTEN gene: Implications for molecular diagnostic testing - *Miss Stacey Sandell, DJ Bunyan, JF Harvey*

(2.32) A Potential Case of Revertant Mosaicism in Familial Retinoblastoma - *Miss Kelly Price, E A Price, K Kolkiewicz, R Patel, E Rosser, D Bunyan, Z Onadim*

(2.33) A years experience of using MS-MLPA supported by MS-HRM for detection of 11p15 methylation abnormalities in diagnostic testing for Beckwith-Wiedemann and Silver-Russell syndromes - *Dr Cath Willoughby, R Scott, S Mansour, R Taylor*

(2.34) The first two years of a diagnostic service for Marfan syndrome and other Type I Fibrillinopathies at the SW Thames Regional Genetic Laboratory - *Mrs Louise Kiely, S Butler, S Cottrell, G Arno, A Child, R Taylor*

(2.35) Identification of a new locus for non-syndromic recessive optic atrophy on chromosome 20 - *Mr James Poulter, DF Gilmour, IM Carr, AJ Churchill, E Sheridan, CP Bennett, HC Ardley, M Ali, CF Inglehearn, C Toomes*

(2.36) Identification and RQ-PCR monitoring of CML patients with rare variant BCR-ABL transcripts - *Mr Christopher Bowles, J Mason, S Akiki, M Griffiths*

(2.37) Identification of aberrant imprinting at 14q32.2 in a patient giving rise to a upd(14)pat-like phenotype - *Dr Natalie Trump, M Suri, N Shannon, A.L Sharif, GS Cross*

(2.38) Pontocerebellar hypoplasia caused by mutations in RARS2; the first British case - *Dr Julia Rankin, R Brown, WB Dobyns, J Harington, J Patel, M Quinn, G Brown*

(2.39) Our inheritance, our future: review of the Genetics Laboratories Yorkshire and Northern Collaboration (GenLYNC) in Leeds and Newcastle - *Dr David Cockburn, D Bourn, R Charlton, A Curtis, C Chu, M Wright*

(2.40) MLPA analysis to detect large deletions and duplications of SDHB, SDHC and SDHD: interesting findings since the establishment of a diagnostic service - *Ms Eleanor Rattenberry, R Woodward, N Bradshaw, L Izatt, A Wren, S Aylwin, J Bell, P Rehal, E Maher, F Macdonald*

(2.41) Results of Parkinson disease genetic analysis; spectrum of mutations detected and prevalence of dominant vs recessive mutation - *Mr Pierre Foskett, R. Sud, O. Taiwo, V.S. Gibbons, M.G. Sweeney, N.W. Wood, A. Haworth, M.B. Davis*

(2.42) TGFBR2 mutation analysis in Marfanoid patients with ascending aortic aneurysm - *Dr Gavin Arno, P Comeglio, S Hasso, AH Child*

(2.43) A four year audit of MSH6 screening and tumour tissue analysis results in the Edinburgh molecular genetics laboratory - *Mrs Louise Slater, N Williams, N Andrew, P Westwood, D. U Baty, M.E. Porteous, J.P. Warner, T Bradley*

(2.44) Patients with microdeletions upstream of the FOX transcription factor gene cluster at 16q24.1 have similar phenotypes to those with whole-cluster microdeletions, suggesting the presence of upstream critical regulatory element - *Dr Charles Shaw-Smith, R Sandford, S Bhatt, M Storer, Z Xia, P Sen, V Martin, L Willatt, I Simonic, J Paterson, S Mehta, S Gribble, E Prigmore, D Rajan, T Fitzgerald, L Shaffer, S-W Cheung, NP Carter, CX Langston, P Stankiewicz*

(2.45) Patient heterozygous for Friedreich ataxia GAA repeat expansions in the full and premutation size ranges – could this suggest a diagnosis of Friedreich ataxia? - *Dr Andrew Purvis, F White, F Coyne, R Mountford*

(2.46) Cystic Fibrosis Neonatal Screening - An Update and Interesting Results - *Dr Louise Bourdon, D Patel, M Pereira, SJ Payne*

(2.47) Mitochondrial DNA depletion syndromes – molecular diagnosis and future developments - *Dr Conrad Smith, J Evans, N Ashley, A Seller, C Fratter, J Poulton*

(2.48) A false negative result for Myotonic Dystrophy type 2 using triple primed PCR - *Mr Nicholas Parkin, O Wood, D Robinson, J Harvey*

(2.49) Mutations in the Rab Escort Protein (REP-1) gene in UK Choroideremia patients - *Mr James O'Sullivan, N Hart-Holden, T Colclough, A O'Grady, S Ramsden, R Elles, GCM Black*

(2.50) Complex Imprinting Disorders - *Dr Rebecca Poole, J Bliek, IK Temple, DJG Mackay*

(2.51) Extended analysis of possible PLA2G6 deletions in 2 patients with Infantile Neuroaxonal Dystrophy - Miss Danielle Crompton, PR Pauline Rehal, MK Manju Kurian, FM Fiona MacDonald

(2.52) Elucidating the aetiology of Prader-Willi syndrome: deletion of the HBII-85 class of snoRNA is associated with hyperphagia, obesity and hypogonadism - *Mr Adam de Smith, C Purmann, RG Walters, RJ Ellis, SE Holder, MM Van Haelst, AF Brady, UL Fairbrother, M Dattani, JM Keogh, E Henning, GSH Yeo, S O'Rahilly, P Froguel, IS Farooqi, AIF Blakemore*

(2.53) Genetic analysis of MEVF mutations in Iranian Familial Mediterranean Fever patients - *Dr Mandana Hasanzad, M Hasanzad, N Almadani, A Kariminejad, H Najmabadi*

(2.54) Autosomal recessive primary microcephaly: Importance of complete mutation analysis - *Dr Sally Halsall, H Martin, V Burkitt, P Seymour, G Woods, R Treacy*

(2.55) Stickler syndrome and the vitreous phenotype: correlation with mutations in COL2A1 and COL11A1 - *Dr Allan Richards, H Martin, K Oakhill, H Rai, B Treacy, J Whittaker, A Poulson, M Snead*

(2.56) Discrepancies in CAG repeat results for Huntington's disease: a European Huntington's Disease Network analysis from Jan 2004 to Dec 2007 - *Dr Oliver Quarrell, O Handley, C Dumoulin, GB Landwehrmeyer*

(2.57) Definitive Diagnosis of Glycogen Storage Disease (GSD) by Molecular Analysis - *Dr Nick Beauchamp, A Dalton, R J Kirk, S Tanner, M J Sharrard*

(2.58) Clinical and Molecular Characterisation of HCM in Egypt: - *Dr Heba Kassem, M Saber Ayad, H Farza, I Olivotto, F Cecchi, MH Yacoub, BA HCM Consortium*

(2.59) Molecular genetic testing for LMNA-Related Dilated Cardiomyopathy - *Dr Judith Hudson, E Graham, C Healey, M Buddles, K Bushby, D Bourn*

(2.60) Evaluation of a diagnostic service for haemophilia A incorporating the LightScanner high resolution melt analysis system - *Dr Kate Sergeant, V Wilson, J Hanley, K Talks, A Curtis*

(2.61) Developmental delay MLPA screening: an audit and review of the service delivered by the Edinburgh laboratory - *Miss Tara Azam, FH Sharkey, N Williams, AK Lampe, WWK Lam, MEM Porteous, DR Fitzpatrick, JP Warner*

(2.62) The development of a diagnostic service for familial exudative vitreoretinopathy (FEVR) - *Dr Eilidh Jackson, D Cockburn, R Charlton, D Gilmour, C Toomes*

(2.63) Investigating somatic mosaicism in FAP - *Miss Natalie Bibb, Z Allen, L Sutherland, SJ Payne*

(2.64) Diagnosis of Zellweger Syndrome by Biochemical, Enzymatic and Molecular Analysis: a Case Study - *Mr Richard Kirk, C A Reed, S Mehta, S Olpin, N Manning, A Dalton*

(2.65) Assessment of candidate imprinted genes in the human term placenta - *Dr Caroline Daelemans, ME Ritchie, S Abu-Amero, IM Sudbery, P Stanier, MS Forrest, P Deloukas, S Tavare, GE Moore, I Dunham*

(2.66) HHEX-IDE genotype associated with birthweight in South Asians - *Miss Laura Towns, M Attard, A J Walley, U L Fairbrother*

(2.67) Genes in the ACAD family show preliminary evidence of association with polygenic obesity. - *Dr Julian Asher, AJ Walley, P Froguel, P Froguel, AIF Blakemore*

(2.68) Epidermal Growth Factor Receptor and K-Ras mutations in patients with Non Small Cell Lung Cancer - *Mr Hood Mugalaasi, J Davies, L Medley, D Talbot, R Butler, R Brito,*

(2.69) Homozygosity mapping as a novel tool to identify patients with Wolcott-Rallison syndrome among infants with permanent neonatal diabetes - *Dr Jayne Minton, O Rubio-Cabezas, A-M Patch, SF Flanagan, E Edghill, K Hussain, AT Hattersley, S Ellard*

(2.70) A standardised framework for the validation and verification of diagnostic molecular genetic tests - *Mr Chris Mattocks, G Matthijs, M Morris*

(2.71) The development of a sequencing service to detect mutations in the FRMD7 gene causing X-linked idiopathic infantile nystagmus - *Mr Lewis Darnell, M G Thomas, J G Barwell, P Vasudevan, I Gottlob, G S Cross*

(2.72) Implementation of and initial results from a UKGTN CPVT1 Mutation Screening Service - *Dr Emma Howard, W Newman, CJ Garratt, RG Elles, AJ Wallace*

(2.73) CNVs in Obesity: Uncovering a New Level of Genomic Variation - *Miss Julia Sarah El-Sayed Moustafa, LJM Coin, AJ de Smith, RG Walters, JL Buxton, JE Asher, D Meyre, C Dina, P Froguel, P Froguel, AIF Blakemore*

(2.74) Assignment of trisomic genotypes using the Sequenom iPLEX MALDI TOF assay reveals transmission disequilibrium of two SNP loci in the Type VI Collagen gene cluster associated with CHD in Down's Syndrome. - *Ms Anne Trewick, P Froguel, LJM Coin, AIF Blakemore*

(2.75) Gene expression profiling of human oocytes and embryo blastocysts - *Miss Georgia Kakourou, S Jaroudi, S Gotts, A Doshi, P Serhal, JC Harper, JD Delhanty, SB SenGupta*

(2.76) Identification and validation of novel genomic CNVs associated with Type 2 diabetes - *Dr Jessica Buxton, LJM Coin, RG Walters, J Andersson, AJ de Smith, JS El-Sayed Moustafa, R Sladek, P Froguel, AIF Blakemore*

(2.77) Massively parallel sequencing of the X chromosome coding exons for the identification of novel X-linked disease genes - *Dr Patrick Tarpey, R Smith, D Turner, L Mamanova, C Scott, A Whibley, R Bowman, N Rahman, L Biesecker, G Black, J Campbell, R Stevenson, CE Schwartz, PA Futreal, FL Raymond, MR Stratton*

POSTER PRESENTATIONS

3. Cytogenetics and Molecular Cytogenetics

(3.01) Double partial deletions of proximal 21q and distal 22q in an adult mentally retarded patient derived from a maternal translocation - *Dr Farkhondeh Behjati, S Ghasemi Firouzabadi, K Kahrizi, H Yazdan, S Arzhangi, S Banihashemi, M Ataei Kachouei, H Najmabadi*

(3.02) Identification of cryptic genomic imbalances in both de novo and familial balanced chromosome rearrangements using aCGH - *Miss Georgina Hall, L Cooper-Charles, L Silcock, N James, J Morton, I Aligianis, D McMullan, V Davison*

(3.03) Bleomycin Stress Testing for Ataxia Telangiectasia - *Miss Josie Hayes*

(3.04) MYC amplification in double minute chromosomes in a case of acute myeloid leukaemia - *Mr Robert Vince, A Wood, J Burbridge, S Ryley, K Waters*

(3.05) A novel microdeletion of chromosome 10q24.32 in a patient with features similar to renal-coloboma syndrome - *Miss Anna Benson, U Maye, S Smith, R Skillbeck, A Fryer*

(3.06) Multicolour banding (Mbanding) as an adjunct to aCGH or karyotype analysis, for the characterisation of complex chromosome rearrangements - *Miss Susan Bint, AF Davies, R Oke, Z Docherty, C Mackie Ogilvie*

(3.07) Characterisation of a de novo complex chromosome abnormality in a child with global delay – the need for high resolution aCGH. - *Mr Richard J Ellis, E Hawes Collins, C Brooks, S Mountford, N Canham*

(3.08) Recurrent imbalance detected in unrelated individuals with different abnormal phenotypes - *Dr Joo Wook Ahn, SE Walsh, C Mackie Ogilvie*

(3.09) Investigation of patients with unexplained learning difficulties by array CGH – Bristol Genetics Laboratory experience. - *Miss Siobhan Toomey, HM Martin, CS Wragg, E Roberts, T Davies*

(3.10) On the origin of trisomy 21- the evidence from oocyte and polar body analysis - *Prof Maj Hulten, E Iwarsson*

(3.11) On the origin of constitutional aneuploidy: Ovarian aneuploidy mosaicism - *Prof Maj Hulten, S Patel*

(3.12) A Paternally Inherited Unbalanced Insertion of Chromosome 1: Importance of FISH Follow-up for aCGH - *Dr Sandra Edwards, M Glancy, R Wang, R Palmer, E Rosser*

(3.13) Partial Monosomy of Distal 2q and Partial Trisomy of Distal 2p in An Adult Mentally Retarded Patient, Derived from a Paternal Inversion - *Ms Saghar Ghasemi, F Mojahedi, K Kahrizi, H Darvish, M Ataei Kachoui, G Bahrami Monajemi, H Najmabadi, F Behjati*

(3.14) Report of a ring Chromosome 18 in a mentally retarded woman referred for prenatal diagnosis - *Mr Iman Bagherizadeh, A Saberi, N Mohammadkhani, F Mojahedi, Z Hadipour, F Hadipour, A Saremi, Y Shafeghati, F Behjati*

(3.15) An unusual case of mosaicism for two structural rearrangements of chromosome 12 in a phenotypically normal male. - *Miss Gemma Forsyth, J Cunningham, G Hislop, L McMahon, D Goudie, N Pratt*

(3.16) A de novo t(5;16)(16q23.3;p15.32) translocation generating partial trisomy 16q23.3>16qter and p15.32>pter deletion in two siblings - *Dr Khaled Abu-Amero, A Hellani*

(3.17) Chromosomal copy number variation; non-penetrance can be associated with large cytogenetically visible imbalances but becomes more likely when gene content is low - *Mr Mark Bateman, JCK Barber*

(3.18) Detection of subtle BCL2 insertion into IGH: Another case highlighting the limitations inherent in the design of certain commercial FISH probes - *Mr Michael Dyson*

(3.19) Unusual Complexity of a Philadelphia-positive Biphenotypic Leukaemia revealed by Array CGH - *Mr Steve Chatters, H Kempski*

(3.20) Performance Comparison of the Affymetrix SNP6.0 and the Affymetrix Cytogenetics 2.7M Microarrays for Cytogenetic Analysis - *Dr Lorraine Gaunt, MN Needham, G Scopes, L Gaunt*

(3.21) Development of a BAC-FISH test to detect FGFR1 rearrangements in patients with 8p11 Myeloproliferative Syndrome. - *Miss Lauren Jones, D McMullan, A Ely, S Jeffries, N James, L Silcock, M Griffiths*

4. Counselling, Social, Policy and Education

(4.01) Progress in development of national inherited cardiovascular conditions (ICC) services in the light of rapid scientific advances - *Ms Alison Hall, H Burton, C Alberg, A Stewart*

(4.02) Towards implementation of non-invasive prenatal diagnosis in the NHS - *Ms Alison Hall, H Burton, C Wright*

(4.03) DYSCERNE: An Online Educational Tool for Dysmorphology - Examination of a fetus with congenital abnormality - *Dr Ruth Day, S Gardner, P Griffiths, C Harrison, K Strong, D Donnai, B Kerr, K Metcalfe, H Brunner, B Dallapiccola, K Devriendt, M Krajewska-Walasek, N Philip, J Clayton-Smith*

(4.04) How, what and when to tell the children? Factors that influence the communication between parent and child about being a carrier of a balanced chromosome translocation. - *Dr Rachel Iredale, S Owens*

(4.05) DYSCERNE: Developing clinical management guidelines for dysmorphic conditions - *Mrs Pamela Griffiths, K Strong, S Gardner, R Day, C Harrison, D Donnai, B Kerr, K Metcalfe, H Brunner, B Dallapiccola, K Devriendt, M Krajewska-Walasek, N Philip, J Clayton-Smith*

(4.06) What is so special about genes? Exploring the wider implications of genetics and genomics: A Report of the Society for Genomics Policy and Population Health (SGPPH) Spring Conference 2009 - *Ms Alison Hall, L Jader, M Hopkins, S Hogarth, H Burton, A Metcalfe*

(4.07) Nowgen's new Darwin-inspired molecular genetics workshop for school students - *Dr Leah Holmes, K Mathieson, P Finegold, HR Middleton-Price, D Donnai*

(4.08) Development of molecular biology laboratory training courses for health care professionals at the Nowgen Centre, The North West Genetics Knowledge Park - *Dr Mark Leech, F Salway, F Jury, D Carthy, L Gaunt, S Hamilton, A Wallace, R Elles, A Devereau, D vanGent, J Crolla, M Bottomley, K Mathieson, W Ollier, M Yuille, D Donnai, H Middleton-Price*

(4.09) Effective strategies to generate public interest and dialogue in genetics - *Miss Kate Mathieson, P Finegold, LE Holmes, MJ Leech, D Donnai, HR Middleton-Price*

(4.10) Ethical Issues in prevention of genetic diseases in Iran - *Dr Seyed Hamid Jamaldini, VR Yassaee, SMH Ghaderian*

(4.11) A New Insight in Human Genetics in Iranian Students - *Dr Mandana Hasanzad*

(4.12) Influences on Young People's Attitudes to Genetics and Health - *Dr Nicola V Taverner, K Madden, R Iredale*

(4.13) Development of a set of family-friendly leaflets on rare chromosome disorders for use by families and professionals - *Ms Prisca Middlemiss, SL Wynn, M Hulten*

(4.14) Facily communcation regarding genetic testing: why, what, when and why? - *Mrs Kim Chivers-Seymour, A Lucassen, J Addington-Hall, CL Foster*

(4.15) Carers' participation in decision making in a Huntington's disease management clinic - *Miss Donna Davies, AE Rosser, S Sarangi*

5. Cancer Genetics

(5.01) Familial Cholangiocarcinoma: Case Report and Review. - *Dr Alex Henderson, P Jonas, J Burn, P Douglas*

(5.02) The Functional Role of CASP8 D302H and Other Apoptosis Gene Variants in Breast Cancer - *Dr Sushila Rigas, M Parry, M Reed, A Cox*

(5.03) Patient Open Days at the Cancer Genetics Service for Wales - *Dr Rachel Iredale, A Murray*

(5.04) Molecular Genetic Analysis of Gliomas (LOH and MGMT) for Optimisation of Treatment - *Dr Paula Waits, K McDonagh, H Swayer, S Burton-Jones, M Greenslade, K Hopkins, H Norman, S Love, M Williams*

(5.05) Population Based Survey of Cancer Risks in Chromosome 3 Translocation Carriers - *Dr Emma Woodward, AB Skytte, DG Cruger, ER Maher*

(5.06) Analysis of germline variants in CDH1, IGFBP3, MMP1, MMP3, STK15 and VEGF in familial and sporadic renal cell carcinoma - *Dr Christopher Ricketts, MP Zeegers, J Lubinski, ER Maher*

(5.07) The IMPACT study - Identification of men with a genetic predisposition to prostate cancer: targeted screening of BRCA1/2 mutation carriers and controls - *Miss Elizabeth K Bancroft, AV Mitra, EC Page, RA Eeles*

(5.08) Expression profiling of schwannomas - *Prof Gareth Evans, K Hadfield, Y Hey, S Pepper, D Trump, W Newman*

(5.09) Screening guidelines for familial paediatric cancer susceptibility - *Miss Zakia Mansurah, ST Khan, PC Vasudevan, JG Barwell*

(5.10) Spectrum and incidence of BRCA1 and BRCA2 mutations in the Republic of Ireland – An Audit - *Ms Mary Higgins, A Crowley, T McDevitt, N Cody, M Meany, C de Baroid, M Adams, E Berkeley, C Nolan, R Clarke, M Farrell, P Daly, A Green, D Barton*

6. Other

(6.01) Chromosomal Distribution of Human Mitochondrial Genes: A Hypothesis. - *Mr Arvind Arya, R Reena, A Kumar, P K Sharma*

(6.02) An audit of waiting time for contact with All Wales Medical Genetics Service (AWMGS) Prenatal Genetic Services for obstetric patients referred from South Wales with abnormal prenatal genetic test results in 2006 and 2007 - *Dr Mark Tein, A Edwards, MD Robinson, AM Procter*

(6.03) The Baby Bio Bank of London - *Dr Sayeda Abu-Amero, G Moore, L Regan, R Rai, S White, A Miller, W Lo, M Johnson, K Nicolaides, C Williamson, D Peebles, J Whittaker*

(6.04) Designing information for children at risk of cancer predisposition syndrome by using Affective Design - *Miss Joanna Zlotkowska, M Ahmed, T Cassidy, C E Chu*

(6.05) Customer satisfaction in software development - *Mr Ali Qayyum*

(6.06) The First GEN2PHEN Pilot Project: Documenting the bioinformatics requirements of clinical scientists - *Dr Michael Cornell, A Devereau*

(6.07) Whole-genome linkage and association scan in primary, non-syndromic vesicoureteric reflux (VUR) and reflux nephropathy - *Prof Judith Goodship, R Darlay, P Charoen, A Stewart, AS Gullett, HJ Lambert, S Malcolm, SA Feather, AS Woolf, RB Kenda, HJ Cordell*

(6.08) New era for Regional Genetics Laboratories at Guy's - *Dr Stephen Abbs, Z Docherty, M Jackson, S Mohammed*

British Human Genetics Conference 2009

Monday 31 August – Wednesday 2 September

University of Warwick

ON-LINE REGISTRATION

No other method of registration will be accepted – you MUST register on line

Please complete all the sections, check them and then press the submit button – this will then generate an acknowledgement sheet and give details of methods of payment. Do **NOT** resubmit, if you have any amendments please email the administrative office in Birmingham. Please ensure that you give your full contact details – postal address (including post code), telephone number and most importantly your ***email*** address – this will be used to send out information to you prior to the conference. Delegates Packs are being handed out at the Conference.

Please print off the acknowledgement sheet and send (**TOGETHER with PAYMENT**) to:

British Human Genetics Conference 2009 Registration
British Society for Human Genetics, Clinical Genetics Unit, Birmingham Women's Hospital, Edgbaston, Birmingham. B15 2TG

If payment is being made by the Trust, please clearly indicate on registration acknowledgement sheet and send a copy to BSHG office.

PLEASE NOTE

- Completing the on-line registration form commits the registrant to payment, whether or not they attend the conference. Any cancellations must be made (in writing or by email) by 12 August.
- We regret that fees paid cannot be reimbursed if you have to cancel your registration after 12 August.
- We **CANNOT** supply invoices in respect of registration fees

CONFERENCE COSTS

Registration Fees	**Member**	**Non-Member**
Full conference	£310.00	£420.00
Individual days (Tuesday or Wednesday)	£155.00	£210.00
Accommodation (per night)		
Standard Single	£35.00	
En-suite single	£52.00	
Conference Dinner (University of Warwick)	£42.00	
Car Parking FREE (please park in designed car parks 7,8,15 – code needed for exit)		

Delegates' Packs - This year we are asking all delegates to collect their Conference Packs from the Registration Desks in the foyer to the "Big Space". The Registration desks will be open from 15:00-19:00 on Monday 31 August and from 07:30 on Tuesday 1 September. General information, including details of accommodation and map of campus, will be sent to you after the closing date of 12 August by **EMAIL**. For those staying on campus, room keys need to be collected from the "Rootes Building" (situated near to the Arts Centre and the "Big Space").

CLOSING DATE FOR REGISTRATION:

12 August 2009

http://www.bshg.org.uk/registration/bhgc2009.asp

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