

British Human Genetics Conference 2011

Monday – Wednesday 5 - 7 September

University of Warwick

Programme and Registration Booklet

Closing date for registration: 14 August 2011

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

- Fanconi Anaemia
- Reflections on the Psychosocial and Ethical Aspects of Emerging Technologies
- Genetics and Genomics in the Developing World
- Approaches to Complex Disease
- Cancer Pathogenesis and Genomic Architecture
- Variable Phenotypes in Genetic Conditions - Implications for Genetic Counselling
- Somatic Mutation Detection in the Context of Drug Prescription
- Neurodevelopmental Disorders
- Animal Models of Genetic Disorders
- Clinical Management of Genetic Disorders
- Neurogenetics
- *Carter Lecture:*
Professor Veronica Van Heyningen "Making Eyes: Genes and Networks"
- *BSHG Lecture:*
Professor Michael Stratton "Evolution of the Cancer Genome"
- *Great Debate:*
"This house believes that Synthetic Biology should be used to design new forms of life"
- *Novel Insights into the Genetics of Acute Lymphoblastic Leukaemia and Myeloproliferative Neoplasms Workshop*
- *Treatment Strategies for Genetic Disorders Workshop*
- *Medico-Legal Workshop*
- *Education Sessions on Clinical Trials and Next Generation Sequencing*
- *Trade Exhibitor - Seminars*

LATE BREAKING ABSTRACTS
Deadline 12 August 2011
Details on website www.bshg.org.uk

SCIENTIFIC PROGRAMME

Welcome to the 2011 British Society for Human Genetics Conference. The Scientific Programme Committee has been delighted by the high quality abstracts submitted to the meeting reflecting a vibrant genetics research and service delivery environment in the UK. We hope that the programme delivered by outstanding UK and international speakers, including the Carter Lecture by Professor Veronica van Heyningen and the BSHG Lecture by Professor Mike Stratton provide a stimulating and enjoyable meeting. This year we have introduced BSHG prizes for the best posters and are keen that as many people take the opportunity to view these and speak with the presenters at the designated session on Tuesday afternoon. 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 2012 meeting.

William Newman
Chairman, Scientific Programme Committee 2011

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

Posters are in Butterworth Hall and the Mead Gallery. ALL Poster presenters will be asked to stand next to their posters on **TUESDAY 6 September from 14:00 – 15:00**

Carter Lecture – Professor Veronica Van Heyningen on “Making Eyes: Genes and Networks”

4th BSHG Lecture – Professor Michael Stratton on “Evolution of the Cancer Genome”

The Great Debate – Monday evening (19:00-20:00 in The Theatre, Arts Centre). The motion for the debate is “*This house believes that Synthetic Biology should be used to design new forms of life*”

Debaters are Prof Alison Metcalfe, Dr Chris French, Prof Paul Martin and Dr Ainsley Newson
Come and join in what promises to be a very lively debate – don’t forget to indicate on the registration form if you wish to attend.

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 we are again at The University of Warwick (just outside Coventry). The entire conference is being held in the Arts Centre.

ACCOMMODATION

Accommodation has been arranged on campus close to the Arts Centre and Rootes Building. The accommodation comprises single study bedrooms with en-suite facilities (Arthur Vick or Jack Martin halls of residence) and single study bedrooms with shared bathroom facilities (Rootes Residences).

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/howtofindus/>

By Road Detailed information available at the above website

Car Parking Car Parking is FREE but please ensure that you park in Car Parks 6,7,8a,15 otherwise a charge will be levied. (Car Park 7 is nearest to the Arts Centre, Car Park 6 nearest to accommodation and Car Park 15 the furthest away) 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

Wine Tasting – Join us for the wine tasting from 18:00-19:00 on Monday 5 September in both Butterworth Hall and the Mead Gallery. This event is sponsored by some of our Exhibitors and features a range of wines; red, white, as well as charming rosé wines. Don't forget to talk to the exhibitors (and get your card validated for the chance to win a fantastic prize - while you are there! Further details will follow in due course)

Conference Dinner & Dance – Following the rip-roaring success of the conference party last year, we will be staying on campus at the University again in 2011. We'll be starting off with a Kir Royal or a J20 Splash, before a delicious meal featuring a choice of mains including supreme of chicken with truffle and pistachio or baked Cornish tuna with citrus butter and quinoa crust for meat eaters or the vegetarian alternative of vegetable cassoulet with roasted basil infused haloumi. The meal also includes a choice of starters and dessert, and is followed by coffee and single-origin chocolates. Following dinner, a live band will perform all of your dance favourites on centre stage. Be sure to attend the Conference Dinner & Dance - it will be an evening to remember! We look forward to seeing you there!

Sports Facilities

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 are advertised in the Arts Centre (02476 524524). Tickets can be purchased from Box Office in Arts Centre.

REGISTRATION

Registration is via our 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 14 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 14 August.
- we regret that fees cannot be reimbursed if you have to cancel your registration after 14 August.
- we **CANNOT** supply invoices in respect of registration fees
- a **late booking fee of £30** will be levied this year

CONFIRMATION OF REGISTRATION

This year we are asking all delegates to collect their Conference Packs from the Registration Desks in the Arts Centre Upper Foyer. The Registration desks will be open from 17:00-20:00 on Sunday 4 September - from 07:30 on Monday, Tuesday and Wednesday 5-7 September. General information, including details of accommodation, map of campus and travel information, will be sent to you after the closing date of 14 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 opposite to the Arts Centre). Room keys will need to be shown for breakfast.

CPD APPROVAL

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

EMERGENCY TELEPHONE NUMBERS

Conference Office	02476 5 23222
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 – 09:00 Monday 5 September – Room: SS0.10, Social Sciences Building

Annual General Meeting -17:30 Monday 5 September – Room: The Theatre, Arts Centre

Editors Meeting - 07:30 Tuesday 6 September – Room: National Grid, Arts Centre

ASSOCIATION OF GENETIC NURSES AND COUNSELLORS

Members Meeting – 15:15 Monday 5 September – Room: Conference Room, Arts Centre

CLINICAL MOLECULAR GENETICS SOCIETY

Members Extraordinary General Meeting – 12:45 Tuesday 6 September – Room: The Theatre, Arts Centre

OTHER MEETINGS

Clinical Genetics Trainees Meeting – 13:15-14:00 Monday 5 September – Room: The Studio, Arts Centre

Genetic Counsellor Registration Board Meeting – 13:15-14:00 Monday 5 September – Room: SS0.09, Social Sciences Building

Lead Clinicians Meeting – 12:45-13:15 Tuesday 6 September – Room: SS0.09, Social Sciences Building

FUTURE MEETINGS

BRITISH SOCIETY FOR HUMAN GENETICS

UNIVERSITY OF WARWICK 2012 – 17-19 September

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

Sunday 4 September 2011

17:00-20:00 **REGISTRATION** - Upper Foyer, Arts Centre

Monday 5 September 2011

07:30 **REGISTRATION**

09:00-11:00 **BSHG – Council Meeting** – Room: SSO.10, Social Sciences Building

11:25 **CHAIRMAN'S WELCOME** – Room: The Theatre, Arts Centre

11:30-13:15 Symposium: Fanconi Anaemia

Chair: Mr David McDowell / Prof Gareth Evans		Room: The Theatre, Arts Centre
11:35	(SP01)	The A to P of Fanconi anaemia genetics - <i>Prof Christopher Mathew (London)</i>
11:55	(SP02)	Clinical Aspects of Fanconi Anaemia - <i>Dr Marc Tischkowitz (Cambridge)</i>
12:15	(SP03)	Screening for Fanconi Anaemia patients with hand anomalies: an evidence based approach to diagnosis and genetic testing – <i>Dr Ruth Newbury-Ecob (Bristol)</i>
12:35	(SP04)	Clinical Management of patients with Fanconi Anaemia – <i>Dr Stefan Meyer (Manchester)</i>
12:55	(SP05)	Fanconi Anemia Genes and Breast/Ovarian cancer risks - <i>Prof Nazneen Rahman (Sutton)</i>

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

13:15-14:00 **Clinical Genetics Trainees Meeting**
Room: The Studio, Arts Centre

13:15-14:00 **Genetic Counsellors Registration Board Meeting**
Room: SSO.09, Social Sciences Building

14:15-15:15 Concurrent Sessions

Molecular		Room: The Theatre, Arts Centre
Chair: Prof Eamonn Maher/ Dr Ann Curtis		
14:15	(SP06)	Exome sequencing identifies CCDC8 mutations in 3-M syndrome, suggesting CCDC8 contributes in a pathway with CUL7 and OBSL1 to control human growth - <i>Dr Jill Urquhart, D Hanson, PG Murray, J O'Sullivan, S Daly, SS Bhaskar, LG Biesecker, M Skae, C Smith, T Cole, J Kirk, K Chandler, H Kingston, D Donnai, PE Clayton, GCM Black</i>
14:30	(SP07)	First implication of STRA6 mutations in isolated anophthalmia, microphthalmia and coloboma - <i>Prof Andrew Green, J Casey, R Kawaguchi, M Morrissey, H Sun, P McGettigan, J Nielsen, J Conroy, R Regan, P Tormey, M Ni Chroinin, B Kennedy, SA Lynch, S Ennis</i>
14:45	(SP08)	Exome sequencing identifies a DYNC1H1 mutation in a large pedigree with dominant Charcot-Marie-Tooth disease - <i>Dr Sian Ellard, MN Weedon, R Caswell, R Hastings, W Xie, K Paszkiewicz, T Antoniadi, M Williams, C King, L Greenhalgh, R Newbury-Ecob</i>
15:00	(SP09)	Whole-Exome Sequencing Identifies FAM20A Mutations as a Cause of Amelogenesis Imperfecta and Gingival Hyperplasia Syndrome - <i>Mr James O'Sullivan, CC Bitu, SB Daly, JE Urquhart, MJ Barron, SS Bhaskar, H Martelli-Ju'nior, P Eleuterio dos Santos Neto, MA Mansilla, JC Murray, RD Coletta, GCM Black, MJ Dixon</i>

Clinical/Cancer		Room: The Cinema, Arts Centre
Chair: tbc		
14:15	(SP10)	Increased chromosome breakage in lymphocytes exposed to ionizing radiation can be used to distinguish FANCD1 caused by biallelic BRCA2 mutations from more common forms of Fanconi anaemia - <i>Dr Simon Holden, G Stewart, P Byrd, C Turnbull, I Kesterton, F McDonald, P Fulton, C Ridout, A Davidson, H Bye, CG Mathew, AM Taylor</i>
14:30	(SP11)	Mainstreaming initiatives in the North East: innovation, leadership and a new role for tertiary services - <i>Dr Paul Brennan, O Claber, T Brennan, L O'Dair, J Oliver, S Ball, M Miller</i>
14:45	(SP12)	Preliminary analyses of de novo CNVs and sequence variation in the DDD project - <i>Dr Matthew Hurles (on behalf of the DDD project)</i>
15:00	(SP13)	22q11 deletion syndrome – understanding neurodevelopmental disorders in a common CNV - <i>Dr Kate Baker, T Baldeweg, C Chaddock, D Skuse</i>

Cytogenetics		Room: Conference Room, Arts Centre
Chair: Mrs Deborah Morrogh / Mrs Katie Waters		
14:15	(SP14)	DDD: a UK-wide translational genetics study - <i>Dr Caroline Wright</i>
14:35	(SP15)	Two years of first-line cytogenetic testing by array CGH – strategy, overview and findings - <i>Dr Caroline Ogilvie, S Bint, Z Docherty, S Mohammed, JW Ahn</i>
14:55	(SP16)	Accurate measurement of complex copy number variants (CNVs) in association studies: insights from beta-defensins - <i>Prof John Armour, R Pala, S Abu Bakar</i>

Counselling, Social, Policy & Education		Room: Studio, Arts Centre
Chair: Mrs Claire Dolling / Mrs Georgina Hall		
14:15	(SP17)	The Burden and Impact of Retinal Dystrophy on Patients: Lessons for Improving Genetic Eye Services - <i>Dr Ryan Combs, M McAllister, K Payne, G Hall, S Downes, J Lowndes, A Moore, S Devery, G Wright, S Ramsden, G Black</i>
14:30	(SP18)	E-genetics: patient perspectives on using technology in cancer genetics services - <i>Miss Jennifer Hilgart, R Iredale, J Hayward</i>
14:45	(SP19)	Public health implications of personalised screening for cancer: The First Collaborative Oncological Gene-environment Study (COGS) Workshop findings - <i>Dr Susmita Chowdhury, N Pashayan, A Hall, T Dent, G Lyratzopoulos, P Pharoah, H Burton</i>
15:00	(SP20)	Genetic counselling in British Sign Language (BSL) - <i>Ms Rachel Belk, D Trump, A Middleton, AM Young</i>

15:15-16:00 **REFRESHMENT BREAK**

15:15-15:45 **Association of Genetic Nurses and Counsellors Members Meeting**
Room: Conference Room, Arts Centre

16:00-17:30 Concurrent Symposia: Reflections on the Psychosocial and ethical aspects of emerging technologies

Chair: Dr Sue Kenwick / Ms Eva Nazem		Room: The Theatre, Arts Centre
16:00	(SP21)	Scientific introduction to next generation sequencing - <i>Prof Sian Ellard (Exeter)</i>
16:15	(SP22)	Practical Ethical Aspects of Emerging Technologies - <i>Prof Mike Parker (Oxford)</i>
16:30	(SP23)	Implications of emerging technologies for genetic counselling - <i>Dr Christine Patch (London)</i>
16:45	(SP24)	Rapid genetic counselling and testing in newly diagnosed breast cancer patients: a retrospective, observational study - <i>Dr Marijke Wevers (The Netherlands)</i>
17:00	(SP25)	Non-invasive prenatal diagnosis: The patient perspective – <i>Ms Celine Lewis (London)</i>
17:15	Questions and Answers session	

16:00-17:30 Concurrent Symposia: Genetics and Genomics in the Developing World

Chair: Dr Layla Jader / Prof Dhavendra Kumar		Room: Conference Room, Arts Centre
16:00	(SP26)	Genetics and Genomics in the developing world: challenges and opportunities - <i>Prof Dhavendra Kumar (Cardiff)</i>
16:30	(SP27)	Red cell genetic disorders: burden and selective pressures in Africa - <i>Dr Thomas Williams (Kenya)</i>
17:00	(SP28)	Genomic studies of non-communicable diseases and traits in sub-Saharan Africa: opportunities and challenges - <i>Dr Manj Sandhu (Cambridge)</i>

16:00-17:30 Concurrent Symposia: Approaches to Complex Disease

Chair: Dr Alex Blakemore / Prof Nazneen Rahman		Room: Studio, Arts Centre
16:00	(SP29)	Rare variant analysis in large-scale association and sequencing studies - <i>Dr Eleftheria Zeggini (Cambridge)</i>
16:30	(SP30)	Genetics of Coeliac disease - <i>Prof David Van Heel (London)</i>
17:00	(SP31)	Genetic Studies in Rheumatoid arthritis – <i>Prof Jane Worthington (Manchester)</i>

17:30 **BRITISH SOCIETY FOR HUMAN GENETICS – Annual General Meeting**
Room: The Theatre, Arts Centre

18:00-19:00 **Wine Tasting** – Room: Butterworth Hall and the Mead Gallery, Arts Centre

19:00-20:00 Debate

Chair: Prof Alison Metcalfe	Room: The Theatre, Arts Centre
<p>“This house believes that Synthetic Biology should be used to design new forms of life”</p> <p>Setting up the Debate: <i>Dr Chris French (Edinburgh)</i> Debators: <i>Prof Paul Martin (Nottingham), Dr Ainsley Newson (Bristol)</i></p>	

Tuesday 6 September 2011

07:30-08:45 **BSHG Editors Meeting**
Room: National Grid, Arts Centre

07:30 **REGISTRATION**

09:00-10:30 Concurrent Symposia: Cancer pathogenesis and genomic architecture

Chair: Dr Jon Strefford / Miss Sara Dyer		Room: The Theatre, Arts Centre
09:00	(SP32)	Interrogating the Architecture of Cancer Genomes - <i>Dr Peter Campbell (Cambridge)</i>
09:30	(SP33)	Sequence Of Mutational Events And Clonal Architecture Childhood Acute Lymphoblastic Leukaemia - <i>Dr Lyndal Kearney (Sutton)</i>
10:00	(SP34)	Translating genomic insights into treatment approaches in CLL - <i>Prof Thorsten Zenz (Germany)</i>

09:00-10:30 Concurrent Symposia: Somatic Mutation detection in the context of drug prescription

Chair: Ms Jenny Bell		Room: The Cinema, Arts Centre
09:00	(SP35)	The CRUK stratified medicines programme - <i>Dr Phillipe Taniere (Birmingham)</i>
09:30	(SP36)	Somatic mutation detection in the context of drug prescription - <i>Dr Clive Mulatero (Leeds)</i>
10:00	(SP37)	Targeted therapies in the management of Colorectal cancer - <i>Prof Tim Maughan (Oxford)</i>

09:00-10:30 Concurrent Symposia: Neurodevelopmental Disorders

Chair: Prof Angus Clarke / Dr Julia Rankin		Room: Conference Room, Arts Centre
09:00	(SP38)	Rett syndrome, CDKL5 and FOXP1 - <i>Dr Hayley Archer (Cardiff)</i>
09:30	(SP39)	Could this be Angelman Syndrome? - <i>Prof Jill Clayton-Smith (Manchester)</i>
10:00	(SP40)	Delineation of the Pitt-Hopkins syndrome - <i>Dr Jeanne Amiel (France)</i>

09:00-10:30 Concurrent Symposia: Variable phenotypes in genetic conditions – implications for genetic counselling

Chair: Dr Laura Boyes / Mr Peter Marks		Room: Studio, Arts Centre
09:00	(SP41)	Premature ovarian failure and tremor ataxia syndrome in Fragile X - <i>Prof Jeremy Turk (London)</i>
09:20	(SP42)	Alpha-1-Antitrypsin Deficiency (AATD) - <i>Prof Robert Stockley (Birmingham)</i>
09:40	(SP43)	Variability of Phenotypes in Cystic Fibrosis - <i>Dr David Honeybourne (Birmingham)</i>
10:00	(SP44)	Phenotypic Variability and Uncommon Forms of Congenital Adrenal Hyperplasia - <i>Dr Nils Krone (Birmingham)</i>
10:20	Questions and Answers session	

10:30-11:15 **REFRESHMENT BREAK**

10:30-11:00 **NanoString Technologies Seminar – “Digital gene signatures from FFPE- the nCounter enables the bridge, from Research to the Clinic”**
 Room: The Cinema, Arts Centre
 Speaker: *Dr Jim White*

11:15-12:45 EDUCATION SESSIONS

11:15-12:45 **National Institute for Health Research (NIHR)**
 Room: The Theatre, Arts Centre
 Chair: *Prof Peter Farndon (Birmingham)*
 Speakers: *Prof Sir John Burn (Newcastle), Dr Helen Firth (Cambridge), Prof Lyn Chitty (London), Dr Janice McLaughlin (Newcastle)*
 This education session is sponsored by the NIHR Collaborative Group for Genetics in Healthcare (CGGH)

11:15-12:45 **National Genetics Reference Laboratories – Next Generation Sequencing - What’s happening now?**
 Room: Conference Room, Arts Centre
 Speakers: *Mr Hedley Carr (AstraZeneca), Dr Simon Ramsden (Manchester), Ms Antigoni Tzika (Leeds), Mr Sanjeev Bhaskar (Manchester)*

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

12:45-13:15 **Clinical Molecular Genetics Society Members Extraordinary General Meeting**
 Room: The Theatre, Arts Centre

- 12:45-13:15 **Lead Clinicians Meeting**
Room: SSO.09, Social Sciences Building
- 13:00-14:00 **Medico-Legal Workshop**
Room: Conference Room, Arts Centre
Speakers: to be confirmed
- 14:00-16:00 **POSTER VIEWING & TRADE EXHIBITION**
- 15:00-15:30 **Qiagen Seminar**
Room: Conference Room, Arts Centre
Speakers: to be confirmed
- 15:15-16:00 **REFRESHMENT BREAK**

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

Chair: Dr William Newman / Dr Richard Scott		Room: The Theatre, Arts Centre
16:00	(SP45)	Disruption of the pre-replication complex causes a spectrum of primordial dwarfism phenotypes - <i>Dr Louise Bicknell, S Walker, J Schoots, A Klingseisen, T Stiff, A Leitch, S Brown, C Kerzendorfer, CA Martin, P Yeyati, ME Harley, M O'Driscoll, PA Jeggo, E Bongers, AP Jackson</i>
16:15	(SP46)	Using crystallography and biophysics to understand mutations in the Aicardi-Goutières Syndrome Ribonuclease H2 enzyme - <i>Dr Martin Reijns, D Bubeck, LCD Gibson, SC Graham, A Leitch, GS Baillie, EY Jones, AP Jackson</i>

17:15-18:15 CARTER LECTURE

Chair: Dr Peter Turnpenny		Room: The Theatre, Arts Centre
17:15	(SP47)	Making Eyes: Genes and Networks - <i>Prof Veronica van Heyningen (Edinburgh)</i>

- 19:30 **CONFERENCE DINNER & DANCE**
Panorama Suite, Drinks Reception in Chancellor's Suite,
Rootes Building, University of Warwick

Wednesday 7 September 2011

- 07:30 **REGISTRATION**

09:00-10:30 Concurrent Symposia: Neurogenetics

Chair: Dr Jonathan Waters / Mr Dominic McMullan		Room: The Theatre, Arts Centre
09:00	(SP48)	Genetics of Autistic Disorders - <i>Prof David Skuse (London)</i>
09:30	(SP49)	Array CGH in adults with epilepsy - <i>Prof Sanjay Sisodiya (London)</i>
10:00	(SP50)	A de novo paradigm for mental retardation - <i>Dr Lisenka Vissers (The Netherlands)</i>

09:00-10:30 Concurrent Symposia: Clinical Management of Genetic Disorders

Chair: Dr Peter Turnpenny / Dr Charu Deshpande		Room: The Cinema, Arts Centre
09:00	(SP51) Coordinating care for Neurofibromatosis type one (NF1): a population based approach - <i>Dr Susan M Huson (Manchester)</i>	
09:30	(SP52) Ehlers Danlos Syndrome - clinical management in 2011 - <i>Dr Glenda Sobey (Sheffield)</i>	
10:00	(SP53) The clinical and genetic management of Bardet-Biedl Syndrome - <i>Prof Phil Beales (London)</i>	

09:00-10:30 Concurrent Symposia: Animal Models of Genetic Disorders

Chair: Dr Forbes Manson / Dr Andrew Jackson		Room: Conference Room, Arts Centre
09:00	(SP54) Role of Drosophila in the study of complex genetic disorders - <i>Dr Tamar Grossman (USA)</i>	
09:30	(SP55) Zebrafish models for human disease: rare disorders and development of high throughput in vivo imaging tools for phenotype screening applications - <i>Dr Ferenc Mueller (Birmingham)</i>	
10:00	(SP56) Genetic analysis of tetrapod development & disease in Xenopus tropicalis - <i>Dr Lyle Zimmerman (London)</i>	

10:30-11:00 **REFRESHMENT BREAK**

11:00-12:15 Concurrent Sessions

Clinical		
Chair: Dr Julia Rankin/ tbc		Room: The Theatre, Arts Centre
11:00	(SP57) Molecular and pathological characterisation of an inherited condition amongst the Amish involving high myopia and sensorineural hearing loss - <i>Dr Heather Moore-Barton (nee Webb), BA Chioza, Y Matsumoto, HE Cross, K Sakoori, M Ota, K Gurtz, MA Patton, J Aruga, AH Crosby</i>	
11:15	(SP58) Mutations in PRDM5 and ZNF469 cause brittle cornea syndrome by influencing extracellular matrix development and maintenance - <i>Dr Emma Burkitt Wright, HL Spencer, FDC Manson, J Urquhart, N Zoppi, L Porter, KE Chandler, S Banka, J Clayton-Smith, N Khan, M Rohrbach, M Colombi, C Giunta, GCM Black</i>	
11:30	(SP59) Lupus: How much 'complexity' is really (just) genetic heterogeneity? - <i>Prof Yanick Crow</i>	
11:45	(SP60) Reversible Infantile Respiratory Chain Deficiency is a Genetically Heterogenous Mitochondrial Disease - <i>Prof Joanna Poulto, J Uusimaa, H Jungbluth, C Fratter, EMFV Bailey, G Crisponi, L Feng, M Zeviani, I Hughes, E Treacy, J Birks, GK Brown, C Sewry, F Muntoni</i>	
12:00	(SP61) Micro and Martsof Syndromes - <i>Dr Irene Aligianis, M Handley, S Carpanini, D Bem, S Yoshimura, R Nunes-Bastos, F Bond, M Kurian, F Rahman, Y Hadzhiev, S Pasha, G Kirby, A Straatman-Iwanowska, A Cullinane, A McNeill, P Gissen, F Müller, E Maher, F Barr</i>	

Cytogenetics		Chair: Miss Janine Burbridge / Mr Graham Fews	Room: The Cinema, Arts Centre
11:00	(SP62)	Evaluation of MLPA and SISH as two alternative methods for assessing ERBB2 gene amplification status in breast cancer patients - <i>Miss Clare Durajczyk</i>	
11:15	(SP63)	Sequential SNP6.0 microarray analysis in Rai stage 0 CLL patients with either stable or progressive identifies genomic evolution in both cohorts - <i>Dr Jonathan Strefford, M Rose-Zerilli, H Parker, M Rajabali, A Parker, A Gardiner, Z Davis, S Mold, S Glide, T Chaplin, BD Young, DG Oscier</i>	
11:30	(SP64)	Simultaneous t(5;11)(q35;p15) NUP98-NSD1 gene fusion and FLT3-ITD in paediatric acute myeloid leukaemia - <i>Dr Sara Dyer, S Akiki, S Jeffries, K Tawana, N Abou-Zeid, J Borrow, J Caddick, M Velangi, M Griffiths, H Newell</i>	
11:45	(SP65)	A non-tandem duplication of the dystrophin gene: The value of FISH as a follow-up method for imbalances detected by Array CGH - <i>Mrs Joanna McParland, E Blair, R Lumsden, C Noakes, J Evans, K Smith</i>	
12:00	(SP66)	Basic genetic Principles illustrated by Meiotic Chromosome Analysis - <i>Prof Maj Hulten</i>	

Molecular		Chair: Dr David Baty / Dr Fiona Macdonald	Room: Conference Room, Arts Centre
11:00	(SP67)	The Clinical and scientific impact of investigating splicing - <i>Dr Diana Baralle, M Raponi, C Tammaro, D Eccles, S Marks, F Lin, M Lyon, D Robinson</i>	
11:15	(SP68)	Further developments in the use of cell free fetal DNA for non-invasive prenatal diagnosis of single gene disorders: digital PCR allows diagnosis of sickle cell disorder - <i>Prof Lyn Chitty, TCR McDonnell, A Barrett</i>	
11:30	(SP69)	Investigation of existing methods for diagnostic EGFR Mutation Testing in non-small cell Lung Cancer Patients and exploration fo the utility of Next Generation Sequencing - <i>Miss Victoria Crowe</i>	
11:45	(SP70)	Cafe Variome - gene mutation data clearinghouse - <i>Dr Owen Lancaster, RK Hastings, R Dalglish, D Atlan, GA Thorisson, RC Free, A Webb, AJ Brookes</i>	
12:00	(SP71)	Use of Next Generation Sequencing in the diagnosis of Retinitis Pigmentosa and associated Retinal Dystrophies - <i>Mr James O'Sullivan, SB Daly, SS Bhaskar, B Nicholas, S Ramsden, GCM Black</i>	

Cancer		Chair: tbc	Room: Studio, Arts Centre
11:00	(SP72)	Feasibility and acceptability of offering breast cancer risk estimation in the context of the UK National Health Service Breast Cancer Screening Programme: a new paradigm for cancer prevention - <i>Prof Gareth Evans, S Astley, P Stavrinou, S Sahin, S Ingham, H McBurney, B Eckersley, M Harvie, M Wilson, U Beetles, R Warren, A Hufton, J Warwick, W Newman, I Buchan, J Cuzick, A Howell</i>	
11:15	(SP73)	Association of the TNFRSF10A and TNFRSF10B Gene Region on Chromosome 8 with Breast Cancer Risk - <i>Dr Sushila Rigas, G Elliot, I Brock, M Parry, R Abo, S Knight, G Burghel, WY Lin, MW Reed, NJ Camp, A Cox</i>	
11:30	(SP74)	Fine-Scale mapping of the 11q13 breast cancer susceptibility locus - <i>Dr Maya Ghousaini, S Ahmed, K Michailidou, K Gregory, E Dicks, K Driver, PD Pharoah, DF Easton, AM Dunning</i>	
11:45	(SP75)	Screening and oophorectomy for Familial Ovarian Cancer in a single centre - <i>Prof D Gareth Evans, J Taylor, F Lalloo, R Clayton</i>	
12:00	(SP76)	Analysis of BRCA1 and BRCA2 mutation distributions in a large unselected UK population - <i>Dr Simon Thomas, J Sillibourne, D Ward, C Mattocks, J Harvey</i>	

12:15-13:15 4TH BSHG LECTURE

Chair: Prof Sir John Burn		Room: The Theatre, Arts Centre
12:15	(SP77)	Evolution of the Cancer Genome - <i>Prof Michael Stratton (Cambridge)</i>

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

13:15-14:15 **BSHG Scientific Programme Committee Meeting**
Room: Studio, Arts Centre

14:15-16:15 Concurrent Workshops

Novel Insights into the Genetics of Acute Lymphoblastic Leukaemia and Myeloproliferative Neoplasms		Room: The Theatre, Arts Centre
Chair: Prof Christine Harrison		
14:15	(SP78)	Mutations associated with acquired UPD in MDS/MPN - <i>Prof Nick Cross (Wessex)</i>
14:45	(SP79)	How do we decipher the genetic profile of acute lymphoblastic leukaemia? - <i>Prof Christine Harrison (Newcastle)</i>
15:15	(SP80)	Collaborative GWA studies identify four loci associated with childhood acute lymphoblastic leukaemia risk - <i>Dr Amy Sherborne (Sutton)</i>
15:45	(SP81)	What we know about CRLF2 in Acute Lymphoblastic Leukaemia - <i>Dr Lisa Russell (Newcastle)</i>

Treatment Strategies for Genetic Disorders		Room: Conference Room, Arts Centre
Chair: Prof Julian Sampson/ Dr William Newman		
14:15	(SP82)	New therapies for lysosomal storage disorder - <i>Dr Simon Jones (Manchester)</i>
14:50	(SP83)	RNAi and small molecule therapy development in genetic skin disorders - <i>Prof Irwin McLean (Dundee)</i>
15:25	(SP84)	Gene therapy for eye disease - <i>Prof James Bainbridge (London)</i>
16:00	Questions and Answers session	

16:15 **End of Conference**

Poster presentations

1. Clinical Genetics

- (1.01) Patient Information Collation Service: The Cambridge solution to managing waiting times - *Ms Felicity Wadrup, A Kershaw, S Downing*
- (1.02) The common TMC1 mutation p.Arg34X is not a significant cause of deafness in British Asians - *Dr Claire Searle, LA Mavrogiannis, CP Bennett, RS Charlton*
- (1.03) An OPA1 mutation is associated with a multiple sclerosis-like illness in two siblings - *Dr Wendy Jones, P Rich, AK Saggat, A Al-Memar*
- (1.04) HRAS p.Gly12Val mutations in four patients with neonatal lethal Costello syndrome - *Dr Emma Burkitt Wright, L Bradley, J Shorto, V McConnell, C Gannon, HV Firth, S-M Park, A D'Amore, P Munyard, PD Turnpenny, A Charlton, M Wilson, B Kerr*
- (1.05) Best practice guidelines; are they falling on deaf ears? - *Dr Heather Moore-Barton (nee Webb), HL Hanson, S Mansour*
- (1.06) Merosin-deficient congenital muscular dystrophy: a founder mutation in eight Arab families from Kuwait - *Dr Makia Marafie, I AlSuliman, M Al-Ben Ali, A Mahdi, L Bastaki*
- (1.07) Genotype-phenotype correlation in Polyposis; genotype is a predictor of survival - *Miss Katy Newton, E Mallinson, J Bowen, T Clancy, F Lallo, J Hill, DG Evans*
- (1.08) An audit of surveillance for Wilms tumour in at-risk individuals - *Dr Anjana Kulkarni, A F Brady, C Deshpande, D Ruddy*
- (1.09) Algorithm to guide investigations and referral pathways in hypertrophic cardiomyopathy - *Dr Anjana Kulkarni, L Robert, T Beuser, G Carr-White*
- (1.10) Metachronous colorectal cancer risk in patients with a moderate family history - *Miss Katy Newton, K Green, F Laloo, J Hill, DG Evans*
- (1.11) Colonoscopy screening compliance and outcome in patients with Lynch Syndrome - *Mrs Katy Newton, K Green, F Laloo, DG Evans, J Hill*
- (1.12) A retrospective cohort study to determine the genetic outcome in children born by assisted reproductive technologies - *Dr Heather Moore-Barton (nee Webb), A Marsh, M McEntagart*
- (1.13) Variants of Uncertain Significance in BRCA1: A review of 41 Cambridge families - *Dr Sue Kenwick, K Oakhill, E Thompson, J Drummond, J Whittaker*
- (1.14) Does NSD1 duplication cause a mirror phenotype of Sotos syndrome? - *Dr Kate Baker, A Clarkson, L Willatt, I Simonic, FL Raymond, R Sandford*
- (1.15) Functional assessment of mismatch repair - *Miss Pinar Tulay, S Jaroudi, J Harper, S SenGupta*
- (1.16) Mutations in Zinc-finger E-box binding protein (ZEB2) cause microphthalmia - *Miss Arghavan Gofrehi, R Perveen, D FitzPatrick, W Reardon, W Dobyns, A Mederia, I Cordeiro, EN Hilton, G Black*
- (1.17) New insights into genetic causes of neonatal diabetes mellitus: SLC2A2 mutations can cause neonatal diabetes prior to the appearance of Fanconi-Bickel syndrome - *Dr Francis Sansbury, SE Flanagan, JAL Houghton, E Young, FL Shuixian Shen, S Al-Sinani, A Kariminejad, AM Habeb, MA Abdullah, S Ellard, AT Hattersley*
- (1.18) Marfan syndrome and the Ghent nosologies - which is best? - *Dr John Dean, J Crow, W Lam*
- (1.19) Propionic acidaemia in the Old Order Amish presenting as isolated dilated cardiomyopathy - *Dr Martina Muggenthaler, HE Cross, ER Behr, AH Crosby*
- (1.20) Clinical Utility of the UKGTN DHCR7 service for Smith-Lemli-Opitz syndrome - *Ms Hilary Sawyer, MJ Gable, J Honeychurch, M Greenslade, S Burton-Jones, AY Brown, M Williams*
- (1.21) Array CGH in patients with cleft lip and/or palate and external ear malformations - *Dr Angela Barnicoat, A Shaw, R Palmer, M Bitner-Glindzicz, M Lees, L Chitty*
- (1.22) A novel Xq28 deletion including FAM58A causes Syndactyly, Telecanthus, Anogenital and Renal malformations (STAR syndrome) - *Dr Sarju Mehta, N Thalange, I Simonic*
- (1.23) Distinguishing features of 10q23 deletion as a cause of juvenile polyposis - *Dr Mark Tein, RA Clark, CM Ogilvie*
- (1.24) The Scottish Electronic Records System for Clinical Genetics - *Dr John Dean, M Edwards, W Lam*
- (1.25) Non-invasive prenatal diagnosis using cell free fetal DNA – how far have we got? An update from the RAPID programme - *Prof Lyn Chitty, m HillRAPID Team*
- (1.26) Targeted Capture and Next Generation Sequencing (NGS) of genes involved in Inherited Retinal Degeneration (IRD) - *Dr Morag Shanks, SM Downes, A Kwasniewska, E Packham, P Clouston, A Seller, R Copley, S Lise, J Taylor, K Hudspith, AOM Wilkie, J Ragoussis, AH Nemeth*

- (1.27) Two autosomal recessive conditions in a family with multiple consanguineous relationships - *Dr Gabriela Fuchs, JG Barwell, PC Vasudevan*
- (1.28) Unusual antenatal presentation of Zellweger syndrome confirmed biochemically in a second affected fetus - *Dr Helen Moody, A Male, R Scott, T Jacques, A Offiah, J Rennie, M Jackson*
- (1.29) The wide spectrum of Fraser syndrome and associated conditions: A clinical and molecular analysis of two interesting cases - *Dr Helen Moody, J Hurst, K Nischal, A Slavotinek*
- (1.30) Outcomes following the diagnosis of Turner syndrome: East Midlands & South Yorkshire Anomalies Register (EMSYCAR) 1998-2010 - *Dr Emily Craft, JLS Budd, ES Draper*
- (1.31) Detailed Clinical Features of Naegeli-Franceschetti-Jadassohn Syndrome in a Large Family - *Dr Victoria Harrison, G Ogg, H Stewart*
- (1.32) Pseudohypoparathyroidism type 1b due to paternal uniparental disomy of chromosome 20q - *Dr Abhijit Dixit, M Lever, RL Poole, DJG Mackay, M Suri*
- (1.33) Establishing a comprehensive molecular genetics service for infantile onset epilepsies - *Dr Eleanor Reavey, RE Ellis, SM Zuberi, SR Stenhouse*
- (1.34) X-linked lissencephaly with ambiguous genitalia – a case presentation - *Mrs Sheila Palmer-Smith, LG Rosser, J Tull, R Butler, DT Pilz, N Foulds*
- (1.35) Referral to Clinical Genetics following post mortem examination in foetal losses - *Dr Meena Balasubramanian, S Bourke, N Crawford, M Cohen*
- (1.36) Evaluation of urodele amphibians as animal models - *Dr Marie-Anne O'Reilly*
- (1.37) Reverse genetics: A 2.8 Mb deletion encompassing the NF1 gene in a patient with no clinical features of neurofibromatosis type 1 - *Dr Ana Lisa Taylor Tavares, SM Park, R Armstrong, L Willatt, I Simonic*
- (1.38) A second case of triplication of the Williams-Beuren region with markedly abnormal neurodevelopment and neuroimaging - *Dr Vishakha Phatak, D Gray, E Jones, K Cubitt, N Gayatri, J Thomson*
- (1.39) Born Healthy - A toolkit for assessing needs and developing genetic services in low and middle income countries - *Ms Corinna Alberg, L Nacul, H Burton, G Sagoo, S Chowdhury, S Moorthie, A Hall*
- (1.40) Commissioning of a national diagnostic service for Stickler syndrome - *Dr Allan Richards, A McNinch, K Oakhill, B Treacy, J Whittaker, A Poulson, M Snead*
- (1.41) Learning and behaviour in children with NF1: Data from a population-based study - *Dr Annukka Lehtonen, S Garg, J Green, D Trump, G Evans, S Huson*
- (1.42) Eliciting feedback from our patients – how to improve a service - *Mrs Julia Finch, C Brooks, K Dymant, A Woodward, C Fallon, J Goulding, C Benjamin*
- (1.43) UK GenetICC – Inherited Cardiac Conditions register - *Dr Liz Ormondroyd, E Blair, C McClymont, P McKeown, J McOsker, F Stewart, P Brennan, R Newbury-Ecob, G Stewart, S Mehta, W Newman, K Metcalfe, L Kerzin Storrar, H Watkins*
- (1.44) A large intragenic duplication in the Folliculin (FLCN) gene causing Birt-Hogg-Dube syndrome with an interesting phenotype - *Dr Derek Lim, C Morgan, PK Rehal, F Bond, L Walker, D Halliday, ER Maher, F Macdonald*
- (1.45) Mutation testing and prenatal diagnosis in Neurofibromatosis type one (NF1) - *Dr Susan Huson, E Sach, E Butler, M Crocker, S Wall, J Byron, W Fergusson, A Wallace, L Messiaen*
- (1.46) Patient and public involvement in genetic and developmental medicine research - *Dr Marisha Palm, G Hall, G Black, J Clayton-Smith, H Kitchener, R Albrow, L Sadler, B Starling, K Dack, D Donnai, HR Middleton-Price*
- (1.47) A founder mutation in the SAMHD1 gene identified in the Amish is associated with laryngeal involvement - *Dr Emma Baple, H Cross, B Chioza, M Simpson, R Trembath, M McEntagart, M Patton, A Crosby*
- (1.48) A unique form of Septo-Optic Dysplasia and its rehabilitative management: a case report - *Mr Amar Ajay Chotai, EL Wakeling, MT Dattani, A Bloch-Zupan, RM Winter, SE Holder*
- (1.49) Does genetics add value to the care of paediatric cardiac patients? Our experience of cardiac genetic referrals at Loma Linda University Medical Centre (LLUMC) in the USA, highlighting the need for a protocol for genetic work-ups - *Dr June-Anne Gold, S Ramanathan, L Bailey, J Bartley, R Clark*

2. Molecular Genetics

- (2.01) Association of apolipoprotein AIV gene polymorphism (G360T) with metabolic syndrome in Tehran Lipid and Glucose Study - *Miss Maryam Zarkesh, MS Daneshpour, M Zarkesh, M Hedayati, F Azizi*
- (2.02) Eurasian and Sub-Saharan African Mitochondrial DNA haplogroup influences pseudoexfoliation glaucoma development in Saudi patients - *Dr Khaled Abu-Amero, VM Cabrera, JM Larruga, EA Osman, AM Gonzalez, SA Al-Obeidan*
- (2.03) Validation of the Luminex xTAG Cystic Fibrosis 39 assay for diagnostic testing and newborn screening using dried blood spots - *Dr Melissa Rogers, S Roring, T McDevitt, DE Barton*
- (2.04) DFNB1 markers associated with congenital hearing loss in newborns - *Prof Emilia Severin, C Dragomir, A Stan, TD Stefanescu, A Toma, C Sarafoleanu, L Savu*
- (2.05) Investigation of unusual results from B reaction of Elucigene®'s CF-EU2 kit - *Mrs Elizabeth Johnston, T Campbell, S Mutch, J Field, G Cross*
- (2.06) Case Study: Unusual copy number findings in a Sicilian family with SMA - *Dr Johanna Hunt, C Hardy, R Barber, H Cox, R Coleman, F Macdonald, K Stewart*
- (2.07) Development of an ARMS PCR test for the rapid detection of three Polish BRCA1 founder mutations - *Ms Naomi Bowers, A Wallace*
- (2.08) Mitochondrial Genome Analysis using Next Generation DNA Sequencing - *Dr Suzanne Drury, M Sweeney, C Woodward, M Davis, L Jenkins, N Lench*
- (2.09) GLIS3 gene mutations in patients with syndromic neonatal diabetes - *Dr Jayne Houghton, A Damhuis, R Caswell, S Flanagan, J Crolla, C Shaw-Smith, S Ellard*
- (2.10) Dominant and recessive RRM2B mutations are frequent in familial PEO with multiple mtDNA deletions - *Mr Carl Fratter, P Raman, CL Alston, EL Blakely, K Craig, C Smith, J Evans, A Seller, B Czermin, RDS Pitceathly, MG Hanna, J Poulton, C Brierley, TG Staunton, PD Turpenny, AM Schaefer, PF Chinnery, R Horvath, DM Turnbull, GS Gorman, RW Taylor*
- (2.11) Testing for PMS2 gene variants in hereditary colorectal cancer patients in Scotland - *Dr Nicola Carroll, N Williams, ME Porteous, JP Warner*
- (2.12) Mitochondrial DNA depletion syndrome due to mutations in the MPV17 gene - *Dr Julie Evans, J Uusimaa, C Smith, A Butterworth, K Craig, N Ashley, C Liao, D Kelly, M Samyn, S Rahman, H Stewart, AAM Morris, A Seller, C Fratter, RW Taylor, J Poulton*
- (2.13) Autosomal Recessive (ARO) and Autosomal Dominant Osteopetroses (ADO): A UKGTN diagnostic service for rare genes - *Ms Melanie Pennock, C Steward, A Villa, A Pangrazio, M M Williams*
- (2.14) Development of a Molecular Genetic Diagnostic Service for Radiosensitive-Severe Combined Immunodeficiency Disease - *Ms Shahnaz Bibi, A Taylor, T Linton-Willoughby, CM Cale, KC Gilmour, R Vijzelaar, L Jenkins, N Lench*
- (2.15) A Comprehensive Molecular Genetics Screening Service for Choroideremia - *Ms Tracy Colclough, A O'Grady, N Hart-Holden, J O'Sullivan, S Ramsden, GCM Black*
- (2.16) Epidermal Growth Factor Receptor (EGFR) mutation testing in non-small cell lung cancer (NSCLC) patients - *Mrs Helen Eaton, L Dutton, AJ Wallace, E Howard, L Gaunt, F Blackhall, W Newman*
- (2.17) WFS1-related disorders: an overview of the WMRGL service - *Miss Rachel Coleman, C Hardy, F Macdonald, N Motton, P Rehal*
- (2.18) Identification of a non-pathogenic splice site mutation in the GJB2 (CX26) gene? - *Miss Rachel Coleman, C Hardy, F Macdonald, C Oley, G Vincent*
- (2.19) Design and Optimisation of a Diagnostic Screening Service for Kabuki Syndrome (MLL2 gene) - *Dr Emma Howard, S Osborne, A Wallace, R Raghuram, S Banka, L Gaunt*
- (2.20) Audit of ALMS1 gene screening in Alstrom syndrome patients - *Miss Nicole Motton, C Hardy, Y Wallis, F Macdonald*
- (2.21) Imprinting disorders – finding out why: exploring the causes and consequences of imprinting disorders - *Dr Deborah Mackay, RL Poole, IK Temple*
- (2.22) A family with co-existing SDHB and SDHD mutations causing hereditary paraganglioma syndrome - *Dr Deirdre Cilliers, SM Park, S Kenwick, H Simpson, K Sarson, F Laloo, L Bradley, ER Maher*
- (2.23) Beta-thalassemia molecular study showed IVSI-5 is the most common mutation in Khorasan, Iran - *Prof Kazem Ghodsi, F Mohajer Tehran*
- (2.24) Development of a reliable MLH1 hypermethylation assay protocol from formalin fixed paraffin embedded (FFPE) tissue - *Dr Andrew Wallace, N Jorgenson, K Newton, DGR Evans, F Laloo, J Hill*
- (2.25) Exome sequencing in Patients with rare cortical malformations - *Dr Alistair Pagnamenta, S Lise, V Harrison, U Kini, H Stewart, G Quaghebeur, A Deng, JC Taylor, DC Keays*

- (2.26) Association of intercellular adhesion molecule-1 (ICAM-1) gene polymorphism and squamous cell carcinoma in a population in northern Iran - *Dr Farhad Mashayekhi, Z Salehi*
- (2.27) Estrogen receptor –alpha gene polymorphism and the risk of recurrent spontaneous abortion in an Iranian population - *Dr Farhad Mashayekhi, A Bakhshi*
- (2.28) Autozygosity mapping and linkage analysis identify novel loci for juvenile myoclonic epilepsy - *Dr Kate Everett, AR Al-Tahan, R Robinson*
- (2.29) MDM2 309 polymorphism and idiopathic male infertility in Iran - *Dr Zivar Salehi, F Mashayekhi*
- (2.30) Mitochondrial DNA lineages of African origin confer susceptibility to primary open-angle glaucoma in Saudi patients - *Dr Khaled Abu-Amero, A Gonzalez, E Osman, J Larruga, V Cabrera, S Al-Obeidan*
- (2.31) Manganese superoxide dismutase (MnSOD) gene polymorphism and endometriosis in Iran - *Dr Zivar Salehi, S Khodayari*
- (2.32) - *Dr Zivar Salehi, S Khodayari*
- (2.33) Next Generation Sequencing (NGS) of Genes Associated with Congenital and Neurodegenerative Ataxia - *Dr Alexandra Kwasniewska, ME Shanks, S Lise, MZ Cader, K Talbot, SM Downes, J Ragoussis, AH Nemeth*
- (2.34) Validation of the Asuragen Amplidex FMR1 kit for diagnostic Fragile X testing and further characterization of the WHO FRAX Reference Panel - *Dr Michael Sweeney, K Meaney, DE Barton*
- (2.35) Introduction of a comprehensive genetics service for disorders of pain perception - *Dr Sally-Anne Halsall, R Treacy, CG Woods*
- (2.36) A UKGTN service for Ellis-van Creveld syndrome - *Miss Laura Yarram, T Antoniadi, S Smithson, J Goodship, S Batey, M Williams*
- (2.37) Direct RNA analysis for splice abnormalities as a diagnostic service - *Miss Sophie Marks, F Lin, M Lyon, M Raponi, D Baralle, D Robinson*
- (2.38) Non-invasive Prenatal Detection of Aneuploidy by Targeted Next Generation Sequencing - *Dr Carolyn Dent, LS Chitty, JA Crolla, HE White*
- (2.39) The wider implications of trisomy 21 mosaicism - *Dr Helen White, V Hall, EG Allen, S Sherman, P Jacobs*
- (2.40) The use of hybrid mini-gene assays in a diagnostic setting to assess unclassified variants for splicing defects - *Miss Nicola Ibberson, L Parker, V Moye, R Treacy, J Whittaker, A Richards*
- (2.41) Expansion of a diagnostic service for hypertrophic cardiomyopathy using next generation sequencing - *Dr Sarah Reid, J Woodley, K Kaur, K Thomson, J Broxholme, J Taylor, E Blair, J Taylor, H Watkins*

3. Cytogenetics and Molecular Cytogenetics

- (3.01) Investigation of Subtelomeric Chromosome Abnormalities, Using MLPA in 50 patients with Idiopathic Mental Retardation and its comparison with 50 Control Normal Cases - *Dr Farkhondeh Behjati, K Kahrizi, S Ghasemi Firouzabadi, M Najafi, B Ebrahimzadeh, Z Hadipour, F Hadipour, Kh Jalalvand, S Banihashemi, S Arzhang, I Bahman, SH Jamaladini Ezabady, H Najmabadi*
- (3.02) A familial ins(6;11)(p21.1;p12p14.3) with unexpected phenotypic consequences - *Ms Lori Ekstrom, SA Lynch, J Crolla, T Morris, DR Betts*
- (3.03) Introduction of the CD138 Selection Kit (EasySep) as a method of improving the identification of genetic aberrations from Myeloma patients: our Aberdeen experience - *Dr Kathryn McAuley, D Massie, P Batstone, J Tighe, D Stevenson*
- (3.04) A family with an inherited 15q13.3 microdeletion - *Miss Sian Jose, SM Morgan, DT Pilz, SH Roberts*
- (3.05) Stabilisation of an inverted duplication deletion (8p) by telomere capture from 15q - *Mrs Deborah Barrell, SF Jose, SM Morgan, PW Thompson, SH Roberts*
- (3.06) Are amniotic fluid colonies derived from single precursor cells? - *Mrs Olivia Fox, M Crocker, A Bhagwagar, K Smith*
- (3.07) A novel rearrangement in an atypical myeloproliferative neoplasm with subsequent development of an unrelated clone following transformation to AML - *Mrs Claire Brooks, J Burbridge, S Marks, A Shlebak, S Ryley*
- (3.08) A 10 megabase familial deletion of 5q21-2 and short stature: Does size really matter? - *Dr Lionel Willatt, I Simonic, A Clarkson, M Bateman, RN Sandford*
- (3.09) Multiplex PCR for diagnostic free fetal DNA sexing – A pilot study - *Dr Lionel Willatt, K Yates, R Stephens, R Treacy, FL Raymond*
- (3.10) Prenatal diagnosis of androgenetic/parthenogenetic chimeras - *Dr Celia Donaghue, Q Zheng, A Hills, T Spencer, C Ogilvie, J Waters, K Mann*

- (3.11) A female with XX/XY chimaerism and a balanced translocation - *Mrs Heather Ward, K Chandler, E Burfitt, M Bulman*
- (3.12) Automation, How Much Has it Helped? - *Miss Angela Harris, E Waldron, M Fenlon, S Jeffries, M Griffiths*
- (3.13) Chromosomal abnormalities showed on amenorrhoea patients - *Prof Kazem Ghodsi, F Mohajer Tehran, L Hafizi*
- (3.14) Array CGH as a helpful diagnostic tool for increasing the detection of genetic causes of intellectual disability in children with congenital anomalies and normal karyotype - *Miss Malgorzata Drozniewska, A Bullo, M Bancarz, O Zuffardi, O Haus*
- (3.15) Implementation of Genetix Cytovision GSL-120 and its impact on diagnostic services - *Miss Sophie Laird, C Price, J Crolla*
- (3.16) Discovery of very large mosaic genomic changes by aCGH – confirmation in near-parallel bone marrow karyotyping and FISH) analysis consistent with ALL - *Mrs Deborah Morrough, D Addy, S Chatters, R Palmer, J Kenny, A Barnicoat, H Kempski, JJ Waters*
- (3.17) Evaluation of lineage-specific confined placental sex chromosome mosaicism: 45,X, 46,X,der(Y) and 46,XY in CVS and follow-up placental tissue (post-spontaneous miscarriage) - *Mrs Olivera Spasic-Boskovic, A Hills, C Donaghue, T Spencer, K Mann, JJ Waters*
- (3.18) Two case reports of Feingold syndrome due to microdeletions of MYCN detected by Array-CGH - *Ms Karen Marks, S Mansour, M McEntagart, L Hill*

4. Counselling, Social, Policy and Education

- (4.01) The Process of parental disclosure in Duchenne Muscular dystrophy (DMD) - *Miss Pooja Dasani, A Clarke*
- (4.02) Walsall Community Genetic counselling and awareness Service - *Miss Pooja Dasani, H Cox, R Ayaz*
- (4.03) Young peoples' experience of neurofibromatosis type 1: interviews with health professionals - *Ms Jenny Barke, D Harcourt, J Coad, A Newson*
- (4.04) How should non-invasive prenatal diagnosis for chromosomal abnormalities be implemented? An ethical analysis - *Dr Ainsley Newson, Z Deans*
- (4.05) Professional training in bioinformatics and next generation sequencing - *Dr Angela Davies, TE Hancocks, K Mulryan, A Devereau, A Brass, H Middleton-Price, D Donnai*
- (4.06) Increased demand for genetics education within the medical specialties: a role for e-learning - *Mrs Michelle Bishop, P Rainger, R Newton, P Farndon*
- (4.07) Genetic Testing for Health: a dedicated website to support the ordering of genetic tests by non-genetics specialists - *Mr William Foster, WT Foster, C Bradley, P Rainger, F MacDonald, P Farndon*
- (4.08) Orphanet: more comprehensible information about rare diseases services in the UK - *Dr Idoia Gomez-Paramio, S Ayme, HR Middleton-Price, D Donnai*
- (4.09) Genetic counselling – a review of breast cancer risk and Neurofibromatosis Type 1 in the West of Scotland Regional Genetics Service - *Ms Catherine Watt, N Bradshaw, S Gibson, E Haque, M Longmuir, E Mc Guire, D Meechan, V Murday, L Snadden, E Tobias, R Davidson*
- (4.10) Enhancing knowledge about modern genetic medicine among secondary school students and their teachers in the UK - *Dr Matthew Hickman, P Finegold, LE Holmes, I Starling, K Dack, L Winters, M Palm, A Hall, J Worthington, J Harris, AP Read, D Donnai, HR Middleton-Price*
- (4.11) Increasing awareness of genetic research techniques through a national schools programme delivered by the UK's network of science and discovery centres - *Dr Matthew Hickman, K Dack, P Fidler, M Livingstone, N Stock, SJ Robinson, N Cartwright, LE Holmes*
- (4.12) Development and validation of the Genetic Counselling Outcome Scale (GCOS-24) - *Dr Marion McAllister, AM Wood, G Dunn, S Shiloh, C Todd*
- (4.13) Whole genome sequencing: applications and implications - *Dr Caroline Wright, H Burton, A Pokorska-Bocci, S Moorthie, A Hall, S Sanderson, R Skinner, G Sagoo*
- (4.14) An Investigation into the perceptions of Individuals with Turner Syndrome and their Parents - *Miss Jessica Hensman, A Edwards, K Thirlaway, A Procter*
- (4.15) Inaccuracies in the flow of information about positive BRCA1/2 diagnostic genetic test results from the genetics clinic through the family - *Ms Chris Jacobs, JA Smith, C Dancyger, M Papaconstantinou, S Michie*
- (4.16) Revising our practice: predictive testing for hereditary cancer conditions with a single clinic appointment - *Dr Amy Taylor, F Wadrup, V Wiles*
- (4.17) The perspectives and experiences of young men with Duchenne muscular dystrophy - *Mr Ben Player*

- (4.18) The Clinical Genetics Specialty Certificate Examination – The trainees' perspective - *Dr Alexandra Murray*
- (4.19) Risk perception, communication and response to personalised genetic information in context of personalised screening for cancer of the general population - *Dr Susmita Chowdhury, N Pashayan, A Hall, T Dent, P Pharoah, H Burton*

5. Cancer Genetics

- (5.01) Outcomes from the new integrated mismatch repair deficiency service at Guy's & St Thomas NHS Foundation Trust - *Mrs Gail Norbury, D Ruddy, L Izatt, H Deere, J Freeman, D Moore, N Trump, P Ross, M George, G Pichert, A Shaw, C Jacobs*
- (5.02) Understanding referral pathways to cancer genetics services for ethnic minorities - *Mrs Anna Allford, C Lewis, N Qureshi, J Kai*
- (5.03) Understanding influences on access to cancer genetics services for people from ethnic minorities - *Mrs Anna Allford, C Lewis, N Qureshi, J Kai*
- (5.04) The impact of screening and genetic registration on mortality and cancer incidence in Familial Adenomatous Polyposis - *Miss Katy Newton, E Mallinson, T Clancy, J Bowen, F Laloo, J Hill, DG Evans*
- (5.05) Intragenic Duplication and Deletion of BRCA1 in a single family - *Dr Sue Kenwick, K Oakhill, M Bowman, J Whittaker, H Stewart*
- (5.06) PTPN11 Testing in Patients with Suspected JMML: Unexpected finding of Noonan syndrome - *Miss Kimberley Slowther, S Jeffries, Y Wallis, M Griffiths*
- (5.07) Heritable transmission of a germline MLH1 hypermethylation epimutation in a family with Lynch syndrome - *Mr Stewart Payne, D Patel, D Ruddy, J Cook, J Field, N Shannon, C Eddy, R Harrison*
- (5.08) EGFR mutation testing in non-small cell lung cancer: The Bristol experience - *Miss Charlene Crosby, T Smye-Rumsby, M Plummeridge, J Braybrooke, A Dangoor, E De Winton, P Jankowska, M Williams*
- (5.09) Molecular analysis of gliomas: Aiding treatment decisions - *Miss Charlene Crosby, P Waits, T Smye-Rumsby, L Yarram, M Greenslade, C Herbert, K Hopkins, S Love, M Williams*
- (5.10) Pathological activation of mTOR pathway in sporadic pancreatic neuroendocrine tumours: the immunohistochemical evidence - *Dr Mark Tein, RA Clark, F Campbell, A Foulis, B Jasani, G Williams, S Wozniak, JR Sampson*
- (5.11) Parent-of-origin effect in SDHD hereditary tumours explained by requirement for additional molecular steps - *Dr Edward Tobias, PM Yeap, E Mavraki, A Fletcher, ME Freel, N Bradshaw, VA Murday, C Perry, RS Lindsay*
- (5.12) Germline mutations on the PALB2 gene are more frequent in women with contralate - *Dr Marc Tischkowitz, M Capanu, N Sabbaghian, L Li, MP Vallée, S Tavtigian, P Concannon, W Foulkes, J Bernstein, The WECARE Study Collaborative Group, C Begg*
- (5.13) Extending the Benign and Malignant Phenotypes Associated with Germline DICER - *Dr Marc Tischkowitz, A Bahubeshi, N Hamel, B Pasini, S Asiola, G Baynam, A Overkov, RP Frieder, M Dishop, N Graf, M Ekim, D Bouron-Dal Soglio, J Arseneau, RH Young, N Sabbaghian, A Srivastava, JR Priest, WD Foulkes*
- (5.14) Genetic counselling in a family with simultaneous heterozygosity of SDHD and SDHB mutations – *Miss Kate Sarson, SM Park, S Kenwick, H Simpson, F Laloo, L Bradley, E Maher, D Cilliers*

6. Other

- (6.01) Going Paperless-the GOSH Clinical Genetics experience - *Dr Ajith Kumar, A Kumar, A Bruce, C Skilbeck*
- (6.02) Barriers to developing services for molecular testing for retinal dystrophy - *Ms Georgina Hall, S Ramsden, R Combs, M McAllister, K Payne, S Downes, J Lowndes, A Moore, S Devery, G Wright, G Black*

British Human Genetics Conference 2011

5-7 September 2011

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 2011 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 ask that the trust ensures your name is referenced on the payment details.*

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 14 August.
- We regret that fees paid cannot be reimbursed if you have to cancel your registration after 14 August.
- We **CANNOT** supply invoices in respect of registration fees

CONFERENCE COSTS

Registration Fees

Full conference - 3 Full days
Any 2 Full days
Any 1 Full day

Member

£395.00
£350.00
£180.00

Non-Member

£550.00
£495.00
£250.00

Accommodation (per night)

Standard Single
En-suite Single

£40.00
£57.00

Conference Dinner (University of Warwick)

£45.00

Evening Meals Sunday or Monday night

£10.00

Car Parking FREE (please park in designed car parks 6, 7, 8A,15 – code needed for exit)

Delegates' Packs - We are again asking all delegates to collect their Conference Packs from the Registration Desks which, this year will be situated in the Arts Centre Upper Foyer. The Registration desks will be open from 17:00-20:00 on Sunday 4 September and from 07:30 on Monday 5 September - Wednesday 7 September. General information, including details of accommodation and map of campus, will be sent to you after the closing date of 14 August. For those staying on campus, room keys need to be collected from the "Rootes Building" (situated opposite to the Arts Centre).

CLOSING DATE FOR REGISTRATION:

14 August 2011

<http://www.bshg.org.uk/registration/bhgc2011.asp>

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British Society for Human Genetics

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