

# British Human Genetics Conference 2010

Monday – Wednesday 6 - 8 September

University of Warwick

Programme and Registration Booklet

Closing date for registration: 15 August 2010

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

- Neurogenetics
- Evidence Based Practice for Genetic Counselling
- Genome Informatics
- Prenatal / Congenital Anomalies
- Mitochondria and Disease
- Clinical and Molecular Genetics
- New Clinical Applications of Molecular Genetics Technology
- Basic and Translational Research
- Cancer Genomics and Molecular Diagnostics
- Genomics: where next after genome-wide association studies?
- Mechanisms of Disease
- Genomics and Health Promotion
- *Carter Lecture:*  
Professor Martin Bobrow “Reminiscences and speculations on the role of Genetics in Medicine”
- *BSHG Lecture:*  
Professor Nicholas Katsanis “Mutational Load and Human Genetic Disease”
- *Great Debate:*  
“This house believes that all genetic professionals have a duty to encourage families with inherited diseases to enter into research studies”
- *Young Researchers Forum* – “Non-traditional routes to a research career”
- *Scientists Engaging with the Media Workshop*
- *Research Funding talk from the National Institute for Health Research (NIHR)*
- *Trade Exhibitor - Seminars*

**LATE BREAKING ABSTRACTS**  
**Deadline 13 August 2010**  
Details on website [www.bshg.org.uk](http://www.bshg.org.uk)

## SCIENTIFIC PROGRAMME

The BSHG Scientific Programme Committee welcomes you to the 2010 meeting of the British Society for Human Genetics at the **University of Warwick**. The Scientific Programme Committee has compiled a wide-ranging programme comprising diverse symposia and outstanding UK and international speakers, including the Carter Lecture by Professor Martin Bobrow and the third BSHG Lecture by Professor Nicholas Katsanis. 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 2011 meeting!

**Eamonn Maher**  
*Chairman, Scientific Programme Committee 2010*

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

**Posters** are in Butterworth Hall and the Mead Gallery. Poster presenters will be asked to stand next to their posters on **TUESDAY 7 September**:

**Odd numbers (i.e. 1.01, 1.03 etc)** at 14:00-14:45

**Even numbers (i.e. 1.02, 1.04 etc)** at 14:45-15:30

**Carter Lecture** – Professor Martin Bobrow on "Reminiscences and speculations on the role of Genetics in Medicine"

**Third BSHG Lecture** – Professor Nicholas Katsanis on "Mutational Load and Human Genetic Disease"

**The Great Debate** – Monday evening (19:00-20:00 in The Theatre, Arts Centre). The motion for the debate is "This house believes that all genetic professionals have a duty to encourage families with inherited diseases to enter into research studies"

**Debaters** are Prof Martina Cornel, Dr Hugh Davies, Prof Sir John Burn, Mr Alastair Kent

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.

## YOUNG RESEARCHERS FORUM

### – "NON-TRADITIONAL ROUTES TO A RESEARCH CAREER"

A "Young Researchers Forum" meeting is scheduled for Tuesday lunchtime (13:00-14:00). Last year the topic was "How to obtain a research fellowship" with an eminent panel who had taken this route themselves and now sit on fellowship panels. This year we want to show that it is possible to forge a career in research even if you haven't followed this route and the panel members who will share their experience are Prof Karen Temple (Southampton) and Dr Helen Firth (Cambridge). (Please indicate on the registration form if you wish to attend)

## SCIENTISTS ENGAGING WITH THE MEDIA WORKSHOP

### Monday 6 September 14:15-15:15.

On 13 January 2010, the Dept for Business, Innovation and Skills "Science and the Media" expert group published its final report "Science and the Media: Securing the Future". This addressed some of the problems facing science journalism today and made recommendations for what the Government and others can do to help. Nearly half the science journalists surveyed in the report felt they were "passive recipients" of news, rather than uncovering stories themselves. This tells us that scientists need to be both active and savvy in order to secure timely and accurate media coverage of their research. Delegates attending this workshop will gain insight into some of the challenges and rewards of talking to the media; with tips and pointers on communicating effectively and advice from one of the UK's top science journalists. (Please indicate on the registration form if you wish to attend)

## Research Funding talk from the National Institute for Health Research (sponsored by the NIHR Collaborative Group for Genetics in Healthcare)

### Wednesday 8 September 12:30-13:30

A representative from the National Institute for Health Research Central Commissioning Facility (NIHR CCF) will talk about some of the NIHR research programmes, including Research for Patient Benefit (RfPB). The NIHR Central Commissioning Facility (NIHR CCF) was set up in 2006 to manage and administer research programmes funded by the NIHR and the Department of Health that investigate a range of healthcare matters and assist in how research-based knowledge is applied across all healthcare sectors. (Please indicate on the registration form if you wish to attend)

## TRADE EXHIBITOR SEMINARS

### Monday 6 September

**Roche Diagnostics** – 13:00-14:15 – Cinema, Arts Centre

**“Roche NimbleGen Cytogenetic Solution: Translating Data into Meaning“**

Lisa Shaffer, Signature Genomics: NimbleGen CGX Arrays and Genoglyphix: Instrumental Tools for New Syndrome Identification.

The presentation of Lisa Shaffer will describe the use of microarray-based comparative genomic hybridization (array CGH) for the identification of previously unrecognized genetic disorders and elucidated the etiologies of known syndromes. Using a proprietary database accessed through Signature Genomics lab-developed software program, Genoglyphix, analysis of multiple samples with over-lapping deletions has allowed for a genotype-first approach to the identification of new microdeletion syndromes and delineation of causative genes. From these overlapping deletions, the smallest region of overlap (SRO) is delineated and candidate genes are identified.

For life science research only. Not for use in diagnostic procedures.

NIMBLEGEN is a trademark of Roche.

Other brands or product names are trademarks of their respective holders. (Please register your interest direct with Roche – [dmd.contact@roche.com](mailto:dmd.contact@roche.com))

**NewGene** – 13:00-14:15 - SSO.21 Social Sciences Building

**“Introducing next generation sequencing and high throughput genotyping into clinical diagnostics“**

NewGene is a joint venture between Newcastle Hospitals NHS Foundation Trust and Newcastle University. We have developed a number of clinical diagnostic tests using the Roche GS-FLX and the Sequenom mass spectrometer. This seminar will cover the sequencing and genotyping services that we offer and an insight into the development journey. (Please register your interest direct with NewGene – [Jonathan.Robinson@newgene.org.uk](mailto:Jonathan.Robinson@newgene.org.uk) - all welcome)

### Tuesday 7 September

**InfoQuant** – 13:30-14:30 – Conference Room, Arts Centre

**“An approach to the analysis of aCGH data from multiplex microarray platforms in a clinical laboratory setting using the InfoQuant ‘CGH Fusion’ and ‘oneClickCGH’ software packages“**

The development of aCGH platforms with multiple arrays on a single slide has resulted in significant cost savings and made the use of aCGH as a viable first line test a realistic possibility in clinical cytogenetics laboratories. While the emergence of these array platforms has allowed the streamlining of the laboratory work associated with aCGH there is considerable scope for streamlining data processing and analysis. The use of software which enables the batch processing and documentation of both the array data and sample information is essential for the effective use of this technology in a clinical setting. InfoQuant “CGH Fusion” and “oneClickCGH” have been in use in the North East Thames Regional Cytogenetics Laboratory since January 2010 and have been used for the analysis of over 500 samples. This presentation will demonstrate how this software is currently being used for the analysis of data from the Nimblegen 12x135K microarray to offer aCGH as a first line test for patients with dysmorphic features and developmental delay. (Please register your interest direct with InfoQuant – [natalia@infoquant.com](mailto:natalia@infoquant.com) - all welcome)

**National Genetics Research Laboratories** – 14:00-15:00 - SSO.21 Social Sciences Building

**“Next Generation Sequencing and Clinical Genetics: A Beginner’s Guide“**

This workshop will provide an overview of NGS – existing and upcoming platforms and their data outputs. It will cover how the use of NGS is developing in clinical diagnostics and what the implications are for data handling and analysis. An ideal introduction for all clinical scientists who want to gain or improve their basic understanding of NGS, how it fits into diagnostics and what some of the issues are. (Please register your interest direct with NGRL - [Kathryn.Robertson@cmft.nhs.uk](mailto:Kathryn.Robertson@cmft.nhs.uk) - all welcome)

## 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 6 September in both Butterworth Hall and the Mead Gallery. This event is sponsored by some of our Exhibitors and features six wines, two each of red and white, as well as two charming rose wines, one of which is sparkling. Don't forget to talk to the exhibitors (and get your card validated for the chance to win an iPod), while you are there!

**Conference Party** – Following the rip-roaring success of the conference party last year, we will be staying on campus at the University again in 2010. We'll be starting off with a Bucks Fizz, before a delicious meal featuring supreme of chicken with truffle and pistachio for meat eaters or the vegetarian alternative of vegetable cassoulet with roasted basil infused haloumi. The meal also includes starter, champagne sorbet and dessert, and is followed by coffee and single-origin chocolates. Once again, we will be entertained after dinner by The Poptarts, who provided such excellent live music, resulting in some very exuberant dancing a year ago. We look forward to seeing you there.

### **Lunch on Wednesday 8 September**

Lunch on Wednesday is designed for flexibility (conference finishes at 12:00). Instead of balancing a buffet plate, you will be provided with a scrumptious picnic lunch to enjoy with friends relaxing outside on the beautiful Warwick campus (weather permitting) or to take away with you, if you are rushing to be off.

### **Ramadan**

Once again, this meeting falls during the month of Ramadan. If any delegates are observing a fast during the time of the meeting and require special arrangements for an early breakfast, please contact Dina Kotecha (0121 627 2634)

### **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 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 15 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 15 August.
- we regret that fees cannot be reimbursed if you have to cancel your registration after 15 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 17:00-21:00 on Sunday 5 September - from 07:30 on Monday /Tuesday 6-7 September – from 08:00 on Wednesday 8 September. General information, including details of accommodation, map of campus and travel information, will be sent to you after the closing date of 15 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 – 17:00 Sunday 5 September - Room SS0.10 Social Sciences Building*

*Annual General Meeting -17:30 Monday 6 September – The Theatre, Arts Centre*

### ASSOCIATION OF GENETIC NURSES AND COUNSELLORS

*Members Meeting – 15:15 Tuesday 7 September – Conference Room, Arts Centre*

## OTHER MEETINGS

### Monday 6 September

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

**Genetic Counselling Registration Board** – 13:00-14:00 – *SSO.10 Social Sciences Building*

### Tuesday 7 September

**Lead Clinicians Meeting** – 13:30-14:30 - *SSO.09 Social Sciences Building*

**CGS Clinical Governance Committee Meeting** – 13:30-14:30 - *SSO.08 Social Sciences Building*

## FUTURE MEETINGS

### BRITISH SOCIETY FOR HUMAN GENETICS

UNIVERSITY OF WARWICK	2011 – 5-7 September
	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](mailto:bshg@bshg.org.uk) Website: [www.bshg.org.uk](http://www.bshg.org.uk)

# Sunday 5 September 2010

17:00-21:00 **REGISTRATION** - Upper Foyer, Arts Centre

17:00-19:00 **BSHG – Council Meeting** – Room: SSO.10, Social Studies Building

# Monday 6 September 2010

07:30 **REGISTRATION**

## 09:30-11:00 Concurrent Symposia

<b>Neurogenetics</b>		<b>Chair: Dr Julia Rankin / Dr Andrew Jackson</b>	<b>Room: The Theatre, Arts Centre</b>
09:30	(SP01)	An overview of the Genotype and Phenotype of Neurotransmitter Disorders – <i>Dr Manju Kurian (Birmingham)</i>	
09:45	(SP02)	Tubulin Genes and Cortical Malformations - <i>Dr Daniela Pilz (Cardiff)</i>	
10:00	(SP03)	The future of diagnostics in cerebellar ataxias – <i>Dr Andrea Nemeth (Oxford)</i>	
10:15	(SP04)	Molecular Diagnosis of congenital cranial dysinnervation disorders – <i>Dr Elizabeth Young (Exeter)</i>	
10:30	(SP05)	Clinical and molecular diagnostics of disorders demonstrating intracranial calcification – <i>Prof Yanick Crow (Manchester)</i>	
10:45	(SP06)	Mendelian disorders of brain growth – <i>Dr Geoff Woods (Cambridge)</i>	

<b>Genome Informatics</b>		<b>Chair: Mr Dominic McMullan / Dr David Gokhale</b>	<b>Room: The Cinema, Arts Centre</b>
09:30	(SP07)	IT tools for interpretation of copy number variation in context of the patient phenotype – <i>Dr Steven Van Vooren, YM Yves Moreau (Leuven, Belgium)</i>	
10:00	(SP08)	Issues in interpreting and using genome-wide sequence data – <i>Dr Richard Durbin (Cambridge)</i>	
10:30	(SP09)	Building resources and tools for genomic medicine – <i>Dr Tim Hubbard (Cambridge)</i>	

<b>Evidence based practice for genetic counselling</b>		<b>Chair: Mrs Cath King / Ms Sheila Slater</b>	<b>Room: Conference Room, Arts Centre</b>
09:30	(SP10)	Overview - the genetic counsellor contribution to developing the evidence for genetic counselling practice - <i>Prof Heather Skirton (Taunton)</i>	
09:50	(SP11)	Development of a coherent, multi-disciplinary research programme – <i>Dr Marion McAllister (Manchester)</i>	
10:05	(SP12)	The role of patient organisations in research to support delivery of genetic counselling – <i>Celine Lewis (London)</i>	
10:20	(SP13)	Working as part of a multidisciplinary research team – <i>Ms Chris Jacobs (London)</i>	
10:35	(SP14)	Applying the research to clinical policies and practice - a European perspective – <i>Dr Christine Patch (London)</i>	
10:50		Panel discussion and questions	

11:00-11:25 **COFFEE**

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

## 11:30-13:00 Symposium: Prenatal/Congenital Anomalies

<b>Chair: Dr Lyn Chitty / Dr Helen Firth</b>		<b>Room: The Theatre, Arts Centre</b>
11:30	(SP15)	Referral Testing Patterns for Prenatal Microarray Testing in North America - Dr Lisa Shaffer (Spokane, Washington, USA)
12:00	(SP16)	Selfish mutations in spermatogenesis and paternal age effects - Prof Andrew OM Wilkie (Oxford), A Goriely, GAT McVean, E Rajpert-De Meyts
12:30	(SP17)	Fetal MR for CNS malformations - Prof Paul Griffiths (Sheffield)

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

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

13:00-14:00 **Genetic Counsellor Registration Board Meeting**  
Room: SSO.10 Social Sciences Building

13:00-14:15 **Roche Diagnostics Seminar – Roche NimbleGen Cytogenetic Solutions: Translating Data into Meaning**  
Room: Cinema, Arts Centre  
Speaker: Lisa Shaffer (Signature Genomics)

13:00-14:15 **NewGene Seminar – Introducing next generation sequencing and high throughput genotyping into clinical diagnostics**  
Room: SSO.21 Social Sciences Building  
Speakers: Dr Ann Curtis and Dr Jonathan Coxhead (Newgene)

## 14:15-15:15 Symposium: Mitochondria and Disease

<b>Chair: Prof Judith Goodship / Dr Julia Rankin</b>		<b>Room: The Theatre, Arts Centre</b>
14:15	(SP18)	Mitochondrial disorders presenting in adult life: new mechanisms and new treatments - Prof Patrick Chinnery (Newcastle)
14:35	(SP19)	The childhood presentation of mitochondrial disease - Dr Robert McFarland (Newcastle)
14:55	(SP20)	The investigation and diagnosis of mitochondrial genetic disease - Prof Rob Taylor (Newcastle)

## 14:15-15:15 Scientists Engaging with the Media

<b>Chair: Dr Jess Buxton (BSHG Press Officer), Dr Vivienne Raper (Science Editor of BioNews)</b>		<b>Room: Conference Room, Arts Centre</b>
Speakers:	Mr Mark Henderson (Science Editor of the Times) and Mr Tom Sheldon (Press Officer at the Science Media Centre)	

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

## 16:00-17:30 Concurrent Symposia

<b>New clinical applications of molecular genetics technology</b>		<b>Chair: Prof Sian Ellard / Dr Jonathan Waters</b> <b>Room: The Theatre, Arts Centre</b>
16:00	(SP21)	Epilepsy genetics: what is common, what is rare? - Prof Sanjay Sisodiya (London)
16:30	(SP22)	New approaches to the molecular dissection of human obesity - Dr Alexandra Blakemore (London)
17:00	(SP23)	Next generation sequencing in the clinic – Dr Joris Veltman (Nijmegen, The Netherlands)

<b>Clinical and Molecular Genetics</b>		<b>Chair: Dr Julian Adlard / Dr Rob Elles</b> <b>Room: The Cinema, Arts Centre</b>
16:00	(SP24)	Fumarate hydratase and mitochondrial predisposition to cancer – <i>Prof Ian Tomlinson (Oxford)</i>
16:30	(SP25)	Biallelic mismatch repair defects – <i>Dr Eamonn Sheridan (Leeds)</i>
17:00	(SP26)	The genetics of nevus count variation, pigmentation and melanoma – <i>Prof Tim Bishop (Leeds)</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**

<b>Chair: Prof Jonathan Montgomery / Prof Anneke Lucassen</b>	<b>Room: The Theatre, Arts Centre</b>
This house believes that all genetic professionals have a duty to encourage families with inherited diseases to enter into research studies	
Debaters	<ul style="list-style-type: none"> <li>- <b>For:</b> Prof Sir John Burn (Newcastle) and Mr Alastair Kent (Genetic Alliance UK)</li> <li>- <b>Against:</b> Prof Martina Cornel (Amsterdam) and Dr Hugh Davies (NRES Ethics Adviser)</li> </ul>

## Tuesday 7 September 2010

07:30 **REGISTRATION**

**09:00-10:30 Concurrent Sessions**

<b>Molecular</b>	<b>Chair: Ms Su Stenhouse / Prof Peter Scambler</b>	<b>Room: The Theatre, Arts Centre</b>
09:00	(SP27) Mutations in TMEM216 perturb ciliogenesis and cause Joubert, Meckel and related syndromes - <i>Miss Clare Logan, EM Valente, S Mougou-Zerelli, JH Lee, JL Silhavy, F Brancati, M Iannicelli, L Travaglini, S Romani, B Illi, M Adams, K Szymanska, JE Lee, S Thomas, E Davis, M Vekemans, N Katsanis, T Attié-Bitach, JG Gleeson, CA Johnson</i>	
09:15	(SP28) Identification of mutations in FLVCR2 in patients with proliferative vasculopathy and hydranencephaly-hydrocephaly syndrome (Fowler syndrome) - <i>Dr Esther Meyer, C Ricketts, NV Morgan, MR Morris, S Pasha, LJ Tee, F Rahman, A Bazin, B Bessieres, P Dechelotte, MT Yacoubi, M Al-Adhani, T Marton, D Tannahill, RC Trembath, C Fallet-Bianco, P Cox, D Williams, ER Maher</i>	
09:30	(SP29) Phospholipase C Beta 1 (PLCB1) Deficiency is associated with Early Onset Epileptic Encephalopathy - <i>Dr Manju Kurian, E Meyer, G Vassallo, NV Morgan, N Prakash, N Hai, S Shuib, F Rahman, E Wassmer, JH Cross, F O'Callaghan, JP Osborne, IE Scheffer, P Gissen, ER Maher</i>	
09:45	(SP30) Mutations in X-linked RPGR can modify the Bardet-Biedl Syndrome phenotype by altering RPGR protein levels - <i>Dr Anna Diaz-Font, AT Moore, AJ Hardcastle, PL Beales</i>	
10:00	(SP31) Mutations in TTC37 cause trichohepatoenteric syndrome (phenotypic diarrhoea of infancy) - <i>Dr Jane Hartley, NC Zachos, B Dawood, M Donowitz, J Forman, RJ Pollitt, NV Morgan, L Tee, P Gissen, WHA Kahr, AS Knisely, S Watson, D Chitayat, IW Booth, S Protheroe, S Murphy, E de Vries, DA Kelly, ER Maher</i>	
10:15	(SP32) Mutations in the dynein assembly factor PF22 cause primary ciliary dyskinesia with absent outer dynein arms - <i>Dr Hannah Mitchison, J Freshour, H Olbrich, H Blau, RM Gardiner, H Mussaffi, H Omran, EMK Chung, DR Mitchell</i>	

<b>Clinical/Cancer</b>		<b>Chair: Dr Lucy Side / tbc</b>	<b>Room: The Cinema, Arts Centre</b>
09:00	(SP33)	A Prospective Study of Children Born to Mothers With Epilepsy; Outcome at Six Years - Prof Jill Clayton-Smith, R Bromley, G Baker, G Mawer, S Banka	
09:15	(SP34)	Advances in the development of new therapies for CYLD mutation carriers: Targeting Trk. - Dr Neil Rajan, R Elliott, CJ Lord, J Burn, M Sieber-Blum, A Ashworth	
09:30	(SP35)	Mutations in HPSE2 cause urofacial syndrome - Dr Sarah Daly, JE Urquhart, H Stuart, M Lewis, B Kerr, D Donnai, AS Woolf, GC Black, WG Newman	
09:45	(SP36)	Fine-Scale mapping of breast cancer loci: From tag SNP to causal variant - Dr Maya Ghousaini, BCAC	
10:00	(SP37)	Clinical investigation of the UK cohort of FLCN mutation positive Birt-Hogg-Dube syndrome patients - Dr Derek Lim, ER Woodward, G Kirby, F Macdonald, L Izatt, L Walker, L Side, SV Hodgson, PJ Morrison, ER Maher	
10:15	(SP38)	What makes you very small? - Dr Andrew Jackson	

<b>Counselling, Social, Policy and Education</b>		<b>Chair: Dr Laura Boyes / Mrs Shagufta Khan</b>	<b>Room: Conference Room, Arts Centre</b>
09:00	(SP39)	Cross-cultural values of 'informed choice' in antenatal genetic screening - Dr Z Tizro, S Ahmed, LD Bryant, D Shickle	
09:15	(SP40)	Attitude of Pakistani parents of affected children and their relatives towards prenatal diagnosis & termination of Pregnancy - Dr Mushtaq Ahmed, S Ahmed, SM Sharif, CE Chu, GR Taylor	
09:30	(SP41)	Trying again; the experience of repeated prenatal diagnosis for a genetic condition - Ms Vicki Wiles, FL Raymond, K Crozier, A Harris, N Hadlett	
09:45	(SP42)	Developing the nursing role: Inherited cardiac conditions (ICC) services utilising BHF Cardiac Genetic Nurses: an evaluation - Dr Maggie Kirk, E Tonkin, A Simpson, D Cohen, A Edwards, M Llewellyn, M Longley	
10:00	(SP43)	Consent and Confidentiality: Issues for genetic practice. - Prof Anneke Lucassen, A Hall	
10:15	(SP44)	CBAVD or mild cystic fibrosis? Identification of a rare, second, CFTR mutation, previously classified as severe, in an otherwise asymptomatic man presenting with congenital bilateral absence of the vas deferens - Mr Peter Marks, H Cox, S Tabrah	

<b>Cytogenetics</b>		<b>Chair: Miss Kath Smith / Ms Tori Anthony-Dubernet</b>	<b>Room: Studio, Arts Centre</b>
09:00	(SP45)	Prenatal microarray testing for fetuses with a structural anomaly on ultrasound scan at Birmingham Women's Hospital - Mr Dominic McMullan, SC Hillman, FS Togneri, GK Hall, SA Larkins, A Evans, D Williams, ER Maher, EV Davison, MK Kilby	
09:15	(SP46)	Chromosome breakage in human preimplantation embryos from carriers of structural chromosomal abnormalities in relation to fragile sites, maternal age and poor sperm factors. - Miss Leoni Xanthopoulou, H Ghevaria, A Mantzouratou, P Serhal, A Doshi, JD Delhanty	
09:30	(SP47)	De novo chromosome deletions and duplications diagnosed by array CGH show an excess of paternally derived cases - Miss Charlene Sibbons, J Crolla, J Morris, P Jacobs, S Thomas	
09:45	(SP48)	Chromosome ploidy status in early colorectal polyps and the viability of whole genome amplified FFPE tissue DNA for aCGH - Miss Gemma Forsyth	
10:00	(SP49)	Review of genetic testing in lipomatous tumours - Mr Paul Roberts, WJ Merchant, S Edward	
10:15	(SP50)	Genome-wide analysis of chromosomal alterations in chronic lymphocytic leukaemia - Dr Jon Strefford, M Rose-Zerilli, A Parker, A Gardiner, BD Young, T Chaplin, DG Oscier	

10:30-11:15

**COFFEE**

11:15-12:45 *Concurrent Symposia*

<b>Basic and Translational Research</b>		<b>Chair: Prof Sir John Burn / tbc</b>	<b>Room: The Theatre, Arts Centre</b>
11:15	(SP51)	Pathway-Based Analysis of Oestrogen Receptor Biology in Breast Cancer Susceptibility – <i>Dr Edison Liu (Singapore)</i>	
11:45	(SP52)	Hereditary spastic paraplegias: membrane traffic and the motor pathway – <i>Dr Evan Reid (Cambridge)</i>	
12:15	(SP53)	mTOR inhibition: from cancer to cognition – <i>Prof Julian R Sampson (Cardiff), D Mark Davies, Ming Hong Shen, Andrew Tee (Prof Julian Sampson's lecture is sponsored by the NIHR Collaborative Group for Genetics in Healthcare)</i>	

<b>Cancer Genomics and Molecular Diagnostics</b>		<b>Chair: Dr Jon Strefford / Dr Teresa Davies</b>	<b>Room: The Cinema, Arts Centre</b>
11:15	(SP54)	SNP array-based genomics in CLL: Lessons learned - <i>Dr Sami Malek (Michigan, USA)</i>	
11:45	(SP55)	Telomere dynamics, instability and fusion during the progression to malignancy - <i>Dr Duncan Baird (Cardiff)</i>	
12:15	(SP56)	Molecular Pathology: genes, chromosomes and personalised medicine - <i>Dr David Gonzalez de Castro (Sutton, Surrey)</i>	

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

13:00-14:00 **Young Researchers Forum: Non-traditional routes to a research career**

Chair: Prof Judith Goodship / Dr Emma Burkitt-Wright  
Room: Foyer to SSO.21, Social Sciences Building

13:30-14:30 **Lead Clinicians Meeting**

Room: SSO.09, Social Sciences Building

13:30-14:30 **CGS Clinical Governance Committee Meeting**

Room: SSO.08, Social Sciences Building

13:30-14:30 **InfoQuant Seminar – An approach to the analysis of aCGH data from multiplex microarray platforms in a clinical laboratory setting using the infoQuant ‘CGH Fusion’ and ‘oneClickCGH’ software packages**

Room: Conference Room, Arts Centre

Speakers: Rodger Palmer (Great Ormond Street, London) and Anton Petrov (Infoquant)

14:00-15:00 **National Genetics Reference Laboratories – Next Generation Sequencing and Clinical Genetics: A Beginners Guide**

Room: SSO.21, Social Sciences Building

14:00-15:30 *Symposium: Genomics and Health Promotion*

<b>Chair: Dr Layla Al-Jader</b>		<b>Room: The Cinema, Arts Centre</b>
14:00		Introduction by chair
14:05	(SP57)	Predicting the risk of coronary heart disease – <i>Dr Tom Dent (Cambridge)</i>
14:25	(SP58)	Family history of heart disease: A qualitative follow-up study on its role in prevention - <i>Dr Paula Saukko (Loughborough)</i>
14:45	(SP59)	Does knowledge of genetic risk motivate people to adopt healthy behaviours? - <i>Prof Susan Michie (London)</i>
15:05		Questions and answer session

15:15-15:45 **Association of Genetic Nurses and Counsellors Members Meeting**

Room: Conference Room, Arts Centre

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

<b>Chair: Dr Frances Flinter / Prof Andrew Green</b>		<b>Room: The Theatre, Arts Centre</b>
16:00	(SP60)	ATRX targets variable number tandem repeats and thereby influences allele-specific expression - <i>Dr Karen Lower, M Law, I Dunham, J Hughes, H Ayyub, D Higgs, R Gibbons</i>
17:00	(SP61)	Fine-scale Survey of X Chromosome Copy Number Variants Underlying Intellectual Disability - <i>Dr Lucy Raymond, AC Whibley, V Plagnol, PS Tarpey, F Abidi, T Fullston, L Willatt, M Shaw, RE Stevenson, G Turner, M Field, A Hackett, CE Schwartz, J Gecz, MR Stratton</i>

**17:15-18:15 CARTER LECTURE**

<b>Chair: Dr Frances Flinter</b>		<b>Room: The Theatre, Arts Centre</b>
(SP62)	Reminiscences and speculations on the role of Genetics in Medicine - <i>Prof Martin Bobrow (Cambridge)</i>	

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

## Wednesday 8 September 2010

08:00 **REGISTRATION**

**08:45-10:15 Symposium: Genomics: where next after genome-wide association studies?**

<b>Chair: Dr Alex Blakemore / tbc</b>		<b>Room: The Theatre, Arts Centre</b>
08:45	(SP63)	Where next for genetics of type-2 diabetes? - <i>Prof Philippe Froguel (London)</i>
09:15	(SP64)	Karyotypes to CNVs to Exomes to Genomes: New genomic technologies, old genetics questions – <i>Dr Matthew Hurles (Cambridge)</i>

9:45-10:15 **COFFEE**

**10:15-11:15 Symposium: Mechanisms of Disease**

<b>Chair: Dr Christine Patch / Prof Eamonn Maher</b>		<b>Room: The Theatre, Arts Centre</b>
10:15	(SP65)	Defects in VIPAR and VPS33B cause Arthrogyriposis, Renal dysfunction and Cholestasis syndrome (ARC) and disturbed apical-basolateral polarity – <i>Dr Paul Gissen (Birmingham)</i>
10:45	(SP66)	Stress and Genes – <i>Prof Jonathan Flint (Oxford)</i>

**11:15-12:00 BSHG LECTURE**

<b>Chair: Dr Christine Patch</b>		<b>Room: The Theatre, Arts Centre</b>
11:15	(SP67)	Mutational Load and Human Genetic Disease – <i>Prof Nicholas Katsanis (Baltimore, USA)</i>

12:30-13:30 **Research Funding talk from the National Institute for Health Research (NIHR)** - Room: SSO.21 Social Sciences Building. Sponsored by the NIHR Collaborative Group for Genetics in Healthcare

12:45-14:00 **LUNCH and End of Conference**

# Poster presentations

## 1. Clinical genetics

- (1.01) Cranial osteolysis, skin changes, growth retardation and developmental delay: a new syndrome? - *Dr Katta Girisha, AD Varghese, V Manju, KV Rajagopal, BS Baliga*
- (1.02) Role of connexin genes (Cx26, Cx30 and Cx31) in deafness genesis in Kirov Region (Russia) - *Ms Elizaveta Sharonova, RA Zinchenko*
- (1.03) Cafe for Routine Genetic Data Exchange (Cafe RouGE) - *Dr Owen Lancaster, D Atlan, GA Thorisson, A Webb, AJ Brookes*
- (1.04) The Genetic Variations that cause Down syndrome and Associated Oro-Dental Disorders - *Prof Emilia Severin, A Stan, S Solonaru, L Savu, D Stanciu*
- (1.05) C propeptide mutations in COL3A1 cause both typical and atypical forms of vascular EDS IV - *Prof Francis Michael Pope, S Holden, DJP Ferguson, P Sawle, M Nesbitt, R Pollitt, A Dalton*
- (1.06) Applying clonal sequencing to genetics testing pathways - *Prof Graham Taylor, CE Chu, E Sheridan, K Cohen, J Morgan, H Fraser, RL Robinson, M Davis, M Sweeney, P Roberts, A Tzika*
- (1.07) An unusual mutation in LMX1B presenting with craniosynostosis - *Mr Samintharaj Kumar, SRF Twigg, JA Hurst, S Tomkins, SA Wall, AOM Wilkie*
- (1.08) Non-invasive prenatal diagnosis (NIPD) for fetal sex in clinical practice: accurate information, fewer invasive tests and no additional health care costs - *Dr Melissa Hill, S Taffinder, K Finning, P Martin, J Hogg, C Meaney, G Daniels, S Morris, L Chitty*
- (1.09) A broad phenotypic spectrum associated with mutation in SHOC2 - *Dr Emma Burkitt Wright, J Shorto, BA Kerr*
- (1.10) Making the case for expanded newborn screening in the UK - *Dr Hilary Burton, S Moorthie, GS Sagoo, JR Bonham*
- (1.11) 7q11.23 microduplication- A recognisable phenotype - *Dr Abhijit Dixit, KE Martin, NM Smith, H Cheema, A Sharif, A Sarkar*
- (1.12) Molecular genetic analysis of familial congenital heart disease (CHD) - *Dr Chirag Patel, NV Morgan, H Cox, ER Maher*
- (1.13) The Deciphering Developmental Disorders (DDD) project - *Dr Helen V Firth, DR Fitzpatrick, M Parker, AP Bevan, M Corpas, E Bragin, S Clayton, J Barrett, ME Hurler, NP Carter*
- (1.14) The Enhanced Genetic Services Project (EGSP) - *Mr Yasar Eltaf, D Williams*
- (1.15) Cat Eye Syndrome: The Northern Ireland experience - *Dr Lisa Bradley, K Murtagh, V McConnell*
- (1.16) A deletion of FGFR2 creating a chimeric IIIb/c exon in a child with Apert syndrome - *Miss Aimee Fenwick, S Bowdin, R Klatt, AOM Wilkie*
- (1.17) X-linked VACTERL-Hydrocephalus syndrome: Further delineation of the phenotype caused by mutations in FANCB - *Dr Simon Holden, J McCauley, N Masand, R McGowan, A Hunter, M Richard, S Rajagopalan, K Gibson, J Robertson, F Vas, S Abbs*
- (1.18) The DYSCERNE project has developed clinical management guidelines for Williams, Angelman, Noonan and Kabuki syndromes - *Prof Jill Clayton-Smith, K Strong, P Griffiths, C Harrison, R Day, S Gardner, D Donnai, B Kerr, K Metcalfe, I van der Burgt, B Dallapiccola, K Devriendt, M Krajewska-Walasek, N Philip*
- (1.19) Enquiring about a family history of cancer in primary care: a pilot project - *Dr Lynn Greenhalgh, C Kightley, L Mitchell, E Jolly, S Skillcorn, A Fazackerley, D Duncan, J Hughes, L Wilson, S Parr, A Kenney, N Cropper, S Reynolds, G Allan*
- (1.20) A multi-disciplinary approach to the diagnosis of ALL - *Dr Jeremy Hancock, M Nobbs, A Palmer, D Delmege, R Hathway, P Archer, P Waits, H Stokes, C Wragg, J Moppett, E Roberts*
- (1.21) Atypical haemolytic uraemic syndrome in one large kindred predisposed by factor H mutation: penetrance data based on mutation testing and family history - *Dr Francis H Sansbury, G Bromilow, C Bingham, R Powell, A Nicholls, RA Oram, L Strain, THJ Goodship, PD Turnpenny*
- (1.22) Tetrasomy 9p: Pre and Postnatally recognisable syndrome - *Dr Tabib Dabir, S McCullough, L Rauch, G Smith, S McKee*
- (1.23) Microduplication at 10q24 causing Split Hand Foot Malformation (SHFM) in an extensive pedigree - *Dr Victoria Harrison, R Williams, L Connell, U Kini, E Blair*
- (1.24) Compound heterozygous deletions involving NRXN1 causing severe MR and epilepsy - *Dr Victoria Harrison, L Connell, M Pike, E Blair*
- (1.25) Clinical phenotypes associated with imbalance at the 16p11.2 autism susceptibility locus - *Dr Neeti Ghali, C Ogilvie, FA Flinter, M Irving*
- (1.26) Clinical phenotypes associated with imbalance at 15q13.3 - *Dr Neeti Ghali, L Nashef, N Elmslie*

- (1.27) Clinical Governance Strategies in Clinical Genetics - *Dr Sarah Smithson, EE Hobson, A Lucassen, G Woods, H Murphy, R Cole, C Giblin, MA James, F Flinter*
- (1.28) Screening for Fragile X syndrome using both clinical checklist and PCR: Preliminary experience in a developing country - *Dr Zarina Abdul Latiff, WC Lim, KB Amin Noordin, Z Othman, SM Then, R Harun, SZ Syed Zakaria, R Jamal*
- (1.29) Interstitial deletion within the short arm of chromosome 3, region p12.3 to p14.2 in a patient with CHARGE syndrome - *Dr Bianca Desouza, RJ Ellis, B Bewes, E Wakeling*
- (1.30) An evaluation of the use of array CGH in a tertiary cleft service - *Dr Helen Linden, L Connell, J McPartland, C Noakes, K Smith, T Goodacre, U Kini*
- (1.31) DYSCKERNE: Results from the full European launch of the Electronic Dysmorphology Diagnostic System (DDS) - *Dr Sara Gardner, R Day, PM 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*
- (1.32) A child with an APC mutation presenting with Gardener-associated fibroma and bilateral renal cysts - *Dr Anjana Kulkarni, A Burke, C Fisher, C Roberts, SJ Payne, E Wakeling*
- (1.33) De novo detection of X chromosome q28 duplication in association with Inflammatory Linear Verrucous Epidermal Naevus (ILVEN) - *Dr Naomi Webber, C Ogilvie, J Mellerio, ST Holden*
- (1.34) Agenesis of the corpus callosum in mosaic tetrasomy 8p - *Dr Brian Wilson, C Harikumar, RB Fisher*
- (1.35) A retrospective phenotypic/genotypic analysis of patients with Noonan Syndrome in Northern Ireland - *Dr Gillian Rea, D MacSorley, C Blane, A Magee*
- (1.36) Novel POLG mutation causing distal myopathy and cachexia - *Dr Robert Pitceathly, JL Holton, I Hargreaves, S Heales, C Woodward, MG Sweeney, MB Davis, J Evans, C Smith, C Fratter, S Rahman, MG Hanna*
- (1.37) A genetic investigation proforma for individuals with bowel polyps and a family history of colorectal carcinoma - *Dr Neeta Lakhani, M Yousaf, J Barwell*
- (1.38) Jumping Hurdles: Experiences of Predictive Testing for Huntington's Disease in Young Adults - *Miss Natalie Ellery, S Sarangi*
- (1.39) Array-CGH in patients with cleft lip and/or palate - *Dr Adam Shaw, R Palmer, A Barnicoat, MB Bitner-Glindzic, VKA Kumar, A Male, J Waters, M Lees, LS Chitty*
- (1.40) The impact of array CGH in Wessex - *Dr Andrew Douglas, JA Crolla, IK Temple*
- (1.41) SureSelect in-solution hybridization target enrichment for whole exome sequencing: coverage and efficacy - *Mr Shibu John, A Roberts, J Ware, S Cook*
- (1.42) Similar neuropsychiatric phenotypes in two patients with periventricular nodular heterotopia and Filamin A gene mutations - *Dr Andrew Fry, MP Kerr, SP Robertson, DT Pilz*
- (1.43) Multidisciplinary clinics at Guy's Hospital - Where we are now, but what does the future hold? - *Ms Sally Watts*
- (1.44) Juvenile Huntington's Disease: history and prevalence - *Miss Nadia Peppia, K O'Donovan, OWJ Quarrell*
- (1.45) Thirty-year trends in prevalence and diagnosis of Trisomy 18 and Trisomy 13 - *Dr Tabib Dabir, A Magee, G Smith, D Grier, A Bradley, P Morrison*
- (1.46) ARC Syndrome – Report of a milder case and genotype-phenotype correlation - *Miss Holly Smith, R Romero, A Straatman-Iwanowska, CK Bruce, AR Cullinane, AS Knisely, P Gissen*
- (1.47) The use of BRUM1 resequencing microarray to identify mutations in patients with cholestatic disorders - *Dr Christopher K Bruce, J Hartley, K McKay, R Brown, U Baumann, P Clayton, E Sturm, B Udd, P McKiernan, I Shah, R Mirza, H Cheema, D McCullen, F MacDonald, ER Maher, AS Knisely, CJ Hendriksz, DA Kelly, P Gissen*

## 2. Molecular Genetics

- (2.01) Molecular Genetic Diagnostic Service for Limb Girdle Muscular Dystrophy 2B/Miyoshi Myopathy - *Miss Eileen Graham, J Hudson, L Stanley, K Bushby, D Bourn*
- (2.02) Locus heterogeneity and novel TSHR mutations in consanguineous families with congenital non-goitrous hypothyroidism - *Dr Hakan Cangul, H Saglam, Z Aycan, T Yakut, T Gulten, NV Morgan, JR Forman, M Karkucak, E Eren, S Cetinkaya, V Bas, K Demir, SA Yuca, GA Kirby, S Pasha, M Kendall, W Hogler, TG Barret, ER Maher*
- (2.03) Genetic Testing for Congenital nonsyndromic deafness (DFNB1) - *Prof Emilia Severin, C Dragomir, A Stan, C Albu, D Albu, DT Stefanescu, L Savu*
- (2.04) False negative result caused by allele drop out in the Caveolin 3 gene - *Dr Louise Stanley, J Hudson, K Bushby*
- (2.05) New molecular services for the diagnosis of neonatal cholestasis - *Miss Kirsten McKay, J Hartley, C Bruce, P Gissen, F Macdonald*

- (2.06) Implementation of nucleophosmin (NPM1) screening in acute myeloid leukaemia (AML) patients - *Miss Rachel Coleman, S Akiki, M Griffiths, F Macdonald*
- (2.07) Atypical 22q11 deletion detected by multiplex ligation-dependent probe amplification (MLPA) in patients referred for Prader-Willi Syndrome testing - *Ms Karen Meaney, B O'Hici, SA Lynch, DE Barton*
- (2.08) Validation of a novel mutation screening strategy for Familial Hypercholesterolaemia: LIPOchip, a DNA-array based system - *Mr Jonathan Callaway, O Wood, E Cross, AC Skinner, JF Harvey*
- (2.09) Testing for digenic inheritance in SCL26A4 heterozygotes - *Ms Ann-Marie Differ, K Patel, M Bitner-Glindzicz, N Lench, L Jenkins*
- (2.10) Analysis of the type 5 collagen genes in a cohort of patients with Classic Ehlers Danlos Syndrome - *Ms Rebecca Pollitt, M Nesbitt, G Sobey, M Pope, B Cornejo, A Morsman, A Dalton*
- (2.11) Development of a molecular genetic diagnostic service for Waardenburg syndrome Type 1 and Type 3 - *Miss Fiona McKay, L Jenkins, M Bitner-Glindzicz, N Lench*
- (2.12) Identification of two patients who are double heterozygotes for Familial Hypercholesterolaemia and Familial Defective Apolipoprotein B-100 - *Miss Sarah Fielding, A Taylor, K Patel, G Bayly, A Waise, A Luvai, G Norbury, S Humphries, L Jenkins, N Lench*
- (2.13) Improved first level mutation screening for Autosomal Dominant Hypercholesterolaemia using the Fluidigm BioMark™ platform - *Miss Sarah Fielding, A Taylor, L Jenkins, N Lench*
- (2.14) Solid Phase Sequence Capture vs PCR: Targeted Enrichment of Muscular Dystrophy genes for Next Generation Sequencing - *Mr Andrew Frey, J Coxhead, A Curtis*
- (2.15) Mutations in Autosomal Recessive Osteogenesis Imperfecta - *Dr Isabel Mandy Nesbitt, AC Morsman, B Cornejo, F van Dijk, A Dalton, R Pollitt*
- (2.16) Epidermal Growth Factor Receptor (EGFR) Mutation Testing in non-small cell lung cancer (NSCLC) patients - *Dr Andrew Wallace, E Howard, L Dutton, RG Elles, W Newman*
- (2.17) Reducing the need to Southern blot for fragile X; an evaluation of the Asuragen AmplideX FMR1 kit - *Miss Nicola Ibberson, JA Drummond, R Treacy*
- (2.18) Autozygosity mapping and linkage analysis identify two novel loci for juvenile myoclonic epilepsy on chromosomes 4q32-q35 and 5q31-q33 - *Dr Kate Everett, K Nikali, AR Altahan, R Robinson, RM Gardiner*
- (2.19) Homozygous gene deletion of KCNE1 resulting in Jervell and Lange-Nielsen Syndrome - *Dr Melanie Proven, K McGuire, K Thomson, E Blair, R Belk, D Umapathy, D Trump, A Seller*
- (2.20) Familial hypercholesterolaemia: a 1 year audit of molecular testing at Bristol - *Miss Laura Yarram, S Burton-Jones, M Balasubramani, G Bayly, M Williams*
- (2.21) Implementation of a Diagnostic Service for Niemann-Pick Type C Testing - *Dr Johanna Hunt, F Macdonald, J Bell, P Gissen*
- (2.22) Novel mutations within the zinc finger domains of GLI3 in patients affected by Greig cephalopolysyndactyly syndrome (GCPS) - *Ms Caroline Archer, M Oldridge, A Seller, T Lester*
- (2.23) Analysis of an Interesting Fragile X Family - *Miss Sarah Fuller, GS Wilkie, J Tolmie, S Stenhouse*
- (2.24) The importance of PHOX2B sequence analysis and family studies in Congenital Central Hypoventilation Syndrome; primarily a polyalanine repeat disorder - *Mrs Sarah Burton-Jones, DE Fransen van de Putte, C Faulkner, L Yarram, JC Evans, T Patrick, P Lunt, P Fleming, EK Bijlsma, M Williams*
- (2.25) Facilitating implementation of NIPD: a modified protocol to detect the universal fetal DNA marker RASSF1A - *Dr Helen White, CL Dent, VJ Hall, JA Crolla, LS Chitty*
- (2.26) Screening for fusion genes involving PDGFRA or PDGFRB in patients with eosinophilia-associated myeloproliferative neoplasms (Eos-MPN) using the Fluidigm BioMark real-time PCR system and 48.48 Dynamic Array - *Dr Helen White, DJ Scott, J Huggett, VJ Hall, K Waghorn, M Lyons, R Sanders, CA Foy, NCP Cross*
- (2.27) Establishment of the 1st World Health Organization International Genetic Reference Panel for quantitation of BCR-ABL mRNA - *Dr Helen White, P Matejtschuk, P Rigsby, P Metcalfe, NCP Cross*
- (2.28) A comprehensive molecular genetic screening service for retinoblastoma - *Mrs Anna O'Grady, N Hart-Holden, T Colclough, S Ramsden, T Cole, R Elles, GC Black*
- (2.29) Assigning pathogenicity to mitochondrial tRNA gene mutations - *Mr John Yarham, JL Elson, EL Blakely, CL Alston, DM Turnbull, R McFarland, RW Taylor*
- (2.30) Ciliopathy-related BBS proteins regulate the actin cytoskeleton independent of ciliary function - *Dr Victor Hernandez, PL Beales*
- (2.31) mTOR inhibition reduces Bbs mouse obesity - *Dr Victor Hernandez, PI Beales*

- (2.32) Evaluation of Clinical Diagnostic Potential of Next Generation Sequencing using Hypertrophic Cardiomyopathy as a Model - *Dr Jenny Taylor, K Kaur, J Broxholme, R Copley, J Thistleton, S Reid, E Ormondroyd, A Seller, E Blair, I Ragoussis, H Watkins*
- (2.33) Non invasive prenatal fetal sex determination – service experience and quality assurance - *Dr Helene Schlecht, N Charlesworth, SC Ramsden, RG Elles, Z Deans*
- (2.34) A National Commissioning Group (NCG) funded variant Neurofibromatosis type 1 (NF1) mutation testing service primarily based on cDNA analysis – the first year's experience - *Miss Emma Sach, EL Howard, AJ Wallace, RG Elles, DGR Evans, SM Huson*
- (2.35) Development of an Asian-specific Newborn Screening Assay for Cystic Fibrosis using Luminex xTAG™ technology - *Dr Martin Schwarz, C Charlton, MJ Schwarz*
- (2.36) Genetic testing in phaeochromocytoma/paraganglioma referrals- a seven year audit - *Ms Eleni Mavraki, M McConachie, D Baty*
- (2.37) A diagnostic service for X-linked lymphoproliferative syndrome type 2 - *Miss Kunjan Patel, A Taylor, K Gilmour, C Cale, L Jenkins, N Lench*
- (2.38) Sequencing the DMD gene – review of results from 300+ cases - *Mrs Jo McCauley, H Storey, E Ashton, F Muntoni, S Yau, S Abbs*
- (2.39) Introduction of Semi-Automated Mutation Surveyor (SAMS) sequence analysis to the Cambridge Molecular Diagnostic Laboratory - *Mr Edward Thompson, S Halsall, H Martin, K Oakhill, R Treacy, J Whittaker, D Edge, C Tysoe*
- (2.40) A Friedreich Ataxia patient with a novel point mutation - *Ms Therese Bradley, A Cooke, R McWilliam, S Stenhouse*
- (2.41) Can genotype help predict phenotype in SCN1A related epilepsies? - *Miss Rachael Birch, E Reavey, A Brunklus, S Stenhouse, SM Zuberi*
- (2.42)  $\alpha$  - cardiac myosin heavy chain (MYH6) mutations affecting myofibril formation are associated with congenital heart defects - *Dr Javier T Granados Riveron, TK Ghosh, M Pope, J Eason, F BuLock, C Thornborough, EP Kirk, D Fatkin, MP Feneley, RP Harvey, JAL Armour, JD Brook*
- (2.43) Improving Non-invasive Prenatal Determination of Fetal Sex: A Comparison of Two DNA Extraction Methods - *Dr Angela Barrett, LS Chitty*
- (2.44) Identification of novel somatic mosaic EFNB1 variants in patients affected with craniofrontonasal syndrome - *Miss Helen Lord, A Seller, B Friedman, A Barnicoat, A Wilkie, T Lester*
- (2.45) Diagnostic Testing for CHARGE Syndrome - Three years since service implementation - *Dr Matthew Smith, K Slowther, C Crosby, E Perrott, P Rehal, C Hardy, F Macdonald*
- (2.46) Genetic ophthalmology in focus: clinical testing for Optic Atrophy 1 and Familial Exudative Vitreoretinopathy - *Dr Lampros Mavrogiannis, EF Jackson, DF Gilmour, K Prescott, C Toomes, CF Inglehearn, DJ Cockburn, RS Charlton*
- (2.47) Non-invasive prenatal diagnosis of single gene disorders: an aid to the confirmation of fetal ultrasound findings - *Dr Cathy Meaney, AN Barrett, LS Chitty*
- (2.48) A novel role for the obesity-associated gene FTO in Beta-Catenin stabilisation and ciliogenesis - *Dr Daniel Osborn, S Christou, S Mukherjee, M Ferrante, R Roccasecca, I Barosso, D Stemple, P Beales*
- (2.49) Uniparental disomy of chromosome 14: A decade of testing at the Oxford Regional Molecular Genetics Laboratory - *Mr Anthony O'Rourke, S Man, U Kini, M Tabiner, A Steele, T Lester, A Seller, J Hurst, A Lloyd-Jani*
- (2.50) Molecular Analysis of PMS2 - a UKGTN service offered in Leeds - *Mrs Jennifer Simmonds, RL Robinson, CE Chu, RC Charlton*
- (2.51) Analysis of the Glucocerebrosidase (GBA) and GTP Cyclohydrolase 1 (GCH1) genes in a UK cohort of familial Parkinson's disease patients - *Mr Piers Fulton, DJ Nicholl, A.J Lewthwaite, F Macdonald, J Bell, Y Wallis, M Smith, K. McKay*
- (2.52) Development of NMD Gene Chips for the complete DNA diagnosis of Inherited Neuromuscular Disorders - *Dr Shu Yau, S Cirak, V Castleman, S Abbs, F Muntoni*
- (2.53) Diagnostic Molecular Genetic Testing for Urea Cycle Disorders – Review of a New UK Service - *Dr Liz Allen, NJ Beauchamp, NJ Holmes, AT Crewe, S Man, S Olpin, M Sharrard, A Dalton, RJ Kirk*
- (2.54) A hypomorphic Ift80 genetrapped mouse line phenocopies short rib polydactyly syndromes owing to defects in hedgehog signalling - *Ms Suzanne Rix, V Hernandez Hernandez, P Scambler, PL Beales*
- (2.55) Analysis of eNOS genetic polymorphism in the Iranian patients with coronary artery disease - *Miss Leili Rejali, M Fathi, A Minasian, P Hashemi, R Mohammad Hassani, A Najafi, MR Sarzaeem, D Javidi, SH Sezavar, SH Jamaladini, M Hasanzad*
- (2.56) Clinical and molecular investigation of autosomal recessive TMEM126A-associated optic atrophy and auditory neuropathy (ARTOAN) - *Dr Esther Meyer, M Michaelides, LJ Tee, AG Robson, F Rahman, S Pasha, LM Luxon, AT Moore, ER Maher*

- (2.57) A method for measuring allele-specific RNAi knock-down in SCA7 patient lymphoblasts - *Ms Danielle Berkowitz, J Scholefield, MS Weinberg, MJA Wood, LJ Greenberg*
- (2.58) A review of the clinical criteria and molecular tests offered by the All Wales Medical Genetic Service (AWMGS) for mental retardation with epilepsy syndromes - *Mr Lazarus Lazarou, A Clarke, D Pilz, J Sampson, P Davies, L Rosser, R Butler*
- (2.59) Screening for KRAS and EGFR mutations in circulating cell free tumour DNA from patients with non-small cell lung carcinoma - *Dr Carolyn Gokhale, J Sherwood, A Wallace, R Elles, G Ellison*
- (2.60) The role of Epidermal Growth Factor (EGF) polymorphism in Pre-eclampsia - *Mrs Ayat Sayed, S Chappell, L Morgan*
- (2.61) Variable severity of early onset CMT2 with compound heterozygous Mitofusin 2 mutations - *Dr James Polke, M Laura, VS Gibbons, D Pareyson, F Taroni, M Milani, C DeVile, MG Sweeney, MB Davis, MM Reilly*
- (2.62) Clonal sequencing of BRCA1 and BRCA2: evaluation of a new diagnostic service - *Dr Helen Lindsay, N Camm, HI Fraser, JE Morgan, RL Robinson, R Charlton, CE Chu, GR Taylor*
- (2.63) Neurologic Features of Genetically Defined Lipoid Proteinosis - *Dr Khaled Abu-Amero, S AlRashid, I Al-Orainy, D Oystreck, M Salih, TM Bosley*
- (2.64) Development Of A Molecular Genetic Diagnostic Service For FHL1-Related Diseases - *Dr Judith Hudson, E Graham, R Pinkett, L Grady, K Bushby, D Bourn*
- (2.65) PCR assay for FMR1 used to determine the incidence of expansions in early menopause in the Breakthrough Generations Study - *Dr Claire Bennett, JN Macpherson, PA Jacobs, N Orr, A Ashworth, AJ Swerdlow, A Murray*
- (2.66) Next generation genetic diagnosis of metabolic disorders - *Miss Frances Smith, T Cullup, E Howard, DJ Turner, A Coffey, C Deshpande, M Champion, R McFarland, R Horvath, ZMA Chrzanowska-Lightowlers, RW Taylor, S Abbs, SC Yau*
- (2.67) Strategies for high-throughput gene identification in consanguineous families - *Dr David Parry, BE Hayward, IM Carr, CV Logan, RS Charlton, CF Inglehearn, E Sheridan, GR Taylor, C Toomes, AF Markham, DT Bonthron, CA Johnson*
- (2.68) An Association Study of Bipolar Disorder Type 1 and the CLINT1 Gene - *Mr Kelvin Yan, J Garcia, H Lee-Miller, P Montoya, MC Lopez, J Bedoya, C Palacio, C Lopez, M Ospina-Duque, B Kremeyer, A Ruiz-Linares*
- (2.69) Familial Hypercholesterolemia testing in Scotland - *Mr Stephen Tennant, C Bell, C Brown, K Kelly, H Hailey, H Gregory, J Series, R Finnie, M van Mourik, W Simpson, Z Miedzybrodzka*
- (2.70) A pilot study investigating pathophysiology of late-onset Anderson-Fabry disease using genetic and functional tools - *Dr Hatim Ebrahim, R Baker, A Mehta, D Hughes*
- (2.71) A Diagnostic service for Insulin Receptoropathies - *Emma Gilroy, S Waller, I Delon, RJL Treacy, D Halsall, R Semple*
- (2.72) OPA1 plus: phenotypic variation in dominantly inherited optic atrophy attributable to genetic background - *Prof Joanna Poulton, K Morten, A Williams, I Fearnley, L Rosser, J Lowndes, N Ashley, C Fratter, M Votruba*

### 3. Cytogenetics and Molecular Cytogenetics

- (3.01) Investigation of aneusomy of chromosome 17 and its influence on prognosis (some clinical and immunohistochemical parameters) amongst some Iranian women with sporadic breast cancer - *Dr Farkhondeh Behjati, M Ataee Kachoui, E Keyhani, M Karbasian, M Kadivar, M Karimlou, F Larti, S Ghasemi Firozabadi*
- (3.02) The 12q14 microdeletion syndrome; 6 new cases confirming the role of HMGA2 in growth - *Dr Freddie Sharkey, N Foulds, AC Thuresson, AL Collins, G Anneren, BO Hedberg, CA Delaney, J Iremonger, CM Murray, E Maher, JA Crolla, C Costigan, W Lam, D FitzPatrick, R Regan, S Ennis, SA Lynch*
- (3.03) Atypical 22q11 deletion detected by multiplex ligation-dependent probe amplification (MLPA) in patients referred for Prader-Willi Syndrome testing - *Ms Karen Meaney, B O'Hici, SA Lynch, DE Barton*
- (3.04) Unintentional 'stem cell transplant' following a liver transplant in a patient with monosomy 7 AML - *Mrs Paula Page, M Rindl, R Doak, M Griffiths, D Kelly, M Velangi*
- (3.05) An 18 year old male with ALL and two extra intrachromosomal copies of RUNX1 at relapse: triplication or iAMP21? - *Dr Karen Sims, S Jeffries, M Strachan, C Almond, A Moorman, M Griffiths*
- (3.06) Three Cases of Acute Myeloid Leukaemia with t(5;11)(q35;p15) NUP98/NSD1 Fusion Transcripts and FLT3-ITD - *Dr Nervana Abou-Zeid, J Walker, S Begum, S Whelton, N Geddes, S Dyer, B Lai, R Evans, S Jeffries, M Velangi, S Akiki, M Griffiths*
- (3.07) Genome-Wide Single Nucleotide Polymorphism analysis reveals a novel translocation associated with CDKN2A and TP53 gene deletions in a case of Acute Myeloid Leukaemia - *Dr Nervana Abou-Zeid, C Gould, M Griffiths*
- (3.08) Prenatal diagnosis of Prader-Willi syndrome by MLPA - *Miss Emma Watson, C Lambert, A Callegari, E Holgado, S Liddle*
- (3.09) A case of CML with granulocytic sarcoma - *Miss Edith Terrenoire, S Rose, E Huxley, S Akiki, R Evans, J Walker, J Mason, S Tabrah, S Jeffries, P Page, M Griffiths*

- (3.10) Detecting Mosaicism by Microarray CGH: a mosaic partial duplication of 12p with a tissue distribution similar to the i(12p) in Pallister-Killian syndrome - *Mr Christopher Wragg, H Martin, R Newbury-Ecob, S Toomey, C Delmege, S Holman, R Wragg, E Roberts*
- (3.11) A new-born with anophthalmia and microdeletion of 1q21.1. The severe end of the clinical spectrum or a coincidental finding? - *Dr Lionel Willatt, I Simonic, A Clarkson, C Dunn, RN Sandford*
- (3.12) A CML patient with a cryptic insertion of BCR into both chromosomes 9 - *Mr Graham Halford, S Rose, P Page, M Griffiths*
- (3.13) Further evidence of overgrowth and craniosynostosis in the chromosome 9q22 microdeletion syndrome - *Mr Rodger Palmer, M Lees, D Morrogh, A Shaw, R Wang, J Waters*
- (3.14) 13q deletion size predicts disease progression and time to first treatment in patients with chronic lymphocytic leukaemia - *Dr Jon Strefford, M Rose-Zerilli, A Parker, BD Young, T Chaplin, DG Oscier, JC Strefford*
- (3.15) CD138 Selection Kit (Easy Sep) - a simple method for concentrating plasma cells for routine Myeloma FISH - *Dr Christine Waterman, A Fisher, S Laird, C Pattimore, B Pearce, R Washbourne, F Ross*
- (3.16) The role of rings in the genetic instability of soft tissue tumours - *Miss Josie Hayes, P Roberts, J Williams*
- (3.17) Three cases of reciprocal duplications of common deletion syndromes - *Miss Sheetal Monji, A Callegari, A Fairclough, KG Masters, E Senkardesler, EC Svennevik, V Walbanke, C Vince, TA Ballard, S Joseph*
- (3.18) Multi-platform comparison for arrayCGH hardware and software for use in diagnostic constitutional applications and new high throughput workflow - *Dr Shuwen Huang, J Charlton, S Jones, D Ward, J Crolla*
- (3.19) Solid Tissue Diagnostic Service: a molecular cytogenetics strategy - *Ms Mary Glancy, F Sharkey, C Gregory, M Shade, C Delaney, N Williams, J Warner, M Evans, E Maher*
- (3.20) Formation of a Novel der(5)t(5;14)(q35;q32) associated with HOX11L2 regulation in one case of paediatric T-Cell Acute Lymphoblastic Leukaemia - *Mr Edward Atack, P Talley, K Smith, A Eastwood, S Scott, A Vora, G Fitzgibbon, M Needham, L Gaunt, E Maltby*
- (3.21) An audit of the cytogenetic findings and referral patterns for 7089 patients referred with infertility or reduced fertility in the West Midlands Region of the UK between 2000 and 2010 - *Mr Graham Fews, F O'Neill, SA Larkins, EV Davison*
- (3.22) Two cases of single kit abnormalities identified by MLPA multi- telomere testing and confirmed by aCGH - *Miss Jennifer Corfield, C Wragg, S Toomey, H Martin, E Clements, P Turnpenny, R Hastings, E Roberts*
- (3.23) Deletions of the long arm of chromosome 20 in adult patients with suspected B cell disorders - *Mrs Rhian White, H Habboush, A Salamat, M Pobjoy, K Kardara, H Lewis, PW Thompson, SH Roberts*
- (3.24) Translocation t(3;5) in a female patient with Acute Myeloid Leukaemia (AML): a case report - *Miss Julie Turbitt, P Batstone, J Tighe, D Stevenson*
- (3.25) Tertiary Trisomy in Prenatal Diagnosis-a case report - *Dr Abhijit Kulkarni, M Vast, A Hussain, J Taylor*
- (3.26) Familial deletion 17q21.32-17q21.33 with haploinsufficiency of the HOXB cluster associated with proportionate short stature and mild microcephaly, learning difficulties and hypoplastic clavicles - *Dr Peter Turnpenny, JA Crolla, PD Turnpenny*
- (3.27) De novo deletion 6q25.3, encompassing the ARID1B gene, in a child with developmental delay, autistic spectrum disorder, hypotonia, hirsutism, mild dysmorphic features and hydronephrosis - *Dr Peter Turnpenny, JA Crolla, PD Turnpenny*
- (3.28) Apparently contiguous deletion/duplication imbalances detected by oligonucleotide array CGH - *Dr Caroline Mackie Ogilvie, JW Ahn, D Tindall, AF Davies, RP Hall*
- (3.29) A de novo Marker Chromosome derived from 9p in a Patient with Autism: Genotype-Phenotype Correlation - *Dr Khaled Abu-Amero, A Helani, M Salih, M Seidahmed, T Elmalik, G Zidan, T Bosley*
- (3.30) Atypical Williams-Beuren syndrome deletion in a patient presenting with global developmental delay and dysmorphic features - *Mr Richard Ellis, AJ Brady, S Mountford*
- (3.31) Automating FISH analysis across all sample types with the GSL-120 automated scanner - *Mrs Victoria Anthony-Dubernet, W King, A Marsh, L Hill, K Marks, J Taylor*
- (3.32) A case of a satellite stalk insertion into the long arm of chromosome 21 shown to have an imbalance by array CGH - *Miss Emma Shearing, K Smith, O Quarrell*
- (3.33) Establishment of a rapid aneuploidy/QF-PCR service in Edinburgh Cytogenetics Laboratory - *Dr Carol Delaney, D Carmichael, L Benson, J Iremonger, F Sharkey, A Broadwell, M Glancy, M Shade, E Maher*

## 4. Counselling, Social, Policy and Education

- (4.01) Familial Hypercholesterolaemia cascade testing: a sociological study - *Miss Jane Miller*
- (4.02) The views of health professionals regarding the management and care of families affected by sudden arrhythmic death syndrome - *Dr Kate Brain, B Henderson, D Kumar, M Nicol, R Iredale, A Clarke*
- (4.03) A survey of follow up for cancer syndrome mutation carriers - *Miss Anita Bruce, L Side, C Berlin*
- (4.04) Genomic Medicine: An Independent Response to the House of Lords Science and Technology Committee Report - *Dr Caroline Wright, C Alberg, H Burton, A Hall, R Zimmern*
- (4.05) A discursive investigation into the moral and ethical challenges of professionals developing guidelines relating to the genetic testing of children - *Miss Hannah Shipman, S Sarangi*
- (4.06) Tackling birth defects in low and middle income countries - *Dr Luis Nacul, H Burton, S Moorthie*
- (4.07) An effective model for teaching genetics to general practitioners - *Mrs Michelle Bishop, S Burke, R Newton, P Farndon*
- (4.08) Epidermolysis Bullosa - exploring the experiences of affected individuals and families of living with an inherited skin condition - *Miss Sarah Gibson, M Wilcox, PE Price, AJ Clarke*
- (4.09) Fragmented Biosociality: Familial Meanings of Neurofibromatosis Type 1 - *Mr Daniele Carrieri, HR Farrimond, SE Kelly, PD Turnpenny*
- (4.10) Non-invasive prenatal diagnosis of single gene disorders: An exploration of views and preferences gathered from the people who will use these tests - *Ms Cecilia Compton, M Hill, C Lewis, S Morris, L Chitty*
- (4.11) Personal Genomes and Tomorrow's Doctors - *Ms Francesca Harrington, H R Dorkins*
- (4.12) Psychosocial and ethical issues in pre-symptomatic testing for Inherited Cardiac Conditions (ICCs) - *Dr Liz Ormondroyd, S Oates, E Blair, M Parker, H Watkins*
- (4.13) Facilitating young people's exploration of the \$1000 genome - *Miss Flo Ticehurst, CN Anderson, J Thomas*
- (4.14) An updated genetics education framework for UK nurses - *Prof Maggie Kirk, E Tonkin, H Skirton, M Longley, K McDonald*
- (4.15) Provision of support within a Regional Genetics Service for BRCA carriers; results from a patient information evening - *Miss Emma Williams, A Bruce, L Side, E Rosser*
- (4.16) Using Google Analytics to inform strategy for the 'Telling Stories, Understanding Real Life Genetics' health professional education resource - *Dr Rhian Morgan, E Tonkin, M Kirk, Telling Stories Project Team*
- (4.17) Comparison of two array CGH platforms as tools for the investigation of childhood developmental delay - *Dr Susan McNerlan, P McGrattan, S Heggarty, PJ Erwin, M Humphreys*
- (4.18) Where there are no genetic services: a study of patients suffering from Usher Syndrome in Bangladesh - *Miss Eshika Haque, SR Rozario*
- (4.19) Publicising screening initiatives aimed at university students through the use of the Internet and Social Networking Media - *Mr David Bodansky, J Copitch, S Freedman, B Foxler, D Blank, N Silver, L Korb, A Sher, J Bomsztyk, I Ellis*
- (4.20) An innovative Methodology for Examining the Process of Family Communication of Genetic Test Results - *Ms Chris Jacobs, J Smith, C Dancyger, M Wallace, S Michie*
- (4.21) Influencing how genetics is taught in UK secondary schools: The Nowgen Schools Genomics Programme - *Dr Leah Holmes, P Finegold, I Starling, K Dack, A Hall, J Worthington, J Harris, AP Read, HR Middleton-Price, D Donnai*
- (4.22) Looking Forward Involving young people with Cystic Fibrosis (CF) in developing an educational arts package exploring CF and gene therapy - *Dr Leah Holmes, KW Southern, B Chapman, L Morton, H Kitchen, M Haig, K Wiltshire, ME Palm, AM Smith, K Dack, I Starling*

## 5. Cancer Genetics

- (5.01) Challenges of patient stratification in drug development - what diagnostic tests will your department need to support in the future? - *Dr Mireille Cantarini*
- (5.02) Involving patients at the Cancer Genetics Service for Wales - *Miss Jennifer Hilgart, R Iredale, A Murray*
- (5.03) Developing the StoryBank: Patients' stories about their journey through the Cancer Genetics Service for Wales - *Miss Jennifer Hilgart, R Iredale, L Mundy*
- (5.04) A Diagnostic Service for Familial Platelet Disorder with Propensity to Develop Acute Myeloid Leukaemia (FPD/AML) - *Dr Kerry Wall, J Mason, S Akiki, M Griffiths*

- (5.05) An audit of Li Fraumeni syndrome and Li-Fraumeni like syndrome - *Dr Lara Khoury, SF Douglas, P Brennan, A Henderson*
- (5.06) BRCA1 and 2 Carrier Survey within South Wales - *Miss Jackie Wedderburn, K Bailey, E Kirk, MT Rogers, A Murray*
- (5.07) A novel colorectal cancer susceptibility SNP in the EIF3H promoter influences patient survival and response to treatment - *Mr Christopher Smith, CG Smith, D Fisher, R Harris, TS Maughan, S Idziaszczyk, V Moskvina, J Colley, A Meade, C Bonnet, H West, R Kaplan, JP Cheadle*
- (5.08) Review of Retinoblastoma families and RB1 mutational analysis in the West of Scotland Genetics service - *Miss Sarah Gibson, V Murday*
- (5.09) Juvenile Polyposis Case Study. A 5'UTR Deletion in BMPR1A: Pathogenic or Not? - *Miss Tina Bedenham, T Cranston, D Eccles, K Lachlan, A Seller*
- (5.10) Withdrawn
- (5.11) Assessing the Functional Role of Caspase-8 Gene Variants in Breast Cancer - *Dr Sushila Rigas, M Parry, MW Reed, N Camp, A Cox*
- (5.12) Investigation of the Birt-Hogg-Dubé tumour suppressor gene (FLCN) in familial and sporadic colorectal cancer - *Mr Mike Nahorski, DHK Lim, L Martin, JP Gille, K McKay, PK Rehal, HM Ploeger, M van Steensel, IP Tomlinson, F Latif, FH Menko, ER Maher*
- (5.13) Evaluation of tumour testing criteria for Lynch syndrome - *Miss Kate Simon, AV Kumar, L Side*
- (5.14) DICER1 syndrome – a pleiotropic tumor predisposition syndrome - *Dr Ingrid Slade, C Bacchelli, H Davies, F Abbaszadeh, R Barfoot, A Murray, S Hanks, C Stiller, MR Stratton, J Douglas, N Rahman*
- (5.15) The influence of modifications of HSF1 activity on human carcinoma cells growth - *Ms Maria Zamkova, P Kopnin, N Khromova, B Kopnin*
- (5.16) Novel one-stop multidisciplinary follow-up clinic significantly improves cancer risk management in BRCA1/2 carriers - *Dr Louise Izatt, C Jacobs, I Jacobs, U Menon, R Manchanda, M Johnson, H Hamed, C Firth, M Evison, A Tutt, L de Silva, C Langman, G Pichert*
- (5.17) Caspase-8 gene SNPs in prostate cancer susceptibility: a replication study - *Miss Marina Parry, G Elliott, R Abo, NJ Camp, DE Neal, JL Donovan, FC Hamdy, A Cox*
- (5.18) Lynch syndrome: Tumour incidence in 179 West Midlands Families with Confirmed DNA Mismatch Repair Mutations - *Dr Nicola Cooper, ER Maher, C Bowles, J Bell, P Rehal, KR Ong, C Chapman, C McKeown, T Cole, F MacDonald*
- (5.19) Adhering to annual screening guidelines in patients with Von-Hippel-Lindau Disease; results from a five year follow up period - *Dr Jamie Wilson, G Crawford, DM Eccles*
- (5.20) An investigation of the role of a promotor polymorphism in the BCL2 gene in BCL2 protein expression and survival from breast cancer - *Dr Claire Searle, I Brock, S Cross, A Cox*
- (5.21) Mutation Analysis of HIF-prolyl hydroxylases (PHD/EGLN) in Inherited Pheochromocytoma and Renal Cell Carcinoma - *Dr Dewi Astuti, C Ricketts, R Chowdury, MA McDonough, D Gentle, G Kirby, S Schlisio, PJ Ratcliffe, CJ Schofield, F Latif, ER Maher*

## 6. Other

- (6.01) Clinical Coding of Rare and Genetic Diseases - *Dr Michael Cornell, M Milicic-Brandt, A Devereau*
- (6.02) Professional Training for the New Era of Genetic Medicine - *Dr Angela Davies, K Mulryan, D Donnai, H Middleton-Price*
- (6.03) Assessment of kidney lesions using micro-MRI and histology in preclinical trials for tuberous sclerosis - *Dr Ming Hong Shen, Y Zhang, M Kalogerou, J Sampson*
- (6.04) Therapeutic targeting the loss of the Birt-Hogg-Dubé suppresser gene - *Dr Xiaohong Lu, J Fenton, A Reiman, L Seabara, W Wei, F Latif, E Maher*
- (6.05) Enrichment of Resequencing Targets using Droplet-based PCR - *Dr Steve Picton*
- (6.06) Overview of GEN2PHEN software developments for analysis and management of genetic variant data - *Dr Michael Cornell, G Dobson, A Devereau*
- (6.07) The Sir Jules Thorn Charitable Trust "International Resource for Autozygosity Mapping" - *Dr CA Johnson, I Carr, CV Logan, D Parry, D Bonthron, R Charlton, C Inglehearn, E Sheridan, G Taylor, M Ali, C Toomes, B Hayward, A Markham*

# British Human Genetics Conference 2010

6-8 September 2010  
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 2010 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 15 August.
- We regret that fees paid cannot be reimbursed if you have to cancel your registration after 15 August.
- We **CANNOT** supply invoices in respect of registration fees

## CONFERENCE COSTS

### Registration Fees

	Member	Non-Member
Full conference	£350.00	£475.00
Individual days (Monday or Tuesday)	£175.00	£240.00
Individual days (Wednesday)	£120.00	£150.00

### Accommodation (per night)

Standard Single	£37.00
En-suite Single	£55.00

**Conference Dinner** (University of Warwick) £42.00

**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 17:00-21:00 on Sunday 5 September and from 07:30 on Monday 6 September and Tuesday 7 September. General information, including details of accommodation and map of campus, will be sent to you after the closing date of 15 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:

**15 August 2010**

<http://www.bshg.org.uk/registration/bhgc2010.asp>

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