British Human Genetics Conference 2007

Monday-Wednesday, 17-19 September

University of York

PROGRAMME AND REGISTRATION BOOKLET

Closing date for registration: 17 August 2007

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

- Developmental and Paediatric Genetics
- Genomic / RNA
- Counselling / Ethics
- Communicating and Managing Risk
- Mechanisms of disease / Cardiovascular
- Interphase, networks and evolution
- Complex disease genetics
- Fusions, translocations and databases
- Mechanisms of disease II
- Consanguinity and genetic disorders
- Genetics of Lymphoma and solid tumours
- National Genetic Reference Laboratories – Achievements and Plans
- *Carter Lecture*
  Professor Doug Higgs on "The role of Human Genetics in our understanding of how genes are switched on and off"
- *Great Debate:*
  "The Ups and Downs of Gene Databanks"
- *Researchers Forum*

LATE BREAKING ABSTRACTS

Deadline 1 September 2007

Details on website www.bshg.org.uk

SCIENTIFIC PROGRAMME

The BSHG Scientific Programme Committee welcomes you to the 2007 meeting of the British Society for Human Genetics. The SPC has compiled a wide-ranging programme comprising diverse symposia and outstanding international speakers, including the Carter Lecture by Professor Doug Higgs. 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 2008 meeting!

Eamonn Maher
Chairman, Scientific Programme Committee 2007

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

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

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

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

Carter Lecture - Professor Doug Higgs on "The role of Human Genetics in our understanding of how genes are switched on and off"

The Great Debate is being held again on Monday evening (19:00-20:00 in Room PX001, Exhibition Centre). The motion for the debate is "The Ups and Downs of Gene Databanks"

Debators are Prof Marcus Pembrey, Prof Paul Martin, Prof Steven Bain

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

RESEARCHERS FORUM

A "Young Researchers Forum" meeting is scheduled for Tuesday lunchtime. This session provides an opportunity for trainees, PhD students and junior faculty to meet with senior academics and discuss issues relevant to junior academic researchers. Places are limited. In order to reserve a place (and lunch!) please notify Ruth Cole (york2007@bshg.org.uk) if you intend to attend. In addition, please feel free to email suggestions for topics for discussion that you think would be of general interest.

ABSTRACT BOOKLET

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

CONFERENCE VENUE

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

ACCOMMODATION

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

TRAVEL

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

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

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

We can provide a 3-day parking permit if delegates wish, the cost of the 3-day permit is £11. The Pay and Display charges are £2 for 4 hours, £4 for 10 hours and £5 for 24 hours (Monday-Friday). If you pay for your parking daily, you must remember to put a new ticket on in time or you will be clamped - the release fee is £50.00.

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

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

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

SOCIAL PROGRAMME

Wine Tasting Reception - Start Monday evening with the traditional wine tasting reception (18:00-19:00 Exhibition Centre). View the posters and visit the trade exhibitors whilst enjoying a wine tasting. This year there will be prizes for three competition winners of £100 each of books, again generously donated by Scion Publishers.

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

Conference Party - This year we are having a trip to the races for our conference party, so get your gladrags on and join us for a sumptuous 4-course meal and virtual race night entertainment. Coaches will carry you from the University of York to the racecourse (and back). The food and wine is usually superb at York racecourse and the racing games are lots of fun!

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 17 August. Do NOT resubmit, if you have any amendments please email the administrative office. Please ensure that you give your full postal address (including post code) as this is the only information that will be used to send out your delegates pack.

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

CONFIRMATION OF REGISTRATION

The Conference Pack, containing a map, Programme and Abstract Booklet, accommodation details, tickets for evening meals, a car parking permit and a receipt, will be posted at the end of August.

CPD APPROVAL

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

EMERGENCY TELEPHONE NUMBERS

Information Desk during Conference hours: 01904 434552
Goodricke Porters' Lodge: 01904 433100 (for delegates staying in James College)
Langwith Porters' Lodge: 01904 433400

COUNCIL AND SOCIETY MEETINGS

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

ASSOCIATION OF GENETIC NURSES AND COUNSELLORS
Business Meeting - 13:30 Tuesday 18 September - *Room PX001*

ASSOCIATION OF CLINICAL CYTOGENETICISTS
Annual General Meeting - 14:00 Tuesday 18 September - *Central Hall*

OTHER MEETINGS

Junior Liaison Committee Session – CPD, how do you record yours? – 13:15 Tuesday 17 September – Central Hall
An informal session on CPD highlighting the types of activities that you can record. We shall be launching the example CPD portfolio (which is already on the website) and providing examples of how you might complete it, should you choose to use it. There will be opportunity to discuss other aspects of CPD and the HPC, along with other issues which may be of concern for members of the profession. This session is aimed at members employed at band 7 or below. Email any questions in advance to jlc@cytogenetics.org.uk

FUTURE MEETINGS

BRITISH SOCIETY FOR HUMAN GENETICS

UNIVERSITY OF YORK 15-17 September 2008
14-16 September 2009
13-15 September 2010

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: york2007@bshg.org.uk Website: www.bshg.org.uk

PROGRAMME

Sunday 16 September 2007

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

Monday 17 September 2007

08:30-10:30 **BSHG - Council Meeting** - Room G/020 (Goodricke College)

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

11:30-13:00 Symposium – Developmental and Paediatric Genetics

Chair:			**Central Hall**
11:30	(SP01)	Cornelia de Lange syndrome and the Cohesinopathies: Insights into the Etiology of Developmental Disorders *- Dr Ian Krantz (Philadelphia, USA)*	
12:00	(SP02)	X linked dominant diseases: the example of the Oral-facial-digital type I (OFDI) and microphthalmia with linear skin defects (MLS) syndromes *– Prof Brunella Franco (Naples, Italy)*	
12:30	(SP03)	Clinical and molecular features of Ras-related disorders *– Prof Eric Legius (Leuven, Belgium)*	

13:00-14:15 LUNCH

14.15-15:15 Concurrent Sessions

Cancer	**Chair:**		**Central Hall**
14:15	(SP04)	Morphology and syndromes in children with cancer *- Prof Raoul Hennekam, JHM Merks, HM Ozgen, HN Caron, AH Zwinderman*	
14:30	(SP05)	CAPP2: a randomised chemoprevention trial of aspirin and resistant starch in Lynch Syndrome *- Prof John Burn, DT Bishop, J-P Mecklin, F Macrae, G Moeslein, S Olschwang, MLS Bisgaard, R Ramesar, J Jass, HT Lynch, G Barker, F Elliott, JCM Mathers*	
14:45	(SP06)	Tuberous Sclerosis 2000 Study *- Prof John Yates, C MacLean, A Humphrey, PF Bolton*	
15:00	(SP07)	HIF1 alpha mediated angiogenesis and mammalian Target of Rapamycin (mTOR) activation: implications for treatment of tuberous sclerosis *- Dr Andrew Tee, SC Land, EA Dunlop, M Davies, MH Shen, JR Sampson*	

Mechanisms of Disease 1	**Chair:**		**Exhibition Centre Room PX001**
14:15	(SP08)	Mutation Analysis of CHRNA1, CHRNB1, CHRND and RAPSN Genes in Multiple Pterygium Syndrome/Fetal Akinesia Patients *- Dr Julie Vogt, B Harrison, H Spearman, J Cossins, S Vermeer, L N ten Cate, N Morgan, D Beeson, E Maher*	
14:30	(SP09)	Alpha Cardiac Actin and Atrial Septal Defects *- Dr Jacqueline Eason, JT Granados Riveron, M Pope, FA Bu'Lock, J Cox, TE Robinson, JD Brook*	
14:45	(SP10)	A homoplasmic mitochondrial DNA variant (m.7472A>C) influences the phenotype of the pathogenic m.7472Cins MTTS1 mutation *- Miss Helen Swalwell, EL Blakely, MM Davidson, R Sutton, K Tonska, M Elstner, L He, T Taivassalo, DM Turnbull, RG Haller, RW Taylor*	
15:00	(SP11)	Inherited Creutzfeldt-Jakob disease: phenotypes, genotypes and public health implications *- Dr Sarah Smithson, S Leonard, MA James, C King, RA Newbury-Ecob, SJ Wroe, J Collinge, S Mead*	

Counselling	**Chair:**		**Exhibition Centre Room PL001**
14:15	(SP12)	The Restricted Growth Experience; Quality of Life and Barriers to Participation *- Dr Michael Wright, S Thompson, TW Shakespeare*	
14:30	(SP13)	Adolescent Carrier Testing in Practice : The Impact of Legal Rulings and Problems with 'Gillick competence' *- Dr Maggie Gregory, P R Boddington*	
14:45	(SP14)	Children's understanding of testing for a genetic illness *- Miss Fiona Ulph, C Glazebrook, E Townsend*	
15:00	(SP15)	Funding for NHS Genetic Services *- Dr Frances Flinter, C Patch, P Mills, G Nightingale*	

Constitutional Cytogenetics Chair:		Exhibition Centre Room PL002
14:15	(SP16)	Implementation of the Oxford diagnostic array CGH service *- Mrs Lyndsey Connell, AM Crocker, KK Smith*
14:30	(SP17)	Detailed mapping of chromosome 3p25 deletions *- Mrs Salwati Shuib, E Rattenberry, D McMullan, F Rahman, M Zatyka, C Chapman, EV Davison, F Latif, ER Maher*
14:45	(SP18)	8p23.1 microduplication syndrome: a novel genomic condition with unexpected complexity revealed by array CGH *- Dr John Barber, VK Maloney, S Huang, D J Bunyan, L Cresswell, E Kinning, A Benson, T Cheetham, J Wyllie, SA Lynch, S Zwolinski, L Prescott, Y Crow, R Morgan, E Hobson*
15:00	(SP19)	Two siblings with severe mental retardation and a novel inherited ~4Mb microdeletion of 11p11.2 – ? A new locus for mental retardation. *- Dr Jenny Carmichael, L Willatt, G Parkin, A Clarkson, I Simonic, C Shaw-Smith, E Reid, CG Woods*

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

16:00-17:30 Concurrent Symposia – Genomic / RNA

Chair:		Central Hall
16:00	(SP20)	Therapeutic Gene Silencing in Huntington's Disease *– Prof Neil Aronin (Massachusetts, USA)*
16:30	(SP21)	Fragile X associated tremor ataxia syndrome: Modifiers and mechanisms *– Dr David Nelson (Houston, Texas)*
17:00	(SP22)	RNA – mediated disease *– Prof Maurice Swanson (Florida, USA)*

16:00-17:30 Concurrent Symposia – Counselling / Ethics

Chair:		Exhibition Centre Room PX001
16:00	(SP23)	Conditional parenthood: the ethics of prenatal and preimplantation selection *- Dr Helen Watt (Linacre Centre for Ethics, London)*
16:30	(SP24)	The burden of choice – the parental perspective on antenatal genetic testing *- Ms Jane Fisher (ARC, London)*
17:00	(SP25)	Procreative beneficience and disability: is there a moral obligation to create children with the best chance of the best life? *- Prof Julian Savulescu (Oxford Uehiro Centre for Practical Ethics)*

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

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

19:00-20:00 The Great Debate

Chair: **Exhibition Centre Room PX001**

The Ups and Downs of Gene Databanks

Professor Marcus Pembrey, Prof Paul Martin, Prof Steven Bain

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

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

PROGRAMME

PROGRAMME

Tuesday 18 September 2007

09:00-10:30 Concurrent Sessions

Mechanisms of Disease 2		Chair:	Central Hall
09:00	(SP26)	Mutations in UPF3B, a member of the nonsense-mediated mRNA decay surveillance complex, cause Lujan-Fryns and FG phenotypes and non-syndromic mental retardation *- Patrick Tarpey, FL Raymond, R Smith, G Turner, RE Stevenson, CE Schwartz, PA Futreal, MR Stratton, J Gécz and behalf of the Genetic of Learning Disability (GOLD) study*	
09:15	(SP27)	Nuclear gene mutations impacting mitochondrial genome maintenance *- Miss Joanna Stewart, T Harrower, RW Taylor, G Hudson, H Houlden, G Warner, R De Silva, D O'Donovan, L Findlay, PF Chinnery*	
09:30	(SP28)	Mutations in the mitochondrial DNA gamma polymerase (POLG) may cause Alpers syndrome, mitochondrial DNA depletion and apparent non-syndromic status epilepticus: implications for valproate therapy *- N Ashley, S Jayawant, J Parr, A O'Rourke, C Smith, K Morten, S Adams, M Narasimhan, C Fratter, J Poulton,*	
09:45	(SP29)	Mutations in the RAF1 gene are associated with hypertrophic cardiomyopathy in Noonan syndrome *- Dr Abdur Razzaque, T Nishizawa, Y Komoike, H Yagi, M Furutani, R Amo, T Higashinakagawa, R Matsuoka*	
10:00	(SP30)	Complement C3 variant increases risk of age-related macular degeneration *- Prof John Yates, T Sepp, BK Matharu, JC Khan, DA Thurlby, H Shahid, DG Clayton, C Hayward, J Morgan, AF Wright, AM Armbrecht, B Dhillon, IJ Deary, E Redmond, AC Bird, AT Moore*	
10:15	(SP31)	Genetic Linkage of Adolescent Idiopathic Scoliosis (AIS) in the British Population *- Ms Louise Ocaka, C Zhao, JK O'Dowd, AH Child*	

Communicating and Managing Risk			Chair: Exhibition Centre, Room PX001
09:00	(SP32)	Communicating and Managing Risk – the example of breast cancer *– Prof Gareth Evans (Manchester)*	
09:18	(SP33)	Managing familial ovarian cancer risk *– Dr Usha Menon (UCLH)*	
09:36	(SP34)	Communicating risk to patients *– Prof Susan Michie (UCL)*	
09:54	(SP35)	Conceptual Issues of Risk *– Prof Scott Campbell (Nottingham)*	
10:12	(SP36)	Genetic Risk Communication and Behaviour change *– Prof Theresa Marteau (London)*	

Clinical		Chair:	Exhibition Centre, Room PL001
09:00	(SP37)	Paediatric outcome from birth onwards after preimplantation genetic diagnosis *- Ms Alison Lashwood, T El-Toukhy, F Flinter, F Kavalier, D Kanabar*	
09:15	(SP38)	Prospective Register of Outcomes Of Free-fetal DNA testing (PROOF) – results of the first year's audit. *- Dr Lyn Chitty, G Daniels, K Finning, J Hogg, B Ladd, P Martin, C Meaney, G Norbury, L Thomasson*	
09:30	(SP39)	It's in your hands – a combined clinical, molecular and developmental approach to the diagnosis of radial ray defects *- Dr Ruth Newbury-Ecob, A Sharif, M Logan*	
09:45	(SP40)	Mutation in FGF3 causes absent inner ear development and microtia *- Dr Maria Bitner-Glindzicz, P Rutland, W Pagarkar, D Saunders*	
10:00	(SP41)	Pitt-Hopkins syndrome – clinical report *- Dr Sergio Sousa, A Cabral, C Zweier, M Venancio, A Rauch, J Saraiva*	
10:15	(SP42)	6q24 transient neonatal diabetes – more than just diabetes *- Prof Karen Temple, EJ Davies, DO Robinson, DJG Mackay*	

Cytogenetics: Array and Oncology			Chair: Exhibition Centre, Room PL002
09:00	(SP43)	The European Cytogenetic Initiative (ECI): Molecular karyotyping of 120 patients with unexplained mental retardation by 500K SNP mapping arrays *- Ms Eleanor Rattenberry, DJ McMullan, JM Walker, L Brueton, EV Davison, A Dufke, BBA de Vries, M Bonin, S Jacobs, A Riess, O Altug-Teber, H Enders, T Kleefstra, S Vermeer, N Van Slobbe-Knoers, O Riess, JA Veltman*	
09:15	(SP44)	Interpretation of array CGH data: adding to the growing list of copy number variants in healthy subjects *- Dr Alexandra Blakemore, A.J. de Smith, A Tsalenko, N Sampas, A Scheffer, A Yamada, P Tsang, A Ben-Dor, Z Yakhini, R Ellis, L Bruhn, S Laderman, P Froguel*	
09:30	(SP45)	Disruption of PAX5 in patients with acute lymphoblastic leukaemia and dicentric chromosomes *- Dr Qian An, S.L. Wright, Z.J. Konn, A.V. Moorman, C.J. Harrison, J.C. Strefford*	
09:45	(SP46)	Philadelphia-negative clonal haematopoiesis is a significant feature of dasatinib therapy for chronic myeloid leukaemia *- Ms Valeria AS de Melo, D Milojkovic, JS Khorashad, D Marin, JM Goldman, JF Apperley, AG Reid,*	
10:00	(SP47)	Genomic profiling reveals multiple genetic targets in ETV6-RUNX1 positive acute lymphoblastic leukaemia (ALL). *- Dr Jon Strefford, H Worley, K.J. Barber, S.L. Wright, A.V. Moorman, M. Case, J. Irving, A. Hall, C.J. Harrison*	
10:15	(SP48)	B lymphoid growth factors significantly increase chromosome quality in cytogenetic cultures of ALL *- Mr Andrew Fisher, A Blair, P Strike, FM Ross*	

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

11:15-12:45 Concurrent Symposia – Mechanisms of disease/Cardiovascular

Chair:		Central Hall
11:15	(SP49)	Marfan syndrome and related disorders: from molecules to medicine *– Prof Hal Dietz (Baltimore, USA)*
11:45	(SP50)	Genetic and Molecular mechanisms causing hypertrophic cardiomyopathay *– Prof John Seidman (Boston, USA)*
12:15	(SP51)	Prospects for treatment of Duchenne Muscular Dystrophy *– Dr Francesco Muntoni (London)*

11:15-12:45 Concurrent Symposia – Interphase, networks and evolution

Chair:		Exhibition Centre, Room PX001
11:15	(SP52)	Interphase Chromosome Territories *– Prof Uwe Claussen (Jena, Germany)*
11:45	(SP53)	Regulation of the human genome: folding and mixing chromosomes *– Prof Job Dekker (Worcester, MA, USA)*
12:15	(SP54)	Human neocentromeres and evolutionary-new centromeres *– Prof Mariano Rocchi (Bari, Italy)*

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

13:00 **Young Researchers Forum –** Room: G/020 (Goodricke College)

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

14:00 **ASSOCIATION OF CLINICAL CYTOGENETICISTS – Annual General Meeting** (Central Hall)

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

Chair: Prof Richard Trembath		Central Hall
16:00	(SP55)	RNF135, a gene within the 17q11 NF1-microdeletion region, is responsible for a new autosomal dominant overgrowth syndrome with reduced penetrance *- Dr Deirdre Cilliers, J Douglas, K Tatton-Brown, B Bernhard, J Burn, SM Huson, DJ Josifova, D Lacombe, M Malik, S Mansour, E Reid, V Cormiere-Daire, T Cole, N Rahman*
16:15	(SP56)	Evc is a positive mediator of Ihh signalling *- Dr Judith Goodship, HJ Blair, A Wilson, Y-N Liu, ME Rodriguez-Andres, MJ Blanco, H Peters, C Miles, VL Ruiz-Perez*
16:30-17:15	**Late Breaking Research**	

17:15-18:15 THE CARTER LECTURE

Chair: Dr Alan Fryer **Central Hall**	
(SP57)	**The role of human genetics in our understanding of how genes are switched on and off** *– Professor Doug Higgs*

19:00 **CONFERENCE PARTY** – York Racecourse

PROGRAMME

PROGRAMME

Wednesday 19 September 2007

09:00-10:30 Concurrent Symposium – Complex Disease Genetics

Chair:			Central Hall
09:00	(SP58)	tbc – *Prof Doug Easton (Cambridge)*	
09:30	(SP59)	Tackling common diseases – how has the genome project helped? – *Prof Tim Aitman (London)*	
10:00	(SP60)	tbc – *Dr Rob Sladek (Quebec, Canada)*	

09:00-10:30 Concurrent Symposium – Fusions, translocations and databases

Chair:			Exhibition Centre, Room PX001
09:00	(SP61)	TMPRS22 fusions in solid tumours – *Prof Olli Kallioniemi (Turku, Finland)*	
09:30	(SP62)	Causes and consequences of chromosomal translocations in leukaemias and epithelial cancers – *Prof Terry Rabbitts (Leeds)*	
10:00	(SP63)	Database of cancer cytogenetics and gene fusion - *Prof Felix Mitelman (Lund, Sweden)*	

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

11:15-12:45 Symposium – Mechanisms of Disease II

Chair:			Central Hall
11:15	(SP64)	Genetic diseases of cell junctions in the skin – *Prof John McGrath (KCL)*	
11:45	(SP65)	Molecular Genetics of Cardiac Tumors: A model of Cardiac Growth and Structure – *Prof Craig Basson (New York, USA)*	
12:15	(SP66)	Genetic factors in neurodegeneration with brain iron accumulation – *Prof Susan Hayflick (Portland, USA)*	

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

14:00-16:00 Workshops

National Genetic Reference Laboratories – Achievements and Plans **Chair:** **Central Hall**

Consanguinity and Genetic Disorders	Chair:		Exhibition Centre Room PX001
14:00		Introduction and Overview – *Prof Richard Trembath (London)*	
14:10	(SP67)	Cultural and religious issues in the UK – *Dr Alison Shaw (Oxford)*	
14:25	(SP68)	Autosomal recessive disorders, the Middle East perspective – *Dr Lihadh Al-Gazali (Abu Dhabi)*	
Mapping genetic diseases in consanguineous families			
14:40	(SP69)	Primary Microcephaly – *Dr Andrew Jackson (Edinburgh)*	
14:52	(SP70)	Eye Disorders – *Dr Irene Aligianis (Birmingham)*	
15:04	(SP71)	Aicardi Goutieres syndrome – *Dr Yanick Crow (Leeds)*	
15:16	(SP72)	Multiple pterygium syndromes: Phenotype, Genotype and the role of Acetylcholine Receptor Dysfunction – *Dr Louise Brueton (Birmingham)*	
15:28	(SP73)	Monogenic Obesity Syndrome - *Dr Saddaf Farooqi (Cambridge)*	
15:40	(SP74)	Autosomal Recessive Disorders in the Irish Travellers – *Prof Andrew Green (Dublin)*	
15:52		Summing up – *Prof Eamonn Maher (Birmingham)*	

Genetics of Lymphoma and Solid Tumours		Chair:	Exhibition Centre Room PX001
14:00	(SP75)	From chromosome to transcriptome: comparative genome - wide profiling of malignant lymphomas – *Prof Reiner Siebert (Kiel, Germany)*	
	(SP76)	Molecular Genetic Studies of Rhabdomyosarcomas – *Dr Janet Shipley (Sutton)*	
	(SP77)	Genetic Advances in uveal melanoma – *Dr Karen Sisley (Sheffield)*	
	(SP78)	Chromosome Translocations in Breast Cancer – *Dr Paul Edwards (Cambridge)*	

16:00 **TEA**

Poster presentations

1. Clinical genetics

(1.01) The Importance of a FBN1 Mutation Database for Screening Relatives in Marfan syndrome and Ectopia Lentis. - *Dr Anne Child, G Arno, A Kioteskoglou, P Comeglio*

(1.02) Familial adrenocortical carcinoma associated with HNPCC: A causal link - *Dr N. Serena Nik-Zainal, L.J. Walker, L.C. Happerfield, M.J. Arends, J.S. Paterson*

(1.03) Inverted duplication of 41.6 Mb of 1q characterised by array CGH and review of distal 1q partial trisomy - *Dr Meena Balasubramanian, J C K Barber, M N Collinson, S Huang, V K Maloney, N Foulds*

(1.04) Disease frequency of Inborn Errors of Metabolism in the Irish Traveller Community - *Dr Anne Marie Murphy, EP Treacy, AA Monavari, PD Mayne, SA Lynch*

(1.05) The genetic basis of Oesteogenesis Imperfecta in the Irish Traveller population - *Dr Jennifer Walsh, D Lambert, DM Baldridge, R Morello, D Eyre, B Lee, AJ Green*

(1.06) Notch 3 gene mutation associated with Sneddon's syndrome- an extension of the phenotype of cerebral autosomal dominant arteriopathy with subcortical infarcts and leukoencephalopathy [CADASIL] - *Dr Dhavendra Kumar, J Holroyd, I Frailing, I Ferguson,*

(1.07) A developmental approach to understanding fragile X syndrome: the influence of environmental and genetic factors on behavior problems in patients with fragile X syndrome - *Dr Marjaneh Akbarzadeh, Mash Akbarzadeh.Reza, Russ Akbarzadeh.Reyhaneh*

(1:08) Behavioural phenotypes in genetic syndromes: a window onto the biology of behaviour - *Dr Marjaneh Akbarzadeh, Mash Akbarzadeh.Reza, Russ Akbarzadeh.Reyhaneh*

(1:09) Advances in research on the fragile X syndrome: analyzing gene – brain – behaviour relationships in neurodevelopmental effects - *Dr Reyhaneh Akbarzadeh, Mash Akbarzadeh.Reza, Russ Akbarzadeh.Marjaneh*

(1:10) The behavioural neurogeneticsof fragile X syndrome - *Dr Reyhaneh Akbarzadeh, Mash Akbarzadeh.Reza, Russ kbarzadeh.Marjaneh*

(1:11) Towards outcome measures for clinical genetics services: a triangulation of methods - *Dr Marion McAllister, K Payne, S Nicholls, R MacLeod, D Donnai, L Davies*

(1:12) Paediatric oncology referrals to clinical genetics - *Dr Tracy Briggs, H Murphy, H McDowell, KL Greenhalgh*

(1:13) Cerebroretinal Microangiopathy with Calcification and Cysts (CRMCC) - *Dr Tracy Briggs, GMH Abdel-Salam, P Baxter, N Bishop, D Chityat, WK Chong, I Hughes, KK Nischall, GI Rice, JBP Stephenson, R Surtees, JF Talbot, NN Tehrani, JL Tolmie, C Toomes, MS van der Knaap, YJ Crow*

(1:14) DYSCERNE - A European network of Centres of Expertise for dysmorphology - *Mrs Pamela Griffiths, D Donnai, B Kerr, K Metcalfe, H Brunner, B Dallapiccola, K Devriendt, M Krajewska-Walasek, N Philip, R Day, J Taylor, K Strong, J Clayton-Smith*

(1:15) SPENCD: another 'immunoosseous' dysplasia; normal AGS1-4 sequence in an affected female - *Dr Yanick Crow, GI Rice, V Navarro,*

(1:16) Beta myosin heavy chain gene mutations in hypertrophic, dilated and restrictive cardiomyopathy patients - *Mr Taranjit Singh Rai, Dr Ajay Bahl, Mrs Monica Bhardwaj, Dr Balvinder Singh, Prof. Madhu Khullar*

(1:17) ACE I/D polymorphism in Indian patients with hypertrophic cardiomyopathy and dilated cardiomyopathy - *Prof Madhu Khullar, TS Rai, PS Dhandapany, TS Ahluwalia, A Bahl, B Singh*

(1:18) Searching the genome by phenotype; new developments in DECIPHER http://decipher.sanger.ac.uk - *Dr Helen V Firth, RM Pettett, AP Bevan, S Richards, S van Vooren, H Fiegler, NP Carter*

(1:19) Investigation and management of the fetus with radial anomalies - *Dr Katrina Prescott, V Tripathi, P Boyd, J Hurst*

(1:20) Not the NF Clinic - *Dr Angus Dobbie, D Friend*

(1:21) Evaluating service development: a community based genetic service for consanguineous families of South Asian Origin - *Ms Naz Khan, R Mcleod, J Benson, V Hollings, H Kingston*

(1:22) Spectrum of dominant and recessive glycine substitutions in the COL7A1 gene in dystrophic forms of epidermolysis bullosa - *Dr Lu Liu, L Ozoemena, P Dopping-Hepenstal, P Love, J Mellerio, J McGrath*

(1:23) A case of familial duodenal atresia; do mutations within the Fgf10 signalling pathway underlie its pathogenesis? - *Dr Katie Snape, JM Milunsky, L Izatt*

(1:24) Coronary artery aneurysm: an unusual complication of Marfan syndrome - *Dr Michael Cunnington, IF Purcell, E Cross, JA Goodship*

(1:25) Monozygotic twins with MELAS - *Dr Yuko Morita, A Kondo, S Izumi, T Uesugi, H Takahashi, S Takagi*

(1:26) Etiological heterogeneity of trigonocephaly: a retrospective study - *Dr Usha Kini, J Byren, S Wall, A Wilkie, J Hurst*

(1:27) Alveolar Capillary Dysplasia: a further case report - *Dr Usha Kini, K McCormick, S Gould*

(1:28) The Perspectives and Experiences of Genetic Counsellors and Linkworkers Working Together in Clinical Genetic Consultations - *Miss Gaya Matta, T Clancy*

(1:29) COS outperforms other models for prediction of mutation status in BRCA1 and BRCA2 - *Dr Hassan Roudgari, N. E. Haites*

(1:30) A 20p subtelomeric deletion in association with an epilepsy phenotype has identified potential epilepsy candidate genes - *Dr Virginia Clowes, A Clarkson, S Mehta, L Willatt*

(1:31) Incomplete Denys-Drash Syndrome with multiple congenital anomalies: a case report - *Dr Virginia Clowes, E Roper, J Paterson, HV Firth*

POSTER PRESENTATIONS

(1:32) MEN2 screening dilemmas in a family with a novel Ret mutation in the MEN2 susceptibility region of the gene, a family history of Hirschsprung disease, and no family history of MEN2-related tumours - *Dr Virginia Clowes, C Shaw-Smith, H Simpson, S Ball, C Acerini*

(1:33) Duplication of the Williams-Beuren locus in three family members, with variable penetrance and clinical features. - *Dr Simon Holden, C Donaghue, A Hills, AF Davies, JW Ahn, C Mackie Ogilvie*

(1:34) DNA missing in action: Deafness with X-linked agammaglobulinaemia is the clue to a contiguous gene deletion syndrome - *Dr Catherine McWilliam, C Longman, J Duncan, R Hague*

(1:35) Bipolar disease in a patient carrying an apparently balanced reciprocal translocation with a breakpoint in 11q24.2, a candidate locus for schizophrenia - *Dr Simon Holden, A F Davies, S Bint, A Dunlop, R Blennerhassett, R C Trembath, W Reardon*

(1:36) Case series of 1q21.1 microdeletion: Genomic rearrangement of uncertain significance? - *Dr Serena Nik-Zainal, G Parkin, A Clarkson, M Bateman, I Simonic, S Mehta, H Firth, L Willatt*

(1:37) Natural history of Williams syndrome: a report of 2 adult patients - *Dr Tabib Dabir, Alex Magee*

(1:38) A novel splice site mutation in P53 associated with ovarian cancer and sarcoma of the inferior vena cava - *Dr V K Ajith Kumar, N Sodha, R Eeles, R Stein, J Stein, J Lederman, A Male*

(1:39) Transmitted duplications of 8p23.1-8p23.2 associated with speech delay, autism and learning difficulties - *Ms Mary T Glancy, A Barnicoat, R Vijeratnam, S de Souza, J Gilmore, S Huang, V K Maloney, N S Thomas, D J Bunyan, A Jackson, J C K Barber*

(1:40) Down Syndrome with achondroplasia : a rare combination - *Dr Tabib Dabir, B McCrossan, A Sands, L Sweeney, A Magee*

(1:41) Back To The Future - A Patient's Journey: A case report on new, current and previous techniques used for prenatal diagnosis for an affected female patient with Ornithine Transcarbamylase (OTC) Deficiency - *Dr Derek Lim, S Sharif*

(1:42) "Atypical" Silver Russell Syndrome and 11p15 imprinting abnormalities - *Dr Katrina Tatton-Brown, R Scott, T Homfray, B Thilaganathan, N Rahman, S Mansour*

(1:43) Congenital Haemangiolymphangioma: A series of five prenatally diagnosed cases - *Dr Fiona Connell, T Homfray, B Thilaganthan, A Bhide, I Jeffrey, R Hutt, P Mortimer, S Mansour*

(1:44) Camptodactyly of the index fingers, cleft palate, choanal stenosis and coccygeal appendage in two siblings: a new autosomal recessive syndrome? - *Dr Melissa Lees, K Writzl*

(1:45) A combined genetic breast clinic for women at high genetic risk of developing breast cancer in North-East Scotland - *Dr Helen Gregory, E Smyth*

(1:46) Congenital Synspondylsim - a rare case of vertebral segmentation defect - *Dr Siddramappa Patil, M Bhat, S Rao, J Prasad*

(1:47) A patient with partial chromosome 16 trisomy mosaicism - *Dr Kristiina Avela, J Kirjavainen, S Knuutila, R Salonen*

(1:48) Auditing of antenatal diagnoses – What is good practice? - *Dr Sarah Ball, T Hutchin, G Gray, L Jenkinson, MA Preece*

(1:49) The Ghent nosology and Fibrillin 1 mutation detection in the diagnosis of Marfan Syndrome - *Dr John Dean, P Khau van Kien, M Claustres, R Howarth, J Harvey*

(1:50) Management of Neurofibromatosis 1 Patients by Genetics Centres in the South West of Britain: An Audit - *Dr Caroline Pottinger, S Sharif, L Side*

(1:51) Predictive Testing for Carney Complex in a Child - *Dr Deirdre Donnelly, S McKee*

(1:52) Unexpected Detection of PRKAR1A Deletion - *Dr Moira Blyth, K Temple, J Crolla, V Maloney, S Huang*

(1:53) Fowler syndrome: a clinical and neuropathological study of six further cases - *Dr Denise Williams, K Kalyanasun, R Scott, T Marton, C Patel, L Brueton, W Martin, P Cox*

(1:54) Weyers Acrofacial Dysostosis: The first reported family in the UK - *Dr Derek Lim, S Sharif, H Cox*

(1:55) Neural tube defects in 21st century: Is Northern Ireland changing? - *Dr Tabib Dabir, F Stewart*

(1:56) Is assessment of lipid phenotype needed for cascade genetic screening in Familial Hypercholesterolaemia Families? - *Miss Alison Taylor, RDG Neely, S Fairgrieve, D Wang, S Humphries, G Norbury*

(1:57) Premature ovarian failure in Fragile X syndrome – counseling dilemmas in a large family - *Dr Carol Chu, C Bennett, A Dobbie, H Powell, C Falconer*

(1:58) Arterial calcification and cystic renal disease- aspects of the same condition or separate entities - *Dr Katrina Tatton-Brown, I Jeffrey, S Mansour*

(1:59) Interpretation of a complex FSHD result in an Asian family in relation to initially apparent compound heterozygosity - *Dr Joanna Prothero, P Lunt, F Muntoni, A Manzur, S O'Shea, M Williams, E Wakeling*

(1:60) Another nail tale: volar nail-like finding associated with interstitial deletions of Chromosome 6q - *Dr Joanna Prothero, E Hawes-Collins, RJ Ellis, R Gupta, AF Brady*

(1:61) Del(14)(q22q23) associated with anophthalmia, craniofacial abnormalities and brain malformations; case report and molecular cytogenetic analysis of an emerging contiguous gene syndrome - *Dr Simon Holden, NK Sawyer, J Gilmore, D Bunyan, M Dattani, JRO Collin, AF Davies, N Ragge*

(1:62) Preimplantation Genetic Diagnosis for inherited cancer predisposition – Different Strategies - *Ms Seema Dhanjal, T Mamas, S Jaroudi, G Kakourou, S SenGupta*

(1:63) Variation in Wnt-7a is unlikely to be a cause of familial Congenital Talipes Equinovarus - *Dr G Liu, J Inglis, A Cardy, D Shaw, S Sahota, L Sharp, Z Miedzybrodzka*

(1:64) Congenital fibrosis of extraocular muscles, developmental delay and laterality problems: report of a new association - *Dr Adonis Ioannides, E Rosser*

(1:65) A Leap in the LEOPARD Phenotype? - *Dr Alison Male, M Bitner-Glindzicz, J Short, N Sebire, A Magee, J Harper, R Hennekam*

(1:66) How Isolated are Isolated Antenatal Anomalies? - *Dr Moira Blyth, D Wellesley, L Styles*

(1:67) Genetic variation in RYR1 and disease pathology - *Dr Danielle Carpenter, R Robinson,, R Quinell, C Ringrose, P Booms, D Iles, PJ Halsall, D Steele, P Hopkins, M-A Shaw*

(1:68) Gaucher disease type IIIC in two Indian cousins: unusual phenotype in a rare disorder- *Dr Meenakshi Bhat, SJ Patil, S Shetty, S Shah, H Matalia, Sujay Prasad, P Lissy*

(1:69) First use in Wales of audit tool for genetic services - *Prof Angus Clarke, B. D. Hendicott*

(1:70) A Scottish patient with Juvenile Paget's disease due to compound heterozygosity in the TNFRSF11B gene - *Dr Ayesha Ahmed, S Mumm, M Whyte, J Tolmie, P Galloway*

(1:71) Novel 1q23.3q24.1 microdeletion causes short stature, cardiac abnormalities and developmental delay - *Dr Sarju Mehta, C Shaw-Smith, G Parkin, I Simonic, L Willatt*

(1:72) Clinical and Molecular analysis of Adams Oliver Spectrum of disorders: A European Study - *Dr Deborah Ruddy, W Wyuts, W Reardon, M Zenker, R Trembath*

(1:73) Costello syndrome: clinical and molecular characterisation of 7 Portuguese patients - *Dr Ana Berta Sousa, A. Medeira, B. Filipe, H.G. Santos, I. Gaspar, M. Venâncio, M. Martins, I. Cordeiro*

(1:74) Severe form of split-foot deformity with mandibulofacial dysostosis – Paterson-Stevenson-Fontaine syndrome - *Dr Sergio Sousa, L Ramos, J Sa, J Saraiva*

(1:75) Ophthalmo-acromelic syndrome - case report - *Dr Lina Ramos, S Sousa, C Paiva, F Ramos, J Saraiva*

(1:76) A recognisable 6p deletion syndrome and a locus for Perthes' disease? - *Dr Lily Islam, KS Waters, J Ragoussis, KD MacDermot*

(1:77) A phenotype in three patients with a novel interstitial 2p25.1-p25.3 deletion - *Dr Alison Kraus, R Fisher, B Taylor, A Nkrumah, C Chu,*

(1:78) Mosaicism for a PKD1 gene mutation revealed through family genetic analysis for a living related donor transplant for autosomal dominant polycystic kidney disease (APKD) - *Dr Peter Lunt, C Dolling, Y Patel, L Meredith, Athena Diagnostics, A Gardner, A Connor, C Dudley*

(1.79) Fingerprints: a new look at Naegeli-Franceschetti-Jadassohn syndrom – *Helen Murphy, J Lugassy, E Sprecher, JA McGrath, JL Verbov*

2. Molecular Genetics

(2.01) An audit of exon copy number variants detected by MLPA in BRCA1 and BRCA2 - *Mr Will King, A Johnson-Marshall, L Ormshaw, L Walton, FL Lavender, R Taylor*

(2.02) Development of a diagnostic test for MYH associated polyposis (MAP) - *Ms Sian Davies, PM Bond, A Curtis*

(2.03) Point Mutation Screening Service for Spinal Muscular Atrophy - *Ms Rebecca Brown, Shu Yau, G Haebich, S Abbs*

(2.04) Mutation of junctophilin type 2 associated with hypertrophic cardiomyopathy - *Mr Yoshihisa Matsushita, T Furukawa, H Kasanuki, M Nishibatake, Y Kurihara, A Ikeda, N Kamatani, H Takeshima, R Matsuoka*

(2.05) Marfan syndrome: CSCE as a rapid sensitive technique for screening the FBN1 gene - *Miss Catharina Yearwood, M Schijvenaars, D Ward, C Mattocks, J Harvey*

(2.06) Retention of mutated proteins in the endoplasmic reticulum and their degradation by ERAD is the most common mechanism of monogenic loss-of-function diseases - *Dr Bassam Ali, BR Ali*

(2.07) Potential for a false positive myotonic dystrophy result due to a restriction site polymorphism - *Miss Sandra Ramos, J Short, S Spiden, S Cottrell, R Taylor, R Poh*

(2.08) MLPA analysis to detect large deletions and duplications in Familial Hypercholesterolaemia - *Mr Brendan Martin, D Wang, A Taylor, S Humphries, G Norbury*

(2.09) Novel loss of function mutations in Asian kindreds with hereditary spastic paraplegia and thin corpus callosum (SPG11) - *Ms Laura Baumber, S Patel, E Kinning, P Critchley, AH Nemeth, K Talbot, ER Maher, RC Trembath*

(2.10) Genome-wide analyses in a consanguineous family with autosomal recessive spondyloepimetaphyseal dysplasia and abnormal dentition exclude known genes and provide evidence for a further SEMD locus - *Ms Laura Baumber, E Kinning, ER Maher, RC Trembath*

(2.11) An audit of the first five years of a diagnostic service for Noonan/LEOPARD Syndrome - *Mr John Short, R Poh, R Taylor*

(2.12) Stiochiometric imbalance in the receptor complex contributes to dysfunctional BMPR-II signalling in pulmonary arterial hypertension - *Dr Talat Nasim, RC Trembath*

(2.13) X-linked Idiopathic Infantile Nystagmus Associated with a Missense Mutation in FRMD7 - *Dr Alan Shiels, TM Bennett, JM King, JB Prince, L Tychsen,*

(2.14) Using synthetic MLPA probes to detect deletions in X-linked diseases - *Dr Rachel Smith, S Waller, H Martin, J Whittaker*

(2.15) Developing a diagnostic service for screening ABCA3, SFTPB and SFTPC genes in patients with Hereditary Pulmonary Surfactant Deficiency - *Miss Caroline Archer, Q Mok, G Norbury*

(2.16) Mutational analysis of patients with early onset retinal dystrophies - *Dr Donna Mackay, P Moradi, RH Henderson, AR Webster, AT Moore*

(2.17) A Multicentre Technology Assessment of the Abbott Fragile X Assay - *Dr Andrew Wallace, D Barton, PA van Bunderen, J Duncan, J Dunlop, S Man, J MacPherson, G Monaghan, J McLuskey, G Norbury, Y Patel, H Powell, V Race, M Sweeney, E Thompson, R Treacy, MM Weiss, N Williams, HE White, B Wymer*

(2.18) Long QT syndrome patients with double heterozygous and compound heterozygous mutations - *Ms Karen Livesey, K Thomson, J Thistleton, H Lord, E Blair, A Seller*

(2.19) Large and complex GALT deletion in galactosaemia patients - *Mrs Sarah Burton-Jones, H. Sawyer, M. Gable, P. Briones, A. Morris, P. Lee, M. Cleary, M. Williams,*

(2.20) POLG and PEO1 gene mutations in autosomal disorders of mitochondrial DNA maintenance - *Dr Conrad Smith, C Fratter, A O'Rourke, I Dow, H Lord, J Poulton, A Seller*

(2.21) iPLEX mass array, MLPA and high throughput sequencing provide a comprehensive mutation detection service for Familial Hypercholesterolaemia - *Dr Colin Graham, WT Wright, SV Heggarty, C Byrne, IS Young, DP Nicholls*

POSTER PRESENTATIONS

(2.22) A novel test for the detection of inversion mutations - *Miss Wendy Roworth, DJ Bunyan, DO Robinson, JF Harvey*

(2.23) A High Frequency of Morquio Syndrome type A in a Genetic Isolate in an immigrant population - *Dr Sarah Ball, T Hutchin, S Khan, A Ahmad, A Chakrapani, C Hendriksz, MA Preece, P Kampanis, A Cooper, G Gray*

(2.24) DNA analysis for Non Ketotic Hyperglycinaemia in the UK population - *Dr Sarah Ball, T Hutchin, S Kure, J Van Hove, G Gray*

(2.25) No evidence for RYR1 epigenetic silencing in patients with MH susceptibility - *Dr Rachel Robinson, D Carpenter, P Booms, D Iles, D Steele, PJ Halsall, PM Hopkins*

(2.26) A fast-throughput service for Familial Hypertrophic and Dilated Cardiomyopathy using High resolution melt curve analysis on the Lightscanner - *Miss Jessica Thistleton, K Thomson, M Wilson, K Livesey, J McKinney, E Blair, H Watkins, A Seller*

(2.27) Importance of RB1 Promoter Hypermethylation in Retinoblastoma Genetic Screening - *Ms Sugera Hashim, KS Price, EA Price, K Pritchard, Z Onadim*

(2.28) A Molecular Diagnostic Service for Craniofrontonasal Syndrome - *Dr Louise Williams, AW O'Rourke, J Hurst, L Basel, A Seller, AOM Wilkie, T Lester*

(2.29) Diagnostic service for congenital adrenal hyperplasia – development of sequencing and dosage methods improves mutation detection accuracy and turnaround time - *Mr Ian Berry, M Mohammed, S Bibi, J Schouten, R Charlton*

(2.30) Case Report: Two pathogenic, NF1 gene mutations identified in DNA from the peripheral blood of a child with cafe au lait patches, juvenile myelomonocytic leukaemia and familial macrocephaly - *Mr Hood Mugalaasi, KK Mantripragada, G Spurlock, JE Morton, MJ MacDonald, M Upadhyaya, R Butler*

(2.31) An improved service for Preimplantation Genetic Diagnosis (PGD) for Huntington disease by haplotyping - *Miss Jane Trussler, C Black, A Lashwood, P Braude, P Renwick*

(2.32) Dyggve-Melchior-Clausen syndrome: Molecular characterisation of dymeclin in bone development and disease - *Ms Celine Denais, CL Dent, J Hoyle, E Kinning, RC Trembath*

(2.33) Locus Heterogeneity in Autosomal Recessive Congenital Cataracts - *Dr Esther Meyer, F Rahman, S Pasha, T Forshew, AT Moore, NV Morgan, ER Maher*

(2.34) A study of the Androgen Receptor Gene Polymorphism and the Level of Expression of the Androgen receptor in Androgenetic Alopecia among Egyptians - *Dr Heba Kassem, R. Abou El Seoud, A. El Shafaei, H. Ibrahim, M. El Ramly*

(2.35) Atypical methylation pattern in twins considered to be affected with Angelman syndrome - *Dr Valerie Wilson, S Tennant, C Kettle, A Stewart, A Curtis, D Bourn*

(2.36) Aicardi-Goutieres Syndrome – Introduction of a Diagnostic Service for a Rare Recessive Disorder - *Miss Teresa Patrick, YJ Crow, K Flintoff, A Sharpe, R Charlton*

(2.37) Neonatal screening for cystic fibrosis in the Oxford genetics Laboratory: A review of the first year of testing - *Miss Helen Lord, L Williams, T Lester, A Roberts, J Kay, I Smith, A Thomson, A Seller*

(2.38) Implementation of the national CF newborn screening programme – the Leeds experience one year on - *Mrs Sarah Shepherd, H Lindsay, L Mavrogiannis, L Shapiro, K Brownlee, C Chu, D Cockburn, R Charlton,*

(2.39) Cystic Fibrosis Newborn Screening: Development, Implementation and 6 Months Experience in the Bristol Genetics Laboratory - *Dr Claire Faulkner, H Sawyer, M Greenslade, T Simmons, R Salamanca, J Worgan, P Grundy, A Gardner, S Maund, M deHora, A Brown, H Kemp, M Williams*

(2.40) A single tube assay for the 6 most common inherited spinocerebellar ataxias - *Ms Aisha Ansari, T Azam, T Bradley, A Diamond, N Williams, J Warner*

(2.41) MUTYH-associated polyposis mutation screening; the Cardiff experience - *Miss Sarah Maund, N Jones, R Butler, JR Sampson*

(2.42) Carrier testing in a spinal muscular atrophy family; untangling the genotypes - *Miss Ruth Lewis, J Fenton-May, AJ Clarke, D Cairns, F White, R Butler*

(2.43) The potential application of Multiple Displacement Amplification for Preimplantation Genetic Diagnosis for Haemoglobinopathies in the UK - *Dr Sachin Wani, B Wild, M Petrou*

(2.44) A novel variant in the HFE gene leads to a false positive result - *Miss Marta Pereira, S.J. Payne*

(2.45) First live birth of an unaffected child in the UK following PGD for Type 2 Gaucher Disease - *Miss Colleen Lynch, S Fishel, MRH Hughes, M Studt, J Kitchen, S Brown*

(2.46) Loss of MLH1 expression due to promoter methylation in cases referred for molecular investigations for HNPCC - *Ms Samantha Fisher, L Hawkes, M Novelli, SJ Payne*

(2.47) Estimating carrier risks by linkage in a DMD family with a triple X female - *Dr Dr. Aileen Butler, DE Barton*

(2.48) Two sisters with Rett syndrome and different deletions in the MECP2 gene - *Dr Lazarus Lazarou, SA McKee, L Rosser, D Cooper, J Hughes, R Butler*

(2.49) A summary of CDKL5 gene screening in Cardiff 2006 to 2007 - *Mr Mark Stephens, R Ellwood-Thompson, HL Archer, LP Lazarou, R Butler*

(2.50) GJB2 testing in hearing impaired British Asians: referral frequency, detection rate, and mutation spectrum - *Dr Lampros Mavrogiannis, C Bosomworth, R Birmingham, M L I Esmyot, S M Sharif, J S Rowland, D J Cockburn, R S Charlton, C P Bennett*

(2.51) The E477X MYH-causing mutation is a major contributor of colorectal cancer in the Gujarati population - *Miss Farrah Khawaja, SJ Payne*

(2.52) Novel mutations in the CYP4v2 gene associated with Bietti crystalline corneoretinal dystrophy - *Dr Stephanie Halford, L Ocaka, D Mackay, S Peirson, A Moore, A Webster*

(2.53) Implementation of a new cDNA screening strategy as an adjunct to the West Midlands Regional Genetics Breast Cancer Service – *Emma Sach, R Barber, J Bell, F Macdonald, Y Wallis*

(2.54) Molecular Genetic Testing of the Sarcoglycanopathies at the Northern Genetics Service – *Mr Daniel Routledge, J Hudson, A Curtis*

(2.55) Unusual EcoR1/EagI digest pattern in a permutation carrier of Fragile X (A): a counselling and testing dilemma – *Lazarus Lazarou, M MacDonald, Q Roberts, D Pilz, A Cowe, R Butler*

(2.56) Possible expansion of an intermediate FMR-1 CGG repeat allele to a full mutation in one generation – *Jo Martindale, B Wymer, Maria Panayi, A Dalton*

(2.57) An unusual case of somatic mosaicism in a mildly ataxic 84 year old man referred for ataxia testing – *Helen Stuart, M Panayi, J Martindale, A Dalton*

(2.58) Audit of Microsatellite Instability testing procedures in moderate risk bowel cancer families – *Danielle Crompton, D Bourn, L Strain*

3. Cytogenetics and Molecular Cytogenetics

(3.01) A child with two isodicentric Y chromosomes resulting in tetrasomy Yp and nullisomy Yq - *Dr Hazel J Clouston, HJ Rolf, OW Quarrell, S Williams*

(3.02) Cytogenetic evaluation of Ewing sarcoma family of tumour cell lines - *Miss Josie Hayes, S Burchill, P Roberts*

(3.03) Investigation of Chromosome breakage as a risk factor in the development of sporadic breast cancer amongst Iranian women - *Ms Forough Mojtahedi, K. Kahrizi, F. Seirati, E. Keyhani, Sh. Akhlaghpour, M. Karimloo, S. Ghasemi Firouzabadi, I. Bagherizadeh, F. Behjati*

(3.04) Pseudo hermaphrodism in an Iranian family with Mental Retardation - *Ms Saghar Ghasemi Firouzabadi, K. Kahrizi, H. Najmabadi, F. Mojtahedi, Y. Heshmati, M. Fakhri, R. Vazifehmand, F. Behjati*

(3.05) Rearrangement-specific breakpoint clustering at chromosome band 3q26: implications for EVI1 FISH in myeloid malignancy - *Ms Valeria AS de Melo, M Vetter, J Howard, DR Betts, H Mazzullo, EP Nacheva, JF Apperley, AG Reid*

(3.06) Two cases of unexpected findings at CVS - *Miss Anne Marie Lloyd, J M Tracey, K L Gaunt*

(3.07) Does amniocentesis in the mid-trimester cause congenital talipes equinovarus? - *Dr Zosia Miedzybrodzka, AH Cardy, N Torrance, D Clark, L Sharp*

(3.08) Partial trisomy 13: A case report, verification of the phenotype and review of the literature - *Dr Geoff Smith, D McManus, V McConnell*

(3.09) Cheap as CHIPS: Array CGH with 1Mb resolution for a reagent cost of under £100 per test - *Dr Lionel Willatt, G Parkin, I Simonic, J Whittaker*

(3.10) Copy Number Variation: Discrepancies between Array and FISH Studies - *Dr Lionel Willatt, G Parkin, A Clarkson, I Simonic, J Whittaker*

(3.11) A study of genetics, clinical and epidemiology in microcephally children referred to Emam Reza Hospital, Mashhad, Iran. - *Dr Kazem Ghodsi, A Mohammad Zadeh, A Shah farhat, F Mohajer Tehran, M Saied Zadeh, A Taghavi*

(3.12) Reduction of maternal lymphocytes in bloodstained amniotic fluid by centrifugation and subsequent QF-PCR anuploidy testing - *Miss Philippa May, H Thomas, R Scott, K Mann*

(3.13) Terminal deletion of an inherited 8p paracentric inversion – broken recombinant or independent deletion? - *Dr Carolyn Dunn, M Bateman, D Baralle, L Willatt, I Simonic*

(3.14) The presence of a ring chromosome and acentric fragment derived from chromosome 3 with mitotic stabilisation via a neocentromere. - *Mrs Lyndsey Croft, K J Glover, G A Fews, E V Davison*

(3.15) A large non-heterochromatic neo-centromeric marker with unidentifiable origin - *Dr Christopher Kettle, S W Hellens, A Stewart, M J Wright, S A Zwolinski*

(3.16) Three clinical pictures of Palllister Killian syndrome - *Miss Marta Pinto, A Jardim, E Matoso, A Mascarenhas, P Paiva, J Ferrao, Equipa de DPN da MDM, Equipa de DPN da MBB, J Sa, I Marques Carreira,*

(3.17) Incidence of common microdeletions and microduplications in 658 patients with developmental delay, dysmorphism and/or congenital abnormalities - *Dr Kathy Mann, JW Ahn, H Thomas, C Donaghue, A Hills, C Mackie Ogilvie*

(3.18) Acquired idic(X)(q11) in a 5-year-old girl with Schwachman-Diamond - *Miss Adele Reynolds, J Corfield, T Kus, M Cummins, C Kitchen, L Burvill-Holmes, E Roberts, T Davies*

(3.19) Frequency of triploidy and mosaicism in prenatal diagnosis of chromosome abnormalities in chorionic villus and amniocyte cultures - *Dr Sajayan Joseph, T Ballard*

(3.20) A report on a rare jumping translocation involving chromosome reqion 1q12 in a patient with myeloid disease - *Ms Rachel Ray, L Brown, D Ladon, B Czepulkowski, GM Mufti*

(3.21) Chromosomal abnormalities, risk factors and clinical manifestations study of Down's syndrome in North East Iran, 2005-2006 - *Dr Kazem Ghodsi, F Mohajer Tehran, G Mamoori, E Hosainpoor, M Saeid Zadeh, A Taghavi, H Boskabadi*

(3.22) Case Study: Characterisation of a rare, familial interchromosomal insertion - *Mrs Siobhan Toomey, L Burvill-Holmes, E Roberts, S Smithson, N Leo, C Delmege, T Davies*

(3.23) Case Study: A Vanishing Twin in a Chorionic Villi Sample - *Mrs Helena Hennessey, S Henderson, J McCarthy, J Moore, J MacGregor, A Palmer, M Jones, D Delmege, T Davies*

(3.24) Analysis of 22q11 deletion in foetuses with increased nuchal translucency using the Vysis TUPLE1 probe - *Miss Abigail Whitworth, I Barnes, D Anumba, E Maltby*

(3.25) Diagnosis of Nijmegen breakage syndrome raises questions regarding inheritance - *Mrs Meenakshi Minnis, H Ward, K Metcalfe, L Gaunt*

(3.26) A QF-PCR Audit - "Unfogging the Future"? - *Miss Susan Hamilton, KL Gaunt*

(3.27) Introduction of the Cytocell Chromoprobe Multiprobe CLL Panel as a routine diagnostic service - *Miss Louisa Wilson, L Gallagher, D Stevenson*

(3.28) Distal 4q interstitial deletion of 8-10Mb with no discernible phenotypic effect - *Mr Mark Bateman, S Mehta, L Willatt, L Sparnon, E Selkirk, C Bedwell, I Simonic*

(3.29) Looking for predictors of aCGH quality: a retrospective study of the outcome of 38 Agilent 185K oligonucleotide arrays - *Mr Laura Bisoni, JB Studd, M Buch, AIF Blakemore, RJ Ellis*

(3.30) Subtelomeric MLPA as an Adjunct to Karyotyping - *Mr Ben Holgado, C Lambert, A Callegari, S Joseph, R Palmer, R Wang, T Ballard, S Liddle, L Levett*

(3.31) De Novo Complex Chromosome rearrangement (CCR) - 46,XX,t(4;5;7) associated with developmental delay and hypotonia – Cytogenetics, FISH and SKY analysis. - *Dr Jayarama Kadandale, RR Devi, P Swaroop, A Aravind, P Rajitha,*

POSTER PRESENTATIONS

(3.32) Developing an arrayCGH service for constitutional cytogenetics – initial insights. - *Dr Joo Wook Ahn, SE Walsh, K Mann, C Mackie Ogilvie*

(3.33) A pregnancy with an abnormal heterogeneous karyotype suggestive of mosaic variegated aneuploidy - *Ms Keiko Asakura, C.T. Maliszewska, J. Berg, N.R. Pratt,*

(3.34) Gene Paucity and Pervasive CNV are Indicators of the Chromosome Anomalies - *Dr Art Daniel*

(3.35) Micro-array comparative genomic hybridisation (array CGH) analysis of over 90 unrelated patients with dysmorphic features and mental retardation – *SW Hellens, SA Zwolinski, MJ Wright, MP Splitt*

(3.36) Deletion and duplication of the same two BAC clones at 16p13.11 in two patients with distinctive phenotypes – *MP Splitt, SW Hellens, E Jones, T Montgomery, SA Zwolinski*

4. Counselling, Social, Policy and Education

(4.01) Development of a Set of Patient Leaflets about Genetics and Genetic Testing for Use in Genetic Clinics Across Europe - *Ms Celine Lewis, HS Dr Heather Skirton, DC Dr Domenico Coviello, AC Mr Alastair Kent*

(4.02) Education of genetic counselling for medical students in Japan - *Dr Akane Kondo, Y Morita, N Uchida, S Izumi, M Mikami*

(4.03) Finding a route through the maze of current information and services for individuals and families affected by or at risk of six rare genetic conditions: the patients' perspective. - *Mrs Anna Allford, M Winter*

(4.04) Genetic Led Multidisciplinary Clinics as Communities of Practice - *Ms Sally Watts,*

(4.05) Dedicated primary care genetic counsellors: Effect on uptake of genetic services. - *Ms Marion Turnbull, JL Wiggins, M Lees, L Chitty,*

(4.06) Development of a culturally sensitive translation protocol - *Dr Amy Hunter, A Kessling, M Winter, P Mehta*

(4.07) Cancer in the family-see your geneticist,! Who will go and who won't - *Dr Susan Holloway, B Bernhard, J Campbell, H Campbell, WW Lam*

(4.08) Mainstreaming Genetics – A Guide for the Design of a Service Development Blueprint - *Dr Ian Ellis, C Benjamin, M Wardle, A Douglas*

(4.09) International developments in public health genomics - *Dr Philippa Brice, R Zimmern, H Burton, A Stewart*

(4.10) Family Communication about Cystic Fibrosis from the Mother's Perspective: An Exploratory Study - *Miss Nicola Coates, F M Gregory, E P Parsons, A J Clarke*

(4.11) SGPPH: Society for Genomics Policy and Public Health - *Dr Mair Crouch, H Burton, A Hall, M Hopkins, L Jader, A Kent, M Kroese, I Rafi, C Patch*

(4.12) Consanguineous marriage and its relevance to congenital abnormalities: Parental consanguinity as a cause for increased incidence of births defects. - *Dr Reza Akbarzadeh, Mash Akbarzadeh. Reza, Molec Asadzadeh. Jamshid*

(4.13) Public health perspectives on prevention of morbidity and mortality for genetics disease. - *Dr Reza Akbarzadeh, Mash Akbarzadeh. Reza, Molec Asadzadeh. Jamshid*

(4.14) Study on informational support for prenatal examination by medical professionals of a prefecture in Japan - *Prof Shun-ichiro Izumi, A Kondo, N Uchida, Y Morita, M Ishii, K Wada, H Yokoyama, M Mizoguchi*

(4.15) 21st Century Education for Trainees: Bringing Clinical Genetics Up To Date - *Dr Ayesha Ahmed, C McWilliam, J Tolmie*

(4.16) Boy Genius: Exploring Genetics through Interactive Theatre for Young People - *Miss Flo Ticehurst, B Williams, P Gibbins, L Osborn*

(4.17) Developing a voice recognition system for clinical correspondence in a regional clinical genetics service - *Dr Helen Stewart, S Durell, E Collier, G Fazil*

(4.18) Making the most of learning opportunities in the genetics clinic - *Dr Melissa Martyn, J Haydon, D Williams, S Price, P*

(4.19) Development of molecular biology laboratory training courses for health care professionals at the Nowgen Centre, The North West Genetics Knowledge Park - *Dr Mark Leech, F Salway, F Jury, P Day, D Carthy, D Donnai, H Middleton-Price*

(4.20) Homozygous Huntington's; ethical implications of an unexpected diagnosis - *Dr Frances Elmslie, S Rose, S Spiden, C Willoughby, R Taylor, M Peterson*

(4.21) Receiving genetic information: patients' experiences and preferences - *Mrs Sarah Burke, C Bennett, J Bedward, P Farndon*

(4.22) The Development of a Combined role within Breast Cancer Care and Genetic Services in Bradford - *Mrs Sakina Iqbal G A Karbani, C E Chu, W Case, L Rowett, N Dharni*

(4.23) Enhancing genetics care in practice – workforce competences for non-genetics healthcare professionals - *Dr Catherine Bennett, S Kaur, L Gough, J Haydon, E Tonkin, A Daly, A Eaton, P Farndon*

(4.24) Introducing antenatal screening for haemoglobinopathies in low prevalence areas: lessons for Primary Care? - *Mrs Lynne Mathers, C M E McKeown*

(4.25) Intermediate Fragile X gene expansions: a review of genetic counselling practice and exploration of family experience - *Mrs Sally Taffinder, AJ Barnicoat, JV Bailey*

(4.26) Seeking Practice Development Unit Accreditation as a means of Improving Service Delivery - *Mrs Cath Kightley, J Birch, G Mannion, L Wall, C Higgins, I Ellis, A Fryer, G Core, H Chin, P Harris, C Benjamin, K Dymant, J Daly, T Bryning, S Coulson, G Buckle*

(4.27) A CVS false negative trisomy 18 QF-PCR result with a difference! - *Miss Nicole Motton, S.K. Allen, S. Whelton, E. Huxley, G. Hardy, S.A. Larkins, E.V. Davison*

(4.28) South Wales FH Forum – Improving information & policy input for families - *Miss Buddug Williams, D Townsend, P Rowlands, A Rees, J Williams, S Matthews, I McDowell*

(4.29) Exploring the Patient's Perspective using Drama and a Graphic Novel - *Dr Helen Middleton-Price, K Mathieson, P Dean, A Davison, A Sellyn, P Finegold, D Donnai*

(4.30) ScotGEN – A Scottish National Network to Improve Education in Clinical Genetics. - *Dr Jonathan Berg, S Sutherland, W Lam, M Van Mourik, G Scott, N Bradshaw, W Eboh, M Gray, R Cetnarskyj, P Porteous*

(4.31) An Archive for British Human Genetics - *Prof Peter S Harper, P Keelan*

(4.32) Towards Statutory Regulation of Genetic Counsellors in the United Kingdom – an update on the work of the Genetic Counsellor Statutory Regulation Steering Group (GCSRSG) – *Janet Birch, G Hall, C Barnes, S Durell, P Finnemore, C Jacobs, C Patch*

5. Cancer Genetics

(5.01) The long-term information and support needs of BRCA1/2 mutation carriers - *Dr Rachel Iredale, V Hunt*

(5.02) Improved BrCa testing: Implications for service delivery - *Ms Matta Gaya, JL Wiggins, D Ruddy, A Male, VKA Kumar*

(5.03) IGF-1 is a modifier of disease risk in Hereditary non-polyposis colorectal cancer - *Mr Stuart Reeves, RJ Scott, D Rich, CJ Meldrum, K Colyvas, G Kurzawski, J Suchy, J Lubinski*

(5.04) Psychological evaluation of familial ovarian cancer screening (PsyFOCS): A study outline - *Dr Kate Brain, D Lancastle*

(5.05) Premature chromosome condensation, Microcephaly and Mental Retardation: A report of two large consanguineous Iranian families - *Prof Farkhondeh Behjati, S. Ghasemi Firouzabadi, K. Kahrizi, M. Grshasbi, M.M. Motazacker, S.E. Esmaeili Nieh, S.S. Abedini, R. Vazifehmand, H.H. Ropers, H. Najmabadi*

(5.06) Juvenile Polyposis Syndrome (JPS) - a molecular diagnostic service - *Ms Tina Bedenham, T Cranston, K Gibson, A*

(5.07) An investigation into whether genetics services are meeting the needs of women who are at or near population risk for breast cancer - *Dr Nicola Taverner, F Pelz, R Iredale*

(5.08) Inherited mutations in BRCA1 are much more frequent than in BRCA2 amongst young breast cancer cases - *Ms Victoria N. Hammond, S. M. Gerty, J. Sillibourne, D. Ward, C. Mattocks, P. Simmonds, N. Graham, M. Armstrong, D. M. Eccles*

(5.09) Complex BCL2/IGH/MYC rearrangements in diffuse large B-cell lymphoma - *Mr Mark Sales, JJP Cunningham, NM Kernohan, GD Smith, CT Maliszewska, NR Pratt*

(5.10) Insertion of IGH into MYC in Burkitt Lymphoma identified by sequential FISH on paraffin sections - *Ms Joan Cunningham, MJ Sales, NM Kernohan, L Christie, CT Maliszewska, NR Pratt*

(5.11) The Effect of Dietary Energy Restriction On Gene Expression in Normal Breast and Subcutaneous Adipose Tissues of Overweight Women at Increased Breast Cancer Risk - *Dr Kai Ren Ong, AH Sims, M Harvie, RB Clarke, A Howell*

(5.12) Epigenomics Identifies Novel Tumour Suppressor Genes and Methylation Markers for Renal Cell Carcinoma - *Dr Mark Morris, D Gentle, C Ricketts, RE Banks, M Wiesner, T Kishida, M Yao, F Latif, ER Maher,*

(5.13) Should clinical genetics departments confirm every diagnosis before assessing risk in familial cancer? - *Ms Molly McEwan, V Hunt, EC Kivuva*

(5.14) Setting up a joint genetics/pathology service for immunohistochemistry and microsatellite instability testing for HNPCC – *service issues and initial case studies - Mr Ian Berry, R Charlton, N Scott, L Strain*

(5.15) Mutation analysis of FH and FLCN and Renal Cell Carcinoma (RCC) Susceptibility - *Dr Christopher Ricketts, ER Woodward, MR Morris, RS Houlston, PJ Morrison, F Latif, ER Maher*

(5.16) Screening for Familial Ovarian Cancer: Poor survival of BRCA1/2 related cancers - *Prof D Gareth Evans, KN Gaarenstroom, D Stirling, A Shenton, L Maehle, A Dørum, M Steel, F Lalloo, M Porteous, A van Asperen C, P Moller*

6. Other

(6.01) Molecular modeling and dynamics studies on implications of mutations in human cardiac beta myosin - *Mr Taranjit Singh Rai, M Datt, A Bahl, B Singh, M Khullar*

(6.02) Challenges for Genetic Testing in the UK - *Dr Peter Lunt*

British Human Genetics Conference 2007

Monday-Wednesday, 17-19 September

University of York

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 postal address (including post code) as this is the only information that will be used to send out your delegates pack.

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

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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 17 August.
- We regret that fees paid cannot be reimbursed if you have to cancel your registration after 17 August.
- We cannot supply invoices in respect of registration fees

CONFERENCE COSTS

Registration Fees	**Member**	**Non-Member**
Monday, Tuesday, Wednesday 17-19 September	£286.00	£358.00
Individual days	£108.00	£142.00
Accommodation (per night)		
Standard Single	£42.00	
En-suite single	£56.00	
Conference Dinner (York Racecourse)	£39.00	
Evening Meals (Sunday and Monday only)	£15.00 (per night)	
Car Parking	£11.00 (Three day permit)	

UK Delegates - your Delegates pack will be sent to you at the end of August.

Overseas Delegates – general information, including details of accommodation, will be sent to you at the end of August – your Delegates pack will be available at the conference from Sunday 16 September at 4.00p.m.

Closing date for registration:

17 August 2007

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