



JOINT MEETING  
**UK / Dutch Clinical Genetics Societies  
& Cancer Genetics Group  
Spring Conference**

15<sup>th</sup> - 16<sup>th</sup> March 2012

Life Conference and Banqueting, Centre for Life,  
Times Square, Newcastle upon Tyne NE1 4EP

# Information, Programme and Registration

## Scientific Programme

The Clinical Genetics Society and Cancer Genetics Group Scientific Programme Committees from the UK and the Netherlands are once again holding this joint meeting in the UK, this time in Newcastle at the 'Centre for Life'.

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

This meeting is preceded on 14<sup>th</sup> March by a Dysmorphology Club Meeting also at the Centre for Life, Newcastle.

## Venue

The venue for the event is the Life Conference Centre which forms part of the International Centre for Life. Life is a pioneering science village based in the heart of Newcastle upon Tyne and is home to the region's award winning Science Centre, Newcastle University Institute of Genetic Medicine and the Northern Genetics Service.

[www.life.org.uk](http://www.life.org.uk)

Life is directly adjacent to the Central Station and easily accessible by public transport. Metro trains run directly from the airport and a bus runs directly from the DFDS ferry port. For those driving, several car parks are also close by, including the Times Square Car Park.

[www.life.org.uk/visit-us/times-square-car-park](http://www.life.org.uk/visit-us/times-square-car-park)

## Accommodation

There is a *Dedicated Website* for booking Accommodation [www.conferencebookings.co.uk/delegate/NGIGENETICS2012](http://www.conferencebookings.co.uk/delegate/NGIGENETICS2012)

## Travel

Life Conference & Banqueting is situated in the heart of Newcastle upon Tyne, one of Europe's most cosmopolitan cities. Newcastle has a vibrant mix of the contemporary, cultural, stylish and historic and is famed across the world for its captivating and hospitable welcome. For more information about Newcastle Gateshead visit: [www.visitnewcastlegateshead.com](http://www.visitnewcastlegateshead.com)

Life is located right next to Newcastle Central Station and has its own car park nearby.

### **By car**

Life is situated a few minutes drive from the A1(M), the main route between the UK capital cities of London and Edinburgh. There are over 400 secure car parking spaces available at Times Square Car Park.

### **By train**

Life is located just 200 metres from Newcastle Central Station and the Metro underground station. Metro trains connect all areas of Tyne & Wear, including Sunderland, North Tyneside, South Tyneside, Gateshead and Newcastle International Airport. Regular Metro trains can transport visitors between Life and Newcastle Airport within 30 minutes.

### **By air**

Newcastle International Airport is just 6 miles from the city centre, 15 minutes by car, and has connecting scheduled flights to all major UK and many European airports.

### **By foot**

Newcastle is a very compact city and can be easily walked from one side to the other, in less than 15 minutes.

## Conference Dinner

Overlooking the River Tyne, the Hilton Newcastle-Gateshead Hotel is a short walk from the conference venue and city centre. A three course locally themed meal with wine will be followed by coffee, special guest 'speaker', Baba Brinkman, and a band and disco. Baba is a hugely entertaining hip hop artist, writer, and actor who will present from The Rap Guide to Evolution, a smash hit at the Edinburgh Fringe and around the world. Following this, the band will feature a local celebrity of our own.

[www.hilton.co.uk/newcastlegateshead](http://www.hilton.co.uk/newcastlegateshead)

## Registration

Registration is via the BSHG website – no other method of registration will be accepted. Onsite Registration will be available at the conference, at a higher rate. 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 27 February**. 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.

### **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 24 February 2012 (23:59).
- we regret that fees cannot be reimbursed if you have to cancel your registration after 24 February 2012 (23:59).
- If you require an invoice then please email [bshg@bshg.org.uk](mailto:bshg@bshg.org.uk) to request this giving full details to whom the invoice should be directed, including order numbers.
- If Payment is to be made by your Trust, please clearly indicate this on the auto-generated acknowledgement sheet and send a copy of this to the BSHG office.

## Confirmation of Registration

This year we are asking all delegates to collect their Conference Packs from the Registration Desks at the venue. These will be open from 07:30 on 15 & 16 March. **Please Note:** For those attending the Dysmorphology Meeting you will also need to collect your packs on the 15<sup>th</sup> or 16<sup>th</sup>. General information, including joining Instructions, map of campus and travel information, will be sent to you after the closing date of 24 February by EMAIL.

## CPD Approval

The conference has CPD approval by the Royal Colleges.

## Abstract Booklet

For those who cannot attend the meeting but wish to order a copy of the Abstract Booklet, these will be available from the BSHG Office at a cost of £5.00 each, cheques to be made payable to the 'British Society for Human Genetics'.

## Emergency Telephone Numbers

+44 (0) 7748 291 429 - Dr Alex Henderson (Newcastle)

## Meetings

**DYSMORPHOLOGY CLUB MEETING** – 10:00 Wednesday 14 March, £20

### CLINICAL GENETICS SOCIETY

*Annual General Meeting* – 12:10 Thursday 15 March

### CANCER GENETICS GROUP

*Steering Group Meeting* – 07:30 Friday 16 March

## Future Meetings

### CLINICAL GENETICS SOCIETY

*2013 Spring Conference* – Thursday 14 March 2013, School of Oriental and African Studies (SOAS), London

### CANCER GENETICS GROUP

*tbc*

## Conference Office

### **British Society for Human Genetics**

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[www.clingensoc.org](http://www.clingensoc.org)

[www.ukcgg.org](http://www.ukcgg.org)



## Thursday 15 March 2012

### *Joint Programme*

- 07:30            **REGISTRATION** – Centre for Life
- 09:50            **WELCOME**
- 10:00-11:45    **SYMPOSIUM** “Emerging Therapies for Genetic Disorders”  
*Chairs: Dr Bill Newman (Manchester)/ Dr Grazia Mancini (Rotterdam)*
- 10:00            Introduction “Therapy for Clinical Geneticists” - *Prof Sir John Burn (Newcastle)*
- 10:15            “Atypical Haemolytic Uraemic Syndrome” - *Prof Tim Goodship (Newcastle)*
- 10:40            “Targeted therapies in neurofibromatosis: successful treatment of NF2 related schwannomas with bevacizumab in the national NF2 service”  
- *Prof Gareth Evans (Manchester)*
- 11:05            “The immune-biology of DNA mismatch repair deficient tumour cells: On the road towards a preventive vaccine” - *Prof Dr Magnus von Knebel Doeberitz (Heidelberg, Germany)*
- 11:45-12:15    **Submitted ABSTRACTS**  
*Chairs: Dr Bill Newman (Manchester)/ Dr Grazia Mancini (Rotterdam)*
- 12:15            **CGS Annual General Meeting**
- 12:45            **Lunch**        Sponsored by NewGene Ltd.
- 13:45-15:00    **SYMPOSIUM** “Epigenetics”  
*Chairs: Prof Sir John Burn (Newcastle)/ tbc*
- 13:45            “The epigenome as an integrator of environmental and germline perturbation”  
- *Dr Caroline Relton (Newcastle)*
- 14:10            “Genetic-based epigenetics: unusual cases of familial cancer”  
- *Dr Megan Hitchins (Sydney) Sponsored by QuantuMDx*
- 14:35            “Epigenetic modification of chromatin structure in intellectual disability”  
- *Dr Tjitske Kleefstra – (Nijmegen)*
- 15:00-15:30    **Dutch and UK SpR presentations / Robin Winter prize**  
*Chairs: Dr Sarah Smithson (Bristol)/ Prof Nine Knoers (Utrecht)*
- 15:30            **Tea**
- 16:00-17:00    **Dutch and UK SpR presentations / Robin Winter prize (continued)**  
*Chairs: Dr Sarah Smithson (Bristol)/ Prof Nine Knoers (Utrecht)*
- 17:00-17:45    **Invited Talk** “The Founders of Medical Genetics” by *Prof Peter Harper (Cardiff)*  
*Chairs: Prof Angus Clarke (Cardiff)/ Dr Connie Schrandt-Stumpel (Maastricht)*
- 19:30-Late     **Conference Dinner at the Hilton Hotel, Newcastle Gateshead, NE8 2AR**  
**Robin Winter Prize Giving Ceremony**  
Sponsored by the NIHR Collaborative Group for Genetics in Healthcare



## Friday 16 March 2012

### *Clinical Genetics Programme*

- 07:30            **REGISTRATION** – Centre for Life
- 08.30-09.00    **Sponsored Talk** “tbc”  
*Sponsored by Novartis Pharmaceuticals UK Ltd.*
- 09:00-10:45    **SYMPOSIUM** “Neurogenetics and Mitochondrial Disorders”  
*Chairs: Prof Volker Straub (Newcastle)/ Prof Dr Martijn Breuning (Leiden)*
- 09:00            “Antisense Oligonucleotide treatment for genetic diseases”  
- Prof Dr Gertjan van Ommen (Leiden)
- 09:30            “New insights into the genetic and epigenetic concepts of FSHD”  
- Prof Dr Silvere van der Maarel (The Netherlands)
- 10:00            “Mitochondrial encephalomyopathies: diagnosis, disease mechanisms and possible therapies; Part I” - Dr Rita Horvath (Newcastle)
- 10:30            “Mitochondrial encephalomyopathies: diagnosis, disease mechanisms and possible therapies; Part II”- Dr Eva Morava (Nijmegen)
- 11:00            **Coffee**
- 11:30-12:45    **Presentation by CGS INTERNATIONAL FELLOW**  
**Submitted ABSTRACTS**  
*Chairs: Dr Peter Turnpenny (Exeter)/ Dr Ineke van der Burgt (Nijmegen)*
- 12:45            **Lunch**
- 13:45-15:00    **SYMPOSIUM** “Teratology”  
*Chairs: Dr Laura Yates (Newcastle)/ Prof Dick Lindhout (Utrecht)*
- 13:45            Introduction “Teratology in the genetics clinic” - Dr Laura Yates (Newcastle)
- 14:00            “Fetal Alcohol Syndrome in The Netherlands” - Dr Jan Cobben (Amsterdam)
- 14:25            “Growing prevalence of diabetes: implications for the next generation”  
- Prof Judith Goodship (Newcastle)
- 14:50            “Antiepileptic Drugs and Teratogenicity” - Prof Jill Clayton-Smith (Manchester)
- 15:15-16:00    **Submitted ABSTRACTS**  
*Chairs: Dr Laura Yates (Newcastle)/ Prof Dick Lindhout (Utrecht)*
- 16:00            **Tea**
- 16:30            **END of CONFERENCE**



## Friday 16 March 2012

### *Cancer Genetics Programme*

- 07:30           **REGISTRATION** – Centre for Life
- 07:30-08:45    **Steering Group Meeting** - Room tbc
- 08.30-09.00    **Sponsored Talk** “tbc”  
*Sponsored by Novartis Pharmaceuticals UK Ltd.*
- 09:00-11:30    **SYMPOSIUM** “Hereditary predisposition to Renal/pancreatic tumours”  
*Chairs: Prof Ros Eeles (London)/ Dr Andrew Cuthbert (Birmingham)*
- 09:00           “Birt Hogg Dube/ hereditary leiomyomatosis and renal cell carcinomas (BHD/HLRCC)”  
- *Dr Fred Menko (Amsterdam)*
- 09:30           “Paraganglioma/Von Hippel Lindau – management” - *Dr Steven Ball (Newcastle)*
- 10:00           “Pancreatic Neuroectodermal tumours” - *Dr Juan Valle (Manchester)*
- 10:30           “Genetics of Pancreatic Cancer” - *Dr Marc Tischkowitz (Cambridge)*
- 11:00           “Imaging of the Kidney and Pancreas” - *Dr Maria Sheridan (Leeds)*
- 11:30           **Coffee**
- 11:50-12:50    **DISCUSSION** “Re: Guidelines Screening/Management”  
*Chairs: Dr Lucy Side (London)/ Dr Alan Donaldson (Bristol)*
- Short summaries of current consensus (if any) from UK and Dutch presenters
- a) Renal Carcinoma Guidelines; *Dr Paul Brennan (UK) and Dr Fred Menko (NL)*
- b) Paraganglioma Guidelines; *Dr Fiona Laloo (UK) and Dr Fred Hes (NL)*
- c) Pancreatic Guidelines; *tbc (UK) and Dr Irma Kluijt (NL)*
- 12:50           **Lunch**
- 13:50-14:50    **DIFFICULT CASES open Discussion**  
Please bring difficult case management examples or issues to discuss  
*Chairs: Prof Anneke Lucassen (Southampton)/ Dr Fiona Laloo (Manchester)*
- 15:15-17:00    **Submitted ABSTRACTS**  
*Chairs: Ms Chris Jacobs (London)/ Prof Anneke Lucassen (Southampton)*
- 16:45           **Closing Comments**
- 17:00           **END of CONFERENCE**