



# CLINICAL GENETICS SOCIETY

## Spring Conference 2006

Thursday and Friday 23<sup>rd</sup> - 24<sup>th</sup> March

at the **Boyd Orr Building**, University of Glasgow

### SCIENTIFIC PROGRAMME

The programme of spoken presentations is outlined later in this leaflet. The Scientific Presentations are being held in the Boyd Orr Lecture Theatre 1. There are NO posters at this meeting.

### GENERAL INFORMATION

This year the Clinical Genetics Society spring conference has moved to Glasgow for a two day meeting together with a conference dinner and ceilidh on the Thursday evening.

**Venues:** The conference is being held at the Boyd Orr Building, which is located on University Avenue. The conference dinner is being held at the Winter Gardens, Glasgow Green.

**Emergency Messages:** 0141 330 5301 Emergency messages during the day, can be left with the University Janitors reception – they will then pass these on to the Conference Registration Desk.

**Registration:** please complete and return your Registration Form to the Conference Office, together with your payment by Monday 27<sup>th</sup> February 2006. Your conference pack will be sent to you approximately two weeks in advance of the conference.

**Abstract Booklet:** members not able to attend the meeting may order a copy of the Abstract Booklet from the BSHG office (price £3.50; cheques made payable to "Clinical Genetics Society" please) before 27<sup>th</sup> February 2006.

**Continuing Professional Development:** this meeting is recognised for Continuing Professional Development.

**Car Parking:** Car Parking is very limited around the University. There is off road parking and pay and display car parks on Byres Road.

**Accommodation:** A separate sheet giving information on hotel accommodation and a map showing location of hotels and the University of Glasgow, is enclosed.

**Conference Dinner:** The Conference Dinner is being held at the Peoples Palace, Winter Gardens, Glasgow Green. Coaches will leave at 19:00 (from two pick up points – Hotel Grosvenor Hotel No 3 on map and Novotel No 11 on map). The evening will commence with a drinks reception starting at 19:30 in the "Garden Area". A four course dinner with wine and coffee will be served at 20:00 followed by a ceilidh. A cash bar will be open from 7.30-midnight. Coaches will leave at midnight and 00:30 and both coaches will go to both of the pick up points.

### ANNUAL GENERAL MEETING 2006

To be held at 10:15 on Friday 24<sup>th</sup> March in the Boyd Orr Lecture Theatre 1

**Information, Programme and Registration Form**

# PROGRAMME - 23<sup>rd</sup> March 2006

10:00 Registration and Coffee

## Symposium: Genetics and Dermatology

- 11:00 (1) **Overview of the genodermatoses**- Celia Moss  
 11:25 (2) **Cutaneous Mosaicism: patterns and mechanisms** - Rudolph Happle  
 11:50 (3) **tbc** - Irwin McLean  
 12:15 COFFEE

## Submitted Abstracts

- 12:30 (4) **Penetrance data for Huntington's disease results in the 36-39 CAG repeat range: a multicentre direct observational study.** – Oliver Quarrell  
 12:40 (5) **NICE if you have the cash: funding the NICE breast cancer family history recommendations** – Paul Brennan  
 12:50 (6) **SDHB founder mutation in the West of Scotland** – Nicola Bradshaw  
 13:00 (7) **Inherited association of breast and colorectal cancer: limited role of CHEK2 compared to high-penetrance genes** – Gareth Evans  
 13:10 LUNCH

## Symposium: Human Cloning

- 14:10 (8) **Developmental competence of human in vitro aged oocytes as host cells for human somatic cell nuclear transfer** - Vanessa Hall  
 14:55 (9) **Stem Cells - media hype or miracle cure** - Alison Murdoch  
 15:40 TEA

## Submitted abstracts - Competing for the CGS Prize

- 16:10 (10) **PTEN related disorders: a national study** - Katherine Lachlan  
 16:20 (11) **Mapping autosomal dominant progressive limb-girdle myopathy with bone fragility to chromosome 9p21-p22: A novel locus for a musculoskeletal syndrome** – Sarju Mehta  
 16:30 (12) **A mutation creating a functional upstream initiation codon in the SOX9 5' UTR causes campomelic dysplasia** – Diana Johnson  
 16:40 (13) **Interruptions and structural rearrangements in myotonic dystrophy repeat expansions: unusual genotypes with classical phenotypes** – Jenny Thompson  
 16:50 (14) **A series of 7 Oxford patients with duplication of 22q11.2. The Oxford contribution to our understanding of an emerging phenotype** –Duncan Rourke

## Conference Dinner and Ceilidh – Peoples Palace, Winter Gardens, Glasgow Green

- 19:30 DRINKS RECEPTION  
 20:00 CONFERENCE DINNER AND CEILIDH



# PROGRAMME - 24<sup>th</sup> March 2006

## Symposium: Phenotypic Variability in Single Gene Disorders

- 09:00 (15) **Mutations of the fibroblast growth factor receptors: a rich tapestry of mechanisms and phenotypes**- Andrew Wilkie
- 09:25 (16) **“Splitting” or “lumping” of retinal dystrophies?** - Graeme Black
- 09:50 (17) **PTEN related disorders – many names, one condition** - Karen Temple
- 10:15 AGM
- 10:45 COFFEE

## Submitted Abstracts

- 11:15 (18) **The transmembrane protein meckelin (MKS3) is mutated in Meckel-Gruber syndrome**  
– Colin Johnson
- 11:25 (19) **Recurrence of Fanconi Anaemia in male siblings: confirmation of X-linked inheritance**  
–Ruth Newbury-Ecob
- 11:35 (20) **Cardiomyopathy as a cause of death in carriers of Duchenne and Becker muscular dystrophy**  
– Susan Holloway
- 11:45 (21) **Implementing the linked newborn and antenatal Sickle Cell and Thalassaemia population screening programme in England** - Allison Streetly
- 11:55 (22) **IT requirement for clinical genetics services: a national consensus** – Greta Westwood
- 12:05 LUNCH

## Symposium: Cancer Genetics – 10 years on

- 13:15 (23) **The evolution of clinical cancer genetics** - Diana Eccles
- 14:00 (24) **A clinical update of HNPCC/Lynch syndrome** - Hans Vasen
- 14:45 TEA

## Submitted Abstracts

- 15:15 (25) **Lowe (Oculo-cerebro-renal) syndrome: Lessons learned from 3 new cases** – Kathryn Leask
- 15:25 (26) **Aplasia cutis congenita, transverse terminal limb defects and falciform retinal non-attachment: Further evidence of a distinct syndrome of vascular malformation** – Joanna Prothero
- 15:35 (27) **Ligneous Conjunctivitis: a distressing disorder caused by defective fibrinolysis**  
– Jenny Thompson
- 15:45 (28) **Age-related macular degeneration, smoking and the complement factor H variant Y402H**  
– John Yates
- 15:55 FINISH



Programme

**CLINICAL GENETICS SOCIETY – SPRING CONFERENCE – 23<sup>rd</sup> - 24<sup>th</sup> March 2006**  
 Boyd Orr Building, University of Glasgow

# REGISTRATION FORM

Closing Date for Receipt of Registration Forms in Birmingham

## Monday 27<sup>th</sup> February 2006

Surname ..... Forenames ..... Title ..... M/F .....

Address .....

Daytime Telephone ..... Email .....

My name and place of work should appear on my badge as .....

I am the presenter of the spoken presentation shown as number ..... in the enclosed programme.

	BSHG member	Guest/Non-member	Total
<b>REGISTRATION – CGS CONFERENCE</b> <i>(includes abstract booklet, sandwich lunch, tea and coffee)</i>			
Thursday 23rd March 2006			
Registration received before 27th February	£35.00	£45.00	
Registration received after 27th February	£45.00	£55.00	
Friday 24th March 2006			
Registration received before 27th February	£35.00	£45.00	
Registration received after 27th February	£45.00	£55.00	
<b>CONFERENCE DINNER – 23rd March 2006 at</b> Peoples Palace, Winter Gardens, Glasgow Green <i>(including drinks reception, four course dinner with wine and ceilidh)</i>		£40.00	
Dietary requirements .....			
<b>TOTAL</b>			<b>£</b>

## PAYMENTS

I enclose a cheque payable to the CLINICAL GENETICS SOCIETY for £.....

Please return this form to the Conference Office at the address below - don't forget to keep a photocopy for your records!

Your delegates pack will be sent to you two weeks before the conference. Please note that invoices CANNOT be supplied in respect of registration fees.

*We regret that fees CANNOT be reimbursed if you have to cancel your registration after 27th February 2006*

**PLEASE RETURN YOUR FORM TO THE CONFERENCE OFFICE:**

Clinical Genetics Society, Clinical Genetics Unit, Birmingham Women's Hospital, Edgbaston, Birmingham. B15 2TG  
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 Registered Charity No: 274959