THE EUROPEAN SOCIETY
OF HUMAN GENETICS

EUROPEAN HUMAN GENETICS CONFERENCE 2015

joint with the
British Society of Genetic Medicine

June 6-9, Glasgow, Scotland, United Kingdom

Final Programme

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GENERAL WELCOMING ADDRESS

Dear Colleagues and Friends,

The last time the ESHG was held in the UK was in 2003, in Birmingham. I don't believe it has ever been held in Scotland and a great deal has changed in genetics since 2003, so it gives me the greatest of pleasure, on behalf of the British Society for Genetic Medicine (BSGM) to welcome you to Glasgow, for the 2015 Conference.

Glasgow is Scotland's largest city and is renowned for its culture, style and the friendliness of its people. Glasgow offers a blend of internationally-acclaimed museums and galleries, stunning architecture, vibrant nightlife, fantastic shopping and a diverse array of restaurants and bars. Vibrant and energetic, Glasgow enjoys a year-round buzz with an arts scene that regularly produces cutting-edge productions and attracts high-profile exhibitions that led to the city being crowned European City of Culture in 1990.

Glasgow is also notable for its great scientists. It was a Glaswegian Charles Macintosh who patented the invention for waterproof cloth in 1823 and the first Mackintosh coats were made in the family's textile factory, Charles Macintosh and Co. of Glasgow. Hopefully you won't need yours in June.

Other great Glaswegians include Joseph Lister, pioneer of antiseptic surgery and James Watt, the great engineer that the 'Watt' was named after, who started the industrial revolution, so Glasgow knows how to innovate.

The 2015 conference promises to be inspirational, just as always, and will provide the latest in the Genomics Revolution.

Welcome to Glasgow!

Angela Douglas

Local Host
President of the British Society of Genetic Medicine

GENERAL COMMITTEES - BOARD - ORGANISATION

European Society of Human Genetics

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European Human Genetics Conference 2015

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GENERAL ACKNOWLEDGEMENTS-FUTURE MEETINGS

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- AAAS/Science
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- Affymetrix
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- QIAGEN Bioinformatics
- Roche
- Sistemas Genómicos
- Sophia Genetics
- Source BioScience
- Swift Biosciences
- Thermo Fisher Scientific
- People Make Glasgow
- Visit Scotland

Future European Human Genetics Conferences

European Human Genetics Conference 2016
Barcelona, Spain
May 21 – 24, 2016

1967 – 2017: 50th Anniversary of the ESHG
The European Human Genetics Conference 2017
Copenhagen, Denmark
May 27 – 30, 2017

European Human Genetics Conference 2018
Milan, Italy
June 16 – 19, 2018

CME Credits

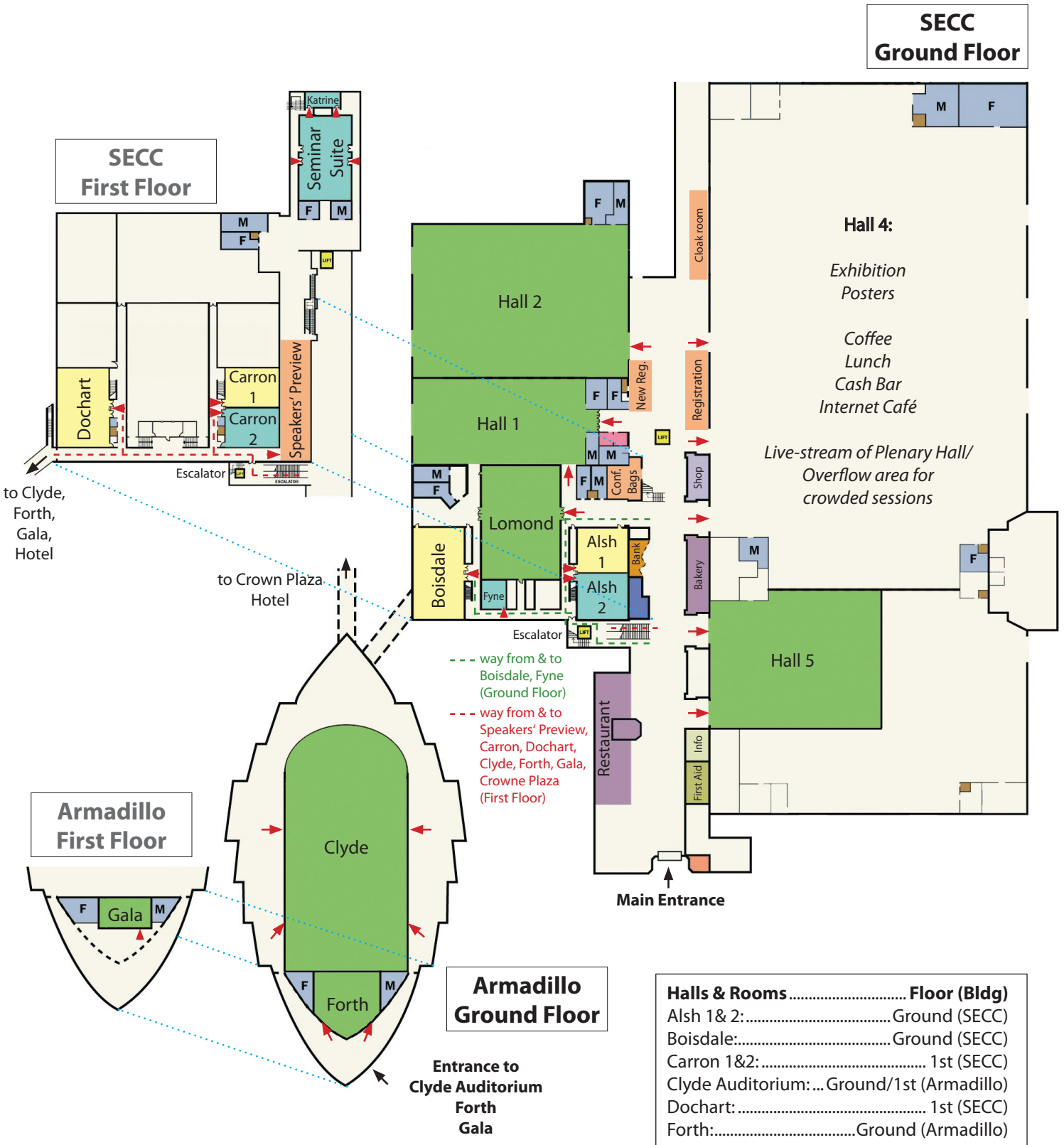
The European Society of Human Genetics is accredited by the European Accreditation Council for Continuing Medical Education (EACCME) to provide the following CME activity for medical specialists. The EACCME is an institution of the European Union of Medical Specialists (UEMS), www.uems.net.

The European Human Genetics Conference 2015 is designated for a maximum of **21 hours of European external CME credits**. Each medical specialist should claim only those hours of credit that he/she actually spent in the educational activity.

EACCME credits are recognized by the American Medical Association towards the Physician's Recognition Award (PRA). To convert EACCME it to AMA PRA category 1 credit, contact the AMA.

IMPORTANT NOTICE
Please note that taking pictures or filming during the sessions is forbidden (no matter if done with a camera or a mobile phone). Persons who will not observe this rule will be excluded from the session by the chairpersons.

GENERAL FLOORPLAN



Other Locations	Floor (Bldg)
Bank	Ground (SECC)
Cloak Room	Ground (SECC)
Conference Bags	Ground (SECC)
Speakers' Preview	1st (SECC)
Registration	Ground (SECC)
Restaurant	Ground (SECC)
Shops	Ground (SECC)

Lecture Halls

Corporate Satellites

Business Meetings

Halls & Rooms	Floor (Bldg)
Alsh 1& 2:	Ground (SECC)
Boisdale:	Ground (SECC)
Carron 1&2:	1st (SECC)
Clyde Auditorium:	Ground/1st (Armadillo)
Dochart:	1st (SECC)
Forth:	Ground (Armadillo)
Fyne:	Ground (SECC)
Gala:	1st (Armadillo)
Hall 1:	Ground (SECC)
Hall 2:	Ground (SECC)
Hall 4/Exhibition/Posters:	Ground (SECC)
Hall 5:	Ground (SECC)
Katrine:	1st (SECC)
Lomond:	Ground (SECC)
Seminar Suite:	1st (SECC)

GENERAL SESSION TYPE DESCRIPTIONS

Plenary Sessions (PL1 - PL5)

The plenary sessions are the most prestigious sessions of the congress. These are exhaustive reviews of major subjects and state of the art techniques within the specialty, addressed to all participants. Speakers in plenary sessions are invited and are among the most renowned in their field of expertise.
Plenary sessions are scheduled at “prime time” in the programme, unopposed to other activities in order to achieve maximal attendance. Speaking time varies: 15 minutes for talks in PL2, 30 minutes in PL1 & PL3, and 45 minutes in PL4 & PL5.

Concurrent Symposia (S01 – S16)

The symposia are sessions in which invited speakers share new results on a given topic with other researchers. The aim is to reflect and compare data with other, perhaps contradictory, results and to discuss new hypotheses and concepts for further research with well established colleagues.
In every concurrent symposium three 30-minute lectures will be presented. They provide an update and understanding of new developments and innovations in a certain area.

Educational Sessions (ES1 – ES9)

The Scientific Committee of the ESHG determines topics for these 90 minutes sessions which will best serve the *educational* needs of the attendees. Particular care is taken to ensure that these sessions address basic issues and focus on the educational aspect. These sessions are *not intended for experts* in the respective fields but are designed to give a general basic introduction to a particular topic.

Concurrent Sessions (C01 – C23)

The most notable and exciting work from all abstracts submitted to the conference will be honoured with an oral presentation in these sessions. Presenters are expected to explain their work and answer questions from the audience. Speaking time for concurrent session is 15 minutes including time for discussion. Papers marked with an asterisk are candidates for the ESHG Young Investigator Awards.

Poster Viewing with Authors

Posters are numerically the major scientific presentations of the meeting. Most attendees bring a poster showing data and progress with their personal research. Posters offer an excellent opportunity for people interested in a particular topic to meet and exchange ideas and network with other researchers. Posters should NOT be used to advertise a product or service. Like a paper, a poster abstract should detail the focus of the presentation and the way(s) in which it contributes to the body of knowledge in its field.
Times marked “Poster Viewing with Authors” should be used for communication and interaction with the poster authors, who are requested to be at their posters at these times. Posters will be on display throughout the whole conference for free poster viewing (Saturday-Monday).
Posters bearing a rosette have received a high score during the peer review process and are considered the best posters submitted by young investigators. They are the candidates for the ESHG poster awards.

Workshops (WS01 – WS16)

Workshops are sessions in which the speakers are expected to share their personal experience in a field, either clinical or basic with the audience. These sessions are addressed to participants who wish to acquire practical knowledge on a specific subject, and therefore an interactive discussion during or at the end of the workshop is expected.

Corporate Satellites (CS01-CS23)

There are a number of company satellites planned within the main conference programme. Sponsors are approved as reputable and relevant by the Scientific Programme Committee, but the detailed content of the presentations is proposed directly by the sponsors and under their responsibility. Neither the ESHG nor the organisers have endorsed the content in any way.

Saturday, June 6, 2015

Time	Clyde	Hall 5	Hall 2	Lomond	Hall 1	Forth	Boisdale	Alsh 1	Carron 1
10.00 – 10.30	*Coffee Break / Posters / Exhibition*								
10.30 – 12.00	**ES1** CRISPR-Cas9	**ES2** From Genes to Networks	**ES3** Translational Cancer Genetics	**ES4** Care for Rare Diseases	**WS1** A case that changed my life as a geneticists	**WS2** Galaxy			
12.15 – 13.45	*Lunch break / Posters / Exhibition*						**CS01** Complete Genomics Satellite	**CS02** Personalis Satellite	**CS03** Sistemas Genómicos Satellite
14.00 – 14.30	**Opening Welcome Addresses**								
14:30 – 16.00	**PL1** Opening Plenary Session								
16:00 – 16.30	*Vitamin break / Posters / Exhibition*								
16.30 – 18.00	**PL2** What's New? Highlights Session								
18.00 – 18.30	*Coffee break / Posters / Exhibition*								
18.30 – 20.00	**C01** NIPT	**C02** Improvement in genome sequencing and functional studies	**C03** Novel genes causing intellectual disability	**C04** The many faces of cancer mutations	**C05** Cardiovascular disorders	**C06** Neuromuscular disorders			
20.15 – 21.45	*Opening Networking Mixer at the Glasgow Science Centre*								

Session Types:

Plenary Session	Symposium	Concurrent Session	Workshop	Educational Session	Corporate Satellite

IMPORTANT NOTICE :
Please note that taking pictures or filming during the sessions is forbidden (no matter if done with a camera or a mobile phone). Persons who will not observe this rule will be excluded from the session by the chairpersons.

Sunday, June 7, 2015

Time	Clyde	Hall 5	Hall 2	Lomond	Hall 1	Forth	Gala	Boisdale	Dochart	Alsh 1	Carron 1
08.30 – 10.00	**S01** Big Data Genomics and Human Knock-outs	**ES5** Automating Clinical Genetics	**S02** Genetic testing in Children (Joint with the ASHG)	**S03** Epigenetic Basis of Disease	**S04** Spliceosome-opathies						
10.00 – 10.30	*Coffee break / Poster viewing / Exhibition*										
10.30 – 11.30	**Poster viewing with presenters** (poster numbers starting with "PS")										
11.45 – 13.15	**ES6** My vision on Genomic medicine	*Poster viewing / Lunch break / Exhibition*						**CS04** Thermo Fisher Scientific Satellite	**CS05** Affymetrix Satellite	**CS06** Sophia Genetics Satellite	**CS07** QIAGEN Satellite
13.30 – 15.00	**C07** Reproductive Genetics	**C08** Integrative OMICS approaches in common traits	**C09** Genetic susceptibility to cancer development	**C10** Neurogenetic disorders	**C11** Skeletal disorders	**C12** Sensory disorders					
15.00 – 15.30	*Vitamin break / Poster viewing / Exhibition*										
15.30 – 17.00	**WS03** NGS in clinics	**WS04** Dysmorpho-logy 1	**WS05** Pre-conception carrier testing	**WS06** Blurred boundaries between clinic and research	**WS07** EBMG: What can we do to facilitate you to become a registered genetic professional?	**WS08** Ensembl Highlights: What's New in Accessing our Genomes?	**WS09** Global Alliance for Genomics and Health	**CS08** Complete Genomics Satellite	**CS09** AstraZeneca Satellite	**CS10** Multiplicom Satellite	**CS11** NuGEN Technologies Satellite
17.00 – 17.30	*Coffee break / Poster viewing / Exhibition*										
17.30 – 19.00	**S05** Reproductive genetics and "Chromosome therapy"	**S06** International data sharing initiatives	**ES7** Imprinting-related Disorders	**S07** Mouse Phenotyping for clinical research	**S08** Telomeres in Human Disease						
19.00 – 20.30							**ESHG Membership Meeting**	**CS12** Illumina Satellite	**CS13** Centogene Satellite	**CS14** Natera Satellite	**CS15** Thermo Fisher Scientific Satellite

Session Types

Plenary Session	Symposium	Concurrent Session	Workshop	Educational Session	Corporate Satellite

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Monday, June 8, 2015

Time	Clyde	Hall 5	Hall 2	Lomond	Hall 1	Forth	Gala	Boisdale	Dochart	Alsh 1	Carron 1
08.30 – 10.00	**S09** Evolution of the cancer genome: Clinical implications	**S10** From rare to common variants in cardiovascular diseases (joint with ESC)	**S11** Non-coding DNA and human disease	**S12** Mitochondria and Genetic Disease	**ES8** Palliative Care of Genetic Conditions						
10.00 – 10.30	*Coffee break / Poster viewing / Exhibition*										
10.30 – 11.30	**Poster viewing with presenters** (poster numbers starting with "PM")										
11.45 – 13.15	*Poster viewing / Lunch break / Exhibition*							**CS16** Agilent Technologies Satellite	**CS17** Oxford Gene Technology Satellite	**CS18** Roche Sequencing Satellite	**CS19** LGC Satellite
13.30 – 15.00	**C13** Fundamental insights in structural genomics	**C14** Challenges in genetic counselling	**C15** Network and functional analysis in intellectual disability	**C16** Growth failure and microcephaly	**C17** Epigenetic control of gene expression	**C18** Metabolic and renal disorders					
15.00 – 15.30	*Vitamin break / **Poster removal** / Exhibition*										
15.30 – 17.00	**WS10** Practical Bioinformatics Whole exome sequence analysis	**WS11** Dysmorphology 2	**WS12** Reproductive genetics	**WS13** Clinical Cancer Genetics	**WS14** Copy Number Variant Interpretation and Classification	**WS15** Genome Browser UCSC	**WS16** The genetics clinic of the future	**CS20** Thermo Fisher Scientific Satellite	**CS21** Cartagenia Satellite	**CS22** QIAGEN Bioinformatics Satellite	**CS23** EMQN Satellite
17.00 – 17.30	*Coffee break / **Poster removal** / Exhibition*										
17.30 – 19.00	**S13** Therapeutic Strategies for Genetic Diseases	**ES9** Mutation Prediction Tools	**S14** Genome Regulation	**S15** Somatic Mutation Detection and Interpretation	**S16** Evolution and Disease						
19.30	*Networking Party at the Merchant Square at own expense*										

Session Types:

Plenary Session	Symposium	Concurrent Session	Workshop	Educational Session	Corporate Satellite

IMPORTANT NOTICE :
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Tuesday, June 9, 2015

Time	Clyde	Hall 2	Lomond	Hall 1	Forth
09.00 – 10.30	**PL3** **Interactive Debate: Should all geneticists have their genome sequenced?**				
10.30 – 11.00	*Coffee break in Hall 5*				
11.00 – 12.30	**C19** **Diagnostic NGS**	**C20** **Current issues in genet(h)ics**	**C21** **Multiple congenital anomaly syndromes**	**C22** **Statistical genetics and bioinformatics**	**C23** **Movement and motor disorders**
12.30 – 13.30	*Lunch break in Hall 5*				
13.30 – 14.15	**PL4** **Mendel Lecture The neurexin enigma - from synapse formation toschizophrenia**				
14.15 – 15.45	**PL5** **Closing Plenary** **ESHG Award Lecture** **- ESHG Education Award** **- EJHG-NPG Awards** **- Young Investigator & Poster Awards** **- Closing**				

Session Types:

Plenary Session	Symposium	Concurrent Session	Workshop	Educational Session

IMPORTANT NOTICE :
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PROGRAMME POSTER TOPICS-TECHNICAL INFORMATION

Poster Topics

P01 Reproductive Genetics/Prenatal Genetics ..... P01.01 - P01.91
P02 Sensory disorders (eye, ear, pain) ..... P02.01 - P02.62
P03 Internal organs & endocrinology (lung, kidney, liver, gastrointestinal) ..... P03.01 - P03.41
P04 Skeletal, connective tissue, ectodermal and skin disorders ..... P04.01 - P04.79
P05 Cardiovascular disorders ..... P05.01 - P05.85
P06 Metabolic and mitochondrial disorders ..... P06.01 - P06.70
P07 Immunology and hematopoetic system ..... P07.01 - P07.25
P08 Intellectual Disability ..... P08.01 - P08.73
P09 Neurogenetic and psychiatric disorders ..... P09.001 - P09.138
P10 Neuromuscular disorders ..... P10.01 - P10.40
P11 Multiple Malformation/anomalies syndromes ..... P11.001 - P11.139
P12 Cancer genetics ..... P12.001 - P12.148
P13 Basic mechanisms in molecular and cytogenetics ..... P13.01 - P13.41
P14 New diagnostic approaches, technical aspects & quality control ..... P14.001 - P14.109
P15 Personalized/Predictive Medicine and Pharmacogenomics ..... P15.01 - P15.37
P16 Omics/Bioinformatics ..... P16.01 - P16.63
P17 Epigenetics and Gene Regulation ..... P17.01 - P17.37
P18 Genetic epidemiology/Population genetics/Statistical methodology and evolutionary genetics ..... P18.01 - P18.96
P19 Genetic counselling/Education/public services ..... P19.01 - P19.59
P20 Psychological/Ethical/legal issues ..... P20.01 - P20.28

Technical Information for Presenters of Posters

Posters will be on **display** from	Saturday, June 6, (09.30 hrs) to Monday, June 8 (17.30 hrs)
Poster mounting will be possible on:	Saturday, June 6, from 09.30 hrs onwards
Removal will be mandatory on:	Monday, June 8, from 13.30 hrs - 17.30 hrs (strict)

Access after Monday, 17:30 hrs is not possible! Please note that posters not removed until this time will be taken down by the staff of the conference centre and will not be stored or sent to the authors after the meeting.
You can find your poster board number in the author index of the Poster Listing available at the “poster help desk” or you can ask for assistance at the “poster help desk” located at the entrance “4A” to Hall 4 or at the two information points located in the exhibition / poster area.

Presence at Posters
In order to enable discussion and interaction with other participants, it is mandatory for you or one of your group to be at your poster board between:
• 10.30 and 11.30 hrs on Sunday, June 7 for posters with poster board numbers starting with “PS” (e.g. PS03.01, PS04.03)
or
• 10.30 and 11.30 hrs on Monday, June 8 for posters with poster board numbers starting with “PM” (e.g. PM07.02, PM08.04)

If it is not possible for you or one of your group to be present during the above stated times, please leave a note on your poster board detailing the times when you will be present at the board.

Technical Information for Presenters of Talks

- All rooms will be equipped with data projection.
- It is essential that you load and view your presentation in the Speakers’ Preview/Media check (**1st Floor**) not later than 2 hours in advance (30 minutes for the first morning talks).
- The lecture rooms are exclusively equipped with Windows-PCs (no MACs). In case you absolutely need to use your own laptop or notebook, please contact the Speakers’ Preview well in advance of your talk to check compatibility.
- Please bring a USB-key or CD-ROM all formatted for Windows® (PC). You may want to carry a second key/CD as a back-up in case there is any insoluble technical problem.
- File Format: Microsoft® Power Point 2007™ presentation formatted for Windows® (PC) only. (Operating system: Windows 7®)
- Preferred Resolution: XGA (1024 x 768 pixel)

SCIENTIFIC

SCIENTIFIC PROGRAMME

Saturday, June 6, 2015

PROGRAMME

PROGRAMME SATURDAY, JUNE 6

Time	Clyde	Hall 5	Hall 2	Lomond	Hall 1	Forth
10.30 - 12.00	**ES1 CRISPR-Cas9** Chair: D. Grinberg, M. Bailey	**ES2 From genes to networks** Chair: A. Fry, K. van Steen	**ES3 Translational cancer genetics** Chair: E. Woodward, A. Dalton	**ES4 Care for rare diseases** Chair: F. Palau, H. Burton	**WS01. A case that changed my life as a geneticist (TEDEx format)** Organisers: H. Brunner; M. Macek Jr.	**WS02. NGS Variant analysis with Galaxy** Organiser: D. Clements
10.30	**ES1.1** **Applications of CRISPR-Cas9 for Genome Engineering** **Le Cong**; *Cambridge, MA, United States*	**ES2.1** **Leveraging molecular networks to reveal pathways underlying complex diseases** **Daniel Marbach**; *Lausanne, Switzerland*	**ES3.1** **Inherited and acquired kidney cancers: opportunities for targeted therapeutic approaches** **Mariaflavia F. Di Renzo**; *Candiolo, Italy*	**ES4.1** **Patient perspective's to rare diseases** **Yann Le Cam**; *Paris, France*	For the First time, the ESHG conference offers a session dedicated to storytelling in Medical Genetics. We have invited professionals in medical genetics to submit a short video in which they describe an event or moment when genetics made a difference and had a profound impact on how they see the field of Medical Genetics. These stories can be happy, sad, informative or funny. The only requirement was that they are real experiences of yourself or your colleagues.	Galaxy is a free and open source data integration and analysis platform for life sciences research (http://www.galaxyproject.org). This workshop will briefly introduce the Galaxy platform and then walk through a live multi-step variant calling analysis using human data and Galaxy's rich tool set. The analysis will highlight Galaxy's collaboration, publishing, reproducibility, and visualisation features. Previous experience using Galaxy is helpful, but not required. All workflows and analyses from the workshop will be made publicly available at https://www.usegalaxy.org
11.15	**ES1.2** **CRISPR-Cas9: biological roles, mechanisms, evolution and applications** **Emmanuelle Charpentier**; *Braunschweig, Germany*	**ES2.2** **Gene co-expression networks** **Luis Serrano**; *Barcelona, Spain*	**ES3.2** **From inherited breast/ovarian cancer to PARP inhibitors and beyond** **William Foulkes**; *Montreal, Canada*	**ES4.2** **European rare disease policies- what does it really mean for planning services?** **Kate Bushby**; *Newcastle upon Tyne, United Kingdom*		
12.00 - 14.00	*Lunch break / Posters / Exhibition*					

Detailed Workshop programmes (when submitted by the organisers) can be found in the "ESHG Bulletin" in the conference bag.

PROGRAMME SATURDAY, JUNE 6

Time	Clyde
14.00 - 14.30	**Opening & Welcoming Addresses** Chair: A. Douglas, H. Kääriäinen
	Welcoming Addresses by Helena Kääriäinen *President of the ESHG* Angela Douglas *President of the British Society of Genetic Medicine (BSGM), Local host* Bailie Nina Baker *Representative of the Lord Provost of Glasgow*
14.30 - 16.00	**Opening Plenary Session PL1** Chair: A. Douglas, H. Kääriäinen
14.30	**PL1.1** **Chromosome conformation and long-distance gene regulation** *N. Benabdallah, S. Bhatia, I. Williamson,* **Wendy Bickmore***;* *Edinburgh, United Kingdom*
15.00	**PL1.2** **Deciphering Developmental Disorders** **Matthew Hurles***;* *Cambridge, United Kingdom*
15.30	**PL1.3** **Ribonucleotides embedded in genomic DNA** **Andrew Jackson***;* *Edinburgh, United Kingdom*
16.00 - 16.30	*Vitamin break / Posters / Exhibition*
16.30 - 18.00	**Plenary Session PL2. Highlights - What's new?** Chair: H. Kääriäinen, B. Wirth
16.30	**PL2.1** **De novo mutations in *PLXND1* and *REV3L* cause Möbius syndrome** **Laura Tomas Roca***, A. Tsaalbi-Shtylik, J.G. Jansen, M.K. Singh, J.A. Epstein, U. Altunoglu, H. Verzijl, L. Soria, E. van Beusekom, T. Roscioli, Z. Iqbal, C. Gilissen, A. Hoischen, A.P.M. de Brouwer, C. Erasmus, D. Schubert, H. Brunner, A. Pérez Aytés, F. Marin, P. Aroca Tejedor, H. Kayserili, A. Carta, N. de Wind, G.W. Padberg, H. van Bokhoven;* *Nijmegen, Netherlands*
16.45	**PL2.2** **Beyond the ACMG 56: Parental choices and initial results from a comprehensive WGS-based search for predictive secondary variants in children** **M Stephen Meyn**, *N. Monfared, C. Marshall, D. Merico, D.J. Stavropoulos, R.Z. Hayeems, M. Szego, R. Jobling, M. Gardia, G.D. Bader, M. Brudno, R.D. Cohn, R. Zlotnik-Shaul, C. Shuman, P.N. Ray, S. Bowdin;* *Toronto, Canada*
17.00	**PL2.3** **Spotlight on the pathogenesis of Kabuki syndrome** *N. Bögershausen, I. Tsai, E. Pohl, P. Simsek Kiper, F. Beleggia, F.E. Percin, K. Keupp, A. Matchan, E. Milz, Y. Alanay, H. Kayserili, Y. Liu, S. Banka, A. Kranz, M. Zenker, D. Wieczorek, N. Elcioglu, P. Prontera, S. Lyonnet, T. Meitinger, F. Stewart, D. Donnai, T.M. Strom, K. Boduroglu, G. Yigit, Y. Li, N. Katsanis,* **Bernd Wollnik***;* *Cologne, Germany*
17.15	**PL2.4** **Disruptions of topological chromatin domains cause pathogenic rewiring of gene-enhancer interactions** **Darío G. Lupiáñez***, K. Kraft, V. Heinrich, P. Krawitz, F. Brancati, E. Klopocki, D. Horn, H. Kayserili, J. Opitz, R. Laxova, F. Santos-Simarro, B. Gilbert-Dussardier8, L. Wittler, M. Borschiwer, S. Haas, M. Osterwalder, M. Franke, B. Timmermann, J. Hecht, M. Spielmann, A. Visel, S. Mundlos;* *Berlin, Germany*
17.30	**PL2.5** **A germline homozygous loss-of-function mutation in the base excision repair gene NTHL1 causes adenomatous polyposis and colorectal cancer** **Robbert D.A. Weren***, M.J.L. Ligtenberg, C.M. Kets, R.M. de Voer, E.T.P. Verwiel, L. Spruijt, W.A.G. van Zelst-Stams, M.C. Jongmans, C. Gilissen, J.Y. Hehir-Kwa, A. Hoischen, J. Shendure, E.A. Boyle, E.J. Kamping, I.D. Nagtegaal, B.B.J. Tops, F.M. Nagengast, A. Geurts van Kessel, J.H.J.M. van Krieken, R.P. Kuiper, N. Hoogerbrugge;* *Nijmegen, Netherlands*
17.45	**PL2.6** **The genetic handicap principle: a severely deleterious mutation can be tolerated if the genome-wide mutation load is sufficiently low** **Konstantin Popadin**, *S. Peischl, R. Sailani, A. Letourneau, F. Santoni, M. Garieri, S. Nikolaev, D. Meyer, L. Excoffier, S. Antonarakis;* *Geneva, Switzerland*
18.00 - 18.30	*Coffee break / Poster / Exhibition*

Presentations highlighted by an asterisk (*) and a grey background are from Young Investigator Award Finalists.
City and country refer to the affiliation of the presenting author.

PROGRAMME SATURDAY, JUNE 6

Time	Clyde	Hall 5	Hall 2	Lomond	Hall 1	Forth
18.30 - 20.00	**C01 NIPT** Chair: D. Wellesley, H. Skirton	**C02 Improvement in genome sequencing and functional studies** Chair: H. Scheffer, M. Hurles	**C03 Novel genes causing intellectual disability** Chair: C. Wright, T.E. Prescott	**C04 The many faces of cancer mutations** Chair: J. Schmidtke, J. Adlard	**C05 Cardiovascular disorders** Chair: A.J. Clarke, E. Blair	**C06 Neuro-muscular disorders** Chair: B. Melegh, E. Reid
18.30	**C01.1** **Implementation of a non-invasive prenatal testing (NIPT) for aneuploidy service in an NHS diagnostic laboratory** **Lyn S. Chitty**, *F.J. McKay, S. Mason, C. Boustred, K. Lo, L. Jenkins, R. Daley, M. Hill, C. Lewis, S. Drury, J. Fisher, T. Verhoef, S. Morris; London, United Kingdom*	**C02.1** **Single cell analysis "simplification" dramatically increases complexity: considerations in technique, quality control, analysis, and possibilities for translation to the clinic** **Lisa D. White**, *J.D. Landua, L. Simon, M.T. Bernardi, D. Xavier, C.A. Shaw, M.T. Lewis; Houston, United States*	**C03.1** ***De novo* and familial *DDX3X* mutations are associated with X-linked intellectual disability and a diverse phenotypic spectrum** **Lot Snijders Blok***, *E. Madsen, M. Reijnders, H. Venselaar, C. Helsmoortel, C. Gillissen, A. Hoischen, L. Vissers, T. Koemans, W. Wissink, E.E. Eichler, C. Romano, H. Van Esch, C. Stumpel, M. Vreeburg, E. Smeets, B. van Bon, M. Shaw, J. Gecz, M. Bienek, C. Jensen, B. Loeys, A. van Dijck, A.M. Innes, N. Di Donato, S.G. Mehta, K. Tatton-Brown, D. Baralle, A. Henderson, S. Dijkstra, J. Schieving, S. Haas, H. Brunner, F. Kooy, C. van Roozendaal, R. Pfundt, V. Kalscheuer, N. Katsanis, T. Kleefstra; Nijmegen, Netherlands*	**C04.1** **Mosaic loss of chromosome Y (LOY) in peripheral blood is associated with smoking, shorter survival and increased risk of cancer** **Lars A. Forsberg**, *C. Rasi, M. Lönn, H. Davies, M. Ingelsson, V. Giedraitis, L. Lannfelt, N.N.C. Cross, D. Absher, P.K.E. Magnusson, C. Lindgren, A.P. Morris, D. Cesarini, M. Johannesson, E. Tiensuu, L. Lind, N.L. Pedersen, E. Ingelsson, J. Dumanski; Uppsala, Sweden*	**C05.1** **MFAP5 loss-of-function mutations underscore the involvement of matrix alteration in the pathogenesis of Familial Thoracic Aortic Aneurysms and Dissections** **Mathieu Barbier**, *M. Gross, M. Aubart, N. Hanna, K. Kessler, D. Guo, L. Tosolini, B. Ho-Tin-Noe, E. Regalado, M. Varret, M. Abifadel, O. Milleron, S. Odent, S. Dupuis-Girod, L. Faivre, T. Edouard, Y. Dulac, T. Busa, L. Gouya, D. Milewicz, G. Jondeau, C. Boileau; Paris, France*	**C06.1** **Neurogenetic disease diagnostics by targeted capture and next generation sequencing** **Nigel G. Laing**, *K. Yau, R. Allcock, R. Ong, K. Mina, G. Ravenscroft, M. Cabrera, R. Gooding, C. Wise, P. Sivadorai, D. Trajanoski, V. Atkinson, S. Wagner, K.J. Nowak, R.M. Duff, P.J. Lamont, M.R. Davis; Nedlands, Australia*
18.45	**C01.2** **TRIDENT: or monitored NIPT implementation in the Netherlands** **Erik A. Sistermans**, *G.H. Schuring-Blom, B.H.W. Faas, E.M.J. Boon, C.J. Bax, A.B.C. Coumans, A.T.J.I. Go, K. Huijsdens-van Amsterdam, M.V.E. Macville, D. van Opstal, E. Pajkrt, B. Sikkema-Raddatz, R.F. Suijkerbuijk, J.M.G. van Vugt, M.M. Weiss, G.C.M.L. Page-Christiaens, D. Oepkes; Amsterdam, Netherlands*	**C02.2** **Large-scale genotyping of polymorphic inversions in the human genome** **Sergi Villatoro***, *R. Zaurin, M. Gayà-Vidal, C. Giner-Delgado, D. Vicente-Salvador, D. Izquierdo, M. Oliva, L. Pantano, M. Puig, M. Cáceres; Bellaterra (Barcelona), Spain*	**C03.2** **De novo and recurrent PPP2R5D and PPP2R1A missense mutations cause protein phosphatase 2A dysfunction and intellectual disability** **Dorien Haesen***, *V. Janssens, L.E.L.M. Vissers, S. Mehta, M.J. Parker, M. Wright, J. Vogt, S. McKee, J.L. Tolmie, N. Cordeiro, T. Kleefstra, M.H. Willemsen, M.R.F. Reijnders, S. Berland, E. Hayman, E. Lahat, E.H. Brilstra, K.L.I. van Gassen, E. Zonneveld-Huijssoon, C.I. de Bie, A. Hoischen, E.E. Eichler, R. Holdhus, V.M. Steen, S.O. Døskeland, M.E. Hurles, D.R. FitzPatrick, T. DDD-study, G. Houge; Leuven, Belgium*	**C04.2** **SNP-SNP interaction analysis of NF-kB signaling pathway on breast cancer survival** **Maral Jamshidi***, *R. Fagerholm, S. Khan, K. Aittomäki, D.E. Easton, P. Hall, C. Blomqvist, M.K. Schmidt, H. Nevanlinna, B. C.A.C (Breast Cancer Association Consortium); Helsinki, Finland*	**C05.2** **Mutations in a TGFβ ligand, TGFB3, cause syndromic aortic aneurysms and dissections** **Elisabeth Gillis***, *A.M. Bertoli-Avella, H. Morisaki, J.M.A. Verhagen, E. Gallo, B.P.T. Kruithof, S. Laga, A.J. Doyle, G. Oswald, M. Lammens, C. Evers, K. Devriendt, M. Dumoulein, J. Timmermans, I. Rodrigus, G. Baynam, M. Kempers, J. Saenen, E.M. Van Craenenbroeck, K. Minatoya, R. Matsukawa, T. Tsukube, N. Kubo, M. Goumans, J.W. Roos-Hesselink, I.M.B.H. van de Laar, H.C. Dietz, L. Van Laer, T. Morisaki, M.W. Wessels, B.L. Loeys; Antwerp, Belgium*	**C06.2** **The SMCHD1 mutation spectrum in Facioscapulohumeral muscular dystrophy** **Marlinde L. van den Boogaard***, *R.J.L.F. Lemmers, P.J. van der Vliet, J. Balog, B. Bakker, S.J. Tapscott, S. Sacconi, R. Tawil, S.M. van der Maarel; Leiden, Netherlands*
19.00	**C01.3** **Non-invasive prenatal diagnosis; expansion from de novo to autosomal recessive disorders using congenital adrenal hyperplasia as an example** **Suzanne Drury**, *K. Lo, C. Boustred, F. McKay, S. Mason, P. Twiss, S. Edwards, M. Hill, C. Lewis, R. Daley, L. Jenkins, L. Chitty; London, United Kingdom*	**C02.3** **Large-scale single-molecule sequencing of tandem repeats on the human X chromosome** **Alena Zablotskaya***, *G. Peeters, W.I.M. Meert, K.J. Verstrepen, G. Froyen, J.R. Vermeesch; Leuven, Belgium*	**C03.3** **Mutations in genes encoding components of protein phosphatase 2A (PP2A) cause human overgrowth and intellectual disability** **Chey Loveday**, *K. Tatton-Brown, M. Clarke, I. Westwood, A. Renwick, E. Ruark, E. Ramsay, R. van Montfort, N. Rahman; London, United Kingdom*	**C04.3** **Towards understanding the genomic architecture of cancer genomes** **Ernest T. Lam***, *A.R. Hastie, M.B. Imielinski, C. Zhang, J. Wala, Z. Dzakula, H. Cao; San Diego, United States*	**C05.3** **Exome-chip meta-analysis identifies novel associations of coding variants with cardiac conduction in 62,251 adults of European descent from the Cohorts for Heart and Aging Research in Genomic Epidemiology (CHARGE) Consortium.** **Yalda Jamshidi**, *B.P. Prins, C. Liu, J. van Setten, L. Hall, F. Radmanesh, CHARGE Consortium Exome-Chip EKG Working Group; London, United Kingdom*	**C06.3** **Plastin 3, a human protective modifier is highly upregulated in iPSC-derived motoneurons in asymptomatic individuals and rescues spinal muscular atrophy in mice** *M. Peters, L. Heesen, S. Hosseini Barkooie, M. Peitz, A. Kaczmarek, E. Janzen, O. Brüstle,* **Brunhilde Wirth**; *Cologne, Germany*

Presentations highlighted by an asterisk (*) and a grey background are from Young Investigator Award Finalists.

PROGRAMME SATURDAY, JUNE 6

Time	Clyde	Hall 5	Hall 2	Lomond	Hall 1	Forth
cont.	**C01 NIPT**	**C02 Improvement in genome sequencing and functional studies**	**C03 Novel genes causing intellectual disability**	**C04 The many faces of cancer mutations**	**C05 Cardiovascular disorders**	**C06 Neuro-muscular disorders**
19.15	**C01.4** **Non-invasive prenatal diagnosis (NIPD) of Duchenne and Becker muscular dystrophies (DMD/BMD) by relative haplotype dosage** **Michael Parks***, *S. Court, S. Cleary, S. Clokie, J. Hewitt, D. Williams, T. Cole, F. MacDonald, M. Griffiths, S.K. Allen;* *Birmingham, United Kingdom*	**C02.4** **The value of long-read single molecule sequencing in diagnostics** *S.Y. Anvar, H. Buermans, R. Vossen, M. Liem,* **Monique Losekoot**, *T. van der Straaten, H. van der Klift, J. Wijnen, J. Swen, D. Peters, J.T. den Dunnen;* *Leiden, Netherlands*	**C03.4** **De novo mutations in BCL11A cause developmental delay: additional implications of the BAF SWI/SNF complex in intellectual disability and autism** **Cristina Dias***, *J.A. Hurst, S. Joss, S.E. Holder, G. Sánchez-Andrade, S.J. Sawiak, S. Lee, P. Liu, M.E. Hurles, D.D.D. Deciphering Developmental Disorders Study, D.W. Logan;* *Hinxton, Cambridge, United Kingdom*	**C04.4** **Molecular classification of diffuse cerebral gliomas using genome- and transcriptomewide profiling.** *M. Weller, R.G. Weber, E. Willscher, V. Riehmer, B. Hentschel, M. Kreuz, J. Felsberg,* **Ulrike Beyer**, *H. Wirth, K. Kaulich, J. Steinbach, C. Hartmann, D. Gramatzki, J. Schramm, M. Westphal, G. Schackert, M. Simon, T. Martens, J. Boström, C. Hagel, M. Sabel, D. Krex, J.C. Tonn, W. Wick, S. Noell, U. Schlegel, B. Radlwimmer, T. Pietsch, M. Loeffler, A. von Deimling, H. Binder, G. Reifenberger, German Glioma Network;* *Hannover, Germany*	**C05.4** **A genome-wide association study of nonsyndromic mitral valve prolapse and functional studies of risk loci provide insight into underlying biological mechanisms** **Nabila Bouatia-Naji**, *C. Dina, N. Tucker, R.A. Norris, D. Milan, S. Slaugenhaupt, R.A. Levine, J. Schott, A.A. Hagège, X. Jeunemaitre;* *Paris, France*	**C06.4** **Analysis of the Gdap1 knockout mice reveals calcium homeostasis and mitochondrial dynamics defects in the Charcot-Marie-Tooth disease pathogenesis** **Azahara Civera-Tregón***, *P. Juárez, M. Barneo-Muñoz, S. Fernández-Lizarbe, D. Pla-Martin, J. Zenker, C. Cuevas-Martín, M. Sánchez-Aragó, J. Forteza-Vila, J.M. Cuezva, R. Chrast, F. Palau;* *Valencia, Spain*
19.30	**C01.5** **Incidental findings of genome wide non-invasive fetal aneuploidy detection (NIPT): presymptomatic identification of maternal cancers** **Nathalie Brison***, *K. Van Den Bogaert, P. Brady, L. Dehaspe, I. Wlodarska, F. Amant, P. Vandenberghe, T. de Ravel, H. Peeters, H. Van Esch, K. Devriendt, E. Legius, J.R. Vermeesch;* *Leuven, Belgium*	**C02.5** **Comparison of exome and genome sequencing technologies for the complete capture of protein coding regions** **Stefan H. Lelieveld***, *M. Spielmann, S. Mundlos, J.A. Veltman, C. Gilissen;* *Nijmegen, Netherlands*	**C03.5** **De novo loss-of-function mutations in WAC in the 10p12p11 critical region cause intellectual disability** **Margot R.F. Reijnders***, *D. Lugtenberg, M. Fenckova, E.K. Bijlsma, B.W.M. van Bon, A.T. Vulto- van Silfhout, D.G.M. Bosch, E.E. Eichler, H.C. Mefford, R. Pfundt, H.G. Yntema, P.F. de Vries, J.A. Veltman, B.B.A. de Vries, A. Hoichen, A. Schenck, T. Kleefstra, L.E.L.M. Vissers;* *Nijmegen, Netherlands*	**C04.5** **Vaccination with monocyte-derived dendritic cells in Lynch syndrome patients: vigorous T cell responses to neoantigen frameshift-derived peptides.** **Nicoline Hoogerbrugge**, *H. Westdorp, G. Schreibelt, K. Bol, M. Welzen, J. Krieken, T. Bisseling, M. Ligtenberg, W. Gerritsen, C. Figdor, I. Vries;* *Nijmegen, Netherlands*	**C05.5** **Recessive mutations in matrix metallopeptidase 21 (MMP21) cause heterotaxy in humans** **Anne Guimier***, *G. Gabriel, F. Bajolle, M. Tsang, M. Schwartz, A. Noll, L. Smith, H. Yagi, C. Saunders, C. Baker, M. Oufadem, N. Miller, K. Peterson, I. Thiffault, N. Klena, C. Bole-Feysot, P. Nitschke, S. Lyonnet, L. de Pontual, S. Murray, D. Bonnet, S. Kingsmore, J. Amiel, P. Bouvagnet, C. Lo, C. Gordon;* *Paris, France*	**C06.5** **Junctophilin-1 expression levels could modify the effects of GDAP1 mutations in Charcot-Marie-Tooth disease** **Eduardo Calpena***, *V. Lopez, V. Lupo, T. Sevilla, F. Palau, I. Galindo, C. Espinós;* *Valencia, Spain*
19.45	**C01.6** **Non-invasive prenatal testing for aneuploidy and beyond: challenges of responsible innovation in prenatal screening - an ESHG/ASHG position statement** **Wybo Dondorp**, *G. de Wert, C.G. Van El, M.C. Cornel;* *Maastricht, Netherlands*	**C02.6** **A significant proportion of de novo point mutations arise post-zygotically** *R. Acuna-Hidalgo, T. Bo, M. Kwint, M. van de Vorst, M. Pinelli, J.A. Veltman, H. Alexander, L.E.L.M. Vissers,* **Christian Gilissen**; *Nijmegen, Netherlands*	**C03.6** **A novel syndrome of learning disability and obesity caused by 6q16 deletions encompassing the essential neurogenesis factor POU3F2 (Brn2) helps to delineate the neuro-endocrine pathway for body-mass control** **Siddharth Banka**, *P. Kasher, K.E. Schertz, M. Thomas, S. Annunziata, M. Ballesta, P. Campeau, J.L. Eaton, T. Granata, E. Guillén-Navarro, A. Jackson, C.E. Laverriere, A. Liedén, O.V. Marcos, A. Nordgren, C. Pantaleoni, C. Pebrel-Richard, F.L. Sciacca, C. Sarret, R. Wright, B. Kerr, E. Glasgow;* *Manchester, UK*	**C04.6** **Through the looking glass: the reversion of EMT** **Patricia Oliveira***, *J. Carvalho, S. Rocha, M. Azevedo, A. Vieira, D. Ferreira, N. Mendes, I. Reis, J. Vinagre, A. Heravi-Moussavi, J. Nunes, J. Lima, V. Maximo, A. Burleigh, C. Roskelley, F. Carneiro, R. Seruca, J. Paredes, D. Huntsman, C. Oliveira;* *Porto, Portugal*	**C05.6** **Somatic/mosaic mutations are an important cause of sporadic vascular anomalies.** **Miikka Vikkula**, *A. Mendola, J. Soblet, M. Schlögel, M. Amyere, P. Brouillard, N. Limaye, L.M. Boon;* *Brussels, Belgium*	**C06.6** **CCDC174 mutation underlies a syndrome of hypotonia and psychomotor developmental delay with abducens nerve palsy** **Michael Volodarsky***, *H. Lichtig, T. Leibson, Y. Sadaka, K. Leibson, L. Gradstein, Z. Shorer, R. Shaco-Levy, D. Frank, O.S. Birk;* *Beer-Sheva, Israel*
20.15	*Networking Mixer in the Glasgow Science Centre*					

SCIENTIFIC

SCIENTIFIC PROGRAMME

Sunday, June 7, 2015

PROGRAMME

PROGRAMME SUNDAY, JUNE 7

Time	Clyde	Hall 5	Hall 2	Lomond	Hall 1
08.30 - 10.00	**S01 Big data genomics and human knock-outs** Chair: E. Sheridan, S. Ripatti	**ES05 Automating clinical genetics** Chair: D. Fitzpatrick, M. Suri	**S02 ESHG-ASHG Building Bridges Session: Genetic testing in children** ***Joint with the ASHG*** Chair: J. McInerney, P. Borry	**S03 Epigenetic basis of disease** Chair: K. Temple, M. Nyström	**S04 Spliceosome-opathies** Chair: R. O'Keefe, W. Newman
08.30	**S01.1** **Integrative analysis of 80,000 whole exome sequencing and the Human Knock-out Project** **Daniel McArthur**; *Boston, United States*	**ES5.1** **GeneConsult, Phenomizer, Face2gene with short presentations how they work and test cases to compare the tools** **Peter Robinson**; *Berlin, Germany*	**S02.1** **Whole genome sequencing in newborn screening? A Statement on the continued importance of targeted approaches in newborn screening programmes** **Heidi C. Howard**, *B.M. Knoppers, M.C. Cornel, E. Wright Clayton, K. Sénécal, P. Borry, European Society of Human Genetics, the P3G International Paediatric Platform, the Human Genome Organisation, the PHG Foundation;* *Uppsala, Sweden*	**S03.1** **Heritable germline epimutations in humans** **Bernhard Horsthemke**; *Essen, Germany*	**S04.1** **Spliceosome biology: Understanding causes and consequences of splicing mutations** **Mikko J. Frilander**; *Helsinki, Finland*
09.00	**S01.2** **Identification of a large set of rare complete human knockouts** **Patrick Sulem**; *Reykjavik, Iceland*	**ES5.2** **Clinical Face Phenotype Space: Using standard facial imaging to aid diagnosis of genetic syndromes** *M. Alvi, Q. Ferry, J. Steinberg, C. Webber, D.R. FitzPatrick, C.P. Ponting, A. Zisserman,* **Christoffer Nellaker**; *Oxford, United Kingdom*	**S02.2** **The 2014 ASHG Statement on Genetic testing in Children and Adolescence** **Jeff Botkin**; *Salt Lake City, United States*	**S03.2** **Maternal obesity during pregnancy and offspring later life disease** **Rebecca Reynolds**; *Edinburgh, United Kingdom*	**S04.2** **Spliceosome and development in human** **Jeanne Amiel**; *Paris, France*
09.30	**S01.3** **SISu project: 200,000 near complete sequences of Finns** **Aarno Palotie**; *Helsinki, Finland*		**S02.3** **Carrier testing in children and adolescents** **Sylvia A. Metcalfe**; *Parkville Victoria, Australia* ESHG Building Bridges Session ASHG "Towards finding global agreement on topical discussions in genetics"	**S03.3** **Regional activation of the cancer genome by long range epigenetic remodelling** **Susan Clark**; *Sydney, Australia*	**S04.3** **Spliceosome and cancer** **Philipp A. Greif**; *München, Germany*
10.00 - 10.30	*Coffee Break / Poster viewing / Exhibition*				
10.30 - 11.30	**Poster viewing with presenters (poster numbers starting with "PS")**				
11.30 - 13.30	*Lunch break / Posters / Exhibition*				

PROGRAMME SUNDAY, JUNE 7

	Clyde
11.45 - 13.15	**ES06 My vision on genomic medicine** Chair: J. Burn, J. Veltman
	ES6.1 **The 100,000 Genomes Project, Bringing Personalised Medicine Into Healthcare** **Mark Caulfield**; *London, United Kingdom*
	ES6.2 **My vision on genomic medicine** **Anne Wojcicki**; *Mountain View, United States*

PROGRAMME SUNDAY, JUNE 7

Time	Clyde	Hall 5	Hall 2	Lomond	Hall 1	Forth
13.30 - 15.00	**C07 Reproductive Genetics** Chair: D. Plaseska-Karanfilska, D. McMullan	**C08 Integrative OMICS approaches in common traits** Chair: M. Perola, S. Eyre	**C09 Genetic susceptibility to cancer development** Chair: A. Carrió, J. Bell	**C10 Neurogenetic disorders** Chair: D. Pilz, M. Nöthen	**C11 Skeletal disorders** Chair: F. Ramos, M. Wright	**C12 Sensory disorders** Chair: L. Hoefsloot, N. Canki-Klain
13.30	**C07.1** **Does paternal imprinting of FOXF1 on 16q24.1 explain maternal UPD(16) phenotype?** **Avinash V. Dharmadhikari**, *B. Carofino, J.J. Sun, P. Szafranski, R. Ray, M.J. Justice, M.E. Dickinson, P. Stankiewicz; Houston, United States*	**C08.1** **Context-specific eQTLs identify hormonal effects in obese Finnish men** **Arthur Ko***, *R.M. Cantor, B. Pasaniuc, E. Nikkola, M. Alvarez, A.J. Lusis, M. Civelec, M. Boehnke, F.S. Collins, K.L. Mohlke, J. Kuusisto, M. Laakso, P. Pajukanta; Los Angeles, United States*	**C09.1** **High yield of causative mutations by whole exome sequencing in selected individuals with childhood cancer** **Marjolijn Jongmans***, *E. Waanders, M. Ligtenberg, E. Kamping, P. Hoogerbrugge, M. Oldenrode-Berends, D. Koolen, G. van Santen, M. van Belzen, D. Mordaunt, A. Kattamis, E. de Bont, R. Kuiper, N. Hoogerbrugge; Nijmegen, Netherlands*	**C10.1** **Whole genome sequencing reveals the mutation characteristics in Autism Spectrum Disorder** **Ryan K. Yuen***, *B. Thiruvahindrapuram, D. Merico, S. Walker, K. Tammimies, N. Hoang, C. Chrysler, T. Nalpathamkalam, G. Pellecchia, Y. Liu, M.J. Gazzellone, L. D'Abate, E. Deneault, J.L. Howe, R.S. Liu, A. Thompson, M. Zarrei, M. Uddin, C.R. Marshall, R.H. Ring, L. Zwaigenbaum, P.N. Ray, R. Weksberg, M. Carter, B. Fernandez, W. Roberts, P. Szatmari, S.W. Scherer; Toronto, Canada*	**C11.1** **Mutations in a novel dynein-2 light chain, TCTEX1D2, cause Jeune Asphyxiating Thoracic Dystrophy (JATD) with incomplete penetrance** **Miriam Schmidts***, *Y. Ho, C. Cortes, C. Huber, D. Mans, K. Boldt, C.A. Johnson, M. Ueffing, H. Kayserili, D. Krakow, U. Consortium, P.L. Beales, L. Al Gazali, C. Wicking, V. Cormier-Daire, R. Roepman, H. Mitchison, G. Witman; Nijmegen, Netherlands*	**C12.1** **A novel disorder reveals Clathrin Heavy Chain-22 is essential for human pain and touch development** **Mike S. Nahorski***, *L. Al-Gazali, J. Hertecant, D.J. Owen, G. Borner, Y. Chen, C. Benn, O. Carvalho, S.S. Shaikh, A. Phelan, M. Robinson, S. Royle, G.C. Woods; Cambridge, United Kingdom*
13.45	**C07.2** **Next-gen cytogenetics in prenatal diagnosis: lessons learned with balanced de novo rearrangements** **Cynthia C. Morton**, *Z. Ordulu, T. Kammin, C. Hanscom, V. Pillalamarri, J.L. Andujar, B.B. Currall, J.F. Gusella, M.E. Talkowski; Boston, United States*	**C08.2** **Genetic variants affect expression of nearly all genes, but only in a specific context** **Daria V. Zhernakova***, *The BIOS consortium; Groningen, Netherlands*	**C09.2** **Integration of somatic and germline exome data to evaluate pathogenicity of rare variants in cancer predisposition genes** **Shawn Yost**, *M. Clarke, E. Ruark, N. Rahman; London, United Kingdom*	**C10.2** **Identification of a common set of microRNAs deregulated in Autism Spectrum disorders** *L. Nguyen, M. Lepleux, M. Makhlouf, C. Martin, J. Fregeac, A. Phillipe, F. Ferron, B. Gepner, C. Rougeulle, Y. Humeau,* **Laurence Colleaux***; *Paris, France*	**C11.2** **Mutations in DVL1 cause an osteosclerotic form of Robinow Syndrome** **Stephen Robertson**, *K. Bunn, P. Daniel, H. Rosken, A. O'Neill, S. Cameron-Christie, D. Markie, H. Brunner, H. Kunst, A. Lai; Dunedin, New Zealand*	**C12.2** **Exome sequencing of ataxia-blindness patients identifies atypical Brown-Vialetto-Van Laere syndrome-2 (BVVLS2) presentation and identifies PEX6 as the SCAR3 (MIM#271250) gene** **Claire Guissart***, *E. Acar, H. Topaloglu, B. Leheup, S. Ferdinandusse, M. Koenig; Montpellier, France*
14.00	**C07.3** **Targeted prenatal screening as a successful and fast approach in cases with increased nuchal translucency and/or abnormal ultrasound** **Pascal Joset**, *A. Baumer, M. Papic, S. Papuc, M. Zweier, S. Azzarello-Burri, D. Niedrist, L. Gogoll, B. Oneda, K. Steindl, A. Rauch; Schlieren-Zurich, Switzerland*	**C08.3** **Pedigree-Associated Genetics and Recent Environment Make Important Contributions to Metabolic Syndrome Traits.** **Charley Xia***, *C. Amador, J. Huffman, H. Trochet, A. Campbell, G. Scotland, D. Porteous, N. Hastie, C. Hayward, V. Vitart, P. Navarro, C.S. Haley; Edinburgh, United Kingdom*	**C09.3** **Expanding the mutation spectrum and phenotype of Polymerase Proofreading-Associated Polyposis (PPAP): novel and previously reported POLE variants** **Maurizio Genuardi**, *M. Calicchia, M. Ciavarella, B. Riboli, P. Cavalli, M. Castori, P. Grammatico, E. Lucci-Cordisco; Rome, Italy*	**C10.3** **Rare variants in GABAA receptor genes in Rolandic epilepsy and related syndromes** **Eva M. Reinthaler***, *B. Dejanovic, D. Lal, M. Semtner, Y. Merkler, A. Reinhold, D.A. Pittrich, C. Hotzy, J. Altmüller, A. Kawalia, M.R. Toliat, EuroEPINOMICS Consortium, GABA receptor study group, P. Nürnberg, H. Lerche, M. Nothnagel, H. Thiele, T. Sander, J.C. Meier, G. Schwarz, B.A. Neubauer, F. Zimprich; Vienna, Austria*	**C11.3** **Mutations in ZAK cause autosomal recessive split foot malformation in humans and complex hindlimb defects in mice** **Naeimeh Tayebi***, *N. Kakar, M. Spielmann, C. Leettola, S. Kühl, G. Nürnberg,, N. Sowada, J. Altmüller, D. Lupianez, R. Flöttmann,, M. Radenz, H. van Bokhoven, C. Schwartz, H. Thiele, P. Nürnberg, M. Kühl, J. Bowie, C. Kubisch, S. Ahmad, S. Mundlos, G. Borck; Berlin, Germany*	**C12.3** **Heimler Syndrome is caused by unique hypomorphic mutations in the peroxisome biogenesis genes PEX1 and PEX6** *I. Ratbi, K.D. Falkenberg, M. Sommen, N. Al-Sheqaih, S. Guaoua, J.E. Urquhart, K.E. Chandler, S.G. Williams, N.A. Roberts, M. El Alloussi, G.C. Black, S. Ferdinandusse, H. Ramdi, A. Heimler, A. Fryer, S. Lynch, N. Cooper, K. Ong, C.E. Smith, C.F. Inglehearn, A.J. Mighell, J.A. Poulter, M. Tischkowitz, S. Davies, A. Sefiani, A.A. Mironov, W.G. Newman, H.R. Waterham,* **Guy Van Camp**; *Antwerp, Belgium*

Presentations highlighted by an asterisk * and a grey background are from Young Investigator Award Finalists.

Time	Clyde	Hall 5	Hall 2	Lomond	Hall 1	Forth
cont.	**C07 Reproductive Genetics**	**C08 Integrative OMICS approaches in common traits**	**C09 Genetic susceptibility to cancer development**	**C10 Neurogenetic disorders**	**C11 Skeletal disorders**	**C12 Sensory disorders**
14.15	**C07.4 Comprehensive carrier genetic test using next-generation DNA sequencing in infertile couples wishing to conceive trough assisted reproductive technologies (ART)** **Trinitat M. Alberola**, *J. Martin, A. Asan, Y. Yuting, B. Rodriguez-Iglesias, J. Jimenez, Y. Xin, C. Simon; Paterna (Valencia), Spain*	**C08.4 Genome-wide study for metabolic phenotypes identifies 62 loci and elucidates the metabolic context of LPA in coronary heart disease** **Johannes Kettunen**, *A. Demirkan, H.H.M. Draisma, T. Haller, R. Rawal, A. Vaarhorst, A.J. Kangas, L. Lyytikäinen, M. Pirinen, R. Pool, A. Sarin, P. Soininen, T. Tukiainen, Q. Wang, P. Würtz, N. Amin, M. Beekman, J. Deelen, K. van Dijk, J. Hottenga, E.M. van Leeuwen, T. Lehtimäki, E. Mihailov, R.J. Rose, A.J.M. de Craen, L. Bogl, C. Gieger, M. Kähönen, M. Perola, M.J. Savolainen, A. Verhoeven, J. Viikari, G. Willemsen, D.I. Boomsma, C.M. van Duijn, J. Eriksson, A. Jula, M. Järvelin, J. Kaprio, A. Metspalu, O. Raitakari, V. Salomaa, P.E. Slagboom, M. Waldenberger, M. Ala-Korpela, S. Ripatti; Oulu, Finland*	**C09.4 Germline mutations in patients with hereditary breast and ovarian cancer establish ERCC2 as a cancer susceptibility gene.** **Andreas Rump**, *A. Benet-Pages, S. Schubert, R. Janavicius, K. Hackmann, E. Betcheva-Krajcir, L. Mackenroth, J. Lehmann, A. Nissen, J. Altmueller, H. Thiele, N. Di Donato, B. Klink, J. Kuhlmann, A. Tzschach, K. Kast, P. Wimberger, E. Holinski-Feder, A. Meindl, S. Emmert, E. Schrock; Dresden, Germany*	**C10.4 Hyperexcitability or electrical silencing: *de novo* loss- or gain-of-function mutations in *KCNA2* cause epileptic encephalopathy** *S. Syrbe, U. Hedrich, E. Riesch, T. Diémié, S. Müller, R. Møller, B. Maher, L. Hernandez-Hernandez, M. Synofzik, H. Caglayan, M. Arslan, J. Serratosa, M. Nothnagel, P. May, R. Krause, H. Löffler, K. Detert, T. Dorn, H. Vogt, G. Krämer, L. Schöls, P. Mullis, T. Linnankivi, A. Lehesjoki, K. Sterbova, D. Craiu, D. Hoffman-Zacharska, C. Korff, Y. Weber, M. Steinlin, S. Gallati, A. Bertsche, M. Bernhard, A. Merkenschlager, W. Kiess, EuroEPINOMICS RES consortium, M. Gonzalez, S. Züchner, A. Palotie, A. Suls, P. De Jonghe, I. Helbig, S. Biskup, M. Wolff, S. Maljevic, R. Schüle, S. Sisodiya, S. Weckhuysen, H. Lerche,* **Johannes Lemke**; *Leipzig, Germany*	**C11.4 Spondyloenchondro-dysplasia: The ex-panding phenotype of TRAP deficiency** **Tracy A. Briggs***, *G.I. Rice, Y.J. Crow; Manchester, United Kingdom*	**C12.4 An in-frame deletion in *FOXL1* identifies the first gene causing autosomal dominant otosclerosis** *N. Abdelfatah, A. Mostafa, S.G. Stanton, M.B. Lucas, A. Griffin, V. Booth, C. Rowley, J.E. Besaw, L. Tranebjærg, N. Dahl Rendtorff, K.A. Hodgkinson, L.A. Little, A. Sangamanatha, S. Agrawal, L. Parnes, A. Batten, J. Houston, D. Galutira, T. Benteau, C. Penney, C. Negrijn,* **Terry-Lynn Young**; *St. John's, Canada*
14.30	**C07.5 Non-manifesting AHI1 truncations indicate localized loss-of-function tolerance in a severe Mendelian disease gene** *S.M. Elsayed, J.B. Phillips, R. Heller, M. Thoenes, E. Elsobky, G. Nürnberg, P. Nürnberg, S. Seland, I. Ebermann, J. Altmüller, H. Thiele, M. Toliat, F. Körber, X. Hu, Y. Wu, M.S. Zaki, G. Abdel-Salam, J. Gleeson, E. Boltshauser, M. Westerfield,* **Hanno J. Bolz**; *Ingelheim, Germany*	**C08.5 Systematic identification of downstream trans-effects for 1,300 known disease associated SNPs** **Marc Jan Bonder**, *R. Luijk, BBMRI-NL BIOS Consortium; Groningen, Netherlands*	**C09.5 Tumour risks and genotype-phenotype-proteotype analysis in ~800 patients with germline mutations in the succinate dehydrogenase subunit genes SDHB, SDHC and SDHD** **Katrina A. Andrews***, *D.B. Ascher, D.E.V. Pires, L. Vialard, N. Bradshaw, L. Izatt, A. Kumar, F. Lalloo, R. Irving, J. Cook, T. Cole, D. Goudie, M. McConachie, R. Lindsay, C. Perry, J. Adlard, V. Murday, S. Stewart, E. Woodward, E.R. Maher; Cambridge, United Kingdom*	**C10.5 Cysteine Correction of NOTCH3: exon skipping as a potential therapeutic strategy for CADASIL** **Julie W. Rutten***, *H.G. Dauwerse, D.J.M. Peters, A. Goldfarb, R.R. Klever, H. Venselaar, S. Verbeek, A.M.J.M. van den Maagdenberg, G.B. van Ommen, A.M. Aarstma-Rus, S.A.J. Lesnik Oberstein; Leiden, Netherlands*	**C11.5 Brachyolmia with amelogenesis imperfecta can be caused by a defect in the TGFbeta signaling pathway** **Agnes M. Bloch-Zupan**, *M. Huckert, C. Stoetzel, S. Morkmued, V. Laugel-Haushalter, V. Geoffroy, J. Muller, F. Clauss, M.K. Prasad, F. Obry, Y. Alembik, S. Soskin, J. Hemmerlé, J. Weickert, B. Dabovic, D.B. Rifkin, A. Dheedene, E. Boudin, O. Caluseriu, M. Cholette, R. McLeod, R. Antequera, M. Gellé, L. Jacquelin, I. Bailleul-Forestier, M. Manière, W. Van Hul, D. Bertola, P. Dollé, A. Verloes, G. Mortier, H. Dollfus; Strasbourg, France*	**C12.5 Submicroscopic deletions at 13q32.1 cause congenital microcoria** **Lucas Fares Taie***, *S. Gerber, A. Tawara, A. Ramirez-Miranda, J. Douet, H. Verdin, A. Guilloux, J. Zenteno, H. Kondo, H. Moisset, B. Passet, K. Yamamoto, M. Iwai, T. Tanaka, Y. Nakamura, W. Kimura, C. Bole-Feysot, M. Vilotte, S. Odent, J. Vilotte, A. Munnich, A. Regnier, N. Chassaing, E. De Baere, I. Raymond-Letron, J. Kaplan, P. Calvas, O. Roche, J. Rozet; Paris, France*
14.45	**C07.6 How to design expanded carrier screening panels? Results of an interview study with European geneticists** **Davit Chokoshvili**, *S. Janssens, D. Vears, A. De Paepe, P. Borry; Leuven, Belgium*	**C08.6 Integrated analysis of human and bacterial genomes in relation to BMI and blood lipid metabolites.** **Alexandra Zhernakova**, *M. Bonder, M. Cenit, E. Tigchelaar, J. Dekens, J. Marczynska, F. Imhann, R. Weersma, T. Poon, R. Xavier, D. Gevers, L. Franke, M. Hofker, C. Wijmenga, J. Fu; Groningen, Netherlands*	**C09.6 Germline SMAD9 Mutation Destabilizes PTEN: Exome Sequencing Reveals a Novel Susceptibility Gene For Hamartomatous Polyposis and Gastrointestinal Ganglioneuromas** **Joanne Ngeow**, *W. Yu, L. Yehia, C. Eng; Singapore, Singapore*	**C10.6 De novo deleterious genetic variations target a biological network centered on Aβ peptide in early-onset Alzheimer disease** *A. Rovelet-Lecrux, C. Charbonnier, D. Wallon,* **Gaël Nicolas***, *M.N.J. Seaman, C. Pottier, S.Y. Breusegem, P. Prakash Mathur, P. Jenardhanan, K. Le Guennec, A.S. Mukadam, O. Quenez, S. Coutant, S. Rousseau, A. Richard, A. Boland, J. Deleuze, T. Frebourg, D. Hannequin, D. Campion; Rouen, France*	**C11.6 Pentosan Polysulfate: New Mechanistic Insights and Treatment of the Mucopolysaccharidoses** **Calogera M. Simonaro**, *S. Tomatsu, M. Frohbergh, M. Haskins, A. Solyom, E. Schuchman; New York, United States*	**C12.6 A molecular network surrounding dysregulated H3K9 di-methylation in PRDM5-associated disease** **Louise F. Porter***, *G.G. Galli, S. Williamson, J. Selley, D. Knight, N. Elcioglu, M. Elcioglu, A. Lund, R. Bonshek, G. Black, F. Manson; Manchester, United Kingdom*
15.00 - 15.30	*Vitamin break / Poster viewing / Exhibition*					

PROGRAMME SUNDAY, JUNE 7

Time	Clyde	Hall 5	Hall 2	Lomond	Hall 1	Forth	Gala
15.30 – 17.00	**WS03. NGS in the Clinic*** Organiser: J. Veltman	**WS04. Dysmorphology 1*** Organisers: D. Donnai, S. Douzgou	**WS05. Pre-conception carrier testing** Organisers: L. Henneman, U. Kristoffersson	**WS06. Blurred boundaries between clinic and research** Organisers: M. Macek Jr., T. Clancy	**WS07. European Board of Medical Genetics: What can we do to facilitate you to become a registered genetic professional?** Organisers: F. Ramos, D. Coviello	**WS08. Ensembl Highlights: Accessing Genomes*** Organisers: D.R. Carvalho-Silva, A. Zadissa	**WS09. Global Alliance for Genomics and Health** Organisesa: P. Goodhand, M. Lawler
	In this workshop several European experts will present and discuss examples in which diagnostic next generation sequencing (both targeted, exome and genome) was performed, highlighting both challenges and solutions. In addition, people from the audience may bring forward challenging cases to be discussed (please bring max 2 ppt-slides and be present 15 minutes before start of workshop).	The organisers of the dysmorphology workshop invite clinicians to submit rare known and unknown cases with dysmorphic syndromes before the workshop. Please bring a short case presentation on a USB stick from 15:00 - 15:30 hrs to the lecture room. Maximum time for presentation: 5'.	Pre-conception carrier testing is the detection of carrier status of recessive disorders to facilitate informed reproductive decision-making by identifying individuals or couples at risk of having an affected child. Carrier screening on population level detects carrier status in persons who do not have an a priori increased risk of having a child with a certain disease based on their or their partners' personal or family history. This workshop will discuss recent decades of experiences with carrier screening and challenges on the introduction of expanded screening panels.	Clinical care and research are often thought of as separate activities, but the distinction between them can be unclear in practice. Patients/ families do not always recognise the difference between tests for clinical care and those for research. Clinicians/ researchers often see involvement in research as being part of good clinical care and have an expectation that research will benefit participants directly. This workshop will explore the advantages, disadvantages and tensions of this blurred boundary in genomics.	In this workshop we will discuss the work of the European Board of Medical Genetics. The registration systems now operating for clinical laboratory geneticists and genetic nurses and counsellors will be discussed, alongside the European curriculum for specialists in medical genetics. This will be an interactive session and we will be seeking feedback from those who have undergone the registration process and suggestions for development of the work of the EBMG in future.	This interactive workshop is aimed at all attendees of ESHG 2015 who already use Ensembl or would like to familiarise themselves with the genome browser. Attendees will be able to follow along with the demonstrations in this workshop if they wish, and therefore you should bring a fully-charged WiFi enabled laptop.	This workshop will present an overview of the Global Alliance, including overall goals and Working Group progress. The second part of the workshop will be a short presentation to initiate a panel discussion on the topic of community engagement. Specifically, how can the Global Alliance better engage the many different communities (researchers, clinicians, patients, and others) with whom – and for whom – we work.
17.00 – 17.30	*Coffee break / Poster viewing / Exhibition*						

Detailed Workshop programmes (when submitted by the organisers) can be found in the "ESHG Bulletin" in the conference bag.

*Interactive workshops - your input is solicited. See details in the ESHG Bulletin for more information.

*In selected workshops, a voting system will be made available to the audience. Connect to the wifi „voting_ *workshopnumber*" (e.g. „voting_ws03") with any WIFI-capable device (laptop, tablet, phone) and open an internet browser. The voting form will be displayed accordingly.*

PROGRAMME SUNDAY, JUNE 7

Time	Clyde	Hall 5	Hall 2	Lomond	Hall 1
17.30 - 19.00	**S05 Reproductive genetics and "Chromosome therapy"** Chair: J. Vermeesch, U. Maye	**S06 International data sharing initiatives** Chair: A. Hall, J. Veltman	**ES07 Imprinting-related disorders** Chair: E. Maher, G. Houge	**S07 Mouse phenotyping for clinical research** Chair: P. Robinson	**S08 Telomeres in human disease** Chair: J.B. Vannier, M. Genuardi
17.30	**S05.1** **Using XIST to Silence Trisomy 21: Implications for Cell and Chromosome Therapy** **Jun Jiang**, *Y. Jing, G. Cost, J. Chiang, H. Kolpa, A. Cotton, D.M. Carone, B.R. Carone, M. Byron, P.D. Gregory, C.J. Brown, F.D. Urnov, L.L. Hall, J.B. Lawrence;* *Worcester, United States*	**S06.1** **DECIPHER** **Helen Firth**; *Cambridge, United Kingdom*	**ES7.1** **Imprinting and long noncoding RNAs in health and disease** **Marisa S. Bartolomei**; *Philadelphia, United States*	**S07.1** **The International Mouse Phenotyping Consortium: New insights into the genetic and molecular bases of disease** **Steve Brown**, *International Mouse Phenotyping Consortium;* *Harwell, United Kingdom*	**S08.1** **Constitutional and somatic variations in telomerase reverse transcriptase and human cancer** **Rajiv Kumar**; *Heidelberg, Germany*
18.00	**S05.2** **Having developed an accurate noninvasive prenatal test for aneuploidies- What else can we work on?** **K. C.Allen Chan**; *Hong Kong, China*	**S06.2** **Sharing Data in Cancer Genomics; Lessons from the International Cancer Genome Consortium** **Sean Grimmond**; *Glasgow, United Kingdom*	**ES7.2** **Diagnosing imprinting-related disorders** **Karen I. Temple**; *Southampton, United Kingdom*	**S07.2** **Investigating genetic diseases with intellectual disability in the mouse** **Yann Herault**, *The GENCODYS Network;* *Illkirch, France*	**S08.2** **The role of telomeres in aging** **Christian Bär**; *Madrid, Spain*
18.30	**S05.3** **Status and outcome of randomized trials for aneuploidy screening preimplantation embryos** **Jan Traeger-Synodinos**; *Athens, Greece*	**S06.3** **The Challenge of the Global Variome** **John Burn**; *Newcastle upon Tyne, United Kingdom*		**S07.3** **Deciphering the genetic and epigenetic role in metabolic diseases** **Martin Hrabé de Angelis**; *Munich, Germany*	**S08.3** **Novel insights into the telomere syndromes** **Inderjeet Dokal**; *London, United Kingdom*

	Gala
19.15 – 20.15	**ESHG Membership Meeting** **All ESHG members welcome!**

SCIENTIFIC PROGRAMME

Monday, June 8, 2015

PROGRAMME MONDAY, JUNE 8

Time	Clyde	Hall 5	Hall 2	Lomond	Hall 1
08.30 - 10.00	**S09 Evolution of the cancer genome: clinical implications** Chair: J. Machado, V. Murday	**S10 From rare to common variants in cardiovascular diseases** ***(joint with the European Society of Cardiology)*** Chair: R. Newbury-Ecob, X. Jeunemaitre	**S11 Non-coding DNA and human disease** Chair: J. Ferrer, Y. Crow	**S12 Mitochondria and genetic disease** Chair: F. Palau, R. Taylor	**ES08 Palliative care of genetic conditions** Chair: A. Bruce, T. Clancy
08.30	**S09.1** **The AML Genome(s)** **Timothy J. Ley**, *on behalf of the Genomics of Acute Myeloid Leukemia Program Project Grant, and The Genome Institute;* *St. Luis, United States*	**S10.1** **Genomics and Hypertension** **Anna Dominiczak**; *Glasgow, United Kingdom*	**S11.1** **Retrotransposons and human disease** **Jose Luis Garcia-Pérez**, *S. Morell, E. Blanco-Jimenez, S. Amador-Cubero;* *Granada, Spain*	**S12.1** **Mitochondria in neurodegeneration** **Eric Schon**; *New York, United States*	**ES8.1** **Wishes for the end of life in Huntington's Disease** **Suzanne Booij**, *A. Tibben, R.A.C. Roos, D.P. Engberts;* *Nijmegen, Netherlands*
09.00	**S09.2** **Reconstruction of clonal composition in cancer** **Ville Mustonen**; *Cambridge, United Kingdom*	**S10.2** **Ten Years Later: How The Pcsk9 Gene Discovery Affects the Diagnosis and Treatment of Hypercholesterolemia** **Catherine Boileau**; *Paris, France*	**S11.2** **CNVs of noncoding cis-regulatory elements in human disease** **Eva Klopocki**; *Würzburg, Germany*	**S12.2** **Mitochondrial dynamics in the pathophysiology of genetic disease** **Luca Scorrano**; *Padua, Italy*	**ES8.2** **End of life decision making in neonates** **Eduard Verhagen**; *Groningen, Netherlands*
09.30	**S09.3** **Genomic medicine to tailor cancer drugs** **Nicola Normanno**; *Naples, Italy*	**S10.3** **Genetic variation in APOC3, plasma triglycerides and risk of ischemic cardiovascular disease** *A. Jørgensen, R. Frikke-Schmidt, B.G. Nordestgaard,* **Anne Tybjaerg-Hansen**; *Copenhagen, Denmark*	**S11.3** **A Novel Dicer1-miR328-Bace1 Signaling Axis Controls Ageing- and Obesity-Induced Brown Fat Dysfunction** *M. Oliverio, J.C. Brüning,* **Jan W. Kornfeld**; *Cologne, Germany*	**S12.3** **Gene therapy for mitochondrial neurogastrointestinal encephalomyopathy (MNGIE)** **Ramon Martí**; *Barcelona, Spain*	
10.00 - 10.30	*Coffee break / Poster viewing / Exhibiton*				
10.30 - 11.30	**Poster viewing with presenters (poster numbers starting with "PM")**				
11.30 - 13.30	*Lunch break / Posters / Exhibition*				

PROGRAMME MONDAY, JUNE 8

Time	Clyde	Hall 5	Hall 2	Lomond	Hall 1	Forth
13.30 - 15.00	**C13 Fundamental insights in structural genomics** Chair: S. Lyonnet, W. Yue	**C14 Challenges in genetic counselling** Chair: F. Forzano, L. Boyes	**C15 Network and functional analysis in intellectual disability** Chair: K. Devriendt, L. Raymond	**C16 Growth failure and microcephaly** Chair: A. Jackson, H. Brunner	**C17 Epigenetic control of gene expression** Chair: I. Ceccherini, K. Temple	**C18 Metabolic and renal disorders** Chair: P. Kroisel, S. Banka
13.30	**C13.1** **Human-specific gene evolution and diversity of the chromosome 16p11.2 autism CNV** **Giuliana Giannuzzi**, *X. Nuttle, M.H. Duyzend, P.H. Sudmant, O. Penn, G. Chiatante, M. Malig, J. Huddleston, L. Denman, L. Harshman, J. Chrast, C. Baker, A. Raja, K. Penewit, F. Antonacci, A. Reymond, E.E. Eichler; Lausanne, Switzerland*	**C14.1** **External Quality Assessment of Genetic Counselling: experiences with the first pilot assessment** **Conny M.A. van Ravenswaaij-Arts**, *C. van Asperen, E. Dequeker, L. Tranebjaerg, L. Garavelli, B. Peterlin, B. Cope, H. Skirton, R. Hastings, ESHG Genetic Services Quality Committee; Groningen, Netherlands*	**C15.1** **Genome-wide association study of 200,000 individuals identifies 18 genome-wide significant loci and provides biological insight into human cognitive function** **Tonu Esko***, *on the behalf of Social Science Genetic Association Consortium (SSGAC); Tartu, Estonia*	**C16.1** **Systematic evaluation of patients with idiopathic short stature using whole exome sequencing** **Christian T. Thiel**, *N.N. Hauer, S. Schuhmann, E. Schöller, M.T. Wittmann, S. Uebe, A.B. Ekici, H. Sticht, H. Dörr, A. Reis; Erlangen, Germany*	**C17.1** **RNF12 is essential for X-inactivation in female mouse embryonic stem cells, is required for female mouse development, and might be a target for future therapies to treat X-linked disorders in females: evidence from a mouse knockout model** **Tahsin Stefan S. Barakat***, *J. Gribnau; Rotterdam, Netherlands*	**C18.1** **Disassembly of MINOS complex by *CHCHD10* mutations promotes loss of mitochondrial cristae with defects in mitochondrial genome maintenance and apoptosis** *E. Genin, M. Plutino, S. Bannwarth, E. Villa, E. Cisneros-Barroso, M. Roy, B. Ortega-Vila, K. Fragaki, F. Lespinasse, E. Pinero-Martos, G. Augé, D. Moore, F. Burté, S. Lacas-Gervais, Y. Kageyama, P. Yu-Wai-Man, H. Sesaki, J. Ricci, C. Vives-Bauza,* **Véronique Paquis-Flucklinger**; *Nice, France*
13.45	**C13.2** **The impact and activity of mobile elements within the genome** **Jayne Y. Hehir-Kwa**, *D. Thung, V. Guryev, W.P. Kloosterman, T. Marschall, K. Ye, J.A. Veltman; Nijmegen, Netherlands*	**C14.2** **Hereditary breast and ovarian cancer syndrome: successful, large-scale implementation of a group-based approach to genetic counseling.** **Patrick R. Benusiglio**, *M. Di Maria, A. Jouinot, B. Claret, D. Boinon, D. Lejri, O. Caron; Villejuif, France*	**C15.2** **Systematic phenotype-based deconvolution of intellectual disability disorders into biologically coherent modules** **Christiane Zweier**, *K. Kochinke, B. Nijhof, M. Fenckova, P. Cizek, F. Honti, S. Keerthikumar, M.A.W. Oortveld, T. Kleefstra, J.M. Kramer, C. Webber, M.A. Huynen, A. Schenck; Erlangen, Germany*	**C16.2** **Mutations in the core NHEJ components LIG4 and XRCC4 result in microcephalic primordial dwarfism** **Jennie E. Murray***, *M. van der Burg, H. Ijspeert, P. Carroll, Q. Wu, T. Ochi, A. Leitch, E.S. Miller, B. Kysela, A. Jawad, A. Bottani, F. Brancati, M. Cappa, V. Cormier-Daire, C. Deshpande, E. Ali Faqeih, G. Graham, E. Ranza, T.L. Blundell, A.P. Jackson, G.S. Stewart, L.S. Bicknell; Edinburgh, United Kingdom*	**C17.2** **Pattern of X chromosome inactivation across human tissues - insights from population-scale and single-cell RNA sequencing** **Taru Tukiainen***, *A. Villani, A. Kirby, D. DeLuca, R. Satija, A. Byrnes, J. Maller, T. Lappalainen, The GTEx Project Consortium, A. Regev, K. Ardlie, D. MacArthur; Boston, United States*	**C18.2** **COQ4 mutations cause a broad spectrum of mito-chondrial disorders associated with CoQ10 deficiency** **Laura Kremer***, *G. Brea-Calvo, T.B. Haack, D. Karall, A. Ohtake, F. Invernizzi, R. Carrozzo, S. Dusi, C. Fauth, S. Scholl-Bürgi, E. Graf, U. Ahting, N. Resta, N. Laforgia, D. Martinelli, D. Verrigni, Y. Okazaki, M. Kohda, P. Freisinger, T. Strom, T. Meitinger, C. Lamperti, A. Lacson, P. Navas, J. Mayr, E. Bertini, K. Murayama, M. Zeviani, D. Ghezzi, H. Prokisch; Neuherberg, Germany*
14.00	**C13.3** **Chromosomal contacts connect loci associated with autism, BMI and head circumference phenotypes** *M. Loviglio, M. Leleu, G. Giannuzzi, K. Mannik, E. Migliavacca, I. Roberts-Caldeira, I. van der Werf, 16p11.2 European Consortium, J.S. Beckmann, S. Jacquemont, J. Rougemont,* **Alexandre Reymond**; *Lausanne, Switzerland*	**C14.3** **Experiences of systematic genetic testing involving women recently diagnosed with epithelial ovarian cancer: a qualitative study** **Hannah E. Shipman**, *M. Tischkowitz, S. Flynn, C. MacDonald-Smith, N. Hulbert-Williams, GTEOC Study team; Cambridge, United Kingdom*	**C15.3** **9.6% of mouse gene knockouts show abnormal neuroanatomy: a resource to identify genes and gene networks involved in ID in human** *B. Yalcin,* **Anna Mikhaleva**, *V.E. Vancollie, M. Kannan, H. Whitley, A. Edwards, C. Wagner, J. Estabel, C.J. Lelliott, J.K. White, Sanger Mouse Genetics Project, D.J. Adams, D.A. Keays, J. Flint, Y. Herault, A. Reymond; Lausanne, Switzerland*	**C16.3** **Loss-of-Function Mutations in WDR73 Are Responsible for Microcephaly and Steroid-Resistant Nephrotic Syndrome: Galloway-Mowat Syndrome** **Estelle Colin***, *E. Huynh Cong, G. Mollet, A. Guichet, O. Gribouval, C. Arrondel, O. Boyer, L. Daniel, M. Gubler, Z. Ekinci, M. Tsimaratos, B. Chabrol, N. Boddaert, A. Verloes, A. Chevrollier, N. Gueguen, V. Desquiret-Dumas, M. Ferré, V. Procaccio, L. Richard, B. Funalot, A. Moncla, D. Bonneau, C. Antignac; Angers, France*	**C17.3** **Genome wide DNA promoter methylation: Differences in human subcutaneous vs. omental visceral adipose tissue** **Maria Keller***, *L. Hopp, X. Liu, K. Rohde, M. Klös, A. Dietrich, M. Schön, D. Gärtner, T. Lohmann, M. Dreßler, M. Stumvoll, P. Kovacs, H. Binder, M. Blüher, Y. Böttcher; Leipzig, Germany*	**C18.3** **MCT1 deficiency impairs ketone utilization and causes profound ketoacidosis upon catabolic stress** *P. van Hasselt, S. Ferdinandusse, G. Monroe, J. Ruiter, M. Turkenburg, M. Geerlings, K. Duran, M. Harakalova, B. van der Zwaag, A. Monavari, I. Okur, M. Sharrard, M. Cleary, N. O'Connell, V. Walker, E. Rubio Gozalbo, M. de Vries, G. Visser, R. Houwen, J. van der Smagt, N. Verhoeven-Duif, R. Wanders,* **Gijs van Haaften**; *Utrecht, Netherlands*

PROGRAMME MONDAY, JUNE 8

Time	Clyde	Hall 5	Hall 2	Lomond	Hall 1	Forth
cont.	**C13 Fundamental insights in structural genomics**	**C14 Challenges in genetic counselling**	**C15 Network and functional analysis in intellectual disability**	**C16 Growth failure and microcephaly**	**C17 Epigenetic control of gene expression**	**C18 Metabolic and renal disorders**
14.15	**C13.4** **Single-cell allele specific expression (ASE) in T21: a novel approach to understand Down syndrome.** **Georgios Stamoulis***, *P. Makrythanasis, F. Santoni, A. Letourneau, M. Guipponi, M. Garieri, N. Panousis, E. Falconnet, P. Ribaux, C. Borel, S.E. Antonarakis; Geneva, Switzerland*	**C14.4** **Sharing information with children and young people about adult-onset inherited conditions: Using evidence to improve services for parents and their children** **Karen Forrest Keenan**, *L. McKee, Z. Miedzybrodzka; Aberdeen, United Kingdom*	**C15.4** **Finding new connections in the transcriptional regulation of Lysine-specific demethylase 5C (KDM5C) a disease gene involved in syndromic and non-syndromic XLID** **Agnese Padula***, *L. Poeta, C. Shoubridge, A. Ranieri, K. Helin, J. Gecz, C. Schwartz, M.V. Ursini, H. vanBokhoven, M.G. Miano; Naples, Italy*	**C16.4** **Mutations in PLK4, encoding a master regulator of centriole biogenesis, and its substrate, TUBGCP6, cause microcephaly, growth failure and retinopathy** **Louise S. Bicknell**, *C. Martin, A. Klingseisen, I. Ahmad, M.S. Hussain, A. Leitch, G. Nurnberg, M.R. Toliat, J. Murray, D. Hunt, F. Khan, Z. Ali, S. Tinschert, J. Ding, C. Keith, M.E. Harley, P. Heyn, R. Mueller, I. Hoffman, V. Cormier-Daire, H. Dollfus, L. Dupuis, A. Bashamboo, K. McElreavey, A. Kariminejad, R. Mendoza-Londono, A.T. Moore, A. Saggar, C. Schlechter, R. Weleber, H. Thiele, J. Altmuller, W. Hohne, M.E. Hurles, A.A. Noegel, S.M. Baig, P. Nurnberg, A.P. Jackson; Edinburgh, UK*	**C17.4** **Mapping genetic and epigenetic factors influencing human hippocampal gene expression** **Andrea Hofmann***, *H. Schulz, A. Ruppert, S. Herms, K. Pernhorst, C. Wolf, N. Kerbalai, O. Stegle, D. Czamara, S. Sivalingam, A. Hillmer, B. Pütz, A. Woitecki, S. Schoch, A.J. Forstner, B. Müller-Myhsok, M.M. Nöthen, T. Sander, A. Becker, P. Hoffmann, S. Cichon; Bonn, Germany*	**C18.4** **Rare non-synonymous variations in the human ferroportin iron transporter gene (haemochromatosis type 4): the quest for causal mutations** *I. Callebaut, S. Pissard, C. Kannengiesser, V. Gérolami, C. Ged, F. Cartault, J. Rochette, C. Ka, C. Férec,* **Gérald Le Gac**; *Brest, France*
14.30	**C13.5** **High incidence of mosaic chromosomal aneuploidies in human cell lines: a quantification of the frequency of the phenomenon** **Eftychia S. Dimitriadou***, *M. Zamani Esteki, N. Van der Aa, T. Voet, J.R. Vermeesch; Leuven, Belgium*	**C14.5** **Attitudes towards returning data to participants in sequencing research** **Anna Middleton**, *C. Wright, H. Firth, M. Hurles, M. Parker, on behalf of the DDD study; Cambridge, United Kingdom*	**C15.5** **HCFC1 is a dosage sensitive transcriptional coregulator of neurodevelopment that influences neural progenitor and neuronal cell function** **Lachlan A. Jolly**, *L.S. Nguyen, D. Domingo, Y. Sun, S. Barry, M. Hancarova, P. Plevova, M. Vlckova, M. Havlovicova, V.M. Kalscheuer, C. Graziano, T. Pippucci, Z. Sedlacek, E. Bonora, J. Gecz; Adelaide, Australia*	**C16.5** **Mutations in TUBGCP4 alter microtubule organization via the γ-tubulin ring complex γTuRC in autosomal recessive microcephaly with chorioretinopathy.** **Sophie Scheidecker**; *Strasbourg, France*	**C17.5** **Analysis of monoallelic expression in human individual cells revealed novel imprinting genes.** **Christelle Borel**, *F. Santoni, M. Garieri, E. Falconnet, P. Ribaux, S.E. Antonarakis; Geneva, Switzerland*	**C18.5** **Companion diagnostics by comprehensive targeted NGS with evidence for a threshold model in a cohort of 605 patients with atypical haemolytic uremic syndrome and hereditary glomerulopathies** *M. Grohmann, N. Bachmann, M. Hiersche, T. Eisenberger, H.J. Bolz, T. Ring, B. Hohenstein, C. Mache, M.J. Kemper, C.S. Haas, N. Heyne, R.P. Wüthrich, F. Thaiss, B. Tönshoff, L. Pape, M. Wiesener, J. Menne, G. Walz,* **Carsten Bergmann**; *Ingelheim, Germany*
14.45	**C13.6** **Chromothripsis in healthy individuals affects multiple protein-coding genes and can result in severe congenital abnormalities in offspring** **Mirjam S. de Pagter***, *M.J. van Roosmalen, A.F. Baas, I. Renkens, K.J. Duran, E. van Binsbergen, M. Tavakoli-Yaraki, R. Hochstenbach, L.T. van der Veken, E. Cuppen, W.P. Kloosterman; Utrecht, Netherlands*	**C14.6** **Population-based Preconception Carrier Screening: how do potential users view a preconception test for 70 severe autosomal recessive diseases?** **Mirjam Plantinga**, *E. Birnie, S. Kaplan, M.A. Verkerk, A.M. Lucassen, A.V. Ranchor, I.M. van Langen; Groningen, Netherlands*	**C15.6** **Clinical and experimental evidence establish a link between *KIF7* and *C5orf42*-related ciliopathies** **Reza Asadollahi***, *J.E. Strauss, M. Zenker, O. Beuing, S. Edvardson, O. Elpeleg, P. Joset, T.M. Strom, D. Niedrist, B. Oneda, S. Azzarello-Burri, M. Papik, A. Baumer, K. Steindl, A. Schinzel, E.T. Stoeckli, A. Rauch; Zurich-Schlieren, Switzerland*	**C16.6** **From whole exome sequencing to functional studies in syndromic microcephaly: using zebrafish for variant testing** **Francesca Cristofoli***, *E.E. Davis, K. Devriendt, H. Peeters, H. Van Esch, J.R. Vermeesch; Leuven, Belgium*	**C17.6** **Novel method reveals a large number of expression quantitative trait loci (eQTLs) influencing transcript levels in a Parent-of-origin fashion** **Aaron F. McDaid**, *T. Esko, L. Franke, Z. Kutalik; Lausanne, Switzerland*	**C18.6** **Common and rare variants associated with kidney stones and biochemical traits** **Asmundur Oddsson***, *P. Sulem, H. Helgason, V. Edvardsson, G. Thorleifsson, G. Sveinbjornsson, E. Haraldsdottir, G.I. Eyjolfsson, O. Sigurdardottir, I. Olafsson, G. Masson, H. Holm, D.F. Gudbjartsson, U. Thorsteinsdottir, O.S. Indridason, R. Palsson, K. Stefansson; Reykjavik, Iceland*
15.00 - 15.30	*Vitamin break / Poster removal / Exhibition*					

Presentations highlighted by an asterisk (*) and a grey background are from Young Investigator Award Finalists.

PROGRAMME MONDAY, JUNE 8

Time	Clyde	Hall 5	Hall 2	Lomond	Hall 1	Forth	Gala
15.30 - 17.00	**WS10. Practical Bioinformatics Whole Exome Sequence analysis** Organiser: P. Robinson	**WS11. Dysmorphology 2*** Organisers: D. Donnai, S. Douzgou	**WS12. Reproductive genetics** Organiser: J. Vermeesch	**WS13. Clinical Cancer Genetics*** Organisers: M. Genuardi, D. Stoppa-Lyonnet	**WS14. Copy Number Variant Interpretation and Classification*** Organisers: N. de Leeuw, C. van Ravenswaaij-Arts	**WS15: Genome Browser UCSC*** Organiser: R. Kuhn	**WS16. Research, clinic and everyday life: new roles for patients and citizens** Organisers: H. Kääriäinen, T. Vrijenhook
	No description available as per date of printing.	The organisers of the dysmorphology workshop invite clinicians to submit rare known and unknown cases with dysmorphic syndromes before the workshop. Please bring a short case presentation on a USB stick from 15:00 - 15:30 hrs to the lecture room. Maximum time for presentation: 5'.	In this workshop an overview will be provided on the status of different approaches for embryo freezing, preimplantation genetic diagnosis and preimplantation genetic aneuploidy screening. Participants who want to bring forward own work can contact the organizer. We will engage in panel discussion on the current status of the field, the challenges and discuss the likely clinical future.	This workshop will provide a forum for ESHG meeting attendants involved in clinical cancer genetics practice. We will share and discuss peculiar cases to highlight unusual aspects of known syndromes or to gain insights and advice on unsolved issues related to diagnosis, counseling, follow up, or laboratory findings. Presentations made on a voluntary basis.	The aim of this workshop is to focus on various aspects of copy number variant (CNV) interpretation and classification in a diagnostic setting. We will talk about multi-, intra- and intergenic CNVs detected by genome wide array analysis, but also CNV detection in Whole Exome Sequencing data will be included. We will use illustrative cases from our own diagnostic laboratories to have an interactive discussion on the more challenging findings, including low-penetrant, recurrent Copy Number Variants (CNVs) and structurally rearranged chromosomal imbalances as well as patients with compound heterozygous variants in a recessive disease gene. We will have an app-based feedback system available for this interactive session, so please bring your smart phone, tablet or laptop.	The UCSC Genome Browser is a widely used visualization tool for genomic information. This workshop will demonstrate how to upload your own information, including output from high-throughput sequencing experiments; intersect data from multiple tables, including your own; and interpret variants using the Variant Annotation Integrator. The Genome Browser-in-a-Box may be downloaded to bring the full functionality of the Genome Browser to the desktop, eliminating the need to upload potentially sensitive data. Participants should bring a laptop.	This workshop acknowledges the revolution in clinical genetics and genetic research due to technical innovations, genetics becoming part of nearly every medical specialty, and patients and citizens becoming more active in getting and producing health related data. Short presentations aim at raising discussion focusing on how to benefit from new developments and possibly direct them.
17.00 - 17.30		*Coffee break / Poster removal / Exhibition*					

Detailed Workshop programmes (when submitted by the organisers) can be found in the "ESHG Bulletin" in the conference bag.

*Interactive workshops - your input is solicited. See details in the ESHG Bulletin for more information.

In selected workshops, a voting system will be made available to the audience. Connect to the wifi „voting_*workshopnumber" (e.g. „voting_ws14") with any WIFI-capable device (laptop, tablet, phone) and open an internet browser. The voting form will be displayed accordingly.*

PROGRAMME MONDAY, JUNE 8

Time	Clyde	Hall 5	Hall 2	Lomond	Hall 1
17.30 - 19.00	**S13 Therapeutic strategies for genetic diseases** Chair: G. Neri, J. Sampson	**ES09 Mutation prediction tools** Chair: P. Robinson, S. Abbs	**S14 Genome regulation** Chair: G. Houge, J. Blow	**S15 Somatic mutation detection and interpretation** Chair: A. Rauch, R. Butler	**S16 Evolution and disease** Chair: A. Read, H. Dollfuss
17.30	**S13.1** **Nonsense suppression strategies to treat ocular malformations** **Cheryl Y. Gregory-Evans**, *X. Wang, K.M. Wasan, K. Gregory-Evans; Vancouver, Canada*	**ES9.1** **Functional prediction of DNA sequence changes** **Sean Tavtigian**; *Salt Lake City, United States*	**S14.1** **Spatial organization of genomes** **Bing Ren**; *La Jolla, United States*	**S15.1** **How much of *de novo* is meiotic?** **Pawel Stankiewicz**; *Houston, United States*	**S16.1** **The human Y chromosome in evolution and disease** **Chris Tyler-Smith**; *Hinxton, United Kingdom*
18.00	**S13.2** **Therapeutic targeting of the mTOR pathway** **Julian R. Sampson**; *Cardiff, United Kingdom*	**ES9.2** **Protein structures to advance therapeutic discoveries** **Wyatt Yue**; *Oxford, United Kingdom*	**S14.2** **Regulatory RNAs and eQTLs** **Manolis Dermitzakis**; *Geneva, Switzerland*	**S15.2** **Selfish mosaicism: impact of somatic mutations occurring in the paternal germline** **Anne Goriely**; *Oxford, United Kingdom*	**S16.2** **Ancient pathogen genomics of re-emerging infectious diseases** **Johannes Krause**; *Jena, Germany*
18.30	**S13.3** **The Use of AAV in Cardiomyopathy** **Hélène Puccio**; *Illkirch, France*		**S14.3** **Mutations in regulatory domains in human disease** **Stanislas Lyonnet**, *C.T. Gordon, S. Benko, A. Pelet, J. Amiel; Paris, France*	**S15.3** **Somatic mutations in monozygotic twins** **Eline Slagboom**; *Leiden, Netherlands*	**S16.3** **Evaluating human genetic (and epigenetic) adaption to pathogen pressures** **Lluis Quintana-Murci**; *Paris, France*
19.30	*Networking Party at the Merchant Square (at own expense)*				

SCIENTIFIC

SCIENTIFIC PROGRAMME

Tuesday, June 9, 2015

PROGRAMME

Time	Clyde
09.00 - 10.30	**PL3 Interactive Debate: Should all geneticists have their genome sequenced?** *Moderators:* *M.C. Cornel,* *J. Veltman*
	Panelists: • **Carsten Bergmann**; *Freiburg, Germany* • **Wendy Bickmore**; *Edinburgh, United Kingdom* • **Kate M.D. Bushby**; *Newcastle upon Tyne, United Kingdom* • **Heidi Howard**; *Uppsala, Sweden* • **Gijs Santen**; *Leiden, The Netherlands* *A voting system will be made available to the audience. Connect to the wifi „voting_pl3" with any WIFI-capable device (laptop, tablet, phone) and open an internet browser. The voting form will be displayed accordingly.*
10.30 - 11.00	*Coffee Break in Hall 5*

PROGRAMME TUESDAY, JUNE 9

Time	Clyde	Hall 2	Lomond	Hall 1	Forth
11.00 - 12.30	**C19 Diagnostic NGS** Chair: M. Breuning, S. Ellard	**C20 Current issues in genet(h)ics** Chair: A. Middleton, P. Borry	**C21 Multiple congenital anomaly syndromes** Chair: J. McGaughran, R. Newbury-Ecob	**C22 Statistical genetics and bioinformatics** Chair: A. Devereau, I. Prokopenko	**C23 Movement and motor disorders** Chair: D. Lev, S. Mehta
11.00	C19.1 **Large-scale, high-throughput testing of cancer predisposition genes using the TruSight Cancer panel** **Shazia Mahamdallie**, *E. Ruark, A. Fowler, M. Münz, V. Cloke, A. George, S. Seal, G. Lunter, N. Rahman, Mainstreaming Cancer Genetics Programme; London, United Kingdom*	C20.1 **Ethical and legal challenges of genomic cloud computing** **Edward S. Dove**, *M. Phillips, Y. Joly, A. Tassé, B.M. Knoppers; Montreal, Canada*	C21.1 **Recurrent de novo p.Arg83Cys mutations in the acetyl CoA binding site of NAA10 are associated with atypical Cornelia de Lange syndrome** **Morad Ansari**, *N. Akawi, H. Bengani, A.M. Meynert, I. Parenti, J. Pozojevic, D.C. Soares, C. Martin, A. Blatnik, H. Kayserili, S. Avci, S. Joss, K. Tatton-Brown, F. Elmslie, M. Suri, S. Mansour, K.S. Wendt, E. Watrin, F.J. Kaiser, M.S. Taylor, M.E. Hurles, D.R. FitzPatrick; Edinburgh, United Kingdom*	C22.1 **The secrets of GWAS are written in the reads** **Claes Wadelius**, *M. Cavalli, G. Pan, H. Nord, O. Wallerman, E. Wallén Arzt, O. Berggren, I. Elvers, M. Eloranta, L. Rönnblom, K. Lindblad Toh; Uppsala, Sweden*	C23.1 **TBK1 mutations cause amyotrophic lateral sclerosis and fronto-temporal dementia** **Thomas Wieland***, *A. Freischmidt, B. Richter, W. Ruf, V. Schäffer, K. Müller, N. Marroquin, F. Nordin, A. Hübers, P. Weydt, S. Pinto, R. Press, J. Dorst, E. Graf, T. Meyer, A.S. Winkler, J. Winkelmann, M. de Carvalho, D.R. Thal, M. Otto, T. Brännström, A.E. Volk, P. Sarvari, D.Y.R. Stainier, P. Kursula, K.M. Danzer, P. Lichtner, I. Dikic, T. Meitinger, A.C. Ludolph, P.M. Andersen, J.H. Weishaupt, T.M. Strom; Neuherberg, Germany*
11.15	C19.2 **Increasing accessibility and affordability of genetic testing through targeted clinical exome sequencing** **Stephen Abbs**, *H. Martin, K. Brugger, I. Delon, O. Spasic-Boskovic, G. Sagoo, F. Rodger, R. Littleboy, S. Mehta, S. Park, R. Armstrong, G. Woods, S. Holden, J. Campbell, C. Bennett, R. Sandford, E. Maher; Cambridge, United Kingdom*	C20.2 *In utero* **treatment of Down syndrome - proceed with care** **Guido M.W.R. de Wert**, *W.J. Dondorp; Maastricht, Netherlands*	C21.2 **STAG1 haploinsufficiency is responsible for a new cohesinopathy with intellectual disability and characteristic facial features in four unrelated individuals** **Daphné Lehalle**, *A. Masurel-Paulet, A. Mosca-Boidron, M. Deardorff, H. Olivie, J. Thevenon, M. Willemsen, C. Zweier, A. Rauch, C. Gilissen, P. Callier, C. Thauvin-Robinet, L. Faivre; Dijon, France*	C22.2 **Optimal ancestry-matched imputation of GWAS association summary statistics using large reference panel of sequenced individuals** **Sina Rüeger**, *Z. Kutalik; Lausanne, Switzerland*	C23.2 **PMPCA Mutations cause Abnormal Mitochondrial Protein Processing in Patients with Non-Progressive Cerebellar Ataxia** *R. Jobling, M. Assoum, O. Gakh, S. Blaser, J.A. Raiman, C. Mignot, E. Roze, A. Dürr, A. Brice, N. Lévy, C. Prasad, T. Paton, A.D. Paterson, N. Roslin, C.R. Marshall, J. Desvignes, N. Roëckel-Trevisiol, S. Scherer, G.A. Rouleau, A. Mégarbané, G. Isaya,* **Valerie Delague**, *G. Yoon; Marseille, France*
11.30	C19.3 **The RD-Connect platform includes the first 360 analysed exomes linked to phenotypic data and integrates user-friendly tools for rare disease variant prioritization** *D. Piscia,* **Steven Laurie***, *A. Cañada, J. Fernández, C. Kingswood, J. Desvignes, M. Thompson, R. Kaliyaperumal, E. van der Horst, S. Lair, P. Sernadela, A. Topf, I. Zaharieva, M. Girdea, M. Brudno, A. Blavier, R. Thompson, H. Lochmüller, M. Bellgard, J. Paschall, P. Lopes, J. Oliveira, M. Roos, P. ,t Hoen, V. de la Torre, A. Valencia, D. Salgado, C. Béroud, S. Beltran, I. Gut; Barcelona, Spain*	C20.3 **Should children's carrier results be reported following diagnostic WES/WGS?** **Danya F. Vears***, *K. Sénécal, J. Massie, P. Borry; Leuven, Belgium*	C21.3 **Wiedemann-Steiner Syndrome: Expanding the phenotypic spectrum associated with KMT2A (MLL) mutations** **Wendy D. Jones***, *M. McEntagart, C. Deshpande, T. Deciphering Developmental Disorders Project, M.A. Simpson, M. Hurles, J. Barrett; Cambridge, United Kingdom*	C22.3 **A novel method and software tool for genome-wide multi-phenotype analysis of rare variants** **Marika Kaakinen***, *R. Mägi, K. Fischer, M. Järvelin, A.P. Morris, I. Prokopenko; London, United Kingdom*	C23.3 **Spinocerebellar ataxia type 28, from molecular hypothesis to human therapy** *F. Maltecca, E. Baseggio, F. Consolato, D. Mazza, P. Podini, A. Puliti, F. Codazzi, A. Quattrini,* **Giorgio Casari**; *Milan, Italy*

Presentations highlighted by an asterisk (*) and a grey background are from Young Investigator Award Finalists.

Time	Clyde	Hall 2	Lomond	Hall 1	Forth
cont.	**C19 Diagnostic NGS**	**C20 Current issues in genet(h)ics**	**C21 Multiple congenital anomaly syndromes**	**C22 Statistical genetics and bioinformatics**	**C23 Movement and motor disorders**
11.45	**C19.4** **Copy Number Analysis using Exon-level aCGH and Exome Sequencing in over 3,000 Parent-Offspring Trios from the Deciphering Developmental Disorders Project** **Tomas W. Fitzgerald***, *J. McRae, D. de Vries, M. Hurles;* *Cambridge, United Kingdom*	**C20.4** **Informed consent for whole exome sequencing in pediatric disease diagnostics: parental decision-making processes, their ethical relevance and implications for policy development** **Candice Cornelis***, *M. van Summeren, I. Bolt, A. Tibben, W. Dondorp, M. van Haelst, A. Bredenoord, M. Düwell, N. Knoers;* *Utrecht, Netherlands*	**C21.4** **Mutations in the endothelin receptor type A cause mandibulofacial dysostosis with alopecia via a maxillary to mandibular transformation** **Chris T. Gordon**, *N. Weaver, R. Zechi-Ceide, E. Madsen, A. Tavares, M. Oufadem, Y. Kurihara, I. Adameyko, A. Picard, S. Breton, S. Pierrot, M. Biosse-Duplan, N. Voisin, C. Masson, C. Bole-Feysot, P. Nitschké, M. Delrue, D. Lacombe, M. Guion-Almeida, P. Moura, D. Garib, A. Munnich, P. Ernfors, R. Hufnagel, R. Hopkin, H. Kurihara, H. Saal, D. Weaver, N. Katsanis, S. Lyonnet, C. Golzio, D. Clouthier, J. Amiel;* *Paris, France*	**C22.4** **The Exomiser suite for exome prioritization of human disease genes** **Damian Smedley**, *J. Jacobsen, S. Kohler, A. Oellrich, K. Wang, C. Mungall, N. Washington, S. Bauer, D. Seelow, P. Krawitz, C. Gilissen, M. Haendel, S.E. Lewis, P.N. Robinson;* *Cambridge, United Kingdom*	**C23.4** **Homozygous truncating mutations in WDR73 cause a severe nephrocerebellar syndrome, part of the Galloway Mowat syndrome spectrum** **Emma L. Baple***, *R. Jinks, E. Puffenberger, B. Harding, P. Crino, A. Fogo, O. Wenger, H. Wang, B. Xin, A. Koehler, M. McGlincy, L. Tran, M. Provencher, J. Smith, S. Al Turki, B. Chioza, R. Maroofian, G. Harlalka, M. Hurles, S. Gerety, H. Cross, A. Heaps, M. Morton, L. Stempak, F. Hildebrandt, C. Sadowski, J. Zaritsky, K. Campellone, H. Morton, K. Strauss, A.H. Crosby;* *Southampton, United Kingdom*
12.00	**C19.5** **Small exonic CNVs as causes of primary immunodeficiencies** **Asbjørg Stray-Pedersen**, *H.S. Sorte, T. Gambin, P.S. Samarakoon, S. Gu, L.R. Forbes, I. Chinn, Z.H.C. Akdemir, O.K. Rødningen, B. Yuan, P.M. Boone, A. Patel, S.J. Penney, W. Wiszniewski, S.N. Jhangiani, D. Muzny, R.A. Gibbs, R. Lyle, J.S. Orange, J.R. Lupski;* *Houston, United States*	**C20.5** **A Human Rights Approach to International Data Sharing?** **Bartha Maria Knoppers**, *E. Dove;* *Montreal, Canada*	**C21.5** **Mutations in transcription factor ZBTB20 cause tall stature, macrocephaly, cognitive deficits, diabetes, progressive muscle wasting and deafness** **Viviana Cordeddu***, *B. Redeker, E. Stellacci, A. Jongejan, A. Fragale, T. Bradley, M. Anselmi, A. Ciolfi, S. Cecchetti, V. Muto, L. Bernardini, M. Azage, D. Carvalho, A. Espay, A. Male, A. Molin, R. Posmyk, C. Battisti, A. Casertano, D. Melis, A. van Kampen, F. Baas, M. Mannnens, G. Bocchinfuso, L. Stella, M. Tartaglia, R. Hennekam;* *Rome, Italy*	**C22.5** **Allele specific expression reveals common and rare regulatory variation acting in human substantia nigra and putamen** **Karishma D'Sa***, *A. Ramasamy, S. Guelfi, J. Vandrovcova, J.A. Botía, D. Trabzuni, J.R. Gibbs, C. Smith, M. Matarin, V. Varghese, P. Forabosco, The UK Brain Expression Consortium (UKBEC), J. Hardy, M.E. Weale, M. Ryten;* *London, United Kingdom*	**C23.5** **Mutations in PDE10A, resulting in a loss of PDE10A activity cause a hyperkinetic movement disorder in humans and in a mouse model.** **Eamonn G. Sheridan**, *R. Hinttala, J. Uusimaa, M. Kurian, N. Brandon, C. Diggle;* *Leeds, United Kingdom*
12.15	**C19.6** **Whole genome sequencing as a clinical diagnostic tool for heterogeneous Mendelian disease** **Jamie M. Ellingford***, *S. Barton, S. Bhaskar, S.G. Williams, P.I. Sergouniotis, J. O'Sullivan, J.A. Lamb, R. Perveen, G. Hall, W.G. Newman, P.N. Bishop, S.A. Roberts, S. Bayliss, S.C. Ramsden, A.H. Nemeth, G.C.M. Black;* *Manchester, United Kingdom*	**C20.6** **What's in it for me? A critical analysis of the notion of personal utility in genomic testing** **Eline M. Bunnik***; *Rotterdam, Netherlands*	**C21.6** **Mutations impairing GSK3-mediated MAF phosphorylation cause cataract, deafness, intellectual disability, seizures, and a Down syndrome-like facies.** **Marcello Niceta***, *E. Stellacci, K.W. Gripp, G. Zampino, M. Kousi, M. Anselmi, A. Traversa, A. Ciolfi, D. Stabley, A. Bruselles, V. Caputo, S. Cecchetti, S. Prudente, M.T. Fiorenza, C. Boitani, N. Philip, D. Niyazov, C. Leoni, T. Nakane, K. Keppler-Noreuil, S.R. Braddock, G. Gillessen-Kaesbach, A. Palleschi, P.M. Campeau, B.H.L. Lee, C. Pouponnot, L. Stella, G. Bocchinfuso, N. Katsanis, K. Sol-Church, M. Tartaglia;* *Rome, Italy*	**C22.6** **Evidence for directional dominance on complex traits relating to size and cognition in a wide range of human populations** **Peter Joshi***, *T. Esko, H. Matteson, N. Eklund, I. Gandin, A. Jackson, T. Nutile, C. Schurmann, O. Polasek, J.F. Wilson;* *Edinburgh, United Kingdom*	**C23.6** ***PLP1* mutations affecting *PLP1/DM20* alternative splicing causes Hypomyelination of Early Myelinating Structures** **Sietske H. Kevelam***, *J.R. Taube, R.M.L. van Spaendonk, E. Bertini, K. Sperle, M. Tarnopolsky, D. Tonduti, E.M. Valente, L. Travaglini, E.A. Sistermans, G. Bernard, C.E. Catsman-Berrevoets, C.D.M. van Karnebeek, J.R. Østergaard, R.L. Friederich, M. Fawzi, J.H. Schieving, M. Tarailo-Graovac, S. Orcesi, M.E. Steenweg, C.G.M. van Berkel, Q. Waisfisz, T.E.M. Abbink, M.S. van der Knaap, G.M. Hobson, N.I. Wolf;* *Amsterdam, Netherlands*
12.30 - 13.30	*Lunch Break in Hall 5*				

PROGRAMME TUESDAY, JUNE 9

Time	Clyde
13.30 - 14.15	**Plenary Session PL4** **Mendel Lecture** Chair: F. Ramos, B. Wirth
13.30	**PL4.1** **The neurexin enigma - from synapse formation to schizophrenia** **Thomas Südhof**; *Stanford, United States* *Introduction by Brunhilde Wirth*
14.15 - 15.45	**Plenary Session PL5** **ESHG Award and Closing Session** Chair: F. Ramos, B. Wirth
14.15	**PL5.1** **ESHG Award Lecture** **Svante Pääbo**; *Leipzig, Germany* *Laudation by Helena Kääriäinen*
15.00	**Awards Ceremony** **ESHG Education Award awarded to Heather Skirton** *Laudation by Aad Tibben* **EJHG-NGP Awards** **ESHG Young Investigator Awards:** • **ESHG Young Investigator Awards** for Outstanding Science • **Isabelle Oberlé Award** for an outstanding presentation in the field of genetics of mental retardation • **Lodewijk Sandkuijl Award** for an outstanding presentation in the field of complex disease genetics and statistical genetics • **Vienna Medical Academy Award** for an outstanding presentation in translational genetic research/therapy of genetic diseases • **Mia Neri Award** for an outstanding presentation in the field of cerebral cancer **ESHG Poster Awards in clinical research and basic science** **Closing**

At the end of the Closing Plenary Session, three Apple iPads mini will be drawn among the attendees, who have had their badges scanned at the entrance of the Clyde Auditorium.

PROGRAMME

PROGRAMME INFORMATION

CORPORATE SATELLITE MEETINGS

BUSINESS MEETINGS

YOUNG INVESTIGATOR AWARD CANDIDATES

POSTER AWARD CANDIDATES

INFORMATION

CS01 Complete Genomics, Saturday, June 6, 2015, 12.15-13.45 hrs, Boisdale, Ground Floor — Stands # 364 & 436

Advances in Genome Sequencing Technologies

Complete Genomics, a leader in whole human genome sequencing, has used its proprietary technology to sequence over 20,000 whole human genomes. Complete has now developed a new generation of products using its high-throughput sequencing technology that will redefine and fully enable genome sequencing. Join us for lunch and a preview of the first truly integrated sequencing solution with game-changing performance, full automation and scalability. Come learn about our new and future offerings designed to address the needs of the research and clinical community.

CS02 Personalis, Saturday, June 6, 2015, 12.15-13.45 hrs, Alsh 1, Ground Floor — Stand # 338

Advancing Discovery and Diagnostics for Inherited Disease and Cancer: Why Accuracy Matters

Speakers:
- *Jon Beck[1], FRCPath*
- *Deanna Church[1], PhD*
- *Gemma Chandratillake[1], MPhil, PhD, MS CGC*

[1]Personalis, Inc., Menlo Park, CA, USA

Personalis is a leading genomics-based clinical diagnostic laboratory that provides researchers and clinicians advanced sequencing and interpretation services for inherited genetic disease and cancer. Our focus on accuracy extends from our sequencing platform through our bioinformatics pipeline and biomedical interpretation. Examples of how we are addressing issues of accuracy in genomic sequencing and interpretation will be discussed. Cases in which our augmented exome test proved successful in making diagnoses of Mendelian disease that would have been missed by conventional exome and/or gene panel testing will be presented.

Guest Presentation:
Mosaic Long QT in the Neonatal Intensive Care Unit
Euan Ashley, D.Phil., Associate Professor, Medicine and Genetics; Director, Stanford Center for Inherited Cardiovascular Disease, Stanford University School of Medicine, Stanford, CA, USA
It is increasingly appreciated that augmented exomes can cover the non-coding content once accessible only by WGS. Such platforms also offer sufficient sequencing depth to confidently call mosaic variants and this can be critical clinically. This presentation will focus on such a case from the neonatal intensive care unit at Lucile Packard Children's Hospital.

CS03 Sistemas Genómicos, Saturday, June 6, 2015, 12.15-13.45 hrs, Carron 1, First Floor — Stand # 224

Deciphering the genetic bases of intellectual disability. From the beginning to the end.

Chaired by: Alejandro Romera from the Medical Genetics Unit of Sistemas Genómicos

Diagnosing the origin of intellectual disability (ID) is still a long and complex process that affects more than half the patients with a clinical diagnosis of ID. With Next-Generation Sequencing, it is now possible to study simultaneously all the genes associated with autosomal and X chromosome-linked disorders with ID to offer a prognosis and adequate genetic counselling to the families. This symposium will guide you on how to develop your own Next-Generation Sequencing analyses in order to study the genetic origin of intellectual disability and on how to interpret your data.

How to develop and prepare your own Next-Generation Sequencing studies
Roger Rovira, Market Development Manager for the UK, Sistemas Genómicos

Intellectual disability as an example
Dan Diego, Medical Genetics Lab Manager, Sistemas Genómicos

Analysis and interpretation of your data
Oscar Rodriguez, Department of Bioinformatics, Sistemas Genómicos

PROGRAMME CORPORATE SATELLITES

CS04 Thermo Fisher Scientific, Sunday, June 7, 2015, 11.45-13.15 hrs, Boisdale, Ground Floor — Stand # 320

Advances in Applied Biosystems™ and Ion Torrent™ technologies transforming human genetics research

The expectation from our customer is that every time we launch a new technology, we look also for ways to expedite their analysis so they can focus on what they do best: connecting the dots and understand the science behind their research.
In this corporate satellite you'll learn how we connect scientists and their data using the latest Applied Biosystems and Ion Torrent products for genetic analysis and the Thermo Fisher Cloud™.
Our panel of speakers will discuss the latest product updates, with Mike Lelivelt Director of Bioinformatics & Software Products at Thermo Fisher Scientific hosting the session and connecting the technologies to the cloud software solutions.

Participants in the panel discussion include:
- *Dr. Andy Felton, Ion Torrent Product Management Leader, Thermo Fisher Scientific*
- *Dr. Martin Storm, Applied Biosystems™ Product Management Leader, Thermo Fisher Scientific*
- *Inger Jonasson, Uppsala Genome Center, Sweden*
- *Damien Luk, Applied Biosystems Product Management Leader, Thermo Fisher Scientific*

CS05 Affymetrix, Sunday, June 7, 2015, 11.45-13.15 hrs, Dochart, First Floor — Stand # 530

Karyotyping is not enough...

Karyotyping is not enough for constitutional cytogenetics; this fact has been well established as newer technologies have helped gain further insight into the human genome. These newer technologies are also having a profound impact on unravelling the cancer genome in both liquid and solid tumors.

Hear users' views on how Affymetrix® CytoScan® Cytogenetics Suite is enabling researchers to analyze postnatal and prenatal constitutional samples with more confidence than with any other traditional or array-based technology, and how OncoScan® FFPE Assay Kit facilitates whole-genome copy number analysis for accurate tumor profiling of highly degraded FFPE samples.

Speakers include:
Paul Boutros, PhD, Department of Medical Biophysics, University of Toronto, Canada
Identifying Drivers and Biomarkers of Localized Prostate Cancer

Oskar A. Haas, Univ.-Prof. Dr., Childrens Cancer Research Institute, Vienna, Austria
Array Analyses of Childhood Leukemia: Copy Number Aberrations and Beyond

Tord Jonson, PhD, Department of Clinical Genetics, University Hospital, Lund, Sweden
CytoScan HD Analysis in Lund - Yesterday, Today, and Tomorrow

Lunch and refreshments will be provided. Spaces are limited; please arrive early to avoid disappointment. Visit us on stand # 530.

CS06 Sophia Genetics, Sunday, June 7, 2015, 11.45-13.15 hrs, Alsh 1, Ground Floor — Stand # 230

Routine NGS-based Diagnostics: How to achieve top analytical performance and guarantee quality

Recipes of a clinical grade algorithm to support routine NGS-based diagnostics
Dr Zhenyu Xu, CTO, Sophia Genetics, Lausanne, Switzerland
Sufficient coverage of target region is a well known prerequisite for accurate variant detection. In addition, issues related to variants exposed to the end of reads, proper trimming of the primer sequences or repetitive genomic regions also introduce limitations and biases. Understanding these limitations in the combination of sequencing platform, enrichment technology, gene panel and sample type are key to ensure accurate variant detection. Here we introduce our platform Sophia DDM that supports a clinical grade algorithm containing over 150 paths or pipelines, taking into account the above limitations.

Validation of a routine NGS diagnostic test using Sophia DDM data analysis platform
Dr Christine Bell, Clinical Scientist, Department of Medical Genetics, NHS Grampian, Aberdeen, Scotland
Method transfer from Sanger to NGS is desired to increase throughput of BRCA1/2 sequencing in a diagnostic setting. We have used 100 samples with distinct sequence variations using the BRCA1/2 Ion AmpliSeq panel on the PGM to validate this methodology. Analysis has been performed using Sophia DDM - specifically tailored to this technology configuration. Results of this validation will be presented. Sophia DDM is user friendly and saves considerable time in analysis. The system also benefits from version control, particularly useful for diagnostic labs in view of accreditation requirements.

CS07 QIAGEN, Sunday, June 7, 2015, 11.45-13.15 hrs, Carron 1, First Floor — Stand # 746

Advancements in ccfDNA isolation: standardized methods for non-invasive prenatal testing and oncology

Isolation and analysis of circulating cell free DNA (ccfDNA) in plasma has become an important tool for non-invasive prenatal testing and oncology. However, several challenges remain due to the limited concentration of ccfDNA in samples. Good handling practices as well as standardized and rapid ccfDNA extraction protocols can help overcome some of these challenges. Here, we present a newly developed automated solution for extracting ccfDNA from plasma samples on the QIAsymphony® SP. The new ccfDNA extraction protocol on the QIAsymphony® SP revealed a highly efficient recovery of total ccfDNA and fetal ccfDNA compared to the QIAamp® Circulating NA Kit. Linearity was shown for ccfDNA yield from 2-6 ml plasma input. A high efficiency for extraction of low copy numbers of ccfDNA is given. Compatibility of eluates to manifold downstream assays was confirmed for different PCR and methylation-sensitive assays, library preps and subsequent NGS-based analysis of chromosomes. Concluding, the novel protocol enables automated ccfDNA recovery from up to 4 ml plasma and up to 96 samples per QIAsymphony® SP run in 6 hours combined with high recovery efficiency.
The applications presented here are for research use only. Not for use in diagnostic procedures.

Speaker:
Marco Polidori, PhD in Cell Biology, Application Scientist, LifeTechnologies (3 years), Global Product Manager, Sample Preparation Technologies, QIAGEN GmbH (4 years)

CS08 Complete Genomics, Sunday, June 7, 2015, 15.30-17.00 hrs, Boisdale, Ground Floor — Stands # 364 & 436

Clinical Applications of Whole Genome Sequencing

Whole genome sequencing (WGS) is having a major impact on our understanding of health and disease. Complete Genomics continues to collaborate with key opinion leaders in the field of clinical genomics to demonstrate the potential improvements that whole genome sequencing applications may offer to physicians, patients, and payers. The primary goal of these studies is to compare WGS to the standard of care and determine the impact on clinical and economic outcomes. In this satellite symposium, you will hear from clinical researchers using whole genome sequencing and their views on the potential to improve patient care.

CS09 AstraZeneca, Sunday, June 7, 2015, 15.30-17.00 hrs, Dochart, First Floor — Stand # 548

BRCA testing in the new therapeutic era

Welcome and introduction, *Nazneen Rahman*[1]

Who to test?, *Dominique Stoppa-Lyonnet*[2]

How to test?, *Angela George*[1]

Challenges in the classification and clinical management of variants of uncertain clinical significance in BRCA1 and BRCA2, *Encarna Gómez García*[3]

Pooling data: The BRCA Challenge of the Global Alliance for Genomics and Health, *John Burn*[4]

Q&A, *All*

Summary and close, Nazneen Rahman[1]

[1]The Royal Marsden NHS Foundation Trust, London, UK
[2]Curie Institute and University of Paris Descartes, Paris, France
[3]Maastricht University Medical Centre, Maastricht, The Netherlands
[4]Institute of Genetic Medicine, International Centre for Life, Newcastle upon Tyne, UK

CS10 Multiplicom, Sunday, June 7, 2015, 15.30-17.00 hrs, Alsh 1, Ground Floor — Stand # 634

Integrated germline MASTR™ solutions for clinical diagnostics and Clarigo™ solution for NIPT

Cost effective integration of precise diagnostic tests in clinical routine practice that are easy to implement, with a fast turnaround time continue to be challenging for molecular genetics laboratories.
In our satellite symposium, we will share the experience when implementing germline MASTR tests and Clarigo: a decentralized lab solution for non-invasive prenatal aneuploidy testing in combination with most currently used massively parallel sequencing systems.

Targeted next generation sequencing of 51 genes involved in primary electrical disease predisposing for Sudden Cardiac Death
Dorien Proost, University of Antwerp, Department Medical Genetics, Antwerp, Belgium

Clarigo™: a decentralized lab solution for non-invasive prenatal aneuploidy testing
Prof. Dr. Jurgen Del-Favero, CTO, Multiplicom N.V., Niel, Belgium

Breast and Ovarian Cancer diagnosis: The Next Level
Noor Remmerie, PhD, R&D Manager, Multiplicom N.V., Niel, Belgium

PROGRAMME CORPORATE SATELLITES

CS11 NuGEN Technologies, Sunday, June 7, 2015, 15.30-17.00 hrs, Carron 1, First Floor — Stand # 650

An efficient method for identifying mutations and gene fusions in clinically important samples using targeted RNA sequencing

Single Primer Enrichment Technology (SPET), a novel approach for targeted resequencing of genomic DNA or cDNA for targeted RNA analysis, is suitable for a wide range of target sizes from a few kilobases to over 10 megabases. The method uses a single targeting probe that hybridizes to the target region and then extends through the region of interest. The approach eliminates the difficulty of designing specific PCR primers and maintains high specificity of recovered target sequences in the final library. The application of the technique is highly flexible and suitable for use in the targeted analysis of a wide range of genomic markers including mutations, SNP's, indels, gene fusions, alternately spliced transcripts and CNVs. We will describe how SPET is employed to detect variants from gDNA derived from fresh and formalin-fixed paraffin embedded (FFPE) samples using a cancer panel design of 344 cancer-related genes and will show data for detection of known and novel gene fusions using a panel of 500 cancer genes. Lastly, the application of SPET to characterize gene fusions potentially involved in the biogenesis of sarcomas (malignant soft tissue tumors) will be discussed.

Presenters:
- *Steve Kain, Ph.D., Director, NuGEN Technologies, San Carlos, CA, USA*
- *Bastiaan Tops, Ph.D., Clinical Scientist in Molecular Pathology, UMC St Radboud, Nijmegen, the Netherlands*
- *Klaas Kok, Ph.D., Scientist, Department of Genetics, University of Groningen Medical Center, Groningen, the Netherlands*

CS12 Illumina, Sunday, June 7, 2015, 19.00 – 20.30 hrs, Boisdale, Ground Floor — Stand # 726

Unlocking the Genome: Advancing our understanding of human genetics

Advances in Illumina genomic technologies are catalyzing an unprecedented explosion in the knowledge of genetics and its role in disease. Many groups are utilizing arrays and next generation sequencing technologies to study genes, exomes and even large scale whole genome sequencing projects. Please join us as we highlight some of these groundbreaking developments in human genetics and review the latest advancements in our portfolio of genomic solutions.

Complimentary wine and cheese will be served.

Pre-registration is encouraged, please plan to arrive early as space is limited.

CS13 CENTOGENE, Sunday, June 7, 2015, 19.00 – 20.30 hrs, Dochart, First Floor — Stand # 360

CENTOGENE revolutionizes the clinical interpretation of rare disease genetic variants

CentoMD® is a comprehensive and unique repository of genetic variants, including a huge number of unpublished pathogenic variants gathered from patients worldwide. By analyzing over 2,300 genes across a multi-cultural and ethnically diverse population, this extensive database enables physicians, researchers and geneticists to search, select and filter through variations in genes, genetic transcripts and mutations, tailoring their search to specific medical needs.

With CentoMD®, physicians now have a virtual encyclopedia of genetic information at their fingertips, allowing them to diagnose and treat their patients in a much more efficient, speedy and targeted manner.

Diagnosing a patient with a rare disease is a complex task. The majority of detectable genetic variants have up to now not been properly described.

CentoMD® fills an important gap currently existing in the clinical interpretation of novel genetic variants. Previously, it was a monumental task sifting through an enormous amount of medical data for detecting the right genotype/phenotype correlation when diagnosing rare disease cases.

By combining precise clinical genetic information from more than 90 countries with the actual clinical patient case corresponding to that data, CentoMD® greatly standardizes and expedites the medical interpretation of these variants.

CS14 Natera, Sunday, June 7, 2015, 19.00 – 20.30 hrs, Alsh 1, Ground Floor — Stand # 430

2q.11.2 Clinical Experience with over 20,000 SNP-based NIPTs and Prenatal Management

Speaker:
Trudy McKanna, MS, CGC Manager, Medical Science Liaisons for Natera, San Carlos, CA, USA

You are cordially invited to this Natera lecture about the latest advances in non-invasive prenatal screening.

CS15 Thermo Fisher Scientific, Sunday, June 7, 2015, 19.00 – 20.30 hrs, Carron 1, First Floor Stand # 320

CRISPR-based Genome Editing Tools: New Applications and Streamlined Workflows

Jon D. Chesnut, PhD - Thermo Fisher Scientific, Carlsbad, CA, USA

CRISPR-Cas9 is rapidly evolving as the tool of choice for genome editing in mammalian cells. The delivery of Cas9 and synthesis of guide RNA (gRNA) remain as steps that limit overall efficiency and general ease of use. Here we describe novel methods for rapid synthesis of gRNA and delivery of Cas9 protein/gRNA complexes into a variety of cells. This workflow enables highly efficient genome editing and biallelic knockout of multiple genes in hard-to-transfect cells in as little as three to four days. The reagent preparation and delivery to cells requires no plasmid manipulation so is amenable for high throughput, multiplexed genome-wide cell engineering.

Further, we will show data using lentivirus-based CRISPR delivery for high-throughput screening of mammalian cell populations. We are creating gene family-specific arrayed libraries of CRISPR-lenti particles that will enable high throughput, arrayed gene knockout screens using various cell types.

These two CRISPR-based gene-editing platforms represent the latest in the rapid evolution of editing tools for mammalian genomes by simplifying the cell engineering workflow and providing a pre-designed, ready to use platform for efficient compound screening in mammalian cell lines.

CS16 Agilent Technologies, Monday, June 8, 2015, 11.45-13.15 hrs, Boisdale, Ground Floor Stand # 446

The New Frontier in Human Genetics

Detection of somatic mosaicism in children with multiple primary tumors using molecular-barcoding
Jiannis Ragoussis, Department of Human Genetics, McGill University and Genome Quebec Innovation Centre, Montreal, Canada
Mutations in DICER1 gene are associated with rare cancer syndromes, DICER1 Syndrome, pleuropulmonary-blastoma-(PPB), familial-tumor-and-dysplasia-syndrome (PPB-FTDS). Tissues were screened for DICER1 RNase IIIb mutations using a custom PCR-based array and a novel HaloPlexHS panel incorporating molecular barcodes identifying low-frequency mutations.

The use of chromosomal microarrays in prenatal research
Juan C. Cigudosa, C.S.O, NIMGenetics and Molecular Cytogenetics, Spanish National Cancer Research Center (CNIO), Madrid, Spain
Several publications and guidelines outline the use of microarrays in prenatal clinical research. The design of array-CGH investigating copy-number-variations in fetal-origin-DNA is important. This design should compile conditions of theoretical and practical resolution, and also involve knowledge of the phenotypic impacted for a given CNV to enable clinical research.

Highlighting both copy-number and sequence-variants by a single next generation sequencing assay
Orsetta Zuffardi, Department of Molecular Medicine, University of Pavia and IRCCS Foundation Policlinico San Matteo, Pavia, Italy
Detection of genomic variants is pivotal to highlighting genetic bases of common and rare diseases. WES offers a good alternative for detecting coding SNVs and indels, but less useful to detect CNVs. We present a good compromise to meet this inescapable requirement.

CS17 Oxford Gene Technology, Monday, June 8, 2015, 11.45-13.15 hrs, Dochart, First Floor Stand # 544

The Next Generation of Microarrays: Identifying a Broader Range of Genetic Syndromes Using Exon - Focussed Array Designs

Chair: *James Clough, Executive Vice President Commercial, OGT, Oxford, UK*

Presentations:
Complementing the Medical Exome with Arrays for CNV Detection
Professor Madhuri Hegde, PhD, FACMG, Professor of Human Genetics, Emory University School of Medicine, Atlanta, GA, USA

Combining Advanced Array Design with the Deciphering Developmental Disorders (DDD) Study Data
Duarte Molha, Senior Computational Biologist, OGT, Oxford, UK

While microarrays are now firmly established as the first-tier test for a range of genetic disorders, it is imperative to incorporate the latest genomic research to further drive their utility. OGT is at the forefront of translating this research into advanced array designs that allow the accurate detection of a broader range of genetic syndromes. At this workshop you will hear how Emory University School of Medicine utilises the CytoSure Medical Research Exome array — a novel array design incorporating over 4600 hand-curated, medically-relevant genes — to complement their NGS workflows, providing gold-standard CNV detection at the exon level in rare genetic diseases. In addition, discover how OGT, in partnership with the Wellcome Trust Sanger Institute, has utilised the data from the Deciphering Developmental Disorders (DDD) Study along with most recent updates from the ICCG (formerly ISCA) to develop the new CytoSure Constitutional array — the most advanced array design available for identifying developmental disorders.

PROGRAMME CORPORATE SATELLITES

CS18 Roche Sequencing, Monday, June 8, 2015, 11.45-13.15 hrs, Alsh 1, Ground Floor — Stand # 336

Roche NimbleGen SeqCap Target Enrichment Systems: Empowering translational research in oncology & rare genetic disorders

Join us for a workshop where Dr. Sudipto Das, from the University College in Dublin, Ireland, Dr. David Gonzalez, from the Institute of Cancer Research, London, UK, and Dr. Keith Gomez, from Royal Free London NHS Foundation Trust, London, UK, will present their clinical research applications of targeted sequencing using Roche NimbleGen target enrichment technologies. Focused approaches to the identification and classification of genetic variants are critical to developing informative, reproducible and cost-effective next-generation sequencing methods needed for clinical research applications.

Dr. Das will cover work using SeqCap EZ Choice, SeqCap Epi and a novel amplicon-based enrichment strategy in studying the molecular basis of breast cancer.

Dr. Gonzalez will discuss his work developing a targeted enrichment panel to elucidate the underlying genetic basis of hemato-oncological disease.

Dr. Gomez will focus on using custom target enrichment panels to streamline and understand the genetics of rare inherited bleeding and platelet disorders.

CS19 LGC, Monday, June 8, 2015, 11.45-13.15 hrs, Carron 1, First Floor — Stand # 349

Functional validation of genetic variation in population genomics

Speakers:
Michael Nicholas Weedon, Associate Professor, Royal Devon & Exeter Hospital, University of Exeter Medical School, UK
Using common genetic variants to distinguish between Type 1, Type 2 and monogenic diabetes in young adult patients

Mikael Kubista, TATAA Biocenter, CEO, TATAA Biocenter, Gothenburg, Sweden, and Head of Department, BTU, CAS, Prague, Czech Republic
Quality control and performance assessment of qPCR instruments

Jim Huggett, PhD, Science Leader, Nucleic Acid Metrology, LGC, London Twickenham, UK
dPCR and qPCR application / standardisation / MIQE

CS20 Cartagenia, Monday, June 8, 2015, 15.30-17.00 hrs, Dochart, First Floor — Stand # 330

Implementing NGS in a clinical setting: automation, data-sharing and guidelines

Speakers:
Berivan Baskin, PhD, Uppsala University Hospital, Sweden
Implementing ACMG/AMP guidelines in routine NGS variant assessment

Marielle van Gijn, PhD, University Medical Center Utrecht, the Netherlands
NGS panels in clinical diagnostics: analysis, interpretation, compliance and data-sharing

Jonatan Taminau, PhD, Cartagenia, Belgium
How labs confidently interpret, report and share genomic variants: introduction to the Cartagenia Bench LabTM platform

Running Next Generation Sequencing in clinical genetics, prenatal diagnosis, or somatic variant analysis?

Join our ESHG 2015 seminar or stop by at the Cartagenia booth (#330) to discover how the Cartagenia Bench Lab NGS software platform helps you implement the new ACMG/AMP guidelines in your clinical variant assessment workflow.

- Set up lab report automation and build variant assessment pipelines with ease
- Build an internal knowledge base and integrate public content
- Implement guidelines in a clinically robust fashion – traceable and scalable

Learn how clinical labs use the Bench platform to set up interpretation and reporting pipelines for inherited disease and cancer, and automate their reporting workflow. This seminar will show case studies, discuss how labs can implement the ACMG guidelines on an automated variant assessment and reporting platform, and allow the lab to deal with growing NGS assay volumes.

CS21 Thermo Fisher Scientific, Monday, June 8, 2015, 15.30-17.00 hrs, Boisdale, Ground Floor — Stand # 320

Advances in applications for variant detection & quantitation using Applied Biosystems™ and Ion Torrent™ systems

In this corporate satellite you will hear the practical experiences from leading laboratories on a range of applications including mutation discovery and verification using orthogonal technologies and how they perform routine data interpretation workflows in order to achieve their goals faster, better and with less workload.
Applications to be presented include:

- **Pre-implantation genetic screening on the Ion Torrent sequencing platform**
  Speaker tbc
- **Personalised clinical genomics using Ion Torrent sequencing**
  Dr. Corina Shtir, Enterprise Genomics Group, Thermo Fisher Scientific
- **Using AmpliSeq™ Colon and Lung Panel and QuantStudio™ 3D Genotyping Assays for the detection of rare mutations in cfDNA of lung cancer research samples**
  Dr. Jose Luis Costa, Institute of Molecular Pathology and Immunology, University of Porto, Porto, Portugal
- **Experience using the latest QuantStudio 3/5 Real-Time PCR for fusion transcript detection and quantification**
  Dr. Csab Bodorf, Semmelweis Medical University, Budapest, Hungary
- **Validation of fragment analysis CE-IVD assays for inherited genetic diseases on the 3500DX Genetic Analyser,**
  Dr. Greg Fitzgibbon, Product Development Manager, Elucigene Diagnostics, UK

CS22 QIAGEN Bioinformatics, Monday, June 8, 2015, 15.30-17.00 hrs, Alsh 1, Ground Floor — Stand # 244

Solving the data analysis bottleneck- see how leading sequencing institutions, such as Mt Sinai and the Rigshospitalet NGS core, use QIAGEN Bioinformatics to rapidly move from raw data to valuable insights

Speakers:
- *Lars Jønson. M.Sc., Ph.D., Head of NGS Core Unit, Center for Genomic Medicine, Copenhagen, Denmark*
- *John Martignetti, Associate Professor at the Icahn School of Medicine at Mount Sinai, New York, USA*

DNA and RNA NGS methods make it easy to generate large amounts of data. Making sense of that data, arriving at actionable insights that can inform follow up experiments or recommendations treatment options can be difficult. QIAGEN Bioinformatics, powered by CLC bio, Ingenuity and BIOBASE, provides a complete sample to insight solution that addresses the data analysis bottleneck. Its industry-leading applications for the analysis, interpretation and reporting of biological data work with the most comprehensive and trusted content resources available. It is through the unique combination of content plus content-aware analytics that QIAGEN Bioinformatics consistently delivers real, measurable value through improved false positive and case solve rates. Join our satellite meeting to see first-hand how leading sequencing institutions, such as Mount Sinai and the Rigshospitalet NGS core, are using QIAGEN Bioinformatics to rapidly move from raw data to valuable insights.

CS23 EMQN, Monday, June 8, 2015, 15.30-17.00 hrs, Carron 1, First Floor — Stand # 124

Improving the quality of patient care: lessons learnt from 16 years of EMQN external quality assessment (EQA)

Meeting programme:
EQA scheme summary reports:
a. Pilot EQA scheme for Next Generation Sequencing (NGS) – 20 mins.
b. Lung Cancer EQA scheme – 20 mins.
c. Phenylketonuria (PKU) EQA scheme – 20 mins.
Open forum Q&A session

Chairs:
- *Prof. David Barton, National centre for Medical Genetics, Our Lady's Hospital, Dublin, Ireland*
- *Dr Simon Patton, EMQN, St Mary's Hospital, Manchester, UK*

Speakers:
- *Dr Simon Patton, EMQN, St Mary's Hospital, Manchester, UK*
- *Mrs Nicola Wolstenholme, EMQN, St Mary's Hospital, Manchester, UK*
- *Dr Martina Witsch-Baumgartner, Department of Medical Genetics, University of Innsbruck, Innsbruck, Austria*

GENERAL BUSINESS AND ANCILLARY MEETINGS

As per date of printing.

Saturday, June 6, 2015

08:30-09:30	B01	Fetal Genomics group CGS	Carron 2	closed
09:00-10:30	B02	UEMS Board Meeting	Seminar Suite	closed
10:00-13:00	B03	Meeting of the Int. Ehlers-Danlos Syndrome Consortium	Alsh 2	closed
10:00-13:00	B04	EuroGentest Subcommittee for Professional Guidelines	Katrine	closed
10:45-13:30	B05	ESHG PPPC Meeting	Carron 2	closed
11:00-13:00	B06	UEMS Section Meeting	Seminar Suite	closed
12:00-14:00	B07	ESHG EUGT Quality sub committee meeting	Fyne	closed
13:15-15:15	B08	EU cancer research network meeting	Alsh 2	closed
14:30-18:00	B09	European Cytogenetic Guidelines review meeting	Seminar Suite	closed
16:30-18:00	B10	Global Alliance members' meeting	Fyne	closed
17:30-18:30	B11	SPC Meeting BSGM	Carron 2	closed

Sunday, June 7, 2015

10:00-11:00	B12	European Genetic Nurses and Counsellors Meeting	Seminar Suite	closed
10:00-11:15	B13	European Journal of Medical Genetics Editorial Board	Carron 2	closed
10:00-11:45	B14	Building Bridges ESHG/ASHG Meeting	Alsh 2	closed
11:00-12:30	B15	EBMG branch board Clinical Laboratory Geneticists (CLG)	Fyne	closed
11:30-13:30	B16	NEQAS/CEQAS participants Meeting	Gala	open
11:30-13:30	B17	National Human Genetics Societies Meeting	Seminar Suite	closed
11:30-13:30	B18	IT Leads Meeting BSGM	Carron 2	closed
12:00-13:00	B19	A beginner's guide to bioinformatics for genetic counsellors	Forth	open to counsellors
12:00-13:30	B20	Council Meeting BSGM	Alsh 2	closed
19:15-20:15	B21	ESHG Membership Meeting	Gala	open to members

Monday, June 8, 2015

10:00-12:00	B22	EBMG General Assembly	Alsh 2	closed
10:15-11:15	B23	EJHG Editorial Board Meeting	Carron 2	closed
10:30-13:30	B24	Assessor Training Meeting	Katrine	closed
11:30-12:00	B25	AGM BSGM	Forth	closed
11:30-13:30	B26	eRare EuroMicro Consortium Meeting	Carron 2	closed
12:00-13:00	B27	ESHG Board Meeting II	Seminar Suite	closed
12:00-13:30	B28	ESHG Education Committee Meeting	Alsh 2	closed
12:15-18:15	B29	Journal of Community Genetics Editorial Board	Fyne	closed
12:30-13:00	B30	AGNC	Gala	closed
13:00-13:30	B31	GCRB	Gala	closed

Tuesday, June 9, 2015

12:15-13:15	B32	ESHG SPC Meeting	Seminar Suite	closed

Disclaimer
Ancillary and satellite meetings shall not state or imply endorsement of or support by the ESHG of the event, organiser, products or services presented in any verbal statements or printed/electronic media before, after and during the presentations.

PROGRAMME ESHG AND YOUNG INVESTIGATOR AWARDS

ESHG Award

The ESHG Award, formerly "Mauro Baschirotto Award", was founded in 1992 and is presented by the European Society of Human Genetics during its annual European Human Genetics Conference in recognition of individual achievement in human genetics.

Award Holders

1992 Lore **Zech**
1993 Pierre **Maroteaux**
1994 Mary **Lyon**
1995 Jean **Weissenbach**
1996 Malcolm **Ferguson-Smith**
1997 Leena **Peltonen**
1998 Jean-Louis **Mandel**
1999 Pat **Jacobs**
2000 Dirk **Bootsma**
2001 Robin **Winter**
2002 Albert **de la Chapelle**
2003 Peter S. **Harper**
2004 Bernhard **Horsthemke**
2005 Stylianos **Antonarakis**
2006 Veronica **van Heyningen**
2007 Andrea **Ballabio**
2008 Arnold **Munnich**
2009 Kari **Stefansson**
2010 Sir Alec **Jeffreys**
2011 GertJan B. **van Ommen**
2012 Peter **Lichter**
2013 Felix **Mitelman**
2014 Sir Michael **Stratton**
2015 Svante **Pääbo**

ESHG Young Investigator Awards

The Scientific Programme Committee has shortlisted presenters for the **ESHG Young Investigator Award**. The profiles as well as a short interview of the finalists can be found on the next pages.
The committee will judge finalists' presentations during the conference.

The following awards will be presented to the winners in the closing ceremony on Tuesday, June 9, 2015 at 14.15 hrs:

- A total of four **ESHG Young Investigator Awards** are granted for outstanding research by young scientists presented as a spoken contribution at the conference.
- The **Isabel Oberlé Award** is awarded yearly since 2002 for best presentation by a young scientist on research concerning the genetics of mental retardation.
- The **Lodewijk Sandkuijl Award** was instituted in 2004 to be awarded to the author of the best presentation at the ESHG conference within the field of complex disease genetics and statistical genetics.
- The **Vienna Medical Academy Award** (funded by our conference organiser VMA since 2012) will be awarded to the best presentation in translational genetic research/therapy of genetic diseases.
- The **Mia Neri Award** (funded by the Mia Neri Foundation) will be awarded to the best presentation in cerebral cancer research.

All winners will receive prize money in the amount of EUR 500 and a complementary ESHG online membership for one year.

Talks of YIA finalists are highlighted by an asterisk (*) as well as a grey background in the detailed scientific programme.

On the next pages you will find short self-presentations of the candidates.

PROGRAMME YOUNG INVESTIGATOR AWARD CANDIDATES

We have asked the candidates to answer the following questions:
Q1: Date and city of birth
Q2: What is your current position?
Q3: Why did you choose a career in genetics?
Q4: What is so interesting about the research you are presenting at ESHG 2015?

Katrina Andrews
Cambridge, United Kingdom

Talk: C09.5 Tumour risks and genotype-phenotype-proteotype analysis in ~800 patients with germline mutations in the succinate dehydrogenase subunit genes SDHB, SDHC and SDHD
Session: C09 Genetic susceptibility to cancer development
Date: Sunday, June 7, 2015, 13:30 hrs
Q1: 27/11/1988, Guildford, UK
Q2: Academic foundation doctor, Cambridge University Hospitals NHS Foundation Trust
Q3: I have chosen to apply for a career in clinical genetics because I enjoy the challenge of communicating risk and uncertainty, diagnosing rare disorders and constantly adapting my practice as new technologies come into play.
Q4: In a large cohort of mutation positive patients with paraganglioma/phaeochromocytoma, we are able to show how different mutations within different proteins of the same succinate dehydrogenase enzyme complex can result in dramatically different phenotypes. This will be important not only for genetic counselling, but also for designing tailored screening protocols.

Reza Asadollahi
Zurich-Schlieren, Switzerland

Talk: C15.6 Clinical and experimental evidence establish a link between KIF7 and C5orf42-related ciliopathies
Session: C15 Network and functional analysis in intellectual disability
Date: Monday, June 8, 2015, 13:30 hrs
Q1: Yazd, Iran
Q2: MD-PhD, Postdoctoral Fellow
Q3: My primary entry into genetics goes back to my MD thesis which was about polymorphisms of IL10 gene and risk of breast cancer. Later, I was very fortunate to have the opportunity to continue my studies in the field of medical genetics with the support of my inspiring mentor, Anita Rauch. Now that genetics/genomics is revolutionizing the entire field of medicine for precise risk assessment, personalized diagnostics and targeted treatment, I am more than sure that I am on the right track.
Q4: By combining the clinical presentation of patients, whole exome or targeted sequencing results and experimental data from chicken embryos, we established the link between KIF7 and C5orf42-related ciliopathies. We evidenced, for the first time, the role of C5orf42 in craniofacial development, pathfinding of commissural axons and neural circuit formation.

Emma Baple
Southampton, United Kingdom

Talk: C23.4 Homozygous truncating mutations in WDR73 cause a severe nephrocerebellar syndrome, part of the Galloway Mowat syndrome spectrum
Session: C23 Movement and motor disorders
Date: Tuesday, June 9, 2015, 11:00 hrs

Tahsin Stefan Barakat
Rotterdam, Netherlands

Talk: C17.1 RNF12 is essential for X-inactivation in female mouse embryonic stem cells, is required for female mouse development, and might be a target for future therapies to treat X-linked disorders in females: evidence from a mouse knockout model
Session: C17 Epigenetic control of gene expression
Date: Monday, June 8, 2015, 13:30 hrs
Q1: 25/05/1984, Meerbusch, Germany
Q2: At present, I am a postdoctoral research fellow at the University of Edinburgh, in the MRC Centre for Regenerative Medicine. Here I try to understand how embryonic stem cells maintain their self-renewal and differentiation capacity. At a long term, insights in the mechanisms regulating this pluripotency might help in developing methods to alter cell fate, and might help to develop novel stem cell based therapies.
Q3: During my Medical Studies in Rotterdam, The Netherlands, I noticed that I am very fascinated by mechanisms underlying disease. In particular genetic mechanisms underlying inherited disorder always attracted my attention. Especially being fascinated by the X chromosome inactivation process and the many influences this process can have on disease phenotypes in females, I embarked on a Ph.D. project which aimed to understand the regulation of this important epigenetic process. At the long term, I plan to train as a clinical geneticist, and combine clinical related work with my research, trying to develop novel stem cell based therapies for inherited disorders.
Q4: My research has contributed to a better understanding of the regulation of the initiation of the female specific X chromosome inactivation (XCI) process. In particular, I identified the E3 ubiquitin ligase RNF12 (RLIM) as a crucial activator of the XCI process which functions by degrading the embryonic stem cell pluripotency factor REX1, thereby providing evidence for a direct link between regulation of XCI and the pluripotency network in embryonic stem cells. To test the role of RNF12 in vivo, I have created a novel RNF12 knockout mouse model. This model emphasizes the importance of RNF12 in the regulation of XCI, as homozygous RNF12 deficient female mice are lethal due to absence of XCI initiation. Surprisingly, in Rnf12+/- female mice, which are healthy, we found evidence for a lack of XCI in many different cell types in vivo. Hence, these peculiar female mice are one of the first examples of placental mammals with a lack of XCI, and bi-allelic X-linked gene expression. Since these animals do not have an obvious disease phenotype, it might now be possible to develop X chromosome reactivation methods to diminish the disease phenotype of females suffering from X-linked disorders due to unfavorable skewing of XCI and inactivation of the wild type X chromosome. These X reactivation methods, combined with stem cell based therapies, might become a new future treatment modality for X-linked disorders.

Tracy Briggs
Manchester, United Kingdom

Talk: C11.4
Spondyloenchondrodysplasia: The expanding phenotype of TRAP deficiency
Session: C11 Skeletal disorders
Date: Sunday, June 7, 2015, 13:30 hrs

Q1: Halifax, UK. 18-06-77

Q2: I am an NIHR Academic Clinical Lecturer at The University of Manchester, UK. My time is split 50:50 between my laboratory-based research, which focuses particularly on immunogenetics, and my clinical training at The Manchester Centre for Genomics Medicine, St Mary's Hospital.

Q3: I have always found genetics fascinating. I am particularly passionate about the opportunity to combine clinical genetic medicine and research. I love the challenge of testing hypotheses and investigating new questions. When the answers have the potential to translate directly to my patients, and those of my colleagues, I find this very rewarding.

Q4: I have been working on Spondyloenchondrodysplasia for the past eight years. During this period we have identified the causative gene and moved towards understanding the functional biology. I will present data which demonstrate the highly pleiotropic nature of the condition, including an absence of immune disease in some gene positive cases. I will show that, whilst most cases demonstrate elevated interferon-stimulated gene expression, this is not universal, and patients with normal levels may provide a clue towards effective therapy.

Nathalie Brison
Leuven, Belgium

Talk: C01.5 Incidental findings of genome wide non-invasive fetal aneuploidy detection (NIPT): presymptomatic identification of maternal cancers
Session: C01 NIPT
Date: Saturday, June 6, 2015, 18:30 hrs

Q1: July 6, 1983, Kortrijk, Belgium

Q2: I'm a postdoc in the Centre for Human Genetics, Leuven (Belgium). I'm responsible for the diagnostic NIPT workflow in the Clinical Cytogenetics lab.

Q3: How can even the smallest change in DNA sequence or copy number cause disease in one person, and have almost no phenotypic effect in another? How can we accurately predict phenotypic outcome in newborns or at later stages in life? Finding clues using pre/postnatal testing on the edge of research and routine diagnostics is the challenge I am eager to pursue. The answers we can give using novel techniques in the rapidly evolving field of Clinical Genetics can make a huge difference, not only for Science, but for a person's quality of life, for a couple, for a whole family.

Q4: The presence of cell-free fetal DNA in the maternal circulation has allowed for the development of different methods for non-invasive detection of fetal chromosomal imbalances. Non-invasive prenatal testing (NIPT) thus avoids miscarriages cause by invasive sampling of fetal material. We developed and validated an innovative, fast, cost efficient workflow and high throughput analysis pipeline for NIPT. This optimized genome-wide analysis pipeline overcomes some of the technical and biological causes of false positive or false negative results which resulted in very high sensitivities and specificities for trisomy 21, 18 and 13 detection in over 6000 pregnancies. Moreover, it seems to create opportunities to detect other chromosomal abnormalities in addition to the traditional trisomies, among which other (partial) fetal aneuploidies, clinically relevant maternal incidental findings and even presymptomatic cancer in some pregnant women…

Eline Bunnik
Rotterdam, Netherlands

Talk: C20.6 What's in it for me? A critical analysis of the notion of personal utility in genomic testing
Session: C20 Current issues in genet(h)ics
Date: Tuesday, June 9, 2015, 11:00 hrs

Q1: Leidschendam, 14 October 1982

Q2: post-doc researcher

Q3: Genetics is a fast-moving field, which confronts citizens, patients and physicians with ever-evolving ethical challenges, which must be handled proactively and constructively. I can put my training in philosophy and ethics to use in genetics, and thus contribute to responsible innovation in genetics-based medicine and technology.

Q4: I will critically address the often-used but rarely studied notion of personal utility. When is a genomic test personally useful? Clearly defined, the notion of personal utility can be a great tool to distinguish meaningful from meaningless tests, to justify a liberal but responsible genomic testing offer, and to help ensure the progress of genetics research and technology.

Eduardo Calpena
Valencia, Spain

Talk: C06.5 Junctophilin-1 expression levels could modify the effects of GDAP1 mutations in Charcot-Marie-Tooth disease
Session: C06 Neuromuscular disorders
Date: Saturday, June 6, 2015, 18:30 hrs

Q1: February 1986, Alicante (Spain)

Q2: PhD student in the the Program in Rare and Genetic Diseases at Centro de Investigación Príncipe Felipe (CIPF) in Valencia (Spain).

Q3: Rare disease awakened my interest for human genetics. An exciting field is not only to discover new genes involved in Mendelian genetic disorders but also to identify genetic factors that may modulate the disease phenotype to explain the variable clinical expression. In fact, I really believe that characterization of genetic modifiers is the key to discover important pathways for new therapies.

Q4: We have recently described the JPH1 gene as a genetic modifier in Charcot-Marie-Tooth (CMT) disease, one of the

PROGRAMME YOUNG INVESTIGATOR AWARD CANDIDATES

most common inherited neurological disorders. In this work we have identified new variants in the JPH1 gene which affect its expression and that could modify the effects of GDAP1 mutations in CMT disease. We have used Drosophila models to demonstrate how altered junctophilin expression levels modify the effects of Gdap1-related neural degeneration. Moreover, the Drosophila model has allowed us to discover new pathways related to junctophilin.

Azahara Civera-Tregón
Valencia, Spain

Talk: C06.4 Analysis of the Gdap1 knockout mice reveals calcium homeostasis and mitochondrial dynamics defects in the Charcot-Marie-Tooth disease pathogenesis
Session: C06 Neuromuscular disorders
Date: Saturday, June 6, 2015, 18:30 hrs

Estelle Colin
Angers, France

Talk: C16.3 Loss-of-Function Mutations in WDR73 Are Responsible for Microcephaly and Steroid-Resistant Nephrotic Syndrome: Galloway-Mowat Syndrome
Session: C16 Growth failure and microcephaly
Date: Monday, June 8, 2015, 13:30 hrs
Q1: 22/08/1979, Brest, France
Q2: Currently I am working as clinical geneticist and PhD student at the University Hospital of Angers, France (UMR INSERM 1083 – CNRS 6214).
Q3: Everything is a matter of meeting. Initially I am a pediatrician and during my training I met the team in which I currently work. The whole team showed me the importance of the links between the clinic and the laboratory. The considerations that may exist between phenotype and genotype or the arrival of new technology which allows a better understanding of pathological mechanisms. The practice of clinical genetics also request an important dialogue with patients and their families that challenges perpetually our practice.
Q4: We have shown that WDR73 is the first gene involved in Galloway-Mowat syndrome. And our work underlines the critical function of WDR73, which was yet an unknown gene, in both neurons and podocytes via an important role in neuronal cell survival and an involvement in the organization of microtubule networks. Also this research should allow a genetic counseling for all the concerned families with this rare and severe disease.

Laurence Colleaux
Paris, France

Talk: C10.2 Identification of a common set of microRNAs deregulated in Autism Spectrum disorders
Session: C10 Neurogenetic disorders
Date: Sunday, June 7, 2015, 13:30 hrs

Viviana Cordeddu
Rome, Italy

Talk: C21.5 Mutations in transcription factor ZBTB20 cause tall stature, macrocephaly, cognitive deficits, diabetes, progressive muscle wasting and deafness
Session: C21 Multiple congenital anomaly syndromes
Date: Tuesday, June 9, 2015, 11:00 hrs
Q1: 28-06-1974 Sassari
Q2: I am in-staff molecular biologist at the Department of Hematology Oncology and Molecular Medicine, Italian National Health Institute, Rome, Italy.
Q3: I have been fascinated with the molecular mechanisms involved in human disease since my first studies at the University. Medical genetics was my natural choice for my scientific track.
Q4: Beside the identification of a new disease gene implicated in a human developmental disorder, the Primrose syndrome, this discovery provide a fascinating evidence of the diverse clinical impact of mutations affecting different domain in the same protein and differentially affect protein stability and function.

Candice Cornelis
Utrecht, Netherlands

Talk: C20.4 Informed consent for whole exome sequencing in pediatric disease diagnostics: parental decision-making processes, their ethical relevance and implications for policy development
Session: C20 Current issues in genet(h)ics
Date: Tuesday, June 9, 2015, 11:00 hrs
Q1: 23-11-1984, Washington, D.C., U.S.A.
Q2: PhD candidate at University Medical Center Utrecht and the Ethics Institute of Utrecht University in the Netherlands.
Q3: As an ethicist in training, what drew me to genetics are all the interesting moral questions that accompany scientific development and technological innovation within the field. For example, those regarding novel sequencing technologies: How can we safeguard persons' well-informed decision-making for these techniques? What should return of results policies for unsolicited findings look like?
Q4: My research shows how important the context-specific factors of persons' situations are for understanding why parents make certain (disclosure) decisions concerning clinical sequencing for their child. In turn, understanding the different kinds of context-specific factors that can play a role in decision making helps us sharpen our answers to moral questions of policy development for using these techniques.

Francesca Cristofoli
Leuven, Belgium

Talk: C16.6 From whole exome sequencing to functional studies in syndromic microcephaly: using zebrafish for variant testing
Session: C16 Growth failure and microcephaly
Date: Monday, June 8, 2015, 13:30 hrs

Q1: 07/07/1987 San Daniele del Friuli (Italy)
Q2: PhD candidate student at the Laboratory for Cytogenetics and Genome Research, Departmet of Human Genetics, KU Leuven, Belgium
Q3: My interest in genetics dates back to middle school, I was already very fascinated by DNA and chromosomes, a perfect combination of beautiful structure and rational replication - translation mechanisms which are the foundation of living organisms. My curiosity and passion for human genetics in particular was instilled by a former University cytogenetics professor who made me realize the importance of understanding more in depth the mechanisms underlying genetic disorders in order to concretely help people.
Q4: Although the genetic causes of an increasing number of disorders encompassing microcephaly have been identified, a great number of still unexplained syndromes exist. The research I am presenting at ESHG is interesting first of all because we display the results we obtained by whole exome sequencing on a cohort of patients presenting sporadic syndromic forms of microcephaly. Secondly, since variant interpretation is always challenging when analyzing WES data, we also present data obtained using the zebrafish model to define pathogenicity of some of the candidate variants identified.

Mirjam de Pagter
Utrecht, Netherlands

Talk: C13.6 Chromothripsis in healthy individuals affects multiple protein-coding genes and can result in severe congenital abnormalities in offspring
Session: C13 Fundamental insights in structural genomics
Date: Monday, June 8, 2015, 13:30 hrs

Q1: 18-12-1982 Vlissingen (Netherlands)
Q2: PhD student
Q3: Ever since I first learned about genetics, it's importance and impact it has interested me. Even though we've learned so much over the last decades, there is always more to be discovered. What inspires me is providing families with genetic answers to 'what and how' while potentially facilitating treatment options/ development as well.
Q4: Chromothripsis has devastating effects on chromosomal architecture and has been linked to cancer and congenital defects. We have identified copy number balanced chromothripsis in healthy females. In all cases, this directly impacted reproduction. The occurrence of balanced chromothripsis in healthy individuals may remain undetected by frequently used diagnostic tools and can have direct clinical implications.

Cristina Dias
Hinxton, Cambridge, United Kingdom

Talk: C03.4 De novo mutations in BCL11A cause developmental delay: additional implications of the BAF SWI/SNF complex in intellectual disability and autism
Session: C03 Novel genes causing intellectual disability
Date: Saturday, June 6, 2015, 18:30 hrs

Q1: 14/07/1975, Toronto, Canada
Q2: Fellow of the Wellcome Trust PhD Programme for Clinicians (Wellcome Trust Sanger Institute and University of Cambridge)
Q3: Early in my medical training I learned significant numbers of children with chronic and/or debilitating disorders have a rare disease of genetic aetiology. Understanding the underlying pathophysiology of rare disease contributes to the well-being of patients and provides valuable insight into common disease. I am committed to integrating scientific research into the multidisciplinary approach to clinical care, contributing to the goal of aiding the establishment of long-term prognosis and therapy for rare disease.
Q4: BCL11A has been extensively studied for its roles in malignancies and hematopoiesis. We are just beginning to discover how important it is in neurodevelopment, through the identification of patients with intellectual disability and autism with mutations in BCL11A. In modeling biological processes and behavior in mammalian models, we are increasing our understanding of patients, also our overall understanding of neurodevelopment and cognition.

Eftychia Dimitriadou
Leuven, Belgium

Talk: C13.5 High incidence of mosaic chromosomal aneuploidies in human cell lines: a quantification of the frequency of the phenomenon
Session: C13 Fundamental insights in structural genomics
Date: Monday, June 8, 2015, 13:30 hrs

Q1: 22/02/1983, Thessaloniki, Greece
Q2: Postdoctoral Researcher in the Centre for Human Genetics, Laboratory for Cytogenetics and Genome Research, KU Leuven, Belgium
Q3: During my late high-school years already I was fascinated by human chromosomes. Later, as a student in Applied Biology and Biotechnology, I got even more curious to understand how their function and structure can be linked to human disease, which led me to choose for a carrier in Human Genetics. Today, the mysterious nature of chromosomes still intrigue me and unravelling the causes and mechanisms underlying chromosomal instability as well as understanding the possible consequences of such events on human health has become my everyday life.
Q4: Whereas chromosome segregation is thought to occur with high fidelity, our study shows that large segmental chromosomal imbalances occur in fibroblast- and EBV-cell lines, with a frequency 100-400 times higher than thus far estimated. The analysis of the both daughter cells following a single cell division together the implementation of haplarithmisis, a novel concept that enables very accurate concurrent haplotyping and copy-number profiling

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in unprecedented detail at the single-cell resolution and in a genome-wide fashion, result in high-confidence characterization of the imbalances. Our findings have important practical and theoretical consequences: they put in question the efficacy of the DNA repair mechanisms and control checkpoints and imply that the detected imbalances may underlie the first steps towards cancer progression.

Jamie Ellingford
Manchester, United Kingdom

Talk: C19.6 Whole genome sequencing as a clinical diagnostic tool for heterogeneous Mendelian disease
Session: C19 Diagnostic NGS
Date: Tuesday, June 9, 2015, 11:00 hrs
Q1: 16/08/1990, Chester, UK
Q2: I am a 2nd year PhD student at the University of Manchester. I am completing my PhD in Genetic Medicine and Clinical Bioinformatics at the Manchester Centre for Genomic Medicine, a collaboration between the University of Manchester and the Central Manchester NHS Foundation Trust.
Q3: I knew that a research career in human genetics would be a fast-paced, challenging, competitive, and as a result an extremely exciting area of medical research, but it is the real-life and everyday application of clinical genetics that has inspired me to build a career in genetics.
Q4: My research is at the cutting edge of the application of genetics knowledge to the clinical care of patients. It assesses the advantage of using whole genome sequencing techniques in the clinic, and reveals the diversity of disease-causing genetic variation that this technology can detect.

Tonu Esko
Tartu, Estonia

Talk: C15.1 Genome-wide association study of 200,000 individuals identifies 18 genome-wide significant loci and provides biological insight into human cognitive function
Session: C15 Network and functional analysis in intellectual disability
Date: Monday, June 8, 2015, 13:30 hrs

Lucas Fares Taie
Paris, France

Talk: C12.5 Submicroscopic deletions at 13q32.1 cause congenital microcoria
Session: C12 Sensory disorders
Date: Sunday, June 7, 2015, 13:30 hrs
Q1: 22/03/1977, Mar del Plata (Argentina)
Q2: I'm a Postdoctoral Research Fellow at the Imagine Institut of Paris
Q3: During my studies of biochemistry I was captivated by the contribution of genetics in health and disease. I am particularly interested in the identification of news genes in order to understand mechanism s underlying diseases.
Q4: Congenital Microcoria is a very rare disease associated with strong myopia and glaucoma. This is the first report that identifies the genetic anomaly causing this particular disease. Moreover our work should provide further insights into the development of the anterior chamber of the eye, which anomalies are often responsible of severe visual loss, due, for example, to glaucoma.

Tomas Fitzgerald
Cambridge, United Kingdom

Talk: C19.4 Copy Number Analysis using Exon-level aCGH and Exome Sequencing in over 3,000 Parent-Offspring Trios from the Deciphering Developmental Disorders Project
Session: C19 Diagnostic NGS
Date: Tuesday, June 9, 2015, 11:00 hrs

Elisabeth Gillis
Edegem - Antwerp, Belgium

Talk: C05.2 Mutations in a TGFβ ligand, TGFB3, cause syndromic aortic aneurysms and dissections
Session: C05 Cardiovascular disorders
Date: Saturday, June 6, 2015, 18:30 hrs
Q1: April 1st, 1990
Q2: I am a PhD Student at the University of Antwerp, Belgium
Q3: "We have the power to imagine better" is what JK Rowling once said about helping people. And I am convinced that we, as researchers, can not only imagine better, but actually play a significant role in treating diseases. And that process starts at the genetic level, by unravelling that basis the road is set to find proper treatment.
Q4: To this day, we keep identifying mutations in new genes (in this case TGFB3) within the TGF2/ pathway that cause different thoracic aortic aneurysm syndromes, such as Marfan syndrome and Loeys-Dietz syndrom

Anne Guimier
Paris, France

Talk: C05.5 Recessive mutations in matrix metallopeptidase 21 (MMP21) cause heterotaxy in humans
Session: C05 Cardiovascular disorders
Date: Saturday, June 6, 2015, 18:30 hrs
Q1: 25/06/1982, Paris, France
Q2: PhD student, IMAGINE institut, Paris
Q3: MD specialized in Paediatrics, I m mostly interested in genetics of congenital malformations and I have found in Genetics, an enthusiastic and fascinating way to link medicine/ clinical skill and research.
Q4: Our study is about the identification of MMP21, a matrix metallopeptidase family member, as a novel gene implicated in heterotaxy and congenital heart defects in human, providing new insights into regulation of left right asymmetry during embryonic development.

Claire Guissart
Montpellier, France

Talk: C12.2 Exome sequencing of ataxia-blindness patients identifies atypical Brown-Vialetto-Van Laere syndrome-2 (BVVLS2) presentation and identifies PEX6 as the SCAR3 (MIM#271250) gene
Session: C12 Sensory disorders
Date: Sunday, June 7, 2015, 13:30 hrs
Q1: 3/3/1986, Montpellier, France
Q2: I am a PhD student, PharmD and graduate teaching assistant at the Regional University Hospital Center of Montpellier, France, where I am involved in the research and diagnosis of autosomal recessive cerebellar ataxias by exome sequencing.
Q3: Genetics is a fascinating field that offers me the opportunity to work on a constantly evolving matter.
Therefore, I am motivated by the potential application of my work to patients.
Q4: We therefore postulate that PEX6 is the gene defective at the SCAR3/SCABD1 locus at 6p21 (OMIM# 211530) and we propose that patients with ataxia, deafness, optic atrophy and mutations in SLC52A2 present with the SCABD type 2 entity (SCABD2).
Distinguishing the two entities at the molecular level is of high importance since patients with mutations in SLC52A2 can be ameliorated by riboflavin supplementation.

Dorien Haesen
Leuven, Belgium

Talk: C03.2 De novo and recurrent PPP2R5D and PPP2R1A missense mutations cause protein phosphatase 2A dysfunction and intellectual disability
Session: C03 Novel genes causing intellectual disability
Date: Saturday, June 6, 2015, 18:30 hrs
Q1: 19/05/1988, Genk, Belgium
Q2: PhD-student
Q3: I more or less 'accidently' ended up in this discipline, through a group of clinical geneticists, coordinated by Prof. Gunnar Houge, who discovered a potentially novel genetic cause of intellectual disability, and contacted my supervisor for collaboration to characterize the biochemical and functional consequences of the mutations.
Q4: Phosphatases, such as PP2A, are still too often considered as the 'ugly ducklings of cell signaling'. We characterised a PP2A syndrome causing ID, through de novo mutations in two different PP2A genes, but giving rise to a common dysfunction via a novel mechanism. This opens perspectives for improved treatment options.

Andrea Hofmann
Bonn, Germany

Talk: C17.4 Mapping genetic and epigenetic factors influencing human hippocampal gene expression
Session: C17 Epigenetic control of gene expression
Date: Monday, June 8, 2015, 13:30 hrs
Q1: 19/09/1983, Ratingen, Germany
Q2: I'm a postdocteral researcher in bioinformatics at the Institute of Human Genetics, University of Bonn, Germany
Q3: Genetics is a fascinating scientific field and provides an exciting opportunity to combine my interest in biology, statistics and programming. I hope systems genetics approaches will help bridging the gap between basic molecular mechanisms and clinical translation.
Q4: Brain QTL studies are hampered by the restricted accessibility of relevant tissue. We have access to a unique sample of pre-mortem human hippocampus tissue and systematically mapped genetic and epigenetic effects on gene expression. Our study provides a valuable resource for functional SNP annotation and will help guiding the interpretation of GWAS hits in complex brain disorders.

Maral Jamshidi
Helsinki, Finland

Talk: C04.2 SNP-SNP interaction analysis of NF-kB signaling pathway on breast cancer survival
Session: C04 The many faces of cancer mutations
Date: Saturday, June 6, 2015, 18:30 hrs
Q1: 21.09.1981, Ahvaz
Q2: M.Sc., PhD candidate
Q3: I enjoy the logical reasoning behind genetics. Biology, mathematics, and art have always been my favorite topics and they come together in the field of genetics beautifully.
Q4: For a complex disease such as breast cancer it is possible that a single SNP is not independently critical in the biological function underlying the initiation or progression of the disease, but multiple loci might jointly exert a greater impact. Furthermore, aberrant regulation of the NF-κB pathway has been shown in breast cancer, however, the impact of the genetic variation in the pathway on patient prognosis has been little studied. We investigated the NF-κB activating pathway for association of the genetic variation in 75 genes involved in the pathway with breast cancer prognosis. Assessing two-way SNP-SNP interaction survival analyses, we found two pairs of genetic variations, i.e. rs5996080 and rs7973914, and rs17243893 and rs57890595, corresponding to five NF-κB activating pathway genes, i.e. BAFFR and TNFR1/TNFR3, and TRAF2 and TRAIL-R4 (respectively), with interactive effect on survival after breast cancer. These results suggest a role for these genetic loci and their plausible target genes in the progression of the disease and patient survival. However, further validation and functional studies are needed, also for establishing their clinical impact. Additionally, our study highlights the utility of genetic interaction analyses on breast cancer survival.

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Wendy Jones
Cambridge, United Kingdom

Talk: C21.3 Wiedemann-Steiner Syndrome: Expanding the phenotypic spectrum associated with KMT2A (MLL) mutations
Session: C21 Multiple congenital anomaly syndromes
Date: Tuesday, June 9, 2015, 11:00 hrs
Q1: Maidstone, UK. 10th September 1976
Q2: Clinical PhD Student, Wellcome Trust Sanger Institute
Q3: In health and in disease humans are all different, and there is no other medical specialty or academic discipline that drills as deep down to the fundamental cause of these differences as genetics does. I relish both the molecular science and helping families find and understand the genetic cause of their difficulties.
Q4: Wiedemann-Steiner syndrome (WSS) resulting from KMT2A mutations is proving to be one of the more common rare diseases so why hasn't it been more recognised before recent times? This is the largest known study to date of individuals with this condition, so what really is the phenotype associated with germline KMT2A mutations?

Peter Joshi
Edinburgh, United Kingdom

Talk: C22.6 Evidence for directional dominance on complex traits relating to size and cognition in a wide range of human populations
Session: C22 Statistical genetics and bioinformatics
Date: Tuesday, June 9, 2015, 11:00 hrs
Q1: 13/4/65 Brussels
Q2: PhD Candidate Centre for Population Health Sciences, University of Edinburgh
Q3: I am interested in understanding and predicting the genetic basis of complex traits, especially human longevity. I believe large scale population studies and sequencing and computing technologies are about to give us the power to elucidate these effects.
Q4: Using genomic data on over 350,000 subjects, we show conclusively for the first time that body size and cognition but not 12 other traits are subject to directional dominance across the whole genome, that this effect is trans-continental and unlikely to be due to confounding. As directional dominance is forecast to arise for traits under directional selection, we conclude that height and cognition have been subject to directional selective pressure, but risk factors for cardiovascular disease have not, Our study thus answers long standing questions on the alleleic architecture of these complex traits and insight into evolution.

Marjolijn Jongmans
Nijmegen, Netherlands

Talk: C09.1 High yield of causative mutations by whole exome sequencing in selected individuals with childhood cancer
Session: C09 Genetic susceptibility to cancer development
Date: Sunday, June 7, 2015, 13:30 hrs
Q1: 13-12-1977 Roosendaal
Q2: Clinical Geneticist
Q3: As a medical student I attended a surgery of a child with a cleft lip. I realized that I was much more intrigued by the information in her file about the syndrome she had and how her clinical geneticist came to this diagnosis, than by the surgery. At that moment I knew that I wanted to work in clinical genetics.
Q4: I will present our experiences with germline exome sequencing in children with cancer. Our data confirm that the clinical definitions of syndromes are often to strict and that NGS answers many questions but raises at least as many.

Marika Kaakinen
London, United Kingdom

Talk: C22.3 A novel method and software tool for genome-wide multi-phenotype analysis of rare variants
Session: C22 Statistical genetics and bioinformatics
Date: Tuesday, June 9, 2015, 11:00 hrs
Q1: 18.9.1982, Oulunsalo, Finland
Q2: Postdoctoral Marie Curie Fellow
Q3: My undergraduate training is in statistics and epidemiology, but my first research experience already involved analysis of genetic data. I immediately got interested in the complexity and challenges this rapidly moving field offers for everyday research. I am very excited to be involved in research trying to understand the code we carry in our cells and to use that information for improving public health.
Q4: Humans have about 25,000 genes but a lot more phenotypes can be defined for each of us. It is obvious that some genes affect multiple phenotypes. We have developed a method and a software tool that can help to detect such multi-phenotype effects for genetic variants that are rare in the population, to address the so-called missing heritability issue.

Maria Keller
Leipzig, Germany

Talk: C17.3 Genome wide DNA promoter methylation: Differences in human subcutaneous vs. omental visceral adipose tissue
Session: C17 Epigenetic control of gene expression
Date: Monday, June 8, 2015, 13:30 hrs
Q1: 15.05.1987, Leipzig
Q2: I am a PhD student at the Integrated Research and Treatment Center (IFB) for Adiposity Diseases at the University of Leipzig, Germany. I work in a junior research group which is mainly interested in functional genetics of obesity.
Q3: My interest for genetics and especially epigenetics already started during my time as a Master student (Nutrition Sciences) at the University of Vienna. I am still fascinated by the idea that it might be possible to change the transcriptional activity of our genes due to a change in environmental conditions.
Q4: The data I will present at the conference is to my knowledge one of the first large and comprehensive dataset of genome-wide DNA methylation and mRNA expression comparing paired samples of human subcutaneous and omental visceral adipose tissue in lean and obese individuals, which helped to select novel candidate genes.

Sietske Kevelam
Amsterdam, Netherlands

Talk: C23.6 PLP1 mutations affecting PLP1/DM20 alternative splicing causes Hypomyelination of Early Myelinating Structures
Session: C23 Movement and motor disorders
Date: Tuesday, June 9, 2015, 11:00 hrs
Q1: 30-07-1985, Nijmegen
Q2: I am a PhD student working at the departments of Child Neurology and Medical Genome Analysis.
Q3: During medical school I became fascinated by the contribution of genetics in disease and in health. Using the next-generation sequencing techniques we can now identify the genetic cause of more and more rare disorders. This results in a broader knowledge of novel cellular and molecular mechanisms and most importantly will give patients and their families answers and help them cope with their diseases. Working in a field that both enhances our understanding of human biology and disease and has an essential interaction with the patients and families is extremely rewarding.
Q4: The focus of my research is on inherited childhood white matter disorders. These are rare disorders often resulting in severe neurological deficits and an early death. These patients present with a specific MRI-pattern. We use this MRI-pattern to form homogeneous patients groups, which helps tremendously in finding the common mutated gene with whole-exome sequencing (WES). In this study of a group of male patients with a novel MRI phenotype we identified unusual intronic mutations in a known gene. This discovery of these mutations indicates that long-distance intronic regions can be involved in disease. Also, caution is warranted as these regions are not covered with WES.

Arthur Ko
Los Angeles, United States

Talk: C08.1 Context-specific eQTLs identify hormonal effects in obese Finnish men
Session: C08 Integrative OMICS approaches in common traits
Date: Sunday, June 7, 2015, 13:30 hrs
Q1: August 24th, 1989. Ames, USA
Q2: PhD. Student
Q3: Our DNA contains information about who we are and where we are from. My goal as a geneticist is to untangle the genetic code in order to predict and prevent diseases. I can't think of anything more exciting and rewarding than to understand life itself and help improve others' lives.
Q4: I will present our work on context-specific eQTLs as a form of gene and environment interactions in obesity. We investigated the adipose transcriptional regulation in 566 men and discovered that many estrogen pathway genes are regulated by DNA variants only in obese men implicating the importance hormone effect in obesity.

Laura Kremer
Neuherberg, Germany

Talk: C18.2 COQ4 mutations cause a broad spectrum of mitochondrial disorders associated with CoQ10 deficiency
Session: C18 Metabolic and renal disorders
Date: Monday, June 8, 2015, 13:30 hrs
Q1: 05.09.1986 Rodalben, Germany
Q2: 3rd year PhD student
Q3: I hope understanding the genetic cause of diseases helps paving the way for the better understanding of the pathomechanism and eventually the development of therapeutic intervention.
Q4: Mitochondrial disorders are genetically and clinically extremely heterogeneous making proper diagnosis very challenging. Exome sequencing has now revolutionized the field and proven as a powerful and reliable tool to identify disease causing mutations and helping to understand mitochondrial physiology.

Ernest Lam
San Diego, United States

Talk: C04.3 Towards understanding the genomic architecture of cancer genomes
Session: C04 The many faces of cancer mutations
Date: Saturday, June 6, 2015, 18:30 hrs
Q1: Nov 6, 1986 in Hong Kong
Q2: Senior Scientist, Computational Biology at BioNano Genomics.
Q3: Since I was young, I have been very interested in cancer biology. I had great mentors in high school and college that

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introduced me to research. I was exposed to the research environment and realized it was my passion. In particular, there was key advances in genetics that drew me to the field.
Q4: Cancer genomes often harbor complex structural abnormalities; therefore, understanding the driving forces of cancer remains a challenge. Taking advantage of both next-generation sequencing and genome mapping gives a more comprehensive view of a cancer genome.

Steven Laurie
Barcelona, Spain

Talk: C19.3 The RD-Connect platform includes the first 360 analysed exomes linked to phenotypic data and integrates user-friendly tools for rare disease variant prioritization
Session: C19 Diagnostic NGS
Date: Tuesday, June 9, 2015, 11:00 hrs
Q1: 04/04/1973, Edinburgh
Q2: Senior Data Analyst, Data Analysis Team, CNAG, Barcelona
Q3: It is a fascinating field and may allow me to make some contribution towards helping others.
Q4: The RD-Connect project is a huge international endeavour, and upon completion will facilitate the rapid integration and interpretation of clinically relevant data, accelerating time to diagnosis and development of novel treatments for rare diseases.

Stefan Lelieveld
Nijmegen, Netherlands

Talk: C02.5 Comparison of exome and genome sequencing technologies for the complete capture of protein coding regions
Session: C02 Improvement in genome sequencing and functional studies
Date: Saturday, June 6, 2015, 18:30 hrs
Q1: 02-01-1987, Voorburg, The Netherlands
Q2: I am a bioinformatics PhD student at the Genomics Disorders Group in the Radboud University Medical Centre Nijmegen
Q3: A career in genetics gives me the opportunity me to combine the fields of biology, computer science and statistics to analyse large genomic datasets. This will help us better understand how mechanisms in disease work and provide important answers to patients and their families.
Q4: We investigated whether whole genome sequencing offer improved coverage of coding regions compared to whole exome sequencing, and compared single-base coverage for a large set of exome and genome samples. Our findings will guide laboratories to make an informed decision on which sequencing platform and coverage to choose.

Darío Lupiáñez
Berlin, Germany

Talk: PL2.4 Disruptions of topological chromatin domains cause pathogenic rewiring of gene-enhancer interactions
Session: PL2 "What's New?" Highlights Session
Date: Saturday, June 6, 2015, 18:30 hrs
Q1: 04-11-81 Algeciras (Spain)
Q2: Postdoctoral researcher at the Max Planck Institute for Molecular Genetics
Q3: I have been always intrigued about how life operates, and genetics has the key to answer this question. In that sense, genetics gives me the opportunity to understand biological processes and how they relate to disease and evolution.
Q4: Our research focuses on the 3D folding of the genome in the nucleus. We demonstrate how structural variations can affect this genomic organization and cause developmental disorders in humans. Consequently, we present a model to predict the pathogenic effects of structural variations.

Jennie Murray
Edinburgh, United Kingdom

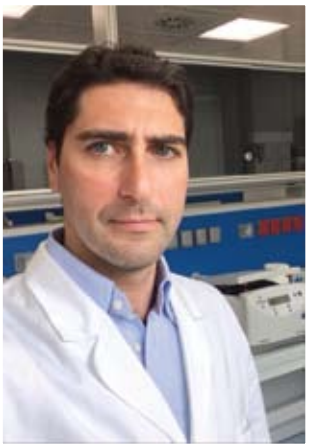
Talk: C16.2 Mutations in the core NHEJ components LIG4 and XRCC4 result in microcephalic primordial dwarfism
Session: C16 Growth failure and microcephaly
Date: Monday, June 8, 2015, 13:30 hrs

Mike Nahorski
Cambridge, United Kingdom

Talk: C12.1 A novel disorder reveals Clathrin Heavy Chain-22 is essential for human pain and touch development
Session: C12 Sensory disorders
Date: Sunday, June 7, 2015, 13:30 hrs
Q1: 11/07/1987, Great Yarmouth, UK
Q2: I am currently a Post Doctoral Research Associate at the Cambridge Institute of Medical Research, University of Cambridge.
Q3: I chose a career in genetics having secured a placement in a human genetics laboratory one summer which really opened my eyes as to what fun a career in research could be. I thought that medicine would eventually be revolutionized by next generation genetic technologies, and really wanted to be a part of that. It has been fascinating to work on the more molecular biological aspects of human disease genetics. The identification of causative gene mutations in numerous rare syndromes have provided novel and often unexpected insights into basic molecular biology and I am excited to see many of these insights now directly translating back into the clinic.
Q4: Individuals who suffer from Mendelian disorders of painlessness are unable to sense any type of pain anywhere in their body.

Despite only a few genes having been identified to date, they are now directly translating into the development of novel analgesics for people feeling excess pain. My talk will describe a novel cause of painlessness; mutations in the CLTCL1 gene encoding the second clathrin heavy chain (CHC22), a developmental role for CHC22 in pain and touch neuron development and early insights into the role of CHC22 in endosomal trafficking. My hope is that investigations into CHC22 function might provide similar advances in the treatment of pain.

Marcello Niceta
Roma, Italy



Talk: C21.6 Mutations impairing GSK3-mediated MAF phosphorylation cause cataract, deafness, intellectual disability, seizures, and a Down syndrome-like facies.
Session: C21 Multiple congenital anomaly syndromes
Date: Tuesday, June 9, 2015, 11:00 hrs
Q1: 01-04-1975
Q2: Research Fellow/PhD Student
Q3: I do believe there is always a genetic explanation permitting us to understand how life can exist at all levels of its complexity.
Q4: New genetic technologies capable of advancing knowledge on complex medical conditions.

Gaël Nicolas
Rouen, France

Talk: C10.6 De novo deleterious genetic variations target a biological network centered on Aβ peptide in early-onset Alzheimer disease
Session: C10 Neurogenetic disorders
Date: Sunday, June 7, 2015, 13:30 hrs
Q1: 21th July 1984, Le Havre, Normandy, France
Q2: MD, PhD student
Q3: To identify the molecular bases of rare diseases with Mendelian inheritance as well as complex disorders. To better understand the pathophysiology of rare diseases. To be able to report this to the patients and their families. To provide them genetic counselling and personalized medicine.
Q4: We highlight the role of de novo mutations in early onset Alzheimer's disease (EOAD). While autosomal dominantly-inherited forms of the disease are well characterized, very few is known about sporadic EOAD. Using a two-step procedure (array-CGH followed by whole exome sequencing of patient-unaffected parents trios), we identified the first de novo APP duplication and a de novo PSEN1 pathogenic variant in two patients. Beyond this proof of concept, we identified further de novo variants that fell into a biological network linked to the Abeta peptide, the agregation of which being a key pathogenic event in AD. Finally, we demonstrate the functional role for two of them. Despite the difficulty to recruit AD trios due to age structures of the pedigrees and the genetic heterogeneity of the disease, exploring the de novo paradigm in this adult onset disease using this strategy allowed us to highlight the role of de novo pathogenic events, the putative involvement of new genes in AD genetics and the key role of Abeta network alteration in AD.

Asmundur Oddsson
Reykjavik, Iceland

Talk: C18.6 Common and rare variants associated with kidney stones and biochemical traits
Session: C18 Metabolic and renal disorders
Date: Monday, June 8, 2015, 13:30 hrs
Q1: 14.5.2015, Akureyri, Iceland
Q2: Research associate
Q3: Modern sequencing technology has pushed back the frontier of biological knowledge. Therefore, genetics is the field today were biology can be practised at the leading edge.
Q4: Whole-genome sequencing of 2,636 Icelanders and imputation into >100k long-range phased individuals and their relatives has created a unique resource of genetic information. Making use of this resource allowed us to identify both rare and common variants associating with kidney stone disease.

Patricia Oliveira
Porto, Portugal

Talk: C04.6 Through the looking glass: the reversion of EMT
Session: C04 The many faces of cancer mutations
Date: Saturday, June 6, 2015, 18:30 hrs
Q1: November 13th, 1982 in Coimbra, Portugal
Q2: Currently I am a postdoctoral research fellow at the Expression Regulation in Cancer Group at Ipatimup in Porto, Portugal.
Q3: Genetics is the most powerful mechanism for Life as we know it. Just as a comet, Genetics travels through time and space, across populations and within organisms. And as a comet, Genetics can crash and burn, originating remarkable evolutionary leaps or catastrophic diseases, such as Cancer. It is this inherent duality of Genetics and all its (epi)layers, that drew me to this field, in the unwavering hope of finding novel mechanisms to fight Cancer.
Q4: Our findings are above all else, challenging, both in terms of our hypothesis as well as in current state of the art. Unlike EMT which has been extensively associated with cancer progression, MET is an underdog process, often viewed as a mirror of EMT. We hypothesized and proved that MET is a permissive process with a particular transcriptional signature, manipulating cellular plasticity generating heterogeneity. In fact, cellular heterogeneity is a common phenomenon observed in human tumour samples, thought to underlie drug resistance, a major pitfall in current cancer treatment regimens. Our findings highlight MET as a significant process and pinpoint novel biological pathways relevant to understand tumour heterogeneity and cancer progression.

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Agnese Padula
Naples, Italy

Talk: C15.4 Finding new connections in the transcriptional regulation of Lysine-specific demethylase 5C (KDM5C) a disease gene involved in syndromic and non-syndromic XLID
Session: C15 Network and functional analysis in intellectual disability
Date: Monday, June 8, 2015, 13:30 hrs
Q1: 22/08/1988 Naples
Q2: PhD student
Q3: Because since i started to study this subject at the university, i was fascinated by this topic, and i would like to study it in more deep looking also at its applications.
Q4: I think is interesting that i present a research in which i show a transcriptional path including different genes that cause similar phenotypes.

Michael Parks
Birmingham, United Kingdom

Talk: C01.4 Non-invasive prenatal diagnosis (NIPD) of Duchenne and Becker muscular dystrophies (DMD/BMD) by relative haplotype dosage
Session: C01 NIPT
Date: Saturday, June 6, 2015, 18:30 hrs
Q1: 03/06/1985 in Verona, Italy
Q2: Developmental Scientist at West Midlands Regional Genetics Laboratories
Q3: The world of genetics has fascinated me since my first biology class in high school. The profound and at times fatal impact that our genes have on our lives has constantly fueled my first for knowledge and understanding of the human genome. Driven by an unrelenting desire to use and improve my skills to help others, I am now in the position to make a real difference in people's lives through my research.
Q4: Non-invasive prenatal diagnosis is revolutionizing the field of prenatal genetics. By working at the forefront of this field, my research provides novel insights into the implementation of NIPD for single gene disorders in a clinical setting.

Louise Porter
Manchester, United Kingdom

Talk: C12.6 A molecular network surrounding dysregulated H3K9 di-methylation in PRDM5-associated disease
Session: C12 Sensory disorders
Date: Sunday, June 7, 2015, 13:30 hrs
Q1: 15/10/1979, London
Q2: Clinical research fellow in ophthalmology and genetics
Q3: I have chosen a career in ophthalmo-genetics as it provides a stimulating patient-focussed clinical and research environment in which to advance diagnosis, understanding of pathogenesis, and therapies in rare diseases.
Q4: The study of clinical samples from patients with brittle cornea syndrome type 2 (BCS2) has provided an opportunity to analyze the impact of mutations affecting a transcription factor, PRDM5, on a repertoire of associated epigenetic modifiers. I propose a role for defective interaction of repressive complexes and H3K9 di-methylation in BCS2. My observed dysregulation of epigenetic regulatory mechanisms in BCS2 suggests that epigenetic modifications may be a more widespread disease mechanism in inherited eye disease.

Margot Reijnders
Nijmegen, Netherlands

Talk: C03.5 De novo loss-of-function mutations in WAC in the 10p12p11 critical region cause intellectual disability
Session: C03 Novel genes causing intellectual disability
Date: Saturday, June 6, 2015, 18:30 hrs
Q1: 27-07-1989, Roosendaal
Q2: PhD student in Clinical Genetics
Q3: Although a lot already has been discovered in genetics, even more is still unknown. I really like to be part of the research team who tries to unravel small pieces of the unsolved puzzle of human genetics. With this new knowledge, we are able to give more patients a diagnosis and to further improve the care for them.
Q4: WAC is one of the genes which is already for years a candidate gene for intellectual disability. These days, many different genetic tests are available to identify additional patients. We used several of these techniques and performed functional studies in Drosophila to further establish to role of WAC in the development of intellectual disability. With de novo mutations in WAC in ten patients, we show that these mutations result in a new intellectual disability syndrome with a broad clinical spectrum.

Eva Reinthaler
Vienna, Austria

Talk: C10.3 Rare variants in GABAA receptor genes in Rolandic epilepsy and related syndromes
Session: C10 Neurogenetic disorders
Date: Sunday, June 7, 2015, 13:30 hrs
Q1: 26/05/1983, Rohrbach, Austria
Q2: Postdoc
Q3: I like to work with the complex genetic information of human beings. I am fascinated by the diversity of genetic alterations and its contribution to health and disease and how information is transferred from generation to generation. I am fascinated by the fact that changes in our DNA can cause a disease in one person and have almost no effect in the other.
Q4: We report a mutation screening of 18 GABA receptor genes in familial and sporadic idiopathic focal childhood epilepsies patients. We show a statistical association and functional evidence of mutations in GABRG2 with typical and atypical Rolandic epilepsy. This illustrates that GABAergic mechanisms participate in the etiology of idiopathic focal epilepsies.

Julie Rutten
Leiden, Netherlands

Talk: C10.5 Cysteine Correction of NOTCH3: exon skipping as a potential therapeutic strategy for CADASIL
Session: C10 Neurogenetic disorders
Date: Sunday, June 7, 2015, 13:30 hrs
Q1: 8/1/1984, Breda, The Netherlands
Q2: PhD student at the departments of Human and Clinical Genetics, Leiden University Medical Center in the Netherlands.
Q3: During my training as a medical doctor, I was caught by the impact which hereditary diseases have on the life of not only the patient, but also the patient's family. Advances in the diagnosis of genetic diseases, but also the prospect of future therapies, make this a very interesting, relevant and exciting field to work in both as a medical doctor and as a scientist
Q4: What I find most interesting about this research are the various aspects that come into play when developing a therapy for a relatively unknown disease such as CADASIL. We have developed a therapeutic approach which is based on antisense-mediated exon skipping. We use exon skipping in an innovative way, namely to restore cysteine residues in the NOTCH3 protein, in order to prevent protein accumulation. Development of this strategy requires knowledge of the mutation characteristics and the pathogenic mechanism in CADASIL. Also, we developed a mouse model which recapitulates the disease, and are working on the development of biomarkers and read outs in mouse and man. These various aspects make the research versatile and challenging. During this conference, I will present our pre-clinical proof of concept studies on this exon skipping approach for CADASIL.

Miriam Schmidts
Nijmegen, Netherlands

Talk: C11.1 Mutations in a novel dynein-2 light chain, TCTEX1D2, cause Jeune Asphyxiating Thoracic Dystrophy (JATD) with incomplete penetrance
Session: C11 Skeletal disorders
Date: Sunday, June 7, 2015, 13:30 hrs

Lot Snijders Blok
Nijmegen, Netherlands

Talk: C03.1 De novo and familial DDX3X mutations are associated with X-linked intellectual disability and a diverse phenotypic spectrum
Session: C03 Novel genes causing intellectual disability
Date: Saturday, June 6, 2015, 18:30 hrs
Q1: 24-4-1987, Sneek
Q2: Clinical Geneticist in training
Q3: I'm intruiged by the complexity of the human genome and that there are still a lot of things we don't understand. And next to that, I love the fact that complex molecular mechanisms and patient care come together in this field every day.
Q4: At the moment I'm working on the DDX3X gene, a X-linked gene in which mutations can cause a complex neurodevelopmental phenotype in females and in males. What I think is very interesting about this gene is that different missense mutations that are close to each other in the genome are associated with different X-linked inheritance patterns. And also interesting: while we are working on it, mutations in DDX3X turn out to be a very frequent cause of intellectual disability in girls.

Georgios Stamoulis
Geneva, Switzerland

Talk: C13.4 Single-cell allele specific expression (ASE) in T21: a novel approach to understand Down syndrome.
Session: C13 Fundamental insights in structural genomics
Date: Monday, June 8, 2015, 13:30 hrs
Q1: 4/3/1986, Thebes, Greece
Q2: PhD student at Stylianos Antonarakis' laboratory in the University of Geneva, Switzerland, at the Department of Genetic Medicine and Development
Q3: Since I was a student at school I was always fascinated by health and life sciences, because I was intrigued by the complexity of how our body works. I decided to study Genetics and continue my career in the field by doing an MSc in Medical Genetics and now a PhD in Genetics, because I strongly believe that our genome is the book of life, which contains the answers to the all the questions related to our health and disease state. I believe genetic research will shed light in many unanswered questions and will open a new era in medicine in the near future by generating the new field of personalized medicine.
Q4: In this study we explore the allele specific expression (ASE)

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on a single cell level in Trisomy 21 (Down syndrome) and common aneuploidies for the first time, using transcriptome studies in single cells. In our study we used a pair of monozygotic twins discordant for T21 and mosaic cells from affected individuals with other common aneulopidies in order to eliminate the interindividual variability in expression profile. Such studies can help to reveal important biological insights regarding the cellular impact of aneuploidy and elucidate the fundamental mechanisms of gene dosage.

Naeimeh Tayebi
Berlin, Germany

Talk: C11.3 Mutations in ZAK cause autosomal recessive split foot malformation in humans and complex hindlimb defects in mice
Session: C11 Skeletal disorders
Date: Sunday, June 7, 2015, 13:30 hrs
Q1: 27.08.1979-Yazd, Iran
Q2: Currently I am a PhD student in Molecular Human Genetics in Max Planck Institute for Molecular Genetics, Berlin, Germany
Q3: I have always been excited by studying human genetic disorders in order to realize what the cause of this specific disorder is and if the phenotype of the patients is unusual with unknown pathogenic cause , how I can address what the genetic cause of this strange phenotype is and how I can help the families to prevent from the same genetic disease in next pregnancy.
Q4: I am interested in exploring how genes relate to skeletal malformation. According to our cohort, no molecular diagnosis has been made for nearly 70% of split-hand-foot malformation cases. Therefore, finding and investigating of novel genes in these malformations are fascinating. In this presentation, I would like to present a novel gene, ZAK, that the mutations within this gene were identified in two families who suffered from split-foot malformation and hearing loss. To date, the precise biological functions of ZAK are not yet quite understood and so far, no evidence has suggested that ZAK gene has a role in limb development. In addition, the most interesting part of my presentation is generating a modified mouse line using CRISPR-Cas system in order to find the mechanism of ZAK in limb bud development.

Laura Tomas Roca
Nijmegen, Netherlands

Talk: PL2.1 De novo mutations in PLXND1 and REV3L cause Möbius syndrome
Session: PL2 "What's New?" Highlights Session
Date: Saturday, June 6, 2015, 18:30 hrs
Q1: 01.10.1984, Murcia, Spain
Q2: Postdoc
Q3: Genetics is the basic of life
Q4: I am showing for the first time the etiology of Möbius syndrome.

Taru Tukiainen
Boston, United States

Talk: C17.2 Pattern of X chromosome inactivation across human tissues - insights from population-scale and single-cell RNA sequencing
Session: C17 Epigenetic control of gene expression
Date: Monday, June 8, 2015, 13:30 hrs
Q1: March 10, 1983, Helsinki, Finland
Q2: I'm a research fellow at the Massachusetts General Hospital and the Broad Institute of MIT and Harvard, in Boston.
Q3: Genetics beautifully combines biology, medicine, statistics, and technology.
Q4: We're exploring X chromosome inactivation on several layers of biology – from population to single cells, across multiple tissue types, between and within individuals – utilizing RNA sequencing and the capability of this technology to capture the transcriptome at base-pair resolution.

Marlinde van den Boogaard
Leiden, Netherlands

Talk: C06.2 The SMCHD1 mutation spectrum in Facioscapulohumeral muscular dystrophy
Session: C06 Neuromuscular disorders
Date: Saturday, June 6, 2015, 18:30 hrs
Q1: 06/02/1989, Woerden, The Netherlands
Q2: I am working as a PhD student in the department of Human Genetics, Leiden University Medical Center, Leiden, The Netherlands
Q3: For me genetics is like a giant puzzle, and many more pieces need to be put together. I find it fascinating to participate in research on the genetic mechanisms behind diseases. I believe that unravelling these genetic mechanisms will be very important for the development of therapies.
Q4: Facioscapulohumeral muscular dystrophy (FSHD) is characterized by extreme inter- and intrafamilial clinical variation in onset, progression and severity. Our research aims at deciphering the molecular basis for this variability and to identify modifiers of disease severity. This will have prognostic value and it is to be expected that it will provide new opportunities for therapeutic intervention.

PROGRAMME YOUNG INVESTIGATOR AWARD CANDIDATES

Danya Vears
Leuven, Belgium

Talk: C20.3 Should children's carrier results be reported following diagnostic WES/WGS?
Session: C20 Current issues in genet(h)ics
Date: Tuesday, June 9, 2015, 11:00 hrs
Q1: 12/05/1981 Melbourne
Q2: PhD student
Q3: I have always loved genetics as a field. After undertaking a Masters in Genetic Counselling, I became very passionate about the ethical issues relating to genetic testing in children. This drove me to commence my PhD in this field and pursue a career in bioethics.
Q4: While whole genome/exome technology is an exciting new world which has opened up unique research opportunities, it also creates many new ethical challenges, particularly relating to how to manage genetic information and what should be reported to patients. As children cannot be involved in these decisions themselves, the use of their results from whole genome/exome sequencing for diagnostic purposes requires careful consideration.

Michael Volodarsky
Beer-Sheva, Israel

Talk: C06.6 CCDC174 mutation underlies a syndrome of hypotonia and psychomotor developmental delay with abducens nerve palsy
Session: C06 Neuromuscular disorders
Date: Saturday, June 6, 2015, 18:30 hrs
Q1: Feb 1981, Saint-Petersburg
Q2: PhD student
Q3: Genes are the driving force of our appearance, behavior and existence. Rare mutations may alter their proper function, revealing the dark side of genes. I chose this career because hunting these mutations leads to exciting discoveries and enable testing for many carrier families.
Q4: The presented research enables a glance into a rare genetic event. The same mutation was found in a narrow haplotype shared by two families belonging to different ethnic groups. Moreover, a novel hypotonia causing gene is presented, acting in a still not fully understood pathway.

Sergi Villatoro
Bellaterra (Barcelona), Spain

Talk: C02.2 Large-scale genotyping of polymorphic inversions in the human genome
Session: C02 Improvement in genome sequencing and functional studies
Date: Saturday, June 6, 2015, 18:30 hrs
Q1: January 27th, 1977, Sabadell, Barcelona (Spain)
Q2: Right now, I am working as specialist technician and simultaneously I am trying to get my PhD degree in my spare time.
Q3: Because, since I heard about genes and how they might determine phenotypic traits and/or they could be involved in illness, always I wanted to figure out this relationship. In particular, I am interested in deciphering how structural variation could alter human features and occasionally leading to disease.
Q4: My research opens the door for a new knowledge in a kind of variation, inversions, that have barely studied so far, especially due to the technical difficulties to detect them. Besides, thanks to the new method that we have developed to genotype inversions in a high-throughput way is possible carry out population's studies and obtain information about their geographical distribution, evolutionary importance and determine the functional consequences of these inversions. Finally, this new source of information might shed light on long standing questions like missing heritability and the genetic architecture of complex traits.

Robbert Weren
Nijmegen, Netherlands

Talk: PL2.5 A germline homozygous loss-of-function mutation in the base excision repair gene NTHL1 causes adenomatous polyposis and colorectal cancer
Session: PL2 "What's New?" Highlights Session
Date: Saturday, June 6, 2015, 18:30 hrs
Q1: 13-12-1985, Heythuysen, the Netherlands
Q2: I am currently working as a PhD student at the Department of Human Genetics, Radboud university medical center, Nijmegen, the Netherlands.
Q3: I have always been intrigued by the fact that heritable traits underlie different phenotypes, especially regarding human health. I started a career in genetics to unravel why individuals/families, without a known genetic cause, show high-penetrant predisposition to develop specific diseases. By studying cancer genetics, I hope to provide answers to the question why some individuals are more prone to develop (colon) cancer in their lives and, thereby, make a significant contribution to improve the genetic counseling and clinical management of these patients.
Q4: Adenomatous polyposis, the constitutive development of adenomas in the colon and rectum, is strongly associated with heritable germline aberrations. However, a large subset of adenomatous polyposis patients remains unexplained. We recently have shown, for the first time, that germline aberrations in NTHL1 underlie high-penetrant predisposition to the development of adenomatous polyposis and colorectal cancer in an autosomal recessive manner.

PROGRAMME YOUNG INVESTIGATOR AWARD CANDIDATES

Thomas Wieland
München, Germany

Talk: C23.1 TBK1 mutations cause amyotrophic lateral sclerosis and fronto-temporal dementia
Session: C23 Movement and motor disorders
Date: Tuesday, June 9, 2015, 11:00 hrs
Q1: 12.06.1986, Scheibbs(Austria)
Q2: I am a PhD student at the Institute of Human Genetics at the Helmholtz Zentrum München.
Q3: In my opinion the field of human genetics and especially of Next-Generation Sequencing presented the most interesting and challenging opportunities for me as a bioinformatician.
Q4: Using rare variant association tests on exome sequencing data, we were able to identify TBK1 as a new susceptibility gene for ALS. This will help to learn more about the molecular basis of the disease.

Charley Xia
Edinburgh, United Kingdom

Talk: C08.3 Pedigree-Associated Genetics and Recent Environment Make Important Contributions to Metabolic Syndrome Traits.
Session: C08 Integrative OMICS approaches in common traits
Date: Sunday, June 7, 2015, 13:30 hrs
Q1: 23-10-1989
Q2: I am a 2nd-year PhD student of University of Edinburgh.
Q3: My grandfather is a Parkinson disease patient and my grandma has diabetes and hypertension, all of which are partly heritable. Seeing them suffered, I've made my mind to devote myself to helping people like them, including myself, to stop suffering from or minimize the probability to suffer from heritable diseases.
Q4: We found that current environment as couples and variants not in LD with SNP array but in LD with pedigree are important for human complex traits related to metabolic syndrome, in addition to SNP effects. Our findings reveal a plausible trait architecture as well as point to appropriate models for future studies.

Ryan Yuen
Toronto, Canada

Talk: C10.1 Whole genome sequencing reveals the mutation characteristics in Autism Spectrum Disorder
Session: C10 Neurogenetic disorders
Date: Sunday, June 7, 2015, 13:30 hrs

Alena Zablotskaya
Leuven, Belgium

Talk: C02.3 Large-scale single-molecule sequencing of tandem repeats on the human X chromosome
Session: C02 Improvement in genome sequencing and functional studies
Date: Saturday, June 6, 2015, 18:30 hrs
Q1: 30 September 1986, Minsk
Q2: PhD student
Q3: I got fascinated by the other Universe which is hidden inside our cells, and its laws of life.
Q4: At the conference I am presenting how we perform a large-scale genotyping of tandem repeats and search for disease associated repeat expansions in familial cases, where traditional techniques failed to detect causal variation. This allows to approach a large pool of phenotypically important genetic variation in humans that remains understudied.

Daria Zhernakova
Groningen, Netherlands

Talk: C08.2 Genetic variants affect expression of nearly all genes, but only in a specific context
Session: C08 Integrative OMICS approaches in common traits
Date: Sunday, June 7, 2015, 13:30 hrs
Q1: 14-04-1988, St. Petersburg, Russia
Q2: PhD student, Genetics Department, University Medical Center Groningen
Q3: For me studying genetics is a way to learn how Life is organized and how it works.
Q4: I present a project based on RNA-sequencing of 2,116 healthy Dutch samples. We found that expression of most genes are regulated by genetic variants, often only in a specific context.

PROGRAMME POSTER AWARD FINALISTS

ESHG Poster Awards

For the eighth time, the ESHG proposes the ESHG Poster award for the best posters presented by Young Investigators at the meeting. The two winners (one in clinical the other in basic research) will receive a prize money of EUR 500. The five honorable mentions receive a complementary ESHG online membership for one year.

The ESHG Scientific Programme Committee has selected a number of candidates for the ESHG Poster Award. Candidate posters can be identified by a rosette on the board.

ESHG Poster Award Candidates

PS01.09
Expanded carrier screening of 311,688 individuals: the case for going beyond CF
I. S. Haque, *G. A. Lazarin, M. Raia, H. Bellerose, E. A. Evans, J. Goldberg;*
Counsyl, South San Francisco, CA, United States.

PS01.43
Non-Invasive Prenatal Testing for the most common aneuploidies (trisomies 21, 18, and 13) using a semiconductor-sequencing platform: a French multicenter pilot study
S. Brun[1], P. Gueguen[2], L. El Khattabi[3], N. Chatron[4], J. Nectoux[5], S. Schutz[2], J. Pipoli da Fonseca[6], E. Guichoux[7], A. Sorlin[8], M. Quere[9], J. Boudjarane[10], C. Bonnet[8], F. Letourneur[6], C. Schluth-Bolard[4], P. Jonveaux[8], C. Bardel[11], V. Paquis-Fluckinger[9], S. Bannwarth[9], B. Arveiler[12], M. Goossens[13], C. Badens[10], J. Dupont[3], **C. Rooryck**[12], *D. Sanlaville[4], C. Ferec[2], M. Vidaud[5];*
[1]Maternité Centre Aliénor d'Aquitaine, CHU Bordeaux, Bordeaux, France, [2]Laboratoire de génétique moléculaire, INSERM U1078, CHRU de Brest, Brest, France, [3]Service de Cytogénétique, APHP-HUPC, INSERM U1016, Université Paris Descartes, Paris, France, [4]HCL, Service de Génétique, UCBL1, Lyon, France, [5]Service de Biochimie et Génétique Moléculaire, HUPC Hôpital Cochin, Paris, France, [6]Plateforme génomique – Inserm U1016, Paris, France, [7]Plateforme Génome Transcriptome de Bordeaux, INRA Cestas, Bordeaux, France, [8]Service de génétique-CHRU Nancy-INSERM U954-Université de Lorraine, Nancy, France, [9]Service de Génétique Médicale, Hôpital de l'Archet II, CHU de Nice, Nice, France, [10]Département de génétique médicale, CHU Timone, Marseille, France, [11]HCL, Service de Biostatistique, CNRS UMR 5558, UCBL1, Lyon, France, [12]Service de Génétique Médicale, CHU Bordeaux, Université Bordeaux, Bordeaux, France, [13]Service de Biochimie et Génétique Moléculaire, HUPC Hôpital Cochin, Bordeaux, France.

PS02.05
SF3B2, a novel candidate gene for autosomal dominant retinitis pigmentosa, encodes a component of the U2 small nuclear ribonucleoprotein
C. Van Cauwenbergh[1], *M. Karlstetter[2], K. Vleminckx[1,3], G. Manes[4], T. Langmann[2], C. Hamel[4], European Retinal Disease Consortium (ERDC), B. P. Leroy[5,6], F. Coppieters[1], E. De Baere[1];*
[1]Center for Medical Genetics, Ghent University, Ghent, Belgium, [2]Department of Ophthalmology, University of Cologne, Cologne, Germany, [3]Department of Biomedical Molecular Biology, Ghent University, Ghent, Belgium, [4]INSERM U1051, Institut des Neurosciences de Montpellier, Montpellier, France, [5]Dept of Ophthalmology, Ghent University Hospital, Ghent, Belgium, [6]Division of Ophthalmology, The Children's Hospital of Philadelphia, Philadelphia, PA, United States.

PM02.30
NGS revealed PSIP1/LEDGF as a new gene causing sensorineural progressive hearing loss and variable eye phenotypes
G. Girotto[1], *D. I. Scheffer[2], A. Morgan[1], D. Vozzi[3], D. Vuckovic[4], E. Rubinato[1], M. Di Stazio[1], E. Muzzi[5], S. Pensiero[6], A. B. Giersch[7], J. Shen[7], N. Robertson[7], C. Morton[7], D. P. Corey[2], P. Gasparini[8];*
[1]Dep.Rep.Sciences,Dev.Pub.Health;IRCCS-Burlo Garofolo-Children Hospital-University of Trieste, Trieste, Italy, [2]Harvard Medical School-Howard Hughes Medical Institute, Department of Neurobiology, 220 Longwood Avenue Boston, MA 2115, MA, United States, [3]Medical Genetic Institute for Maternal and Child Health- IRCCS "Burlo Garofolo", Trieste, Italy, [4]University of Trieste-Department of Medical, Surgical and Health Sciences, Trieste, Italy, [5]Audiology and ENT Unit, Department of Pediatrics, Institute for Maternal and Child Health - IRCCS Burlo Garofolo, Trieste, Italy, [6]Ophthalmology Unit, Department of Pediatrics, Institute for Maternal and Child Health - IRCCS Burlo Garofolo, Trieste, Italy, [7]Harvard Medical School and Brigham and Women's Hospital, Department of Pathology, Boston, MA, United States, [8]University of Trieste-Department of Medical, Surgical and Health Sciences/ Medical Genetic Institute for Maternal and Child Health-IRCCS "Burlo Garofolo", Trieste, Italy.

PM03.10
Prioritization of candidate variants using targeted next generation sequencing of 208 candidate genes in congenital anomalies of the kidney and urinary tract
N. Nicolaou[1], S. Pulit[1], I. J. Nijman[1], G. Monroe[2], W. F. J. Feitz[3], M. F. Schreuder[4], A. M. van Eerde[1], J. C. Giltay[1], R. H. Giles[5], E. Cuppen[1], E. M. H. F. Bongers[6], N. V. A. M. Knoers[1], K. Y. Renkema[1], **G. van Haaften**[7];
[1]Department of Medical Genetics, University Medical Center Utrecht, Utrecht, Netherlands, [2]Medical Genetics, UMC UtrechtDepartment of Medical Genetics, University Medical Center Utrecht, Utrecht, Netherlands, [3]Department of Urology, Radboudumc Amalia Children's Hospital, Radboud university medical center, Nijmegen, Netherlands, [4]Department of pediatrics, Radboudumc Amalia Children's Hospital, Radboud university medical center, Nijmegen, Netherlands, [5]Department of Nephrology and Hypertension, University Medical Center Utrecht, Utrecht, Netherlands, [6]Department of Human Genetics, Radboud university medical center, Nijmegen, Netherlands, [7]UMC Utrecht, Utrecht, Netherlands.

PROGRAMME POSTER AWARD FINALISTS

PS04.23
Genetic heterogeneity and clinical variability in musculocontractural Ehlers-Danlos syndrome caused by impaired dermatan sulfate biosynthesis
D. M. Syx[1], *T. Van Damme*[1], *S. Symoens*[1], *M. C. Maiburg*[2], *I. van de Laar*[3], *J. Morton*[4], *M. Suri*[5], *M. Del Campo*[6], *I. Hausser*[7], *T. Hermanns-Lê*[8], *A. De Paepe*[1], *F. Malfait*[1];
[1]*Center for Medical Genetics, Ghent, Belgium,* [2]*University Medical Center Utrecht, Department of Medical Genetics, Utrecht, Netherlands,* [3]*Department of Clinical Genetics, Erasmus Medical Center, Rotterdam, Netherlands,* [4]*Clinical Genetics Unit, Birmingham Women's Hospital, Birmingham, United Kingdom,* [5]*Notttingham Clinical Genetics Service, Nottingham City Hospital, Nottingham, United Kingdom,* [6]*Area de Genetica Clinica y molecular. Hospital Vall d'Hebron, Barcelona, Spain,* [7]*Institute of Pathology, University Clinic Heidelberg, Heidelberg, Germany,* [8]*Department of Dermatopathology, Liège University Hospital, Liège, Belgium.*

PM04.24
The Ehlers-Danlos syndrome type VI spectrum: a genetically heterogeneous group of clinically overlapping conditions
T. Van Damme, *D. Syx, S. Symoens, A. De Paepe, F. Malfait; Center for Medical Genetics, Ghent University Hospital, Ghent, Belgium.*

PM04.54
Targeted sequencing of the Paget's disease associated 14q32 locus identifies several missense coding variants in RIN3 that predispose to Paget's disease of bone
M. Vallet[1], *D. C. Soares*[2], *S. Wani*[1], *A. Sophocleous*[1], *J. Warner*[3], *D. M. Salter*[1], *S. H. Ralston*[1], *O. M. E. Albagha*[1];
[1]*University of Edinburgh, Institute of Genetics and Molecular Medicine, Edinburgh, United Kingdom,* [2]*University of Edinburgh, Centre for Molecular Medicine, Edinburgh, United Kingdom,* [3]*South East Scotland Clinical Genetic Service, Centre for Genomic and Experimental Medicine, Western General Hospital, Edinburgh, United Kingdom.*

PM04.58
Whole Exome Sequencing as a novel tool for the detection of modifier genes in Pseudoxanthoma elasticum
E. Y. G. De Vilder[1,2,3], *F. Van Nieuwerburgh*[4], *D. Deforce*[4], *L. Martin*[5], *G. Lefthériotis*[6], *P. Coucke*[1], *A. De Paepe*[1], *O. M. Vanakker*[1];
[1]*Center for Medical Genetics, Ghent University Hospital, Ghent, Belgium,* [2]*PhD Fellow of the Research Foundation – Flanders, Ghent, Belgium,* [3]*Department of Ophthalmology, Ghent University Hospital, Ghent, Belgium,* [4]*Department of Pharmaceutics, Laboratory of Pharmaceutical Biotechnology, Ghent University, Ghent, Belgium,* [5]*Department of Dermatology, Angers University Hospital, Angers, France,* [6]*Department of Vascular Physiology and Sports Medicine, Angers University, Angers, France.*

PM04.76
Type III collagen is important for collagen fibrillogenesis and for dermal and cardiovascular development
S. D'hondt[1], *B. Guillemyn*[1], *S. Symoens*[1], *W. Toussaint*[2], *L. Vanhoutte*[2], *R. De Rycke*[2], *P. Coucke*[1], *B. Lambrecht*[2], *P. Segers*[3], *A. De Paepe*[1], *S. Janssens*[2], *M. Bertrand*[2], *F. Malfait*[1];
[1]*Center for Medical Genetics, UGent, Ghent, Belgium,* [2]*Inflammation Research Center, UGent, Ghent, Belgium,* [3]*Institute Biomedical Technology, UGent, Ghent, Belgium.*

PM05.30
A loss-of-function mutation in the haptoglobin gene is associated with a decrease in serum haptoglobin and an increase in non-HDL cholesterol and cardiovascular risk
E. Bjornsson[1,2], *P. Sulem*[1], *H. Holm*[1,3], *A. Helgadottir*[1,2], *H. Helgason*[1,2], *S. Gretarsdottir*[1], *A. Oddsson*[1], *R. P. Kristjansson*[1], *I. Olafsson*[3], *D. F. Gudbjartsson*[1,2], *G. Thorgeirsson*[2,3], *U. Thorsteinsdottir*[1,2], *K. Stefansson*[1,2];
[1]*DeCODE Genetics, Reykjavik, Iceland,* [2]*University of Iceland, Reykjavik, Iceland,* [3]*Landspitali University Hospital, Reykjavik, Iceland.*

PM05.38
The type of variants at the COL3A1 gene associates with the phenotype and severity of vascular Ehlers-Danlos syndrome
J. M. Albuisson[1,2,3], *M. Frank*[1,2], *B. Ranque*[4,5,3], *L. Golmard*[1,3], *J. Mazzella*[1], *L. Bal-Theoleyre*[6], *A. Fauret*[1,3], *T. Mirault*[1,3], *N. Denarie*[1], *E. Mousseaux*[7,3], *P. Boutouyrie*[8,2,3], *J. Fiessinger*[1,3], *J. Emmerich*[1,3], *E. Messas*[1,3], *X. Jeunemaitre*[1,2,3];
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PM06.22
Impaired mitochondrial RNA processing in HSD10 disease
A. J. Deutschmann[1], *A. Amberger*[1], *J. A. Mayr*[2], *S. Oerum*[3], *W. W. Yue*[3], *J. Zschocke*[1];
[1]*Division of Human Genetics, Innsbruck, Austria,* [2]*Paracelsus Medical University Salzburg, Salzburg, Austria,* [3]*University of Oxford, Oxford, United Kingdom.*

PM06.66
Thioredoxin 2 deficiency causes early-onset neurodegeneration
E. Holzerová[1,2], *K. Danhauser*[3], *L. S. Kremer*[1], *C. Terrile*[1], *T. B. Haack*[1], *H. Prokisch*[1,2], *F. Distelmaier*[3];
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PROGRAMME POSTER AWARD FINALISTS

PS06.53
High prevalence of monogenic obesity in super obese individuals undergoing bariatric surgery
S. I. M. Alsters[1], *A. M. Yiorkas*[1], *M. Mueller*[1], *A. Sosinsky*[1], *A. Zekavati*[1], *N. H. Ramzi*[1], *N. A. Nor Hashim*[1], *J. Murphy*[1], *H. S. Chahal*[1], *S. Purkayastha*[1], *A. R. Ahmed*[1], *M. M. van Haelst*[2], *C. W. le Roux*[3], *J. L. Buxton*[4], *R. G. Walters*[5], *A. I. F. Blakemore*[1];
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PM07.12
A DGKE intronic mutation explains genetically unsolved cases of familial atypical hemolytic uremic syndrome
C. Mele[1], *M. Lemaire*[2,3], *P. Iatropoulos*[1], *R. Piras*[1], *E. Bresin*[1], *S. Bettoni*[1], *D. Bick*[4,5], *D. Helbling*[4], *R. Veith*[5], *E. Valoti*[1], *R. Donadelli*[1], *L. Murer*[6], *M. Neunhäuserer*[7], *M. Breno*[1], *V. Frémeaux-Bacchi*[8], *R. Lifton*[2], *G. Remuzzi*[1,9], *M. Noris*[1];
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PM08.70
Fast and effective genome editing to study dominant de novo mutations: the WDR45 example
C. A. Biagosch[1,2], *S. Hensler*[1,2], *D. Janik*[3], *F. Neff*[3], *L. Becker*[4,5], *W. Wurst*[5,6], *T. Meitinger*[1,2], *H. Prokisch*[1,2];
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PS09.097
The PCSK6 intronic region associated with handedness controls expression of a novel shorter isoform
R. J. Shore[1], *K. Pettigrew*[1], *R. Diaz*[1], *Y. Xu*[1], *E. Wootton*[1], *L. Covill*[2], *W. Brandler*[2], *J. B. Talcott*[3], *D. F. Newbury*[2], *A. Monaco*[2], *J. Stein*[4], *S. Paracchini*[1];
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PM09.128
Dominance and recessiveness, two faces of the same coin? Illustration with two new genes in autosomal dominant spinocerebellar degenerations
M. Coutelier[1,2], *A. Durr*[3,4], *L. Burglen*[5,6,7], *C. Goizet*[8,9], *F. Habarou*[10], *D. Rodriguez*[5,6,11], *S. Morais*[1,12], *J. Konop*[1], *S. Chantot-Bastaraud*[5,7], *C. Rougeot*[5,13], *I. Alonso*[12], *C. Tallaksen*[1], *R. Schule*[14,15,16], *M. Janin*[10], *M. Cournelle*[17], *P. Coutinho*[12,18], *M. Milh*[19], *A. Toutain*[20], *A. Afenjar*[5,11], *S. Zuchner*[14], *G. Rouleau*[21], *G. Nicholson*[22], *J. Saudubray*[4], *F. Darios*[3], *J. Leal de Loureiro*[12,18], *D. Héron*[4], *C. Ottolenghi*[10], *F. Mochel*[3,4], *A. Brice*[3,4], *G. Stevanin*[1]; [1]*ICM, NEB, Sorbonne UPMC Univ Paris 06, INSERM, UMRS_1127, CNRS 7225 & EPHE, Paris, France,* [2]*Lab of Human Molecular Genetics, de Duve Inst, UCL, Brussels, Belgium,* [3]*ICM, NEB, Sorbonne UPMC Univ Paris 06, INSERM, UMRS_1127, CNRS 7225, Paris, France,* [4]*APHP, Genetics & Cytogenetics, Pitié-Salpêtrière, Paris, France,* [5]*Centre de Réf 'Malformations & maladies congénitales du cervelet', Paris-Lyon-Lille, France,* [6]*INSERM U1141, Paris, France,* [7]*APHP, Armand-Trousseau Hosp., Dep of Genetics, Paris, France,* [8]*Univ. Bordeaux, Labo Maladies Rares: Génétique & Métabolisme, EA4576, Bordeaux, France,* [9]*CHU Pellegrin, Génétique Méd., Bordeaux, France,* [10]*Metabolic Biochem. Lab, Necker-Enfants Malades, APHP & Descartes Univ, Paris, France,* [11]*APHP, Armand Trousseau Hosp, Neuropediatrics, UPMC Univ Paris 06, Paris, France,* [12]*UnIGENe, IBMC, I3S, ICBAS, Univ. do Porto, Porto, Portugal,* [13]*Hospices Civils de Lyon, HFME, Neuropédiatrie, Bron, France,* [14]*Dep of Human Genetics & Inst. for Human Genomics, Univ of Miami Miller School of Med., Miami, FL, United States,* [15]*Ctr for Neurology & Hertie Inst.for Clinical Brain Res, Eberhard- Karls-Univ., Tübingen, Germany,* [16]*DZNE, Eberhard-Karls- Univ., Tübingen, Germany,* [17]*CH du Pays d'Aix, Pédiatrie, Aix-en-Provence, France,* [18]*Neurologia, CH de Entre o Douro e Vouga, S.Maria da Feira, Portugal,* [19]*APHM, Neurologie pédiatrique, Hôp de la Timone, Marseille, France,* [20]*Génétique, Hôp Bretonneau, CHU, Tours, France,* [21]*Montreal Neurol. Inst & Hosp, Neurology & Neurosurgery, McGill U, Montreal, QC, Canada,* [22]*Northcott Neuroscience Lab, ANZAC, Mol.Medicine Lab, Concord Hospital; Univ.of Sydney, Sydney, Australia.*

PS11.063
Clinical utility of exome sequencing as a first-tier molecular test in infants suspected of having a monogenic disorder
Z. Stark[1], *T. Tan*[1,2], *B. Chong*[1], *G. Brett*[1], *P. Yap*[1], *M. Walsh*[1], *D. Amor*[1,3], *R. Savarirayan*[1,2], *G. McGillivray*[1], *A. Yeung*[1], *P. Ekert*[4,2], *C. Theda*[5], *S. Cowie*[1], *H. Peters*[6,2], *A. Boneh*[6,2], *J. Yaplito-Lee*[6], *M. Ryan*[6,2], *R. Leventer*[6,2], *I. Macciocca*[7], *N. Thorne*[7], *Melbourne Genomics Health Alliance, C. Gaff*[7,8], *S. White*[1,2];
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PROGRAMME POSTER AWARD FINALISTS

PS11.107
Efficient clearance of progerin through autophagy induction and SRSF-1 downregulation under MG132 treatment in Hutchinson-Gilford progeria syndrome
K. Harhouri[1], *C. Navarro*[1], *D. Depetris*[1], *M. Mattei*[1], *X. Nissan*[2], *P. Cau*[1,3], *A. De Sandre-Giovannoli*[1,4], *N. Lévy*[1,4];
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PM11.124
TMEM-107 is anchored to ring-like subdomains of the transition zone (TZ) membrane and organizes the TZ recruitment of ciliopathy transmembrane proteins
A. Bruel[1,2], *N. Lambacher*[3], *J. Van Dam*[4], *G. Slaats*[5], *K. Szymanska*[6], *J. Kennedy*[3], *K. Gaff*[3], *C. Johnson*[6], *R. Giles*[5], *T. Attie-Bitach*[7,8,9], *V. Cormier-Daire*[7,8,9], *S. Saunier*[8,10], *L. Burglen*[11,12], *L. Faivre*[2,1], *J. Rivière*[1,2], *M. Huynen*[4], *C. Thauvin-Robinet*[1,2], *O. Blacque*[3];
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PM12.122
Therapy response monitoring in patient with prostate cancer using plasma-Seq approach
J. Belic;
Institute of Human Genetics, Graz, Austria.

PS15.05
Non-invasive genomic profiling of bladder cancer using urinary cfDNA
F. S. Togneri[1], *R. T. Bryan*[2], *D. G. Ward*[2], *J. M. Foster*[3], *A. J. Devall*[2], *P. Wojtowicz*[1], *S. Alyas*[1], *F. Ramos Vasques*[1], *A. Oumie*[4], *N. D. James*[5], *K. K. Cheng*[6], *M. P. Zeegers*[7], *N. Deshmukh*[2], *B. O'Sullivan*[8], *P. Taniere*[8], *K. G. Spink*[4], *D. J. McMullan*[1], *M. Griffiths*[1];
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PM15.20
The plant cytokine kinetin as a potential therapeutic agent to correct CFTR splicing defects
L. Straniero[1,2], *G. Soldà*[3,2], *V. Rimoldi*[2], *R. Asselta*[3,2], *S. Duga*[3,2];
[1]University of Milan, Milan, Italy, [2]Humanitas Clinical and Research Center, Rozzano, Italy, [3]Humanitas University, Rozzano, Italy.

PS16.13
Genome-wide association study of 41 circulating cytokines
A. V. Ahola-Olli[1], *J. Kettunen*[2], *P. Würtz*[3], *N. Pitkänen*[4], *K. Aalto*[1], *M. Salmi*[1], *A. Havulinna*[1], *V. Salomaa*[5], *T. Lehtimäki*[6], *S. Jalkanen*[1], *O. Raitakari*[1];
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PM16.20
Transcriptome analysis of mouse ES cells carrying a human chromosome 21
A. Letourneau[1], *J. Groet*[2], *F. Santoni*[1], *C. Gehrig*[1,3], *M. Guipponi*[1,3], *C. Borel*[1], *V. L. J. Tybulewicz*[4], *E. M. C. Fisher*[5], *D. Nizetic*[2,6], **S. E. Antonarakis**[1,3,7];
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PROGRAMME POSTER AWARD FINALISTS

PM16.36
Clinical metabolomic profiling for the diagnosis of inborn errors of metabolism & undifferentiated genetic phenotypes
P. S. Atwal[1], *M. Miller*[1], *T. Donti*[1], *A. D. Kennedy*[2], *A. D. Eckhart*[2], *J. E. Wulff*[2], *M. V. Milburn*[2], *J. A. Ryals*[2], *A. L. Beaudet*[1], *Q. Sun*[1], *V. R. Sutton*[1], *S. H. Elsea*[1];
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PS16.59
Resolving Complex Structural Genomic Rearrangements using a Randomized Approach
X. Zhao, *S. B. Emery, J. M. Kidd, R. E. Mills;*
University of Michigan, Ann Arbor, MI, United States.

PM16.60
Sexpression analysis of >1,700 Finnish individuals reveals sex-dependent transcriptional differences in whole blood for immune system processes, response to stress and lipid metabolism
A. Joensuu[1,2], *M. Nuotio*[1,2], *V. Salomaa*[2], *T. Lehtimäki*[3], *O. Raitakari*[4,5], *M. Perola*[2,1,6], *J. Kettunen*[7,2,8];
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PM18.20
Common CNVs in a population-based cohort reveal several associations in the transcriptome and consecutive changes in the metabolome
H. Mattsson[1,2], *A. Joensuu*[1,2], *V. Salomaa*[1], *A. J. Kangas*[3], *P. Soininen*[3,4], *M. Ala-Korpela*[4,5,6], *J. Kettunen*[1,3,4], *M. Perola*[1,2,7];
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PS18.23
Exome-sequencing in a large family-based and population-based study identifies a large-effect missense variant associated with depression
N. Amin, *F. de Vrij, R. Brouwer, J. van Rooij, A. G. Uitterlinden, W. F. J. van IJcken, S. Kushner, H. Tiemeier, C. M. van Duijn;*
Erasmus MC, Rotterdam, Netherlands.

PS18.43
Regional variation in health-related traits in Scotland: genes or environment?
C. Amador[1], *A. Spiliopoulou*[1], *J. Huffman*[1], *A. Campbell*[1], *D. Porteous*[1], *G. Scotland*[2], *N. Hastie*[1], *V. Vitart*[1], *C. Hayward*[1], *P. Navarro*[1], *C. S. Haley*[1,3];
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PM18.78
Strategies to improve the performance of rare variant rare disease association studies by optimizing the selection of controls
N. Zhu[1], *V. Heinrich*[1], *T. Dickhaus*[2], *J. Hecht*[3], *P. Robinson*[1], *S. Mundlos*[1], *T. Kamphans*[4], *P. Krawitz*[1];
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PM20.06
Storage and future use of consumers' samples and data in direct-to-consumer genetic testing companies offering whole genome sequencing
E. Niemiec[1,2], *H. C. Howard*[3];
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INFORMATION

GENERAL INFORMATION

REGISTRATION FEES

NETWORKING EVENTS

CORPORATE EXHIBITION

INFORMATION GENERAL INFORMATION

IMPORTANT NOTICE :
Please note that taking pictures or filming during the sessions is forbidden (no matter if done with a camera or a mobile phone). Persons who will not observe this rule will be excluded from the session by the chairpersons.

Conference Venue
SECC - Scottish Exhibition and Conference Centre
Exhibition Way, Glasgow G3 8YW, Scotland, United Kingdom
www.secc.co.uk

Badges
Participants should collect name badges from the conference registration desk. As only registered participants will be permitted to attend the scientific sessions, the exhibition and poster areas, you are required to wear your badge when entering and while remaining in the congress venue.
Accompanying persons and exhibitors will also receive badges to allow access to the appropriate areas.
Lost badges can be replaced at the registration desk. However, a handling fee of EURO 25.- will be charged.

Bank services - Money matters
Banks are generally open weekdays and Saturdays between 8.00/9.00 to 17.00/18.00 hrs and are closed on Sundays.
There are multiple bank machines (ATMs) open 24 hours a day throughout the city (as well as in the conference venue) which accept all major international bankcards.
The official currency of the United Kingdom is the British Pound (GBP). Note that Scottish banks print their own versions.
Major credit cards are widely accepted, but please always check beforehand.

Cancellations and Refunds
Notice of cancellation had to be made in writing by email or fax to the Congress Office.
The policy for refunding registration fees is as follows:
Written cancellation received:
- before April 1, 2015: 75% refund
- between April 1 and May 11, 2015: 25% refund
- after May 11, 2015: no refund

The date of the email/fax ID is the basis for considering refunds. Refunds will be made after the congress.

Car Parking
The multi-storey car park (MSCP) at the SECC, operated by City Parking (Glasgow) LLP, is the main parking area used for events at the venue. This facility is located at 10 Stobcross Road, Glasgow, G3 8YW.
On-site pay machines are located on level 2 (walkway level) and ground floor main foyer of the car park. Payment can be made by cash or credit/debit card. Tariff Rates in the MSCP Monday to Sunday are: 1 hour: £3.50; 2-12 hours: £7.00; 13 hours: £10.50; 14-24 Hours: £14.00. Visitors can either pre-pay after arrival or pay before exiting.

Certificate of Attendance
Certificates of attendance will be issued at the registration desk.

Climate
The average temperatures in June in Glasgow are 17°C (high) and 9°C (low). The average number of rainy days is 20 with an average rainfall of 70 mm in June.

Cloakroom and Luggage
A cloakroom and luggage storage are available close to the registration area and is free of charge.

CME credits
The European Society of Human Genetics is accredited by the European Accreditation Council for Continuing Medical Education (EACCME) to provide CME activities for medical specialists. The EACCME is an institution of the European Union of Medical Specialists (UEMS), www.uems.net
The European Human Genetics Conference has been granted **21 European CME credits (ECMEC)**.
EACCME credits are recognized by the American Medical Association towards the Physician's Recognition Award (PRA). To convert EACCME credit to AMA PRA category 1 credit, contact the AMA.
The EACCME credit system is based on 1 ECMEC per hour with a maximum of 3 ECMECs for half a day and 6 ECMECs for a full-day event.

Coffee Breaks
During the session breaks, refreshments (coffee, tea and water) will be served free of charge to participants wearing name badges. On Saturday, Sunday and Monday coffee and lunch bags will be served in the exhibition area (Hall 4), and on Tuesday in Hall 5 (the exhibition is closed on Tuesday). See also *Lunch and Refreshments.*

INFORMATION GENERAL INFORMATION

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Download the ***ESHG 2015 Conference App*** for iOS and Android from iTunes App Store and Google Play Store.

Currency
The official currency in the United Kingdom is the Pound Sterling (GBP).
1 GBP = 1,35 EUR = 1,38 USD = 1,83 CAD = 181 JPY = 1,41 CHF = 1,92 AUD as per May 4, 2015.

Drinking water
The tap water in Glasgow can be used without concern.

Eating Out in Glasgow
Glasgow has a number of great places to have lunch or dinner. Check the following websites for more infomation
http://www.tripadvisor.co.uk/Restaurants-g186534-Glasgow_Scotland.html
http://www.timeout.com/glasgow/restaurants/the-best-restaurants-in-glasgow
Please note that these websites should serve as indication only. The ESHG is not endorsing any of the stated opinions or listed restaurants.

Electricity Supply
230 V - 50Hz AC, using Type G (BS 1363) three-pin (rectangular) plugs and sockets.

Emergency Services
European Emergency Number: 112, alternatively 999.

Exhibition Opening Hours

Saturday, June 6	09.30 - 18.30 hrs
Sunday, June 7	09.00 - 17.30 hrs
Monday, June 8	09.00 - 17.30 hrs
Tuesday, June 9	CLOSED

GSM Cell Phone Roaming
GSM cell/mobile phone roaming is available without any problems for all major international providers. It is advisable to inquire beforehand or online at your provider which roaming company in the UK offers the cheapest tariffs.

Insurance
By registering to the ESHG 2015 participants agree that neither the organising committee nor the congress office assume any liability whatsoever. Participants are requested to make their own arrangements for health and travel insurance.

Internet and Printing Facilities
WiFi access and terminals with printing facilities are available at the venue. Network ID: *eshg2015*, password: *eshg2015*

Language
The official language of the congress will be English (no simultaneous translation).

Lunch and Refreshments
Lunch tickets for lunch boxes had to be pre-ordered - they cannot be purchased on site. Please note that lunch tickets are not refundable.
Lunch boxes can be picked up from 11.30 - 13.30 hrs at the coffee points in the exhibition (on Tuesday June 9 in Hall 5 from 12.30-13.30 hrs). A cash bar is also available in the exhibition area.

Message Board
Message boards will be available in the registration area.

Pharmacies
Most pharmacies are generally open weekdays from 9.00-18.00 hrs, Saturdays from 9.00-13.00, some until 17.00 hrs and closed on Sundays.

Poster Removal
The organisers cannot assume any liability for loss or damage of posters displayed in the poster area. Posters that will not be removed by Monday, June 8, 2015, 17.30 hrs, will be removed by the staff and will not be kept or mailed to the author after the meeting, but will be discarded.

Speakers' Preview
Equipment for a final check of the sequence of your presentation is available in the Speakers' preview on **the first Floor of the SECC**. All presenters should bring their electronic presentation to the Speakers' preview not later than 2 hours before the start of the session (30 minutes for the first morning sessions).

INFORMATION GENERAL INFORMATION

Registration Desk Opening Hours

Saturday, June 6	08.00 - 20.00 hrs
Sunday, June 7	08.00 - 19.00 hrs
Monday, June 8	08.00 - 19.00 hrs
Tuesday, June 9	08.30 - 14.30 hrs

Safety - Crime

Glasgow can be considered just as safe as other comparable cities in the UK or Europe. Use of common sense is however (always) required. Unfortunately, experience has shown that some basic precautionary measures should always be kept in mind in any city:

- Do not carry important items like flight tickets, passports etc. with you when visiting the conference or strolling through the city, leave them in the hotel safe during your stay. Rather carry a Xerox copy of your passport or an identity card with you.
- Try not to carry all documents, money, credit cards and other essential items and valuables in one bag. If it is lost or stolen, everything will be gone and might be difficult to replace on short notice, especially passports and visa to return to your country of residence.
- Take off your name badge when leaving the conference centre.
- In heavily frequented tourist zones, be aware of attemps of scam and pickpocketing.

Shops

Shops in Glasgow city centre are normally open from 9.00 to 17.30 hrs (Monday to Saturday), some retail stores are now open until 18.00 hrs. Many stores will remain open until 20.00 hrs on Thursdays. More and more shops in the city centre are opening on Sundays, generally from 12.00 to 17.00 hrs. All major credit cards are generally accepted, but it is not possible to pay with foreign banknotes.

Smoking Policy

The ESHG 2015 is officially a “No-smoking-Conference”. Note that smoking is banned in all public places, including restaurants and bars.

Social Media Guidelines

The ESHG supports the use of social media around the European Human Genetics Conference to network with your colleagues and friends attending the meeting. Please do however respect the ESHG social media guidelines, including the following:

Dos:

- Follow ESHG on Twitter (**@eshgsociety**) and use the **#ESHG15** meeting hashtag to follow the latest updates and join in the conversation about the ESHG 2015.
- Follow ESHG on Facebook at **facebook.com/eshg.org**.
- You may blog or tweet about content of the talks, unless the speaker explicitly asks you not to share either the entire talk or specific details or slides.
- Communicate in a respectful and considerate way, and show your criticism in a fair, constructive and professional manner.
- Do remember that people who will read your postings or tweets are not necessarily genetic professionals, but also patients, policymakers, members of the media, and the general public.

Don’ts:

- The use of photography, video, or other type of recording devices in oral sessions (plenary, educational, concurrent sessions and symposia) and poster sessions at the ESHG 2015 is strictly prohibited. Hence posting pictures or videos of these sessions on any social media platform, blogs, or websites, etc., is also strictly prohibited.
- Do not capture or re-distribute data presented at the ESHG 2015 as this may jeopardize the subsequent publication of the data in a scientific journal. Powerpoint presentations must not be photographed under any circumstances. Do respect journal embargo policies and the work of your colleagues!
- Refrain from engaging in personal attacks or showing rude behaviour.

Offensive and disrespectful behaviour, sales-oriented, self-promoting, or otherwise inappropriate comments will not be tolerated.
Individuals should not post copyrighted or trademarked material or material protected by other intellectual property rights. The views and opinions posted on ESHG’s social media do not necessarily reflect the views, opinions, or policies of the ESHG, its Board or membership. The ESHG reserves the right to remove comments it deems to be inappropriate.

Staff

If you should have any questions, the congress staff (recognizable by a yellow badge and a black polo shirt) will be pleased to help you.

Taxis
There are two types of taxis in Glasgow. Taxis and Private Hire Cars. Only Taxis should be hired on a there and then basis (waived at and stopped on the street). There are also numerous taxi ranks in the city.
Private Hire Cars must be pre-booked. Taxi and Private Hire Car both have identification marks (Licence Plate Number and Registration Number).
People do normally tip taxi drivers unless there has been an issue – if it's a low fare than normally rounding it up to the nearest pound is fine, and if you've traveled a bit further a couple of pounds is all you need to tip.

Telephone calls
The country code of the United Kingdom is 44 and the area code for Glasgow is 141, followed by a 7 digit number. To call abroad, dial 00 before the country code.

Time Zone
Glasgow's time zone is Greenwich Mean Time (GMT) or one hour ahead of GMT, known as British Summer Time (BST), during the summer months.

Tipping
There are no hard and fast rules for tipping in Glasgow. If you are happy with the service, a 10-15% tip is customary, particularly in restaurants or cafés. Tipping in bars is not expected. For taxi fares, it's usual to round up to the nearest pound.

Tourist Information Centres (Visit Scotland Information Centre)
Visit Scotland's Information Centre in Glasgow is situated at 170 Buchanan Street, Glasgow, G1 2LW. For more info visit their website: http://www.visitscotland.com/info/services/glasgow-information-centre-p332751.

Travelling - Accessibility - Public Transportation
Directions from Glasgow City Centre to the SECC (Scottish Exhibition and Conference Centre)
By train – journey time 4 minutes
Direct train services are available from Glasgow Central train station to Exhibition Centre train station.
Trains leave Glasgow Central from the lower-level platform 17. Information screens at the platform indicate the incoming train and destinations.
Exiting Exhibition Centre train station, turn right and take the covered walkway, which ends at the SECC (Scottish Exhibition and Conference Centre).
Timetable and fare information is available on the ScotRail website: http://www.scotrail.co.uk

Special delegate offer – once you have collected your delegate badge, show this at the train station to purchase a Conference Rover Ticket. This costs £5 and will allow you to travel by train within the Conference Zone for up to 5 days (covering train travel between the SECC and the city centre).
This ticket also gives you a 50% discount on train fares in Scotland outside the Conference Zone, should you wish to explore further after the conference.

Directions from the SECC to Glasgow City Centre
By train – journey time 4 minutes walk + 4 minutes on train
Exit the SECC and turn left. Take the covered walkway which leads to the Exhibition Centre train station. Trains to the city centre (Glasgow Central) leave from platform 1.
Information screens at the platform indicate the incoming train and destinations.
Timetable and fare information is available on the ScotRail website: http://www.scotrail.co.uk/

By taxi – journey time 5 minutes
Exit the SECC and turn right. A taxi rank is available outside the Clyde Auditorium building.
Should you wish to book a taxi in advance, call +44 (0) 141 429 7070.

V.A.T.
The VAT rate is 20%.

WIFI
Wifi is available throughout the conference venue. Network ID: eshg2015, password: eshg2015

INFORMATION REGISTRATION FEES

Registration fees Payment received:	***before March 31, 2015 (reduced rate)***	***between March 31 & May 11, (normal rate)***	***after May 11, 2015 and on site***	***Day tickets on site***
ESHG Members	EUR 300.-	EUR 400.-	EUR 450.-	EUR 150.-
Non-Members	EUR 450.-	EUR 550.-	EUR 600.-	EUR 200.-
Postgraduate Trainees ESHG Members[1]	EUR 200.-	EUR 300.-	EUR 350.-	EUR 125.-
Postgraduate Trainees Non- Members[1]	EUR 300.-	EUR 400.-	EUR 450.-	EUR 150.-
Counsellors/Gen.Nurses ESHG Members[2]	EUR 200.-	EUR 300.-	EUR 350.-	EUR 125.-
Counsellors/Gen.Nurses Non-Members[2]	EUR 300.-	EUR 400.-	EUR 450.-	EUR 150.-
Students[3]	EUR 100.-	EUR 150.-	EUR 200.-	EUR 100.-
Guests[4]	EUR 85.-	EUR 85.-	EUR 85.-	N/A
	Tickets	Students		
Networking Party	EUR 49.-	EUR 29.-		

[1]Applies to MSc./PhD students. Please provide a confirmation signed by the head of department at the moment of your registration. Confirmations handed in at a later stage cannot be considered.

[2]Applies to non-MD/PhD-Counsellors.

[3]Applies to undergraduate students. Please provide a copy of a Student's ID or a confirmation signed by the head of department at the moment of your registration. Confirmations handed in at a later stage cannot be considered.

[4]Guest registration is only available for family members of registered participants. The fee includes admission to the Networking Mixer (Saturday) and the poster exhibition, no admission to scientific sessions. Guest badges will be coloured differently.

Please see also the General Terms & Conditions for participants: https://www.eshg.org/termsandconditions2015.0.html

What is covered by the registration fee?

Participants:
- Admission to all scientific sessions, exhibition and networking mixer
- Electronic abstract book and printed programme
- Coffee/Tea during breaks from Saturday, June 6 to Tuesday, June 9

Guests (family members only):
- Access to the poster exhibition and the networking mixer (*no admission to scientific sessions!*)

Payment of Registration fees may be made in cash (in Euro or Pound Sterling) or by credit/debit card (in Euro, we accept Diners Club, Mastercard, VISA, American Express and Maestro).

Please note
The reduced registration fee is only applicable, if it has been credited to the congress account before the deadline. Registering before March 31 (or May 11), 2015 without performing the actual payment is not sufficient to benefit from the reduction.

Cancellations and Refunds
Notice of cancellation had to be made in writing by registered letter or fax to the Congress Office.
The policy for refunding registration fees is as follows:
Written cancellation received:
- Before April 1, 2015: 75% refund
- Between April 1 and May 11, 2015: 25% refund
- After May 11, 2015: no refund

The date of the email or fax ID was the basis for considering refunds. Refunds will be made after the congress.

INFORMATION NETWORKING EVENTS

Opening Networking Mixer

Saturday, June 6, 2015, 20.15 - 21.45 hrs - Glasgow Science Centre (50 Pacific Quay, Glasgow G51 1EA)

Network with your colleagues at this mixer following the first group of concurrent sessions on Saturday evening. Drinks and small snacks will be offered.
The networking mixer is free of charge, however admission is only possible for registered participants and registered guests. Two pipers will guide you from the SECC over the Millenium Bridge to the Glasgow Science Centre.

ESHG Networking Party

Monday, June 8, 2015, 19.30 hrs - *Merchant Square*

Join us for a party evening at "*Merchant Square*" in down town Glasgow with dancing, a live band and DJ entertainment. Flat footwear is recommended to join the famous Scottish Ceilidh dance.
Entrance fees include finger food, 5 drinks*, (non alcoholic, beer or wine), music entertainment and a whisky tasting. Cocktails and liquors are avaible at cost.

Ticket: EUR 49.-
Students: EUR 29.-

Please note that only a limited number of tickets can be purchased on a first-come-first-served basis at the onsite registration desk.

Tickets will be checked at the entrance. There will be strictly no access without the entrance ticket!

** The limitation of included drinks complies with the requirements of UK licencing board.*

INFORMATION EXHIBITION

Exhibition Management

Name	**ROSE INTERNATIONAL** Exhibition Management & Congress Consultancy bv
Address	P.O. Box 93260 NL-2509 AG The Hague The Netherlands
Telephone	+31 (0)70 383 89 01
Fax	+31 (0)70 381 89 36
E-mail	eshg@rose-international.com

Exhibition & Poster Area – Hall 4 – Dates & Opening Hours

Saturday, June 6, 2015	09.30 – 18.30 hrs
Sunday, June 7, 2015	09.00 – 17.30 hrs
Monday, June 8, 2015	09.00 – 17.30 hrs
Tuesday, June 9, 2015	CLOSED

Posters – Mounting, Viewing & Removal Schedules

Poster presentations will be held in the exhibition hall from June 6-8. Poster mounting, viewing and removal times are:

Saturday, June 6, 2015	09:30 – 18.30 hrs	Poster mounting / viewing
Sunday, June 7, 2015	09.00 – 17.30 hrs	Poster viewing
Monday, June 8, 2015	09.00 – 17.30 hrs	Poster viewing
Monday, June 9, 2015	13.30 – 17.30 hrs	Poster removal - Strict

Posters not removed by 17.30 hrs on Monday June 8, will be taken down and will not be stored or sent to authors after the meeting but discarded.

Floor Plan – Exhibition & Poster Topics

You will find the floor plan of the Exhibition and Poster Topics in your conference bag in the ESHG Bulletin 2015.

Exhibition Catalogue & Corporate Satellites

All further information on exhibitors and the products and services they offer as well as the Corporate Satellites, can be found in the Exhibition Catalogue & Corporate Satellites book in your conference bag.

Corporate Satellites short programmes are also printed in this Final Programme, section ESHG Scientific Information.

Lead Retrieval System used by Exhibitors (exhibitors and corporate satellites)

A growing number of companies uses a so-called Lead Retrieval System in order to record visitors to their stand and satellite.

Note the following please:
- Companies using the device will ask permission to scan the barcode on your badge.
- This barcode gives the company access to your contact details as follows:
  1. Your name and postal address.
  2. Your e-mail address.

Thank you for your understanding and cooperation.