ESHG

THE EUROPEAN SOCIETY
OF HUMAN GENETICS

EUROPEAN HUMAN GENETICS
CONFERENCE 2014

in conjunction with the

European Meeting on
Psychosocial Aspects of Genetics 2014 (EMPAG)

and the

Italian Society of Human Genetics (SIGU)

May 31 - June 3, Milan, Italy

Final Programme

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GENERAL WELCOMING ADDRESS

Dear Colleagues and Friends,

It was in 1997 in Genoa when the European Society of Human Genetics met in Italy for the last time, so it was time to be back. On behalf of the Italian Society of Human Genetics (SIGU), I cordially welcome you to the European Human Genetics Conference 2014 in Milan.

Milan, known as the industrial capital of Italy, also treasures a notable artistic as well as scientific tradition. Just think of Leonardo da Vinci, the visionary genius, author of the Codex Atlanticus and of the Last Supper, that you will be able to admire in the Biblioteca Ambrosiana and in the Refectory of S. Maria delle Grazie, respectively. In that tradition, Milan is today the place of many universities as well as a host of research institutes, providing the right climate for an international scientific meeting, such as the ESHG Conference.

Likewise, the Italian Society of Human Genetics will contribute to the success of the Conference with a large participation of its members and with all possible measures of support.

Human genetics continues to progress at an unprecedented pace and every annual meeting promises to bring new data to the attention of the scientific community, fostering productive debates and further progress. In this respect, the Milan Conference will not differ from previous ones. Therefore, I expect a numerous and enthusiastic participation. I can promise that your stay will be a memorable one.

Bienvenuto a Milano!

Antonio Amoroso
President, Societa Italiana di Genetica Umana and Local Host of the ESHG 2014

Dear colleagues,

On behalf of the EMPAG Scientific Programme Committee, we are glad to welcome colleagues with an interest in psychological, social and ethical issues to the European Meeting on Psychosocial Aspects of Genetics (EMPAG) 2014 in Milan. Our meetings have an international reputation as a key event for those involved in psychosocial research as well as those providing clinical services.

The 14th EMPAG is again held in conjunction with the European Human Genetics Conference. It offers an excellent and comprehensive programme of interest to a range of healthcare professionals and academics including genetic counsellors, psychologists, social workers, medical sociologists, epidemiologists, genetic nurses, clinical geneticists, scientist and ethicists. There are EMPAG plenary sessions, poster presentations and workshops as well as joint EMPAG/ESHG symposia and educational sessions. We have aimed for a balance between research and practice and to provide time for interaction and discussion.

Participants of the European Conference of Human Genetics are welcome in all EMPAG sessions and EMPAG participants have access to all ESHG sessions.

We are delighted that you have joined us here in Milan for an exciting EMPAG meeting!

Elisabetta Razzaboni & Tara Clancy
Co-chairs of the EMPAG Scientific Programme Committee

GENERAL COMMITTEES - BOARD - ORGANISATION

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Further information on structure and organisation can be found on the website **www.eshg.org**

European Human Genetics Conference 2014

Conference Organisation, Abstract Management

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GENERAL ACKNOWLEDGEMENTS-FUTURE MEETINGS

The European Human Genetics Conference gratefully acknowledges the support of the following companies (list correct as per date of printing):

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- SCIEX Separations
- Wiley

Future European Human Genetics Conferences

European Human Genetics Conference 2015
Glasgow, United Kingdom
June 6 – 9, 2015

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

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 2014 is designated for a maximum of **20 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.

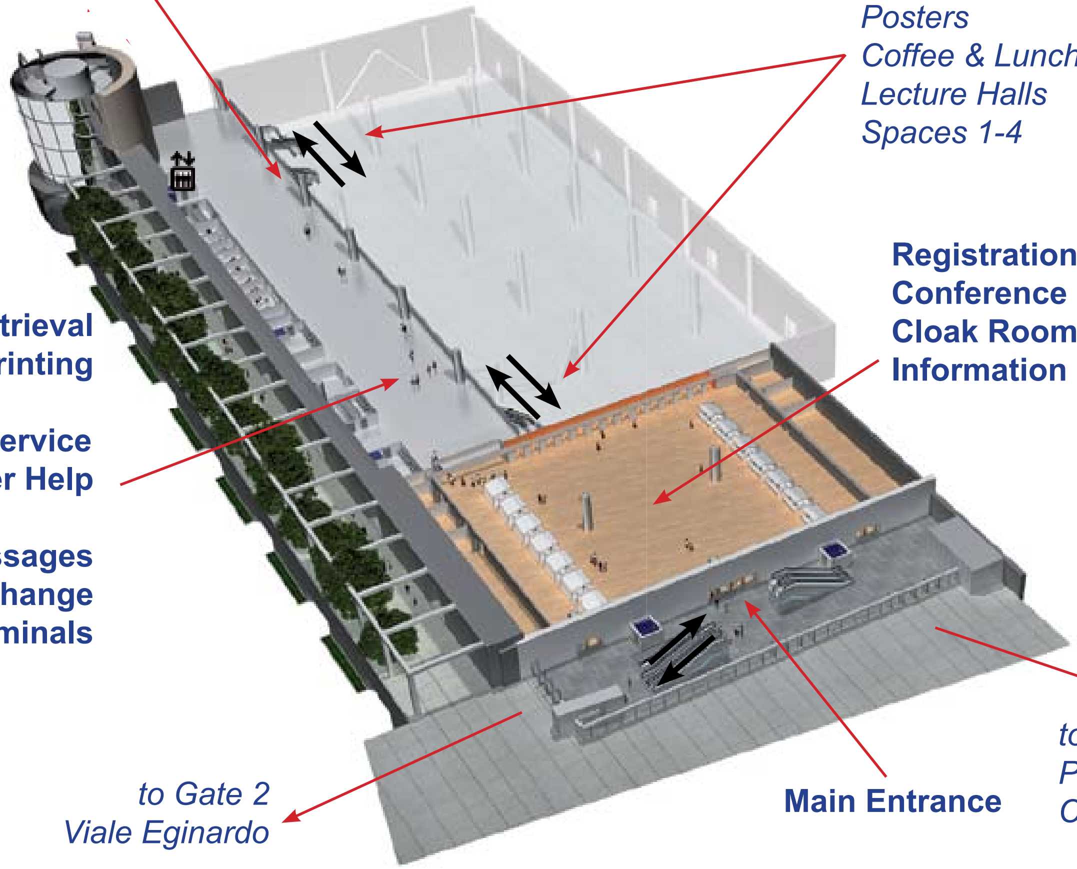
EACCME credits are recognized by the American Medical Association towards the Physician's Recognition Award (PRA). To convert EACCME it to AMA PRA category 1 credit, contact the AMA.

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). Chairpersons are allowed to exclude from the session, persons who will not observe this rule.

GENERAL FLOORPLAN

LEVEL 1



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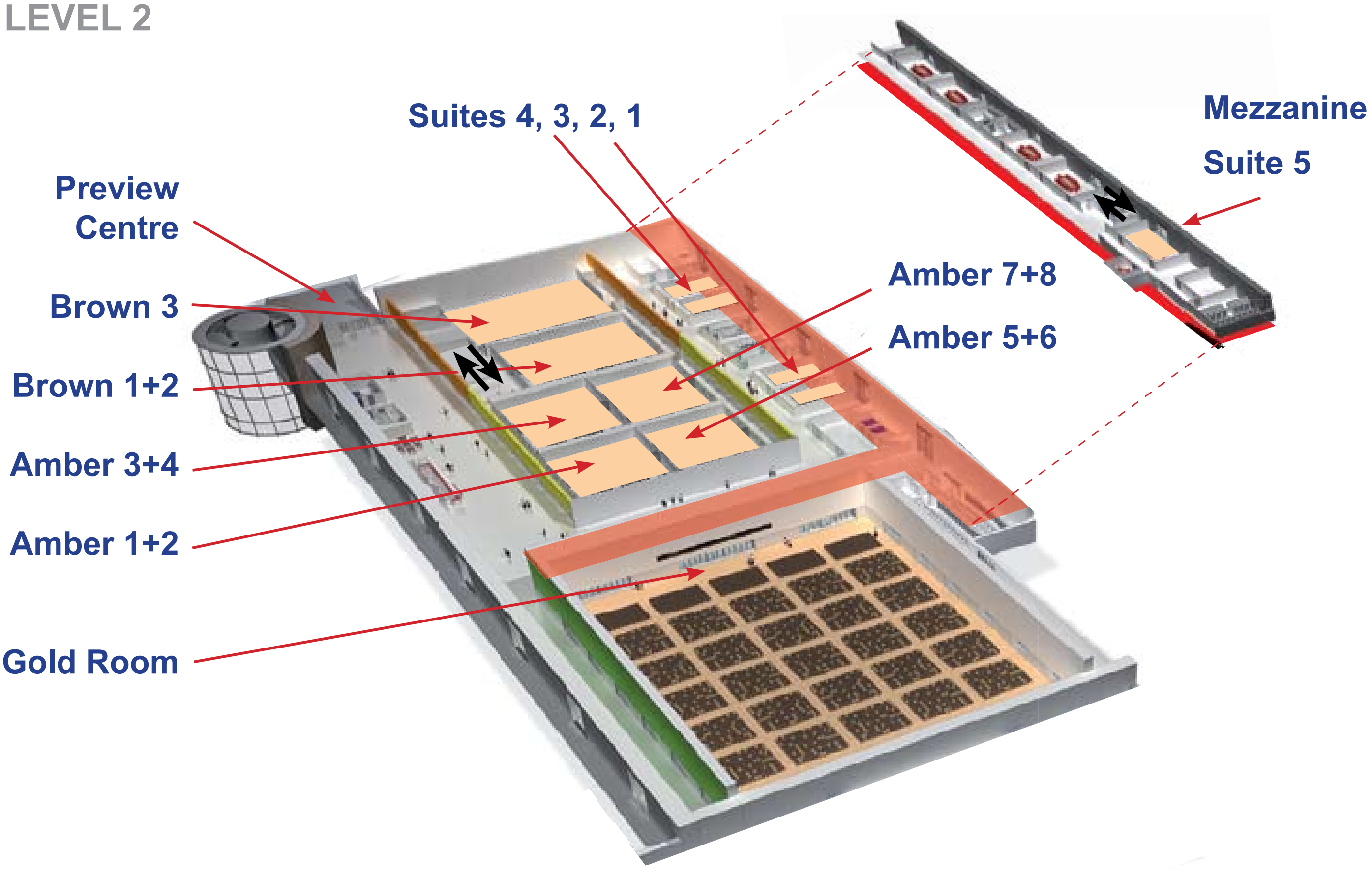

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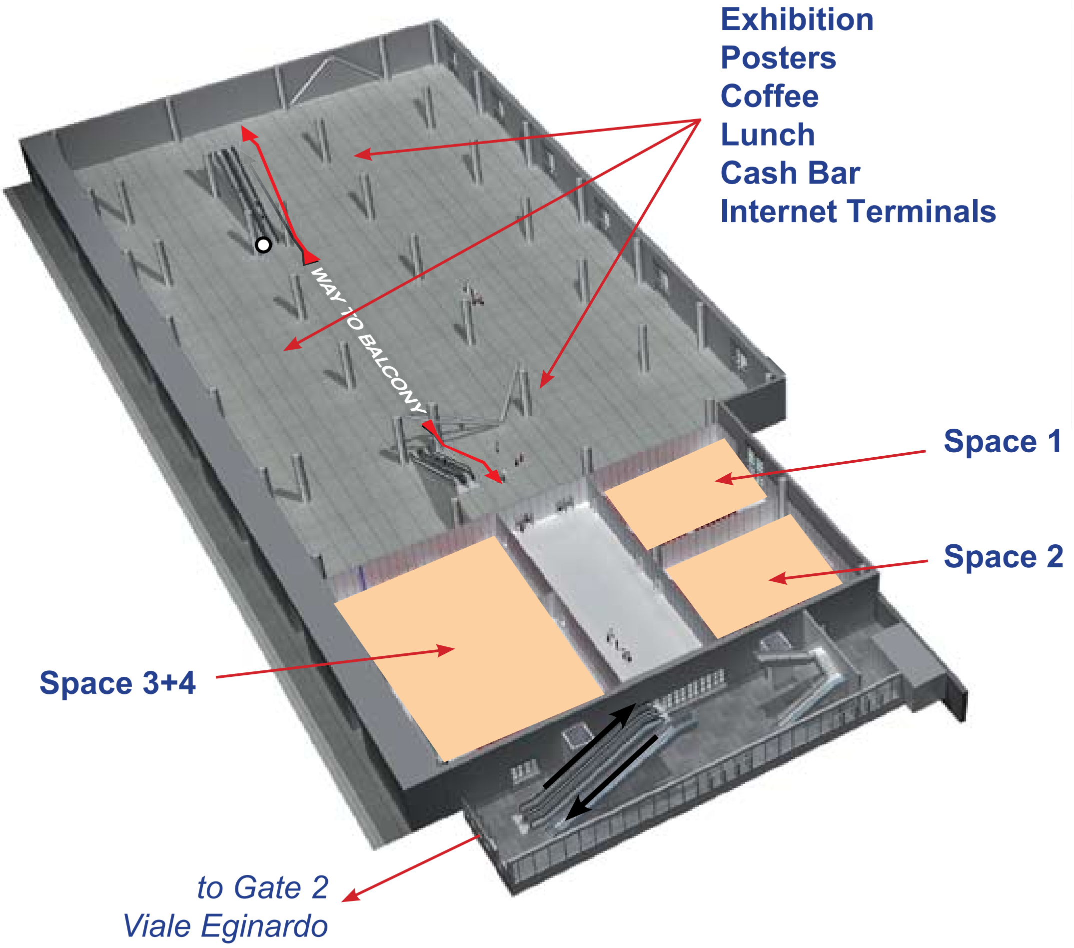

GENERAL PROGRAMME AT A GLANCE-SATURDAY

Saturday, May 31, 2014

Time	Gold Room	Space 3+4	Brown 3	Brown 1+2	Space 1	Space 2	Amber 3+4	Amber 7+8	Suite 5
10.30 – 12.00	**ES1** **The platelets planet: from diagnosis to therapy of inherited thrombocytopenias**	**ES2** **Genetic prediction scores in common diseases: are they of any value?**	**ES3** **What's new in Next Generation Sequencing?**	**ES4** **DNA repair and genomic instability**	**WS01** **Disease of the year: Rasopathies**				
12.15 – 13.45	*Lunch Break / Posters / Exhibition*						**EWS1** **The impact of risk reducing surgery**	**CS01** **Myriad Genetics Satellite**	**CS02** **Personalis Satellite**
14.00 – 14.30	**Opening Welcome Addresses**								
14:30 – 16.00	**PL1** **Opening Plenary Session**						**EPL1** **Psychosocial issues in cancer genetics**		
16:00 – 16.30	*Vitamin Break / Posters / Exhibition*								
16.30 – 18.00	**PL2** **What's New? Highlights Session**						**EPL2** **Reproductive decision making**		
18.00 – 18.30	*Coffee Break / Posters / Exhibition*								
18.30 – 20.00	**C01** **Prenatal testing**	**C02** **Personalized medicine and pharmacogenomics**	**C03** **Intellectual disability**	**C04** **Cardiovascular disorders**	**C05** **Genes and development 1**	**C06** **Functional and computational genomics**	**EPL3** **Genomic testing: psychosocial and ethical issues**		
20.00 – 21.30	*Opening Networking Mixer at the MiCo*								

Session Types:

Plenary Session	Symposium	Concurrent Session	Workshop	Educational Session	EMPAG Sessions	Corporate Satellite

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GENERAL PROGRAMME AT A GLANCE-SUNDAY

Sunday, June 1, 2014

Time	Gold Room	Space 3+4	Brown 3	Brown 1+2	Space 1	Space 2	Amber 3+4	Amber 5+6	Amber 7+8	Suite 5
08.30 – 10.00	**S01 Towards Genomic Personalised Medicine**	**S02 Functional genomics**	**S03 Neuronal Migration disorders**	**S04 Computational Analysis of Gene Networks**	**S05 Early development and preimplantation genetics**	**ES5 Mosaicism in human disease**	**EES 1 Responding to guilt and shame**			
10.00 – 10.30	*Coffee break / Poster viewing / Exhibition*									
10.30 – 11.30	**Poster viewing with presenters** (poster numbers ending with “S”)									
11.45 – 13.15	*Poster viewing / Lunch break / Exhibition*						**EPL4 Family Dynamics** (11.30-12.15)	**CS3 Affymetrix Satellite**	**CS04 Life Technologies Satellite**	**CS05 BIOBASE Satellite**
13.30 – 15.00	**C07 Implementation of NGS in diagnostics**	**C08 Cancer genetics**	**C09 Common neurological disease**	**C10 Bone and skeletal patterning**	**C11 Statistical genetics**	**C12 Sensory disorders**	**EPL5 Access to genetic services and testing**			
15.00 – 15.30	*Vitamin break / Poster viewing / Exhibition*									
15.30 – 17.00	**WS02 Dysmorphology 1**	**WS03 ENSEMBL**	**WS04 Practical Bioinformatics: Whole exome sequence analysis**	**WS05 Quality assurance**	**WS06 Community genetics - Clinical Genetic Services in 2025**	**WS07 Preimplantation genetic diagnosis**	**EPL6 Facilitating communication about genetic information**	**CS06 PerkinElmer Satellite**	**CS07 Cartagenia Satellite**	**CS08 Multiplicom Satellite**
17.00 – 17.30	*Coffee break / Poster viewing / Exhibition*									
17.30 – 19.00	**S06 Risk perception and risk communication,** joint with EMPAG	**S07 Therapy for human genetic diseases**	**S08 Population genetics in a globalized world**	**S09 Advances and new challenges in genetics of cardiovascular diseases,** joint with the Eur.Soc. of Cardiology	**S10 New Mutational Mechanisms**	**ES6 How to be successful in rare disease gene Identification**				
19.00 – 20.30							**ESHG Membership Meeting**	**CS09 AstraZeneca Satellite**	**CS10 Illumina Satellite**	**CS11 Natera Satellite**

Session Types

Plenary Session	Symposium	Concurrent Session	Workshop	Educational Session	EMPAG Sessions	Corporate Satellite

GENERAL PROGRAMME AT A GLANCE-MONDAY

Monday, June 2, 2014

Time	Gold Room	Space 3+4	Brown 3	Brown 1+2	Space 1	Space 2	Amber 3+4	Amber 5+6	Amber 7+8	Suite 5
08.30 – 10.00	**S11** **Rare copy number variants in common traits**	**S12** **Epigenetic basis of disease**	**S13** **Non-invasive prenatal testing,** joint with EMPAG	**S14** **Rapid genome diagnostics**	**ES7** **From mutation identification to therapy**					
10.00 – 10.30	*Coffee break / Poster viewing / Exhibition*									
10.30 – 11.30	**Poster viewing with presenters** (poster numbers ending with “M”)									
11.45 – 13.15	*Poster viewing / Lunch break / Exhibition*						**EPL7** **Autonomy and consent** (11.30-12.15)	**CS12** **Agilent Technologies Satellite**	**CS13** **Abbott Molecular Satellite**	**CS14** **LGC Satellite**
13.30 – 15.00	**C13** **Innovation in genetic services**	**C14** **Genetics of complex traits**	**C15** **Novel genes in neurogenetic disorders**	**C16** **Genes and development 2**	**C17** **Metabolic and mitochondrial disorders**		**EPL8** **Psychosocial issues in prenatal & preimplantation diagnosis**			
15.00 – 15.30	*Vitamin break / **Poster removal** / Exhibition*									
15.30 – 17.00	**WS08** **Dysmorphology 2**	**WS09** **Genome Browser UCSC**	**WS10** **Analysis, interpretation and reporting of array data**	**WS11** **Clinical Cancer Genetics Club**	**WS12** **Preconception and prenatal screening**	**WS13** **Next Generation Sequencing**	**EPL9** **Lessons learned and new issues in predictive testing**		**CS15** **Life Technologies Satellite**	**CS16** **QIAGEN Satellite**
17.00 – 17.30	*Coffee break / **Poster removal** / Exhibition*									
17.30 – 19.00	**S15** **Networks and pathways in genetic diseases**	**S16** **The new RNA world**	**S17** **Tumour heterogeneity**	**S18** **A novel class of disease of lipid metabolism**	**S19** **Three sequencing approaches in complex disease**	**ES8** **Current developments in legal aspects of genetics: Untangling the law and what it means for you**	**EES 2** **Qualitative and quantitative methods in psychosocial research**			
19.00 – 20.30								**CS17** **BGI Satellite**	**CS18** **Fluidigm Satellite**	**CS19** **Roche Sequencing Satellite**
20.30	*Networking Party at the Old Fashion Club*									

Session Types:

Plenary Session	Symposium	Concurrent Session	Workshop	Educational Session	EMPAG Sessions	Corporate Satellite

GENERAL PROGRAMME AT A GLANCE-TUESDAY

Tuesday, June 3, 2014

Time	Gold Room	Space 3+4	Brown 3	Brown 1+2	Space 1
09.00 – 10.30	**PL3** **ESHG-ASHG "Bulding Bridges Session":** **Debate: What IF... (Incidental Findings), an interactive Debate,** joint with EMPAG				
10.30 – 11.00	*Coffee break (Level 1 & 2)*				
11.00 – 12.30	**C18** **Large scale genomics**	**C19** **Internal organs**	**C20** **Basic mechanisms in genetics**	**C21** **Rasopathies and CDG**	**C22** **Returning results: Ethical and legal issues,** joint with EMPAG
12.30 – 13.30	*Lunch Break (Level 1 & 2)*				
13.30 – 14.15	**PL4** **Mendel Lecture**				
14.15 – 15.45	**PL5** **Closing Plenary** **ESHG Award Lecture** **- ESHG Honorary Award** **- EJHG-NPG Awards** **- Young Investigator & Poster Awards** **- Closing**				

Session Types:

Plenary Session	Symposium	Concurrent Session	Workshop	Educational Session	EMPAG Sessions	Corporate Satellite

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GENERAL BUSINESS AND ANCILLARY MEETINGS

As per date of printing.

Saturday, May 31, 2014

08.30 - 10.30	UEMS Board Meeting	Amber 1+2	closed
11.00 - 14.00	ESHG Genetic Services Quality Committee Meeting	Suite 2	closed
12.15 - 13.45	UEMS Section Meeting	Amber 1+2	closed
13.00 - 15.45	ESHG PPPC Meeting	Suite 1	closed
13.00 - 15.00	eRare EuroMicro	Suite 3	closed

Sunday, June 1, 2014

10.00 - 11.00	European Genetic Nurses and Counsellors Meeting	Amber 1+2	closed
10.00 - 14.00	Int.Soc. of Community Genetics Founding Members meeting	Suite 1	closed
10.00 - 17.00	Patient Representatives Meeting	Suite 3	closed
11.30 - 13.30	National Human Genetics Societies Meeting	Amber 1+2	closed
11.30 - 13.30	European Journal of Medical Genetics, Ed. Board Meeting	Suite 2	closed
12.15 - 13.30	EMPAG SPC Meeting	Amber 3+4	closed
15.00 - 17.30	Network use of NGS in autoinflammatory diagnostics: an explorative meeting	Amber 1+2	closed
15.15 - 16.15	EJHG Editorial Board Meeting	Suite 1	closed
16.30 - 17.30	Informed Consent Meeting	Suite 2	closed
19.15 - 20.15	ESHG Membership Meeting	Amber 3+4	members

Monday, June 2, 2014

10.00 - 12.00	European Board of Medical Genetics Meeting	Suite 2	closed
10.15 - 12.15	ESHG Education Committee Meeting	Suite 1	closed
10.30 - 12.15	Committee meeting of the COST Action BM1208 - Network of Congenital Imprinting Disorders - EUCID.net	Space 2	closed
10.30 - 11.30	CEQAS Participants Meeting	Amber 1+2	closed
12.15 - 13.15	ESHG Board Meeting II	Amber 1+2	closed
15.30 - 17.00	Int. Federation of Human Genetics Societies - IFHGS Board Meeting	Amber 1+2	closed
15.30 - 17.00	Journal of Community Genetics Meeting	Suite 1	closed

Tuesday, June 3, 2014

12.15 - 13.15	ESHG SPC Meeting	Suite 1	closed
14.15 - 19.45	Hirschsprung Consortium Meeting	Suite 1	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.

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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 – S19)

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 – ES8)

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 – C22)

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 a * are candidates for one of 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 – WS13)

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.

EMPAG Sessions (EPL1 – EPL9, EES1 – EES2)

Every other year, the ESHG holds its annual meeting in conjunction with the European Meeting on Psychosocial Aspects of Genetics, that has a special programme focus on Genetic Counsellors and Nurses in Plenaries, Workshops and Educational Sessions, as well as joint ESHG-EMPAG Sessions. ESHG attendees are welcome to attend the EMPAG sessions and vice-versa.

Corporate Satellites (CS01-CS19)

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.

SCIENTIFIC

SCIENTIFIC PROGRAMME

Saturday, May 31, 2014

PROGRAMME

PROGRAMME SATURDAY, MAY 31

Time	Gold Room	Space 3+4	Brown 3	Brown 1+2	Space 1
10.30 - 12.00	**ES1 The platelets planet: from diagnosis to therapy of inherited thrombocytopenias** Chair: M. Seri	**ES2 Genetic prediction scores in common diseases: are they of any value?** Chair: S. Ripatti	**ES3 What‘s new in Next Generation Sequencing?** Chair: J. Veltman	**ES4 DNA repair and genomic instability** Chair: M. Genuardi	**WS01.** **Disease of the year: Rasopathies** Organisers: G. Neri, M. Tartaglia
10.30	**ES1.1** **Genetics of familial forms of thrombocytopenia** ***Anna Savoia***; *Trieste, Italy*	**ES2.1** **Using prediction scores in cardiovascular medicine** ***Samuli Ripatti***; *Helsinki, Finland*	**ES3.1** **Novel sequencing approaches in genetic disease research** ***Alexander Hoischen***; *Nijmegen, Netherlands*	**ES4.1** **Protein replacement system: the case of polymerase-delta and MLH1 mutations in colon cancer.** ***Josef Jiricny***; *Zurich, Switzerland*	RASopathies are a family of syndromes including Noonan, CFC an Costello syndrome, plus related disorders neurofibromatosis 1 and Legius syndrome. Their clinical similarities are due to the fact the causal mutant genes all encode proteins belonging to the same RAS-ERK signaling pathway. New insights into the molecular pathogenesis of these disorders may be conducive to new treatments.
11.15	**ES1.2** **Diagnosis and management of inherited thrombocytopenias** ***Carlo L. Balduini***, *P. Noris, A. Pecci; Pavia, Italy*	**ES2.2** **The benefits of using genetic information to design prevention trials** ***Aroon Hingorani***; *London, United Kingdom*	**ES3.2** **Single cell genome and transcriptome sequencing** ***Joakim Lundeberg***; *Stockholm, Sweden*	**ES4.2** **Aging and cancer: The impact of DNA damage** ***Jan H.J. Hoeijmakers***; *Rotterdam, Netherlands*	
12.00 - 14.00	*Lunch break / Posters / Exhibition / Satellites*				

Detailed Workshop programmes (as submitted by the organisers) can be found in the “ESHG Bulletin” in the conference bag.

PROGRAMME SATURDAY, MAY 31

Time	Gold Room
14.00 - 14.30	**Opening & Welcoming Addresses** Chair: A. Amoroso, H. Brunner
	Welcoming Addresses by Han Brunner *President of the ESHG* Antonio Amoroso *President of the Italian Society of Human Genetics (SIGU), Local host* Tara Clancy *Co-Chair of the European Meeting on Psychosocial Aspects of Genetics*
14.30 - 16.00	**Opening Plenary Session PL1** Chair: A. Amoroso, H. Brunner
14.30	**PL1.1** **RASopathies. The other face of RAS signalling dysregulation** ***Marco Tartaglia***; *Rome, Italy*
15.00	**PL1.2** **Evolution of the HD gene** ***Elena Cattaneo***; *Milan, Italy*
15.30	**PL1.3** **Genetic engineering of hematopoietic stem cells for the treatment of inherited diseases** ***Alessandra Biffi***; *(Change of presenter!)* *Milan, Italy*
16.00 - 16.30	*Vitamin break / Posters / Exhibition*
16.30 - 18.00	**Plenary Highlights Session PL2. What's new?** Chair: H. Brunner, B. Wirth
16.30	**PL2.1** **Early-Onset Stroke and Vasculopathy Associated with Mutations in ADA2** ***Ivona Aksentijevich***, *Q. Zhou, A.K. Ombrello, D. Yang, A.V. Zavialov, R. Sood, M. Boehm, D.L. Kastner;* *Bethesda, United States*
16.45	**PL2.2** **Disrupted auto-regulation of SNRPB causes cerebro-costo-mandibular syndrome** ***Danielle C. Lynch******, *T. Revil, J. Schwartzentruber, E.J. Bhoj, A.M. Innes, R.E. Lamont, E.G. Lemire, B.N. Chodirker, J.P. Taylor, E.H. Zackai, D.R. McLeod, E.P. Kirk, J. Hoover-Fong, L. Fleming, R. Savarirayan, .. Care4Rare Canada, J. Majewski, A. Jerome-Majewska, J.S. Parboosingh, F.P. Bernier;* *Calgary, Canada*
17.00	**PL2.3** **The First 100 patients diagnosed by whole-exome sequencing through FORGE Canada: Insights for Clinical Translation** ***Sarah L. Sawyer***, *C.L. Beaulieu, T. Hartley, D. Bulman, J. Majewski, FORGE Canada Consortium, K.M. Boycott;* *Ottawa, Canada*
17.15	**PL2.4** **Transcriptomes of individual cells** ***Christelle Borel***, *P.G. Ferreira, M. Garieri, F.A. Santoni, O. Delaneau, E. Falconnet, P. Ribaux, P. Makrythanasis, M. Guipponi, E.T. Dermitzakis, S.E. Antonarakis;* *Geneva, Switzerland*
17.30	**PL2.5** **Chromosome X-wide association analysis discovers new loci for complex traits including a height locus not dosage compensated between men and women** ***Taru Tukiainen******, *M. Pirinen, A. Sarin, C. Ladenvall, J. Kettunen, T. Lehtimäki, M. Lokki, M. Perola, J. Sinisalo, E. Vlachopoulou, J.G. Eriksson, L. Groop, A. Jula, M. Järvelin, O.T. Raitakari, V. Salomaa, S. Ripatti;* *Boston, United States*
17.45	**PL2.6** **Genome sequencing identifies major causes of severe intellectual disability** ***Christian Gilissen******, *J.Y. Hehir-Kwa, D.T. Thung, M. Van de Vorst, B.W.M. van Bon, M.H. Willemsen, M. Kwint, I. Janssen, A. Hoischen, R. Leach, R. Klein, R. Tearle, T. Bo, R. Pfundt, H.G. Yntema, B.B.A. De Vries, T. Kleefstra, H.G. Brunner, L.E.L.M. Vissers, J.A. Veltman;* *Nijmegen, Netherlands*
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 affilitation of the presenting author.

PROGRAMME SATURDAY, MAY 31

Time	Gold Room	Space 3+4	Brown 3	Brown 1+2	Space 1	Space 2
18.30 - 20.00	**C01 Prenatal testing** Chair: F. Forzano, P. Borry	**C02 Personalized medicine and pharmacogenomics** Chair: M. Gennarelli, M. Nöthen	**C03 Intellectual disability** Chair: F. Gurrieri, T. Prescott	**C04 Cardiovascular disorders** Chair: A. Renieri, J. McGaughran	**C05 Genes and development 1** Chair: C. Romano, M. Breuning	**C06 Functional and computational genomics** Chair: V. Nigro, G. Houge
18.30	**C01.1** **Clinical implementation of non-invasive prenatal aneuploidy detection** ***Nathalie Brison****, *B. Bayindir, P. Brady, L. Dehaspe, S. Ardui, J. Van Houdt, H. Van Esch, E. Legius, T. De Ravel, K. Devriendt, J.R. Vermeesch;* *Leuven, Belgium*	**C02.1** **A novel variant in the SLC9A9 gene influences disease activity in interferon-beta treated multiple sclerosis patients** ***Melissa Sorosina****, *F. Esposito, C. Guaschino, G. Liberatore, A. Osiceanu, V. Martinelli, D. Brassat, G. Comi, P.L. De Jager, F. Martinelli Boneschi;* *Milan, Italy*	**C03.1 Dominant β-catenin mutations cause a recognizable syndrome with intellectual disability, and are associated with learning deficits and structural and functional brain abnormalities in mice** ***Marjolein H. Willemsen****, *V. Tucci, T. Kleefstra, A. Hardy, I. Heise, S. Maggi, W. Wissink-Lindhout, A. Vulto-van Silfhout, B. de Vries, Z. Iqbal, H. Brunner, W. Nillesen, H. Yntema, H. Hilton, M. Simon, S. Tsaftaris, H. van Bokhoven, A. Constestabile, T. Nieus, A. Raimondi, B. Greco, D. Cantatore, L. Gasparini, L. Berdondini, A. Bifone, J. Veltman, L. Peart-Vissers, A. Gozzi, S. Wells, P. Nolan; Nijmegen, Netherlands*	**C04.1** ***EIF2AK4* mutations cause pulmonary veno-occlusive disease, a recessive form of pulmonary hypertension** ***Melanie Eyries***, *D. Montani, B. Girerd, C. Perret, A. Leroy, C. Lonjou, N. Chelghoum, F. Coulet, D. Bonnet, P. Dorfmüller, E. Fadel, O. Sitbon, G. Simonneau, D. Tregouët, M. Humbert, F. Soubrier;* *Paris, France*	**C05.1** **Compound inheritance of a low-frequency promoter deletion and a null mutation in a new gene causes Burn-McKeown syndrome (BMKS)** ***Dagmar Wieczorek***, *W.G. Newman, T. Wieland, T. Berulava, M. Kaffe, D. Falkenstein, C. Beetz, S. Douzgou, J. Clayton-Smith, S.B. Daly, S.G. Williams, S. Bhaskar, J. Urquhart, B. Anderson, J. O'Sullivan, O. Boute, E. Graf, J.C. Czeschik, A.J. van Essen, F. Hazan, A. Hing, A. Kuechler, J. Lemke, C. Marques Lourenco, U. Hehr, B. Horsthemke, T. Meitinger, J. Burn, H. Lüdecke, T.M. Strom;* *Essen, Germany*	**C06.1** **Resolving variants of unknown significance through reanalysis of 4,978 public RNA-seq samples** ***Patrick Deelen****, *D.V. Zhernakova, M. van der Sijde, J. Karjalainen, J.K. van der Velde, M. de Haan, K.M. Abbott, C. Wijmenga, R.J. Sinke, M.A. Swertz, J. Fu, L. Franke;* *Groningen, Netherlands*
18.45	**C01.2** **Clinical Validation of Noninvasive Prenatal risk assessment for fetal sex chromosome aneuploidies in maternal plasma using Direct ANalysis of Selected Regions (DANSR™) assays** *K.H. Nicolaides, T. Musci, C. Struble, E. Wang, J. Hooks, A. Syngelaki, M. del Mar Gil, A. Oliphant,* ***Adam Wolfberg****;* *San Jose, United States*	**C02.2** **High throughput sequencing in sporadic forms of steroid-resistant nephrotic syndrome: heterogeneous genetic alterations can predict resistance to treatments** ***Aldesia Provenzano****, *B. Mazzinghi, F. Becherucci, L. Giunti, G. Sansavini, F. Ravaglia, R. Roperto, S. Farsetti, E. Benetti, M. Rotondi, L. Murer, L. Lasagni, M. Materassi, P. Romagnani, S. Giglio;* *Firenze, Italy*	**C03.2** ***De Novo* loss of function mutations in *SETD5*, a novel methyltransferase gene within the 3p25 microdeletion syndrome critical region, cause intellectual disability** ***Keren J. Carss****, *D. Grozeva, O. Spasic-Boskovic, M.J. Parker, H. Archer, H.V. Firth, S. Park, N. Canham, S.E. Holder, M. Wilson, A. Hackett, M. Field, J.A.B. Floyd, UK10K Consortium, M.E. Hurles, F.L. Raymond;* *Hinxton, Cambridgeshire, United Kingdom*	**C04.2 Rare variants in *NR2F2* cause congenital heart defects in humans** *S. Al Turki, A.K. Manickaraj,* ***Catherine L. Mercer****, *S. Gerety, M.P. Hitz, S. Lindsay, L.C.A. D'Alessandro, G.J. Swaminathan, J. Bentham, A.K. Arndt, J. Breckpot, J. Low, B. Thienpont, H. Abdul-Khaliq, C. Harnack, K. Hoffmann, H.H. Kramer, S. Schubert, R. Siebert, O. Toka, C. Cosgrove, H. Watkins, A.M. Lucassen, I.M. O'Kelly, A.P. Salmon, F.A. Bu'Lock, J. Granados-Riveron, K. Setchfield, C. Thornborough, J.D. Brook, B. Mulder, S. Klaassen, S. Bhattacharya, K. Devriendt, D.F. FitzPatrick, S. Mital, M.E. Hurles, D.I. Wilson;* *Southampton, United Kingdom*	**C05.2** **Genetic studies of mosaic birth defects affecting the skin by next-generation DNA sequencing** ***Jean-Baptiste Rivière***, *J. St-Onge, Y. Duffourd, J. Courcet, Fédération des Centres Labellisés « Anomalies du Développement » (FeCLAD), Filière des Maladies Rares en Dermatologie (FIMARAD), Société française de Foetopathologie (SOFFOET), L. Faivre, B. Demeer, P. Vabres;* *Dijon, France*	**C06.2** **The long non-coding RNA landscape of autoimmune diseases** ***Cisca Wijmenga***, *I. Ricaño-Ponce, Y. Li, B. Hrdlickova, D. Zhernakova, J. Karjalainen, P. Deelen, S. Withoff, L. Franke, V. Kumar;* *Groningen, Netherlands*
19.00	**C01.3** **mtDNA mutations variously impact mtDNA maintenance throughout the human embryofetal development** ***Sophie Monnot***, *S. Rondeau, P. Vachin, E. Herzog, B. Bessieres, N. Gigarel, D. Samuels, L. Hesters, N. Frydman, G. Chalouhi, M. Rio, A. Rotig, A. Benachi, L. Salomon, A. Munnich, J. Bonnefont, J. Steffann;* *Paris, France*	**C02.3 Personalized thiopurine dosing based on TPMT genotyping reduces leucopenia occurrence and results in cost-savings in IBD patients; results from a randomized trial in the Netherlands** ***Marieke J.H. Coenen***, *D.J. de Jong, C.J. van Marrewijk, L.J.J. Derijks, S.H. Vermeulen, D.R. Wong, O.H. Klungel, A.L.M. Verbeek, P. Hooymans, W.H.M. Peters, R.H.M. te Morsche, TOPIC recruitment team, W.G. Newman, H. Scheffer, H. Guchelaar, B. Franke;* *Nijmegen, Netherlands*	**C03.3** **Genetic heterogeneity in Hyperphosphatasia with Mental Retardation Syndrome due to mutations in PGAP3, a member of the GPI anchor synthesis pathway** ***Denise Horn***, *Y. Murakami, C. Daumer-Haas, B. Fischer, J. Hecht, U. Kölsch, S. Leiz, Y. Maeda, D. Mitchell, J. Phillips, S. Mundlos, P.N. Robinson, P.M. Krawitz;* *Berlin, Germany*	**C04.3** **Loss of alpha1 beta1 soluble guanylate cyclase, the major nitric oxide receptor, leads to moyamoya and achalasia** *D. Hervé, A. Philippi, R. Belbouab, M. Zerah, S. Chabrier, S. Collardeau-Frachon, F. Bergametti, A. Essongue, E. Berrou, V. Krivosic, C. Sainte-Rose, E. Houdart, F. Adam, K. Billiemaz, M. Lebret, S. Roman, S. Passemard, G. Boulday, A. Delaforge, S. Guey, X. Dray, P. Brouckaert, M. Bryckaert,* ***Elisabeth Tournier-Lasserve****;* *Paris, France*	**C05.3** **A point mutation in STIM1 (p.R304W) is associated with Stormorken syndrome** ***Gilles Morin***, *N. Ortiz Bruechle, A. Rabbind Singh, C. Knopp, G. Jedraszak, M. Elbracht, D. Brémond-Gignac, K. Hartmann, H. Sevestre, P. Deutz, D. Hérent, P. Nürnberg, B. Roméo, K. Konrad, M. Mathieu-Dramard, J. Oldenburg, E. Bourges-Petit, Y. Shen, K. Zerres, H. Ouadid-Ahidouch, J. Rochette;* *Amiens, France*	**C06.3** **Population Scale Comprehensive Identification and Analysis of Complex Structural Variation Using Nanochannel Array** *H. Dai, A. Hastie, E. Lam, W. Andrews, T. Anantharaman, A. Pang, M. Saghibini, H. Sadoski, H. VanSteenhouse, M. Austin, X. Yang, T. Dickinson, Z. Dzakula, M. Xiao, P. Kwok,* ***Han Cao****;* *San Diego, United States*

PROGRAMME SATURDAY, MAY 31

Time	Gold Room	Space 3+4	Brown 3	Brown 1+2	Space 1	Space 2
cont.	**C01 Prenatal testing** Chair: F. Forzano, P. Borry	**C02 Personalized medicine and pharmacogenomics** Chair: M. Gennarelli, M. Nöthen	**C03 Intellectual disability** Chair: F. Gurrieri, T. Prescott	**C04 Cardiovascular disorders** Chair: A. Renieri, J. McGaughran	**C05 Genes and development 1** Chair: C. Romano, M. Breuning	**C06 Functional and computational genomics** Chair: V. Nigro, G. Houge
19.15	**C01.4** **Whole-genome single-cell haplotyping, a generic method for preimplantation genetic diagnosis** **_Masoud Zamani Esteki*_**, _E. Dimitriadou, L. Mateiu, C. Melotte, N. Van der Aa, P. Kumar, R. Das, J. Cheng, E. Legius, Y. Moreau, S. Debrock, T. D'Hooghe, P. Verdyck, M. De Rycke, K. Sermon, J. Vermeesch, T. Voet;_ _Leuven, Belgium_	**C02.4** **Genome-wide identification and phenotypic validation of loss of function mutations** **_Leslie G. Biesecker_**, _K. Lewis, D. Ng, J. Johnston, J. Mullikin;_ _Bethesda, United States_	**C03.4** **The significance of small copy number variants in neurodevelopmental disorders** **_Reza Asadollahi*_**, _B. Oneda, P. Joset, S. Azzarello-Burri, D. Bartholdi, K. Steindl, M. Vincent, J. Cobilanschi, H. Sticht, R. Baldinger, R. Reissmann, I. Sudholt, C.T. Thiel, A.B. Ekici, A. Reis, E.K. Bijlsma, J. Andrieux, A. Dieux, D. FitzPatrick, S. Ritter, A. Baumer, B. Latal, B. Plecko, O. Jenni, A. Rauch;_ _Zurich, Switzerland_	**C04.4** **From Identification of Differing TIE2 Mutations with Distinct Cellular Characteristics in Four Types of Venous Anomalies towards a Murine Model and a Therapeutic Pilot Study** _N. Limaye, J. Soblet, M. Uebelhoer, M. Natynki, E. Boscolo, L. Eklund, J. Bischoff, L.M. Boon,_ **_Miikka S. Vikkula_**_;_ _Brussels, Belgium_	**C05.4** **TashT is a novel mouse model that phenocopies both the variable penetrance and male sex-bias of Hirschsprung's disease** **_Nicolas Pilon_**, _K.F. Bergeron, T. Cardinal, A.M. Touré, D.W. Silversides;_ _Montréal, Canada_	**C06.4** **Chromatin loops and CNVs: the complex spatial organization of the 16p11.2 locus** **_Maria Nicla Loviglio*_**, _M. Leleu, N. Gheldof, E. Migliavacca, K. Mannik, J. Beckmann, S. Jacquemont, J. Rougemont, A. Reymond;_ _Lausanne, Switzerland_
19.30	**C01.5** **Scenarios for implementation of noninvasive prenatal testing (NIPT) for Down syndrome in a national health care system** **_Elke Mersy*_**, _C.E.M. de Die-Smulders, A.B.C. Coumans, L.J.M. Smits, G.M.W.R. de Wert, S.G.M. Frints, J.A. Veltman;_ _Maastricht, Netherlands_	**C02.5** **The SickKids Genome Clinic: Developing and evaluating a pediatric model for individualized genomic medicine** **_M S. Meyn_**, _S. Bowdin, N. Monfared, D. Merico, D.J. Stavropoulos, M. Girdea, R. Hayeems, M. Szego, G. Bader, R.D. Cohn, J.A. Anderson, R. Zlotnik-Shaul, M. Brudno, C. Shuman, C.R. Marshall, P.N. Ray;_ _Toronto, Canada_	**C03.5** **Rare large CNVs are associated with intellectual disability, education level, and female fertility in general population** **_Alexandre Reymond_**, _K. Männik, R. Mägi, A. Mace, A. Maillard, H. Alavere, A. Kolk, L. Leitsalu, A. Ferreira, M. Noukas, J.S. Beckmann, S. Jacquemont, Z. Kutalik, A. Metspalu;_ _Lausanne, Switzerland_	**C04.5** **A high yield of variants with a putative role as modifiers in patients with hypertrophic cardiomyopathy** **_Sara Bardi_**, _F. Girolami, M. Benelli, B. Tomberli, E. Contini, G. Marseglia, C. Pescucci, G. Castelli, A. Fornaro, F. Cecchi, I. Olivotto, F. Torricelli;_ _Florence, Italy_	**C05.5** **WNT pathway downregulation and Cornelia de Lange Syndrome** _A. Pistocchi, G. Fazio, L.R. Bettini, A. Cereda, L. Ferrari, F. Cotelli, A. Biondi, A. Selicorni, V. Massa,_ **_Anna Marozzi_**_;_ _Milan, Italy_	**C06.5** **Informing rare disease mechanisms: informatics for the International Mouse Phenotyping Consortium** **_Terrence F. Meehan_**, _on behalf of the MPI2 Consortium;_ _Hinxton, Cambridge, United Kingdom_
19.45	**C01.6** **Whole genome sequencing and analysis in prenatal screening: ethical reflection** **_Guido de Wert_**, _W. Dondorp;_ _Maastricht, Netherlands_	**C02.6** **Collaboration to integrate genomics into clinical care: a demonstration evaluation** **_Clara Gaff_**, _N. Thorne, I. Macciocca, P. Waring, S. Forrest, P. Ekert, I. Winship, T. Lockett, M. South, A. Sinclair, Melbourne Genomics Health Alliance;_ _Melbourne, Australia_	**C03.6** **Altered neuronal network in iPSC derived cortical neurons from patients with _MECP2_ duplication syndrome** _S. Nageshappa, C. Carromeu, I. Espuny-Camacho, C. Bagni, C. Verfaillie, C. Carvalho, M. Ramocki, J. Lupski, P. Vanderhaeghen, A. Muotri,_ **_Hilde Van Esch_**_;_ _LEUVEN, Belgium_	**C04.6** **Causal relationship of body mass index with cardiometabolic traits and events: a Mendelian randomization analysis** **_Michael V. Holmes*_**, _L.A. Lange, T. Palmer, M.B. Lanktree, IBC BMI Mendelian Randomization Group, E.E. Schadt, F.W. Asselbergs, A.P. Reiner, B.J. Keating;_ _Philadelphia, United States_	**C05.6** **Trio-based exome sequencing in ten unrelated cases of atypical CdLS** **_Morad Ansari_**, _A. Meynert, H. Bengani, D. Braunholz, D.C. Soares, R.C.M. Hennekam, H. Kayserili, S. Avci, E. Wakeling, J. Tolmie, K. Tatton-Brown, M. Splitt, T. Homfray, A.F. Brady, S.G. Mehta, A. Ross, F.J. Kaiser, M.S. Taylor, D.R. FitzPatrick;_ _Edinburgh, United Kingdom_	**C06.6** **Strategies for Exome Prioritization of Human Disease Genes** **_Damian Smedley_**, _S. Kohler, A. Oellrich, J. Jacobsen, Sanger Mouse Genetics Group, 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_
20.00	_Networking Mixer in the MiCo_					

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

SCIENTIFIC

SCIENTIFIC PROGRAMME

Sunday, June 1, 2014

PROGRAMME

PROGRAMME SUNDAY, JUNE 1

Time	Gold Room	Space 3+4	Brown 3	Brown 1+2	Space 1	Space 2
08.30 - 10.00	**S01 Towards Genomic Personalised Medicine** Chair: P. Pignatti, A. Rauch	**S02 Functional genomics** Chair: G. Matullo, J. Barrett	**S03 Neuronal Migration disorders** Chair: A. Brusco, G. Neri	**S04 Computational Analysis of Gene Networks** Chair: S. Banfi, N. Robinson	**S05 Early development and preimplantation genetics** Chair: L. Stuppia, J. Vermeesch	**ES5 Mosaicism in Human Disease** Chair: L. Larizza
08.30	**S01.1** **From rare disease to management of common disorders** ***Marshall Summar***; *Washington, United States*	**S02.1** **Variation and genetic control of chromatin in humans** ***Bart Deplancke***; *Lausanne, Switzerland*	**S03.1** **Pontocerebellar hypoplasia** ***Kerstin Kutsche***; *Hamburg, Germany*	**S04.1** **Disease, networks and epistasis** ***Caleb Webber***; *Oxford, United Kingdom*	**S05.1** **Dynamic blastomere behaviour** ***Renee Reijo Pera***; *Stanford, United States*	**ES5.1** **Genomic View of Mosaicism and Disease** ***Nancy B. Spinner***, *L.K. Conlin; Philadelphia, United States*
09.00	**S01.2** **Breast cancer genes: beyond BRCA1 and BRCA2** ***Paul Pharoah***; *Cambridge, United Kingdom*	**S02.2** **Control of gene expression in disease** ***Michel Georges***; *Liège, Belgium*	**S03.2** **The neurobiology of lissencephal** ***Anthony Wynshaw-Boris***; *Cleveland, United States*	**S04.2** **Understanding molecular mechanisms of human disease mutations and coding variants through 3D protein networks** ***Haiyuan Yu***; *Ithaca, United States*	**S05.2** **24 chromosome copy number analysis for preimplantation genetic screening** ***Alan H. Handyside***; *Cambridge, United Kingdom*	**ES5.2** **Revertant mosaicism in skin disease** ***Marcel F. Jonkman***, *A.M.G. Pasmooij; Groningen, Netherlands*
09.30	**S01.3** **Age-related Macular Degeneration** ***Caroline Klaver***; *Rotterdam, Netherlands*	**S02.3** **Computational challenges in single-cell transcriptomics** ***John Marioni***; *Hinxton, United Kingdom*	**S03.3** **Neuronal migration defects associated with mutations in tubulins and MT-related proteins** *L. Broix, K. Poirier, Y. Saillour, N. Bahi-Buisson,* ***Jamel Chelly***; *Paris, France*	**S04.3** **From protein networks to disease mechanisms** ***Roded Sharan***; *Tel Aviv, Israel*	**S05.3** **Preimplantation genetic diagnosis** ***Thierry Voet***; *Leuven, Belgium*	
10.00 - 10.30	*Coffee Break / Poster viewing / Exhibition*					
10.30 - 11.30	**Poster viewing with presenters (poster numbers ending with "S")**					
11.30 - 13.30	*Lunch break / Posters / Exhibition / Satellites*					

PROGRAMME SUNDAY, JUNE 1

Time	Gold Room	Space 3+4	Brown 3	Brown 1+2	Space 1	Space 2
13.30 - 15.00	**C07 Implementation of NGS in diagnostics** Chair: F. Girolami, K. Devriendt	**C08 Cancer genetics** Chair: G. Gasparre, A. Carrió-Ybáñez	**C09 Common neurological disease** Chair: D. Tiziano, B. Peterlin	**C10 Bone and skeletal patterning** Chair: A. Percesepe, H. Kääriäinen	**C11 Statistical genetics** Chair: L. Salviati, M. Perola	**C12 Sensory Disorders** Chair: A. Sensi, M. Dündar
13.30	**C07.1** **LysoPlex: an efficient strategy to study the role of lysosomal-autophagic-endocytic pathway** **Giuseppina Di Fruscio***, *A. Schulz, R. De Cegli, M. Mutarelli, M. Savarese, V. Singhmarwah, M. Filocamo, D. Di Bernardo, S. Banfi, T. Braulke, V. Nigro, A. Ballabio;* *Naples, Italy*	**C08.1** **Smc1a cohesin gene mutations in colorectal precancerous lesions** **Francesco Cucco***, *A. Servadio, V. Gatti, P. Bianchi, L. Mannini, A. Prodosmo, E. De Vitis, G. Basso, A. Friuli, L. Laghi, S. Soddu, G. Fontanini, A. Musio;* *Pisa, Italy*	**C09.1** **Functional analysis of SHANK2 mutations identified in schizophrenia patients** **Slavil Peykov***, *S. Berkel, T. Böckers, K. Weiss, G. Schratt, S. Cichon, M. Rietschel, M. Noethen, G. Rappold;* *Heidelberg, Germany*	**C10.1 PLS3 mutations in X-linked osteoporosis and fractures: unraveling a new bone regulatory pathway** **Dimitra Micha***, *F.S. van Dijk, M.C. Zillikens, M. Riessland, C.L.M. Marcelis, C.E. de Die-Smulders, J. Milbradt, A.A. Franken, A.J. Harsevoort, K.D. Lichtenbelt, M.E. Rubio-Gozalbo, H.E. Pruijs, R. Zwertbroek, Y. Moutaouakil, J. Egthuijsen, M. Hammerschmidt, R. Bijman, C. Semeins, A.D. Bakker, V. Everts, J. Klein-Nulend, N. Campos-Obando, A. Hofman, G.J. te Meerman, A.J.M.H. Verkerk, A.J.M.H. Uitterlinden, A. Maugeri, E.A. Sistermans, Q. Waisfisz, H. Meijers-Heijboer, B. Wirth, M.E.H. Simon, G. Pals; Amsterdam, Netherlands*	**C11.1** **Polygenic risk for ADHD is associated with impaired educational achievement and lower IQ in the general population** **Evangelia Stergiakouli***, *J. Martin, M.L. Hamshere, A. Thapar, D.M. Evans, N.J. Timpson, G. Davey Smith;* *Bristol, United Kingdom*	**C12.1** **Next generation sequencing as a reliable and efficient technique to identify mutations in patients with retinal dystrophies** **John Neidhardt**, *N. Glöckle, A. Tiwari, T. Besnard, J. Lemke, S. Kohl, S. Biskup, W. Berger, B. Wissinger;* *Schlieren, Switzerland*
13.45	**C07.2** **Comparing Clinical Exome Sequencing versus Whole Exome Sequencing for monogenic diseases and undiagnosed patients** **Pascal Joset**, *M. Papik, K. Steindl, S. Papuc, M. Vincent, L. Gogoll, D. Niedrist, B. Oneda, A. Baumer, A. Rauch;* *Schlieren-Zurich, Switzerland*	**C08.2 Comprehensive annotation of splice junctions supports pervasive alternative splicing at the BRCA1 locus: a report from the ENIGMA consortium** *Mara Colombo (co-first author), M.J. Blok (co-first author), P. Whiley, M. Santamariña, S. Gutiérrez-Enríquez, A. Romero, P. Garre, A. Becker, L.D. Smith, G. De Vecchi, R.D. Brandão, D. Tserpelis, M. Brown, A. Blanco, S. Bonache, M. Menéndez, C. Houdayer, C. Foglia, J.D. Fackenthal, D. Baralle, B. Wappenschmidt, K. ConFaB, E. Díaz-Rubio, T. Caldés, L. Walker, O. Díez, A. Vega, A.B. Spurdle,* **P. Radice**, *M. de la Hoya; Milano, Italy*	**C09.2** **Exome sequencing of familial parkinsonism in Scandinavia** **Emil K. Gustavsson***, *I. Guella, C. Szu-Tu, D.M. Evans, C. Thompson, H.E. Sherman, H. Han, C. Vilarino-Guell, M.K. Lin, F.T. Pishotta, M. Toft, K. Wirdefeldt, A.C. Belin, M.S. Petersen, J. Aasly, M.J. Farrer, GEOPD Consortium;* *Vancouver, Canada*	**C10.2** **Mutations in plastin 3 cause osteoporosis with fractures. Overexpression of PLS3 and other F-actin bundling proteins influence skeletal development in zebrafish and mice** *J. Milbradt, M. Dimitra, M. Riessland, N. Hamann, F. van Dijk, M. Peters, N. Mendoza Ferreira, M.S. Hosseini Barkooie, E. Janzen, M.C. Zillikens, M. Hammerschmidt, A. Niehoff, G. Pals,* **Brunhilde Wirth**; *Cologne, Germany*	**C11.2** **Polygenic risk score analysis shows shared genetic aetiology between AN and five other psychiatric disorders** **Laura M. Huckins***, *K.S. Mitchell, L. Thornton, WTCCC3 Consortium, GCAN Consortium, D.A. Collier, P.F. Sullivan, C.M. Bulik, E. Zeggini;* *Hinxton, United Kingdom*	**C12.2** **New Hereditary hearing loss (HHL) genes/mutations identified by High throughput sequencing and genotyping in the Italian and Qatari populations.** **Giorgia Girotto***, *D. Vozzi, E. Rubinato, A. Morgan, K. Abdulhadi, D. Vuckovic, M. Di stazio, A. d'Eustacchio, M. La Bianca, R. Badii, P. Gasparini;* *Trieste, Italy*
14.00	**C07.3** **One generic automated workflow for both Sanger and ion semiconductor sequencing in routine DNA diagnostics** **Kornelia Neveling**, *A. Diekstra, E. Bosgoed, A. Rikken, B. van Lier, E.J. Kamsteeg, M. Tychon, R.C. Derks, R.A. van Soest, A.R. Mensenkamp, H. Scheffer, M.R. Nelen;* *Nijmegen, Netherlands*	**C08.3** **Germline mutations in MAP3K6 predispose to gastric cancer** **Daniel Gaston***, *S. Hansford, C. Oliveira, M. Nightingale, H. Pinheiro, C. Macgillivray, P. Kaurah, A.L. Rideout, P. Steele, G. Soares, W. Huang, S. Whitehouse, S. Blowers, M.A. LeBlanc, H. Jiang, W. Greer, M.E. Samuels, A. Orr, C.V. Fernandez, J. Majewski, M. Ludman, S. Dyack, L.S. Penney, C.R. McMaster, D. Huntsman, K. Bedard;* *Halifax, Canada*	**C09.3 Genome-wide analysis of microRNA coding genes in bipolar disorder** **Andreas J. Forstner**, *A. Hofmann, T.W. Mühleisen, M. Leber, T.G. Schulze, J. Strohmaier, F. Degenhardt, J. Treutlein, M. Mattheisen, R. Breuer, S. Meier, S. Herms, P. Hoffmann, A. Lacour, S.H. Witt, A. Reif, B. Müller-Myhsok, S. Lucae, W. Maier, M. Schwarz, A. Pfennig, M. Bauer, M. Hautzinger, S. Moebus, P.M. Czerski, J. Hauser, J. Lissowska, N. Szeszenia-Dabrowska, P. Brennan, J.D. McKay, A. Wright, P.B. Mitchell, J.M. Fullerton, P.R. Schofield, N.G. Martin, G. Babadjanova, M. Alda, P. Grof, G.A. Rouleau, G. Turecki, C. Laprise, F. Rivas, F. Mayoral, M. Kogevinas, M. Grigoroiu-Serbanescu, G. Schratt, T. Becker, M. Rietschel, S. Cichon, M.M. Nöthen; Bonn, Germany*	**C10.3** **XYLT1 mutations in Desbuquois dysplasia type 2** **Celine Huber**, *C. Bui, Y. Alanay, B. Tuysuz, C. Bole-Feysot, J. Leroy, G. Mortier, P. Nitschke, V. Cormier-Daire;* *PARIS, France*	**C11.3** **Efficient estimation of pairwise genetic correlations between hundreds of quantitative traits from population samples of thousands of individuals** **Matti Pirinen**, *C. Benner, T. Lehtimäki, J.G. Eriksson, O.T. Raitakari, M. Järvelin, V. Salomaa, S. Ripatti;* *Helsinki, Finland*	**C12.3** **Disclosure of false disease genes - an underestimated potential of targeted and genomewide NGS: The example of MYO1A and deafness type DFNA48** *T. Eisenberger, N. Di Donato, S.M. Baig, C. Neuhaus, A. Beyer, E. Decker, C. Bergmann,* **Hanno J. Bolz**; *Ingelheim, Germany*

PROGRAMME SUNDAY, JUNE 1

Time	Gold Room	Space 3+4	Brown 3	Brown 1+2	Space 1	Space 2
cont.	**C07 Implementation of NGS in diagnostics** Chair: F. Girolami , K. Devriendt	**C08 Cancer genetics** Chair: G. Gasparre, A. Carrió-Ybáñez	**C09 Common neurological disease** Chair: D. Tiziano, B. Peterlin	**C10 Bone and skeletal patterning** Chair: A. Percesepe, H. Kääriäinen	**C11 Statistical genetics** Chair: L. Salviati, M. Perola	**C12 Sensory Disorders** Chair: A. Sensi, M. Dündar
14.15	**C07.4** **Setting sequencing thresholds for the use of next generation sequencing as a diagnostic tool** *Y. Sun, M.J.V. Hoffer, C.A.L. Ruivenkamp, J.T. den Dunnen, **Gijs W.E. Santen**;* *Leiden, Netherlands*	**C08.4** **Germline mutations in SUFU cause Gorlin syndrome and redefine the risk associated with childhood medulloblastoma.** ***William G. Newman**, M.J. Smith, C. Beetz, S. Williams, Z. Bholah, B. Anderson, S.B. Daly, J. Urquhart, J. O'Sullivan, A. Kelsey, S. Bhaskar, D.G. Evans; Manchester, United Kingdom*	**C09.4** **Imbalance between excitation and inhibition in Neurons derived from *MECP2*, *CDKL5* and *FOXG1* iPSCs** ***Ilaria Meloni**, T. Patriarchi, S. Amabile, A. Bartolini, D. Yasui, E. Calcagno, C. Lo Rizzo, F. Ariani, F. Mari, M. Mencarelli, J.W. Hell, A. Renieri;* *Siena, Italy*	**C10.4** **Mutations in endothelin 1 cause auriculocondylar syndrome and isolated question mark ears** ***Chris T. Gordon**, F. Petit, P. Kroisel, L. Jakobsen, R. Zechi-Ceide, M. Oufadem, C. Bole-Feysot, P. Nitschke, A. Munnich, S. Lyonnet, M. Holder-Espinasse, J. Amiel;* *Paris, France*	**C11.4** **The influence of genotype and phenotype data quality control on SNP based heritability estimates within and across studies.** ***Jouke-Jan Hottenga**, I. Fedko, G. Willemsen, E.J.C.N. de Geus, B. Penninx, D.I. Boomsma;* *Amsterdam, Netherlands*	**C12.4** **AON intravitreal injections to manipulate splicing in retinal cells** ***Xavier Gerard****, I. Perrault, A. Munnich, J. Kaplan, J. Rozet;* *Paris, France*
14.30	**C07.5** **EuroGentest guidelines for diagnostic next generation sequencing** ***Gert Matthijs**, M. Alders, P. Bauer, A. Corveleyn, S. Eck, I. Feenstra, V. Race, H. Scheffer, E. Sistermans, E. Souche, M. Sturm, M. Weiss, H. Yntema, and the Participants to the EuroGentest workshop on Diagnostic NGS Guidelines;* *Leuven, Belgium*	**C08.5** **Evaluation of anticancer chemotherapy genotoxicity using a new p53 functional assay in human lymphocytes** ***Jean-Michel M. Flaman**, E. Kasper, Y. Zerdoumi, F. Soubigou, G. Bougeard, T. Frebourg;* *Rouen, France*	**C09.5** **Left/right asymmetry genes are associated with handedness and appear relevant for neurodevelopmental disorders** ***Silvia Paracchini**, W. Brandler, A. Morris, D. Evans, S. Ring, J. Stein, A. Monaco, J. Talcott, S. Fisher, C. Webber;* *St Andrews, United Kingdom*	**C10.5** **Defects in TAPT1, involved in Axial Skeletal Patterning, Cause a Complex Lethal Recessive Disorder of Skeletal Development** ***Sofie Symoens****, A. Barnes, F. Malfait, K. Vleminckx, W. Steyaert, D. Syx, E. Parthoens, M. Biervliet, G. Gillessen-Kaesbach, J. De Backer, H. Bächinger, A. De Paepe, J.C. Marini, P.J. Coucke;* *Ghent, Belgium*	**C11.5** **Co-regulated transcripts associated to cooperating eSNPs define bi-fan motifs in human gene networks** *Anat Kreimer, **I. Pe'er;*** *New York, United States* *(Change of presenter!)*	**C12.5** **Mutations in the tricarboxylic acid cycle enzyme, Aconitase 2,cause either isolated or syndromic optic neuropathy with encephalopathy and cerebellar atrophy** ***Metodi D. Metodiev**, S. Gerber, L. Hubert, D. Chretien, X. Gérard, P. Amati, N. Boddaert, A. Kaminska, I. Desguerre, J. Kaplan, A. Munnich, A. Rötig, J. Rozet, C. Besmond; Paris, France*
14.45	**C07.6** **Clinical exome sequence performance for reporting secondary genetic findings** ***Eric Londin**, P. Clark, M. Sponziello, L. Kricka, P. Fortina, J.Y. Park;* *Philadelphia, United States*	**C08.6** **Functional analysis of mismatch repair gene variants of uncertain significance and their possible contribution to Lynch syndrome** ***Mariann Kasela****, R. Tricarico, J. Kantelinen, G. Gorelli, M. Genuardi, M. Nyström;* *Helsinki, Finland*	**C09.6** **Exome sequencing to disclose potential new pathogenetic variants in Rett patients without mutations in the known Rett genes** *M. Marchi, F. Cogliati, D. Gentilini, I. Cracco, D. Cittaro, M. Pintaudi, A. Vignoli, L. Giordano, E. Veneselli, B. Ben Zeev, L. Larizza, **Silvia Russo**;* *Milano, Italy*	**C10.6** **ZIC1 mutations cause coronal craniosynostosis and learning disability** ***Stephen R.F. Twigg**, J.A.C. Goos, I. Westbury, S.J. McGowan, M. van Dooren, A.M.W. van den Ouweland, P.J. van der Spek, .. 500 Whole-Genome Sequences (WGS500) Consortium, S.A. Wall, I.M.J. Mathijssen, E. Pauws, A.O.M. Wilkie; Oxford, United Kingdom*	**C11.6** **Inferring the human embryonic selection via genomic data** ***Konstantin Popadin**, P. Makrythanasis, S.E. Antonarakis;* *Geneva, Switzerland*	**C12.6** **Isolated foveal hypoplasia with secondary nystagmus and low vision is associated with a homozygous SLC38A8 mutation** ***Yonatan Perez****, L. Gradstein, H. Flusser, B. Markus, I. Cohen, Y. Langer, M. Marcus, T. Lifshitz, R. Kadir, O.S. Birk;* *Beer Sheva, Israel*
15.00 - 15.30	*Vitamin break / Poster viewing / Exhibition*					

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

PROGRAMME SUNDAY, JUNE 1

Time	Gold Room	Space 3+4	Brown 3	Brown 1+2	Space 1	Space 2
15.30 – 17.00	**WS02 Dysmorphology 1** Organisers: D. Donnai; J Clayton-Smith; S. Douzgou	**WS03 ENSEMBL** Organisers: A. Zadissa; E. Pritchard	**WS04 Practical Bioinformatics: Whole exome sequence analysis** Organisers: N. Robinson	**WS05 Quality assurance** Organisers: E. Dequeker; M. Morris	**WS06 Community genetics - Clinical Genetic Services in 2025** Organisers: M. Cornel; U. Kristoffersson	**WS07 Preimplantation genetic diagnosis** Organisers: J. Vermeesch; E. Iwarsson
	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 14:45 - 15:15 hrs to the lecture room. Maximum time for presentation: 5 minutes.	This workshop is organised and will be presented by Emily Pritchard and Amonida Zadissa, from Ensembl. This workshop is aimed at attendees already using Ensembl, including the bioinformatics community. Participants can follow along with the demonstrations in this workshop if they wish, and for this purpose should bring a fully-charged WiFi enabled laptop. For questions about the workshop, contact Emily Pritchard: emily@ebi.ac.uk.	In this workshop, we will explain the bioinformatics pipelines and algorithms needed to successfully analyse exome data. We will go over the major steps involved, providing intuitive explanations of initial quality control steps, alignments and variant calling, filtering, and gene prioritization for diagnostics and novel disease-gene identification. The workshop is intended for researchers and clinicians who need to understand the process and results of bioinformatics analysis, but additional material will be available upon request for bioinformaticians who need to know the nitty gritty details.	Quality assurance is established as central to clinical genetic testing. For this workshop we have selected poster presentations illustrating the importance of both proactive and retroactive approaches, with an emphasis on the identification, isolation and prevention of potential errors.	Technological changes will change genetic service provision. What will be the task of clinical geneticists and genetic counsellors in 2025 and will primary care, paediatricians, oncologists, public health care and cardiologists integrate much of the current tasks of clinical geneticists? We will discuss scenario's with geneticists from a diversity of countries. *A voting system will be made available to the audience. Connect to the wifi „voting_ws06“ with any WIFI-capable device (laptop, tablet, phone) and open an internet browser. The voting form will be displayed accordingly.*	The workshop will discuss some of the hot topics in PGD/PGS and will be in the form of a debate. The following questions will be addressed: - Preimplantation genetic screening: a fallacy or the holy grail towards improved IVF outcome. - Preimplantation genetic diagnosis: The beginning of the end of rare Mendelian inherited disorders? - When to opt for PGD/PGS: the patient dilemma. - Is PGS leading to a two tiered health care system? *A voting system will be made available to the audience. Connect to the wifi „voting_ws07“ with any WIFI-capable device (laptop, tablet, phone) and open an internet browser. The voting form will be displayed accordingly.*
17.00 – 17.30	*Coffee break / Poster viewing / Exhibition*					

Detailed Workshop programmes (as submitted by the organisers) can be found in the “ESHG Bulletin” in the conference bag.

PROGRAMME SUNDAY, JUNE 1

Time	Gold Room	Space 3+4	Brown 3	Brown 1+2	Space 1	Space 2
17.30 - 19.00	**S06 Risk perception and risk communication,** joint with EMPAG Chair: B. Dallapiccola, T. Clancy	**S07 Therapy for human genetic diseases** Chair: M. De Marchi, M. Seri	**S08 Population genetics in a globalized world** Chair: G. Romeo, P. de Bakker	**S09 Advances and new challenges in genetics of cardiovascular diseases**, joint with the European Society of Cardiology Chair: A. Ferlini, X. Jeunemaitre	**S10 New Mutational Mechanisms** Chair: I. Ceccherini, L. Larizza	**ES6 How to be successful in rare disease gene Identification** Chair: D. Grinberg
17.30	**S06.1** **Risk is More Than a Number: About Risks and People's Perceptions of Genetic Risks** ***Danielle R.M. Timmermans***; *Amsterdam, Netherlands*	**S07.2** **Epithelial stem cell in cell and gene therapy** ***Michele De Luca***; *Modena, Italy* *(Change of sequence!)*	**S08.1** **Demographic inference from identity by descent** ***Itsik Pe'er;*** *New York, United States*	**S09.1** **Twenty-five years of research in sarcomeric cardiomyopathies and therapeutic perspectives** ***Hugh Watkins***; *Oxford, United Kingdom*	**S10.1** **Chromotrypsis** ***Edwin Cuppen***, *W. Kloosterman;* *Utrecht, Netherlands*	**ES6.1** **Strategies for rare disease gene discovery in the era of next-generation sequencing** ***Fowzan S. Alkuraya*** *King Faisal Specialist Hospital and Alfaisal University, Riyadh, Saudi Arabia* ***Kym M. Boycott*** *Children's Hospital of Eastern Ontario Research Institute, University of Ottawa, Ottawa, Canada*
18.00	**S06.2** **Risk perception: what could be at stake in multiple genetic testing?** ***Claire M. Julian-Reynier***; *Marseille, France*	**S07.1** **Gene therapy of human genetic diseases with AAV vectors** ***Alberto Auricchio***; *Napoli, Italy* *(Change of sequence!)*	**S08.2** **Insights into European genetic history at fine geographic scales using haplotype-based approaches** ***Simon Myers***; *Oxford, United Kingdom*	**S09.2** **Mendelian Randomization** ***Michael V. Holmes***; *Philadelphia, United States*	**S10.2** **Kataegis: a mutation signature identified through whole-genome sequencing of human cancers** ***Serena Nik-Zainal***, *L.B. Alexandrov, B.J. Taylor, Y. Wu, D. Wedge, C. Rada, P.J. Campbell, M. Neuberger, M.R. Stratton;* *Cambridge, United Kingdom*	
18.30	**S06.3** **Risk Communication Methods for Helping Patients Understand the Risks and Benefits of Genetic Testing** ***Angie Fagerlin***; *Ann Arbor, United States*	**S07.3** **Therapeutic targeting of Phosphatidylinositol-3-kinase/AKT/mTOR signalling in segmental overgrowth disorders** ***Rob Semple***; *Cambridge, United Kingdom*	**S08.3** **The role of population isolates in understanding genetic and complex diseases** ***Paolo Gasparini***; *Trieste, Italy*	**S09.3** **Genetic testing in the clinical arena, current and future perspectives** ***Philippe Charron***; *Paris, France*	**S10.3** **Medulloblastoma links chromothripsis with TP53 mutations** ***Jan O. Korbel***; *Heidelberg, Germany*	

	Amber 3+4
19.15 – 20.15	**ESHG Membership Meeting** **All ESHG members welcome!**

SCIENTIFIC PROGRAMME

Monday, June 2, 2014

PROGRAMME MONDAY, JUNE 2

Time	Gold Room	Space 3+4	Brown 3	Brown 1+2	Space 1
08.30 - 10.00	**S11 Rare copy number variants in common traits** Chair: S. D'Alfonso, B. Newman	**S12 Epigenetic basis of disease** Chair: A. Riccio, M. Nyström	**S13 Non-invasive prenatal testing**, joint with EMPAG Chair: C. Rosatelli, E. Iwarsson	**S14 Rapid genome diagnostics** Chair: A. Pizzuti, D. Fitzpatrick	**ES7 From mutation identification to therapy** Chair: G. Neri
08.30	**S11.1** **Copy number alterations in skin disorders** ***Xue Zhang***; *Beijing, China*	**S12.1** **The Epigenetic Basis of Common Human Disease** ***Andrew P. Feinberg***; *Baltimore, United States*	**S13.1** **State of the Art of Non-Invasive Prenatal Testing** ***Lyn S. Chitty***; *London, United Kingdom*	**S14.1** **Developments in rapid DNA sequencing technology** ***John Tyson;*** *Newcastle upon Tyne, United Kingdom*	**ES7.1** **From Mutations in the Few to Drugs for the Many** ***Michael R. Hayden***; *Petah-Tikva, Israel*
09.00	**S11.2** **Congenital heart disease** ***Bernard Keavney***; *Manchester, United Kingdom*	**S12.2** **Intergenerational epigenetic programming in a mouse model of undernutrition** ***Anne Ferguson-Smith***; *Cambridge, United Kingdom*	**S13.2** **Noninvasive prenatal testing creates an opportunity for antenatal treatment of Down syndrome** ***Diana W. Bianchi***, *T. Tarui, M. Ferres, J. Pennings, D. Slonim, F. Guedj; Boston, United States*	**S14.2** **DNA sequencing in neonatal intensive care units** ***Stephen Kingsmore***; *Kansas City, United States*	**ES7.2** **Genetic, cell biological and clinical interrogation of disease-causing CFTR mutations informs strategies for future drug discovery** ***Christine E. Bear***, *S. Molinski, T. Gonska, L. Huan, B. Baskin, I. Janahi, P.N. Ray; Toronto, Canada*
09.30	**S11.3** **Copy number variants are a common cause of short stature** ***Christian T. Thiel***, *A. Reis, H. Dörr, A. Rauch; Erlangen, Germany*	**S12.3** **Cancer Genetics and Epigenetics: Two Sides of the Same Coin?** ***Peter A. Jones***; *Grand Rapids, United States*	**S13.3** **Clinical and social implications of NIPT** ***Kelly E. Ormond***; *Stanford, United States*	**S14.3** **Impact of rapid DNA sequencing on diagnostic and public health microbiology** ***Claudio U. Köser***; *Cambridge, United Kingdom*	
10.00 - 10.30	*Coffee break / Poster viewing / Exhibiton*				
10.30 - 11.30	**Poster viewing with presenters (poster numbers ending with "M")**				
11.30 - 13.30	*Lunch break / Posters / Exhibition / Satellites*				

PROGRAMME MONDAY, JUNE 2

Time	Gold Room	Space 3+4	Brown 3	Brown 1+2	Space 1
13.30 - 15.00	**C13 Innovation in genetic services** Chair: P. Grammatico, D. Coviello	**C14 Genetics of complex traits** Chair: M. Clementi, M. Nöthen	**C15 Novel genes in neurogenetic disorders** Chair: E. Valente, D. Lev	**C16 Genes and development 2** Chair: F. Lalatta, Y. Alanay	**C17 Metabolic and mitochondrial disorders** Chair: V. Capra, N. Canki-Klain
13.30	**C13.1** **Stratified cancer screening in Europe using genomic information: conclusions and recommendations from the COGS project** ***Thomas H.S. Dent***, *S. Chowdhury, A. Hall, N. Pashayan, P.D.P. Pharoah, H. Burton;* *Cambridge, United Kingdom*	**C14.1** **Insights into the genetic architecture of anthropometric traits using whole genome sequence data** ***Eleftheria Zeggini***, *UK10K consortium;* *Hinxton, United Kingdom*	**C15.1** **BCAP31 mutations cause a new X-linked syndrome with deafness, dystonia, central hypomyelination and disorganization of the Golgi apparatus** *P. Cacciagli, J. Sutera-Sardo, A. Borges-Correia, J. Roux, I. Dorboz, J. Desvignes, C. Badens, M. Delepine, M. Lathrop, P. Cau, N. Lévy, N. Girard, P. Sarda, O. Boespflug-Tanguy,* ***Laurent Villard****;* *Marseille, France*	**C16.1** **A congenital disorder of glycosylation, with lymphopenia, neutropenia, and skeletal dysplasia, caused by mutations in the gene encoding phosphoglucomutase 3 (PGM3).** ***Asbjorg Stray-Pedersen***, *P.H. Backe, H.S. Sorte, L. Mørkrid, N.Y. Chokshi, H. Erichsen, T. Gambin, K.B.P. Elgstøen, M. Bjørås, M. Wlodarski, M. Krüger, S. Jhangiani, D.M. Muzny, A. Patel, K. Raymond, G.S. Sasa, R.A. Krance, C.A. Martinez, S.M. Abraham, C. Speckmann, S. Ehl, P. Hall, L. Forbes, E. Merckoll, J. Westvik, G. Nishimura, C.F. Rustad, T.G. Abrahamsen, A. Rønnestad, L.T. Osnes, T. Egeland, O.K. Rødningen, C.R. Beck, E. Boerwinkle, R.A. Gibbs, J.R. Lupski, J.S. Orange, E. Lausch, I. Hanson;* *Houston, United States*	**C17.1** **A dominant mutation in CHCHD10 causes neurodegenerative disorder with mitochondrial DNA instability** ***Veronique Paquis-Flucklinger***, *S. Bannwarth, S. Saadi, A. Chaussenot, E. Genin, K. Fragaki, L. Berg-Alonso, S. Lacas-Gervais, V. Serre, A. Verschueren, C. Rouzier, G. Augé, C. Cochaud, F. Lespinasse, J. Pouget;* *Nice, France*
13.45	**C13.2** **Expanding access to genetic counseling for hereditary breast and ovarian cancer with telephone delivery: A cluster randomized noninferiority trial** ***Anita Y. Kinney***, *K.M. Butler, M.D. Schwartz, J.S. Mandelblatt, K.M. Boucher, L.M. Pappas, A. Gammon, W. Kohlmann, S.L. Edwards, A.M. Stroup, K.G. Flores, R.A. Campo;* *Albuquerque, United States*	**C14.2** **Genome of the Netherlands imputation identifies seven new loci for quantitative ECG traits in meta-analysis of 30,000 samples** ***Jessica van Setten******, *N. Verweij, M.N. Niemeijer, S. Trompet, H. Mbarek, M. Eijgelsheim, J.J. Hottenga, J.A. Kors, E.M. van Leeuwen, P.W. Macfarlane, A. Hofman, B.H. Stricker, J.W. Jukema, C.M. van Duijn, D.I. Boomsma, E.J.C. de Geus, P. van der Harst, P.I.W. de Bakker, A. Isaacs;* *Utrecht, Netherlands*	**C15.2** **Mutations in KPTN Cause Macrocephaly, Neurodevelopmental Delay, and Seizures** ***Emma L. Baple******, *R. Maroofian, B. Chioza, M. Izadi, H.E. Cross, S. Al-Turki, K. Barwick, A. Skrzypiec, R. Pawlak, K. Wagner, R. Coblentz, T. Zainy, M.A. Patton, S. Mansour, P. Rich, B. Qualmann, M.E. Hurles, M.M. Kessels, A.H. Crosby;* *Exeter, United Kingdom*	**C16.2** **Lenz-Majewski syndrome: disturbed phosphatidylserine metabolism causes intellectual disability and a sclerosing bone dysplasia** ***Sérgio B. Sousa******, *D. Jenkins, E. Chanudet, G. Tasseva, E. Bliss, M. Ishida, J. Sá, J.M. Saraiva, A. Barnicoat, R. Scott, A. Calder, D. Wattanasirichaigoon, K. Chrzanowska, M. Simandlová, L. Van Maldergem, A. Hing, M. Silengo, G. Glenn Anderson, J. Docker, M. Ryten, J. Pereira, K. Mills, P. Clayton, P. Stanier, P. Beales, J.E. Vance, G.E. Moore;* *Coimbra, Portugal*	**C17.2** **Decoding Mitochondrial Disorders using Exome Sequencing** ***Laura Kremer******, *T. Haack, R. Kopajtich, B. Haberberger, C. Biagosch, T. Wieland, E. Graf, T. Schwarzmayr, T. Strom, P. Freisinger, T. Klopstock, T. Meitinger, W. Sperl, J. Mayr, H. Prokisch;* *Neuherberg, Germany*
14.00	**C13.3** **New approaches to bridge the gap between genetics research and primary health care in Ireland** ***Sally Ann Lynch***, *R. O' Shea, R. O' Shea, J. Turner, A. Ward, M. Byrne, J. Casey;* *Dublin 12, Ireland*	**C14.3** **Genome-wide association analysis identifies a new gene involved in salt perception and liking** ***Antonietta Robino***, *N. Pirastu, C. Mansfield, D. Hwang, D.R. Reed, P. Gasparini;* *Trieste, Italy*	**C15.3** **REPS1 is a novel gene of Neurodegeneration with Brain Iron Accumulation** ***Anthony B. Drecourt******, *N. Boddaert, I. Desguerre, D. Chretien, A. Munnich, A. Rötig;* *Paris, France*	**C16.3** **Homozygous *FIBP* truncating mutation in a new multiple congenital anomalies syndrome with overgrowth, macrocephaly, Iris coloboma, and learning disabilities** ***Christel Thauvin-Robinet***, *D. Picot, L. Duplomb-Jego, J. Thevenon, B. Terriat, D. Minot, J. St-Onge, Y. Duffourd, P. Vabres, J. Rivière, L. Faivre;* *Dijon, France*	**C17.3** **Lentivial vector based hematopoietic stem cell gene therapy mediates sustained expression of functional thymidine phosphorylase in mitochondrial neurogastrointestinal encephalopathy mouse model** ***Rana Yadak******, *J. Torres-Torronteras, Y. Cámara, E. Bogaerts, G. de Ruijter, R. Martı, N. van Til, G. Wagemaker, I. de Coo;* *Rotterdam, Netherlands*

PROGRAMME MONDAY, JUNE 2

Time	Gold Room	Space 3+4	Brown 3	Brown 1+2	Space 1
cont.	**C13 Innovation in genetic services** Chair: P. Grammatico, D. Coviello	**C14 Genetics of complex traits** Chair: M. Clementi, M. Nöthen	**C15 Novel genes in neurogenetic disorders** Chair: E. Valente, D. Lev	**C16 Genes and development 2** Chair: F. Lalatta, Y. Alanay	**C17 Metabolic and mitochondrial disorders** Chair: V. Capra, N. Canki-Klain
14.15	**C13.4** **Unanticipated results in whole exome study: we‘ve still a lot to learn** ***Cecile Skrzynia***, *J.M. O‘Daniel, D. Marchuk, K. Lee, J.S. Berg, J.P. Evans;* *Chapel Hill, United States*	**C14.4** **ImmunoSeq: Discovery of novel rare variants implicated in autoimmune and inflammatory diseases by targeting regulatory regions in immune cells** ***Andréanne Morin****, *T. Kwan, L. Letourneau, K. Tandre, M. Eloranta, V. Arseneault, M. Caron, A. Madore, G. Bourque, A. Montpetit, A. Syvanen, L. Ronnblom, M.G. Lathrop, C. Laprise, T. Pastinen;* *Montréal, Canada*	**C15.4** **Interferon type 1 response regulator *USP18* is mutated in severe pseudo-TORCH syndrome** ***Marije Meuwissen****, *R. Schot, G. Oudesluijs, S. Tinchert, L. van Unen, D. Heijsman, M. Lequin, M. Kros, R. Willemsen, R. Brouwer, W. van IJcken, R. de Coo, J. Dudink, A. Bertoli Avella, F. Verheijen, G. Mancini;* *Rotterdam, Netherlands*	**C16.4** **Hidden mutations in Cornelia de Lange Syndrome (CdLS) - Limitations of Sanger sequencing in molecular diagnostics** ***Frank J. Kaiser***, *D. Braunholz, J. Eckhold, J. Pozojevic, K. Wendt, E. Watrin, H. Rieder, G. Gillessen-Kaesbach;* *Lübeck, Germany*	**C17.4** **Deletion of a distant-acting enhancer near *C16ORF91* underlies recessive congenital diarrhea** ***Danit Oz-Levi****, *I. bar Joseph, T. Olender, D. Marek-Yagel, A. Alkelai, E.K. Ruzzo, P. Tatarsky, H. Reznik-Wolf, C. Hartman, R. Shamir, R. Kleta, D.B. Goldstein, E. Pras, L.A. Pennacchio, D. Lancet, Y. Anikster;* *Rehovot, Israel*
14.30	**C13.5** **The stepping stone approach towards the Genetics Clinic of the Future** *D. Schuurbiers, G. Bertier, M. Radstake, P. Bauer, C. Bock, P. Borry, A. Bredenoord, A. Brookes, X. Estivill, R. Hennekam, L. Johnston, H. Kääriäinen, N. Knoers, F. Lescai, R. Maalman, F. Nielsen, G. van Ommen, C. Oosterwijk, J. Paschall, B. Prainsack, J. Saarela, R. Sudbrak, M. Swertz, H. Teare,* ***Terry Vrijenhoek****;* *Utrecht, Netherlands*	**C14.5** **Exome array analysis in >30,000 Europeans establishes a functional role for G6PC2 and identifies novel coding variants influencing glycaemic traits** ***Anubha Mahajan***, *X. Sim, H.J. Ng, A.K. Manning, M.A. Rivas, H.M. Highland, A.E. Locke, N. Grarup, H.K. Im, A.P. Morris, J.B. Meigs, C.M. Lindgren, A.L. Gloyn, on behalf of T2D-GENES and GoT2D consortia;* *Oxford, United Kingdom*	**C15.5** **Loss of CTNND2 is associated with borderline intellectual dysfunction in humans and neuronal migration defects in zebrafish** *W. Hofmeister, D. Nilsson, A. Topa, B. Anderlid, F. Vezzi, V. Wirta, M. Nordenskjöld, E. Syk Lundberg,* ***Anna Lindstrand****;* *Stockholm, Sweden*	**C16.5** **RNA Polymerase II activity is affected at the promoter regions in SMC1A-mutated Cornelia de Lange Syndrome cells** ***Linda Mannini***, *S. Bilodeau, C. Amato, V. Quarantotti, F. Cucco, I.D. Krantz, A. Musio;* *Pisa, Italy*	**C17.5** **Genetic testing leads clinical care in neonatal diabetes: a new paradigm** ***Elisa De Franco****, *S.E. Flanagan, J.A.L. Houghton, H. Lango Allen, R. Caswell, D.J.G. Mackay, K.I. Temple, S. Ellard, A.T. Hattersley;* *Exeter, United Kingdom*
14.45	**C13.6** **Teaching Genomic Medicine to Physicians - this is our responsibility as medical geneticists** ***Idit Maya***, *L. Basel-Vanagaite, E. Taub, A. Koifman, D.M. Behar, R. Tomashov-Matar, R. Sukenik-Halevi, D. Marom, A. Reches, M. Shaohat;* *Petah Tikva, Israel*	**C14.6** **Transethnic association study of IBD identifies novel risk loci and shows pervasive sharing of genetic risk factors across populations** ***Jimmy Z. Liu****, *S. van Sommeren, R.K. Weersma, C.A. Anderson, The International IBD Genetics Consortium;* *Hinxton, United Kingdom*	**C15.6** **Novel (ovario) leukodystrophy related to *AARS2* mutations** ***Sietske H. Kevelam****, *C. Dallabona, D. Diodato, T.B. Haack, L. Wong, G.S. Salomons, E. Baruffini, L. Melchionda, C. Mariotti, T.M. Strom, T. Meitinger, H. Prokisch, K. Chapman, A. Colley, H. Rocha, K. Őunap, R. Schiffmann, E. Salsano, M. Savoiardo, E. Hamilton, T.E.M. Abbink, N.I. Wolf, I. Ferrero, C. Lamperti, M. Zeviani, A. Vanderver, D. Ghezzi, M.S. van der Knaap;* *Amsterdam, Netherlands*	**C16.6** **In silico and functional characterization of KMT2D/MLL2 missense mutations as causative in Kabuki syndrome** ***Pasquelena De Nittis****, *L. Micale, B. Augello, C. Fusco, A. Romano, B. Piccinni, M. Pellico, B. Mandriani, C. Rinaldi, A. Di Lauro, T. Verri, L. Zelante, G. Merla;* *San Giovanni Rotondo, Italy*	**C17.6** **Safety and efficacy of pravastatin and zoledronate association in Hutchinson-Gilford Progeria: two-years treatment results of a phase II, open label, single arm clinical trial (ClinicalTrials.gov #NCT00731016)** ***Annachiara De Sandre-Giovannoli***, *S. Sigaudy, P. Bourgeois, S. Miloudi, P. Roll, G. Gorincour, J. Gentet, F. Sabatier, L. Arnaud, N. André, P. Cau, R. Truillet, E. Jouve, J. Micallef, N. Lévy;* *Marseille, France*
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 2

Time	Gold Room	Space 3+4	Brown 3	Brown 1+2	Space 1	Space 2
15.30 - 17.00	**WS08 Dysmorphology 2** Organisers: D. Donnai; J Clayton-Smith S. Dougzou	**WS09 Genome Browser UCSC** Organiser: R. Kuhn	**WS10 Analysis, interpretation and reporting of array data** Organisers: N. de Leeuw; C. van Ravenswaaij-Arts	**WS11 Clinical Cancer Genetics Club** Organisers: M. Genuardi; D. Stoppa-Lyonnet	**WS12 Preconception and prenatal screening** Organisers: M. Macek Jr.; T.H. Bui	**WS13 Next Generation Sequencing** Organisers: J. Veltman
	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 14:45 - 15:15 hrs to the lecture room. Maximum time for presentation: 5 minutes.	This advanced workshop on the UCSC Genome Browser will feature new navigation features; use of the Table Browser for data-mining, including intersections and filtering; saving/sharing sessions; Custom Tracks and Track Data hubs. Data-handling for high-throughput sequencing datasets will discussed, including support of user-hosted large datasets in BAM, VCF, bigBed and bigWig formats. Previous familiarity with the Browser is useful, but not necessary.	The aim of this workshop is to focus on various aspects of array analysis, interpretation and reporting in a diagnostic setting. We will use illustrative cases from our own diagnostic laboratories to discuss the more challenging findings, including low-penetrant, recurrent Copy Number Variants (CNVs) and imbalances on the X-chromosome as well as SNP genotype information leading to the disease cause. We plan to have an app-based feedback system available for this interactive session, so please bring your smart phone, tablet or laptop. Participants are invited to send questions, comments or suggestions related to this topic by e-mail to Nicole.deLeeuw@radboudumc.nl before June 1, 2014. *A voting system will be made available to the audience. Connect to the wifi „voting_ws09“ with any WIFI-capable device (laptop, tablet, phone) and open an internet browser. The voting form will be displayed accordingly.*	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 by attendants who can contribute cases or case series with the above outlined characteristics are strongly encouraged. Each presentation should last not more than 5 minutes (maximum 3 slides).	The objective of this workshop is to discuss pro and cons of the latest advances in noninvasive prenatal testing (NIPT), demonstrate its utility in the general obstetrical population and discuss the complementary nature of preconception and neonatal screening in the case of cystic fibrosis.	In this workshop several European experts will present and discuss patients for which diagnostic next generation sequencing (both targeted and exome) 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).
17.00 - 17.30	*Coffee break / Poster removal / Exhibition*					

Detailed Workshop programmes (if submitted by the organisers) can be found in the “ESHG Bulletin” in the conference bag.

PROGRAMME MONDAY, JUNE 2

Time	Gold Room	Space 3+4	Brown 3	Brown 1+2	Space 1	Space 2
17.30 - 19.00	**S15 Networks and pathways in genetic diseases** Chair: P. Finelli, F. Palau	**S16 The new RNA world** Chair: G. Casari, A. Amoroso	**S17 Tumour heterogeneity** Chair: L. Chessa, J. Machado	**S18 A novel class of disease of lipid metabolism** Chair: G. Ferrero, A. Brice	**S19 Three sequencing approaches in complex disease** Chair: F. Macciardi, J. Veltman	**ES8 Current developments in legal aspects of genetics: Untangling the law and what it means for you** Chair: M. Cornel
17.30	**S15.1** **Signaling networks in the auditory sensory cells unveiled by hereditary deafness** ***Christine Petit***; *Paris, France*	**S16.1** **SINEUPs: a new functional class of antisense non-coding RNAs that activate translation** ***Stefano Gustincich***; *Trieste, Italy*	**S17.1** **Cancer genetic heterogeneity: implications for therapy responsiveness and acquisition of therapy resistance** ***Sandra Misale***; *Candiolo, Italy* *(change of presenter)*	**S18.3** **Update on lipidomic approaches in disorders affecting complex lipids metabolism: the example of cardiolipin** ***Frederic M. Vaz***; *Amsterdam, Netherlands* *(Change of sequence!)*	**S19.1** **Whole genome sequencing of 4000 individuals provides insight into genetic architecture of complex traits** ***Nicole Soranzo***; *Hinxton, United Kingdom*	**ES8.1** **New Proposals for the Regulation of in vitro Diagnostic Devices (IVDs)** ***David E. Barton***, *S. Hogarth*; *Dublin, Ireland*
18.00	**S15.2** **Genes and cellular pathway of Fanconi's anemia** ***Jordi Surrallés***; *Barcelona, Spain*	**S16.2** **Molecular function of the repetitive (epi)genome in normal physiology and in disease** ***Davide Gabellini***; *Milan, Italy*	**S17.2** **Non-cell autonomous interactions promote sub-clonal heterogeneity** ***Andriy Marusyk***, *D. Tabassum, V. Almendro, P. Altrock, F. Michor, K. Polyak*; *Boston, United States*	**S18.2** **Disorders of phospholipids, sphingolipids and fatty acids biosynthesis** ***Fanny Mochel***; *Paris, France*	**S19.2** **Using transcriptome sequencing to understand mechanisms of disease** ***Tuuli Lappalainen***; *New York, United States*	**ES8.2** **Data protection regulation** ***David Townend***; *Maastricht, Netherlands*
18.30	**S15.3** **Analysis of signalling pathways in Tbx1 mutants identifies a novel mechanism in coronary artery Morphogenesis** ***Peter J. Scambler***, *S. Ivins, J. Chappell, J. Suntharalingham, B. Vernay, T. Mohun*; *London, United Kingdom*	**S16.3** **The SMN complex: RNA processing and motor neuron disease** ***Livio Pellizoni***; *New York, United States*	**S17.3** **Circulating tumor cells: Detection, biology and clinical implications** ***Klaus Pantel***; *Hamburg, Germany*	**S18.1** **Glyco-lipophobia: association with disorders of glycolipid and glycosylphosphatidylinositol anchor synthesis** ***Hudson H. Freeze***; *La Jolla, United States* *(Change of sequence!)*	**S19.3** **High resolution genetic analysis to detect variants associated with quantitative traits and diseases in the founder Sardinian population** ***Francesco Cucca***; *Sassari, Italy*	
20.30	*Networking Party at the Old Fashion Club*					

SCIENTIFIC

SCIENTIFIC PROGRAMME

Tuesday, June 3, 2014

PROGRAMME

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PROGRAMME TUESDAY, JUNE 3

Time	Gold Room
09.00 - 10.30	**ESHG-ASHG Building Bridges Session PL3: “Towards finding global agreement on...” What IF... (Incidental Findings), an interactive Debate,** joint with EMPAG
	Diagnostic exome and genome sequencing data can be interrogated for clinically relevant variants other than those relevant for a diagnostic request. There are different opinions on the way to deal with these „incidental findings“ in the clinic, on the potential benefits and risks to patients, on patient autonomy and on the obligation of laboratories to report these findings. These will be debated with representatives from both sides of the Atlantic. *Moderator: Han Brunner, The Netherlands* *Discussants:* • *Angus Clarke, Cardifff, United Kingdom* • *Martina Cornel, Amsterdam, The Netherlands* • *Robert Green, Boston, United States* • *Stephen Kingsmore, Kansas City, United States* • *Marjolein Kriek, Leiden, The Netherlands* • *Arnold Munnich, Paris, France* *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 on Level 1 & 2*

PROGRAMME TUESDAY, JUNE 3

Time	Gold Room	Space 3+4	Brown 3	Brown 1+2	Space 1
11.00 - 12.30	**C18 Large scale genomics** Chair: O. Zuffardi, H. Scheffer	**C19 Internal organs** Chair: M. Zollino, B. Melegh	**C20 Basic mechanisms in genetics** Chair: B. Franco, S. Lyonnet	**C21 Rasopathies and CDG** Chair: F. Sangiuolo, K. Writzl	**C22 Returning results: Ethical and legal issues,** joint with EMPAG Chair: F. Faravelli, M. Cornel
11.00	**C18.1** **Molecular Inversion Probe based Resequencing Identifies Recurrently Mutated Genes in Intellectual Disability** ***Alexander Hoischen***, *L.E.L.M. Vissers, B.P. Coe, C. Gilissen, A. Vulto-Silfhout, J. Schuurs-Hoeijmakers, M. van de Vorst, M. Steehouwer, P. de Vries, C. Baker, K. Witherspoon, B.J. O'Roak, J. Shendure, H.G. Brunner, C. Romano, B.B.A. de Vries, J.A. Veltman, E.E. Eichler;* *Nijmegen, Netherlands*	**C19.1** **Constitutive Activation of PRKACA in Adrenal Cushing's Syndrome** ***Thomas Schwarzmayr****, *F. Beuschlein, M. Fassnacht, G. Assié, D. Calebiro, C.A. Stratakis, A. Osswald, C.L. Ronchi, T. Wieland, S. Sbiera, F.R. Faucz, K. Schaak, A. Schmittfull, O. Barreau, D. Vezzosi, M. Rizk-Rabbin, U. Zabel, E. Szarek, P. Salpea, A. Forlino, A. Vetro, O. Zuffardi, C. Kisker, S. Diener, T. Meitinger, M.J. Lohse, M. Reincke, J. Bertherat, T.M. Strom, B. Allolio;* *Neuherberg, Germany*	**C20.1** **Single cell allele-specific expression (ASE) in Down syndrome and common aneuploidies** ***Georgios Stamoulis****, *P.G. Ferreira, P. Makrythanasis, F. Santoni, M. Guipponi, M. Garieri, E. Falconnet, P. Ribaux, E.T. Dermitzakis, C. Borel, S.E. Antonarakis;* *Geneva, Switzerland*	**C21.1** **Mutations in POGLUT1, encoding protein O-glucosyltransferase 1, cause autosomal dominant Dowling-Degos disease** ***Fitnat B. Basmanav****, *A. Oprisoreanu, S.M. Pasternack, H. Thiele, G. Fritz, J. Wenzel, L. Größer, M. Wehner, S. Wolf, C. Fagerberg, A. Bygum, J. Altmüller, A. Rütten, L. Parmentier, L. El Shabrawi-Caelen, C. Hafner, P. Nürnberg, R. Kruse, S. Schoch, S. Hanneken, R.C. Betz;* *Bonn, Germany*	**C22.1** **The impact of reporting exome and whole genome sequencing: Predicted frequencies of primary, secondary and incidental findings based on modelling** ***Leslie Burnett***, *L.C. Ding, R.M. Lew, D. Chesher, A.L. Proos;* *Sydney, Australia*
11.15	**C18.2** **Efficient molecular diagnosis of Intellectual Disability: targeted High throughput exon sequencing of 217 ID genes detects causative mutations in at least 26 of 106 tested patients** ***Jean Louis Mandel***, *C. Redin, J. Muller, B. Gérard, B. Jost, M. Dumas, S. Le Gras, the French clinical genetics of ID consortium, D. Bonneau, H. Dollfus, Y. Alembik, E. Flori, V. Drouin Garraud, D. Lacombe, M. Doco Fenzy, P. Sarda, D. Geneviève, P. Edery, B. Isidor, L. Olivier Faivre, A. Piton;* *Illkirch, CU Strasbourg, France*	**C19.2** **A mutation in SEC61A1 causes autosomal dominant interstitial kidney disorder associated with anemia and growth retardation** ***Bart Loeys***, *N. Bolar, C. Golzio, C. Van Hemelrijk, A. Hoischen, J. Huyghe, A. Raes, E. Matthys, E. Sys, M. Gubler, C. Antignac, M. Azou, G. Van Camp, S. Kmoch, A. Bleyer, J. Vande Walle, G. Mortier, H. Brunner, L. Van Laer, N. Katsanis;* *Antwerp, Belgium*	**C20.2** **Distinct properties of de novo mutations from whole genome sequencing of 50 patient-parent trios** ***Michele Pinelli***, *B. Tan, J.M. van de Vorst, R. Leach, R. Klein, L.E.L.M. Vissers, H.G. Brunner, J.A. Veltman, A. Hoischen, C. Gilissen;* *Nijmegen, Netherlands*	**C21.2** **The phenotypic spectrum of SHOC2 c.4A>G (p.Ser2Gly)** ***Christina Lissewski****, *L. Mazzanti, G. Zampino, V. Cordeddu, E. Burkitt-Wright, A. De Luca, C. Rossi, I. van der Burgt, A. Verloes, B. Dallapiccola, B.D. Gelb, B. Kerr, K. Kutsche, H. Cavé, M. Tartaglia, M. Zenker;* *Magdeburg, Germany*	**C22.2** **Defending the child's right to an open future concerning genetic information** ***Annelien L. Bredenoord****, *M.C. de Vries, J.J. van Delden;* *Utrecht, Netherlands*
11.30	**C18.3** **Comprehensive NGS based diagnostics in over 1000 patients with epileptic disorders** ***Isabelle Steiner***, *M. Doecker, A.C. Russ, J. Juengling, K. Reicherter, J. Hoffmann, S. Fehr, F. Battke, J. Lemke, H. Lerche, S. Biskup, K. Hoertnagel;* *Tübingen, Germany*	**C19.3** **TJP2 deficiency: a new cholestatic liver disease** ***Melissa Sambrotta****, *S. Strautnieks, E. Papouli, P. Rushton, B.E. Clark, D.A. Parry, L. Brett, C.V. Logan, L.J. Newbury, B.M. Kamath, S. Ling, T. Grammatikopoulos, B.E. Wagner, J.C. Magee, R.J. Sokol, G. Mieli-Vergani, University of Washington Center for Mendelian Genomics, J.D. Smith, C.A. Johnson, S. Davison, P. McClean, M.A. Simpson, A.S. Knisely, L.N. Bull, R.J. Thompson;* *London, United Kingdom*	**C20.3** **Study of the regulatory landscape of *SHOX* in 503 LWD and ISS cases uncover a key role of the upstream *cis*-regulatory element CNE-3** *H. Verdin, A. Fernández Miñán, K. De Leeneer, L. Borms, S. Janssens, B. Callewaert, F. Malfait, A. Kariminejad, K. De Waele, J. De Schepper, I. François, A. Dheedene, B. Menten, J.L. Gómez-Skarmeta,* ***Elfride De Baere****; *Ghent, Belgium*	**C21.3** **Heterozygous germline mutations in *A2ML1* are associated with a disorder clinically related to Noonan syndrome** ***Helger G. Yntema***, *L.E.L.M. Vissers, M. Bonetti, J. Paardekooper Overman, W.M. Nillesen, S.G.M. Frints, J. de Ligt, G. Zampino, A. Justino, J.C. Machado, M. Schepens, H.G. Brunner, J.A. Veltman, P. Gros, J.L. Costa, M. Tartaglia, J. den Hertog, I. van der Burgt;* *Nijmegen, Netherlands*	**C22.3** **Implementation of a duty-to-recontact system in molecular and clinical genetics: perspectives from professionals and patients** ***Mirjam Plantinga***, *W. Lamers, A.V. Ranchor, M.A. Verkerk, E. Birnie, I.M. van Langen;* *Groningen, Netherlands*

PROGRAMME TUESDAY, JUNE 3-CONTINUED

Time	Gold Room	Space 3+4	Brown 3	Brown 1+2	Space 1
cont.	**C18 Large scale genomics** Chair: O. Zuffardi, H. Scheffer	**C19 Internal organs** Chair: M. Zollino, B. Melegh	**C20 Basic mechanisms in genetics** Chair: B. Franco, S. Lyonnet	**C21 Rasopathies and CDG** Chair: F. Sangiuolo, K. Writzl	**C22 Returning results: Ethical and legal issues,** joint with EMPAG Chair: F. Faravelli, M. Cornel
11.45	**C18.4** **Planar cell polarity gene mutations contribute to the etiology of human Neural Tube Defects** ***Patrizia De Marco****, E. Merello, G. Piatelli, A. Cama, Z. Kibar, V. Capra;* *Genova, Italy*	**C19.4** **Identification and functional characterization of ESR2, a new disease gene for 46,XY disorders of sex development (DSD)** ***Dorien Baetens****, T. Guran, L. De Cauwer, L. Looijenga, K. De Bosscher, M. Cools, E. De Baere;* *Ghent, Belgium*	**C20.4** **Pseudoautosomal region 1 length polymorphism in the human population** ***Martin A. Mensah****, M.S. Hestand, M.H.D. Larmuseau, M. Isrie, N. Vanderheyden, M. Declercq, E.L. Souche, J. Van Houdt, R. Stoeva, H. Van Esch, K. Devriendt, T. Voet, R. Decorte, P.N. Robinson, J.R. Vermeesch;* *Leuven, Belgium*	**C21.4** **A mutation in PAK3 with a dual molecular effect deregulates the RAS/MAPK pathway and drives an X-linked syndromic phenotype** ***Pamela Magini****, T. Pippucci, I. Tsai, S. Coppola, E. Stellacci, A. Bartoletti-Stella, D. Turchetti, C. Graziano, G. Cenacchi, I. Neri, D.M. Cordelli, V. Marchiani, R. Bergamaschi, G. Gasparre, G. Neri, L. Mazzanti, A. Patrizi, E. Franzoni, G. Romeo, D. Bordo, M. Tartaglia, N. Katsanis, M. Seri;* *Bologna, Italy*	**C22.4** **International views on sharing incidental findings from whole genome research** ***Anna Middleton****, M. Parker, C. Wright, H. Firth, E. Bragin, M. Hurles, O. DDD Project;* *Cambridge, United Kingdom*
12.00	**C18.5** **Clinical exome sequencing for cerebellar ataxia and spastic paraplegia reveals novel gene-disease associations and uncovers unanticipated rare disorders** ***Erik-jan Kamsteeg****, B.P. van de Warrenburg, S.T. de Bot, M.A.A.P. Willemsen, S. Vermeer, M.I. Schouten, R. Meijer, M. Pennings, C. Gilissen, H. Scheffer;* *Nijmegen, Netherlands*	**C19.5** ***LRP5*** **variants associated with development of polycystic kidney and liver disease** ***Wybrich R. Cnossen****, R.H.M. te Morsche, A. Hoischen, C. Gilissen, H. Venselaar, S. Mehdi, C. Bergmann, M. Losekoot, M.H. Breuning, D.J.M. Peters, J.A. Veltman, J.P.H. Drenth;* *Nijmegen, Netherlands*	**C20.5** **Comparative proteomic analysis of different fragile X syndrome cell lines** *S. Lanni, F. Palumbo, M. Goracci, G. Mancano, A. Vitali, V. Marzano, F. Iavarone, F. Vincenzoni, M. Castagnola, P. Chiurazzi,* ***Elisabetta Tabolacci****, G. Neri;* *Rome, Italy*	**C21.5** **Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukaemogenesis** ***Francesca Pantaleoni****, M. Jaiswal, E. Flex, S. Martinelli, M. Strullu, E.K. Fansa, A. Caye, A. De Luca, F. Lepri, L. Pannone, S. Paolacci, G. Bocchinfuso, C. Rossi, A. Farrotti, O. Fenneteau, B. Brethon, P. Cianci, E. Di Schiavi, A. Selicorni, B. Dallapiccola, I.C. Cirstea, L. Stella, M. Zenker, B.D. Gelb, H. Cavé, M.R. Ahmadian, M. Tartaglia;* *Roma, Italy*	**C22.5** **Newborn screenings and whole genome sequencing: the real need of a genuine public involvement** ***Marta Tomasi****, A. Santosuosso;* *Trento, Italy*
12.15	**C18.6** **WES detects disease causing SNVs and CNVs in Primary immunodeficiencies** ***Hanne S. Sorte****, A. Stray-Pedersen, P.S. Samarakoon, L. Forbes, T. Gambin, O.K. Rødningen, I.C. Hanson, L.M. Noroski, C. Davis, F. Seeborg, S.K. Nicholas, J.W. Caldwell, N.Y. Chokshi, D. Bayer, C.R. Beck, T.J. Vece, W. Wiszniewski, S.J. Penney, S.N. Jhangiani, D. Muzny, L.O. Mæhle, A. Patel, H.C. Erichsen, T.G. Abrahamsen, J. Buchner, G.E. Tjonnfjord, P. Aukrust, L.T. Osnes, M.A. Kulseth, D.E. Undlien, W.T. Shearer, B. Fevang, R.A. Gibbs, R. Lyle, J.S. Orange, J.R. Lupski;* *Oslo, Norway*	**C19.6** **Digenic model in Alport syndrome** ***Maria Antonietta Mencarelli****, M. van Geel, H. Storey, C. Fallerini, L. Dosa, M. Antonucci, F. Cetta, A. van den Wijngaard, S. Yau, F. Mari, M. Bruttini, F. Ariani, K. Dahan, B. Smeets, F. Flinter, A. Renieri;* *Siena, Italy*	**C20.6** **RNA-DNA Differences in Endoplasmic Reticulum Stress Response** ***Allison L. Richards****, S. Liu, Z. Zhu, V.G. Cheung;* *Ann Arbor, United States*	**C21.6** **A New Mouse Model for Costello Syndrome** ***Tania Sorg****, B. Arveiler, M. Birling, G. Bou About, M. Champy, F. Dupuy, I. Goncalves, M. Jagla, H. Jacobs, H. Meziane, G. Pavlovic, N. Philip, F. Radvanyi, R. Rossignol, M. Roux, S. Sigaudy, Y. Herault, D. Lacombe;* *Illkirch, France*	**C22.6** **Current Developments in the Regulation of Direct-to-Consumer Genetic Testing in Europe** ***Louiza M. Kalokairinou****, H.C. Howard, P. Borry;* *Leuven, Belgium*
12.30 - 13.30	*Lunch Break on Level 1 & 2*				

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

PROGRAMME TUESDAY, JUNE 3

Time	Gold Room
13.30 - 14.15	**Plenary Session PL4** **Mendel Lecture** Chair: H. Kääriäinen, B. Wirth
13.30	**PL4.1** **Gene Targeting into the 21st Century: Mouse Models of Human Diseases from Cancer to Neuropsychiatric Disorders** ***Mario Capecchi***; *Salt Lake City, United States*
14.15 - 15.45	**Plenary Session PL5** **ESHG Award and Closing Session** Chair: H. Kääriäinen, B. Wirth
14.15	**PL5.1** **Signatures of Mutational Processes in Human Cancer** *Sir* ***Michael Stratton***; *Hinxton, United Kingdom* *Laudation by Han Brunner*
15.00	**Awards Ceremony** **ESHG Honorary Award awarded to Jean Jacques Cassiman** *Laudation by Helena Kääriäinen* **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 **ESHG Poster Awards** **Closing**

At the end of the final Plenary Session, 3 Apple iPads mini will be drawn within the attendees having had their badges scanned at the entrance of the hall.

SCIENTIFIC

SCIENTIFIC PROGRAMME

WORKSHOPS

SATELLITES

CORPORATE SATELLITES

TECHNICAL INFORMATION

SCIENTIFIC INFORMATION

YOUNG INVESTIGATOR AWARD CANDIDATES

POSTER AWARD FINALISTS

PROGRAMME

Scottish Exhibition and Conference Centre

ESHG 2015 Glasgow

Scotland, United Kingdom, June 6-9

EUROPEAN SOCIETY OF HUMAN GENETICS

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See you in Glasgow

at the European Human Genetics Conference 2015

PROGRAMME WORKSHOPS-SATELLITES

Workshops

Detailed information on workshops can be found in the “ESHG Bulletin” in the conference bag.

Saturday, May 31, 2014, 10.30 - 12.30 hrs

WS01 Disease of the year: Rasopathies (G. Neri, M. Tartaglia)	Space 1

Sunday, June 1, 2014, 15.30 - 17.00 hrs

WS02 Dysmorphology 1* (D. Donnai, J. Clayton-Smith, S. Douzgou)	Gold Room
WS03 ENSEMBL* (A. Zadissa, E. Pritchard)	Space 3+4
WS04 Practical Bioinformatics: Whole exome sequence analysis (N. Robinson)	Brown 3
WS05 Quality assurance (E. Dequeker, M. Morris)	Brown 1+2
WS06 Community genetics - Clinical Genetic Services in 2025 (M. Cornel & U. Kristoffersson)	Space 1
WS07 Preimplantation genetic diagnosis (J. Vermeesch, E. Iwarsson)	Space 2

Monday, June 2, 2014, 15.30 - 17.00 hrs

WS08 Dysmorphology 2* (D. Donnai, J. Clayton-Smith, S. Douzgou)	Gold Room
WS09 Genome Browser UCSC* (R. Kuhn)	Space 3+4
WS10 Analysis, interpretation and reporting of array data* (N. de Leeuw & C. van Ravenswaaij-Arts)	Brown 3
WS11 Clinical Cancer Genetics Club (M. Genuardi & D. Stoppa-Lyonnet)	Brown 1+2
WS12 Preconception and prenatal screening (M. Macek Jr., T.H. Bui)	Space 1
WS13 Next Generation Sequencing* (J. Veltman)	Space 2

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

Official satellite meetings open to all participants

As per date of printing.

Saturday, May 31, 2014

SIGU High School Workshop
09.00 - 13.30 hrs Space 2

Sunday, June 1, 2014

Introduction to using Encode data for your analysis Workshop
11.30 - 13.00 hrs Space 3+4

Monday, June 2, 2014

Telegenetics in practice
12.15 - 13.15 hrs Suite 3

EUCID.net satellite Meeting
12.30 - 13.15 hrs Space 2

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.

GENERAL
SATURDAY
SUNDAY
MONDAY
TUESDAY
PROGRAMME
SC. INFO & YIA
EMPAG
INFORMATION

PROGRAMME CORPORATE SATELLITES

Myriad Genetics – Saturday, May 31, 2014, 12.15 – 13.45 hrs - Amber 7 & 8 – Level 2 Stand # 220

Multi-gene Panel Testing for Hereditary Cancer: Opportunities and Challenges for the Laboratory and Clinic

Lunch bags will be provided.

Chairman: *James Mackay, University College London - London, United Kingdom*

The challenge: Who to test? The opportunity: A multi-syndrome panel approach
Karen Copeland, Myriad Genetics GmbH - Zurich, Switzerland

The opportunity: NGS technology for multi-gene panels. The challenge: Ensuring optimal sensitivity and specificity in technical analysis and interpretation for clinical use
Karla Bowles, Myriad Genetics Laboratories - Salt Lake City, USA

The challenge: Integrating multi-gene/multi-syndrome genetic testing into clinical practice.
The opportunity: Improving cancer risk stratification for optimal patient management from three clinical perspectives:

Onco-geneticist perspective from referral practice in UK
James Mackay, University College London - London, United Kingdom

Oncologist perspective from referral and internal practice in Switzerland
Rudolf Morant, Brustzentrum ZeTuP AG - St. Gallen, Switzerland

Oncologist perspective from internal practice in US
Julia Smith, New York University - New York City, USA

Personalis – Saturday, May 31, 2014, 12.15 – 13.45 hrs - Suite 5 – Level 2 Mezzanine Stand # 486

The Personalis ACE Exome™: for Discovery Research and Clinical Diagnostics

Speaker: *Jonathan Beck, Personalis, Menlo Park, California, USA*

Personalis stands out as the provider of the most complete exome currently available, targeting more than 7,800 genes of highest biomedical relevance, and finishing these genes towards 100% coverage. Personalis provides customers with a world-class end-to-end service, from experimental design to sample receipt, through to phenotype-driven expert analysis and delivery of intuitive and actionable reports. All sample processing occurs within a state-of-the-art CLIA and CAP accredited environment that can provide as little as 8 week turn-around-times. The ACE Exome targets features that either perform poorly (incomplete or entirely absent from standard exomes), or are related to susceptibility, drug response, or structural variation in regions outside of the established exome (deeply intronic or intergenic). Our enhanced exome utilises optimized sample preparation and probe design significantly expanding the footprint of the exome. Annotation and interpretation of results makes use of unique, manually-curated content alongside public databases. From large cancer research studies using ACE exome with structural variant analysis, to individual pediatric congenital diagnostics with rapid turnaround, whatever your need, Personalis can provide the solution. Details regarding all aspects of our ACE Platform, service, new products and answers to your questions will be presented by the Personalis team in the workshop.

Affymetrix – Sunday, June 1, 2014, 11.45 – 13.15 hrs - Amber 5 & 6 – Level 2 Stand # 364

Find out what others are missing...

Researchers are demonstrating that a high density, whole genome approach is necessary to provide the most comprehensive results by identifying additional, clinically significant cytogenetic information not routinely seen with karyotyping and FISH.

Hear users' views on how Affymetrix® CytoScan® Cytogenetics Suite is enabling researchers to detect and 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.

Fiona Sara Togneri, BSc, West Midlands Regional Genetics Laboratory, Birmingham, UK
Affymetrix OncoScan™: MIP assay: a robust, reliable multiplex tool for detecting actionable aberrations in solid tumors

Beatrice Oneda, PhD, Institute of Medical Genetics, University of Zürich, Switzerland
Increased prevalence of pathogenic findings using high resolution chromosomal microarrays in foetuses

Massimo Carella, PhD, IRCCS Casa Sollievo della Sofferenza, San Giovanni Rotondo, Italy
High density SNP array as an investigational test for postnatal referrals: from large to single gene rearrangements

Lunch and refreshments will be provided. Spaces are limited; please arrive early to avoid disappointment. Visit us on stand 364.

PROGRAMME CORPORATE SATELLITES

BIOBASE – Sunday, June 1, 2014, 11.45 – 13.15 hrs - Suite 5 – Level 2 Mezzanine Stand # 446

NGS in Clinical Use and Diagnostics

Margherita Mutarelli, PhD, Bioinformatician, Telethon Institute of Genetics and Medicine
Maria Iascone, PhD, Lab Genetica Medica, AO Papa Giovanni XXIII, Bergamo
Frank Schacherer, CTO, BIOBASE GmbH

Advances in next generation sequencing have opened the door to using sequencing of genes, exomes and in some cases whole genomes as a powerful tool in the diagnostic process for patients suffering from rare inherited or de novo disease. While NGS sequencing provides great promise as a means of identifying the causal variants distinct to an individual's personal genome, the challenge of sifting through tens of thousands of variants to identify the few that are relevant to the disease state observed remains. Reductionist methods of filtering variants have become part of the standard process employed in sequence analysis. Such methods typically rely on the removal of common variants, synonymous variants and, in the case of cancers, the removal of germ-line variants. Attempts are then made to characterize the remaining variants by algorithmically predicted deleteriousness, known molecular function, etc. We will discuss the use of HGMD®, the Human Gene Mutation Database, in the clinical interpretation of targeted sequencing and exome analysis results and how it has been used in NGS variant analysis pipeline for interpreting patient data.

Life Technologies – Sunday, June 1, 2014, 11.45 – 13.15 hrs - Amber 7 & 8 – Level 2 Stands # 102 & 548

Application of Digital Next Generation Sequencing in Clinical Research

This satellite will focus on the analysis and interpretation of clinical research data in hematology, solid tumours and inherited disease. Leading scientists present their data using Ion Torrent™ Semiconductor Sequencing technology including:

- **Development and comparative analysis of an Ion AmpliSeq™ gene panel for AML analysis**
- **Targeted analysis of RNA gene fusions in lung tumour FFPE samples**
- **Identification and validation of diagnostic/prognostic markers and therapeutic targets in oncology**
- **Integrated analysis for chronic kidney disease**

Speakers include:

José Luis Costa, PhD, IPATIMUP, Portugal
Jacqueline Schoumans, PhD, Centre Hospitalier Universitaire Vaudois (CHUV), Lausanne, Switzerland
Amy Jayne McKnight, PhD, Faculty of Medicine, Health and Life Sciences, Queen's University Belfast

Cartagenia – Sunday, June 1, 2014, 15.30 – 17.00 hrs - Amber 7 & 8 – Level 2 Stand # 376

Clinical Applications of CNV and NGS Interpretation Pipelines

Are you implementing Next Generation Sequencing or arrays for rare disease, ID, prenatal diagnosis, oncology screening or somatic analysis?
Are you looking to grow your volumes and wondering about variant assessment pipelines and report automation?
Do you want to learn more on securely reaching out to referrers for electronic assay requisition and lab reporting?

If so, join Cartagenia's satellite symposium and drop by our booth (# 376) for a demonstration. Learn how clinical labs use the Bench platform to set up interpretation and reporting pipelines in various clinical domains, automate diagnostic workflows, improve turn-around times, and generate high-quality clinical variant assessments and lab reports.

Prof. Berivan Baskin PhD, FACMG, FCCMG, Department of Clinical Genetics, Uppsala University Hospital, Sweden
Implementation of targeted NGS in a clinical diagnostic lab

Prof. Avni Santani, PhD, Division of Genomic Diagnostics, Children's Hospital of Philadelphia, USA
Integration of NGS data into clinical diagnostics: interpretation and compliance for targeted panels and whole exome sequencing data

Steven Van Vooren, PhD, Cartagenia Inc, Cambridge MA, USA
Combined analysis of CNV and NGS data on a pediatric case: the Cartagenia Bench Lab platform

PROGRAMME CORPORATE SATELLITES

Multiplicom – Sunday, June 1, 2014, 15.30 – 17.00 hrs - Suite 5 – Level 2 Mezzanine Stand # 328

Advances of MASTR™ in Routine Clinical Diagnostics

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 seminar, experts in the clinical field will share their experience when implementing germline and somatic MASTR™ tests for clinical diagnosis of congenital disorders and cancer in combination with current massively parallel sequencers.

Program:
- **Implementing CFTR diagnostic testing**
- **Clinical routine diagnostic testing with EGFR, GIST and SOMATIC 1 MASTR™**
- **Validation of HCM and ADH MASTR™ testing for routine diagnostics**
- **New developments of MASTR™ integrated approach for personalized medicine**

PerkinElmer – Sunday, June 1, 2014, 15.30 – 17.00 hrs - Amber 5 & 6 – Level 2 Stand # 520

Innovative Solutions for Molecular Genetics

Please join us at our satellite meeting to hear more about PerkinElmer's latest innovations including our solutions for automated DNA/RNA isolation, and optimized KRAS/NRAS mutation detection for cost and time efficient RAS testing. We will also present PerkinElmer's integrated sample preparation workflow solutions utilizing chemagen Technology for nucleic acid extraction and talk about solutions to eliminate processing bottlenecks presented by today's sequencing technologies.

The Agenda

Research Applications
15:30 Nucleic Acid Isolation – Combined Best-In-Class Technologies
16:00 Integrated Sample Prep and QC Solutions for Accelerating Translational Genomics

IVD Applications
16:30 Optimized KRAS/NRAS Mutation Detection

Visit us also at the stand #520 to learn more about our complete product offering!

AstraZeneca – Sunday, June 1, 2014, 19.00 – 20.30 hrs - Amber 5 & 6 – Level 2 Not exhibiting

BRCA to the Future: Towards Best Testing Practice in the Era of Personalised Healthcare

Chair: *Ettore Capoluongo, Laboratory of Clinical Molecular and Personalised Diagnostics, Department of Diagnostics and Laboratory Medicine, Teaching and Research Hospital 'A. Gemelli', Rome, Italy*

The biological effects and clinical implications of BRCA mutations: where do we go from here?
Dominique Stoppa-Lyonnet, Curie Institute and University of Paris Descartes, Paris, France

New challenges for BRCA testing: a view from the diagnostic laboratory
Andrew Wallace, Genomic Diagnostics Laboratory, Manchester Centre for Genomic Medicine, St Mary's Hospital, Manchester, UK

Options for BRCA testing models: best practices and multidisciplinary collaboration
Nicoline Hoogerbrugge, Radboud University Medical Center, Nijmegen, The Netherlands

Refreshments will be provided.

AstraZeneca, Alderley Park, Macclesfield, Cheshire, UK. April 2014 – ATLAS 155,514.011 – Expiration date 10th June 2014.

Illumina – Sunday, June 1, 2014, 19.00 – 20.30 hrs - Amber 7 & 8 – Level 2 Stand # 458

Illumina Workshop

Please join us as we highlight groundbreaking developments in research from around the world and review the latest advancements in our portfolio of genomic solutions for Cancer, Genetic & Infectious Disease, and Reproductive Health.

Complimentary wine and cheese will be served.

No pre-registration required, however space is limited, so please arrive early.

PROGRAMME CORPORATE SATELLITES

Natera – Sunday, June 1, 2014, 19.00 – 20.30 hrs - Suite 5 – Level 2 Mezzanine Stand # 358

PanoramaTM Goes Micro

Please join us for canapes and cocktails where we will discuss the SNP-based Panorama™ NIPT that now screens for microdeletions. We will review the published clinical trial results that demonstrate Panorama's high accuracy for trisomy 21, trisomy 18, trisomy 13, monosomy X and triploidy.

We will also discuss the microdeletion included in the Panorama™ screen, which are:
- common and severe
- of equal risk across all maternal ages
- often undiagnosed

Learn how these proven advances in NIPT screening can enhance the level of prenatal care offered to your patients.

Elizabeth Valenti, M.S., CGC, Natera, San Carlos, CA, USA
Powered by SNPs

Melissa Stosic, M.S., CGC, Natera, San Carlos, CA, USA
Panorama™ is going Micro

Megan Hall, Ph.D, Natera, San Carlos, CA, USA
Show Me the Data!

Abbott Molecular – Monday, June 2, 2014, 11.45 – 13.15 hrs - Amber 7 & 8 – Level 2 Stand # 528

Recent Advances in Detection of Expanded FMR1 Alleles

Talk 1 **Introduction to Abbott Molecular's "PCR Tools for FMR1"**
Paul Kyle, Abbott Molecular, Wavre, Belgium

Talk 2 **The Changing Paradigm of Testing for Expanded Alleles of FMR1**
Dr Monica Basehore, Greenwood Genetic Centre, Greenwood, SC, USA

Talk 3 **A Pilot Study for Prenatal and Preconceptional Detection of Expanded FMR1 Alleles in the Balearic Islands**
Dr Damain Heine-Suñer, Hospital Son Espaces, Mallorca, Spain

Agilent Technologies – Monday, June 2, 2014, 11.45 – 13.15 hrs - Amber 5 & 6 – Level 2 Stand # 420

Advances in Clinical Research Applications Using Target Capture for Next- Generation Sequencing and Chromosomal Microarray Analysis

Screening for Oral Precancer by Next-Gen Sequencing of Brush Biopsies
Prof. Ruud H Brakenhoff, Tumor Biology Section, VU University Medical Center, Amsterdam, The Netherlands
Oral squamous cell carcinomas arise in preneoplastic mucosal fields characterized by tumor-associated genetic changes. Here we employed targeted Next Gen sequencing in cytological samples. Challenges discussed include the low amount of DNA isolated, mutational noise and costs.

Complementing Next-Generation Sequencing with Exon-centric Microarray for a Comprehensive Analysis of Autism Patients: The Greenwood Genetic Center Experience
Alka Chaubey, PhD, FACMG, Cytogenetics Laboratory, Greenwood Genetic Center, Greenwood, USA
Traditional screening methods have not significantly impacted clinical yield due to high genetic heterogeneity associated with autism spectrum disorder. We developed a targeted NGS Panel and a custom microarray, demonstrating the nature of both technologies to provide a more comprehensive genetic analysis of autism.

Effective Detection of Genetic Disease by Computational Phenotype Analysis of the Disease-Associated Genome
Dr. Tomasz Zemojtel, Institute for Medical and Human Genetics, Charité-Universitätsmedizin Berlin, Germany
We established a combined approach that targets variants in 2755 Mendelian-disease- genes and computational method that determines on pathogenicity and semantic similarity of phenotype profiles described by Human Phenotype Ontology. Thus, this approach provides the means for quick and effective method for detection.

PROGRAMME CORPORATE SATELLITES

LGC – Monday, June 2, 2014, 11.45 – 13.15 hrs - Suite 5 – Level 2 Mezzanine Stand # 468

Functional Validation of Genetic Variation in Population Genomics

Finding and characterizing type 2 diabetes genes by genomic and physiological studies
Niels Grarup, Assistant Professor, MD, PhD - The Novo Nordisk Foundation Center for Basic Metabolic Research, Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark

Sample size in genetic research of blood pressure: How big is big enough?
Dr. Folkert W. Asselbergs, Consultant cardiologist, UMC Utrecht, The Netherlands, University College London, UK, and Durrer Center for Cardiogenetic Research, Netherlands Heart Institute, The Netherlands

Genetic variation in host pathway for triage of women with Chlamydia trachomatis **based subfertility: translation into public health**
Prof. Dr. Servaas A. Morré, Associate Professor, Head of the Institute of Public Health Genomics, Department Genetics of Cell Biology, University of Maastricht, The Netherlands

Life Technologies – Monday, June 2, 2014, 15.30 – 17.00 hrs - Amber 7 & 8 – Level 2 Stands # 102 & 548

Transforming Clinical Research

Join our Satellite: **"Chip-based Digital PCR Applications and New Frontiers in Multiplexing qPCR"**

Listen to leading scientists sharing their research using next generation digital PCR and qPCR technologies:

- *Dr. Francisco Cifuentes, Life Sciences Solutions, Thermo Fisher Scientific*
  Transforming clinical research with high-performance digital PCR on the QuantStudio™ 3D Digital PCR System

- *Mme N. Vasseur, Faculté de Médecine et Pharmacie, Université de Rouen, France*
  Plasma cell-free DNA and fraction of circulating KRAS mutation as prognostic biomarkers in patients with metastatic colorectal cancer

- *TBD, Life Sciences Solutions, Thermo Fisher Scientific*
  Empower clinical research with new full-spectrum TaqMan® Multiplex PCR solution

QIAGEN – Monday, June 2, 2014, 15.30 – 17.00 hrs - Suite 5 – Level 2 Mezzanine Stand # 350

How to Ensure Valuable Insights and Results from Sample Preparation to NGS Data Analysis

How to ensure valuable insights and results from sample preparation to NGS data analysis
Jason T. Gammack, VP, Advanced Genomics Commercial Operations, QIAGEN Redwood City, CA, USA

Fast and accurate identification of disease related variants in a Glioblastoma cohort
Jos de Graaf, PhD, Head Next Generation Sequencing Unit, Translational Oncology (TRON), Mainz, Germany

TBC CLC Platform in routine genetic testing
Dr. Ina Vogl, Scientist, Center of Human Genetics and Laboratory Diagnostics (AHC), Munich, Germany

Novel variants in PIGQ, PGAP3 and PIGY further implicate the GPI pathway in the pathogenesis of neurodevelopmental abnormalities
Dr. Alistair Pagnamenta, The Wellcome Trust Center for Human Genetics, University of Oxford, UK

NB. Seating is limited, so first come, first served. Refreshments will be provided after the symposium. We look forward to meeting you.

PROGRAMME CORPORATE SATELLITES

BGI – Monday, June 2, 2014, 19.00 – 20.30 hrs - Amber 5 & 6 – Level 2 Stand # 536

Applications of Next-Gen Sequencing in Human Disease Research and Clinical Diagnostics

In the emerging era of personalized medicine, major pharmaceutical companies and leading research institutes increasingly rely on next-gen sequencing (NGS) technologies and analytical tools to facilitate human disease research and develop clinical diagnostics solutions.

Partnering with a team of world-renowned genomics leaders, this workshop is led by world's leading genomics institution BGI to introduce the applications of its state-of-the-art trans-omics technologies and bioinformatics tools in disease research, pre-natal testing, and reproductive health.

Chair: *Joyce Peng, Ph.D., Marketing Director, BGI Tech Americas & Europe*

Rick Tearle, Ph.D., Senior Field Applications Scientist, Complete Genomics, CA, USA
Challenges in Whole Human Genome Sequencing

Francesco Lescai, Ph.D., Associate Professor of Aarhus University, Denmark
Rare and *De-novo* Variation in Psychiatric Disorders in the Faroe Islands

Tze Kin Lau, Ph.D., Chairman of The Chinese Fetal Medicine Foundation, China
Application of Next Generation Sequencing in Prenatal Testing for Fetal Chromosomal Abnormality

Yutao Du, Ph.D., VP of BGI Health, Director of Clone and Genetic Engineering Platform, BGI, China
New Opportunity of Next Generation Sequencing in Human Assisted Reproduction.

Fluidigm – Monday, June 2, 2014, 19.00 – 20.30 hrs - Amber 7 & 8 – Level 2 Stand # 336

Programme to be announced.

Roche Sequencing – Monday, June 2, 2014, 19.00 – 20.30 hrs - Suite 5 – Level 2 Mezzanine Stand # 368

Clinical Research Applications of Exome Capture and Custom Targets for High-Throughput Sequencing

Please join us for our exciting Next-Generation Sequencing (NGS) workshop, where

Dr. Sabrina Giglio, from the University of Florence and Dr. Gema Garcia, from Montpellier University Hospital (INSERM) 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. Giglio will discuss her work applying focused sequencing to the study of a variety of known clinical genotypes for pediatric Glioblastoma, and the identification of novel mutations associated with early-onset diabetes and Multiforme Nephrotic syndrome. The work in her laboratory is pioneering the use of exome sequencing to screen for these variants and the characterization of candidate genes underlying these diseases to potentially improve downstream targeted therapeutic strategies.

Dr. Garcia will address the specifics of Usher syndrome, with targeted sequencing used as the primary method for discovery and screening of genomic DNA variants. Presenting recent research, she will show the utility of targeted sequencing in characterizing the complex structural variations involved in USH.

PROGRAMME POSTER TOPICS-TECHNICAL INFORMATION

Poster Topics

P01. Reproductive Genetics/Prenatal Genetics	P01.002-128
P02. Sensory disorders (eye, ear, pain)	P02.01-49
P03. Internal organs & endocrinology (lung, kidney, liver, gastrointestinal)	P03.01-49
P04. Skeletal, connective tissue, ectodermal and skin disorders	P04.01-73
P05. Cardiovascular disorders	P05.01-67
P06. Metabolic and mitochondrial disorders	P06.01-61
P07. Immunology and hematopoietic system	P07.01-43
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P10. Neuromuscular disorders	P10.01-42
P11. Multiple Malformation/anomalies syndromes	P11.001-154
P12. Cancer genetics	P12.001-143
P13. Basic mechanisms in molecular and cytogenetics	P13.01-49
P14. New diagnostic approaches, technical aspects & quality control	P14.01-97
P15. Personalized/Predictive Medicine and Pharmacogenomics	P15.01.-39
P16. Omics/Bioinformatics/Epigenetics	P16.01-78
P17. Genetic epidemiology/Population genetics/Statistical methodology and evolutionary genetics	P17.01-95
P18. Genetic counselling/Education/public services	P18.01-48
EP. EMPAG Posters	EP01-52

Technical Information for Presenters of Posters

Posters will be on **display** from	Saturday, May 31 (08:30 hrs) to Monday, June 2 (17:30 hrs)
Poster mounting will be possible on:	Saturday, May 31, from 08:30 hrs onwards
Removal will be mandatory on:	Monday, June 2, from 13.30 hrs - 17.30 hrs (strict).

Access after this time is not possible! Please note that posters not removed until then 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 in the Poster Listing available at the poster help desk or you can ask for assistance at the poster help desk on the balcony (Level 1) or at the two information points 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 1 for posters with poster board numbers ending with an “S”
(e.g. P04.01-S, P04.03-S)
or
• 10.30 and 11.30 hrs on Monday, June 2 for posters with poster board numbers ending with an “M”
(e.g. P07.02-M, P07.04-M)

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- and overhead projection (no slides).
- It is essential that you load and view your presentation in the media check/preview centre (**Level 2**) preferably in the morning of the day your talk is scheduled, but 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 preview centre 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)

PROGRAMME ESHG AWARD - YOUNG INVESTIGATORS

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. The laureate receives a cheque of EUR 1.500.- to cover the expenses of participating in the meeting.

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**

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 3, 2014 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) will be awarded to the best presentation in translational genetic research/therapy of genetic diseases.

All winners will receive prize money in the amount of EUR 500 and a complementary ESHG online membership for 1 year.

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

**Download the new
ESHG 2014 Conference App
for iOS and Android
devices from the
iTunes App Store or
Google Play Store**

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 2014?

Reza Asadollahi
Schlieren-Zurich, Switzerland

Talk: C03.4 The significance of small copy number variants in neuro-developmental disorders
Session: C03 Intellectual disability
Date: Saturday, May 31, 2014, 18:30 hrs.

Q1: Yazd, Iran
Q2: MD-PhD Fellow
Q3: Medical genetics is a remarkable field. This is due to the close interaction of medicine and science for molecular characterization of genetic disorders in order to help individual patients.
Q4: In a large cohort of patients with neuro-developmental disorders of unknown cause, we investigated the diagnostic relevance of genome-wide rare CNVs <500 kb and highlighted their inherent potential for discovery of new conditions.

Dorien Baetens
Ghent, Belgium

Talk: C19.4 Identification and functional characterization of ESR2, a new disease gene for 46,XY disorders of sex development (DSD).
Session: C19 Internal organs
Date: Tuesday, June 3, 2014, 11:00 hrs.

Q1: 10/15/1989, Dendermonde, Belgium
Q2: I am a PhD student at the Center of Medical Genetics in Ghent University Hospital.
Q3: My interest for genetics started in high school. I found it intruiging that one single change in our DNA could have such a great impact. At university, my knowledge in the field grew and so did the fascination. I learned that genetics are a rapidly changing field with new techniques and new mechanisms. Besides that, it is very motivating that genetic research is so closely linked to the clinical setting. Genetic results can improve patient care and life quality.
Q4: We identified a possible new disease genes for a rare disorder called Disorders of Sex Development (DSD). Despite the low prevelance of these disorders, it is important to identify the underlying molecular cause. Studying abnormal sexual development, can help us to understand pathways that are important for normal development and they can improve our knowledge about more frequent reproductive disorders such as premature ovarian failure. Identification of the molecular cause of DSD can also lead to a refined diagnose, a more accurate prognosis on fertility and improved patient management.

Emma Baple
Exeter, United Kingdom

Talk: C15.2 Mutations in KPTN Cause Macrocephaly, Neurodevelopmental Delay, and Seizures
Session: C15 Novel genes in neurogenetic disorders
Date: Monday, June 2, 2014, 13:30 hrs.

Q1: 5/3/1978, Epsom, United Kingdom
Q2: Specialist trainee in Clinical Genetics and Honorary Clinical Research Fellow, University of Exeter
Q3: Genetics is probably the most rapidly advancing scientific field and thus one of the most exciting to be a part of. I have always aspired to a career as a clinical academic within genetics, helping to maximize the clinical benefits of cutting edge genetic research by bridging the gap between basic science and mainstream medicine.
Q4: The research findings that I will present identify KPTN as a molecule fundamental to normal human brain growth and development. This study is part of a wider community genetics project based within the Ohio Amish community. It illustrates well the significant translational benefits of such work to both the Amish community and the wider population.

Fitnat Basmanav
Bonn, Germany

Talk: C21.1 Mutations in POGLUT1, encoding protein O-glucosyltransferase 1, cause autosomal dominant Dowling-Degos disease
Session: C21 Rasopathies and CDG
Date: Tuesday, June 3, 2014, 11:00 hrs.

Q1: 6/2/1982, Ankara, Turkey
Q2: PhD student at the Institute of Human Genetics, University of Bonn, Germany
Q3: I decided to be a researcher in this field because I have always been fascinated by how much genetics can explain about what we are as the human kind as well as who we are as unique individuals each…
Q4: It is very exciting that we can explain about one third of the cases in our large cohort of Dowling-Degos disease (DDD) patients by the mutations we identified in this novel gene and that we generated information on the functional outcomes of some of these mutations. The gene we identified is from the Notch pathway and the involvement of this pathway in DDD is very intriguing and creates new opportunities of research for us. I am also delighted that we were able to define a gene-phenotype correlation in this disease for the first time which will be very useful in genetic screening and diagnostic testing.

Annelien Bredenoord
Utrecht, Netherlands

Talk: C22.2 Defending the child's right to an open future concerning genetic information.
Session: C22 Returning results: Ethical and legal issues (joint ESHG/EMPAG session)
Date: Tuesday, June 3, 2014, 11:00 hrs.
Q1: 8/1/1979, Utrecht, The Netherlands
Q2: Associate Professor of Medical Ethics
Q3: I examine the ethical issues in novel biomedical technology. I am particularly fascinated by the rapid developments in regenerative medicine and stem cells, genetics/genomics and biobanking and the associated ethical and societal challenges: how to translate biomedical innovations from basic research into clinical care and society in an ethically sound way? I strongly believe that ethical parallel research can contribute to sustainable, ethically sound innovation in those important but often also controversial fields.
Q4: There has been a discussion regarding the ethical acceptability of genetic testing of children for years, resulting in a firm majority view that minors should only be tested for early onset disorders where treatment or preventive options exist. The emergence of next-generation sequencing seems to challenge this consensus. This may have serious consequences for future autonomy rights of children, their so-called 'right to an open future'. I would like to use this presentation to discuss with the audience whether this is the direction we should aim at.

Nathalie Brison
Leuven, Belgium

Talk: C01.1 Clinical implementation of non-invasive prenatal aneuploidy detection
Session: C01 Prenatal testing
Date: Saturday, May 31, 2014, 18:30 hrs.
Q1: 7/6/1983, Kortrijk, Belgium
Q2: I'm a postdoc in the Clinical Cytogenetics lab in the Centre for Human Genetics, Leuven (Belgium).
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 methods for non-invasive detection of fetal chromosomal aneuploidies. Non-invasive prenatal testing (NIPT) thus avoids miscarriages due to invasive sampling of fetal material. We developed and validated an innovative, fast, cost efficient workflow and high throughput analysis pipeline for NIPT. This approach resulted in 100% specificity and sensitivity for trisomy 21 and 18 detection and has been clinically implemented and accredited. Moreover, optimization of the initial analysis pipeline seems to create opportunities to detect other chromosomal abnormalities in addition to the traditional trisomies...

Keren Carss
Hinxton, Cambridgeshire, United Kingdom

Talk: C03.2 De Novo loss of function mutations in SETD5, a novel methyltransferase gene within the 3p25 microdeletion syndrome critical region, cause intellectual disability
Session: C03 Intellectual disability
Date: Saturday, May 31, 2014, 18:30 hrs.
Q1: 2/13/1985, Norwich, United Kingdom
Q2: PhD student (4th year), at the
Wellcome Trust Sanger Institute, Cambridge, UK.
Q3: I am interested in identifying variants that cause rare genetic diseases. This has allowed me to study the biology underlying a range of phenotypes, using both 'wet lab' and computational approaches. I enjoy this diversity, and am motivated by the potential application of my work to patients, who are often desperate to know the cause of their disease.
Q4: A high proportion of people with intellectual disability (ID) do not have likely causative variants in genes known to be involved in ID. Therefore, with each new gene discovered, the chances of a patient receiving a diagnosis increases. In this study, we find that loss of function mutations in <i>SETD5</i> are a relatively common cause of ID. Additionally, our data suggest that perturbation of SETD5 function is likely to account for many of the features of 3p25 microdeletion syndrome.

Wybrich Cnossen
Nijmegen, Netherlands

Talk: C19.5 LRP5 variants associated with development of polycystic kidney and liver disease
Session: C19 Internal organs
Date: Tuesday, June 3, 2014, 11:00 hrs.
Q1: 5/26/1985
Q2: I am working as PhD student with a focus on polycystic liver disease. My project is a collaboration between the laboratory of the Department Gastroenterology and Hepatology of professor Joost Drenth, and the Genomic Disorders Group of professor Joris Veltman at the Radboud university medical center. We apply different strategies to identify novel genes associated with cystogenesis and perform functional analyses.
Q3: Rare liver disorders have my special interest. Polycystic liver disease is one of those. Many genetic factors related to (progressive) development of multiple cysts are yet unknown. Secondly, I really enjoy performing research in collaboration with colleagues from different disciplines such as technicians, biologists and bioinformatics. Sharing knowledge brings us together to the next level.
Q4: Isolated polycystic liver disease (PCLD) and autosomal dominant polycystic liver disease (ADPKD) are the 2 major polycystic liver diseases. The genetic cause is unexplained in the ma-

PROGRAMME YOUNG INVESTIGATOR AWARD CANDIDATES

jority (~80%) of PCLD patients. Recently, we identified 4 unique variants on the LRP5 gene in PCLD families by exome sequencing (PNAS 2014 March 24). Almost all ADPKD patients harbor a PKD1 or PKD2 mutation, but some cases are still unlinked. Here, we present unique and rare LRP5 variants associated with ADPKD.

Francesco Cucco
Pisa, Italy

Talk: C08.1 Smc1a cohesin gene mutations in colorectal precancerous lesions
Session: C08 Cancer genetics
Date: Sunday, June 1, 2014, 13:30 hrs.
Q1: 7/9/1984, San Benedetto del Tronto (AP), Italy
Q2: PhD student
Q3: During my undergraduated studies I have always been interested in genetics. I attended at several curricular and extra-curricular genetics courses and seminars. In particular I have always been fascinated by how DNA variants can lead to genetic disorders.
Q4: The implication of cohesin and its alterations in early steps of colorectal cancer. We also defined the role of cohesin mutations in the tumorigenesis.

Elisa De Franco
Exeter, United Kingdom

Talk: C17.5 Genetic testing leads clinical care in neonatal diabetes: a new paradigm
Session: C17 Metabolic and mitochondrial disorders
Date: Monday, June 2, 2014, 13:30 hrs.
Q1: 12/24/1986, Bra, Italy
Q2: Research Associate
Q3: I chose a career in genetics as I am convinced that the DNA holds most of the answers to unsolved questions in human biology and I strongly believe genetics will soon make an impact on everybody's lives.
Q4: We studied the impact of genetic testing in the world largest cohort of neonatal diabetes patients (n=1020). Our results show that next-generation sequencing has changed clinical practice: now the genetic result guides clinicians' choices on patients' treatment and clinical management.

Pasquelena De Nittis
San Giovanni Rotondo, Italy

Talk: C16.6 In silico and functional characterization of KMT2D/MLL2 missense mutations as causative in Kabuki syndrome
Session: C16 Genes and development 2
Date: Monday, June 2, 2014, 13:30 hrs.
Q1: 7/10/1988, San Giovanni Rotondo (FG), Italy
Q2: I am currently a postgraduate trainees at the Medical Genetics Unit of "Casa Sollievo della Sofferenza" Hospital (FG, Italy).
Q3: I think that it is so fascinating to have curiosity in the genetic basis of disease, being genetics the basis of biological systems and to link interests in molecular mechanisms with clinical implications. The up-to-date knowledges of cellular and developmental systems raises the possibility to study pathogenesis of diverse genetic disease, as well as to generate cells of different lineages for future personalized therapies, using patient-specific input cells. I think that it is motivating for my future career.
Q4: Kabuki syndrome is a rare syndrome, caused mainly by mutations in KMT2D and UTX genes. Among the KMT2D mutations we identified some missense variants. In this work we propose to estimate the real deleterious effect of KMT2D missense variants by an analysis with bioinformatic tools and functional assays, being the final effect of mutation in causative gene, a main issue in diagnostic counseling.

Patrick Deelen
Groningen, Netherlands

Talk: C06.1 Resolving variants of unknown significance through reanalysis of 4,978 public RNA-seq samples
Session: C06 Functional and computational genomics
Date: Saturday, May 31, 2014, 18:30 hrs.
Q1: 5/30/1986, Rotterdam, The Netherlands
Q2: I'm a bioinformatics PhD student at the Genomics Coordination Center of the Genetics Department at the University Medical Center Groningen, the Netherlands.
Q3: The field of genetics allows me to put my curiosity, interest in genetics and statistical and programming skills to good use. I love tackling complex problems and dealing with the increasing amounts of datasets. I hope that the knowledge I acquire will be useful for gaining insight in the understanding of the genetic basis of diseases.
Q4: For many mutations it remains unclear how they cause disease. I developed an algorithm that can integrate and extract genotypes from >1,000 public RNA-seq experiments, which enables allele specific expression analysis. I found that many rare mutations affect gene expression levels, illustrating the power of mining public RNA-seq data.

Giuseppina Di Fruscio
Naples, Italy

Talk: C07.1 LysoPlex: an efficient strategy to study the role of lysosomal-autophagic-endocytic pathway
Session: C07 Implementation of NGS in diagnostics
Date: Sunday, June 1, 2014, 13:30 hrs.
Q1: 6/13/1987, Naples, Italy
Q2: I am a PhD student at Professor Nigro's laboratory and I am currently working on the molecular characterization of lysosomal diseases and disorders related to autophagy. In particular, I have contributed to the development of LysoPlex, a novel targeted NGS tool investigating more than 800 genes selected by computational approaches and involved in the lysosomal-autophagic pathway.
Q3: I chose to attend the biotechnology faculty driven by a strong curiosity about the mechanisms causing genetic diseases and, in particular, responsible for the relationship between the genotype and phenotype. I find it very interesting to observe how any little change in the DNA code can lead to a pathology in an organism.
Q4: LysoPlex is the first platform able to investigate a high number of genes predicted to be related to the lysosomal-autophagic pathway. Its widespread use could allow us to uncover the role of these cellular functions in health and disease.

Anthony Drecourt
Paris, France

Talk: C15.3 REPS1 is a novel gene of Neurodegeneration with Brain Iron Accumulation
Session: C15 Novel genes in neurogenetic disorders
Date: Monday, June 2, 2014, 13:30 hrs.
Q1: 5/18/1987, Conflans sainte Honorine, France
Q2: I'm Ph.D student at the IMAGINE Institut, Paris, France
Q3: I like to study life mecanisms and especially the link between genetics and desease. But the most important is that my work can help people.
Q4: Using Exome sequencing od DNA from patients with NBIA (Neurodegeneration with Brain Iron Accumilation)we identified mutations in a new gene linked to this disease. These patients are caracterised by iron accumulation in the brain and in skin fibroblasts. Currently, we are trying to dissect the molecular mechanism behind iron accumulation in the patients. By studying this we hope to identify a drug target that will allow us to limit the progression of the disease.

Daniel Gaston
Halifax, Canada

Talk: C08.3 Germline mutations in MAP3K6 predispose to gastric cancer
Session: C08 Cancer genetics
Date: Sunday, June 1, 2014, 13:30 hrs.

Xavier Gerard
Paris, France

Talk: C12.4 AON intravitreal injections to manipulate splicing in retinal cells
Session: C12 Sensory disorders
Date: Sunday, June 1, 2014, 13:30 hrs.
Q1: 1/1/1984, Valence, France
Q2: Postdoctoral fellowship
Q3: To develop therapeutic approaches
Q4: The manipulation of mRNA splicing in retinal cells after an antisense oligonucleotides intravitreal injection.

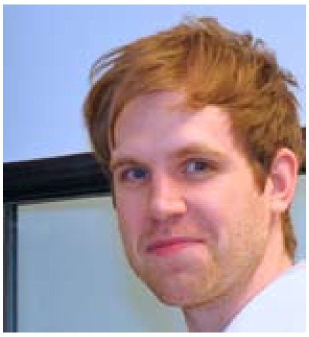
Christian Gilissen
Nijmegen, Netherlands

Talk: PL2.6 Genome sequencing identifies major causes of severe intellectual disability
Session: PL2 What's new? Highlights Session
Date: Saturday, May 31, 2014, 4:30:00 PM hrs.
Q1: 4/13/1980, Geleen, The Netherlands
Q2: I'm a postdocteral researcher in bioinformatics
Q3: My first experience with genetics was by accident after losing a coin-toss for an internship assignment with a colleague. However, after my first experiences I got very excited about my field because there were so many new fundamental things to discover, while at the same time my work really affected people's lives and helped patients.
Q4: This research presents the first real application of Whole Genome Sequencing (WGS) in the clinic. We find that de novo mutations are the major cause of severe intellectual disability and that by using WGS we can identify all different types of genomic variation in a single test thereby providing a diagnosis for the majority of patients.

PROGRAMME YOUNG INVESTIGATOR AWARD CANDIDATES

Giorgia Girotto
Trieste, Italy

Talk: C12.2 New Hereditary hearing loss (HHL) genes/mutations identified by High throughput sequencing and genotyping in the Italian and Qatari populations.
Session: C12 Sensory disorders
Date: Sunday, June 1, 2014, 13:30 hrs.
Q1: Venice, Italy
Q2: Postdoctoral Research Fellow, PhD, University of Trieste/ IRCCS Burlo Garofolo, Italy
Q3: Since I started my thesis in Genetics, I had no idea where the road would take me. Then, I have been intrigued by the complex mechanism of human body that are driven by genetics rules
Q4: Hearing loss is the most frequent sensory defect affecting humans and according to the World Health Organization, worldwide, more than 250 million of people have disabling hearing loss. Considering the large number of people affected, the limited potential of available therapies and the vast genetic heterogeneity, there is an unmet need to discover new genes/alleles involved and to develop new preventive strategies and therapeutic approaches. Thanks to our multistep strategy and the use of high-throughput technologies, we were able to characterize at molecular level several families affected by Hereditary Hearing Loss identifying also new genes. This means a substantial increase in our understanding of the physiology of hearing and will be the pre-requisite for additional functional studies as well as for putative gene-specific therapeutic approaches. Furthermore, the definition of accurate molecular epidemiology data, is an essential step towards the development of diagnostic algorithms and protocols



Emil Gustavsson
Vancouver, Canada

Talk: C09.2 Exome sequencing of familial parkinsonism in Scandinavia
Session: C09 Common neurological disease
Date: Sunday, June 1, 2014, 13:30 hrs.
Q1: 11/8/1981, Stockholm, Sweden
Q2: PhD student at the Centre for Applied Neurogenetics, University of British Columbia and Norwegian University of Science and Technology
Q3: Studying neuroscience of disease made me realize that we work with the consequences of disease whereas genetics might answer how and why the pathogenesis may arise and propagate within families.
Q4: Working within a homogeneous population where environmental factors and clinical history has been documented over a long period of time makes it powerful to investigate the genetic contribution of disease. Using exome sequencing within families in close relation to the neurologists is a fast and powerful method to understand the molecular ethiology of parkinsonism in these families.

Michael Holmes
Philadelphia, United States

Talk: C04.6 Causal relationship of body mass index with cardiometabolic traits and events: a Mendelian randomization analysis
Session: C04 Cardiovascular disorders
Date: Saturday, May 31, 2014, 18:30 hrs.
Q1: 6/23/1978, Glasgow, United Kingdom
Q2: Assistant Professor, Department of Surgery, Perelman School of Medicine, University of Pennsylvania, USA
Q3: My interest lies in exploiting genomic data to make inferences about causal mechanisms in disease aetiology
Q4: I will present findings from a Mendelian randomization analysis of body mass index on cardiometabolic traits and events. The findings show that body mass index has wide-ranging causal effects on multiple traits that are harmful to cardiovascular health. Since body mass index is a modifiable trait, these findings highlight the importance of weight management for optimizing cardiovascular health at the population level.

Laura Huckins
Hinxton, United Kingdom

Talk: C11.2 Polygenic risk score analysis shows shared genetic aetiology between AN and five other psychiatric disorders
Session: C11 Statistical genetics
Date: Sunday, June 1, 2014, 13:30 hrs.
Q1: 3/21/1989, Oxford, UK
Q2: PhD Student, Wellcome Trust Sanger Institute
Q3: I chose a career in genetics because I wanted to understand the role played by genetics in our thoughts and emotions. My PhD focuses on the genetics and functional mechanisms of eating disorders, and involves not only statistical genetics, and teasing out polygenic or epigenetic aetilogy, but also the study of mouse behaviour and the functional effect of putative AN genes. I am always fascinated by the interplay of genetics and societal or environmental factors.
Q4: Anorexia Nervosa (AN) has the highest mortality rate of any psychiatric disorder, yet the disorder is poorly understood, and no effective treatment exists. This research will be the first time a polygenic aetiology has been shown for AN, and the first evidence of cross-disorder genetic architecture between AN and other psychiatric disorders. This research will be a first step to explaining the biological mechanisms underlying AN.

Louiza Kalokairinou
Leuven, Belgium

Talk: C22.6 Current Developments in the Regulation of Direct-to-Consumer Genetic Testing in Europe
Session: C22 Returning results: Ethical and legal issues (joint ESHG/EMPAG session)
Date: , 11:00 hrs.
Q1: 8/17/1987, Heraklion, Greece
Q2: I am a PhD researcher at KU leuven in Belgium. My project focuses on legal, ethical and social aspects of direct-to-consumer genetic testing.
Q3: Genetics is a fast growing field which unravels great possibilities for prevention and treatment of diseases, but at the same time may present numerous ethical and legal implications,as well as policy challenges. I am particularly interested in exploring how innovation and the development of personalized medicine may be promoted without compromising public health and fundamental rights.
Q4: My research aims to present how the proposed Regulation on in vitro diagnostic medical devices may affect direct-to-consumer genetic testing in Europe. The proposed Regulation, if eventually adopted, will impact significantly, among others, the pre-market assessment of genetic tests and render illegal their provision and marketing directly to consumers. These amendments raise questions regarding the appropriate degree of genetic testing regulation.

Mariann Kasela
Helsinki, Finland

Talk: C08.6 Functional analysis of mismatch repair gene variants of uncertain significance and their possible contribution to Lynch syndrome.
Session: C08 Cancer genetics
Date: Sunday, June 1, 2014, 13:30 hrs.
Q1: 10/29/1986, Tallinn, Estonia
Q2: Doctoral student at University of Helsinki
Q3: Since childhood I have been interested in nature and animals. My mother and grandmother are both doctors. I guess since I grew up in this environment it influenced me. In addition I had a really interesting biology teacher who first introduced the field of genetics in the ninth grade. Since then I knew I wanted to study genetics. And I have not regretted my choice.
Q4: The study regarding Lynch syndrome is interesting to me because there are a lot of people all around the world who suffer from this syndrome. I personally feel that our research has a great impact on discovering putative Lynch syndrome variants and help to develop more transparent and efficient way to determine the patients and give them accurate care.

Sietske Kevelam
Amsterdam, Netherlands

Talk: C15.6 Novel (ovario)leukodystrophy related to AARS2 mutations
Session: C15 Novel genes in neurogenetic disorders
Date: Monday, June 2, 2014, 13:30 hrs.
Q1: 7/30/1985, Nijmegen, The Netherlands
Q2: I am currently 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 health and disease. It is exciting to learn more about the underlying genetic mechanisms of diseases and the implications of these insights for human physiology. With the rapidly evolving new techniques we can and will decipher more and more disorders, which will give patients and their families answers and help them cope with their diseases.
Q4: My focus is on exceedingly rare inherited progressive encephalopathies. These patients present with specific patterns of MRI abnormalities. We use such MRI-patterns to form homogeneous groups of patients, which helps tremendously in finding the common mutated gene by exome sequencing. In this study we identified mutations in AARS2 in patients with specific abnormalities of the left-right connections in the corpus callosum and descendings tracts of the brain, and in females ovarian failure.

Anat Kreimer
New York, United States

Talk: C11.5 Co-regulated transcripts associated to cooperating eSNPs define bi-fan motifs in human gene networks
Session: C11 Statistical genetics
Date: Sunday, June 1, 2014, 13:30 hrs.

Laura Kremer
Neuherberg, Germany

Talk: C17.2 Decoding Mitochondrial Disorders using Exome Sequencing
Session: C17 Metabolic and mitochondrial disorders
Date: Monday, June 2, 2014, 13:30 hrs.
Q1: 9/5/1986, Rodalben, Germany
Q2: Second year PhD student at Institute of Human Genetics at the Helmholtz Zentrum München
Q3: The identification of disease causing mutation eases the understanding of the pathological phenotype and holds promise for the developement of therapeutic approaches. Therefore I chose to investigate the genetic causes of mitochondrial disorders, which for the most part still lack treatement options.
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.

Christina Lissewski
Magdeburg, Germany

Talk: C21.2 The phenotypic spectrum of SHOC2 c.4A>G (p.Ser2Gly)
Session: C21 Rasopathies and CDG
Date: , 11:00 hrs.
Q1: 4/5/1983, Wilhelmshaven, Germany
Q2: I am a PhD student in the Institute of Human Genetics in Magdeburg, Germany.
Q3: I already knew in school that I wanted to be a scientist. As an AuPair I watched 2 boys and one of them has Noonan syndrome. This made me pick a college class in Genetics. I liked it and decided to study Biology and get my Masters in Genetics. Being able to write my dissertation on Noonan syndrome and related disorders is an added bonus.
Q4: We were able to collect clinical information from a large number of patients with Noonan-like syndrome and a specific mutation (SHOC2 p.S2G). This should give patients and their families a better idea on prognosis and possible rare complictions.

Jimmy Liu
Hinxton, United Kingdom

Talk: C14.6 Transethnic association study of IBD identifies novel risk loci and shows pervasive sharing of genetic risk factors across populations
Session: C14 Genetics of complex traits
Date: Monday, June 2, 2014, 13:30 hrs.
Q1: 3/19/1986, Harbin, China
Q2: PhD student at the Wellcome Trust Sanger Institute
Q3: The analysis of modern genomic datasets requires a unique blend of biology, statistics, mathematics and computer science. I am interested in how these come together to help us better understand disease, and ultimately translation into more effective therapies.
Q4: Fewer than 5% of genetic association studies have been performed with non-European samples. Our work on the genetics of inflammatory bowel disease (IBD) represents the largest of its type in South and East Asian populations. In addition to discovering risk loci, our study for the first time enables well-powered unravelling of both the similarities and differences in the genetic architecture of IBD between European and Asian populations.

Maria Nicla Loviglio
Lausanne, Switzerland

Talk: C06.4 Chromatin loops and CNVs: the complex spatial organization of the 16p11.2 locus
Session: C06 Functional and computational genomics
Date: Saturday, May 31, 2014, 6:30 hrs.
Q1: 3/29/1986, Altamura(Bari)-Italy
Q2: PhD student at CIG - University of Lausanne
Q3: I choose a career in genetics because I love the idea of getting a deeper understanding about all the elements concurring in the definition of the phenotype. Furthermore, I strongly believe that advancements in the medical genetics field can have a great impact on people's life.
Q4: I think that the study of chromatin organization provides an additional layer of complexity to the understanding of the complex mechanisms regulating gene expression, likely contributing to disease phenotype.

Danielle Lynch
Calgary, Canada

Talk: PL2.2 Disrupted auto-regulation of SNRPB causes cerebro-costo-mandibular syndrome
Session: PL2 What's new? Highlights Session
Date: Saturday, May 31, 2014, 4:30:00 PM hrs.
Q1: 9/23/1989, Vancouver, Canada
Q2: PhD student in at the University of Calgary
Q3: Working in genetics allows me to satisfy both my love of molecular biology and my curiosity about what we are made of as humans.
Q4: This research reveals the long sought-after gene causing cerebro-costo-mandibular syndrome (CCMS). Our discovery that CCMS is caused by mutatations in a core spliceosomal component invites questions on the likely very nuanced role of splicing in development. We also provide the first example of de-regulation of spliceosome-mediated mRNA decay in disease.

Pamela Magini
Bologna, Italy

Talk: C21.4 A mutation in PAK3 with a dual molecular effect deregulates the RAS/MAPK pathway and drives an X-linked syndromic phenotype
Session: C21 Rasopathies and CDG
Date: Tuesday, June 3, 2014, 11:00 hrs.
Q1: 1/26/1982, Mondavio (Pesaro-Urbino), Italy
Q2: Postdoctoral fellow

Q3: I chose genetics as a natural continuation of my academic studies in biology. After my first stage in a genetic laboratory I was sure that this would have been my career: what could be more fascinating than a code of only four bases that regulates the life of entire organisms?
Q4: The most interesting finding of our research is that we concretely demonstrated that a single mutation in a gene had a double functional effect, depriving the encoded protein of its biological activity and conferring it an uncontrolled dominant-negative function. This combination caused a more severe phenotype compared to simple loss-of-function mutations in the same gene, suggesting that what is generally defined as “variable expressivity” could have a molecular explanation that should be investigated.

Maria Antonietta Mencarelli
Siena, Italy

Talk: C19.6 Digenic model in Alport syndrome
Session: C19 Internal organs
Date: , 11:00 hrs.
Q1: 11/8/1979, Chianciano Terme, Italy
Q2: PhD Student and Consultant
Q3: Medical genetics offers me the opportunity to work on a constantly evolving matter that ranges a broad spectrum of medical disciplines from Pediatrics to Oncology going through Neurology and Prenatal Diagnosis.
Q4: The work that I present identifies a new mechanism of inheritance in a well known Mendelian disease, possibly leading to the characterization of potential modifying factors that can have relevant implications in genetic counselling.

Martin Mensah
Berlin, Germany

Talk: C20.4 Pseudoautosomal region 1 length polymorphism in the human population
Session: C20 Basic mechanisms in genetics
Date: , 11:00 hrs.
Q1: 5/25/1988, Berlin, Germany
Q2: I am a final year medical student at the Charité Berlin writing my theseis in genetics in a corporation with KU Leuven.
Q3: I have already been fascinated by human biology and especially genetics during my highschool time. That encouraged me to study medicine with the aim of becoming a genetecist.
Q4: We have found the first polymorphism of PAR1’s length, which had apparently been formed by NAHR. This is a totally new aspect of sex chromosomal evolution. Interestingly, this NAHR had been mediated by a homology of just a few hundred bp length.

Elke Mersy
Maastricht, Netherlands

Talk: C01.5 Scenarios for implementation of noninvasive prenatal testing (NIPT) for Down syndrome in a national health care system
Session: C01 Prenatal testing
Date: Saturday, May 31, 2014, 6:30 hrs.
Q1: 8/18/1986, Kortrijk, Belgium
Q2: Currently, I am a medical doctor working as a full time researcher on new developments in non-invasive prenatal testing at the Clinical Genetics department of the Maastricht University Medical Center in the Netherlands.
Q3: I have always been interested in the fundamentals of genetics and the technical developments. Most importantly, I am interested in the translation of these technologies for the benefit of patients.
Q4: Addition of non-invasive prenatal testing (NIPT) into the national Down syndrome screening programs will result in important advantages for pregnant women. However, as one might suspect, this is a complicated process and requires decision-making about the timing of the test and the combination with other tests. To provide an overview of the pros and cons of different NIPT implementation strategies, we combined a decision-analytic model and an ethical exploration.

Marije Meuwissen
Rotterdam, Netherlands

Talk: C15.4 Interferon type 1 response regulator USP18 is mutated in severe pseudo-TORCH syndrome
Session: C15 Novel genes in neurogenetic disorders
Date: Monday, June 2, 2014, 13:30 hrs.
Q1: 7/16/1980, Leidschendam, The Netherlands
Q2: After finishing my training as a clinical geneticist and my PhD at the Erasmus University Medical Center Rotterdam, I am currently working as a Clinical Geneticist at the University Hospital Brussels.
Q3: I love the combination of the technical, molecular part and the communicative aspects of the field, together with the challenge of fitting different pieces of a puzzle.
Q4: We identified a novel autosomal recessive cause of “pseudo-TORCH” syndrome with in addition severe cerebral hemorrhage and a dramatic course in the affected patients. We identified USP18 mutations, leading to an upregulation of the IFN type I signalling. Although this is also observed secondary to viral infections and Aicardi- Goutières syndrome, this is the first time that “pseudo-TORCH” and this signalling pathway are directly linked.

PROGRAMME YOUNG INVESTIGATOR AWARD CANDIDATES

Dimitra Micha
Amsterdam, Netherlands

Talk: C10.1 PLS3 mutations in X-linked osteoporosis and fractures: unraveling a new bone regulatory pathway
Session: C10 Bone and skeletal patterning
Date: Sunday, June 1, 2014, 13:30 hrs.

Q1: 6/1/1905, Stockholm, Sweden
Q2: Post doc in the Clinical Genetics department of the Vrije Universiteit medical center in Amsterdam Netherlands.
Q3: I believe genetics holds the key for the cure of many diseases. Understanding disease genetics is a very important step towards the development of new treatment approaches.
Q4: We discovered PLS3 mutations to be a monogenetic cause of X-linked recessive osteoporosis. A rare PLS3 variant associated with osteoporosis in the general population. This provides a completely novel bone regulatory pathway responsible for the development of this disease which will open new therapeutic avenues in the future.

Andréanne Morin
Montréal, Canada

Talk: C14.4 ImmunoSeq: Discovery of novel rare variants implicated in autoimmune and inflammatory diseases by targeting regulatory regions in immune cells
Session: C14 Genetics of complex traits
Date: Monday, June 2, 2014, 13:30 hrs.

Q1: 1/27/1988, Chicoutimi, Québec, Canada
Q2: I am a PhD candidate in the Human Genetics department, McGill University, Montreal, Canada. I am under the co-supervision of Dr. Tomi Pastinen (McGill University, Montreal) and Dr. Catherine Laprise (Université du Québec à Chicoutimi, Chicoutimi)
Q3: Science and research in health have always interested and intrigue me. I met researchers at a young age and found what they were doing fascinating. From that point, my interest for research in health just kept growing. The incredible progress of genetics and genomics in the past few years and the high discovery potential really attracted me. I am working on the genomics of complex traits, more precisely on the impact of rare variants in the development of autoimmune and inflammatory diseases. Working in a field which is still unexplored and where there is still so much to discover and understand is very motivating for me to be part of.
Q4: My project is the design of the ImmunoSeq, a novel way to interrogate rare non-coding variants. By targeting and sequencing non-coding regulatory regions of immune cells, we think that we can find potentially causal variants for different autoimmune and inflammatory diseases in a cost effective manner. Preliminary results show the potential impact of rare variants on gene expression regulation. This unique approach will potentially help to better identify relevant disease mechanisms of autoimmune and inflammatory complex trait.

Danit Oz-Levi
Rehovot, Israel

Talk: C17.4 Deletion of a distant-acting enhancer near C16ORF91 underlies recessive congenital diarrhea
Session: C17 Metabolic and mitochondrial disorders
Date: Monday, June 2, 2014, 13:30 hrs.

Q1: 9/14/1983, Kfar Saba Israel
Q2: PhD student in genetics
Q3: I wanted to study genetic diseases in order to be able to help decipher undiagnosed conditions that might provide prenatal screening options for families seeking such services. It is of critical importance to learn the mechanisms of rare diseases in order to improve our understanding of drug therapies and treatment, especially in cases where there are not too many patients with a particular disease, a situation that leaves them bereft of answers.
Q4: We have identified the disease casuing variant for a rare form of congenital diarrhea in an intergenic region using exome sequencing alone. The sophisticated bioinformatic analysis not only led to the identification of a deleted intergenic region, but also defined it as a putative enhancer, that when deletd is causing the disease.

Francesca Pantaleoni
Roma, Italy

Talk: C21.5 Activating mutations in RRAS underlie a phenotype within the RASopathy spectrum and contribute to leukaemogenesis
Session: C21 Rasopathies and CDG
Date: Tuesday, June 3, 2014, 11:00 hrs.

Yonatan Perez
Beer Sheva, Israel

Talk: C12.6 Isolated foveal hypoplasia with secondary nystagmus and low vision is associated with a homozygous SLC38A8 mutation
Session: C12 Sensory disorders
Date: Sunday, June 1, 2014, 13:30 hrs.

Q1: 9/23/1983, Beer-Sheva, Israel
Q2: PhD student, The Morris Khan Laboratory of Human Genetics at the National Institute of Biotechnology in the Negev, Department of Genetics, Faculty of Health Sciences, Ben-Gurion University, Beer Sheva, Israel.
Q3: Genetics in general has always fascinated me because of its complexity, beauty and elegance. I still find the basic concepts of phenotypic information transferred from generation to generation via a basic molecular structure fascinating. My passion for the field of human genetics in particular is driven by my curiosity and interest in medical sciences and because it allows me to be in a unique position, having the great privilege of influencing people's quality of life.
Q4: My research enabled insights into normal eye development, as well as identification of the molecular basis of a human disease. I have demonstrated that a homozygous mutation in SLC38A8 causes isolated hypoplasia of the fovea, a pit in the

retina that is essential for sharp vision. Interestingly, I have shown that SLC38A8 resides within the nuclear membrane, suggesting a role different than that predicted for similar putative amino acid transporters.

Slavil Peykov
Heidelberg, Germany

Talk: C09.1 Functional analysis of SHANK2 mutations identified in schizophrenia patients
Session: C09 Common neurological disease
Date: Sunday, June 1, 2014, 13:30 hrs.
Q1: 9/30/1984, Sofia, Bulgaria
Q2: PhD student at the Institute of Human Genetics (Heidelberg)
Q3: I always wanted to learn how the information stored in our genomes can be read, understood and finally re-written in a ways that answer questions about mechanisms of different disorders and potentially fix different mistakes in our DNA.
Q4: This is the first report showing an association of the SHANK2 gene to schizophrenia. Our information completes the story of the SHANK gene family and suggests that all three members are playing role in both major neurological disorders ASD and SCZ.

Aldesia Provenzano
Firenze, Italy

Talk: C02.2 High throughput sequencing in sporadic forms of steroid-resistant nephrotic syndrome: heterogeneous genetic alterations can predict resistance to treatments
Session: C02 Personalized medicine and pharmacogenomics
Date: Saturday, May 31, 2014, 6:30 hrs.
Q1: 8/11/1982, Cosenza Italy
Q2: PhD student
Q3: Genetics has always fascinated me because is like explore the universe of diseases with a magnifying glass. I was always interested to search the cause of biological processes behind human diseases and genetics gives the opportunity to find novel genes or mechanisms to improve the knowledge in this field. My interest has grown since I work in a children's hospital, it's amazing understand the physiopathology of genetic conditions because it can bring to the development of personalized therapies.
Q4: Our study demonstrated that a genetic test for children affected by nephrotic syndrome is very helpful for their management. In particular the resistance to immunosuppressive treatments in these patients is frequently associated with mutations in podocyte genes. The genetic results may help clinicians to establish a personalized therapy to each patients

Allison Richards
Ann Arbor, United States

Talk: C20.6 RNA-DNA Differences in Endoplasmic Reticulum Stress Response
Session: C20 Basic mechanisms in genetics
Date: , 11:00 hrs.
Q1: 5/3/1988, New York, United States
Q2: I am a graduate student in my 4th year.
Q3: Genetics offers me the opportunity to study individual differences in human phenotypes. I am interested in characterizing the extent of this natural variation and taking advantage of it to determine the mechanistic basis of human diseases.
Q4: My research highlights the role of RNA processing, such as canonical RNA editing and other types of RNA-DNA sequence differences, in regulating cellular response to stress.

Melissa Sambrotta
London, United Kingdom

Talk: C19.3 TJP2 deficiency: a new cholestatic liver disease
Session: C19 Internal organs
Date: , 11:00 hrs.
Q1: 11/16/1985, Italy
Q2: PhD student in liver molecular genetics at King's College London
Q3: I honestly believe that genetics is going to change the face of medicine in the next few years. The Human Genome Project was just the beginning. At the moment I am studying Mendelian diseases, which represent high penetrance variants in the genome. I have previously studied common variants which predispose to disease. I think that the variation on the human genome between these extremes is going to unravel many of the explanations of human disease in the decade. I want to contribute to this work, and really understand the role of genetics in physiology and pathophysiology.
Q4: The research that I'm going to present is focused on the importance of tight junction complexes in liver disease. Recently, through the application of next generation sequencing, we identified novel mutations in tight junction protein 2 involved in the aetiology of a rare Mendelian liver disorder known as progressive familial intrahepatic cholestasis, which arises in early childhood causing severe liver damage, followed by death if no liver transplantation has occurred.

Thomas Schwarzmayr
Neuherberg, Germany

Talk: C19.1 Constitutive Activation of PRKACA in Adrenal Cushing's Syndrome
Session: C19 Internal organs
Date: Tuesday, June 3, 2014, 11:00 hrs.

Melissa Sorosina
Milan, Italy

Talk: C02.1 A novel variant in the SLC9A9 gene influences disease activity in interferon-beta treated multiple sclerosis patients
Session: C02 Personalized medicine and pharmacogenomics
Date: Saturday, May 31, 2014, 6:30 hrs.
Q1: 11/14/1985, Bergamo, Italy
Q2: I have recently completed my PhD at the San Raffaele Hospital in Milan where I'm continuing to work as post doc fellow.
Q3: I've always been fascinated by science, especially by neurology and genetics. After the master's degree I started to work on neurological complex disorders allowing me to improve the knowledge and increase the interest on this topic. This is a highly dynamic field and a career in genetics will be full of exciting challenges.
Q4: I'm going to present a pharmacogenetic study including almost 1,000 multiple sclerosis patients which led us to the identification of a SNP associated with the response to interferon-beta. This finding was further corroborated with functional experiments, supporting the involvement of this variant in the interferon-beta pathways.

Sérgio Sousa
Coimbra, Portugal

Talk: C16.2 Lenz-Majewski syndrome: disturbed phosphatidylserine metabolism causes intellectual disability and a sclerosing bone dysplasia
Session: C16 Genes and development 2
Date: Monday, June 2, 2014, 13:30 hrs.
Q1: 12/31/1977, Coimbra, Portugal
Q2: Medical Geneticist
Q3: Curiosity is definitely part of the reason - wanting to know why. Working in genetics and especially dysmorphology is a permanent challenge. One never knows where the story will take us and the journey is often surprising.
Q4: First, the phenotype – a striking, progressive and very specific pattern of malformations. Secondly, the discovery of the first human disease caused by disturbed phosphatidylserine synthesis, and part of the growing group of diseases of the phospholipid metabolism. Lastly, the characterisation of one of the few examples of conditions caused by gain-of-function mutations affecting an enzyme.

Georgios Stamoulis
Geneva, Switzerland

Talk: C20.1 Single cell allele-specific expression (ASE) in Down syndrome and common aneuploidies.
Session: C20 Basic mechanisms in genetics
Date: , 11:00 hrs.
Q1: 3/4/1986, Thebes, Greece
Q2: PhD student at Stylianos Antonarakis' laboratory in the Department of Genetic Medicine and Development, University of Geneva, Switzerland
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) 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 inter-individual variability in expression profile. Through this study we aim to improve the understanding of the molecular basis of Down syndrome and other common aneuploidies.

Evangelia Stergiakouli
Bristol, United Kingdom

Talk: C11.1 Polygenic risk for ADHD is associated with impaired educational achievement and lower IQ in the general population
Session: C11 Statistical genetics
Date: Sunday, June 1, 2014, 13:30 hrs.
Q1: 5/2/1982, Larissa, Greece
Q2: I am a postdoctoral researcher at the MRC Integrative Epidemiology Unit at the University of Bristol where I am using genetic epidemiology and statistical genetics methods to investigate genetic factors influencing complex disorders. I am especially interested in the genetics of psychiatric disorders and traits and genetic factors influencing sexually dimorphic psychiatric traits.
Q3: I decided to pursue a career in genetics when I first learnt at school that the DNA code is written using only 4 different nucleotides encoding the instructions for the development and functioning of any organism. I never cease to be fascinated by the potential of genetics to improve our health.
Q4: I am investigating polygenic risk scores, which are aggregates of common genetic variants associated with ADHD. My study highlights the importance of ADHD genetic scores for individuals from the general population without the disorder. Higher genetic scores for ADHD are associated with worse educational outcomes and lower IQ even when people do not have the disorder.

YOUNG INVESTIGATOR AWARD CANDIDATES

Sofie Symoens
Ghent, Belgium

Talk: C10.5 Defects in TAPT1, involved in Axial Skeletal Patterning, Cause a Complex Lethal Recessive Disorder of Skeletal Development
Session: C10 Bone and skeletal patterning
Date: Sunday, June 1, 2014, 13:30 hrs.
Q1: 7/29/1978, Ghent, Belgium
Q2: Post-doctoral researcher - supervisor Connective Tissue Lab (focus on Osteogenesis imperfecta and Ehlers-Danlos syndrome)
Q3: The combination of research and clinical diagnosis is according to me a very exciting combination. Trying to find the causal underlying genetic defect of heritable (connective tissue) syndromes is very intriguing since it learns us more on general biology questions and also it helps to understand the underlying pathogenic pathways of disease.
Q4: Osteogenesis imperfecta is a heritable brittle bone disease with variable clinical severity. Although almost patients are genetically unraveled, a certain proportion still remains in whom no causal defect can be found. Identification of novel genetic causes not only sheds more light on the disease itself, but also reveals important processes or pathways in normal bone formation. The gene we have identified encodes TAPT1, a protein with until now unknown function. We showed that TAPT1 is important for cilium formation, thereby implying that correct cilium formation and signalling is crucial for normal embryonic bone formation.

Taru Tukiainen
Boston, United States

Talk: PL2.5 Chromosome X-wide association analysis discovers new loci for complex traits including a height locus not dosage compensated between men and women
Session: PL2 What's new? Highlights Session
Date: Saturday, May 31, 2014, 4:30:00 PM hrs.
Q1: 3/10/1983, Helsinki, Finland
Q2: Research Fellow at the Analytic and Translational Genetics Unit, Massachusetts General Hospital and the Broad Institute, Boston, USA
Q3: Genetics is a beautiful combination of biology and statistics
Q4: Our study emphasizes the value of including the X chromosome in large-scale genetic association studies of complex traits, which thus far have often focused only on autosomal variation. ChrX is not only a stretch of DNA but the loci that escape from X chromosome inactivation, and hence are not dosage compensated between men and women, provide another and a particularly fascinating dimension to ChrX association studies.

Jessica van Setten
Utrecht, Netherlands

Talk: C14.2 Genome of the Netherlands imputation identifies seven new loci for quantitative ECG traits in meta-analysis of 30,000 samples.
Session: C14 Genetics of complex traits
Date: Monday, June 2, 2014, 13:30 hrs.
Q1: 1/10/1987, Heerlen, The Netherlands
Q2: PhD student at the University Medical Center Utrecht, department of Medical Genetics
Q3: I want to understand the biological processes underlying diseases without the restriction of focusing on a specific cell or protein. Instead, working with genome-wide data provides the opportunity to test millions of variants simultaneously. I enjoy working on various traits in a field that is in constant development.
Q4: We meta-analyzed association results of 30,000 samples for four quantitative ECG traits and identified seven novel loci, using Genome of the Netherlands as an imputation reference panel. We show that the use of larger and more accurate imputation reference panels allow us to identify novel SNP-disease associations.

Terry Vrijenhoek
Utrecht, Netherlands

Talk: C13.5 The stepping stone approach towards the Genetics Clinic of the Future
Session: C13 Innovation in genetic services
Date: Monday, June 2, 2014, 13:30 hrs.
Q1: 6/24/1979, Delft, The Netherlands
Q2: Faculty and Staff Advisor
Q3: The possibility to sequence anyone's DNA at high speed and low costs puts genetics in an increasingly central position in health care. It is crucial that health care policy be adapted to this changing landscape. I hope to make a significant contribution to the genetics-based health care agenda for the future.
Q4: The consortium that I am representing is working towards the Genetics Clinic of the Future. We take an approach that is based on 'radical interdisciplinarity'; we bring together disciplines that are generally wide apart to jointly identify the design principles of genome data infrastructures as genomic technologies mature and become integrated in routine diagnostic procedures and health management systems.

PROGRAMME YOUNG INVESTIGATOR AWARD CANDIDATES

Marjolein Willemsen
Nijmegen, Netherlands

Talk: C03.1 Dominant β-catenin mutations cause a recognizable syndrome with intellectual disability, and are associated with learning deficits and structural and functional brain abnormalities in mice
Session: C03 Intellectual disability
Date: Saturday, May 31, 2014, 6:30 hrs.
Q1: 2/28/1981, Nijmegen The Netherlands
Q2: Clinical geneticist in training
Q3: First of all, I like the opportunity to combine and link genetic research with the care for patients and their families. It is also a pleasure to collaborate with many different disciplines in the lab and in the clinics. Furthermore, it is great to be part of the fast moving and exciting field of genetics.
Q4: I will present a group of patients representing a novel recognizable intellectual disability syndrome caused by dominant mutations in the gene CTNNB1. In addition I will show the results of the functional studies that we have performed in parallel in a mouse mutant, illustrating the consequences of beta-catenin dysfunction through development and into adulthood.

Rana Yadak
Rotterdam, Netherlands

Talk: C17.3 Lentivial vector based hematopoietic stem cell gene therapy mediates sustained expression of functional thymidine phosphorylase in mitochondrial neurogastrointestinal encephalopathy mouse model
Session: C17 Metabolic and mitochondrial disorders
Date: Monday, June 2, 2014, 13:30 hrs.
Q1: 3/23/1986, Nablus, Palestine
Q2: PhD student
Q3: Better understanding of genes leads to better understanding of a disease leads to better treatment strategies eventually leading to a better life.
Q4: Mitochondrial neurogastrointestinal encephalomyopathy (MNGIE) is an autosomal recessive multisystemic disease. Different kinds of pathogenic mutations in the thymidine phosphorylase (TP) gene are responsible for the biochemical imbalances in the nucleaoside levels that leads to the alterations in the function of mitochondria in the affected tissues. Our target is to optimize a Lenti-viral vector based hematopoietic stem cell gene therapy protocol to introduce a functional copy of the human TP gene in a MNGIE mouse model, aiming for a safe and long term correction of the biochemical imbalances.

Masoud Zamani Esteki
Leuven, Belgium

Talk: C01.4 Whole-genome single-cell haplotyping, a generic method for preimplantation genetic diagnosis
Session: C01 Prenatal testing
Date: Saturday, May 31, 2014, 6:30 hrs.
Q1: 8/19/1984,
Q2: PhD student
Q3: When I was a high-school student and was introduced to the amazing laws of inheritance formulated by my hero (Mendel), I was so fascinated by the fact that simple and limited resources lead to such a discovery and established the fundamentals of Genetics, even before DNA double helix was part of equation! Later on, when I learned about the 'chromosome theory' by Morgan that I found the inheritance more complex. Furthermore, I realized that how solving wonderful logic problems, by these two great scientists, defined the actual inheritance. As a graduate student, I started my research in the young and fascinating field of single-cell genomics. A combination of these factors leads me to apply fundamental genetics at the single-cell level and develop novel genome screening tools. Currently, I'm interested in development and application of these for basic research, e.g. to study the genetic basis of early development in human. Importantly, I am committed to translate these genome screening tools into the clinic.
Q4: We developed and validated a genome-wide genome screening approach as a generic method for preimplantation genetic diagnosis. The method allows selecting for single Mendelian up to various Mendelian traits at once, as well as for a combination of ancient genetic variants conferring susceptibility to complex diseases, which are increasingly being discovered in large-scale genome-wide association studies. We anticipate single-cell haplotyping will standardize PGD practice.

PROGRAMME POSTER AWARD FINALISTS

ESHG Poster Awards

For the fourth time, the ESHG proposes the ESHG Poster award for the 7 best posters presented by Young Investigators at the meeting. Finalists receive a complementary ESHG online membership for 1 year. The 2 winners will receive prize money of EUR 500.

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

P01.091-S Identification of rare CNVs involving genes acting in oocyte maturation and differentiation in a cohort of patients affected by Primary Ovarian Insufficiency

I. Bestetti[1,2], *C. Castronovo*[1], *M. Crippa*[1], *R. Rossetti*[3], *A. Pistocchi*[2], *C. Caslini*[2], *C. Sala*[4], *D. Toniolo*[4], *L. Persani*[3,5], *A. Marozzi*[2], *P. Finelli*[1]*;*
[1]*Laboratory of Medical Cytogenetics and Molecular Genetics, IRCCS Istituto Auxologico Italiano, Milano, Italy,* [2]*Department of Medical Biotechnology and Translational Medicine, University of Milan, Milano, Italy,* [3]*Laboratory of Endocrine and Metabolic Research and Division of Endocrine and Metabolic Diseases, IRCSS Istituto Auxologico Italiano, Milano, Italy,* [4]*Division of Genetics and Cell Biology, San Raffaele Research Institute and Vita Salute University, Milano, Italy,* [5]*Department of Clinical Sciences and Community Health,University of Milan, Milano, Italy.*

P02.10-M Hearing and ageing: a complex genomic strategy leading to new genes/variants identification in European and Central Asian populations

D. Vuckovic[1], *S. Dawson*[2], *M. P. Concas*[3], *M. Ciullo*[4], *T. Nutile*[4], *S. Cappellani*[5], *M. Pirastu*[3], *P. Gasparini*[1,5], *G. Girotto*[1]*;*
[1]*Dep. Medical Sciences - University of Trieste, Trieste, Italy,* [2]*UCL Ear Institute, University College London, London, United Kingdom, 3Institute of Population Genetics, National Council of Research, Sassari, Italy, 4Inst. Genetics and Biophysics 'A.Buzzati-Traverso', CNR, Naples, Italy, 5Inst. Maternal and Child Health-IRCCS 'Burlo Garofolo', Trieste, Italy.*

P02.37-S Homozygous deletion of glutamate receptor gene *GRID2* causes new human hotfoot mutant phenotype, characterized by early-onset cerebellar ataxia and retinal dystrophy

K. Van Schil[1], *M. Karlstetter*[2], *F. Meire*[3], *M. Bauwens*[1], *H. Verdin*[1], *F. Coppieters*[1], *E. Scheiffert*[2], *N. Deconinck*[4], *T. Langmann*[2], *E. De Baere*[1]*;*
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P03.12-M New genetic abnormalities underlying chronic intestinal pseudo-obstruction (CIPO)

F. Bianco[1], *L. Cordeddu*[2], *M. D'Amato*[2], *M. Bamshad*[3], *L. Francescatto*[3], *V. Stanghellini*[1], *G. Lindberg*[2], *Z. Mungan*[4], *C. Graziano*[1], *T. Pippucci*[1], *N. Katsanis*[5], *M. Seri*[1], *G. Romeo*[1], *R. De Giorgio*[1], *E. Bonora*[1]*;*
[1]*University of Bologna, St Orsola Malpighi, Bologna, Italy,* [2]*Karolinska Institutet, Stockholm, Sweden,* [3]*Center for Mendelian Disorders,University of Washington, Seattle, WA, United States,* [4]*Koc University School of Medicine, Istanbul, Turkey,* [5]*Depts of Cell Biology and Pediatrics,Duke University, Durham, NC, United States.*

P03.24-M Targeted sequencing of 208 candidate genes in 460 CAKUT patients facilitates the inclusion of a novel gene set in diagnostics

N. Nicolaou[1], *I. J. Nijman*[1], *S. van Lieshout*[1], *G. Monroe*[1], *A. M. van Eerde*[1], *W. F. Feitz*[2], *I. A. L. M. van Rooij*[3], *L. F. M. van der Zanden*[3], *N. Roeleveld*[3,4], *E. M. H. F. Bongers*[5], *R. H. Giles*[6], *E. Cuppen*[1], ***K. Y. Renkema***[1], *N. V. A. M. Knoers*[1]*;*
[1]*Medical Genetics, UMC Utrecht, Utrecht, Netherlands,* [2]*Urology, Radboud university medical center, Utrecht, Netherlands,* [3]*Health Evidence, Radboud university medical center, Nijmegen, Netherlands,* [4]*Paediatrics, Radboud university medical center, Nijmegen, Netherlands,* [5]*Genetics, Radboud university medical center, Nijmegen, Netherlands,* [6]*Nephrology and Hypertension, UMC Utrecht, Utrecht, Netherlands.*

P04.52-M A spectrum of disorders are associated with somatic mutations in *PIK3CA*, encoding the p110α catalytic subunit of phosphatidylinositol-4,5-bisphosphate 3-kinase

V. E. R. Parker[1], *A. Luchetti*[1], *H. Martin*[1], *I. Isaac*[1], *M. J. Lindhurst*[2], *J. Sapp*[2], *K. Keppler-Noreuil*[2], *L. G. Biesecker*[2], *E. R. Maher*[1], *R. K. Semple*[1]*;*
[1]*Cambridge University Hospitals NHS Trust, Cambridge, United Kingdom,* [2]*National Human Genome Research Institute (NHGRI)/NIH, Bethesda, MD, United States.*

P06.05-S Dissecting the genetic architecture of loci with established effects on multiple cardiometabolic phenotypes

L. Marullo[1], *T. O. Kilpeläinen*[2], *B. K. Cornes*[3], *J. Dupuis*[4], *C. Scapoli*[1], *R. J. F. Loos*[5], *J. B. Meigs*[3], *A. P. Morris*[6], *I. Prokopenko*[7], *on behalf of the XC-Pleiotropy Group;*
[1]*The Department of Life Sciences and Biotechnologies, University of Ferrara, Ferrara, Italy,* [2]*The Novo Nordisk Foundation Center for Basic Metabolic Research, Faculty of Health and Medical Sciences, University of Copenhagen, Copenhagen, Denmark,* [3]*General Medicine Division, Massachusetts General Hospital, Boston, MA, United States,* [4]*Department of Biostatistics, Boston University School of Public Health, Boston, MA, United States,* [5]*The Icahn School of Medicine at Mount Sinai, New York, NY, United States,* [6]*Department of Biostatistics, University of Liverpool, Liverpool, United Kingdom,* [7]*Department of Genomics of Common Disease, Imperial College London, London, United Kingdom.*

PROGRAMME POSTER AWARD FINALISTS

P06.55-S A homozygous mutation in the transcription factor THAP11 in a patient with methylmalonic aciduria and a severe neurological phenotype

A. Brebner[1], *H. Yu*[2], *D. Watkins*[1], *V. Adoue*[1,3], *T. Pastinen*[1,3], *F. Skovby*[4], *T. H. Shaikh*[2], *D. S. Rosenblatt*[1];

[1]*McGill University, Montreal, QC, Canada,* [2]*University of Colorado School of Medicine, Aurora, CO, United States,* [3]*McGill University and Genome Quebec Innovation Centre, Montreal, QC, Canada,* [4]*The Juliane Marie Centre, Rigshospitalet, Copenhagen, Denmark.*

P07.22-M SNP variants in MHC are associated with sarcoidosis susceptibility and subgroups - a joint case-control association study in four European populations

A. Wennerström[1,2,3], *E. Lahtela*[2], *V. Anttila*[4,5,3], *J. Grunewald*[6], *C. van Moorsel*[7], *M. Petrek*[8], *A. Eklund*[6], *J. Grutters*[7], *V. Kolek*[8], *L. Padyukov*[9], *A. Pietinalho*[10], *M. Ronninger*[11], *M. Seppänen*[12], *O. Selroos*[13], *M. Lokki*[14];

[1]*National Institute for Health and Welfare (THL) Public Health Genomics Unit, Helsinki, Finland,* [2]*Transplantation Laboratory, Haartman Institute, University of Helsinki, Finland, Helsinki, Finland,* [3]*University of Helsinki The Institute for Molecular Medicine Finland (FIMM) Biomedicum, Helsinki, Finland,* [4]*Analytical and Translational Genetics Unit, Department of Medicine, Massachusetts General Hospital and Harvard Medical School, Boston, MA, United States,* [5]*Program in Medical and Population Genetics, Broad Institute of MIT and Harvard, Cambridge, MA, United States,* [6]*Respiratory Medicine Unit, Department of Medicine Solna and CMM Karolinska Institutet and Karolinska University Hospital, Solna, Sweden,* [7]*Department of Pulmonology, St Antonius Hospital Nieuwegein, and Heart and Lung Center University Medical Center Utrecht, Utrecht, Netherlands,* [8]*Laboratory of Immunogenomics and Immunoproteomics, Faculty of Medicine and Dentistry, Palacky University Olomouc, Olomouc, Czech Republic,* [9]*Rheumatology Unit, Department of Medicine, Karolinska Institutet and Karolinska University Hospital, Stockholm, Sweden,* [10]*Raasepori Health Care Centre, Raasepori, Finland,* [11]*Respiratory Medicine Unit, Department of Medicine Solna and CMM, Karolinska Institutet and Karolinska University Hospital, Solna, Finland,* [12]*10 Immunodeficiency Unit, Division of Infectious Diseases, Department of Medicine, Helsinki University Central Hospital, Helsinki, Helsinki, Finland,* [13]*Semeco AB, Vejbystrand, Sweden,* [14]*Transplantation Laboratory, Haartman Institute, University of Helsinki, Helsinki, Finland.*

P07.34-M Functional analysis of genetic risk factors for canine SLE-related disease complex and identification of genetic risk factors for human SLE

F. H. G. Farias[1], *M. Wilbe*[2], *S. V. Kozyrev*[1], *D. Leonard*[3], *H. Bremer*[4], *J. Dahlqvist*[1], *A. Hedlund*[4], *G. R. Pielberg*[1], *U. Gustafson*[2], *M. Eloranta*[3], *H. Hansson-Hamlin*[4], *G. Andersson*[2], *L. Rönnblom*[3], *K. Lindblad-Toh*[1,5];

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P08.10-M Functional studies of ARX mutants linked to neurophenotypes and Application of rescue strategies targeting KDM5C down-regulation

L. Poeta[1], *A. Padula*[1], *C. Shoubridge*[2], *S. Zucchelli*[3], *F. Fusco*[1], *S. Filosa*[1,4], *P. Collombat*[5], *K. Helin*[6], *L. Altucci*[1,7], *M. Lioi*[8], *S. Gustincich*[3], *J. Gecz*[2], *M. Ursini*[1], *M. Miano*[1];

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P08.17-S TALEN-mediated mutagenesis as a tool to generate disease models for diseases caused by dominant de novo mutations

C. A. Biagosch[1,2], *S. Hensler*[3], *R. Kühn*[3], *T. Meitinger*[1,2], *H. Prokisch*[1,2];

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P08.18-M *NR2F1* mutations cause optic atrophy with intellectual disability

D. G. M. Bosch[1,2], *F. N. Boonstra*[2], *C. Gonzaga-Jauregui*[3], *M. Xu*[3], *J. de Ligt*[1], *S. Jhangiani*[3], *W. Wiszniewski*[3,4], *D. M. Muzny*[3], *H. G. Yntema*[1], *R. Pfundt*[1], *L. E. L. M. Vissers*[1], *L. Spruijt*[1], *E. A. W. Blokland*[1], *C. Chen*[3,4], *Baylor-Hopkins Center for Mendelian Genomics, R. A. Lewis*[3,4], *S. Y. Tsai*[3], *R. A. Gibbs*[3], *M. Tsai*[3], *J. R. Lupski*[3,4], *H. Y. Zoghbi*[3,4,5], *F. P. M. Cremers*[1], *C. P. Schaaf*[3,4], *B. B. A. de Vries*[1];

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P08.73-S Mutations in the P54NRB/NONO gene cause a novel syndromic XLID with a slender built-macrocephaly gestalt

M. Langouet[1], *M. Rio*[1], *S. Moutton*[1], *K. Siquier-Pernet*[1], *C. Bole-Feysot*[1], *N. Cagnard*[1], *P. Nitschke*[1], *A. Munnich*[1], *D. Mircsof*[2], *P. Seebeck*[3], *S. Brown*[2], *J. Amiel*[1], *L. Colleaux*[1];

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P09.038-M Rare Copy Number Variants underlying Genetic Epilepsy: a regional study

R. F. Oliveira[1,2], *C. Noakes*[3], *A. Smith*[3], *R. Candlin*[3], *E. Blair*[2], *R. Gibbons*[4], *J. Hurst*[5], *A. Nemeth*[2], *J. Poulton*[6], *S. Price*[2], *D. Shears*[2], *H. Stewart*[2], *J. Roberts*[3], *C. Campbell*[3], *U. Kini*[2];

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Clinical Genetics, Oxford University Hospitals NHS Trust, Oxford, United Kingdom, [3]Oxford Medical Genetics Laboratories, Oxford University Hospitals NHS Trust, Oxford, United Kingdom, [4]MRC Molecular Haematology Unit, Weatherall Institute of Molecular Medicine, Oxford University Hospitals NHS Trust, Oxford, United Kingdom, [5]NE Thames Genetics Service, Great Ormond Street Hospital NHS Foundation Trust, Great Ormond Street, London, United Kingdom, [6]Nuffield Department of Obstetrics and Gynaecology, Oxford University Hospitals NHS Trust, Oxford, United Kingdom.

P09.051-S Exome sequencing reveals mutations of a solute carrier gene in an autosomal recessive form of epileptic encephalopathy of the first days of life

J. Thevenon[1], *F. Feillet[2], M. Milh[3], J. St-Onge[4], Y. Duffourd[5], A. Roubertie[6], E. Raffo[7], C. Jugé[4], D. Héron[8], B. Isidor[9], D. Sanlaville[10], S. Wahlen[11], V. Darmency-Stamboul[12], A. Masurel-Paulet[1], C. Thauvin-Robinet[1], A. Toutain[13], G. Lesca[14], S. Odent[15], C. Philippe[16], L. Olivier-Faivre[1], J. Rivière[4];*

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P09.094-M A cell reprogramming-based approach to study 7q11.23 gene dosage imbalances in Williams Beuren syndrome and autism spectrum disorder

S. Atashpaz[1], *A. Adamo[1], P. Germain[1], J. Chenoweth[2], G. D'Agostino[1], M. Zanella[1], P. Prontera[3], C. Unger[4], P. W. Andrews[4], G. Pruneri[1], B. Hamilton[5], G. Merla[6], R. D. McKay[2], G. Testa[1];*

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P11.112-M Mutations in a new gene cause a novel overgrowth syndrome with macrocephaly, hypoglycemia, enlarged ventricles, mild/moderate intellectual disability and recurrent inflammatory diseases

J. Tenorio[1,2,3], *A. Mansilla[4], M. Valencia[5], V. Martínez-Glez[1], V. Romanelli[1], P. Arias[1], N. Castrejón[6], F. Poletta[7], E. Guillén-Navarro[8], G. Gordo[1], E. Mansilla[1], F. García-Santiago[1], I. González-Casado[9], E. Vallespín[1], M. Palomares[1], M. Mori[1], F. Santos-Simarro[1], S. García-Miñaur[1], L. Fernández[1], R. Mena[1], S. Benito-Sanz[1], A. del Pozo[1], J. Silla[1], K. Ibañez[1], E. López-Granados[1], A. Martín-Trujillo[10], D. Montaner[11], The SOGRI Consortium, K. E. Heath[1], Á. Campos-Barros[1], J. Dopazo[11], J. Nevado[1], D. Monk[10], V. L. Ruiz-Pérez[5], P. Lapunzina[1,2,3];*

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P12.020-M TAp73α regulates Otx1 expression during breast cancer stem cells differentiation and in response to cisplatin treatment

I. S. Pagani[1,2], *C. Pirrone[1], E. Amelotti[1], A. Terrinoni[2], F. Bernassola[2], A. G. Sanarico[2], M. Agostini[3], F. Pasquali[1], F. Lo Curto[1], I. Zucchi[4], E. Candi[2], G. Melino[2,3], G. Porta[1];*

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P12.040-M Identification of novel candidate genes for early-onset colorectal cancer susceptibility

R. M. de Voer[1], *M. Hahn[1], R. D. A. Weren[1], A. R. Mensenkamp[1], W. A. van Zelst-Stams[1], L. Spruijt[1], M. Kets[1], J. Zhang[1], C. Gilissen[1], M. Tychon[1], R. Derks[1], H. K. Schackert[2], A. Geurts-van Kessel[1], N. Hoogerbrugge[1], M. J. L. Ligtenberg[1], R. P. Kuiper[1];*

[1]Radboud university medical centre, Nijmegen, Netherlands, [2]Carl Gustav Universität Dresden, Dresden, Germany.

P12.041-S Whole-exome sequencing identifies rare coding variants in new predisposition genes for familial colorectal cancer

C. Esteban-Jurado[1], *M. Vila[1], P. Garre[2], J. Lozano[1], A. Pristoupilova[3], S. Beltrán[3], J. Muñoz[1], T. Ocaña[1], F. Balaguer[1], S. Franch[1], J. Piqué[1], A. Castells[1], A. Carracedo[4], C. Ruiz-Ponte[4], A. Abulí[5], X. Bessa[6], M. Andreu[6], The EPICOLON consortium, L. Bujanda[7], T. Caldés[2], S. Castellví-Bel[1];*

[1]IDIBAPS, CIBEREHD, Hospital Clínic, Barcelona, Spain, [2]Hospital Clínico San Carlos, Madrid, Spain, [3]CNAG, Barcelona,

Spain, [4]FPGMX, CIBERER, UInversity of Santiago de Compostela, Santiago de Compostela, Spain, [5]IDIBAPS, Hospital del Mar, Barcelona, Spain, [6]Hospital del Mar, Barcelona, Spain, [7]Hospital Donostia, CIBEREHD, San Sebastián, Spain.

P12.043-S Towards personalized cellular adoptive immunotherapy targeting immunogenic neo-antigens in microsatellite unstable colorectal cancers
P. Maby[1]*, M. Hamieh*[1]*, D. Tougeron*[2]*, B. Mlecnik*[3]*, G. Bindea*[3]*, H. Angell*[3]*, T. Fredriksen*[3]*, N. Elie*[4]*, A. Drouet*[1]*, E. Fauquembergue*[1]*, J. Mauillon*[5]*, R. Sesboüé*[1]*, J. Galon*[3]*, T. Frebourg*[6]*, J. Latouche*[6]*;*
[1]Inserm U1079, Rouen, France, [2]Department of Gastroenterology, University Hospital, Poitiers, France, [3]Inserm U872, Laboratory of Integrative Cancer Immunology, Paris, France, [4]Imaging Core Facility, CMABIO, University Hospital, Caen, France, [5]Department of Genetics, Rouen University Hospital, Rouen, France, [6]Inserm U1079 and Department of Genetics, University Hospital, Rouen, France.

P12.044-M Fourfold increased detection of Lynch syndrome by raising age limit for tumour genetic testing from 50 to 70 years is cost-effective
A. S. Sie[1]*, A. R. Mensenkamp*[1]*, E. M. M. Adang*[2]*, M. J. L. Ligtenberg*[3]*, N. Hoogerbrugge*[1]*;*
[1]Department of Human Genetics, Radboud university medical center, Nijmegen, Netherlands, [2]Department of Health Evidence, Radboud university medical center, Nijmegen, Netherlands, [3]Department of Human Genetics and Department of Pathology, Radboud university medical center, Nijmegen, Netherlands.

P12.061-S Impaired Th17 mucosal host defense against *Helicobacter pylori* in an early-onset gastric cancer patient with a homozygous germline variant in *MYD88*
I. P. Vogelaar*, M. J. L. Ligtenberg, R. S. van der Post, M. C. M. Kets, R. M. de Voer, T. J. G. Jansen, L. Jacobs, G. Schreibelt, H. J. M. van Krieken, J. M. de Vries, M. G. Netea, N. Hoogerbrugge;*
Radboud university medical center, Nijmegen, Netherlands.

P12.088-M Genetic variants in the interleukin locus at 1q32.1 as markers of melanoma survival
J. Rendleman*, C. Adaniel, E. Kern, N. Fleming, M. Krogsgaard, D. Polsky, R. Berman, R. Shapiro, A. Pavlick, Y. Shao, I. Osman, T. Kirchhoff;*
New York University, New York, NY, United States.

P12.114-M Development of Acquired Resistance to Anti-EGFR Therapy in Colorectal Cancer Identified by Whole-Genome Plasma DNA Sequencing
S. Mohan[1]*, E. Heitzer*[1]*, P. Ulz*[1]*, I. Lafer*[1]*, S. Lax*[2]*, M. Auer*[1]*, M. Pichler*[3]*, A. Gerger*[3]*, F. Eisner*[3]*, G. Hoefler*[4]*, T. Bauernhofer*[3]*, J. B. Geigl*[1]*, M. R. Speicher*[1]*;*
[1]Institute of Human Genetics, Medical University of Graz, Graz, Austria, [2]Department of Pathology,General Hospital Graz West, Graz, Austria, [3]Division of Oncology, Medical University of Graz, Graz, Austria, [4]Institute of Pathology, Medical University of Graz, Graz, Austria.

P14.14-M Profiling circulating miRNAs in plasma samples of celiac disease patients
R. C. Almeida[1]*, S. Vriezinga*[2]*, V. Kumar*[1]*, A. Stachurska*[1]*, R. Modderman*[1]*, U. Võsa*[3,4]*, L. Mearin*[2]*, C. Wijmenga*[1]*, S. Withoff*[1]*;*
[1]University of Groningen, University Medical Center Groningen, Dept. of Genetics, Groningen, Netherlands, [2]Leiden University Medical Center, Leiden, Netherlands, [3]Estonian Genome Center, University of Tartu, Tartu, Estonia, [4]Department of Biotechnology, Institute of Molecular and Cell Biology, University of Tartu, Tartu, Estonia.

P14.89-S Direct trans-differentiation of skin fibroblasts for functional testing of unclassified variants
J. Pals[1,2]*, K. van der Kuij*[2]*, D. Micha*[1]*, A. Maugeri*[1]*, G. Pals*[1]*, M. J. Baars*[3]*, V. Everts*[2]*, B. Zandieh Doulabi*[2]*;*
[1]VU medical center, Amsterdam, Netherlands, [2]ACTA, Amsterdam, Netherlands, [3]AMC, Amsterdam, Netherlands.

P16.55-S Epigenome-wide analysis identified highly significant age-related DNA methylation changes
A. Russo[1,2]*, S. Guarrera*[1,2]*, G. Fiorito*[1,2]*, C. Di Gaetano*[1,2]*, F. Rosa*[1,2]*, A. Allione*[1]*, F. Modica*[1]*, L. Iacoviello*[3]*, M. Giurdanella*[4]*, R. Tumino*[4]*, S. Grioni*[5]*, V. Krogh*[5]*, A. Mattiello*[6]*, S. Panico*[6]*, P. Vineis*[1,7]*, C. Sacerdote*[1,8]*, G. Matullo*[1,2]*;*
[1]Human Genetics Foundation, Torino, Italy, [2]University of Turin, Turin, Italy, [3]Catholic University, Campobasso, Italy, [4]"Civile-M.P. Arezzo" Hospital, Ragusa, Italy, [5]Fondazione IRCSS Istituto Nazionale dei Tumori, Milano, Italy, [6]Federico II University, Napoli, Italy, [7]Imperial College London, London, United Kingdom, [8]CPO-Piemonte, Turin, Italy.

P17.20-M High frequency of hypoketotic hypoglycemia associated CPT1A mutation in Northeast Siberia caused by positive selection
A. Cardona[1]*, F. Clemente*[1]*, C. Inchley*[1]*, L. Pagani*[1]*, M. Metspalu*[2,3]*, M. Vicente*[1]*, M. Mitt*[2]*, R. Magi*[2]*, T. Antao*[4]*, Z. Faltyskova*[1]*, C. Eichstaedt*[1]*, G. Jacobs*[5,1]*, D. Lawson*[6]*, Y. Xue*[7]*, Q. Ayub*[7]*, E. Willerslev*[8]*, R. Villems*[2,3]*, A. Vidal-Puig*[1]*, C. Tyler-Smith*[7]*, M. Derenko*[9]*, B. Malyarchuk*[9]*, J. Wee*[10]*, M. Syafiq Abdullah*[11]*, T. Kivilsid*[1,3]*;*
[1]University of Cambridge, Cambridge, United Kingdom, [2]University of Tartu, Tartu, Estonia, [3]Estonian Biocentre, Tartu, Estonia, [4]University of Oxford, Oxford, United Kingdom, [5]University of Southampton, Southampton, United Kingdom, [6]University of Bristol, Bristol, United Kingdom, [7]Wellcome Trust Sanger Institute, Hinxton, United Kingdom, [8]University of Copenhagen, Copenhagen, Denmark, [9]Institute of Biological Problems of the North, Magadan, Russian Federation, [10]National Cancer Centre, Singapore, Singapore, [11]RIPAS Hospital, Bandar Seri Begawan, Brunei Darussalam.

P17.25-S Exome sequencing revealing Nunavik Inuit specific variants in genes regulate lipid metabolism
S. Zhou[1], *L. Xiong*[2], *P. Xie*[1], *A. Ambalavanan*[1], *C. Bourassa*[1], *A. Dionne-Laporte*[1], *D. Spiegelman*[1], *N. Dupré*[3], *M. Dubé*[4], *P. Dion*[1], *G. A. Rouleau*[1]*;*
[1]*Montreal Neurological Institute, Montreal, QC, Canada,* [2]*Centre de recherche, Institut universitaire en santé mentale de Montréal, Montreal, QC, Canada,* [3]*Department of Neurology, Université Laval, Quebec, QC, Canada,* [4]*Pharmacogenomics Centre, Montreal Heart Institute, Montreal, QC, Canada.*

P17.26-M GWAS and candidate gene analysis highlight many novel loci associated to food preferences
N. Pirastu[1,2], *M. Kooyman*[3], *M. Traglia*[4], *A. Robino*[1,2], *S. M. Willems*[3], *G. Pistis*[4], *N. Amin*[3], *C. Sala*[4], *L. C. Karssen*[3], *C. Van Duijn*[3,5], *D. Toniolo*[4], *P. Gasparini*[1,2]*;*
[1]*Università degli Studi di Trieste, Trieste, Italy,* [2]*IRCCS Burlo Garofolo, Trieste, Italy,* [3]*Genetic Epidemiology Unit, Department of Epidemiology, Erasmus Medical Center, Rotterdam, Netherlands,* [4]*Division of Genetics and Cell Biology, San Raffaele Scientific Institute, Milan, Italy,* [5]*Centre for Medical Systems Biology, Leiden University Medical Center, Leiden, Netherlands.*

P17.45-S A signal near FRMD4A is associated with lower extremity arterial disease in patients with type 2 diabetes in GoDARTS
N. R. van Zuydam[1], *C. N. A. Palmer*[2], *H. M. Colhoun*[2], *SUMMIT;*
[1]*University of Oxford, Oxford, United Kingdom,* [2]*University of Dundee, Dundee, United Kingdom.*

P17.50-M Genetic markers predicting menopausal age associate with diabetes and lipid traits in 11864 Finns
A. Joensuu[1,2], *J. Kettunen*[1,2], *S. Ripatti*[1,2,3], *J. Sinisalo*[4], *M. S. Nieminen*[4], *M. Lokki*[5], *A. Jula*[6], *V. Salomaa*[2], *M. Perola*[2,1,7], *K. Auro*[2,1]*;*
[1]*Institute for Molecular Medicine Finland (FIMM), Helsinki, Finland,* [2]*National Institute for Health and Welfare (THL), Helsinki, Finland,* [3]*Hjelt Institute, University of Helsinki, Helsinki, Finland,* [4]*Heart and Lung Center HUCH, Helsinki University Central Hospital, Helsinki, Finland,* [5]*Haartman Institute, University of Helsinki, Helsinki, Finland,* [6]*National Institute for Health and Welfare (THL), Turku, Finland,* [7]*University of Tartu, Tartu, Estonia.*

P17.82-M Genetic survival modeling with large-scale population cohorts
C. Benner[1], *M. Pirinen*[1], *E. Tikkanen*[1,2], *S. Ripatti*[1,2,3]*;*
[1]*Institute for Molecular Medicine Finland, University of Helsinki, Helsinki, Finland,* [2]*Hjelt Institute, University of Helsinki, Helsinki, Finland,* [3]*Wellcome Trust Sanger Institute, Hinxton, Cambridge, United Kingdom.*

SCIENTIFIC PROGRAMME

EUROPEAN MEETING ON PSYCHOSOCIAL ASPECTS OF GENETICS - EMPAG 2014

PROGRAMME EMPAG 2014 - SATURDAY

Saturday, May 31, 2014

12:15 - 13:45 **EMPAG Workshop: The impact of risk reducing surgery** **Room Amber 3+4**

14:00 - 14:30 **Opening and welcoming addresses, joint with ESHG** **Gold Room**
Chair: A. Amoroso, H. Brunner

14:30 - 16:00 **EPL1 - EMPAG Plenary Session: Psychosocial issues in cancer genetics** **Room Amber 3+4**
Chair: D. Turchetti, E. Razzaboni

14:30 **EPL1.1 The impact of total gastrectomy upon e-cadherin carriers: experiences of eating**
Nina Hallowell, *S. Badger, S. Richardson, R. Fitzgerald, C. Caldas, J. Lawton;*
Edinburgh, United Kingdom

14:45 **EPL1.2 Impact of rapid genetic counselling and testing on primary surgery and psychosocial well-being in newly diagnosed breast cancer patients: Findings from a randomized controlled trial**
Marijke R. Wevers, *M.G.E.M. Ausems, E.M.A. Bleiker, E.J.T. Rutgers, A.J. Witkamp, D.E.E. Hahn, T. Brouwer, R.B. van der Luijt, F.B.L. Hogervorst, T. van Dalen, E.B. Theunissen, B. van Ooijen, M.A.J. de Roos, P.J. Borgstein, B.C. Vrouenraets, E. Vriens, W.H. Bouma, H. Rijna, J.P. Vente, S. Verhoef, N.K. Aaronson;*
Amsterdam, Netherlands

15:00 **EPL1.3 Disclosure of psychosocial research results: a randomized study among GENEPSO-Ψ cohort participants**
Julien Mancini, *E. Le Cozannet, N. Resseguier, E. Bureau, A.D. Bouhnik, C. Lasset, E. Mouret-Fourme, C. Noguès, C. Julian-Reynier;*
Marseille, France

15:15 **EPL1.4 Prevalence and detection of psychosocial problems in cancer genetic counseling**
W. Eijzenga, ***Eveline M. Bleiker***, *D.E.E. Hahn, L.E. van der Kolk, G.N. Sidharta, N.K. Aaronson;*
Amsterdam, Netherlands

15:30 **EPL1.5 Developing a group programme for BRCA1/2 mutation carriers who underwent prophylactic mastectomy**
Mariska den Heijer, *J. Gopie, A. Tibben;*
Rotterdam, Netherlands

16:00 - 16:30 *Coffee break*

16.30 - 18:00 **EPL2 - EMPAG Plenary Session: Reproductive decision making** **Room Amber 3+4**
Chair: L. Godino , S. Riedijk

16:30 **EPL2.1 Ok for us, not for them: Patients and genetic counsellors' experiences of NIPT and views on wider use**
Angela Effa, *E. Alexander, S.E. Kelly, L. Kerzin-Storrar;*
Manchester, United Kingdom

16:45 **EPL2.2 Non-invasive prenatal testing (NIPT): opinions and interest among pregnant women in a country with relative low uptake of prenatal screening**
Rachel V. van Schendel, *D.R.M. Timmermans, W.J. Dondorp, E. Pajkrt, J.H. Kleinveld, L. Henneman;*
Amsterdam, Netherlands

17:00 **EPL2.3 Received information and knowledge about Down syndrome among pregnant women and their partners coming for a first trimester combined (CUB) test? - Do they have the knowledge to make the decision**
Charlotta Ingvoldstad, *E. Ternby, G. Annerén, P. Lindgren, O. Axelsson;*
Solna, Sweden

17:15 **EPL2.4 Diagnosis Down syndrome: a cross-cultural study of family experiences**
Marcia L. Van Riper;
Chapel Hill, United States

17:30 **EPL2.5 Dynamics of prenatal screening: blurring boundaries between normative frameworks**
Wybo Dondorp, *G. De Wert;*
Maastricht, Netherlands

17:45 **EPL2.6 Stigma and reproduction: the place of stigma in reproductive decisions**
Angus J. Clarke;
Cardiff, United Kingdom

18:00 - 18:30 *Coffee break*

PROGRAMME EMPAG 2014 - SUNDAY

18.30 - 20:00 **EPL3 - EMPAG Plenary Session: Genomic testing: psychosocial and ethical issues** **Room Amber 3+4**
Chair: R. Moldovan, C. Bjorvatn

18:30 **EPL3. How do research participants perceive "uncertainty" in genomic sequencing?**
Barbara B. Biesecker, *W. Klein, L.G. Biesecker, P.K. Han;*
Bethesda, United States

18:45 **EPL3.2 Discussing clinical utility; The role of patients and their families**
Simone van der Burg, *L. Krabbenborg;*
Nijmegen, Netherlands

19:00 **EPL3.3 Variants in Practice Study (VIP): High risk women's responses to receiving genetic test results for genomic variants associated with breast cancer risk**
Mary-Anne Young, *P. James, G. Mitchell, L. Forrest, S. Sawyer, N. Hallowell;*
Melbourne, Victoria, Australia

19:15 **EPL3.4 To Disclose, or Not to Disclose? The Context Matters**
Vasiliki Rahimzadeh, *D. Avard, K. Sénécal, B.M. Knoppers, D. Sinnett;*
Montreal, Canada

19:30 **EPL3.5 Comparing the views of Australian parents, paediatricians and genetic health professionals about disclosure of genomic results**
Erin Turbitt, *J. Halliday, D. Amor, S. Metcalfe;*
Parkville, Australia

19:45 **EPL3.6 The experiences and views of health care professionals and researchers regarding the feedback of results in the context of next generation sequencing in oncology**
H. Howard, A. Mahalatchimy, ***Alexandra Soulier***, *A. Blassime, A. Cambon-Thomsen;*
Toulouse, France

20:00 - 21:30 *Networking Mixer at the MiCo Convention Centre*

Sunday, June 1, 2014

08:30 - 10:00 **EMPAG EES1 - EMPAG Educational Session: Responding to guilt and shame** **Room Amber 3+4**
Chair: E. Razzaboni

EES1.1 Responding to guilt and shame in clinical consultations
Clare Baguley;
Manchester, United Kingdom

10:00 - 10:30 *Coffee Break, Free Poster Viewing, Exhibition*

10:30 - 11:30 **Poster Viewing with Authors (poster numbers ending with „S")**

11:30 - 12:15 **EPL4 - EMPAG Plenary Session: Family Dynamics** **Room Amber 3+4**
Chair: M. Franiuk, N. Hallowell

11:30 **EPL4.1 Parental influences on decision making in Duchenne/Becker clinical trials**
Holly L. Peay, *B.B. Biesecker, J.V. Bowie, H. Scharff, K. Nagaraju, J. Piacentino, A. Tibben;*
Richmond, United States

11:45 **EPL4.2 The impact on children and parents of participation in clinical research trials for Morquio A syndrome and Sanfilippo A syndrome**
Deborah L. Holliday, *M. Farag, C. Breen, S. Jones, T. Clancy;*
Leeds, United Kingdom

12:00 **EPL4.3 Why do parents request carrier testing in their healthy children? A comparison of genetic health professionals' and parents' views**
Danya F. Vears, *C. Delany, J. Massie, L. Gillam;*
Parkville, Australia

12:15 - 13:30 *Lunch, Free Poster Viewing, Exhibition*

PROGRAMME EMPAG 2014 - SUNDAY

13:30 - 15:00 **EPL5 - EMPAG Plenary Session: Access to genetic services and testing** **Room Amber 3+4**
Chair: I. Blanco, M. Cornel

13:30 **EPL5.1 What is the role of genetic counsellors? A systematic review of evidence**
Heather Skirton*, C. Cordier, C. Ingvoldstad, N. Taris, C. Benjamin;*
Plymouth, United Kingdom

13:45 **EPL5.2 Referral for breast cancer genetic counseling among Turkish and Moroccan patients in the Netherlands**
Jessica E. Baars*, A.M. van Dulmen, M. Velthuizen, E.B.M. Theunissen, B.C. Vrouenraets, A.N. Kimmings, T. van Dalen, B. van Ooijen, A.J. Witkamp, M.G.E.M. Ausems;*
Utrecht, Netherlands

14:00 **EPL5.3 Genetic counselling for Indigenous populations: an exploratory study from the perspective of Australian genetic health professionals**
Lyndon Gallacher*, M. Sahhar, I. Macciocca, E. Kowal;*
Oxford, United Kingdom

14:15 **EPL5.4 Attitudes toward consumer-targeted genetic testing in Japan**
Kaori Muto*, A. Nagai, H. Hong, Z. Yamagata;*
Tokyo, Japan

14:30 **EPL5.5 Predictors of adverse psychological reactions to receipt of direct-to-consumer genome-wide profiling results**
K.M. Broady, ***Kelly E. Ormond****, N.J. Schork, E.J. Topol, A.J. Butte, C.S. Bloss;*
Stanford, United States

15:00 **EPL5.6 “It is a very lonely path”: Exploring experiences of establishing a genetic support group in Victoria, Australia**
Louisa Di Pietro*, E. Swain, L. Forrest, M. Sahhar;*
Parkville, Melbourne, Australia

15:00 - 15:30 *Vitamin Break*

15:30 - 17:00 **EPL6 - EMPAG Plenary Session: Facilitating communication about genetic information** **Room Amber 3+4**
Chair: T. Clancy

15:30 **EPL6.1 Co-designing an Intervention to facilitate family communication about inherited genetic conditions (IGC).**
Emma Rowland*, S. Hutchison, C. Jackson, L. Longworth, M. McAllister, R. Macleod, C. Patch, F. Ulph, A. Metcalfe;*
London, United Kingdom

15:45 **EPL6.2 A randomised controlled trial of a genetic counselling intervention to enhance family communication - the GIF study**
Jane L. Halliday*, S.A. Metcalfe, M. Aitken, S.M. Donath, C.L. Gaff, I.M. Winship, M.B. Delatycki, L.L. Skene, J.M. Hodgson; Parkville, Australia*

16:00 **EPL6.3 “What would you like to know?” Patients‘ attitudes towards communication of incidental findings emerging from new sequencing technologies**
Lea Godino*, G. Rodella, G. Severi, M. Mirra, G. Lanzoni, M. Romagnoli, A. Tranchina, G. Tortora, C. Graziano, A. Wischmeijer, M. Seri, D. Turchetti;*
Bologna, Italy

16:15 **EPL6.4 Genomic investigations: health care professional (HCP) and family experiences of managing incidental information in clinical practice**
Gillian Crawford*, A. Fenwick, A. Lucassen;*
Southampton, United Kingdom

16:30 **EPL6.5 „Very often the answer‘s not black or white“: Exploring communication in paediatric clinical genetic consultations**
Jean Paul*, S. Metcalfe, L. Stirling, J. Hodgson;*
Melbourne, Australia

16:45 **EPL6.6 Communicating oncogenetic information: do gastroenterologists and surgeons discuss heredity with their patients and, if so, what and how?**
K.F.L. Douma, E. Dekker, E.M.A. Smets, ***Cora M. Aalfs****;*
Amsterdam, Netherlands

17:00 - 17:30 *Coffee Break, Free Poster Viewing, Exhibition*

PROGRAMME EMPAG 2014 - MONDAY

17:30 - 19:00 **S06. Risk perception and risk communication, joint with ESHG** **Gold Room**
Chair: B. Dallapiccola, T. Clancy

17:30 **S06.1 Risk is more than a number: About risks and probabilities and people‘s preceptions of genetic risks**
Danielle Timmermans*, Amsterdam, The Netherlands*

18:00 **S06.2 Risk perception: What could be at stake in multiple genetic testing?**
Claire Julian-Reynier*, France*

18:30 **S06.3 Methods of communicating complex statistical information**
Angie Fagerlin*, United States*

Monday, June 2, 2014

08:30 - 10:00 **S13. Non-invasive prenatal testing, joint with ESHG** **Brown 3**

8:30 **S13.1 State of the Art of Non-Invasive Prenatal Testing,**
Lyn S. Chitty*, United Kingdom*

9:00 **S13.2 Noninvasive prenatal testing creates an opportunity for antenatal treatment of Down syndrome**
Diana W. Bianchi*, United States*

9:30 **S13.3 Clinical and social implications of NIPT**
Kelly Ormond*, United States*

10:00 - 10:30 *Coffee break*

10:30 - 11:30 **Poster Viewing with Authors (poster numbers ending with „M“)**

11:30 - 12:15 **EPL7 - EMPAG Plenary Session: Autonomy and consent** **Room Amber 3+4**
Chair: A. L Bredenoord

11:30 **EPL7.1 Consent and confidentiality in clinical genetics: a qualitative study**
Sandi Dheensa*, A. Fenwick, A. Lucassen;*
Southampton, United Kingdom

11:45 **EPL7.2 Autonomy and emotions: Professional challenges in seeking consent to genetic testing**
Hannah E. Shipman*, A.J. Clarke;*
Cardiff, United Kingdom

12:00 **EPL7.3 Randomized controlled trial of a telephone-based peer support program for female carriers of a BRCA1 or BRCA2 mutation: Impact on psychological distress**
Bettina Meiser*, V. White, M. Young, A. Farrelly, M. Jefford, S. Ieropoli, J. Duffy, I. Winship;*
Randwick, Australia
Bettina Meiser, *V. White, M. Young, A. Farrelly, M. Jefford, S. Ieropoli, J. Duffy, I. Winship;*
Randwick, Australia

12:15 - 13:30 *Lunch, Free Poster Viewing, Exhibition*

PROGRAMME EMPAG 2014 - MONDAY

13:30 - 15:00 **EPL8 - EMPAG Plenary Session: Psychosocial issues in prenatal & preimplantation diagnosis** **Room Amber 3+4**
Chair: L. Henneman, N. Hallowell

13:30 **EPL8.1 Women's experiences following a prenatal diagnosis of fetal abnormality: The PeTALS project**
Jan M. Hodgson*, M.A. Menezes, S.A. Metcalfe, J.L. Halliday, J. Fisher, K. Petersen, C. Hickerton, B.J. McClaren;*
Melbourne, Australia

13:45 **EPL8.2 Experiences of young Huntington's disease carriers and their partners soliciting a prenatal and/or pre-implantation genetic diagnosis: a qualitative study**
Ariane J. Van Tongerloo*, A.M. De Paepe;*
Ghent, Belgium

14:00 **EPL8.3 Difficult decisions in prenatal diagnosis - patients' experiences of decision-making under uncertainty, and the implications for expanding the offer of prenatal testing.**
Samantha Leonard*;*
Bristol, United Kingdom

14:15 **EPL8.4 Offering a choice between 5 Mb and 0.5 Mb prenatal whole genome SNP array analysis: are pregnant couples able of making informed decisions?**
Sanne L. van der Steen*, K.E.M. Diderich, S.R. Riedijk, J. Verhagen-Visser, L.C.P. Govaerts, M. Joosten, M.F.C.M. Knapen, F.A.T. de Vries, D. Van Opstal, M.I. Srebniak, A. Tibben, R.H. Galjaard;*
Rotterdam, Netherlands

14:30 **EPL8.5 SNP Array in prenatal diagnosis; first impressions on the psychological impact of receiving a susceptibility locus s a test result**
Judith Visser*, L.P.C. Govaerts, S.L. van der Steen, K.E.M. Diderich, M. Joosten, M.F.C.M. Knapen, F.A.T. de Vries, D. Van Opstal, M.I. Srebniak, A. Tibben, S.R. Riedijk, R.H. Galjaard;*
Rotterdam, Netherlands

14:45 **EPL8.6 Professional views about prenatal aCGH-testing**
Shiri Shkedi-Rafid*, A. Fenwick, D. Wellesley, A.M. Lucassen;*
Southampton, United Kingdom

15:00 - 15:30 *Vitamin break*

15:30 - 17:00 **EPL9 - EMPAG Plenary Session: Lessons learned and new issues in predictive testing** **Room Amber 3+4**
Chair: T. Clancy, F. Forzano

15:30 **EPL9.1 Predictive testing for Huntington Disease: Lessons learned from 24 years' experience**
Fiona H. Richards*, M.J. Wilson;*
Westmead, Australia

15:45 **EPL9.2 Patient views on the delivery of predictive test counselling services for Huntington's Disease**
Mary E. Jones*, R. MacLeod;*
Manchester, United Kingdom

16:00 **EPL9.3 Quality issues in genetic counselling practice for presymptomatic testing: a European Delphi study**
Milena Paneque*, J. Sequeiros, H. Skirton;*
Porto, Portugal

16:15 **EPL9.4 Experiences and implications of young women undergoing predictive BRCA testing under the age of 30**
Kate Brunstrom*, A. Murray, M. McAllister;*
Cardiff, United Kingdom

16:30 **EPL9.5 The experiences of BRCA1/2 mutation positive women in Northern Norway**
Nina Strømsvik*, M. Myklebust, E. Gjengedal;*
Tromsø, Norway

17:00 **EPL9.6 Genetic test declining and high personal colorectal cancer risk perception in DNA mismatch repair gene mutation families**
Louisa Flander*, A. Ugoni, L. Keogh, H. Niven, A. Rutstein, A. Ko Win, D. Ait Ouakrim, C. Gaff, M. Jenkins, I. Winship; Parkville, Australia*

17:00 - 17:30 *Coffee Break*

PROGRAMME EMPAG 2014 - TUESDAY

17:30 - 19:00 **EES2 - EMPAG Educational Session: Qualitative and quantitative methods in psychosocial research** **Room Amber 3+4**
Chair: B. Ignacio, C. Bjorvatn

EES2.1 Qualitative and quantitative methods in psychosocial research
K. O'Doherty,
Guelph, Canada

***Bettina Meiser**;*
Randwick, Australia

20:30 *Networking party*

Tuesday, June 3, 2014

09:00 - 10:30 **ESHG-ASHG Building Bridges Session PL3:** **Gold Room**
„Towards finding global agreement on..."
What IF... (Incidental Findings), an interactive Debate - joint with ESHG

Moderator: **Han Brunner, The Netherlands**

Discussants:

- ***Angus Clarke**, United Kingdom*
- ***Martina Cornel**, The Netherlands*
- ***Robert Green**, United States*
- ***Stephen Kingsmore**, United States*
- ***Marjolijn Kriek**, The Netherlands*
- ***Arnold Munnich**, France*

10:30 - 11:00 *Coffee break*

11:00 - 12:30 **C22 - Returning results: Ethical and legal issues, joint with ESHG** **Space 1**
Chair: F. Faravelli, M. Cornel

11:00 **C22.1 The impact of reporting exome and whole genome sequencing: Predicted frequencies of primary, secondary and incidental findings based on modelling**
***Leslie Burnett**, L.C. Ding, R.M. Lew, D. Chesher, A.L. Proos;*
Sydney, Australia

11:15 **C22.2 Defending the child's right to an open future concerning genetic information.**
***Annelien L. Bredenoord**, M.C. de Vries, J.J. van Delden;*
Utrecht, Netherlands

11:30 **C22.3 Implementation of a duty-to-recontact system in molecular and clinical genetics: perspectives from professionals and patients**
***Mirjam Plantinga**, W. Lamers, A.V. Ranchor, M.A. Verkerk, E. Birnie, I.M. van Langen;*
Groningen, Netherlands

11:45 **C22.4 International views on sharing incidental findings from whole genome research**
***Anna Middleton**, M. Parker, C. Wright, H. Firth, E. Bragin, M. Hurles, O. DDD Project;*
Cambridge, United Kingdom

12:00 **C22.5 Newborn screenings and whole genome sequencing: the real need of a genuine public involvement**
***Marta Tomasi**, A. Santosuosso;*
Trento, Italy

12:15 **C22.6 Current Developments in the Regulation of Direct-to-Consumer Genetic Testing in Europe**
***Louiza M. Kalokairinou**, H.C. Howard, P. Borry;*
Leuven, Belgium

End of Meeting

GENERAL INFORMATION

INFORMATION

GENERAL INFORMATION

REGISTRATION FEES

NETWORKING EVENTS

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). Chairpersons are allowed to exclude from the session, persons who will not observe this rule.

Also note that Monday, June 2 is a public holiday in Italy.

Conference Venue

MiCo - Milano Congressi
Gate 2 - South Wing
Viale Eginardo
20149 Milan
Italy
www.micmilano.it

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 between 8.00/8.30 to 13.00/13.30 and 14.30/15.00 to 16.00/16.30 hrs and are closed over the weekend. Some branches are open from 9.00-12.00 hrs on Saturdays. There are multiple bank machines (ATMs) open 24 hours a day throughout the city which accept all major international bankcards. The official currency of Italy is the Euro (€). 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, 2014: 75% refund
- between April 1 and May 9, 2014: 25% refund
- after May 9, 2014: no refund

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

Car Parking

From any of the ring roads circling Milan follow the signs to Fieramilanocity, or to any of the large Park & Ride car parks located close to these Metro stops: Cascina Gobba (Green Line), San Donato (Yellow Line), Famagosta (Green Line), Bisceglie (Red Line), Lampugnano (Red Line).

Certificate of Attendance

Certificates of attendance will be issued at the registration desk.

Climate

In June, the weather in Milan is generally nice (average 20°C, high: 26°C, average low: 13°C). Please make sure to protect yourself from possible sunburns. Milan has an average rainfall of 93 mm over 12 days in June.

Cloakroom and Luggage

A cloakroom and luggage storage are available in the registration area.

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 **20 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.

GENERAL INFORMATION

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 boxes will be served in the exhibition area, on Tuesday on Levels 1 and 2 (the exhibition is closed on Tuesday).

Conference App

Download the ***ESHG 2014 Conference App*** for iOS and Android from iTunes App Store and Google Play Store.

Currency

The official currency of Italy is the Euro (€). 1 EUR = 1,38 USD = 0,82 GBP = 1,51 CAD = 141 JPY = 1,22 CHF = 1,48 AUD as per April 29, 2014. Other currencies.

Drinking water

The tap water in Milan can be used without concern.

Eating Out in Milan

Milan is surely one of the gastronomic capitals of Italy, but like in every city heavily frequented by tourists, having an excellent meal for a fair price is just as easily possible, as getting mediocre food for not so little money, depending on the choice of location.
Guide to eating in Milan:
http://www.cntraveller.com/guides/europe/italy/milan/where-to-eat
http://www.hostelworld.com/guides/city-guides/milan/italy/eating-out
http://www.lonelyplanet.com/italy/milan/restaurants
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

220-240 V - 50Hz AC, using CEI 23-50, CEI 23-5, some (older) sockets will not accept CEE 7/7 plugs, however in modern installations multiple standard sockets have been used.

Emergency Services

European Emergency Number: 112. (Alternatively, Ambulance – 118; Fire – 115; State Police – 113; Carabinieri – 112; 911 is redirected to 112.)

Exhibition Opening Hours

Saturday, May 31:	08.30 - 18.30 hrs
Sunday, June 1:	08.00 - 17.30 hrs
Monday, June 2:	08.00 - 17.30 hrs
Tuesday, June 3:	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 Italy offers the cheapest tariffs.

Insurance

By registering to the ESHG 2014 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

Internet (WiFi) access and terminals with printing facilities are available at the venue.

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 at the coffee points in the exhibition. A cash bar is also available in the exhibition area.

INFORMATION GENERAL INFORMATION

Message Board

Message Boards are available on the balcony on Level 1.

Pharmacy

Most pharmacies are open during normal trading hours, a rotational service is in place. The following pharmacies are open 24/7: Stazione Centrale - Gallerie Partenze, Phone: 02 6690735; Piazza Duomo 21 (corner Via S. Pellico), Phone: 02 878668.

Poster Removal

The organisers cannot assume any liability for loss or damage of posters displayed in the poster area. Posters that will not have been removed by Monday, June 2, 2014, 17.30 hrs, will be removed by the staff and will not be kept or mailed to the author after the meeting.

Preview Centre

Equipment for a final check of the sequence of your presentation is available in the preview centre on **Level 2**. All presenters should bring their electronic presentation to the preview room preferably in the morning of the day of the talk, but not later than 2 hours before the start of the session (30 minutes for the first morning sessions).

Registration Desk opening hours

Saturday, May 31:	08.00 - 20.00 hrs
Sunday, June 1:	08.00 - 19.00 hrs
Monday, June 2:	08.00 - 19.00 hrs
Tuesday, June 3:	08.30 - 15.45 hrs

Safety - Crime

Milan can be considered safe compared to other cities of the same size. Use of common sense is however 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

Most shops are open from 9.30-12.30 and 15.30-19.30 hrs, from Tuesday to Sunday. Bigger (and department) stores stay open all day. Most shops close on Sunday and re-open on Monday afternoon from about 15.30-19.30, except food stores, which re-open on Monday morning but close again for the afternoon. All major credit cards are generally accepted, but it is not possible to pay with foreign banknotes.

Smoking Policy

The ESHG 2014 is officially a “No-smoking-Conference”. Note that smoking is banned in public buildings, restaurants and bars.

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

As in most Italian cities, taxis in Milan are not cheap. Fares are at a fixed price of EUR 3,20 per pick-up on weekdays, 5,20 on Sunday, 6,20 after 21.00 hrs and on public holidays, *plus* EUR 1.06 per kilometre. The meter should only be started as you set off. Round up the tip to the nearest euro. Taxis cannot be hailed in the street. There are ranks (a white sign marked with a black ‘TAXI’) at Piazza del Duomo, Teatro La Scala and Castello Sforzesco, outside all airport terminals and at the train stations. Taxis will also be waiting at the entrance of the conference centre at Gate 2. Most taxi drivers speak a little English and are usually only too happy to make recommendations of sights, shops and restaurants. If you speak to them in Italian, they will rapidly become your new best friend.
Avoid bogus taxi drivers at the airports; they often over-charge by as much as 600 percent. Always go to the ranks outside the terminals. Licensed and metered taxis are white with yellow and black signs on top.

INFORMATION GENERAL INFORMATION

Telephone calls

The country code of Italy is 39 and the area code for Milan is 02. If calling a number in Milan from within Italy (including Milan!), dial 02 before the subscriber number.

Tipping

Tipping is quite flexible in Milan as the ‘coperto’ (cover/service charge) is automatically added in the bill. However, if you are happy with the service then tipping the staff is acceptable.Taxi drivers, theatre and cinema usherettes, luggage handlers are also given a token amount as a tip for their services, but you are not compelled to do so.

Tourist Information Centres

The Milan Tourism Office will have a desk in the registration area of the conference centre.
Website: http://www.tourism.milan.it

Travelling - Accessibility - Public Transportation

Homepage of the Milan Public Transports: http://www.atm.it
How to reach MiCo by:

Buses & Trams

- For Gate 2 „Viale Eginardo / Viale Scarampo“ entrance:
  Bus No. 78 – „Eginardo/Colleoni“ stop
- For Gate 17 „Piazzale Carlo Magno / Via Gattamelata“ entrance:
  Bus no. 78 – get off at „Colleoni/Gattamelata“
  or
  Tram no. 27 – get off at „Piazza 6 Febbraio“

Metro

Red Line 1:

For Gate 2 - „Viale Eginardo / Viale Scarampo“ entrance: get off at the “Amendola” stop – 700 m from the Congress Centre, or at “Lotto” approx. 800 m.

For Gate 17 - „Piazzale Carlo Magno / Via Gattamelata“ entrance: get off at the “Cadorna” stop, exit the subway and go to the railroad station above : take the first train departing and get off at the “Domodossola” stop – just 600 m from the Congress Centre

V.A.T.

The VAT rate is 22%, 10% on food.

Download the new
ESHG 2014 Conference App
for iOS and Android
devices from the
iTunes App Store or
Google Play Store

INFORMATION REGISTRATION FEES

Registration fees Payment received:	***before March 31, 2014 (reduced rate)***	***between March 31 & May 9, 2014 (normal rate)***	***after May 9, 2014 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/termsandconditions2014.0.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, May 31 to Tuesday, June 3

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 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 9), 2014 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, 2014: 75% refund
- Between April 1 and May 9, 2014: 25% refund
- After May 23, 2014: 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, May 31, 2014, 20.00 - 21.30 hrs - MiCo (conference venue)

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.

ESHG Networking Party

Monday, June 2, 2014, 20.30 hrs - Old Fashion Club

Join us for a party evening at “The Old Fashion Club” in down town Milan with dancing, a live band and DJ entertainment.

Address: Viale Alemagna, 6, 20121 Milan (the club is located in a part of the Triennale Building in Parco Sempione)

Directions: Take metro no. 1 (red line) get o at station „Cadorna“. Walk accross Piazzale Luigi Cadorna, turn left to Via Pietro Paleocapa. Walk along the park until you reach the Triennale di Milano Building. Walk past it and turn right at the corner of the building and walk until you reach the Old Fashion Club entry.

Entrance fees include finger food, non alcoholic drinks, beer and wine, live and DJ music. Cocktails and liquors are avaible at cost.

Ticket: EUR 49.-
Students: EUR 29.-
Please note that a only limited number of tickets can be purchased on a first-com-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!

INFORMATION EXHIBITION

Exhibition Organiser

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 – Level 0 – South Wing – Dates & Opening Hours

Saturday, May 31, 2014	08.30 – 18.30 hrs
Sunday, June 1, 2014	08.00 – 17.30 hrs
Monday, June 2, 2014	08.00 – 17.30 hrs
Tuesday, June 3, 2014	closed

Posters – Mounting, Viewing & Removal Schedules

Poster presentations will be held in the exhibition hall from May 31 – June 2. Poster mounting, viewing and removal times are:

Saturday, May 31, 2014	08:30 – 18.30 hrs	Poster mounting / viewing
Sunday, June 1, 2014	08.00 – 17.30 hrs	Poster viewing
Monday, June 2, 2014	08.00 – 17.30 hrs	Poster viewing
Monday, June 3, 2014	13.30 – 17.30 hrs	Poster removal

Please note that posters not removed by 17.30 hrs on Monday June 2, will be taken down by the staff of the conference centre and will not be stored or sent to the authors after the meeting.

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 2014.

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 can also be found on pages 46-51.

Lead Retrieval System used by Exhibitors

A growing number of exhibitors will be using a so-called Lead Retrieval System on their stands.
Note the following please:

- exhibitors who use the device will ask permission to scan the barcode on your badge
- this barcode gives this exhibitor access to your contact details as follows:
    - name and full postal address
    - e-mail address

Thank you for your understanding and cooperation.