THE EUROPEAN SOCIETY
OF HUMAN GENETICS

EUROPEAN HUMAN GENETICS CONFERENCE 2013

June 8 - 11, Paris, France

Final Programme

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GENERAL WELCOMING ADDRESS

Dear Colleagues,
Dear Friends,

For the third time in the 46 years of existence of the ESHG, in June 2013, the European Human Genetics Conference has returned to Paris. On behalf of the French and the European Societies of Human Genetics, I am honoured to welcome you in our city of lights.

The history of Paris dates back to more than 2000 years ago when it was a small village on the islands of the river Seine. The river is indeed the mother of Paris and Parisians, and has witnessed their glorious history and culture. Paris, now a city of world importance as the historical, political, intellectual and artistic centre of France, also has long-standing scientific and educational traditions. This is why Paris is proud to host a conference that, I am confident, will continue in the successful series of excellent ESHG meetings covering the latest developments in the field of human genetics.

Since we are witnessing an unprecedented time of technological and scientific evolutions, I am sure that the sessions and symposia will be filled with exciting and up-to-date talks, educational lectures and distinguished speakers, hopefully making the conference a success from both the scientific as well as the social points of view.

I sincerely hope that you will enjoy your visit to Paris and a meeting that we wish to be of interest for both clinicians and research scientists.

Bienvenue à Paris!

With my best regards … et toute mon amitié!

Stanislas Lyonnet

President of the ESHG

GENERAL COMMITTEES-BOARD-ORGANISATION

European Society of Human Genetics

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

European Human Genetics Conference 2013

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):

- AAAS/Science
- Abbott Molecular
- Affymetrix
- Agilent Technologies
- BIOBASE
- BioDiscovery
- Bio-Rad Laboratories
- Cartagenia
- Circulation: Cardiovascular Genetics – An American Heart Association Journal
- CLC bio
- DNA Genotek
- Duzen Laboratories Group
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- Emory Genetics Laboratory
- Fluidigm
- Illumina
- IntegraGen
- Integrated DNA Technologies
- Life Technologies
- Multiplicom
- Myriad Genetics
- Natera
- Nature Publishing Group
- PerkinElmer
- Roche Applied Science
- Theradiag

Future European Human Genetics Conferences

European Human Genetics Conference 2014, joint with EMPAG 2014
Milano, Italy
May 31 – June 3, 2014

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

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 2013 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.

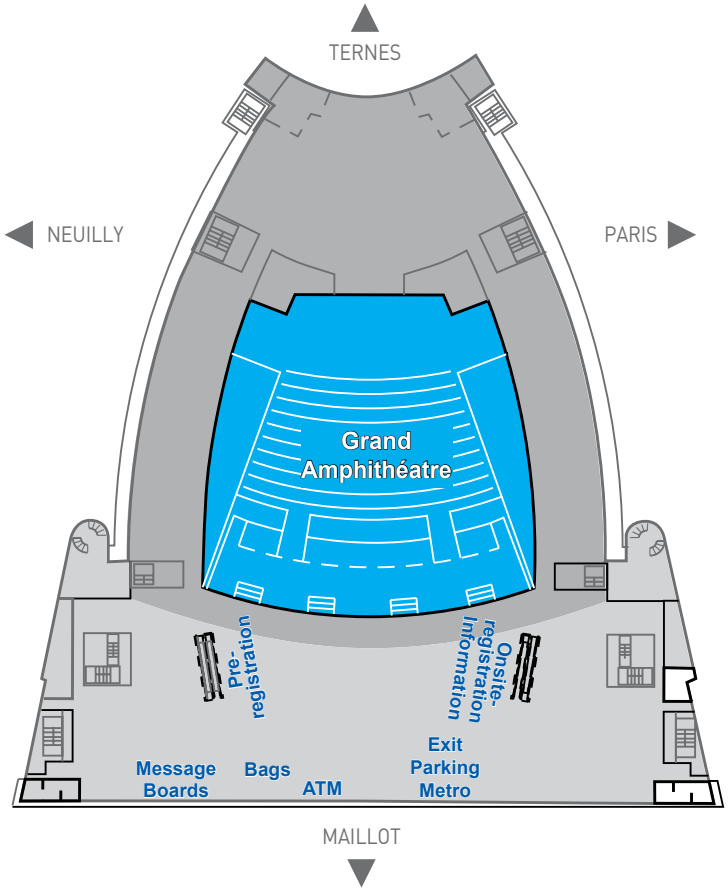
IMPORTANT NOTICE

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

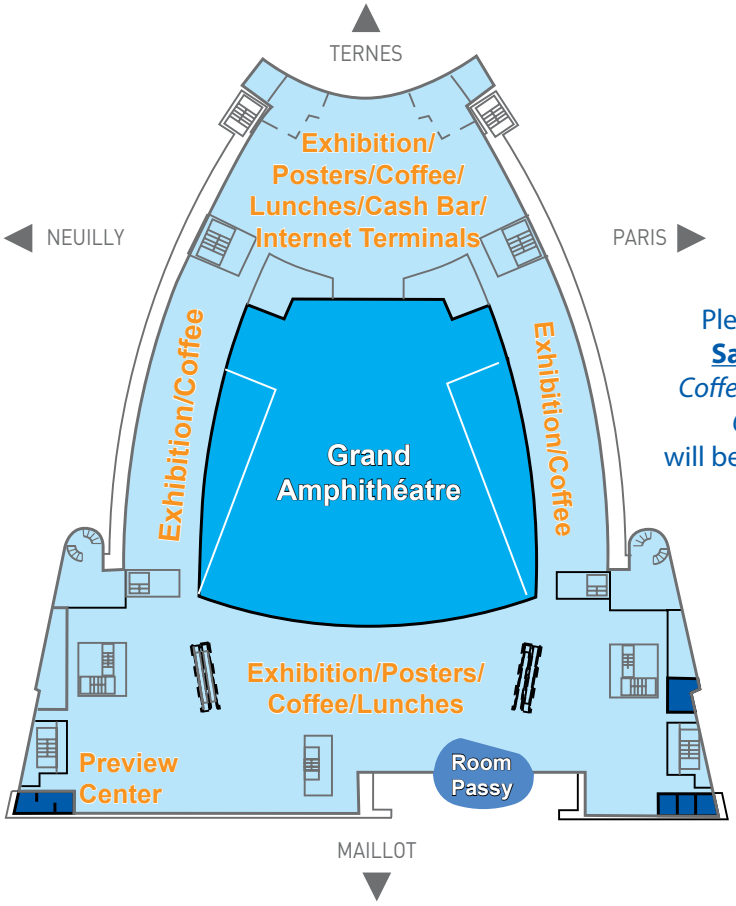
Level 0

Registration Area
Grand Amphithéatre
Exit
Parking (-1)
Metro Station (-1)



Level 1

Grand Amphithéatre
Room Passy
Exhibition
Posters
Coffee
Lunches
Cash bar
Internet Café

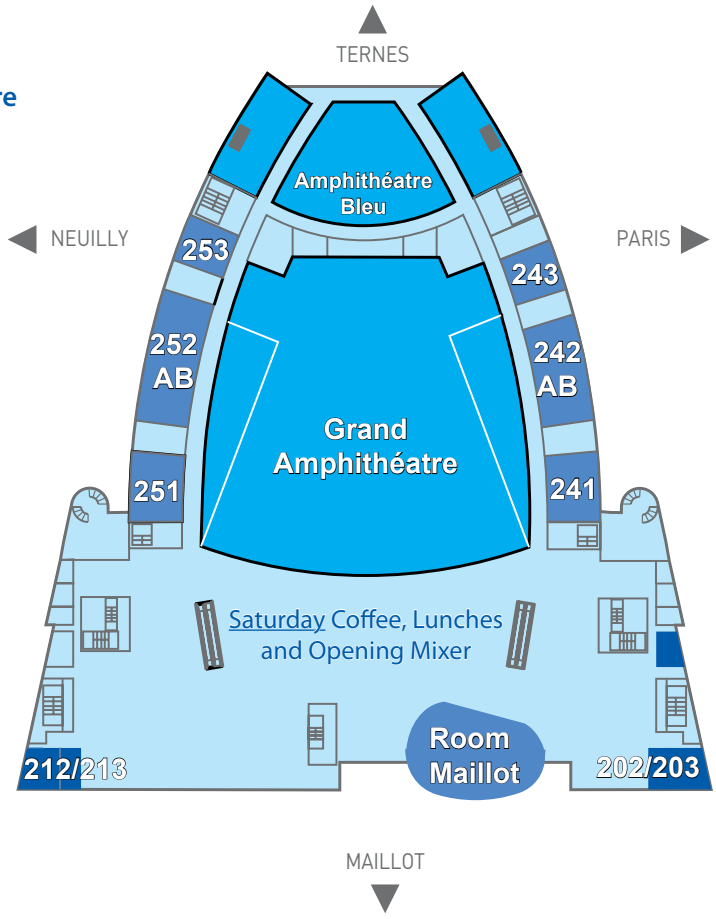


Please note that on **Saturday, June 8** *Coffee*, *Lunches* and the *Opening Mixer* will be located on **level 2**.

GENERAL FLOORPLAN

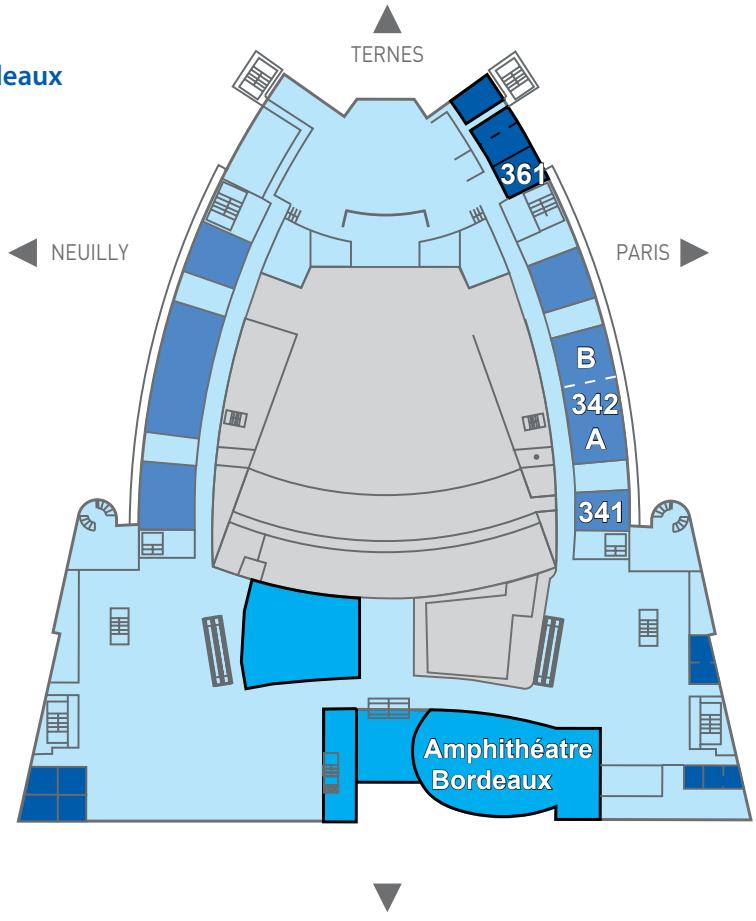
Level 2

Grand Amphithéatre
Amphithéatre Bleu
Rooms 202-253
Room Maillot



Level 3

Amphithéatre Bordeaux
Rooms 341-361



Saturday, June 8, 2013

Time	Grand Amphithéatre	Amphithéatre Bordeaux	Room Maillot	Room 251	Room 212/213	Room 202/203
11.45 – 13.15				**CS01** **Fluidigm Satellite**	**CS02** **INTEGRAGEN Satellite**	**CS03** **BIOBASE Satellite**
13.15 – 14.00	*Lunch Break* *Poster Mounting: 12.00-14.00 hrs (Strict)*					
14.00 – 15.30		**ES1** **Performance and future of next generation sequencing (what's next?)**	**ES2** **Prenatal and Preimplantation Genetic Screening**		**CS04** **Theradiag Satellite**	**CS05** **DNA Genotek Satellite**
15.30 – 16.00	*Coffee Break*					
16:00 – 16.30	**Opening** **Welcome Addresses**					
16.30 – 18.00	**PL1** **Opening Plenary Session**					
18.00 – 18.30	*Coffee Break*					
18.30 – 20.00	**PL2** **What's New?**					
20.00 – 21.30	*Opening Networking Mixer at the Palais des Congrès*					

Session Types:

Plenary Session	Symposium	Concurrent Session	Workshop	Educational Session	Corporate Satellite

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Sunday, June 9, 2013

Time	Grand Amphithéatre	Amphithéatre Bleu	Amphithéatre Bordeaux	Room Maillot	Room 252AB	Room 242AB	Room 243	Room 251	Room 241	Room 202/203
08.30 – 10.00	**S01** Chromatin organisation and gene expression	**S02** From genes to treatment in multifactorial diseases	**S03** Gene regulation in cancer	**S04** Emerging topics in neurobiology	**ES3** Tweeting about Genomics					
10.00 – 10.30	*Coffee break / Poster viewing / Exhibition*									
10.30 – 11.30	**Poster viewing with presenters** (odd poster numbers)									
11.40 – 13.10	**WS01** Next Generation Sequencing in clinical practice - Filtering & reporting	**WS02** Debate: Hot topics in Pre-implantation genetic testing	**ES4** Cancer risk in developmental syndromes	*Poster viewing / Lunch break / Exhibition*	**WS03** UCSC Genome Browser I	*Poster viewing / Lunch break / Exhibition*		**CS06** Affymetrix Satellite	**CS07** Life Technologies Satellite	**CS08** Myriad Genetics Satellite
13.15 – 14.45	**C01** Structural variation and de novo mutations	**C02** Genotype phenotype correlation	**C03** Prenatal diagnosis	**C04** Cancer predisposition	**C05** Functional Genomics	**C06** Neuromuscular disorders: From genes and modifiers to function and therapy	**C07** Metabolic and mitochondrial disorders			
14.45 – 15.15	*Vitamin break / Poster viewing / Exhibition*									
15.15 – 16.45	**WS04** Dysmor-phology Workshop 1	**WS05** Analysis, interpretation and reporting of array data	**WS06** Clinical Cancer Genetics Club	**WS07** Quality assurance	**WS08** Biomedical Data Analysis with Galaxy	**WS09** Inequalities in genetic services		**CS09** Roche Applied Science Satellite	**CS10** Cartagenia Satellite	**CS11** Multiplicom Satellite
16.45 – 17.15	*Coffee break / Poster viewing / Exhibition*									
17.15 – 18.45	**S05** Interpreting NGS data	**S06** Cancer genetics	**ES5** Epilepsies	**S07** Genetics of skin diseases and new therapies	**S08** Evolution of organs					
18.45								**CS12** Illumina Satellite	**CS13** PerkinElmer Satellite	
19.00 – 20.15				**ESHG** Membership Meeting						

Sesion Types

Plenary Session	Symposium	Concurrent Session	Workshop	Educational Session	Corporate Satellite

Monday, June 10, 2013

Time	Grand Amphithéatre	Amphithéatre Bleu	Amphithéatre Bordeaux	Room Maillot	Room 252AB	Room 242AB	Room 243	Room 251	Room 241	Room 202/203
08.30 – 10.00	**S09** New therapeutic highlights in rare diseases	**S10** Reproductive genetics and newborn screening	**S11** Animal Models for Human Diseases	**S12** Judging our genes	**ES6** Retinal dystrophies: news and views					
10.00 – 10.30	*Coffee break sponsored by Abbott Molecular / Poster viewing / Exhibition*									
10.30 – 11.30	**Poster viewing with presenters** (even poster numbers)									
11.40 – 13.10	**ES7** NGS in the clinic	**ES8** How to get published in the EJHG	*Poster viewing / Lunch break / Exhibition*					**CS14** Agilent Technologies Satellite	**CS15** Abbott Molecular Satellite	**CS16** CLC Bio Satellite
13.15 – 14.45	**C08** Intellectual disability: Gene discovery and dysfunction	**C09** Population-based sequencing	**C10** Genetic Counselling, Education and Public Services	**C11** Molecular mechanisms in tumorigenesis	**C12** Connective tissue disorders: Gene identification	**C13** Neuro-degenerative disorders: From gene discovery to therapy	**C14** Sensory disorders and Immunological disorders			
14.45 – 15.15	*Vitamin break / Poster viewing / Exhibition*									
15.15 – 16.45	**WS10** Dysmor-phology Workshop 2	**WS11** Prenatal diagnoses	**WS12** DNA Dysmorphology	**WS13** Community genetics	**WS14** UCSC Genome Browser II			**CS17** Roche Applied Science Satellite	**CS18** Life Technologies Satellite	**CS19** BioDiscovery Satellite
16.45 – 17.15	*Coffee break / Poster viewing / Exhibition*									
17.15 – 18.45	**S13** Stem cells and IPS cells in genetic diseases	**S14** Chromosomal (in)stability	**ES9** Where do we come from?	**S15** The interrelated world of drugs and genes	**S16** The expanding world of the primary cilia					
20.00	*Networking Party at the Museum of Natural History*									

Session Types:

Plenary Session	Symposium	Concurrent Session	Workshop	Educational Session	Corporate Satellite

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Tuesday, June 11, 2013

Time	Grand Amphithéatre	Amphithéatre Bleu	Amphithéatre Bordeaux	Room 342AB	Room 252AB	Room 251
09.00 – 10.30	**PL3** **Large Scale Cohorts Studies to Identify Novel Highly Penetrant Genetic Disease Causing Variants**					
10.30 – 12.00	*Coffee break / Poster viewing / Exhibition*					
11.00 – 12.30	**C15** **NGS-based diagnostics**	**C16** **Developmental syndromes**	**C17** **Basic mechanisms in cytogenetics and molecular genetics**	**C18** **Big GWAS**	**C19** **Internal organs and endocrinology: Gene identification and function**	**C20** **Neurodevelopmental and neuropsychiatric disorders**
12.30 – 13.30	*Lunch / Poster removal (12.30-13.30 hrs) / Exhibition*					
13.30 – 14.15	**PL4** **Mendel Lecture**					
14.15 – 15.45	**PL5** **Closing Plenary** **ESHG Award Lecture** **- ESHG Education Award** **- EJHG Awards** **- Young Investigator & Poster Awards** **- Closing**					

Session Types:

Plenary Session	Symposium	Concurrent Session	Workshop	Educational Session	Corporate Satellite

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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 – S16)

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

Educational Sessions (ES1 – ES9)

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

Concurrent Sessions (C01 – C20)

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.

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.
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 – WS14)

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

Corporate Satellites (CS01-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.

GENERAL BUSINESS AND ANCILLARY MEETINGS

As per date of printing.

Saturday, June 8, 2013

09.00 - 14.00	B8.1 ESHG Board Meeting I	342A	closed
10:00 - 13:00	B8.2 An International Consortium for the Ehlers-Danlos Syndrome	253	closed
13.00 - 15.45	B8.3 ESHG PPPC Meeting	341	closed

Sunday, June 9, 2013

10.00 - 11.00	B9.1 European Genetic Nurses and Counsellors Meeting	Passy	closed
10.00 - 11.30	B9.2 Human Variome Project special consultation for the WHO	341	open
10.00 - 11.40	B9.3 ESHG Genetic Services Quality Comittee	253	closed
11.15 - 13.15	B9.4 National Human Genetics Societies Meeting	342A	closed
12.30 - 14.30	B9.5 Elsevier Editorial Board Meeting	253	closed
15.15 - 16.15	B9.6 EJHG Editorial Board Meeting	341	closed
19.00 - 20.00	B9.7 ESHG Membership Meeting	Maillot	members only

Monday, June 10, 2013

10.00 - 12.00	B10.1 European Board of Medical Genetics	341	closed
10.15 - 12.15	B10.2 ESHG Education Committee	253	closed
10:30 - 12:30	B10.3 SENSGENE, French Network for Genetics in Sensorial Conditions	342A	closed
11.00 - 13.00	B10.4 UEMS Section Meeting	252AB	open
12.00 - 13.00	B10.5 Meeting of the Editorial Board of Human Heredity	341	closed
12.15 - 13.15	B10.6 ESHG Board Meeting II	Passy	closed
13.00 - 15.30	B10.7 Journal of Community Genetics Editorial Board Meeting	253	closed
14.30 - 17.30	B10.8 ENGAGE-European Network for Genetic&Genomic Epidemiology Project Meeting	341	closed

Tuesday, June 11, 2013

12:00 - 16:30	B11.1 COST Action BM1208 Steering committee meeting	361	closed
12.15 - 13.15	B11.2 ESHG SPC Meeting	341	closed
15.00 - 18.00	B11.3 How to reach European consensus on reporting unsolicited findings & unknown variation	341	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.

SCIENTIFIC

SCIENTIFIC PROGRAMME

Saturday, June 8, 2013

PROGRAMME

PROGRAMME SATURDAY, JUNE 8

Time	Amphithéatre Bordeaux	Room Maillot
14.00 - 15.30	**ES1.** **Performance and future of next generation sequencing (what's next?)**	**ES2.** **Prenatal and Preimplantation Genetic Screening**
	ES1.1 **Sequencing at The Wellcome Trust Sanger Institute in the year 2013** ***Michael Quail***; *Cambridge, United Kingdom*	**ES2.1** **Integration of Microarray Technology into Prenatal Diagnosis** ***Ron Wapner***; *New York, United States*
	ES1.2 **Performance and Improvements in Analyzing Next Generation Sequencing Technologies** *H. Tilgner, D. Sharon, D. Xie, V. Kuleshov, R. Chen, D. Pushkarev, K. Karczewski, A.P. Boyle, T. Blauwkamp, M. Kertesz, R. Chen, H. Lam, M. Pratt, G. Bartha, J. Harris, J. West,* ***Mike Snyder****;* *Stanford, United States*	**ES2.2** **Advances in embryo selection for optimizing IVF outcome** ***Santiago Munné***; *Livingston, United States*
15.30 - 16.00	*Coffee break* *NOTE: Saturday coffee breaks are located on* ***level 2*** *to be accessed via escalators or directly from the Grand Amphitheatre*	

PROGRAMME SATURDAY, JUNE 8

Time	Grand Amphithéatre
16.00 - 16.30	**Opening & Welcoming Addresses** Chair: Hélène Dollfus, Stan Lyonnet
	Welcoming Addresses by Stanislas Lyonnet *President of the ESHG* Hélène Dollfus *President of the French Society of Human Genetics, Local host* Heather Skirton *Chair of the European Board of Medical Genetics*
16.30 - 18.00	**Opening Plenary Session PL1** Chair: Hélène Dollfus, Stan Lyonnet
16.30	**P1.1** **Integrating chromosome structure and function during X-chromosome inactivation** ***Edith Heard***; *Paris, France*
17.00	**P1.2** **Signaling transcription factors explode dogmas in brain development and disease** ***Alain Prochiantz***; *Paris, France*
17.30	**P1.3** **Life-threatening infectious diseases of childhood: single-gene inborn errors of immunity?** ***Jean-Laurent Casanova***; *New York, United States*
18.00 - 18.30	*Coffee break* *NOTE: Saturday coffee breaks are located on **level 2** to be accessed via escalators or directly from the Grand Amphitheatre*
18.30 - 20.00	**Plenary Session PL2. What's new?** Chair: Hélène Dollfus, Brunhilde Wirth
18.30	**P2.1** **Mutations of TCF12, encoding a basic-helix-loop-helix partner of TWIST1, are a frequent cause of coronal craniosynostosis** ***Vikram P. Sharma****, A.L. Fenwick, M.S. Brockop, S.J. McGowan, J.A.C. Goos, A.J.M. Hoogeboom, A.F. Brady, O. Jeelani, S. Lynch, J.B. Mulliken, D.J. Murray, J.M. Phipps, E. Sweeney, S.E. Tomkins, L.C. Wilson, S. Bennett, R.J. Cornall, J. Broxholme, A. Kanapin, D. Johnson, S.A. Wall, P.J. van der Spek, I.M.J. Mathijssen, R.E. Maxson, S.R.F. Twigg, A.O.M. Wilkie; *Oxford, United Kingdom*
18.45	**P2.2** **C-terminal deletions of the *AUTS2* locus cause distinct syndromic features and cognitive impairment** ***Els Voorhoeve***, *G. Beunders, C. Golzio, L. Pardo, J. Rosenfeld, M. Talkowski, I. Simonic, A. Lionel, S. Vergult, R. Pyatt, J. van de Kamp, A. Nieuwint, M. Weiss, P. Rizzu, D. Posthuma, L. Verwer, H. Meijers-Heijboer, B. Menten, G. Mortier, S. Scherer, E. Eichler, S. Girirajan, N. Katsanis, A. Groffen, E. Sistermans;* *Amsterdam, Netherlands*
19.00	**P2.3** **MED4: a suicide gene to explain low penetrance in retinoblastoma patients** *C. Dehainault, A. Garancher, L. Castéra, I. Aerts, F. Doz, L. Lumbroso, R. Montes-de-Oca, G. Almouzni, D. Stoppa-Lyonnet, C. Pouponnot, M. Gauthier-Villars,* ***Claude Houdayer****;* *Paris, France*
19.15	**P2.4** **Van Maldergem syndrome is caused by defective cadherin receptor-ligand interactions leading to dysregulation of neuroprogenitor cell proliferation and differentiation** ***Stephen Robertson***, *S. Cappello, M. Gray, S. Lange, M. Einsiedler, I. Burtscher, Z. Jenkins, T. Morgan, N. Preitner, V. Morrison, N. DiDonato, L. van Maldergem, T. Neuhann, R. Newbury-Ecob, M. Swinkells, P. Terhal, L. Wilson, P. Zwijnenburg, A. Sutherland-Smith, D. Markie, M. Simpson, S. Mansour, M. Goetz;* *Dunedin, New Zealand*
19.30	**P2.5** **BMN111, a CNP analogue, potential novel investigational therapy for achondroplasia** ***Laurence Legeai-Mallet***, *N. Kaci, J. Peng, C. Benoist-Lasselin, T. Oppeneer, L. Tsuruda, C.A. O' Neill, F. Di Rocco, A. Munnich, F. Lorget;* *Paris, France*
19.45	**P2.6** **Sequencing-based GWAS on peripheral blood monocyte counts in the SardiNIA cohort** ***Maristella Steri***, *A. Mulas, M. Zoledziewska, C. Sidore, G. Pistis, F. Danjou, E. Porcu, M. Marongiu, F. Busonero, M.G. Piras, M. Lobina, F. Reinier, R. Berutti, M.F. Urru, A. Angius, C.M. Jones, D. Schlessinger, G.R. Abecasis, S. Sanna, F. Cucca;* *Monserrato, Cagliari, Italy*
20.00 - 21.30	**Opening Networking Mixer in the Palais des Congrès, Level 2**

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

SCIENTIFIC

SCIENTIFIC PROGRAMME

Sunday, June 9, 2013

PROGRAMME

PROGRAMME SUNDAY, JUNE 9

Time	Grand Amphithéatre	Amphithéatre Bleu	Amphithéatre Bordeaux	Room Maillot	Room 252AB
08.30 - 10.00	**S01.** **Chromatin organisation and gene expression** Chair: Damien Sanlaville, Michel Vekemans	**S02.** **From genes to treatment in multifactorial diseases** Chair: Florence Demenais, Aarno Palotie	**S03.** **Gene regulation in cancer** Chair: Marc Billaud, Minna Nyström	**S04.** **Emerging topics in neurobiology** Chair: Judith Melki, Francesco Palau	**ES3.** **Tweeting about genomics**
08.30	**S01.1** **Evolutionary pressures and gene expression regulation** ***Yoav Gilad***; *Chicago, United States*	**S02.1** **The search for bone and joint genes, and what to do with them?** ***André G. Uitterlinden***; *Rotterdam, Netherlands*	**S03.1** **Lynch syndrome as a model of mutations and epimutations in cancer** ***Päivi Peltomäki***; *Helsinki, Finland*	**S04.1** **The role of microglia in synaptogenesis** ***Cornelius Gross***; *Monterotondo, Italy*	**ES3.1** **Understanding Genomics. Trends in Science Communication** ***Anne M. Dijkstra***; *Enschede, Netherlands*
09.00	**S01.2** **Chromosomal rearrangements and gene expression** ***Alexandre Reymond***; *Lausanne, Switzerland*	**S02.2** **Inflammatory bowel disease: From genes to clinical impact?** ***Carl Anderson***; *Cambridge, United Kingdom*	**S03.2** **Epigenetic Programming of the Cancer Phenotype** ***Jean-Pierre Issa***; *Philadelphia, United States*	**S04.2** **The role of glia in neurodegenerative diseases** ***Don W. Cleveland***; *La Jolla, United States*	**ES3.2** **Science in the media. Lost in translation** ***David M. Secko***; *Montréal, Québec, Canada*
09.30	**S01.3** **Variation in Gene Regulation, Chromatin States and Protein Levels across Human Individuals and Populations** *M. Kasowski, S. Kyriazopoulou-Panagiotopoulou, F. Grubert, J.B. Zaugg, A. Kundaje, L. Wu, S. Candille, Y. Liu, L. Jiang, D. Xie, A. Boyle, Q. Zhang, F. Zakharia, D.V. Spacek, J. Li, L.M. Steinmetz, J.B. Hogenesch, M. Kellis, S. Batzoglou, H. Tang,* ***Mike Snyder****,* *Stanford, United States*	**S02.3** **From genetics to translation in SCD** ***Rob Graham***; *South San Francisco, United States*	**S03.3** **Insights into oncogenesis from cancer predisposition genes** ***Nazneen Rahman***; *London, United Kingdom*	**S04.3** **Molecular genetics of axon degeneration** *J. Gilley, L. Conforti, R. Adalbert, S. Milde,* ***Michael Coleman***; *Cambridge, United Kingdom*	
10.00 - 10.30	*Coffee Break / Poster viewing / Exhibition*				
10.30 - 11.30	**Poster viewing with presenters (odd poster numbers)**				
11.40 - 13.10	**WS01.** **Next Generation Sequencing in clinical practice - Filtering and reporting** Organisers : Joris Veltman, Gert Matthijs	**WS02.** **Debate: Hot topics in preimplantation genetic testing** Organisers : The H. Bui, Damien Sanlaville	**ES4.** **Cancer risk in developmental syndromes**		**WS03.** **UCSC Genome Browser I** Organiser: Robert M. Kuhn
	In this workshop clinical geneticists and laboratory specialists from 3 European laboratories will illustrate their diagnostic NGS approach and results obtained so far by discussing practical examples that demonstrate the power of the approach as well as challenges encountered. An open discussion with all participants will follow these presentations.	In the last two decades, the use of preimplantation genetic testing has increased dramatically. Genetic testing strategies and diagnostic accuracy continues to improve, but not without some controversies. In this Debate three PGD experts have been asked to give their views and debate on some “hot topics” in preimplantation genetic testing	**ES4.1** **Cancer risk in overgrowth syndromes** ***Andrea Riccio***; *Naples, Italy* **ES4.2** **Cancer risk in RASopathies** ***Karen Gripp***; *Wilmington, United States*		The introductory session on the UCSC Genome Browser will focus on the conceptual paradigm of the Genome Browser, navigation and searching, viewing your own data in Custom Tracks and saving Sessions for re-use and sharing with colleagues.We will share some useful features of the Browser that are not obvious. Please bring laptops.

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

PROGRAMME SUNDAY, JUNE 9

Time	Grand Amphithéatre	Amphithéatre Bleu	Amphithéatre Bordeaux	Room Maillot	Room 252AB	Room 242AB	Room 243
13.15 - 14.45	**C01. Structural variation and de novo mutations** Chair: Peter Kroisel, Cédric LeCaignec	**C02. Genotype phenotype correlation** Chair: Koen Devrient, Sylvie Odent	**C03. Prenatal diagnosis** Chair: Pascal Borry, Anne Moncla (tbc)	**C04. Cancer predisposition** Chair: Claude Houdayer, Agnes Bloch-Zupan	**C05. Functional Genomics** Chair: Claude Ferec, Benoit Arveiller	**C06. Neuromuscular disorders: From genes & modifiers to function & therapy** Chair: Yasemin Alanay, Nicolas Levy	**C07. Metabolic and mitochondrial disorders** Chair: Bela Melegh, Agnès Rötig
13.15	**C01.1 *De novo* mutations in the Genome of the Netherlands** ***Laurent C. Francioli****, P. Polak, W. Kloosterman, S. Sunyaev, P.I.W. de Bakker; Utrecht, Netherlands	**C02.1 From acrodysotosis to acroscyphodysplasia : phenotypic spectrum of PDE4D and PRKAR1A mutations through the study of 26 cases.** ***Caroline Michot***, C. Le Goff, Y. Alanay, G. Baujat, E. Blair, O. Boute, B. Gilbert-Dussardier, A. Goldenberg, B. Isidor, H. Kayserili, E. Kinning, M. Le Merrer, M. Simon, B. Tuysuz, A. Verloes, A. Munnich, V. Cormier-Daire; Paris, France	**C03.1 Highly multiplexed targeted single-nucleotide polymorphism (SNP) amplification and sequencing as a method for identifying fetal chromosomal disorders from maternal cell-free DNA** ***Bernhard Zimmermann***, M. Banjevic, M. Hill, P. Lacroute, M. Dodd, S. Sigurjonsson, P. Lau, D. Prosen, N. Chopra, A. Ryan, M. Hall, S. McAdoo, Z. Demko, B. Levy, M. Rabinowitz; San Carlos, United States	**C04.1 Mutations in SMARCE1 cause a novel disorder of multiple spinal meningiomas** ***William G. Newman***, M.J. Smith, J. O'Sullivan, S.S. Bhaskar, K.D. Hadfield, G. Poke, J. Caird, S. Sharif, D. Eccles, D. Fitzpatrick, D. Rawluk, D. DuPlessis, D.G. Evans; Manchester, United Kingdom	**C05.1 Post GWAS analysis of a BCL11A intronic region to define its role in regulating HbF levels.** ***Francesca Anedda***, S. Sanna, I. Asunis, G. Usala, D. Fabrice, C.A. Caria, L. Perseu, A. Loi, A. Cabriolu, L. Porcu, M.G. Marini, M.F. Marongiu, C. Sidore, R. Berutti, M. Pala, A. Angius, F. Busonero, A. Maschio, S. Satta, F. Demartis, L. Maccioni, R. Nagaraja, G. Abecasis, D. Schlessinger, M.S. Ristaldi, R. Galanello, P. Moi, F. Cucca, S. Sanna, M. Uda; Monserrato, Italy	**C06.1 The disease mechanisms of FSHD1 and FSHD2 converge at the level of somatic expression of DUX4** ***Silvere M. van der Maarel***, R. Lemmers, B. Bakker, R. Tawil, S. Sacconi, D. Miller, S. Tapscott; Leiden, Netherlands	**C07.1 New diagnostic paradigms for mitochondriopathies** ***Laura S. Kremer****, T.B. Haack, C.A. Biagosch, R. Kopajtich, B. Haberberger, T. Wieland, T. Schwarzmayr, A. Walther, T.M. Strom, T. Klopstock, M. Zeviani, R. Taylor, A. Rötig, A. Munnich, J. Smeitink, P. Freisinger, T. Meitinger, H. Prokisch; Neuherberg, Germany
13.30	**C01.2 Type 2 Diabetes strongly increases risk for the precancerous state of clonal mosaicism** ***Amélie Bonnefond****, B. Skrobek, S. Lobbens, E. Eury, S. Cauchi, O. Lantieri, B. Balkau, E. Riboli, M. Marre, G. Charpentier, L. Yengo, P. Froguel; Lille, France	**C02.2 Broadening the clinical spectrum to be ascribed to EFTUD2 haploinsufficiency** ***Jeanne Amiel***, F. Petit, M. Oufadem, V. Malan, J. Andrieux, G. Goudefroye, J. Alessandri, G. Baujat, C. Baumann, O. Boute, R. Caumes, C. Decaestecker, B. Delobel, K. Dieterich, L. Faivre, D. Gaillard, A. Goldenberg, M. Gonzales, D. Lacombe, S. Marlin, M. Mathieu-Dramard, S. Mehta, L. Pasquier, I. Simonic, A. Verloes, M. Vekemans, A. Munnich, M. Holder-Espinasse, L. de Pontual, S. Lyonnet, T. Attie-Bitach, C. Gordon; Paris, France	**C03.2 Diagnostic accuracy for the non-invasive prenatal detection of common autosomal aneuploidies** ***Sebastian Groemminger****, M. Stumm, M. Entezami, K. Haug, C. Blank, M. Wüstemann, B. Schulze, G. Raabe-Meyer, M. Hempel, M. Schelling, E. Ostermayer, S. Langer-Freitag, T. Burkardt, R. Zimmermann, T. Schleicher, B. Weil, U. Schöck, Y. Kumar, W. Hofmann; Konstanz, Germany	**C04.2 Mosaic *PPM1D* mutations are associated with predisposition to breast and ovarian cancer** ***Katie M. Snape****, E. Ruark, P. Humburg, T. Breast Ovarian Cancer Susceptibility Collaboration, T. Wellcome Trust Case Control Consortium, C. Turnbull, J. Reis-Filho, A. Ashworth, A. Antoniou, C.J. Lord, P. Donnelly, N. Rahman; Sutton, United Kingdom	**C05.2 Transcriptome and genome sequencing uncovers functional variation in human populations** *T. Lappalainen, M. Sammeth, M.R. Friedlander, P.A.C. 't Hoen, J. Monlong, M.A. Rivas, M. González-Porta, N. Kurbatova, T. Griebel, P.G. Ferreira, M. Barann, T. Wieland, L. Greger, M. van Iterson, J. Almlof, P. Ribeca, I. Pulyakhina, D. Esser, T. Giger, A. Tikhonov, M. Sultan, G. Bertier, D.G. MacArthur, M. Lek, E. Lizano, H.P.J. Buermans, H. Kilpinen, I. Padioleau, T. Schwarzmayr, O. Karlberg, H. Ongen, S.B. Montgomery, M.I. McCarthy, T. Strom, H. Lehrach, S. Schreiber, R. Sudbrak, A. Carracedo, S.E. Antonarakis, R. Haesler, A. Syvanen, G. van Ommen, A. Brazma, T. Meitinger, P. Rosenstiel, R. Guigo, I.G. Gut, X. Estivill,* ***Emmanouil T. Dermitzakis****; Geneva, Switzerland*	**C06.2 Plastin 3 ameliorates spinal muscular atrophy via delayed axon pruning and improves neuromuscular junction functionality** *B. Ackermann, S. Kröber, L. Torres-Benito, A. Borgmann, M. Peters, S. Hosseini Barkooie, R. Tejero, M. Jakubik, J. Schreml, J. Milbradt, T.F. Wunderlich, M. Riessland, L. Tabares,* ***Brunhilde Wirth****; Cologne, Germany*	**C07.2 ER Mannosidase I deficiency: An unexpected CDG-II with intellectual disability and dysmorphic features** ***Gert Matthijs***, D. Rymen, R. Peanne, V. Race, L. Sturiale, D. Garozzo, P. Mills, P. Clayton, J. Jaeken, F. Foulquier; Leuven, Belgium
13.45	**C01.3 De novo mutations in the autophagy gene encoding WDR45 (WIPI4) cause static encephalopathy of childhood with neurodegeneration in adulthood** ***Naomichi Matsumoto***, T. Nishimura, K. Muramatsu, H. Kodera, S. Kumada, K. Sugai, E. Kasai-Yoshida, N. Sawaura, H. Nishida, A. Hoshino, F. Ryujin, S. Yoshioka, H. Arakawa, M. Kato, N. Mizushima, H. Saitsu; Yokohama, Japan	**C02.3 Delineation of the clinical spectrum of *RNU4ATAC*-related microcephalic osteodysplastic primordial dwarfism type I syndrome: an international cohort** ***Patrick Edery***, E. Alix, M. B. Bober, A. Labalme, R. Touraine, S. Nampoothiri, I. Borg, P. Jouk, S. Berland, A. Toutain, E. Steichen, J. Attia, F. Dijoud, A. Vasilijevic, A. Fournier, C. Poizat, C. Marcaillou, C. A. Wise, L. Guibaud, D. Sanlaville, F. Rousseau, F. Clerget-Darpoux, A. Leutenegger; Lyon, France	**C03.3 Clinical Performance Comparison of the Harmony(TM) Prenatal Test and First Trimester Combined Screening in a General Pregnancy Population** *K. Nicolaides, A. Syngelaki, G. Ashoor, T. Musci, E. Wang,* ***Ken Song****; San Jose, United States*	**C04.3 Germline mutations of inhibin in early-onset ovarian cancer** ***Isabelle Tournier***, R. Marlin, K. Walton, F. Charbonnier, S. Coutant, C. Spurrell, M. Vezain, H. Roman, J. Tinat, J. Sabourin, D. Vaur, M. King, C. Harrison, T. Frebourg; Rouen, France	**C05.3 Genetic regulation of lincRNA and protein-coding genes expression variation - similarities and differences** ***Konstantin Popadin***, M. Gutierrez-Arcelus, T. Lappalainen, E. Dermitzakis, S.E. Antonarakis; Geneva, Switzerland	**C06.3 The neuronal endopeptidase ECEL1 is associated with autosomal recessive distal arthrogryposis** *Klaus Dieterich*, S. Quijrano-Roy, N. Monnier, J. Zhou, J. Fauré, D. Avila Smirnow, R. Carlier, C. Laroche, P. Marcorelles, S. Mercier, A. Mégarbané, S. Odent, N. Romero, D. Sternberg, I. Marty, B. Estournet, P. Jouk, J. Melki, J. Lunardi; Grenoble, France*	**C07.3 Mutations in nuclear-encoded components of mitochondrial respiratory chain complex III & IV cause apoptosis-driven developmental defects, a new mitochondrial phenotype in vertebrates** ***Alessia Indrieri****, V. van Rahden, V. Tiranti, I. Conte, M. Morleo, D. Iaconis, G. Chesi, A. Romano, R. Tate, I. D'Amato, I. Maystadt, S. Demuth, A. Zvulunov, I. Ferrero, P. Goffrini, P. Bovolenta, K. Kutsche, M. Zeviani, B. Franco; Naples, Italy*

PROGRAMME SUNDAY, JUNE 9-CONTINUED

Time	Grand Amphithéatre	Amphithéatre Bleu	Amphithéatre Bordeaux	Room Maillot	Room 252AB	Room 242AB	Room 243
cont.	**C01. Structural variation and de novo mutations**	**C02. Genotype phenotype correlation**	**C03. Prenatal diagnosis**	**C04. Cancer predisposition**	**C05. Functional Genomics**	**C06. Neuromuscular disorders: From genes and modifiers to function & therapy**	**C07. Metabolic and mitochondrial disorders**
14.00	**C01.4** **Mapping of two human genomes with a single molecule nanochannel array platform for genome-wide structural variation analysis and de novo sequence assembly** ***Pui Y. Kwok**, Y.Y.Y. Lai, A.C.Y. Mak, E.T. Lam, J. Silbert, T.P. Kwok, J.W. Li, A.K.Y. Leung, J.J.K. Wu, A.K.Y. Yim, A. Poon, C. Chu, C. Lin, M. Requa, A. Hastie, T. Anantharaman, H. Van-Steenhouse, H. Dai, F. Trintchouk, M. Saghbini, M. Austin, K. Haden, H. Cao, S.M. Yiu, K.Y. Yip, T.F. Chan, M. Xiao; San Francisco, USA*	**C02.4** **Baraitser-Winter syndrome due to ACTB/G1 mutations: delineation of the spectrum in 34 cases** ***Alain Verloes**, J. Masliah-Planchon, N. Di Donato, J. Allanson, M. Baraitser, H. Brunner, N. Chassaing, A. David, K. Devriendt, V. Drouin-Garraud, F. Faravelli, F. Juliano, M. Kempers, D. Lacombe, A. Lin, G. Mancini, C. Marques Lourenço, G. Morin, M. Nezarati, M. Nowaczyk, J. Ramer, S. Osimani, N. Philip, M. Pierpont, M. Rossi, C. Rusu, Y. Sznajer, N. Templin, V. Uliana, B. Van bon, C. Van Ravenswaaij, J. Rivière, A. Fry, A. Hoischen, W.B. Dobyns, D. Pilz; Paris, France*	**C03.4** **Comprehensive Chromosome Screening in PGD and PGS - Ethical Challenges** ***Kristien Hens**, W. Dondorp, G. de Wert; Maastricht, Netherlands*	**C04.4** **Parkinson's disease and melanoma: a common genetic pathway linked to *PARKIN* inactivation** ***Hui-Han Hu**, N. Dumaz, S. Lesage, L. Michel, V. Descamps, S. Mourah, C. Lebbé, N. Basset Seguin, M. Bagot, A. Bensussan, L. Deschamps, M. Leccia, A. Tsalamlal, R. Sivaramakrishna, S. Klebe, R. Kumar, C. Kannengiesser, A. Couvelard, B. Grandchamp, T. Luc, A. Brice, N. Soufir; Paris, France*	**C05.4** **Coordinated effects of sequence variation on DNA binding, chromatin structure, and transcription** ***Andreas R. Gschwind**, H. Kilpinen, S.M. Waszak, S.K. Raghav, R.M. Witwicki, A. Orioli, E. Migliavacca, M. Wiederkehr, M. Gutierrez-Arcelus, N. Panousis, A. Yurovsky, T. Lappalainen, L. Romano-Palumbo, A. Planchon, D. Bielser, J. Bryois, I. Padioleau, G. Udin, S. Turnheer, D. Hacker, L.J. Core, J.T. Lis, N. Hernandez, B. Deplancke, A. Reymond, E.T. Dermitzakis; Lausanne, Switzerland*	**C06.4** **Constitutive activation of STIM1 causes tubular aggregate myopathy** *J. Laporte, F. Chevessier, A. Maues De Paula, S. Attarian, D. Hantaï, K. Ghorab, N. Levy, M. Krahn, B. Eymard, M. Bartoli, **Johann Böhm**; Strasbourg, France*	**C07.4** **Mutation of the iron-sulfur cluster assembly IBA57 gene causes lethal myopathy and encephalopathy.** ***Nikhita Ajit Bolar**, A.V. Vanlander, C. Wilbrecht, N. Van der Aa, J. Smet, B. De Paepe, G. Vandeweyer, F. Kooy, F. Eyskens, E. De Latter, G. Delanghe, P. Govaert, J.G. Leroy, R. Lill, R. Van Coster, L. Van Laer, B. Loeys; Antwerp, Belgium*
14.15	**C01.5** **Exome sequencing in sporadic cases of schizophrenia identifies de novo protein-altering mutation in candidate genes** ***Michel Guipponi**, F. Santoni, C. Gehrig, M. Rotharmel, M. Cuenca, O. Guilin, D. Dikeos, G. Papadimitriou, A. Méary, F. Schürhoff, S. Jamain, M. Leboyer, D. Rujescu, D. Campion, A. Malafosse, S.E. Antonarakis; Geneva, Switzerland*	**C02.5** **A comprehensive analysis of a cohort of Cornelia de Lange syndrome cases.** ***Morad Ansari**, R. Aldridge, G.V. Poke, K. Williamson, T. Homfray, R.C.M. Hennekam, D.R. FitzPatrick; Edinburgh, United Kingdom*	**C03.5** **The challenge of preconceptional, preimplantation, and prenatal genetic diagnoses of mitochondrial DNA disorders** ***Sophie Monnot**, N. Gigarel, P. Vachin, E. Herzog, P. Burlet, N. Frydman, A. Benachi, G. Chalouhi, Y. Ville, R. Frydman, A. Lebre, A. Rotig, D.C. Samuels, C. Elie, A. Munnich, J. Bonnefont, J. Steffann; Paris, France*	**C04.5** **Loss of a Regulatory Element May Determine Endometrial Cancer Risk in *EPCAM* Deletion Carriers** ***Richarda M. de Voer**, E. Verwiel, M. Donna, E. van Wijk, L. Vreede, R. Freixas, K. Wu, I. Nagtegaal, N. Hoogerbrugge, A. Geurts van Kessel, M. Ligtenberg, R. Kuiper; Nijmegen, Netherlands*	**C05.5** **Deciphering vertebrate regulatory grammar using high-throughput in vivo functional assays** *R.P. Smith, L. Taher, S.J. Riesenfeld, R.P. Patwardhan, I. Ovcharenko, K.S. Pollard, J. Shendure, **Nadav Ahituv**; San Francisco, United States*	**C06.5** **Myotonic dystrophy CTG expansion affects synaptic vesicle proteins, neurotransmission and mouse behavior** *O. Hernández-Hernández, C. Guiraud-Dogan, G. Sicot, A. Huguet, S. Luilier, E. Steidl, S. Saenger, C. Chevarin, A. Nicole, B. Buisson, J. Bizot, M. Hamon, S. Humez, G. Bassez, F. Metzger, L. Buée, A. Munnich, N. Sergeant, G. Gourdon, **Mário Gomes-Pereira**; Paris, France*	**C07.5** **Exome sequencing Reveals Mutated NUBPL in Patients with Complex I Deficiency and a Distinct MRI Pattern** ***Sietske H. Kevelam**, R.J. Rodenburg, N.I. Wolf, P. Ferreira, R.J. Lunsing, L.G. Nijtmans, A. Mitchell, H.A. Arroyo, D. Rating, A. Vanderver, C.G.M. van Berkel, T.E.M. Abbink, P. Heutink, M.S. van der Knaap; Amsterdam, Netherlands*
14.30	**C01.6** **The genome structure of the Dutch population** ***Victor Guryev**, W. Kloosterman, L.C. Francioli, J.Y. Hehir-Kwa, T. Marschall, A. Schoenhuth, M. Moed, E. Lameijer, A. Abdellaoui, S. Koval, F. Hormozdiari, J. de Ligt, N. Amin, F. van Dijk, L. Karssen, H. Mei, E.E. Eichler, K. Ye; Groningen, Netherlands*	**C02.6** **Treacher Collins Syndrome: clinical and molecular study based on a series of 135 patients** ***Corinne Collet**, M. Vincent, S. Marlin, D. Martin-Coignard, C. Coubes, A. David, S. Lyonnet, C. Vilain, A. Dieux-Coeslier, M. Holder, B. Isidor, M. Jacquemont, S. Julia, D. Lacombe, V. Layet, S. Naudion, S. Odent, L. Pasquier, S. Pelras, N. Philip, G. Pierquin, F. Prieur, N. Aboussair, T. Attié-Bitach, G. Baujat, H. Dollfus, B. Doray, P. Edery, F. Giuliano, A. Goldenberg, C. Goizet, A. Guichet, L. Lambert, B. Le Heup, J. Martinovic, S. Mercier, C. Mignot, M. Moutard, M. Perez, H. Randrianaivo, K. Szakszon, A. Toutain, A. Verloes, J. Vigneron, E. Sanchez, J. Puechberty, J. Laplanche, P. Sarda, D. Geneviève; Paris, France*	**C03.6** **Exome sequencing of 27 trios to identify genetic causes of fetal abnormalities** ***Keren J. Carss**, S. Hillman, E. Maher, P. Vijayarangakannan, D. Stemple, M. Kilby, M. Hurles; Hinxton, Cambridgeshire, United Kingdom*	**C04.6** **Suppressor-tRNA Restores Functional E-Cadherin Expression in *Cdh1* Mutant Cancer Cells: A Potential Approach to Treat Hereditary Diffuse Gastric Cancer** ***Renata Bordeira-Carriço**, D. Ferreira, D. Mateus, H. Pinheiro, A. Pêgo, R. Seruca, M. Santos, C. Oliveira; Porto, Portugal*	**C05.6** **Novel genetic variants associated with alternative polyadenylation and expression of noncoding transcripts** ***Daria V. Zhernakova**, H. Westra, E. de Klerk, A. Mastrokolias, S. Amini, Y. Ariyurek, R. Jansen, B.W. Penninx, J.J. Hottenga, G. Willemsen, E.J. de Geus, D.I. Boomsma, J.H. Veldink, L.H. van den Berg, C. Wijmenga, J.T. den Dunnen, G.B. van Ommen, P.A.C. 't Hoen, L. Franke; Groningen, Netherlands*	**C06.6** **Nanoparticles as delivery systems for antisense oligoribonucleotides: biodistribution studies and definition of the release kinetic in intraperitoneally and orally treated mdx mice** *M. Falzarano, C. Passarelli, K. Sparnacci, M. Laus, P. Bonaldo, P. Braghetta, J. vanDeutekom, **Alessandra Ferlini**; Ferrara, Italy*	**C07.6** **Pioglitazone prevents mitochondria dysfunction and halts axonal degeneration in a mouse model of X-adrenoleukodystrophy** ***Aurora Pujol**, L. Morato, J. Galino, M. Ruiz, M. Portero-Otin, R. Pamplona, I. Ferrer; Barcelona, Spain*
14.45 - 15.15	*Vitamin break / Poster viewing / Exhibition*						

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

PROGRAMME SUNDAY, JUNE 9

Time	Grand Amphithéatre	Amphithéatre Bleu	Amphithéatre Bordeaux	Room Maillot	Room 252AB	Room 242AB
15.15 – 16.45	**WS04. Dysmorphology Workshop 1** Organisers: Dian Donnai; Jill Clayton-Smith	**WS05. Analysis, interpretation and reporting of array data** Organisers: Nicole de Leeuw; Cornelia van Ravenswaaij-Arts	**WS06. Clinical Cancer Genetics Club** Organisers: Maurizio Genuardi; Dominique Stoppa-Lyonnet	**WS07. Quality assurance** Organisers: Elisabeth Dequeker; Mike Morris	**WS08. Biomedical Data Analysis with Galaxy** Organisers: Anton Nekrutenko; Enis Afgan	**WS09. Inequality in Genetic Services** Organisers: Stan Lyonnet; Jörg Schmidkte
	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 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.		Galaxy is an open, web-based platform for data intensive biological research that enables non-bioinformaticians to create, run, tune, and share their own bioinformatic analyses. This workshop will show participants how to integrate data and perform simple and complex analysis within Galaxy. It will also demonstrate how Galaxy enables reproducibility in bioinformatics, and how to use visualization to refine and drive analysis, all within the Galaxy Framework. Finally, the workshop will highlight how researchers can install Galaxy on local computational resources, or using computational cloud providers such as Amazon Web Services.	Inequalities in the provision of genetic services exist in many parts of the world including Europe, and extend from sub-populations to whole countries. This workshop thus discusses the situation in underprivileged groups within countries, including migrants and displaced persons, and underserved countries in Northern Western and Eastern Europe, and looks out for action that could be taken.
16.45 – 17.15	*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 9

Time	Grand Amphithéatre	Amphithéatre Bleu	Amphithéatre Bordeaux	Room Maillot	Room 252AB
17.15 - 18.45	**S05.** **Interpreting NGS data** Chair: Catherine Boileau, Joris Veltman	**S06.** **Cancer genetics** Chair: Thierry Frébourg (tbc), Carla Oliveira	**ES5.** **Epilepsies**	**S07.** **Genetics of skin diseases and new therapies** Chair: Didier Lacombe, Giovanni Neri	**S08.** **Evolution of organs** Chair: Corinne Antignac, Jean-Louis Mandel
17.15	**S05.1** **Analytical challenges of using next-generation sequencing to unlock complex disease** ***Mark Daly***; *Boston, United States*	**S06.1** **Identification of the gene for mixed polyposis syndrome: the end of a 50-year journey** ***Ian Tomlinson***; *Oxford, United Kingdom*	**ES5.1** **Spectrum of monogenic forms and clinical importance of de novo mutations** ***Stéphanie Baulac***; *Paris, France*	**S07.1** **iPS and their therapeutic potentiel for keratinizing disorders** ***Dennis Roop***; *Aurora, United States*	**S08.1** **Dissecting the effects of selection in the human genome: the case of immunity to infection** ***Luis Quintana-Murci***; *Paris, France*
17.45	**S05.2** **Diagnostic exome sequencing in genetically heterogeneous disease** ***Lisenka E.L.M. Vissers***; *Nijmegen, Netherlands*	**S06.2** **The role of IL7R in childhood T-cell acute lymphoblastic leukemia** ***João T. Barata***; *Lisboa, Portugal*	**ES5.2** **The role of genetic susceptibility factors in epilepsy and their clinical relevance** ***Ingo Helbig***; *Kiel, Germany*	**S07.2** **Netherton syndrome and links to eczema** ***Alain Hovnanian***; *Paris, France*	**S08.2** **Gene dosage sensitivity and copy-number evolution** ***Aoife McLysaght***; *Dublin, Ireland*
18.15	**S05.3** **Prioritizing disease-causing variation by genomic data fusion** ***Yves Moreau***; *Leuven, Belgium*	**S06.3** **The role of exosomes in cancer-cell communication, dissemination, and therapy-resistence** ***Lorraine O'Driscoll***; *Dublin, Ireland*		**S07.3** **Artificial Skin** ***Marcela del Rio***; *Madrid, Spain*	**S08.3** **Evolution of vision** ***Detlef Arendt***; *Heidelberg, Germany*

	Room Maillot
19.00 – 20.00	**ESHG Membership Meeting** **All ESHG members welcome!**

SCIENTIFIC

SCIENTIFIC PROGRAMME

Monday, June 10, 2013

PROGRAMME

PROGRAMME MONDAY, JUNE 10

Time	Grand Amphithéatre	Amphithéatre Bleu	Amphithéatre Bordeaux	Room Maillot	Room 252AB
08.30 - 10.00	**S09.** **New therapeutic highlights in rare diseases** Chair: Arnold Munnich (tbc), Brunhilde Wirth	**S10.** **Reproductive genetics and newborn screening** Chair: The-Hung Bui, Julie Steffann	**S11.** **Animal Models for Human Diseases** Chair: Peter Robinson, Sandrine Humbert	**S12.** **Judging our genes** Chair: Francesca Forzano, Dominique Stoppa-Lyonnet	**ES6.** **Retinal dystrophies: news and views**
08.30	**S09.1** **Adaptive immunodeficiency and gene therapy treatment** ***Alain Fischer***; *Paris, France*	**S10.1** **Noninvasive whole genome sequencing** ***Stephen Quake***; *Stanford, United States*	**S11.1** **Mousemodels of Down syndrome** ***Yann Hérault***; *Illkirch, France*	**S12.1** **The geneticist's perspective** ***Andrew Read***; *Manchester, United Kingdom*	**ES6.1** **Inherited retinal dystrophies: insights & challenges behind the rainbow** ***Frans P.M. Cremers***; *Nijmegen, Netherlands*
09.00	**S09.2** **Antisense therapy in SMA** ***Adrian Krainer***; *Cold Spring Harbor, United States*	**S10.2** **New approaches in preimplantation screening** ***Dagan Wells***; *Oxford, United Kingdom*	**S11.2** **Protein Degradation in Health and Disease** ***Thorsten Hoppe***; *Cologne, Germany*	**S12.2** **Narrowing down the free-will compass? A social and ethical perspective** ***Mairi Levitt***; *Lancaster, United Kingdom*	**ES6.2** **Inherited retinal disease - management and therapies** ***Andrew R. Webster***; *London, United Kingdom*
09.30	**S09.3** **Stem-cell gene therapy for the Wiskott-Aldrich Syndrome** ***Christoph Klein***; *Munich, Germany*	**S10.3** **Newborn screening; what is possible, what do we want?** ***Bridget Wilcken***; *Sydney, Australia*	**S11.3** **From bedside to bench: a Fragile X patient mutation yields insights into the functional specialization of the FXR protein family.** *Z. Okray,* ***Bassem Hassan***; *Leuven, Belgium*	**S12.3** **Narrowing down the free-will compass? A legal perspective** ***Amedeo Santosuosso***; *Pavia, Italy*	
10.00 - 10.30	***Coffee break sponsored by Abbott Molecular*** */ Poster viewing / Exhibiton*				
10.30 - 11.30	**Poster viewing with presenters (even poster numbers)**				

	Grand Amphithéatre	Amphithéatre Bleu
11.40 - 13.10	**ES7.** **NGS in the clinic**	**ES8.** **How to get published in the European Journal of Human Genetics (EJHG)**
	ES7.1 **Introducing next generation technology to clinical scientists** ***Caroline Wright***; *Cambridge, United Kingdom* **ES7.2** **Bioinformatic challenges of applying NGS in the clinic** ***Christian Gilissen***; *Nijmegen, Netherlands*	**ES8.1** **How to get published in the European Journal of Human Genetics** ***GertJan van Ommen***; *Leiden, Netherlands*

PROGRAMME MONDAY, JUNE 10

Time	Grand Amphithéatre	Amphithéatre Bleu	Amphithéatre Bordeaux	Room Maillot	Room 252AB	Room 242AB	Room 243
13.15 - 14.45	**C08. Intellectual disability: Gene discovery and dysfunction** Chair: Didier Lacombe, Dorit Lev	**C09. Population-based sequencing** Chair: Tayfun Ozcelik, Laurent Abel	**C10. Genetic Counselling, Education and Public Services** Chair: Segolène Aymé, Heather Skirton	**C11. Molecular mechanisms in tumorigenesis** Chair: Olivier Delattre (tbc), Borut Peterlin (tbc)	**C12. Connective tissue disorders: Gene identification** Chair: Domenico Coviello, Laurence Olivier-Faivre	**C13. Neurodegenerative disorders: From gene discovery to therapy** Chair: Martijn Breuning, Alexandra Durr	**C14. Sensory disorders and Immunological disorders** Chair: Patrick Calvas, Nina Canki-Klain
13.15	**C08.1 Mutations in the microtubule-associated protein EML1/Eml1 lead to ectopic progenitors during cortical development and heterotopia in mouse and human** ***Françoise Phan Dinh Tuy***, *M. Kielar, C. Lebrand, K. Poirier, R. Olaso, S. Bizzotto, K. Boutourlinsky, N. Bahi-Buisson, A. Le Moing, C. de Juan, V. Borrell, P. Berquin, W. Carpentier, E. Welker, J. Chelly, A. Croquelois, F. Francis; Paris, France*	**C09.1 Exome sequencing of 2,000 Danish individuals and the role of rare coding variants in type 2 diabetes** ***Thomas Sparso****, *K. Lohmueller, Q. Li, E. Andersson, T. Korneliussen, A. Albrechtsen, K. Banasik, N. Grarup, I. Hallgrimsdottir, K. ´Kiil, T. Kilpeläinen, N. Krarup, T. Pers, G. Sanchez, T. Jørgensen, A. Sandbæk, T. Lauritzen, K. Kristiansen, S. Brunak, Y. Li, T. Hansen, J. Wang, R. Nielsen, O. Pedersen; Copenhagen, Denmark*	**C10.1 From personal genetic counseling to public health screening: The BRCA Opportunity** ***Sari Lieberman****, *A. Tomer, A. Ben-Chetrit, O. Olsha, T. Zalut, A. Lahad, E. Levy-Lahad; Jerusalem, Israel*	**C11.1 The 3D topographic mapping of genetic variations in treatment naïve advanced ovarian cancer** ***Mirjam S. de Pagter****, *M. Hoogstraat, G.A. Cirkel, J. Kreeftmeijer, C.C. Lee, E. Levandowsky, T. Guy, K. Duran, R. van ,t Slot, G.N. Jonges, S. van Lieshout, M.P.J. Lolkema, P.O. Witteveen, R.P. Zweemer, M.J. Koudijs, I.J. Nijman, E.E. Voest, T.T. Harkins, E. Cuppen, W.P. Kloosterman; Utrecht, Netherlands*	**C12.1 Congenital poikiloderma, fatty infiltration of muscles and pulmonary fibrosis: a new syndrome caused by a new gene** ***Sandra Mercier****, *S. Küry, N. Khumalo, D. Houniet, E. Salort-Campana, N. Bodak, V. Cormier-Daire, G. Shaboodien, J. Mussini, A. David, S. Barbarot, B. Keavney, B. Mayosi, S. Bézieau; Nantes, France*	**C13.1 Alteration of lipid metabolism in hereditary spastic paraplegia 26** *E. Mundwiller, A. Boukhris, R. Schule, J.L. Loureiro, C. Marques Lourenço, I. Rekik, M.A. Gonzalez, P. Charles, J. Gauthier, A. Ferbert, M. Gaussen, A. Caballero Oteyza, S. Forlani, C. Mhiri, L. Schols, G. Rouleau, W. Marques Junior, C. Depienne, A. Brice, F. Darios, A. Durr, S. Zuchner,* ***Giovanni Stevanin****; Paris, France*	**C14.1 *ALDH1A3* mutations cause recessive anophthalmia and microphthalmia** ***Lucas Fares Taie****, *S. Gerber, N. Chassaing, J. Clayton-Smith, S. Hanein, E. Silva, M. Serey, V. Serre, X. Gerard, C. Baumann, G. Plessis, B. Demeer, D. Bremond-Gignac, L. Brétillon, C. Bole-Feyssot, P. Nitschke, P. Nitschke, A. Munnich, S. Lyonnet, P. Calvas, J. Kaplan, N. Ragge, J. Rozet; Paris, France*
13.30	**C08.2 Mutations in *DEAF1* cause intellectual disability with severe speech impairment** ***Anneke T. Vulto-van Silfhout****, *B. Nijhof, P.J. Jensik, C. Zweier, J. de Ligt, B.W.M. van Bon, D. Lugtenberg, J.A. Veltman, H. van Bokhoven, H.G. Brunner, A. Rauch, L.E.L.M. Vissers, M.W. Collard, A. Schenck, B. Menten, B.B.A. de Vries; Nijmegen, Netherlands*	**C09.2 Haplotype sharing reveals fine-scale demographic history** ***Pier Francesco Palamara****, *T. Lencz, A. Darvasi, I. Pe'er, The Genome of the Netherlands Consortium; New York City, United States*	**C10.2 Drivers, barriers and opportunities for genetic testing services in emerging economies: the GenTEE (Genetic Testing in Emerging Economies) project** ***Irmgard Nippert****, A. Christianson, D.D.G. Horovitz, R. Kamal Raouf, C.D. Padilla, V. Penchaszadeh, A. Rajab, I.C. Verma, N. Zhong, L. Gribaldo, U. Kristoffersson, J. Schmidtke; Münster, Germany*	**C11.2 The Genomic Landscape of Somatic Mutations in Subtypes of Germinal-Center derived B-cell Lymphomas** *M. Schlesner, J. Richter, O. Ammerpohl, S.H. Bernhart, A. Borkhardt, B. Brors, B. Burkhardt, A. Claviez, M. Dreyling, S. Eberth, J. Eils, R. Eils, S. Haas, M. Hansmann, K. Hezaveh, J. Hoell, S. Hoffmann, M. Hummel, D. Karsch, W. Klapper, J. Korbel, U. Kostezka, M. Kreuz, D. Kube, R. Küppers, D. Langenberger, C. Lawerenz, E. Leich, D. Lenze, P. Lichter, M. Loeffler, P. Moeller, B. Radlwimmer, S. Radomski, M. Rohde, P. Rosenstiel, A. Rosenwald, M. Rosolowski, M. Schilhabel, S. Schreiber, P.F. Stadler, M. Szczepanowski, L. Trümper, M. Weniger,* ***Reiner Siebert****; Kiel, Germany*	**C12.2 TBX4 mutations (small patella syndrome) are associated with childhood-onset pulmonary arterial hypertension** ***Wilhelmina S. Kerstjens-Frederikse****, E.M.H.F. Bongers, M.T.R. Roofthooft, E.M. Leter, J.M. Douwes, A. Van Dijk, A. Vonk-Noordegraaf, K.K. Dijk-Bos, L.H. Hoefsloot, E.S. Hoendermis, J.J.P. Gille, B. Sikkema-Raddatz, R.M.W. Hofstra, R.M.F. Berger; Groningen, Netherlands*	**C13.2 *WDR45 de novo* mutations cause of a clinically distinct, X-linked subtype of NBIA** ***Tobias B. Haack****, *P. Horgath, A. Gregory, M.C. Kruer, T. Wieland, T. Schwarzmayr, A. Walther, L. Sanford, M.A. Kurian, B. Garavalgia, N. Nardocci, V. Tiranti, T.M. Strom, T. Meitinger, S.J. Hayflick, H. Prokisch; Munich, Germany*	**C14.2 Mutations in the Nuclear NAD+ synthesising enzyme *NMNAT1* cause Leber congenital amaurosis with early-onset severe macular atrophy and optic atrophy** ***Sylvain Hanein****, I. Perrault, X. Zanlonghi, V. Serre, M. Nicouleau, S. Defoort-Delhemmes, N. Delphin, L. Fares Taie, S. Gerber, O. Xerri, C. Edelson, A. Glodenberg, A. Duncombe, G. LeMeur, C. Hamel, E. Silva, P. Nitschke, P. Calvas, A. Munnich, O. Roche, H. Dollfus, J. Kaplan, J. Rozet; Paris, France*
13.45	**C08.3 Mutations in TTI2 reveal a role for Triple T complex in human brain development** ***Maéva Langouët****, *A. Saadi, G. Rieunier, S. Moutton, K. Siquier-Pernet, M. Fernet, P. Nitschke, A. Munnich, M. Stern, M. Chaouch, L. Colleaux; Paris, France*	**C09.3 Signatures of selection in the Genome of the Netherlands Project** ***Clara C. Elbers*****, *S.L. Pulit*, L.C. Francioli, A. Menelaou, P.I.W. de Bakker, Genome of the Netherlands Consortium, * these authors contributed equally; Utrecht, Netherlands*	**C10.3 Incidental findings in research: National Health Service Research Ethics Committee member perspectives.** ***Leigh M. Jackson****, *L. Goldsmith, A. O'Connor, H. Skirton; Plymouth, United Kingdom*	**C11.3 FAS/FASL pathways is impaired in chordoma and is involved in notochord development and regression** ***Luca Ferrari****, A. Pistocchi, A. Calastretti, L. Libera, N. Boari, G. Canti, P. Mortini, F. Cotelli, P. Riva; Milan, Italy*	**C12.3 Mutations in SNRPE, which encodes a core protein of the spliceosome, cause autosomal-dominant hypotrichosis simplex** *S.M. Pasternack, M. Refke, E. Paknia, H. Hennies, T. Franz, N. Schäfer, A. Fryer, M. van Steensel, E. Sweeney, M. Just, C. Grimm, R. Kruse, C. Ferrándiz, M.M. Nöthen, U. Fischer,* ***Regina C. Betz****; Bonn, Germany*	**C13.3 A stop mutation in WDR81 causes microcephaly with variable penetrance.** ***Sebahattin Cirak****, *B. Peterson, Y. Chang, A. ElSharawy, P. Alexandre, K. Schoner, A. Pagenstecher, G. Gestri, S. Suren, D. Gerrelli, A. Volk, P. Nurnberg, S. Wilson, J. Clarke, B. Albrecht, W. Huttner, D. Morris-Rosendahl, G. Uyanik, A. Franke, T. Voit; Washington, United States*	**C14.3 Identification of new genes for Hereditary Hearing Loss (HHL) using linkage studies and Whole Exome Sequencing analysis** *D. Vozzi,* ***Giorgia Girotto****, F. Faletra, K. Abdulhadi, D. Vuckovic, A. D'Eustacchio, M. Khalifa Alkowari, R. Badii, P. Gasparini; Trieste, Italy*

Time	Grand Amphithéatre	Amphithéatre Bleu	Amphithéatre Bordeaux	Room Maillot	Room 252AB	Room 242AB	Room 243
cont.	**C08. Intellectual disability: Gene discovery & dysfunction**	**C09. Population-based sequencing**	**C10. Genetic Counselling, Education and Public Services**	**C11. Molecular mechanisms in tumorigenesis**	**C12. Connective tissue disorders: Gene identification**	**C13. Neurodegenerative disorders: From gene discovery to therapy**	**C14. Sensory disorders and Immunological disorders**
14.00	**C08.4 Involvement of kinesin family members KIF4A and KIF5C in intellectual disability and synaptic function** *M.H. Willemsen, W. Ba, K. Poirier, W.M. Wissink-Lindhout, L.E.L.M. Peart-Vissers, A.P.M. de Brouwer, H. van Bokhoven, J. Chelly, V. Kalscheuer, N. Nadif-Kasri,* ***Tjitske Kleefstra****; Nijmegen, Netherlands*	**C09.4 The impact of genetic variation on lipid traits from whole exome sequences of 10,000 individuals: the T2D-GENES Consortium** ***Inga Prokopenko****, H.M. Highland, X. Sim, A. Mahajan, A. Manning, M. Rivas, G. Atzmon, S. Choi, B.K. Cornes, A. Lokke, J.C. Florez, P. Fontanillas, N. Palmer, E.R. Gamazon, I. Huh, H.K. Im, J. Kim, Y.J. Kim, C.M. Lindgren, T.M. Teslovich, T.M. Frayling, J. Dupuis, J.B. Meigs, A.P. Morris; London, United Kingdom*	**C10.4 Preferences for priority setting criteria in genetic testing: a discrete choice experiment** ***Franziska Severin****, W. Hess, J. Schmidtke, A. Mühlbacher, P. Meyer, W. Rogowski; Neuherberg, Germany*	**C11.4 SDHB mutations link pheochromocytoma/paraganglioma malignancy to epithelial to mesenchymal transition, both in human tumors and in SDHB-/- chromaffin cells** ***Céline Loriot****, M. Domingues, A. Berger, N. Burnichon, C. Martinelli, L. Vescovo, E. Letouzé, J. Sael, L. Larue, A. Gimenez-Roqueplo, J. Favier; Paris, France*	**C12.4 Gain-of-function mutations in the mechanically activated ion channel PIEZO2 cause distal arthrogryposis type 5** ***Gunnar Houge****, B. Coste, R.L. Maas, A. Hoischen, S.R. Sunyaev, A. Patapoutian; Bergen, Norway*	**C13.4 Chromatin structure, transcription and CAG instability in Huntington's disease** *A. Goula, A. Stys, J. Chan, Y. Trottier, R. Festenstein,* ***Karine Merienne****; Strasbourg, France*	**C14.4 A founder mutation in ULFIN, a new gene on chromosome 16q22.1, in patients with spinocerebellar ataxia type 4 (SCA4)** ***Diana Braunholz****, F. Kähllitz, I. Braenne, Y. Hellenbroich, S. Tennstedt, P. Buse, P. Seibler, G. Gillessen-Kaesbach, J. Erdmann, C. Zühlke, F. Kaiser; Lübeck, Germany*
14.15	**C08.5 Abnormal expression of sex biased genes in PCDH19-female limited epilepsy and intellectual disability (PCDH19-FLE) suggests a role for neurosteroid hormones.** *C. Tan, C. Shard, K. Hynes, E. Douglas, L.S. Nguyen, M. Corbett, G. Buchanan, E. Ranieri, R. Guerrini, C. Marini, S.F. Berkovic, I.E. Scheffer,* ***Jozef Gecz****; North Adelaide, Australia*	**C09.5 Exome sequence analysis of type 2 diabetes in over 10,000 samples from five ancestry groups: the T2D-GENES Consortium.** ***Manuel A. Rivas****, T.M. Teslovich, A. Morris, P. Fontanillas, A. Mahajan, X. Sim, J. Flannick, N.P. Burtt; Oxford, United Kingdom*	**C10.5 Effects of multifaceted oncogenetics training for general practitioners** ***Elisa J.F. Houwink****, L. Henneman, A.M. Muijtjens, S.R. van Teffelen, J. Rethans, L. van der Jagt, G. Dinant, C. van der Vleuten, C.T. Schrander-Stumpel, H. Meijers-Heijboer, M.C. Cornel; Amsterdam, Netherlands*	**C11.5 MicroRNAs as possible initiators and drivers for microsatellite unstable colorectal tumours** ***Nizar El-Murr****, M. Svrcek, K. Wanherdrick, T. Lesuffleur, A. Duval; Paris, France*	**C12.5 Defective initiation of glycosaminoglycan synthesis due to mutations in B3GALT6 causes a pleiotropic connective tissue disorder with severe alterations in proteoglycan assembly and collagen fibrillogenesis** ***Fransiska Malfait****, A. Kariminejad, T. Van Damme, C. Gauche, D. Syx, F. Mehri-Soussi, S. Gulberti, S. Symoens, S. Van Hauwaert, B. Bozorgmehr, M. Kariminejad, I. Hausser, A. Huysseune, S. Fournel-Gigleux, A. De Paepe; Ghent, Belgium*	**C13.5 A large genomic deletion upstream of the lamin B1 gene (LMNB1) likely causes adult-onset autosomal dominant leukodystrophy due to alteration of the regulatory landscape of LMNB1.** ***Elisa Giorgio****, D. Robyr, E. Di Gregorio, D. Lacerenza, G. Vaula, D. Impériale, C. Atzori, A. Brusco, S. Antonarakis, A. Brussino; Torino, Italy*	**C14.5 U1 snRNP interference with polyadenylation - a new pathomechanism for unclassified 3'UTR mutations** ***Jörg Langemeier****, M. Radtke, C. Klein, J. Bohne; Hannover, Germany*
14.30	**C08.6 Identification of Single-minded 2 (Sim2) binding sites by ChIP-Seq; understanding of the regulatory network of chromosome 21 transcription factors** ***Audrey Letourneau****, G. Cobellis, E. Falconnet, A. Vannier, F. Santoni, M. Guipponi, C. Borel, S.E. Antonarakis; Geneva, Switzerland*	**C09.6 Lessons learned from the NHLBI-Exome Sequencing Project (ESP)** ***Suzanne M. Leal****, on behalf of NHLBI-ESP; Houston, United States*	**C10.6 The psychological impact of cryptic chromosomal abnormalities diagnosis announcement.** *F. Houdayer, M. Gargiulo, M. Frischmann, A. Labalme, E. Decullier, M. Cordier, S. Dupuis-Girod, G. Lesca, M. Till, D. Sanlaville, P. Edery,* ***Massimiliano Rossi****; Bron, France*	**C11.6 Western diet and *Mlh1* mutation predispose colonic mucosa to early inactivation of the Wnt signaling antagonist *Dickkopf-1*** ***Marjaana Pussila****, L. Sarantaus, D. Dermadi Bebek, S. Valo, N. Reyhani, S. Ollila, E. Päivärinta, P. Peltomäki, M. Mutanen, M. Nyström; Helsinki, Finland*	**C12.6 Identification of INPPL1 mutations in Opsismodysplasia** ***Celine Huber****, E. Faqeih, D. Bartholdi, C. Bole-Feysot, Z. Borochowitz, D.P. Cavalcanti, A. Frigo, P. Nitschke, J. Roume, H.G. Santos, S.A. Shalev, A. Superti-Furga, A. Delezoide, K.M. Girisha, M. Wright, M. Le Merrer, A. Munnich, V. Cormier-Daire; PARIS, France*	**C13.6 Interferon-beta induces clearance of mutant ataxin-7 and improves locomotion in SCA7 knock-in mice** ***Annie Sittler****, A. Chort, S. Alves, M. Marinello, B. Dufresnois, J. Dornbierer, C. Tesson, M. Latouche, D.P. Baker, M. Barkats, K. El Hachimi, M. Ruberg, A. Janer, G. Stevanin, A. Brice; Paris, France*	**C14.6 Ion transporter deficiency predisposes to pyogenic bacterial infection by partial oxidative burst defect in granulocytes** ***Marjorie Hubeau****, G. Vogt, F. Conti, A. Grant, L. Abel, P. Gros, M. Cellier, C. Picard, J. Bustamante, J. Casanova; Paris, France*
14.45 - 15.15	*Vitamin break / Poster viewing / Exhibition*						

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

PROGRAMME MONDAY, JUNE 10

Time	Grand Amphithéatre	Amphithéatre Bleu	Amphithéatre Bordeaux	Room Maillot	Room 252AB
15.15 - 16.45	**WS10. Dysmorphology 2** Organisers: Dian Donnai; Jill Clayton-Smith	**WS11. Prenatal Diagnostic** Organisers: Milan Macek Jr.; Joris Vermeesch	**WS12. DNA Dysmorphology** Organisers: Hans Scheffer, PhD; Ove Bruland; Bjørn-Ivar Haukanes	**WS13. Community genetics** Organisers: Martina C. Cornel; Ulf Kristoffersson	**WS14. UCSC Genome Browser II** Organiser: Robert M. Kuhn
	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.	The rapid progress of array and the next generation of sequencing-based (NGS) techniques opened new possibilities in the field of prenatal diagnosis / testing. Non-invasive prenatal testing (NIPT) offers noninvasive approach to prenatal aneuploidy screening. Currently, there are several alternative approaches, the entire field is technology driven and there is a need for synthesis and sharing of best practice. Workshop participants will find the most recent information on evolving NIPT technologies, their validation and implementation in clinical practice.	A major challenge of the application of high throughput techniques including next generation sequencing in clinical genetic diagnostics is the interpretation of identified variants, and linking these to the clinical phenotype. In this workshop practical approaches will be illustrated and different cases will be discussed.	Theme: Genetics in Medicine Genetics is moving from research to patient care, not only to clinical genetics but to mainstream medicine. Four lectures will illustrate challenges of translation, with ample time for discussion. Regulation of medicines and implementing NGS are touched upon. The citizens' perspective is reflected in two papers on informed consent.	The advanced workshop on the UCSC Genome Browser will feature use of the Table Browser for data-mining, including intersections and filtering, advanced 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 not necessary. Please bring laptops.
16.45 - 17.15	*Coffee break / Poster viewing / Exhibition*				

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

PROGRAMME MONDAY, JUNE 10

Time	Grand Amphithéatre	Amphithéatre Bleu	Amphithéatre Bordeaux	Room Maillot	Room 252AB
17.15 - 18.45	**S13.** **Stem cells and IPS cells in genetic diseases** Chair: Anne-Lise Bennaceur Griscelli, Alexis Brice	**S14.** **Chromosomal (in) stability** Chair: Philippe Jonveaux, Joris Vermeesch	**ES9.** **Where do we come from?**	**S15.** **The interrelated world of drugs and genes** Chair: Paul de Bakker, Arnold Munnich (tbc)	**S16.** **The expanding world of the primary cilia** Chair: Serge Amselem, Hélène Dollfus
17.15	**S13.1** **Human pluripotent stem cells for modeling genetic diseases** ***Oliver Brüstle***; *Bonn, Germany*	**S14.1** **DNA repair and microcephaly** ***Mark O'Driscoll***; *Brighton, United Kingdom*	**ES9.1** **A genetics history of our species** ***Evelyne Heyer***; *Paris, France* **ES9.2** **Tracing back geographic origine and phenotypes using genetic data** ***Manfred Kayser***; *Rotterdam, Netherlands*	**S15.1** **Drugs and genes - an overview** ***Ann K. Daly***; *Newcastle upon Tyne, United Kingdom*	**S16.1** **Extreme ciliary phenotypes** ***Tania Attie***; *Paris, France*
17.45	**S13.2** **iPS for modelling diseases and drug screening : the example of Steinertmyotonic dystrophy** *Cécile Martinat;* *Evry, France*	**S14.2** **Apparently balanced chromosome rearrangements in human development** ***Cynthia C. Morton***, *J. Rosenfeld, A.M. Lindgren, S. Pereira, I. Blumenthal, C. Chiang, L.G. Shaffer, J.F. Gusella, M.E. Talkowski;* *Boston, United States*		**S15.2** **An immunological basis for pharmacogenetic associations in the MHC** ***James McCluskey***, *Patricia T. Illing, Julian P. Vivian, NadineL. Dudek, Lyudmila Kostenko, Zhenjun Chen,, Mandvi Bharadwaj, John J. Miles, Lars Kjer-Nielsen, Stephanie Gras, Nicholas A. Williamson,, Scott R. Burrows, Anthony W. Purcell, Jamie Rossjohn,, Monash University and QIMR, QLD;* *Melbourne, Australia*	**S16.2** **Primary cilia -kidney disease - DNA Repair** ***Thomas Benzing***; *Cologne, Germany*
18.15	**S13.3** **IPS cells to model genetic diseases and individual variability.** *M. Brimpari, F. Rouhani, N. Kumasaka, a. Bradley, D. Gaffney,* ***Ludovic Vallier***; *Cambridge, United Kingdom*	**S14.3** **Chromosomal mosaicisms in aging and cancer** ***Jan Dumanski***; *Uppsala, Sweden*		**S15.3** **Genetic risk mirrors outcome of anti-TNF therapy in Multiple Sclerosis** ***Lars Fugger***; *Oxford, United Kingdom*	**S16.3** **Moving into and inside cilia: the awesome power of diffusion** *D.K. Breslow, F. Ye, E.F. Koslover, A.J. Spakowitz, W. Nelson,* ***Maxence V. Nachury***; *Stanford, United States*
20.00	*Networking Dinner & Party at the Museum of Natural History*				

SCIENTIFIC

SCIENTIFIC PROGRAMME

Tuesday, June 11, 2013

PROGRAMME

Attend ASHG in Boston

Discover the Clinical Track at the 2013 ASHG Annual Meeting

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  - A Renaissance in Gene Therapy: New Tools and Clinical Trials
  - DNA Damage Response Network Defects and Cancer Predisposition: Where One Plus One Does Not Equal Two
  - Informed Consent for Whole Genome Sequencing: Experience and Implications for Practice
  - Multimodal Treatment of Lysosomal Storage Diseases as a Portal to Emergent Genetic Therapies
  - Whole Genome Sequencing For Every Baby? Where Diagnostic and Screening Applications Collide
  - Design, Content and EMR Integration of Clinical Sequencing Reports

Clinically Focused Workshops

- Diagnostic Dilemmas: Rare and Unknown Cases
- Navigating Clinical Genomic Resources

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PROGRAMME TUESDAY, JUNE 11

Time	Grand Amphithéatre
09.00 - 10.30	**Plenary Session PL3** **Large Scale Cohorts Studies to Identify Novel Highly Penetrant Genetic Disease Causing Variants** Chair: Françoise Clerget-Darpoux, David Fitzpatrick
09.00	**P3.1** **Deciphering Developmental Disorders Project** ***Matt Hurles***; *Cambridge, United Kingdom*
09.30	**P3.2** **Duke Fetal and Neonatal Cohort Study** ***Nicholas Katsanis***; *Durham, United States*
10.00	**P3.3** **Ethical implications of Whole Genome Sequencing in medicine** ***Jane Kaye***; *Oxford, United Kingdom*
10.30 - 11.00	*Coffee break / Poster viewing / Exhibition*

PROGRAMME TUESDAY, JUNE 11

Time	Grand Amphithéatre	Amphithéatre Bleu	Amphithéatre Bordeaux	Room 342AB	Room 252AB	Room 251
11.00 - 12.30	**C15. NGS-based diagnostics** Chair: Hans Scheffer, Michel Goossens	**C16. Developmental syndromes** Chair: Anne Cambon-Thomsen, Nicole Philip	**C17. Basic mechanisms in cytogenetics and molecular genetics** Chair: Martine Doco-Fenzy, Alexandre Reymond	**C18. Big GWAS** Chair: Philippe Froguel (tbc), Markus Perola	**C19. Internal organs and endocrinology: Gene identification and function** Chair: Dominique Bonneau (tbc), Ana Carrió	**C20. Neuro-developmental and neuropsychiatric disorders** Chair: Thomas Bourgeron (tbc), Markus Nöthen
11.00	**C15.1 FGFR related anomalies of foramen magnum : phenotypic-genotypic correlation** ***Federico Di Rocco***, *C. Collet, M. Duplan-Biosse, M. Zerah, C. Sainte-Rose, V. Cormier-Daire, A. Munnich, L. Legeai-Mallet, E. Arnaud; Paris, France*	**C16.1 Patterns and Rates of Exonic *de novo* Mutations in Sporadic Hirschsprung Disease Patients** ***Hongsheng Gui****, *D. Schriemer, B.J.L. Eggen, R.M.W. Hofstra, W. van IJcken, M. van den Hout, P. Griseri, I. Matera, I. Ceccherini, A. Pelet, J. Amiel, S. Lyonnet, M. Garcia-Barcelo, P.K. Tam, M. Ruiz-Ferrer, G. Antiñolo, S. Borrego, C. Berrios, A. Chakravarti; Hong Kong, Hong Kong*	**C17.1 Natural variation in the histone demethylase, KDM4C, influences transcriptional regulation and cell growth** ***Brittany L. Gregory****, *V.G. Cheung; Philadelphia, United States*	**C18.1 Meta-analysis of 233,000 individuals identifies sex- and age-dependent genetic associations for obesity traits** ***Zoltán Kutalik***, *T. Winkler, S. Chu, J. Czajkowski, T. Fall, A. Justice, T.O. Kilpeläinen, Y. Lu, R. Mägi, M. Graff, K.E. North, I.M. Heid, R.J. Loos; Lausanne, Switzerland*	**C19.1 Mutations in *PIK3R1* cause syndromic insulin resistance and lipodystrophy** *C. Thauvin-Robinet, L. Duplomb, M. Avila, J. St-Onge, M. Le Merrer, B. Le Luyer, D. Héron, M. Mathieu-Dramard, P. Bitoun, S. Odent, J. Amiel, D. Picot, V. Carmignac, J. Thevenon, P. Callier, J. Petit, J. Capeau, C. Vigouroux, O. Lascols, F. Huet, L. Faivre,* ***Jean-Baptiste Rivière****; Dijon, France*	**C20.1 RNA foci in C9FTD/ALS patients sequester RNA binding proteins and subsequently alter downstream splicing and expression of their RNA targets.** ***Veronique V. Belzil****, *J. Chew, W. Lee, L. Petrucelli; Jacksonville, United States*
11.15	**C15.2 New genes in epilepsy and its co-morbidities: a linkage and whole exome sequencing approach.** ***Leanne M. Dibbens***, *B. de Vries, S. Donatello, K.R. Smith, M. Bahlo, B.L. Hodgson, S. Chintawar, J. Serratosa, F. Andermann, E. Andermann, A.M.J.M. van den Maagdenberg, M. Pandolfo, S.F. Berkovic, I.E. Scheffer, S.E. Heron; Adelaide, Australia*	**C16.2 A novel chromosomal breakage syndrome caused by a missense mutation in a gene from the SMC5/6 complex** ***Magdalena Harakalova****, *S.N. van der Crabben, M.P. Hennus, J.M. van Montfrans, I. Renkens, K. Duran, M. van Roosmalen, S. van Lieshout, P.M. van Hasselt, S.W. Terheggen-Lagro, P. Terhal, T. Letteboer, R. Hochstenbach, K. Gaiser, E. Kuijk, I.J. Nijman, N. Knoers, W. Kloosterman, E. Cuppen, G. van Haaften; Utrecht, Netherlands*	**C17.2 Enrichment of uniparental disomy events detected in the Deciphering Developmental Disorders rare disease study** ***Daniel A. King****, *M.E. Hurles; Hinxton, United Kingdom*	**C18.2 A causal association between vitamin D status & blood pressure: a Mendelian Randomization study in up to 150,846 individuals** ***Karani S. Vimaleswaran****, *D.J. Berry, A. Cavadino, P. van der Harst, G. Grimnes, A.K. Zaineddin, C. Lu, A. Couto Alves, M.H. de Borst, A. Wong, E. Tikkanen, M. Mangino, K.A. Jablonski, I.M. Nolte, B.L. Langdahl, D.K. Houston, T.S. Ahluwalia, P.J. van der Most, D. Pasko, L. Zgaga, J. Heinrich, E. Thiering, F.G.R. Fowkes, C. Ohlsson, K. Michaëlsson, T.M. Frayling, T. Sørensen, S.B. Kritchevsky, L. Rejnmark, L.K. Billings, T.D. Spector, T. Lehtimäki, D. Kuh, S.E. Humphries, C. Cooper, J.G. Eriksson, W. März, C. Power, M. Kumari, H. Brenner, R. Jorde, H. Snieder, T.J. Wang, A.D. Hingorani, S. Pilz, J.C. Whittaker, M.R. Järvelin, E. Hyppönen; London, United Kingdom*	**C19.2 Genome sequencing identifies mutations causing pancreatic agenesis in a novel *PTF1A* enhancer** ***Sian Ellard***, *M.N. Weedon, I. Cebola, A. Patch, E. de Franco, S.E. Flanagan, S. Rodriguez-Segui, J.A.L. Houghton, H. Lango Allen, C. Shaw-Smith, R. Caswell, A. Murray, P. Ravassard, L. Vallier, J. Ferrer, A.T. Hattersley; Exeter, United Kingdom*	**C20.2 Rapid identification of autosomal recessive and X-chromosomal mutations in small sibling families** ***Janneke H.M. Schuurs-Hoeijmakers****, *A.T. Vulto-van Silfhout, L.E.L.M. Vissers, I.I.G.M. van de Vondervoort, B.W.M. van Bon, J. de Ligt, C. Gilissen, J.Y. Hehir-Kwa, K. Neveling, M. del Rosario, G. Hira, S. Reitano, A. Vitello, P. Failla, D. Greco, M. Fichera, O. Galesi, T. Kleefstra, M.T. Greally, C.W. Ockeloen, M.H. Willemsen, E.M.H.F. Bongers, I.M.H. Janssen, R. Pfundt, J.A. Veltman, C. Romano, M.A. Willemsen, H. van Bokhoven, H.G. Brunner, B.B.A. de Vries, A.P.M. de Brouwer; Nijmegen, Netherlands*
11.30	**C15.3 Targeted sequencing of GPI anchor synthesis pathway genes identifies mutations in PGAP2 as a new cause of hyperphosphatasia with mental retardation** ***Peter M. Krawitz****, *Y. Murakami, A. Rieß, M. Hietala, U. Krüger, Z. Na, T. Kinoshita, S. Mundlos, P.N. Robinson, J. Hecht, D. Horn; Berlin, Germany*	**C16.3 SPRTN deficiency causes a novel segmental progeroid syndrome with chromosomal instability** ***Davor Lessel****, *J. Oshima, J. Lopez-Mosqueda, I. Marinovic-Terzic, M. Philipp, R. Fertig, S. von Ameln, M. Degoricija, H. Thiele, G. Nürnberg, P. Nürnberg, G.M. Martin, C.M. Aalfs, K. Ramadan, J. Terzic, I. Dikic, C. Kubisch; Ulm, Germany*	**C17.3 Exonic splicing mutations in Mendelian disorders: more prevalent than currently estimated** *O. Soukarieh, D. Di Giacomo, S. Krieger, T. Frébourg, M. Tosi, P. Gaildrat,* ***Alexandra Martins****; Rouen, France*	**C18.3 Large-scale genome-wide association meta-analysis using imputation from the dense 1000 Genomes Project identifies novel susceptibility loci for glycemic and obesity traits: ENGAGE Consortium report** ***Momoko Horikoshi***, *R. Mägi, I. Surakka, A. Sarin, A. Mahajan, L. Marullo, T. Ferreira, T. Esko, C.M. Lindgren, A.P. Morris, M.I. McCarthy, S. Ripatti, I. Prokopenko; Oxford, United Kingdom*	**C19.3 Somatic mutations in ATP1A1 & ATP2B3 lead to aldosterone-producing adenomas & secondary hypertension** ***Thomas Wieland****, *F. Beuschlein, S. Boulkroun, A. Osswald, H.N. Nielsen, U.D. Lichtenauer, D. Penton, V.R. Schack, L. Amar, E. Fischer, A. Walther, P. Tauber, T. Schwarzmayr, S. Diener, E. Graf, B. Allolio, B. Samson-Couterie, A. Benecke, M. Quinkler, F. Fallo, P. Plouin, F. Mantero, T. Meitinger, P. Mulatero, X. Jeunemaitre, R. Warth, B. Vilsen, M. Zennaro, T.M. Strom, M. Reincke; Neuherberg, Germany*	**C20.3 Associations between gene expression and phenotypes in 16p11.2 rearrangements** ***Katrin Männik***, *E. Migliavacca, A. Macé, G. Gianuzzi, M.N. Loviglio, F. Zufferey, N.D. Beckmann, L. Harewood, L. Hippolyte, A.M. Maillard, V. Siffredi, R.M. Witwicki, G. Didelot, J.S. Beckmann, Z. Kutalik, S. Jacquemont, A. Reymond; Lausanne, Switzerland*

PROGRAMME TUESDAY, JUNE 11-CONTINUED

Time	Grand Amphithéatre	Amphithéatre Bleu	Amphithéatre Bordeaux	Room 342AB	Room 252AB	Room 251
cont.	**C15. NGS-based diagnostics**	**C16. Developmental syndromes**	**C17. Basic mechanisms in cytogenetics and molecular genetics**	**C18. Big GWAS**	**C19. Internal organs and endocrinology: Gene identification and function**	**C20. Neurodevelopmental and neuropsychiatric disorders**
11.45	**C15.4 Diagnostic exome sequencing to elucidate the genetic basis of likely recessive disorders in consanguineous families** ***Periklis Makrythanasis****, *M. Nelis, F.A. Santoni, M. Guipponi, A. Vannier, F. Béna, S. Gimelli, E. Stathaki, S. Temtamy, A. Mégarbané, A. Masri, M.S. Aglan, M.S. Zaki, A. Bottani, S. Fokstuen, L. Gwanmesia, K. Aliferis, M.E. Bustamante, G. Stamoulis, S. Psoni, S. Kitsiou-Tzeli, H. Frissyra, E. Kanavakis, N. Al-Allawi, A. Sefiani, S. Al-Hait, S.C. Elalaoui, N. Jalkh, L. Al-Gazali, F. Al-Jasmi, H. Chaabouni Bouhamed, E. Abdalla, D.N. Cooper, H. Hamamy, S.E. Antonarakis; Geneva, Switzerland*	**C16.4 Mutations in HDAC8 cause a clinically recognizable Cornelia de Lange Syndrome (CdLS)-like disorder** ***Frank J. Kaiser**, D. Braunholz, M. Ansari, S. Lynch, N. DiDonato, J. Eckhold, M. Gil-Rodríguez, J. Pié, F. Ramos, N. Revencu, I. Krantz, G. Gillessen-Kaesbach, D. FitzPatrick, M. Deardorff; Lübeck, Germany*	**C17.4 New insights into human germline chromothripsis: underlying mechanisms and definition** ***Lusine Nazaryan****, *M. Bak, A. Lind-Thomsen, C. Halgren, E. Stefanou, C. Hansen, K.F. Henriksen, M. Bugge, J. Lespinasse, G. Houge, N. Tommerup; Copenhagen, Denmark*	**C18.4 The ‚Genome of The Netherlands' outperforms ‚1000 genomes' as a reference set for imputing rare variants in the European population** ***Patrick Deelen****, *F. van Dijk, L. Francioli, J. Hottenga, E. van Leeuwen, M. Kattenberg, L. Karssen, K. Estrada, E. Kreiner-Møller, F. Rivadeneira, A. Kanterakis, H. Westra, A. Menelaou, D. van Enckevort, .. Members of the GoNL consortium, L. Franke, P. de Bakker, C. Wijmenga, M. Swertz; Groningen, Netherlands*	**C19.4 Mutation-dependent recessive inheritance in NPHS2-associated steroid-resistant nephrotic syndrome. Beyond Mendel's laws** ***Kálmán Tory**, D.K. Menyhárd, F. Nevo, O. Gribouval, A. Kerti, P. Stráner, C. Arrondel, T. Tulassay, G. Mollet, A. Perczel, C. Antignac; Paris, France*	**C20.4 *GATAD2B* loss-of-function mutations cause a recognizable syndrome with intellectual disability and are associated with learning deficits and synaptic undergrowth in *Drosophila*** ***Marjolein H. Willemsen****, *B. Nijhof, M. Fenckova, W.N. Nillesen, E.M.H.F. Bongers, A. Castells Nobau, L. Asztalos, E. Viragh, B.W.M. van Bon, J.A. Veltman, H.G. Brunner, B.B.A. de Vries, J. de Ligt, Z. Asztalos, H.G. Yntema, H. van Bokhoven, D.A. Koolen, L.E.L.M. Vissers, A. Schenck, T. Kleefstra; Nijmegen, Netherlands*
12.00	**C15.5 Exome Sequencing in the Diagnostics of non-motile Ciliopathies (113 cases)** ***Miriam Schmidts****, *E. Chanudet, V. Plagnol, F. Lescai, H. Jungbluth, G. Haliloglu, H. Kayserili, M.E. Hurles, P.J. Scambler, U. Consortium, P.L. Beales, H.M. Mitchison; London, United Kingdom*	**C16.5 A homozygous PDE6D mutation in Joubert syndrome impairs targeting of farnesylated INPP5E protein to the primary cilium** ***Sophie Thomas**, K.J. Wright, S. Le Corre, A. Mocalizzi, M. Romani, A. Abhyankar, J. Saada, I. Perrault, J. Amiel, J. Litzler, E. Filhol, N. Elkhartoufi, M. Kwong, J. Casanova, N. Boddaert, W. Baehr, S. Lyonnet, A. Munnich, L. Burglen, N. Chassaing, F. Encha-Ravazi, M. Vekemans, J.G. Gleeson, E. Valente, P.K. Jackson, I.A. Drummond, S. Saunier, T. Attié-Bitach; Paris, France*	**C17.5 Whole-genome sequencing analysis of human induced pluripotent stem cells uncovers lineage-manifested CNVs** ***Alexander E. Urban**, *A. Abyzov, J. Mariani, D. Palejev, M. Haney, Y. Zhang, L. Tomasini, A. Szekely, S.M. Weissman, M. Gerstein, F. Vaccarino; Palo Alto, United States*	**C18.5 Genome-wide association study identifies common variation associated with congenital heart disease** ***Judith Goodship**, *H. Cordell, J. Bentham, A. Topf, D. Zelenika, S. Heath, C. Mamasoula, D. Brook, S. Bhattacharya, D. Winlaw, K. Devriendt, S. Mital, A. Postma, M. Lathrop, M. Farrall, B. Keavney; Newcastle upon Tyne, United Kingdom*	**C19.5 A recurrent homozygous missense mutation in TTC21B encoding the ciliary protein IFT139 unexpectedly causes steroid-resistant nephrotic syndrome** ***Evelyne Huynh Cong****, *A. Bizet, S. Woerner, E. Filhol, O. Gribouval, S. Thomas, F. Silbermann, C. Bole-Feysot, P. Nitschke, G. Canaud, J. Hachicha, M. Gubler, G. Mollet, S. Saunier, C. Antignac; Paris, France*	**C20.5 Disruption of Methyl CpG Binding Protein 5 contributes to a spectrum of psychopathology and neurodevelopmental abnormalities** ***Jennelle C. Hodge**, *E. Mitchell, V. Pillalamarri, T.L. Toler, F. Bartel, H.M. Kearney, Y.S. Zou, W. Tan, C. Hanscom, S. Kirmani, R.R. Hanson, S.A. Skinner, C. Rogers, D.B. Everman, E. Boyd, C. Tapp, S.V. Mullegama, D. Keelean-Fuller, C.M. Powell, S.H. Elsea, C.C. Morton, J.F. Gusella, B. DuPont, A. Chaubey, A.E. Lin, M.E. Talkowski; Rochester, United States*
12.15	**C15.6 Detection of clinically relevant copy number variants with whole exome sequencing** ***Joep de Ligt****, *P.M. Boone, R. Pfundt, L.E.L.M. Vissers, T. Richmond, J. Geoghegan, K. O'Moore, N. de Leeuw, C. Shaw, H.G. Brunner, J.R. Lupski, J.A. Veltman, J.Y. Hehir-Kwa; Nijmegen, Netherlands*	**C16.6 *Integrator Complex Subunit 8* mutations associated with abnormal brain development and spliceosomal defect** ***Renske Oegema****, *R. Schot, D. Heijsman, L. van Unen, S. Kheradmand Kia, J. Hoogeboom, A. Kremer, F.W. Verheijen, P. van der Spek, R. Hofstra, M. Fornerod, G.M.S. Mancini; Rotterdam, Netherlands*	**C17.6 Modelling and rescuing neurodevelopmental defect of Down syndrome using induced pluripotent stem cells from monozygotic twins discordant for trisomy 21** ***Youssef Hibaoui****, *i. Grad, a. Letourneau, m.r. Sailani, s. Dahoun, f.a. Santoni, s. Gimelli, m.l. Guipponi, m. Pelte, f. Bena, s.e. Antonarakis, a. Feki; Geneva, Switzerland*	**C18.6 Common genetic variants predispose to a rare disease with high risk of sudden cardiac death** ***Richard Redon**, *J. Barc, Y. Mizusawa, C.A. Remme, J.B. Gourraud, F. Simonet, P.J. Schwartz, L. Crotti, P. Guicheney, A. Leenhardt, C. Antzelevitch, E. Schulze-Bahr, E.R. Behr, J. Tfelt-Hansen, S. Kaab, H. Watanabe, M. Horie, N. Makita, W. Shimizu, P. Froguel, B. Balkau, O. Lantieri, M. Gessler, D. Roden, V.M. Christoffels, H. Le Marec, A.A. Wilde, V. Probst, J.J. Schott, C. Dina, C.R. Bezzina; Nantes, France*	**C19.6 Characterization of the large patient cohort of the International Inflammatory Bowel Disease Genetics Consortium (IIBDGC)** ***Isabelle Cleynen**, *o. (IIBDGC); Cambridge, United Kingdom*	**C20.6 Analysis of copy number variations at 15 schizophrenia-associated loci in a large, independent cohort** ***Elliott Rees****, *J.T.R. Walters, L. Georgieva, A.R. Isles, K.D. Chambert, A. Richards, G. Davies, S.E. Legge, J.L. Moran, S.A. McCarroll, M.C. O' Donovan, M.J. Owen, G. Kirov; Cardiff, United Kingdom*
12.30 - 13.30	Lunch break / Poster Removal / Exhibiton					

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

PROGRAMME TUESDAY, JUNE 11

Time	Grand Amphithéatre
13.30 - 14.15	**Plenary Session PL4** **Mendel Lecture** Chair: Han Brunner, Brunhilde Wirth
13.30	**P4.1** **Mendel Lecture: Rett syndrome and MECP2 Disorders: From the Clinic to Genes and Neurobiology** ***Huda Zoghbi****;* *Houston, United States*
14.15 - 15.45	**Plenary Session PL5** **ESHG Award and Closing Session** Chair: Han Brunner, Brunhilde Wirth
14.15	**P5.1** **ESHG Award Lecture** ***Felix Mitelman****;* *Lund, Sweden* *Laudation by Thé-Hung Bui*
15.00	**Awards Ceremony** **ESHG Education Award awarded to Peter Farndon** *Laudation by Dian Donnai* **EJHG Nature 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**

A set of 3 Apple iPads mini will be drawn within the attendees of the final plenary session.

SCIENTIFIC

SCIENTIFIC PROGRAMME

WORKSHOPS

SATELLITES

CORPORATE SATELLITES

TECHNICAL INFORMATION

SCIENTIFIC INFORMATION

YOUNG INVESTIGATOR AWARD CANDIDATES

POSTER AWARD FINALISTS

PROGRAMME

PROGRAMME WORKSHOPS-SATELLITES

Workshops

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

Sunday, June 9, 2013, 11.40 - 13.15 hrs

WS01 NG sequencing in clinical practice, Filtering & reporting (J. Veltman& J.Vermeesch)	Grand Amphithéatre
WS02 Debate: Hot topics in preimplantation genetic testing (T.H. Bui & D. Sanlaville)	Amphithéatre Bleu
WS03 UCSC Genome Browser I (R. Kuhn)	Room 252AB

Sunday, June 9, 2013, 15.15 - 16.45 hrs

WS04 Dysmorphology Workshop 1 (D. Donnai & J. Clayton-Smith)	Grand Amphithéatre
WS05 Analysis, interpretation and reporting of array data (N. de Leeuw & C.van Ravenswaaij-Arts)	Amphithéatre Bleu
WS06 Clinical Cancer Genetics Club (M. Genuardi & D. Stoppa-Lyonnet)	Amphithéatre Bordeaux
WS07 Quality assurance (E. Dequeker & M. Morris)	Room Maillot
WS08 Biomedical Data Analysis with Galaxy (A. Nekrutenko & E. Afgan)	Room 252AB
WS09 Inequality in Genetic Services (S. Lyonnet & J. Schmidtke)	Room 242AB

Monday, June 10, 2013, 15.15 - 16.45 hrs

WS10 Dysmorphology 2 (D. Donnai & J. Clayton-Smith)	Grand Amphithéatre
WS11 Prenatal Diagnostic (M. Macek Jr. & J. Vermeesch)	Amphithéatre Bleu
WS12 DNA Dysmorphology (H. Scheffer, O. Bruland & B. Haukanes)	Amphithéatre Bordeaux
WS13 Community genetics (M. Cornel & U. Kristoffersson)	Room Maillot
WS14 UCSC Genome Browser II (R. Kuhn)	Room 252AB

Official satellite meetings open to all participants

As per date of printing.

Saturday, June 8, 2013

ESHG Satellite workshop on External Quality Assessment of Genetic Counselling
09:00 – 12:45 hrs, Room Maillot
For a maximum of 200 participants from clinical genetics.
Separate registration necessary.

HGVS: Clinical Applications of Next Generation Sequencing
08:30 – 16:00 hrs, Hotel Concorde La Fayette

Satellite Meeting on Telehealth in (clinical) Genetics
14:00 - 15:30 hrs, Room 342A

Sunday, June 9, 2013

Satellite Meeting Future Panel on Public Health Genomics (FP7 PACITA project)
11:40 - 13:10 hrs, Room 242AB

Monday, June 10, 2013

Beyond GWAS: Biological and Clinical Insights from Research in European Biobanks
A Symposium Supported by the ENGAGE Consortium
11:30 – 13:10 hrs, Room 242AB
No extra registration necessary! The meeting is open to all ESHG participants.

"Innovative Techniques in Genome Diagnostics" by EuroGentest (II)
11:40 - 13:10 hrs, Room Maillot
No extra registration necessary! The meeting is open to all ESHG participants

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.

BIOBASE – Saturday 8 June 2013, 11.45 – 13.15 hrs - Room 202/203 – Level 2 Stand # 622

Identification of causal variants in type 2 diabetes (T2D) and obesity using NGS and BIOBASE's HGMD® Professional

Speakers *Julien Philippe, CNRS, Institut Pasteur, Lille, France*
Frank Schacherer, CTO, BIOBASE

Molecular characterization of monogenic forms of T2D/obesity is critical for diagnosis and research. NGS has opened new ways to develop innovative methods for rapid identification of known forms of T2D/obesity but also to identify novel genetic etiologies.

We developed a method for a highly sensitive molecular diagnosis of 43 forms of monogenic diabetes or obesity, based on microdroplet PCR enrichment and NGS. We assessed this technology in 40 patients carrying known causal mutations. Except for a variant, we re-identified all causal mutations, associated with an almost perfect sequencing of the targets. We only failed to call a highly complex indel. In 3 patients, we detected other mutations with a putatively deleterious effect.

After screening patients for known genes, some of them remain uncharacterized molecularly. Whole exome sequencing is a powerful approach to discover new loci linked to new signaling pathways involved in the physiopathology of T2D/obesity. Through whole-exome we identified a new MODY gene: KCNJ11.

These high throughput techniques lead to the identification of hundreds of genetic variants which need validation, good calling and annotations. Therefore, HGMD® reporting all published causal variants and Genome TraxTM prioritizing these variants are essential tools.

Fluidigm – Saturday 8 June 2013, 11.45 – 13.15 hrs - Room 251 – Level 2 Stand # 174

Break through the bottlenecks: How simplified workflows tackle the challenges in high-throughput sample preparation, clinical research, and heterogeneous stem cell populations

Investigating Cell Fate Specification at Single-Cell-Resolution
Julia Tischler, PhD, Postdoctoral Researcher, Wellcome Trust/Cancer Research UK Gurdon Institute, Cambridge, UK

Use of a Fluidigm target enrichment strategy for NGS analysis of BRCA1/BRCA2/TP53 genes in DNA extracted from both peripheral blood and FFPE tissue
Jennifer Lickiss, Clinical Scientist, West Midlands Regional Genetics Laboratory, Birmingham Women's Hospital, Birmingham, UK

Genetic research using the Fluidigm Integrated Fluidic Circuits
Dani Bercovich, PhD, Scientific Director, Galil Genetic Analysis, Ltd., Kazerin, Israel

IntegraGen – Saturday 8 June 2013, 11.45 – 13.15 hrs - Room 212/213 – Level 2 Stand # 252

Valuable exome and transcriptome sequencing: from complex samples to highly accurate results and analysis

In this workshop, we will address these challenges through the shared experience of some customer's case studies:

IntegraGen Genomic Services: a one-stop shop to integrate robust NGS processes
Emmanuel Martin, Director of Genomic Services, IntegraGen, Evry, France

Exome sequencing reveals additional mutations in sporadic Juvenile MyeloMonocytic Leukemia, but not in syndromic JMML
Professor Hélène Cavé, Hôpital Robert-Debré, Paris-Diderot University, France

Exome sequencing identifies DEPDC5 mutations as a major cause for familial focal epilepsies
Dr Stéphanie Baulac, INSERM U975 CRICM, La Pitié-Salpêtrière, Paris, France

ERIS, an online exome data analysis tool
Emmanuel Martin, IntegraGen, Evry, France

Exome analysis in colorectal cancer
Professor Pierre Laurent-Puig, INSERM U775, Paris Descartes University, France

IntegraGen is your trustful partner for genomic projects: www.integragen.com.

PROGRAMME CORPORATE SATELLITES

DNA Genotek – Saturday 8 June 2013, 14.00 – 15.30 hrs - Room 202/203 – Level 2 Stand # 672

Novel applications in complex biomarker analysis – telomere analysis, exome sequencing and methylation profiling

DNA Genotek is pleased to host three renowned experts representing industry and academia who will share their insights into complex biomarker analysis using novel technologies, highlighting why quality samples are important.

Calvin B. Harley, PhD, President and Chief Scientific Officer of Telome Health, is a world-renowned expert in telomere biology and aging. Dr. Harley co-founded Telome Health with Nobel Prize winner Dr. Elizabeth Blackburn and will speak about how leveraging telomere analysis can increase the life span of normal cells.

William H. Biggs, PhD, Senior Director Genetics and Genomics of Aviir Corporation, will speak about bringing an Inherited Cardiovascular Disorder test to market using target sequencing technologies**.**

Paul Arnold, MD, PhD, FRCPC, Associate Professor at the University of Toronto, and a Scientist in the Genetics and Genome Biology program and Centre for Brain and Behaviour at SickKids hospital, will speak on the use of methylation analysis in assessment and pharmacological treatment of children with Obsessive-Compulsive Disorders (OCD).

Theradiag and Asuragen – Saturday 8 June 2013, 14.00 – 15.30 hrs - Room 212/213 – Level 2 Stand # 136

Fragile X diagnosis : CGG sizing and methylation status

Fragile X Syndrome (FXS) is caused by the expansion of CGG repeats in the *FMR1* gene and is the most common known genetic cause of autism and inherited mental retardation. Fragile X carriers may be at risk of Fragile X-associated tremor/ataxia syndrome (FXTAS) or primary ovarian insufficiency (FXPOI). Current in-house techniques such as PCR or Southern Blot are often laborious, time consuming or limiting in terms of resolution.

Asuragen and Theradiag are pleased to present the AmplideX™ *FMR1* PCR product range for professional use in clinical laboratories. These devices allow the amplification of the FMR1 gene, including full length and CGG peaks for each allele, provides superior sensitivity than Southern blotting, resolution of female zygosity, detection of interrupting AGG sequences and low level mosaicisms. For determination of methylation status, particularly for large pre-mutations, AmplideX™ *FMR1* mPCR (methylation-specific PCR) helps biologists and physicians in confirming Fragile X Syndrome.

We would be delighted to welcome you to our lunchtime session **"Fragile X Diagnosis: CGG Sizing and Methylation Status"** with
Dr. Elles Boon, Leiden University Medical Center, Leiden, the Netherlands and
Dr. Gaëtan Lesca, Hospices Civils de Lyon, Lyon, France

For additional information please visit our booth number 136 or contact us at amarie@theradiag.com or dmukherjee@asuragen.com.

Affymetrix – Sunday 9 June 2013, 11.40 – 13.10 hrs - Room 251 – Level Stand # 688

Find out what others are missing...

With the rapid adoption of microarrays for cytogenetic analysis, researchers have shown 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® high density arrays are enabling cytogenetic researchers to detect and analyze constitutional and cancer chromosomal aberrations, with more confidence than with any other traditional or array-based technology.

The utility of CytoScan® HD for analyzing hematological malignancies and pre-natal samples
Stuart Schwartz, PhD, Laboratory Corporation of America, Burlington, Vermont, USA

The necessity of high quality SNP information in array analysis and interpretation
Rolph Pfundt, PhD, Radboud University Nijmegen Medical Centre, Nijmegen, the Netherlands

Generating high quality whole genome copy number data using limited DNA from FFPE with Affymetrix's OncoScan™ Solution
Stéphane Minvielle, PhD, Centre de Recherche en Cancérologie, Nantes-Angers, France

Lunch and refreshments will be provided. Spaces are limited; please arrive early to avoid disappointment. Visit us at stand number 688.

Life Technologies – Sunday 9 June 2013, 11.40 – 13.10 hrs - Room 241 – Level 2 Stand # 400

Rapid mutation analysis using Ion PGM™ and Ion Proton™ sequencing

Leading scientists present their research on inherited diseases using Ion PGM™ and Ion Proton™ semi-conductor sequencing applications.

Dr Marjolijn Ligtenberg, UMC St Radboud, Nijmegen, the Netherlands
Comprehensive screening of BRCA1/2 genes on the Ion PGM™ system, using an Ion AmpliSeq™ community designed gene panel

Peter Ray, PhD, Hospital for Sick Children, Toronto, Canada
Verification and validation of CFTR mutation detection; comparison of Ion PGM™ data to Sanger sequencin

Adam Ameur, PhD, Uppsala Genome Centre, Uppsala, Sweden
New mutation discovery using massively parallel exome and transcriptome sequencing on the Ion Proton™ Sequencer

Dr. Martin Krahn, Laboratoire de Génétique Moléculaire, Hôpital d'enfants de la Timone, Marseille, France
Application of Ion Proton™ exome sequencing for genetic diagnosis of myopathies

Dr. Dominique Vidaud, Department of Genetics and Development of Skeletal Muscles, Cochin Hospital, Paris, France
Next generation sequencing for NF1 molecular analysis

Myriad Genetics – Sunday 9 June 2013, 11.40 – 13.10 hrs - Room 202/203 – Level 2 Stand # 610

Cancer Gene Panels: The Future of Hereditary Cancer Testing

Identifying individuals at increased risk for hereditary cancer prompts enhanced cancer surveillance and early detection with the potential to reduce disease specific morbidity and mortality. Targeted gene testing is usually guided by the differential diagnosis of cancer syndromes. However, hereditary cancer syndromes have significant genetic heterogeneity which often requires testing multiple genes. By testing numerous possible causal genes simultaneously, hereditary cancer gene panels reduce the time and cost compared to sequential gene testing and improve the sensitivity of mutation detection.

As a pioneer in hereditary cancer testing, Myriad Genetics is pleased to introduce a Next Generation Sequencing-based test to identify the multiple genes implicated in hereditary and familial cancer risk.

Agenda **Introduction**
Phenotypic Overlap of the Common Hereditary Cancer Syndromes
Myriad's Hereditary Cancer Panel
Designing Variant Classification for a Hereditary Cancer Panel
Prevalence of Mutations in Patients Referred for HBOC: Implications for Panel Approach
Criteria for Clinically Robust Commercial Application of NGS

Speakers *Richard Wenstrup, M.D., Chief Medical Officer, Myriad Genetic Laboratories, Inc., Salt Lake City, USA*
Karen Copeland, M.S., C.G.C. Director-International Medical Affairs, Myriad Genetics GmbH, Zurich, Switzerland
Brian Allen, M.S., C.G.C. Senior Manager, Clinical Development, Myriad Genetic Laboratories, Inc., Salt Lake City, USA
Julie M. Eggington, Ph.D., Variant Classification Program Director, Myriad Genetic Laboratories, Inc., Salt Lake City, USA

Cartagenia – Sunday 9 June 2013, 15.15 – 16.45 hrs - Room 241 – Level Stand # 670

NGS and Arrays in postnatal and prenatal clinical practice

Are you implementing Next Generation Sequencing in routine clinical practice? Are you looking to automate and grow your Array activities for clinical assessment of copy number variants? Are you expanding your activities to prenatal testing? If so, you must be wondering how you're going to set up lab workflow, manage all that data, efficiently extract the clinically relevant information, and produce high-quality lab reports. Learn from experts in the field on state of the art approaches, evidence based medicine, diagnostic standard of care, robust clinical interpretation support, and get updated on current research.

Prof. Ronald Wapner, MD, Maternal-Fetal Medicine Division, Columbia University Medical Center, NY, USA
Prenatal microarray testing in clinical care

Prof. Dr. Gert Matthijs, Center for Human Genetics, Leuven University Hospitals, Belgium
Next Generation Sequencing in Diagnostics: gene panels or exomes?

Steven Van Vooren, PhD, Cartagenia, Belgium
The Cartagenia BENCH platform: Confidently store, interpret, report and share genomic variants

Learn about how Cartagenia helps you to confidently interpret, store, share and report genomic variants. See how the BENCHlab platform supports robust structural and molecular variant analysis, interpretation and reporting pipelines.

PROGRAMME CORPORATE SATELLITES

Multiplicom – Sunday 9 June 2013, 15.15 – 16.45 hrs - Room 202/203 – Level 2 Stand # 416

Broad clinical diagnostic applications of MASTR™ kits for germline and somatic mutations with current MPS technologies

Multiplicom's MASTR™ technology (Multiplex Amplification of Specific Targets for Resequencing) is widely used by genetic centres and molecular pathology laboratories. Several applications are used in the diagnostic routine. We bring the latest information and data from the field.

Status of Standardization of breast cancer diagnostics using NGS (BRCA MASTR)
Prof. Dr. Alfons Meindl, Klinik Rechts der Isar, Munich, Germany

Applications in oncogenetics of MASTR on PGM
Dr. Pierre Hutter, Hôpital du Valais (HVS) – Institut Central (ICHV), Sion, Switzerland

First results with the custom-designed Stargardt MASTR panel on the PGM platform
Dr. rer. nat. Helmut Roth, University of Regensburg, Institute of Human Genetics, Regensburg, Germany

MASTR based analysis of major cardiomyopathies
Dr. Pascale Richard, Hôpital Pitié Salpêtrière, Paris, France

Recent developments in MASTR technology for clinical diagnostics
Prof. Jurgen Del-Favero, Multiplicom, Niel, Belgium

Roche Applied Science – Sunday 9 June 2013, 15.15 – 16.45 hrs - Room 251 – Level Stand # 110

Roche real-time qPCR Workshop

The NEW LightCycler® 96 System
Hannes Kirzinger, Roche Diagnostics, Germany

The LightCycler® 96 System is a novel touchscreen-based bench-top instrument for up to 96 samples. It supports basic and advanced qPCR applications without the need for calibration, color compensation or routine maintenance. Experiments can be handled via USB stick, LAN or a connected PC. RDML-compatible result files fully support current PCR best practice guidelines. The presentation includes data on new HRM and qualitative detection analysis modules.

DNA hypermethylation is an earlier and more penetrant event than mutation in colorectal carcinoma
Prof. Béla Molnár, Semmelweis University, Budapest, Hungary

A project aiming at the identification of novel hypermethylated genes in colon carcinoma is presented.
Microsatellite status, KRAS and BRAF mutational status were assessed in samples obtained from fresh-frozen biopsies. Gene expression levels were studied by whole genomic mRNA expression arrays. Genes with decreased expression and CpG island in their promoters were studied by PCR and HRM analysis on a LightCycler® 480 System and sequenced on the GS Junior System. Results show that colorectal carcinoma is dominantly characterized by epigenetic rather than genetic alterations.

LightCycler® is a trademark of Roche.

Illumina – Sunday 9 June 2013, 18.45 – 20.15 hrs - Room 251 – Level Stand # 420

Empowering Genomic Science – Latest Advances in Next-Generation Sequencing and Genotyping Technologies

Advancements in our understanding of genetics have the potential to change the practice of medicine and enable genomics-based healthcare. With streamlined workflows and advanced bioinformatics tools, there's never been a better time to explore genomic technologies for research and translational/clinical needs. This workshop will highlight the latest advances in Illumina's sequencing and array solutions, plus our family of data analysis options.

PerkinElmer – Sunday 9 June 2013, 18.45 – 20.15 hrs - Room 241 – Level Stand # 196

Innovative total solutions for molecular genetics

Join us at our satellite meeting to hear about PerkinElmer's latest innovations including our solutions for automated DNA/RNA isolation, and newest techniques for molecular genetics. Visit us also at the stand #196 to learn more about our complete product offering, e.g. Next Generation Sequencing sample preparation solutions provided by Caliper, a PerkinElmer company based in Waltham, MA, US.

1. **Accelerating medical research with fully automated, large volume DNA extractions**
   Mr. Ron den Boer from the Erasmus MC, University Medical Center, Rotterdam, the Netherlands
2. **Prenatal BACs-on-Beads: after a two years worldwide experience**
   Prof. François Vialard, Poissy-Saint Germain Hospital, University of Versailles-St. Quentin, France
3. **New Developments in Fragile X testing**
   Dr. Mack Schermer, R&D Director, Molecular Diagnostics, R&D, PerkinElmer Inc, Waltham, MA, USA
4. **One step closer to cost effective Fragile X syndrome population carrier screening; validation of a novel approach to assessing repeat size**
   Dr.Tanya N. Nelson, PhD, FCCMG, Acting Director/Division Head, Molecular Genetics Laboratory/Laboratory Genetics, Dept. of Pathology & Lab Medicine, Children's & Women's Health Centre of BC., Vancouver, Canada

Complimentary wine and sandwiches will be served.

Abbott Molecular – Monday 10 June 2013, 11.40 – 13.10 hrs - Room 241 – Level 2 Stand # 496

Update on PCR strategies for the detection of expanded alleles of the FMR1 gene and their methylation status

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

Talk 2 **Use of PCR Tools for FMR1 in Routine Laboratory Testing**
Dr Monica Basehore, Greenwood Genetic Centre, Greenwood, SC, USA

Talk 3 **Update on Detection of FMR1 Promoter Methylation Status Detection Using a Laboratory Developed High Resolution Melt Assay Design**
Aaron Hamilton, Celera, San Francisco, CA, USA

Agilent Technologies – Monday 10 June 2013, 11.40 – 13.10 hrs - Room 251 – Level Stand # 100

Advances in Human Genetics Research using market leading Target Enrichment and Comparative Genome Hybridization applications

The "Motor" experience: an innovative Next Generation workflow for research on neuromuscular disorders
Marco Savarese, Second University of Naples, Telethon Institute of Genetics and Medicine, Italy

We are validating a NGS-based workflow for a complete and exhaustive analysis of samples related to neuromuscular disorders. Using Haloplex, SureSelect All Exon kit and RNA Target Enrichment System, our preliminary results demonstrate the ability of NGS to make significant progress in understanding these pathologies.

Targeted resequencing results in a significant increase in identifying genetic causes of cardiomyopathies
Rowida Almomani, University Medical Center Groningen, the Netherlands

We constructed a targeted enrichment kit (Agilent SureSelect) using 48 genes associated with hereditary cardiomyopathies. We determined whether the sensitivity, specificity and robustness of targeted NGS are equal to those of Sanger Sequencing (SS). An improved kit targeting 55 genes was constructed and implemented.

The whole tumor genome – opening new perspectives for clinical research
Frédéric Chibon, Institut Bergonié, Bordeaux, France

We use CGH on FFPE sample to distinguish between two mimicking tumors, detect and discriminate amplifications from targeted therapy gains, determine whether tumor events are related or independent. Genome complexity analysis will help better define the genome of sarcoma subtypes.

PROGRAMME CORPORATE SATELLITES

CLC bio – Monday 10 June 2013, 11.40 – 13.10 hrs - Room 202/203 – Level Stand # 562

Identification and comparison of genetic variants - How to manage your human genetics data analysis

Program
- **Implementation of NGS in diagnostics**
- **Somatic mutations and germline variants in human colon cancers**
- **Automatic identification and annotation of somatic mutations in cancer**
- **Resequencing workflows and comparative analyses of large-scale ngs data**

Guest speakers
- *Dr. Phillip J. Buckhaults, PhD, Associate Professor, Department of Medicine, Division of Hematology & Oncology,*
- *The University of Alabama at Birmingham, USA*
- *Dr. Kasper Thorsen, M.Sc., Ph.D., Molecular Diagnostics Laboratory, Aarhus University Hospital Skejby, Denmark*

Speakers
- *Dr. Anika Joecker, PhD, Senior Bioinformatics Specialist, CLC bio*
- *Dr. Holger Karas, PhD, Senior Field Application Specialist, CLC bio*

NB. Seating is limited, so first come, first served. Lunch is complimentary. We look forward to meeting you.

BioDiscovery – Monday 10 June 2013, 15.15 – 16.45 hrs - Room 202/203 – Level Stand # 536

New! Nexus 7 - A Single Platform for Copy Number and Sequence Variation Analysis with Built-in Reference Data (ISCA, AGRE, and TCGA)

BioDiscovery Nexus Copy Number has enabled users to efficiently detect, visualize, and interpret copy number and allelic event changes across many application areas for several years. With the newly released Nexus Copy Number version 7, sequence variations can now be interpreted alongside copy number changes for an integrated view of genomic aberrations. Built-in reference data (such as ISCA, AGRE, DGV, TCGA, etc.) facilitates fast and accurate interpretation providing a complete solution to routine clinical sample analysis in addition to research applications. See what's new in Nexus Copy Number 7 and hear from current users on how Nexus Copy Number has streamlined their workflow.

Agenda

SNP array data reveal homozygous regions on chromosome 9 leading to identification of novel disease-causing mutations
Dr. Alistair Pagnamenta, The Wellcome Trust Centre for Human Genetics, University of Oxford, Oxford, UK

Clinical assessment of CNVs on chromosome 16 using Nexus Copy Number and large population matched datasets
Dr. Ingrid Simonic, Genetics Laboratories, Cambridge University, Cambridge, UK

Nexus Copy Number 7 enhances genomic variation detection with integrated analysis of CNV and sequence variations
Dr. Soheil Shams, BioDiscovery Inc., Hawthorne, CA, USA

Life Technologies – Monday 10 June 2013, 15.15 – 16.45 hrs - Room 241 – Level 2 Stand # 400

Go beyond the limits of real-time PCR - Next generation digital PCR technology

Dr. Iain Russell, Life Technologies

Digital PCR is an extremely promising method for increasing accuracy, precision and sensitivity of nucleic acid quantification beyond the capabilities of traditional real-time qPCR. This is done by combining PCR assays with a system to partition a sample into a set of reactions which number is close to, or more than, the total number of target molecules in that sample. The combination of these functions makes it possible to calculate the number of target molecules present by counting the number of reactions with or without amplification. Many novel applications are enabled by this new approach, including reference free absolute quantification and high sensitivity rare-allele quantification.

We will be presenting a live demonstration of the QuantStudio™ 3D Digital PCR System, a new digital PCR platform from Life Technologies. This chip based system enables collection of up to 20,000 data points per sample run. The workflow has been optimised for simplicity, minimising the risk of sample cross contamination, and minimising sample loss. We will review the basics of digital PCR theory, look at exciting research projects from scientists across Europe, discuss the capabilities of the QuantStudio™ 3D Digital PCR System, and demonstrate the steps involved in a full digital PCR experiment.

Roche Applied Science – Monday 10 June 2013, 15.15 – 16.45 hrs - Room 251 – Level Stand # 110

Roche Sequencing Solutions Workshop - Driving Next-Generation Sequencing from Research to Routine

Clinical Research for the Genetic Testing of Familial Hypercholesterolemia Using Next-Gen Sequencing
Alain Carrie, M.D., Centre de Génétique Moléculaire et Chromosomique, Hôpital de la Pitié Salpêtrière, Paris, France

Implementation of Targeted Sequencing Technologies for Cardiological and Neurological Disorders in a Clinical Research Setting
Prof. Dr. Frank Baas, M.D, Ph.D., Academic Medical Center, University of Amsterdam, Amsterdam, the Netherlands

Latest Advancements in Roche Sequencing Solutions
Mark Driscoll, Ph.D., International Product Manager, Roche Sequencing Solutions, Branford, CT, United States

Next-generation sequencing is driving groundbreaking advancements to improve human health and disease. Roche Sequencing Solutions portfolio of target enrichment and next-generation sequencing systems are helping scientists solve complex life science challenges and laying the foundation for Personalized Healthcare by enabling better understanding of cancer, infectious diseases and other complex conditions. Join the Roche Sequencing Solutions workshop to learn how researchers are leading efforts to drive next-generation sequencing from clinical research to routine applications using NimbleGen SeqCap EZ Libraries and the 454 GS FLX+ System and GS Junior System. Get the latest information on advancements in the Roche Sequencing Solutions portfolio.

PROGRAMME POSTER TOPICS-TECHNICAL INFORMATION

Poster Topics

Technical Information

Posters will be on display from	**Sunday, June 9, 08.30 to Tuesday, June 11, 12.15 hrs**	
Poster mounting will be possible on:	**Saturday, June 8, from** **Sunday, June 9, from**	**12.00 - 14.00 hrs (strict) and** **08.30 hrs**
Removal will be possible on:	**Tuesday, June 11, from**	**12.30 hrs - 13.30 hrs (strict)**

Please note that posters not removed until then will be taken down by the staff of the conference center and will be discarded. Access to the exhibition hall will be prohibited after 13.30 hrs for security reasons.

You will find your **poster board number** in the author index at the end of the electronic abstract book, the poster manual (available at the poster desk) or ask the poster desk for assistance.

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 9** for **posters with odd numbers**
  (e.g. P04.01, P04.03 - this refers to your final poster board number - not the abstract control number!)
  or
- **10.30 and 11.30 hrs on Monday, June 10** for **posters with even numbers**
  (e.g. P07.02, P07.04 - this refers to your final poster board number not the abstract control number!)

If this is not possible, please leave a note on your poster board detailing the times when you will be present at the board.

Spoken Presentations - Projection and Technical Setting

- 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 room (**Level 1, Room 111/112/113**) preferably in the morning of the day your talk is scheduled, but not later than 2 hours in advance.
- The lecture rooms are exclusively equipped with Windows-PCs (no Macintosh machines). In case you absolutely need to use your own laptop or notebook, please contact the preview center 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)

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**

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 11, 2013 at 14.00 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.

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

PROGRAMME AWARD FINALISTS

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 2013?

Nikhita Ajit Bolar
Antwerp, Belgium

Talk: C07.4 Mutation of the iron-sulfur cluster assembly IBA57 gene causes lethal myopathy and encephalopathy.
Session: C07 Metabolic and mitochondrial disorders
Date: Sunday, June 9, 2013, 13:15 hrs

Q1: 15th June 1988, Mangalore, India
Q2: PhD student at the Centre for Medical Genetics at University of Antwerp.
Q3: I was interested, quite earlier on, in the field of genetics and this led me to pursuing both, my bachelors and masters degrees in the field of human genetics. My inquisitiveness and interest coupled with the dynamics and challenges of the constantly evolving field of genetics, along with impacting the knowledge of understanding disease mechanism and pathology, was the motivating factor to choose a career in research in this area.
Q4: This research, in particular, was extremely interesting because it's a story that starts from the clinic and goes through a circle ending back at the clinic. It allowed the integration of different fundamental and clinical research techniques, including computational tools which together, helped identify mutations in a novel gene(IBA57) and elucidate its role in severe myopathy and encephalopathy. Although challenging, it was wonderful to see how the results from each technique contributed to a little piece to the puzzle. This research impacted patient health providing them further insight into their disorder. There is much left to be understood and the results have opened new doors to more questions that need to be answered, but this is exactly what makes research interesting. It's not very often that you can see how your research impacts patient health positively and I am quite grateful for the opportunity to be a part of such exciting research.

Veronique Belzil
Jacksonville, United States

Talk: C20.1 RNA foci in C9FTD/ALS patients sequester RNA binding proteins and subsequently alter downstream splicing and expression of their RNA targets.
Session: C20 Neurodevelopmental and neuropsychiatric disorders
Date: Tuesday, June 11, 2013, 11:00 hrs

Amélie Bonnefond
Lille, France

Talk: C01.2 Type 2 Diabetes strongly increases risk for the pre-cancerous state of clonal mosaicism
Session: C01 Structural variation and de novo mutations
Date: Sunday, June 9, 2013, 13:15 hrs

Q1: 11 May 1984 (Grenoble, France)
Q2: I am currently a postdoctoral fellow in the CNRS unit #8199 (Lille, France) headed by Prof Philippe Froguel. I am head of the next-generation sequencing group. My own research is focused on the contribution of rare genetic events to the risk of type 2 diabetes and obesity.
Q3: It is so fascinating and motivating to try to identify the cause of a genetic disease! Furthermore, our discoveries can radically change life of the mutated patients with more appropriate treatment and therapies.
Q4: Large clonal mosaic events (CME) were shown to be associated with age and cancer risk. We have shown a strong effect of type 2 diabetes (T2D) and its complications on CME development. Given the increased risk of cancer in CME carriers, our results may have profound clinical implications for the management of patients with severe T2D.

Renata Bordeira-Carriço
Porto, Portugal

Talk: C04.6 Suppressor-tRNA Restores Functional E-Cadherin Expression in Cdh1 Mutant Cancer Cells: A Potential Approach to Treat Hereditary Diffuse Gastric Cancer
Session: C04 Cancer predisposition
Date: Sunday, June 9, 2013, 13:15 hrs
Q1: 29-10-1982 Lisbon, Portugal
Q2: I am currently applying for post-doctoral fellowships through CEDOC – Chronic Diseases Research Centre from FCM Nova, Lisbon.
Q3: Many diseases, including cancer, are related with genetic alterations. Genetics is the basis of biological systems. By understanding the genetic basis of each disease, we can develop tools more specific, effective and appropriate for each patient. I believe the future of medicine will be intrinsically related with the molecular profile of patients and diseases, as we are evolving towards more personalized treatments.
Q4: This work, developed during my PhD at IPATIMUP, is based on manipulation of suppressor-tRNAs to recognize premature stop codons and restore protein expression. We demonstrate, for the first time, efficient recovery of a functional full-length protein, from a nonsense-mutated allele, using a suppressor-tRNA in gastric cancer cells. Importantly, this new strategy may be applied to other genetic diseases, being especially significant for inherited cancer syndromes.

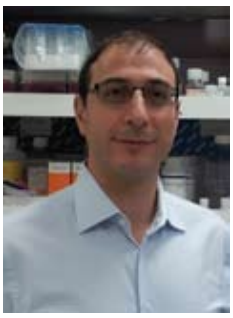
PROGRAMME AWARD FINALISTS

Diana Braunholz
Lübeck, Germany

Talk: C14.4 A founder mutation in ULFIN, a new gene on chromosome 16q22.1, in patients with spinocerebellar ataxia type 4 (SCA4)
Session: C14 Sensory disorders and Immunological disorders
Date: Monday, June 10, 2013, 13:15 hrs
Q1: 15.02.1984 Leipzig, Germany
Q2: post-Doc, Institut für HumangenetiK Lübeck (Germany)
Q3: Especially in the field of human genetics I think to have the best possibilities to combine my interests in molecular mechanisms with clinical aspects. My main interest here is to understand the phenotypical consequences of specific mutations on physiological level. The field of human genetics is strongly and fast evolving, which contributes to help people and I really like being part of this progress.
Q4: Within the last month of my PhD time, which I successfully finished very recently, it started to expand my field of interest. The genetics of spinocerebellar ataxia (SCA), in particular SCA type 4, has a longstanding history in the department of Human Genetics in Lübeck. Using genome sequencing analysis and first functional follow-up experiments we could identify a mutation in a hitherto unknown protein-coding gene. Furthermore we could detect this mutation in a couple of patients and even two large families with SCA4 while this mutation is unknown in all databases available and could be excluded in a very large control cohort. The functional characterization of this new protein as well as experimental investigations delineating the physiological effects of the identified mutation draws my special interest.

Keren Carss
Hinxton, Cambridgeshire, United Kingdom

Talk: C03.6 Exome sequencing of 27 trios to identify genetic causes of fetal abnormalities
Session: C03 Prenatal diagnosis
Date: Sunday, June 9, 2013, 13:15 hrs
Q1: 13th February 1985, Norwich, UK
Q2: PhD student (3rd year) at the Wellcome Trust Sanger Institute, Cambridge, UK.
Q3: The genetic causes of many rare, monogenic diseases remain unknown. Patients are often desperate to know the cause of these conditions, even though there is usually no treatment. I aim to identify genetic variants that cause these diseases, and therefore improve diagnostic rates for patients.
Q4: Exome sequencing is an effective tool for identifying genetic causes of abnormal fetal development. This is really exciting because it will empower affected families to make more informed reproductive decisions, and it may pave the way for exome sequencing to be introduced into prenatal diagnostic labs on a wider scale.



Sebahattin Cirak
Washington, United States

Talk: C13.3 A stop mutation in WDR81 causes microcephaly with variable penetrance.
Session: C13 Neurodegenerative disorders: From gene discovery to therapy
Date: Monday, June 10, 2013, 13:15 hrs
Q1: 14.06.1970; Bayburt/Turkey
Q2: Research Associate, MD
Q3: I am a paediatrician and during my clinical practice I was confronted with many kids who suffered of rare inherited diseases. But only a few had a molecular genetic diagnosis, leading to frustration and also inability of future family planning. Therefore I decided to investigate the genetic basis of inherited diseases with focus on neurogenetics disorders. Accurate molecular genetic diagnosis is obligatory today as targeted pesonalized molecular therapies are entering clinical practice (Exon skipping therapies for Duchenne Muscular Dystrophy).
Q4: We found a novel gene for microcephaly which seems to be located in the nucleus, which may indicate a novel mechanism of disease. We also observed remarkable clinical variability. The gene discovery made pre-natal testing possible and one family has now a healthy child.

Joep de Ligt
Nijmegen, Netherlands

Talk: C15.6 Detection of clinically relevant copy number variants with whole exome sequencing
Session: C15 NGS-based diagnostics
Date: Tuesday, June 11, 2013, 11:00 hrs
Q1: 20-Sep-1986 Boskoop, NL
Q2: PhD student
Q3: The recent advances in sequencing technologies enabled researchers to start using Genomewide techniques in patients. This was a drive for me to go into genetics as this provided me an opportunity to do research which could improve diagnostic procedures.
Q4: We show that copy number variation (CNV) detection from whole exome sequencing data identifies most rare clinically relevant CNVs. And can be an improvement over low resolution microarrays used in routine diagnostics.

Mirjam de Pagter
Utrecht, Netherlands

Talk: C11.1 The 3D topographic mapping of genetic variations in treatment naïve advanced ovarian cancer
Session: C11 Molecular mechanisms in tumorigenesis
Date: Monday, June 10, 2013, 13:15 hrs
Q1: 18-12-1982 Vlissingen, The Netherlands
Q2: PhD student
Q3: DNA has fascinated me since I had my first lesson on the subject in school.
Q4: We have performed extensive multi-level profiling of treatment-naïve material obtained from multiple primary and metastatic sites of three ovarian cancer patients. Our data highlights extreme intra-tumor heterogeneity, and demonstrate the substantial diversity of the evolutionary patterns of epithelial ovarian cancer.

PROGRAMME AWARD FINALISTS

Richarda de Voer
Nijmegen, Netherlands

Talk: C04.5 Loss of a Regulatory Element May Determine Endometrial Cancer Risk in EPCAM Deletion Carriers
Session: C04 Cancer predisposition
Date: Sunday, June 9, 2013, 13:15 hrs
Q1: 19/03/1984, Breda, The Netherlands
Q2: I am currently a postdoc in the Cancer Genomics Group at the Department of Human Genetics of the Radboud University Medical Centre, Nijmegen, The Netherlands.
Q3: I have always been intrigued by the fact that single genetic defects can predispose individuals to develop cancer. As the field of genetics is continuously evolving with innovative technologies, it is an extremely challenging work environment where I have found the perfect mix between fundamental and translational science.
Q4: Constitutional 3'end deletions of EPCAM cause epigenetic silencing of the downstream MSH2 gene in EPCAM-expressing tissues, leading to Lynch syndrome. As we learn more about these 3' end deletions, we see that the length of these deletions may have clinical implications for their carriers with respect to the type of tumors they may develop. It is an example of tissue specific cancer risk by the loss of a potential enhancer element which adds to our understanding of cancer predisposition.

Klaus Dieterich
Grenoble, France

Talk: C06.3 The neuronal endopeptidase ECEL1 is associated with autosomal recessive distal arthrogryposis
Session: C06 Neuromuscular disorders: From genes and modifiers to function and therapy
Date: Sunday, June 9, 2013, 13:15 hrs
Q1: Febrary 2 1978, Berlin, Germany
Q2: Assistant professor and clinical geneticist at the University Hospital of Grenoble/ France and doctoral fellow in cell biology at the Grenoble Institut of Neurosciences (Inserm U836).
Q3: Better understand the relationship between tiny abberations taking place at the molecular level and their sometimes dramatic macroscopic consequences during the embryonic and foetal development was certainly one of the main triggers for me to choose genetics. The challenge to recognize specific phenotypes and predict their molecular bases was hence another reason.
Q4: Distal arthrogryposis (DA) is one of the most frequent clinical presentations of Arthrogryposis multiplex congenita. Yet until now molecular analyses failed to show mutations in known genes in approximately 60-70% of all cases. Our results show that ECEL1 is a frequent cause of DA and implies a new physiopathological mecanism.

Patrick Deelen
Groningen, Netherlands

Talk: C18.4 The 'Genome of The Netherlands' outperforms '1000 genomes' as a reference set for imputing rare variants in the European population
Session: C18 Big GWAS
Date: Tuesday, June 11, 2013, 11:00 hrs
Q1: 1986-05-30, 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
Q3: The field of genetics allows me to put my computer knowledge and creativity to good use when studying interesting and challenging biological research questions. I aim to contribute to the knowledge about genetics during my PhD and hopefully many years beyond.
Q4: We show that the Genome of The Netherlands (GoNL), a population based sequencing effort of 769 individuals, can also be used to enhance non-Dutch GWAS's. The genotype imputation quality, when comparing to 1000G imputation, is not only enhanced in Dutch samples but also in British and Italian samples.

Clara Elbers
Utrecht, Netherlands

Talk: C09.3 Signatures of selection in the Genome of the Netherlands Project
Session: C09 Population-based sequencing
Date: Monday, June 10, 2013, 13:15 hrs
Q1: 27/02/1980 Utrecht, the Netherlands
Q2: Postdoc
Q3: I've always been interested in the heritability of traits.
Q4: The genome of the Dutch population reflects the history of the Netherlands at a molecular level.

PROGRAMME AWARD FINALISTS

Nizar El-Murr
Paris, France

Talk: C11.5 MicroRNAs as possible initiators and drivers for microsatellite unstable colorectal tumours
Session: C11 Molecular mechanisms in tumorigenesis
Date: Monday, June 10, 2013, 13:15 hrs
Q1: June 23, 1987 - Beirut, Lebanon
Q2: I am a PhD student at the INSERM's Saint-Antoine Research Center, affiliated to the Pierre-and-Marie-Curie University in Paris.
Q3: My admiration for genetics started very early, even before entering college. As a young boy, I was fascinated by its logic, as a PhD student I am thrilled by its complexity. The technology, the prospect of a better understanding of human disease and the ever evolving challenges we face keep me motivated.
Q4: My work focuses on the interrelationship between genetics and epigenetics in microsatellite unstable colorectal cancers. For the first time, we propose a model where microRNA deregulation in tumour microenvironments and microRNA sequence alterations in tumour cells might constitute early and late events in the development of colorectal neoplasms, respectively.

Lucas Fares Taie
Paris, France

Talk: C14.1 ALDH1A3 mutations cause recessive anophthalmia and microphthalmia
Session: C14 Sensory disorders and Immunological disorders
Date: Monday, June 10, 2013, 13:15 hrs

Laurent Francioli
Utrecht, Netherlands

Talk: C01.1 De novo mutations in the Genome of the Netherlands
Session: C01 Structural variation and de novo mutations
Date: Sunday, June 9, 2013, 13:15 hrs
Q1: 23.03.1983, Lausanne, Switzerland
Q2: I am a 3rd year PhD student in Paul de Bakker's lab at the UMC Utrecht in the Netherlands.
Q3: I find it fascinating that all life's information is coded in DNA and am thrilled to work on understanding it.
Q4: We present here the largest set of directly observed de novo mutations to date and unveil some of their key properties contributing to genomic evolution. Moreover, computing mutation rates along the genome help establish a baseline for de novo mutation burden in healthy individuals.

Elisa Giorgio
Torino, Italy

Talk: C13.5 A large genomic deletion upstream of the lamin B1 gene (LMNB1) likely causes adult-onset autosomal dominant leukodystrophy due to alteration of the regulatory landscape of LMNB1.
Session: C13 Neurodegenerative disorders: From gene discovery to therapy
Date: Monday, June 10, 2013, 13:15 hrs
Q1: 29/05/1984, Savona, Italy
Q2: PhD Student in Human Genetics
Q3: I love solving puzzles, and human genetics provides the most interesting and charming.
Q4: I will describe a position effect mutation causing overexpression of LMNB1 gene and mimicking LMNB1 duplication as cause of Autosomal Dominant Leukodystrophy. This work further emphasize that genomic alterations that change the regulatory landscapes are important determinants of Mendelian phenotypes and are probably an underestimated entity.

Brittany Gregory
Philadelphia, United States

Talk: C17.1 Natural variation in the histone demethylase, KDM4C, influences transcriptional regulation and cell growth
Session: C17 Basic mechanisms in cytogenetics and molecular genetics
Date: Tuesday, June 11, 2013, 11:00 hrs
Q1: 02/24/1984, Traverse City, Michigan, USA
Q2: I am pursuing a combined doctorate in veterinary medicine and genetics at the University of Pennsylvania, USA.
Q3: I became interested in genetics while working with animal models of human disease. I learned that while there are many biological similarities across species, it is equally important to understand the biological differences. This can also be said for differences among individuals within a species. Genetics interests me because it asks how DNA sequences influence biological differences among individuals, such as differences in disease susceptibility.
Q4: The research that I am presenting highlights how individual variation can be used to learn about biological function. We used natural variation in a histone demethylase gene, KDM4C, in order to learn how KDM4C is regulated, how KDM4C regulates the expression of other genes, and how KDM4C has a role in cell proliferation. This work also shows how targeting KDM4C could benefit cancer treatment.

PROGRAMME AWARD FINALISTS

Sebastian Grömminger
Konstanz, Germany

Talk: C03.2 Diagnostic accuracy for the non-invasive prenatal detection of common autosomal aneuploidies
Session: C03 Prenatal diagnosis
Date: Sunday, June 9, 2013, 13:15 hrs
Q1: 18.03.1983 Radolfzell am Bodensee, Germany
Q2: Scientist Diagnostics & Research at LifeCodexx AG, Konstanz, Germany.
Q3: From my point of view genetics has the greatest potential to uncover the biology of human beeings
Q4: The use of next generation sequencing technology to provide non-invasive genetic testing provides early access to a broad spectrum of genetic information. Using this techniqe for prenatal testing is just the starting point of an evolving field of genetic testing that may cover a variety of genetically based clinical pictures

Andreas Gschwind
Lausanne, Switzerland

Talk: C05.4 Coordinated effects of sequence variation on DNA binding, chromatin structure, and transcription
Session: C05 Functional Genomics
Date: Sunday, June 9, 2013, 13:15 hrs

Hongsheng Gui
Hong Kong, Hong Kong

Talk: C16.1 Patterns and Rates of Exonic de novo Mutations in Sporadic Hirschsprung Disease Patients
Session: C16 Developmental syndromes
Date: Tuesday, June 11, 2013, 11:00 hrs
Q1: March 09, Jiujiang
Q2: PhD student
Q3: I found genetics the most interesting to me when studied biology in undergraduate time. I enjoy the later research life for forensic genetics and statistical genetics.
Q4: The disease I am working on can be serve as a model for genetic study on complex diseases.

Tobias Haack
Munich, Germany

Talk: C13.2 WDR45 de novo mutations cause of a clinically distinct, X-linked subtype of NBIA
Session: C13 Neurodegenerative disorders: From gene discovery to therapy
Date: Monday, June 10, 2013, 13:15 hrs
Q1: 05.03.1982 Überlingen
Q2: PostDoc / MD
Q3: A career in genetics allows me to think about questions I am interested in a stimulating setting = fun.
Q4: The major forms of NBIA (neurodegeneration with brain iron accumulation)are autosomal recessive inherited. X-chromosomal WDR45 mutations occur de novo and together with a role of the beta-propeller family protein in autophagy it suggests both, a new pathomechanism and a new pathway in the pathogenesis of disorders characterized by extensive iron accumulation in the basal ganglia.

Magdalena Harakalova
Utrecht, Netherlands

Talk: C16.2 A novel chromosomal breakage syndrome caused by a missense mutation in a gene from the SMC5/6 complex
Session: C16 Developmental syndromes
Date: Tuesday, June 11, 2013, 11:00 hrs
Q1: 24-01-1984, Humenne, Slovakia
Q2: I am currently finishing my PhD training at the Department of Medical Genetics, University Medical Center Utrecht, The Netherlands. The titel of my thesis is: 'The next-generation of disease gene discovery'.
Q3: During my medical studies I realized that many diseases just happen without our contribution - we have it in our genes. I find it extremely fascinating to find out where exactly, what is the functional consequence and the continuously increasing impact on patient's care and treatment.
Q4: We characterized a novel chromosomal breakage syndrome with fatal pulmonary failure in a Dutch family that was negative for all known genes. Thanks to next-generation sequencing we were able detect the responsible gene. This gene is for the first time linked to a human disease and we are currently working on further functional characterization.

Kristien Hens
Maastricht, Netherlands

Talk: C03.4 Comprehensive Chromosome Screening in PGD and PGS - Ethical Challenges
Session: C03 Prenatal diagnosis
Date: Sunday, June 9, 2013, 13:15 hrs
Q1: 11/09/1975, Gent (Belgium)
Q2: I am currently working as a postdoctoral researcher at the Maastricht University.
Q3: My field of research is bioethics, especially ethical questions surrounding genetics and reproductive technologies. I chose these specific fields because they allow me to both study exciting new technologies and reflect on their societal and ethical implications.
Q4: Discussions about genetic screening of embryos have often focussed on designer babies and selecting smart children. I have investigated how technologies that allow comprehensive screening of an embryo's chromosomes already raise difficult dilemmas and questions regarding the role of the professional in assisted reproduction and the extent of parental autonomy. These issues need further reflection as these technologies are being implemented today.

PROGRAMME AWARD FINALISTS

Youssef Hibaoui
Geneva, Switzerland

Talk: C17.6 Modelling and rescuing neurodevelopmental defect of Down syndrome using induced pluripotent stem cells from monozygotic twins discordant for trisomy 21
Session: C17 Basic mechanisms in cytogenetics and molecular genetics
Date: Tuesday, June 11, 2013, 11:00 hrs
Q1: Dijon, France
Q2: Postdoctoral researcher at the Stem Cell laboratory and the Department of Genetic Medicine and Development, Geneva University
Q3: Genetics have proven extremely informative for the study of many diseases investigated using induced pluripotent stem cells (iPSCs). Indeed, the combination of genetics and iPSCs is an exciting and fast moving area of current studies. I believe that these approaches will advance our understanding of human diseases by deciphering the mechanisms involved in the pathogenesis.
Q4: We report the generation and characterization of induced pluripotent stem cells (iPSCs) derived from monozygotic twins discordant for trisomy 21. Since the rest of the genome is identical between the two twins, this provides a unique model to study the effect of the supernumerary chromosome 21. This iPSC-based model is an innovative way to study Down syndrome (DS) neurodevelopment as it offers an unprecedented opportunity to study DS early embryonic development and enable the study of the detailed pathogenetic mechanisms by which the extra copy of chromosome 21 leads to DS phenotypes. We found in particular that neural cells derived from DS iPSCs recapitulate the neurodevelopmental features of DS as revealed through genetic and functional studies. Moreover, the finding that we can target pathogenetic pathways by both pharmacological and hairpin RNA silencing approaches allows a proof-of-principle for potential screening tests using iPSC technology. This should provide the basis for designing new therapeutic approaches for DS patients.

Elisa Houwink
Amsterdam, Netherlands

Talk: C10.5 Effects of multifaceted oncogenetics training for general practitioners
Session: C10 Genetic Counselling, Education and Public Services
Date: Monday, June 10, 2013, 13:15 hrs
Q1: 13-12-1973, Haarlem, The Netherlands
Q2: General practitioner and PhD Candidate
Q3: I started my science work in genetics during my medical study rotation in Boston, USA with Prof Dr Carol Warner in 1998 where I worked on preimplantation embryo development and genetic influences. This is really where I started to find my passion in genetics and see how genotype malfunction can influence phenotype. Further on I worked on Rett syndrome with Dr Carolyn Schanen at UCLA, where we found out about DNAtranscription chaos through mutations in the methyl CpG binding 2 gene and helped parents when I was able to diagnose the genetic mutation responsible for their child's impairments.
Q4: Being able to translate genetics innovations and finally bring them to the clinic by educating physicians on genetics is very fulfilling. Finally it is possible to bring everything learnt in the laboratory effectively to the physician, patient and their family where it belongs.

Hui-Han HU
Paris, France

Talk: C04.4 Parkinson's disease and melanoma: a common genetic pathway linked to PARKIN inactivation
Session: C04 Cancer predisposition
Date: Sunday, June 9, 2013, 13:15 hrs
Q1: 17/12/1983 Taipei Taiwan
Q2: I'm a Taiwanese student in France. This is the last year of my PhD program.
Q3: For me genes are like ordering forms which save and transfer customers'(body's) needs to a factory for producing and assembling components (amino acids) in a functional furniture (proteins). They are the source of mistakes that cause a disease. Fixing the mistakes from the biginning may prevent the occurrence of the disease.
Q4: Melanoma and Parkinson's disease are both considered as multifactorial diseases. Identification of genetic risk factors for both diseases and high incidence of co-occurrence of both diseases in an individual have been carried out and mentioned for a long time. This noteworthy work identifies a common genetic pathway that may explain the epidemiological association between these two diseases.

Marjorie Hubeau
Paris, France

Talk: C14.6 Ion transporter deficiency predisposes to pyogenic bacterial infection by partial oxidative burst defect in granulocytes
Session: C14 Sensory disorders and Immunological disorders
Date: Monday, June 10, 2013, 13:15 hrs

Evelyne Huynh Cong
Paris, France

Talk: C19.5 A recurrent homozygous missense mutation in TTC21B encoding the ciliary protein IFT139 unexpectedly causes steroid-resistant nephrotic syndrome
Session: C19 Internal organs and endocrinology: Gene identification and function
Date: Tuesday, June 11, 2013, 11:00 hrs
Q1: 04/16/1987, Sarcelles (France)
Q2: PhD student
Q3: Genetic diseases affect a large number of patients but little is known about the physiopathology of many of them. Until today, few therapies are available to treat them. Research in this field gives me the opportunity to find novel genes and to improve the understanding of the mechanisms involved in Nephrotic syndrome. My results will hopefully contribute to find new therapeutic targets in order to elaborate treatments.
Q4: My work underlines the critical function of IFT139 in both podocytes and tubular epithelial cells, via ciliary transport but also microtubule network necessary for cell differenciation, migration and proper organization/maintenance of intercellular junction. Indeed, it shows for the first time that a ciliary protein, previously found in isolated Nephronophtisis, could also be implicated in Nephrotic syndrome.

PROGRAMME AWARD FINALISTS

Alessia Indrieri
Naples, Italy

Talk: C07.3 Mutations in nuclear-encoded components of mitochondrial respiratory chain complex III and IV cause apoptosis-driven developmental defects, a new mitochondrial phenotype in vertebrates
Session: C07 Metabolic and mitochondrial disorders
Date: Sunday, June 9, 2013, 13:15 hrs
Q1: 23/12/1980 Cosenza, Italy
Q2: PostDoc
Q3: Since I was a child, my dream was to become a good scientist. I enthusiastically witnessed the possibility to investigate our genetic determinant to better understand how our body works.
Q4: Studing a rare genetic disease we uncovered the existence of a new group of mitochondrial diseases in which impairment of the last segment of the mitochondrial respiratory chain results in developmental tissue-specific defects. Our results indicate an essential role of the mitochondrial respiratory chain in eye and brain development.

Leigh Jackson
Plymouth, United Kingdom

Talk: C10.3 Incidental findings in research: National Health Service Research Ethics Committee member perspectives.
Session: C10 Genetic Counselling, Education and Public Services
Date: Monday, June 10, 2013, 13:15 hrs

Sietske Kevelam
Amsterdam, Netherlands

Talk: C07.5 Exome sequencing Reveals Mutated NUBPL in Patients with Complex I Deficiency and a Distinct MRI Pattern
Session: C07 Metabolic and mitochondrial disorders
Date: Sunday, June 9, 2013, 13:15 hrs
Q1: 30-07-1985Nijmegen
Q2: I am currently a PhD student working at the departments of Child Neurology and Medical Genome Analysis.
Q3: With the new genetic techniques we can solve the genetics of rare diseases. It is fantastic to be able to give patients and families answers and help them cope with their diseases. It is also fantastic to achieve better understanding of human physiology through determining the cause of diseases.
Q4: My focus is on exceedingly rare inherited encephalopathies. Finding the causal gene with exome sequencing for one patient remains challenging. We show that specific abnormalities on brain MRI can help tremendously in defining homogenous patient groups, for which finding the common mutated gene by exome sequencing has a better chance.

Daniel King
Hinxton, United Kingdom

Talk: C17.2 Enrichment of uniparental disomy events detected in the Deciphering Developmental Disorders rare disease study
Session: C17 Basic mechanisms in cytogenetics and molecular genetics
Date: Tuesday, June 11, 2013, 11:00 hrs
Q1: March 24th, 1983. New York, USA
Q2: PhD Student
Q3: In my early clinical training, I developed an enthusiastic hope that the availability and interpretation of DNA sequencing data would improve diagnostic accuracy in our patients with rare, often cryptic diseases. I decided I needed to improve my computational skills to take a leadership role in bioinformatics software development. This PhD studentship has been a fantastic opportunity to work with scientists and physician scientists in developing new software tools that assist the diagnostic process in our rare disease study.
Q4: Our novel computational method detects an important inheritance aberration called Uniparental Disomy in family trios with high accuracy, and the first method capable of exome-based detection. We describe some of the UPD events we found in our patients, and show how UPD detection has helped bring diagnoses to these children.

Peter Krawitz
Berlin, Germany

Talk: C15.3 Targeted sequencing of GPI anchor synthesis pathway genes identifies mutations in PGAP2 as a new cause of hyperphosphatasia with mental retardation
Session: C15 NGS-based diagnostics
Date: Tuesday, June 11, 2013, 11:00 hrs

Laura Kremer
Neuherberg, Germany

Talk: C07.1 New diagnostic paradigms for mitochondriopathies
Session: C07 Metabolic and mitochondrial disorders
Date: Sunday, June 9, 2013, 13:15 hrs
Q1: 05.09.1986, Rodalben, Germany
Q2: PhD student
Q3: Identifying the mutation underlying a particular disease is essential for the understanding of the corresponding mechanistic pathway as well as the development of therapeutic 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. This hopefully paves the way for new treatement options

PROGRAMME AWARD FINALISTS

Maéva Langouët
Paris, France

Talk: C08.3 Mutations in TTI2 reveal a role for Triple T complex in human brain development
Session: C08 Intellectual disability: Gene discovery and dysfunction
Date: Monday, June 10, 2013, 13:15 hrs
Q1: 4 June 1988
Q2: I am a second year PhD student.
Q3: When I was in first year of university I chose to continue in the genetic field because it was the one I was the most interested in. Then I developed a high interest in hospital-related work, in collaboration with clinicians, investigating genetic causes of human conditions.
Q4: In addition to explaining the affected patient's phenotype, the mutations identified in the TTI2 gene offer new clues about the molecular networks that may be disrupted in ID patients. These mutations also provide new insights on those networks that are involved in brain development and the establishment of cognitive functions.

Davor Lessel
Ulm, Germany

Talk: C16.3 SPRTN deficiency causes a novel segmental progeroid syndrome with chromosomal instability
Session: C16 Developmental syndromes
Date: Tuesday, June 11, 2013, 11:00 hrs
Q1: 05.06.1981, Zagreb, Croatia
Q2: Resident, training in medical genetics
Q3: As a medical doctor I have chosen to pursue a career in medical genetics as I found it to be the most exciting and emerging area in medicine, which additionally offers me a unique opportunity to combine scientific research of studying the human nature with clinical practice in various fields of medicine.
Q4: Driven by sustained increase in lifespan, age-related diseases represent a major challenge for health care systems. Studies of rare, monogenic syndromes with signs of premature aging offer a unique opportunity to better understand the "physiological" aging.I will be presenting identification of the genetic cause of a novel progeroid syndrome in just a single patient, supported by extensive in-vitro and in-vivo analyses and providing functional links to known progeroid syndromes.

Audrey Letourneau
Geneva, Switzerland

Talk: C08.6 Identification of Single-minded 2 (Sim2) binding sites by ChIP-Seq; understanding of the regulatory network of chromosome 21 transcription factors
Session: C08 Intellectual disability: Gene discovery and dysfunction
Date: Monday, June 10, 2013, 13:15 hrs
Q1: 06.04.1985, Le Mans, France
Q2: PhD student at the University of Geneva Medical School, Switzerland
Q3: I've always been intrigued by the complexity of our genome and I think it is fascinating to explore the mechanisms involved in the genotype/phenotype relationships. I believe that, in the near future, genetics will play a key role in medicine.
Q4: In this study, we show that the functional characterization of human chromosome 21 genes is essential to improve the understanding of the molecular bases of Down syndrome.

Sari Lieberman
Jerusalem, Israel

Talk: C10.1 From personal genetic counseling to public health screening: The BRCA Opportunity
Session: C10 Genetic Counselling, Education and Public Services
Date: Monday, June 10, 2013, 13:15 hrs
Q1: 24/10/1974Haifa, Israel
Q2: Genetic counselor, PhD student
Q3: I chose to pursue a career in genetics since I believe that today genetics is impacting all fields of health and science. And has enormous influence on diseases and variance. As a genetic counselor I have the opportunity create the interface between science and people, to help improve their knowledge and decision making.
Q4: Our research is intended to examine the ability to switch the use of BRCA testing as a tool for individuals at risk to the population level. The study aims to increase prevention, therefor reducing morbidity of breast and ovarian cancer. We study the feasibility and costs of this intervention.

Céline Loriot
Paris, France

Talk: C11.4 SDHB Mutations link pheochromocytoma/ paraganglioma malignancy to epithelial to mesenchymal transition, both in human tumors and in SDHB-/- chromaffin cells
Session: C11 Molecular mechanisms in tumorigenesis
Date: Monday, June 10, 2013, 13:15 hrs
Q1: 30/06/1987, Longjumeau (France)
Q2: PhD student, 3rd year
Q3: I choose a career in both genetics and cancer, and I think they are closely related. I'm convicted that genetics are roots of everything and especially in genetically determined cancers. I believe that personalized medicine is future and to do that we absolutely need genetics.
Q4: The most interesting point of my research is that in the first developed cellular model of Sdhb deficient cells, we found the same activated pathway that in human tumors. It is a crucial point because this molecular pathway allows to explain invasive capacities of these cells, and thus the aggressiveness of tumors, which at the moment remains misunderstood

PROGRAMME AWARD FINALISTS

Periklis Makrythanasis
Geneva, Switzerland

Talk: C15.4 Diagnostic exome sequencing to elucidate the genetic basis of likely recessive disorders in consanguineous families
Session: C15 NGS-based diagnostics
Date: Tuesday, June 11, 2013, 11:00 hrs
Q1: 22-03-1978, Athens
Q2: Post-doc at Stylianos Antonarakis' laboratory in the Department of Genetic Medicine and Development, University of Geneva, Switzerland
Q3: Because genetics is life's cornerstone
Q4: The power of modern genetics to diagnose previously undiagnosed patients and the inversion of our way to approach and learn the genes' function, starting from the gene and ending up to the phenotype.

Sandra Mercier
Nantes, France

Talk: C12.1 Congenital poikiloderma, fatty infiltration of muscles and pulmonary fibrosis: a new syndrome caused by a new gene
Session: C12 Connective tissue disorders: Gene identification
Date: Monday, June 10, 2013, 13:15 hrs
Q1: 05/10/1978, Chambray-lès-Tours (France)
Q2: MD-PhD in clinical genetics in the Medical Genetics Department, University Hospital, Nantes (France) . I have also joined the UMR1089 research team, INSERM, Atlantic Gene Therapy Institute (AGTI), that works on gene and cell therapies in neuromuscular disorders.
Q3: Genetics provides great opportunities for gene identification as we can see with the NGS revolution and for the understanding of pathological mechanisms. By a better knowledge of the pathology underlying disorders, I hope we will be able to cure more and more genetic diseases in the years to come.
Q4: It all began from a sporadic case of a boy with congenital poikiloderma and muscle weakness. We identified a new gene in this new syndrome that was a major step for the patients. It is my hope that all our efforts to understand the disease will lead to a treatment as soon as possible.

Lusine Nazaryan
Copenhagen, Denmark

Talk: C17.4 New insights into human germline chromothripsis: underlying mechanisms and definition
Session: C17 Basic mechanisms in cytogenetics and molecular genetics
Date: Tuesday, June 11, 2013, 11:00 hrs
Q1: 23.12.1980
Q2: PhD-student
Q3: The interest towards genetics has began since high school, from Mendelian inheritance and more complex gene interactions. Subsequently, the interest had been growing along with my deeper studies at the University (2000-2003), when Human Genome project has been just finished. With great amazement I discovered a new huge "universe" full of mysteries, which inspired me a lot to be involved in the studies of Human Genetics.
Q4: Chromothripsis is a very interesting phenomenon discovered quit recently (2011)and there are many open questions regarding the possible mechanisms leading to these catastrophic localized rearrangements in cancer and germline. By reviewing all reported germline cases we define and classify chromothripsis, as well as propose a possible mechanism for the initiation of chromosome shattering.

Renske Oegema
Rotterdam, Netherlands

Talk: C16.6 Integrator Complex Subunit 8 mutations associated with abnormal brain development and spliceosomal defect
Session: C16 Developmental syndromes
Date: Tuesday, June 11, 2013, 11:00 hrs
Q1: 26-04-1981, Zaanstad
Q2: CLinical geneticist in training at the dept. of Clinical Genetics, Erasmus MC, Rotterdam, the Netherlands.
Q3: As a clincian, I want to understand the disease mechanism in my patients, from genotype to phenotype. Rapidly evolving new technologies enable us to make new discoveries and change the field of medicine.
Q4: For the fist time we show that defects of the Integrator Complex, involved in processing of the major spliceosome snRNAs, can lead to human disease.

Pier Francesco Palamara
New York City, United States

Talk: C09.2 Haplotype sharing reveals fine-scale demographic history
Session: C09 Population-based sequencing
Date: Monday, June 10, 2013, 13:15 hrs
Q1: June 1982 in Rome, Italy
Q2: PhD candidate at Columbia University, USA.
Q3: During my studies of artificial intelligence and cognitive robotics I became fascinated by the process of automatic learning from data, and focused my studies on machine learning. I then had a chance to work on large genetic datasets, and decided to continue my research in the field of statistical genetics.
Q4: We show that the length and frequency of identical-by-descent haplotypes in large datasets of purportedly unrelated individuals can be used to reconstruct demographic events that occurred in the past tens to few hundreds of generations, where other methods are underpowered. Our research provides a new tool for DNA-based investigation of recent historical events and can be used to support studies of evolutionary and medical genetics.

PROGRAMME AWARD FINALISTS

Marjaana Pussila
Helsinki, Finland

Talk: C11.6 Western diet and Mlh1 mutation predispose colonic mucosa to early inactivation of the Wnt signaling antagonist Dickkopf-1
Session: C11 Molecular mechanisms in tumorigenesis
Date: Monday, June 10, 2013, 13:15 hrs
Q1: 30.08.1980 Oulu
Q2: PhD student
Q3: I am fascinated by the tremendous wisdom and diversity hidden in the simple chemical structure of DNA
Q4: Our research highlights the interplay between genome, epigenome, and environment in colon tumorigenesis and provides new insight of respective genes and alterations as biomarkers for diagnostic, prognostic and therapeutic applications in humans.

Elliott Rees
Cardiff, United Kingdom

Talk: C20.6 Analysis of copy number variations at 15 schizophrenia-associated loci in a large, independent cohort
Session: C20 Neurodevelopmental and neuropsychiatric disorders
Date: Tuesday, June 11, 2013, 11:00 hrs
Q1: 23/11/85 Swansea
Q2: PhD student at the MRC Centre for Neuropsychiatric Genetics and Genomics, Institute of Psychological Medicine and Clinical Neurosciences, Cardiff University
Q3: With the recent technological advances in genetics, I was excited by the opportunity of applying my training in bioinformatics to real data in order to gain a greater understanding of the biology underlying human disease.
Q4: Several copy number variants (CNVs) have been implicated in the aetiology of schizophrenia. However, due to their rarity, it is not yet clear whether they are all true risk factors for the disorder. In the largest independent schizophrenia sample to date, we provide the most accurate estimates of risk from individual CNVs and also estimate the total burden of susceptibility conferred by all CNVs.

Manuel Rivas
Oxford, United Kingdom

Talk: C09.5 Exome sequence analysis of type 2 diabetes in over 10,000 samples from five ancestry groups: the T2D-GENES Consortium.
Session: C09 Population-based sequencing
Date: Monday, June 10, 2013, 13:15 hrs
Q1: October 6, 1985 in Managua, Nicaragua.
Q2: PhD Student.
Q3: I was always interested in mathematics and found that genetics was a field where mathematics can have a very strong impact on how we think about health, medicine, and disease. I am intrigued by the possibilities of what we can learn from genetic discoveries related to disease: 1) understanding the genetic architecture of disease, 2) pinpointing genes and functional pathways that play a role in disease risk, and 3) defining novel therapeutic hypotheses with the goal of translating genetics to therapies.
Q4: The research I am presenting at ESHG 2013 is one of the first studies to zoom in on rare variants and the role they play in common diseases. It is a major undertaking where we have made progress in our ability to: 1) process and analyze raw sequencing data, 2) characterize the landscape of genetic variation in multiple ethnicities, 3) predict the impact of genetic variants on gene function, and 4) assess association of rare variants to phenotype.

Jean-Baptiste Rivière
Dijon, France

Talk: C19.1 Mutations in PIK3R1 cause syndromic insulin resistance and lipodystrophy
Session: C19 Internal organs and endocrinology: Gene identification and function
Date: Tuesday, June 11, 2013, 11:00 hrs
Q1: Oct 22, 1983, Bordeaux, France
Q2: I am a molecular geneticist at Dijon University Hospital
Q3: I started working in human genetics at 17 years old as a summer student in Guy Rouleau's lab. Since then, I could not imagine another career choice. Genetics offers the opportunity to advance our understanding of the human genome and to make a difference in the life of people.
Q4: We identified the genetic cause of a rare syndromic form of insulin resistance and lipoatrophy. These findings may help deciphering the molecular mechanisms underlying more prevalent forms of insulin resistance and may have direct implications for the clinical management of affected individuals.

PROGRAMME AWARD FINALISTS

Miriam Schmidts
London, United Kingdom

Talk: C15.5 Exome Sequencing in the Diagnostics of non-motile Ciliopathies (113 cases)
Session: C15 NGS-based diagnostics
Date: Tuesday, June 11, 2013, 11:00 hrs
Q1: 21/07/1978 in Loerrach, Germany
Q2: Clinical Research Fellow and PhD student
Q3: I am fascinated by the way genetic information determines such complex processes as building entire organisms. From a medical point of view, I think we can learn a lot about disease pathomechanisms from genetics (not only regarding genetic diseases). And I very much enjoy the way research and clinical care is linked in genetics!
Q4: I work on the molecular basis of ciliary diseases, mainly a rare skeletal disorder called Jeune Syndrome which often leads to death. Although cilia, representing hair like cellular extensions, have been known to exist on nearly every cell type for decades, they have long been neglected and have only been recently acknowledged to play crucial roles in development and health maintenance. Proteins encoded for by ciliary genes are quite abundantly expressed throughout the human organism, but mutations in those genes can cause quite different phenotypes from obesity, cystic kidneys, brain malformations, retinal degeneration to chondrodysplasias. Vice versa, the same disease can be caused by mutations in different genes implying more complex regulatory mechanisms. With the new sequencing techniques combined with functional follow-up, the ciliary field has made huge advances which have also massively increased the knowledge about ciliary diseases. So for me these are very exciting times!

Janneke H. Schuurs-Hoeijmakers
Nijmegen, The Netherlands

Talk: C20.2 Rapid identification of autosomal recessive and X-chromosomal mutations in small sibling families
Session: C20 Neurodevelopmental and neuropsychiatric disorders
Date: Tuesday, June 11, 2013, 11:00 hrs

Vikram Sharma
Oxford, United Kingdom

Talk: P2.1 Mutations of TCF12, encoding a basic-helix-loop-helix partner of TWIST1, are a frequent cause of coronal craniosynostosis
Session: PL2 What's New?
Date: Saturday, June 8, 2013, 18:00 hrs
Q1: 03/08/1981, Nagpur, India
Q2: Clinical Research Fellow in Clinical Genetics/Craniofacial Surgery and 2nd year DPhil student, University of Oxford
Q3: Craniofacial malformation and its genetic basis has always fascinated me. As a surgeon working closely with geneticists, I have seen the positive impact a formal molecular genetic diagnosis can have on patient care. It improves the accuracy of genetic counselling and recurrence risk, guides post-operative management and provides a tangible 'label' for patients.
Q4: My research uniquely marries molecular genetics with surgery. It describes a fascinating story encompassing novel disease gene discovery by next-generation sequencing, identification of numerous further mutations and strong corroborating functional evidence. It has been translated into national testing strategies in the UK, thereby improving diagnostic outcomes for craniofacial patients.

Katie Snape
Sutton, United Kingdom

Talk: C04.2 Mosaic PPM1D mutations are associated with predisposition to breast and ovarian cancer
Session: C04 Cancer predisposition
Date: Sunday, June 9, 2013, 13:15 hrs
Q1: 16/02/1979, Cambridge, England
Q2: Clinical Research Fellow, Department of Genetics and Epidemiology,Institute of Cancer Research, London, UK
Q3: I have always been fascinated by the molecular aetiology of disease which gives insight into the biological pathways underlying pathological processes, and identifies new therapeutic approaches. I was particularly drawn to cancer genetics, since identifying individuals who are genetically predisposed to cancer development allows the implementation of personalised screening and management strategies to reduce mortality.
Q4: This research has identified a new class of genetic defect associated with cancer predisposition; a mosaic cancer predisposition marker that is genetic, but not hereditary, and is detectable in blood but not the tumours it is associated with. This opens the possibility of exciting new mechanisms of cancer development and the potential for novel biomarkers for disease.

Thomas Sparso
Copenhagen, Denmark

Talk: C09.1 Exome sequencing of 2,000 Danish individuals and the role of rare coding variants in type 2 diabetes
Session: C09 Population-based sequencing
Date: Monday, June 10, 2013, 13:15 hrs

Karani Vimaleswaran
London, United Kingdom

Talk: C18.2 A causal association between vitamin D status and blood pressure: a Mendelian Randomization study in up to 150,846 individuals
Session: C18 Big GWAS
Date: Tuesday, June 11, 2013, 11:00 hrs
Q1: 06/05/1977, Chennai, India
Q2: Research Associate, Institute of Child Health, University College London, London, United Kingdom &
Study coordinator of D-CarDia Collaboration
http://www.ucl.ac.uk/ich/research-ich/mrc-cech/research/studies/D-CarDia
Q3: Since I was in high school, I loved genetics. Of all the biological and medical fields, Genetics is the one that attracted me the most because for me understanding the way in which genetic mutations/polymorphisms contribute to complex traits is really fascinating. I anticipated back then that Genetics was supposed to be the potential science of the future. So I chose this field and I contributed in projects which related to Genetic Epidemiology and Molecular Genetics. However, I found

Genetic Epidemiology to be more interesting and hence, I will continue in this field with the aim of identifying novel genetic markers and pathways relating to complex traits using genetic epidemiological strategies.
Q4: Observational studies have shown inconsistent associations between low vitamin D status and hypertension. Also, randomised controlled trials of vitamin D supplementation in humans have produced inconsistent effects on cardiovascular outcomes. So, we have used genetic variants as proxy markers for life-long differences in vitamin D status to test for the causal association with hypertension. This approach, commonly entitled Mendelian Randomisation, exploits the concept that the individual genotypes are assigned randomly at meiosis, thereby remaining unaffected by underlying disease processes and meaning that genetic variants are largely independent of non-genetic confounding factors.
Our study is interesting as it provides further support for important non-skeletal effects of vitamin D and also promoting vitamin D as a potentially interesting target for preventive interventions.

Anneke Vulto-van Silfhout
Nijmegen, Netherlands

Talk: C08.2 Mutations in DEAF1 cause intellectual disability with severe speech impairment
Session: C08 Intellectual disability: Gene discovery and dysfunction
Date: Monday, June 10, 2013, 13:15 hrs
Q1: July 23rd 1985 in Eindhoven, the Netherlands
Q2: I am a PhD student in clinical genetics at the department of Human Genetics of the Radboud University Medical Centre in Nijmegen, the Netherlands. The topic of my PhD is the identification of novel causes for intellectual disability.
Q3: During my medical studies I became interested in genetics, because I was intrigued by the diverse and complex problems of the patients and the possibilities genetics could offer to these patients and their families. I think it is very exciting to work in a field where clinic and research are working so closely together and that is so rapidly evolving.
Q4: We identified a novel gene in three patients with intellectual disability using next generation sequencing and investigated with different functional tests the role of this gene and the mutations in the intellectual disability of the patients. In this way we show that mutations in DEAF1 are a new cause for intellectual disability.

Thomas Wieland
München, Germany

Talk: C19.3 Somatic mutations in ATP1A1 and ATP2B3 lead to aldosterone-producing adenomas and secondary hypertension
Session: C19 Internal organs and endocrinology: Gene identification and function
Date: Tuesday, June 11, 11:00 hrs
Q1: June 12th 1986 in Scheibbs/Austria
Q2: I am a third year PhD student in bionformatics at the Institute of Human Genetics at the Helmholtz Center Munich. Currently, I'm working mostly on the analysis of Next-Generation Sequencing experiments.
Q3: In my opinion the field of genetics and especially of Next-Generation Sequencing presented the most interesting and challenging opportunities for me as a bioinformatician.
Q4: Using exome sequencing, we identified dominant somatic alterations in two members of the ATPase gene family which are responsible for autonomous aldosterone secretion in roughly 7% of aldosterone-producing adenomas. This shows that exome sequencing is a promising tool to identify causal mutations in adenomas

Marjolein Willemsen
Nijmegen, Netherlands

Talk: C20.4 GATAD2B loss-of-function mutations cause a recognizable syndrome with intellectual disability and are associated with learning deficits and synaptic undergrowth in Drosophila
Session: C20 Neurodevelopmental and neuropsychiatric disorders
Date: Tuesday, June 11, 11:00 hrs
Q1: February 28, 1981
Q2: I'm a clinical geneticist in training at the department of Human Genetics in Nijmegen, The Netherlands. Last year I defended my thesis entitled ''Making Headway with the Molecular and Clinical Definition of Rare Genetic Disorders with Intellectual Disability''
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 four individuals representing a novel clinically recognizable intellectual disability syndrome. The first two individuals were identified by whole exome sequencing. Identification of two additional individuals with highly similar phenotypes and complementary functional studies in Drosophila ultimately further established this novel syndrome. This illustrates the importance of close collaboration between clinical and lab specialties, and the need for careful collection, storage and sharing of phenotype and genotype data.

Daria Zhernakova
Groningen, Netherlands

Talk: C05.6 Novel genetic variants associated with alternative polyadenylation and expression of noncoding transcripts
Session: C05 Functional Genomics
Date: Sunday, June 9, 13:15 hrs

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

P01.017 The genetic study of intracranial aneurysm in the Japanese population
S. Low[1], *A. Takahashi*[1], *M. Kubo*[2], *Y. Nakamura*[3];
[1]*Laboratory for Statistical Analysis, Center for Genomic Medicine, RIKEN, Tokyo, Japan,* [2]*Laboratory for Genotyping Development, Center for Genomic Medicine, RIKEN, Yokohama, Japan,* [3]*Laboratory of Molecular Medicine, Human Genome Center, Institute of Medical Science, The University of Tokyo, Tokyo, Japan.*

P01.031 TAPT1 and CREB3L1 interfere with normal type I collagen secretion and further expand the genetic spectrum of recessive Osteogenesis Imperfecta
S. Symoens[1], *F. Malfait*[1], *S. D'hondt*[1], *B. Callewaert*[1], *A. Dheedene*[1], *W. Steyaert*[1], *D. Syx*[1], *E. Parthoens*[2], *H. Bächinger*[3,4], *H. Kayserili*[5], *A. De Paepe*[1], *P. Coucke*[1];
[1]*Center for Medical Genetics Ghent, Ghent University Hospital, Ghent, Belgium,* [2]*Department of Biomedical Molecular Biology, Ghent University, Ghent, Belgium,* [3]*Research Department, Shriners Hospitals for Children, Portland, OR, United States,* [4]*Department of Biochemistry and Molecular Biology, Oregon Health & Science University, Portland, OR, United States,* [5]*Department of Medical Genetics, Istanbul Medical Faculty, Istanbul University, Istanbul, Turkey.*

P01.049 Myhre syndrome-causing SMAD4 mutations result in disorganization of extracellular matrix that is corrected by losartan treatment.
P. Piccolo[1], *P. Mithbaokar*[1], *V. Sabatino*[1], *J. Tolmie*[2], *M. Schiaffino*[3], *M. Filocamo*[4], *D. Melis*[5], *G. Andria*[5], *N. Brunetti-Pierri*[1,5];
[1]*Telethon Insitute of Genetics and Medicine, Naples, Italy,* [2]*Ferguson-Smith Department of Clinical Genetics, Yorkhill Hospital, Glasgow, United Kingdom,* [3]*Clinica Pediatrica, Istituto G. Gaslini, Genoa, Italy,* [4]*Centro di Diagnostica Genetica e Biochimica delle Malattie Metaboliche, Istituto G. Gaslini, Genoa, Italy,* [5]*Department of Translational Medical Sciences, Federico II University of Naples, Naples, Italy.*

P01.055 The SMAD-binding domain of SKI: a hotspot for de novo mutations causing Shprintzen-Goldberg syndrome.
D. Schepers[1], *A. J. Doyle*[2,3], *J. J. Doyle*[2], *S. L. Bessling*[2], *L. Gillis*[1], *P. Willems*[4], *S. Mansour*[6], *M. Simpson*[6], *H. Fryssira*[7], *G. R. Mortier*[1], *A. J. M. Hoogeboom*[8], *A. S. Mc Callion*[2,9], *H. C. Dietz*[2,3], *L. Van Laer*[1], *B. L. Loeys*[1];
[1]*Center of Medical Genetics, Faculty of Medicine and Health Sciences, University of Antwerp and Antwerp University Hospital, Antwerp, Belgium,* [2]*McKusick-Nathans Insititute of Genetic Medicine, Johns Hopkins University School of Medicine, Baltimore, MD, United States,* [3]*Howard Hughes Medical Institute, Baltimore, MD, United States,* [4]*GENDIA, GENetic DIAgnostic Network, Antwerp, Belgium,* [5]*SW Thames Regional Genetics Service, St George's, Univerity of London, London, United Kingdom,* [6]*Department of Medical and Molecular Genetics, Division of Genetics and Molecular Medicine, King's College London School of Medicine, London, United Kingdom,* [7]*Department of Medical Genetics, National and Kapodistrian University of Athens Medical School, Athens, Greece,* [8]*Department of Clinical Genetics, Erasmus Medical Center, Rotterdam, Netherlands,* [9]*Department of Molecular and Comparative Pathobiology, Johns Hopkins University School of Medicine, Baltimore, MD, United States.*

P01.101 Comprehensive characterization of a zebrafish model for pseudoxanthoma elasticum role reveals a role for the abcc6 transporter in cardiovascular development
M. J. Hosen, *A. Willaert, P. J. Coucke, A. De Paepe, O. M. Vanakker;*
Center for Medical Genetics, Ghent University Hospital, Ghent, Belgium.

P01.117 Antisense mediated exon skipping gene therapy for dystrophic epidermolysis bullosa
S. Turczynski[1,2], *M. Titeux*[1,2], *L. Tonasso*[3], *A. Hovnanian*[1,2,4];
[1]*INSERM U781 - Imagine Institute of Genetic Diseases, Paris, France,* [2]*University Paris Descartes Sorbonne Cité, Paris, France,* [3]*CNRS UMR5288, Toulouse, France,* [4]*Department of Genetics, Necker Hospital, Paris, France.*

P02.008 14 new patients with 6q16 deletion: a new minimal critical region and first fetopathological data
L. El Khattabi[1,2], *E. Pipiras*[1,3], *S. Drunat*[2], *F. Guimiot*[2,3], *J. Andrieux*[4], *C. Baumann*[2], *S. Bouquillon*[5], *A. L. Delezoide*[2,3], *B. Delobel*[6], *F. Demurger*[7], *H. Dessuant*[8], *C. Dubourg*[9], *C. Dupont*[2], *L. Faivre*[10], *M. Holder-Espinasse*[11], *S. Jaillard*[12], *H. Journel*[13], *S. Lyonnet*[14], *V. Malan*[14], *A. Masurel*[10], *N. Marle*[10], *C. Missirian*[15], *A. Moerman*[16], *A. Moncla*[15], *S. Odent*[17], *A. Ravel*[18], *S. Romana*[14], *A. C. Tabet*[2], *M. Vadulga*[19], *M. Vermelle*[20], *A. Verloes*[2,3], *B. Benzacken*[1,2,3], *A. Delahaye*[1,3];
[1]*Laboratoire d'Histologie-Embryologie-Cytogénétique-BDR-CECOS, Hôpital Jean Verdier, AP-HP, Université Paris 13, Bondy, France,* [2]*Département de Génétique et Biologie du développement, Hôpital Robert Debré, AP-HP, Université Paris VII, Paris, France,* [3]*U676, Inserm, Paris, France,* [4]*Laboratoire de Génétique Médicale, Hôpital Jeanne de Flandre, CHRU de Lille, Lille, France,* [5]*Institut de Génétique Médicale, Hôpital Jeanne de Flandre, CHRU de Lille, Lille, France,* [6]*Centre de Génétique Chromosomique, Saint-Vincent de Paul, GHIC, Lille, France,* [7]*Service de Génétique Clinique CLAD-Ouest-CHU Rennes, Université Rennes1, Rennes, France,* [8]*Département de Cytogénétique, Laboratoire Biomnis, Paris, France,* [9]*Laboratoire de Génétique Moléculaire, CHU Pontchaillou, UMR 6290 CNRS, IGDR, Faculté de Médecine, Université de Rennes 1, Rennes, France,* [10]*Département de Génétique, CHU Dijon, Dijon, France,* [11]*Clinical Genetics Department, Guy's Hospital, Great Maze Pond, London SE1 9RT, United Kingdom,* [12]*Laboratoire de Cytogénétique et Biologie Cellulaire, CHU Pontchaillou, Rennes, France,* [13]*Service de Génétique Médicale, Centre Hospitalier Bretagne Atlantique, Vannes, France,* [14]*Département de Génétique, Université Paris Descartes, INSERM U-781, Hôpital Necker-Enfants Malades, AP-HP, Paris, France,* [15]*Laboratoire de Génétique Chromosomique, CHU Timone enfants, AP-HM, Marseille, France,* [16]*Service de Génétique Clinique, Hôpital Jeanne de Flandre, CHRU de Lille, Lille, France,* [17]*Service de Génétique Clinique CLAD-Ouest-CHU Rennes, université Rennes1, Rennes, France,* [18]*Institut Jérôme Lejeune, Paris, France,* [19]*Laboratoire de Génétique, EA 4368, CHU de Nancy, Nancy-Université Henri Poincaré, Vandoeuvre les Nancy, France,* [20]*Service de Neuropédiatrie, CHRU de Lille, Lille, France.*

P02.083 A comprehensive study on Kabuki syndrome: diagnostics, modeling, and therapy
B. Mandriani[1,2], *L. Micale*[1], *C. Fusco*[1], *B. Augello*[1], *M. Pellico*[1], *F. Zucchetti*[3], *S. Maitz*[3], *E. Biamino*[4], *M. Silengo*[4], *A. Selicorni*[3], *L. Zelante*[1], ***G. Merla***[1,5];
[1]*IRCCS Casa sollievo della Sofferenza, San Giovanni Rotondo, Italy,* [2]*University of Brescia, Brescia, Italy,* [3]*Clinica Pediatrica, Università Milano Bicocca, Milano, Italy,* [4]*University of Torino, Torino, Italy,* [5]*University of Trieste, Trieste, Italy.*

P02.100 ZEB2 zinc-finger missense mutations lead to hypomorphic alleles and a mild Mowat-Wilson syndrome
J. Ghoumid[1,2], *L. Drévillon*[1], *M. Alavi-Naini*[3], *N. Bondurand*[2], *M. Rio*[4], *A. Briand-Suleau*[1], *M. Nasser*[1], *L. Goodwin-Swahs*[5], *P. Raymond*[1], *C. Yanicostas*[3], *M. Goossens*[1,2], *S. Lyonnet*[4,6], *D. Mowat*[7], *J. Amiel*[4,6], *N. Soussi-Yanicostas*[3], *I. Giurgea*[1,2];
[1]*AP-HP, Hôpital Henri Mondor, Service de Biochimie et Génétique, Créteil, France,* [2]*Inserm U955 équipe 11, Créteil, France,* [3]*Inserm U676, Paris, France,* [4]*Département de Génétique, Hôpital Necker Enfants-Malades, Paris, France,* [5]*Department of Clinical Genetics, Nepean Hospital, Sydney, Australia,* [6]*Inserm U781, Paris, France,* [7]*Department of Medical Genetics, Sydney Children's Hospital, Sydney, Australia.*

P03.02 Identification of Novel Homozygous Deletions in Consanguineous Pedigrees as a Shortcut to Candidate Gene Discovery in Hereditary Blindness

K. Van Schil[1], T. De Ravel[2], B. P. Leroy[3,1], H. Verdin[1], F. Coppieters[1], F. Meire[4], E. De Baere[1];
[1]Center for Medical Genetics, Ghent University Hospital, Ghent, Belgium, [2]Center for Human Genetics, Leuven University Hospitals, Leuven, Belgium, [3]Center for Medical Genetics, Department of Ophthalmology, Ghent University Hospital, Ghent, Belgium, [4]Department of Ophthalmolgy, Huderf, Brussels, Belgium.

P03.36 Antisense oligonucleotide-mediated exon skipping to treat 10% of Leber congenital amaurosis cases.

X. Gerard[1,2,3], I. Perrault[1], S. Hanein[1], E. Silva[4], K. Bigot[5], S. Defoort-Delhemmes[6], M. Rio[1], A. Munnich[1], D. Schermann[3], J. Kaplan[1], A. Kichler[2,3], J. Rozet[1];
[1]INSERMU781, Fondation IMAGINE, Paris Descartes University, Paris, France, [2]Genethon, Evry, France, [3]CNRS UMR8151-INSERMU1022, Paris, France, [4]IBILI, Faculty of Medicine, University of Coimbra, Department of Ophthalmology, Centro Hospitalar Universitário de Coimbra, Coimbra, Portugal, [5]CERTO, Paris, France, [6]Exploration de la Vision & Neuro-ophtalmologie, CHU Lilles, Lilles, France.

P04.25 Genetic variants affecting the expression of DRAM2 at 1p13.3 are associated with acute myocardial infarction with different effects for STEMI and NSTEMI

P. Salo[1], J. Sinisalo[2], J. Kettunen[3,1], A. Havulinna[4], A. Sarin[3,1], T. Hiekkalinna[1,3], S. Ripatti[3,1], P. J. Karhunen[5,6], H. Huikuri[7], M. Lokki[8], V. Salomaa[4], M. Nieminen[2], M. Perola[9,3,10];
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P04.51 Urofacial syndrome is a genetically and phenotypically heterogeneous condition caused by recessive mutations in HPSE2 and LRIG2.

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P05.071 A novel recessive intellectual disability syndrome caused by GPI-anchor deficiency

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P05.084 Loss of FMR2 further emphasises the link between deregulation of immediate early response genes FOS and JUN and intellectual disability

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P05.096 KCNT1 is the major gene causing malignant migrating partial seizures in infancy

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P05.102 The French cohort of *MECP2* duplication patients: clinical delineation of 45 affected patients

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P07.13 Pharmacokinetic studies of 2'-O-methyl phosphorothioate antisense oligonucleotides in mdx mice

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PROGRAMME POSTER AWARD FINALISTS

P08.40 A whole genome sequencing strategy to identify novel genes for frontotemporal lobar degeneration

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P09.19 Identification of the Cia27 quantitative trait gene

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P10.20 DHTKD1 mutations cause 2-aminoadipic and 2-oxoadipic aciduria and suggest a therapeutic strategy for glutaric aciduria type 1

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P10.21 Exploration of the impact of low-frequency and rare coding variation in the genetic architecture of type 2 diabetes susceptibility

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P10.25 Haplotype-based regulation of frataxin by microRNAs in Friedreich Ataxia

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P10.32 Genomic, transcriptomic, and lipodomic profiling demonstrates the benefits of extreme phenotype approach and highlights the role of inflammation in individuals low HDL-cholesterol

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P11.007 Exome sequencing identifies potential new candidate genes for colorectal adenomatous polyposis

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P11.009 Exome sequencing identifies drivers of progression of Transient Myeloproliferative Disorder to Acute Megakaryoblastic Leukemia in children with Down Syndrome

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P11.015 Unexpected diagnosis of Fanconi anemia with biallelic FANCD1/BRCA2 mutations associated with early onset colorectal tumors in adulthood: potential role of pre-mRNA splicing in phenotypic variability.

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P11.076 Immune infiltration in choroidal melanomas

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P11.084 Copy number variation analysis in 222 patients with colorectal polyposis reveals potential new causative candidate genes

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P11.103 Carriership of pathogenic *BLM* alleles is associated with early-onset colorectal cancer

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P11.107 Delineating the PMS2 cancer risk

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P11.152 Modulation of CDX2 expression by the RNA-binding protein MEX3A: impact on intestinal differentiation and stemness

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P11.157 Exome Sequencing Identifies Recurring *FLT3* N676K Mutations in Core Binding Factor Leukemia

S. Opatz[1,2,3], *H. Polzer*[1,4], *T. Herold*[1], *N. P. Konstandin*[1], *B. Ksienzyk*[1], *E. Zellmeier*[1], *S. Vosberg*[1,4], *A. Graf*[5], *S. Krebs*[5], *H. Blum*[5], *K. Hopfner*[6], *P. M. Kakadia*[6], *S. Schneider*[1], *A. Dufour*[1], *J. Braess*[7], *M. C. Sauerland*[8], *W. E. Berdel*[8], *T. Büchner*[8], *B. J. Woermann*[9], *W. Hiddemann*[1,3,4], *K. Spiekermann*[1,3,4], *S. K. Bohlander*[6,4], *P. A. Greif*[1,2,3];
[1]Klinikum LMU Munich, Munich, Germany, [2]German Cancer Research Centre (DKFZ), Heidelberg, Germany, [3]German Cancer Consortium (DKTK), Munich, Germany, [4]Helmholtz Zentrum München, Munich, Germany, [5]Gene Center LMU Munich, Munich, Germany, [6]Philipps University Marburg, Marburg, Germany, [7]St. John-of-God Hospital Regensburg, Regensburg, Germany, [8]Universität Münster, Münster, Germany, [9]German Society of Hematology and Oncology, Berlin, Germany.

P11.174 Genetic variation in immuno-modulatory genes as markers of melanoma recurrence-free and overall survival.

J. Rendleman, S. Shang, J. Shields, C. Adaniel, N. Fleming, R. Shapiro, R. Berman, A. Pavlick, D. Polsky, Y. Shao, I. Osman, ***T. Kirchhoff****;*
New York University School of Medicine, New York, NY, United States.

P11.197 Multiple primary malignant tumours: Clinical comparison of patients with and without detected germline mutations in cancer susceptibility genes.

J. Whitworth*, E. Maher;*
Birmingham Women's Hospital, Birmingham, United Kingdom.

P11.224 Novel mutations in spliceosomal gene *PRPF8* show ring sideroblast, proliferative, and missplicing phenotype in patients with myeloid neoplasms

A. Kurtovic-Kozaric[1], *H. Makishima*[1], *B. Przychodzen*[1], *J. Singh*[2], *M. Konarska*[3], *R. A. Padgett*[2], *J. P. Maciejewski*[1];
[1]Department of Translational Hematology and Oncology Research, Taussig Cancer Institute, Cleveland Clinic, Cleveland, OH, United States, [2]Department of Molecular Genetics, Lerner Research Institute, Cleveland Clinic, Cleveland, OH, United States, [3]Rockefeller University, New York, NY, United States.

P13.07 Breakpoint mapping in a family with balanced translocation t(1;12) and learning difficulties and cerebral infarctions drawn from the Finnish national registry of balanced rearrangements

T. M. Luukkonen[1,2], *M. Pöyhönen*[3,4], *R. Salonen*[5], *P. Ellonen*[1], *A. Palotie*[1,3,6], *J. Terwilliger*[7,8,2], *T. Varilo*[3,2];
[1]Institute for Molecular Medicine Finland FIMM, Helsinki, Finland, [2]National Institute for Health and Welfare, Helsinki, Finland, [3]Dept. of Medical Genetics, Haartman Institute, University of Helsinki, Helsinki, Finland, [4]Dept. of Clinical Genetics, Helsinki University Central Hospital, Helsinki, Finland, [5]Dept. of Medical Genetics, Norio-centre, Rinnekoti Foundation, Helsinki, Finland, [6]Wellcome Trust Sanger Institute, Hinxton, Cambridge, United Kingdom, [7]Dept. of Genetics and Development, Dept. of Psychiatry, Columbia Genome Center, Columbia University, New York, NY, United States, [8]Division of Medical Genetics, New York State Psychiatric Institute, New York, NY, United States.

P13.40 Targeted high-throughput sequencing of 220 genes identifies a high proportion of causative mutations in 50 patients with undiagnosed intellectual disability

C. Redin[1,2], *J. Lauer*[3], *S. Le Gras*[4], *V. Geoffroy*[4], *A. Creppy*[1,3], *Y. Herenger*[3], *Y. Alembik*[5], *M. Doco-Fenzy*[6], *B. Doray*[5], *P. Edery*[7], *S. El Chehadeh*[8], *E. Flori*[9], *G. Lesca*[7], *A. Masurel*[8], *B. Gerard*[3], *J. Muller*[1,3], *B. Jost*[4], *L. Olivier-Faivre*[8,10], *J. Mandel*[1,2,3], *A. Piton*[1,2];
[1]Department of Translational Medicine and Neurogenetics, IGBMC, Illkirch Graffenstaden, France, [2]Chaire de Génétique Humaine, Collège de France, Paris, France, [3]Laboratoire de Diagnostic Génétique, Hôpitaux Universitaires de Strasbourg, Strasbourg, France, [4]Microarray and Sequencing Platform, IGBMC, Illkirch Graffenstaden, France, [5]Service de Génétique Médicale, Hôpitaux universitaires de Strasbourg, Strasbourg, France, [6]Biology Section, Department of Genetics, University Hospital of Reims, Reims, France, [7]Laboratoire de Cytogénétique Constitutionnelle, Service de Génétique, Centre de Biologie et de Pathologie Est, Hospices Civils de Lyon, Bron, France, [8]Centre de génétique et Centre de Référence Anomalies du développement et Syndromes malformatifs, Hôpital d'Enfants, CHU Dijon, Dijon, France, [9]Service de cytogénétique, Hôpitaux universitaires de Strasbourg, Strasbourg, France, [10]Génétique des anomalies du développement (GAD) EA 4271, Faculté de Médecine, Université de Bourgogne, Dijon, France.

P13.57 Targeted "ciliome" sequencing for gene identification in nephronophthisis and associated ciliopathies

P. Krug[1,2,3], *E. Filhol*[1,2,3], *C. Masson*[3,2], *M. Parisot*[2], *C. Bole-Feysot*[2], *P. Nitschké*[3,2], *I. Perrault*[4,2,3], *R. Salomon*[1,2,5], *T. Attié-Bitach*[6,2,3], *K. Tory*[7,1,2], *C. Antignac*[1,2,8], *C. Jeanpierre*[1,2,3], *S. Saunier*[1,2,3];
[1]Inserm U983, Paris, France, [2]Imagine Institute, Paris, France, [3]Université Paris Descartes, Sorbonne Paris Cité, Paris, France, [4]Inserm 781, Paris, France, [5]Department of Paediatric Nephrology, AP-HP, Necker Hospital, Paris, France, [6]Inserm U781, Paris, France, [7]Ist Department of Pediatrics, Semmelweis University, Budapest, Hungary, [8]Department of Genetics, AP-HP, Necker Hospital, Paris, France.

P15.70 Transcriptome sequence genomic analysis in three tissues of a twin cohort reveals complex causes of allelic expression

A. Buil[1,2,3], *A. Brown*[4], *A. Viñuela*[5], *M. Davies*[5], *K. Small*[5], *M. Gallardo*[6], *D. Glass*[5], *M. Blasco*[6], *R. Durbin*[4], *T. D. Spector*[5], *E. T. Dermitzakis*[1,2,3];
[1]University of Geneva, Geneva, Switzerland, [2]Institute of Genetics and Genomics in Geneva, Geneva, Switzerland, [3]Swiss Institute of Bioinformatics, Geneva, Switzerland, [4]Wellcome Trust Sanger Institute, Hinxton, United Kingdom, [5]King's College London, London, United Kingdom, [6]Spanish National Cancer Centre, Madrid, Spain.

PROGRAMME POSTER AWARD FINALISTS

P15.74 Characterising the genetic architecture of the miRNA response to Mycobacterium tuberculosis

K. J. Siddle[1], *L. Tailleux*[2], *Y. Nédélec*[3], *E. Patin*[1], *L. Barreiro*[3], *L. Quintana-Murci*[1];
[1]*Institut Pasteur, Unit of Human Evolutionary Genetics, Paris, France,* [2]*Institut Pasteur, Unit of Mycobacterial Genetics, Paris, France,* [3]*CHU Sainte-Justine, University of Montreal, Montreal, QC, Canada.*

P16.057 Assessing the contribution of chromosome X to complex traits: Two new loci for height and evidence for incomplete dosage compensation

T. Tukiainen[1], *M. Pirinen*[1], *A. Sarin*[1,2], *C. Ladenvall*[3], *J. Kettunen*[1,2], *J. Eriksson*[4,5,6], *A. Jula*[7], *L. Groop*[3], *V. Salomaa*[4], *O. T. Raitakari*[8,9], *M. Järvelin*[10,11], *S. Ripatti*[1,2,12];
[1]*Institute for Molecular Medicine Finland (FIMM), Helsinki, Finland,* [2]*Unit of Public Health Genomics, National Institute for Health and Welfare, Helsinki, Finland,* [3]*Department of Clinical Sciences, Diabetes and Endocrinology, Lund University and Lund University Diabetes Centre, CRC at Skåne University Hospital, Malmö, Sweden,* [4]*Department of Chronic Disease Prevention, National Institute for Health and Welfare, Helsinki, Finland,* [5]*Department of General Practice and Primary Healthcare, University of Helsinki, Helsinki, Finland,* [6]*Unit of General Practice, Helsinki University Central Hospital, Helsinki, Finland,* [7]*Population Studies Unit, Department of Chronic Disease Prevention, National Institute for Health and Welfare, Helsinki, Finland,* [8]*Department of Clinical Physiology and Nuclear Medcine, Turku University Hospital, Turku, Finland,* [9]*Research Centre of Applied and Preventive Cardiovascular Medicine, Turku, Finland,* [10]*Department of Epidemiology and Biostatistics, Faculty of Medicine, Imperial College London, London, United Kingdom,* [11]*Institute of Health Sciences, Biocenter Oulu, University of Oulu, Oulu, Finland,* [12]*Hjelt Institute, University of Helsinki, Helsinki, Finland.*

P16.098 Investigating the Effect of Sequencing Depth on Low Coverage Next Generation Sequencing Association Studies

T. S. Shah, *J. Z. Liu, Y. Luo, M. O. Pollard, C. A. Anderson;*
Wellcome Trust Sanger Institute, Cambridge, United Kingdom.

P18.08 Clarifying assent

N. A. A. Giesbertz, *A. L. Bredenoord, J. J. M. van Delden;*
UMC Utrecht, Julius Center, Utrecht, Netherlands.

P18.52 Reflecting on earlier experiences with unsolicited findings: Points to consider for next generation sequencing and informed consent in diagnostics

T. Rigter[1], *L. Henneman*[1], *U. Kristoffersson*[2], *A. Hall*[3], *H. G. Yntema*[4], *P. M. Borry*[5], *H. Tönnies*[6], *Q. Waisfisz*[7], *M. W. Elting*[7], *W. J. Dondorp*[8], *M. C. Cornel*[1];
[1]*VU University Medical Center, Department of Clinical Genetics, Section of Community Genetics and the EMGO+ Instittute for Health and Care Research, Amsterdam, Netherlands,* [2]*Lund University, Department of Clinical Genetics, University and Regional Laboratories, Region Skane, Lund, Sweden,* [3]*PHG foundation, Cambridge, United Kingdom,* [4]*Radboud University Nijmegen Medical Centre, Department of Human Genetics, Nijmegen, Netherlands,* [5]*Centre for Biomedical Ethics and Law, Department of Public Health, Leuven, Belgium,* [6]*Robert Koch-Institut, Geschäftstelle Gendiagnostik-Kommission, Berlin, Germany,* [7]*VU University Medical Center, Department of Clinical Genetics, Amsterdam, Netherlands,* [8]*Maastricht University, Department of Healt, Ethics and Society, Research Schools CAPHRI and GROW, Maastricht, Netherlands.*

P18.75 The marketing of gene therapy medicinal products: What are the regulatory challenges?

A. Mahalatchimy[1,2], *W. Meng*[1,3], *A. Cambon-Thomsen*[1], *A. M. Duguet*[1], *E. Rial-Sebbag*[1];
[1]*UMR1027, Inserm and Université de Toulouse III - Paul Sabatier, Toulouse, France,* [2]*IRDEIC Université Toulouse 1 Capitole, Toulouse, France,* [3]*Shandong University, School of law, Jinan, China.*

P18.79 Telemedicine use in Clinical Genetics; a European survey

E. Otten[1], *E. Birnie*[1], *A. V. Ranchor*[2], *I. M. Van Langen*[1];
[1]*Department of Clinical Genetics, University of Groningen, University Medical Center Groningen, Groningen, Netherlands,* [2]*Department of Health Psychology, University of Groningen, University Medical Center Groningen, Groningen, Netherlands.*

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.

Conference Venue
Palais des Congrès
2 Place de la Porte Maillot
75017 Paris
France

Badges
Participants should collect name badges from the conference registration desks. As only registered participants will be permitted to attend the scientific sessions, the Exhibition and poster areas, you are kindly asked to wear your badge when entering the congress venue.
Guests 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 9.00 - 17.15 hrs and are closed over the weekend. There are multiple bank machines (ATMs) open 24 hours a day throughout the city which accept all major international bank cards. One is located in the conference center. The official currency of France is the Euro (€). Major credit cards are widely accepted, but please always check beforehand.

Cancellations and Refunds
Notice of cancellation had to be made before the conference 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, 2013: 75% refund
- between April 1 and May 23, 2013: 25% refund
- after May 23, 2013: no refund

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

Car Parking
The Palais des Congrès de la Porte Maillot has plenty of parking facilities. Note that the traffic in Paris is sometimes considerable and public transportation is definitely recommended, especially given that the venue has its own subway station.

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

Climate
The weather in June is generally nice (average high: 22°C, average low: 13°C), evenings may be cooler and of course occasional rain showers have to be expected.

Cloakroom and Luggage
A cloakroom and luggage storage is available. Persons with reduced mobility needing to store luggage should contact the information counter for assistance. Price: EUR 2 per garment, EUR 3 per luggage.

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.

INFORMATION GENERAL INFORMATION

Coffee Breaks
During the session breaks refreshments (coffee, tea and lemonade) will be served free of charge in the exhibition area to participants wearing name badges.

Currency
The official currency of France is the Euro (€). 1 EUR = 1,30 USD = 0,84 GBP = 1,32 CAD = 127 JPY = 1,22 CHF = 1,26 AUD as per date of printing, for orientation purposes only. No responsibility is taken for the correctness of this information.

Drinking water
The tap water in France can be used without concern. People have however the habit of buying bottled drinking water.

Eating Out in Paris
As one would expect, Paris is definitely one of the world's gastronomic capitals when it comes to french and international cuisine, 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.
A guide to eating in Paris: http://www.placesinfrance.com/eating_in_paris_restaurants.html
Eating and drinking in Paris: http://www.timeout.com/paris/en/eating-drinking
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 NF C 61-314 sockets (includes also CEE 7/7, 7/16 and 7/17 plugs).

Emergency Services Phone Numbers
European Emergency Number: 112. (Alternatively, Police: 17; Hospital-based Ambulance (SAMU): 15; Fire Service-based Ambulance: 18; Fire: 18).

Exhibition Opening Hours
Sunday, June 9: 08.30 – 18.00 hrs
Monday, June 10: 08.30 – 18.00 hrs
Tuesday, June 11: 09.00 – 13.30 hrs

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 with your provider which roaming company in France offers the cheapest tariffs.

Insurance
In registering for the ESHG 2013 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. Login data are as follows: SSID: ESHG2013, password: eshg2013

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

Lunch and Refreshments
Vouchers for lunch boxes had to be pre-ordered and cannot be purchased on site. Please note that lunch vouchers are not refundable.

Message Centre
A Message Board is available in the registration area on level 0.

Pharmacy
Most pharmacies are open during regular trading hours, between 8.00 and 20.00 hrs (some 21.00 hrs). Closed on Sunday.

INFORMATION GENERAL INFORMATION

Poster Removal
The organisers cannot assume any liability for loss or damage of posters displayed in the poster area. Posters that were not removed after the end of the meeting on Tuesday, June 11, 2013, 13.30 hrs, will be removed by the staff and will not be kept or mailed to the author after the meeting.

Preview Room
Equipment for a final check of the sequence of your presentation is available in the preview room 111/112/113 on the first floor, room 111/112/113. 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, June 8	08.30 – 20.00 hrs
Sunday, June 9	07.45 – 18.45 hrs
Monday, June 10	07.45 – 18.45 hrs
Tuesday, June 11	08.30 – 15.30 hrs

Safety - Crime
Paris can be considered a safe city 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 photocopy 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 center.
- In heavily frequented tourist zones, be aware of attemps of scam and pickpocketing (especially in the metro).
You may also notice patrols of military dressed in camouflage uniform. These soldiers are part of the “Plan Vigipirate”, an anti-terrorist vigilence plan.

Shops
Large shops and department stores are open between 9.00 and 19.00 hrs from Monday to Saturday. Large department stores have a late closure on one day per week. All shops, with the exception of small souvenir or food and convenience stores, are closed on Sunday. All major credit cards are generally accepted, but it is not possible to pay with foreign banknotes.

Smoking Policy
The ESHG 2013 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
Taxis in Paris can be considered as relatively cheap, as long as the traffic situation is adequate. Taxis will generally take only 3 persons in a regular car, hence do indicate at the time of reservation in case you require a van. Also a number of taxis DO NOT accept credit cards, please check this before boarding. If you need a receipt ask for “un reçu s’il vous plait”.
A white/green light on the roof indicates the availability, a red/orange light means that the taxi is occupied.

Telephone calls
The country code of France is 33 and the area code for Paris is 1. If calling a number in Paris from within France (including Paris!), dial 01 before the subscriber number.

Tipping
By law, 15% for service is included in the price. It is common to leave (part of) the change, when paying cash (coins). Do not add anything to your credit card bill, as this will not be passed on to the waiters.

INFORMATION GENERAL INFORMATION

Tourist Information Centres
A list of welcome centers can be found here: http://en.parisinfo.com/where-to-find-us/.
Website of the Paris visitor bureau: http://en.parisinfo.com

Travelling - Accessibility - Public Transportation
The Palais des Congrès has its own railway station ("Porte Maillot") on line 1 & RER C.
Paris has an extensive metro system, which will allow you to get practially everywhere, mostly quicker than by car, given the sometimes heavy traffic in Paris.
See this article for lots of information on the Parisian public transportation system:
http://goparis.about.com/od/transportation/ss/Metro_and_Buses.htm

Travelling from and to Paris Airports
Roissy Charles de Gaulle (CDG) is connected via
- Train RER B (blue) to Denfert-Rochereau, Saint-Michel-Notre-Dame & Gare du Nord, every 10-15 minutes. Fare: EUR 9,10 (full fare)
- RoissyBus shuttle to Opéra, every 15-20 minutes. Fare: EUR 10.-
- Air France Bus Line 2 to Palais des Congrès and Etoile, every 30 minutes. Fare: Single: EUR 17.-, Return: EUR 29,-

Paris Orly (ORY) is connected via
- Train RER B (blue) to Gare du Nord, Chatelet Les Halles, St-Michel Notre Dame, Luxembourg & Denfert Rochereau, every 10-15 minutes. Fare: EUR 10,90 (full fare).
- Orlybus shuttle to Denfert-Rochereau, every 20-30 minutes. Fare: EUR 7,20
- Air France Bus Line 1 to Gare Montparnasse, Invalides, Étoile, every 30 minutes. Fare: Single: EUR 12.-, Return: EUR 20,-

V.A.T.
The VAT rate is 19,6% (7% on non-alcoholic drinks and food).

INFORMATION REGISTRATION FEES

Registration fees Payment received:	***before March 31, 2013 (reduced rate)***	***between March 31 & May 23, 2013 (normal rate)***	***after May 23, 2013 and on site***	***Day tickets***
ESHG Members	EUR 300.-	EUR 400.-	EUR 460.-	EUR 170.-
Non-Members	EUR 415.-	EUR 525.-	EUR 600.-	EUR 200.-
Postgraduate Trainees*	EUR 225.-	EUR 280.-	EUR 320.-	EUR 130.-
Students**	EUR 150.-	EUR 180.-	EUR 215.-	EUR 85.-
Guests	EUR 85.-	EUR 85.-	EUR 85.-	N/A
	Networking Dinner	Networking Party	Networking Package	
Members, Non-Members, Guest	EUR 59.-	EUR 29.-	EUR 79.-	
Students/Postgrad. Trainees	EUR 39.-	EUR 19.-	EUR 54.-	

* Applies to MSc./PhD Students, research students. Please provide a confirmation signed by the head of department **at the moment of your registration** by fax to +43 1 407 82 74 or together with the hardcopy of the registration form. **Confirmations handed in at a later stage cannot be considered**.

** Please provide a copy of a Student's ID or a confirmation signed by the head of department **at the moment of your registration** by fax to +43 1 407 82 74 or together with the hardcopy of the registration form. **Confirmations handed in at a later stage cannot be considered**.

***Guest registration is **only available for family members of registered participants**. The fee includes admission to the Networking Mixer (Saturday) and the exhibition, no admission to scientific sessions. Guest badges are orange.

What is covered by the registration fee?

Participants:
- Admission to all scientific sessions, exhibition and networking mixer
- Electronic abstract book and printed programme
- Coffee/Tea during breaks from Saturday, June 8 to Tuesday, June 11

Guests (family members only):
- Access to the 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 23), 2013 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, 2013: 75% refund
- Between April 1 and May 23, 2013: 25% refund
- After May 23, 2013: no refund

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

INFORMATION NETWORKING EVENTS

Opening Networking Mixer

Saturday, June 8, 2013, 20.00 - 21.30 hrs - Palais des Congrès de Paris (conference venue)

Network with your colleagues at this mixer following the second plenary session 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 Dinner and Party

Monday, June 10, 2013, 20.00 hrs - Muséum National d'Histoire Naturelle, Grande Galerie de l'evolution / Jardin des Plantes

Join us for a networking dinner (20:00 - 22:30) at the Muséum National d'Histoire Naturelle, Grande Galerie de l'evolution and / or the subsequent networking party (22:30 - 02:00) in the beautiful adjacent Jardin des Plantes. The entrance fees for these unique Paris experiences are:

- Dinner ticket: EUR 59.- (reduced student fee: EUR 39.-)
- Party ticket: EUR 29.- (reduced student fee: EUR 19.-)
- Combi ticket for Dinner and Party: EUR 79.- (reduced student fee: EUR 54.-)

The entrance fee for the dinner includes the access to the Muséum National d'Histoire Naturelle, Grande Galerie de l'evolution, a french style gala buffet, drinks and music.
The entrance fee for the party includes the access to the party area in the Jardin the Plantes, all drinks, a live band and a DJ.

Note: Tickets for the dinner are limited, please register early. The party is sold out.

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

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 Areas – Level 1 – Dates & Opening Hours

Sunday, 9 June 2013	08.30 – 18.00 hrs
Monday, 10 June 2013	08.30 – 18.00 hrs
Tuesday, 11June 2013	09.00 – 13.30 hrs

Posters – Mounting, Viewing & Removal Schedules

Posters will be accissible in the exhibition hall, Level 1, from June 9 – 11. Note the below schedule please.

Poster mounting	Saturday, 8 June 2013	12.00 – 14.00 hrs	STRICT
Poster mounting	Sunday, 9 June 2013	as of 08.30 hrs	
Poster viewing	Sunday, 9 June 2013	08.30 – 18.00 hrs	
Poster viewing	Monday, 10 June 2013	08.30 – 18.00 hrs	
Poster viewing	Tuesday, 11 June 2013	09.00 – 12.30 hrs	
Poster removal	Tuesday, 11 June 2013	12.30 – 13.30 hrs	STRICT

After 13.30 hrs no access to poster areas due to safety regulations.

Please note that posters not removed by 13.30 hrs on Tuesday 11 June will be taken down by the staff of the conference center 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*".

Exhibition Catalogue

All further informatin on exhibitors and the products and services they offer can be found in the Exhibition Catalogue in your conference bag.

Lead Retrieval System used by Exhibitors

Again this year, 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.