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Sunday, 28 May | 19.15 – 20.45 | Room Ballerup
TOUGH TARGETS, SIMPLE GENOTYPING.

New solutions for CRISPR, mitochondrial disease, neurology, and pharmacogenetic research

Saturday, 27 May
12.15–13.45 hrs
Birmingham Room

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EUROPEAN HUMAN GENETICS CONFERENCE 2017
Bella Center | Copenhagen, Denmark | May 27 - 30

PROGRAMME
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Dear Colleagues and Friends,

On behalf of the board of the European Society of Human Genetics, I would like to cordially welcome you to the European Human Genetics Conference 2017 in Copenhagen, Denmark.

The ESHG 2017 marks the 50th Anniversary of the first ESHG Conference which took place in Copenhagen in 1967. In commemoration of this event, we have chosen the Bella Center in Denmark’s capital as the venue for our meeting.

Denmark is not only renowned for the Little Mermaid, Hans Christian Andersen and Niels Bohr, but also for Danish Design and the “New Nordic Cuisine”. Landing at Copenhagen airport, you will have a wonderful view of “The Bridge” connecting Denmark and Sweden. Copenhagen is one of the most vivid cities in northern Europe, although it is also referred to as Northern Europe’s cosiest capital; a city full of street cafés, design shops and some of the best restaurants in Scandinavia. From the winding streets of the beautiful old town and the grand royal palaces to the city’s cutting-edge buildings and attractions, Copenhagen is a great mix of the old and the new world.

Copenhagen is also known for being an international hub for science. It is number one in Europe in terms of clinical trials per capita and number two in the world for developing biotechnology. In fact, COBIS, the Copenhagen Bio Science Park, was named the world’s best biotech incubator in 2011. Furthermore, the newly built Copenhagen Science City with its Niels Bohr Building are significant investments of the Danish government to enhance science and to attract the world’s best scientists.

The combination of history, culture and science led us to choose Copenhagen as the best place to celebrate our 50th Anniversary. Among other highlights, the ESHG 2017 conference provides the latest findings in the field of basic and applied human genetics. Thanks to the excellent programme committee selecting the best speakers and outstanding presentations we are looking forward to a highly inspirational meeting.

Velkommen til København!

Olaf Riess, MD
President
European Society of Human Genetics
European Society of Human Genetics

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T: +46 8 5465 1500
E: confirmation@mci-group.com
## General Acknowledgements-Future Meetings

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

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### Future European Human Genetics Conferences

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

- **European Human Genetics Conference 2019**
  - Gothenburg, Sweden
  - June 15 – 18, 2019

- **European Human Genetics Conference 2020**
  - Berlin, Germany
  - June 6 – 9, 2020

### 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 2017 is designated for a maximum of **22 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.

### Download the ESHG 2017 Conference App

**for iOS**

![QR Code for iOS](QRCode.png)

**for Android**

![QR Code for Android](QRCode.png)

### Important Notice

Please note that taking pictures or filming during the sessions is forbidden (no matter if done with a camera or a mobile phone). Persons who will not observe this rule will be excluded from the session by the chairpersons.
GENERAL GET THE MOST OUT OF THE ESHG 2017

Get the most out of your ESHG Meeting!

We are glad to announce the following features which might contribute to your positive experience of the ESHG conference.

The ESHG 2017 Congress App

Do you always want to be up-to-date? The ESHG Congress App will guide you through the programme day by day or by session type, will show available profiles of the speakers and help you to find exhibitors by name or by service provided. Add papers or entire sessions to your mobile calendar, receive push messages with important reminders and give feedback on talks or sessions. Our staff at the General Information desk will help you in case of questions.

Available for iOS and Android in your App and Play Stores. Search for ESHG 2017 Congress.

Commenting

Do you have a specific comment on the running presentation? To discuss with colleagues, know that many attendees will be using twitter with the hashtags #eshg2017 #sessionnumber (e.g. #eshg2017 #S01).

For all sessions, remember to use the discussion microphones in aisles of the lecture halls to direct your questions to the speakers.

e-Posters & Best Posters

For the second time, a number of posters will be presented as e-Posters at 40 e-Poster stations in the exhibition hall. The list of available posters can be viewed on any of these screens. From there, each e-Poster can be selected for viewing. Use the zoom-in, zoom-out function to focus on specific parts of the e-Posters and the navigation icons to browse though the multiple slide posters.

This year, the 19 Best Posters were selected for a short oral presentation (2 minutes) in the Concurrent Session C03, on Saturday, May 27, 18.30 - 19.10 hrs. After the presentation of all posters, the authors and the audience will proceed to the electronic posters in front of the lecture hall for discussion with the authors for the remainder of the session.

The list of e-Posters is also available on https://2017.eshg.org/index.php/abstracts-2/eposters.

Live streaming and on-demand webcast of selected sessions

For the second time, all Educational Sessions will be available as webcast after the meeting. So in case you are interested in a Symposium and a parallel Educational session, no worries, you can watch it at home or whenever you have time.

As usual, the Plenaries on Tuesday (including the Building Bridges Session joint with the ASHG) and the Symposium joint with the European Society of Cardiology (ESC) will be available as live webcast and as on-demand streaming after the conference.

The following sessions are planned to be available:
- E1-E13
- PL1, PL3, PL4 & PL5
- S15

Note that the actual availability of the individual talks depends on the consent of the speakers.

Live stream in the exhibition

The plenary lecture hall is equipped with a live transmission possibility to the Live stream area in the exhibition. The programme of the room Aarhus will be transmitted to this area during exhibition opening hours.

Poster viewing with authors

Posters will be discussed in 4 groups (A-D), 10.15 – 11.15 hrs and 16.45 – 17.45 hrs both on Sunday and Monday.

We hope that this will stimulate the interaction at the posters and increase convenience for the presenters.

All posters will remain on display from Saturday to Monday during exhibition opening hours.

Young Investigators in Focus

The workshop ‘W11 . Career development and funding opportunities for young investigators’ on Sunday aims directly at young investigators attending the conference.

Two types of fellowships were allocated to young investigators from European and Non-European countries. In total the ESHG awarded more than 100 fellowships to young investigators in 2017.

You might also be interested to know that the Scientific Programme Committee decided to have at least 30% of its members aged under 40 years by 2020.

Post-doc Young Investigator Award Winners of 2016 have been invited to co-chair a session at this year’s conference.

Have a look at this year’s candidates on page 55 ff.
**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 (E01 – E13)**

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

**Concurrent Sessions (C01 – C23)**

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

**Poster Viewing with Authors**

Posters are numerically the major scientific presentations of the meeting. Most attendees bring a poster showing data and progress with their personal research. Posters offer an excellent opportunity for people interested in a particular topic to meet and exchange ideas and network with other researchers. Posters should NOT be used to advertise a product or service. Like a paper, a poster abstract should detail the focus of the presentation and the way(s) in which it contributes to the body of knowledge in its field.

Times marked “Poster Viewing with Authors” should be used for communication and interaction with the poster authors, who are requested to be at their posters at these times. Posters will be on display throughout the conference for free poster viewing (Saturday-Monday). Posters bearing a rosette have received a high score during the peer review process and are considered the best posters submitted by young investigators. They are the candidates for the ESHG poster awards.

**Workshops (W01 – W18)**

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-CS25)**

There are a number of company satellites planned within the main conference programme. Sponsors are approved as reputable and relevant by the Scientific Programme Committee, but the detailed content of the presentations is proposed directly by the sponsors and under their responsibility. Neither the ESHG nor the organisers have endorsed the content in any way.
Saturday, May 27, 2017

Time Aarhus Copenhagen Cannes Alicante Amsterdam Cologne Cambridge Ancona Birmingham Belgrade

08.15 – 10.15
Sponsored Session E01 Sequencing

10.30 – 12.00
E02 CRISPR/Cas9 genome editing to model disease

W01 NGS in the Clinic
E03 50 Shades of Cancer Genetics
E04 Channelopathies
E05 Imprinting-related disorders
W02 A case that changed my life as a geneticist
W03 Rareconnect: Connecting rare disease patient organisations
W04 UCSC Genome Browser

12.15 – 13.45
Lunch break / Posters / Exhibition

14.00 – 14.30
Opening Welcome Addresses

14.30 – 16.00
PL1 Opening Plenary Session

16.00 – 16.30
Fruit break / Posters / Exhibition

16.30 – 18.00
PL2 What’s New? Highlights Session

18.00 – 18.30
Coffee break / Posters / Exhibition

18.30 – 20.00
C01 Personalized Medicine and Pharmacogenomics
C02 Neurogenetics 1
C03 Best Posters Session
C04 Epigenetics and Gene Regulation
C05 Skin and Bones
C06 ELSI genomics

20.00 – 21.45
Opening Networking Mixer at the Bella Center (Centerhall, ground floor)

Session Types:
- Plenary Session
- Symposium
- Concurrent Session
- Workshop
- Educational Session
- Sponsored Session
- Corporate Satellite

IMPORTANT NOTICE:
Please note that taking pictures or filming during the sessions is forbidden (no matter if done with a camera or a mobile phone). Persons who will not observe this rule will be excluded from the session by the chairpersons.
### Monday, May 29, 2017

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**Networking Party at the Øksenhallen (at own expense - ticket required)**
### Tuesday, May 30, 2017

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<td><strong>C20</strong> Molecular syndromology</td>
<td><strong>C21</strong> Cardiovascular disorders</td>
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#### Session Types:
- Plenary Session
- Symposium
- Concurrent Session
- Workshop
- Educational Session
- Corporate Satellite

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

**Posters will be on display from:** Saturday, May 27, (09.30 hrs) to Monday, May 29 (17.45 hrs)

**Poster mounting will be possible on:** Saturday, May 27, from 09.30 hrs onwards

**Removal will be mandatory on:**
- Groups A-C: Monday, May 29, 2017 from 16.45 hrs
- Group D: Monday, May 29, 2017: 17.45 - 18.00 hrs

You can find your poster board number in the author index of the Poster Listing available at the “Poster Help Desk” located at the entrance to the Exhibition Hall or at the two information points located in the poster area.

**Access after Monday, 18.00 hrs is not possible!** Safety regulations in place for the exhibition break-down do not allow participants in the hall after this time. Please note that posters not removed until this time will be taken down by the staff of the conference centre. They will be available for (unsupervised) pickup until Tuesday, 16.00 hrs, but will not be stored afterwards or sent to the authors after the meeting.

**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:
- Poster Group A: 10.15-11.15 hrs on Sunday, May 28 for posters with board numbers ending with “A” (e.g. P01.01A)
- Poster Group B: 16.45-17.45 hrs on Sunday, May 28 for posters with board numbers ending with “B” (e.g. P01.01B)
- Poster Group C: 10.15-11.15 hrs on Monday, May 29 for posters with board numbers ending with “C” (e.g. P01.01C)
- Poster Group D: 16.45-17.45 hrs on Monday, May 29 for posters with board numbers ending with “D” (e.g. P01.01D)

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

### Technical Information for Presenters of E-Posters

**Schedule for display and upload**

Electronic Posters will be on display from Saturday, May 27 (09:30 hrs) to Monday, May 29 (17:45 hrs).

The upload of the poster file will be possible in the Preview Centre from Friday, May 26 from 14:00 hrs onwards (during conference times).

### Technical Information for Presenters of Talks

- All rooms will be equipped with data projection.
- It is essential that you load and view your presentation in the Preview centre not later than 2 hours in advance (30 minutes for the first morning talks).
- The lecture rooms are exclusively equipped with Windows-PCs (no MACs). In case you absolutely need to use your own laptop or notebook, please contact the Speakers’ Preview well in advance of your talk to check compatibility.
- Please bring a USB-key or CD-ROM all formatted for Windows® (PC). You may want to carry a second key/CD as a back-up in case there is any insoluble technical problem.
- File Format: Microsoft® PowerPoint 2007™ presentation formatted for Windows® (PC) only.
- Preferred Resolution: XGA (1024 x 768 pixel)
- Screen format: 4:3
SCIENTIFIC PROGRAMME
Saturday, May 27, 2017
Programme Saturday, May 27

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Esprit Yildirim, Istanbul, Turkey

The Insulin Producing Bacterial Cell
Emre Akbas, Kadikoy/Istanbul, Turkey

My future job Abiey Abedin
Tehran, Iran

Genetics is part of my DNA
Chantal van Wyk, Bloemfontein, South Africa

Genetics: Legal and Ethical Limitations
Iulia-Teodora Perva, Timisoara, Romania

Multidisciplinary approach in treating patients
Anastasia Vasina, Saint-Petersburg, Russia

The secret of success: exome sequencing over candidate gene approach
Yeserin Yildirim, Istanbul, Turkey

Brain channelopathies
Renzo Guerrini, Florence, Italy

Overview on imprinting related disease
Deborah J.G. Mackay, Southampton, United Kingdom

We invited professionals in medical genetics to tell us about an event or moment when genetics made a difference and had a profound impact on how they see the field of Medical Genetics.

Integrating the Human Phenotype Ontology into an online patient community. Looking towards patient self-phenotyping on RareConnect in combination with Phenotips, Orphan Buske, Sick Children’s Hospital, Toronto

The Orphanet experience of integrating the Human Phenotype Ontology into its database
Annie Olry, Orphanet

An update on progress and recent developments from the Matchmaker Exchange initiative
Kym Boycott, University of Ottawa

The UCSC Genome Browser is a widely used platform for access to genomic data provided by laboratories around the world and for display of user data alongside it.

New tools are constantly being developed by our team, and this workshop will demonstrate the latest offerings (Data Integrator, display modes tailored to whole-exome sequencing and RNA-seq, Genome Browser-in-a-Box).

Previous familiarity with the Genome Browser is recommended, but not required. Laptops strongly encouraged.
**PROGRAMME SATURDAY, MAY 27**

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<td>Welcoming Addresses by</td>
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<td>Olaf Riess</td>
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<td>President of the Danish Society of Human Genetics</td>
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<td>PL1.50 Years of ESHG</td>
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<td>PL1.1 A brief history of how we got here</td>
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<td>PL1.3 The Future: Solved Problems and Persisting Challenges</td>
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<td>18.00</td>
<td>Enhancer composition and dosage control developmental gene expression</td>
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<td>Anja J. Will*</td>
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<td></td>
<td>Berlin, Germany</td>
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<tr>
<td>16.45</td>
<td>PL2.2 Quantifying the impact of rare coding variation across the phenotypic spectrum</td>
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<td>Andrea Ganna*</td>
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<td>Cambridge, United States</td>
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<td>17.00</td>
<td>De novo gain-of-function mutations in the epigenetic regulator SMCHD1 cause Bosma arhinia microphthalmia syndrome</td>
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<td></td>
<td>Chris T. Gordon</td>
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<td>Paris, France</td>
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<tr>
<td>17.15</td>
<td>Genetic variation in the Estonian population: a pharmacogenomic study of adverse drug reactions using electronic health records</td>
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<td>Kristi Krebs*</td>
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<td>T. Tasa, M. Kals, R. Mägi, T. Eksa, A. Metspalu, J. Viil, L. Milani;</td>
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<td>Tartu, Estonia</td>
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<tr>
<td>17.30</td>
<td>From pathogenic mechanism to a therapeutic approach in Spinocerebellar Ataxia 38 (SCA38)</td>
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<td>Torino, Italy</td>
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<td>17.45</td>
<td>Analysis of de novo mutation clustering identifies candidate disease genes in neurodevelopmental disorders due to likely gain-of-function and dominant-negative mechanisms</td>
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<td>Nijmegen, Netherlands</td>
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<td>18.00</td>
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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.
18.30 - 20.00
Copenhagen - Aarhus, Copenhagen, Cannes, Alicante, Amsterdam, Cologne

18.30
C01.1 The role of Next-generation sequencing in tumours in Adolescents and young adults (AYA) with advanced solid tumors participating in phase I trials Terri P. McLeigh*, R. Sundar, N. Diamantis, S. Kaye, U. Banerjei, J. Lopez, J. de Bono, W. van der Graaf, A.J. George, Sutton, United Kingdom

C01.2 Allele-specific silencing as therapeutic strategy for disorders due to gene duplication: a proof-of-principle in Autosomal Dominant LeukoDystrophy (ADLD), Elisso Giorgio*, A. Bartollett-Stella, A. Brussoni, C. Mancini, S. Cavallero, M. Ferrero, E. Di Gregorio, E. Pozzi, E. Riberi, L. Guappini, P. Cortelli, S. Capoleti, A. Brusco, Torino, Italy

C02
C02.1 Best Posters Session
Chair: Gunnar Hougé
Joris Veltman

C02.2 CHRNA7 CNVs: shared clinical phenotypes mediated by differing molecular mechanisms Madelyn A. Gillentine*, J. Yin, S. Cummock, J.J. Kin, A. Bajic, C.P. Schoaf; Houston, United States

20.00

19.00
C03.1 A first genome-wide systems genetics approach identifies risk loci and pathways for cardiometabolic susceptibility, Vasiliki Matzaraki*, M. Jaeger, R.A. Gamba, S. Smeekens, M. Oosting, F. van de Veen, L.J., M.G. Heijna, C. Wijmenga, V. Kumar; Groningen, Netherlands

C03.2 Generating large-scale datasets for interpreting regulatory variants Martin Kircher, F. Inoue, C. Xiong, B. Martin, N. Ahituv, J. Shendure; Berlin, Germany

20:30-21:00: Opening of ESHG Awards Gala

C04
C04.1 Polymer physics predicts the effects of structural variants on chromatin architecture Dario G. Lupiáñez, S. Bianco, A.M. Chanetello, C. Amann-Zoberi, C. Kraft, R. Schöpf, L. Wittler, G. Andrey, M. Vingron, A. Pambasa, S. Mundlos, M. Nicodem; Berlin, Germany

C04.2 Changes in chromatin interaction dynamics at the PITX1 locus cause congenital limb malformations Matte Spielmann, B. Kraggsteen, C. Paliou, R. Schöpf, V. Heinich, L. Harabula, D. Lupianez, M. Franke, M. Hochradel, K. Kraft, J. Jerkov, L. Wittler, S. Mundlos, G. Andrey; Berlin, Germany

C04.3 Generating large-scale datasets for interpreting regulatory variants Martin Kircher, F. Inoue, C. Xiong, B. Martin, N. Ahituv, J. Shendure; Berlin, Germany

C05


21.00
C06.1 Uncertainty about carrier results from exome sequencing: A randomized controlled trial of disclosure Barbara B. Biesecker, K. Lewis, K.L. Umstead, J. Johnston, L.G. Biesecker; Bethesda, United States

C06.2 Recommendations for the reporting of results from diagnostic next generation sequencing Danya F. Vears*, K. Sénécal, A.J. Clarke, H.G. Yntema, M. Jackson, L. Lovrecic, A. Pitts, K.L.I. Von Gassen, B.M. Knappes, P. Barøy; Leuven, Belgium

C06.3 Legal framework for genomic data sharing in view of the new EU General Data Protection Regulation Mahsa Shabani*, P. Barøy; Leuven, Belgium
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<tr>
<td>19.15</td>
<td>Aarhus</td>
<td><strong>C01.4 Whole genome sequencing yields medically significant secondary variants in ~25% of a paediatric cohort</strong>&lt;br&gt;Participants of Estonian Genome Center: P. Polubothu, T. Elko, A. Metspalu, Neeme Tonisson, Tartu, Estonia.</td>
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<tr>
<td>19.45</td>
<td>Tartu</td>
<td><strong>C01.6 Genetics-first analysis of high-risk variants for breast, ovarian and prostate cancer in participants of Estonian Genome Center</strong>&lt;br&gt;Authors: M. Palovets, L. Leitsalu, A. Reigo, T. Nikopensius, K. Vanikull, M. Kals, P. Padirk, T. Elko, A. Metspalu, Neeme Tönisson, Tartu, Estonia.</td>
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<tr>
<td>18.30</td>
<td>CO3</td>
<td>Best Posters Session</td>
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<tr>
<td>19.10</td>
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<td>Chair: Gunnar Houge, Joris Veltman</td>
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<tr>
<td>P09.005A</td>
<td>snRNU12 mutation presents a new genetic cause of congenital AR-cerebellar ataxia. Minor spliceosome machinery associated inherited genetic diseases M. Elsdorf, N. Chadha, T. Ben Omran, H. Karmel, K. Ibrahim, J. Malek, K. Suhre, M. Ross, Alice K. Abd Al Ameen; Doha, Qatar</td>
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<tr>
<td>P14.002B</td>
<td>Tracing the dark matter: investigating the prevalence of exonic copy number and structural variants in Mendelian disorders Swaroop Aradhya, R. Trusty, J. Paul, M. Kenneemer, E. Gafni, S. Fay, S. Lincoln, R. Nussbaum; San Francisco, United States</td>
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<tr>
<td>C01.006C</td>
<td>Next generation phenotyping in Emanuel and Pallister Killian syndrome using computer-aided facial dysmorphology analysis of 2D photos Thomas Liehr, N. Acquarola, K. Pyle, S. St-Pierre, M. Rinholm, I. Schreyer; Jena, Germany</td>
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<tr>
<td>P20.003C</td>
<td>Which policies work?: Comparative approaches to regulating life insurer use of genetic information from the UK, Canada, and Australia Anya Prince; Chapel Hill, United States</td>
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<tr>
<td>P20.005B</td>
<td>Plasmin 3 regulates bone development and maintenance through the NFκB pathway in osteoclasts Inês Carvalho*, J. Freixo, M. Marques, M. Cardoso, J. Fino, C. Alves, B. Marques, M. Talkowski, H. Correia, C. Morton, D. David; Lisbon, Portugal</td>
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<tr>
<td>P17.001A</td>
<td>Characterization of the expression of the imprinted Kcnk9-gene in specific brain regions and the phenotypic analysis of Kcnk9-knock-out mice Alexis Cooper*, M. Linke, F. Lesage, S. Schweiger, U. Zechner; Mainz, Germany</td>
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<tr>
<td>P09.001A</td>
<td>Mutations in CoA Synthase cause pontocerebellar hypoplasia Tessa van Dijk*, S. Ferdinandussen, J.P.N. Ruiter, I.B. Mathijssen, J.S. Parboosingh, M. Alders, A.M. Innes, E.J. Meijers-Heijboer, F. Bernier, R. Larmont, F. Baas; Amsterdam, Netherlands</td>
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<tr>
<td>P19.01A</td>
<td>Reconciling non-directivity and the counselors’ preference in prenatal counseling Sanne L. van der Steen*, I.M. Bakkeren, R.H.J. Galjaard, M.G. Polak, J.B. Busschbach, S.R. Riedijk, A. Tibben; Rotterdam, Netherlands</td>
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<tr>
<td>P13.02B</td>
<td>Cryptic complexity and disease candidate genes identified in de novo apparently balanced translocations using whole-genome mate-pair sequencing Constantin Aristidou, M.M. Mhroojy, M. Bak, A. Theodosiou, V. Christophidou-Anastasidou, N. Skordis, N. Tommerup, C. Sismani; Nicosia, Cyprus</td>
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<tr>
<td>P01.001A</td>
<td>Copy number variation profile in the placental and parental genomes of recurrent pregnancy loss families Laura Kasak*, K. Rull, S. Söder, M. Luan; Tartu, Estonia</td>
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<td>P16.04D</td>
<td>High-throughput clonal analysis of tumors with droplet microfluidics Dennis Eastburn, M. Pellegrino, S. Treusch, A. Sciambi; South San Francisco, United States</td>
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<tr>
<td>P12.004D</td>
<td>Impact Of Genome Variation Analysis Of CRISPR Screens And Mapping Variant Data To Disease Biology A. Aytemur, Leigh Brody, N. Humphry-Kirkov; London, United Kingdom</td>
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<tr>
<td>P09.006B</td>
<td>TBCD mutations cause autosomal recessive early childhood-onset neurodegenerative encephalopathy Noriko Miyake, T. Chihara, M. Miura, H. Shimizu, A. Kakita, N. Matsumoto; Yokohama, Japan</td>
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The 19 Best Posters were selected for a short presentation (2 minutes) in the lecture hall.

After the presentation of all posters (approximately at 19:10 hrs), the authors and the audience will proceed to the electronic posters in front of the lecture hall for discussion with the authors for the remainder of the session (until 20.00 hrs).
SCIENTIFIC PROGRAMME
Sunday, May 28, 2017
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<tr>
<td>08.30 - 10.00</td>
<td>S01 Single cell studies: From technology to biology Chair: Thierry Voet Henna Tyynismaa</td>
<td>S02 One gene, many phenotypes Chair: Erza Maria Valente Elsebet Ostergaard</td>
<td>S03 Novel Treatment Options Chair: Yanick J. Crow Asbjorg Stray-Pedersen</td>
<td>S04 From Association to Causality in complex diseases Chair: Samuli Ripatti Anders Banglam</td>
<td>E06 Bioethics for dummies Chair: Francesca Forzano</td>
<td>E07 Pharmacogenomics in the clinic Chair: William G. Newman</td>
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<tr>
<td>08.30</td>
<td>S01.1 Single cell RNAseq-based characterisation of adult stem cells Bart Deplancke: Lausanne, Switzerland</td>
<td>S02.1 Filaminopathies Stephen P. Robertson: Dunedin, New Zealand</td>
<td>S03.1 Emerging targeted drug therapies in skeletal dysplasias Ravi Savarirayan: Melbourne, Australia</td>
<td>S04.1 Genetic architecture of coronary artery disease, a common and complex disorder Sekar Kathiresan: Boston, United States</td>
<td>E06.1 Gene editing, NIPT Martina C. Cornell: Amsterdam, Netherlands</td>
<td>E07.1 Pharmacogenomics Knowledge for Personalized Medicine Teri Klein: Stanford, United States</td>
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<tr>
<td>09.00</td>
<td>S01.2 Towards single-cell proteomics: Unraveling cell populations in health and disease by single-cell mass cytometry Bernd Bodenmiller: Zurich, Switzerland</td>
<td>S02.2 Laminopathies Nicolas Levy: Marseille, France</td>
<td>S03.2 Gene therapy of myotubular myopathy Anna Buj-Bello: Genethon, France</td>
<td>S04.2 Efficient fine-mapping of genome-wide association study results Matti Pirinen: Helsinki, Finland</td>
<td>E06.2 Balancing public health &amp; biomedical ethics: The case of newborn screening Yvonne Bombard: Toronto, Canada</td>
<td>E07.2 Implementation of pharmacogenomics in the clinic Munir Firmaheem: Liverpool, United Kingdom</td>
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<tr>
<td>09.15</td>
<td>S01.3 Single-cell genomics Gioele La Manno: Stockholm, Sweden</td>
<td>S02.3 Disruption of Na⁺/K⁺-ATPase by neurological disease mutations and rescue by second-site mutation Bente Vilsen, R. Holm, C.P. Rann, M.S. Toustrup-Jensen, A.P. Einholm, V.R. Schack: Aarhus, Denmark</td>
<td>S03.3 Pronuclear transfer to prevent mitochondrial DNA disease (Mito therapy) Mary Herbert: Newcastle, United Kingdom</td>
<td>S04.3 Integration of eQTL and GWAS to find susceptibility genes for complex traits Bogdan Pasaniuc: Los Angeles, United States</td>
<td>E06.3 Corporate Satellites (see page 46-48 for details)</td>
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<td>10.00 - 10.15</td>
<td>Coffee Break / Poster viewing / Exhibition</td>
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<td>10.15 - 11.15</td>
<td>Poster viewing with presenters and coffee - GROUP A</td>
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<td>11.15 - 12.45</td>
<td>Lunch break / Posters / Exhibition</td>
<td>B-Halls</td>
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C07 Novel genomics technologies
Chair: Johan den Dunnen
Michael Banks

C08 Neuromuscular Diseases
Chair: Marta Bertoli Sabine Gribneng

C09 Molecular Mechanisms of Disease
Chair: Olaud Rednening niel Tomsenup

C10 GWAS: Resolving Missing Causality
Chair: Philippus C. Patalsis Anne Tjibjarg-Hansen

C11 Sensory disorders
Chair: Trine E. Prescott Dragana Vuckovic

C12 Engaging Patients in Genomics
Chair: Helena Kaaisen Birgite R. Dinesh

C07.1 Mosaic mutation detection using single molecule molecular inversion probes (smMIPs) for autoinflammatory disorders
Utrecht, Netherlands

C08.1 Biallelic mutations in the myopallidial gene, MYPN are associated with childhood-onset, slowly progressive nemaobstrinopathy
Koichi Matsuoka, S. Yokohama, Japan

C08.2 Neurocaldelta as a novel protective modifier for spinal muscular atrophy: A full story from gene identification to therapy

C08.3 Dissecting the causal mechanism of X-linked dystonia-parkinsonism by integrating genome and transcriptome assembly

C09.1 X chromosome inactivation in human single cells

C10.1 Systems Genetics and Transcriptome analysis on Circulating Proteins
D.V. Zhernakova, U. Vosa, A. Claringbould, M.J. Bander, A. Kurlishkoy, S. Sanna, B. Alasanovska, K. A. Boer, F. Kuipers, L. Franke, C. Wijmenga, A. Zehnavoraka, Jangyu Fu; Groningen, Netherlands

C11.2 A homozygous variant in mitochondrial RnaP subunit PRORP is associated with Perrault Syndrome characterised by hearing loss and primary ovarian insufficiency

C12.1 SEQUEP: Preferences and representations from patients and parents with regard to the use of Next-Generation Sequencing technologies in medical genetics.

C07.2 Ultra-sensitive detection of mosaic mutations in blood DNA of healthy individuals provides new insights into age-related clonal hematopoiesis
Rocio Acuna Hidalgo, H. Senjgu, M. Steenhouwer, M. van der Vorst, A.J. Veltman, C. Gisslen, A. Haischer, M. Nijmegen, Netherlands

C08.3 Whole genome characterization of array defined clusters of CNVs reveals two distinct complex rearrangement subclasses generated through either non-homologous repair or template switching

C10.2 The deCODE replication server, a resource for the replication of published genome-phenotype associations

C11.3 Rare genetic variants in MEPE cause congenital facial paresis with stapes fixation, and are associated with otosclerosis

C12.2 Children with a rare chromosome disorder. How have UK families’ experiences of diagnosis and counseling changed over the ten year period 2003 – 2013?
Ala Szczepura, S. Wynn, B. Searle, A.J. Khan, T. Palmer, D. Biggersstaff, J. Elliott, M. Hulten; Coventry, United Kingdom

C30.1 Neurodevelopmental malformations are associated with child hood-onset, slowly progressive nemaobstrinopathy
Naomi M. Matsuyama, S. Yokohama, Japan

C30.2 Morbidity risk of chromosomal breakpoints in topological domains enriched in non-exonic conserved elements

C30.3 Quantifying the role of paralogous genes in tissue selective hereditary diseases
R. Barsir, H. Neshem, I. Hekelimen, O. Bashia, H. Sharon, L. Alifandri, L. No- vack, Ester Eger-Lotem; Beer-Sheva, Israel

C30.4 Assessing the causal role of body mass index on cardiovascular health in young adults: a Mendelian randomization and recall-by-phylogeny analysis

C31.2 Rare genetic variants in MEPE cause congenital facial paresis with stapes fixation, and are associated with otosclerosis

C31.3 Rare genetic variants in MEPE cause congenital facial paresis with stapes fixation, and are associated with otosclerosis
Presentations highlighted by an asterisk * and a grey background are from Young Investigator Award Finalists.

**PROGRAMME SUNDAY, MAY 28**

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<td>cont.</td>
<td>C07</td>
<td>Novel genomics technologies</td>
<td>C08</td>
<td>Neuromuscular Disorders</td>
<td>C09</td>
<td>Molecular Mechanisms of Disease</td>
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**13.45**

**C07.4** MAPPING AND PHASING OF STRUCTURAL VARIATION IN PATIENT GENOMES USING NONPORE SEQUENCING


**C08.4** APPLICATION OF EXOME SEQUENCING TECHNOLOGIES TO 1,000 PATIENTS AFFECTED BY LIMB-GIRDLE WEAKNESS OF UNKNOWN ORIGIN

Katherine Johnson*, A. Tafol, M. Bertoli, L. Phillips, A. Blain, E. Ennsri, M. Lek, L. Xu, T. Muller, E. Valkanas, D.G. MacArthur, V. Straub; Newcastle upon Tyne, United Kingdom

**C09.4** BIALLIC MUTATIONS OF PRUNE-1 ARE CAUSING PEHO-LIKE SYNDROME WITH MICROCEPHALIC AND NEURODEVELOPMENTAL IMPAIRMENT


**C10.4** MAPPING-MAPPING ANALYSIS OF 158 BREAST CANCER RISK LOCI FROM ONCOARRAY DATA

Laura Fachal*, J. Allen, M. Ghoussia, J. Beesley, J.S. Carroll, G. Chennova-Trench, J. Simard, P. Kraft, D.F. Easton, A. Dunning; Cambridge, United Kingdom

**C11.4** NATURALLY-OCCURRING EXON-SKIPPING ALLOWS Bypassing complete CEP290 loss-of-function in individuals with unusually mild retinal disease


**14.00**

**C07.5** ENRICHMENT OF UNAMPLIFIED DNA AND LONG-READ SMRT SEQUENCING TO UNLOCK REPEAT EXPANSION DISEASES

Christoph J. Koenig; Y. Tian, D. Greenberg, T.A. Clark; Menlo Park, United States

**C08.5** AUTOSOMAL RECESSIVE MYOPATHY ASSOCIATED WITH CATARACTS CAUSED BY MUTATIONS IN THE GENINE INPP5K, IN INOSITOL PHOSPHATASE

Andreas Roos, M. Wiersner, D. Cox, R. Barreni, D. Hathazi, L. Swan, H. Lochmüller, J. Senderak; Newcastle upon Tyne, United Kingdom

**C09.5** A HUMAN DEVELOPMENTAL SYNDROME CAUSED BY GERMINE MUTATION TO A HISTONE H4 GENE HIGHLIGHTS THE IMPORTANCE OF H4K91 DNA DAMAGE RESPONSE AND CELL CYCLE CONTROL

Federico Tessadori, J. Gallay, J. Hurlst, M. Massink, K. Duran, D. van Gasseren, R. Scott, J. Bakker, G. van Haasen; Utrecht, Netherlands

**14.15**

**C06.6** CLIP-CAP: COMBINED LONG-INSERT PAIRED-END AND CAPTURE SEQUENCING, A NOVEL METHOD FOR THE ANALYSIS OF COMPLEX GENOMIC ABERRATIONS

Carolin Purmann, X. Zhu, D. Paliwog, J. Bernstein, J.F. Hallmayer, A.E. Urban; Palo Alto, United States

**C08.6** HOMOZYGOUS VARIANTS IN LMOD1 AND MYLK CAUSE MACUGIESTIC MICROCONOL INTESTINAL HYPERPERISTALSIS SYNDROME BY DISRUPTION OF SMOOTH MUSCLE CONTRACTILITY


**14.30** - **15.00**

**C09.6** THE CILIOPATHY PROTEIN TAPEID3/ KIAA0586 PLAYS A ROLE UPSTREAM OF RAB8 ACTIVATION IN OUTER SEGMENT FORMATION AND MAINTENANCE IN ZEBRAFISH RETINAL PHOTORECEPTORS

I. Ojeda Naharros, F. Gritzanos, J. Zang, M. Giltay, J. Hurst, M. Massink, J. Beesley, J.S. Carroll, G. Chennova-Trench, J. Simard, P. Kraft, D.F. Easton, A. Dunning; Cambridge, United Kingdom

**C10.6** GENOME-WIDE INFERRED GENOME-WIDE ASSOCIATION STUDIES AS A NOVEL STRATEGY TO IDENTIFY THE RISK LOCUS IN 158 BREAST CANCER PATIENTS

Laura Fachal*, J. Allen, M. Ghoussia, J. Beesley, J.S. Carroll, G. Chennova-Trench, J. Simard, P. Kraft, D.F. Easton, A. Dunning; Cambridge, United Kingdom

**C11** NEW DIAGNOSTIC BIOMARKERS FOR PEROXISOME BIORREGENESIS DISORDERS REVEALED BY UNTARGETED METABOLOMIC PROFILING

Sarah H. Elsaa, L. Hubert, T. Donit, M. Ventum, M. Miller, M. Bravenor, M. Base, W. Rizzo, R. Jones, A. Maser, D. Sun, A. Kennedy, M. Wangler; Houston, United States

**11.26**

**C07** ENGAGING PATIENTS IN GENOMICS

Michelle Bishop, E. Miller, A. McPherson, A. Pope, A. Selle; Birmingham, United Kingdom

**C08** GENOMICS EDUCATION AT SCALE

Michelle Bishop, E. Miller, A. McPherson, A. Pope, A. Selle; Birmingham, United Kingdom

**C09** ENGAGING PATIENTS IN GENOMICS

Michelle Bishop, E. Miller, A. McPherson, A. Pope, A. Selle; Birmingham, United Kingdom

**C10** GENOMICS EDUCATION AT SCALE

Michelle Bishop, E. Miller, A. McPherson, A. Pope, A. Selle; Birmingham, United Kingdom

**C11** GENOMICS EDUCATION AT SCALE

Michelle Bishop, E. Miller, A. McPherson, A. Pope, A. Selle; Birmingham, United Kingdom

**C12** GENOMICS EDUCATION AT SCALE

Michelle Bishop, E. Miller, A. McPherson, A. Pope, A. Selle; Birmingham, United Kingdom

**14.30** - **15.00**

**Fruit break / Poster viewing / Exhibition**
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<td>15.00 - 16.30</td>
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<td>15.00 Welcome and opening remarks Malte Spielmann</td>
<td>15:00 Welcome and short history of syndrome workshops Dian Donnai</td>
<td>Prenatal diagnosis of fetal structural abnormalities in the PAGE project: results and reflections on exome sequencing in 400 trios Jenny Lord</td>
<td>The Welcome Trust Sanger Institute, Wellcome Genome Campus</td>
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<td>Opening comments: Genetics in an isolated population like Finland: a different basis for genomic medicine? Helena Kääriäinen</td>
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<tr>
<td>15:10 The Changing Face of Syndrome Diagnosis Jill Clayton-Smith</td>
<td>Genomic Approaches to Prenatal Diagnostics Michael E. Talkowski</td>
<td>The pharmacogenomics workshop will consider some of the advances in pharmacogenomics that are moving this discipline more into the mainstream. The opportunities afforded by pre-emptive testing, point of care testing and linked electronic health care records. The workshop will be interactive with opportunities for participants to share their own experiences of delivering clinical pharmacogenomics; the barriers encountered and opportunities to overcome these.</td>
<td>Diversity and Inclusion in Genomic Research: Why the uneven progress? Charles Rotimi</td>
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<tr>
<td>15:30 What is the future of a Dysmorphology Clinic: open discussion Sofia Douzgou &amp; Joris Veltman</td>
<td>Microarray as standard analysis for a pregnant population at increased risk for aneuploidy. Results and their implication on a change to NIPF as standard analysis. Ida Vogel</td>
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<td>Increasing the involvement of diverse populations in genomics-based health care – Lessons from Haemoglobinopathies Helen Merridith Robinson</td>
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<tr>
<td>15:45 till end Case presentations</td>
<td>Non-invasive prenatal diagnosis for single-gene disorders Natalie Chandler</td>
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<td>Panel discussion</td>
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<tr>
<td>15:45 Break</td>
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<td>15:50 Assessment of variants Martin Kircher, Dominik Seelow</td>
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<tr>
<td>16.10 Challenges of interpreting non-coding variants Martin Kircher, Dominik Seelow</td>
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<td>16.20 Questions and participant feedback Malte Spielmann, Martin Kircher, Dominik Seelow</td>
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**Coffee break / Poster viewing / Exhibition**

**Poster viewing with presenters and coffee - GROUP B**

**Programme Sunday, May 28**

**ESHG 2017 | Copenhagen, Denmark | www.eshg.org**
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<tr>
<th>Time</th>
<th>Aarhus</th>
<th>Athens</th>
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<tbody>
<tr>
<td>17.45</td>
<td><strong>S05</strong></td>
<td><strong>S06</strong></td>
<td><strong>S07</strong></td>
<td><strong>S08</strong></td>
<td><strong>E08</strong></td>
<td><strong>E09</strong></td>
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<tr>
<td>- 19.15</td>
<td>3D genome architecture: non-coding variants and human disease</td>
<td>Treatment-Focused Genetic Testing in Cancer</td>
<td>Still the golden age of chromosomes</td>
<td>New technologies in Neurogenetics</td>
<td>Multi-omics data integration</td>
<td>Phakomatosis Update</td>
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<tr>
<td></td>
<td>Chair: Malte Spielmann</td>
<td>Chair: Conxi Lazaro Anne-Marie Gerdes</td>
<td>Chair: Erik Iwarsson Karen Brøndum Nielsen</td>
<td>Chair: Yanick J. Crow Zeynep Tümer</td>
<td>Chair: Lude Franke</td>
<td>Chair: Hélène J. Dollfus</td>
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<tr>
<td>17.45</td>
<td><strong>S05.1</strong></td>
<td><strong>S06.1</strong></td>
<td><strong>S07.1</strong></td>
<td><strong>S08.1</strong></td>
<td><strong>E08.1</strong></td>
<td><strong>E09.1</strong></td>
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<td></td>
<td>A 3D Code in the Human Genome</td>
<td>Circulating tumor DNA in cancer monitoring</td>
<td>The molecular pathogenesis of trisomy 21</td>
<td>3D analysis of commissural systems with light sheet microscopy</td>
<td>Functional Genomics</td>
<td>Neurofibromatosis Update</td>
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<td></td>
<td>Erez Lieberman Aiden; Houston, United States</td>
<td>Ellen Heitzer; Graz, Austria</td>
<td>Stylianos E. Antonarakis; Geneva, Switzerland</td>
<td>Alain Chédotal; Paris, France</td>
<td>Phillip Beales; London, United Kingdom</td>
<td>D.R. Gareth Evans; Manchester, United Kingdom</td>
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<tr>
<td>18.15</td>
<td><strong>S05.2</strong></td>
<td><strong>S06.2</strong></td>
<td><strong>S07.2</strong></td>
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<td><strong>E08.2</strong></td>
<td><strong>E09.2</strong></td>
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<td></td>
<td>Long Range regulation of mammalian gene expression</td>
<td>Next-generation sequencing: a change of paradigm in molecular diagnostics of cancer</td>
<td>Mosaic loss of chromosome Y - not that normal benign phenomenon after all</td>
<td>Brain imaging genetics in neurodevelopmental disorders</td>
<td>Methods of integrating genomics data</td>
<td>Tuberous Sclerosis Complex Update</td>
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<td>Doug Higgs; Oxford, United Kingdom</td>
<td>David González de Castro; Belfast, United Kingdom</td>
<td>Lars Forsberg; Uppsala, Sweden</td>
<td>Barbara Franke; Nijmegen, Netherlands</td>
<td>Marylyn Ritchie; Danville, United States</td>
<td>Sergiusz Jozwiak; Warsaw, Poland</td>
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<td>18.30</td>
<td><strong>S05.3</strong></td>
<td><strong>S06.3</strong></td>
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<td></td>
<td>Hox gene regulation in development and disease</td>
<td>Precision cancer medicine: translating laboratory studies into improvements in patient care</td>
<td>Introducing the emerging era of ‘Cytogenomics’</td>
<td>Speaker and title to be announced</td>
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<td>Denis Duboule; Geneva, Switzerland</td>
<td>Gabriel Capella; Barcelona, Spain</td>
<td>Michael Tałkowski; Boston, United States</td>
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See https://2017.eshg.org/index.php/late-changes/ for speaker updates after the printing deadline.
SCIENTIFIC PROGRAMME

Monday, May 29, 2017
Stop by booth 264 to learn more about what ASHG has to offer and enter to win prizes!

www.ashg.org

Join ASHG + ESHG and save!

Read and publish high-quality genetics research

ASHG 2017
ORLANDO · OCTOBER 17-21, 2017
Abstract Deadline June 7, 2017

Stop by booth 264 to learn more about what ASHG has to offer and enter to win prizes!

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<th>E11</th>
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</table>
| 08.30 - 10.00 | **E10**  Whole-genome haplotyping methods for human embryo selection  
Chair: Thierry Voet | **S09** Explaining phenotypic variability  
Chair: Enza Maria Valente Asbjørg Stray-Pedersen | **S10** Population and evolutionary genetics  
Chair: Maria Jesus Sobrido Robert Lyle | **S11** Cancer immunogenetics  
Chair: Jose C. Machado Emma Tham | **S12** Genetics and Microbiome  
Chair: Lude Franke | **E11** Strategies to avoid sudden cardiac death  
Chair: Xavier Jeunemaitre |
| 08.30 | **E10.1** Karyo- and Meio-mapping for human embryo selection  
Dagan Wells: Nuffield, United Kingdom | **S09.1** Karyo- and Meio-mapping for human embryo selection  
Dagan Wells; Nuffield, United Kingdom | **S10.1** Genetic time travel  
Johannes Krause; Jena, Germany | **S11.1** Next-generation immunotherapies for colorectal cancer  
Noel de Miranda; Leiden, Netherlands | **S12.1** Microbiome host-pathogen interactions  
Raminck Xavier; Boston, United States | **E11.1** Sudden Cardiac Death in the Young  
Christopher Sensarnian; Sydney, Australia |
| 09.00 | **E10.2** Haplartihmisis for human embryo selection  
Joris R. Vermeesch: Leuven, Belgium | **S09.2** Genetic and epigenetic regulation of repetitive DNA in relation to disease  
Silvère M. van der Maarel; Leiden, Netherlands | **S10.2** Peopling of the world  
Eske Willerslev; Copenhagen, Denmark | **S11.2** Dissecting tumor-immune cell interactions using genomics tools  
Zlatko Trojanoski; Innsbruck, Austria | **S12.2** Host-microbe interaction  
Jeroen Raes; Leuven, Belgium | **E11.2** Recommendations for the management of sudden cardiac death  
Florence Fellmann; Lausanne, Switzerland |
| 09.30 | **E10.3** Multiple molecular diagnoses underlie some cases of apparent phenotypic expansion  
Jennifer E. Posey, E. Karaca, Z.H. Coban Akdemir, X. Song, T. Harel, S. Jhanganian, Y. Bayram, V. Bahrambeigi, D. Muzy, R.A. Gibbs, J.R. Lupski; Houston, United States | **S09.3** Multiple molecular diagnoses underlie some cases of apparent phenotypic expansion  
Jennifer E. Posey, E. Karaca, Z.H. Coban Akdemir, X. Song, T. Harel, S. Jhanganian, Y. Bayram, V. Bahrambeigi, D. Muzy, R.A. Gibbs, J.R. Lupski; Houston, United States | **S10.3** The origins of farming  
Mark Thomas; London, United Kingdom | **S11.3** Adaptive T cell therapy  
Thomas Blankenstein; Berlin, Germany | **S12.3** Genetics of the microbiome  
Alexandra Zhernakova; Groningen, Netherlands | |
### PROGRAMME MONDAY, MAY 29

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### C13
**Novel diagnostic guidelines for prediction of variant splice-donor or splice-acceptor dency derived from a set of 313 combined in silico and in vitro studies: an international collaborative effort**


### C14
**Admixture mapping identifies Inuux aneuploidy loci associated with metabolic traits in the Greenlandic population**

Victor Yakimov*, L. Skote, A. Koch, B. Saborg, M. Andersson, S.W. Michelsen, M.L. Pedersen, F. Giller, M. Melbye, F. Feuresn. Copenhagen, Denmark

### C15
**Diagnostic value of non-invasive prenatal testing (NIPT) using genomic imprinting profiling (GIPan)**


### C16
**Recurrent de novo missense mutations in small GTPase gene RAB11B cause severe intellectual disability and a distinctive brain phenotype**


### C17
**The optimal cancer risk assessment for hereditary cancers**

PROGRAMME MONDAY, MAY 29

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<td>C13.4</td>
<td>C14.4</td>
<td>C15.4</td>
<td>C16.4</td>
<td>C17.4</td>
<td>C18.4</td>
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<tr>
<td>13.45</td>
<td>Mutation spectrum of NOD2 reveals recessive inheritance as a main driver of Early Onset Crohn’s Disease</td>
<td>Farming in Estonia was introduced by Early Bronze Age migrants from the Steppe</td>
<td>Clinical implementation of non-invasive prenatal diagnosis (NIPD) for single gene disorders</td>
<td>A syndromeic neurodevelopmental disorder is caused by de novo disruption of the pro tease regulatory subunit PSMD12</td>
<td>A somatic mutational signature in different tumor types associated with biallelic germline NTH1L mutations</td>
<td>Genome-wide association study identifies two novel loci associated with female stress and urgency urinary incontinence</td>
</tr>
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</table>

Podocytes differentiated from urine renal precursor as a tool for Alport syndrome diagnosis and for assessing therapeutic strategies based on patient-derived cells

14.00 C13.5 Podocytes differentiated from urine renal precursor as a tool for Alport syndrome diagnosis and for assessing therapeutic strategies based on patient-derived cells

Sergio Daga*, M. Bal-dassarri, C. Lo Rizzo, C. Fallarini, V. Imperatore, I. Longa, E. Fusianni, F. Aria-ni, M.A. Mencarelli, F. Mari, A.M. Pinto, A. Renieri; Siena, Italy

14.15 C13.6 Machine learning models for the characterization of genes associated with adult brain diseases

Juan A. Botla, S. Guell, K. D’as, J. Vandrovcova, J. Hardy, M. Wiele, M. Ryden; London, United Kingdom

14.30 - 15.00 Fruit break / Poster removal / Exhibition

Presentations highlighted by an asterisk (*) and a grey background are from Young Investigator Award finalists.
Programme Monday, May 29

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<tr>
<td>15.00 - 16.30</td>
<td>W12 Big Data in Human Genetics: opportunities and challenges</td>
<td>Organisers: Kristel Van Steen Bertram Muellen-Mynsk</td>
<td>W13 Dysmorphology 2*</td>
<td>W14 Copy Number Variation Interpretation and Classification</td>
<td>W15 Multi-gene hereditary cancer panels: balancing clinical utility and research interest</td>
<td>W16 Ensuring the quality of genetic counseling</td>
<td>W17 Quality assurance in interpretation and reporting in genome wide diagnostics</td>
<td>W18 ENSEMBL GENCODE</td>
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</table>

**Invited speakers / case studies:**
- **Big data in genetic studies:**
  - **Challenging light phenotyping with clinical dissection:** Cathryn Lewis, Genetic Epidemiology and Statistics, King's College London, UK

**Workshops:**
- **The workshops will discuss complex undiagnosed cases with distinctive features and patients with known diagnoses which are particularly educational and demonstrate new clinical information or genomic mechanisms.** Cases for presentations should be brought to the auditorium in the break before the workshop starts. Places for presentation are allocated on a first come, first serve basis. Each presenter is asked to give a concise outline of their case and demonstrate the relevant features in a short (approximately 6 slides) PowerPoint presentation. The discussion of the case will then be facilitated by the workshop chairs with comments invited from experts in the audience. No photographs of slides should be taken in the session.

**Challenges of high-dimensional data for machine learning in genomics:**
- Chloé-Agathe Azencott, Centre for Computational Biology (CIBIO), Mines ParisTech, Institut Curie and Inserm,

**Interactive brainstorming:**
- Take-home messages and future perspectives for human geneticists

**Sessions:**
- "Interactive roundtable discussion" (20 mins)
- "CNVs with the VEP API" (30 mins)
- "居室 genome browser" (20 mins),
- "Ensembl genome browser" (20 mins),
- "Introduction to the ENSEMBL GENCODE API" (30 mins),
- "The aim of this workshop is to focus on various aspects of copy number variant (CNV) interpretation and to discuss differentiating points in a diagnostic setting. We will talk about multi-, intra- and intergenic CNVs detected by genome wide array analysis, but also CNV detection in Whole Exome Sequencing data will be included. We will use illustrative cases from our own diagnostic laboratories to have an interactive discussion on the more challenging findings, including retracted-penetrant, recurrent CNVs and structurally rearranged chromosomal imbalances as well as patients with compound heterozygous variants in a recessive disease gene. This workshop uses the voting system in the ESHG conference app. Go to "Interactive > "All Sessions" > "W14..."

**Poster viewing:**
- With presenters and coffee - GROUP D
| Time  | Aarhus                          | Athens                                      | Alicante                                      | Antwerp                                      | Room 115+116 | Ancona
|-------|---------------------------------|---------------------------------------------|-----------------------------------------------|----------------------------------------------|--------------|--------
| 17.45 | **S13** Next generation clinical genetics<br>Chair: Gunnar Houge, Valtteri Wirta | **S14** Organoid models: The Maxi Impact Of Mini Organs<br>Chair: Brunilde Wirth, Lars A. Larsen | **S15** ESHG / ESC JOINT Symposium: Polygenic Cardiovascular traits<br>Chair: Xavier Jeunemaitre, Michael Christiansen | **E12** The evolution of genetic counseling: Lessons learned from psychotherapy<br>Chair: Sam Riedijk | **E13** Network Medicine<br>Chair: Kristel Van Steen | **S16** Autophagy in health and disease<br>Chair: Brunella Franco, Trine Prescott |
| 17.45 | **S13.1** Integrating the Phenomic and Genomic Architectures of Developmental Disorders<br>David FitzPatrick, Edinburgh, United Kingdom | **S14.1** Dissecting human and chimpanzee cerebral organoids using single-cell RNA-seq<br>J. Camp, S. Kanton, F. Badsha, F. Mara-Bermudez, S. Pålbo, W. Huttner, Barbara Treutlein, Leipzig, Germany | **S15.1** Implications of understanding the genetic basis of coronary artery disease<br>Nilesh J. Samani, Leicester, United Kingdom | **E12.1** The added value of psychotherapy in the genetic counselling process<br>Ramona Moldovan, Cluj-Napoca, Romania | **E13.1** Mining biological networks<br>Nataša Pržulj, London, United Kingdom | **S16.1** Autophagy gets to the bone<br>Carmine Settembre, Naples, Italy |
| 18.15 | **S13.2** The clinical geneticists' perspective on exome sequencing<br>Anita Rauch, Zurich, Switzerland | **S14.2** Common mechanisms between Zika virus-induced and inherited microcephaly in human brain organoids<br>Jay Gopalakrishnan, Cologne, Germany | **S15.2** Genetics of arterial blood pressure: from common to rare variants in the general population<br>Patricia Munroe, London, United Kingdom | **E12.2** Genetics and Family Dynamics: Navigating the Sometimes Bumpy Road to Effective Communication<br>Susan H. McDaniel, Rochester, United States | **E13.2** Cellular Networks and Human diseases<br>Amitabh Sharma, Boston, United States | **S16.2** Autophagy in metabolic processes<br>Romeo Ricc, Illkirch, France |
| 18.30 | **S13.3** Fast-WES for neonates, how useful is it really?<br>Gijs Santen, Leiden, Netherlands | **S14.3** Liver organoids for the study of liver biology and disease<br>Meritxell Huch, Cambridge, United Kingdom | **S15.3** The genetic architecture of type 2 diabetes<br>Philippe Froguel, Lille, France | **E12.3** Autophagy in neurodegeneration and ageing<br>Nektarios Tavernarakis, Crete, Greece |
| 20.00 | Networking Party at the Bksenhallen (at own expense - Ticket required) | | | | | |
SCIENTIFIC PROGRAMME

Tuesday, May 30, 2017
EUROPEAN HUMAN GENETICS CONFERENCE 2018

MiCo | Milan - Italy | June 16 - 19

GENERAL INFORMATION

The European Society of Human Genetics promotes research in basic and applied human and medical genetics and facilitates contact between all persons who share these aims.

This international conference (now in its 51st year) is a forum for all workers in human and medical genetics to review advances and develop research collaborations. The conference has become one of the premier events in the field of human genetics with over 3,000 delegates, more than 250 oral presentations, 18 workshops and 12 educational sessions. The ESHG conference is where the latest developments in human genetics are discussed and where professionals from all parts of human genetics meet.

PROGRAMME OVERVIEW

Invited Plenary lectures and Symposia | ESHG Award and Mendel lectures | Educational Track throughout the meeting | Workshops | Concurrent sessions from submitted abstracts | Poster presentations of submitted abstracts | Young Scientist and Poster Awards | Conference Fellowships for young researchers | Fellowships of Excellence | Fellowships for National Societies | Corporate Satellites | Over 160 exhibitors from all over the world

FURTHER DETAILS

The website 2018.eshg.org will open in October 2017.

Abstract Submission

Online via 2018.eshg.org. Closing date: Friday, February 9, 2018

Scientific & Administrative Conference Secretariat

ESHG 2018 c/o Vienna Medical Academy
Alser Strasse 4, 1090, Vienna, Austria
Tel: +43 1 405 13 83 11
Email: conference@eshg.org

Exhibition, Sponsoring, Corporate Satellites

Rose International
P.O.Box 93260, 2509 AG The Hague, The Netherlands
Tel: +31 70 383 8901
Email: eshg@rose-international.com

INFORMATION FOR INCOMING DELEGATES

CHRISTINE PATCH
President
European Society of Human Genetics

www.eshg.org | @eshgsociety | #eshg2018
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<th>Time</th>
<th>Session</th>
<th>Title</th>
<th>Speaker</th>
<th>Location</th>
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<tbody>
<tr>
<td>09.00</td>
<td>PL3</td>
<td>ESHG-ASHG Building Bridges Debate: Ethical and Legal Discussions - Past, Present &amp; Future</td>
<td>Joris Veltman, Francesca Forzano, Peter Scacheri</td>
<td>Aarhus</td>
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<tr>
<td>09.00</td>
<td>PL3.1</td>
<td>Reflecting on ethics in genetics: The past, present and future</td>
<td>Ruth Chadwick;</td>
<td>Manchester, United Kingdom</td>
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<tr>
<td>09.10</td>
<td>PL3.2</td>
<td>ELSI issues and the implementation of genetics in clinical practice</td>
<td>Mats Hansson;</td>
<td>Uppsala, Sweden</td>
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<tr>
<td>09.20</td>
<td>PL3.3</td>
<td>The Evolution of Genetic Counseling: Effectively Meeting Our Clients' Needs</td>
<td>Barbara B. Biesecker;</td>
<td>Bethesda, United States</td>
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<tr>
<td>09.30</td>
<td>PL3.4</td>
<td>From Medical Genetics to Applied Genomics: Implications for Human Geneticists' Core Goals and Values</td>
<td>Eric Juengst;</td>
<td>Chapel Hill, United States</td>
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<td>09.40</td>
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<td>Interactive discussion with the audience</td>
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<td>Coffee Break</td>
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### PROGRAMME TUESDAY, MAY 30

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<td>Toril Fagerheim</td>
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<td>11.30</td>
<td>C20 Molecular.........</td>
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<td>C21 Cardiovascular...</td>
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<td>Hertz Hans Sheffler</td>
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Presentations highlighted by an asterisk (*) and a grey background are from Young Investigator Award finalists.
Programme Tuesday, May 30

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<tr>
<th>Time</th>
<th>Aarhus</th>
<th>Copenhagen</th>
<th>Cannes</th>
<th>Alicante</th>
<th>Amsterdam</th>
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</table>
| 11.45 | **C19.4 High-resolution variant filtering empowers clinical interpretation and provides insights into variant penetrance and population specificity**


| 12.00 | **C20.4 KIAA1109 variants are associated with a severe disorder of brain development and arthrogryposis**


**C20.4 Copy number variants account for at least 2% of non-syndromic cardiomyopathies**

Frank Honti*, G. Beaman, M. Edwards, T. Monk, S. Wilkinson, L. Brett, S. Cook, J.S. Ware, W.G. Newman, D. Morris-Rosendahl; London, United Kingdom

**C22.4 Adipose cis-eQTL variants at enhancer-promoter interaction circuits regulate obesity genes**


**C23.4 WD40-repeat 47 is essential for brain development via microtubule-mediated processes and autophagy**


**C21.5 A major beneficial effect of angiotensin II receptor blockade for preventing spontaneous aortic rupture in a new mouse model of vascular Ehlers Danlos syndrome**

E. Fontaine, J. Faugeroux, J. Lavelle-Ferreiras, F. Vignal, A. Gianfermi, H. Nematallah, P. Brunevaid, J. Haddouch, E. Messias; Xavier Jeunemaître; Paris, France

**C22.5 Trans-eQTL analysis in 25,000 individuals reveals clear differences between diseases in the types and number of causally involved biological pathways**

Annie Clárignolbo*, U. Viito, eQTLGen Consortium; T. Esko, L. Franke; Groningen, Netherlands

**C23.5 ATPase-deficient ATAD3A alters mitochondrial dynamics in hereditary spastic paraplegia**


**C23.6 Mutation of ribosomal RNA-processing protein 7 homolog A (RRP7A) causes autosomal recessive microcephaly with intellectual disability**


**C21.6 Patterns of co-occurrence of congenital heart defects follows distinct patterns**


**C22.6 Men with LOY and cells without the Y chromosome - transcripts and functional effects studied in 6000 single cells by RNA sequencing using the 10X Chromium platform**

Jonatan Halvorsen, M.D. Fernow, H. Davies, C. Rasi, J.P. Dumanski, L.A. Forsberg; Uppsala, Sweden

**C23.6 Mutation of ribosomal RNA-processing protein 7 homolog A (RRP7A) causes autosomal recessive microcephaly with intellectual disability**


| 12.15 | **C19.5 External Quality Assessment of Clinical Genetics: from pilot assessment to full EQA scheme**


**C20.5 Heterozygous BMP2 mutations leading to haploinsufficiency cause a recognisable human syndrome comprising short stature, palatal anomalies, congenital heart disease and skeletal malformations**


**C21.5 Trans-eQTL analysis in 25,000 individuals reveals clear differences between diseases in the types and number of causally involved biological pathways**

Annie Clárignolbo*, U. Viito, eQTLGen Consortium; T. Esko, L. Franke; Groningen, Netherlands

**C23.5 ATPase-deficient ATAD3A alters mitochondrial dynamics in hereditary spastic paraplegia**


**C23.6 Mutation of ribosomal RNA-processing protein 7 homolog A (RRP7A) causes autosomal recessive microcephaly with intellectual disability**


| 13.30 | Lunch Break

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**ESHG 2017 | COPENHAGEN, DENMARK | WWW.ESHG.ORG**
**PROGRAMME TUESDAY, MAY 30**

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<td>13.30</td>
<td><strong>Plenary Session PL4</strong></td>
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<td>14.15</td>
<td>Chair: Christine Patch, Joris A. Veltman</td>
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<td>13.30</td>
<td><strong>PL4.1</strong></td>
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<td>Reading and Writing Genomes</td>
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<td>George Church:</td>
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<td>Boston, United States</td>
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<td>Introduction by Joris A. Veltman</td>
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<td>14.15</td>
<td><strong>Plenary Session PL5</strong></td>
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<td>ESHG Award and Closing Session</td>
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<td>15.45</td>
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<td><strong>PL5.1</strong></td>
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<td>X-chromosome structure and epigenetic dynamics during X inactivation</td>
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<td>Edith Heard:</td>
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<td>Paris, France</td>
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<td>Laudation by Joris A. Veltman</td>
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<tr>
<td>15.00</td>
<td><strong>Plenary Session PL6</strong></td>
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<td>15.45</td>
<td>Awards Session</td>
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**ESHG Education Award to Ségolène Aymé**
Laudation by Christine Patch

**EJHG-NGP Awards**

**ESHG Young Investigator Awards:**
- **ESHG Young Investigator Awards** for Outstanding Science
- **Isabelle Oberlé Award** for an outstanding presentation in the field of genetics of mental retardation
- **Lodewijk Sandkuijl Award** for an outstanding presentation in the field of complex disease genetics and statistical genetics
- **Vienna Medical Academy Award** for an outstanding presentation in translational genetic research/theraphy of genetic diseases
- **Mia Neri Award** for an outstanding presentation in the field of childhood cancer

**ESHG Poster Awards in clinical research and basic science**

Closing

At the end of the Awards Plenary Session, three Apple iPads mini will be drawn among the attendees, who have had their badges scanned at the entrance of the Plenary Hall before the start of the afternoon plenary sessions.
PROGRAMME INFORMATION

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CORPORATE SATELLITE MEETINGS
BUSINESS MEETINGS
YOUNG INVESTIGATOR AWARD CANDIDATES
POSTER AWARD CANDIDATES
## PROGRAMME SPONSORED SESSION, Saturday, May 27

**Saturday, May 27, 08.15 - 10.15 hrs**

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<td>Sponsored Educational Session E01</td>
<td>Joris A. Veltman</td>
<td>Aarhus</td>
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<td>Sequencing, sponsored by Illumina</td>
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<tr>
<td>08.15</td>
<td><strong>E01.1</strong> The Future of Genomic Medicine</td>
<td>Elaine Mardis, Columbus</td>
<td>United States</td>
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<td>08.45</td>
<td><strong>E01.2</strong> Deep Sequencing of 10,000 Human Genomes</td>
<td>Amalio Telenti, San Diego</td>
<td>United States</td>
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<td>09.15</td>
<td><strong>E01.3</strong> Increasing the diagnostic yield of genome-wide sequencing for rare diseases</td>
<td>Kym M. Boycott, Ottawa</td>
<td>Canada</td>
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<td>09.45</td>
<td><strong>E01.4</strong> Medical genome sequencing in the 100,000 Genomes Project Rare Disease Programme</td>
<td>Richard H. Scott, London</td>
<td>United Kingdom</td>
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| CS24 NimaGen | Belgrade | 53 |
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CS01 – Canon BioMedical, Saturday, May 27, 2017, 12.15–13.45 hrs, Room Birmingham

Tough Targets. Simple Genotyping. — New Solutions for CRISPR, Mitochondrial Disease, Neurology, and Pharmacogenetic Research

Dana Pfister, Product Manager, Canon BioMedical, Rockville, USA

Do you have targets that are difficult to genotype? Are you starting new projects and looking for a fast convenient genotyping solution?

Whether screening CRISPR clones, exploring pharmacogenetic interactions, detecting mitochondrial disease targets, or distinguishing genotypes for neurological disorders, PCR followed by high-resolution melting (HRM) is a fast, reliable, and inexpensive genotyping method to get the answers you need for your experiments.

Canon BioMedical has developed a library of common and unique targets for identifying single nucleotide polymorphisms (SNPs), indels, and large deletions. Unlike most other genotyping techniques, HRM analysis can genotype a single sample in an hour. The Novallele™ assays can be run on any HRM-capable thermocycler.

Attend our session to learn how the Novallele assays can help you with your research.

Novallele assays are for Research Use Only. Not for use in diagnostic procedures.

CS02 – BD Life Sciences, Saturday, May 27, 2017, 12.15–13.45 hrs, Room Belgrade

Chair: Richard Henfrey, Dr, BD Genomics, Oxford, UK

Bringing Innovative Technologies Together: An Integrated Workflow for Single Cell Analysis Uncovering Hidden Events in a Solid Tumour

Part I: Wieland Keilholz, Dr, BD Genomics, Heidelberg, Germany

BD Genomics offers a suite of products that enable broad biological research through a fully integrated workflow. Tissue dissociation combined with FACS analysis based on complex surface marker panels is capable of detecting cellular heterogeneity within a tissue sample. Furthermore, in conjunction with FACS sorting technology and RNA-Seq on the sorted cells, the picture is sharpened by linking proteomic and transcriptomic data. A case study on PDX tumour samples will be presented.

Part II: Claire Gibney, Dr, BD Genomics, Oxford, UK

An overview of BD Genomics technologies will be presented, including FACS sorting of single cells into plates using the BD FACSMelody™, combined with BD™ Precise assays for whole transcriptome/targeted gene expression. The BD™ Resolve platform will be introduced, demonstrating single-cell gene expression analysis for hundreds/tens of thousands of single cells. Finally, the latest advances using the BD CLIC™ automated library preparation instrument for targeted and whole genome sequencing will be covered.

Sunday, May 28, 11.15 - 12.45 hrs


New products to enable the discovery of de novo and germline mutations

Announcing four new products that will advance our understanding of human genetics and disease.

Sanger Sequencing in Molecular Pathology and Genetics
Dr Luca Quagliata, Basel University Hospital, Switzerland

Targeted Next Generation Sequencing in inherited disease research: example from Noonan syndrome
Adam Ameur PhD, Uppsala University, Sweden

Application to rare diseases
Morten Dunø PhD, Rigshospitalet, Copenhagen, Denmark

Advanced carrier screening research: pan-ethnic high throughput mutation and genomic variation detection
Doron Behar MD PhD, Gene by Gene and Igentify, Haifa, Israel

Implementation of BRCA Oncomine panel for germline and somatic variant analysis
Enrico Tagliafico MD, PhD, Center for Genome Research, Department of Medical and Surgical Sciences, University of Modena and Reggio Emilia, Italy

Lunch will be served. Find out more at thermofisher.com/eshg17. Visit booth #438.
CS04 – Agilent Technologies, Sunday, May 28, 2017, 11.15–12.45 hrs, Room Brussels

Alissa: The next evolution of Cartagenia applications & how the Netherlands is leading the way

Dr. Maartje Vogel, Clinical Laboratory Geneticist, Department of Pathology, The Netherlands Cancer Institute, Amsterdam, NL
Development of Molecular diagnostic techniques at the Netherlands Cancer Institute

We will discuss how we are investigating the use of Next Generation Sequencing for diagnostic use at the Netherlands Cancer Institute. We will describe how we are carrying out test validation, workflow automation, report standardization and data integration for clinical decision making and translational research.

Dr. Kristin Abbott, PhD, Molecular Geneticist, NGS analysis pipeline coordinator, Department of Genetics, University Medical Center, Groningen, NL
Data sharing- How to make the most of your data through collaborations

Most will agree that data-sharing is essential to a successful diagnostic service. The question is how to implement such an initiative. In the Netherlands we have created a national (VKGL) working group to coordinate, with the help of Cartagenia, a concerted data-sharing effort. Here, I will show how the Dutch data-sharing initiative works within Cartagenia, and how the shared data can be used to improve overall NGS analysis.

CS05 – QIAGEN, Sunday, May 28, 2017, 11.15–12.45 hrs, Room Berlin

Transforming your biological samples into actionable insights

Using seamlessly integrated preanalytical, next-generation sequencing and bioinformatics solutions, and leveraging expertise in translational and clinical research to refine our understanding of human genetics and diseases.

Chairs: Anja Wild and Phoebe Loh, QIAGEN, Hilden, Germany

11.15–11:20 Welcome

11:20–11:50 Molecular analysis of thyroid nodules – detection of gene mutations and fusion genes by DNA/RNA sequencing
Dr. Egbert Schulze, Molecular Genetics Laboratory, Heidelberg, Germany

11:50–12:15 Circulating Cell Free DNA Pre-Analytics: Importance of ccfDNA Stabilization and Extraction for Liquid Biopsy Applications
Dr. Dominic O’Neil, QIAGEN, Hilden, Germany

12:15–12:45 Integrative approach to biomarker discovery by performing comparative analysis of two cancers, using genetics and transcriptomics from RNA sequencing data
Jean-Noel Billaud, QIAGEN Bioinformatics, Redwood City, USA

12:40–12:45 Closing

CS06 – NIPD Genetics, Sunday, May 28, 2017, 11.15–12.45 hrs, Room Birmingham

New avenues in non-invasive prenatal testing

NIPD Genetics has developed a novel targeted non-invasive prenatal test (NIPT) that allows the non-invasive detection of numerous genetic syndromes and single gene diseases by analyzing fetal DNA in maternal circulation with unparalleled accuracy. This technology is currently marketed as the VERACITY new generation non-invasive prenatal test.

Our novel NIPT methodology captures and counts cfDNA fragments from selected genomic regions at very high read-depths using targeted capture enrichment technology and proprietary analytical methods and bioinformatics. VERACITY considers the complexities of the human genome, and is designed to avoid problematic genomic regions that reduce test sensitivity and specificity. VERACITY has demonstrated the ability to detect fetal aneuploidies, deletions/duplications and point mutations with unparalleled accuracy.

Speakers: Prof. Philipppos Patsalis, Distinguished Professor, The Cyprus Institute of Neurology and Genetics, Founder and CEO, NIPD Genetics, Nicosia, Cyprus
Dr. George Koumbaris, Chief Scientific Officer, NIPD Genetics, Nicosia, Cyprus
Dr. Jurate Kasnauskiené, Associate Professor, Vilnius University, Vilnius, Lithuania
PROGRAMME CORPORATE SATELLITES

Sunday, May 28, 11.15 - 12.45 hrs

CS07 – Face2Gene, Sunday, May 28, 2017, 11.15–12.45 hrs, Room Belgrade

Face2Gene Research: Accelerating Clinical Genomic Discoveries

Facial Analysis & Phenotyping Technology Lead to Rare Disease Discoveries

Speakers: Prof. Peter Krawitz, Institut für Medizinische Genetik und Humangenetik – Charité, Berlin, Germany
Nicole Fleischer, Director of Research Collaboration, FDNA, Boston, United States

Sunday, May 28, 15.00 - 16.30 hrs

CS08 – Roche Sequencing Solutions – Sunday, May 28, 2017, 15.00–16.30 hrs, Room Ballerup

Real world experience with cell-free DNA testing - clinical and laboratory perspectives on Non-invasive prenatal testing (NIPT)

Join us to hear top experts in cell-free DNA testing present the latest data and describe real world experience with NIPT in their laboratory. During this 90 minute symposium, you will see an updated meta-analysis of clinical data for aneuploidy screening using cell-free DNA testing, learn about different technological approaches to NIPT, and hear about a laboratory’s experience in acquiring and implementing a microarray based cell-free DNA technology.

Chair: Maximilian Schmid, MD, Roche Sequencing Solutions, San Jose, USA
Speakers: Maria Del Mar Gil, MD, PhD, Hospital Universitario de Torrejón, Madrid, Spain
Junaid Shabbeer, PhD, FACMG, Roche Sequencing Solutions, San Jose, USA
Francesca Grati, PhD, ErCLG, Toma Advanced Biomedical Assays S.p.A., Busto Arsizio, Italy

CS09 – Sophia Genetics, Sunday, May 28, 2017, 15.00–16.30 hrs, Room Brussels

Dr. Zhenyu Xu, Chief Technology Officer, Sophia Genetics, Switzerland

NGS-based Diagnostics: Challenges in Germline and Somatic Variants Identification

Reliable variants identification using Next-Generation Sequencing (NGS) is challenging and complex in routine genetic diagnostics. Sufficient coverage of target region is a well-known prerequisite of accurate variant detection. However, there are other issues to overcome for ensuring correct variant identification. For example, variants exposed to the end of reads, proper trimming of the primer sequences, special care of repetitive regions, etc. In oncology, we need also to consider the quality of the FFPE samples, the limited starting material and the mixture of somatic and germline cells, that complicate even further the variant detection. Here I introduce the efforts that we made to tackle those different problems, which can influence the performance of different gene enrichment assay used in oncology. The experimental design principles and the general power of limit detection for a somatic NGS assay will be discussed. Moreover, I will present the analytical performance of two capture-based library preparation kits, covering the most common mutated genes involved in Myeloid and Hereditary Cancers. Examples of the routine usage of these tests in a clinical setting will be also provided.

CS10 – Bluebee & Lexogen, Sunday, May 28, 2017, 15.00–16.30 hrs, Room Berlin

Complete and cost-efficient solution for differential gene expression analysis with RNA-Seq: from library prep to automated data analysis results

QuantSeq 3’ mRNA-Seq has already been recognized as one of the fastest, most cost-efficient, and accurate methods for gene expression profiling using next generation sequencing. The data analysis is based on counting of the transcripts, which is straightforward and fast in comparison to standard RNA-Seq.

The QuantSeq data analysis pipeline has been automated and is available on the highly secure cloud-based Bluebee genomics analysis platform free of charge for any QuantSeq customer. Data evaluation - quality control, mapping, and gene read counting - can be performed by any user, also those without experience in bioinformatics. A differential gene expression analysis workflow has been implemented, adding a valuable tool to complete the analysis pipeline, which is accurate, fast, affordable, and bioinformatically not challenging.

In this talk, QuantSeq and the Bluebee platform will be introduced and the end-to-end solution will be showcased. The user will share experience with the pipeline: costs, time savings, and scientific conclusions based on accurate data analysis will be discussed.

Chairs and speakers: Markus Dueringer, Director of Sales, Bluebee, Rijswijk, the Netherlands
Brigit Steinmetz, Product Manager, Lexogen, Vienna, Austria
CS11 – Integrated DNA Technologies, Sunday, May 28, 2017, 15.00–16.30 hrs, Room Birmingham

### High performance NGS target enrichment and CRISPR genome editing solutions

The world leader in custom nucleic acid synthesis, Integrated DNA Technologies (IDT) offers a growing portfolio of genomics products for use in research and clinical applications. At this workshop, we will share our latest developments in next-generation sequencing target enrichment and CRISPR genome editing.

One presentation, **High performance NGS solutions for research and the clinic**, provides insight into how xGen® Exome, predesigned panels, gene capture probes, and innovative universal blockers and adapters have made targeted sequencing truly efficient and informative. We’ll present independent studies comparing multiple commercially available systems for various applications and disease areas.

Another presentation, **Precision genome editing using Cas9 and Cpf1 ribonucleoprotein (RNP) complexes**, demonstrates that CRISPR nucleases plus guide RNAs are the preferred tools for genome editing due to their simplicity and efficiency. Learn how IDT scientists optimized CRISPR RNAs by testing different lengths and modifications, increasing overall genome editing potency.

This workshop will be hosted by IDT’s Chief Scientific Officer, **Mark Behlke (US), MD, PhD**, with presentations from **Dr. Behlke himself**, **Mirna Jarosz (US), PhD, Director of NGS Scientific Applications at IDT**, and **Andrea Gärtner (Switzerland), PhD, Application Scientist from Sophia Genetics**.

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CS12 – Multiplicom, Sunday, May 28, 2017, 15.00–16.30 hrs, Room Belgrade

### Making the difference in genetic diagnostics: Multiplicom solutions

Multiplicom, part of Agilent Technologies, provides your lab with a broad range of Next-Generation Sequencing solutions, which guide you towards the most reliable results. During our Symposium we will be talking about how we make a difference in genetic diagnostics. One of our key speakers is Orlando Diez from Vall D’Hebron University Hospital. He will explain the diverse spectrum of causal mutations in hereditary cancer families and show some remarkable results. Furthermore, Katrien De Mulder from the Sint-Lucas will elaborate on how their lab has implemented NIPT via the Clarigo solutions in their workflow. Last but not least we will be talking about how our new Reporter software ensures the correct analysis and enables quality control from sample to results. Come to our booth no. 662 for a demonstration of our new software and discover how we guide your lab towards better results. Every step of the way.

**Topics:**
- **BRCA MASTR Dx and MASTR Reporter: a tester experience**
  - Loubna Nachate, Technicienne laboratoire biologie moleculaire, Institut Jean-Godinot, Reims, France
- **Diverse spectrum of causal mutations in hereditary cancer families**
  - Dr. Orland Diez – Val d’Hebron, Institute of Oncology, Barcelona, Spain
- **Clarigo: a decentralized NIPT solution**
  - Dr. Katrien De Mulder – General hospital Sint-Lucas, Ghent, Belgium

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CS13 – Illumina, Sunday, May 28, 2017, 15.00–20.45 hrs

### Implementing whole-genome sequencing in clinical routine for diagnostics of rare inherited diseases

- **Valtteri Wirta, Ph.D, Head of Clinical Genomics Facility at Science for Life Laboratory, Stockholm, Sweden**

### The Human Functional Genomics Project: towards understanding human immune function

- **Prof. Cisca Wijmenga, Professor of Human Genetics at the Department of Genetics, University Medical Center Groningen, the Netherlands**

Complimentary wine and cheese will be served.
**Sunday, May 28, 19.15 - 20.45 hrs**

**CS14 – Congenica, Sunday, May 28, 2017, 19.15–20.45 hrs, Room Birmingham**

**Analysing Genetic Inherited Disorders: Lessons from Leading Women’s and Children’s Hospitals**

In this Corporate Satellite Seminar, you will hear about the practical advances in genome and exome interpretation from Birmingham Women’s and Children’s Hospital and Great Ormond Street Hospital, London; two international centres of excellence in healthcare.

Sapientia is a clinical genome analysis and interpretation platform used to diagnose rare inherited diseases. It is used to support diagnosis at major Children’s Hospitals as well as large-scale national studies such as the UK 100K Genomes Project.

What you will learn:

- Lessons learnt when using Sapientia to analyse prenatal clinical genomes and exomes
- Practical examples from Great Ormond Street Hospital London and Birmingham Women’s and Children’s Hospital
- Sapientia case study: providing clinical interpretation services for the UK 100K Genomes Project

Speakers include:

- Dr Dominic McMullan, Consultant Clinical Scientist, WMRGL Birmingham Women’s and Children’s Hospital NHS Foundation Trust, Birmingham, UK
- Natalie Chandler, Senior Clinical Scientist, Great Ormond Street Hospital for Children NHS Foundation Trust, London, UK
- Laura Reed, Pre-Registered Clinical Scientist, Congenica, Cambridge, UK
- Gavin Stone, VP Marketing at Edico Genome, La Jolla, USA
  
Gavin designed DRAGENTM Bio-IT Processor, the world’s first next-generation sequencing bioinformatics chip.


**Find the most suitable genomic data repository for your needs**

*Manuel Corpas, Scientific Lead at Repositive, Cambridge, United Kingdom*

Researchers rely on acquiring external data to validate, benchmark and supplement research findings. Funders require researchers to make their datasets accessible for further reuse. The goal of this workshop is to make researchers in genetics/genomics aware of the existing challenges with genomic data access and reuse, and to present a number of tools and resources that researchers can use directly for simplifying their data access workflows.

Manuel Corpas, Scientific Lead at Repositive, will run this workshop. Manuel has done pioneering work in exploring his personal genome, through direct to consumer genomic testing and online international collaboration. He crowdfunded the DNA sequencing for both himself and his family, and he was the first to publish the complete collection of genomic data for his family online as Open Access. Manuel was previously Project Leader for plant and animal genomes at TGAC (now Earlham Institute), and his earlier roles included Sanger, EBI (European Bioinformatics Institute) and the Spanish National Bioinformatics Centre. Alongside his role at TGAC/Earlham, Manuel was also the ELIXIR-UK Technical Coordinator and board director of the International Society for Computational Biology (ISCB).

**Monday, May 29, 11.15 - 12.45 hrs**

**CS16 – Agilent Technologies, Monday, May 29, 2017, 11.15–12.45 hrs, Room Ballerup**

**Exomes and arrays: why they are here to stay!**

*Dr. Jill Urquhart, Manchester Centre for Genomic Medicine, Central Manchester University Hospitals NHS Foundation Trust, Saint Mary’s Hospital, UK*

**The UKExome**

Commercial exome panels often fail to meet the individual requirements of researchers or countries. Here we describe why we pursued a UK specific exome and the possibilities it offers for clinical disease research.

*Dr. Maria Lascone, ASST Ospedale Papa Giovanni XXIII Bergamo, Italy*

**Exome Sequencing in routine Clinical research of rare diseases**

NGS set in the last years a new frontier in molecular characterization of rare diseases. Exome sequencing coupled with efficient and modern tools for data analysis and interpretation represents today the ideal link between clinical cases reports.

*Dr. Gaelle Pierron, Coordinator of the Somatic Genetics Unit, Marie Curie Institute, Paris, France*

Dr. Gaelle Pierron will describe her work with Agilent’s comparative genomics hybridization (CGH)s arrays.
PROGRAMME CORPORATE SATELLITES

Monday, May 29, 11.15 - 12.45 hrs

CS17 – Centogene, Monday, May 29, 2017, 11.15–12.45 hrs, Room Brussels

Start the proper interpretation of your genetic data - now!

The importance of unbiased genetic and clinical data compilations
Prof. Arndt Rolfs, CEO, CENTOGENE AG, Rostock, Germany

Detailed phenotyping in neurogenetic diseases as a key for NGS data interpretation in diagnostics
Prof. Katja Lohmann, University of Lübeck, Lübeck, Germany

Diagnostics need highly qualified mutation databases - what about CentoMD®
Dr. Gabriela Oprea, Director Digital Data Products, CENTOGENE AG, Rostock, Germany

Next generation interpretation of big data
Prof. Peter Bauer, COO, Centogene AG, Germany

About CENTOGENE
As one of the most diversified and largest genetic testing companies worldwide we are dedicated to transforming the science of genetic information into solutions and hope for patients and their families. CENTOGENE’s goal is the rapid medical diagnosis of inherited diseases, provided at the earliest possible moment as we turn analytical information into actionable results for physicians, patients and pharmaceutical partners. Our commitment to the global medical community is an early and precise diagnosis for continuous improvement of therapeutic options for each individual patient.

CS18 – Asuragen, Monday, May 29, 2017, 11.15–12.45 hrs, Room Berlin

Fragile X Screening: Two Perspectives

Although accurate and high-throughput fragile X carrier screening tests are available, they are under-utilized by the general public due to lack of awareness. As a mother of a child who has fragile X syndrome, Ilana Garber will share her insights of why she believes women should have the opportunity to have carrier screening performed, even if there is ‘no family history’. The workshop will also discuss responsible clinical implementation of carrier screening across populations: the risks, benefits, and limitations.

Presented by: Ilana Garber, West Hartford, CT, USA

CS19 – Sistemas Genómicos – Monday, May 29, 2017, 11.15–12.45 hrs, Room Birmingham

New diagnostic tools: From embryo to adult

Chairman: Javier Benitez PhD, Head Human Cancer Genetics Programme, Spanish National Cancer Centre (CNIO), Madrid, Spain

New advances in genomics enable the study of the genetic bases of many diseases or the individualized response to specific treatments. This analysis currently starts along the embryo stage and continues up to adult life.

In this session we will have the opportunity to learn from three scientists who work in three important biomedical areas.

a) Human embryos trying to improve the detection of aneuploidies in single blastomeres.

b) New exome developments that permit simplifying the diagnostic processes.

c) Different approaches in pharmacogenetics and pharmacogenomics in order to speed the results and cover different options and situations in the clinical practice.

Talk 1: Whole genome sequencing in single human embryo cells for aneuploidy detection
Xavier Vendrell Montón, PhD, Head of the Reproductive Genetics Unit at Sistemas Genómicos, Valencia, Spain

Talk 2: Exome screening for genetic diseases
Juan Carlos Triviño, PhD, Head of bioinformatics department of Sistemas Genómicos, Valencia, Spain

Talk 3: Looking for the correct technology in precision medicine
Diana Valero Hervás, PhD, Pharmacist specialized in Immunology at Sistemas Genómicos Laboratory of Molecular Diagnostics, Valencia, Spain
Monday, May 29, 11.15 - 12.45 hrs


**From discovery to understanding: new CRISPR technologies that enable your functional genetic research**

This session will inform genetics researchers involved in gene target identification and functional analysis studies on the state of the art of screening technologies.

Join us during lunch to hear about new product announcements that will advance our understanding of human genetics and disease.

**Discovery: automated high throughput solutions for DNA and RNA isolation for demanding applications**

We present new solutions for maximising genomic analysis through sequential recovery of DNA and RNA from the same FFPE samples including genetic testing of tumour-derived cell-free DNA with a high-throughput processing platform.

**Validation: using Sanger sequencing to facilitate genome editing workflows**

We demonstrate the use of Sanger sequencing to: determine the efficiency of genome editing in transformed cultures, confirm successful edits in transformed cultures, including screening secondary clones for successful editing and determine the frequency of SNP changes in clones isolated from secondary cultures.

**Understanding: New CRISPR libraries and new capabilities with award-winning solutions**

Hear about how the new award-winning Invitrogen™ LentiArray CRISPR Libraries expand the application of CRISPR-Cas9 technology into high throughput applications for functional genomics screening.

Lunch will be served.

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Monday, May 29, 15.00 - 16.30 hrs

CS21 – NanoString Technologies, Monday, May 29, 2017, 15.00–16.30 hrs, Room Brussels

**More insights for less sample with 3D Biology™ Technology: Multiplex, multi-analyte digital detection of mRNA, DNA, Fusion Genes, and Proteins**

**Digital Spatial Profiling of FFPE tissue using 3D Biology™ Technology**

Niroshan Ramachandran, Ph.D., Senior Director Product Management, NanoString Technologies, Seattle, USA

**Simultaneous detection of Single Nucleotide Variants and Gene Fusions from a single NSCLC patient sample**

Dr. Noemi Reguart, Hospital Clinic Barcelona, Spain

**Immune Profiling of Solid Tumors using NanoString’s new efficient high throughput PlexSet™ Reagents**

Dr. Maggie Cheang, The Institute of Cancer Research, Clinical Trials and Statistics Unit, Belmont, Surrey, UK

**Multiplexed gene fusion detection in non-small cell lung cancer using NanoString technology**

Prof. Johan Staaf, Oncology and Pathology, Lund University, Sweden

Coffee and cakes will be provided.


CS22 – 10x Genomics, Monday, May 29, 2017, 15.00–16.30 hrs, Room Berlin

**Your Sequencer - Our Solutions - Powerful Discovery**

10x Genomics meets the critical need for long range, structural and cellular information, with an innovative system that transforms short-read sequencing technologies. Our Chromium™System supports comprehensive genomics and high-throughput single cell transcriptomics. It enables researchers to discover previously inaccessible genomic information at unprecedented scale, including phased structural variants, phased single nucleotide variants, and dynamic gene expression of individual cells—while leveraging their existing sequencing systems and workflows.

So join our Corporate Satellite to learn more about our technology, from sample preparation through to downstream analysis, and discover how 10x Genomics can help to power your discoveries.

**The Chromium System: Technology & Applications**

Jill Herschleb, Staff Scientist, 10x Genomics, Pleasanton, USA

**Closing the Loop: Informatic Solutions**

Paul Ryvkin, Senior Scientist, 10x Genomics, Pleasanton, USA

**Identifying the downstream consequences of genetic risk factors through single-cell eQTL analysis**

Prof. Dr. Lude Franke, Department of Genetics, University Medical Centre Groningen, the Netherlands
CS23 – Agilent Technologies, Monday, May 29, 2017, 15.00–16.30 hrs, Room Birmingham

**Gene Editing - New Frontiers, new discoveries, new possibilities**

*Dr. Michael Bassik, Ph.D, Assistant Professor, Department of Genetics, Stanford University, USA*

Parallel shRNA and CRISPR/Cas9 Screens Reveal Biology of Stress Pathways and Identify Novel Drug Targets

*Dr. Ana Banito, Research fellow at the Scott Lowe Laboratory, Cancer Biology & Genetics Program, Memorial Sloan-Kettering Cancer Center (MSKCC), New York, USA*

**Defining epigenetic dependencies in synovial**

Synovial sarcomas, driven by a key oncogenic event in unidentified progenitor cells, reveal how global epigenetic changes underlie oncogenic programs; however, the targets of SS18-SSX and the mechanism of its recruitment to chromatin are still unknown. By performing an shRNA depletion screen, we identify an epigenetic dependencies and a new target for synovial sarcoma treatment. We outline how our research approaches could serve as a blueprint for studying dependencies of numerous translocation-driven tumors with yet unknown targets, and unclear molecular function.

CS24 – NimaGen, Monday, May 29, 2017, 15.00–16.30 hrs, Room Belgrade

**NGS Sequencing using smMIP captures: Status Quo 2017**

Chair and introduction: *Joop Theelen, B.Sc., NimaGen, Nijmegen, the Netherlands*

15:00 **Accurate and fast targeted re-sequencing by MIPs - research applications**

*Alexander Hoischen, PhD, Radboudumc, Nijmegen, the Netherlands*

An overview how MIPs evolved, have been used for candidate gene sequencing and novel applications based on smMIPs for low level mosaic mutations.

15:20 **smMIP sequencing in a diagnostic setting: BRCA as a model**

*Arjen Mensenkamp, PhD, Radboudumc, Nijmegen, the Netherlands*

Implementation of smMIPs, from a research tool to a diagnostic setting, with BRCA genes as a model.

15:40 **SeqNext - customised solutions for smMIP sequencing analysis**

*Peter Kirchmeier, PhD, JSI medical systems, Ettenheim, Germany*

Analysis of smMIP generated data with SeqNext to detect FFPE artefacts, CNVs and somatic mutations. Subsequent variant interpretation with varSEAK, the new variant database for Shared Experience And Knowledge.

16:10 **EasySeq NGS Targeted Resequencing kits: smMIP based sequencing of BRCA genes and Oncology Hotspots**

*Joop Theelen B.Sc., NimaGen, Nijmegen, the Netherlands*

NimaGen launched a range of smMIP based NGS target enrichment kits, with a number of unique features, consisting of only 5 steps with an extremely low hands-on-time.

CS25 – Fabric Genomics, Monday, May 29, 2017, 15.00–16.30 hrs, Room Barcelona

**Accurate and Rapid Clinical Whole-Genome Sequence interpretation with Fabric Genomics’ clinical platform**

*Vanisha Mistry, Ph.D., Fabric Genomics, London, UK*

For every clinical lab that wants to develop a comprehensive, high-throughput genomic testing program, Fabric Genomics enables you to deliver the highest quality clinical interpretation of patient genetic information at scale. Clinical NGS testing is expanding from panels to exomes to whole genomes.

We will discuss the key interpretation and reporting capabilities needed to launch and scale clinical NGS testing, including the need for advanced computational algorithms to maximize diagnostic yield. We will highlight examples of genomic testing transforming medical care, such as the 100,000 Genomes Project and Rady Children’s Institute for Genomic Medicine, which has a goal of rapid genome analysis with a 24 hour turnaround time from blood sample to result. Rady uses Fabric Genomics STAT, which delivers comprehensive annotations on whole genome data in less than an hour.

Fabric Genomics is setting the standard for how health systems are using whole genomes to increase diagnosis rates and save lives.
**PROGRAMME BUSINESS AND ANCILLARY MEETINGS**

As per date of printing.

### Friday, May 26, 2017

<table>
<thead>
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<tr>
<td>09.00 – 13.00 hrs</td>
<td>ESHG Executive Board Meeting</td>
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<tr>
<td>13.30 – 18.00 hrs</td>
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### Saturday, May 27, 2017

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<td>08.00 – 09.00 hrs</td>
<td>UEMS and EBMG Clinical Genetics Boards Meeting</td>
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<tr>
<td>09.00 – 11.00 hrs</td>
<td>UEMS Section Meeting</td>
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<td>09.00 – 13.00 hrs</td>
<td>HSCR Consortium Meeting II</td>
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<tr>
<td>09.00 – 13.30 hrs</td>
<td>ESHG Quality Subcommittee Meeting</td>
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<tr>
<td>10.45 – 13.30 hrs</td>
<td>ESHG PPPC Meeting</td>
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<tr>
<td>12.15 – 13.45 hrs</td>
<td>ESHG-SpringerNature Meeting</td>
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<td>13.30 – 20.00 hrs</td>
<td>GENTURIS Meeting (Part II)</td>
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### Sunday, May 28, 2017

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<tr>
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<td>COST Action CHIP ME WG1 (Part I)</td>
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<tr>
<td>10.00 – 12.00 hrs</td>
<td>European Genetic Nurses and Counsellors (GNGC) Meeting</td>
<td>Room 20</td>
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<td>11.00 – 13.00 hrs</td>
<td>EBMG Clinical Laboratory Geneticists (CLG) Meeting</td>
<td>Room 17</td>
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<td>11.00 – 13.00 hrs</td>
<td>National Human Genetics Societies Meeting</td>
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<td>GenIDAda Advisory Board Meeting</td>
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<td>13.00 – 20.00 hrs</td>
<td>GENTURIS Meeting (Part II)</td>
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<td>14.30 – 16.00 hrs</td>
<td>eRare EuroMicro Consortium Meeting</td>
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<td>16.30 – 18.00 hrs</td>
<td>Building Bridges ESHG/ASHG Meeting</td>
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<td>16.30 – 18.00 hrs</td>
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### Monday, May 29, 2017

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<td>ESHG Education Committee Meeting</td>
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<td>10.00 – 12.00 hrs</td>
<td>European Board of Medical Genetics General Assembly</td>
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<td>11.00 – 13.00 hrs</td>
<td>CEQAS, EMON and UK NEQAS for Molecular Genetics</td>
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<td>11.45 – 12.45 hrs</td>
<td>ESHG Board Meeting II</td>
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<td>13.15 – 14.45 hrs</td>
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<td>13.15 – 15.00 hrs</td>
<td>FP7 II consortium BBMRI-LPC – Steering Group Meeting</td>
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<td>European Journal of Medical Genetics (EJM) Board Meeting</td>
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<td>15.00 – 16.30 hrs</td>
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<td>16.30 – 18.00 hrs</td>
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### Tuesday, May 30, 2017

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<td>12.30 – 16.00 hrs</td>
<td>GG2020 – Part II</td>
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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.
**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 ESHG Award Lecture is held by Edith Heard on Tuesday, May 30, 2017 at 14.15 hrs in Hall Aarhus. An interview can be found on the next page.

**Award Holders**

<table>
<thead>
<tr>
<th>Year</th>
<th>Name</th>
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<tr>
<td>2017</td>
<td>Edith Heard</td>
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<td>Stefan Mundlos</td>
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<td>Pierre Maroteaux</td>
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<td>1992</td>
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**Mendel Lecturers**

Since 2006 the European Human Genetics Conference closes with the lecture of a distinguished speaker. In 2009 this lecture was officially named “Mendel Lecture”.

The Mendel Lecture is held by George Church on Tuesday, May 30, 2017 at 13.30 hrs in Hall Aarhus.

**Mendel Lecturers**

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<thead>
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<tr>
<td>2017</td>
<td>George Church</td>
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<td>Sir Adrian Bird</td>
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<td>Thomas Südhof</td>
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<td>Mario Capecchi</td>
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<td>Mary Claire King</td>
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<td>Sir John Burn</td>
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<td>Leroy Hood</td>
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<td>2007</td>
<td>Aaron J. Ciechanover</td>
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<td>2006</td>
<td>Sydney Brenner</td>
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*The Mendel Prize was designed by Swedish geneticist Alicia Bergsten*
Edith Heard

Edith Heard is the Head of Genetics and Developmental Biology Unit, Institut Curie and Professor at the Collège de France. She will be giving the ESHG Award lecture on Tuesday, May 30, 2017 at 14:15 hrs. She talked to Mary Rice about her life and work.

Born into a bilingual Greek/English family, and now working in Paris, Edith Heard says that she learned to be adaptable at an early age. "I was brought up in central London amidst lots of heated Greek political discussions with members of my mother’s family who were staying with us in exile – it was the era of the Greek colonels – while my father was quietly engineering in his garage downstairs".

"So I would often hide and read a book. This meant that I learned to concentrate wherever I was, and also to think about two different topics in two different languages more or less simultaneously," she says.

Heard’s father was an electrical engineer. "For him science was physics or engineering; he didn’t consider biology to be real science. My mother inspired me to care a lot about how things work, and also about people." Her interest in biology didn’t begin until she was at Cambridge, having previously been more attracted to mathematics. "I realised that I was fascinated by all the unknowns in biology, and the buzz that was there in the mid 1980s when molecular biology, genetics and developmental biology were exploding was very exciting. I was lucky enough to come across many great and inspiring scientists in Cambridge."

As a post doc, she started working in the field of X-chromosome inactivation. "I did not realise at the time how lucky I was to be working on such a beautiful biological problem that opened up so many questions and fields. Working on X inactivation means that one can work on development, gene regulation, chromatin, non-coding RNAs, and chromosome biology. It has meant that my team and I have explored many different disciplines and it keeps us curious and happy!"

Among the discoveries of which Heard is proudest is the insight obtained by looking at early mouse embryos, where the team uncovered the highly dynamic process of X inactivation, with a wave of silencing, followed by reactivation and then silencing again. "This was unexpected, and an unexpected example of in vivo reprogramming."

Another high point was the discovery of topologically associated domains, or TADs, in collaboration with Job Dekker. "Elephege Nora, a PhD student in our lab, used chromosome conformation capture to explore the X-inaction centre, and we stumbled across these sub-megabase scale domains of chromosome interaction. This totally changed our way of thinking about the locus we were interested in, and also had many repercussions in terms of our understanding of chromosome structure and gene regulatory landscapes in general."

Like so many others, the current state of science support worries her. "The perception – and related funding – that biology must always be related to human health in some way, is disturbing. It seems that, in the last decade or so, curiosity-driven research is much less supported than previously – except for the ERC’s grants which are a blessing for European research. Often, however, research has to be focused on improving the human condition, or else applicable commercially in order to attract funding. Things will turn around, though – I am sure we will realise (yet again) that it is only curiosity-driven research that can lead to discoveries that will be applicable to human health."

Even so, Heard feels she is lucky to have ended up as a scientist. "I almost became a musician and I used to think that I would have liked to be a historian. But working in science is a truly fascinating job, and I also enjoy watching the emergence of young scientists and seeing the leaps in understanding that are happening in biology."

A downside to this fascination, perhaps, is that she has little time for other interests. "I like music and art, and I love to read. Although I have no concrete plans for retirement at present, I don’t want it to be too late. And when it comes I would definitely like to do more of these things, as well as to watch my family evolve, write a book, spend more time in the Mediterranean where I have family roots, and maybe try to help the world in some way. Like many people, at present I am watching the news and worrying about our future….."

The subject of Heard’s lecture will be her lab’s work on trying to understand one of the most fundamental questions in biology: how do you shut down genes and how do you turn them back on again in a developmental context using the inactive X chromosome? "Our work on chromosome organisation has led us to some exciting new avenues and we are now exploring the process of X inactivation in the context of chromosome dynamics. Is chromosome folding into TADs a cause or a consequence of gene activation, and when and how does this happen in a chromosomal context?"

Answering this key question takes time. Such is her curiosity for further knowledge that it looks as though an early retirement is unlikely to appeal to Edith Heard.
ESHG Young Investigator Awards

The Scientific Programme Committee has shortlisted presenters for the ESHG Young Investigator Awards. The committee will judge finalists’ presentations during the conference.

The following awards will be presented to the winners in the closing ceremony on Tuesday, May 30, 2017 at 15.00 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 since 2012) will be awarded to the best presentation in translational genetic research/therapy of genetic diseases.

- The Mia Neri Award (established by the Mia Neri Foundation in 2015) will be awarded to the best presentation in childhood cancer research.

All winners will receive prize money in the amount of EUR 500, a complementary ESHG online membership for one year as well as a free participation in next year’s conference.

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

ESHG Poster Awards

The ESHG proposes the ESHG Poster Awards for the best posters presented by Young Investigators at the meeting. The two winners (one in clinical, the other in basic research) will receive a prize money of EUR 500, a complementary ESHG online membership for one year as well as a free participation in next year’s conference.

The five honorable mentions receive a complementary ESHG online membership for one year.

The ESHG Scientific Programme Committee has selected a number of candidates for the ESHG Poster Award based on the score of their submission after peer review. Candidate posters can be identified by a rosette on the board.

Presentation of the Candidates

On the next pages you will find short self-presentations of the candidates for Young Investigator and Poster Awards.
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 2017?

Raul Aguirre-Gamboa
Groningen, Netherlands

Talk: C22.3 Deconvolution of whole blood eQTLs into rare immune-subpopulations uncovers key players of immune mediated diseases
Concurrent Session C22 Systems Genetics
Date & Time: Tuesday, May 30, 2017, 11:30 hrs, Alicante

Q1: 29/08/1986, Monterrey, México.
Q2: PhD student at the Genetics Department, UMC Groningen
Q3: High-throughput genomics and the advances of statistical methods to analyze this huge amount of information brought genetics to the spotlight of personalized medicine and drug development. Contributing to this rapidly developing field is very rewarding, as it could potentially help improve our healthcare system and therefore the quality of life of patients suffering from complex genetic diseases.
Q4: Our talk focuses on a computational approach that detects cell type specific expression quantitative trait loci (eQTLs) by solely using expression from bulk whole blood RNA-seq data. In the context of complex diseases and their genetic risk factors, knowing in which cell type a risk factor is more likely to exert an eQTL effect its a crucial step towards understanding the pathogenesis of these diseases.

Tatsiana Aneichyk
Boston, United States

Talk: C08.3 Dissecting the causal mechanism of X-linked dystonia-parkinsonism by integrating genome and transcriptome assembly
Concurrent Session C08 Neuromuscular Disorders
Date & Time: Sunday, May 28, 2017, 13:30 hrs, Athens

Q1: 22/03/1985, Minsk, Belarus
Q2: Postdoctoral Fellow, Massachusetts General Hospital/ Harvard Medical School
Q3: I want to understand how the DNA, being a molecule containing just four components, can lead to a creation of a human being. My ability to use my knowledge of statistics, programming and algorithms to drive new discoveries of the ways the life is built is my main motivator.
Q4: X-linked dystonia parkinsonism is a Mendelian disorder that has been linked to a founder haplotype over 20 years ago, but no progress has been made since then in uncovering pathogenic mechanism. With new technologies, we are able to scrutinize this disease and move the research forward while making unexpected discoveries.

Daniel Bader
Munich, Germany

Talk: C13.3 Genetic diagnosis of Mendelian disorders via RNA sequencing
Concurrent Session C13 Innovative Variant Interpretation
Date & Time: Monday, May 29, 2017, 13:30 hrs, Aarhus

Q1: Wertheym, Germany
Q2: PhD student
Q3: Genetics, or for me rather bioinformatics focusing on disease characterization always kept me fascinated. At the end of my studies I wanted to understand diseases and ultimately help people. With this project I found the perfect match.
Q4: We developed a general pipeline to improve genetic diagnosis of Mendelian disorders. This project is exemplary for successful interdisciplinary collaboration between bioinformaticians, biochemists and clinicians.

Iris Barny
Paris, France

Talk: C11.4 Naturally-occuring exon-skipping allows bypassing complete CEP290 loss-of-function in individuals with unusually mild retinal disease
Concurrent Session C11 Sensory disorders
Date & Time: Sunday, May 28, 2017, 13:45 hrs, Alicante

Q1: February 27, 1992 - Paris, France
Q2: I’m a PhD student at the Laboratory of Genetics in Ophtalmology (LGO) in Imagine Institute, Paris, France
Q3: Research in human genetics is both complex and fascinating. Understanding the molecular mechanisms leading to diseases, that are so different on rarity and severity scale, is clearly a challenge to undertake, and for which I want to be part of. For me, the most motivating aspects of this field is the opportunity to make my contribution in treatment of patients affected from such illnesses.
Q4: Analyzing fibroblasts from two individuals having biallelic truncating CEP290 mutations but unexpectedly moderate visual dysfunction, I observed naturally occurring splicing of mutant exons and expression of minimally shortened CEP290 protein. This provides strong rational to my PhD project which assesses antisense oligonucleotides to bypass CEP290 truncation in blind LCA10 infants.

Miriam Bauwens
Ghent, Belgium

Talk: C11.5 Hidden genetic variation in Stargardt disease: novel copy number variations, cis-regulatory and deep-intronic splice variants of the ABCA4 locus
Concurrent Session C11 Sensory disorders
Date & Time: Sunday, May 28, 2017, 14:00 hrs, Alicante

Q1: 16-07-1986, Sint-Niklaas
Q2: PhD student at the Center for Medical Genetics Ghent
Q3: During my master studies in biomedical science, I became fascinated by the complexity of the human genome and intrigued by the link between genetic variation and disease. When I was
given the opportunity to start a PhD in this field, I gladly took it :-) Q4: My research focuses on unraveling the importance of non-coding mutations in a form of inherited blindness: Stargardt disease. Finding these hidden mutations provides these patients with a molecular diagnosis, which is a prerequisite for inclusion in the currently ongoing gene therapy trials. Also, identifying non-coding mutations enables us to explore new therapeutic approaches and will hopefully result in a better future for these patients.

Louise Benarroch
Paris, France
Talk: C05.5 Cross-mapping analysis identifies 9 modifier loci in Marfan syndrome
Concurrent Session C05 Skin and Bones
Date & Time: Saturday, May 27, 2017, 19:30 hrs, Amsterdam
Q1: 06.06.1989, Montreuil sous bois
Q2: PhD student
Q3: I chose genetics because, for me, genetics is a fascinating and thrilling domain of research. Genetics is in a perpetual evolution and everyday new discoveries are made. Understanding the mechanisms and interactions at this level could allow us to elucidate many mysteries of biology.
Q4: Cross-mapping is an innovative strategy based on crossing the results of several studies on different population. Indeed, a locus identified by using different methods will be considered as a potential regulatory locus. This strategy allow us to make the best use of all the data available and also to avoid creating a replication cohort, which is the main limitation when studying rare disease.

Christian Benner
Helsinki, Finland
Talk: C10.5 Prospects of fine-mapping causal genetic variants using summary statistics from genome-wide association studies
Concurrent Session C10 GWAS: Resolving Missing Causality
Date & Time: Sunday, May 28, 2017, 14:00 hrs, Cannes
Q1: April 10, 1982 in Leipzig/Germany
Q2: PhD student of Matti Pirinen at the University of Helsinki
Q3: I had to do an internship during undergraduate studies in statistics. I excluded many fields of study and genetics remained. I went for the internship, liked it and stuck with human genetics because its full of great problems that require skills in designing and implementing efficient statistical models and algorithms.
Q4: Several statistical methods have recently been introduced to fine-map genomic regions using GWAS summary statistics. Common to all methods is that they require Linkage Disequilibrium (LD) information between variants. Since the hope has been that LD information from publicly available reference panels could replace the original genotype data in fine-mapping analyses, we evaluate how estimation of LD from reference panels performs compared to the original individual-level GWAS data. Come see our presentation to hear about new software and good practices to more efficiently exploit summary statistics in fine-mapping analyses.

Stefania Benonisdottir
Reykjavik, Iceland
Talk: C18.3 Whole-genome sequencing identifies associations of sequence variants with clinically relevant urinary disease markers
Concurrent Session C18 Internal organs
Date & Time: Monday, May 29, 2017, 13:30 hrs, Amsterdam
Q1: 11th January, 1985, Reykjavik.
Q2: Research Scientist at the Statistic department at deCODE genetics.
Q3: Living in Iceland, a country where genealogy is considered a national hobby, the initiation in 1996 of large genetic studies by deCODE put human genetics at a central point of interest for many of my generation. I got familiarized with genetics during my master studies, applying statistical genetics to animal data in my MSc project to search for pairs of relatives in an Icelandic DNA-registry of fin whales. I subsequently shifted my focus to human genetics and, in 2014, had the unique opportunity to join deCODE. There, nationwide phenotypic and genotypic data enable us to examine associations underlying almost all observable traits and diseases. Thus far I have authored three published manuscripts and participated in numerous projects on common and rare human diseases. This has made me aware of how diverse the field of genetics can be and I am excited to continue my work.
Q4: Our study is based on a data set that consists of urine dipstick measurements from a large fraction of the Icelandic population. Whole-genome sequencing allows us to test these urinary markers for association with over 30 million variants, down to very low minor allele frequencies. Examining rare sequence variants is intriguing since they can lead to discoveries of geographic clusters, and the thorough Icelandic genealogical records make it possible to trace their time of creation or introduction into the population. Working with traits based on the chemical analysis of urine is also very interesting as it is one of the most widely used clinical tests, and the results can be considered as a natural variation in the population rather than a binary indication of disease status. By assessing the association of the detected variants with other phenotypes, we have the ability to uncover genetic links to diseases and shed light on their biology. In addition, this study emphasizes the role of genetic studies in the identification of drug targets.

Rufus Cartwright
London, United Kingdom
Talk: C18.4 Genome wide association study identifies two novel loci associated with female stress and urgency urinary incontinence
Concurrent Session C18 Internal organs
Date & Time: Monday, May 29, 2017, 13:45 hrs, Amsterdam
Q1: 27 February 1977, London
Q2: Specialist Trainee, Obstetrics and Gynaecology, Imperial College London
Q3: I wanted to pursue the genetic basis of the common pelvic floor disorders that affect my female patients.
Q4: I found it exciting to apply techniques developed for much more widely researched conditions to something as hidden and as stigmatising as incontinence.
Q1: 24 September 1986, Bolton.
Q2: PhD student
Q3: I became interested in genetics when I worked at a company developing genetic assays for personalised medicine. I like that it is a fast paced field where the technology and the scope of what can be achieved is constantly improving. I believe that genetics will be a driving force in many future medical advances.
Q4: The genetic syndrome I work on is extremely rare but the separate clinical features of the syndrome are much more common. Discoveries linked to this syndrome can make the genetic diagnosis of subsequent patients easier and may inform on the mechanisms behind more common genetic diseases.

Laura Fachal
Cambridge, United Kingdom

Talk: C10.4 Fine-mapping analysis of 158 breast cancer risk loci from OncoArray data
Concurrent Session C10 GWAS: Resolving Missing Causality
Date & Time: Sunday, May 28, 2017, 13:45 hrs, Cannes

Q1: 13/06/1980 Lugo, Spain
Q2: Postdoctoral Researcher at Centre for Cancer Genetic Epidemiology, University of Cambridge
Q3: I became fascinated with genetics while I was studying at high school. Later, whilst undertaking an MSc in Biotechnology, I had the opportunity to join the Genomic Medicine group in Santiago de Compostela. I love the multidisciplinary nature of the work, and the opportunities to learn new techniques. I started studying mendelian disorders and transitioned to the genetic epidemiology of complex disorders.
Q4: More than one hundred breast cancer independent signals have been identified through GWAS, but only one third of these regions have been studied in detail. Evaluating more than 150 loci together allows us to identify additional risk signals, as well as common epigenetic marks, along with transcription factors binding sites enriched at those loci. This aids us in elucidating the hidden genetic causes of breast cancer and piece together the complex and interactive genetic mechanisms underlying these risks.
Andrea Ganna
Cambridge, United States

Talk: PL2.2 Quantifying the impact of rare coding variation across the phenotypic spectrum
Plenary Session PL2 What’s New? Highlights
Session
Date & Time: Saturday, May 27, 2017, 16:45 hrs, Aarhus

Q1: 10th November 1985, Varese, Italy
Q2: Postdoctoral fellow
Q3: I’m always been fascinated about exploring large-scale datasets and implement novel statistical methods. Genetic is the perfect training territory since it requires data-analysis abilities and innovative approaches. Moreover, since it is mostly determined at birth, it can provide information about causality that no other risk factor can provide. Therefore is a unique tool for answering epidemiological questions.
Q4: We explored for the first time an area of the allele frequency spectrum that was not possible to explore before. Only by using such large-scale dataset we can now understand what is the impact of this very deleterious group of variants and how they can affect a person’s life. It is quite interesting that we can see an effect on number of hospital visits and educational attainment. This indicates that the effect of these very rare variants is detectable on population-level and, once we will have a better understanding about the driving signals, these results might have a real clinical impact.

Elisa Giorgio
Torino, Italy

Talk: C02.1 Allele-specific silencing as therapeutic strategy for disorders due to gene duplication: a proof-of-principle in Autosomal Dominant LeukoDystrophy (ADLD).
Concurrent Session C02 Neurogenetics 1
Date & Time: Saturday, May 27, 2017, 18:30 hrs, Copenhagen

Q1: Savona, 29/05/1984
Q2: I’m a post-doc researcher at the University of Turin, Dep. of Medical Sciences, Medical Genetics Unit (Italy).
Q3: Being a researcher in human genetics is not one single job, it is thousands of jobs simultaneously. On the one day, I’m a scientist who can change the life of people, an academic who can contribute to the advancement of science, but also a ‘glasswarewasher’, a secretary, a talent scout, a manager, a technician, a psychologist, a fundraiser, a teacher and a student… What more exciting and charming job could anyone choose?
Q4: My project represents a proof of principle for the use of Allele Specific-RNA interference (ASP-RNAi) as therapeutic strategy in disorders associated with gene duplication. In this study, I used ASP-RNAi to treat Autosomal Dominant adult-onset demyelinating Leukodystrophy (ADLD), a hereditary, progressive and fatal disorder caused by lamin B1 (LMNB1) duplication, in in vitro pre-clinical studies.

Madelyn Gillentine
Houston, TX, United States

Talk: C02.2 CHRNA7 CNVs: shared clinical phenotypes mediated by differing molecular mechanisms
Concurrent Session C02 Neurogenetics 1
Date & Time: Saturday, May 27, 2017, 18:45 hrs, Copenhagen

Q1: March 16, 1991
Dallas, TX, USA
Q2: I am a graduate student in Dr. Christian Schaaf’s lab at Baylor College of Medicine in the Dept. of Human and Molecular Genetics.
Q3: I was inspired to pursue a career in genetics because I have two older brothers with autism spectrum disorder of differing severity. That, combined with my love of biology, made genetics the perfect career path!
Q4: We are really excited about this work because it identifies a mechanism for 15q13.3 duplications, focusing on CHRNA7, in induced pluripotent stem cells (iPSCs) derived from patients with 15q13.3 CNVs. While the genomics and mRNA expression of CHRNA7 in duplication iPSC and differentiated neural progenitor cells (NPCs) suggest that we would have increased alpha7 nicotinic acetylcholine receptors (nAChRs) at the membrane, we’ve found that both 15q13.3 deletion and duplication NPCs have decreased alpha7 nAChR functionality. We’ve made steps in determining why this occurs in duplication cells and deletion cells. Most interesting, this suggests that similar therapeutics could be utilized for both groups of probands, which have begun to be tested in clinical trials.

Jakob Goldmann
Nijmegen, Netherlands

Talk: C14.2 Clustered de novo mutations with large intra-mutational distance contribute to the maternal age effect
Concurrent Session C14 Population Genetics and Ancient DNA
Date & Time: Monday, May 29, 2017, 13:15 hrs, Athens

Q1: 28.02.1987, Hamburg, Germany
Q2: I am PhD student in the Human Genetics Department of Nijmegen, the Netherlands in the Genome Research Section. I work in the bioinformatic group of Christian Gilissen.
Q3: Genetics is rocket science! Week by week, we get to know more about our genome and about the way that it works or sometimes struggles to work. Year by year, new technical innovations make possible what seemed science fiction before. Yet, we are still only scratching the surface of many biological problems. The role of genetics will continue to grow. In these times, working in genetics is extremely exciting.
Q4: Every single human being has a unique genome. This enormous diversity is established by de novo mutations, that are present in every newborn child, but not in its parents. Some of these mutations occur very close to each other on the same chromosome, separated by less than 20 kbp. The group of these clustered mutations has properties different from the rest of de novo mutations. We investigated these mutations and found that they are associated with oocyte aging. They contribute a significant fraction of all aging-associated mutation in the female germline.
Judith Grolleman
Nijmegen, Netherlands

Talk: C17.4 A somatic mutational signature in different tumor types associated with biallelic germline NTHL1 mutations
Concurrent Session C17 Hereditary Cancer
Date & Time: Monday, May 29, 2017, 13:45 hrs, Alicante

Q1: 09-11-1991, Zutphen, The Netherlands
Q2: PhD student at the Department of Human Genetics, Radboud University Nijmegen Medical Centre, Nijmegen, The Netherlands.
Q3: I’ve always been fascinated by the fact that aberrations in the DNA can affect the whole function and phenotype of an organism.
Q4: The research I am presenting enables to determine the tumour phenotype of patients based on the mutation pattern of a tumour; from DNA level to patient (care).

Tanya Halbersma-Konings
Groningen, Netherlands

Talk: C06.4 Recontact about clinically significant variant reclassifications in cardiogenetics; patient experiences
Concurrent Session C06 ELSI genomics
Date & Time: Saturday, May 27, 2017, 19:15 hrs, Cologne

Q1: 08-06-1985, Leiderdorp
Q2: I’m a PhD-student on the subject of re-contact in Clinical Genetics and a clinical geneticist in training at the University Medical Center Groningen in the Netherlands.
Q3: Genetic information can greatly influence the lives of patients and their family members. My goal is to give patients personalized information, help make choices (that are right for them) and promote communication with family members and medical professionals, USING DIGITAL HEALTH AS A (POSSIBLE) SOLUTION (both in research and patient care).
Q4: Our genetic knowledge and diagnostic technologies are increasing rapidly. This could lead to new genetic information that is of medical relevance to former patients. But how should we re-contact these patients ALSO REGARDING CONSTRAINTS IN WORKFORCE? WE NEED EMPIRICAL EVIDENCE TO PROVE FEASIBILITY (OID) In order to answer that question empirical evidence is needed.

Frederike Harms
Hamburg, Germany

Talk: C16.2 Mutations in EBF3 disturb transcriptional profiles and cause intellectual disability, ataxia, and facial dysmorphism
Concurrent Session C16 Intellectual Disability
Date & Time: Monday, May 29, 2017, 13:15 hrs, Cannes

Q1: 25.05.1990, Lüneburg, Germany
Q2: I’m a PhD-student on the subject of cardiac genetics; patient experiences
Q3: I chose a career in genetics because I am very interested in identifying the genetic cause of rare diseases and thereby help the patients and their families to find answers to their questions. At the institute I am working, we are not focussing on certain diseases. Instead, I am always confronted with new phenotypes which gives room for my knowledge about genes, proteins, and pathways to grow steadily. My work also involves a broad spectrum of genetic, biochemical, and cell biological methods. So, I am not limited to explain the genetic cause of the patient’s phenotype but also get deeper insight into protein function and a variety of signaling cascades. As I decided to become a scientist, I am very proud to be nevertheless part of the patient care at our hospital (as I am participating in genetic counseling of patients included in my research projects) and I hope that some of my findings will lead to therapeutic interventions in the future.
Q4: Next-generation sequencing allowed us to identify the EBF3 gene, encoding a member of the early B-cell factor transcription factor family, as a novel disease gene for intellectual disability, speech delay, ataxia, and facial dysmorphism.
Steven Harrison  
Cambridge, United States  

Talk: **C19.2 ClinGen Sequence Variant Interpretation Work Group recommendations for ACMG-AMP guideline specification**  
Concurrent Session C19 Diagnostic variant interpretation and quality control  
Date & Time: Tuesday, May 30, 2017, 11:15 hrs, Aarhus

Q1: May 30, 1984; Pleasanton, California, USA  
Q2: Clinical Molecular Genetics Fellow  
Q3: My interest in genetics began in high school when I first drew a pedigree and attempted to understand the genetics of my family. My interest has increased as I’ve learned more about the utility of clinical genetics and the impact it has on the care of patients with genetic disorders.  
Q4: The ACMG-AMP variant interpretation guidelines provide an evidence-based framework to classify variants; however further guidance on application of the guidelines is critical to move towards consistency in variant interpretations and transparency in classification rationale.

Gaëlle Hayot  
Illkirch-Graffenstaden, France  

Talk: **C23.1 Complex cis-interaction is responsible for the craniofacial and neuroanatomical defects of the 4p16.1 copy number variant.**  
Concurrent Session C23 Neurogenetics 2  
Date & Time: Tuesday, May 30, 2017, 11:00 hrs, Amsterdam

Q1: 27/04/1989, Paris  
Q2: PhD student in the Institute for Genetics and Molecular and Cellular Biology (IGBMC), Strasbourg, France  
Q3: The complexity of the genome amazes me. I am especially interested in understanding how some mutations cause human diseases while others are a motor of evolution.  
Q4: Using zebrafish, we are dissecting a rare CNV that has been linked to Autism Spectrum Disorder (ASD). We show that in that CNV, the different phenotypes of the patients are caused by different major drivers.

Frank Honti  
London, United Kingdom  

Talk: **C21.4 Copy number variants account for at least 2% of nonsyndromic cardiomyopathies**  
Concurrent Session C21 Cardiovascular disorders  
Date & Time: Tuesday, May 30, 2017, 11:45 hrs, Cannes

Q1: 5th Dec. 1982, Cezovce, Slovakia  
Q2: Bioinformatics Analyst at the Royal Brompton Hospital and Honorary Teaching Fellow in Genomic Medicine at Imperial College London  
Q3: Having been a curious kid, I created my first genetically modified plants with colchicine at the age of 15. Afterwards, I studied biochemistry and genetics and I feel very lucky to work in genetic diagnostics, which interests me most.  
Q4: We became very good at detecting tiny mutations from sequencing data, but knew that exon deletions and duplications were being overlooked. In contrast to earlier studies, my research uncovers a significant burden and enrichment of rare CNVs in cardiomyopathy patients, revealing the clinical utility of CNV calling in these conditions. In addition, I have developed a method that produces fewer false calls, decreasing the number of costly variant confirmations. I will present several clinically-significant CNVs and tell you how to find them.

Leigh Jackson  
Exeter, United Kingdom  

Talk: **C12.4 The European Gen-Equip project to create accessible resources for genetics education in primary care: an account of the process, the challenges and the successes.**  
Concurrent Session C12 Engaging Patients in Genomics  
Date & Time: Sunday, May 28, 2017, 13:45 hrs, Amsterdam

Q1: 28/05/1984 Plymouth, United Kingdom  
Q2: Lecturer in Genomic Medicine, University of Exeter  
Q3: I was always fascinated by science from an early age and decided I wanted to be a forensic scientist. As I grew older, I became captivated by the wonders of evolution and genetics and decided to pursue that instead. I have always had a passion for helping others to understand science.  
Q4: Now that we stand on the edge of an avalanche of genomic information cascading into health services, it is imperative that the primary care clinicians on the frontline understand enough about this often uncertain information to be able to best help their patients. Gen-Equip is creating high-quality free educational resources throughout Europe to assist the mainstreaming of genetics in the clinic.

Katherine Johnson  
Newcastle upon Tyne, United Kingdom  

Talk: **C08.4 Application of exome sequencing technologies to 1,000 patients affected by limb-girdle weakness of unknown origin**  
Concurrent Session C08 Neuromuscular Disorders  
Date & Time: Sunday, May 28, 2017, 13:45 hrs, Athens

Q2: Research Associate at Newcastle University  
Q3: I was always interested in science at school. I think it remarkable how seemingly small and insignificant DNA is, yet it is the molecular code for all living things – I love how nature is so intricate and finely-tuned. When I was able to specialise for my undergraduate degree, I knew that genetics was the only career for me. It isn’t just the subject of my degree and now my job, it is one of my biggest passions.  
Q4: To the best of our knowledge, we have gathered the largest ever cohort of patients with an unexplained limb-girdle weakness phenotype. These debilitating diseases are relatively rare in the general population, but collectively affect many people and can ultimately precipitate premature mortality. By sequencing the exomes of the affected individuals, we are able to decipher the true genetic cause of rare muscle diseases, explaining how single DNA changes have such huge consequences.
**Hákon Jónsson**
Reykjavík, Iceland

**Talk:** C15.1 Aging oocytes accelerate regional sequence diversity in humans and African apes

*Concurrent Session C15 Reproductive Genetics*

*Date & Time: Monday, May 29, 2017, 13:00 hrs, Copenhagen*

Q1: Sturlugata 8
Q2: Researcher at deCODE Genetics, working on de novo mutations.
Q3: Initially coming from a mathematical background, I was fascinated by the unique synergy of statistics, biology and computing in the interdisciplinary field of genomics. Sparked by this scientific curiosity, I pursued a Phd in evolutionary genomics at the University of Copenhagen, which I completed in 2015.
Q4: De novo mutations are the building blocks of evolution generated in a sex specific manner in the parental germlines. In our research we phased DNMs, allowing us to accurately estimate age-effects and mutational spectra in both germlines. We show a profound impact of maternal age on the regional sequence diversity of human populations and divergence between African great ape species.

**Mariann Koel**
Tartu, Estonia

**Talk:** C15.2 Interactome between embryo trophectoderm cells and endometrial epithelial and stromal cells: novel insights into implantation process in human

*Concurrent Session C15 Reproductive Genetics*

*Date & Time: Monday, May 29, 2017, 13:15 hrs, Copenhagen*

Q1: 2nd of July in Põlva, Estonia
Q2: I am PhD student in University of Tartu and researcher in the Competence Centre on Health Technologies.
Q3: My real passion is reproductive biology of humans. It is interesting to explore the early days of human development and shed light to the molecular mechanisms of the pregnancy. I also believe that the understanding of molecular causes of fertility is crucial for understanding life in general and therefore I chose it for my specialty.
Q4: Knowing that genetic disorders can be driven by a single variation within the 3 billion base pairs of the human genome is fascinating. By discovering novel disease-causing variants, we can therefore help understanding the molecular basis of the diseases and promote the development of drugs.

**Kristi Krebs**
Tartu, Estonia

**Talk:** PL2.4 Genetic variation in the Estonian population: a pharmacogenomic study of adverse drug reactions using electronic health records

*Plenary Session PL2 What’s New? Highlights*

*Date & Time: Saturday, May 27, 2017, 17:15 hrs, Aarhus*

Q1: 09.01.1990 Tallinn, Estonia
Q2: I am a second year doctoral student in Estonian Genome Center
Q3: I wanted to learn how genetics defines us in so different levels and how it can be used to study or predict different traits, diseases, and even history. I thought understanding more about genetics is crucial for understanding life in general and therefore I chose it for my specialty.
Q4: We study adverse drug reactions using Electronic health records for the first time in Estonian population. Our study shows that using e-health system is a powerful way to study genomics and proposes a way to identify individuals potentially at risk for unexpected drug response.

**Kilan Le Guennec**
Rouen, France

**Talk:** C02.3 17q21.31 duplication causes prominent Tau-related Dementia with Increased MAPT expression

*Concurrent Session C02 Neurogenetics 1*

*Date & Time: Saturday, May 27, 2017, 19:00 hrs, Copenhagen*

Q1: 06/20/1989 DOMONT, FRANCE
Q2: Ph.D Student
Q3: Initially coming from a mathematical background, I was fascinated by the unique synergy of statistics, biology and computing in the interdisciplinary field of genomics. Sparked by this scientific curiosity, I pursued a Phd in evolutionary genomics at the University of Copenhagen, which I completed in 2015.
Q4: We describe here a novel clinico-pathological entity driven by a genomic duplication of the 17q21.31 locus, encompassing the MAPT /Tau gene. Duplication carriers have increased MAPT/Tau expression and neurodegeneration associated with an extensive Tau pathology, leading to early-onset dementia.

**Stefan Lelieveld**
Nijmegen, Netherlands

**Talk:** PL2.6 Analysis of de novo mutation clustering identifies candidate disease genes in neurodevelopmental disorders due to likely gain-of-function and dominant-negative mechanisms

*Plenary Session PL2 What’s New? Highlights*

*Date & Time: Saturday, May 27, 2017, 17:45 hrs, Aarhus*

Q1: 2nd January 1987, Voorburg
Q2: I am a bioinformatics PhD student at the Genomics Disorders Group in the Radboud University Medical Centre Nijmegen
Q3: Working as a bioinformatician in genetics gives me the opportunity to work on the intersection of biology, computer science and statistics. I believe genetics is increasingly becoming a data-driven discipline where the analysis of large genomic datasets will give us more insight on how mechanisms of disease work and provide important answers to patients and their families.
Q4: I believe that our findings are of great interest as we developed a novel statistical method to identify disease genes within large trio studies that can easily be applied by others. Moreover, the application of our method to public datasets identifies novel candidate genes for neurodevelopmental disorders and suggests a larger role for disease mechanisms other than haploinsufficiency than previously thought. I hope that the methodology shown in this study will encourage fellow researchers to use the presented...
approach on their in-house cohorts. In addition, I believe that the identification of novel candidate genes will lead to new insights into the disease mechanisms and potentially in the long term, to therapy.

Vasiliki Matzaraki
Groningen, Netherlands

Talk: C01.3 A first genome-wide systems genetics approach identifies risk loci and pathways for candidaemia susceptibility
Concurrent Session C01 Personalized Medicine and Pharmacogenomics
Date & Time: Saturday, May 27, 2017, 19:00 hrs, Aarhus

Q1: 14 April 1984, Xanthi, Greece
Q2: I am a PhD student and have started my project in September 2014.
Q3: I am fascinated by the wealth of information that is hidden in our DNA and I wanted to learn why some individuals are more susceptible to a disease compared to others. If we understand this process, we may be able to develop new treatments and to prevent disease.
Q4: The interactions between environment and our genome have evolutionarily shaped our genome. Exploring the functional implications of DNA variants in the context of a phenotype is more insightful and, therefore, I am capturing these interactions in the context of an infectious disease (candidaemia) by investigating multiple molecular phenotypes, which may individually or together cause the infection.

Terri McVeigh
Sutton, United Kingdom

Talk: C01.1 The role of Next-generation sequencing in tumours in Adolescents and young adults (AYA) with advanced solid tumors participating in phase I trials
Concurrent Session C01 Personalized Medicine and Pharmacogenomics
Date & Time: Saturday, May 27, 2017, 18:30 hrs, Aarhus

Q1: 25/07/1986, Sligo, Ireland
Q2: Clinical Research Fellow in Cancer Genetics, Royal Marsden NHS Trust, London UK
Q3: Clinical genetics is incredibly varied, and always challenging - if the science isn't difficult enough, there will often be an ethical or psychosocial issue to deal with to keep things interesting!
Q4: Adolescents and young adults are an interesting group - with unique needs and risk factors. Identifying germline defects in such individuals is crucial, as most will have young siblings, children, or even young parents that all may be at risk of cancer. Somatic defects in these individuals are also noteworthy, as they may represent targets novel therapeutic agent. This study highlights the importance of profiling both the soma and the germline of these patients for their benefit, and the benefit of the wider family.

M. Loretto Munoz-Venegas
Lübeck, Germany

Talk: C21.3 Generalized compound heterozygosity analysis highlights associated loci for coronary artery disease in genetic and exome data
Concurrent Session C21 Cardiovascular disorders
Date & Time: Tuesday, May 30, 2017, 11:30 hrs, Cannes

Q1: 31/07/1981; Cape Town, South Africa
Q2: Postdoc at Institute for Cardiogenetics, University of Lübeck, Germany
Q3: I don’t think I consciously chose a career in genetics, it feels like it chose me. It all started when, at a very young age, I accompanied my mom to her lab at work one Saturday morning and looked through my first ("grown-up") microscope – I was hooked to the sciences and my curiosity was peaked. Since then, especially during my graduate studies, I have tried to pursue the questions I found most interesting; putting in the long hours and hard work to find their answers. Therefore, I would say that following my passion has been an influential guide throughout my career path.
Q4: In our study, we wanted to contribute to the current search for the genetics of coronary artery disease (CAD), by using the generalized form of double compound heterozygosity (GCDH) to detect the genetic associations caused by compound heterozygosity in European ancestry genetic and exome data. From our results, SNPs found to be genome-wide significant (≤5x10^{-8}) using the single-SNP based method were confirmed by GCDH; while others were only identified by using GCDH (≤1.25x10^{-10}). Thus hinting towards possible underlying mechanisms of CAD.

Lusine Nazaryan-Petersen
Copenhagen, Denmark

Talk: C09.3 Whole genome characterization of array defined clustered CNVs reveals two distinct complex rearrangement subclasses generated through either non-homologous repair or template switching
Concurrent Session C09 Molecular Mechanisms of Disease
Date & Time: Sunday, May 28, 2017, 13:30 hrs, Copenhagen

Q1: 23, December, 1980, Yerevan (Armenia)
Q2: Post doc.
Q3: I got my fascination in genetics in my high school while I was resolving simple tasks to figure out the patterns of inheritance.
Q4: We NGS sequenced clusterd CNVs to confirm whether or not they can be defined as chromothripsis or chromoanasynthesis, as well as we looked at the breakpoint-junctions to predict the potencial mechanisms for those complex rearrangements.
Q1: The recent technical advances in sequencing have opened up new and interesting opportunities in human genetic research, where suddenly we have population-scale genotype/phenotype information. After finishing a PhD were the main focus was on making and interpreting mouse knockout models I saw an opportunity to apply that experience in the field of human genetics.

Q2: Our research is focused on Williams-Beuren syndrome and disease, and human disease research has always been the most interesting area of study for me. Q3: One of the pillars of genome wide association studies (GWAS) is the replication of genotype-phenotype association. We have created a resource that will allow for assessing of published GWAS genotype-phenotype associations in the Icelandic population. This work will benefit the GWAS research community as a whole and should be of some interest.

Q4: Describing a completely novel genetic syndrome by combining the data from exome sequencing, model organisms and patients’ phenotypes is always exciting. Even more, I am fascinated how the modern data sharing efforts made our discovery possible by making international collaboration truly efficient. Finally, the patients involved in our study received the long-waited molecular diagnoses.

Asmundur Oddsson
Reykjavik, Iceland
Talk: C10.2 The deCODE replication server, a resource for the replication of published genotype-phenotype associations
Concurrent Session C10 GWAS: Resolving Missing Causality
Date & Time: Sunday, May 28, 2017, 13:15 hrs, Cannes

Q1: 14th of may 1980
Q2: Research associate at deCODE genetics in Reykjavik Iceland
Q3: The recent technical advances in sequencing have opened up new and interesting opportunities in human genetic research, where suddenly we have population-scale genotype/phenotype information. After finishing a PhD were the main focus was on making and interpreting mouse knockout models I saw an opportunity to apply that experience in the field of human genetics.

Q4: One of the pillars of genome wide association studies (GWAS) is the replication of genotype-phenotype association. We have created a resource that will allow for assessing of published GWAS genotype-phenotype associations in the Icelandic population. This work will benefit the GWAS research community as a whole and should be of some interest.

Paula Ortiz-Romero
Barcelona, Spain
Talk: C21.2 Epigallocatechin-3-gallate prevents cardiac hypertrophy in a Williams-Beuren syndrome mouse model
Concurrent Session C21 Cardiovascular disorders
Date & Time: Tuesday, May 30, 2017, 11:15 hrs, Cannes

Q1: 27th of February, 1994, Barcelona
Q2: I am a PhD student in the Genetics Unit at Universitat Pompeu Fabra (Barcelona).
Q3: Science, and specifically biology, has always been my main interest when deciding about my professional career. What I find more attractive about genetics is that it is closely related to health and disease, and human disease research has always been the most interesting area of study for me.

Q4: Our research is focused on Williams-Beuren syndrome and cardiovascular alterations associated to it. Our results revealed a molecule that could be used as a therapeutic agent. Moreover, the description of the pathway that is being affected allows the identification of alternative targets that may have the same therapeutic effect.

Sander Pajusalu
Tartu, Estonia
Talk: C02.5 BZRAP1 (RIM-BP1) mutations cause a novel autosomal recessive dystonia syndrome
Concurrent Session C02 Neurogenetics 1
Date & Time: Saturday, May 27, 2017, 19:30 hrs, Copenhagen

Q1: April 7, 1988, Tartu, Estonia.
Q2: Clinical geneticist in training at Tartu University Hospital and PhD student at the Institute of Clinical Medicine, University of Tartu, Estonia
Q3: As a medical doctor, I am convinced that every patient deserves a clear personal diagnosis which then should serve as the basis for personalized care. Working in a genetics clinic provides me the ability to work towards this dream by searching for disease causing genetic variants, counselling the patients, and doing research.

Q4: Describing a completely novel genetic syndrome by combining the data from exome sequencing, model organisms and patients’ phenotypes is always exciting. Even more, I am fascinated how the modern data sharing efforts made our discovery possible by making international collaboration truly efficient. Finally, the patients involved in our study received the long-waited molecular diagnoses.

Petros Patsali
Nicosia, Cyprus
Talk: C01.2 Less is More: knockdown of the aberrant HBB IVSI-1 10(G>A) mRNA restores HBB expression and enhances gene therapy by gene addition in primary erythroid cells
Concurrent Session C01 Personalized Medicine and Pharmacogenomics
Date & Time: Saturday, May 27, 2017, 18:45 hrs, Aarhus

Q1: 13/01/1987, Paralimni, Cyprus
Q2: I am a postdoctoral researcher in the department of Molecular Genetics Thalassaemia at The Cyprus Institute of Neurology and Genetics (CING)
Q3: Since the early stage of my biology undergraduate studies, I have always been fascinated by the intricacy of biology and the role of genetics in the fine balance between health and disease. Genetics is now a rapidly evolving field, approaching a comprehensive understanding of diseases and offering unique opportunities to develop novel therapies, such as those based on gene addition and genome editing.

Q4: We address aberrant β-globin mRNA as a factor in the disease pathology of β-thalassaemia by using RNAi, which merely removes aberrant transcripts but which in contrast to antisense-mediated splice repair can be applied as a single, permanent treatment, suitable for clinical translation. Planning to use RNAi merely to boost β-globin gene addition, we were surprised to see that RNAi not only enhances gene addition, but that it outperforms the latter when applied alone, by increasing endogenous β-globin levels. Our findings emphasise the importance of patient genotype, towards truly personal and more effective gene therapy.
Antoine Paul
Paris, France

Talk: C11.1 FDXR mutations cause sensorial neuropathies, a new mitochondrial Fe-S disease.
Concurrent Session C11 Sensory disorders
Date & Time: Sunday, May 28, 2017, 13:00 hrs, Alicante

Q1: 7th September 1987, Paris
Q2: I am currently working as an intern specialized in ENT.
Q3: I spent one year working in genetic as I am very interested in congenital genetic deafness. Correlating clinical and genetic data is key to improve our knowledge about physiopathology, and will moreover help in enhancing the care of affected children, in term of screening, diagnosis and treatment.
Q4: Our work provided understanding of the genetic cause in children suffering from auditory and optic neuropathies. To date, few is known about genetic cause of auditory neuropathy. Less is known about the rare cases of auditory associated with optic neuropathies. Studying the gene responsible for this particular association may provide a better understanding of the synaptic or neuronal transmission in sensorineural conditions.

Olga Plotnikova
Moscow, Russian Federation

Talk: C22.2 Expression insights into the human miRNA-mRNA interactome
Concurrent Session C22 Systems Genetics
Date & Time: Tuesday, May 30, 2017, 11:15 hrs, Alicante

Q1: 18 march 1993, Moscow (Russia)
Q2: PhD student
Q3: I have always had a strong interest in research and challenging tasks that can help people. As my way in science, I chose to apply bioinformatics methods for proceeding and understanding big biology data. I believe that it helps to reveal some new insights at the regulation level of the human genome. Rapid development in genetics inspires me and reminds how complex is human genome.
Q4: “Expression insights into the human miRNA-mRNA interactome”. In my research, I present different miRNA groups that could be biomarkers associated with adverse prognostic features in cancer and other severe pathologies. Only about 1% of mRNAs are actively engaged in miRNA interactions. Furthermore, we identified several coding mRNAs with a substantial sponge effect, including AGO1, which function may reflect the competition and resultant coevolution of mRNAs and miRNAs.

Margot Reijnders
Nijmegen, Netherlands

Talk: C16.3 Recurrent de novo missense mutations in small GTPase gene RAB11B cause severe intellectual disability and a distinctive brain phenotype
Concurrent Session C16 Intellectual Disability
Date & Time: Monday, May 29, 2017, 13:30 hrs, Cannes

Q1: July 27th 1989, Roosendaal (The Netherlands)
Q2: PhD student
Q3: During my medicine study, I was always fascinated by underlying genetic causes of diseases: very small defects in the human genome can have big consequences for the patient. This, as well as the fact that a large field within genetics is still unknown and has to be discovered, did me choose for a career in genetics.
Q4: Parents of five patients have searched for years for an explanation of the developmental delay in their child. Not only the introduction of WES in diagnostics, but also the increasing possibilities to share data with clinicians and researchers all over the world, allowed us to find the cause of the problems in the children: all had missense mutations in the RAS-GTPase RAB11B. Because only two different mutations were present in the five patients, we were able to focus our study on the functional effects of these specific missense mutations. Since RAB11B amino acids and domains are highly conserved among other members of the RAS-family, our results could potentially be used for other RAS-GTPases as well, of which many still await to be discovered.

Lehti Saag
Tartu, Estonia

Talk: C14.4 Farming in Estonia was introduced by Early Bronze Age migrants from the Steppe
Concurrent Session C14 Population Genetics and Ancient DNA
Date & Time: Monday, May 29, 2017, 13:45 hrs, Athens

Q1: 30.04.1991; Tartu, Estonia
Q2: PhD student
Q3: I have been interested in biology, evolution and archaeology since I was little so archaeogenetics and ancient DNA studies are the perfect study area for me.
Q4: Ancient DNA studies are extremely important for obtaining information about the demographic history of the world. My research from the Baltics (Estonia) shows very interesting results of hunter-gatherers being directly followed by Steppe ancestry farmers, skipping the first wave of farming into Europe from Anatolia.
Q1: My interests have always centred on biology and medicine. Medical Genetics, Poznan University of Medical Sciences
Q2: PhD student in Mathematical Genomics and Medicine at the University of Cambridge, supervised by Matt Hurles at the Wellcome Trust Sanger Institute.
Q3: I started off studying mathematics. When I was introduced to the concept of genome sequencing around 2009, I was blown away by how rapidly the technology had been progressing and all of the amazing research already being done. I swapped my degree from Mathematics and Economics to Mathematics and Biology and got involved with research at my University, starting first in mathematical modelling of the yeast mitotic spindle, then moving into cancer genetics and analysing gene expression microarrays. I found the confluence of mathematics, computer science, and genetics to be incredibly exciting and a perfect fit for my skills and interest. While the role mathematics and computer science has grown in many other fields, the prospect of having a tangible impact on people’s health and wellbeing that comes along with research in genetics is a powerful motivation.
Q4: Exome sequencing (sequencing the protein-coding parts of the genome) has been incredibly successful at identifying genes which, when disrupted by a mutation, result in severe developmental disorders. However, still more than half of families with a rare developmental disorder remain without a diagnosis. There has been great speculation about the degree to which mutations outside of protein-coding genes, in regulatory elements, contribute to developmental disorders. My research as a part of the Deciphering Developmental Disorders project, based on genetic data from almost 8,000 families, is the first study to show that mutations in regulatory elements do contribute to developmental disorders. Moreover, we can make an estimate for the proportion of all unsolved cases that are explained by mutations in these elements with a dominant mechanism (only 1-3%) as well as the proportion of sites within a regulatory element that, when mutated, cause a disorder with a dominant mechanism (far fewer than in genes). These estimates have important implications for how whole genome sequencing studies should be conducted and analysed in the future. In particular, that more work on understanding the ‘grammar’ of regulatory elements is crucial and that several hundred thousand families would be needed to comprehensively identify disease-associated regulatory elements.

Mahsa Shabani
Leuven, Belgium

Talk: C06.3 Legal framework for genomic data sharing in view of the new EU General Data Protection Regulation
Concurrent Session C06 ELStI genomics
Date & Time: Saturday, May 27, 2017, 19:00 hrs, Cologne

Q1: 09.07.1986 Oldenburg, Germany
Q2: PhD student in the research group of Prof. Dr. Wirth at the Institute of Human Genetics in Cologne, Germany
Q3: Genetics and especially human genetics is an exciting discipline for me, because unraveling the functional relevance of genes and the mechanisms underlying genetic disease phenotypes fascinate me, as it provides insights in the fundamental processes of our bodies. With the development of modern molecular, biological and high-throughput sequencing methods in the last decades, the field of human genetics is rapidly evolving and continuously pushing the frontiers of human knowledge, thus making it an extremely interesting work field.
Q4: Our research field aims at the identification of new genetic modifiers for SMA, a very common genetic neuromuscular disorder. By analyzing the genome of rarely existing genetic discordant families, we identify differentially regulated genes in individuals who are protected from SMA. Unravelling the functional role of these modifying genes allows us not only to find impaired processes in SMA, as recently shown for endocytosis, but also deepens our understanding of the complex SMA pathogenesis. Our pivotal goal is to translate this knowledge to further establish novel therapies to counteract disease phenotype.

Patrick Short
Hinxton, United Kingdom

Talk: C02.6 De novo mutations in regulatory elements cause neurodevelopmental disorders
Concurrent Session C02 Neurogenetics
Date & Time: Saturday, May 27, 2017, 19:45 hrs, Copenhagen

Q1: September 10th, 1991, Summit, New Jersey, USA
Q2: PhD student in Mathematical Genomics and Medicine at the University of Cambridge, supervised by Matt Hurles at the Wellcome Trust Sanger Institute.
Q3: I am fascinated with the potentials of genomic data and how they could revolutionize medical diagnosis and drug development. Nevertheless, there are certain ethical, legal and social challenges associated with processing genomic data, which need to be addressed by adequate mechanisms and policies. Working on ethical and legal aspects of genetic testing and genomic research provided me with an opportunity to contribute to a very dynamic interdisciplinary field, which is at a crossroad of science and society.
Q4: Adopting adequate privacy safeguards is paramount when processing genomic data for research or clinical purposes. One of the major legal instruments for personal data protection in the EU is the new General Data Protection Regulation (GDPR), which has entered into force in May 2016 and repealed the Directive 95/46/EC, with an ultimate goal of enhancing effectiveness and harmonization of personal data protection in the EU. My research discusses the rules for processing genomic data for research purposes in view of new GDPR.

Magdalena Socha
Poznań, Poland

Talk: C05.4 A 10q24.32 duplication causes bilateral femoral hypoplasia through formation of a novel sub-TAD
Concurrent Session C05 Skin and Bones
Date & Time: Saturday, May 27, 2017, 19:15 hrs, Amsterdam

Q1: 30.08.1984 Koszalin
Q2: Assistant, Chair and Department of Medical Genetics, Poznan University of Medical Sciences
Q3: My interests have always centred on biology and medicine. As a biologist I was drawn to the depth and breadth of the field...
of medical genetics and the countless challenges it creates. I have come to discover that the more I learn and understand about genetics the deeper I want to explore it.

Q4: What makes this study captivating is the identification of a novel cause of a unique and extremely rare malformation - femoral hypoplasia. Moreover, despite the fact that on the genomic level the causative aberration is very similar to the duplications reported in the split hand/foot malformation cases our patients’ phenotype is strikingly different.

Ana Sophia Valente
Porto, Portugal

Talk: C04.5 Cis-Regulatory Noncoding Elements, the hidden master weavers of CDH1 expression: lessons from HDGC patients
Concurrent Session C04 Epigenetics and Gene Regulation
Date & Time: Saturday, May 27, 2017, 19:30 hrs, Alicante

Q1: 20-08-1993, Porto, Portugal
Q2: I am a research fellow at the Expression Regulation in Cancer Group at the Institute for Research and Innovation in Health (i3S), Porto, Portugal
Q3: For me, the choice of a career in the genetics field was not a conscious decision or a plan that I followed. During my studies I did a couple internships in different areas and then I was very lucky to have a wonderful mentor who „contaminated” me with his passion for genetics. And I’m still here, highly enthusiastic for what comes next in this fast-developing field.
Q4: Hereditary Diffuse Gastric Cancer (HDGC) is a syndrome with high mortality rate that remains genetically unexplained in 50% of cases. With this research we try to understand the genetic cause in these cases by looking at the noncoding portion of the genome. If confirmed, our data could dramatically improve the diagnosis and management of these HDGC patients and their families.

Hanne Valgaeren
Edegem, Belgium

Talk: C11.3 Rare genetic variants in MEPE cause congenital facial paresis with stapes fixation, and are associated with otosclerosis
Concurrent Session C11 Sensory disorders
Date & Time: Sunday, May 28, 2017, 13:30 hrs, Alicante

Q1: 24/03/1990 Boom
Q2: Last-year PhD student at the Human Molecular Genetics group of the Center of Medical Genetics, University of Antwerp & Antwerp University Hospital
Q3: Genetics has always fascinated me. From the introduction to Mendel’s laws in secondary school throughout my Biomedical Sciences study at the University of Antwerp, this has only increased. It is fascinating how much information is encoded in our genes. When my PhD-position became available, I did not hesitate to apply in the hope of making a contribution to the field.
Q4: The research we present here has been performed in a large collaboration, and has led to the identification of one of the first otosclerosis genes, which is a break-through for the field. Furthermore, since we screened the gene in more than one thousand patients and more than one thousand controls, our study has sufficient power to unambiguously define its role in otosclerosis development.

Danya Vears
Leuven, Belgium

Talk: C06.2 Recommendations for the reporting of results from diagnostic next generation sequencing
Concurrent Session C06 ELSI genomics
Date & Time: Saturday, May 27, 2017, 18:45 hrs, Cologne

Q1: 12/05/1981 Melbourne, Australia
Q2: I am a postdoctoral research fellow with the Centre for Biomedical Ethics and Law, KU Leuven in Belgium where I am exploring ethical issues related to diagnostic uses of next generation sequencing.
Q3: I have been drawn to the field of human genetics since high school. As soon as I learned there was a profession which combined genetics, psychology and helping people, namely genetic counselling, I knew I had found my niche! I am a trained genetic counsellor but I love exploring people's perspectives on ethical issues relating to genetics and genomics so qualitative research has become my focus.
Q4: Although next generation sequencing has been around for quite a few years, researchers and clinicians are still in doubt about which findings to report following diagnostic testing and guidelines by professional bodies are not very explicit on this issue. I organised a working group of experts in this field in order to develop some useful recommendations to address this need in order to assist laboratories in their decision making about the reporting of variants.

Arnau Vich Vila
Groningen, Netherlands

Talk: C18.2 The microbiome of inflammatory bowel disease and irritable bowel syndrome - a case-control study of 1792 individuals
Concurrent Session C18 Internal organs
Date & Time: Monday, May 29, 2017, 13:15 hrs, Amsterdam

Q1: 29/08/1988, Sabadell, Barcelona (Catalunya)
Q2: PhD Student
Q3: Genetics is a fascinating field. I think it was fascinating to me the challenge of understanding as something as basic as the interplay of nucleotide pairs can become something as complex as long populations and interactions of living organisms. I also like genetics because it is a stimulating field that evolves quickly.
Q4: We study two of the most common gastrointestinal (GI) disorders; Irritable bowel syndrome (IBS) and inflammatory bowel disease (IBD). In previous studies, we found that high IBD genetic risk score is associated with a decrease in the genus Roseburia in the gut microbiota of healthy controls without gut complaints. Now by using microbial whole-genomes sequencing, higher taxonomical resolution can be reached and microbial functional pathways can be inferred. In this large case-control study we identified species- and strain-level microbial signatures associated with IBD and IBS, using stool samples from 355 IBD patients, 421 IBS patients and 1025 population controls. Strikingly, a substantial overlap between the gut microbiomes of patients with IBD and IBS compared to controls was observed. Nevertheless, we could distinguish IBD from IBS, using the high resolution microbial signatures as a biomarker. Our non-invasive stool test performed much better than the currently used fecal calprotectin test. Our study shows the importance understanding the gut microbiome and the opportunities for clinical treatments.
**Norine Voisin**  
Lausanne, Switzerland

**Talk:** C20.3 Variants in the degron motif of AFF3 cause a multisystem disorder with skeletal dysplasia and severe neurologic involvement  
**Concurrent Session C20 Molecular syndromology**  
**Date & Time:** Tuesday, May 30, 2017, 11:30 hrs, Copenhagen

**Q1:** January 19th, 1991 at Aubergenville, France  
**Q2:** PhD student  
**Q3:** I have always been interested in rare disorders and their mechanisms.  
**Q4:** Besides expanding the number of individuals with CHOPS syndrome, caused by mutations in AFF4, I will report on individuals with de novo mutations in AFF3. AFF3 has never been associated with pathologies before, and the new syndrome described here, including intellectual disability and skeletal dysplasia, is distinct from CHOPS syndrome.

**Kaitlin Wade**  
Bristol, United Kingdom

**Talk:** C10.3 Assessing the causal role of body mass index on cardiovascular health in young adults: a Mendelian randomization and recall-by-genotype analysis  
**Concurrent Session C10 GWAS: Resolving Missing Causality**  
**Date & Time:** Sunday, May 28, 2017, 13:30 hrs, Cannes

**Q1:** 12/05/1990 in St. Albans, UK  
**Q2:** Research Associate at the Integrative Epidemiology Unit based within the School of Social and Community Medicine at the University of Bristol.  
**Q3:** The ability to use multiple ‘omics’ data within a conventional epidemiological setting plus the skills and knowledge I acquired throughout my PhD motivated me to pursue a career in genetic epidemiology, applying and developing these complex methods to explore the causal relevance of adiposity on adverse health.  
**Q4:** With this innovative study design, using complementary multivariable regression, Mendelian randomization (MR) and recall-by-genotype (RbG) analyses, results strongly suggest a causal role of higher BMI resulting in adverse cardiovascular health in young adults. The consistency between results found from RbG with 418 participants to those from MR with 7,909 participants suggests this approach is valid and statistically very efficient. Furthermore, the RbG method predominantly allowed the collection of extremely precise cardiovascular phenotypes that would otherwise not have been possible in sample sizes required for MR.

**Nicola Whiffin**  
London, United Kingdom

**Talk:** C19.4 High-resolution variant filtering empowers clinical interpretation and provides insights into variant penetrance and population-specificity  
**Concurrent Session C19 Diagnostic variant interpretation and quality control**  
**Date & Time:** Tuesday, May 30, 2017, 11:45 hrs, Aarhus

**Q1:** 25th October 1988 in Derby, UK  
**Q2:** I am currently a Bioinformatics Research Associate at the MRC London Institute of Medical Sciences and the National Heart and Lung Institute at Imperial College London.  
**Q3:** It was as an undergraduate, studying Natural Sciences, that I was introduced to genetics. At that moment, I decided it was what I wanted to pursue. From then on, I have always followed the aspect of my work that I enjoy the most. During my PhD I stumbled upon bioinformatics, and was encouraged to learn to code. I have never looked back. Genetics, to me, is the perfect mix of the sciences; I find it fascinating and my work has a real clinical impact.  
**Q4:** Clinical genetic testing can be crucial in disease diagnosis and management of patients and their relatives; however, it can be tricky to determine which variants are disease-causing. Allele frequency is a key discriminator, as variants that are common in the general population cannot be causing a rare disease. We have taken this idea further, really digging down into disease architecture to determine ‘how common is too common?’ I am going to talk about how our statistically robust frequency thresholds add power to interpreting genetic variants, removing many candidate variants from contention but preserving variants that are truly disease-causing.

**James Whitworth**  
Cambridge, United Kingdom

**Talk:** C17.3 The optimal cancer genetics testing tool? - Diagnostic whole genome sequencing in research participants with multiple primary tumours  
**Concurrent Session C17 Hereditary Cancer**  
**Date & Time:** Monday, May 29, 2017, 13:30 hrs, Alicante

**Q1:** 25th October 1988 in Derby, UK  
**Q2:** Clinical research fellow - Department of Medical Genetics, University of Cambridge.  
**Q3:** It was as an undergraduate, studying Natural Sciences, that I was introduced to genetics. At that moment, I decided it was what I wanted to pursue. From then on, I have always followed the aspect of my work that I enjoy the most. During my PhD I stumbled upon bioinformatics, and was encouraged to learn to code. I have never looked back. Genetics, to me, is the perfect mix of the sciences; I find it fascinating and my work has a real clinical impact.  
**Q4:** Clinical genetic testing can be crucial in disease diagnosis and management of patients and their relatives; however, it can be tricky to determine which variants are disease-causing. Allele frequency is a key discriminator, as variants that are common in the general population cannot be causing a rare disease. We have taken this idea further, really digging down into disease architecture to determine ‘how common is too common?’ I am going to talk about how our statistically robust frequency thresholds add power to interpreting genetic variants, removing many candidate variants from contention but preserving variants that are truly disease-causing.
**Programme Young Investigator Award Candidates**

**Anja Will**  
Berlin, Germany  
**Talk:** PL2.1 Enhancer composition and dosage control developmental gene expression  
*Plenary Session PL2 What’s New? Highlights Session*  
*Date & Time: Saturday, May 27, 2017, 16:30 hrs, Aarhus*

Q1: 31.01.1986 Pirna, Germany  
Q2: I am a PhD Student at the Max-Planck-Institute of Molecular Genetics in Berlin.  
Q3: For me, genetics is a fundamental element of life as the genome of every organism is unique. It is achieved by variations in the genome that might be tolerated but can also result in disease as these variations might influence genome organization and gene regulation. I was always interested in how genes are regulated and how faulty regulation might be associated with disease.  
Q4: Gene expression is regulated by enhancers, which commonly arrange in clusters. To date, enhancer function has been assessed by deletion studies only, without considering the effects of increased enhancer dosage or alterations in the composition of enhancer clusters - modifications that are common in human disease. I studied this phenomenon at the Ihh locus, at which variable non-coding duplications have been associated with the human phenotypes Craniosynostosis and Syn(poly)dactyly.

**Rosa Woldegebriel**  
University of Helsinki, Finland  
**Talk:** C23.5 ATPase-deficient ATAD3A alters mitochondrial dynamics in hereditary spastic paraplegia  
*Concurrent Session C23 Neurogenetics 2*  
*Date & Time: Tuesday, May 30, 2017, 12:00 hrs, Amsterdam*

Q1: 1989, Helsinki  
Q2: Student  
Q3: Science can make a real difference in the world, and that is what I am passionate about. Genetics is the most fascinating and endless source of discovery and I could not think of a better career than genetics combined with neuroscience - neurogenetics. These two complex and intertwined fields can help us understand how genetic changes contribute to neurological diseases. My goal is to participate in the discovery of new genetic mutations and studying their molecular mechanisms. The ultimate goal is find cure for complex neurological diseases.  
Q4: The discovery of association between a gene and a specific disease is crucial for understanding of the molecular mechanisms of a disease and how it can be treated in the future. This is especially important for diseases such as hereditary spastic paraplegia, which is extremely heterogeneous. We have identified a family with a mutation in mitochondrial protein ATAD3A. Molecular studies showed that mitochondrial networks and dynamics were severely disturbed. We show the association between ATAD3A and dominant HSP with intrafamiliar variability. With these findings and future studies into the protein, it will be useful for clinical diagnosis, for understanding the cellular mechanisms of this family of diseases and for potential future treatment.

**Victor Yakimov**  
Copenhagen, Denmark  
**Talk:** C14.3 Admixture mapping identifies Inuit ancestry loci associated with metabolic traits in the Greenlandic population  
*Concurrent Session C14 Population Genetics and Ancient DNA*  
*Date & Time: Monday, May 29, 2017, 13:30 hrs, Athens*

Q1: September 9th, 1987 — Sofia, Bulgaria  
Q2: Bioinformatician  
Q3: Living things are these messy, incredibly complicated, but ultimately knowable pieces of chemical machinery. I think genetics is the best way to determine what each of the different parts do.  
Q4: Isolated populations living in extreme conditions face unique selective pressures, and this should be reflected in their genetic profile. Looking at local ancestry is a way to directly examine this.
Hassan Abolhassani
Huddinge, Sweden
Poster: **P07.01A Application of next generation sequencing in the conundrum of primary antibody deficiency**
Poster Session P07 Immunology and hematopoietic system
Presence at the poster: Sunday, May 28, 2017, 10.15 - 11.15 hrs, Poster Area

Q1: 1986-09-17, Tehran, Iran
Q2: PhD student of Clinical Immunology
Q3: Genetic diagnosis of patients with inborn immune disorders is currently “Achilles Heel” in the field of clinical immunology and more than 80% of molecular defects underlying these phenotypes are unknown. This process is also necessary for targeted therapy and family consulting and long term prognosis estimation.
Q4: After more than 15 years comprehensive clinical, immunologic and basic studies in the field of primary antibody deficiency we have developed the most efficient and successful pipeline in the genetic diagnosis of these patients identifying approximately 80% of the pathogenesis.

Rodrigo Almeida
Curitiba, Brazil
Poster: **P12.001A Integrative approach and eQTL analysis identify breast cancer risk genetic variants affecting microRNA binding sites.**
Poster Session P12 Cancer genetics
Presence at the poster: Sunday, May 28, 2017, 10.15 - 11.15 hrs, Poster Area

Q1: 09/11/1980 Bom Jesus da Lapa, Brazil
Q2: Currently I’m a postdoc researcher in the Molecular Epidemiology Department at the Leiden University Medical Center.
Q3: For me genetics is one of the most exciting field of out time. I choose a career in genetics because I liked the idea to understand one of the most fundamentals discipline in biology. Besides, there are numerous opportunities in the field.
Q4: In this „Big Data Era“, I’ll present a data integration approach that shows how to interpreted non-coding genetic variants associate to breast cancer, which can regulate microRNAs target genes expression. Therefore, this variants could have an effect in the development of the disease.

Anna Babayan
Hamburg, Germany
Poster: **P12.002B Dissecting cancer evolution by liquid biopsy and single cell genomic interrogation**
Poster Session P12 Cancer genetics
Presence at the poster: Sunday, May 28, 2017, 16.45 - 17.45 hrs, Poster Area

Q1: 25-05-1984, Meerbusch, Germany
Q2: Postdoc
Q3: I have Always been interested in mechanisms of disease, and during my Medical training, I was fascinated by genetics. After doing a PhD in epigenetics on X inactivation, I wanted to learn more about gene regulation by non-coding elements, and therefore I have continued my postdoctoral research in that area, using embryonic stem cells as a model system
Q4: Although most of the genome is non-coding, it still very hard to predict those non-coding elements that are functional. The work presented here established a novel, quantitative and genome-wide approach to identify functional enhancers, by combining chromatin-immunoprecipitation with a massively-parallel-reporter-assay. This yielded novel information about the characteristics of functional enhancers, which might be useful to extrapolate to other areas of genetics to further crack the non-coding genome code.

Stefan Barakat
Rotterdam, Netherlands
Poster: **P17.02B Functional dissection of the enhancer network in human embryonic stem cells by ChiP-STARR-seq**
Poster Session P17 Epigenetics and Gene Regulation
Presence at the poster: Sunday, May 28, 2017, 16.45 - 17.45 hrs, Poster Area

Q1: 29-10-1982 Lisbon
Q2: Post-doctoral researcher at Vertebrate Development and Regeneration Group at I3S/IBMC, Porto, Portugal
Q3: I see the future of Medicine lying on Genetics, since is the basis of biological systems, and a vast majority of diseases is caused by genetic alterations. By understanding the genetic basis of disease we can develop more specific and effective tools to prevent disease and to treat patients in a more personalized manner.
Q4: Genetic research in disease has been focusing mostly in gene coding sequence. However, cis-regulatory elements(CREs), required for proper gene expression, can have a detrimental impact on disease when mutated. We are interested in exploring the role of these elements in the proper expression/function of cancer-related genes and their contribution to pancreatic cancer development. CREs might constitute new susceptibility alleles and/or potential therapeutical targets, with important impact on clinical management.

Renata Bordeira-Carício
Porto, Portugal
Poster: **P17.03C Unveiling the regulatory landscapes of genes involved in pancreatic cancer using a zebrafish model**
Poster Session P17 Epigenetics and Gene Regulation
Presence at the poster: Monday, May 29, 2017, 10.15 - 11.15 hrs, Poster Area

Q1: 29-10-1982 Lisbon
Q2: Post-doctoral researcher at Vertebrate Development and Regeneration Group at I3S/IBMC, Porto, Portugal
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Henrike Heyne
Leipzig, Germany
Poster: P09.004D Elucidating the spectrum of protein-altering de novo variants in neurodevelopmental disorders with epilepsy
Poster Session P09 Neurogenetic and psychiatric disorders
Presence at the poster: Monday, May 29, 2017, 16.45 - 17.45 hrs, Poster Area

Q1: 24th September 1986, Berlin (Germany)
Q2: postdoctoral research fellow mentored by Mark Daly

Analytical Translational Genetics Unit, Massachusetts General Hospital (Boston, USA) / Broad Institute of Harvard and MIT (Cambridge, USA)

Q3: In human genetics as a relatively young medical discipline, research and clinical application are tightly linked. It is exciting to work in such a quickly evolving field.
Q4: Our study provides the first comprehensive picture of the genetic architecture of neurodevelopmental disorders with epilepsy and will likely substantially impact clinical definitions of epilepsy entities as well as design of diagnostic testing applications.

Louiza Kalokairinou
Leuven, Belgium
Poster: P20.01A Regulation of direct-to-consumer genetic testing in Europe: a fragmented landscape
Poster Session P20 Psychological/Ethical/legal issues
Presence at the poster: Sunday, May 28, 2017, 10.15 - 11.15 hrs, Poster Area

Q1: 27.11.1985, Fort Riley, Kansas, USA
Q2: Doctoral student in Molecular Human Genetics, Institute of Human Genetics, Mainz, Germany
Q3: We are all driven by the same questions: Who are we, where did we come from and how are we still alive? We want to decipher the code of life and the mechanisms of disease. A research career in this field is fascinating, illuminating and so rewarding. I am intrigued to find out how our environment shapes us within our epigenetics and how novel techniques will assist us in the process.
Q4: Do not underestimate epigenetics. The research I am presenting provides an epigenetic resolution underlying the mechanism and elucidation of the Birk-Barel mental retardation dysmorphism syndrome. Our epigenetic drug administration in mice show a promising approach for the treatment of the disease.

Laura Kasak
Tartu, Estonia
Poster: P01.001A Copy number variation profile in the placental and parental genomes of recurrent pregnancy loss families
Concurrent Session P01 Reproductive Genetics/Prenatal Genetics
Presence at the poster: Sunday, May 28, 2017, 10.15 - 11.15 hrs, Poster Area and in the concurrent session C03 Best Posters Session, Saturday, May 27, 18.30-20.00 hrs, Cannes

Q1: 30.12.1987 Tartu, Estonia
Q2: I am currently a final year PhD student at the Institute of Molecular and Cell Biology, University of Tartu, Estonia
Q3: Since the first year of my undergraduate studies I was fascinated with human genetics. Fortunately, I was soon welcomed at the human molecular genetics research group of Professor Maris Laan. One of our interests is the placental genome structure and function in such a quickly evolving field.
Q4: Our study provides the first comprehensive picture of the genetic architecture of neurodevelopmental disorders with epilepsy entities as well as design of diagnostic testing applications.

Q4: We have recently shown an extensive load of somatic copy number variations (CNVs) in the human placental genome with the highest fraction detected in normal term pregnancies. In the current study we hypothesized that insufficient promotion of CNVs may impair placental development and lead to recurrent pregnancy loss. Our results may have a number of perspective implications and suggest that early placental development may need a burst of
somatic genomic rearrangements to guarantee active proliferation, migration and invasion of trophoblasts.

Svenja Kohler
Lübeck, Germany
Poster: **P10.01A Col6A2 p.G283E: antisense-induced mRNA knockdown as a possible treatment strategy**
*Poster Session P10 Neuromuscular disorders*
*Presence at the poster: Sunday, May 28, 2017, 10.15 - 11.15 hrs, Poster Area*

Q1: 18.06.1993 Rodalben
Q2: MD student

Janine Milbradt
Cologne, Germany
Poster: **P04.02B Plastin 3 regulates bone development and maintenance through the NFκB pathway in osteoclasts**
*Concurrent Session P04 Skeletal, connective tissue, ectodermal and skin disorders*
*Presence at the poster: Sunday, May 28, 2017, 16.45 - 17.45 hrs, Poster Area and in the concurrent session C03 Best Posters Session, Saturday, May 27, 18.30-20.00 hrs, Cannes*

Q1: 20.10.1984 in Berlin, Germany
Q2: PhD student at the Institute of Human Genetics in Cologne (RG BProf. Dr. Brunhilde Wirth)
Q3: I was specifically interested in human genetics because I wanted to produce valuable data that could be beneficial one day for the treatment of various diseases affecting people worldwide.
Q4: We found a novel protein interaction affecting the NFkB pathway which could not only have importance for therapeutic aspects in osteoporosis, but may also have crucial impact on other areas in human genetics, for example immunologic or neurologic diseases.

Antonino Montalbano
Heidelberg, Germany
Poster: **P04.03C Retinoic acid catabolizing enzyme CYP26C1 is a genetic modifier in SHOX deficiency**
*Concurrent Session P04 Skeletal, connective tissue, ectodermal and skin disorders*
*Presence at the poster: Monday, May 29, 2017, 10.15 - 11.15 hrs, Poster Area and in the concurrent session C03 Best Posters Session, Saturday, May 27, 18.30-20.00 hrs, Cannes*

Q1: 24/06/1986, Palermo
Q2: Postdoc at Prof. Neville Sanjana’s lab, New York Genome Center and New York University, New York, USA.
Q3: It is a very exciting time to be a geneticist. Reduction of the sequencing costs is leading to the generation of increasing amounts of data that need to be analyzed. There is an urgent need of new generation scientists able to work with those data to understand how changes in DNA sequences can lead to disease. I want to be part of the new generation of scientists, able to combine genetics, bioinformatics, and wet lab experiments to unravel mechanisms of disease with the ultimate goal to discovery novel diagnostic markers and new therapeutic targets.
Q4: The research I am presenting at the conference used genetic analyses, whole genome linkage analysis and whole exome sequencing, to identify candidate genetic modifiers. Functional analysis were performed to characterize the mechanisms leading to alteration of the disease. This study is one among the few that identified the mechanism of disease modifications by genetic factors in human. Identifying disease modifiers may enable the accurate prediction of disease progression and improve therapeutic development.

Anna Morgan
Trieste, Italy
Poster: **P02.01A Next Generation Sequencing (NGS) followed by in vitro and in vivofunctional studies revealed new genes for both Hereditary (HHL) and Age Related Hearing Loss (ARHL).**
*Poster Session P02 Sensory disorders (eye, ear, pain)*
*Presence at the poster: Sunday, May 28, 2017, 10.15 - 11.15 hrs, Poster Area*

Q1: 22/10/1988, Conegliano (TV), Italy
Q2: Post Doc at the Department of Medicine, Surgery and Health Sciences, University of Trieste
Q3: Since my first Genetics course during University, I started realizing how Genetics contribute to many aspects of human life. This intuition prompted me to dedicate my research activity to this field. In particular, I am interested in uncovering the molecular mechanisms leading to genetic diseases and in understanding the link between DNA mutations and human phenotypes.
Q4: My work focuses on the study of both monogenic and complex forms of hearing loss (HL). HL is the most common sensory disorder in human and, despite the numerous advances in sequencing technologies, it is not always possible to reach a clear molecular diagnosis. The aim of my research project is to identify new HL-genes, thus helping in understanding the biology of the hearing system and providing hints for new therapeutic approaches.

Lam Son Nguyen
Paris, France
Poster: **P09.003C Role of miR-146a in the differentiation and neural lineage identity determination of human neural stem cells: relevance for autism spectrum disorders**
*Poster Session P09 Neurogenetic and psychiatric disorders*
*Presence at the poster: Monday, May 29, 2017, 10.15 - 11.15 hrs, Poster Area*

Q1: 9th April 1984
Q2: Post-doctoral Researcher
Q3: I choose a career in genetics because there is few therapy available currently for genetic disorders, especially for brain disorders. I believe that I can make a change.
Q4: My work explores the contribution of an epigenetic factor, namely microRNA, in neurodevelopmental disorders. This work is interesting because it demonstrates a causative role of microRNA in these diseases. More importantly, treatment with synthetic miRNA constructs by nasal spray has been proven to successfully reverse disease symptom in rat models. This brings treatment for genetic brain disorders one step closer to reality.
**Ron Nudel**  
Roskilde, Denmark  
Poster: **P16.01A GeneHancer: Genome-wide integration and scoring of enhancers and target genes in GeneCards**  
*Poster Session P16 Omics/Bioinformatics*  
*Presence at the poster: Sunday, May 28, 2017, 10.15 - 11.15 hrs, Poster Area*

**Manon Oud**  
Nijmegen, Netherlands  
Poster: **P01.002B Validation and application of a novel integrated genetic screening method to a cohort of 1,112 men with idiopathic azoospermia or severe oligozoospermia**  
*Poster Session P01 Reproductive Genetics/ Prenatal Genetics*  
*Presence at the poster: Sunday, May 28, 2017, 16.45 - 17.45 hrs, Poster Area*

**Gundula Povysil**  
Linz, Austria  
Poster: **P16.03C panelcn.MOPS: CNV detection in targeted NGS panel data for clinical diagnostics**  
*Poster Session P16 Omics/Bioinformatics*  
*Presence at the poster: Monday, May 29, 2017, 10.15 - 11.15 hrs, Poster Area*

**Sonika Rathi**  
Hyderabad, India  
Poster: **P13.01A Molecular genetic and functional analysis indicate novel genes involvement in retinopathy of prematurity**  
*Poster Session P13 Basic mechanisms in molecular and cytogenetics*  
*Presence at the poster: Sunday, May 28, 2017, 10.15 - 11.15 hrs, Poster Area*

**Neus Roca-Ayats**  
Barcelona, Spain  
Poster: **P15.01A A GGPS1 mutation found by WES in three sisters with bisphosphonate-associated atypical femoral fractures**  
*Poster Session P15 Personalized/Predictive Medicine and Pharmacogenomics*  
*Presence at the poster: Sunday, May 28, 2017, 10.15 - 11.15 hrs, Poster Area*

**Lara Rodriguez Laguna**  
Madrid, Spain  
Poster: **P11.001A Somatic activating PIK3CA mutations cause CLAPO syndrome**  
*Poster Session P11 Multiple Malformation/ anomalies syndromes*  
*Presence at the poster: Sunday, May 28, 2017, 10.15 - 11.15 hrs, Poster Area*

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**Q1:** 12-09-1992, Nijmegen  
**Q2:** PhD student  
**Q3:** I have been fascinated by DNA ever since I visited a Science Museum in Amsterdam as a child. The fascination only grew during my Biology studies and I decided to learn more by doing a PhD in genetics.  
**Q4:** Our study shows that it is possible to consolidate the detection of different types of genetic variation causing male infertility while increasing the diagnostic yield and detection precision at the same or lower price compared to currently used methods. This can greatly help both research and diagnostics in the field of male infertility.

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**Q1:** 29/04/1984, Indore, India  
**Q2:** Research Associate  
**Q3:** From my higher education I found genetics very interesting and logical. I realized genetics is the background of most of the biomedical Research.  
**Q4:** The genetics involvement in ROP is not clear till now. My research not only identified novel genes in ROP pathogenesis but also whole mechanism and finally biomarkers which can be used for the progression of the diseases.

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**Q1:** 24th January 1991, Vic (Spain)  
**Q2:** PhD student at the University of Barcelona  
**Q3:** I got fascinated by Genetics during my studies in Biochemistry, although it had already caught my attention before. I am interested in how life works at molecular level and in deciphering the mechanisms that underlie diseases. Genetics is a multidisciplinary field that evolves a lot, which make it challenging and enable me to learn continuously.  
**Q4:** Atypical femoral fractures (AFFs) are a rare but devastating type of fracture possibly associated with long-term bisphosphonate (BP) therapy, the main treatment for osteoporosis and cancer-related bone disease. This is the first WES performed in AFF patients and a novel mutation in GGPS1 was identified, which totally impairs its function. The encoded enzyme, GGPPS, is involved in the mevalonate pathway and it is known to be inhibited by BPs. This study opens the door to risk assessment tools to personalize the therapy.

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**Q1:** May, 18th 1989, Madrid, Spain.  
**Q2:** PhD student.  
**Q3:** My vocation for genetics arises from my curiosity and passion...
for medicine, diseases and technology. Having the chance to gain knowledge in the natural development and evolution of diseases, and being able to apply it to patients, was what lead me to choose genetics as my professional career.

Q4: This paper describes for the first time the genetic cause of a rare disease of which there was little knowledge. In addition, since the work includes this syndrome as a part of a larger known spectrum, the existing knowledge regarding future possibilities of treatment, pathogenic mechanism and counselling with reproductive intentions, it is automatically applied to CLAPO syndrome improving its personalized advice. Finally by working in a hospital service, I could personally observe the importance for patients and their families to have a specific diagnosis after years of uncertainty and anxiety.

Francesco Russo
Copenhagen, Denmark

Poster: P16.02B Comorbidity landscape of the Danish patient population affected by chromosomal abnormalities
Poster Session P16 Omics/Bioinformatics
Presence at the poster: Sunday, May 28, 2017, 16.45 - 17.45 hrs, Poster Area

Q1: September 20, 1985, Catania (Italy)
Q2: PhD student at Novo Nordisk Foundation Center for Protein Research, University of Copenhagen
Q3: I am a molecular biologist and genetics is a new field for me. I started around one year ago studying the Klinefelter Syndrome, an opportunity to learn more about complex diseases. It was love at first sight! After few months our work was accepted for publication. A great recognition! I hope to work more in this field and hopefully help people with my little contribution.
Q4: My research presents a comprehensive epidemiological study of the entire Danish population affected by chromosomal abnormalities. We have unique data consisting of the national patient registry which includes 6.9 million patients. We discovered more than 9,000 patients affected by chromosomal abnormalities and we found specific comorbidities for each chromosomal abnormality but also interesting overlaps between diseases.

Shiri Shkedi-Rafid
Jerusalem, Israel

Poster: P19.02B Pregnant genetic counselors in the era of advanced genomic tests: What do the experts test prenatally?
Poster Session P19 Genetic counselling/Education/public services
Presence at the poster: Sunday, May 28, 2017, 16.45 - 17.45 hrs, Poster Area

Q1: 18 March 1975, Jerusalem, Israel
Q2: Genetic counselor at the Center for Clinical Genetics, Hadassah Hebrew University Medical Center Director, MsC program in genetic counseling. Faculty of Medicine, the Hebrew University of Jerusalem
Q3: Genetic counseling is a unique multidisciplinary field. I was fascinated by the combination of human interaction, never ending learning, and by the many research opportunities.
Q4: It has frequently been argued that knowledge and probabilistic/statistical understanding are essential to allowing pregnant women to make sound, informed decisions about which genetic tests to perform during pregnancy.

Genetic counselors, unlike most women who take prenatal diagnostic tests, are both highly knowledgeable about the various outcomes of genetic tests, and trained in statistics and concepts of probability. They are also a unique group in the sense that they do not oppose the selection of future generations on the basis of their health prospects, as this is their professional practice. Moreover, their practice overexposes them to genetic anomalies and risk. Based on their specific features, it can be argued that pregnant genetic counselors are highly capable of making informed decisions as to which tests, and which test results, they wish to receive during their own pregnancies. Yet it is not clear whether they would be early adopters of new technologies, or prefer to practice caution, since they encounter in their work both the advantages and disadvantages of advanced genomic testing. The novelty of our study is that it adds to the existing literature, which has focused either on medical experts’ professional views, or on public preferences and experience, by shedding light on the actions of professionals as patients.

Arthur Sorlin
Dijon, France

Poster: P04.01A Postzygotic dominant-negative mutations of RHOCause a mosaic neuroectodermal syndrome
Concurrent Session P04 Skeletal, connective tissue, ectodermal and skin disorders
Presence at the poster: Sunday, May 28, 2017, 10.15 - 11.15 hrs, Poster Area and in the concurrent session C03 Best Posters Session, Saturday, May 27, 18.30-20.00 hrs, Cannes

Q1: 28/09/1988, Firminy (France)
Q2: PhD student and medical geneticist in training
Q3: As a medical doctor, genetics is a fascinating field to work in, as it deals with cutting-edge science, precision medicine, and major ethical questions. I am convinced that advances in genetics will shape the medicine of tomorrow, and will highly improve patients’ life. Q4: Using next generation sequencing, we identified the cause of a recognizable, previously undescribed mosaic syndrome. We reported the first gene firmly associated with hypomelanosis of Ito. Contrary to other mosaic syndromes, caused by activating mutations, we demonstrated that our variants have a dominant-negative effect, providing a new mechanism for lethal mutations surviving only by mosaicism.

Sanne van der Steen
Rotterdam, Netherlands

Poster: P19.01A Reconciling non-directivity and the counselors’ preference in prenatal counseling
Concurrent Session P19 Genetic counselling/Education/public services
Presence at the poster: Sunday, May 28, 2017, 10.15 - 11.15 hrs, Poster Area and in the concurrent session C03 Best Posters Session, Saturday, May 27, 18.30-20.00 hrs, Cannes

Q1: 4-02-1989, The Hague, Netherlands
Q2: PhD Candidate
Q3: As a psychologist, my interest for people goes further than the science of the brain; I am fascinated by the building blocks of humankind.
Q4: In my presentation I will reveal how the counselors’ own preference for a test influences the patients’ choice, putting the concept of non-directivity in genetic counseling in a new light.
Tessa van Dijk  
Amsterdam, Netherlands

Poster: **P09.001A Mutations in CoA Synthase cause pontocerebellar hypoplasia**
Concurrent Session P09 Neurogenetic and psychiatric disorders
Presence at the poster: Sunday, May 28, 2017, 10.15 - 11.15 hrs, Poster Area and in the concurrent session C03 Best Posters Session, Saturday, May 27, 18.30-20.00 hrs, Cannes

Roel Wouters  
Utrecht, Netherlands

Poster: **P20.02B A duty to hunt for pathogenic mutations? Ethical considerations regarding routine screening of genomic data**
Poster Session P20 Psychological/Ethical/legal issues
Presence at the poster: Sunday, May 28, 2017, 16.45 - 17.45 hrs, Poster Area

Q1: 17 February 1991 in Heerlen, The Netherlands
Q2: I am a PhD candidate in medical ethics, focussing on the responsible introduction of next generation sequencing in oncology.
Q3: I think that genetics, as one of the cornerstones of personalized medicine, has the potential to fundamentally change the way we organize health care in the 21st century. As a young medical doctor, I expect that these developments in genetics will enable me to offer my future patients better treatment options.
Q4: I present an ethical analysis on the question as to whether geneticists should routinely screen genomic sequencing data for a list of pathogenic mutations (also referred to as a duty to hunt). This research provides insights that are relevant for a lot of professionals who are confronted with large quantities of data but ask which parts of these data should be analyzed.

Shachar Zuckerman  
Jerusalem, Israel

Poster: **P19.03C Reproductive experiences, medical concerns and moral attitudes among preimplantation genetic diagnosis (PGD) users: Synthesis of qualitative and quantitative analysis**
Poster Session P19 Genetic counselling/Education/public services
Presence at the poster: Monday, May 29, 2017, 10.15 - 11.15 hrs, Poster Area
Registration and Opening Hours

Opening Hours Registration and Preview Centre
Friday, May 26  14.00 – 19.00 hrs
Saturday, May 27  07.30 – 20.15 hrs
Sunday, May 28  08.00 – 19.30 hrs
Monday, May 29  08.00 – 19.30 hrs
Tuesday, May 30  08.30 – 14.00 hrs

Opening Hours Exhibition
Friday, May 26  CLOSED!
Saturday, May 27  09.30 – 18.30 hrs
Sunday, May 28  09.00 – 17.45 hrs
Monday, May 29  09.00 – 17.45 hrs
Tuesday, May 30  CLOSED!

Badges
Participants should collect name badges from the conference registration desk. As only registered participants will be permitted to attend the scientific sessions, the exhibition and poster areas, you are required to wear your badge when entering and while remaining in the congress venue.

Accompanying persons and exhibitors will also receive badges to allow access to the appropriate areas. Lost badges can be replaced at the registration desk. However, a handling fee of EURO 25.- will be charged.

Cancellations and Refunds
Notice of cancellation had to be made in writing by email or fax to the Congress Office.

The policy for refunding registration fees is as follows:

- Written cancellation received:
  - before April 1, 2017: 75% refund
  - between April 1 and May 7, 2017: 25% refund
  - after May 7, 2017: no refund

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

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

Insurance
By registering to the ESHG 2017 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. The conference fee does not include insurance.

Programme

MobileApp
The “ESHG 2017 Congress” mobile app with full programme and other useful information is available for download in your App- or Playstore.

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

Poster Removal
The organisers cannot assume any liability for loss or damage of posters displayed in the poster area. Removal times for the different groups:

Groups A-C: Monday, May 29, 2017 from 16:45 hrs
Group D: Monday, May 29, 2017: 17:45 – 18:00 hrs (strict)

Posters that will not be removed by Monday, May 29, 2017, 18.00 hrs, will be removed by the staff, will not be kept or mailed to the author after the meeting, but will be discarded.

Live Streaming in the Exhibition Hall
The plenary lecture hall is equipped with a live transmission possibility to the Live stream area in the exhibition. The programme of the room Aarhus will be transmitted to this area during exhibition opening hours.

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

Lunch and Refreshments
Lunch tickets for lunch boxes had to be pre-ordered – they cannot be purchased on site. Please note that lunch tickets are not refundable. Lunch boxes can be picked up from 11.30 – 13.30 at the coffee points in the exhibition. A cash bar is also available in the exhibition area.
**Venue**

**Conference Venue**
BCC – Bella Center Copenhagen
Center Blvd. 5
2300 Copenhagen S, Denmark

**Travelling – Accessibility – Public Transportation**

**From the Airport**

*By taxi – journey time 10 minutes*

You can find taxi stands just outside terminal 3. The trip by taxi from Copenhagen Airport to the BCC takes approximately 10 minutes depending on traffic. It costs between DKK 150 and DKK 200 (EUR 20 – EUR 27).

*By bus – journey time 15 minutes*

The Airport shuttle service operates every 30 minutes between 6am and 11pm and runs between Copenhagen Airport and the BCC-hotels, AC Hotel Bella Sky Copenhagen (which is directly connected to Bella Center Copenhagen) and Crowne Plaza Copenhagen Towers. You can find the schedule on the following website. It costs DKK 15 (EUR 2) each way (to be paid at check-in / check-out at the hotel receptions) and the fifteen seats are filled on a first come, first served principle. At the airport information desk you will find the hotel's iPad with a guide to the shuttle parking spot by Terminal 2 from where you can get on the bus. When departing the hotel you order the shuttlebus in the hotel reception.

*By train and metro – journey time 30 min*

Regional trains also run from the airport to Ørestad Station. Then you can either walk or take the metro (1 stop in direction Vanløse) and walk to the Bella Center from there.

**To the city**

*By Metro – journey time 15 min*

If you take the metro, the center of Copenhagen is just a few minutes away. The Metro line M1 runs between Vanløse and Vestamager. The metro station “Bella Center” is located by Bella Center’s east entrance.

ESHG 2017 is pleased to offer participants with a reduced Travel Pass for the conference.
Unlimited Public Transportation by Bus, Train and Metro. The Travel Pass offers unlimited public transportation in all of Greater Copenhagen. Get it here and receive it as an e-mail and an SMS ticket on your mobile phone.

**Advantages**

- Get the ticket online and make use of the public transportation the moment you arrive in the city
- Airport to city transportation included
- Easy online ordering

Notice: This offer is only valid for online ordering and requires a minimum of 2 days

**How to use**

You do not need to show your ticket (e-mail or SMS) before entering Bus, Train or Metro, but you need to have your ticket available for control checks.

http://www.travelpass.dk/conferences/eshg-2017/

**Car Parking**

There is a parking lot right in front of the conference venue.

**Cloakroom and Luggage**

A cloakroom and luggage storage are available close to the registration area.

**WIFI**

WiFi is available throughout the conference venue. Network ID: eshg2017, password: eshg2017

**Staff**

If you should have any questions, the congress staff (recognizable by a pink lanyard and the usual black polos) will be pleased to help you.

**Conference Policy**

IMPORTANT NOTICE:

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

**Language**

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

**Smoking Policy**

The ESHG 2017 is officially a “No-smoking” Conference. Note that smoking is banned in public buildings and private businesses – including restaurants, pubs, shops, public transport, entertainment venues and workplaces. The only exception from the ban is for establishments with an area less than 40 square metres, which do not serve fresh food.
Social Media Guidelines
The ESHG supports the use of social media around the European Human Genetics Conference to network with your colleagues and friends attending the meeting. Please do however respect the ESHG social media guidelines. The views and opinions posted on ESHG’s social media do not necessarily reflect the views, opinions, or policies of the ESHG, its Board or membership. The ESHG reserves the right to remove comments it deems to be inappropriate.

Movements of Attendees - Improving your future experience
The ESHG is excited about new technologies, especially those enabling us to evaluate and improve the quality and overall experience of the ESHG Conference.

As part of this continuing improvement, some participants will be asked to carry a small pendant. These little tags will allow us to refine our services for the ESHG Conference 2018 in Milan and ensure your needs are fully met.

The magic of counting
The tags enable us to understand the attractiveness of lectures and certain activities during the conference. They simply count visited areas – that’s it.

Don’t worry and enjoy the conference
The counting process in the background works via bluetooth receptors without collecting any sort of personal information – FULLY ANONYMOUSLY. Counting is only done within the lecture halls and the exhibition and poster area (not outside the conference centre) and is solely to improve future ESHG Conferences. And no, we will not be counting visits to the restaurant or the smoking area...

Why are we doing this?
Identifying hot (or not so hot) topics, no matter if during lectures or in the poster area, will help us improve the planning of the programme and contribute to avoiding crowded (or empty) lecture halls in future venues.

Please return the tag!
We kindly ask you to RECYCLE the tag in one of the collection boxes when you exit the congress for the last time, as they will be reused.
Thank you very much for your support and enjoy the conference!

Copenhagen – General Information

Bank services – Money matters
Banks are generally open from Monday to Friday from 09:00 – 17:00, on Thursdays until approximately 18:00 (closed on Saturdays and Sundays). There are multiple bank machines (ATMs) open 24 hours a day throughout the city which accept all major international bankcard. The official currency of Denmark is the Danish Krone (DKK). Major credit cards are widely accepted, but please always check beforehand. When paying by credit card for your shopping, you will be asked to show identification. Please bring your PIN code and have an ID with you all the time, otherwise they may refuse to accept your credit card as payment.
Travel checks and Euro cheques can be cashed at a fee in most of the banks.

Currency
The official currency in Denmark is the Danish Krone (DKK).
1 DKK = 0,13 EUR = 0,12 GBP = 0,14 USD = 0,19 CAD = 16,22 JPY = 0,14 CHF = 0,19 AUD as per date of printing.

V.A.T.
The VAT rate in Denmark is 25%.

Emergency Services
European Emergency Number: 112

Pharmacies – Medical Emergencies
Regular pharmacy opening hours are weekdays from 9.00-17:00 hrs. Obviously a number of pharmacies is open 24/7. A list of 24-hour pharmacies and Emergency Hospitals can be found here.

Safety – Crime
Copenhagen remains a relatively safe, secure city. However, use of common sense is (always) required, as in any large city. Experience has shown that some basic precautionary measures should always be kept in mind in any city:
– Do not carry important items like flight tickets, passports etc. with you when visiting the conference or strolling through the city, leave them in the hotel safe during your stay. Rather carry a Xerox copy of your passport or an identity card with you.
– Try not to carry all documents, money, credit cards and other essential items and valuables in one bag or purse. If it is lost or stolen, everything will be gone and might be difficult to replace on short notice, especially passports and visa to return to your country of residence.
– Take off your name badge when leaving the conference centre.
– In heavily frequented tourist zones and the metro at rush hour, be aware of attempts of scam and pickpocketing.
– Do not respond to anybody unknown to you who comes up to you on the street engaging you in a conversation, no matter how safe they appear to be.
Telephone calls
The country code of Denmark is 45. Denmark does not use city codes. To call abroad, dial 00 before the country code.

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

Time Zone
Copenhagen’s time zone is Central European Summer Time (CEST), GMT+2.

Climate
The average temperatures end of May in Copenhagen are 16°C (high) and 7°C (low). The average number of rainy days is 14 with an average rainfall of 40 mm in May.
Weather forecast for Copenhagen

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

Electricity Supply
The power supply in Denmark is 220 V. Most electric outlets adhere to the continental standard (Schuko). Appliances from North America require a transformer and British ones an adaptor for the two-pin continental plugs in use in Denmark.

Shops
Shops in Copenhagen city centre are normally open as follows:

- Monday – Thursday: 10:00 – 18:00
- Friday: 10:00 – 19:00
- Saturday: 10:00 – 16:00
- Sunday: 12:00 – 16:00

All major credit cards are generally accepted. Some places might charge a fee when accepting foreign credit cards as payment. Note that you are usually required to show an ID when using your credit card.

Eating Out in Copenhagen
One of the main characteristics of Copenhagen is its nightlife and gastronomy. You can choose among all kinds of restaurants for all price ranges. Copenhagen offers a wide range of food markets as well, where you can enjoy good, sustainable street food and drinks.
The opening hours of bars, night clubs and discotheques vary, but the most are usually open until 04:00 hours (some even longer).
For further details on restaurants in Copenhagen, please visit their website.

Tipping
According to Danish law, tips for waiters have to be included in the prices indicated in restaurants. However, opinions on tipping vary. Some sources say it is common to leave up to 10% to the waiter, if you are satisfied with the service. Others say, that it is not customary to tip at all, as wages are adequately high. Hence, you may choose to tip or not at your discretion.

Tourist Information Centres
Copenhagen Visitor Service is situated Vesterbrogade 4, across the street from the main entrance to Tivoli Gardens and just around the corner from Copenhagen Central Station.

Wonderful Copenhagen has a stand in the registration area, where you can get your free city map of Copenhagen and tips for your free time.

Download the ESHG 2017 Conference App

For iOS

For Android
### Registration Fees

**Payment received:**

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<tr>
<th>Registration fees</th>
<th>before March 31, 2017 (reduced rate)</th>
<th>from April 1 to May 7, 2017 (normal rate)</th>
<th>after May 7, 2017 and on site</th>
<th>Day tickets on site</th>
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<tbody>
<tr>
<td>ESHG Members</td>
<td>EUR 300.-</td>
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<tr>
<td>Networking Evening at own expense</td>
<td>EUR 55.-</td>
<td>EUR 35.-</td>
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</table>

Participants/Guests

| Participants                        | EUR 55.-                              | EUR 35.-                                 |                             |                    |

* Applies to MSc./PhD students working towards a degree in human genetics or an associated field. 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 via email to conference(at)eshg.org. Confirmations handed in at a later stage cannot be considered.

† Applies to non-MD/PhD Counsellors.

‡ Applies to undergraduate students. Please provide a copy of a Student’s ID or a confirmation signed by the head of department at the moment of your registration by fax to +43 1 407 82 74 or via email to conference(at)eshg.org. Confirmations handed in at a later stage cannot be considered.

§ Applies to a selected list of countries.

Please see also the General Terms & Conditions for participants: https://2017.eshg.org/index.php/general-terms-and-conditions

### What is covered by the registration fee?

**Participants:**

- Admission to all scientific sessions, exhibition and networking mixer
- Printed programme and other conference material
- Coffee/Tea during breaks from Saturday, May 27 to Tuesday, May 30

**Guests (family members only):**

- Access to the poster exhibition and the networking mixer (no admission to scientific sessions)

### Payment of Registration fees

Payment of Registration fees may be made in cash in Euro or Danish Crowns 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 also been paid to the congress account meeting the according deadlines. Registering without performing an actual payment will automatically set your balance to the fee applicable onsite.

### Cancellations and Refunds

Notice of cancellation had to be made in writing by email or fax to the Congress Office.

The policy for refunding registration fees is as follows:

- Written cancellation received:
  - Before April 1, 2017: 75% refund
  - Between April 1 and May 7, 2017: 25% refund
  - After May 7, 2017: no refund

The cancellation will not be effective until a written acknowledgement from the ESHG Conference Registration Department is received. In the case of over-payment or double payment, refund requests must be made in writing and sent to the ESHG Conference Registration Department by email.

No refunds will be granted for unattended events or early termination of attendance, in case of cancellation of speakers, lack of space in the conference room or any other incidents during the conference, which are beyond the control of the conference organisers.

Participants are requested to make their own arrangements for health and travel insurance. The conference fee does not include insurance.

No exceptions to the refund policy can be made, including health or family issues, however, we welcome substitute delegates at any time. By registering to the ESHG 2017 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. The conference fee does not include insurance.
**INFORMATION NETWORKING EVENTS**

### Opening Networking Mixer

**Saturday, May 27, 2017, 20.00 - 21.30 hrs - Bella Center, Centerhall**

Network with your colleagues at this mixer following the first group of concurrent sessions on Saturday evening. Drinks and small snacks will be offered.

*The networking mixer is free of charge, however admission is only possible for registered participants and registered guests.*

### ESHG Networking Party

**Monday, May 29, 2017, 20.00 hrs - 0ksenhallen**

Join us for a party evening at the “0ksenhallen” with dancing, music entertainment. Entrance fees include food, drinks, (non alcoholic, beer or wine), music entertainment.

**Ticket:** EUR 55.-  
**Students:** EUR 35.-

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

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

Individual arrival and departure, no transfers are provided.

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<td>294</td>
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<tr>
<td>Carl Zeiss</td>
<td>544</td>
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<tr>
<td>Zymo Research Europe</td>
<td>122</td>
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INFORMATION EXHIBITION PLAN

LOCTURE HALLS C

GENERAL SATURDAY SUNDAY MONDAY TUESDAY

COFFEE & CASHBAR

COFFEE & LUNCH

AWARDS

INFORMATION
INFORMATION EXHIBITION

Exhibition & Poster Area - Hall C – Dates & Opening Hours

Friday, May 26: CLOSED
Saturday, May 27: 09.30 – 18.30 hrs
Sunday, May 28: 09.00 – 17.45 hrs
Monday, May 29: 09.00 – 17.45 hrs
Tuesday, May 30: CLOSED

Products & Services Index

The Index of Products and Services may be found in the ESHG 2017 Conference App. Download the App, for iOS or Android, from iTunes App Store and Google Play Store.

Exhibition Catalogue & Corporate Satellites

The Exhibition Catalogue & Corporate Satellites book, in your conference bag, lists exhibitors with further information on the companies and products, as well as invites to the corporate satellites, and their programmes.

Floor Plan – Exhibition & Poster Topics

You will find the floor plan of the Exhibition and Poster Topics on the next page and on a separate A4 sheet in your conference bag.

Posters – Mounting, Viewing & Removal Schedules

Poster presentations will be held in the Exhibition & Poster Area, from May 27 – 30. Poster mounting, viewing and removal times are:

Saturday, May 27, 2017: 09.30 – 18.30 hrs Poster mounting / viewing
Sunday, May 28, 2017: 09.00 – 17.45 hrs Poster viewing
Monday, May 29, 2017: 09.00 – 17.45 hrs Poster viewing
13.30 – 17.45 hrs Poster removal Groups A-C
17.45 – 18.00 hrs Poster removal Group D - Strict

Posters not removed by 18.00 hrs on Monday May 29, will be taken down and will not be stored or sent to authors after the meeting but discarded.

Coffee Breaks, Cash Bar, Lunch

Official coffee breaks, as per the final programme, will be held in the Exhibition hall on Saturday, Sunday and Monday. Also outside the official coffee breaks times there will be free coffee and tea in the exhibition hall. The Cash Bar in the Exhibition hall is open during exhibition opening hours. The menu includes sandwiches, smørrebrød, salads, croissants, warm snacks such as burgers and hot dogs, drinks, special coffees. The menu is available at the Cash Bar. Payment in cash (DKK, EURO) or credit card.

Pre-ordered lunch bags will be available during lunch breaks at the left and right coffee break areas (the ones without the Cash Bar). Lunch bags cannot be ordered on-site.

Lead Retrieval System used by Companies

Many companies will be using a so-called Lead Retrieval System on their stands and at the entrance to their corporate satellites. Note the following please:
- companies using the device will ask permission to scan the barcode on your badge
- this barcode gives this company access to your contact details as follows:
  - name and full postal address
  - e-mail address

Thank you for your understanding and cooperation.

Exhibition Management

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