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Dear Colleagues and Friends,

On behalf of the board of the European Society of Human Genetics, it is my privilege to invite you to the annual European Human Genetics Conference, which in 2019 will take place in the beautiful city of Gothenburg, Sweden, from the 15th to the 18th of June. As a Norwegian I am especially pleased to welcome you to a meeting in Scandinavia that will let you experience the famous Scandinavian mid-summer. If you have time I recommend to stay a few extra days to experience the magnificent Swedish west coast.

The purpose of our annual meetings is threefold: to give you excellent science, to teach you human genetics, and to provide an optimal environment for making new contacts and finding new friends. Our Scientific Programme Committee (SPC) has worked hard to invite the best speakers from various sectors of human genetics, and to select the best abstracts for platform presentations. The scientific content is thus a nice and balanced mixture of invited talks (the concurrent symposia) and selected abstract presentations (the concurrent sessions). Throughout the meeting we also provide educational talks for those of you that are not that scientific, or that need to get updated or get an overview on various topics (that probably means all of you). These educational talks are also given by invited speakers, all with expert general knowledge of their subject.

A unique and usually fondly memorable event of our conferences is the Monday evening party. This is not only arranged for the young but also for those that want to feel young – and those that just want to meet and talk to friends in a relaxed atmosphere. As a bonus you get good food and excellent music – for an affordable and subsidized price.

As you hopefully have noticed, ESHG is everything you need in human genetics, whatever your professional background is. Our exhibitors also enjoy this plurality, showcasing their range of products for both clinical and laboratory work. We hope to see as many of you as possible in Gothenburg, and hope you will return back home with the feeling that the ESHG meeting is a major meeting in human genetics that you really do not want to miss.

Gunnar Houge
President
European Society of Human Genetics

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**Swedish Society of Medical Genetics and Genomics**

Dear Colleagues and Friends,

On behalf of the board of the Swedish Society of Medical Genetics and Genomics, it is my privilege to welcome you to the annual European Human Genetics Conference 2019 in the city of Gothenburg, Sweden.

I am pleased to invite you to this event in Scandinavia and grateful for the work by ESHG and the programme committee that has resulted in a very interesting program with a balanced mix of educational talks and the latest in human genetics, both basic and applied. Thus, we are looking forward to a highly inspirational meeting.

Gothenburg is the largest non-capital in the Nordics and located on the beautiful west coast of Sweden and I hope that you also have a chance to explore it in conjunction with the meeting and, if the weather permit, to enjoy the bright summer evenings. However, one must go much further north in Sweden to experience midnight sun. Finally, the meeting is an opportunity to meet old friends, new colleagues and making new collaborations.

Magnus Burstedt
President of the Swedish Society of Medical Genetics and Genomics
European Society of Human Genetics

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EXHIBITION, SPONSORING AND CORPORATE SATELLITES

ROSE INTERNATIONAL

Exhibition Management and Congress Consultancy bv

Jantie de Roos, Flora van Laer

MCI Amsterdam
Schipluidenlaan 4, 1062 HE
Amsterdam, The Netherlands
E: eshg-hotels@mci-group.com

Hotel Accommodation

MCI Amsterdam
Schipluidenlaan 4, 1062 HE
Amsterdam, The Netherlands
E: eshg-hotels@mci-group.com
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 2020**
Berlin, Germany
June 6 – 9, 2020

**European Human Genetics Conference 2021**
Glasgow, United Kingdom
June 12 – 15, 2021

**European Human Genetics Conference 2022**
Vienna, Austria
June 11 – 14, 2022

CME Credits

The European Human Genetics Conference (ESHG 2019), Gothenburg, Sweden, 15/06/2019-18/06/2019 has been accredited by the European Accreditation Council for Continuing Medical Education (EACCME®) with 27 European CME credits (ECMEC®s). Each medical specialist should claim only those hours of credit that he/she actually spent in the educational activity.

Through an agreement between the Union Européenne des Médecins Spécialistes and the American Medical Association, physicians may convert EACCME® credits to an equivalent number of AMA PRA Category 1 Credits™. Information on the process to convert EACCME® credit to AMA credit can be found at www.ama-assn.org/education/earn-credit-participation-international-activities. Live educational activities, occurring outside of Canada, recognised by the UEMS-EACCME® for ECMEC®s are deemed to be Accredited Group Learning Activities (Section 1) as defined by the Maintenance of Certification Program of the Royal College of Physicians and Surgeons of Canada.

Important Notice

In order to obtain CME credits, participants seeking these credits will need to scan their badges daily at the General Information Desk located at the registration area. Please note that if you miss to scan your badge you will not receive the credits accredited for the day.

Download the ESHG Mobile App!

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 2019 Mobile App**

Do you always want to be up-to-date? The ESHG Mobile App will guide you through the programme day by day or by session type, will make available profiles of speakers and delegates 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.

*Available for iOS and Android in your App and Play Stores. Search for European Society of Human Genetics or ESHG.*

**Young Investigators in Focus**

A workshop ('W03 How to enhance your career - How to present - How to network') on Saturday aims directly at young investigators attending the conference.

To promote attendance of young scientists from Central and Eastern European Countries as well as other regions of the world, the ESHG awarded more than 100 fellowships this year.

All young investigators who have an oral presentation compete for 8 YI awards which will be announced in the award session on Tuesday afternoon. The best scored posters from YI compete for 7 poster awards handed out in the same session.

Young Investigator Award Winners of 2018 have been invited to co-chair a session at this year’s conference. Have a look at the 2019 candidates online and from page 58 onwards.

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.

**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 #eshg2019 #sessionnumber (e.g. #eshg2019 #S01). You may also chat with colleagues via the ESHG Mobile App.

*For all sessions, remember to use the discussion microphones in aisles of the lecture halls.*

**e-Posters & Best Posters**

A number of posters will be presented as e-Posters at e-Poster stations in the exhibition hall. The list of available posters can be viewed on any of these screens. From there, they 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 through the multiple slide posters.

This year, the 30 Best Posters were selected for a short presentation (3 minutes) in two Concurrent Sessions – C15 on Sunday and C23 on Monday. These sessions will take place at the live stream area in the exhibition hall. After the presentation, participants will have the opportunity to approach the authors of each poster for questions at the e-Poster area.

**Live streaming and on-demand webcast of selected sessions**

All Educational Sessions will be available as webcast after the meeting. In case you are interested in a Symposium and a parallel Educational Session, no worries, you can watch the Educational Session at home or whenever you have time.

As usual, the Plenaries on Tuesday as well as the ESHG-ASHG Building Bridges Session will be available as live webcast and as on-demand streaming after the conference.

The following sessions are planned to be available:
- E01 - E14
- PL3 & PL4
- S17

*Note that the actual availability of the 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 livestream area in the exhibition. The programme in Hall C will be transmitted to this area during exhibition opening hours.

**Poster viewing with authors**

Posters will be discussed in 4 different groups, at 10.15 – 11.15 hrs and 16.45 – 17.45 hrs both on Sunday and Monday to offer enough interaction between the authors and the audience.

*All posters will remain on display from Saturday to Monday.*

**IMPORTANT NOTICE**

Please note that taking pictures or filming during the sessions is allowed, unless explicitly requested otherwise by the presenter.
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, and 45 minutes in PL3 + PL4.

Concurrent Symposia (S01 – S20)
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 – E14)
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 – C29)
The most notable and exciting work from all abstracts submitted to the conference will be honored with an oral presentation in these sessions. Presenters are expected to explain their work and answer questions from the audience. Speaking time for concurrent session is 15 minutes including time for discussion.

Poster Viewing with Authors
Posters are numerically the major scientific presentations of the meeting. Most attendees bring a poster showing data and progress with their personal research. Posters offer an excellent opportunity for people interested in a particular topic to meet and exchange ideas and network with other researchers. Posters should NOT be used to advertise a product or service. Like a paper, a poster abstract should detail the focus of the presentation and the way(s) in which it contributes to the body of knowledge in its field. Times marked “Poster Viewing with Authors” should be used for communication and interaction with the poster authors, who are requested to be at their posters at these times. Posters will be on display throughout the 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.

“ELPAG” Track (E03, W02, C07, S01, S14, C14, W09, S06, C22, W16, C29, A1)
The “ELPAG” Track (Ethical, Legal and Psychosocial Aspects in Genetics) has a special focus on the psychosocial impact of genetics and genetic counselling. They are not only intended for Genetic Counsellors and Genetic Nurses, but address issues of relevance to all experts in the field of Human Genetics. All sessions related to this track can be identified with the following tag: ELPAG

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

**Saturday, June 15, 2019**

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<th>HALL C</th>
<th>K2+K3</th>
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<tr>
<td>08.00 - 10.00</td>
<td><strong>E01</strong></td>
<td>New Technologies</td>
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<td>10.00 - 12.00</td>
<td><strong>W01</strong></td>
<td>NGS in the Clinic - Mistakes and Quality Assurance</td>
<td><strong>E02</strong></td>
<td>Epigenetics and cancer</td>
<td><strong>W02</strong></td>
<td>Prenatal Diagnosis</td>
<td><strong>E03</strong></td>
<td>Bridging genomic discoveries and personalized medicine</td>
<td><strong>W03</strong></td>
<td>How to enhance your career - How to network</td>
<td><strong>E04</strong></td>
<td>Gene Expression Resources</td>
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<td>14.00 - 16.00</td>
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<td>Opening Plenary Session</td>
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<td>18.00 - 20.00</td>
<td><strong>C01</strong></td>
<td>Novel diagnostic approaches</td>
<td><strong>C02</strong></td>
<td>3D gene regulation</td>
<td><strong>C03</strong></td>
<td>Neurogenetic and psychiatric disorders</td>
<td><strong>C04</strong></td>
<td>Fertility</td>
<td><strong>C05</strong></td>
<td>Developmental disorders 1</td>
<td><strong>C06</strong></td>
<td>Cellular dysfunctions</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 allowed, unless requested otherwise by the presenter.
### Sunday, June 16, 2019

#### TIME | HALL C | K2+K3 | F1+F2+F3 | F4+F5 | G2+G3 | K1 | H2 | EXHIBITION HALL - LIVE STREAM AREA | A3 | A1 | A5 | A2 | A4
---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
**08.30 - 10.00** | **S01** | **E05** | **S02** | **E06** | **S03** | **S04** | **An update on kidney research** |
08.30 - 10.00 | Updates in psychiatric genetics | The longer the better? Third generation sequencing technologies | Finding the strengths that make cancer cells weak | Pharma-cogenomic testing for personalized medicine | RNA mis-splicing dynamics, diagnosis and treatment |
**10.00 - 10.15** | **Coffee Break / Free Poster Viewing / Exhibition** |
**10.15 - 11.15** | **Poster Viewing with authors and coffee - group A** |
10.15 - 10.30 | Poster Viewing with authors and coffee - group A |
10.30 - 10.45 | Poster Viewing with authors and coffee - group A |
**11.15 - 12.45** | **Lunch Break / Free Poster Viewing / Exhibition** |
**12.45 - 13.00** | **Coffee Break / Free Poster Viewing / Exhibition** |
**13.00 - 14.30** | **Poster Viewing with authors and coffee - group A** |
13.00 - 15.00 | Oxford Nanopore Technologies Satellite |
15.00 - 15.15 | QIAGEN Satellite |
15.15 - 15.30 | Agilent Technologies Satellite |
15.30 - 15.45 | Thermo Fisher Scientific Satellite |
15.45 - 16.00 | Asuragen Satellite |
16.00 - 16.15 | Fabric Genomics Satellite |
**14.30 - 16.30** | **Fruit Break / Free Poster Viewing / Exhibition** |
14.30 - 15.00 | Oxford Nanopore Technologies Satellite |
15.00 - 15.15 | QIAGEN Satellite |
15.15 - 15.30 | Agilent Technologies Satellite |
15.30 - 15.45 | Thermo Fisher Scientific Satellite |
15.45 - 16.00 | Asuragen Satellite |
16.00 - 16.15 | Fabric Genomics Satellite |
**16.30 - 16.45** | **Coffee Break / Free Poster Viewing / Exhibition** |
**16.45 - 17.45** | **Poster Viewing with authors and coffee - group B** |
16.45 - 17.15 | Oxford Nanopore Technologies Satellite |
17.15 - 17.30 | QIAGEN Satellite |
17.30 - 17.45 | Agilent Technologies Satellite |
**17.45 - 19.15** | **Coffee Break / Free Poster Viewing / Exhibition** |
17.45 - 19.15 | Oxford Nanopore Technologies Satellite |
19.15 - 20.45 | ESHG Membership Meeting
## GENERAL PROGRAMME AT A GLANCE - MONDAY

### Monday, June 17, 2019

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<td>S09</td>
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<td>ELPAG Award Lecture</td>
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<td>08.30</td>
<td>Variant interpretation and high-throughput functional assays</td>
<td>Multidimensional nuclear organization</td>
<td>From genome wide association study to mechanisms: fine-mapping</td>
<td>Meiosis: factory of genetic variation</td>
<td>“De novo” developments in epilepsy</td>
<td>Congenital disorders of glycosylation</td>
<td>ELPAG Award Lecture</td>
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<td>10.00</td>
<td>Personalized &amp; Predictive Medicine</td>
<td>Genetic mechanisms in cancer</td>
<td>Therapies</td>
<td>From genome architecture to RNA biology</td>
<td>Neuromuscular and neurodegenerative disorders</td>
<td>Internal organs</td>
<td>Ethical, policy and psychosocial aspects in genomics</td>
<td>Best Posters Session 2</td>
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<td>Dysmorphology II</td>
<td>CopyNumber Variant Interpretation and Classification</td>
<td>Molecular Newborn screening vs. newborn testing</td>
<td>European Reference Networks - What is in it for me?</td>
<td>Opportunistic or non opportunistic genetic screening?</td>
<td>Using the EnsemblVEP for analysing variants in rare and common disease</td>
<td>Pharmacogenomics in practice</td>
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<td>E12</td>
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<td>S16</td>
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<td>16.30</td>
<td>Genome First Testing in Pediatrics</td>
<td>Understanding mutations to detect cancer</td>
<td>Debate: Genomics and the Media</td>
<td>Oligogenic inheritance</td>
<td>Regulatory Landscapes</td>
<td>Methods for genetic epidemiology</td>
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### Satellite Sessions
- Roche Sequencing Solutions Satellite
- Agilent Technologies Satellite
- Sistemas Genómicos Satellite
- Thermo Fisher Scientific Satellite

### Exhibition Hall - Live Stream Area
- A3
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<td>E13</td>
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<tr>
<td>09.00</td>
<td>ESHG-ASHG building bridges: Global collaboration to advance the use of genomics in health</td>
<td>Our genetic history and its phenotypic consequences</td>
<td>Treating rare genetic disease</td>
<td>Genetic innovations in reproductive medicine</td>
<td>Epigenetics and early development</td>
<td>Understanding human disease through animal models</td>
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<td>Mosaics</td>
<td>Bioinformatics and multiomics</td>
<td>Mitochondrial disorders</td>
<td>Developmental disorders 2</td>
<td>Late breaking abstracts</td>
<td>Stakeholder perspectives in cancer genetics</td>
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<td>ESHG Award Lecture</td>
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<td>Award Session</td>
<td>ESHG-SN Citation Awards</td>
<td>Young Investigator Awards</td>
<td>Poster Awards</td>
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<tr>
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<td>Award Session</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 allowed, unless requested otherwise by the presenter.
**General Poster Topics - Technical Information**

**Poster Topics**

P01 Reproductive genetics - Prenatal genetics ..........................................................  P01.01 - P01.94
P02 Sensory disorders (eye, ear, pain) ........................................................................ P02.01 - P02.62
P03 Internal organs & endocrinology (lung, kidney, liver, gastrointestinal) ........ P03.01 - P03.52
P04 Skeletal, connective tissue, ectodermal and skin disorders .........................  P04.01 - P04.76
P05 Cardiovascular disorders ..................................................................................  P05.01 - P05.63
P06 Metabolic and mitochondrial disorders ............................................................  P06.01 - P06.61
P07 Immunology and hematopoietic system ............................................................  P07.01 - P07.33
P08 Intellectual disability .........................................................................................  P08.01 - P08.66
P09 Neurogenetic and psychiatric disorders .............................................................  P09.001 - P09.128
P10 Neuromuscular disorders ..................................................................................  P10.01 - P10.37
P11 Multiple Malformation/anomalies syndromes ................................................... P11.01 - P11.89
P12 Cancer genetics .................................................................................................. P12.001 - P12.155
P13 Basic mechanisms in molecular and cytogenetics ............................................ P13.01 - P13.23
P15 Personalized/predictive medicine - Pharmacogenomics ................................. P15.01 - P15.49
P16 Omics - Bioinformatics .................................................................................... P16.01 - P16.85
P17 Epigenetics - Gene Regulation ........................................................................ P17.01 - P17.43
P18 Genetic epidemiology - Population genetics - Statistical methodology - Evolutionary genetics .................................................................  P18.01 - P18.83
P19 Genetic counselling - Services - Education ....................................................... P19.01 - P19.37
P20 Psychological and social issues in genetics ....................................................... P20.01 - P20.11
P21 Lay beliefs and public understanding of genetics - Access to genetic services P21.01 - P21.09
P22 Ethical issues in genetics ................................................................................... P22.01 - P22.12
P23 Legal implications of advances in genetics ....................................................... P23.01 - P23.05
P24 Other relevant ELPAG issues in genetics .......................................................... P24.01 - P24.07

**Technical Information for Presenters of Posters**

Posters will be on display from: Saturday, June 15, (09.30 hrs) to Monday, June 17 (17.45 hrs)
Poster mounting will be possible on: Saturday, June 15, from 09.30 hrs onwards
Removal will be mandatory on: Groups A-C: Monday, June 17, 2019: 16.45 – 17.45 hrs (strict!)
Group D: Monday, June 17, 2019: 17.45 – 17.50 hrs (strict!)

You can find your poster board number in the ESHG 2019 mobile app (see page 7) or in the ESHG 2019 website programme planner.
Access after Monday, June 17, 17.45 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 members to be at your poster board between:
Poster Group A: 10.15 – 11.15 hrs on Sunday, June 16 for posters with board numbers ending with “A” (e.g. P01.01A)
Poster Group B: 16.45 – 17.45 hrs on Sunday, June 16 for posters with board numbers ending with “B” (e.g. P01.01B)
Poster Group C: 10.15 – 11.15 hrs on Monday, June 17 for posters with board numbers ending with “C” (e.g. P01.01C)
Poster Group D: 16.45 – 17.45 hrs on Monday, June 17 for posters with board numbers ending with “D” (e.g. P01.01D)

If it is not possible for you or one of your group members 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.
Please note that taking pictures is allowed, unless requested otherwise by the presenter.

**Technical Information for Presenters of e-Posters**

Schedule for display and upload
Electronic Posters will be on display from Saturday, June 15 (09.30 hrs) to Monday, June 17 (17.45 hrs).
The upload of the e-Poster file will be possible in the Preview Centre from Friday, June 14 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 upload and view your presentation in the Preview centre not later than 2 hours in advance (30 minutes for the first morning talks or the day prior to your talk if possible).
- The lecture rooms are exclusively equipped with Windows-PCs (no MACs). In case you absolutely need to use your own laptop or notebook, please contact the Preview Centre well in advance of your talk to check compatibility.
- Please bring a USB-key all formatted for Windows® (PC). You may want to carry a second key as a back-up in case there is an insoluble technical problem.
- File Format: Microsoft® PowerPoint 2007™ (or newer) presentation formatted for Windows® (PC) only.
- Preferred Resolution: 1920 x 1080 pixel
- Screen format: 16:9
SCIENTIFIC PROGRAMME
SATURDAY, JUNE 15, 2019
**Programme Saturday, June 15**

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</table>
| 08.00 - 10.00   | New Technologies Session E01  
(See page 44 for details) |       |          |       |       |      |      |
| 10.00 - 10.30   |        |       |          |       |       |      |      |
| 10.30 - 12.00   | Coffee Break / Poster Mounting / Exhibition | W01   | E02     | W02   | E03   | W03  | E04  |
|                 |        | NGS in the Clinic - Mistakes and Quality Assurance  
Organisers: Guis Santen, Helger Yntema, Weronika Gutowska-Ding | Epigenetics and cancer  
Chair: Carla Oliveira | Prenatal Diagnosis  
Organisers: Ida Vogel, Joris Vermeesch, Sam Riedijk | Bridging genomic discoveries and personalized medicine  
Chair: Edward Dove | How to enhance your career - How to present - How to network  
Organiser & Moderator: Roy Sheppard | From genetics to new medicines: The Open Targets Genetics Portal  
Organiser: Maya Ghoussaini |
| 10.30-10.45     | 10.30-10.35 Welcome and presentation of the workshop  
Ida Vogel | 10.35-10.45 Prenatal screening in US anno 2019  
Ronald Wagner, United States | 10.45-10.55 Prenatal screening in Denmark anno 2019  
Olav Bjarn Petersen, Denmark | 10.55-11.05 Prenatal screening in Belgium anno 2019  
Joris Vermeesch, Belgium | 11.05-11.15 Diversity in prenatal screening  
Sam Riedijk, The Netherlands | 11.15-11.35 Panel discussion: Can we ever obtain international guidelines on prenatal diagnosis after 2019?  
11.35-12.00 Discussion with the audience |
| 10.30-11.15     | Cancer Epigenetics: From DNA to RNA modifications  
Francois Fuk, Univ of Brussels, Brussels, Belgium | Preparing for genomic medicine: A real world demonstration of health system change  
Clara Gaff, Melbourne, Australia | Personalised Medicine and clinical practice: What it means for patients and healthcare delivery  
William Newman, Manchester, United Kingdom | Do you brighten a room when you walk in, or when you leave?  
What do your colleagues say about you behind your back? | Using expression data to understand the genetics of disease  
Alexis Battle, Baltimore, United States | Open Targets Genetics: Integrating evidence from genome-wide associations and functional genomics to identify and prioritise drug targets  
The workshop will focus on Open Targets Genetics (genetics.opentargets.org), a portal that uses a wide range of biological resources including NHGRI-EBI GWAS Catalog, UK Biobank, GTEx, pQTL and chromatin interaction features enabling users to access an interactive visualisation of summary statistics plots along with fine-scale mapping, trait co-localisation, and gene prioritisation. Participants will be able to use Open Targets Genetics to visualise and establish links between genes, variants, and diseases, find shared susceptibility loci between different traits, investigate molecular trait-disease colocalisation and prioritise drug targets for their disease of interest.  
Workshop Speakers: Maya Ghoussaini, Ellen Schmidt, Edward Mountjoy, Gareth Peat |
| 11.15-12.00     | Common mistakes and challenges in clinical NGS QC/QA  
Christophe Roos, Eufomatics Oy | New for details) |      |      |      |      |
| 11.15-12.00     | Discussion with statements “How to use NGS mistakes to help patients” | E02.2 Cell-type heterogeneity and systems epigenomics of cancer  
Andrew E. Teschendorff, London, United Kingdom | E02.1 Cancer Epigenetics: From DNA to RNA modifications  
Francois Fuk, Univ of Brussels, Brussels, Belgium | E03.1 Preparing for genomic medicine: A real world demonstration of health system change  
Clara Gaff, Melbourne, Australia | E02.2 Cell-type heterogeneity and systems epigenomics of cancer  
Andrew E. Teschendorff, London, United Kingdom | E02.1 Cancer Epigenetics: From DNA to RNA modifications  
Francois Fuk, Univ of Brussels, Brussels, Belgium |      |
| 12.00           | Lunch Break / Free Poster Viewing / Exhibition |      |      |      |      |      |      |
| 14.00           | Corporate Satellites  
(see page 45 for details) |      |      |      |      |      |      |

**General Information**

- **AWARDS**
- **SATELLITES**
- **MONDAY**
- **TUESDAY**
- **SUNDAY**
- **GENERAL**

**Programme Saturday, June 15**

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<td>08.00 - 10.00</td>
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<td>10.30 - 12.00</td>
<td>Coffee Break / Poster Mounting / Exhibition</td>
<td>W01</td>
<td>E02</td>
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<td>E04</td>
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| 10.30-10.45 | Common mistakes and challenges in clinical NGS QC/QA  
Christophe Roos, Eufomatics Oy |        |       |          |       |       |      |      |
| 10.45-11.00 | Bio-informatic learnings from the 100,000 genomes project  
Michael Muller, Genomics England |        |       |          |       |       |      |      |
| 11.00-11.15 | Presentation of questionnaire results  
11.15-12.00 |        |       |          |       |       |      |      |
| 12.00 | Lunch Break / Free Poster Viewing / Exhibition |       |       |          |       |       |      |      |
| 14.00 | Corporate Satellites  
(see page 45 for details) |       |       |          |       |       |      |      |
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<th>TIME</th>
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| 14.00 - 14.30 | **PL0**  
**Welcoming Address**  
Live streamed in room K2+K3  
Chairs: Gunnar Houge, Joris Veltman |
| 14.30 - 16.00 | **PL1**  
**Opening Plenary lecture**  
Live streamed in room K2+K3  
Chairs: Gunnar Houge, Joris Veltman |
| 14.30 | **PL1.1**  
**Genetic epidemiology of colorectal cancer - from discovery to prevention**  
Ulrike Peters, Seattle, United States |
| 15.00 | **PL1.2**  
**Single Cell transcriptional analysis of early human embryo development and stem cells**  
Frederik Lanner, Stockholm, Sweden |
| 15.30 | **PL1.3**  
**Targeted therapy in patients with PIK3CA-related overgrowth syndrome**  
Guillaume Canaud, Necker Enfants Malades hospital, Paris, France |
| 16.00 - 16.30 | Fruit Break / Free Poster Viewing / Exhibition |
| 16.30 - 18.00 | **PL2**  
**'What's New?' Highlight Session**  
Live streamed in room K2+K3  
Chairs: Gunnar Houge, Joris Veltman |
| 16.30 | **PL2.1**  
**The single-cell transcriptional landscape of mammalian organogenesis**  
Malte Spielmann, Max Planck Inst for Molecular Genetics, Berlin, Germany |
| 16.45 | **PL2.2**  
**Chromatin 3D interactions mediate genetic effects on gene expression**  
Olivier Delaneau, Dept of Computational Biology, Univ of Lausanne, Lausanne, Switzerland |
| 17.00 | **PL2.3**  
**Insights from the largest genetic study of sporadic and recurrent miscarriage**  
Triin Laisk, Dept of Obstetrics and Gynecology, Univ of Tartu, Tartu, Estonia |
| 17.15 | **PL2.4**  
**Discovery and characterisation of 49 novel genetic disorders from analysing de novo mutations in 31,058 parent child trio exomes**  
Joanna Kaplanis, Wellcome Sanger Inst, Cambridge, United Kingdom |
| 17.30 | **PL2.5**  
**Loss of MAENLI, a newly characterized lncRNA, results in limb specific inactivation of EN1 and a dorsal dimelia limb phenotype**  
Lila Allou, RG Development & Disease, Max Planck Inst for Molecular Genetics, Berlin, Germany |
| 17.45 | **PL2.6**  
**Whole-genome sequencing of rare disease patients in a national healthcare system**  
Lucy Raymond, Cambridge Inst for Medical Res, Cambridge, United Kingdom |
| 18.00 - 18.30 | Coffee Break / Free Poster Viewing / Exhibition |

Presentations highlighted by a grey background are from Young Investigator Award finalists. Institute, city and country refer to the affiliation of the presenting author.

**Late Programme Changes**

All contents are up-to-date as per date of printing.

For changes in the scientific programme which occurred after the printing deadline, please consult the website: [https://2019.eshg.org/index.php/programme/late-programme-changes/](https://2019.eshg.org/index.php/programme/late-programme-changes/)
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| A national approach to rapid genomic diagnosis in acute paediatrics  
Zornitza Stark, Victorian Clinical Genetics Service, Melbourne, Australia |
| C01.1     |         |       |          |       |       |      |       |
| C01.2     |         |       |          |       |       |      |       |
| Effectiveness of integrated interpretation of exome and corresponding transcriptome data in detecting splicing variants of recessive disorders  
Mamiko Yamada, Keio Univ Sch of Med, Shinyaiku, Japan |
| C02       |         |       |          |       |       |      |       |
| CRISPR-engineered serial genomic inversions lead to tissue-specific architectural stripes, ectopic gene expression and congenital limb malformations  
Andreas Magg, Max Planck Inst for Molecular Genetics, Berlin, Germany |
| C02.1     |         |       |          |       |       |      |       |
| C02.2     |         |       |          |       |       |      |       |
| Functional dissection of TADs reveals non-essential and instructive roles in regulating gene expression  
Daniel M. Ibrahim, Max Planck Inst for Molecular Genetics, Berlin, Germany |
| C02.3     |         |       |          |       |       |      |       |
| C03       |         |       |          |       |       |      |       |
| Neurogenetic and psychiatric disorders  
Chairs: Olaf Riss, Olaud Raudning |
| C03.1     |         |       |          |       |       |      |       |
| Intrinsic expansion of an ATTTC pentamer in the STARD7 gene underlies Familial Adult Myoclonic Epilepsy linked to chromosome 2 (FADE2).  
Josef Gecc, The Univ of Adelaide, Adelaide, Australia |
| C03.2     |         |       |          |       |       |      |       |
| SINEUPs technology: a new route to possibly treat haploinsufficiency-induced Epilepsy and Autism Spectrum Disorders (ASDs)  
Michele Arnoldi, Ctr for Integrative Biology, Univ of Trento, Trento, Italy |
| C04       |         |       |          |       |       |      |       |
| Fertility  
Chairs: Danja Plasjeska-Karanfilska, Permin Tarring |
| C04.1     |         |       |          |       |       |      |       |
| Ectopic expression of CCG repeats leads to impaired response to gonadotropin hormones and reduced fertility with age in a mouse model of the FMRI premutation  
David L. Nelson, Duncan Neurological Res Inst, Houston, United States |
| C04.2     |         |       |          |       |       |      |       |
| Proteomics and single-cell RNA analysis of Akap4-knockout mice model the indispensable role of Akap4 in spermatogenesis  
Na Li, Guangzhou Inst of Pediatrics, Guangzhou Women and Children's Medical Ctr, Guangzhou Medical Univ, Guangzhou, China |
| C04.3     |         |       |          |       |       |      |       |
| C05       |         |       |          |       |       |      |       |
| Developmental disorders 1  
Chairs: Cecilie Rustad, Sergio Sousa |
| C05.1     |         |       |          |       |       |      |       |
| Human and mouse gene essentiality screens allow to identify candidate genes for developmental disorders  
Violeta Munoz Fuentes, EMBL-EBI, Cambridge, United Kingdom |
| C05.2     |         |       |          |       |       |      |       |
| C06       |         |       |          |       |       |      |       |
| Cellular dysfunctions  
Chairs: Rune Østern, Birute Tumiene |
| C06.1     |         |       |          |       |       |      |       |
| DTYMK deficiency is the cause of a novel vanishing brain disease  
Jo M. Vanoeven, Dept of Clinical Genetics, Maastricht Univ Medical Ctr+, Maastricht, Netherlands |
| C06.2     |         |       |          |       |       |      |       |
| DEGS1 Mutation causes sphingo-lipidosis  
Vadin A. Dolgin, The Morris Kahn Lab of Human Genetics, Natl Inst for Biotechnology in the Negev and Faculty of Health Sciences, Ben-Gurion Univ of the Negev, Beer Sheva, Israel |
| C07       |         |       |          |       |       |      |       |
| Gene editing and reproduction  
Chairs: Angus John Clarke, Danya Vears |
| C07.1     |         |       |          |       |       |      |       |
| Experts' opinions on genome editing in humans: a collective construction of a disruptive technology  
Virginia Romano, Uppsala Univ, Uppsala, Sweden |
| C07.2     |         |       |          |       |       |      |       |
| How will new reproductive genetic technologies change genetically at-risk couples' reproductive decision making? Views on NIPD and genetic modification  
Ivy van Dijke, Amsterdam UMC, Amsterdam Reproduction and Development Res Inst, Amsterdam, Netherlands |
| C07.3     |         |       |          |       |       |      |       |
| Perspectives of a Genetic Disease Community and Genetic Professionals on Germline Gene Editing  
Vince L. Bonham, Natl Human Genome Res Inst, Bethesda, United States |
| 19.00     |         |       |          |       |       |      |       |
| Chromosome conformation capture (HiC) combined with whole genome sequencing for the detection and functional interpretation of complex genomic rearrangements in developmental disease  
Uira S. Melo, MPI, Berlin, Germany |
| C03.3     |         |       |          |       |       |      |       |
| Brain somatic mutations associated with aging contribute to dysregulation of Tau phosphorylation in Alzheimer's disease  
Jun Sung Park, Biomedical Science and Engineering Interdisciplinary Program, Korea Advanced Inst of Science and Technology (KAIST), Daejeon, Korea, Republic of Korea |
| C03.4     |         |       |          |       |       |      |       |
| Discovery of selfish mutations expanding in the male germline with duplex sequencing  
Renato Salazar, Inst of Biophysics, Johannes Kepler Univ, Linz, Austria |
| C05.3     |         |       |          |       |       |      |       |
| Towards the treatment of Cantu syndrome  
Helen I. Roessler, Dept of Genetics, Ctr for Molecular Med, Univ Medical Ctr Utrecht, Utrecht, Netherlands |
| C05.4     |         |       |          |       |       |      |       |
| C06.3     |         |       |          |       |       |      |       |
| MAGT1-CDG vs. XMEM: two faces of a novel glycosylation disorder  
Eline Blommaert, Dept of Human Genetics, KU Leuven, Leuven, Belgium |
| C06.4     |         |       |          |       |       |      |       |
| C07.3     |         |       |          |       |       |      |       |
| Perspectives of a Genetic Disease Community and Genetic Professionals on Germline Gene Editing  
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<td>C01.4</td>
<td>Expanding Next Generation Phenotyping on clinical notes and hand radiographs</td>
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<td>Guy Nadav, FDN Inc, Boston, United States</td>
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<td>C01.5</td>
<td>Using UK Biobank to assess the pathogenicity, penetrance and expressivity of monogenic disease variants</td>
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<td>Caroline F. Wright, Inst of Biomedical and Clinical Science, Exeter, United Kingdom</td>
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<td>C02</td>
<td>3D gene regulation</td>
<td>19.30</td>
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<td>C02.4</td>
<td>Characterization of GB2 cis-regulatory elements in the DFN1B1 locus</td>
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<td>Anais Le Nabec, Univ Brest, Inserm, EFS, UMR 1078, GGB, Brest, France</td>
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<td>C02.5</td>
<td>Novel insights into molecular mechanisms in X-linked dystonia-parkinsonism (XDP)</td>
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<td>Jelena Pozojevic, Section for Functional Genetics, Inst of Human Genetics, Lubeck, Germany</td>
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<td>C03</td>
<td>Neurogenetic and psychiatric disorders</td>
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<td>19.45</td>
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<td>C03.4</td>
<td>Loss of neutral sphingomyelinase-3 (SMPD4) links neurodevelopmental disorders to cell cycle and nuclear envelope anomalies</td>
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<td>Daphne J. Smits, Dept of Clinical Genetics, Erasmus Univ Medical Ctr, Rotterdam, Netherlands</td>
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<td>C03.5</td>
<td>Exploring the impact of CHD2 mutations on DNA double strand break (DSB) repair via non-homologous end joining (NHEJ) using Cas9 and Nanopore sequencing in human induced pluripotent stem cells (hiPSC)</td>
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<td>Ian Tully, Neuroscience and Mental Health Res Inst, Cardiff, United Kingdom</td>
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<td>C03.6</td>
<td>De novo mutations in TAO1 cause neurodevelopmental disorders</td>
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<td>Marija Dubovic Mahlow, Inst of Neurogenetics, Lubeck, Germany</td>
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<td>Fertility</td>
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<td>C04.4</td>
<td>Exome sequencing reveals de novo mutations and deletions in severe idipathic male infertility</td>
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<td>Manon S. Oud, Dept of Human Genetics, Radboudumc, Nijmegen, Netherlands</td>
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<td>C04.5</td>
<td>A restricted spectrum of KMT2D variants cause a multiple malformations disorder distinct from Kabuki syndrome</td>
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<td>Sara Cuvertino, Div of Evolution and Genomic Sciences, Sch of Biological Sciences, Faculty of Biology, Med, and Health, The Univ of Manchester, Manchester, United Kingdom</td>
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<td>C05</td>
<td>Developmental disorders 1</td>
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<td>C05.4</td>
<td>Deciphering altered inhibitor G-protein signaling in the cardiac dysfunction syndrome underlying Intellectual Developmental Disorder with Cardiac Arrhythmia (IDDOCA) syndrome</td>
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<td>Pasqueline De Nittis, Ctr for Integrative Genomics, Lausanne, Switzerland</td>
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<td>C06</td>
<td>Cellular dysfunctions</td>
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<td>C06.4</td>
<td>Implication of LRPS variants in familial hypercholesterolemia</td>
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<td>Youngha Gheleb, LUTS INSERM U1148, Paris, France</td>
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<td>C07</td>
<td>Gene editing and reproduction</td>
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<td>C07.4</td>
<td>National implementation of genome-wide non-invasive prenatal testing as a first-tier screening test in the Netherlands: evaluation of women’s perspectives</td>
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<tr>
<td>Karuna R. van der Mieij, Dept of Clinical Genetics, Amsterdam UMC, Vrije Univ Amsterdam, Amsterdam, Netherlands</td>
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Presentations highlighted by a grey background are from Young Investigator Award finalists.
SCIENTIFIC PROGRAMME
SUNDAY, JUNE 16, 2019
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<td>- 10.00</td>
<td>ELPAG</td>
<td>The longer the better? Third generation sequencing technologies Chair: Zeynep Tumer Finding the strengths that make cancer cells weak Chair: Jose Luis Costa Hildegunn Vetti Pharmacogenomic testing for personalized medicine Chair: Vita Dolzan RNA mis-splicing dynamics, diagnosis and treatment Chairs: Siren Bergland Elfride de Baere An update on kidney research Chairs: Jens Michael Hertz Charlotte von der Lippe</td>
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<td>Common and rare variants in psychiatric disorders Chairs: Ramona Moldovan Enza Maria Valente How long do we need? The relative value of emerging sequencing technologies in genomic medicine Chair: Mike Talkowski, New York, United States Tumour suppressor function restoration: role in tumour reversal and response to treatment Scott Lowe, New York, United States Integrating pharmacogenomics into personalized drug treatment Magnus Ingelman-Sundberg, Stockholm, Sweden Minor spliceosome and disease Mikko J. Frilander, Univ of Helsinki, Helsinki, Finland Polycystic kidney disease and ciliopathies Carsten Bergmann, Dept of Med, Univ Hosp Freiburg, Freiburg, Germany</td>
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<td>S01.2</td>
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<td>S02.2</td>
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<td>Genome wide patterns of structural mutation and selection guide for identification of cis-regulatory variants E05.2 Delineating the structure of chromosome rearrangements using multiple WGS technologies Chairs: Mike Talkowski, New York, United States Myc in Cancer: targeting an engine, not a driver Gerard Evan, Cambridge, United Kingdom Pharmacogenomics based personalized drug treatment across world populations Andrea Gaedigk, Children's Mercy Kansas City, Kansas City, United States Dynamic mutations and RNA mis-splicing in disease Maurice Swanson, Univ of Florida, Gainesville, United States New insights in the genetics of hereditary nephrotic syndromes Corinne Antignac, Paris, France</td>
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<td>Updates in clinical applications of psychiatric genetics Jehannine Austin, Univ of British Columbia, Vancouver, Canada Immunotherapy in cancer Karine Serre, Inst de Medicina Moleculares, ILM, Lisbon, Portugal Restoring splicing defects by antisense oligonucleotide therapy Rob Collin, Nijmegen, Netherlands CRISPR Gene Editing in human organoids for inherited renal diseases Benjamin Freedman, Seattle, United States</td>
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<td>Coffee Break / Free Poster Viewing / Exhibition Poster Viewing with authors and coffee (Group A) Lunch break / Free Poster Viewing / Exhibition Corporate Satellites (see page 45 for details)</td>
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### Programme Sunday, June 16

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<td>13.00 - 14.30</td>
<td>C08</td>
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<td>C12</td>
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<td>C14</td>
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<td>C08.1</td>
<td>Increased risk at first trimester screening: trisomies are not everything, but the risk for an atypical chromosomal aberration is low. Experiences from the Swedish Pregnancy Register</td>
<td>C09.1</td>
<td>Germine genetic variation drives the somatic landscape of tumors</td>
<td>Chairs: Noah Zaiten, UCL, Los Angeles, United States</td>
<td>C10.1</td>
<td>Sequence variants associated with resistant hypertension mechanisms affecting potassium levels</td>
<td>Chairs: Vinicio Tragante, deCODE Genetics/Amgen, Reykjavik, Iceland</td>
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<td>C08.2</td>
<td>C09.2</td>
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<td>ESHG 2019</td>
<td>Gothenburg, Sweden</td>
<td><a href="http://www.eshg.org">www.eshg.org</a></td>
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<td>13.45</td>
<td><strong>C08.4</strong></td>
<td>Systematic evaluation of prenatal and pediatric diagnostic yields from whole-genome sequencing in 8,954 individuals</td>
<td>Chelsea Lowther, Ctr for Genomic Med, Boston, United States</td>
<td><strong>C09.5</strong></td>
<td>Non-invasive prenatal diagnosis of sickle cell disease by next generation sequencing of cell-free DNA</td>
<td>Julia C van Campen, Genetics Labs, Guy's and St. Thomas' NHS Fdn, London, United Kingdom</td>
<td><strong>C10.5</strong></td>
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<tr>
<td>14.00</td>
<td><strong>C09.6</strong></td>
<td>Application of genomics and cognitive technology in precision oncological medicine</td>
<td>Gloria Ribas, Medical Genetics Unit, Sistemas Genomicos, Valencia, Spain</td>
<td><strong>C10.6</strong></td>
<td>Genetics of human plasmalipidome and its link to cardiovascular diseases</td>
<td>Rubina Tabassum, Inst for Molecular Med Finland, HI-LIFE, Univ of Helsinki, Helsinki, Finland</td>
<td><strong>C10.7</strong></td>
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<tr>
<td>14.15</td>
<td><strong>C08.6</strong></td>
<td>Prevalence and clinical outcome of mosaicism in uncultured chorionic villus samplings after chromosomal microarray</td>
<td>Ida Charlotte Bay Lund, Dept of Clinical Genetics, Aarhus Univ Hosp, Aarhus N, Denmark</td>
<td><strong>C11.5</strong></td>
<td>Defective DNA polymerase a-primase leads to X-linked intellectual disability associated with severe growth retardation, microcephaly and hypogonadism.</td>
<td>Hilde Van Esch, Ctr for Human Genetics, LEUVEN, Belgium</td>
<td><strong>C11.6</strong></td>
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<tr>
<td>14.30</td>
<td>Fruit break / Free Poster Viewing / Exhibition</td>
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<tr>
<td>13.00 - 14.30</td>
<td><strong>C15</strong> Best Posters Session 1</td>
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<tr>
<td></td>
<td>Chairs: Joris Veltman, Alexandre Reymond</td>
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<tr>
<td><strong>P03.05A</strong></td>
<td>Gastrointestinal dysfunction in autism spectrum disorder: New insights from the Foxp1+/--mouse with altered gut motility and achalasia</td>
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<td>Gudrun Rappold, Dept of Human Molecular Genetics, Inst of Human Genetics, Univ of Heidelberg, Heidelberg, Germany</td>
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<tr>
<td><strong>P04.41A</strong></td>
<td>Identification and characterization of microRNA-149, a candidate for orofacial clefting.</td>
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<td>Ronja Hollstein, Inst of Human Genetics, Univ of Bonn, Sch of Med &amp; Univ Hosp Bonn, Bonn, Germany</td>
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<tr>
<td><strong>P06.60C</strong></td>
<td>High-throughput metabolomics for early detection of individuals at increased risk for type 2 diabetes</td>
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<td>Jenni Hällfors, Nightingale Health Ltd., Helsinki, Finland</td>
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<td><strong>P07.09A</strong></td>
<td>Increasing fetal hemoglobin by genetic editing the cells of sickle cell disease patients</td>
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<td>Kirmo Wartiovaara, Clinical Genetics, Helsinki Univ Hosp, Helsinki, Finland</td>
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<tr>
<td><strong>P09.012C</strong></td>
<td>Anorexia nervosa genome-wide association study identifies eight loci and implicates psychiatric and metabolic origins</td>
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<td>Christopher Hübel, Karolinska Instt, Stockholm, Sweden</td>
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<tr>
<td><strong>P09.028C</strong></td>
<td>Characterizing cellular heterogeneity of de novo mutations in autism spectrum disorders</td>
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<td>Abdulrahman Y. Ali, Mohammed Bin Rashid Univ of Med and Health Sciences, Dubai, United Arab Emirates</td>
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<td><strong>P10.39C</strong></td>
<td>Analysis of DNA tandem repeats in ALS from Whole Genome Sequencing: Role of FRA10Ac1 gene repeat expansion in ALS</td>
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<td>Lucia Corrado, Univ of Eastern Piedmont UPO, NOvara, Italy</td>
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<td><strong>P13.13A</strong></td>
<td>Gain-of-function mutations in KCNN3 encoding the small-conductance Ca2+-activated K+ channel SK3 cause Zimmermann-Laband syndrome</td>
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<td>Pauline E. Schneeberger, Inst of Human Genetics, Univ Medical Ctr Hamburg-Eppendorf, Hamburg, Germany</td>
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<td><strong>P14.008C</strong></td>
<td>Variants with reduced variant fractions in NGS-based germline diagnostics for hereditary breast and ovarian cancer</td>
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<td>Mirjam Larsen, Ctr for Hereditary Breast and Ovarian Cancer, Ctr for Integrated Oncology (DO), Univ of Cologne, Faculty of Med and Univ Hosp Cologne, Cologne, Germany</td>
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<td><strong>P16.46B</strong></td>
<td>LOY Associated Transcriptional Effect (LATE) in immune cells measured by single cell RNAseq and bulk RNAseq</td>
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<td>Jonas Mattisson, Dept of Immunology, Genetics and Pathology, Uppsala Univ, Uppsala, Sweden</td>
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<td><strong>P16.028D</strong></td>
<td>Gabriella Miller Kids First Data Resource Center: Harmonizing genomic and clinical information to support childhood cancer and structural birth defect research</td>
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<td>Yiran Guo, Children's Hosp of Philadelphia, Philadelphia, United States</td>
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<td><strong>P16.02B</strong></td>
<td>Genetic dysregulation of gene expression and splicing during a ten-year period of human aging</td>
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<td>Brunilda Balliu, Dept of Bioinformatics, UCLA, Los Angeles, United States</td>
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<tr>
<td><strong>P17.13B</strong></td>
<td>An integrated chromatin accessibility and transcriptome landscape of human pre- and post-implantation embryos</td>
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<td>Zhouchun Shang, BGI-Shenzhen, Shenzhen, China</td>
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<td><strong>P17.40A</strong></td>
<td>Disease interpretation of regulatory variants with GeneHancer</td>
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<td>Simon Fishilevich, Weizmann Inst of Science, Rehovot, Israel</td>
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<tr>
<td><strong>P18.67C</strong></td>
<td>The genetics of sleep traits and their links with disease</td>
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<td>Samuel E. Jones, Univ of Exeter Medical Sch, Exeter, United Kingdom</td>
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The 30 Best Posters were selected for a short presentation during two concurrent sessions. In this session, the best posters from different topics will be presented. Please refer to page 14 for more information about the topics. The poster authors will have 3 minutes each to present their most important findings.

After the presentation of all posters (approximately at 13.45 hrs), the authors and the audience will proceed to the electronic posters next to the live stream area for discussion with the authors for the remainder of the session.
16.00 – 16.30

**W05**
Exome sequencing and variant interpretation
Organisers: Christian Gilissen Katlin Samocha

**W06**
Dysmorphology
Organisers: Jill Clayton-Smith Sofia Douzgou Dian Donnai

**W07**
UCSC Genome Browser
Organiser: Robert Kuhn

**W08**
Investigating genotype-phenotype data using the GWAS Catalog
Organisers: Laura Harris Daniel Suveges

**W09**
ELPAG

**W10**
Genomics Quiz
Organisers: Robert Hofstra Julie Mc Guinness Moderator: Roy Sheppard

15.00-15.25
ERC Starting Grant Lude Franke, The Netherlands
15.25-15.50
ERC Consolidator Grant Bart Loey, Belgium
15.50-16.15
ERC from the perspective of LS panels Konstantina Topourdou, Belgium
16.15-16.30
Q&A Session

---

Although exome sequencing is now routinely available both for research and clinical purposes, the interpretation of identified variants remains a major challenge. In this workshop, we will address the available public bioinformatics resources that can help in interpreting variants from exome sequencing, and illustrate their importance by real-life examples.

15.00-15.05
Welcome and opening remarks Christian Gilissen
15.05-15.30
Using gnomAD for variant interpretation Katlin Samocha
15.30-15.50
Predicting the effect of splicing site variants Jeremy McRae
15.50-16.10
Variant interpretation using protein structure and interactions James Stephenson
16.10-16.30
Analysis of CNVs from exome data Ralph Pfundt

---

We invite all those working in the field of syndrome diagnosis, and those who wish to learn more about the art and science of Dysmorphology, to attend this session. Please participate by bringing along short PowerPoint presentations of your distinctive unsolved cases or your instructive solved cases to one of the two Dysmorphology workshops. Even if you do not have cases to bring, we also encourage workshop attendees to share their knowledge of dysmorphology, and broaden genetic mechanisms by participation in the case discussions. As we move further into the genomic era we anticipate more discussion around variant interpretation and so we would also welcome experts in this area to join us. We also welcome “solved” cases that you may have presented as unknowns at the ESHG in previous years, but where you now have an answer. These are very interesting and instructive for the audience.

---

The UCSC Genome Browser continues to expand its feature set and data. The workshop will describe our representation of pre-computed CRISPR guides (including off-target locations), the Genome Aggregation Database (gnomAD) and new data formats:

1) interact, for display of physical interaction data (e.g., SC, Hi-C) or conceptual relationships (e.g., enhancers)
2) barChart, for aggregating data from multiple experiments into a simple, single display

The new Track Collection feature allows multiple RNA-seq datasets to be configured together; to be superimposed on a single axis and to be subtracted on the fly to show the difference between two datasets.

---

Our workshop will cover an introduction to the GWAS Catalog, including the scientific background, the web-based search tools, and programmatic access via our RESTful API. We will include hands-on demonstrations covering different methods for accessing GWAS Catalog data, focusing on the most common use-cases. Participants who bring their own laptops will have a chance to try out the GWAS Catalog online; those wishing to try out the API during the session must have a modern browser such as Chrome or Firefox. We anticipate that some participants, who bring their own laptops, will find it useful to have a command line interface (e.g., Unix terminal).

---

This workshop will explore psychotherapeutic elements in genetic counseling casework that highlight Dr. Seymour Kessler’s clinical scholarship. Attendees will learn about addressing patient suffering: distinctions between shame and guilt, when to execute personal scrutiny, transference and counter-transference, and family transitions. We will discuss the role of the genetic counselor regarding end of life options for those affected with neurodegenerative disease.

---

In an exciting new experiment, 2 teams as well as the audience will test their knowledge of the ESHG, genetics and Gothenburg, using multiple choice questions, performance acts and audience participation, in an hopefully entertaining and educative quiz.
### PROGRAMME SUNDAY, JUNE 16

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<td>17.45 - 19.15</td>
<td><strong>S05</strong></td>
<td>Genome editing</td>
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<td><strong>S05.1</strong></td>
<td>CRISPR single-cell sequencing: Toward functional biology in high throughput</td>
<td>Christoph Bock, CeMM Res Ctr for Molecular Med of the Austrian Acad of Sciences, Vienna, Austria</td>
<td>S05.1</td>
<td>Current understanding of psychiatric genetics research and services amongst mental health service users and their families</td>
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<td>Mosaic loss of chromosome Y (LOY) in leukocytes: from discovery to impact</td>
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<td>17.45</td>
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<td>CRISPR single-cell sequencing: Toward functional biology in high throughput</td>
<td>Christoph Bock, CeMM Res Ctr for Molecular Med of the Austrian Acad of Sciences, Vienna, Austria</td>
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<td>Genetic profiling in primary care: triggers and impact on risk-reducing behaviour</td>
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<td>18.15</td>
<td><strong>S05.2</strong></td>
<td>Therapeutic applications of genome editing to prevent diseases</td>
<td>Kiran Musunuru, Cambridge, United States</td>
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<td>Genetic profiling in primary care: triggers and impact on risk-reducing behaviour</td>
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<td>Genetic profiling in primary care: triggers and impact on risk-reducing behaviour</td>
<td>Nadeem Qureshi, Notingham, United Kingdom</td>
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<td>Mosaic chromosome Y loss, ageing and cancer risk</td>
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<td><strong>S05.3</strong></td>
<td>Advances in therapeutic CRISPR/Cas9 genome editing</td>
<td>Gerald Schwank, IMHS, Zurich, Switzerland</td>
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<td>What will this genetic result mean for my baby?</td>
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<td>19.15 - 20.30</td>
<td><strong>H2</strong></td>
<td>ESHG Membership Meeting</td>
<td>Corporate Satellites</td>
<td>(see page 45 for details)</td>
<td>Corporate Satellites</td>
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### Late Programme Changes

All contents are up-to-date as per date of printing.

For changes in the scientific programme which occurred after the printing deadline, please consult the website: [https://2019.eshg.org/index.php/programme/late-programme-changes/](https://2019.eshg.org/index.php/programme/late-programme-changes/)
SCIENTIFIC PROGRAMME
MONDAY, JUNE 17, 2019
By joining the ESHG, you will gain access to a range of benefits and an international network of specialists, who promote and encourage collaboration and exchange of information within Human and Medical Genetics and Genomics in Europe and the world.

Benefits of joining ESHG as a Member include:

- Subscription to the EJHG, the European Journal of Human Genetics
- Discounted joint memberships with many national societies
- Discounted registration fees for the European Congress of Human Genetics
- Preferential treatment for fellowships at a number of courses
- ESHG Newsletters
- Access to the member area of the ESHG with the membership directory

You may choose between regular, online, collective or joint membership.

Visit the ESHG booth #342 today and enroll as a member or join online at www.eshg.org
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<td>Variant interp-</td>
<td>Multidimensional nuclear organization</td>
<td>From genome wide association study to mechanisms: fine-mapping</td>
<td>Meiosis: factory of genetic variation</td>
<td>De novo developments in epilepsy</td>
<td>Congenital disorders of glycosylation</td>
<td>ELPAG Award Lecture</td>
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<td>From association to causal variant(s): statistical methods for fine-mapping</td>
<td>Genetic diversity and its unexpected impacts on recombination, genome evolution, speciation and fertility in mammals</td>
<td>De novo variants in neurodevelopmental disorders with epilepsy</td>
<td>Genetic heterogeneity in CDGs: where are the patients?</td>
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<td>Leveraging genome-wide association studies in diverse populations to fine-map complex human trait loci</td>
<td>Meiotic recombination, gene conversion and mutation</td>
<td>Parental Mosaicism in “De Novo” Epileptic Encephalopathies</td>
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<td>Ohad Medalia, Univ of Zurich, Zurich, Switzerland</td>
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<td>Irene Tiemann-Boege, Johannes Kepler Univ, Linz, Austria</td>
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<td>Brain somatic mutations in malformations of cortical development with epilepsy</td>
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<td><strong>Mutations in the Golgi protein GBF1 as a novel cause of distal</strong></td>
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<td><strong>Rare heterozygous deleterious GDF6 variants in patients with</strong></td>
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<td><strong>Clinical applicability of the 313-SNP based polygenic risk score</strong></td>
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<td><strong>Safe and efficient personalised TALEN- and CRISPR/Cas9-based</strong></td>
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<td><strong>Recessive mutations in muscle-specific isoforms of FXR1 cause</strong></td>
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<td>High polygenic risk contributes to an early disease onset in common cardiometabolic diseases and cancers</td>
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### PROGRAMME MONDAY, JUNE 17

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<th>TIME</th>
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| 13.00 - 14.30 | **Best Posters Session 2**  
**Chairs:** Joris Veltman, Gunnar Houge  
**P02.56B** Biallelic sequence and structural variants in RAX2 are a novel cause for autosomal recessive inherited rod-dominated retinal disease  
**Stijn Van de Sompele,** Ctr for Medical Genetics, Ghent Univ and Ghent Univ Hosp, Ghent, Belgium  
**P02.32B** Diagnostic yield of whole exome sequencing-based genetic testing for patients with inherited eye diseases  
**Emma Mårtensson,** Blueprint Genetics, Helsinki, Finland  
**P02.50D** Mutations in PLS1, encoding fimbrin, cause autosomal dominant non-syndromic hearing loss (ADNSHL).  
**Anna Morgan,** Univ of Trieste, Trieste, Italy  
**P05.19C** Mutations in genes involved in MAPK pathway cause lymphatic anomalies  
**Dong Li,** Ctr for Applied Genomics, Philadelphia, United States  
**P08.35D** RNFT2, a novel gene causing intellectual disability; functional evidence in Drosophila melanogaster  
**Reza Ataei,** Univ of Social Welfare and Rehabilitation Sciences, Tehran, Iran, Islamic Republic of  
**P08.17B** Biallelic variants in DYNC1I2 cause syndromic microcephaly with intellectual disability, global developmental delay and dysmorphic facial features  
**Erica E. Davis,** Ctr for Human Disease Modeling, Durham, United States  
**P08.63D** Pathogenic WDFY3 variants cause neurodevelopmental disorders and opposing effects on brain size  
**Diana Le Duc,** Univ of Leipzig Medical Ctr, Leipzig, Germany  
**P10.20C** Inactivation of KLHL24 results in myopathy and cardiomyopathy  
**Carola Hedberg-Oldfors,** Univ of Gothenburg, Gothenburg, Sweden  
**P10.12D** Genetic analysis of autosomal dominant motor and sensory neuropathy with proximal dominancy in the lower extremities, urinary disturbance, and paroxysmal dry cough  
**Shiroh Miura,** Div of Respiratory, Neurology and Rheumatology, Dept of Med, Kurume Univ Sch of Med, Kurume, Japan  
**P11.24D** 49 novel recessive candidate genes for intellectual disability and visual impairment in 350 consanguineous families  
**Stylianos E. Antonarakis,** Dept of Genetic Med and Development, Univ of Geneva, Geneva, Switzerland  
**P11.32D** Gating-affecting mutations in KCNK4 cause a recognizable neurodevelopmental syndrome  
**Francesca Clementina C. Radio,** Ospedale Pediatrico Bambino Gesù, IRCCS, Rome, Italy  
**P11.68D** SEC31A mutation affects ER homeostasis, causing neurological syndrome  
**Daniel Halperin,** Ben-Gurion Univ of the Negev, Beer-Sheva, Israel  
**P11.23C** Genomic overlap between neurodevelopmental disorders and congenital heart defects  
**Seyed Ali Safizadeh Shabestari,** Mohammed Bin Rashid Univ of Med and Health Sciences, Dubai, United Arab Emirates  
**P19.03B** Genetic counselling for the Inuit indigenous population of Nunavut, Canada: an exercise in cultural competency  
**Mireille Clouties,** Children’s Hosp of Eastern Ontario, Ottawa, Canada  
**P19.35B** The discussion of uncertainty concerning multigene panel testing during cancer genetic counseling. An observational study.  
**Niki M. Medendorp,** Amsterdam UMC, Amsterdam, Netherlands  

The 30 Best Posters were selected for a short presentation during two concurrent sessions. In this session, the best posters from different topics will be presented. Please refer to page 14 for more information about the topics. The poster authors will have 3 minutes each to present their most important findings.

After the presentation of all posters (approximately at 13.45 hrs), the authors and the audience will proceed to the electronic posters next to the live stream area for discussion with the authors for the remainder of the session.
We invite all those working in the field of syndrome dysmorphology, and those who wish to learn more about the art and science of Dysmorphology, to attend this session. Please participate by bringing along short PowerPoint presentations of your distinctive unsolved cases or your instructive solved cases to one of the two Dysmorphology workshops. Even if you do not have cases to bring, we also encourage workshop attendees to share their experience of dysmorphology and broader genetic mechanisms by participation in the case discussions. As we move further into the genomic era we anticipate more discussion around variant interpretation and so we would also welcome experts in this area to join us. We also welcome "solved" cases that you may have presented as unknowns at the ESHG in previous years, but where you now have an answer. These are very interesting and instructive for the audience.

15.00 - 16.30

Dysmorphology

Organisers: Jill Clayton-Smith, Sofia Douzgou, Diaan Donnai

W12

Copy Number Variant Interpretation and Classification

Organisers: Nicole de Leeuw, Erica Genes, Zeynep Tumer

W13

Molecular Newborn screening vs. newborn testing

Organisers: Asbjorg Stray-Pedersen, Lucy Raymond

W14

European Reference Networks - What is it for me?

Organisers: Conxi Lazaro, Carla Oliveira

W15

Opportunistc or non opportunistic genetic screening

Organisers: Francesca Forzano, Marina Cornel

W16

Using the Ensembl VEP for analysing variants in rare and common disease

Organiser: Emily Perry

W17

Pharmacogenomics in practice

Organisers: Vita Dolzan, Volker Lauschke, Andrea Gaedigk

Corporate Satellites

(see page 45 for details)

The Ensembl Variant Effect Predictor (VEP) allows analysis of variants from sequencing to experiments to identify likely effect of the variants on genes, allowing for the prioritisation of further experiments. This workshop will familiarise the audience with general usage of the VEP, as well as two specialised use-cases: analysis of a short list of variants from GWAS to identify likely indirect effects on genes and analysis of genome wide data to identify variants likely to cause rare disease. Participants who bring their own laptops will have a chance to try out using the VEP online, as well as run VEP jobs using the script, by downloading Docker image.

Workshop Speakers: Emily Perry, Ensembl Outreach Project Leader

Inna Armean, Ensembl Information Bioinformatician

Coffee Break / Free Poster Viewing / Exhibition

Poster Viewing with authors and coffee (Group D)
### PROGRAMME MONDAY, JUNE 17

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| 17.45 – 19.15 | **E11** Genome First Testing in Pediatrics  
Chair: Asbjørg Stray-Pederson | **S13** Understanding mutations to detect cancer  
Chairs: Svetlana Bajalica-Lagercrantz  
Consil Lazaro | **S14** Debate: Genomics and the Media  
Chairs: Birgitte Dinnes  
Francesca Forzano | **E12** Oligogenic inheritance  
Chair: Alexandre Reymond | **S15** Regulatory Landscapes  
Chair: Elfride de Baere  
Martin Kircher | **S16** Methods for genetic epidemiology  
Chairs: Lude Franke  
Johannes Kettunen |
| 17.45         | **E11.1** The landscape of genomic alteration across childhood cancers  
Natalie Jäger, Hopp Children’s Cancer Ctr Heidelberg (KITZ), Heidelberg, Germany | **S13.1** Understanding mutational processes and tumor biology  
Abel Gonzalez-Perez, Inst for Res in Biomedicine, The Barcelona Inst of Science and Technology, Barcelona, Spain | **S14.1** Introduction  
Vivienne Parry, London, United Kingdom | **E12.1** Systematic analysis of genetic interactions: from yeast to human  
Jolanda Van Leeuwen, Univ of Lausanne, Lausanne, Switzerland | **S15.1** Enhancer Logic and Mechanics in Development and Disease  
Ali Shilatifard, Chicago, United States | **S16.1** Leveraging polygenic signals for insight into disease biology  
Hilary Finucane, Cambridge, United States |
| 18.15         |                               | **S13.2** Finding a germline mutation during tumor testing: implications for the patient and the family  
Jeffrey Weitzel, Los Angeles, United States | **S14.2** Genetics and Social Media  
Ellen T. Matloff, My Gene Counsel, New Haven, United States | **E12.2** Epistasis in Cardiac defects  
Bart Loey, Antwerp, Belgium | **S15.2** Regulation of disease-associated gene expression in the 3D genome  
Wouter De Laat, Oncoode & Hubrecht Inst, Utrecht, Netherlands | **S16.2** Genetic instruments in mendelian randomization studies  
George Davey-Smith, Bristol, United Kingdom |
| 18.30         | **E11.2** Rapid NGS for children in intensive care units  
Lucy Raymond, Cambridge, United Kingdom | **S13.3** Liquid biopsy to follow clonal evolution in cancer  
Benedetta Mussolin, Candiolo Cancer Inst – Fondazione Piemontese per l’Oncologia (FPO), Istituto di Ricovero e Cura a Carattere Scientifico (IRCCS), Candiolo, Italy | **S14.3** Human germline genome editing: The public and the pundits  
Francoise Baylis, Dalhousie Univ, Halifax, Canada | **E12.3** | **S15.3** Identifying DNA-regulatory elements in non-traditional model systems  
David Garfield, Berlin, Germany | **S16.3** Large-scale inference of human genetic data  
Manuel Rivas, Stanford, United States |
| 18.45         |                               | **S13.4**                              | **S14.4**                               | **E12.4**                                 | **S15.4**                                   |                              |
| 20.00         | Networking Event (at own expense - ticket required) | | | | | |

**Late Programme Changes**  
All contents are up-to-date as per date of printing.  
For changes in the scientific programme which occurred after the printing deadline, please consult the website:  
SCIENTIFIC PROGRAMME

TUESDAY, JUNE 18, 2019
EUROPEAN HUMAN GENETICS CONFERENCE 2020

53rd Meeting
City Cube | Berlin – Germany | June 6 – 9
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| 09.00 - 10.30 | S17            | ESHG-ASHG Debate: Global collaboration to advance the use of genomics in health  
 Chairs: Kiran Musunuru, Joris Veltman | S18               | Our genetic history and its phenotypic consequences  
 Chairs: Tord Jonson, Matti Pirinen | S19               | Treating rare genetic disease  
 Chairs: Brunella Franco, Asbjørg Stray-Pedersen | E13               | Genetic innovations in reproductive medicine  
 Chair: Maris Laan | S20               | Epigenetics and early development  
 Chairs: Siren Berland, Lucy Raymond | E14               | Understanding human disease through animal models  
 Chair: Jose Luis Costa |
| 09.00         | S17.1          | Challenges and solutions to advance global collaboration to understand, diagnose, and develop therapies for rare diseases  
 Kym M. Boycott, Children’s Hosp of Eastern Ontario, Univ of Ottawa, Ottawa, Canada | S18.1             | Tales of Early Humans, Admixture, and Adaptation  
 Mattias Jakobsson, Uppsala Univ, Uppsala, Sweden | S19.1             | Regeneration of the entire human epidermis using transgenic stem cells  
 Laura De Rosa, Modena, Italy | E13.1             | Novel discoveries of genes implicated in male and female infertility  
 Christophe Arnoult, Univ Grenoble Alpes, Grenoble, France | S20.1             | Single Cell Epigenomic Analysis of the Anatomy and Neuronal Circuitry of the Brain  
 Joseph R. Ecker, Howard Hughes Medical Inst, La Jolla, United States | E14.1             | Animal models of Machado-Joseph disease  
 Luis Perreira de Almeida, Coimbra, Portugal |
| 09.30         | S17.2          | EMBL-EBI and global data integration  
 Nick Goldman, European Bioinformatics Inst, Hinxton, United Kingdom | S18.2             | Timing past admixture events and characterizing their consequences in contemporary human populations  
 Francesco Muntoni, Univ Coll London Great Ormand Street Inst of Child Health, London, United Kingdom | E13.1             | Genetic-epigenetic interactions: mechanistic insights and practical applications  
 Benjamin Tycko, HHMI Ctr for Discovery and Innovation, Nutley, United States | S20.2             | Functional genomics approaches for uncovering the role of regulatory sequences in developmental abnormalities and disease  
 Justin L. Cotney, Genetics and Genome Sciences, UConn Health, Farmington, United States | E14.2             | CRISPR/Cas9 and TALENs fuel genetically engineered clinically relevant Xenopus tropicalis models  
 Kris Vlieghe, Ghent, Belgium |
| 10.00         | S17.3          | The NIH All of us program: Building a national research program of 1 million U.S. participants to advance precision medicine  
 Joshua C. Denny, Nashville, United States | S18.3             | Consequences of population genetic differences in genetic risk prediction across diverse human populations  
 Alicia Martin, Boston, United States | S19.3             | Gene therapy for hemoglobinopathies  
 Giuliana Ferrari, Milan, Italy | E13.2             | Population genetic carrier screening programs for reproductive purposes  
 Joél Zlotogora, Hadassah medical center, Hebrew Univ, Jerusalem, Israel | S20.3             | Functional genomics approaches for uncovering the role of regulatory sequences in developmental abnormalities and disease  
 Justin L. Cotney, Genetics and Genome Sciences, UConn Health, Farmington, United States | E14.2             | CRISPR/Cas9 and TALENs fuel genetically engineered clinically relevant Xenopus tropicalis models  
 Kris Vlieghe, Ghent, Belgium |
<p>| 10.30         | Coffee Break   | (Aisle G, Aisle F, Hall H) | 10.00             |                  |                    |                  |</p>
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<th>G2+G3</th>
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<td>11.00 - 12.30</td>
<td>C24</td>
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<td>C24.1</td>
<td>Analysis of Mosaicism for Sequence and Copy Number Variants in a Broad Diversity of Hereditary Disorders in a Large Clinical Cohort</td>
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<tr>
<td>Chairs: Jill Clayton-Smith Feliciano Ramos</td>
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<td>C24.2</td>
<td>A transcriptome-wide Mendelian randomization study to uncover tissue-dependent regulatory mechanisms across the human phenotype</td>
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<tr>
<td>Chairs: Tom G. Richardson, MRC Integrative Epidemiology Unit, Bristol, United Kingdom</td>
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<tr>
<td>C24.3</td>
<td>Somatic mutation cell lineage analysis reveals progressive clonal determination in human embryo</td>
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<tr>
<td>Chairs: Sara Bizzotto, Boston Children’s Hosp, Dept of Genetics and Genomics, Manton Ctr for Orphan Disease, Boston, United States</td>
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<td>C25.1</td>
<td>Multivariate GWAS of inflammatory markers reveals novel disease associations</td>
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<td>Chairs: Sanni E. Ruotsalainen, Univ of Helsinki, Inst for Molecular Med Human Genetics, Finland, Inst for Molecular Life Sciences, Radboud Univ Medical Ctr, Nijmegen, Netherlands</td>
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<td>C25.2</td>
<td>Analysis of genetic variants through aggregation of homologous human protein domains via MetaDome strongly improves diagnostic prediction of missense variants</td>
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<tr>
<td>Chairs: Laurens Wiel, Dept of Human Genetics, Radboud Inst for Molecular Life Sciences, Radboud Univ Medical Ctr, Nijmegen, Netherlands</td>
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<td>Mutations in the MRPS28 gene encoding the small mitochondrial ribosomal subunit protein b5t1m in a patient with intrauterine growth retardation, craniofacial dysmorphism and multisystemic involvement</td>
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<td>Basal and mutation-driven somatic mutagenesis shape the genome of healthy human cells</td>
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<td>Chairs: Irene Franco, Karolinska Institutet, HUDDINGE, Sweden</td>
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<td>C25.5</td>
<td>Bioinformatics and multomics</td>
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<td>Chairs: Christian Gillisen, Daniel Nilsson</td>
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<td>C26.1</td>
<td>Mutations in LIG3 are a novel cause of mitochondrial neurogastrointestinal encephalomyopathy</td>
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<td>Chairs: Elena Bonora, University of Bologna, Bologna, Italy</td>
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<td>Mutations in POLRMT impair mitochondrial transcription and are associated with a spectrum of mitochondrial disease presentations</td>
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<td>Chairs: Robert W. Taylor, Stanford Univ Sch of Med, Palo Alto, United States</td>
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<td>Mutated epigenetic modifiers in CYLD cutaneous syndrome</td>
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<td>Chairs: Neil Rajan, Inst of Genetic Medicine, Newcastle upon Tyne, United Kingdom</td>
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<td>C26.4</td>
<td>Identification and vocabulary of accessible DNA elements in the human genome</td>
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<td>Chairs: Wouter Meuleman, Altius Institute for Biomedical Sciences, Seattle, United States</td>
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<td>C26.5</td>
<td>Identification and characterization of NEPRO-related skeletal dysplasia resembling cartilage hair hypoplasia</td>
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<td>Chairs: Dhanay Lakshmi Narayanan, Kasturba Medical Coll, Manipal, Manipal Acad of Higher Education, Manipal, Manipal, India</td>
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<tr>
<td>C27.1</td>
<td>Diagnostic utility of genome-wide DNA methylation testing in genetically unsolved patients with suspected hereditary conditions</td>
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<td>Chairs: Bekim Sadikovic, London Health Sciences Ctr, Canada, London, Canada</td>
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<td>C27.2</td>
<td>Multivariates Approach to Diagnosing Undiagnosed Patients</td>
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<tr>
<td>Chairs: Matthew T. Wheeler, Stanford Univ Sch of Med, Palo Alto, United States</td>
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<td>C27.3</td>
<td>A novel ciliary Joseph-Saint-Vaast syndrome-associated protein module regulates axonal post translational modifications and cilium stability</td>
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<tr>
<td>Chairs: Ruxandra Bachmann-Gagescu, University of Zurich-Medical Genetics, Zürich, Switzerland</td>
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<tr>
<td>Chairs: Manel Roussouly, Altius Institute for Biomedical Sciences, Seattle, United States</td>
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<td>C27.5</td>
<td>Modeling the pathological long-range regulatory effects of human structural variation with patient-specific HiPSCs</td>
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<td>Chairs: Magdalena Laugsch, Centre for Molecular Medicine Cologne (CMM), University of Cologne, Cologne, Germany</td>
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<td>C28</td>
<td>Late Breaking abstracts</td>
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<td>Chairs: Inga Prokopenko Anna Lindstrand</td>
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<td>Whole exome sequencing and characterization of coding variation in 49,960 individuals in the UK Biobank</td>
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<td>Chairs: Cristopher Van Hout, Regeneron Pharmaceuticals, Tarrytown, United States</td>
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<td>Communication across generations: disclosure of BRCA cancer risk with young adults</td>
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<td>Chairs: Alison Luk, Young, Ctr for Medical Psychology &amp; Evidence-based Decision-making (CeMPED), Sch of Psychology, The Univ of Sydney, Sydney, Australia</td>
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<td>C29</td>
<td>Stakeholder perspectives in cancer genetics</td>
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<td>Chairs: Luzia Garrido, Ulf Kristoffersson</td>
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<td>C29.1</td>
<td>The public favours healthcare-mediated disclosure of hereditary cancer risk to at-risk relatives: a population-based survey in Sweden</td>
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<td>Chairs: Carolina Hawranek, Dept of Radiation sciences, Oncology, Umeå university, Umeå, Sweden</td>
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<td>C29.3</td>
<td>High-Risk Women's Responses and Understanding of Polygenic Breast Cancer Risk Information</td>
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<tr>
<td>Chairs: Tatiane Yanes, Univ of New South Wales, Sydney, Australia</td>
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### TIME | HALL C | K2+K3 | F1+F2+F3 | F4+F5 | G2+G3 | K1
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**12.00**<br>**C24**<br> Genetic basis of mosaic pigmentary disorders of the skin and how to detect them: the M.U.S.T.A.R.D. cohort’s experience<br> Arthur Sorlin, Ctr de Génétique, CHU Dijon Bourgogne, Dijon, France<br> C24.5<br> Genetic basis of mosaic pigmentary disorders of the skin and how to detect them: the M.U.S.T.A.R.D. cohort’s experience<br> Arthur Sorlin, Ctr de Génétique, CHU Dijon Bourgogne, Dijon, France<br> C25.5<br> A GWAS on data-driven 3D facial phenotypes selected by matching siblings reveals 310 genetic loci<br> Hanne Hoskens, Dept of Human Genetics, KU Leuven, Leuven, Belgium<br> C26.5<br> The homozygous variant c.797G>A/p.(Cys266Tyr) in PISD is associated with a spondyloepimetaphyseal dysplasia with large epiphyses and disturbed mitochondrial function<br> Leonie von Elsner, Inst of Human Genetics, Univ Medical Ctr Hamburg-Eppendorf, Hamburg, Germany<br> **12.15**<br> **C24.6**<br> The Hutchinson-Gilford progeria syndrome mutation is a somatic mutation in chronic kidney disease<br> Maria Eriksson, Karolinska Instt, Dept of Biosciences and Nutrition, Huddinge, Sweden<br> C25.6<br> GestaltMatcher: Identifying the second patient of its kind in the phenotype space<br> Tzung-Chien Hsieh, Inst for Genomic Statistics and Bioinformatics, Bonn, Germany<br> C26.6<br> SSBP1 mutations cause a complex optic atrophy spectrum disorder with mitochondrial DNA depletion<br> Tommaso Pippucci, Medical Genetics Unit, Sant’Orsola-Malpighi Univ Hosp, Bologna, Italy<br> **12.30**<br> Lunch break (Aisle G, Aisle F, Hall H)<br> **13.30**<br> **C27.6**<br> New mechanism for retinal degeneration on chrXq27.1<br> Jessica C. Gardner, UCL Inst of Ophthalmology, 11-43 Bath Street, EC1V 9EL, London, United Kingdom

Presentations highlighted by a grey background are from Young Investigator Award finalists.

### Late Programme Changes
All contents are up-to-date as per date of printing.
For changes in the scientific programme which occurred after the printing deadline, please consult the website: https://2019.eshg.org/index.php/programme/late-programme-changes/
## Programme Tuesday, June 18

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<tr>
<th>TIME</th>
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| 13.30 - 14.15 | PL3                  | Mendel Lecture  
Chairs: Alexandre Reymond, Joris Veltman |
| 13.30 - 13.45 | PL4.1               | A 25 Year Genomic Odyssey  
Chairs: Alexandre Reymond, Joris Veltman  
Laudation by Alexandre Reymond |
| 13.30 - 14.15 | PL3.1               | A 25 Year Genomic Odyssey  
Chairs: Alexandre Reymond, Joris Veltman  
Laudation by Alexandre Reymond |
| 13.30 - 13.45 | PL4.1               | We and our second genome: two key players in common complex diseases  
Chairs: Alexandre Reymond, Joris Veltman  
Laudation by Joris Veltman |
| 15.00 - 15.45 | PL5                 | Award Ceremony        
Chairs: Joris Veltman, Alexandre Reymond |
| 15.00 - 15.45 | PL5.1               | Award Ceremony        
Chairs: Joris Veltman, Alexandre Reymond |

- EJHG-SN Citation Awards
- ESHG Young Investigator Awards:
  - ESHG Young Investigator Awards for Outstanding Science
  - Lodewijk Sandkuijl Award for an outstanding presentation in the field of genetics of intellectual disability
  - Vanguard Medical Academy Award for an outstanding presentation in translational genetic research/therapy of genetic diseases
  - Mia Neri Award for an outstanding presentation in the field of cancer
- ESHG Poster Awards in clinical research and basic science
- Closing remarks

### Late Programme Changes

All contents are up-to-date as per date of printing.

For changes in the scientific programme which occurred after the printing deadline, please consult the website:  
PROGRAMME INFORMATION

SPONSORED SESSION
CORPORATE SATELLITE MEETINGS
BUSINESS MEETINGS
YOUNG INVESTIGATOR AWARD CANDIDATES
POSTER AWARD CANDIDATES
### Saturday, June 15, 08.00 - 10.00 hrs

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<tr>
<td>08.00</td>
<td>E01</td>
<td>New technologies (Sponsored by Illumina)</td>
<td>Chair: Martin Kircher</td>
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<td>08.00</td>
<td>E01.1</td>
<td>Using single cell genomics to understand cell fate decisions</td>
<td>John Marioni</td>
<td>Cambridge, United Kingdom</td>
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<td>08.30</td>
<td>E01.2</td>
<td>Resolving human genetic variation with long-read single-molecule sequencing</td>
<td>Mark J. Chaisson</td>
<td>Univ. of Southern California, Los Angeles, United States</td>
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<td>09.00</td>
<td>E01.3</td>
<td>Advancing single-cell genomics using combinatorial indexing</td>
<td>Andrew Adey</td>
<td>Portland, United States</td>
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<tr>
<td>09.30</td>
<td>E01.4</td>
<td>In vivo optical imaging and insights into human disease</td>
<td>Michelle Digman</td>
<td>Irvine, United States</td>
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## Overview

<table>
<thead>
<tr>
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<th>Room</th>
<th>Stand #</th>
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<td>Collecta</td>
<td>Room A-2 – Level 1</td>
<td>Stand # 428</td>
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<td>Dovetail Genomics</td>
<td>Room A-4 – Level 1</td>
<td>Stand # 532</td>
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<td>MGI, a subsidiary of BGI</td>
<td>Room A-3 – Level 1</td>
<td>Stand # 642 (BGI)</td>
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<td>BluePrint Genetics</td>
<td>Room A-3 – Level 1</td>
<td>Stand # 220</td>
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<td>CENTOGENE</td>
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<td>Stand # 336</td>
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<td>Congenica</td>
<td>Room A-4 – Level 1</td>
<td>Stand # 280</td>
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<td>NimaGen</td>
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<td>Stand # 348</td>
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<td>Stand # 634</td>
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<td>Asuragen</td>
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<td>Stand # 274</td>
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<td>Fabric Genomics</td>
<td>Room A-4 – Level 1</td>
<td>Stand # 374</td>
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<td>Oxford Nanopore Technologies</td>
<td>Room H-2 – Level 2</td>
<td>Stand # 244</td>
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<td>QIAGEN</td>
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<td>Thermo Fisher Scientific</td>
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<td>MSD &amp; AstraZeneca</td>
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<td>NanoString Technologies</td>
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<td>New England Biolabs</td>
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<td>Stand # 320 &amp; 620</td>
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<td>SOPHIA GENETICS</td>
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<td>Stand # 258</td>
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<td><strong>Sunday, June 16, 19.15 - 20.45 hrs</strong></td>
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<td>Bionano Genomics</td>
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Saturday, June 15, 10.00 - 11.30 hrs

**Collecta, Saturday, June 15, 2019, 10.00–11.30 hrs, Room A-2 – Level 1**

**Genetic Profiling and Functional Screening for Drug Target and Biomarker Discovery**

Join Collecta at a Corporate Satellite event at ESHG.

Learn about:
- State-of-the-art tools for high-throughput functional screening with CRISPR and shRNA libraries
- Unique applications of clonal barcode technologies for the study of tumor development
- Targeted expression profiling and its application for single-cell analysis

Presentations:

**Efficient tools to enable drug target and biomarker discovery: a brief introduction to Collecta technologies**

*Paul Diehl, Ph.D., COO, Collecta, Inc.*

Other presenters and topics to be confirmed.

Refreshments will be served.

For more updated information on event speakers and topics and to register, visit [www.collecta.com/eshg2019](http://www.collecta.com/eshg2019)

Collecta is a leading provider of genomic products and services for drug target and biomarker discovery and validation. Our functional genomics portfolio includes services providing gene knockout, knock-in, and knockdown screens; custom and genome-wide CRISPR, RNAi, and barcode libraries; construct services; cell engineering; kits to facilitate functional screening workflow; and targeted expression profiling products and services. Learn more at [www.collecta.com](http://www.collecta.com).

**Dovetail Genomics, Saturday, June 15, 2019, 10.00–11.30 hrs, Room A-4 – Level 1**

**Solving Complex Genomic Challenges with a Multi-Dimensional View of the Genome**

**Reconstructing shattered chromosomes and other applications for Dovetail Hi-C**

*Jannat Ijaz, BSc., Wellcome Sanger Institute, Cambridge, UK*

Chromothripsis is a catastrophic genome reshuffling event in which tens to hundreds of structural variants occur in a single crisis. It is a major driver of cancer development in some tumour types such as sarcomas and some pediatric cancers. Chromothripsis provides a natural experiment to elucidate functional and epigenetic consequences of genome rearrangement. We selected 5 patient-derived esophageal adenocarcinoma organoid models with chromothripsis on which to build reference genome assemblies, coupled with comprehensive chromosome conformation mapping. These organoids contain massively restructured chromosomes, with fragments that have been fused together ranging from tens of bases to megabases. The chromothripsis in these samples affect only a single parental copy and the other parental copy is mainly wild-type, which allows us to reconstruct the chromothriptic chromosomes. We have sequenced these organoid cell lines at high depth using a range of sequencing technologies including Illumina x10, 10X Genomics, PacBio and Dovetail™ Hi-C chromosome capture. Since each technology has its own advantages and disadvantages, integration of the different sequencing methods will provide a more complete picture of the chromothriptic regions. This has allowed us to reconstruct a highly shattered chromosome, which had over 800 rearrangements on just a single chromosome. In turn this has given us a better understanding of the mechanisms of chromothripsis and the generation of structural variants.

**MGI, a subsidiary of BGI, Saturday, June 15, 2019, 10.00–11.30 hrs, Room A-3 – Level 1**

**Towards a Perfect Genome**

The “Perfect Genome” is free of errors, meaning there is no need to validate medically relevant variants by sequencing. With a perfectly read genome, we can be confident that everything that could affect the health of the individual, including all de novo mutations, has been found. In practice, it may be difficult to ever achieve a truly “Perfect Genome”, but a genome with a few errors and some unresolved repeat sequences is very obtainable and for the purposes of improving human health.

In this presentation, we will introduce our “Perfect Genome” solution which employs advanced massively parallel DNA sequencing of “co-barcoded” reads from long genomic DNA molecules, and efficient de novo assembly empowered by these barcoded reads. It will be also presented how DNBSeq™ based NGS technology is applied for large-scale studies of microbiome profiling as well as CleanPlex MGI NGS panels.

**Chair:** Dr Yong Hou, Executive director, BGI-Research, Shenzhen, China

**DNBseq™ and stLFR enabling close to perfect WGS**

*Dr Rade Drmanac, CSO of MGI Tech, San Jose, USA*

**Large scale microbiome profiling in population-based studies**

*Prof Lars Engstrand, Director of Center for Translational Microbiome Research, Karolinska Institute, Stockholm, Sweden*

**CleanPlex for MGI: A New Targeted NGS Solution with Exceptional Coverage Uniformity and Rapid Customization**

*Dr Edward Jan, Director of Product Marketing, Paragon Genomics, Hayward, CA, USA*
Saturday, June 15, 12.15 - 13.45 hrs

BluePrint Genetics, Saturday, June 15, 2019, 12.15–13.45 hrs, Room A-3 – Level 1

Improving diagnostic yield with advances in difficult-to-sequence regions and high resolution CNV detection
12:15-13:00
Emerging opportunities in resolving difficult-to-sequence regions – PKD1 as a case example
Johanna Sistonen, PhD, Head of Clinical R&D, Blueprint Genetics, Helsinki, Finland
13:00-13:45
High resolution CNV detection – a powerful diagnostic tool
Eveliina Salminen, MD, PhD, Associate Laboratory Director, Blueprint Genetics, Helsinki, Finland

Enhancing diagnostic performance in difficult-to-sequence regions is improving patient outcomes. However, there are still many regions in the genome with clinically important variants that are not covered with standard NGS strategies or Sanger sequencing. These regions include genes that have pseudogenes, other highly homologous genomic regions, or consist of longer stretches of repetitive sequences.

The first part of this session presents our experiences with resolving such regions, highlighting PKD1 and the genetic diagnostics of polycystic kidney disease as a case example. The second part of this session will focus on the importance of CNV detection as an integral part of diagnostic testing. Case examples demonstrating the clinical relevance of CNV detection will be presented, with a special focus on ophthalmology.

CENTOGENE, Saturday, June 15, 2019, 12.15–13.45 hrs, Room A-5 – Level 1

Benefits of WGS
Clinical Whole Genome Sequencing: a rising Star
Prof. Peter Bauer, Chief Scientific Officer, CENTOGENE AG, Rostock, Germany
With the adoption of WGS into clinical heath care, CENTOGENE has built bioinformatics and medical solutions, to ensure fast and robust processes. Based on an analysis of more than 1,000 in-house cases, Prof. Bauer will share strategies to optimize diagnostic utility and yield of clinical WGS.

A pediatricians experience with Whole Genome Sequencing
Prof. Yasemin Alanay, Department of Pediatrics, Acibadem Maslak Hospital, Istanbul, Turkey
Explaining clinical pictures in pediatric patients with complex phenotypes can be a challenge. State-of-the-art diagnostic tools such as WGS help you uncovering the unknown. CENTOGENE’s long-term partner Prof. Alanay will present illustrative examples from a series of patients seen at a large pediatric department in Turkey.

Classification, Curation, Re-Classification: we Care
Dr. Gabriela Oprea, Senior Vice President Digital Products, CENTOGENE AG, Rostock, Germany
The steady increase in genetic knowledge makes variant classification remain a highly dynamic process. CENTOGENE utilizes its proprietary database CentoMD™ to document and keep track of all relevant information. Dr. Oprea will outline how this highly curated data is used for standardized variant re-classification and, thereby, for a sustained diagnostic service for patients and their clinicians.

Congenica, Saturday, June 15, 2019, 12.15–13.45 hrs, Room A-4 – Level 1

Improving Diagnosis for Patients with Rare Genetic Disease
Three speakers from leading institutions will outline how they have improved diagnostic yield and case throughput in their clinics, across three different medical specialties.

Using case examples, the speakers will show how they have streamlined diagnosis in fetal abnormalities, epilepsy and complex neuropathy in everyday clinical practice, and for the UK 100,000 Genomes Project.

Talks include:
Clinical Utility of Prenatal Exome Sequencing: Achieving a high diagnostic yield with careful patient ascertainment
Dr Tessa Homfray, Consultant, Medical Genetics, St George’s University Hospital, London, UK

Maximising Diagnostic Yield: Application of Congenica’s Sapientia variant interpretation platform to a cohort of Irish patients with epilepsy and learning disability
Robert Carton, Postdoctoral Research Fellow, Royal College of Surgeons Ireland (RCSI), Dublin, Ireland

The use of Sapientia for the analysis of cases from the 100,000 Genomes project in a clinical diagnostic setting
Sarah Mackenzie, Clinical Scientist, Northern Genetics Service, The Newcastle upon Tyne Hospitals NHS Foundation Trust, Newcastle, UK
**Monday, June 17, 11.15 - 12.45 hrs**

**Agilent Technologies, Sunday, June 16, 2019, 11.15–12.45 hrs, Room A-1 – Level 1**

**Approaches for Precision Medicine in Cancer**

**Fast and high precision cancer samples analysis using SureSelect XT HS / Low Input technology**

*Dr. Raouf Ben Abdelali, Laboratoire CERBA, Saint-Ouen-l’Aumône, France*

Laboratoire CERBA will present optimization and automation of its NGS analysis workflow, using SureSelectXT HS / Low Input, for delivering high quality results from a wide variety of samples types (blood, fresh frozen tissue and FFPE), DNA quality and input, for oncogenetics and onco-hematology panels up to somatic exome and RNAseq analysis.

**Mutational analysis of BRCA1/2 in a group of ovarian cancer patients**

*Bartosz Wasag, PhD, Medical University of Gdansk & University Clinical Centre, Gdansk, Poland*

Both germline and somatic BRCA1/2 alterations induce the sensitivity to PARP1 inhibitors of ovarian cancer patients. Therefore, complex mutational analysis of BRCA1/2 genes is required to identify patients likely to benefit from this treatment. Highly sensitive and cost-effective analysis of BRCA1/2 accessible for routine diagnostics will be presented.

**Precision medicine and the future of clinical cancer genomics**

*Dr. Susie Cooke, Glasgow Precision Oncology Laboratory, University of Glasgow, UK*

Dr. Cooke describes how Precision Panc, a UK-wide clinical trials network for pancreatic cancer, works to translate cutting-edge concepts combining comprehensive genomic profiling of real-world samples with affordability and rapid turnaround, allowing patient stratification, biomarker validation and biomarker discovery as part of each trial.

**Asuragen, Sunday, June 16, 2019, 11.15–12.45 hrs, Room A-2 – Level 1**

**Shining a Light on Dark DNA:**

**Simple and Streamlined Solutions for SMN1/2 Copy Number Determination, HTT Expansions and More…**

This corporate satellite will highlight the rapid expansion of AmplideX® technology to new genomic targets, including those each trigonucleotide repeat expansions, such as HTT, as well as genes where precise copy number resolution is required, including SMN1 and SMN2. Marcia Oliveira, PhD, ErcLg, will review the performance of the AmplideX PCR/CE SMN1/2 Kit (RUO) in testing clinical specimens spanning a broad range of SMN1 and SMN2 copy numbers. Additionally, Ferdinando Squitieri, MD, PhD, will share his experience with the AmplideX PCR/CE HTT Kit (RUO) and will review its ability to quickly and easily detect CAG expansions in HTT, even in samples exceeding 200 CAG repeats. Both presenters will highlight the versatility of AmplideX technology in providing simple, streamlined solutions for the analysis of these targets while leveraging a single, common workflow compatible with broadly installed systems.

**Speakers:**

Marcia Oliveira, PhD, ErcLg, Centro Hospitalar do Porto E.PE, Centro de Genetica Medica Doutar Jacinto de Magalhaes, Unidade de Genetica Molecular, Porto, Portugal

Ferdinando Squitieri, MD, PhD, Head of Neurology CSS- Mendel Institute of Human Genetics, Head of Huntington and Rare Diseases Unit IRCCS, Scientific Officer and Co founder Lega Italiana Ricerca Huntington e Malattie ostrac Foundation, Rome, Italy
**Fabric Genomics, Sunday, June 16, 2019, 11.15–12.45 hrs, Room A-4 – Level 1**

**Applying Artificial Intelligence to Accelerate Interpretation for Genomes and Gene Panels**

The biggest bottleneck to scaling genetic testing is the time spent in interpretation and classification of all variants in a case. Fabric Genomics delivers an end-to-end software platform for genomic analysis and reporting, and includes AI-based methods to accelerate and reduce costs in clinical reporting of whole-genomes, exomes, and gene panels.

Francisco De La Vega, Fabric’s SVP of Genomics, will present data from our new inference engine that leverages deep gene and variant annotation for highly accurate ACMG variant classification for gene panels used for incidental findings, population risk, and newborn screening. Fabric ACE (AI Classification Engine) is embedded into Fabric Enterprise for complete FASTQ-to-clinical report workflow, and allows labs to accelerate accurate variant interpretation, classification, and clinical reporting down to minutes per case.

Mark Yandell, from the University of Utah, will show results from a new AI-layer on top of Fabric’s VAAST and Phevor methods for the diagnosis of rare genetic diseases, that allow the identification of disease genes with unprecedented accuracy and speed.

Learn how Fabric’s proprietary methods, when integrated with our clinical reporting software, enable high-throughput genetic labs to interpret and score variants rapidly, reproducibly, cost-effectively and at scale.

Speakers: Mark Yandell, PhD, Professor of Human Genetics, Co-director USTAR Center for Genetic Discovery, Assoc. Director Program in Personalized Health, University of Utah, Salt Lake City, USA

Francisco M. De La Vega, D.Sc. (Chair), Senior Vice President, Genomics, Fabric Genomics, Oakland, USA

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**Oxford Nanopore Technologies, Sunday, June 16, 2019, 11.15–12.45 hrs, Room H-2 – Level 2**

**Ultra-long read nanopore sequencing and human genetics**

Please join Oxford Nanopore Technologies for an exciting seminar featuring updates from Oxford Nanopore plus user talks from speakers including Alba Sanchis-Juan (University of Cambridge) and Nicolas Chatron (CHU Lyon).

Oxford Nanopore Technologies has developed the world’s first nanopore DNA and RNA sequencing devices. The MinION is a portable, real-time, long-read, low-cost device designed to bring easy biological analyses to anyone, anywhere. The GridION and PromethION devices serve users with larger projects or more samples. Fully characterise human genetic variation by sequencing whole genomes, targeted regions or full-length RNA transcripts. Long nanopore reads enable comprehensive analysis of structural variation, repetitive regions, haplotype phasing, RNA splice variants, isoforms, fusion transcripts and base modifications. Oxford Nanopore Technologies products are currently for Research Use Only. Not for use in diagnostic procedures.

Lunch will be provided. Please register to attend at nanoporetech.com/events/eshg-2019

Confirmed speakers: Alba Sanchis-Juan, University of Cambridge, UK

Nicolas Chatron, CHU Lyon, France

More speakers to be announced soon.

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**QIAGEN, Sunday, June 16, 2019, 11.15–12.45 hrs, Room A-3 – Level 1**

**Unravel the complexities of human genetics with QIAGEN’s Sample to Insight solutions**

Accelerate insights in cancer genetics, cardiovascular disorders, reproductive medicine and hereditary diseases by leveraging integrated preanalytical, next-generation sequencing (NGS) and bioinformatics solutions. QIAGEN's market-leading sample collection, stabilization and extraction products, combined with high-performance NGS solutions and intuitive bioinformatics tools optimize the identification, classification, interpretation and standardized reporting of the most challenging and complex pathogenic variants.

Join us on a fascinating journey of discovery of new groundbreaking biomarkers.

A panel of external and internal experts will report on their latest findings and will be available for discussion during the workshop.

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**Thermo Fisher Scientific, Sunday, June 16, 2019, 11.15–12.45 hrs, Room A-5 – Level 1**

**Genomics into the Clinic**

As the path from genomic research to the clinic becomes clearer, how do scientific and technological innovations combine to bring answers closer? Across complex diseases and genetic disorders, our lunchtime seminar will showcase innovative science and offer a view on how labs will evolve.

Chairperson: Karen Jones, PhD, Thermo Fisher Scientific

Genomics lab of the future: productivity from digital science

Will Geist, VP and General Manager, Thermo Fisher Scientific

FinnGen: a platform for drug target development and precision medicine

Samuli Ripatti, Professor of Biometry, Dept. of Public Health and Institute for Molecular Medicine Finland (FiMM), HILIFE, University of Helsinki; Broad Institute of MIT and Harvard, US

From CMAP to NMAP: an integrated microarray approach to repositioning drugs for neurodegenerative disorders

David Chambers, PhD, Principal Investigator, Lecturer in Functional Genomics & Drug Discovery, Genomics Drug Discovery Unit, Wolfson Centre for Age-Related Diseases (CARO), King's College London, UK

The use of high-resolution exon microarray in Uppsala, Sweden

Ann-Charlotte Thuresson, Associate Professor, Clinical Laboratory Geneticist, Clinical Genetics, Uppsala University Hospital, Uppsala, Sweden

Proof of principle assessment of a next generation sequencing workflow for rapid newborn screening and Cystic Fibrosis testing

Rebecca Thomas, PhD, Elizabeth Sollers PhD, Sheffield Children’s NHS Foundation Trust, UK
Sunday, June 16, 15.00 - 16.30 hrs

MSD & AstraZeneca, Sunday, June 16, 2019, 15.00–16.30 hrs, Room A-5 – Level 1

Not exhibiting

BRCA Testing to Guide Precision Medicine: Strengthening the Foundations and Building for the Future

The Corporate Satellite will give an overview of the importance of biomarker testing in an era of precision medicine, where patient selection becomes ever more important in the treatment of cancer.

The program will give insights into BRCA testing, specifically focusing on optimization of BRCA testing in ovarian cancer, as well as BRCA tumor testing methodologies and related quality requirements. Additionally, the program will provide an outlook on BRCA testing potential to guide precision medicine approaches in additional cancer types, beyond Breast and Ovarian Cancer.

Optimizing Patient Selection in Ovarian Cancer: The Rationale for Tumor BRCA Testing

Dr. Bojana Djordjevic, Gynecologic Pathologist Division of Anatomic Pathology Department of Laboratory Medicine and Molecular Diagnostics Sunnybrook Health Sciences Centre. Associate Professor Department of Laboratory Medicine and Pathobiology University of Toronto, Canada

The Keys to Quality in Tumor BRCA Testing

Professor Ettore Capoluongo, Agostino Gemelli University Polyclinic Foundation (Hormonal Analysis and Clinical Molecular Biology Unit), Rome, Italy

BRCA Testing in Precision Medicine Beyond Ovarian Cancer

Professor Frederik Marmé, Experimental and Translational Gynecologic Oncology, University Medical Center, Mannheim, Germany

NanoString Technologies, Sunday, June 16, 2019, 15.00–16.30 hrs, Room A-3 – Level 1

Stand # 452

Novel Molecular Approaches to Genomic Discovery and Profiling

Digital Spatial Profiling: delivering on the promise of Spatial Genomics

Margaret Hoang, Sr Scientist, NanoString Technologies, Seattle, WA, USA

NanoString GeoMx™ Digital Spatial Profiling is a novel, highly multiplexed assay that digitally characterizes protein and RNA expression from spatially discrete regions of interest (ROIs) within tissue sections. We present our high-plex spatial RNA molecular profiling of ~1,500 immuno-oncology targets coupled to downstream NGS readout to enable high-throughput capacity and scalability. GeoMx™ DSP with NGS readout measures mRNA abundance and has the ability for in situ detection of splice variants. Concordance data with other technologies will also be presented.

Multiplexed Lung Cancer fusions detection without FISH or NGS

Dr. Leon Van Kempen, University Medical Center Groningen, the Netherlands

The detection of gene translocations in lung cancer samples can reveal actionable molecular targets for therapy. Translocations of ALK, ROS1, RET and NTRK are oncogenic drivers of NSCLC. The NanoString nCounter™ platform enables the detection of up to 800 different fusions transcripts, including ALK and other druggable targets such as MET exon 14 skipping. This simple and fast multiplex approach enables the detection of these rare mutations in a single assay in less than 24 hrs. Comparison vs NGS and FISH will be presented.

New England Biolabs, Sunday, June 16, 2019, 15.00–16.30 hrs, Room A-4 – Level 1

Stand # 456

Advancements in NGS Sample Preparation - Enzymatic Methyl-seq, Targeted Sequencing and SNP Genotyping

New England Biolabs is a global leader in developing solutions for Next Generation Sequencing Sample Preparation and continues to push the forefront in providing high quality, robust products to support the clinical application of genomic data. During this workshop we will elucidate this through practical examples demonstrating how these products are being applied to overcome challenges associated with clinical genomics as well as an overview of the latest advancements from NEB.

Chair: Dr. Bjorn Textor, Sr. Application Specialist, New England Biolabs, Germany

Speakers:

- A precise DNA-friendly analysis of cytosine methylation status with EM-seq
  Dr. Vladimir Benes, Head, Genomics Core Facility EMBL Heidelberg, Germany

- NEBNext Direct: Maximizing efficiency and throughput for diverse target enrichment applications
  Kruti Patel, Research Scientist, Directed Genomics, USA

- An overview of the latest advancements from NEB to enable human genetics
  Andrew Barry, Sr. Manager Business Development, New England Biolabs, USA

PerkinElmer, Sunday, June 16, 2019, 15.00–16.30 hrs, Room A-2 – Level 1

Stand # 320 & 620

PerkinElmer innovations in Genetics for Reproductive Health

3:00 p.m. – 3:30 p.m.

Preimplantation testing of aneuploidies (PGT-A) and structural rearrangements (PGT-SR) using automated PG-Seq™ workflow

Jakub Horák, PhD, Repromeda, Brno, Czech Republic

3:30 p.m. – 4:00 p.m.

Vanadis® NIP System*: A new non-amplification and targeted cfDNA technology for Noninvasive Prenatal Testing (NIPT) and beyond

Fredrik Dahl, PhD, PerkinElmer, Stockholm, Sweden

4:00 p.m. – 4:30 p.m.

User perspective on NIPT implementation and clinical experience with Vanadis® NIPT System

TBC

* Products may not be licensed in accordance with the laws in all countries, such as the United States and Canada. Please check with your local representative for availability.
SOPHiA GENETICS, Sunday, June 16, 2019, 15.00–16.30 hrs, Room A-1 – Level 1

Hereditary disorders solutions powered by SOPHiA: overcoming challenges to increase diagnostic yield
Moderator:  Gioia Althoff, PhD, SVP Genomics, SOPHiA GENETICS
The advent of Next-Generation Sequencing (NGS) and cutting-edge genomic applications has greatly improved the ability to rapidly analyze many genes at the same time. Unfortunately, managing the massive amount of generated data to easily identify potentially disease-causing variations has become a major challenge. The session will focus on how SOPHiA enables clinicians to deal with the vast amounts of data coming from targeted and exome applications and accurately detect the variant of interest. You will discover the full capacities of our applications for hereditary disorders and hear customer success stories.

Molecular diagnosis of osteogenesis imperfecta through a targeted gene panel powered by SOPHiA
Assoc. Prof. Sehime Gülsün Temel, MD, PhD, Department of Medical Genetics, Uludag University Hospital, Turkey

SOPHiA Whole Exome Solution: shedding light on complex clinical cases
Georgios Stamoulis, PhD, Clinical Application Product Manager, SOPHiA GENETICS
Dr. Pantelis Constantoulakis, Head of the Molecular Genetics Department, Genotypas-Science Labs S.A., Athens, Greece

Turning raw genomic data into characterized variants using SOPHiA
Zhenyu Xu, PhD, Chief Technology Officer, SOPHiA GENETICS

Sunday, June 16, 19.15 - 20.45 hrs

Bionano Genomics, Sunday, June 16, 2019, 19.15–20.45 hrs, Room A-4 – Level 1

Next-Generation Cytogenetics: High-throughput mapping of structural variation in cancer and genetic disease
The Bionano Saphyr platform for whole genome mapping offers an extremely long-read technology, providing unmatched sensitivity to detect structural variation, genome-wide, at low cost. Our de novo maps can resolve complex repetitive regions, identify Copy Number Variations, and elucidate genome-wide structural variation like balanced/unbalanced translocations, inversions, and indels with high sensitivity and precision.

Hear from two members of world-leading institutions on their use of Bionano optical mapping for diagnosis of leukemias and reproductive and developmental disorders, and their ongoing projects exploring the use of this technology to replace a combination of classical cytogenetic approaches such as karyotyping, FISH and Chromosomal Microarray Analysis (CMA).

Talks will include:
19:15 – 19:45 New developments in long read optical mapping enable novel applications for cancer and genetic disease
Sven Bocklandt, PhD, Director of Scientific Affairs, Bionano Genomics, Inc., San Diego, CA, USA

19:45 – 20:15 Optical mapping enables next generation cytogenetics – applications in medical genetics
Alexander Hoischen, PhD, Assistant Professor, Immuno-Genomics, Radboud University Medical Center, Nijmegen, the Netherlands

20:15 – 20:45 Using next generation mapping to detect balanced as well as unbalanced structural variants in reproductive and developmental diseases
Laïla El Khattabi, PharmD PhD, Associate Professor, APHP Cochin - Paris Descartes University, Paris, France

Illumina, Sunday, June 16, 2019, 19.15–20.45 hrs, Room A-5 – Level 1

Genetic Disease… from Research to Clinical Application
Programme not available at the time of printing of the Final Programme.

Integrated DNA Technologies, Sunday, June 16, 2019, 19.15–20.45 hrs, Room A-2 – Level 1

New NGS solutions to simplify your journey from sample prep to sequencing
Two IDT experts will be introducing our newest innovations for NGS, rhAmpSeq™ Amplicon Sequencing and the Lotus™ DNA Library Preparation Kit.

Beginning with the rhAmpSeq system, we recommend it for any researcher interested in target enrichment by amplification. Based on proprietary rhAmp PCR technology, rhAmpSeq allows for unprecedented specificity when amplifying thousands of targets in a single reaction. In this presentation, you will see data generated from custom rhAmpSeq panels which showcases their uniform coverage and negligible off-target amplification.

Next, we’ll be discussing our new library preparation kit, Lotus. The kit consistently provides uniform coverage across GC content, lowering your sequencing costs, and can be used seamlessly with IDT custom adapters to create high-complexity libraries. Our presentation will showcase the kit’s performance for whole and targeted genome sequencing applications.

Speakers: Elisabeth Gustafson-Wagner, PhD, Manager, Scientific Applications Support, Integrated DNA Technologies, Coralville, Iowa, United States
Nick Downey, PhD, Senior Product Manager, Integrated DNA Technologies, Coralville, Iowa, United States
Monday, June 17, 11.15 - 12.45 hrs

**Agilent Technologies, Monday, June 17, 2019, 11.15–12.45 hrs, Room A-5 – Level 1**

**Developments in NGS Workflows for Human Genetics**

**Integrated NGS-based approaches for breast cancer-related germline mutations detection**

Dr. Valeria D’Argenio, MD, PhD, Department of Molecular Medicine and Medical Biotechnologies, Federico II University and CEINGE Biotecnologie Avanzate, Napoli, Italy

Discussion on data collected from 6 years’ experience in the field of hereditary breast cancer diagnosis using BRCA genes amplicon sequencing and custom panel analysis.

**TLA-based haplotyping & NIPD development**

Prof. Wouter de Laat, Hubrecht Institute-KNAW and University Medical Center Utrecht, Utrecht, the Netherlands

Presenting Monogenic Non-invasive prenatal diagnostics NIPD (MG-NIPD) assay, where blood samples from both parents is used for Targeted Locus Amplification (TLA)-based phasing of selective heterozygous variants.

**The need of short turnaround times for whole exome sequencing (WES) in critically ill children and fetuses with multiple congenital anomalies on ultrasound**

R. J. Sinke, Department of Genetics, UCMG, University of Groningen, the Netherlands

We discuss the potential challenges of implementation of rapid diagnostic WES in critically ill children, aiming to improve clinical care and replace time-consuming and/or invasive diagnostic testing. Trio WES analysis using a custom virtual gene-panel for quick turnaround times, as well as WES in prenatal care to identify the cause of fetal anomalies is discussed. We also tested a fully-automated sample preparation system (Magnis, Agilent) which could reduce turnaround time, simplify and shorten lab-procedures.

**NIPD Genetics, Monday, June 17, 2019, 11.15–12.45 hrs, Room A-1 – Level 1**

**High-Fidelity Diagnostic Solutions for NIPT, Reproductive Health and Oncology**

NIPD Genetics is a European biotechnology company, that designs, develops and delivers innovative genetic tests. We have developed a novel target capture enrichment technology for the analysis of DNA fragments. In 2015, we launched the VERACITY NIPT for the detection of the common autosomal and gonosomal aneuploidies. We then expanded the technology to detect 4 microdeletion syndromes of clinical significance. In 2018, we extended the technology further to detect point mutations, and introduced VERAgene, the only NIPT that simultaneously tests for whole chromosome aneuploidies, microdeletions, and 50 monogenic disorders.

Further research and development led to expanding our technology into detecting indels, marking the launch of new oncology products in hereditary cancer screening and tumor tissue biopsy. The targeted nature of our technology, and the specifically designed analytical methods and bioinformatics ensure high read-depths and unparalleled accuracy across all applications.

NIPD Genetics aims to further expand our technology to applications like carrier screening, preimplantation genetic testing and liquid biopsy.

Speakers:
- PD Dr. rer. nat. Markus Stumm, Head of Laboratory Fachhumangenetiker (GfH), Clinical Laboratory Geneticist (EBMG)
- Medicovery Genetics GmbH, Berlin, Germany
- Alexia Eliades, PhD, Oncology Product Development Leader, NIPD Genetics, Nicosia, Cyprus
- Hari Radhakrishnan, PhD, Chief Business Development Officer, NIPD Genetics, Nicosia, Cyprus

**Roche Sequencing Solutions, Monday, June 17, 2019, 11.15–12.45 hrs, Room A-3 – Level 1**

**NGS in Oncology Today - Pain Points and Solutions**

Chair: Maximilian Schmid, MD, Head of Medical Affairs, Roche Sequencing Solutions, Inc.

**Blood-based therapy monitoring in lung adenocarcinoma**

Holger Sultmann, PhD, Professor, German Cancer Research Center, Germany

**Practical experiences as a pilot with the NAVIFY Mutation Profiler - a pragmatic voice from the bridge deck**

Markus Tiemann, MD, CEO and Managing Director, Institute of Hematopathology Hamburg, Germany

**Oncology NGS: Dreams Delivered?**

Marianne Nicolson, MD, Professor of Medical Oncology, Aberdeen University, Aberdeen Royal Infirmary, UK

A rapid expansion of targeted cancer therapies and molecular biomarkers led to the fast adoption of NGS in the clinic. As a result, clinical laboratories are facing unprecedented challenges in managing and interpreting data from NGS. The session will focus on the varying approaches to adopting NGS testing and highlight technical as well as biological limitations that need to be taken into account in the laboratory implementation of NGS assays. It will cover the implementation of NGS testing for both tissue and liquid biopsies, the latter of which is a clinical research tool with the sensitivity and specificity needed to detect low levels of ctDNA in the plasma. To meet the challenges of NGS data interpretation, a highly curated knowledge base enabling straightforward data interpretation will be discussed.
In this talk we will explore LoopSeq™ sequencing technology and how it is applied to provide additional, previously inaccessible layers of information purification from virtually any source and suitable for desired downstream applications.

KingFisher instruments and MagMAX reagents pair to provide highly versatile, automated magnetic-particle processing for DNA/RNA, protein or cell making their identification very challenging. In this presentation we will hear how the KingFisher system can be used to enrich and identify CTCs.

Automated sample preparation supports their work in this process.

Blood or serum cfDNA is suitable for disease analysis and in a range of research applications. Our academic scientist will give an overview of how Twist Technology: Improving Sequencing on Challenging Samples

During this workshop, presenters will discuss various applications and present data from their work using Twist NGS Target Enrichment Solutions. Downstream sequencing analysis and perform fewer sequencing runs per sample without sacrificing performance, saving them time and money. Due to the precision of these Twist capture probes, researchers are able to considerably improve the accuracy of the downstream sequencing analysis and perform fewer sequencing runs per sample without sacrificing performance, saving them time and money.

Twist Bioscience uses its proprietary DNA synthesis platform to build target enrichment panels for researchers performing next-generation sequencing experiments and analysis. The advent of next generation sequencing (NGS) technology has revolutionized our ability to read the genetic code, bringing about tremendous progress in our understanding of how biology is encoded in DNA and in medical diagnostics. However, while the throughput of NGS has improved by orders of magnitude compared to Sanger sequencing, NGS is limited to reading DNA in short segments of 150-300 nucleotides at a time. Biology, unfortunately, is not encoded in stretches of 150 nucleotides, but in much longer segments of DNA and RNA spanning many thousands of nucleotides. Loop Genomics has developed a synthetic long read sequencing technology that leverages existing Illumina short read sequencers coupled with LoopSeq™ barcoding technology to enable single-molecule, long-read sequencing on any Illumina infrastructure. In this talk we will explore LoopSeq™ sequencing technology and how it is applied to provide additional, previously inaccessible layers of information from Illumina sequencer for a wide variety of sequencing applications as diverse as Microbiome, Transcriptome, Targeted Amplicons and others.

The utility of exome sequencing analysis in solving rare genetic disorders: the Exeter experience

In this session we will learn about the challenge that the bioinformatic analysis represents, the visualization of normal and abnormal sequences and the impact that is going to perform in the clinical practice through some examples based on the novo reciprocal translocations.

Bioinformatic principles of genome analysis

Juan Carlos Triviño Head of bioinformatics department, Sistemas Genómicos, Valencia, Spain

From bioinformatic computation to clinically relevant alterations

Alejandra Pérez Sastre, PhD. Molecular Pharmacology and Cell Biology, Sistemas Genómicos, Valencia, Spain

Genome in apparently balanced de novo translocations in patients with malformations and/or neurodevelopmental disorders

Irene Valenzuela, MD, Clinical Geneticist, Hospital Vall d’Hebron, Barcelona, Spain

A result is only as good as the sample preparation that precedes it.

The satellite will consider solutions to the different challenges in a sample preparation, with a focus on new applications such as the purification of cell-free DNA and the isolation of circulating tumour cells (CTCs).

Magnetic-based enrichment in circulating tumour cells using the KingFisher™ system: new opportunities for liquid biopsies

Rui P.L. Neves, Dr. rer. nat., University Hospital and Medical Faculty of the Heinrich-Heine University, Düsseldorf, Germany

Circulating tumour cells (CTCs) are valuable prognostic biomarkers in different tumours. CTCs are present in blood at extremely low frequencies making their identification very challenging. In this presentation we will hear how the KingFisher system can be used to enrich and identify CTCs.

Towards integrating of targeted next generation sequencing in the clinic

Jacqui Shaw, Prof. Dr., University of Leicester, UK

Cell-free DNA or circulating DNA are degraded DNA fragments found in the bloodstream and can be captured as a biological sample such as whole blood or serum. cfDNA is suitable for disease analysis and in a range of research applications. Our academic scientist will give an overview of how automated sample preparation supports their work in this process.

Using KingFisher to increase reproducibility in new applications using challenging samples

Hannah E. Saunders, MSPH, Scientist at Thermo Fisher Scientific™, Austin, USA

KingFisher instruments and MagMAX reagents pair to provide highly versatile, automated magnetic-particle processing for DNA/RNA, protein or cell purification from virtually any source and suitable for desired downstream applications.

Monday, June 17, 15.00 - 16.30 hrs

LoopSeq Synthetic Long Read Sequencing and Its Applications: from Microbiome to Transcriptome and Beyond

Speaker: Indira Wu, PhD, Director of Molecular Biology, Loop Genomics, San Jose, California, USA

The advent of next generation sequencing (NGS) technology has revolutionized our ability to read the genetic code, bringing about tremendous progress in our understanding of how biology is encoded in DNA and in medical diagnostics. However, while the throughput of NGS has improved by orders of magnitude compared to Sanger sequencing, NGS is limited to reading DNA in short segments of 150-300 nucleotides at a time. Biology, unfortunately, is not encoded in stretches of 150 nucleotides, but in much longer segments of DNA and RNA spanning many thousands of nucleotides.

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Twist Bioscience, Monday, June 17, 15.00-16.30 hrs, Room A-2 – Level 1

Believe in Better: Leading the Way in Target Enrichment

Twist Bioscience uses its proprietary DNA synthesis platform to build target enrichment panels for researchers performing next-generation sequencing experiments and analysis. Due to the precision of these Twist capture probes, researchers are able to considerably improve the accuracy of the downstream sequencing analysis and perform fewer sequencing runs per sample without sacrificing performance, saving them time and money.

During this workshop, presenters will discuss various applications and present data from their work using Twist NGS Target Enrichment Solutions.

Twist Technology: Improving Sequencing on Challenging Samples

Renata Pellegrino, PhD, Technical Director, The Children’s Hospital of Philadelphia, USA

Liquid biopsies in metastatic prostate cancer: challenges, possibilities and clinical applicability

Johan Lindberg, PhD, Senior Researcher, Department of Medical Epidemiology and Biostatistics, Karolinska Institutet, Sweden

The utility of exome sequencing analysis in solving rare genetic disorders: the Exeter experience

Julia Baptista, PhD, Clinical Scientist/Honorary Lecturer, Royal Devon and Exeter NHS Foundation Trust, UK
### PROGRAMME BUSINESS AND ANCILLARY MEETINGS

As per date of printing.

#### Friday, June 14, 2019

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<thead>
<tr>
<th>Time</th>
<th>Meeting</th>
<th>Room</th>
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<tbody>
<tr>
<td>08.00 – 18.00 hrs</td>
<td>UEMS Exams .</td>
<td>R24+R25</td>
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<tr>
<td>09.00 – 13.00 hrs</td>
<td>ESHG Executive Board Meeting</td>
<td>J1</td>
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<tr>
<td>13.30 – 18.00 hrs</td>
<td>ESHG Board Meeting I .</td>
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#### Saturday, June 15, 2019

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<tr>
<td>09.00 – 13.30 hrs</td>
<td>ESHG Quality Subcommittee Meeting</td>
<td>J1</td>
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<tr>
<td>09.00 – 13.30 hrs</td>
<td>ESHG PPPC Meeting</td>
<td>R4</td>
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<tr>
<td>10.00 – 18.30 hrs</td>
<td>EB MG Exams</td>
<td>R5</td>
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<tr>
<td>12.00 – 14.00 hrs</td>
<td>GDPR Code of Conduct for Health Research Meeting</td>
<td>R3</td>
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<tr>
<td>12.15 – 13.45 hrs</td>
<td>ESHG Junior Branch Meeting</td>
<td>H2</td>
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<tr>
<td>16.00 – 18.30 hrs</td>
<td>GDPR Code of Conduct for Health Research Meeting</td>
<td>R3</td>
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#### Sunday, June 16, 2019

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<th>Meeting</th>
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<tbody>
<tr>
<td>08.15 – 10.45 hrs</td>
<td>European Network of Genetic Nurses and Counsellors Meeting</td>
<td>G4</td>
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<tr>
<td>10.00 – 17.00 hrs</td>
<td>EB MG Exams</td>
<td>R5</td>
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<tr>
<td>10.00 – 10.45 hrs</td>
<td>ESHG-SpringerNature Meeting</td>
<td>R4</td>
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<tr>
<td>10.00 – 11.00 hrs</td>
<td>GDPR Code of Conduct for Health Research Meeting</td>
<td>R4</td>
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<tr>
<td>11.00 – 13.00 hrs</td>
<td>National Human Genetics Societies Meeting</td>
<td>G4</td>
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<tr>
<td>11.15 – 12.45 hrs</td>
<td>GENIDA project Advisory Board</td>
<td>J1</td>
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<tr>
<td>11.15 – 13.15 hrs</td>
<td>Genetic Testing Across ERNs Meeting</td>
<td>R4</td>
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<tr>
<td>13.00 – 14.00 hrs</td>
<td>GN GC professional branch meeting</td>
<td>R3</td>
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<tr>
<td>13.30 – 15.15 hrs</td>
<td>EB MG Clinical Laboratory Geneticists (CLG) Meeting</td>
<td>G4</td>
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<tr>
<td>13.50 – 19.30 hrs</td>
<td>GDPR Code of Conduct for Health Research Meeting</td>
<td>R4</td>
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<tr>
<td>16.30 – 18.00 hrs</td>
<td>Building Bridges ESHG/ASHG Meeting</td>
<td>R3</td>
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<tr>
<td>16.30 – 18.00 hrs</td>
<td>ERM-ITHACA Ancillary Meeting</td>
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<tr>
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<td>J1</td>
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<tr>
<td>19.30 – 20.30 hrs</td>
<td>ESHG Membership Meeting</td>
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#### Monday, June 17, 2019

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<tr>
<td>08.00 – 09.00 hrs</td>
<td>EBMG BMGG Boards Meeting</td>
<td>J1</td>
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<td>08.30 – 10.30 hrs</td>
<td>ESHG Education Committee Meeting</td>
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<tr>
<td>09.00 – 10.00 hrs</td>
<td>UEMS SMG Boards Meeting</td>
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<td>10.00 – 13.00 hrs</td>
<td>UEMS Section Meeting</td>
<td>J1</td>
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<tr>
<td>10.00 – 11.00 hrs</td>
<td>EJHG Editorial Board Meeting</td>
<td>R4</td>
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<tr>
<td>10.15 – 11.15 hrs</td>
<td>ESHG/ASHG Leadership</td>
<td>R5</td>
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<tr>
<td>10.30 – 13.00 hrs</td>
<td>GDPR Code of Conduct for Health Research Meeting</td>
<td>R3</td>
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<tr>
<td>11.45 – 12.45 hrs</td>
<td>ESHG Board Meeting II</td>
<td>G4</td>
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<tr>
<td>12.00 – 14.00 hrs</td>
<td>Editorial Board Meeting for the European Journal of Medical Genetics</td>
<td>R4</td>
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<tr>
<td>13.00 – 15.00 hrs</td>
<td>EB MG General Assembly</td>
<td>J1</td>
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<tr>
<td>13.15 – 15.15 hrs</td>
<td>GenQA Educational Rapid Prenatal Aneuploidy Testing Workshop</td>
<td>G4</td>
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<tr>
<td>15.00 – 16.00 hrs</td>
<td>IFHGS Executive Board Meeting</td>
<td>R4</td>
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<tr>
<td>16.00 – 18.00 hrs</td>
<td>ICHG 2021 ISPC</td>
<td>R4</td>
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#### Tuesday, June 18, 2019

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<tr>
<th>Time</th>
<th>Meeting</th>
<th>Room</th>
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<tbody>
<tr>
<td>12.15 – 13.15 hrs</td>
<td>ESHG SPC Meeting</td>
<td>J1</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.
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 on Tuesday, June 18, 2019 at 13.30 hrs in Lecture Hall C.

Mendel Lecturers

2019 Craig Venter
2018 Emanuelle Charpentier
2017 George Church
2016 Sir Adrian Bird
2015 Thomas Südhof

2014 Mario Capecchi
2013 Huda Zoghbi
2012 Evan Eichler
2011 Elizabeth H. Blackburn
2010 Mary Claire King

2009 Sir John Burn
2008 Leroy Hood
2007 Aaron J. Ciechanover
2006 Sydney Brenner

The Mendel Award was designed by Swedish geneticist Alicia Bergsten.

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 on Tuesday, June 18, 2019 at 14.15 hrs in Lecture Hall C.

Award Holders

2019 Cisca Wijmenga
2018 Matthew Hurles
2017 Edith Heard
2016 Stefan Mundlos
2015 Svante Pääbo
2014 Sir Michael Stratton
2013 Felix Mitelman
2012 Peter Lichter
2011 GertJan B. van Ommen
2010 Sir Alec Jeffreys

2009 Kari Stefansson
2008 Arnold Munnich
2007 Andrea Ballabio
2006 Veronica van Heyningen
2005 Stylianos Antonarakis
2004 Bernhard Horsthemke
2003 Sir Peter S. Harper
2002 Albert de la Chapelle
2001 Robin Winter
2000 Dirk Bootsma

1999 Pat Jacobs
1998 Jean-Louis Mandel
1997 Leena Peltonen
1996 Malcolm Ferguson-Smith
1995 Jean Weissinbach
1994 Mary Lyon
1993 Pierre Maroteaux
1992 Lore Zech
Cisca Wijmenga is the Lodewijk Sandkuijl Professor of Human Genetics at the University of Groningen and the University Medical Centre, Groningen, The Netherlands. She will be giving the ESHG Award Lecture on Tuesday, June 18 at 14.15 hrs. She talked to Mary Rice about her life and work.

“I have always been interested in diseases and what causes them, starting as a child when I used to read the medical encyclopaedia at home for fun,” says Cisca Wijmenga. She wanted to become a medical doctor, but at the time entrance to medical school in The Netherlands was drawn by lot, and she missed her chance.

However, she is not someone who is easily discouraged. “For me it always has been important to go into new territories and try completely new things. At moments that is really scary, but it is also the only way forward to make progress.” Encouraged to continue by her parents, she turned to biology. “I was the first person in my family to go to university. My father in particular was very proud of that, as he always wanted to go to university but was not allowed to do so by his parents.”

She soon realised that biology might be an even better fit, as it focused more on disease mechanisms and molecular biology. “I loved molecular biology and biochemistry and eventually ended up in genetics.”

After her first degree at the University of Groningen, she acquired a PhD at Leiden University and then decided that she wanted to continue her career in the US. After several job offers, she decided on a post at the National Human Genome Research Institute at NIH, working with Francis Collins. “He turned out to be the kind of leader I wanted to be myself: always available and enthusiastic about new talent.”

When her time in the States was up, she had to choose between two projects – one predictable and well mapped out in advance, and the other that was not yet in existence. Collins encouraged her to go for the latter, saying that the ability to jump in at the deep end was important in a scientific career. She took his advice and returned to The Netherlands to begin work on complex disease genetics at the University of Utrecht. “This was completely new territory for me.”

Persistence and willingness to take risks are important qualities in scientific research, says Wijmenga, but luck and instinct play a part too. “I simply had a hunch to send off a grant application to look at a genetic relation between autoimmune disease and gluten intolerance.” That hunch paid off, and in 2007 she was the first to show that coeliac disease is genetically very similar to other autoimmune disorders, a hypothesis that is now widely accepted.

A source of great pride is the fact that her Professorship is named after her scientific mentor and friend Lodewijk Sandkuijl, a statistical geneticist who died aged only 49. And she is enthusiastic about the continuing evolution of her work. “I am very happy about the different directions my research has taken. As a PhD student I started to unravel the genetics of a mendelian disorder (FSHD, a rare form of muscular dystrophy), and then moved into the field of complex disease genetics and am now working on the gut microbiome and making complex personalised disease models on tiny chips.”

What pleases her less, though, is the pressure now put on young scientists that makes it far more difficult for them to carry out curiosity driven research and research that takes time. “When the number of publications becomes important for your career then publishing becomes a goal rather than a method to share your results with the scientific community,” she says.

Outside work, Wijmenga likes art and going to museums and art fairs. “I also like hiking, in particular in the mountains. I like bike rides in the beautiful surroundings of Groningen. I love to cook and enjoying reading a wide variety of books.”

In her prize lecture, she will take pleasure in describing the scientific adventure she took that led her from complex disease genetics with a focus on coeliac disease, to the gut microbiome and “the really cool work we just started on gut-on-a-chip to recreate coeliac disease on a chip. With that in place, in future we can study the interaction between host genome, epithelial barrier, immune system and the gut microbiome.”

Retirement? She hasn’t really thought about it yet, partly because she’s too young and partly, one suspects, because she sees a long and exciting scientific adventure still unfolding in front of her.
Professor Craig Venter

Professor Craig Venter is founder, chairman, and CEO of the J. Craig Venter Institute, La Jolla, California, USA. He will be giving the Mendel lecture on Tuesday at 13.00 hrs. He talked to Mary Rice about his life and work.

“From a very young age I was interested in the natural world around me and given that it was the 1950’s parents allowed their kids to just play and explore on their own,” says Craig Venter. “I revelled in that and would spend all day outside. I was also very interested in building things. I was an avid reader of Scientific American and would build things I saw in there. I was not, however, a good student and really hated the school system because it relied on rote memorization and regurgitation of facts which was the worst way for me to learn. In short, my early years were hardly a model of discipline and direction.”

Being drafted into the navy during the Vietnam War changed all that. “Although I was opposed to the war, I had no choice but to join up. I worked in the intensive care ward of a field hospital, where I saw people suffering and dying every day,” he says. “I was one of the lucky ones who served there and returned. It taught me a lot, the first thing being that the worst thing you can lose is your life, but also that taking risks and suffering setbacks are part of moving forward.” And it instilled in him the desire to study medicine.

Following his first degree in biochemistry at the University of California, San Diego, where he studied under the biochemist Nathan O. Kaplan, he received a PhD in physiology and pharmacology; “I very quickly learned to love excelling at school and went on to get my undergrad and PhD in record time,” he says. After moving to become a professor at the State University of New York, Buffalo, he joined the US National Institutes of Health in 1984.

At NIH he developed an innovative DNA sequencing machine and became the first in the world to publish a paper containing data obtained by an automated sequencing method. “My career in science would never be the same again,” he says. Around this time he first became involved in the discussions of a project that would eventually propel his research into the limelight – the Human Genome Project.

Frustrated with the approach and the slow progress of the publicly-funded HGP, he sought funding from the private sector to create Celera Genomics. Using shotgun sequencing, Celera caught up rapidly with the international project, and in 2000 they shared the credit for the mapping of the human genome.

Since that day, Venter has not stopped in his quest for new knowledge. In 2005 he co-founded Synthetic Genomics, a company dedicated to the creation of modified microorganisms to produce clean fuels and biochemical. In 2010, a team of scientists from the company became the first to create ‘synthetic life’, a single-celled organism including, among other things, its own email address.

“The science of synthetic genomics is having and will continue to have a profound impact on human existence, including new chemical and energy generation, human health and medical advances, clean water and food production, positive environmental impact, and possibly even our evolution,” he says.

Retirement is a taboo subject. “I love what I do and see no reason to ever stop – for me, retirement equals death. But I will say that over the last two years I have come to enjoy and see the need for a healthy work/life balance and do things that are non-science related too like sailing and flying.”
ESHG Young Investigator Awards

The Scientific Programme Committee has shortlisted presenters for the ESHG Young Investigator Awards. The committee will judge the finalists’ presentations during the conference. The following awards will be presented to the winners in the closing ceremony on Tuesday, June 18, 2019 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 **Isabelle Oberlé Award** is awarded yearly since 2002 for best presentation by a young scientist on research concerning the genetics of intellectual disability.
- 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 genetics therapy of genetic diseases.
- The **Mia Neri Award** (funded by the Mia Neri Foundation) will be awarded to the best presentation in 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 a grey background in the detailed scientific programme on the previous pages.

We conducted a short interview with each candidate.

These interviews can be found on our website:

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 complimentary 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.

The short interviews with the finalists can be found on the website:
PROGRAMME  YOUNG INVESTIGATOR AWARD CANDIDATES

Saturday, June 15 at 16:30 hrs

PL2.3  Trin Laisk
Tartu, Estonia

PL2.4  Joanna Kaplanis
Cambridge, United Kingdom

PL2.5  Lila Allou
Berlin, Germany

Saturday, June 15 at 18:30 hrs

C01.2  Mamiko Yamada
Shinjuku, Japan

C01.3  Uira Melo
Berlin, Germany

C01.2  Max Schubach
Berlin, Germany

C02.5  Jelena Pozojevic
Lübeck, Germany

C03.2  Michele Arnoldi
Trento, Italy

C03.3  Jun Sung Park
Daejeon, Korea, Republic of

C03.4  Daphne Smits
Rotterdam, Netherlands

C03.5  Ian Tully
Cardiff, United Kingdom

C04.4  Manon Oud
Nijmegen, Netherlands

C04.5  Digumarthi Sudhakar
Habuguda, India

C05.3  Helen Roessler
Utrecht, Netherlands

C05.4  Sara Cuvertino
Manchester, United Kingdom

C06.2  Vadim Dolgin
Beer Sheva, Israel

C06.3  Eline Blommaert
Leuven, Belgium

C06.4  Youmna Ghaleb
Paris, France

C06.5  Pasquelena De Nittis
Lausanne, Switzerland

C07.2  Ivy van Dijke
Amsterdam, Netherlands

C07.4  Karuna van der Meij
Amsterdam, Netherlands

C07.2  Vivien van der Meij
Amsterdam, Netherlands

C07.4  Karuna van der Meij
Amsterdam, Netherlands
PROGRAMME YOUNG INVESTIGATOR AWARD CANDIDATES

Sunday, June 16 at 13.00 hrs

C08.2
Ummi Abdullah
Oxford, United Kingdom

C08.3
Wang Yicong
Shenzhen, China

C08.4
Chelsea Lowther
Boston, United States

C08.5
Julia van Campen
London, United Kingdom

C09.3
Maria Palmieri
Siena, Italy

C09.5
Julika Borde
Cologne, Germany

C10.2
Ilse Luyckx
Edegem, Belgium

C10.3
Ambra Sartori
Geneva, Switzerland

C11.2
Ninon Mounier
Lausanne, Switzerland

C11.3
Hieab Adams
Rotterdam, Netherlands

C11.4
Maarja Lepamets
Tartu, Estonia

C11.5
Ross Byrne
Dublin, Ireland

C12.2
Enrico Konrad
Erlangen, Germany

C12.3
Lot Snijders Blok
Nijmegen, Netherlands

C12.4
Lianmin Chen
Groningen, Netherlands

C12.5
Jonas Bovijn
Oxford, United Kingdom

C13.4
Maria Santos
Madrid, Spain

C13.5
Henrike Heyne
Cambridge, United States

C13.6

C13.7

Monday, June 17 at 13.00 hrs

C16.2
José García Peláez
Porto, Portugal

C16.3
Inge Lakeman
Leiden, Netherlands

C16.4
Nina Mars
Helsinki, Finland

C16.5
Heather Andrighetti
Toronto, Canada

C17.3
Rita Barbosa-Matos
Porto, Portugal

C17.4
Laëtitia Meulemans
Rouen, France

C17.5
Dhanya Ramachandran
Hannover, Germany

C18.2
Stephanie Newman
London, Canada

C18.3
Petros Patsalis
Nicosia, Cyprus

C18.4
Daniel Whisenant
Huddinge, Stockholm, Sweden

C18.5
Susanna Croci
Siena, Italy

C19.2
Maria Pettersson
Solna, Sweden
PROGRAMME

YOUNG INVESTIGATOR AWARD CANDIDATES

Monday, June 17 at 13.00 hrs

- Lisanne Vervoort
  Leuven, Belgium

- Malin Kvamnun
  Stockholm, Sweden

- Mahsa Shabani
  Leuven, Belgium

Tuesday, June 18 at 11.00 hrs

- Sara Bizzotto
  Boston, United States

- Laurens Wiel
  Nijmegen, Netherlands

- Juliette Pulman
  Paris, France

- Tatiane Yanes
  Sydney, Australia

- Justin Rendleman
  New York, United States

- Helge Martens
  Hannover, Germany

- Inga Patarcic
  Berlin, Germany

- Janine Vetsch
  Kensington, Australia

- Natalia Mendoza Ferreira
  Cologne, Germany

- Mathilda Bedin
  Paris, France

- Hanne Hoskens
  Leuven, Belgium

- Dhanya Lakshmi Narayanan
  Manipal, India

- Alison Young
  Sydney, Australia

- Elisa Fernández-Núñez
  Madrid, Spain

- Danya Vears
  Parkville, Australia
PROGRAMME POSTER AWARD FINALISTS

**GENERAL**

- Natalia Pervjakova
  Tartu, Estonia
- Ronja Hollstein
  Bonn, Germany
- Reza Ataeijaliseh
  Tehran, Iran, Islamic Republic of
- Martin Becker
  Solna, Sweden
- Seyed Ali Safizadeh Shabestari
  Dubai, United Arab Emirates
- Bethany Wild
  London, United Kingdom
- Christian Mertes
  Garching, Germany
- Adriaan van der Graaf
  Groningen, Netherlands

**SATURDAY**

- Alexandra Filatova
  Moscow, Russian Federation
- Minttu Marttila
  Helsinki, Finland
- Abdulrahman Ali
  Dubai, United Arab Emirates
- Daniel Halperin
  Beer-Sheva, Israel
- Tamara Simakova
  St. Petersburg, Russian Federation
- Verena Heinrich
  Berlin, Germany
- Bettina Zimmermann
  Basel, Switzerland

**SUNDAY**

- Anna Morgan
  Trieste, Italy
- Diana Le Duc
  Leipzig, Germany
- Moeen Riaz
  Melbourne, Australia
- Pauline Schneeberger
  Hamburg, Germany
- Heidi Marjonen
  Helsinki, Finland
- Simon Fishilevich
  Rehovot, Israel
- Jane Tiller
  Melbourne, Australia

**MONDAY**

- Anna Morgan
  Trieste, Italy
- Dylan Lawless
  Lausanne, Switzerland
- Moeen Riaz
  Melbourne, Australia
- Pauline Schneeberger
  Hamburg, Germany
- Heidi Marjonen
  Helsinki, Finland
- Simon Fishilevich
  Rehovot, Israel
- Jane Tiller
  Melbourne, Australia

**TUESDAY**

- Anna Morgan
  Trieste, Italy
- Stijn Van de Sompele
  Ghent, Belgium
- Alexej Knaus
  Bonn, Germany
- Christopher Hübel
  Stockholm, Sweden
- Guillaume Jouret
  Reims, France
- Kevin Cassinari
  Rouen, France
- Brunilda Balliu
  Los Angeles, United States
- Philip Jansen
  Amsterdam, Netherlands

**SAT话语**

- Nataša Pervjakova
  Tartu, Estonia
- Ronja Hollstein
  Bonn, Germany
- Reza Ataeijaliseh
  Tehran, Iran, Islamic Republic of
- Martin Becker
  Solna, Sweden
- Seyed Ali Safizadeh Shabestari
  Dubai, United Arab Emirates
- Bethany Wild
  London, United Kingdom
- Christian Mertes
  Garching, Germany
- Adriaan van der Graaf
  Groningen, Netherlands

**SATURDAY**

- Alexandra Filatova
  Moscow, Russian Federation
- Minttu Marttila
  Helsinki, Finland
- Abdulrahman Ali
  Dubai, United Arab Emirates
- Daniel Halperin
  Beer-Sheva, Israel
- Tamara Simakova
  St. Petersburg, Russian Federation
- Verena Heinrich
  Berlin, Germany
- Bettina Zimmermann
  Basel, Switzerland

**SUNDAY**

- Anna Morgan
  Trieste, Italy
- Dylan Lawless
  Lausanne, Switzerland
- Moeen Riaz
  Melbourne, Australia
- Pauline Schneeberger
  Hamburg, Germany
- Heidi Marjonen
  Helsinki, Finland
- Simon Fishilevich
  Rehovot, Israel
- Jane Tiller
  Melbourne, Australia

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  Trieste, Italy
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  Ghent, Belgium
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  Bonn, Germany
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  Reims, France
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  Rouen, France
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  Los Angeles, United States
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  Amsterdam, Netherlands

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  Reims, France
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  Rouen, France
- Brunilda Balliu
  Los Angeles, United States
- Philip Jansen
  Amsterdam, Netherlands
INFORMATION

GENERAL INFORMATION
REGISTRATION FEES
NETWORKING EVENTS
CORPORATE EXHIBITION
**Registration and Opening Hours**

**Opening Hours Registration and Preview Centre**
- Friday, June 14: 14.00 – 19.00 hrs
- Saturday, June 15: 07.30 – 20.15 hrs
- Sunday, June 16: 08.00 – 19.30 hrs
- Monday, June 17: 08.00 – 19.30 hrs
- Tuesday, June 18: 08.30 – 14.00 hrs

**Opening Hours Exhibition and Poster Area**
- Saturday, June 15: 09.30 – 18.30 hrs
- Sunday, June 16: 09.00 – 17.45 hrs
- Monday, June 17: 09.00 – 17.45 hrs
- Tuesday, June 18: 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.
Exhibitors also receive badges that allow access to the appropriate areas.
Lost badges can be replaced at the registration desk. However, a handling fee of EURO 50.- 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 4, 2019: 75% refund
- between April 5 and May 23, 2019: 25% refund
- after May 23, 2019: no refund
The date of the email/fax ID is the basis for considering refunds. Refunds will be made after the congress.
No refunds can be made for a cancellation received after May 23, 2019, independent of the reason for the cancellation.
No exceptions to the refund policy can be made, including health or family issues.

**Insurance**
By registering to the ESHG 2019 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.

**IMPORTANT NOTICE**
Certificates of attendance will be issued electronically via email after the conference to all participants.

**Programme**

**ESHG Mobile App**
The mobile app with the full programme and other useful information is be available for download. Please download the ESHG Society app from your App- or Play Stores, which also contains the conference data.

**Preview Centre**
Equipment for a final check of the sequence of your presentation is available in the room G1 – Preview Centre (on the second level).
All presenters should bring their electronic presentation to the Preview Centre not later than 2 hours before the start of the session (30 minutes for the first morning sessions or the day prior to your presentation if possible).

**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, June 17, 2019: 16.45 – 17.45 hrs (strict!)
- Group D: Monday, June 17, 2019: 17.45 – 17.50 hrs (strict!)
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.

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

**Coffee Breaks**
During the exhibition opening hours, refreshments (coffee, tea and water) are served free of charge to participants wearing name badges.
Snacks and Lunch will be served in the exhibition area during designated break times. Outside the official coffee breaks, free coffee and tea are available in the exhibition hall.
On Tuesday the coffee breaks and lunches can be found in Aisle F, Aisle G and Hall H.

**Pre-ordered 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**  
Swedish Exhibition & Congress Center  
Mässans Gata/Korsvägen  
412 94 Göteborg  
Sweden

**Car Parking**  
There is a parking lot right in front of the conference venue accessible through entrance 2.

**Cloakroom and Luggage**  
A cloakroom and luggage storage are available directly at the entrance 2 below registration (Level 0).

**WiFi**  
WiFi is available throughout the conference venue. Network ID: eshg2019, password: eshg2019

**Staff**  
If you should have any questions, the congress staff (recognizable by a pink lanyard) will be pleased to help you.

**Public transport**

**By Air**  
Landvetter Airport  
The Flygbussarna airport coaches run to and from Landvetter airport with departures of up to every 12 minutes (20 min. journey) and stop at Korsvägen/the Swedish Exhibition & Congress Centre, right opposite the hotel. This is the first and last stop when travelling from and to the airport.

**By Train**  
From Gothenburg Central Station take tram 2 (for Krokslätt) or tram 4 (for Mölndal). From Brunnsparken you can also take tram 5 (for Torp). The trams stop on Korsvägen right opposite the hotel. You can find timetables for these trams on the Västtrafik website.

**By Car**  
GPS coordinates: Lat: N 57º 41' 53.06" Long: E 11º 59' 20.01"  
E6/E20 (from the north) and road 45 (from Karlstad): On arriving in Gothenburg, turn off when you see the sign "Mässan Scandinavium Liseberg". Take the first exit on the left, then turn right on to Örgrytevägen and then take the next right exit for the hotel.  
E6/E20 (from the south) and road 40 (from Borås): On arriving in Gothenburg, turn off when you see the sign "Mässan Scandinavium Liseberg". Take the second exit on the right for the Gothia Towers hotel.  
During your stay you can choose from a number of convenient parking options. All parking options are extremely popular, so to guarantee a parking space for your car, you will need to pre-book parking.

**Conference Policy**

**Late programme changes**  
All contents are up-to-date as per date of printing.  
For changes in the scientific programme which occurred after the printing deadline, please consult the website: https://2019.eshg.org/index.php/programme/late-programme-changes/

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

**Smoking Policy**  
The ESHG 2019 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.

**Social Media Guidelines**  
Please see our full policy on our website. https://2019.eshg.org/index.php/mediaonlinepolicy/  
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.
**Sustainability**

The ESHG Conference is committed to seek all possible ways to host an event that bears in mind the responsible use of resources, our environment and the use of sustainable conference materials. Hence, we would like to inform you of the range of “green points” at our 2019 conference.

**Venue and Location**

The city of Gothenburg is currently the world’s most sustainable destination, according to the Global Destination Sustainability Index 2016, 2017 and 2018. Our venue for ESHG 2019, The Swedish Exhibition & Congress Centre, has greatly contributed to this by being part of the Fairtrade City Gothenburg. This means that they aim to serve their guests a large range of fair-trade products, among many others, coffee from a Swedish roastery and the use of sustainable conference materials. Hence, we would like to inform you of the range of “green points” at our 2019 conference.

Furthermore, they endeavour to supply their restaurants with organic, locally produced ingredients on fair terms. This is the basis on which they choose between otherwise equivalent products. Nowadays their wine list also includes many organic options. Also note that, should you have breakfast at the adjacent Gothia Towers Hotel, the honey you will spread on your bread, originates from their own hives located on the roof of one of the towers.

The Swedish Exhibition & Congress Centre cooperates with the *Gothenburg Rescue Mission*, a non-for-profit organisation helping people on the margins of society. As part of this cooperation the congress centre buys products such as jam, marmalade and juices from the Mission’s organic farm. They also donate unsold food and drinks to the Mission’s café and arrange an annual Christmas collection for the homeless and needy.

For more information about the sustainability efforts of our ESHG 2019 venue please visit [https://en svenskamassan.se/explore-us/about-us/sustainability/](https://en svenskamassan.se/explore-us/about-us/sustainability/)

**Congress Materials**

**Bags**

Since 2014 the ESHG Conference has aimed to provide congress bags that are made from durable materials, such as cotton. This is aimed at increasing the re-usability of the bags and decreasing the amount of waste created, and in the best case, making it biodegradable.

**Pens**

Your pen is made from recycled PET-bottles! As previously mentioned, we are committed to using the recourses responsibly. For this reason, we have, for the last 7 years, opted for conference pens made from recycled PET bottles, which would otherwise have ended up as waste.

**Printed Materials**

In this area ESHG has been making hard efforts to reduce its printed materials over time, while ensuring to keep high standards for its conference participants. The ESHG stopped printing the abstract book 8 years ago. This 500+ pages publication totalised around 5 tons of ink and paper each year. Instead we implemented the online programme planner as well as the conference app, which we continue to improve over the years, and have been adopted by our participants. The convenience in using these tools as preparation ahead of the meeting is a quasi standard today, however the fringe benefits of reducing the number of produced items seems less obvious, but definitely contributes to the whole.

Starting with ESHG 2019, we have implemented an “self-printing” approach for registration materials. By using this procedure, we have been able to reduce the registration materials of each participant to a single sheet of paper. Moreover, all participants will receive their confirmation of attendance and CME certificate (for more information on how to obtain CME credits please refer to page 7) as pdf via email after the conference. These initiatives will help us reduce more than 8000 printouts and other materials, having a significant impact in the amount of paper and ink used for the ESHG conference and in consequence largely reducing the CO₂ footprint of the transport of all print work from Vienna to the conference location.

Concerning printing of posters, the ESHG has been introducing e-Posters to its poster exhibition. Currently more than 400 posters are available only in an electronic version located in the exhibition hall next to the live stream. This has not only helped in reducing the amount of printed posters but provide a new experience in terms of interaction between the participants and abstracts being presented.

**Professional Congress Organiser**

The organiser of the European Human Genetics conference, the Vienna Medical Academy GmbH, has been officially certified as “green meeting organiser” by the Federal Ministry of Sustainability and Tourism in Austria. The official label “Green Meeting” is currently only available in Austria, but the Vienna Medical Academy always applies the same standards and strives to ensure that resources are used responsibly and efficiently during the meetings it organises, regardless of their location.

**YOU!**

Keep in mind that you as participant play an important role in the responsible use of the resources at the ESHG conferences. You can help us to do this by recycling your waste, using only the necessary amount of water and eating only those catering items that you know you will completely consume. Furthermore, you can contribute to reduce the amount of waste created by reusing your conference materials, such as conference bag and pen, as long as possible. Together with your efforts and ours we can continue to strive for a more environmentally friendly ESHG conference.
Gothenburg – General Information

Bank services – Money matters
Banks are generally open Mon-Fri 10.00-15.00. Additional opening hours apply in the afternoon at least once a week. All banks are closed on public holidays. Major credit and debit cards are widely accepted in shops, restaurants, hotels and taxis. Restrictions may apply to American Express and Diners. You can use your Visa, MasterCard, Maestro or Cirrus card at any ATM (“bankomat” or “uttagsmaskin”). The monetary unit is the Swedish Krona (SEK). There is no limit on the amount of Swedish and foreign currency taken into Sweden. Some shops, restaurants, museums, hotels and other establishments accepts the Euro but will give you change in kronor.

The official currency in Sweden are Swedish Krona.
1 SEK = 0.09 EUR = 0.08 GBP = 0.10 USD = 0.14 CAD = 11.44 JPY = 0.10 CHF = 0.15 AUD as per May 23, 2019.

V.A.T.
The VAT rate in Swedish is 25%. The ESHG charges VAT on the registration fees. All stated prices charged by the ESHG include VAT.

Emergency Services
European Emergency Number: 112

Pharmacies – Medical Emergencies
There are several different pharmacies (apotek) in Gothenburg and you find one in almost every block. Opening hours between 10.00 and 18.00 and on weekends until 14.00. Apoteket Hjärtat, located in the shopping centre Nordstan, is open until 22.00. Many supermarkets carry non-prescription supplies such as band aid, antiseptics and painkillers. The Medical Information Service (Sjukvårdsrådgivningen) is a 24-hour provider of free healthcare information and consulting. Tel +46 1177. For medical treatment, visit the nearest health centre (vårdbcentrale).
The emergency ward (akutmottagning) is located at the Sahlgrenska University Hospital or Östra Sjukhuset (Eastern Hospital). Bring your passport and your European Health Insurance Card.

Safety – Crime
Gothenburg is 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 public transportation 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
Country code: +46 Gothenburg area code: 031. There is excellent wireless GSM and 3G/UMTS/LTE coverage in Sweden. Americans will need a tri-band phone. Pay phones are rare. They require either a prepaid phone card or a credit/debit card, or Swedish or Euro coins. Phone cards are available at most newsagents and grocery stores.

GSM Cell Phone Roaming
Roaming charges within the European Union have officially been abolished. The EU “roam like at home” rules mean that when you use your mobile phone while travelling outside your home country in any EU country you don’t have to pay any additional roaming charges. With a EU phone contract, you benefit from these rules when calling (to mobile and fixed phones), sending text messages (SMS) and using data services while abroad.

Time Zone
UTC+2 hour. Daylight Saving Time is used from the last Sunday of March to last Sunday of October.

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

Electricity Supply
The electrical current in Sweden is 220 V/50 Hz. Round European-style two-pin Schuko plugs (type F/K) are used.

Taxis
Taxis can be hailed in the streets. Prices vary from company to company. Ask for a fixed price when travelling to or from the airport.
Taxi Göteborg +46 (0)31-65 00 00 • Taxi Kurir +46 (0)31-27 27 27 • Mini Taxi +46 (0)31-14 01 40

Tipping
Most service bills that you receive already include gratuity so tipping isn’t necessary (but always appreciated). If you’re at a restaurant, it is recommended to give a small tip of maximum 10% of your bill.

IMPORTANT NOTICE
In order to obtain CME credits, participants seeking these credits will need to scan their badges daily at the General Information Desk located at the registration area. Please note that if you miss to scan your badge you will not receive the credits accredited for the day.
### INFORMATION

#### REGISTRATION FEES

<table>
<thead>
<tr>
<th>Registration Fees¹</th>
<th>Payment received:</th>
<th>before April 4, 2019 (reduced rate)</th>
<th>from April 4 to May 23, 2019 (regular rate)</th>
<th>after May 23, 2019 and on site</th>
<th>Day Tickets on site</th>
</tr>
</thead>
<tbody>
<tr>
<td>Participants ESHG Members</td>
<td>EUR 330.-</td>
<td>EUR 440.-</td>
<td>EUR 500.-</td>
<td>EUR 165.-</td>
<td></td>
</tr>
<tr>
<td>Participants Non-Members</td>
<td>EUR 500.-</td>
<td>EUR 600.-</td>
<td>EUR 670.-</td>
<td>EUR 230.-</td>
<td></td>
</tr>
<tr>
<td>Postgraduate Trainees ESHG Members²</td>
<td>EUR 220.-</td>
<td>EUR 320.-</td>
<td>EUR 385.-</td>
<td>EUR 145.-</td>
<td></td>
</tr>
<tr>
<td>Postgraduate Trainees Non-Members²</td>
<td>EUR 330.-</td>
<td>EUR 440.-</td>
<td>EUR 500.-</td>
<td>EUR 165.-</td>
<td></td>
</tr>
<tr>
<td>Students⁴</td>
<td>EUR 115.-</td>
<td>EUR 170.-</td>
<td>EUR 220.-</td>
<td>EUR 100.-</td>
<td></td>
</tr>
<tr>
<td>ESHG Members from underprivileged countries⁵</td>
<td>EUR 220.-</td>
<td>EUR 220.-</td>
<td>EUR 220.-</td>
<td>EUR 100.-</td>
<td></td>
</tr>
<tr>
<td>Non-Members from underprivileged countries⁵</td>
<td>EUR 290.-</td>
<td>EUR 290.-</td>
<td>EUR 290.-</td>
<td>EUR 120.-</td>
<td></td>
</tr>
<tr>
<td>Lunch bags/boxes per day⁶</td>
<td>EUR 19.-</td>
<td>EUR 19.-</td>
<td>N/A</td>
<td>N/A</td>
<td></td>
</tr>
<tr>
<td>Participants/Guests</td>
<td></td>
<td></td>
<td></td>
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<tr>
<td>Students/Postgrad. Trainees</td>
<td></td>
<td></td>
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<td></td>
<td></td>
</tr>
<tr>
<td>Networking Evening at own expense</td>
<td>EUR 60.-</td>
<td>EUR 40.-</td>
<td></td>
<td></td>
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</tr>
</tbody>
</table>

¹Registration Fees include 25% Swedish VAT.
²Applies 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 at the registration desk. Confirmations handed in at a later stage cannot be considered.
³Applies to non-MD/PhD-Counsellors.
⁴Applies to undergraduate students. Please provide a copy of a Student’s ID or a confirmation signed by the head of department at the moment of your registration. Confirmations handed in at a later stage cannot be considered.
⁵Applies to a selected list of countries.
⁶Not available onsite.

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.

Please see also the General Terms & Conditions for participants: https://2019.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 Final Programme.
- Coffee/Tea during breaks from Saturday, June 15 to Tuesday, June 18.

**Payment of Registration fees**, may be made in EURO by:
- Credit Cards: Diners Club, Mastercard and Visa
- Cash in Euro

#### IMPORTANT Note

Only payments made in EUR will be accepted at the registration desk. MasterCard, VISA, Diners Club and Maestro cards are accepted both at the registration and in the rest of the conference venue. At the Networking Evening, only credit cards are accepted.

#### Cancellations and refunds

Notice of cancellation had to be made in writing by email or fax to the Congress Office. Registration fees may be refunded as follows:
- Written cancellation received:
  - before April 4, 2019: 75% refund
  - between April 5 and May 23, 2019: 25% refund
  - after May 23, 2019: 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 2019 participants agree that neither the organising committee nor the congress office assume any liability whatsoever.
Opening Networking Mixer

Saturday, June 15, 2019, 20.00 - 22.00 hrs – Halls D and E at the congress venue
Network with your colleagues at this mixer 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.
We would like to thank the City of Gothenburg and Region Västra Götaland for their kind support in hosting this event!

City of Gothenburg

ESHG Networking Evening (at own expense)

Monday, June 17, 2019, 20.00 hrs – Trädgår’n
The networking evening is a great opportunity to meet with friends and colleagues from around the world in a relaxed atmosphere, enjoying the unmatched charm and fascination of Gothenburg. Those who have shared this evening with us in previous years know, one would not want to miss it!

- Ticket: EUR 60.-
- Students: EUR 40.-

Dinner & 3 drinks are included in the price.
Dress code: casual

Please note that payment of extra drinks is only possible with credit/bank cards.

For directions on how to get to the Networking Evening venue please scan the QR code or see the map "Directions to Networking Evening venue" in the ESHG Mobile App.

Make sure to bring your Networking ticket with you! Tickets will be checked at the entrance of the venue.

IMPORTANT NOTICE

In order to obtain CME credits, participants seeking these credits will need to scan their badges daily at the General Information Desk located at the registration area. Please note that if you miss to scan your badge you will not receive the credits accredited for the day.
## List of Exhibitors

<table>
<thead>
<tr>
<th>General</th>
<th>Saturday</th>
<th>Sunday</th>
<th>Monday</th>
<th>Tuesday</th>
<th>Satellites</th>
<th>Awards</th>
</tr>
</thead>
<tbody>
<tr>
<td>10x Genomics</td>
<td>346</td>
<td></td>
<td></td>
<td></td>
<td></td>
<td></td>
</tr>
<tr>
<td>ACTIVE MOTIF</td>
<td>484</td>
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<tr>
<td>ADS Biotech</td>
<td>252</td>
<td></td>
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</tr>
<tr>
<td>Agilent Technologies</td>
<td>634</td>
<td></td>
<td></td>
<td></td>
<td></td>
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</tr>
<tr>
<td>American Society of Human Genetics - ASHG</td>
<td>426</td>
<td></td>
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<td></td>
<td></td>
</tr>
<tr>
<td>Amsterdam UMC, Laboratory of Genome Diagnostics</td>
<td>578</td>
<td></td>
<td></td>
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List correct as per date of printing - Exhibition floor plan is in your conference bag.
## Exhibition & Poster Area – Hall B – Dates & Opening Hours

<table>
<thead>
<tr>
<th>Date</th>
<th>Opening Hours</th>
<th>Activity</th>
</tr>
</thead>
<tbody>
<tr>
<td>Saturday, June 15</td>
<td>09.30 – 18.30 hrs</td>
<td>Poster mounting / viewing</td>
</tr>
<tr>
<td>Sunday, June 16</td>
<td>09.00 – 17.45 hrs</td>
<td>Poster viewing</td>
</tr>
<tr>
<td>Monday, June 17</td>
<td>09.00 – 17.45 hrs</td>
<td>Poster viewing</td>
</tr>
<tr>
<td>Tuesday, June 18</td>
<td>CLOSED</td>
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</table>

## Products & Services Index

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

## Exhibition Catalogue & Corporate Satellites Invites and Programmes

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

## Floor Plan – Exhibition & Poster Topics

You will find the floor plan of the Exhibition and Poster Topics in your conference bag or refer to page 70.

## Posters – Mounting, Viewing & Removal Schedules

Poster presentations will be held in the exhibition hall from 15 – 17 June.

Poster mounting, viewing and removal times are:

<table>
<thead>
<tr>
<th>Date</th>
<th>Opening Hours</th>
<th>Activity</th>
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<td>Poster viewing</td>
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<tr>
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<td>16.45 – 17.45 hrs</td>
<td><strong>Poster removal – Groups A–C (strict!)</strong></td>
</tr>
<tr>
<td></td>
<td>17.45 – 17.50 hrs</td>
<td><strong>Poster removal – Group D only (strict!)</strong></td>
</tr>
</tbody>
</table>

Posters not removed by 17.45 hrs on Monday, June 17 will be taken down and will not be stored or sent to authors after the meeting. 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.

## 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 break times there will be free coffee and tea in the Exhibition hall.

On Tuesday the coffee breaks and lunches can be found in Aisle F, Aisle G and Hall H.

The Cash Bar in the Exhibition hall is open during exhibition opening hours. The menu includes sandwiches, salads, pasta, drinks and special coffees. The menu is available at the Cash Bar. Payment in cash (EUR and SEK) or by credit card.

Pre-ordered lunch bags will be available during lunch breaks at the coffee break areas. 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 MUST to ask permission to scan the barcode on your badge.
  Refusal to have your badge scanned does not entitle a company to deny you access to their corporate satellite and/or to enter an activity at their stand.

- This barcode gives this company access to your contact details as follows (note: only in case you opted for this during the registration process AND/OR if agreed with the company scanning your badge):
  - name and full postal address
  - e-mail address

Thank you for your understanding and cooperation.

## Exhibition & Sponsorship Management

**Name** ROSE INTERNATIONAL

**Address**

P.O. Box 93260
NL-2509 AG The Hague, the Netherlands

**Telephone** +31 (0)70 383 89 01

**Fax** +31 (0)70 381 89 36

**E-mail** eshg@rose-international.com
The standard reference combining ISCN and HGVS nomenclature

**ISCN 2016**

An International System for Human Cytogenomic Nomenclature (2016)

Editors: Jean McGowan-Jordan, Annet Simons, Michael Schmid

Oslo 2019
5th Congress of the European Academy of Neurology
June 29 – July 2
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Neuroinflammation

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