ESHG

EUROPEAN SOCIETY OF HUMAN GENETICS

THE EUROPEAN SOCIETY OF HUMAN GENETICS

EUROPEAN HUMAN GENETICS CONFERENCE 2019

Svenska Mässan | Gothenburg – Sweden | June 15 – 18

PROGRAMME

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SVENSKA MÄSSAN
THE SWEDISH EXHIBITION & CONGRESS CENTRE | GOTHIA TOWERS

ESHG

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PROGRAMME

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Dear Colleagues and Friends,

Gunnar Houge, *President*
European Society of Human Genetics

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, showcasting 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

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

WELCOME

European Society of Human Genetics

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European Human Genetics Conference 2019

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GENERAL ACKNOWLEDGEMENTS - FUTURE MEETINGS

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

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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 CreditsTM. 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!

https://2019.eshg.org/index.php/programme/conference-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 Easten 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 annonced 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 honoured with an oral presentation in these sessions. Presenters are expected to explain their work and answer questions from the audience. Speaking time for concurrent session is 15 minutes including time for discussion.

Poster Viewing with Authors

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

Saturday, June 15, 2019

TIME	HALL C	K2+K3	F1+F2+F3	F4+F5	G2+G3	K1	H2	A3	A1	A5	A2	A4
08.00 - 10.00	**E01** New Technologies											
10.00 - 10.30	*Coffee Break / Poster Mounting / Exhibition*							MGI, a subsidiary of BGI Satellite			Cellecta Satellite	Dovetail Genomics Satellite
10.30 - 12.00	**W01** NGS in the Clinic - Mistakes and Quality Assurance	**E02** Epigenetics and cancer	**W02** Prenatal Diagnosis ELPAG	**E03** Bridging genomic discoveries and personalized medicine ELPAG	**W03** How to enhance your career - How to present - How to network	**E04** Gene Expression Resources	**W04** From genetics to new medicines: The open Targets Genetics Portal					
12.00 - 14.00	*Lunch Break / Free Poster Viewing / Exhibition*							Blueprint Genetics Satellite	Ionis Pharma-ceuticals / AstraZeneca Satellite	CENTOGENE Satellite	NimaGen Satellite	Congenica Satellite
14.00 - 14.30	Opening Welcome Address	Opening Welcome Address *Live Streaming*										
14.30 - 16.00	**PL1** Opening Plenary Session	**PL1** Opening Plenary Session *Live Streaming*										
16.00 - 16.30	*Fruit Break / Free Poster Viewing / Exhibition*											
16.30 - 18.00	**PL2** What's New? Highlight Session from submitted abstracts	**PL2** What's New? Highlight Session from submitted abstracts *Live Streaming*										
18.00 - 18.30	*Coffee Break / Free Poster Viewing / Exhibition*											
18.30 - 20.00	**C01** Novel diagnostic approaches	**C02** 3D gene regulation	**C03** Neurogenetic and psychiatric disorders	**C04** Fertility	**C05** Developmental disorders 1	**C06** Cellular dysfunctions	**C07** Gene editing and repro-duction ELPAG					
20.00 - 22.00	*Opening Networking Mixer (Halls D+E)*											

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** Updates in psychiatric genetics ELPAG	**E05** The longer the better? Third generation sequencing technologies	**S02** Finding the strengths that make cancer cells weak	**E06** Pharma-cogenomic testing for personalized medicine	**S03** RNA mis-splicing dynamics, diagnosis and treatment	**S04** An update on kidney research							
10.00 - 10.15	*Coffee Break / Free Poster Viewing / Exhibition*												
10.15 - 11.15	Poster Viewing with authors and coffee - group A												
11.15 - 12.45	*Lunch Break / Free Poster Viewing / Exhibition*						Oxford Nanopore Technologies Satellite		QIAGEN Satellite	Agilent Technologies Satellite	Thermo Fisher Scientific Satellite	Asuragen Satellite	Fabric Genomics Satellite
13.00 - 14.30	**C08** Prenatal genetics	**C09** Cancer genetics	**C10** Cardio-vascular disorders	**C11** Statistical and population genetics	**C12** Intellectual disability	**C13** Pharmacog-enomics	**C14** Genetic counselling developments ELPAG	**C15** Best Posters Session 1					
14.30 - 15.00	*Fruit Break / Free Poster Viewing / Exhibition*												
15.00 - 16.30	**W05** Exome sequencing and variant interpretation	**W06** Dysmor-phology I	**W07** UCSC Genome Browser	**W08** Investigating genotype-phenotype data using the GWAS Catalog	**W09** A Tribute to Seymour Kessler: Deliberating Psychothera-peutic Work in Genetic Counseling ELPAG	**W10** Genomics Quiz	**W11** European funding schemes for researchers		NanoString Technologies Satellite	SOPHiA GENETICS Satellite	MSD & AstraZeneca Satellite	PerkinElmer Satellite	New England Biolabs Satellite
16.30 - 16.45	*Coffee Break / Free Poster Viewing / Exhibition*												
16.45 - 17.45	Poster Viewing with authors and coffee - group B												
17.45 - 19.15	**S05** Genome editing	**E07** Single-cell transcrip-tomics in the brain	**S06** Thank you for the Variant (a personal utility tale) ELPAG	**E08** Chromosome Y loss and the ageing genome	**S07** Polygenic risk scores coming of age	**S08** Beware of the transposons							
19.15 - 20.45							**ESHG** Membership Meeting				Illumina Satellite	Integrated DNA Technologies Satellite	Bionano Genomics Satellite

Monday, June 17, 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	**E09** Variant interpretation and high-throughput functional assays	**S09** Multidimensional nuclear organization	**S10** From genome wide association study to mechanisms: fine-mapping	**E10** Meiosis: factory of genetic variation	**S11** "De novo" developments in epilepsia	**S12** Congenital disorders of glycosylation	**A1** ELPAG Award Lecture ELPAG						
10.00 - 10.15	*Coffee Break / Free Poster Viewing / Exhibition*												
10.15 - 11.15	**Poster Viewing with authors and coffee - group C**												
11.15 - 12.45	*Lunch break / Free Poster Viewing / Exhibition*								Roche Sequencing Solutions Satellite	NIPD Genetics Satellite	Agilent Technologies Satellite	Sistemas Genómicos Satellite	Thermo Fisher Scientific Satellite
13.00 - 14.30	**C16** Personalized & Predictive Medicine	**C17** Genetic mechanisms in cancer	**C18** Therapies	**C19** From genome architecture to RNA biology	**C20** Neuromuscular and neurodegenerative disorders	**C21** Internal organs	**C22** Ethical, policy and psychosocial aspects in genomics ELPAG	**C23** Best Posters Session 2					
14.30 - 15.00	*Fruit Break / Free Poster Viewing / Exhibition*												
15.00 - 16.30	**W12** Dysmorphology II	**W13** Copy Number Variant Interpretation and Classification	**W14** Molecular Newborn screening vs. newborn testing	**W15** European Reference Networks - What is in it for me?	**W16** Opportunistic or non opportunistic genetic screening? ELPAG	**W17** Using the Ensembl VEP for analysing variants in rare and common disease	**W18** Pharmacogenomics in practice					Twist Bioscience Satellite	Loop Genomics Satellite
16.30 - 16.45	*Coffee Break / Free Poster Viewing / Exhibition*												
16.45 - 17.45	**Poster Viewing with authors and coffee - group D**												
17.45 - 19.15	**E11** Genome First Testing in Pediatrics	**S13** Understanding mutations to detect cancer	**S14** Debate: Genomics and the Media ELPAG	**E12** Oligogenic inheritance	**S15** Regulatory Landscapes	**S16** Methods for genetic epidemiology							
20.00	*ESHG Networking Evening*												

Tuesday, June 18, 2019

TIME	HALL C	K2+K3	F1+F2+F3	F4+F5	G2+G3	K1
09.00 - 10.30	**S17** ESHG-ASHG building bridges: Global collaboration to advance the use of genomics in health	**S18** Our genetic history and its phenotypic consequences	**S19** Treating rare genetic disease	**E13** Genetic innovations in reproductive medicine	**S20** Epigenetics and early development	**E14** Understanding human disease through animal models
10.30 - 11.00	*Coffee Break (Aisle G, Aisle F, Hall H)*					
11.00 - 12.30	**C24** Mosaicisms	**C25** Bioinformatics and multiomics	**C26** Mitochondrial disorders	**C27** Developmental disorders 2	**C28** Late breaking abstracts	**C29** Stakeholder perspectives in cancer genetics ELPAG
12.30 - 13.30	*Lunch Break (Aisle G, Aisle F, Hall H)*					
13.30 - 14.15	**PL 3** Mendel Lecture					
14.15 - 15.00	**PL4** ESHG Award Lecture					
15.00 - 15.45	**PL5** Award Session - EJHG-SN Citation Awards - Young Investigator Awards - Poster Awards - Closing					

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

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 any insoluble technical problem.
- File Format: Microsoft® Power Point 2007™ (or newer) presentation formatted for Windows® (PC) only.
- Preferred Resolution: 1920 x 1080 pixel
- Screen format: 16:9

SCIENTIFIC

SCIENTIFIC PROGRAMME

SATURDAY, JUNE 15, 2019

PROGRAMME

TIME	HALL C	K2+K3	F1+F2+F3	F4+F5	G2+G3	K1	H2
08.00 - 10.00	**New Technologies Session E01** (See page 44 for details)						
10.00 - 10.30	*Coffee Break / Poster Mounting / Exhibition*				**Corporate Satellites** (see page 45 for details)		
10.30 - 12.00	**W01** **NGS in the Clinic - Mistakes and Quality Assurance** Organisers: Gijs Santen, Helger Yntema, Weronika Gutowska-Ding	**E02** **Epigenetics and cancer** Chair: Carla Oliveira	**W02** ELPAG **Prenatal Diagnosis** Organisers: Ida Vogel, Joris Vermeesch, Sam Riedijk	**E03** ELPAG **Bridging genomic discoveries and personalized medicine** Chair: Edward Dove	**W03** **How to enhance your career - How to present - How to network** Organiser & Moderator: Roy Sheppard	**E04** **Gene Expression Resources** Chair: Maria Soller	**W04** **From genetics to new medicines: The Open Targets Genetics Portal** Organiser: Maya Ghoussaini
10.30	**10.30-10.45 Common mistakes and challenges in clinical NGS QC/QA** *Christophe Roos, Euformatics 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 Discussion with statements 'How to use NGS mistakes to help patients'**	**E02.1** **Cancer Epigenetics: from DNA to RNA modifications** **François Fuks,** *Univ of Brussels, Brussels, Belgium*	**10.30-10.35 Welcome and presentation of the workshop** *Ida Vogel* **10.35-10.45 Prenatal screening in US anno 2019** *Ronald Wapner, United States* **10.45-10.55 Prenatal screening in Denmark anno 2019** *Olav Bjørn 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**	**E03.1** **Preparing for genomic medicine: a real world demonstration of health system change** **Clara Gaff,** *Melbourne, Australia*	**Do you brighten a room when you walk in, or when you leave?** **What do your colleagues say about you behind your back?** How you are perceived has a profound effect on your ability to attract professional opportunities into your life. This thought-provoking and entertaining session will provide you with practical ideas, new skills and strategies to help you develop your career. For example, everyone knows that being well-connected and meeting new people is important, but what if you're a quiet type who dreads networking and the 'torture' of making small talk? You can learn how to appear more confident whilst remaining true to yourself. Similarly, the best future opportunities invariably go to those who know how to present their ideas to large and small groups more clearly and confidently. In this packed session, Roy Sheppard will also share the secrets that professional speakers use to deliver engaging and impactful presentations.	**E04.1** **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		**E02.2** **Cell-type heterogeneity and systems epigenomics of cancer** **Andrew E. Teschendorff,** *London, United Kingdom*		**E03.2** **Personalised Medicine and clinical practice: what it means for patients and healthcare delivery** **William Newman,** *Manchester, United Kingdom*		**E04.2** **Tissue-specific enhancer and promoter evolution in mammals** **Paul Flicek,** *Hinxton, United Kingdom*	
12.00 - 14.00	*Lunch Break / Free Poster Viewing / Exhibition*				**Corporate Satellites** (see page 45 for details)		

TIME	HALL C
14.00 - 14.30	**PL0** **Welcoming Address** **Live streamed in room K2+K3** Chairs: Gunnar Houge, Joris Veltman
	Welcoming Addresses by Gunnar Houge, *President of the ESHG* Magnus Burstedt, *President of the Swedish Society of Medical Genetics and Genomics* Håkan Eriksson, *Deputy Lord Mayor on behalf of the City of Gothenburg and Region Västra Götaland*
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/

TIME	HALL C	K2+K3	F1+F2+F3	F4+F5	G2+G3	K1	H2
18.30 - 20.00	**C01** **Novel diagnostic approaches** Chairs: Morten Dunø Philippos Patsalis	**C02** **3D gene regulation** Chairs: Olaf Riess Olaug Rødningen	**C03** **Neurogenetic and psychiatric disorders** Chairs: Hilde van Esch Laura Vandervore	**C04** **Fertility** Chairs: Danja Plaseska-Karanfilska Pernille Tørring	**C05** **Developmental disorders 1** Chairs: Cecilie Rustad Sergio Sousa	**C06** **Cellular dysfunctions** Chairs: Rune Østern Birute Tumiene	**C07** ELPAG **Gene editing and reproduction** Chairs: Angus John Clarke Danya Vears
18.30	**C01.1** **A national approach to rapid genomic diagnosis in acute paediatrics** **Zornitza Stark,** *Victorian Clinical Genetics Service, Melbourne, Australia*	**C02.1** **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*	**C03.1** **Intronic expansions of an ATTTC pentamer in the STARD7 gene underlie Familial Adult Myoclonic Epilepsy linked to chromosome 2 (FAME2).** **Jozef Gecz,** *The Univ of Adelaide, Adelaide, Australia*	**C04.1** **Ectopic expression of CGG repeats leads to impaired response to gonadotropin hormones and reduced fertility with age in a mouse model of theFMR1premutation** **David L. Nelson,** *Duncan Neurological Res Inst, Houston, United States*	**C05.1** **Human and mouse gene essentiality screens allow to identify candidate genes for developmental disorders** **Violeta Munoz Fuentes,** *EMBL-EBI, Cambridge, United Kingdom*	**C06.1** **DTYMK deficiency is the cause of a novel vanishing brain disease** **Jo M. Vanoevelen,** *Dept of Clinical Genetics, Maastricht Univ Medical Ctr+, Maastricht, Netherlands*	**C07.1** **Experts' opinions on genome editing in humans: a collective construction of a disruptive technology** **Virginia Romano,** *Uppsala Univ, Uppsala, Sweden*
18.45	**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, Shinjuku, Japan*	**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*	**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*	**C04.2** **Proteomics and single-cell RNA analysis of Akap4-knockout mice model confirm indispensable role of Akap4 in spermatogenesis** **Na Li,** *Guangzhou Inst of Pediatrics, Guangzhou Women and Children's Medical Ctr, Guangzhou Medical Univ, Guangzhou, China*	**C05.2** **C-type natriuretic peptide improves growth retardation in a mouse model of cardio-facio-cutaneous syndrome associated with a Braf mutation** **Shin-ichi Inoue,** *Dept of Medical Genetics, Tohoku Univ Sch of Med, Sendai, Japan*	**C06.2** **DEGS1 Mutation causes sphingolipidosis** **Vadim 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*	**C07.2** **How will new reproductive genetic technologies change genetically at-risk couples' reproductive decision making? Views on NIPD and gene modification** **Ivy van Dijke,** *Amsterdam UMC, Amsterdam Reproduction and Development Res Inst, Amsterdam, Netherlands*
19.00	**C01.3** **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*	**C02.3** **Saturation mutagenesis of disease-associated regulatory elements** **Max Schubach,** *Berlin Inst of Health (BIH), 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*	**C04.3** **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 Cantú syndrome** **Helen I. Roessler,** *Dept of Genetics, Ctr for Molecular Med, Univ Medical Ctr Utrecht, Utrecht, Netherlands*	**C06.3** **MAGT1-CDG vs. XMEN: two faces of a novel glycosylation disorder** **Eline Blommaert,** *Dept of Human Genetics, KU Leuven, Leuven, Belgium*	**C07.3** **Perspectives of a Genetic Disease Community and Genetic Professionals on Germline Gene Editing** **Vence L. Bonham,** *Natl Human Genome Res Inst, Bethesda, United States*

TIME	HALL C	K2+K3	F1+F2+F3	F4+F5	G2+G3	K1	H2
cont.	**C01** Novel diagnostic approaches	**C02** 3D gene regulation	**C03** Neurogenetic and psychiatric disorders	**C04** Fertility	**C05** Developmental disorders 1	**C06** Cellular dysfunctions	**C07** ELPAG Gene editing and reproduction
19.15	**C01.4** **Expanding Next Generation Phenotyping on clinical notes and hand radiographs** **Guy Nadav,** *FDNA Inc, Boston, United States*	**C02.4** **Characterization of GJB2 cis-regulatory elements in the DFNB1 locus** **Anaïs Le Nabec,** *Univ Brest, Inserm, EFS, UMR 1078, GGB, Brest, France*	**C03.4** **Loss of neutral sphingomyelinase-3 (SMPD4) links neurodevelopmental disorders to cell cycle and nuclear envelope anomalies** **Daphne J. Smits,** *Dept of Clinical Genetics, Erasmus Univ Medical Ctr, Rotterdam, Netherlands*	**C04.4** **Exome sequencing reveals de novo mutations and deletions in severe idiopathic male infertility** **Manon S. Oud,** *Dept of Human Genetics, Radboudumc, Nijmegen, Netherlands*	**C05.4** **A restricted spectrum of KMT2D variants cause a multiple malformations disorder distinct from Kabuki syndrome** **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*	**C06.4** **Implication of LRP6 variants in familial hypercholesterolemia** **Youmna Ghaleb,** *LVTS INSERM U1148, Paris, France*	**C07.4** **National implementation of genome-wide non-invasive prenatal testing as a first-tier screening test in the Netherlands: evaluation of women's perspectives** **Karuna R. van der Meij,** *Dept of Clinical Genetics, Amsterdam UMC, Vrije Univ Amsterdam, Amsterdam, Netherlands*
19.30	**C01.5** **Using UK Biobank to assess the pathogenicity, penetrance and expressivity of monogenic disease variants** **Caroline F. Wright,** *Inst of Biomedical and Clinical Science, Exeter, United Kingdom*	**C02.5** **Novel insights into molecular mechanisms in X-linked dystonia-parkinsonism (XDP)** **Jelena Pozojevic,** *Section for Functional Genetics, Inst of Human Genetics, Lübeck, Germany*	**C03.5** **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)** **Ian Tully,** *Neuroscience and Mental Health Res Inst, Cardiff, United Kingdom*	**C04.5** **CETN1 is associated with spermatogenesis and male fertility: Genetic and functional perspectives** **Digumarthi V. Sudhakar,** *Ctr for Cellular and Molecular Biology, Hyderabad, India*	**C05.5** **De novo C-terminal truncating mutations in MN1 cause a neurodevelopmental syndrome with distinctive facial features** **Chris T. Gordon,** *Inst Imagine, INSERM U1163, Paris, France*	**C06.5** **Deciphering altered inhibitor G-protein signaling in the cardiac dysfunction underlying Intellectual Developmental Disorder with Cardiac Arrhythmia (IDDCA) syndrome** **Pasquelena De Nittis,** *Ctr for Integrative Genomics, Lausanne, Switzerland*	**C07.5** **Implementing non-invasive prenatal testing (NIPT): An interview study of pregnant women's opinions about and experiences with societal pressure, reimbursement and an expanding scope** **Iris M. Bakkeren,** *Erasmus Medical Ctr, Rotterdam, Netherlands*
19.45	**C01.6** **Man vs Machine: Implementing clinically validated automated variant prioritisation with diagnostic performance that equals human experts** **Leslie Burnett,** *Kinghorn Ctr for Clinical Genomics, Garvan Inst of Medical Res, Darlinghurst Sydney, Australia*	**C02.6** **Understanding the roles and the regulation of the Mowat-Wilson Syndrome transcription factor ZEB2 during development and disease** **Andrea Conidi,** *Erasmus MC, Rotterdam, Netherlands*	**C03.6** **De-novo mutations in TAOK1 cause neurodevelopmental disorders** **Marija Dulovic Mahlow,** *Inst of Neurogenetics, Luebeck, Germany*	**C04.6** **Duplication and deletion of key SOX9 enhancers cause sex reversal in humans** **Andrew H. Sinclair,** *Murdoch Children's Res Inst, Melbourne, Australia*	**C05.6** **De novo missense mutations in the X-linked TFE3 gene cause intellectual disability with pigmentary mosaicism and storage disorder-like features** **Daphné Lehalle,** *Equipe GAD, INSERM LNC UMR 1231, Faculté de Médecine, Univ de Bourgogne Franche-Comté, Dijon, France*	**C06.6** **PCYT2 Variants Disrupt Etherlipid Biosynthesis and Cause a Complex Hereditary Spastic Paraplegia** **John H. McDermott,** *Manchester Ctr for Genomic Med, Manchester, United Kingdom*	**C07.6** **The notion of "serious" for genetic disease: To qualify or not, that is the question** **Erika Kleiderman,** *Ctr of Genomics and Policy, McGill Univ, Montreal, Canada*
20.00 - 22.00	***Opening Networking Mixer (Halls D+E)***						

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

SCIENTIFIC

SCIENTIFIC PROGRAMME

SUNDAY, JUNE 16, 2019

PROGRAMME

TIME	HALL C	K2+K3	F1+F2+F3	F4+F5	G2+G3	K1
08.30 - 10.00	**S01** ELPAG **Updates in psychiatric genetics** Chairs: Ramona Moldovan Enza Maria Valente	**E05** **The longer the better? Third generation sequencing technologies** Chair: Zeynep Tümer	**S02** **Finding the strengths that make cancer cells weak** Chairs: Jose Luis Costa Hildegunn Vetti	**E06** **Pharmacogenomic testing for personalized medicine** Chair: Vita Dolzan	**S03** **RNA mis-splicing dynamics, diagnosis and treatment** Chairs: Siren Bergland Elfride de Baere	**S04** **An update on kidney research** Chairs: Jens Michael Hertz Charlotte von der Lippe
08.30	**S01.1** **Common and rare variants in psychiatric disorders** **James Walters,** *Cardiff, United Kingdom*	**E05.1** **How long do we need? The relative value of emerging sequencing technologies in genomic medicine** **Mike Talkowski,** *Cambridge, United States*	**S02.1** **Tumour supressor function restoration: role in tumour reversion and response to treatment** **Scott Lowe,** *New York, United States*	**E06.1** **Integrating pharmacogenomics into personalized drug treatment** **Magnus Ingelman-Sundberg,** *Stockholm, Sweden*	**S03.1** **Minor spliceosome and disease** **Mikko J. Frilander,** *Univ of Helsinki, Helsinki, Finland*	**S04.1** **Polycystic kidney disease and ciliopathies** **Carsten Bergmann,** *Dept of Med, Univ Hosp Freiburg, Freiburg, Germany*
09.00	**S01.2** **Genome wide patterns of structural mutation and selection guide the identification of cis-regulatory structural variants in autism** **Jonathan Sebat,** *La Jolla, United States*		**S02.2** **Myc in Cancer: targeting an engine, not a driver** **Gerard Evan,** *Cambridge, United Kingdom*		**S03.2** **Dynamic mutations and RNA mis-splicing in disease** **Maurice Swanson,** *Univ of Florida, Gainesville, United States*	**S04.2** **New insights in the genetics of hereditary nephrotic syndromes** **Corinne Antignac,** *Paris, France*
09.15		**E05.2** **Delineating the structure of chromosome rearrangements using multiple WGS technologies** **Anna Lindstrand,** *Karolinska Instt, Stockholm, Sweden*		**E06.2** **Pharmacogenomics based personalized drug treatment across world populations** **Andrea Gaedigk,** *Children's Mercy Kansas City, Kansas City, United States*		
09.30	**S01.3** **Updates in clinical applications of psychiatric genetics** **Jehannine Austin,** *Univ of British Columbia, Vancouver, Canada*		**S02.3** **Immunotherapy in cancer** **Karine Serre,** *Insto de Medicina Molecular, JLA, Lisbon, Portugal*		**S03.3** **Restoring splicing defects by antisense oligonucleotide therapy** **Rob Collin,** *Nijmegen, Netherlands*	**S04.3** **CRISPR Gene Editing in human organoids for inherited renal diseases** **Benjamin Freedman,** *Seattle, United States*
10.00 - 10.15	***Coffee Break / Free Poster Viewing / Exhibition***					
10.15 - 11.15	**Poster Viewing with authors and coffee (Group A)**					
11.15 - 13.00	***Lunch break / Free Poster Viewing / Exhibition***				**Corporate Satellites** (see page 45 for details)	

TIME	HALL C	K2+K3	F1+F2+F3	F4+F5	G2+G3	K1	H2
13.00 - 14.30	**C08** **Prenatal Genetics** Chairs: Heidi Marjonen Reiner V. Veitia	**C09** **Cancer genetics** Chairs: Kristiina Aittomäki Samuel Gebre-Medhin	**C10** **Cardiovascular disorders** Chairs: Birgitte Diness Bart Loeys	**C11** **Statistical and population genetics** Chairs: Eleonora Porcu Eava Sliz	**C12** **Intellectual Disability** Chairs: Ilaria Parenti Cecilie Rustad	**C13** **Pharmacogenomics** Chairs: Stein Bergan Johan den Dunnen	**C14** ELPAG **Genetic counselling developments** Chairs: Kinga Hadzsiev Rhona Macleod
13.00	**C08.1** **Increased risk at first trimester screening: trisomies are not everything, but the risk for an atypical chromosome aberration is low. Experiences from the Swedish Pregnancy Register** **Erik Iwarsson,** *Dept of Molecular Med and Surgery, Clinical Genetics Unit, Karolinska Instt and Karolinska Univ Hosp, Stockholm, Sweden*	**C09.1** **Germline genetic variation drives the somatic landscape of tumors** **Noah Zaitlen,** *UCLA, Los Angeles, United States*	**C10.1** **Sequence variants associated with resistant hypertension implicate mechanisms affecting potassium levels** **Vinicius Tragante,** *deCODE Genetics/ Amgen, Reykjavik, Iceland*	**C11.1** **Maximum likelihood method quantifies the overall contribution of gene-environment interaction to complex traits: an application to obesity traits** **Zoltan Kutalik,** *Univ Ctr for Primary Case and Public Health, Lausanne, Switzerland*	**C12.1** **Phenotypic spectrum of novel intellectual disability syndrome due to de novo variants in KMT2E** **Anne O'Donnell-Luria,** *Boston Children's Hosp, Boston, United States*	**C13.1** **Metabolomic consequences of PCSK9 inhibition compared with statin therapy** **Peter Würtz,** *Nightingale Health Ltd., Helsinki, Finland*	**C14.1** **Effect of genetic counseling on adherence to psychotropic medication in people with serious mental illness** **Jehannine Austin,** *Univ of British Columbia, Vancouver, Canada*
13.15	**C08.2** **The PREGCARE study: precision genetic counselling via personalised evaluation of recurrence risk for families with a child affected by a disorder caused by a de novo mutation** **Ummi B. Abdullah,** *Clinical Genetics Group, MRC Weatherall Inst of Molecular Med, Univ of Oxford, Oxford, United Kingdom*	**C09.2** **Germline TP53 mutations: the predominant genetic cause of adrenocortical carcinoma** **Gaëlle Bougeard,** *Dept of Genetics, Normandy Ctr for Genomic and Personalized Med, Normandy Univ, UNIROUEN, Inserm U1245 and Rouen Univ Hosp, Rouen, France*	**C10.2** **Multi-omics approach identifies three novel genes for bicuspid aortic valve related aortopathy** **Ilse Luyckx,** *Ctr for Medical Genetics, Edegem, Belgium*	**C11.2** **Leveraging correlated risks to increase power in Genome-Wide Association Studies** **Ninon Mounier,** *Univ Ctr for Primary Care and Public Health, Lausanne, Switzerland*	**C12.2** **CTCF variants in 31 individuals with a variable neurodevelopmental disorder broaden the mutational and clinical spectrum** **Enrico D. Konrad,** *Inst of Human Genetics, Erlangen, Germany*	**C13.2** **Longitudinal analysis of the gut microbiome reveals dynamic changes in relation to medications & phenotypes** **Lianmin Chen,** *Dept of Genetics, Univ Medical Ctr Groningen, Groningen, Netherlands*	**C14.2** **Psychiatric Genetic Counselling: Efficacy of training and implications for practice** **Kevin A. McGhee,** *Faculty of Science & Technology, Bournemouth Univ, POOLE, United Kingdom*
13.30	**C08.3** **Validation of simultaneous detection of fetal chromosome aneuploidy and monogenic diseases by a novel noninvasive prenatal testing method: Targeted And Genome-wide simultaneous sequencing (TAGs-seq)** **Wang Yicong,** *BGI-Shenzhen, Shenzhen, China*	**C09.3** **Cell free-DNA pinpoints specific clonal expansion at disease progression in solid cancers** **Maria Palmieri,** *Medical Genetics, Univ of Siena, Siena, Italy*	**C10.3** **Investigating atherosclerosis progression through single-cell transcriptional profiling of immune cells of the atherosclerotic plaque** **Ambra Sartori,** *Univ of Geneva, Geneva, Switzerland*	**C11.3** **One and a half million genome wide-association studies of brain morphometry: a proof-of-concept study** **Hieab H. Adams,** *Erasmus MC, Rotterdam, Netherlands*	**C12.3** **De novo variants disturbing the transactivation capacity of POU3F3 cause a characteristic neurodevelopmental disorder** **Lot Snijders Blok,** *Radboud Univ Medical Ctr, Nijmegen, Netherlands*	**C13.3** **Lifelong genetically lowered sclerostin and risk of cardiovascular disease** **Jonas Bovijn,** *Big Data Inst, Univ of Oxford, Oxford, United Kingdom*	**C14.3** **Large scale group genetic counselling: a novel service delivery model** **Jennifer Nuk,** *BC Cancer Hereditary Cancer Program, Vancouver, Canada*

TIME	HALL C	K2+K3	F1+F2+F3	F4+F5	G2+G3	K1	H2
cont.	**C08** Prenatal Genetics	**C09** Cancer genetics	**C10** Cardiovascular disorders	**C11** Statistical and population genetics	**C12** Intellectual Disability	**C13** Pharmacogenomics	**C14** ELPAG Genetic counselling developments
13.45	**C08.4** **Systematic evaluation of prenatal and pediatric diagnostic yields from whole-genome sequencing in 8,954 individuals** **Chelsea Lowther,** *Ctr for Genomic Med, Boston, United States*	**C09.4** **Molecular classification of B-other pediatric B-cell precursor acute lymphoblastic leukemia by DNA methylation and RNA-sequencing** **Jessica Nordlund,** *Molecular Med and Science for Life Lab, Uppsala Univ, Uppsala, Sweden*	**C10.4** **Metabolomic profiling of ANGPTL3 deficiency** **Emmi Tikkanen,** *Nightingale Health Ltd., Helsinki, Finland*	**C11.4** **Genome-wide copy number variant association study reveals several novel disease-associated loci** **Maarja Lepamets,** *Estonian Genome Ctr, Inst of Genomics, Univ of Tartu, Tartu, Estonia*	**C12.4** **De novo variants in MAPK8IP3 cause intellectual disability with variable brain anomalies** **Konrad Platzer,** *Inst of Human Genetics, Univ of Leipzig Medical Ctr, Leipzig, Germany*	**C13.4** **Advanced renal cancer patients with tumor KDM5C mutations show improved response to anti-angiogenic therapy** **Maria Santos,** *Spanish Natl Cancer Res Ctr, Madrid, Spain*	**C14.4** **Genetic counselling experience in Iceland of web-based return of BRCA2 research results** **Vigdís Stefansdottir,** *Landspitali Natl Univ Hosp, Reykjavik, Iceland*
14.00	**C08.5** **Non-invasive prenatal diagnosis of sickle cell disease by next generation sequencing of cell-free DNA** **Julia C. van Campen,** *Genetics Labs, Guy's and St. Thomas' NHS Fndn Trust, London, United Kingdom*	**C09.5** **Polygenic risk scores modify age-dependent breast cancer risk in CHEK2 germline mutation carriers** **Julika Borde,** *Ctr for Hereditary Breast and Ovarian Cancer, Ctr for Integrated Oncology (CIO), Univ of Cologne, Faculty of Med and Univ Hosp Cologne, Cologne, Germany*	**C10.5** **The Future is Now: Genomic Studies Must be Globally Representative** **Kari E. North,** *Dept of Epidemiology, Univ of North Carolina, Chapel Hill, United States*	**C11.5** **Fine-scale population structure and demographic change through time and space in the Netherlands** **Ross P. Byrne,** *Smurfit institute of genetics, Trinity Coll Dublin, Dublin, Ireland*	**C12.5** **Defective DNA polymerase a-primase leads to X-linked intellectual disability associated with severe growth retardation, microcephaly and hypogonadism.** **Hilde Van Esch,** *Ctr for Human Genetics, LEUVEN, Belgium*	**C13.5** **Predicting Functional Effects of Missense Variants in Voltage-Gated Sodium and Calcium Channels** **Henrike O. Heyne,** *Broad Inst of Harvard and MIT, Cambridge, United States*	**C14.5** **The making of the BRCA-chatbot - A patient centered digital counselling tool to support individuals undergoing genetic testing for hereditary breast and ovarian cancer** **Elen Siglen,** *Dept of Medical Genetics, Haukeland Univ Hosp, Bergen, Norway*
14.15	**C08.6** **Prevalence and clinical outcome of mosaicism in uncultured chorionic villus samplings after chromosomal microarray** **Ida Charlotte Bay Lund,** *Dept of Clinical Genetics, Aarhus Univ Hosp, Aarhus N., Denmark*	**C09.6** **Application of genomics and cognitive technology in precision oncological medicine** **Gloria Ribas,** *Medical Genetics Unit, Sistemas Genómicos, Valencia, Spain*	**C10.6** **Genetics of human plasmalipidome and its link to cardiovascular diseases** **Rubina Tabassum,** *Inst for Molecular Med Finland, HiLIFE, Univ of Helsinki, Helsinki, Helsinki, Finland*	**C11.6** **The landscape of pervasive horizontal pleiotropy in human genetic variation is driven by extreme polygenicity of human traits and diseases** **Marie Verbanck,** *The Charles Bronfman Inst for Personalized Med, Icahn Sch of Med at Mount Sinai, New York, United States*	**C12.6** **Non-penetrance of a frameshifting SHANK3 deletion is associated with compensatory mechanisms in both alleles** **Bjørn Ivar Haukanes,** *Dept of Medical Genetics, Bergen, Norway*	**C13.6** **Taurine supplementation as a potential therapy for progressive retinal degeneration due to biallelic pathogenic variants in the Taurine transporter SLC6A6** **Emmanuelle Ranza,** *Dept of Genetic Med and Development, Univ of Geneva, Geneva, Switzerland*	**C14.6** **myKinMatters intervention: developing an online intervention to support patients in communicating relevant health information to at-risk relatives** **Lisa M. Ballard,** *Univ of Southampton, Southampton, United Kingdom*
14.30 - 15.00	***Fruit break / Free Poster Viewing / Exhibition***						

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

TIME	EXHIBITION HALL - LIVE STREAM AREA
13.00 - 14.30	**C15** **Best Posters Session 1** Chairs: Joris Veltman, Alexandre Reymond
	P03.05A **Gastrointestinal dysfunction in autism spectrum disorder: New insights from the Foxp1+/-mouse with altered gut motility and achalasia** **Gudrun Rappold,** *Dept of Human Molecular Genetics, Inst of Human Genetics, Univ of Heidelberg, Heidelberg, Germany*
	P04.41A **Identification and characterization of microRNA-149, a candidate for orofacial clefting.** **Ronja Hollstein,** *Inst of Human Genetics, Univ of Bonn, Sch of Med & Univ Hosp Bonn, Bonn, Germany*
	P06.60C **High-throughput metabolomics for early detection of individuals at increased risk for type 2 diabetes** **Jenni Hällfors,** *Nightingale Health Ltd., Helsinki, Finland*
	P07.09A **Increasing fetal hemoglobin by genetic editing the cells of sickle cell disease patients** **Kirmo Wartiovaara,** *Clinical Genetics, Helsinki Univ Hosp, Helsinki, Finland*
	P09.012C **Anorexia nervosa genome-wide association study identifies eight loci and implicates psychiatric and metabolic origins** **Christopher Hübel,** *Karolinska Instt, Stockholm, Sweden*
	P09.028C **Characterizing cellular heterogeneity of de novo mutations in autism spectrum disorders** **Abdulrahman Y. Ali,** *Mohammed Bin Rashid Univ of Med and Health Sciences, Dubai, United Arab Emirates*
	P10.36C **Analysis of DNA tandem repeats in ALS from Whole Genome Sequencing : Role of FRA10Ac1 gene repeat expansion in ALS** **Lucia Corrado,** *Univ of Eastern Piedmont UPO, NOvara, Italy*
	P13.13A **Gain-of-function mutations in KCNN3 encoding the small-conductance Ca2+-activated K+ channel SK3 cause Zimmermann-Laband syndrome** **Pauline E. Schneeberger,** *Inst of Human Genetics, Univ Medical Ctr Hamburg-Eppendorf, Hamburg, Germany*
	P14.008C **Variants with reduced variant fractions in NGS-based germline diagnostics for hereditary breast and ovarian cancer** **Mirjam Larsen,** *Ctr for Hereditary Breast and Ovarian Cancer, Ctr for Integrated Oncology (CIO), Univ of Cologne, Faculty of Med and Univ Hosp Cologne, Cologne, Germany*
	P16.46B **LOY Associated Transcriptional Effect (LATE) in immune cells measured by single cell RNAseq and bulk RNAseq** **Jonas Mattisson,** *Dept of Immunology, Genetics and Pathology, Uppsala Univ, Uppsala, Sweden*
	P16.08D **Gabriella Miller Kids First Data Resource Center: Harmonizing genomic and clinical information to support childhood cancer and structural birth defect research** **Yiran Guo,** *Children's Hosp of Philadelphia, Phiadelphia, United States*
	P16.02B **Genetic dysregulation of gene expression and splicing during a ten-year period of human aging** **Brunilda Balliu,** *Dept of Biomathematics, UCLA, Los Angeles, United States*
	P17.13B **An integrated chromatin accessibility and transcriptome landscape of human pre- and post-implantation embryos** **Zhouchun Shang,** *BGI-Shenzhen, Shenzhen, China*
	P17.40A **Disease interpretation of regulatory variants with GeneHancer** **Simon Fishilevich,** *Weizmann Inst of Science, Rehovot, Israel*
	P18.67C **The genetics of sleep traits and their links with disease** **Samuel E. Jones,** *Univ of Exeter Medical Sch, Exeter, United Kingdom*

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.

TIME	HALL C	K2+K3	F1+F2+F3	F4+F5	G2+G3	K1	H2	
15.00 - 16.30	**W05** **Exome sequencing and variant interpretation** Organisers: Christian Gilissen Kaitlin Samocha	**W06** **Dysmorphology I** 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 **A Tribute to Seymour Kessler: Deliberating Psychotherapeutic Work in Genetic Counseling** Organiser: Rhona Macleod	**W10** **Genomics Quiz** Organisers: Robert Hofstra Julie McGaughran Moderator: Roy Sheppard	**W11** **European funding schemes for researchers** Organisers: Alexandre Reymond Elfride De Baere	**Corporate Satellites** (see page 45 for details)
	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** *Kaitlin Samocha* **15.30-15.50 Predicting the effect of splicesite 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** *Rolph 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 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.	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., 5C, 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 installed, and may find it useful to have a command line interface (e.g. Unix terminal). Workshop Speakers: Laura Harris, Scientific Curator Daniel Suveges, Bioinformatician Jackie MacArthur, Project Lead	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. Workshop Speakers: Barbara Biesecker, RTI International Aad Tibben, University of Leiden	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.	**15.00-15.25 ERC Starting Grant** *Lude Franke, The Netherlands* **15.25-15.50 ERC Consolidator Grant** *Bart Loeys, Belgium* **15.50-16.15 ERC from the perspective of LS panels** *Konstantina Topouridou, Belgium* **16.15-16.30 Q&A Session**	
16.30 - 16.45	***Coffee Break / Free Poster Viewing / Exhibition***							
16.45 - 17.45	**Poster Viewing with authors and coffee (Group B)**							

TIME	HALL C	K2+K3	F1+F2+F3	F4+F5	G2+G3	K1
17.45 - 19.15	**S05** **Genome editing** Chairs: Malte Spielmann Kirmo Wartiovaara	**E07** **Single-cell transcriptomics in the brain** Chair: Joris Vermeesch	**S06** ELPAG **Thank you for the Variant (a personal utility tale)** Chairs: Charlotta Ingvoldstad Malmgren Celine Lewis	**E08** **Chromosome Y loss and the ageing genome** Chair: Yasemin Alanay	**S07** **Polygenic risk scores coming of age** Chairs: Anders Børglum Franke Lude	**S08** **Beware of the transposons** Chairs: Maris Laan Anna Lindstrand
17.45	**S05.1** **CRISPR single-cell sequencing: Toward functional biology in high throughput** **Christoph Bock,** *CeMM Res Ctr for Molecular Med of the Austrian Acad of Sciences, Vienna, Austria*	**E07.1** **Single cell heterogeneity in human brain and its relation to neurodegenerative diseases** **Raheleh Rahbari,** *Hinxton, United Kingdom*	**S06.1** **Current understanding of psychiatric genetics research and services amongst mental health service users and their families** **David Crepaz-Keay,** *London, United Kingdom*	**E08.1** **Mosaic loss of chromosome Y (LOY) in leukocytes: from discovery to impact** **Lars Forsberg,** *Uppsala, Sweden*	**S07.1** **Polygenic risk scores in genetic epidemiology** **Krista Fisher,** *Tartu, Estonia*	**S08.1** **YY1: an enduring repressor of L1 retrotransposition during human neurodevelopment** **Geoffrey J. Faulkner,** *Univ of Queensland, Brisbane, Australia*
18.15	**S05.2** **Therapeutic applications of genome editing to prevent diseases** **Kiran Musunuru,** *Cambridge, United States*		**S06.2** **Genetic profiling in primary care: triggers and impact on risk-reducing behaviour** **Nadeem Qureshi,** *Nottingham, United Kingdom*		**S07.2** **Polygenic risks and their impact on behavior** **Samuli Ripatti,** *Helsinki, Finland*	**S08.2** **Alu elements and cellular RNA metabolism** **Lynne E. Maquat,** *Univ of Rochester, Rochester, United States*
18.30		**E07.2** **Single cell RNA sequencing in psychiatric disorders** **Jens Hjerling Leffler,** *Karolinska Instt, Stockholm, Sweden*		**E08.2** **Mosaic chromosome Y loss, ageing and cancer risk** **Mitchell J. Machiela,** *Natl Cancer Inst, Rockville, United States*		
18.45	**S05.3** **Advances in therapeutic CRISPR/Cas9 genome editing** **Gerald Schwank,** *IMHS, Zurich, Switzerland*		**S06.3** **What will this genetic result mean for my baby?** **Lidewij Henneman,** *Amsterdam UMC, VUMC, Amsterdam, Netherlands*		**S07.3** **Polygenic risk scores in prostate cancer** **Rosalind Eeles,** *The Inst Of Cancer Res & Royal Marsden NHS Trust, London, United Kingdom*	**S08.3** **Insertion variants at disease risk loci** **Kathleen H. Burns,** *Johns Hopkins Univ Sch of Med, Baltimore, United States*

TIME	H2	Corporate Satellites
19.15 - 20.30	**ESHG Membership Meeting** **All ESHG members welcome!**	(see page 45 for details)

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/

SCIENTIFIC

SCIENTIFIC PROGRAMME

MONDAY, JUNE 17, 2019

PROGRAMME

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TIME	HALL C	K2+K3	F1+F2+F3	F4+F5	G2+G3	K1	H2
08.30 - 10.00	**E09** **Variant interpretation and high-throughput functional assays** Chair: Malte Spielmann	**S09** **Multidimensional nuclear organization** Chairs: Tord Jonson Zeynep Tümer	**S10** **From genome wide association study to mechanisms: fine-mapping** Chairs: Andres Børglum Cecilia Lindgren	**E10** **Meiosis: factory of genetic variation** Chair: Maria Jesus Sobrido	**S11** **De novo developments in epilepsia** Chairs: Christina Fagerberg Karin Writzl	**S12** **Congenital disorders of glycosylation** Chairs: Valerie Cormier-Daire Trine Prescott	**A1** ELPAG **ELPAG Award Lecture**
08.30	**E09.1** **Unraveling the functional impact of thousands of p53 mutations** **Eran Segal,** *Rehovot, Israel*	**S09.1** **Revealing the RNA layer of epigenome** **Sheng Zhong,** *Univ of California San Diego, San Diego, United States*	**S10.1** **From association to causal variant(s): statistical methods for finemapping** **Christian Benner,** *Helsinki, Finland*	**E10.1** **Genetic diversity and its unexpected impacts on recombination, genome evolution, speciation and sterility in mammals** **Simon Myers,** *Oxford, United Kingdom*	**S11.1** **De novo variants in neurodevelopmental disorders with epilepsy** **Johannes Lemke,** *Inst of Human Genetics, Univ of Leipzig, Leipzig, Germany*	**S12.1** **Genetic heterogeneity in CDG: where are the patients?** **Gert Matthijs,** *Leuven, Belgium*	**A1.1** **ELPAG Award Lecture** **Christine Patch,** *Guys and St Thomas' NHS Foundation Trust, London, United Kingdom*
09.00		**S09.2** **The architecture and mechanical properties of the nuclear lamina** **Ohad Medalia,** *Univ of Zurich, Zurich, Switzerland*	**S10.2** **Leveraging genome-wide association studies in diverse populations to fine-map complex human trait loci** **Andrew P. Morris,** *Univ of Manchester, Manchester, United Kingdom*		**S11.2** **Parental Mosaicism in "De Novo" Epileptic Encephalopathies** **Heather C. Mefford,** *Univ of Washington, Seattle, United States*	**S12.2** **CDG therapies** **Eva Morava,** *Mayo Clinic, Rochester, United States*	
09.15	**E09.2** **Understanding the functional effects of coding variation, at scale** **Lea M. Starita,** *Univ of Washington, Seattle, United States*			**E10.2** **Meiotic recombination, gene conversion and mutation** **Irene Tiemann-Boege,** *Johannes Kepler Univ, Linz, Austria*			
09.30		**S09.3** **3D genome organisation in disease: patient-specific chromatin interactions from primary tissue** **Juanma Vaquerizas,** *Münster, Germany*	**S10.3** **Large scale integration of genetic and 'omics' data to find susceptibility genes for obesity and fat distribution** **Sara L. Pulit,** *Univ Medical Ctr Utrecht, Utrecht, Netherlands*		**S11.3** **Brain somatic mutations in malformations of cortical development with epilepsy** **Stéphanie Baulac,** *Inst du Cerveau et de la Moelle, Paris, France*	**S12.3** **Link between Golgi ion homeostasis defects and Congenital Disorders of Glycosylation** **Francois Foulquier,** *CNRS UMR 8576, Univ of Lille, Villeneuve D'Ascq, France*	
10.00 - 10.15	***Coffee Break / Free Poster Viewing / Exhibition***						
10.15 - 11.15	**Poster Viewing with authors and coffee (Group C)**						
11.15 - 13.00	***Lunch break / Free Poster Viewing / Exhibition*** **Corporate Satellites** (see page 45 for details)						

TIME	HALL C	K2+K3	F1+F2+F3	F4+F5	G2+G3	K1	H2
13.00 - 14.30	**C16** **Personalized and predictive medicine** Chairs: John Dermott Hans Ehrencrona	**C17** **Genetic mechanisms in cancer** Chairs: Robert Hofstra Hildegunn Vetti	**C18** **Therapies** Chairs: Josef Gecz Charlotte von der Lippe	**C19** **From genome architecture to RNA biology** Chairs: Ellen Heitzer Oloug Rødningen	**C20** **Neuromuscular and neurodegenerative disorders** Chairs: Elsebet Østergaard André Reis	**C21** **Internal organs** Chairs: Marte Gjøl Haug Ales Maver	**C22** ELPAG **Ethical, policy and psychosocial aspects in genomics** Chairs: Charlotta Ingvoldstad Malmgren Rebecca Ann Pestoff
13.00	**C16.1** **What if we would turn a diagnostic multi-cancer gene panel into a screening tool?** **Helga Westers,** *Univ Medical Ctr Groningen, Groningen, Netherlands*	**C17.1** **Identification of lncRNA-mRNA network(s) that modulate prognosis in hepatocellular carcinoma patients** **Caroline G. Lee,** *Natl Univ of Singapore, Singapore, Singapore*	**C18.1** **AAVHSC15 Packaging Human Phenylalanine Hydroxylase Results in Sustained in vivo Correction of Phenylketonuria Following a Single IV Administration in the Murine Model** **Seemin S. Ahmed,** *Homology Meds Inc, Bedford, United States*	**C19.1** **Phasing of complex genomic rearrangements reveal involvement of both homologous chromosomes in pre- and post-zigotic events** **Claudia M. Carvalho,** *Baylor Coll of Med, Houston, United States*	**C20.1** **Large clinical cohort undergoing simultaneous single nucleotide and copy number variant analysis reveals broad mutation spectrum and high diagnostic yield for neuromuscular disorders** **Emily Decker,** *Invitae, San Francisco, United States*	**C21.1** **Loss-of-function variants in myocardin cause congenital megabladder in humans and mice** **Arjan Houweling,** *Dept of Clinical Genetics, Amsterdam UMC, Vrije Univ Amsterdam, Amsterdam, Netherlands*	**C22.1** **"To find out if it's genetic or not": Motivations, concerns and perceived impact of genome sequencing among young people** **Celine Lewis,** *Great Ormond Street Hosp NHS Fndn Trust, London, United Kingdom*
13.15	**C16.2** **European Landscape of CDH1 germline mutations: a new tool to understand hereditary diffuse gastric cancer (HDGC)** **José García Peláez,** *IPATIMUP/i3s, Porto, Portugal*	**C17.2** **Germline DGCR8 p.E518K alters miRNA profiles and predisposes to thyroid goiter and schwannomatosis** **Barbara Rivera,** *McGill Univ, Montreal, Canada*	**C18.2** **Viral vector therapy as a therapeutic option for peripheral nerve disease associated with metachromatic leukodystrophy.** **Stephanie K. Newman,** *Univ of Western Ontario, London, Canada*	**C19.2** **Cytogenetically detected chromosomal inversions are rarely formed by ectopic recombination between inverted repeats** **Maria Pettersson,** *Karolinska Instt, Stockholm, Sweden*	**C20.2** **Mutations in the Golgi protein GBF1 as a novel cause of distal hereditary motor neuropathy** **Natalia Mendoza Ferreira,** *Inst of Human Genetics, Univ of Cologne, Cologne, Germany*	**C21.2** **Rare heterozygous deleterious GDF6 variants in patients with renal anomalies** **Helge Martens,** *Dept of Human Genetics, Hannover Medical Sch, Hannover, Germany*	**C22.2** **Genetic health professionals' experiences returning results from diagnostic genomic sequencing to patients** **Danya F. Vears,** *Univ of Melbourne, Parkville, Australia*
13.30	**C16.3** **Clinical applicability of the 313-SNP based polygenic risk score for breast cancer risk prediction** **Inge M. Lakeman,** *Leiden Univ Medical Ctr, Leiden, Netherlands*	**C17.3** **Structural variations at CDH1 intronic cis-regulatory elements cause CDH1/E-cadherin loss of function** **Rita Barbosa-Matos,** *IPATIMUP, Inst of Molecular Pathology and Immunology, Univ of Porto, Porto, Portugal*	**C18.3** **Safe and efficient personalised TALEN- and CRISPR/Cas9-based gene correction therapy for β-thalassaemia by non-viral delivery to primary cells** **Petros Patsalis,** *Molecular Genetics of Thalassemia Dept, The Cyprus Inst of Neurology and Genetics, Nicosia, Cyprus*	**C19.3** **Optical mapping of 22q11.2 low copy repeats reveals structural hypervariability** **Lisanne Vervoort,** *Dept of Human Genetics, KU Leuven, Leuven, Belgium*	**C20.3** **Recessive mutations in muscle-specific isoforms of FXR1 cause congenital multi-minicore myopathy** **Elisa Fernández-Núñez,** *Insto de Investigaciones Biomédicas "Alberto Sols" (CSIC-UAM), Madrid, Spain*	**C21.3** **Exome sequencing identifies phenocopies in every fifth solved case in a cohort of 174 patients with hereditary nephropathies** **Korbinian M. Riedhammer,** *Inst of Human Genetics, Klinikum rechts der Isar, Technical Univ of Munich, Munich, Germany*	**C22.3** **Parent experiences with ultra-rapid genomic sequencing in paediatric acute care** **Gemma R. Brett,** *Victorian Clinical Genetics Services, Murdoch Children's Res Inst, Melbourne, Australia*

TIME	HALL C	K2+K3	F1+F2+F3	F4+F5	G2+G3	K1	H2
cont.	**C16** Personalized and predictive medicine	**C17** Genetic mechanisms in cancer	**C18** Therapies	**C19** From genome architecture to RNA biology	**C20** Neuromuscular and neurodegenerative disorders	**C21** Internal organs	**C22** ELPAG Ethical, policy and psychosocial aspects in genomics
13.45	**C16.4** **High polygenic risk contributes to an early disease onset in common cardiometabolic diseases and cancers** **Nina J. Mars,** *Inst for Molecular Med Finland, HiLIFE, Univ of Helsinki, Helsinki, Finland*	**C17.4** **Skipping nonsense to maintain function: the paradigm of BRCA2 exon 12** **Laëtitia Meulemans,** *Normandie Univ, UNIROUEN, Inserm U1245, Normandy Ctr for Genomic and Personalized Med, Rouen, France*	**C18.4** **Therapeutic gene editing for Hutchinson-Gilford progeria syndrome** **Daniel Whisenant,** *Karolinska Instt, Huddinge, Stockholm, Sweden*	**C19.4** **Submicroscopic 13q32.1 deletions causing congenital microcoria modify the regulatory landscape of neighboring genes by enhancer adoption.** **LUCAS Fares Taie,** *INSTITUTE IMAGINE, Paris, France*	**C20.4** **Novel mutations in MYBPC1 associated with myogenic tremor** **Janis Stavusis,** *Latvian Biomedical Study and Res Ctr, Riga, Latvia*	**C21.4** **Novel C-terminal CUBN variants associate with chronic proteinuria and normal renal function** **Mathilda Bedin,** *INSERM U1163, Imagine Inst, Univ Paris Descartes, Paris, France*	**C22.4** **The French FIND study (preliminary results). Psychological effects of actionable secondary findings obtained from exome sequencing in patients/families with undiagnosed rare diseases.** **Françoise Houdayer,** *Genetics Dept, Reference Ctr for Developmental Disorders Sud East, HCL, Bron, France*
14.00	**C16.5** **Clinically actionable results from a multi-gene screening panel in an unselected "healthy" Canadian population** **Heather J. Andrighetti,** *Medcan, Toronto, Canada*	**C17.5** **Genome-wide association study identifies pathways associated with cervical cancer risk** **Dhanya Ramachandran,** *Gynaecology Res Unit, Hannover Medical Sch, Hannover, Germany*	**C18.5** **High efficiency of CRISPR/Cas9 gene editing of T158M-hot spot mutation in MECP2 gene** **Susanna Croci,** *Medical Genetics, Univ of Siena, Siena, Italy*	**C19.5** **First estimation of the scale of canonical 5' splice site GT>GC mutations generating wild-type transcripts and their medical genetic implications** **Jian-Min M. Chen,** *EFS, Univ Brest, Inserm, UMR 1078, GGB, Brest, France*	**C20.5** **Absence of NFASC isoform NF186 causes an autosomal recessive ataxia syndrome** **Malin Kvarnung,** *Dept of Molecular Med and Surgery, Karolinska Instt, Stockholm, Sweden*	**C21.5** **Genome-wide association study of MRI liver iron content in 9,800 individuals yields new insights into its link with hepatic and extrahepatic diseases** **Hanieh Yaghootkar,** *Univ of Exeter, Exeter, United Kingdom*	**C22.5** **Variant data sharing by clinical laboratories through public databases: consent, privacy and further contact for research policies** **Mahsa Shabani,** *KU Leuven, Leuven, Belgium*
14.15	**C16.6** **Population genomic screening of all young adults in a healthcare system: a cost-effectiveness analysis** **Paul Lacaze,** *Dept of Epidemiology and Preventive Med, Sch of Public Health and Preventive Med, Monash Univ, Melbourne, Australia*	**C17.6** **Mitochondrial damage due to a genetic origin explains the autoimmune response that leads to gastric neuroendocrine tumors** **Oriol Calvete,** *Ctr Nacional de Investigaciones Oncologicas (CNIO), Madrid, Spain*	**C18.6** **An open-label, phase 1/2 study of miransertib (ARQ 092), an oral pan-AKT inhibitor, in patients (pts) with PIK3CA-related Overgrowth Spectrum (PROS) and Proteus Syndrome (PS): study design and preliminary results (NCT03094832).** **Chiara Leoni,** *Fondazione Policlinico Univrio A. Gemelli, IRCCS, Univ Cattolica del Sacro Cuore, Rome, Italy*	**C19.6** **Novel regulatory elements control translation of key stress response factors linked to disease** **Justin Rendleman,** *New York Univ, New York, United States*	**C20.6** **Peripheral monitoring of neurodegeneration using cell-free DNA methylation** **Zac Chatterton,** *The Univ of Sydney, Camperdown, Australia*	**C21.6** **Complex compound inheritance of lethal lung developmental disorders due to disruption of the TBX-FGF pathway** **Justyna A. Karolak,** *Dept of Molecular & Human Genetics, Baylor Coll of Med, Houston, United States*	**C22.6** **Are requirements to deposit data in research repositories compatible with the GDPR?** **Deborah Mascalzoni,** *Eurac Res, Bolzano, Italy*
14.30 - 15.00	***Fruit break / Free Poster Viewing / Exhibition***						

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

TIME	EXHIBITION HALL - LIVE STREAM AREA
13.00 - 14.30	**C23** **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*
	P10.13D **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 Respirology, 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 Cloutier,** *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.

TIME	HALL C	K2+K3	F1+F2+F3	F4+F5	G2+G3	K1	H2	
15.00 - 16.30	**W12** **Dysmorphology II** Organisers: Jill Clayton-Smith Sofia Douzgou Dian Donnai	**W13** **Copy Number Variant Interpretation and Classification** Organisers: Nicole de Leeuw Erica Gerkes Zeynep Tümer	**W14** **Molecular Newborn screening vs. newborn testing** Organisers: Asbjørg Stray-Pederson Lucy Raymond	**W15** **European Reference Networks - What is in it for me?** Organisers: Conxi Lazaro Carla Oliveira	**W16** ELPAG **Opportunistic or non opportunistic genetic screening?** Organisers: Francesca Forzano Marina Cornel	**W17** **Using the Ensembl VEP for analysing variants in rare and common disease** Organiser: Emily Perry	**W18** **Pharmacogenomics in practice** Organisers: Vita Dolzan Volker Lauschke Andrea Gaedigk	**Corporate Satellites** (see page 45 for details)
	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 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.	About the workshop: Various aspects of copy number variant (CNV) interpretation and classification in a diagnostic setting will be discussed in this interactive session. The aim of this workshop is to focus on various aspects of copy number variant (CNV) interpretation and classification in a diagnostic setting. We will talk about multi-, intra- and intergenic CNVs detected by genome wide array analysis, but also CNV detection in Whole Exome/Genome Sequencing data will be included. We will use illustrative cases from our own diagnostic laboratories to have an interactive discussion on the more challenging findings, including reduced-penetrant, recurrent CNVs and structurally rearranged chromosomal imbalances as well as patients with compound heterozygous variants in a recessive disease gene. We will have an app-based feedback system available for this interactive session, so please bring your smart phone, tablet or laptop. **Programme Overview:** **15:00-15:30** **Inherited CNVs: the good, the bad, and the (very) rare** Nicole de Leeuw, University Medical Center Nijmegen, Netherlands **15:30-16:00** **Unexpected results with a merciful ending** Erica Gerkes, University Medical Center Groningen, Netherlands **16:00-16:30** **From selected abstracts** moderated by Zeynep Tümer, Kennedy Center, Department of Clinical Genetics, Rigshospitalet, Denmark	New treatment opportunities for congenital disorders challenge old statements and strategies for newborn screening. For many of the severe and treatable congenital disorders no biochemical marker can be used for screening purposes, while genetic testing may rapidly identify disease risk alleles. Effective therapies introduced before manifestation of symptoms may ameliorate disease and result in better long term outcome, also for disorders not regarded as severe and life-threatening, such as early onset retinal dystrophy. In this workshop we want to discuss current status for newborn screening in Europe in light of the implementation of NGS in health care moving towards offering babies without symptoms broad genetic testing. **15:00-15:05 Introduction** *Lucy Raymond, United Kingdom* **15:05-15:20 Genomic sequencing in Newborn Screening Programs – ethical considerations** *Pascal Borry, Belgium* **15:20-15:45 Genomic sequencing of healthy babies – Are we there yet?** *Pankaj Agrawal, United States* **15:45-16:00 NGS in newborn screening in Europe, current status** *Asbjørg Stray-Pedersen, Norway* **16:00-16:30 Panel Discussion** *Pascal Borry (Belgium), Pankaj Agrawal (United States), Lucy Raymond (United Kingdom), Asbjørg Stray-Pedersen (Norway)*	**15.00-15.10 Introduction: What are ERNs and why were they created?** *Birute Tumiene, ESHG Board and ERN Board of Member States, Vilnius University Hospital, Lithuania* **15.10-15.20 The role of European Patient Advocacy Groups (ePAG) in ERNs** *Matt Bolz-Johnson, ERN & Healthcare Advisor EURORDIS* **15.20-15.30 The role of the Genetics Community in ERNs and the Solve-RD link** *Olaf Riess, Co-Coordinator Solve-RD project, Universitätsklinikum Tübingen, Germany* **15.30-15.45 Round Table, Q&A** **15.45-15.55 Clinical patient management system (CPMS), Data collection and registries** *Alesandra Ferlini, Medical Genetics coordinator ERN on neuromuscular diseases (ERN EURO-NMD), Section of Medical Genetics, University of Ferrara, Italy* **15.55-16.05 Knowledge generation: guidelines and implementation** *Nicoline Hoogerbrugge, Coordinator ERN on genetic tumour risk syndromes (ERN GENTURIS), Radboud University Medical Center Nijmegen, The Netherlands* **16.05-16.15 Spreading the knowledge at the National level** *Elke Holinski-Feder, Coordinator of national coordinators ERN GENTURIS, MGZ, Medical Genetics Center Munich, Germany* **16.15-16.30 Round table, Q&A - What is in it for me?**	**15.00-15.15 EU survey on Opportunistic Screening practices and views** *Heidi Howard, Sweden* **15.15-15.30 PPPC-Eurogentest draft recommendations on Opportunistic Screening** *Guido De Wert, The Netherlands* **15.30-15.45 French Society of Predictive and Personalized Medicine Guidelines for reporting secondary findings of genome sequencing in cancer genes** *Pascal Pujol, France* **15.45-16.30 Panel Discussion: screening?** *Sandi Deans, United Kingdom* *Milan Macek, Czech Republic* *Heidi Howard, Sweden* *Guido De Wert, The Netherlands* *Pascal Pujol, France*	The Ensembl Variant Effect Predictor (VEP) allows analysis of variants from sequencing experiments to determine the likely effect of the variants on genes, allowing for the prioritisation for 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 images. Workshop Speakers: *Emily Perry, Ensembl Outreach Project Leader* *Irina Armean, Ensembl Variation Bioinformatician*	This workshop will introduce participants to pharmacogenetic(PGx) resources in particular PharmVar, PharmGKB, CPIC and the PGRN. The focus will be on learning what kind of pharmacogenetic information these resources provide and how to actively engage. To foster interaction presenter will actively engage with participants asking and taking question throughout. The audience will be asked which genes/variants to demonstrate. Resources will be introduced by 2-3 slides and live demonstrations of respective websites will be presented.	
16.30 - 16.45	***Coffee Break / Free Poster Viewing / Exhibition***							
16.45 - 17.45	**Poster Viewing with authors and coffee (Group D)**							

TIME	HALL C	K2+K3	F1+F2+F3	F4+F5	G2+G3	K1
17.45 - 19.15	**E11** **Genome First Testing in Pediatrics** Chair: Asbjørg Stray-Pederson	**S13** **Understanding mutations to detect cancer** Chairs: Svetlana Bajalica-Lagercrantz Conxi Lazaro	**S14** ELPAG **Debate: Genomics and the Media** Chairs: Birgitte Dinness Francesca Forzano	**E12** **Oligogenic inheritance** Chair: Alexandre Reymond	**S15** **Regulatory Landscapes** Chairs: 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*		**S15.2** **Regulation of disease-associated gene expression in the 3D genome** **Wouter De Laat,** *Oncode & 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*			**E12.2** **Epistasis in Cardiac defects** **Bart Loeys,** *Antwerp, Belgium*		
18.45		**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*		**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*
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:
https://2019.eshg.org/index.php/programme/late-programme-changes/

SCIENTIFIC

SCIENTIFIC PROGRAMME

TUESDAY, JUNE 18, 2019

PROGRAMME

ESHG

EUROPEAN SOCIETY OF HUMAN GENETICS

EUROPEAN HUMAN GENETICS CONFERENCE 2020

53rd Meeting

City Cube | Berlin – Germany | June 6 – 9

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TIME	HALL C	K2+K3	F1+F2+F3	F4+F5	G2+G3	K1
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 of 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*
	S17.2 **EMBL-EBI and global data integration** **Nick Goldman,** *European Bioinformatics Inst, Hinxton, United Kingdom*					
	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*					
09.30	**S17.4** **Pulling the Strands Together: MEGA Steps to Drive European Genomics and Personalised Medicine** **Denis DR. Horgan,** *European Alliance for Personalised Med, Bruxelles, Belgium*	**S18.2** **Timing past admixture events and characterizing their consequences in contemporary human populations** **Garrett Hellenthal,** *Univ Coll London, London, United Kingdom*	**S19.2** **Gene therapy for inherited neuromuscular disorders** **Francesco Muntoni,** *Univ Coll London Great Ormond Street Inst of Child Health, London, United Kingdom*		**S20.2** **Genetic-epigenetic interactions: mechanistic insights and practical applications** **Benjamin Tycko,** *HMH Ctr for Discovery and Innovation, Nutley, United States*	
	Debate			**E13.2** **Population genetic carrier screening programs for reproductive purposes** **Joël Zlotogora,** *Hadassah medical center, Hebrew Univ, Jerusalem, Israel*		**E14.2** **CRISPR/Cas9 and TALENs fuel genetically engineered clinically relevant Xenopus tropicalis models** **Kris Vleminckx,** *Ghent, Belgium*
10.00		**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*		**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*	
10.30 - 11.00	*Coffee Break (Aisle G, Aisle F, Hall H)*					

TIME	HALL C	K2+K3	F1+F2+F3	F4+F5	G2+G3	K1
11.00 - 12.30	**C24** **Mosaicisms** Chairs: Jill Clayton-Smith Feliciano Ramos	**C25** **Bioinformatics and multiomics** Chairs: Christian Gillisen Daniel Nilsson	**C26** **Mitochondrial disorder** Chairs: Olaf Bodamer Elsebet Østergaard	**C27** **Developmental disorders 2** Chairs: Marta Bertoli Trine Prescott	**C28** **Late Breaking abstracts** Chairs: Inga Prokopenko Anna Lindstrand	**C29** ELPAG **Stakeholder perspectives in cancer genetics** Chairs: Luzia Garrido Ulf Kristoffersson
11.00	**C24.1** **Analysis of Mosaicism for Sequence and Copy Number Variants in a Broad Diversity of Hereditary Disorders in a Large Clinical Cohort** **Daniel Pineda Alvarez,** *Invitae, San Francisco, United States*	**C25.1** **A transcriptome-wide Mendelian randomization study to uncover tissue-dependent regulatory mechanisms across the human phenome** **Tom G. Richardson,** *MRC Integrative Epidemiology Unit, Bristol, United Kingdom*	**C26.1** **Mutations in LIG3 are a novel cause of mitochondrial neurogastrointestinal encephalomyopathy** **Elena Bonora,** *University of Bologna, Bologna, Italy*	**C27.1** **Diagnostic utility of genome-wide DNA methylation testing in genetically unsolved patients with suspected hereditary conditions** **Bekim Sadikovic,** *London Health Sciences Ctr, Canada, London, Canada*	**C28.1** **Whole exome sequencing and characterization of coding variation in 49,960 individuals in the UK Biobank** **Cristopher Van Hout,** *Regeneron Pharmaceuticals, Tarrytown, United States*	**C29.1** **The public favours healthcare-mediated disclosure of hereditary cancer risk to at-risk relatives: a population-based survey in Sweden** **Carolina Hawranek,** *Dept of Radiation sciences, Oncology, Umeå university, Umeå, Sweden*
11.15	**C24.2** **Uniparental disomy in the Rare Disease Programme of the UK's 100,000 Genomes Project** **Katherine R. Smith,** *Queen Mary Univ of London, London, United Kingdom*	**C25.2** **Multivariate GWAS of inflammatory markers reveals novel disease associations** **Sanni E. Ruotsalainen,** *Inst for Molecular Med Finland, Univ of Helsinki, Helsinki, Finland*	**C26.2** **Mutations in POLRMT impair mitochondrial transcription and are associated with a spectrum ofmitochondrial disease presentations** **Robert W. Taylor,** *Wellcome Ctr for Mitochondrial Res, Newcastle Univ, Newcastle upon Tyne, United Kingdom*	**C27.2** **Multiomics Approach to Diagnosing Undiagnosed Patients** **Matthew T. Wheeler,** *Stanford Univ Sch of Med, Palo Alto, United States*	**C28.2** **A novel ciliary Joubert Syndrome-associated protein module regulates axonemal post translational modifications and cilium stability** **Ruxandra Bachmann-Gagescu,** *University of Zurich- Medical Genetics, Zürich, Switzerland*	**C29.2** **Communication across generations: disclosure of BRCA cancer risk with young adults** **Alison Luk. Young,** *Ctr for Medical Psychology & Evidence-based Decision-making (CeMPED), Sch of Psychology, The Univ of Sydney, Sydney, Australia*
11.30	**C24.3** **Somatic mutation cell lineage analysis reveals progressive clonal determination in human embryo** **Sara Bizzotto,** *Boston Children's Hosp, Dept of Genetics and Genomics, Manton Ctr for Orphan Disease, Boston, United States*	**C25.3** **Analysis of genetic variants through aggregation of homologous human protein domains via MetaDome strongly improves diagnostic prediction of missense variants** **Laurens Wiel,** *Dept of Human Genetics, Radboud Inst for Molecular Life Sciences, Radboud Univ Medical Ctr, Nijmegen, Netherlands*	**C26.3** **Mutations in the MRPS28 gene encoding the small mitoribosomal subunit protein bS1m in a patient with intrauterine growth retardation, craniofacial dysmorphism and multisystemic involvement** **Juliette Pulman,** *UMR1163, Univ Paris Descartes, Sorbonne Paris Cité, Inst IMAGINE, Paris, France*	**C27.3** **Mutated epigenetic modifiers in CYLD cutaneous syndrome** **Neil Rajan,** *Inst of Genetic Med, Newcastle upon Tyne, United Kingdom*	**C28.3** **Index and vocabulary of accessible DNA elements in the human genome** **Wouter Meuleman,** *Altius Institute for Biomedical Sciences, Seattle, United States*	**C29.3** **High-Risk Women's Responses and Understanding of Polygenic Breast Cancer Risk Information** **Tatiane Yanes,** *Univ of New South Wales, Sydney, Australia*
11.45	**C24.4** **Basal and mutagen-driven somatic mutagenesis shape the genome of healthy human cells** **Irene Franco,** *Karolinska Instt, HUDDINGE, Sweden*	**C25.4** **reg2gene: predicting enhancer-gene associations using ensemble learning approaches** **Inga Patarcic,** *MDC BIMSB, Berlin, Germany*	**C26.4** **Brain-on-a-chip - a neurophysiological model of MELAS disease and comorbid psychopathology** **Tamas Kozicz,** *Mayo Clinic, Rochester, United States*	**C27.4** **Identification and characterization of NEPRO-related skeletal dysplasia resembling cartilage hair hypoplasia** **Dhanya Lakshmi Narayanan,** *Kasturba Medical Coll, Manipal, Manipal Acad of Higher Education, Manipal, Manipal, India*	**C28.4** **Modeling the pathological long-range regulatory effects of human structural variation with patient-specific hiPSCs** **Magdalena Laugsch,** *Centre for Molecular Medicine Cologne (CMMC), University of Cologne, Cologne, Germany*	**C29.4** **Families' and healthcare professionals' uncertainties in the era of cancer precision medicine: results from PRISM** **Janine Vetsch,** *Behavioural Sciences Unit, Kids Cancer Ctr, Sydney Children's Hosp, Randwick, Australia*

TIME	HALL C	K2+K3	F1+F2+F3	F4+F5	G2+G3	K1
cont.	**C24** Mosaicisms	**C25** Bioinformatics and multiomics	**C26** Mitochondrial disorder	**C27** Developmental disorders 2	**C28** Late Breaking abstracts	**C29** ELPAG Stakeholder perspectives in cancer genetics
12.00	**C24.5** **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** **Arthur Sorlin,** *Ctr de Génétique, CHU Dijon Bourgogne, Dijon, France*	**C25.5** **A GWAS on data-driven 3D facial phenotypes selected by matching siblings reveals 310 genetic loci** **Hanne Hoskens,** *Dept of Human Genetics, KU Leuven, Leuven, Belgium*	**C26.5** **The homozygous variant c.797G>A/p.(Cys266Tyr) in PISD is associated with a spondyloepimetaphyseal dysplasia with large epiphyses and disturbed mitochondrial function** **Leonie von Elsner,** *Inst of Human Genetics, Univ Medical Ctr Hamburg-Eppendorf, Hamburg, Germany*	**C27.5** **Impact of ALPK1 causative variant in ROSAH syndrome, a newly characterised retinal and multisystem autosomal dominant disorder** **Robyn V. Jamieson,** *Eye Genetics Res Unit, Children's Medical Res Inst, The Children's Hosp at Westmead, Save Sight Inst, Univ of Sydney, Sydney, Australia*	**C28.5** **Cerebral organoids provide insights into brain neurodevelopment disrupted by the 16p11.2 copy number variants in autism** **Lilia Iakoucheva,** *University of California San Diego, La Jolla, United States*	**C29.5** **Advanced cancer patient perspectives on consenting to molecular tumour profiling** **Megan Best,** *Univ of Sydney, Sydney, Australia*
12.15	**C24.6** **The Hutchinson-Gilford progeria syndrome mutation is a somatic mutation in chronic kidney disease** **Maria Eriksson,** *Karolinska Instt, Dept of Biosciences and Nutrition, Huddinge, Sweden*	**C25.6** **GestaltMatcher: Identifying the second patient of its kind in the phenotype space** **Tzung-Chien Hsieh,** *Inst for Genomic Statistics and Bioinformatics, Bonn, Germany*	**C26.6** **SSBP1 mutations cause a complex optic atrophy spectrum disorder with mitochondrial DNA depletion** **Tommaso Pippucci,** *Medical Genetics Unit, Sant'Orsola-Malpighi Univ Hosp, Bologna, Italy*	**C27.6** **New mechanism for retinal degeneration on chrXq27.1** **Jessica C. Gardner,** *UCL Inst of Ophthalmology, 11-43 Bath Street, EC1V 9EL, London, United Kingdom*	**C28.6** **Preclinical validation of allele-specific silencing in patient-derived directly reprogrammed neuronal cell lines as effective treatment for Autosomal Dominant LeukoDystrophy.** **Elisa Giorgio,** *University of Torino-Dep Medical Sciences, Torino, Italy*	**C29.6** **Dimensions of grief and loss for families living with Li Fraumeni Syndrome** **Allison Werner-Lin,** *Sch of Social Policy and Practice, Philadelphia, United States*
12.30 - 13.30	***Lunch break (Aisle G, Aisle F, Hall H)***					

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/

TIME	HALL C
13.30 - 14.15	**PL3** **Mendel Lecture** Chairs: Alexandre Reymond, Joris Veltman
13.30	**PL3.1** **A 25 Year Genomic Odyssey** **Craig Venter,** *J. Craig Venter Inst, San Diego, United States* *Laudation by Alexandre Reymond*
14.15 - 15.00	**PL4** **ESHG Award Lecture** Chairs: Alexandre Reymond, Joris Veltman
15.00	**PL4.1** **We and our second genome: two key players in common complex diseases** **Cisca Wijmenga,** *Lodewijk Sandkuijl Professor of Human Genetics, UMCG Groningen, Groningen, Netherlands* *Laudation by Joris Veltman*
15.00 - 15.45	**PL5** **Award Ceremony** Chairs: Joris Veltman, Alexandre Reymond
	• **EJHG-SN Citation Awards** • **ESHG Young Investigator Awards:** - **ESHG Young Investigator Awards** for Outstanding Science - **Isabelle Oberlé Award** for an outstanding presentation in the field of genetics of intellectual disability - **Lodewijk Sandkuijl Award** for an outstanding presentation in the field of complex disease genetics and statistical genetics - **Vienna Medical Academy Award** for an outstanding presentation in translational genetic research/therapy of genetic diseases - **Mia Neri Award** for an outstanding presentation in the field of cancer • **ESHG Poster Awards** in clinical research and basic science • **Closing remarks**

Late Programme Changes

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PROGRAMME

PROGRAMME INFORMATION

SPONSORED SESSION

CORPORATE SATELLITE MEETINGS

BUSINESS MEETINGS

YOUNG INVESTIGATOR AWARD CANDIDATES

POSTER AWARD CANDIDATES

INFORMATION

Saturday, June 15, 08.00 - 10.00 hrs

TIME	HALL C
08.00 - 10.00	**E01** **New technologies (Sponsored by Illumina)** Chair: Martin Kircher
08.00	**E01.1** **Using single cell genomics to understand cell fate decisions** **John Marioni,** *Cambridge, United Kingdom*
08.30	**E01.2** **Resolving human genetic variation with long-read single-molecule sequencing** **Mark J. Chaisson,** *Univ. of Southern California, Los Angeles, United States*
09.00	**E01.3** **Advancing single-cell genomics using combinatorial indexing** **Andrew Adey,** *Portland, United States*
09.30	**E01.4** **In vivo optical imaging and insights into human disease** **Michelle Digman,** *Irvine, United States*

Overview

Saturday, June 15, 10.00 - 11.30 hrs

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

Stand # 428

Genetic Profiling and Functional Screening for Drug Target and Biomarker Discovery

Join Cellecta 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 Cellecta technologies
Paul Diehl, Ph.D., COO, Cellecta, 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.cellecta.com/eshg2019

Cellecta 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.cellecta.com.

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

Stand # 532

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

Stand # 642 (BGI)

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 — **Stand # 220**

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 — **Stand # 336**

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 — **Stand # 280**

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

Ionis Pharmaceuticals & AstraZeneca, Saturday, June 15, 2019, 12.15–13.45 hrs, Room A-1 – Level 1 — **Not exhibiting**

Bridging The Gap: Translating Genomic Discovery Into Human Therapeutics

Sequencing technology has enabled a revolution in genomic discovery leading to the identification of genes and pathways implicated in many human diseases with great unmet medical needs, paving a path for drug target discovery and development of novel therapeutics.
This free educational luncheon event is open to all ESHG attendees interested in learning about the opportunities that the antisense platform offers in the translation of genomic discoveries into meaningful human therapeutics. A goal for this event is to engage the scientific community and find opportunities to collaborate on novel target discovery to potentially create novel therapeutics to rare and genetic disorders.

Talks will include:

Bridging The Gap: Translating Human Genetics to Therapies for Human Diseases
Chair: Tamar Grossman, PhD, Director, Translational Medicine, Ionis Pharmaceuticals, Carlsbad, USA

Antisense Technology as Means for Bridging the Gap
Chair: Brett Monia, PhD, COO, Ionis Pharmaceuticals, Carlsbad, USA

Applying Antisense Technology to Treat Cardiometabolic Diseases
Regina Fritsche-Danielson, PhD, Senior Vice President, Head of early Cardiovascular, Renal and Metabolism, R&D BioPharmaceuticals, AstraZeneca, Gothenburg, Sweden

Ligand Conjugated Antisense Oligonucleotide (LICA) Strategies for the Pancreas and Beyond
Shalini Andersson, PhD, Head of CVMD iMed DMPK, AstraZeneca, Gothenburg, Sweden

NimaGen, Saturday, June 15, 2019, 12.15–13.45 hrs, Room A-2 – Level 1 — **Stand # 348**

Reverse Complement PCR: The new revolution in NGS targeted library preparation

Speaker: *Daniel Ward, NHS Wessex Regional Genetics Laboratory, Salisbury, UK*

NimaGen will today introduce the new RC-PCR technology, making NGS library prep as simple as a single PCR. With just a single tube, single step reaction for both targeted amplification and NGS indexing.

This new technology solves common issues of amplicon based library prep methods, combined with the easiest workflow on the market: PCR > Pool > Clean > Sequence. By combining target specific amplification and indexing in just one reaction, PCR contamination is eliminated. The improved reaction dynamics of RC-PCR result in high target specificity, reduced primer dimerization and enhanced quantification.

In this meeting, Daniel Ward, one of the inventors of the RC-PCR technology, will present and elucidate the principles of the method. Also, NimaGen will present the first commercially available kit using this method: The Human Exome Sample Tracking and Identification kit for WGS and WES data integrity checking.

Sunday, June 16, 11.15 - 12.45 hrs

Agilent Technologies, Sunday, June 16, 2019, 11.15–12.45 hrs, Room A-1 – Level 1 — **Stand # 634**

Approaches for Precision Medicine in Cancer

Fast and high precision cancer samples analysis using SureSelect XTHS / 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 — **Stand # 274**

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 exhibiting trinucleotide 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.P.E, 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 onlus Foundation, Rome, Italy

Fabric Genomics, Sunday, June 16, 2019, 11.15–12.45 hrs, Room A-4 – Level 1

Stand # 374

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). ACE 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

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

Stand # 244

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.

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

Stand # 546

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.

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

Stand # 538

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 (CARD), 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 transforming 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 METexon 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. Bjoern 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 genetic 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® NIPT 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

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SOPHiA GENETICS, Sunday, June 16, 2019, 15.00–16.30 hrs, Room A-1 – Level 1 **Stand # 258**

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 variants 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, Genotypos-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 **Stand # 332**

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 **Stand # 260**

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 **Stand # 430**

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 — Stand # 634

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 — Stand # 468

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) Medicover 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 — Stand # 552

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 Sültmann, 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.

Sistemas Genómicos, Monday, June 17, 2019, 11.15–12.45 hrs, Room A-2 – Level 1 — **Stand # 286**

How to decode and interact with human genome

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

Knowledge of the human genome provides an understanding of the origin of the human species, the relationships between populations, and the health tendencies or disease risks of humans. Indeed, in the past 20 years knowledge of the sequence and structure of the human genome has revolutionized many fields of study, including medicine, anthropology and forensic. With technological advances that enable inexpensive and expanded access to genomic information, the amount of and the potential applications for the information that is extracted from the human genome is extraordinary.

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

Thermo Fisher Scientific, Monday, June 17, 2019, 11.15–12.45 hrs, Room A-4 – Level 1 — **Stand # 538**

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 (CTS).

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

Loop Genomics, Monday, June 17, 2019, 15.00–16.30 hrs, Room A-4 – Level 1 — **Stand # 574**

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.

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.

Twist Bioscience, Monday, June 17, 2019, 15.00–16.30 hrs, Room A-2 – Level 1 — **Stand # 436**

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

As per date of printing.

Friday, June 14, 2019

Time	Meeting	Room	
08.00 – 18.00 hrs	UEMS Exams	R24+R25	closed
09.00 – 13.00 hrs	ESHG Executive Board Meeting	J1	closed
13.30 – 18.00 hrs	ESHG Board Meeting I	J1	closed

Saturday, June 15, 2019

Time	Meeting	Room	
09.00 – 13.30 hrs	ESHG Quality Subcommittee Meeting	J1	closed
09.00 – 13.30 hrs	ESHG PPPC Meeting	R4	closed
10.00 – 18.30 hrs	EBMG Exams	R5	closed
12.00 – 14.00 hrs	GDPR Code of Conduct for Health Reasearch Meeting	R3	closed
12.15 – 13.45 hrs	ESHG Junior Branch Meeting	H2	open to ESHG members
16.00 – 18.30 hrs	GDPR Code of Conduct for Health Reasearch Meeting	R3	closed

Sunday, June 16, 2019

Time	Meeting	Room	
08.15 – 10.45 hrs	European Network of Genetic Nurses and Counsellors Meeting	G4	open to counsellors
10.00 – 17.00 hrs	EBMG Exams	R5	closed
10.00 – 10.45 hrs	ESHG-SpringerNature Meeting	R3	closed
10.00 – 11.00 hrs	GDPR Code of Conduct for Health Reasearch Meeting	R4	closed
11.00 – 13.00 hrs	National Human Genetics Societies Meeting	G4	closed
11.15 – 12.45 hrs	GENIDA project Advisory Board	J1	closed
11.15 – 13.15 hrs	Genetic Testing Across ERNs Meeting	R4	closed
13.00 – 14.00 hrs	GNGC professional branch meeting	R3	closed
13.30 – 15.15 hrs	EBMG Clinical Laboratory Geneticists (CLG) Meeting	G4	open to public
13.30 – 19.30 hrs	GDPR Code of Conduct for Health Reasearch Meeting	R4	closed
16.30 – 18.00 hrs	Building Bridges ESHG/ASHG Meeting	R3	closed
16.30 – 18.00 hrs	ERN-ITHACA Ancillary Meeting	J1	closed
18.15 – 19.15 hrs	ERN-ITHACA Ancillary Meeting 2	J1	closed
19.30 – 20.30 hrs	ESHG Membership Meeting	H2	open to ESHG members

Monday, June 17, 2019

Time	Meeting	Room	
08.00 – 09.00 hrs	EBMG BMGG Boards Meeting	J1	closed
08.30 – 10.30 hrs	ESHG Education Committee Meeting	R3	closed
09.00 – 10.00 hrs	UEMS SMG Boards Meeting	J1	closed
10.00 – 13.00 hrs	UEMS Section Meeting	J1	closed
10.00 – 11.00 hrs	EJHG Editorial Board Meeting	R4	closed
10.15 – 11.15 hrs	ESHG/ASHG Leadership	R5	closed
10.30 – 13.00 hrs	GDPR Code of Conduct for Health Reasearch Meeting	R3	closed
11.45 – 12.45 hrs	ESHG Board Meeting II	G4	closed
12.00 – 14.00 hrs	Editorial Board Meeting for the European Journal of Medical Genetics	R4	closed
13.00 – 15.00 hrs	EBMG General Assembly	J1	closed
13.15 – 15.15 hrs	GenQA Educational Rapid Prenatal Aneuploidy Testing Workshop	G4	open to public
15.00 – 16.00 hrs	IFHGS Executive Board Meeting	R4	closed
16.00 – 18.00 hrs	ICHG 2021 ISPC	R4	closed

Tuesday, June 18, 2019

Time	Meeting	Room	
12.15 – 13.15 hrs	ESHG SPC Meeting	J1	closed

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

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 Charpentler
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.

ARV swedish design

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 Weissenbach
1994 Mary Lyon
1993 Pierre Maroteaux
1992 Lore Zech

Professor Cisca Wijmenga

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.

Professor Cisca Wijmenga
Lodewijk Sandkuijl Professor of Human Genetics at the University of Groningen and the University Medical Centre, Groningen, The Netherlands.

«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.

Professor Craig Venter
Founder, chairman, and CEO of the J. Craig Venter Institute, La Jolla, California, USA

"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 particpation 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:

https://2019.eshg.org/index.php/abstracts-2/yiacandidates/

ESHG Poster Awards

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

https://2019.eshg.org/index.php/abstracts-2/postercandidates/

Saturday, June 15 at 16:30 hrs

PL2.3

Triin Laisk
Tartu, Estonia

PL2.4

Joanna Kaplanis
Cambridge, United Kingdom

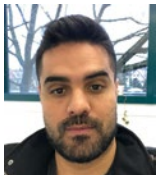
PL2.5

Lila Allou
Berlin, Germany

Saturday, June 15 at 18:30 hrs

C01.2

Mamiko Yamada
Shinjuku, Japan



C01.3

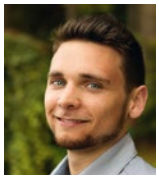
Uira Melo
Berlin, Germany

C02.3

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

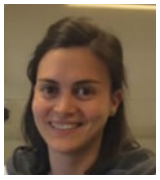
Manon Oud
Nijmegen, Netherlands

C04.5

Digumarthi Sudhakar
Habsiguda, India

C05.3

Helen Roessler
Utrecht, Netherlands



C05.4

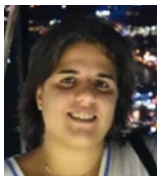
Sara Cuvertino
Manchester, United Kingdom

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

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

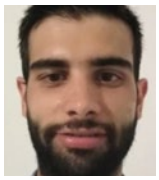
C13.2
Lianmin Chen
Groningen, Netherlands

C13.3
Jonas Bovijn
Oxford, United Kingdom

C13.4
Maria Santos
Madrid, Spain

C13.5
Henrike Heyne
Cambridge, United States

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

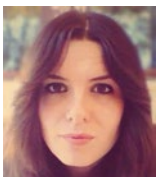
C17.4
Laëtitia Meulemans
Rouen, France

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

Monday, June 17 at 13.00 hrs

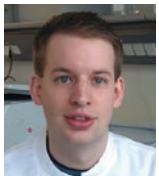
C19.3
Lisanne Vervoort
Leuven, Belgium

C19.6
Justin Rendleman
New York, United States

C20.2
Natalia Mendoza Ferreira
Cologne, Germany

C20.3
Elisa Fernández-Núñez
Madrid, Spain

C20.5
Malin Kvarnung
Stockholm, Sweden



C21.2
Helge Martens
Hannover, Germany

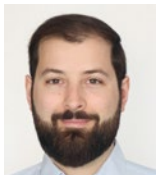
C21.4
Mathilda Bedin
Paris, France

C22.2
Danya Vears
Parkville, Australia

C22.5
Mahsa Shabani
Leuven, Belgium

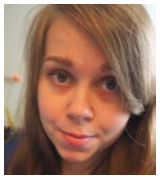
Tuesday, June 18 at 11.00 hrs

C24.3
Sara Bizzotto
Boston, United States



C24.5
Arthur Sorlin
Dijon, France

C25.1
Tom Richardson
Bristol, United Kingdom



C25.2
Sanni Ruotsalainen
Helsinki, Finland

C25.3
Laurens Wiel
Nijmegen, Netherlands

C25.4
Inga Patarcic
Berlin, Germany

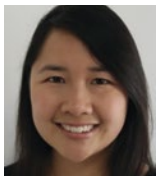
C25.5
Hanne Hoskens
Leuven, Belgium

C25.6
Tzung-Chien Hsieh
Bonn, Germany

C26.3
Juliette Pulman
Paris, France

C26.5
Leonie von Elsner
Hamburg, Germany

C27.4
Dhanya Lakshmi Narayanan
Manipal, India



C29.2
Alison Young
Sydney, Australia

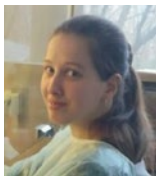
C29.3
Tatiane Yanes
Sydney, Australia

C29.4
Janine Vetsch
Kensington, Australia

PROGRAMME POSTER AWARD FINALISTS

P01.33A

Natalia Pervjakova
Tartu, Estonia



P02.49C

Alexandra Filatova
Moscow, Russian Federation

P02.50D

Anna Morgan
Trieste, Italy

P02.56B

Stijn Van de Sompele
Ghent, Belgium

P04.41A

Ronja Hollstein
Bonn, Germany

P05.52D

Minttu Marttila
Helsinki, Finland

P07.32D

Dylan Lawless
Lausanne, Switzerland

P08.24A

Alexej Knaus
Bonn, Germany

P08.35D

Reza Ataeijaliseh
Tehran, Iran, Islamic Republic of

P08.63D

Diana Le Duc
Leipzig, Germany

P09.008C

Moeen Riaz
Melbourne, Australia

P09.012C

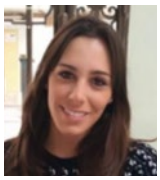
Christopher Hübel
Stockholm, Sweden

P09.027B

Martin Becker
Solna, Sweden

P09.028C

Abdulrahman Ali
Dubai, United Arab Emirates

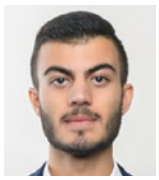


P09.126A

Alba Sanchis-Juan
Cambridge, United Kingdom

P11.03C

Guillaume Jouret
Reims, France



P11.23C

Seyed Ali Safizadeh Shabestari
Dubai, United Arab Emirates

P11.68D

Daniel Halperin
Beer-Sheva, Israel

P13.13A

Pauline Schneeberger
Hamburg, Germany

P14.021D

Kevin Cassinari
Rouen, France

P14.056C

Bethany Wild
London, United Kingdom

P14.067B

Tamara Simakova
St. Petersburg, Russian Federation

P15.39B

Heidi Marjonen
Helsinki, Finland

P16.02B

Brunilda Balliu
Los Angeles, United States

P16.57A

Christian Mertes
Garching, Germany

P17.14C

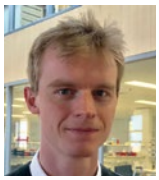
Verena Heinrich
Berlin, Germany

P17.40A

Simon Fishilevich
Rehovot, Israel

P18.29A

Philip Jansen
Amsterdam, Netherlands



P18.42B

Adriaan van der Graaf
Groningen, Netherlands

P21.07B

Bettina Zimmermann
Basel, Switzerland

P23.02B

Jane Tiller
Melbourne, Australia

GENERAL

INFORMATION

GENERAL INFORMATION

REGISTRATION FEES

NETWORKING EVENTS

CORPORATE EXHIBITION

INFORMATION

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

Gothia Towers is located near the Korsvägen stop. Korsvägen is a hub for public transport in Gothenburg, and numerous tram and bus lines stop there. Korsvägen is also the first stop for the airport bus coming from Göteborg Landvetter airport. Note only card payments are accepted at the time of purchasing single tickets in trams and boats. It is not possible to buy tickets in city buses.

The stops Korsvägen and Liseberg are about 100 metres from the Gothia Towers hotel/Conference centre entrance respectively.

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.

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/

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 CO_2 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 (vårdcentral). 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 attemps 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.

Registration Fees[1] Payment received:	*before April 4, 2019 (reduced rate)*	*from April 4 to May 23, 2019 (regular rate)*	*after May 23, 2019 and on site*	*Day Tickets on site*
Participants ESHG Members	EUR 330.-	EUR 440.-	EUR 500.-	EUR 165.-
Participants Non-Members	EUR 500.-	EUR 600.-	EUR 670.-	EUR 230.-
Postgraduate Trainees ESHG Members[2]	EUR 220.-	EUR 320.-	EUR 385.-	EUR 145.-
Postgraduate Trainees Non- Members[2]	EUR 330.-	EUR 440.-	EUR 500.-	EUR 165.-
Counsellors/Gen.Nurses ESHG Members[3]	EUR 220.-	EUR 320.-	EUR 385.-	EUR 145.-
Counsellors/Gen.Nurses Non-Members[3]	EUR 330.-	EUR 440.-	EUR 500.-	EUR 165.-
Students[4]	EUR 115.-	EUR 170.-	EUR 220.-	EUR 100.-
ESHG Members from underprivileged countries[5]	EUR 220.-	EUR 220.-	EUR 220.-	EUR 100.-
Non-Members from underprivileged countries[5]	EUR 290.-	EUR 290.-	EUR 290.-	EUR 120.-
Lunch bags/boxes per day[6]	EUR 19.-	EUR 19.-	N/A	N/A
	Participants/ Guests	Students/ Postgrad. Trainees		
Networking Evening at own expense	EUR 60.-	EUR 40.-		

[1]Registration Fees include 25% Swedish VAT.

[2]Applies to *MSc./PhD students working towards a degree in human genetics or an associated field.* Please provide a confirmation signed by the head of department **at the moment of your registration** at the registration desk. **Confirmations handed in at a later stage cannot be considered.**

[3]Applies to *non-MD/PhD*-Counsellors.

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

[5]Applies to a **selected list of countries.**

[6]Not 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

REGION VÄSTRA GÖTALAND

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

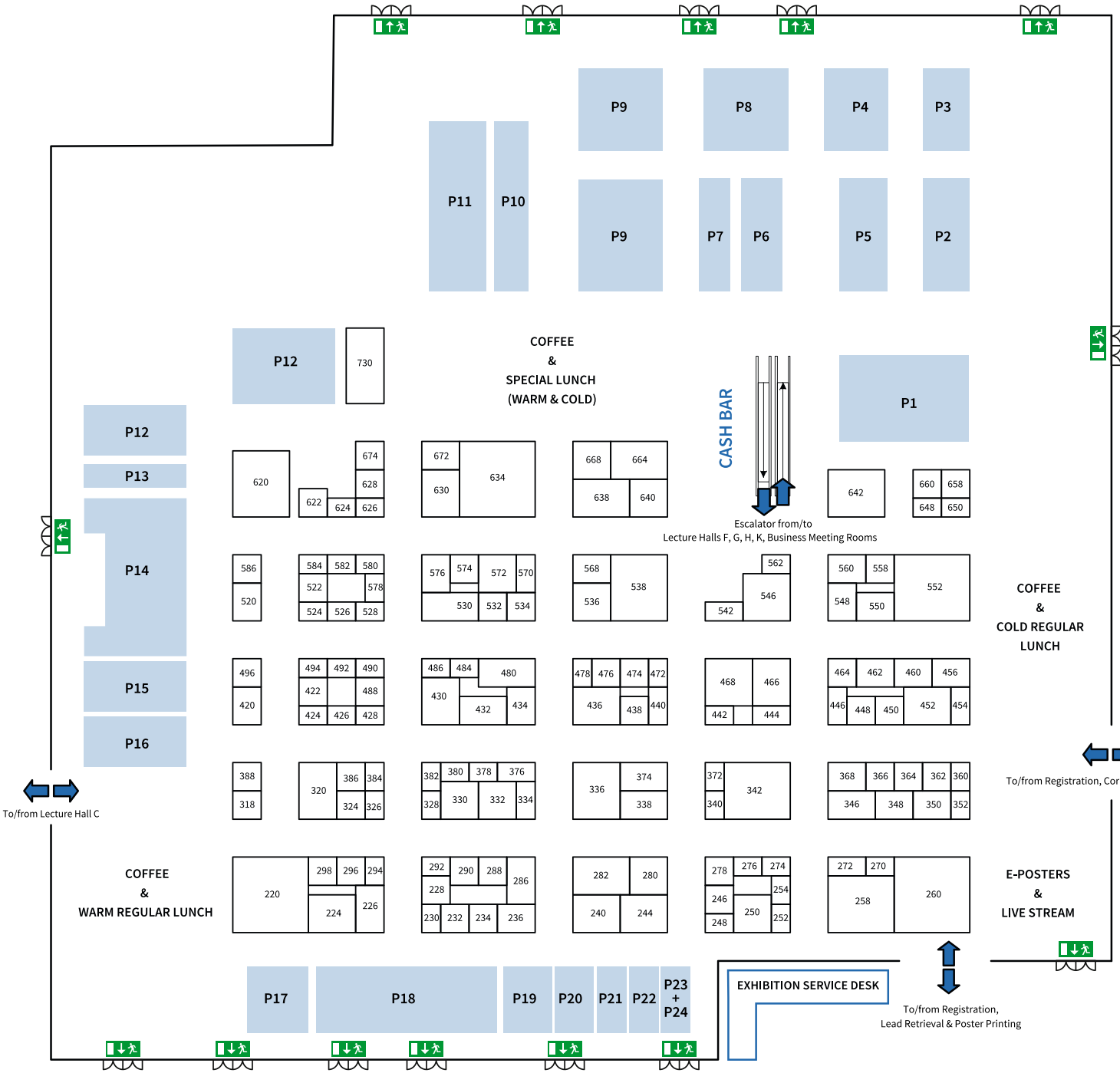
Please note that payment of extra drinks is only possible with credit/bank cards.

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.
https://2019.eshg.org/index.php/myconference/official-events/

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.



POSTER TOPICS

- **P01** Reproductive genetics - Prenatal genetics
- **P02** Sensory disorders (eye, ear, pain)
- **P03** Internal organs & endocrinology (lung, kidney, liver, gastrointestinal)
- **P04** Skeletal, connective tissue, ectodermal and skin disorders
- **P05** Cardiovascular disorders
- **P06** Metabolic and mitochondrial disorders
- **P07** Immunology and hematopoietic system
- **P08** Intellectual disability
- **P09** Neurogenetic and psychiatric disorders
- **P10** Neuromuscular disorders
- **P11** Multiple malformation/anomalies syndromes
- **P12** Cancer genetics
- **P13** Basic mechanisms in molecular and cytogenetics
- **P14** New diagnostic approaches - Technical aspects - Quality control
- **P15** Personalized/predictive medicine - Pharmacogenomics
- **P16** Omics - Bioinformatics
- **P17** Epigenetics - Gene regulation
- **P18** Genetic epidemiology - Population genetics - Statistical methodology - Evolutionary genetics
- **P19** Genetic counselling - Services - Education
- **P20** Psychological and social issues in genetics
- **P21** Lay beliefs and public understanding of genetics - Access to genetic services
- **P22** Ethical issues in genetics
- **P23** Legal implications of advances in genetics
- **P24** Other relevant ELPAG issues in genetics

List correct as per date of printing - Exhibition floor plan is in your conference bag.

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

Saturday, June 15, 2019:	09.30 – 18.30 hrs
Sunday, June 16, 2019:	09.00 – 17.45 hrs
Monday, June 17, 2019:	09.00 – 17.45 hrs
Tuesday, June 18, 2019:	CLOSED

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:

Saturday, June 15, 2019:	09.30 – 18.30 hrs	Poster mounting / viewing
Sunday, June 16, 2019:	09.00 – 17.45 hrs	Poster viewing
Monday, June 17, 2019:	09.00 – 17.45 hrs	Poster viewing
	16.45 – 17.45 hrs	**Poster removal – Groups A–C (strict!)**
	17.45 – 17.50 hrs	**Poster removal – Group D only (strict!)**

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):
  - o name and full postal address
  - o e-mail address

Thank you for your understanding and cooperation.

Exhibition & Sponsorship Management

Name	**ROSE INTERNATIONAL**
	Exhibition Management & Congress Consultancy bv
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	NL-2509 AG The Hague, the Netherlands
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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

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NOTES


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