Presidential Address

by Gunnar Houge, President of the ESHG

Dear ESHG membership,

My year as ESHG president is nearly over, after receiving the president relay baton from Christine Patch, a genetic counsellor. Soon the presidency will pass on to Alexandre Reymond, the first molecular biologist to hold the post in 15 years. I think it is important that our Board, and especially the executive part of it (the “Exec”), reflects that the fundament of our society is teamwork involving all important stakeholders; molecular geneticists, clinicians and genetic counsellors.

My presidential year has been full of interesting tasks. On a very practical and necessary level for the Society, and thanks to the excellent work of Jerome del Picchia and the Vienna Medical Academy (VMA), the transfer of the Society from Belgium to Austria is on track, with our funds in the Netherlands to follow the same route. Revised statutes complying with requirements of Austrian law have been written and will be presented to the Board and Membership in Gothenburg. The aim is to have an Austrian-based society from 2020, making ESHG administration far easier and based in one instead of three countries.

A personal goal was to initiate work on an improved genetic variant classification system, a need expressed by many genetic professionals. A prototype has been made, based on input from an ESHG task force with complementary competencies: Johan den Dunnen (LOVD/HGVS), Hans Schaeffer (Eurogentest), Helen Firth (DECIPHER) and Nicole de Leeuw (molecular cytogenetics). The proposed classification system is two-dimensional and adds a clinical arm to the well-known molecular classification system. The molecular grading addresses parameters like gene function, structure, variability and variant prevalence, with gnomAD as one crucial data source. The clinical arm brings into play parameters like expressivity and penetrance, and the likelihood that the gene implicated is a likely candidate for the phenotype or not. The end result is a combined grade (e.g. 4+2) that is linked to alternative standard results, dependent on the clinical question. The system has been designed for classification of all variants including those associated with likely recessive and hypomorphic alleles. We are currently in the process of real-life testing. This ESHG effort is meant to complement the ACMG/AMP system. I think that a new system which integrates the value of the clinical impression is much needed, to standardise reporting and safeguard our practice by diminishing confusion and creating a common language shared by different genetic laboratories. This is very much work in progress and we would value feedback from ESHG members.

Another goal was to strengthen our educational portfolio in collaboration with the ESHG Education Committee, led by Han G. Brunner, and various ESHG members. Currently, funds of around € 100 000 are used per year for courses and course fellowships, money that are generated by our Journal. We should be most grateful for the excellent work that the EJHG editor Gert-Jan van Ommen and his small staff have done for years to make our Journal attractive and profitable.

There is currently a concern among society-associated medical journals that the proposed Plan S (immediate and free open access) could diminish the income for scientific societies substantially. It remains to be seen what will happen, but we are looking for alternative sources of income for education, e.g. to build up an ESHG educational fund. One other stakeholder that also thinks education is essential for successful implementation of precision medicine, is Illumina, and we have had constructive discussions about this with them. There is a good chance that they will sup-

If you want additional information about the system, please attend the Membership meeting Sunday evening.

Molecular grading (a VUS = 0)

Clinical grading (a VUS = 0)
port new ESHG courses on variant interpretation and prenatal diagnostics, the latter being a course for both gynaecologists and geneticists working in this field. There will be no company strings attached that could jeopardize e.g. a need for CME credits. Such courses may bridge societies, much like the excellent cardiogenetics course arranged by Bart Loeys and William Newman with attendees from both cardiology and genetics societies. In my view, this is a better way of liaising with other societies than having joint sessions at our annual meetings. ESHG also had a record number of members in 2018, passing 3000 for the first time. Naturally, this has increased our income to the benefit of the whole society.

There is also other good news that I want to share with you. This year “ESHG Young” will be established, an initiative of our ‘young’ members! The initiative stems from members of a Facebook group called “Young Geneticist Network” (YGN) with 343 members (per 28.04.19). I hope this group will interact with our Scientific Planning Committee (SPC) in the near future, e.g. suggest speakers, help chairing sessions, do web-related work and arrange workshops.

We are a strong society that should be very vocal when genetics is being misused, as on the Uyghur population in China (see political statement on our web site). Furthermore, ESHG should always strive to promote collaboration between professionals and societies, so that the fruits of the incredible amount of genetic knowledge can benefit everyone, especially the patients. It is our task to provide skilled guidance to politicians or other decision-makers about the implementation of precision medicine: this needs to be clinically relevant and not just an additional burden to our health care systems. Our responsibility is not the “worried well” but real patients and families with real genetic problems, and that is where our focus should be.

It has been an honour to serve as your President. As Vice President for the coming year I will follow up on the variant classification system, awaiting feedback from real life users, and our new educational plans. Have a great Gothenburg meeting!

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Report from the SPC Chair: Welcome to vibrant, sustainable, beautiful and fun Göteborg, home to the 52nd ESHG!

By Joris Veltman, Chair of the Scientific Programme Committee

Göteborg, or Gothenburg for those who prefer the English translated city name, also hosted the ESHG in 2010. No time to look back, however, the pace of change in genetics is continuing to impress us all, never a dull moment, no time to waste!

For the SPC this rapid change offers great opportunities to explore new areas in which genetics is being applied, invite upcoming experts, try out novel ways of exchanging knowledge and keep our participants up-to-date with developments in our field. Speaking of rapid developments, we reintroduced a late-breaking presentation session last year in Milan. While we received more than 80 abstracts we did notice that many of these were more ‘late’ than ‘breaking’ (no offense implied, my own work is often late and rarely breaking...). After discussions in our SPC meeting, we decided to organize this session again this year but schedule it in one of the concurrent slots on Tuesday morning and not during the opening event.

A rapidly changing field also requires an active involvement of young scientists in our meeting. We try to stimulate this in a number of ways. Well-known is the possibility for early-career scientists to win a best presentation award. Great to see that this year 45% (n=75!) of all abstract-selected presentations are given by Young Investigator Award candidates. In addition, winners of this award are asked to chair concurrent sessions in next year’s meeting as a way to promote their involvement and participation. Also, it is great to see the launch of an ESHG Young Ge-
Dear ESHG Members,

As a modern, open society and conference, we want to stimulate knowledge exchange and information sharing. We have therefore decided to change our social media policy to allow the use of photography in oral and poster sessions, unless indicated differently by the author on one or all of their slides or their poster. Also, please promote our meeting on social media using the #ESHG2019 hashtag and provide feedback on the meeting using the @ESHGsocitiy twitter account. We also continue to promote the use of digital media in the form of electronic posters, while at the same time reserving ample of time and space for traditional poster viewing and informal networking. Together with our conference organiser, the SPC is also trying to reduce the CO2 footprint of the conference by focusing, among others, on digital media and publications instead of print work. Both the organising office and the SPC now work almost paperless. This goes hand in hand with a responsible and sustainable use of resources and production of conference material. You will find a page in the conference programme (p. 66) and the report of the Executive Officer where we address the measures taken so far.

Finally, one thing you may have noticed is that we have now completely integrated the European Meeting on Psychosocial Aspects of Genetics in our annual ESHG program. Given the interest in psychosocial aspects of genetics, we decided to move from a biennial joint ESHG-EMPAG meeting with separate tracks to a single integrated programme.

On behalf of the Scientific Programme Committee I wish you all a most interesting and exciting ESHG 2019!

@jorisveltman

PS: We very much welcome suggestions and comments of all attendees. Not every suggestion can be acted on, but each one is welcomed and considered. Please send me or anybody else of the SPC suggestions at any time by email, or approach us at the ESHG. In addition, since 2015 we invite all ESHG members in May/June to propose speakers and topics online for next year’s meeting, including suggestions for new workshops and educational sessions. Please help us to make next year’s meeting in Berlin even better by making use of this possibility!

Report of the Executive Officer

Jerome del Picchia, Executive Officer of the ESHG, on behalf of the ESHG Organising Team

Dear ESHG Members,

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, I would like to inform you of the range of “green points” at our 2019 conference.

Venue and Location

As an organiser it is sometimes difficult to have others implement measures going beyond our influence, as venues will usually comply with local legislation, which varies very massively from country to country. While we try to make the venues understand that this is today an important issue not only for organisers but also for delegates, who will be noticing and pointing out areas where they feel that more can be done, it is often difficult to implement changes on a short term.

Therefore we are glad that 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. They also endeavour to supply their restaurants with organic, locally produced ingredients. On this basis they choose between otherwise equivalent products. Nowadays even their wine list 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 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.

Conference Bags: Since 2014 the ESHG Conference has aimed to provide congress bags made from durable materials, such as cotton. This is aimed at increasing the reusability 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 resources 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: The ESHG has been making hard efforts
to reduce its printed materials over time, while ensuring to keep high standards for its conference participants. We stopped printing the abstract book 8 years ago. This 500+ pages publication totalised around 5 tons of ink and paper each year. Instead we implemented the online programme planner as well as the conference app, which we continue to improve over the years, and have been adopted by our participants. The convenience in using these tools as preparation ahead of the meeting is a quasi standard today, however the fringe benefits of reducing the number of produced items seems less obvious, but definitely contributes to the whole.

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

Concerning printing of posters, the ESHG has slowly 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 GmbH 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. 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.

On behalf of my colleagues and the organising committees, I sincerely hope that you will enjoy this conference which we have tried to organise in the most responsible and sustainable way.

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**Report from the Public & Professional Policy Committee**

*By Francesca Forzano, Chair of the PPPC*  

The Public and Professional Policy Committee (PPPC) is an interdisciplinary Committee of the ESHG with the mission to contribute to the responsible translation of advancements in human genetics, by identifying and discussing the ethical, social and policy issues related to human genetics and its application in research, clinical practice and laboratory genetic services.

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**Personal Comment: Only correct DNA pictures please!**

*By Johan den Dunnen*

Every day I opened the 2018 ESHG app I got annoyed a bit by the first advertisement. Why? Well, the artistic DNA molecule shown turned in the wrong direction. I noticed the same problem in many presentations, ...scientists showing beautiful DNA pictures turning in the wrong direction, ...working in institutes with artistic DNA-based logos turning in the wrong direction. Making a tour through the exhibition area was even more shocking; 2/3 of booths had wrong DNA pictures, on their advertisement, in their logo, on their computer screens. DNA turning in the wrong direction was everywhere.

In my opinion an important tasks of ESHG is “Education”. Being an organisation of genetic experts we should try to get rid of wrong DNA pictures. It seems we need to educate the companies, the scientists, our own members, everybody. My request to the ESHG board to make “wrong DNA pictures” a topic for next year’s ESHG meeting was positively received.

For the 2019 meeting a topic will therefore be “only correct DNA pictures please”. Notes on the topic will be shared with all participants, presenters and sponsors, everybody.

I’ll keep my eyes open in Gothenburg; try to prevent I spot you with wrong DNA.
It addresses these issues and provide guidance through background documents, policy statements, recommendations or other publications, and participating in the public and professional debate around these issues.

The Committee collaborates with a range of other societies and organisations such as ASHG-Social Issues Committee, EMPAG and ESHRE. In 2018-2019 PPCP has been active on the following topics.

**Post-mortem genetic testing**
Dr Florence Fellmann has led a working group composed by PPCP members and experts from cardiology and forensic pathology who had the aim of drafting joint recommendations on post mortem genetic investigations in cases of sudden cardiac death. The recommendations include that medicolegal autopsies should have a dual aim: not only to establish if a death was natural or caused by a criminal act or accident; but also to establish the cause of a natural death, and allow results to be used for health care purposes for the surviving relatives. Sudden cardiac death at a young age should be considered a public health priority, public funding should be allocated for related relevant investigations.

The Board of ESHG has endorsed the recommendations during the meeting in the context of the ESHG conference in Milan in 2018; they are accepted for publication in the European Journal of Human Genetics.

The document has also been endorsed by European Council of Legal Medicine, European Society of Cardiology working group on myocardial and pericardial diseases, European Reference Network for rare, low prevalence and complex diseases of the heart (ERN GUARD-Heart), and Association for European Cardiovascular Pathology.

**Recontacting**
Recommendations on recontacting in clinical genetics have been drafted by PPCP and endorsed by the Board of ESHG during the Milan 2018 meeting. The recommendations have been published online on October 11 2018 in the European Journal of Human Genetics.

A few days later, members of the PPCP have participated to a session on Recontacting in Genetics at the American Society of Human Genetic (ASHG) 2018 Conference in San Diego. Purpose of the session was to compare recommendations on recontacting of various societies, namely the ESHG, the American College of Medical Genetics (ACMG) and the ASHG, from both the diagnostic and research perspective. The recommendations of these latter societies were published respectively on December 22nd 2018 in Genetics in medicine and on Apr 4th 2019 in the American Journal of Human Genetics. A lively debate with a very engaged audience ensued, which demonstrated the relevance of this topic for our communities.

The three societies' statements were all in agreement on the core messages: when new information which might have a significant impact on medical care becomes available, recontacting for updating patients is desirable, although there is currently no duty to do so. Recontacting should be a shared responsibility with the patients, the genetic laboratory and other healthcare professionals involved in the care of the patient. A discussion regarding the possibility of future recontact should occur at the time of consent for genetic testing and should be documented in the medical record.

**WMA Medical Ethics Conference and General Assembly October 2nd-4th 2018**
Some members of PPCP represented the ESHG at the World Medical Association (WMA) Medical Ethics Conference on October 2-4 2018 in Reykjavik, which preceded the WMA General Assembly.

The ESHG has been asked to provide an overview of the preeminent ethical issues our discipline is currently facing. The PPCP organized a session entitled 'Future Challenges in Clinical Genetics', encompassing the following topics: From single gene test to NGS, secondary and incidental findings, opportunistic screening (Angus Clarke); Newborn and population genetic screening (Heidi Howard); Data sharing, Biobanks and Direct to consumer testing (Emmanuelle Rial-Sebbag); Overview of Iceland's experience after data release from deCode (Vigdís Stefánssóttir); Somatic and germline gene editing (Heidi Howard).

PPCP has a standing interest in forensic uses of genetic testing. This year some PPCP members published a response to the forensic genetics policy initiative's report “Establishing Best Practice for Forensic DNA Databases” in Forensic Sciences International Epub July 5 2018. PPCP has contributed to draft ESHG statements regarding conformation of the collection of DNA from ordinary individuals in China as part of an oppressive programme of surveillance and control of the Muslim Uyghur population in Xinjiang, and on the support of a moratorium on germline gene editing.

Currently, PPCP is working on several documents, which aim to provide guidance to our members on the related issues, particularly on Opportunistic Screening, led by Guide de Wert, and Implications of the General Data Protection Regulation for Laboratories and Clinical Genetics practice, led by Emmanuelle Rial-Sebbag, both in collaboration with the Eurogentest Committee. For some of the earlier topics, PPCP members are involved in the implementation of next steps (e.g. germline gene editing, post mortem genetics).

ESHG members having suggestions for new topics to work on, or interested in working on a specific topic as a collaborator are encouraged to contact PPCP via the Chair.

ESHG members interested in becoming a PPCP member are requested to send their CV and a letter of interest stating their expertise and motivation for joining to the Chair.
Members of the PPPC in 2018-2019 were Angus Clarke, Christophe Cordier, Martina Cornel (co-Chair), Guido de Wert, Florence Fellmann, Francesca Forzano (Chair), Sabine Hentze, Heidi Howard, Hülya Kayserili, Béla Melegh, Alvaro Mendes, Markus Perola, Dragica Radojkovic, Emmanuel Rial-Sebbag, Vigdis Stefánsdottir and Carla van El (Secretary-general).

Report of the Education Committee (EduCom) 2018
By Han Brunner, Chair of the EduCom

The 2019 members of the ESHG Education Committee are Inga Prokopenko and Philippos Patсалis (Teach the teacher course), Ed Tobias (teaching materials website), Christophe Cordier (DNA day essay competition), Domenico Coviello (School children’s event), Johan den Dunnen (HGVS course organisation), and Han Brunner (Course portfolio, chair). The EduCom has delegated specific tasks to each of its members. It meets for a telephone conference every 3 months to discuss the progress and plans. Membership of the EduCom is for three years, with tasks shared and then passed on to a new member from the third year. There is the possibility of a 2-year extension.

Domenico Coviello will step down from the Education Committee after Gothenburg. He has been a very active member, and has been influential in setting up the School Children’s activity at the annual meeting from 2014 onwards.

School children’s event:
From 2014, ESHG organises, during its annual congress, a set of practical activities with high school students, which are delivered through the national scientific societies of the host countries. The first event was held in 2014, Milan, Italy, at the congress venue; the second - in 2015, Glasgow, UK, at Glasgow Science Center; in 2016, Barcelona, Spain at the Cosmo Caixa Science Museum, and in 2017, Copenhagen, Denmark, at the conference venue. In 2018, it was held in Milan organized by the Italian society SIGU. In Milan, there were 4 workshops for up to 30 students from Milan and other cities such as Genoa: Walking the chromosome, Healthy or affected, Frome protein to gene, the example of hemoglobinopathies, and DNA detective.

At the moment of writing, there is no clarity as to the organisation of a 2019 school children’s event in Gothenburg.

Teach the Teacher:
Philippos Patsalis and Inga Prokopenko have selected a set of presentations from the educational sessions of the ESHG, and from the European School in Bertinoro. They plan to make these available for attendance online on a specific day of the year, in a “teach the teacher” format, with a short set of questions at the end and a certificate for those who pass. It has proven difficult to attract attention from established online platforms for this. An online channel would be preferable so that teachers can watch content at their leisure. The possibility of using the EUROGEMS website for this will be explored for 2019.

Educational materials:
Prof Edward Tobias (of the University of Glasgow, Scotland, UK) has worked hard, on behalf of the ESHG, creating a website that now provides access to a large number and wide range of educational materials. This new educational website provides direct links to over 70 carefully selected, free, high-quality genetics and genomics educational sources around the world, each with a short summary of its uses and content. Separate detailed, illustrated, sections have been created and individually tailored to different target audiences: students, primary school teachers, secondary teachers, patients and relatives and to genetics professionals, with many resources described on each page. There is also an ethical, legal & social implications section. Ed plans to add a page for primary care in the future.

The linked resources include many useful sites for genetics professionals (e.g. gnomAD, LRG & LOVD databases and HGVS for nomenclature), articles, videos, animations, debates, free genomics-terminology & quiz smartphone apps (http://www.genomicsapps.org) and also many genetics activities for schools such as “DNA origami”, which Ed has actually fully tested, for “quality-control” purposes, of course!

The ESHG Board enthusiastically received and endorsed the website when Ed presented it in detail to them in Milan, June 2018, a link was created directly to it from the main ESHG website and Prof Gunnar Houge informed the National Committee presidents of it.

Since then, Ed has continued to update the links regularly and to add resources, including local initiatives in other languages and, for example, on request, VEP, Varsome and CaVaDa for variant interpretation. The EuroGEMS.org website has already been viewed 4400 times from 89 countries, with approximately 20% of visits from outside
Europe (including over 260 from North America) and an increasing number of visits over time. The most popular page (other than the Home page) is the one for Genetics Professionals, followed, in order, by the pages for Universities & Students; Patients & Families; Secondary Schools; Primary Schools; & Ethical, Legal & Social Implications.

Emailed comments from a range of professionals have included: “I was amazed!”; “Thank you once again for the excellent web resource overview”; “I think this is absolutely brilliant”; & “I am really impressed by your terrific work.” Anonymous written comments from Genomics Masters-level students have included: “The EuroGEMS website was particularly useful”; “The website was wonderful for accessing genetics databases and looking up relevant Genetic conditions”; “EuroGEMS is very useful to navigate through the different sources & databases out there”; & “Excellent. All links have been really useful.”

Ed at Edward.Tobias@glasgow.ac.uk would welcome any comments and also further suggestions for any additional online educational resource that is free-to-access, useful and of high quality, ideally with a note of the source’s target audience, general content and advantages.

DNA day essay contest:
This year’s DNA day question was ‘Why does gene editing work, and why are the expectations for this approach so high?’.

Students for the first time had the possibility to respond to it by a written essay (750 words) or by a video (3 minutes). The winners will be announced at the ESHG meeting in Gothenburg. The EduCom and the executive board will choose and draft an appealing question for next year. This is due by October.

Human Genome Variation Nomenclature course:
ESHG, in collaboration with the Human Variome Project (HVP), organized a first “HGVS nomenclature Teach-the-Teacher course on Friday June 15, 2018 in Milano. The course was supported by 7 fellowships from ESHG with 36 applications from 20 countries. 16 participants (15 countries) were selected with a focus on one participant per country. 7 participants received a fellowship to (partially) cover cost. Participants were educated in order to empower them to prepare and provide similar courses in their country. The course was recorded (sound only) as a basis for generating an e-course to be posted on the HGVS and ESHG web site.

ESHG course portfolio:
The courses that are organized as part of the ESHG course portfolio in 2018-2019 are all doing well. Several have been sold out, and all are well attended with good feedback from students. The ESHG fellowships allow a large number of students from less advantaged countries to participate which reflects the inclusive aims of the society. The current course portfolio is:

- Hereditary Cancer Genetics
- Dysmorphology
- Eye Genetics
- Statistical Genetics
- Clinical Cytogenetics
- Clinical laboratory genetics (beginner’s course)

- The ESHG cardiogenetics course 2019 in Antwerp had a record 61 participants from 18 different countries. The course has been very popular with cardiologists as well as geneticists. It fully reached its educational goals. There was a fantastic and enthusiastic group of participants who on the evaluation forms all loved the content and the format of the course.

- The 10th Dysmorphology in the Genomic Era Course was held in Manchester, from April 9th-11th 2019. The course had 50 delegates from 26 countries. Most were from Europe but there were also delegates from Saudi and Japan. Course format included traditional lectures, interactive sessions, workshops and quizzes. In addition, many participants gave excellent case presentations. New talks this year included skeletal abnormalities and bladder syndromes. All lecturers contributed their expertise for no charge and this year we expanded the external faculty to four (Willie Reardon - Dublin, Daniela Pilz - Glasgow, Abhijit Dixit - Nottingham and Charu Deshpande - London). NGS and large scale studies are influencing clinical practice and most lectures included these new diagnostic approaches and the all-important phenotyping to interpret results. Feedback was very good with some excellent suggestions for the future from participants, they particularly valued the opportunity to meet colleagues from all over the world and many felt that they would not have been able to attend without an ESHG scholarship. Course Organisers Dian Donnai, Jill Clayton-Smith, Siddharth Banka, and Sofia Douzgou hope to offer another course in Spring 2020 perhaps back-to-back with the ESHG Cardiogenetics course.

- The 32nd European School of Genomic Medicine and NGS was held in Bertinoro on 29 April to 4 May 2019. There were 83 students from many different countries and a similar number from remote training centers in Iran (Tehran
and Mashad), Malta, Romania, South Africa, and Turkey who followed the course via online streaming.

Professor Giovanni Romeo who started the first course in medical genetics in 1988 has decided to retire from his organizational involvement with the course after 31 years. He has been instrumental to promoting teaching in Medical and Human Genetics for many years. Giovanni Romeo has made seminal contributions to all aspects of the ESHG. These include not just the School which he founded and organized with great care for so many years in Sestri Levante, Ronzano (Bologna) and now Bertinoro, but also many other courses. An estimated 7000 students attended these various courses over the years. For the course on Genomic Medicine and NGS (formerly Medical Genetics) the number is close to 3000 students. A similar number of students have followed the courses online since 2000. For many of these students, the course has been an inspiration, as well as a fantastic way of connecting with the medical genetics community as a whole. Giovanni Romeo was instrumental in creating the European Journal of Human Genetics of which he was the founding editor, and his role in modernizing the ESHG organization and Society. Giovanni Romeo received the first European Society’s education award in 2005, and the Education award of the ASHG in 2011. For the 2019 course on Genomic Medicine and NGS in Bertinoro, Giovanni Romeo recorded a welcome video for the students. In addition, the first Romeo-McKusick lecture was included in the programme of the 2019 course to honour his dedication and involvement. The 2019 Romeo-McKusick lecture was delivered by professor Andrea Ballabio from TIGEM in Naples. Fittingly, professor Ballabio was also a student on the first course in Medical Genetics in 1988, in Sestri Levante.

There was strong interaction between students and faculty during lectures, workshops, and breaks. The evaluation of the 2018 course was excellent, with average appreciation exceeding 8/10 for almost all lectures and workshops. The topper of 2018 is “How are genetic test results being communicated” by Barlow-Stewart K. et al. EJHG 2019 Feb;27(2):308-316. doi: 10.1038/s41431-018-0279-z. Epub 2018 Oct 24. Altmetric score 76.

For 2020, the Education Committee is interested in attracting new ESHG board members to join the Education Committee: The Education Committee is interested in attracting new active EduCom members. Board members who have a good idea about something that they want to develop regarding education can apply.

**EJHG Highlights 2018**

*by Gertjan van Ommen, Editor in Chief, EJHG*

**Impact Factor**

In 2018, our impact factor (citations measured over 2016-2017) took a dent from 4.29 to 3.64. Our publisher considers this to be due to a statistical quirk due to a sudden surge of papers published in the preceding year to reduce publication backlog from 9 to 3 months. If so, this would be corrected this year, as we have published rather less papers in 2018 than in 2017, 218 vs 271, to remedy that we ‘ate through our available material’ a little too fast. But by far the most important event for our visibility is the appointment of a Social Media Editor, Alisdair McNeill from Sheffield, who has achieved a truly astonishing fact by getting ~2500 followers in the 5 months of his activity from December 2018-April 2019. Elsewhere in this Newsletter, he will introduce himself and highlight his advances more in detail. But let me also stress that the best way to increase our impact remains that our frequent authors submit some of their best research to EJHG.

**Reviewing and time to publication.**

In 2018 we received 704 submissions, against 727 last year; 3.2% less, continuing the small decrease in submissions ongoing for 5 years, probably due to increased competition. Our acceptance rate for 2018 has been 33% like in 2017. On the other hand, the average time to first decision after review, was substantially down to 55 days from the 63 of last year. Papers which are not reviewed are on average returned within 12 days, like last year. As to the review time, I explained last year that what is often – and understandably –perceived by authors as THE review time is actually a composite of the reviewer allocation - and acceptance! – time, and the actual review times. Indeed, actual review times have kept stable over the past years at 14-15 days, but it has become increasingly difficult to get 2-3 reviews in, sometimes requiring us to approach 6-10 reviewers! Using new procedures, online publishing time after acceptance is coming down to 29 days, from 36 days in 2017. The average time to print publication has crept up a bit, to 129 from 95 days in 2017, due to the above mentioned need to build up a bit more buffer.

**Web visibility**

Web page views in 2018 amounted up to 925,626 times. Theopper of 2018 is “How are genetic test results being used by Australian life insurers?” by Barlow-Stewart K. et al. EJHG 2018 Sep;26(9):1248-1256. doi: 10.1038/s41431-018-0198-z. Epub 2018 Jun 11. with 6587 views.

To document web visibility we are monitoring the interest by the Altmetrics score. This is a qualitative and quantitative metric complementary to traditional, citation-based metrics, including (but not limited to) peer reviews on Faculty of 1000, citations on Wikipedia and in public policy documents, discussions on research blogs, mainstream media coverage, bookmarks on reference managers like Mendeley, and mentions on social networks such as Twitter and Facebook. The three Altmetric top scoring papers are:


In 2017, we upgraded the design of the EJHG web page. This is now completed: after logging in at www.nature.com/EJHG, you will find several new links relaying you to ‘Featured articles of the most recent issue’, to the ‘Editor’s choice’ from recent key papers, freely accessible available for one month, to a third page with the ‘Best of EJHG 2017-18’ containing a whole slew of highly downloaded and cited papers from the last two years, and finally a link to our recent Twitter activities.

**EJHG Award**

As every year, EJHG, and Springer/Nature jointly offer a junior authors’ high-citation award. This is given to the top-3 articles published in 2017, with citations counted in the 12 months following print publication. The 1st prize includes a € 500 award and positions 1-3 receive one year free ESHG membership, including an online EJHG subscription, and free registration for that year’s or next year’s meeting. The winners this year, to be honoured in the closing ceremony, are:

1. Dr. D. Trujillano, for the paper titled “Clinical exome sequencing - results from 2,819 samples reflecting 1,000 families”, EJHG 2017 Feb;25(2):176-182. doi: 10.1038/ejhg.2016.146. (Epub 2016 Nov 16), receiving 17 citations.


**EJHG-tube to move to on YouTube**

In 2016 EJHG and Springer/Nature implemented as innovation that authors could send in video abstracts as part of their final submission (or following acceptance). These video summaries were seen as a unique way for authors to present the information in their paper without the constraints of the written word, further enhancing the visibility of their work. However, while we expected high interest the uptake of this has been very modest. Perhaps the outcome was a bit too cumbersome and we are now migrating this to a YouTube page. More about this will be given as instruction to authors when they receive confirmation of acceptance of their manuscript.

Alisdair McNeill becomes EJHG’s first Social Media Editor

I am a Senior Research Fellow & Consultant in Clinical Genetics at the University of Sheffield/Sheffield Children’s Hospital. I was delighted to be appointed as EJHG’s social media editor in December 2018. Since then I have worked alongside the EJHG editorial team to develop and implement a social media strategy for EJHG. EJHG’s social media strategy aims to increase the visibility of papers published in EJHG and stimulate the engagement of the global genomics community with the journal. To achieve this, we have established EJHG Twitter and Facebook profiles. Via these platforms, I post a short summary of each and every paper published in EJHG. First when they are published online and then when they appear in a journal issue.

I believe the key to tweeting about scientific research is to identify the key finding and distil it into 1 or 2 sentences, in order to draw the reader in, by highlighting what unique findings can be learned from the paper. Sometimes this is easy: “Mutations in gene X cause novel syndrome Y”. More often a paper is too complex for this. One approach to these papers is to present the problem (e.g. how do you diagnose condition X if no mutation is found?) and then present a “teaser” of the solution (e.g. the authors used technique Y, read more to find out how”). Sometimes it is tempting to draw in readers with a controversial statement on the paper’s subject matter, for example: “think linkage analysis has had its day? Think again! This paper shows XXX”, or use the newspaper headline approach (“New type of dementia identified”). Since December 2018 we have gained almost 2 500 Twitter followers and this continues to grow at a rate of around 200 per month. This is very encouraging and indicates the strength of the EJHG brand.

In 2019, we will further develop our social media presence. On Twitter we will host monthly journal clubs to discuss an “Editor’s choice” paper. These will enable us to post a series of tweets to explore the paper in more detail and invite questions and comments from the community. We are also going to revamp EJHG tube by asking one of the authors of the monthly journal club paper to make a short video to promote their paper. To add a more rounded feel to our social media content we will live tweet from major medical conferences. We are also going to develop a ‘Featured articles of the most recent issue’ , to the ‘Editor’s choice’ from recent key papers, freely accessible available for one month, to a third page with the ‘Best of EJHG 2017-18’ containing a whole slew of highly downloaded and cited papers from the last two years, and finally a link to our recent Twitter activities.

So why should busy clinicians and academics make time to engage with our social media content? Well, there is ample evidence that use of social media is both a time and cost effective way to promote one’s scientific outputs. For example, the altmetric score, a composite measure of social media interest in a scientific paper, is used by many Universities as a measure of research impact. There is some evidence that high altmetric scores correlate with increased citation rates. Engaging with EJHG social media will also
provide a succinct summary of the latest research in EJHG, helping to guide your reading for Continuing Professional Development (CPD) purposes.

We hope to make EJHG’s social media offering dynamic and interactive. We are open to your suggestions on how to improve, and would particularly welcome your participation in our Twitter journal clubs.

**Annual report ESHG-Eurogentest Committee 2018-19**

*By Hans Scheffer, Chair of EUGT*

The ESHG-Eurogentest committee (EUGT) is the committee dealing with a variety of aspects related to the improvement of quality and service of diagnostic laboratories and clinical services in genetics. Apart from own initiatives and activities EUGT tries to interact with the many international stakeholders in this field, and assist with the implementation of novel applications.

In the past EUGT has been a EU-financed (FP6 & 7) project for many years, initiated by Prof. Jean-Jacques Cassiman and continued by Prof. Gert Matthijs. EUGT continues to be a strong brand name. During the last three years EUGT has been integrating its activities with other ESHG activities, e.g. in collaboration with the ESHG Public and Professional Policy Committee (PPPC).

The main aims of the EUGT steering committee have been (1) to make activities coherent and (2) self-sustainable. With regards to aim (1) the different subcommittees now interact with several stakeholders within and outside the ESHG.

EUGT has now three subcommittees:

(1) The Quality subcommittee chaired by Sandi Deans
(2) The Guidelines subcommittee chaired by Gert Matthijs
(3) The Training subcommittee chaired by Els Dequeker

**Quality**

The objectives of the Quality subcommittee are to:

- Promote harmonisation with EQA providers to reduce poor performance in genetics;
- Review management and governance of EQA providers;
- Establish educational EQAs for genetic counselling;
- Harmonize Quality Management (QM) and service provision within genetics;
- Address quality issues in new-born screening programs;
- Explore quality issues relating to International databases of genetic variants

Els Dequeker of the University of Leuven (Belgium) represents ESHG-EUGT as an observer in the working group of Health Care of European cooperation of Accreditation (EA).

Els Dequeker now also represents ESHG-EUGT on medical device regulation (MDR) issues and represented ESHG-EUGT at a recent meeting at the Joint Research Center (JRC) in Ispra, Italy. One of the conclusions was that expert groups need to be set up for medical devices as well as for IVD kits. Together with representatives of other laboratory disciplines a joint expert group on IVDs will be assembled. Since all genetic tests now are in class C, this is only necessary for future tests that will be classified as class D.

To promote further dissemination of professional expertise across Europe, EUGT closely collaborates with the European Board of Medical Genetics (EBMG).

**Guidelines**

Priority by the Guidelines subcommittee will be given to the development of novel guidelines/recommendations for whole genome sequencing (WGS) in diagnostics. This initiative is undertaken as part of the Horizon2020 Solve-RD project, in close collaboration with other stakeholders including European Reference Networks (ERNs).

Recently a start has been made and a first expert meeting has been held to determine the scope and to prioritize the most urgent issues to be covered in the new diagnostic WGS guidelines/recommendations. These include diagnostic strategy, validation of new platforms and approaches, reporting and bioinformatics. A second expert meeting with participation of in principle all ERNs will be scheduled in autumn. The expert meeting was held in conjunction with a task force organized by Gunnar Houge to develop an improved system on the interpretation of variants of unknown significance (VOUS), in particular to warrant a better match between genome results and clinical phenotype. These two initiatives will result in separate but coherent recommendations.

For policy documents EUGT closely collaborates with the PPCP, e.g. on opportunistic screening.

**Training**

The Training subcommittee initiated plans to further develop training courses and (online) training tools e.g. on quality management aspects in general and on data interpretation using bioinformatic tools. The possibility to join forces with the ESHG Educational committee will be further explored.
Professional disciplines naturally collaborate in daily life and Genetics Technicians (LGTs) as the fourth branch. All four clinicians working in human genetics diagnostics: Laboratory and Genomics (MGG) (representing medical geneticists); GNGCs and Genetic Counsellors (GNGC) and Medical Genetics boards, Clinical Laboratory Geneticists (CLG), Genetic Nurses: After four years Hans Scheffer will stop as steering committee chair. The steering committee has unanimously proposed Gert Matthijs as new steering committee chair. Gert has indicated to be available for two years. For reasons of continuity deputy chairs for the steering committee and all subcommittees will also be installed, as far as that hasn’t happened yet.

We would like to invite enthusiastic colleagues to join the Eurogentest team, and help to stimulate and promote improved quality and service of diagnostic laboratories and counselling units in Clinical/Medical Genetics across Europe. Active recruitment has recently been started. A more extended overview of Eurogentest activities as well as an informal “job description” and contact information are available via the ESHG and Eurogentest websites.

For further information including ongoing activities and changes in the subcommittees we refer to the ESHG and UEGT websites www.eshg.org and www.eurogentest.org.

The UEGT steering committee: Els Dequeker, Sandi Deans, Luca Lovrecic, Holm Graessner, Milan Macek, Hans Scheffer.

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The European Board of Medical Genetics (EBMG), established in 2013 to set professional standards for health professionals working in clinical genetics field in Europe, has as main goal to provide systems to enable professionals to demonstrate their professional competence. From 2014 on the EBMG is an autonomous legal entity with a General Assembly held each year, held during the ESHG Annual Scientific meeting.

The EBMG comprises yet three distinct professional Branch boards, Clinical Laboratory Geneticists (CLG), Genetic Nurses and Genetic Counsellors (GNGC) and Medical Genetics and Genomics (MGG) (representing medical geneticists); there are also efforts to set up a professional branch for technicians working in human genetics diagnostics: Laboratory Genetics Technicians (LGTs) as the fourth branch. All four professional disciplines naturally collaborate in daily life and also coordination across the Branches is well established, even though each Branch has a primary responsibility to serve their own.

In 2018/19, the successful work of the previous years was continued and extended. GNGCs and CLGs have now successfully established the online application system for those wishing to join the EBMG register as initial as well as renewal applicants. This could only be done by the support of the Vienna Medical Academy and namely Jerome del Picchia and Stefan Födermayr: The MGGs works closely together with the UEMS and started a registration system for MDs being specialists in Human Genetics.

All details of the EBMG Branch members and the registration systems are available on the website [https://www.ebmg.eu/413.0.html]. The reports of the three Branch boards are as follows:

**Clinical Laboratory Geneticist Branch Chair: Dr Thomas Liehr**

In 2019 we have 424 registered ErCLGs. In 2018/19 we had 41 applications for registration with the EBMG. 7/41 were unfortunately found to be not eligible, 12 of the remainder 36 candidates were registered already, due to using group 1 or 2 way of application. The 24 further candidates will have to undergo a written and oral test held in Gothenburg, Salzburg, Yerevan, Armenia, Jena, and/or Coimbra. The branch introduced a slightly altered test scheme, including more multiple choice questions than before and an oral exam oriented more towards case scenarios to be discussed. A paper on the rights and duties of CLGs in 50 different countries is in press in EJHG. The data can be enlarged for further countries on https://www.ebmg.eu/910.0.html by sending a notice to Thomas.Liehr@med.uni-jena.de. Also the CLG profession is now state recognized in Finland, France, Greece, Italy, Hungary, Latvia, Northern-Macedonia, Poland, Portugal, Slovenia and UK. This the CLG-branch is contacting different national governments to ask their support in proceeding a request to European Union that CLG may become EU-recognized profession based on approved EU Directive 2005/36/EC - policy developments and Proposal for modernizing the Professional Qualifications Directive - EU Directive 2013/55/EU. The CLG branch has also contributed to planning of courses to support CLG education prior to registration. Further details on the next course are available at: https://www.uniklinikum-jena.de/humangenetik_media/Dokumente/Arbeitsbereiche/fish/Kurse/Announcement.pdf.

Please note that the applicants for 2019-2020 have to submit their data for round 1 on the website between July 15th – October 15th of this year.

**Laboratory Genetics Technicians Branch Chair: Peter Böhm - Denmark**

The LGT-branch started working with support and on initiative of Thomas Liehr. Our first goal is to recruit technicians over Europe who would be willing to actively work in this new branch. Besides, we look forward to identify contacts in each European country, to find out about existing national education schemes concerning human genetics oriented LGTs. Our long term goal is to establish a consented European core cur-

**Report from the European Board of Medical Genetics**

*by Thomas Liehr, Chair of the EBMG*

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riculum for LGT education and if possible a certification system within the frame of the EBMG.

**Medical Genetics and Genomics Branch Chair: Professor Bela Melegh**

During the last year the Branch of Medical Genetics and Genomics (BMGG) again passed a milestone as we took the first step to launch a Continuing Medical Education (CME) registry system, which will be recommended for active practitioners. The draft has been revised so far by our sister EBMG Branches, by the ESHG Exec, and is now under consideration of the UEMS Exec. We hope to be able to launch the EBMG BMGG CME Registry system at our meeting during the ESHG meeting in Gothenburg.

Parallel, after a trial exam in Milan in 2018, the UEMS Section of Medical Genetics scheduled the inaugural, first exam of European Diploma in Medical Genetics and Genomics (EDMGG) in Gothenburg, 14th June 2019. This is a joint project with the ESHG and the EBMGG. This is intended to be a knowledge-based excellence assessment tool for medical geneticists primarily in Europe, although it will be open to candidates worldwide. The EDMGG diploma title and degree will be valid for lifetime. Beside these priorities, we contributed to the Joint Action on Rare Cancers (JARC) WP8 package, the Medical Education.

**Genetic Nurse and Genetic Counsellor Branch Chair: Christophe Cordier**

The Genetic Nurses and Genetic Counsellors Professional Branch had had a very busy year of activities, mainly because of the success of the European certification. We are proud to announce a new paper published in the European Journal of Human Genetic entitled «The recognition of the profession of Genetic Counsellors in Europe» with the full support of the Executive of the ESHG.

There are several updates for the GC&GN registration process.

1. The Grandfather routes are now closed in some countries but for applicants who work in a country without an approved Master course the grandfather clause will continue until October 2020. Please feel free to visit our website: https://www.ebmg.eu/408.0.html.

2. The UK, American, Australian, Canadian and South African Boards already have reciprocity agreements with our European registration system, so that professionals already certificated by those systems and already working for a minimum of one year in Europe can apply for our registration system under the National route with reduced portfolio.

In October 2018 the registration cycle was run for a fifth year. We received a total of 21 notifications of intention to register. This year applicants applied using the National route (n=2), the Normal Route (n=7), the grandfather clause A (n=3), the grandfather clause B (n=2) and the grandfather clause C (n=7). 16 applicants were eligible to submit their full portfolio. The final decision will be announced to the applicant before the ESHG meeting in Gothenburg (Sweden) where we will give the official diplomas.

In September 2019, Dr. Christophe Cordier (Switzerland) will be the past-chair, Deborah Lambert (Ireland) will be the Chair, and Clara Serra (Spain) will be the new chair-elect of the Genetic Nurses and Genetic Counsellors professional branch. Please note that the applicants for 2019-2020 have to submit their intention on the website between July 15th – September 30th of this year.

We would like to thank all the registered GC & GN for supporting the process of professional regulation of genetic counselling in Europe.

More news and updates can be found in our website: https://www.ebmg.eu/408.0.html.

**Conclusion**

According to the statutes I hand over the chair to Ulf Kristoffersson at this year ESHG meeting during general assembly of the EBMG by thanking the members of each Branch for their goodwill and hard work, which has ensured the success of the EBMG.

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**Dear member of the ESHG!**

We are pleased to invite you to the **Annual Membership Meeting 2019**

**at the**

**EUROPEAN HUMAN GENETICS CONFERENCE 2019**

on Sunday, June 16, 2019 from 19.30 – 20.30 hrs

ESHG Conference venue: Swedish Exhibition and Conference Centre Gothenburg Room H2

**AGENDA**

1. Opening by the President of the Society, Gunnar Houge
2. Activity of the Society 2018-2019
4. Presentation of an improved genetic variant classification system, developed by an ESHG task force
5. Discharge of the Board Members for the year 2018-2019
6. Opening by the new President of the Society, Alexandre Reymond
7. Results of election for President-Elect
8. Results Board Member election
9. Progress report transfer seat of the association to Vienna, Austria
10. Site of future European Human Genetics Conferences
11. Major policy questions proposed by Board
12. Future activities

Please find the minutes of the last membership meeting in Milan 2018 in the restricted area: https://www.eshg.org/members.0.html