THE EUROPEAN SOCIETY OF HUMAN GENETICS

NEWSLETTER

No. 32 - June 2018

Presidential Address

by Christine Patch, President of the ESHG

Dear colleagues,

The year of the ESHG runs between the two conferences so what has happened since the very successful celebration of the 50th anniversary of the inception of the Society in Copenhagen.

The extra business of the exec this year has been considering some issues concerning the reorganisation of the Society. As you will be aware at European level there is increasing scrutiny of financial affairs. For a variety of historical reasons the European Society of Human Genetics has a complex structure with the administration and various financial accounts being held in Vienna, Belgium and the Netherlands. The time has come to try and simplify this complexity in order to ensure the necessary compliance with various regulations across Europe. More details about the proposals are provided in the newsletter and will be discussed at the membership meeting.

After the first fifty years this will ensure the Society is in a good position to go forward in the next 50 years bringing together the fast evolving science of human genetics with the science and art of application for patient, population and society benefit.

The main annual business of the Society is the annual conference and this conference goes from strength to strength; growing, but still providing a forum for inter-disciplinarity, mutual communication and hopefully an enjoyable time. This year the conference is jointly with the European Meeting on the Psychosocial Aspect of Genetics which adds another strand to the science presented and the potential conversations that may occur

The work of the Society does between the conferences is of course dependent on the various committees and members of the Society who contribute their expertise to the committees but also those members who contribute to the committee's outputs such as educational courses, policy statements, scientific review to name a few areas. You do this voluntarily and participate actively without you the Society would not be able to function in a way that facilitates its involvement in important areas of science and issues of policy development. So thank you.

I hesitate to mention General Data Protection Regulation (GDPR). It is impacting across all our lives and this has relevance to the organisation of the Society and how your data is processed. It is still not entirely clear how this regulation will impact on the collaborative research endeavour necessary to ensure that the knowledge being generated in the field of human genetics is best used for the benefit of patients, population and society but also that due respect is given to protecting their data and ensuring it is only accessed and used within the appropriate regulatory environment. Of course GDPR is only one regulatory framework for one specific purpose, but it is an example of the challenges in this field.

As the ambitions of the initial large scale sequencing projects start to be realised in the next year and more initiatives are planned, I suspect that conversations which might include issues of; confidentiality, privacy, facilitating knowledge generation, implementation of this knowledge in health systems will become more necessary. This Society because of its scientific expertise and integration with the clinicians from many specialities and backgrounds will have an important role in developing some of these conversations.

During the meeting in Milan I hand over the presidency of the society to Gunnar Houge who with his experience of the organisation of the Society and also his personal qualities is the right person to take the Society forward in these times of change. It was a huge honour to be the first genetic counsellor (and nurse) to be President of this society. It has been enlightening, interesting, enjoyable but most of all a privilege to have played a small part in maintaining the continuity of this important Society. I hope many of you are able to come to Milan as we start planning for the next 50 years.

Christine Patch

ESHG Newsletter No. 32, June 2018
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Welcome to the 51th ESHG in Milan, where genetics mixes smoothly with fashion and football!
Report from the SPC Chair

By Joris Veltman, Chair of the Scientific Programme Committee

Great to be back in Milan, where we last met in 2014 and enjoyed a wonderful conference and left with an appetite for more. On our return we serve you an exciting conference menu, with for early starters on Saturday morning at 8 am an educational Next Generation Sequencing session, and for dessert on Tuesday afternoon the Mendel lecture and ESHG award lecture, this year given by Emmanuelle Charpentier and Matthew Hurles, respectively.

In between, there is a taste for everyone, with 93 hand-picked (invited) and 134 abstract-selected speakers, 1000s of colleagues presenting their work via traditional as well as e-posters, and 100s of life sciences companies showing you their latest assays, lab equipment or software solution.

As always, our programme has a firm focus on education and training, by providing educational sessions which will both introduce and update you, and interactive workshops for you to discuss hot topics in our ever expanding field.

The Scientific Programme Committee is always on the lookout for new items to add to the ESHG conference menu, and improve on your conference experience. This year, all poster presenters can upload their poster as an electronic poster and we scheduled sufficient time for both traditional and e-poster viewing. Also, we decided to add a late-breaking plenary abstract session on Saturday afternoon, which at the time of writing we just opened for submission, so let's see if this brings more exciting and novel work to our meeting. Finally, we will have our first ESHG quiz, which hopefully will turn out to be both entertaining and educative. We hope that this offers you with sufficient opportunities to network, educate and present yourself. Enough food for thought, enjoy!

On behalf of the Scientific Programme Committee I wish you all a most interesting and exciting ESHG 2018!

Joris Veltman
Chair Scientific Programme Committee ESHG

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 Scientific Programme Committee suggestions at any time by email, or approach us at the ESHG. In addition, since 2015 we invite our 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 Gothenburg even better by making use of this possibility!

See www.eshg.org/proposals2019.0.html

The SPC during its meeting in the Offices of the Vienna Medical Academy in March.

Same but different. Report of the Executive Officer

Jerome del Picchia, Executive Officer of the ESHG

Dear ESHG Members,

On behalf of the Executive Board of the European Society of Human Genetics, it is my pleasure to inform you about important plans for the future of the ESHG. These plans have been presented to the Board and the Members at the Membership Meeting in 2017 in Copenhagen, namely to:

- **move the ESHG from Belgium to Austria**
- **dissolve the foundations in the Netherlands**

The Membership has entrusted the Executive to explore the possibilities and to develop a plan accordingly. Subsequently the Executive has met with tax consultants from Belgium, Austria and the Netherlands and has developed a roadmap.

Hopefully we answer the main questions you may have below;

Why should ESHG move?

Currently the ESHG is split over 4 countries. The association is registered in Belgium, the Foundations were established in the Netherlands, the Administration Office and the professional conference organiser are in Austria and the conference is held in another European country each year.

The administration currently has to review 4 different tax and association legislations. Four different tax advisors are taking care of ESHG matters. It would make sense to have all arms of ESHG combined in one location with one tax and legislation system, to have registration, funds and administration in one country, and the conference in a second one. This would also save consultancy costs over time.
The administration office, although well versed in international matters, is of course much more "on top" of the latest evolution of regulations in its own country, hence Austria is the obvious choice. Austria also has a very welcoming attitude towards non-for profit associations, both in legal and tax matters.

How will ESHG move?

Unfortunately moving an association is not as easy as moving a company. The European association laws are not yet harmonised and are still a national matter. So just because an association is recognised as "non-for profit" in one country, it does not automatically mean that it will be in another.
Hence, according to all experts, it will be necessary to dissolve ESHG in Belgium and to create a "new" ESHG in Austria.
The changes for the members will be minimal (only of formal nature). The administrative office remains the same, the statutes will only be changed where Austrian legislation formally requires specific additions, and where the board believes that the old statutes could need some reshaping to make them more suitable for the current times and easier to work with on a daily basis.

Why do we have to change the current statutes?

There are three main reasons:

- The actual dissolution of the association is realistically impossible, as the voting requirements (Art. 19 of the current statutes) cannot be achieved successfully. So this paragraph needs to be amended in a way that makes dissolution democratically achievable.
- If the association was dissolved then all funds would have to be donated to the International Red Cross (also Art. 19). So here the statutes would need to be changed to "an association with a similar goal", so that the new ESHG could eventually retrieve "its own" funds.
- The current statutes lack some essential information on the officers and the official representation of ESHG in any legal business (e.g. when trying to open a back account). Times have changed and so have the laws e.g. on money laundering. Banks do require specific, officially published details on the officers.

Why should ESHG create a Limited Company?

A subsequent plan is then to create "ESHG Ltd" as registered tax-liable limited company in Austria, which would be the official organising entity of the ESHG conferences. As you know, the ESHG conferences are VAT-registered. As mentioned in the second paragraph, this became necessary, as a European country will not necessarily recognise the non-for profit status of an association registered in another country and it has become a more and more complex task to clarify the situation in a different country every year.

By having the conference organised by a VAT registered entity, things are easier, as the VAT-rules are widely harmonised across the EU and very clear.
Also the conference is now able to reclaim the VAT on the expenses, which (although this is obviously not planned) would decrease the volume of a possible loss in the future.
The "limited liability" of this entity would also mean that in case of a disaster with the conference, the funds of the association would not be drained (similar to the current model with the foundations).
This Limited Company would then be liable for corporate tax (just as the Foundations are already now).
The example of a number of other large European Scientific Societies having gone in this direction in recent years, show that this model will "water proof" the tax status of

the ESHG conferences in any country.
Obviously this would by no means make a commercial enterprise out of the ESHG conferences. The limited company would simply be the tax-vehicle to keep the meetings unchallengeable tax-wise, irrespective of local non-profit laws.

What are the next steps?

The next steps are as follows:

1. Vote on changes of current statutes (proposal sent by separate mail) at the Board Meeting in Milan, at the Membership Meeting in Milan, and in a subsequent online vote.
2. In parallel, the Executive is currently drafting the statutes of ESHG "new" and will propose them to the Board, as well as to the Austrian tax authorities for endorsement. Once endorsed, ESHG will be created in Austria. Membership enrolment can hopefully start in 2019.
3. The executive will clarify with the help of the tax experts the most effective way to transfer the current funds from both association and foundations to the new association. It is not yet entirely confirmed, whether taxes would be due or not. It will be the goal to perform this as cost-effectively as possible. It goes without saying that the ESHG will only proceed in a manner that will be 100% safe and legal.
4. Once this is clarified, the dissolution and liquidation of ESHG "old" can proceed.

So please attend the ESHG Membership meeting on Sunday, June 17 at 19:30 hrs in Room Yellow 1+2 of the MiCo in Milan to learn more. We will be happy to answer your questions.

Jerome del Picchia
Executive Officer of the ESHG

EJHG Tube

European Journal of Human Genetics invites you to include a video presentation with your submission to the journal as part of our new initiative **EJHG-Tube**. The video presentation should be included as supplementary material and is a unique way for authors to present the information in their paper and further enhance the visibility of their work by sharing on social media. Through this video authors can convey their findings without the constraints of the written word, plus provide a new and enhanced user experience for readers of the journal.

We accept the following files: .mov, .mpg, .mp3 and mp4. Please see EJHG-Tube at http://www.nature.com/ejhg/videos for our current video presentations and also refer to the journal's Guide to Authors for details on how to submit yours.

Report from the Public and Professional Policy Committee 2017-2018

By Martina Cornel, Chair of the PPPC

Mission

The Public and Professional Policy Committee (PPPC) of ESHG aims to identify and discuss the ethical, social and policy issues related to human genetics and its application in research, clinical practice and laboratory genetic services. It develops guidance through background documents, policy statements, recommendations or other publications to inform, interact with and provide advice to national and international policy makers. The development of the PPPC was recently described here: www.nature.com/articles/ejhg2017160

Germline gene-editing

Now that in the laboratory it appears to be easy, safe, cheap and effective to modify a gene during life or even around the time of conception, the debate on why and when this would (not) be allowed is needed in ESHG but especially also beyond our genetics society. We hope that the work of PPPC will contribute to this debate. In the January 2018 issue of EJHG an agenda setting paper was published listing points to consider for a responsible way forward for gene editing in humans (www.ncbi.nlm.nih.gov/pubmed/29192152). In the April 2018 issue of EJHG recommendations (www.ncbi.nlm.nih.gov/pubmed/29326428) and a background document (www.ncbi.nlm.nih.gov/pubmed/29326429) on germline gene editing, developed together with the European Society of Human Reproduction and Embryology (ESHRE) were officially published.

In many countries germline interventions have been prohibited, sometimes even accompanied by criminal sanctions. What were the arguments behind this legislation, and do these still apply and are they still considered convincing? If a technique can help to avoid serious genetic disorders, in a safe and effective way, would this be a reason to reconsider earlier standpoints? The Clinical Trials Regulation EU No 536/2014 uses the terminology "the subject's germ line genetic identity" in article 90. Would gene therapy implying repair of a mutation change identity? Are the fears related to risks for future generations?

We hope that members of ESHG and national human genetics societies will engage in debate with other stakeholders.

Post-mortem genetic testing

If a relatively young person dies suddenly, post mortem investigations are required in many countries to exclude a crime. In this context, or in the context of a subsequent, often much delayed consultation with relatives the possibility of a genetic disorder that might happen again in the family, especially a cardiomyopathy or an arrhythmia syndrome, may be raised. A DNA test can only be performed on the deceased index case if someone adequately stored a tissue sample for (future) genetic investigations. Current protocols for autopsy today often poorly integrate post-mortem genetic testing. A working group of PPPC led by Dr Florence Fellmann together with experts from cardiology and forensic pathology developed draft recommendations that were posted online for consultation of the ESHG membership and other experts in April 2018. 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 is invited to endorse the recommendations during the Milan meeting. The document is also under consideration for possible endorsement by other scientific societies.

Recontacting

Advances in genomic technologies have led to a reduction in cost, increase in speed of sequencing and increased number of more loosely targeted genetic tests, generating unprecedented amounts of genomic data. Often these genomic data will be stored and in theory they are available to be referred to in the future. A situation often encountered today is that new information about variant-trait associations is discovered, which leads to a reclassification of the variant in either direction (benign or pathogenic).
Is there a responsibility to revisit the genomic data over time? And if yes, who should initiate it? Who should 'recontact' the patient and family if there are new implications of clinical importance? Should this be the patient's clinical care team or the clinical genetics team (who may have had no contact with the patient)? Should it be the genetic diagnostics laboratory, in the case of updates of previous genetic reports? What role do patients play?
These and similar questions were investigated by teams of researchers from a UK consortium including Exeter, Cardiff , Southampton (Daniele Carrieri amongst others) and Groningen (Irene van Langen a.o.) in the Netherlands. Building on their empirical work, and with their collaboration, recommendations on recontacting in clinical genetics have been developed and have been online for ESHG membership consultation in April 2018, and the Board of ESHG will be invited to endorse them in June 2018.
In ethical terms, we all feel a moral obligation to recontact, but due to limited resources we are often or almost always unable to do it for everyone for whom we might have relevant news.
The recommendations highlight a potential role for various stakeholders, including the healthcare professionals with or without genetic background, scientists and patients.
Suitable informatics tools might provide useful platforms to track relevant situations and facilitate the recontacting procedures, but this requires allocation of dedicated resources to build solutions to be integrated into the Electronic Clinical Records and the databases of diagnostic genetics laboratories. Consent

procedures should also progressively integrate the possibility of future contact. Considering the limited budgets for genetics departments in many countries at present, in practice one might "Do the best you can with limited resources".

Members of the PPPC in 2017-2018 were Angus Clarke, Christophe Cordier, Martina Cornel (Chair), Guido de Wert, Florence Fellmann, Francesca Forzano (co-Chair), Heidi Howard, Hülya Kayserili, Béla Melegh, Alvaro Mendes, Markus Perola, Dragica Radojkovic, Emmanuelle 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 2018 members of the ESHG Education Committee are Inga Prokopenko and Philippos Patsalis (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 organization), 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. Progress on each of the tasks is as follows:

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 hoist 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, the ESHG conference will be in Milan again, and the Italian society SIGU will organize this activity in collaboration with ESHG. Domenico Coviello, and his colleagues from Italy have prepared 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
- DNA detective

Domenico will also liaise with the Swedes about the organisation of the 2019 school day event in Gothenburg.

Teach the Teacher

Philippos Patsalis and Inga Prokopenko have selected a set of presentations from the educational sessions of the ECHG, and from the European School in Bertinoro, that will be 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. They are currently working out the best online platform for this. An online channel would be preferable so that teachers can watch content at their leisure.

Educational materials

Ed Tobias has worked hard on a website that contains a large amount and diversity of teaching materials organized by topic and content. It is hoped to make the website available to the community in the near future.

DNA day essay contest

This year's DNA day question was "Comment critically on the proper role (if any) of genetic testing in sport". Students for the first time had the possibility to respond to it by a written essay (750 words) or by a video (3 minutes). There were 126 entries (122 essays and 4 videos) from 18 countries. Most students are from Turkey (35), Italy (24), France (12) and Georgia (12). The winner will be announced at the end of the month of May and at the ESHG meeting in Milano. These numbers are lower than previous years. Christophe Cordier will ask the national societies for help to disseminate this DNAday essay contest in the various countries. 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), organizes a first "HGVS nomenclature Teach-the-Teacher course". Its aim is to train people from a variety of European (and other) countries on HGV nomenclature such that they will then disseminate this knowledge to others in their respective countries. The course is coordinated by Johan den Dunnen, it will be held on Friday June 15 in Milano, and is supported by fellowships from ESHG. Since limited places are available, interested participants had to apply. The course attracted 33 applications from 20 countries; Australia, Egypt, Germany, Hong Kong, Hungary, India, Italy, Malaysia, Moldova, Morocco, Norway, Pakistan, Poland, Portugal, Republic of Macedonia, Romania, Saudi Arabia, Serbia, United Kingdom, United States. Fellowships were available to support some participants to join the course. The entire course will be recorded (sound) and used to develop educational tools on HGVS nomenclature, including an e-learning module. Whether further such courses will be offered in the future depends on the experience with this year's event, and prospects of the "teach the teacher" format.

ESHG course portfolio

The courses that are organized as part of the ESHG course portfolio in 2017-2018 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:

- Clinical Genomics and NGS
- Genetic Counseling
- Cardiac Genetics
- Hereditary Cancer Genetics
- Dysmorphology
- Eye Genetics
- Statistical Genetics
- Clinical Cytogenetics
- Clinical laboratory genetics (beginner's course)

Specific reports were received from a number of the course organizers.

- The 1st and the 2nd editions of the 5-day residential short course ("Introduction to statistical analysis of genome-wide association studies" were held in at the Imperial College London, London, UK. Overall, the course was attended by 78 delegates originating from 34 countries and working in 21 countries, including Belgium, Croatia, Denmark, Egypt, Finland, France, Georgia, Hungary, India, Ireland, Italy, Latvia, Mexico, Netherlands, Norway, Russia, Serbia, Slovenia, Spain, Sweden, United Kingdom. ESHG scholarships were awarded to young researchers from Croatia, Georgia, Hungary, Latvia, Mexico, Russia, Serbia, and Slovenia. All scholarship holders would have not been able to attend the course without the support from the ESHG. The course was attended by researchers at various stages of their career, from resent BSc graduates, PhD students to senior scientists. The course targets the audience willing to understand better the statistical approaches and analytical procedures for genetic association studies into common human traits and diseases. Each course topic is covered by a lecture, followed by a practical exercise, which includes use of the state-of-art software tools and example datasets. The course practicals are available for delegates for a month through internet to master their skills. Feedback fro students was very positive. The 3rd edition of the course will take place on July 2-6 in London, UK.

- The 9th Dysmorphology in the Genomic Era course was held in Manchester, and had 52 delegates from 26 countries including USA, Argentina, India, Malaysia, Saudi and Palestine. ESHG scholarships were awarded to young doctors from Slovenia, Romania, Poland, Hungary, Serbia, Portugal, Lithuania and Czech Republic. Course format included traditional lectures, interactive sessions, workshops and quizzes. In addition many participants gave excellent case presentations. 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 excellent with some excellent suggestions for the future from participants who also commented that, in addition to learning a great deal about dysmorphology, they particularly valued the opportunity to meet colleagues from all over the world. Many felt that they would not have been able to attend without an ESHG scholarship.

-Forty nine delegates attended the third ESHG cardiac genetics training course in Manchester. They represented 24 cardiologists and 25 geneticists (counselors, clinical geneticists and laboratory staff) from 19 countries across Europe and extending beyond to Argentina, Canada and Morocco. The program comprised lectures, case presentations and workshops delivered by 19 experts from across Europe including a keynote lecture on vascular Ehlers Danlos syndrome by Prof Xavier Jeunemaitre, Paris. The feedback was outstanding with delegates especially appreciating the parallel workshops for cardiologists and geneticists on electrophysiology, cardiac imaging and sequence variant interpretation.

- The 30th European School of Genomic Medicine and NGS was held in Bertinoro rom 29 April to 4 May 2018. There were 93 students from many different countries and a similar number form remote training centers in Lebanon, Malta, Iran, Egypt, and Spain, following the course via online streaming. There was strong interaction between students and faculty during lectures, workshops, and breaks. The evaluation of the 2017 course was excellent, with average appreciation exceeding 8/10 for almost all lectures and workshops.

ESHG board members to join the Education Comittee

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 2017

by GertJan van Ommen, Editor in Chief, EJHG

EJHG 25th anniversary

The past year has been a year of celebrations. The ESHG turned 50, with a celebratory meeting in Kopenhagen, where it all started too, and the EJHG turned 25. This was celebrated firstly with a history special from Peter Harper, available at last year's Kopenhagen meeting, going back deep to the ESHG's early past, as well as republishing a collection of noteworthy papers of the past decades. A second Anniversary issue has kept us all busy during the summer and autumn. A large body of key persons from the last 25 ESHG years were asked for their personal recollections of this period, and, with the aid of Mary Rice as guest editor, we compiled this into an 'everything you ever might

learn about the ESHG'. It was published in the late fall of 2017, sent along to members and subscribers and if you are fast, there are a few extra copies available on the Springer/Nature and ESHG stands this years' meeting. Finally, on the EJHG website you can follow a special 25th anniversary video.

Impact Factor

In 2017 our impact factor (citations measured over 2015-2016) went slightly down from 4.58 to 4.29 and the editorial team will do its best with its critical appraisal so we can make up. We have had a number of well-cited papers in 2017 also highly quoted in the social media, so we have our hopes up. But we need the help from the submitting authors, too! Mention your papers on Twitter and Facebook. Predicting public interest is difficult and we are sometimes surprised about at which papers turn out to be hot topics in a given year.

Reviewing and time to publication.

In 2017 we received 727 submissions, against 747 last year, 2.6% less, while last years drop was 5% so perhaps we are levelling off. Our acceptance rate for 2017 has been 33% (2016: 27; 2015:32). The average time to first decision, after review, was 63 days like last year. Papers which are not reviewed are on average returned within 12 days. What is typically – and understandably –perceived by authors as THE review times is actually a composite of the reviewer allocation - and acceptance! – time, and the actual review times. Actual review times have been stable over the past years at 14-15 days, which is quite good indeed, but does not include, the long time it often takes us before we have secured enough confirmed reviewers. In the last years this hovered around 28-25 days, fortunately with a downward trend (25 days in 2017). The average time after acceptance to online publication was 28 days and average time to print publication 68 days. The production department expects to shave off a few days still after the completion of the systems integration of Springer and Nature.

Web visibility

We have a new design of the EJHG web page with a new service: after logging in at www.nature.com/EJHG, you will find a new link 'Editor's Choice: The best of European Journal of Human Genetics 2016-2017' which relays you to a whole slew of highly downloaded and cited papers from the last two years.

Full-text downloads in 2017 amounted up to 1.2 million times. The absolute topper of this year is unquestionably "Genetics of the peloponnesean populations and the theory of extinction of the medieval peloponnesean Greeks", by George Stamatoyannopoulos et al., published in 2017 even, with no less that 17,726 views. Clearly the wrong idea floated in the 1800's that the original Greeks would be extinct sat very high with today's Greeks and has now been conclusively refuted. The second place is for "Genetic evidence for an origin of the Armenians from Bronze Age mixing of multiple populations", by Marc Haber, published in 2015, with 6,025 views, and the third place is for "The phylogenetic and geographic structure of Y-chromosome haplogroup R1a" by Peter Underhill et al. with 5374 views, published in 2014.

EJHG Award

As every year, EJHG, and Nature Publishing Group, as of last year Springer/Nature, jointly offer a junior authors' high-citation award. This is given to the top-3 articles published in 2015, with citations counted in the 12 months following after 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. Alessandro Mussa et al., University of Torino, Turin, Italy. "(Epi)genotype-phenotype correlations in Beckwith-Wiedemann syndrome", published in the European Journal of Human Genetics (2016) 24, pages 183–190, doi: 10.1038/ejhg.2015.88, receiving 16 citations.

2. Anna Middleton et al. , Connecting Science, Cambridge, UK , "Attitudes of nearly 7000 health professionals, genomic researchers and publics toward the return of incidental results from sequencing research", published in the European Journal of Human Genetics (2016) 24, pages 21–29, doi: 10.1038/ejhg.2015.58, receiving 14 citations.

3. Fiona S. Togneri, West Midland Regional Genetics Lab., Birmingham, UK, "Genomic complexity of urothelial bladder cancer revealed in urinary cfDNA", published in the EJHG (2016) 24, pages 1167–1174, doi: 10.1038/ejhg.2015.281, receiving 10 citations.

In addition there is a special award for a guidelines paper by dr Gert Matthijs and coworkers 'Guidelines for diagnostic next-generation sequencing', published in the EJHG (2016) 24, 2–5; doi: 10.1038/ejhg.2015.226. This paper received 22 citations, and since guidelines papers always receive so many more citations that regular ones this has been given a honorary award.

Watch – and use - EJHG-tube: your paper on video

For the prospective authors: In 2016 EJHG and Springer/Nature implemented a major innovation: the EJHG encourages authors to send in video abstracts as part of their final submission (or following acceptance). These video summaries are a unique way for authors to present the information in their paper and further enhance the visibility of their work. Through this video media authors can convey their findings without the constraints of the written word, plus provide a new and enhanced user experience for readers of the journal. We dubbed this 'EJHG-tube'. The first videos are on the website and the format may still evolve with time. We expect that a live rendering of your work may increase interest and citations. The uptake until now has been somewhat hesitantly, so we'd like to reiterate that it does not need to be something very special or contrived: just practise a short ca 4-6' conference talk (which you often will be doing anyway), have some co-authors or other colleagues listen in, and when you are happy with it switch on your iPhone, Samsung, or Android and record it. You may also interlace the narrative with stills of your slides, or even start from commented slides, but it does help seeing the presenter from time to time. Subsequently, just submit the video as a supplementary file togeth-

er with your manuscript. If your paper is accepted, the video will be available for viewing online in the supplemental material. Please note that submitting a video is not obligatory, and its inclusion will not impact editorial decisions. We accept the following files: .mov, .mpg, .mp3 and mp4. For more information please refer to the journal's Guide to Authors.

Annual report ESHG-Eurogentest Committee 2017-18

By Hans Scheffer, Chair of EUGT

The ESHG-Eurogentest committee (EUGT) is the committee dealing with aspects of diagnostic laboratories and services in genetics. Apart from own initiatives and activities EUGT tries to interact with the many international stakeholders in this field. EUGT has four subcommittees:

1. The Quality subcommittee
2. The Guidelines subcommittee
3. The Training subcommittee
4. The Dissemination subcommittee

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 is a strong brand name. During the last three years EUGT has been integrating its activities with other ESHG activities, e.g. educational (courses organized by the Educational Committee) and training activities (organized by Eurogentest) cover similar topics but are dealt with from a different angle.

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. For details see the summaries of the subcommittees below.

The Quality subcommittee objectives 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

In May 2017, a forum meeting was held during the ESHG conference for the European and National genetics EQA providers. This was a productive meeting where common issues including performance criteria, poor performance, critical errors and non-submissions was discussed. An agreement was reached that all the EQA providers would submit their poor performance data to the QSC in future. Consequently in December 2017, seven external quality assessment scheme providers including EMQN, CEQAS, ERNDIM, UK-NEQAS Molecular Genetics and UK NEQAS LI, CF Network, and ISS are now submitting their anonymized poor performance data for all their EQAs to the Quality sub-committee (QSC). The EQA providers are also now providing an explanation and further feedback if the percentage of poor performance is greater than 10%. A second forum meeting of the EQA providers will take place during the 2018 ESHG conference.

A survey of National Governance bodies in Europe revealed that Switzerland and the United Kingdom have advisory bodies to which genetic centre with persistent poor performance were referred. The need for countries to have such similar advisory bodies has been raised at the ESHG National Representatives meeting.

A survey on Newborn screening (and regulatory frameworks) in Europe has been sent to the National representatives, the outcome of which will be made available to all.

In November 2017 the ESHG executive board decided that Els Dequeker of the University of Leuven (Belgium) will represent ESHG-EUGT as an observer in the workgroup of Health Care of European cooperation of Accreditation (EA). This working group meets twice a year with the attendees of all national accreditation bodies of Europe, as well as with representatives of European Scientific Associations. As the ESHG-EUGT committee feels that it is important to be informed on key issues regarding quality management regulations and developments in this field an observer was assigned. Two important topics were discussed in relation to the diagnostic laboratories for genetic testing. Firstly the impact of general data protection regulation (GDPR) and the ISO accreditation process : the accreditation bodies will also review implementation of this regulation in line with the ISO 15189. Secondly a discussion between the national accreditation bodies is ongoing whether interpretation is necessary on genetic data. More specific can a laboratory be accredited without providing interpretation of data? This latest point was discussed in the quality subcommittee of EUGT, and all members agreed that interpretation is necessary. This feedback will be given on the next working group meeting of EA.

The Guideline subcommittee discussed the different types of existing guidelines and their different purposes. The conclusion was that practical guidelines and policy documents have been issued that serve different purposes. For policy documents EUGT closely collaborates with the PPPC. Priority by the Guideline subcommittee will be given to the development of novel guidelines/recommendations for WGS in Diagnostics. These guidelines will also include further recommendations on the interpretation of variants of unknown significance (VUS). This initiative will be undertaken as part of the Horizon2020 Solve-RD project, in close collaboration with other stakeholders including European Reference Networks (ERNs).

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.

The Dissemination subcommittee has updated the EUGT website www.eurogentest.org . We have made several updates to the website, announcing novel events, guidelines, resources and information availability. A start was made to increase our visibility on social networks, predominantly using Twitter to disseminate most recent announcements on the news and information associated with EUGT. In 2017 EUGT successfully organized a workshop integrated in the program of the annual ESHG meeting in Copenhagen entitled: „ Quality assurance in interpretation and reporting in genome wide diagnostics". It was oriented towards diagnostics, with the focus on current challenges and hands-on experience in the interpretation and reporting in the field of diagnostic targeted and exome/genome-wide NGS. In the first part, there were 2 talks (quality control assessment of variant interpretation, the performance analysis of guidelines for interpretation of genetic variants). The second part of WS was a round table discussion on experience and practice of utilizing WES/WGS in clinical practice across EU countries combined with participants' voting on most interesting discrepancies in reporting. There were many participants with very positive feedback. This year again a EUGT workshop has been organized, to be held during the current ESHG meeting in Milan.

Several strategies have been pursued to make EUGT self-sustainable (with a limited ESHG support). One has been to get ESHG-EUGT involved as formal partner with a budget in research projects. EUGT is now partner and WP-leader in the Horizon2020 project Solve-RD. One of the tasks will be to coordinate the development of guidelines for WGS in diagnostics. For this a close collaboration with ERNs will be sought, initially with four ERNs participating in Solve-RD. These guidelines will also include further recommendations on the interpretation of variants of unknown significance (VUS). Strategies to have some EUGT activities sponsored have also been successful. A two day sponsored EUGT symposium is scheduled for 9-10 October 2018 in Nijmegen, preceded by a variant interpretation workshop.

The paper containing EUGT guidelines for diagnostic next generation sequencing, endorsed by the ESHG received an honorary award as "one year after publication most cited paper" in the EJHG:

Gert Matthijs, Erika Souche, Mariëlle Alders, Anniek Corveleyn, Sebastian Eck, Ilse Feenstra, Valérie Race, Erik Sistermans, Marc Sturm, Marjan Weiss, Helger Yntema, Egbert Bakker, Hans Scheffer and Peter Bauer (2016) Guidelines for diagnostic next-generation sequencing, European Journal of Human Genetics 24, p.2–5

The EUGT steering committee has undergone some changes in personnel: Rosalind Hastings stepped down as chair of the Quality subcommittee and her position has been taken over by Sandi Deans, and Anne-Françoise Roux stepped down as chair of the Guideline subcommittee. Her position has been taken over by Gert Matthijs. We would like to express our thanks for the continuous efforts and energy Ros and Anne-Françoise have spent on EUGT activities over an extensive number of years. For further changes in the subcommittees we refer to the ESHG and EUGT websites www.eshg.org and www.eurogentest.org .

Signature and ratification process of the Additional Protocol on Genetic Testing for Health Purposes by the Czech Republic: Impact for member states of the CoE

by Milan Macek Jr, President of the Czech Society of Medical Genetics and Genomics

During the 20th Anniversary of the Oviedo Convention (Convention on Human Rights and Biomedicine), an international conference was held on 24-25 October 2017, in Strasbourg (www.coe.int/en/web/bioethics/20th-anniversary-of-the-oviedo-convention), under the auspices of the Czech Chairmanship of the Committee of Ministers of the Council of Europe (www.coe.int/en/web/congress/cm-czech-republic). On October 24, The Czech Republic representatives signed in Strasbourg the "Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes" (CETS No 203; www.coe.int/en/web/conventions/full-list/-/conventions/treaty/203; DOI: 10.1038/ejhg.2009.84; henceforward termed the „Protocol"). Currently, the ratification process of the Protocol has advanced with all relevant Czech (CZ) Parliament and Senate committees expressing their unambiguous endorsements. Final, hearings in both chambers are expected to take place during second half of May 2018, with an expectation that the ratification process will be completed by early summer 2018.

Between 2014-2016 the Czech Society of Medical Genetics and Genomics (www.slg.cz; CZSGG) had been working on the amendment of Act 373/2011 Coll. (Art 28-29 on Genetic testing) which are now fully in accordance with the Protocol. Our professional body had also involved the Czech National Rare Disease Alliance (www.vzacna-onemocneni.cz) for lobbying of the government to sign the Protocol and start its ratification immediately thereafter. We were also very indebted to bioethics expert Dr. Vera Frankova from the Institute of Medical Humanities at Charles University who provided her expertise when working on the dossier providing legal and ethical justification for the signature of the Protocol. This concerted effort (i.e. involving both experts and patient associations) was aimed not only at the country itself, but also had in mind the broader „Council of Europe-wide" (CoE) context.

In this regard, the ratification of the Protocol by the CZ will express the definite consent of the country to be legally bound by the Protocol and it will make it possible for it

to come into force for all member states of the Council of Europe (www.coe.int). The entry into force of the Protocol requires five ratifications of the Protocol by Council of Europe Members States. Four states have already ratified the Protocol (Moldova, Montenegro, Norway and Slovenia), with the Czech ratification reaching the minimum threshold for its entry into force for the benefit of the field of clinical- / medical genetics and for patients with genetic disorders.

For further information we refer to the website https://www.coe.int/en/web/bioethics/20th-anniversary-of-the-oviedo-convention

Report on the 2nd Course of Basics in human genetic diagnostics

by Nikoleta Christoglou, Biologist MSc, Thessaloniki, Greece.

The 2nd Course of Basics in human genetic diagnostics – A course for Clinical Laboratory Geneticists (CLGs) in education Athens, Greece, September 25-29, 2017

Nikoleta Christoglou, Biologist MSc, Thessaloniki, Greece.

The second course on "Basics in human genetics diagnostics - A course for CLGs in education" organized by Dr. Thomas Liehr (Jena, Germany) and Myrto Poulou (Athens, Greece), took place in Kostis Palamas, Cultural Centre of Athens in Greece the participation of many clinical laboratory geneticists, molecular biologists, biochemists and clinicians from Europe, Asia, and Africa. Overall 49 participants from 17 countries (Armenia, Austria, Bosnia & Herzegovina, Bulgaria, Cyprus, Denmark, Greece, Iran, Latvia, Morocco, Netherlands, Pakistan, Portugal, Romania, Serbia, Singapore, and Spain) were present. The course was mainly supported ESHG and additionally by ADS BIOTEC, Carl Zeiss Jena, Face2Gene, MetaSystems, MRC-Holland and ZytoVision. Almost 25 lecturers gave 44 presentations distributed over 5 days. Daily program started with lectures on theoretical basics of certain topics and continued with workshops followed by analysis and discussion of different cases. Each day finished with a written exam in which the participants were asked to answer to questions and cases concerning the topics of the day. On Tuesday and Friday we had nice dinners in common, included in the course fees, as well as on Friday a walk through Athens old city.

The 1st day was based on human genetics, the history of genetic diagnostics from karyotyping to next generation sequencing, the Mendelian and non-Mendelian inheritance patterns. Moreover, topics as genetic counseling and syndromology, were also covered and in the end we were informed about a new database 'Face2Gene' listing cases of people with syndromes and indicates the correlation that exists between phenotype and genotype. There you can upload a photo of the person you examined and find the risk of percentage of having a syndrome. During the workshop, we worked on family pedigrees, staring with taking family history to how to construct a family pedigree and perform risk assessments. The 2nd day was focused on cytogenetics from the past up to nowadays techniques, guidelines which are used in the laboratory, the ICSN nomenclature in different cases and teratology. In the workshop we practiced on setting up a human GTG-banding based karyotype. The lectures of the 3rd day concerned molecular cytogenetics, molecular genetics, epigenetics and gene therapy. FISH techniques and array-CGH principles and applications were also covered in the workshop, where a variety of such cases were discussed along with how to report a result correctly by using databases. The day we concluded with a lecture on PCR techniques. The 4th day was devoted to a variety of molecular techniques as Non Invasive Prenatal Test (NIPT), MLPA, Sanger sequencing, PGD and Next Generation Sequencing (NGS). In the workshop reporting in Sanger sequencing and in PGD cases was exemplified. The last day of the course continued with molecular genetics and future direction in the field. NGS principles and approaches, cancer genetics, biochemistry genetics and metabolic disorders, neurogenetics and proteomics were shortly presented.

Generally, despite tight schedule of the program, the course was scientifically rather interesting, very well organized, with excellent speakers and presentations. As far as I am concerned I would like to thank for the fellowship (there were 16 full or partially fellowships on the course fees available), and thus for the opportunity to participate in that course which was really educative, especially, for new scientists and clinicians by covering the majority of the techniques and the cases that are faced in a diagnostic cytogenetics or molecular genetics laboratory. Furthermore, the more advanced scientists had the opportunity to refresh their knowledge, as well as to enrich them so as to be updated. In my opinion it was a memorable experience for every participant.

Comment added by Thomas Liehr: My special thanks go to Isabel Marques Carreira (Coimbra, Portugal), Myrto Poulou (Athens, Greece) and Anja Weise (Jena, Germany), taking over (very short-term) my lectures and duties during this course, as I had a broken leg, which hampered my traveling to Athens. Also I thank all lecturers and organizers of this course, providing their time and expertise completely for free, keeping by this the fees for the course as small as possible.

Participants and some of the lecturers in front of Kostis Palamas Building

Report of the EBMG Chair

by Angus Clarke, Cardiff, United Kingdom.

The past year has seen a lot of activity within EBMG, both within the three Professional Branch Boards (PBBs) and within the Executive Committee. The PBBs speak for themselves below; the principal activity of all three relates to the question of certification required for professionals to demonstrate their competence. Two of the PBBs already operate their own processes, with the Genetic Nurses and Genetic Counsellors (GNGC) using the evaluation of a candidate's portfolio of evidence and the European registered Clinical Laboratory Geneticists (ErCLG) using an examination in case there is no national CLG system existing in the candidate's home country. The Clinical and Medical Genetics and Genomics (CMGG) group is also working, along with the associated UEMS branch, to develop a certification system. In addition, the ErCLG PBB is helping to establish a fourth PBB for a group that may be regarded as so far under-professionalised, the Genetics Laboratory Technicians.

The Executive Committee has had some very active discussion about two key topics. These are (i) the role of EBMG itself and how it relates to the work of the three branches, and (ii) how the constitution of EBMG should be implemented and how the Chair of EBMG should be selected. These topics are somewhat connected, in that the Executive Committee could either be a largely supervisory body, ensuring that the PBBs were functioning consistently and transparently, or it could perhaps adopt a more active role, taking more initiative. The former approach would work well with an annual rotation of the role of chair between the three professional groups, while the latter would perhaps benefit from a chair appointed for a longer term and with more independence of the PBBs. And what would work well at one time might need to be revised in a few years: these choices are not made for all time but can be discussed in EBMG General Assemblies and the details can develop organically acording to changing situations.

The implementation of the EBMG Constitution is being developed presently, with nominees of the Board of the ESHG participating in the discussions. We look forward to discussions on the EBMG Constitution and the appointment of the next Chair at the General Assembly of the EBMG in Milan.

Genetic Nurses and Genetic Counsellors (GNGC)

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. Next year will be the fifth year of our branch and we are now preparing the final documentation for accreditation renewal of the professionals registered in the first year of certification. This year, our board is happy to welcome two new members: Joana Bengoa from France (who will replace Emmanuelle Haquet) and Marion McAllister from UK.

We are proud to announce a new paper published in the European Journal of Human Genetic entitled « The perceived impact of the European registration system for genetic counsellors and nurses ».

There are several updates and dates to note for the GC&GN registration process:

The Grandfather routes will close this year in some countries. This route to registration was put in place to recognize the experienced health professionals without an MSc who are practicing as genetic counsellors. Applications under the Grandfather clause are possible for applicants who work in a country where there is an approved Master course that has been running regularly since 2012 can be made October 2018. The grandfather clause for applicants who work in a country without an approved Master course will continue until October 2020.

For MSc Genetic counselling programs requesting the Branch Board's curriculum approval, all MSc programs are now expected to introduce Genomics at their curriculum at the moment of the application for renewal.

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 2017 the registration cycle was run for a fourth year. We received a total of 24 notifications of intention to register, most using the grandfather clause. This year, the board will meet in order to assess those applications on 16th June so we will be able to recognize the successful candidates at the ESHG in Milan.

We now have 69 European registered Genetic Counsellors (64) & Genetic Nurses (5) from Belgium (2), Cyprus (1), France (18), Greece (1), Iceland (1), Ireland (4), Italy (2), Norway (8), Portugal (1), Romania (1), Spain (13), Sweden (1), Switzerland (5) and United Kingdom (11).

More news and updates can be found in our website (https://www.eshg.org/471.0.html).

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

Clinical Laboratory Genetics (CLG)

The European Board of Medical Genetics (EBMG), Professional Branch Board for Clinical Laboratory Geneticists (ErCLG) will have registered ~400 ErCLGs by end of this year. This is a great success already, but we know there are many more colleagues out there who are eligible to apply for this title. So we kindly invite them to register in the 2018/19 year round via https://www.eshg.org/695.0.html between July 15thand October 15th 2018. Also, we have another three member states of the EU their national CLG title is recognized (Hungary, Italy and Slovenia). So we need another six states to apply for CLG to be a recognized profession in EU.

Due to numerous requests in the paper "Expert knowledge on human genetic counselling and chromosomics are necessary for sound genetic laboratory diagnostics (Mol Exp Biol Med 2017;1:1-3), which can be found on our web page, some example questions for the ErCLG test for group 3 applicants have been publishe.

We also wish to announce that a new laboratory genetics technicians (LGTs) branch has been started and seeks members who want to work actively in this new branch.

Clinical and Medical Genetics and Genomics (CMGG)

The Branch focused on the European Diploma in Medical Genetics and Genomics (EDCGG), which is a joint development of the UEMS Section of Medical Genetics and our EBMG Branch. It is intended to be the main knowledge-based assessment tool for Clinical and Medical Genetics, and Genomics training in Europe, with the aim of raising standards in the specialty to world class levels throughout all European countries. A pilot EDMGG examination will be staged in June 2018 as a satellite event of the ESHG in Milan, with the first full inaugural examination to be held in June 2019 in Gothenburg. It will be open to candidates worldwide; the title will be valid for life, though Continuing Medical Education (CME) renewal is recommended for active practitioners, by appropriate record review and approval through official bodies at national level and EACCME, which is an EU Continuing Medical Education (CME) recognition system following on from registration. A similar system of CME following registration is successfully operated by our sister branches in the EBMG. It is planned that both the EDMGG application system and the CME registry will be opened later in 2018. Beside this we worked on the JARC WP8 package (Medical Education).