THE EUROPEAN SOCIETY OF HUMAN GENETICS

NEWSLETTER

No. 30 - May 2017

Presidential Address

by Olaf Riess, President of the ESHG

Dear ESHG members, dear future ESHG members,

We are living in an interesting time. Human genetics is changing medicine visibly, in all disciplines and across the globe. As genetics is developing as an integrated part of the prediction and prevention of diseases, it is also becoming a field of increasing interest to medical insurance companies and a football for politics. In March this year, a Bill of the United States of America that would allow companies to require employees to undergo genetic testing or risk paying a penalty of thousands of dollars, and would let employers see that genetic and other health information, was recently approved by the US House of Representatives Committee on Education and the Workforce. Clearly, when this bill becomes law in the US, it will significantly interfere with genomic health concepts. It is our responsibility to ensure that employees should be tested on their current skills and not on their future health risks in our own countries. I count on our Society to stand up and fight for the interests of our patients and even of healthy individuals when similar issues may come up in European countries as in the US (hopefully this will never happen).

Politically and in research networking terms, Brexit remains one of the major challenges of the next years. Despite the promise of the British government to financially support research that has already started with British partners in the Horizon2020 programme, participation of British scientists beyond the Brexit in 2019 remains unclear. UK's innovation sector and SMEs have received more than €318m from the Horizon 2020 platform, more than any other country. The uncertainty of participation of Great Britain in today's European research calls already lets "mainland" EU researchers consider not integrating British scientists despite their excellence. Thus, the ESHG is proud to have Christine Patch as its next president, in the hope of giving a strong voice as a communicator to keep British scientists integrated into European research funding and also in the recently established 23 European Reference Networks (ERN) for rare diseases. This latest milestone of cross boarder patient care involving more than 300 hospitals, located in 25 EU Member States and Norway, which was announced in Vilnius, Lithuania, in March 2017, now needs to be integrated as a general structure within European research projects. As 80% of all rare disease are caused by genetic factors, ESHG and European human geneticists should become major contributors to these structures.

Integration and not separation is a major interest of the ESHG. We therefore further increased the number of fellowships for training courses (https://www.eshg.org/courses.0.html) and for the ESHG conference in Copenhagen (https://2017.eshg.org/) for Europeans and for scientists from developing countries. We are deeply indebted to the programme committee of the ESHG under the guidance of Joris Veltman, and of the ESHG education committee under the chair of Han Brunner for the courses.

As this is my last presidential address for our Newsletter, I want to thank Karin Writzl (Secretary General), Gunnar Houge (Deputy Secretary-General), Andrew Read (Treasurer), Feliciano Ramos (Vice-President), Christine Patch (President-Elect), and Jerome del Picchia (ESHG Executive Officer) the entire executive board (https://www.eshg.org/56.0.html) for their dedication, a wonderful team of inspiring people working with devotion for our steadily growing society.

Report from the SPC Chair: Welcome to the 50th ESHG in Copenhagen: Back to where we started from!

by Joris A. Veltman, Chair of the ESHG Scientific Programme Committee

After 50 very impressive ESHG meetings there is no reason to assume this year's meeting will be anything less exciting. Technological developments continue to surpass our expectations, from almost routine genome sequencing to the amazing CRISPR revolution of genome editing. With these tools in hand our diagnostic colleagues, basic scientists and clinicians work hand in hand to improve rare disease diagnostics, unravel the genetics of complex traits, study gene regulation and develop and apply novel therapeutic approaches. The Scientific Programme Committee of the ESHG, with invaluable input from the ESHG membership, has invited more than 80 international top scientists to speak at our meeting this year. After the welcoming address on Saturday we will have a special plenary session to look at the past, present and future of the ESHG and the field of human genetics as a whole. Our annual ESHG-ASHG building bridges session on Tuesday will also look at the past, present and future, but in this case focussed on ethical and legal discussions. On Tuesday we will also have our an-

nual Mendel and ESHG award lectures, given this year by eminent scientists George Church and Edith Heard.

A total of 138 out of 2389 submitted abstracts have been selected for oral presentations in the plenary highlight session as well as 22 concurrent sessions. New this year is a special concurrent session in which the 20 best poster candidates will each pitch their poster. Afterwards the attendants will have time to discuss these posters with the presenters at electronic posters outside the session hall. The normal poster sessions remain excellently suited for exchanging information and making new contacts, presenting the wealth of genetic research and diagnostics from around the world. As always, we have tried to make sure there is something for everybody: for clinicians, for people delivering diagnostic services and for researchers. We firmly focus on education and training, and our educational sessions are meant to provide an introduction to the topic whilst providing all attendees with an up to date knowledge of the developments in the particular field. In addition, our workshops are a tool to make the programme more interactive, by allowing the audience to discuss hot topics in research, diagnostics and clinics. Clearly, 50 years of ESHG just marks the starting point of human genetics. Let's take a moment to celebrate and then further shape its bright future together here in Copenhagen!

On behalf of the Scientific Programme Committee I wish you all a most interesting and exciting ESHG 2017!

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. Please help us to make next year's meeting in Milan even better by making use of this possibility!

See www.eshg.org/proposals2017.0.html

Report from the Public and Professional Policy Committee 2016-2017

By Martina Cornel, Chair of the ESHG 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.

No to compulsory DNA testing

In 2016 the Kuwait government, as the first country in the world, wanted to introduce a law calling for the compulsory DNA testing of all its citizens, and also of all visitors, whatever their reason for entering the country. The President of the European Society of Human Genetics and the chair of the PPPC have sent a letter dated 31

August 2016 to His Highness Sheikh Jaber Mubarak Al-Hamad Al-Sabah and to The Council of Ministers of the State of Kuwait asking them to reconsider this law and to amend it so that human DNA is collected for legal purposes only from individuals suspected of having committed serious crimes. In October 2016 the Emir of Kuwait requested the Kuwaiti Prime Minister to reconsider the scope of the law. In May 2017 next steps are expected in court.

In 2017 the US House of Representatives Committee on Education and the Workforce approved a Bill that would allow companies to require employees to undergo genetic testing and disclose the results to their employers, or risk having to make health insurance payments of thousands of dollars extra. This Bill has apparently been integrated into the activities related to the revision of the Affordable Care Act, otherwise known as Obama Care. All 22 Republicans supported the Bill and all 17 Democrats opposed. In the past, however, decisions on whether or not to undergo genetic testing have been the voluntary choices of individuals. Both the Council of Europe and the US law (Genetic Information and Non-Discrimination Act, GINA) uphold this standpoint. The European Society of Human Genetics (ESHG) defends the principle that employees should be employed on the basis of their skills and expertise, and not on their future health risks. Professor Martina Cornel, chair of the PPPC, reaffirmed this view on behalf of the ESHG through a press release dated 16 March 2017. "Transparency is needed on the potential decision to discontinue the GINA. The genetic and health information of individuals needs protection".

Post-mortem genetic testing

In 2016 PPPC worked on genetic testing in the case of sudden cardiac death. Autopsy procedures are generally well described in various European regulations, however, these often poorly integrate post-mortem genetic testing. To address the lack of coordination between different professional domains and improve guidance on post-mortem genetic testing for cardiac disorders, PPPC organized a multidisciplinary Workshop sponsored by the Brocher Foundation and ESHG, on 23-25 November 2016. The workshop consisted of presentations by 12 experts in (forensic) pathology, cardiology, genetics and law, and group work to identify common challenges and draft recommendations. Preliminary 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 outcomes of the Workshop will be presented by Dr Florence Fellmann during the Educational Session "Strategies to avoid sudden cardiac death" (E11), scheduled on Monday May 29, 2017 at 8:30 am. PPPC seeks to collaborate with other relevant scientific societies to discuss and disseminate the results. Recommendations on this topic will be formulated in collaboration with other scientific societies.

Gene-editing

Fast technical developments including CRISPR-CAS9 technology raise many policy questions. Considering the speed of developments and the multiple (potential) applications in different clinical settings, PPPC and the Board considered gene editing a priority to contribute to the dialogue. An agenda setting paper was developed in 2016 and submitted. The PPPC developed recommendations and a background document on germline gene editing together with the European Society of Human Reproduction and Embryology (ESHRE). A first draft of the recommendations were posted on the ESHG website from October 17 to December 2, 2016 and presented at the Annual Congress of the American Society of Human Genetics mid October 2016. In April 2017 both the background document and the recommendations were posted on the ESHG website for consultation and ESHG members were invited by email. The goal of these activities is to contribute to the debate on the current ethical and legal regulations and societal discussions. In many countries germline interventions have been prohibited, sometimes even accompanied by criminal sanctions.

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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? These and other questions require debate beyond our genetics society. We hope that the work of PPPC will contribute to this debate.

Members of the PPPC in 2016-2017 were Caroline Benjamin, Angus Clarke, Martina Cornel (Chair), Guido de Wert, Carla van El (Secretary-general), Florence Fellmann, Francesca Forzano (co-Chair), Heidi Howard, Hülya Kayserili, Bela Melegh, Alvaro Mendes, Markus Perola, Dragica Radojkovic, Emmanuelle Rial-Sebbag.

Report from the ESHG Education Committee

by Han Brunner, Chair of the ESHG Education Committee

ESHG has a mission to promote the practice and knowledge of Human and Medical Genetics in Europe.

The aim of the Education Committee is to foster educational activities in Human and Medical Genetics in Europe. It works by designated taskforces led by one or more Education Committee members who each focus on a specific educational topic. The ESHG Education Committee currently has four core activities;

- The DNA day essay contest for school children coordinated by Christophe Cordier.

The 2016 winner was Maria Sasaki from Cambridge School of Bucharest, Romania. The 2017 question was:

"CRISPR/CAS9 is a powerful new technology to change genetic ma-

terial in all living material including humans, animals and plants. Explain how this technology works. Give your opinion what problems and opportunities this technology should be used for, and what potential uses should not be allowed. Explain why you would make these choices."

- The School Children event at the ESHG annual conference. ESHG liaison Domenico Coviello. Organizer for 2016, Cristina Gonzalez and Conxi Lazaro: School children were told about "Genetics in Art, History and in our times" and learnt about various genomic techniques, including how to find a murderer! They were also taught about Epigenomics. This was a very successful event. Students Organizer for 2017 will be Zeynep Tumer, two classes of a specialised highschool with biomedical focus will visit the ESHG conference on Monday, guided by tutors, and will get a first glimpse at a big international scientific conference.

- A portfolio of ESHG courses held in various locations across Europe, overseen by Han Brunner. These include the Course in Hereditary Cancer Genetics (80 participants, sold out); Preconception, Preimplantation & Prenatal Testing (95 participants); Manchester Dysmorphology Course (45 participants); European Course on Cardiogenetics; Basic & Advanced Course in Genetic Counselling; Next Generation Sequencing (60 participants, sold out); Medical Genetics (50 participants); Basics in Human Genetic Diagnostics; Introduction to the statistical analysis of GWAS; Clinical Cytogenetics.

Courses were generally very well attended and had a good geographical spread. Appreciation was generally very high. The ESHG fellowships greatly promoted the participation of students from lower-income countries.

Except for the Preconception, Preimplantation & Prenatal Testing course all directors have indicated that they would like to continue offering these courses this year. The European School of Genomic Medicine and the next generation sequencing Course will merge in 2017 to create the Genomic Medicine and NGS course. This course is currently sold out at 85 participants.

- A taskforce on the collection of teaching materials for Human and Medical Genetics, to be made available to the genetics Community in Europe, led by Jill Clayton-Smith. This had to be discontinued because Jill was completely absorbed in setting up an ERN on rare syndromes and intellectual disability.

- A taskforce on HGVS nomenclature with the aim of organizing one or 2 educational activities, overseen by Johan den Dunnen. This was approved by the board in 2016. Its activities have been postponed until next year due to organizational reasons.

EJHG Highlights 2016

by GertJan van Ommen, Editor in Chief, EJHG

In 2016, several Editorial policy changes initiated in 2014 have begun to bear fruit:

Impact Factor increase

Raising the bar for sending out papers for review in 2014 has caused our impact factor to increase from 4.225 (2014) through 4.35 (2015) to 4.58. We now rank 30th/165 in 'Genetics and Heredity' (was 36). In two years our acceptance rate has decreased from 37% (2014) through 32% (2015) to 27%. The latter will not yet be seen in the 2016 IF so it'll be interesting to see our next IF. Let's hope we're not doing something wrong and that the IF will further increase in the next few years.

Reducing print backlog to 2-3 months

In 2016, EJHG received 747 submissions in total, down by 5%, which is a trend across the field and probably due to the increase in titles. However, in total 310 papers appeared in print, 5 more than in 2015. At first sight, this seems at odds with raising the bar, but in fact as of 2014 we have increased the publication volume to reduce our print backlog. Up until 2014 this had run up to 9 months, in 2015 this was 6 months and it is now at a healthy 2-3 months. Several smaller improvements were achieved in or turnaround statistics: average time to first decision 63 days (was 68), average time to online after acceptance 28 days (35), with 59% online under 25 days (was 45%).

Web visibility

The journal website has been viewed 1.55 million times in 2016, slightly down from 2015 (1.65 million). Amongst the top 10 highest viewed papers in 2016 are three published in the same year: "Guidelines for diagnostic next-generation sequencing" by G Matthijs et al. (Policy paper, issue 24.1, 10.276 views); "Genetic evidence for an origin of the Armenians from Bronze Age mixing of multiple populations" by M Haber et al. (Article, issue 24.6, 9.336 views) and "Genomic complexity of urothelial bladder cancer revealed in urinary cfDNA" by FS Togneri et al. (Article, issue 24.8, 4.145 views)

EJHG Award

As every year, EJHG, and Nature Publishing Group, 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 are:

1. Dondorp W, de Wert G, Bombard Y, Bianchi DW, Bergmann C, Borry P, Chitty LS, Fellmann F, Forzano F, Hall A, Henneman L, Howard HC, Lucassen A, Ormond K, Peterlin B, Radojkovic D, Rogowski W, Soller M, Tibben A, Tranebjærg L, van El CG, Cornel MC. Non-invasive prenatal testing for aneuploidy and beyond: challenges of responsible innovation in prenatal screening. European Society of Human Genetics.; American Society of Human Genetics. Eur J Hum Genet. 2015 Nov;23(11):1438-50. doi: 10.1038/ejhg.2015.57. Epub 2015 Mar 18.
22 citations.

2. Kaye J, Whitley EA, Lund D, Morrison M, Teare H, Melham K. Dynamic consent: a patient interface for twenty-first century research networks.
Eur J Hum Genet. 2015 Feb;23(2):141-6. doi: 10.1038/ejhg.2014.71. Epub 2014 May 7.
18 citations.

3. Bayindir B, Dehaspe L, Brison N, Brady P, Ardui S, Kammoun M, Van der Veken L, Lichtenbelt K, Van den Bogaert K, Van Houdt J, Peeters H, Van Esch H, de Ravel T, Legius E, Devriendt K, Vermeesch JR.
Noninvasive prenatal testing using a novel analysis pipeline to screen for all autosomal fetal aneuploidies improves pregnancy management.
Eur J Hum Genet. 2015 Oct;23(10):1286-93. doi: 10.1038/ejhg.2014.282. Epub 2015 Jan 14.
13 citations

EJHG-tube: your paper on video!

In 2016 EJHG and Nature implemented a major innovation:

From June 2016 onwards, European Journal of Human Genetics has encouraged authors to include video abstracts as part of their submission. These video summaries should be included as supplementary material and 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 but the format may evolve with time. We expect that a live rendering of your work may increase interest and citations.

The uptake in the first year has been somewhat hesitant, so we would like to stress 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 together 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 from June onwards.

EJHG 25th anniversary

While strictly not in 2016, this column would not be complete without referring to achieving the 25 year milestone in 2017. To celebrate this, as well as the 50th Anniversary of the ESHG, we publish two special historical issues. The first one, available at this Copenhagen meeting, on one hand delves deep into the ESHG's past and on the other hand republishes a collection of noteworthy papers, originally published in the first two decades of our existence. This issue starts with two historic contributions written by Peter Harper. One is a flashback to the Society's first – very different – 25 year episode, from 1967-1992, and the other one is the result of many personal interactions with many pioneers of Human Genetics in Europe. 100 of the first-time geneticists in Europe the field have been interviewed by Peter over the last decades and he has made a selection of this for this special issue. We wish to note that while selection covers as many countries as are represented in his interview series, not all countries are covered, as we have only one Peter Harper. Indeed, to address this and extend the record of first-hand recollections, we shall be happy to hear of individuals who would

be willing to interview additional first-time geneticists, notably in the countries not yet covered.

For the second phase of the ESHG, from 1992 till today, considering that this was truly a Europe-wide group effort, we have decided to consult a large body of key persons from this period, enlisting them to contribute their recollections and views on this episode, in which genetics has moved to the center stage in current health care. We aim to publish this 'ESHG phase 2' compilation in the fall of this year, as a second installment of the 'Anniversary papers'.

Report from the European Board of Medical Genetics

by Heather Skirton, Chair of the EBMG

The European Board of Medical Genetics (EBMG) was formed in 2013 to set professional standards for health professionals working in the clinical genetics field in Europe, and to provide systems to enable professionals to demonstrate their professional competence. The EBMG is an autonomous legal entity, with a set of statutes (from 2014) and under the terms of those statutes, a General Assembly is held each year. For convenience, this is held during the ESHG Annual Scientific meeting. Although there is a need for the EBMG to work autonomously, the connections with the ESHG remain strong.

The EBMG comprises three distinct professional Branch boards, Clinical Laboratory Geneticists (CLGs), Genetic Nurses and Genetic Counsellors (GNGC) and Clinical Genetics and Genomics (CGG) (representing medical geneticists). There is naturally a great of collaboration and co-ordination across the Branches, but each Branch has a primary responsibility to serve their own professional discipline.

In 2016/17, the foundational work of the previous four years was consolidated, but there were also new developments. One of these was the establishment of an online application system for those wishing to join the EBMG register as a Clinical Laboratory Geneticist (CLG), Genetic Nurse or Genetic Counsellor. We had enormous support from the ESHG to do this, and though there were the inevitable teething problems attached to the introduction of any new process (especially it seems those that are designed to make things simpler!), we feel this will reduce the workload of the Branch members and also make the process more straightforward for applicants.

The EBMG has had much financial support for the ESHG to become established, but it is essential that it exercises due diligence and is able to fund its own activities. None of those working for the EBMG receive remuneration of any kind, but the EBMG does have to cover expenses directly connected with assessment of registration applications. For this reason, we have made a decision to alter the registration fees, so that we can cover those expenses. In addition, there was a considerable difference between fees paid by CLGs and those paid by GNGC registrants. To ensure equity across the professions, we have made the fees for the CLG and the GNGC registration comparable.

All details of the EBMG Branch members and the registration systems are available on the website [https://www.eshg.org/413.0.html].

The reports of the three Branch boards are as follows:

Clinical Laboratory Geneticist Branch

Chairs: Dr Thomas Liehr and Dr Isabel Carriera

In 2016/17 we had 84 applications for registration with the EBMG. The process of registration proceeds in two rounds, and six candidates were assessed as ineligible in round 1, another nine in round 2. Forty-two candidates were from group 1 or Group 2 countries and granted registration. Face-to-face examinations for those from group 3 countries will be held, mainly in Copenhagen.

A paper reporting the work of the CLG Branch thus far has been accepted by the EJHG and is in press. It is titled 'European registration process for Clinical Laboratory Geneticists in genetic healthcare'.

The ErCLG has also contributed to planning of courses to support CLG education prior to registration. Further details on the next course are available at: http://www.humangenetik.uniklinikum-jena.de/humangenetik_media/Bilder_Dokumente/Arbeitsbereiche/fish/Greece+2017.pdf.

Clinical Genetics and Genomics Branch

Chair: Professor Bela Melegh

At the General Assembly in 2016 the Medical Branch Board requested a change of Branch name and is now entitled the Clinical Genetics and Genomics Branch. The EBMG remained an indivisible partner of the Section of Clinical Genetics of the European Union of Medical Specialties (UEMS). In collaboration the board completed the document delineating the European Training Requirements (ETR) for Clinical Genetics with supplementary materials (formal, state of art "European Training Requirement", "Description of Clinical Genetics as a Medical Specialty in EU: Aims and objectives for specialist training" and a "Syllabus"). These have already been submitted to the various evaluating boards, including the ESHG Executive Committee and they are now progressing through the UEMS evaluation procedure. These documents will be discussed and voted on for official EU approval at the next meeting of the UEMS to be held at the end of April this year. These credentials are essential for the launch of the European Exam of Clinical Genetics, which we plan to introduce in 2018.

Genetic Nurse and Genetic Counsellor Branch

Chair: Milena Paneque.

Co-chairs:Christophe Cordier and Ramona Moldovan

This year the fourth cohort of genetic nurses and counsellors was registered and the total registered is now 69. In 2016 we received 29 intentions to register, of which 20 were considered eligible and 14 were approved for registration. A further three were not approved and were to re-submit next year.

Notably, most applicants come from countries without a national system for professional certification and were applying mainly using the grandfather clause, which will expire by 2020. The Branch is also proposing a new timeline for GNGC registration system (see website for details) [https://www.eshg.org/408.0.html].

The Branch assessed and approved a new MSc course in Genetic and Genomic Counselling at University of Cardiff. Another important project we were involved in as a Branch was the development of Orphanet Quality Criteria Questionnaire for Genetic Counselling Centres.

In new initiatives this year, we have responded to a demand from masters programmes and individual practitioners outside Europe.

We have developed an associate registration scheme for those working outside Europe who demonstrate competence, and we have also invited Masters programmes outside Europe to apply for assessment as EBMG approved programmes. We have already been contacted by Directors of MSc of Genetic counselling programs outside Europe who are interested into formal assessment of their courses against the European core curriculum for Master's programmes in genetic counselling. The approval of those programmes will enable their graduates to be eligible for registration by the EBMG under the new Associate Route.

We have sought feedback from registrants and a report has been accepted for publication in the EJHG, titled 'The perceived impact of the European Registration system for genetic counsellors and nurses'.

Finally, we would like to thank all the registered GC & GN and their senior colleagues and line managers from European genetic services for supporting the process of professional regulation of genetic counselling.

Conclusion

I retire this year from professional life and will no longer be a member of the EBMG. I thank the members of each Branch for their goodwill and hard work, which has ensured the success of the EBMG. I have huge confidence in handing over the Chairmanship to Professor Angus Clarke, who will be co-opted Chair until the statutory election next year. I am sure he will represent all the Branches equitably and with his usual wisdom and diligence.

My thanks also to the ESHG for having the confidence to begin this endeavour, and Jerome and his team at the VMA for their unfailing co-operation and support.

I am proud to have led the Board through this initial phase of its work. The EBMG was always about making care safer and better for patients and I am certain that the committed professionals working for the Board will ensure it continues to do so.

Heather Skirton
Chair of the EBMG

Recent Developments in Genetics and Medically-Assisted Reproduction: From Research to Clinical Applications

by Milan Macek Jr., former ESHG President and ESHG liaison to ESHRE

On September 20-21; 2016, the third interdisciplinary workshop was held in Amsterdam involving selected experts from the European Society of Human Genetics and European Society of Human Reproduction and Embryology (ESHRE.eu) to evaluate developments in relation the rapid research developments at the intersection between reproductive genetics and assisted reproduction.

The first joint ESHG / ESHRE meeting was held in Seville in 2005 and the resulting consensus paper was titled "The interface between assisted reproductive technologies and genetics; technical, social, ethical and legal issues" (Eur J Hum Genet 2006; 14(5):588). In this paper the experts discussed genetic studies in IVF couples, preimplantation genetic testing, selection of donors based on genetic information, genetic counselling in relation to assisted reproduction, quality and safety procedures, public health policies and published a list of recommendations. The second meeting was held in Brussels in 2012 and the joint paper was titled "Current medical issues in medically assisted reproduction and genetics in Europe; research, clinical practice, ethics, legal issues and policy" (Eur J Hum Genet 2013; 21 Suppl 2:S1). The paper discussed emerging issues related to cross border reproductive care, genetics of male and female infertility, genetic counseling, preimplantation genetic testing, accreditation of genetics laboratories, direct to consumer genetic testing, epigenetics, stem cells, reimbursement issues in assisted reproduction, including ethical and legal issues.

The experts from the third meeting in Amsterdam have highlighted important issues covering the impacts of expanded carrier screening, broad offer of direct-to-consumer genetic testing, voiding of the presumed anonymity of gamete donors by advanced genetic testing, advances in the research of genetic causes underlying male and female infertility, utilisation of massively-parallel sequencing in preimplantation genetic testing and non-invasive prenatal screening, mitochondrial replacement in human oocytes, and additionally, issues related to cross-generational epigenetic inheritance following in vitro fertilisation and germline genome editing. The last topic has also received concurrent attention by members of the ESHG PPPC committee and is currently under public consultation given its importance. As with the previous consensus papers, the third paper will be published in tandem in Hum Reprod and the Eur J Hum Genetics in May 2017 (both under Open Access policy) .

Photo in previous page: Milan Macek, Joep Geraedts, Aafke Van Montfoort, Wybo Dondorp, Sirpa Soini, Heidi Mertes, Inge Liebaers, Stéphane Viville, Kelly Ketterson, Guido Pennings, Karen Sermon, Luca Gianaroli, Martina Cornel, Joyce Harper, Anna Veiga, Kristiina Aittomaki, Kersti Lundin (also part of the group but not in the photo: Guido de Wert, Claudia Spits, Mike Morris and Joris Vermeesch)

Eurogentest activities 2016-2017

by Hans Scheffer, Chair EuroGentest

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 committee
(3) The Training Committee
(4) The Dissemination Committee

Historic background

EUGT has been a EU-financed (Framework 6 & 7) project for many years, initiated by Prof. Jean-Jacques Cassiman and continued by Prof. Gert Matthijs. Since two years ago EUGT has started to integrate its activities with other ESHG activities. These activities sometimes overlap, e.g. educational and training activities cover similar topics but are dealt with from a different angle. So one of the aims for this year has been (1) to make activities coherent and (2) self-sustainable.

With regards to aim (1) the different subcommittees have started to interact with several stakeholders within and outside the ESHG, e.g. the liaison between ESHG-EUGT and GA4GH.

The Quality committee has prepared an action plan in 2015. In short, also last year they interacted with several external quality assessment scheme providers including EMQN, CEQAS, ERNDIM, UK-NEQAS, CF Network, and others. The action plan includes the following issues: (a) EQA and how to deal with poor performance, i.e. EQA providers will be requested to provide an explanation and further feedback if the percentage of poor performance is greater than 10%; (b) How to deal with persistent poor performing laboratories and how to organize a governance structure. Possibly national societies or even the ESHG could play a role in this; (c) quality control in genetic counselling, and (d) quality issues relating to newborn screening.

The Guideline committee recently discussed the different types of existing guidelines and their different purposes. Practical guidelines and policy documents should be distinguished since these serve different purposes. The guidelines section of the EUGT web site has been cleaned and updated. Sandi Deans is now a member of the committee. The aim is to get guidelines endorsed by ESHG, EUGT and/or PPPC. The preparation of informed consent recommendations in collaboration with the PPPC is in progress. Since updated guidelines for CF and CFTR-related disorders are urgently needed, a best practice meeting for CF and CFTR-related diseases will be planned. Several other practical guidelines also require updates, e.g. breast cancer, SMA.

The Training committee initiated plans to develop on line training tools e.g. on bioinformatic data interpretation. An issue here is overlapping activities with the ESHG Educational committee. Preliminary discussions have been started to join forces.

The Dissemination committee continuously works on updating and possibly even redesign of the EUGT web site. Several updates have been made, announcing novel events, guidelines, resources and information availability. The committee also continued its effort to increase our visibility on social networks, predominantly using Twitter to disseminate most recent announcements on the news and information associated with EUGT. In 2016 EUGT successfully organized a Satellite meeting in Barcelona during the yearly ESHG scientific meeting, entitled: Quality assurance in interpretation and reporting in genome wide diagnostics which attracted more than 60 participants. This year the EUGT session will be integrated in the regular scientific program as an educational session.

To make EUGT self-sustainable (with a limited ESHG support) one of the possible strategies has been pursued during the last year: EUGT is included as formal partner with a budget in a recently submitted Horizon2020 project application. EUGT is proposed to be the partner dealing with quality aspects in diagnostic applications. Strategies to have some EUGT activities sponsored or paid for on a subscription basis have been explored, and a business plan will be prepared.

Report from the Executive Officer

by Jerome del Picchia,
ESHG Executive Officer

It is with great pleasure that I welcome you to the 50th Anniversary Meeting of the European Society of Human Genetics in Copenhagen, venue of the first meeting in 1967, and the 17th meeting, which I have had the honour to attend as organiser or Executive Officer.

It has been 17 extremely interesting, challenging and also rewarding years. Together with our organising team (Kristina, Mirjam, Daniela and Manfred from the VMA, our conference organiser, Jantie, Flora and Krista from Rose International, our exhibition and sponsor management, and Elmar from h82, our IT/AV consultant) without which this task would have been impossible to achieve since many years, we look forward to each ESHG conference and its individual characteristics, due to new locations, new technologies, local legislation, and new challenging specificities which we endeavour to overcome in order to make the conference as convenient and memorable as possible. Each country, each venue will show us a number of issues that we have not yet had to deal with and which we are committed to solve. This makes our job a most exciting, diverse and interesting one, and keeps us attentive and always in an engagement to learn, to enhance and to develop a "good product" even further.

Measuring your choices

Within the SPC and the organising team we have always been wondering, whether some of the decisions we have to take on a regular basis (concerning the programme, the room allocation etc.), although based on surveys or on estimations on the attendance in sessions, do indeed reflect the actual truth. It is not a big secret that those estimations on session attendance, which we collect from chairpersons and room assistants, sometimes diverge by up to 100% from each other.

Why is it so important?

Why is it so important to know how many delegates are attending one or another session during the conference? Besides the obvious usefulness in knowing which topics are currently of particular interest to our audience in order to shape the scientific programme accordingly, there is another very essential benefit to it.

One of our primary endeavours is to minimise crowded lecture halls in order to avoid your (understandable) disappointment and to improve your overall experience. The SPC thoroughly discusses the sessions and puts the expected interest in perspective with the available halls to allocate all sessions to the "hopefully" perfect room. As a rule of thumb we will be looking for a venue that has one plenary hall of at least 3,500 seats, one hall with 900-1,000, 4 halls with roughly 500, and finally 2 halls of 300+ seats. In the last years we have been living quite well with these numbers, with very few exceptions of overcrowded rooms, but on rare occasions the allocation (based on estimations) were unfortunately inaccurate.

This also means that already at the very early planning stage of the conference when screening potential venues and eventually selecting the locations, we base the requirements of the size of the lecture halls on these "statistics" or rather estimations. As you have been witnessing the ESHG meetings have grown a lot in recent years. We tripled the amount of attendees and doubled the number of exhibitors in the last 15 years. This seems like a long time, but knowing that we are currently selecting venues 6-10 years in the future, in order to get your preferred places in our preferred time of the year, this relativizes the time span. So to avoid that we overestimate requirements, and to the poentially widen the choice of venues (only a limited number of conference centres in Europe will allow us have our meetings without having to change the setup), we wanted to have a clearer picture.

The future is evidence based

Last year we found a "start-up" based in Vienna that seemed to offer exactly what we need. The system is based on counting participants with the help of a small pendant that is attached to the conference badge.

Participants will be asked at the registration desk if they accept to wear this little chip, one in three delegates needs to do so to make the count statistically relevant. The counting process in the background works via Bluetooth receptors located in the lecture halls and the exhibition and poster areas, not outside the conference centre, and no, neither will we be counting visits to the restaurant nor the smoking area... :-)

No worries

Absolutely no personal information will be collected, as the allocation of the dongle is absolutely randomised «out of a box». You may notice that they have different colours: they will e.g. distinguish young investigators from more senior scientists, and persons having a more clinical or more research oriented focus in their work, that's it. Should you have any worries, you may return the chip to the registration desk at any time.

We kindly ask you to return the tag in one of the collection boxes when you exit the congress for the last time, as they will be reused for other conferences. Thank you very much for your support in our trying to improve your experience at our meetings.

It's a complicated… association world

Those of you involved in an office of your national society will know that the rules by which a non-for profit association is bound, are often very complex and changing over time. Organising a conference in a different country every year, makes things even more complicated, as, just because you are recognised as non-profit in one country, does not automatically mean that you are in another. The ESHG is registered in Belgium, the congress organiser and association management office is based in Austria, and the entity organising the conference is a foundation in the Netherlands. The Executive is currently exploring possibilities to harmonise the ramified structure of these activities and will consult with experts to be able to propose adequate options to board and membership in the future, in order to simplify the workflow and to focus resources more efficiently. We would prefer to invest e.g. in fellowships for young researchers (over 120 have been given out this year) than in tax consulting, now required in 4 different countries on a regular basis.

Cheers

I would also like to invite you to attend the membership meeting on Sunday at 19.15 hrs in room Ancona (if you have not yet renewed your membership for this year, go and see my colleagues Andrea and Karin at the ESHG booth in the exhibition), and to the subsequent anniversary reception for ESHG members.

I wish you a very pleasant and informative ESHG 50th Anniversary meeting and a lot of fruitful interaction with your colleagues!

Jerome del Picchia
ESHG Executive Officer

ESHG

EUROPEAN SOCIETY OF HUMAN GENETICS

EUROPEAN HUMAN GENETICS CONFERENCE 2018

MiCo | Milan - Italy | June 16 - 19

EUROPEAN SOCIETY OF HUMAN GENETICS

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ESHG Course Calendar 2017

Courses co-organized and/or supported by ESHG on www.eshg.org/courses.0.html

2nd European Society of Human Genetics Training Course on Cardiogenetics
Antwerp, Belgium
June 10-13
Organisers: B. Newman, B. Loeys, J. Saenen
http://ngs.uantwerpen.be/cargen2017/

Introduction to the statistical analysis of genome-wide association studies
London, UK
July 4-8
Organisers: P. Froguel, I. Prokopenko, N. Henriquez, P. Murphy
http://www.imperial.ac.uk/school-public-health/study/short-courses/genomic-studies/

12th Course in Clinical Cytogenetics
Goldrain, IT
August 26 - September 2
Organiser: A. Schinzel

Basics in Human Genetic Diagnostics - A course for CLGs in education
Athens, GR
September 25-29
Organiser: T. Liehr
http://www.humangenetik.uniklinikum-jena.de/humangenetik_media/Bilder_Dokumente/Arbeitsbereiche/fish/Greece+2017.pdf

ESGM-ESHG Course Calendar 2017

Courses organized by the ESGM and supported by ESHG on https://www.eshg.org/100.0.html

Clinical Genomics & NGS
Bertinoro
April 30–May 5
Organisers: H. Brunner, C. Gilissen, A. Hoischen, T. Pippucci, G. Romeo, B. Wirth
www.ceub.it/default.asp?id_c=637&id=436#.WFfTmNLhAdU

6th Sardinian International
Summer School - GWAS
Pula
June 12-16
Organisers: F. Cucca, M. Devoto, G. Romeo
http://www.irgb.cnr.it/summerschool2017

Course on Eye Genetics
Bertinoro
September 24-26
Organisers: R. Allikmets, A. Ciardella, B.P. Leroy, M. Seri
www.ceub.it/default.asp?id_c=693&id=436#.WMkCnWczWUk

Conference Calendar

Selection. More meetings on www.eshg.org/633.0.html

June 2017

The 14th Graybill Conference on Statistical Genetics and Genomics
Fort Collins, USA, June 5-7, 2017
http://graybill.wolpe2.natsci.colostate.edu/

14th International Symposium on Variants in the Genome: detection, sequencing & interpretation
Santiago de Compostela, Spain, June 5 - 8, 2017
http://isv.variome.org/

EMBL Course: Whole Transcriptome Data Analysis
Heidelberg, Germany, June 6, 2017
http://www.embl.de/training/events/2017/DAT17-01/index.html

Translational Bioinformatics 2017 - Wellcome Genome Campus conference
Hinxton, UK, June 12-13, 2017
https://coursesandconferences.wellcomegenomecampus.org/events/item.aspx?e=640

July 2017

Mendelian randomization in the age of large-scale accessible genomics data
Bristol, United Kingdom July 11-13, 2017
http://www.mendelianrandomization.org.uk/

International Scientific CDG Symposium 2017 on Congenital Disorders of Glycosylation and related disorders
Leuven Belgium, July 13-14, 2017
https://gbiomed.kuleuven.be/english/research/50000622/international-scientific-cdg-symposium-2017

August 2017

Leena Peltonen School of Human Genomics
Hinxton, Cambridge, UK, August 20-24, 2017
https://coursesandconferences.wellcomegenomecampus.org/events/item.aspx?e=650

October 2017

World Congress on Genetic Counselling
Hinxton, Cambridge, UK, October 4-6, 2017
https://coursesandconferences.wellcomegenomecampus.org/events/item.aspx?e=663

Annual Meeting - American Society of Human Genetics ASHG 2017
Orlando, United States, October 17-21, 2017
http://www.ashg.org/

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Dear member of the ESHG!

We are pleased to invite you to the

Annual Membership Meeting 2017

at the EUROPEAN HUMAN GENETICS CONFERENCE 2017

Sunday, May 28, 2017 at 7.30 – 8.30 p.m.

ESHG Conference venue: Bella Center Copenhagen - **Room Alicante**

AGENDA

1. Opening by the President of the Society, Professor Olaf Riess
2. Activity of the Society 2016-2017
3. Financial Report of the Society 2016
4. Discharge of the Board Members for the year 2016-2017
5. Opening by the new President of the Society, Professor Christine Patch
6. Results of election for President-Elect
7. Results of election for Board Members
8. Membership fees 2018
9. New Membership Categories 2018
10. Site of future European Human Genetics Conferences
11. Budget proposal 2018
12. Major policy questions proposed by Board
13. Future activities

Please find the minutes of the last membership meeting in Barcelona 2016 in the restricted area:
https://www.eshg.org/members.0.html