THE EUROPEAN SOCIETY OF HUMAN GENETICS

NEWSLETTER

No. 35 - November 2020

Presidential Address

by Alexandre Reymond, President of the ESHG 2019-2020

Dear European Society of Human Genetics (ESHG) Members, Dear Friends, Dear Colleagues,

It is challenging to write a presidential report in the midst of a pandemic and its upcoming second wave. Despite the difficulties encountered worldwide and in Europe in particular, we should learn as much as possible from our current situation. In this gloomy moment, the keen observer can perceive some details that allow some optimism. The SARS-CoV-2 outbreak gave back to science, medicine and empirical data the credentials they rightly deserve. Genomicists were quick to sequence its genome, while population geneticists studied how inter-individual differences modulate the response to the infection. Likewise, years of experience in nucleic acid amplification meant that many of our colleagues were recruited for diagnosing the presence of the virus.

Within the society we had to reinvent ourselves. With only eight weeks to go, the Scientific Program Committee (SPC) under the lead of Joris Veltman and with the invaluable help of the Vienna Medical Academy went for broke! Instead of cancelling our Berlin meeting, they decided to transform the 53rd annual European Human Genetics Conference in a virtual event accessible to all for a small fee. My oh my, your response was beyond expectation! Almost all speakers accepted to prerecord their presentations and we registered a record 5561 participants from exactly 100 different countries. These registrants included a greater proportion of human geneticists from Africa and Asia than previous annual conferences suggesting that our education and training mission of the next generation of human geneticists would be better serve with a hybrid annual meeting in the future. Our post-conference survey showed that you overwhelmingly agree with the notion that our next congresses should include such a virtual component. Notwithstanding that physical presence at the meeting has the advantage of one-on-one discussions with colleagues, collaborators and friends, a hybrid event would allow attending the event in spite of economic factors, travel restrictions and/or familial and professional duties. This is why we are already planning for the "best of two worlds" and provide you with the possibility to join future meetings in persons or virtually (covid permitting). In this view, ninety-eight percent of the speakers we have invited for next year conference have accepted our invitation and consented to have their talks streamed and recorded.

Similarly, the ESHG Education committee (Educomm) under the guidance of Bill Newman is seeking new members to fulfill its mission. It is adapting to be able to provide our community in the future not only with traditional on site hands-on courses but also with more webinars and online teachings, as well as apps that facilitate access to free high quality online educational resources in genetics and genomics. For example, they coordinated with the SPC to include a « best-of-the course » session within the upcoming conference program tailored for trainees.

In my last year presidential invitation to join me and the rest of the European human genetics community in Berlin, I wrote that « I am particularly pleased to see that year after year many young human geneticists present their results and compete for our multiple young investigators awards. Their enthusiasm and passion when they take the stage is an integral part of the ESHG meeting." While this could only be virtually conveyed last June, I am particularly pleased to observe that young European geneticists are joining forces and federating themselves under the ESHG-Y label. As a "mother society" it is our duty to help them as best as we can. Hence ESHG-Y will soon become an integral part of our society and be represented by a new sub-committee. ESHG-Y aims to facilitate communication between young human geneticists in Europe and abroad. We can only agree with their statuses that mention that "coming generations are an essential force to meaningful and long-lasting changes in the development of the Human Genetic field worldwide and in Europe in particular." After holding workshops in past conferences, they will for the first time organize an educational session on "human organoids as genetic disease models" during next year conference.

I want to finish this report by mentioning our lasting collaboration with our sister society The American Society of Human Genetics (ASHG). We are traditionally organizing common building bridges sessions at our respective annual conferences to share ideas and compared our views. Just a few days ago participation of our Public and Professional Policy Committee (PPPC) represented by its chair Francesca Forzano at such a session allowed comparing American and European views on variant interpretation and opportunistic genomic screening. While there were some differences in our views, I regard them more as the proof of the dynamism of our discipline in an ever-changing world than deeply ingrained divergences between us. We are easily reconciled over our common goal to help patients and

their families.

I wish you all the best in the months to come and hope to see you in the near future either virtually or in person in Glasgow, in Montreal and/or in Capetown.

Sincerely yours,

Alexandre Reymond

The SPC during its first virtual meeting as perceived in the Offices of the Vienna Medical Academy in March 2020.

Report from the SPC Chair: Making a virtue of necessity

By Joris Veltman, Chair of the Scientific Programme Committee 2020.

Our 53rd annual ESHG conference was to take place in Berlin this year on June 6-9. By the 3rd of April it was 100% clear that there would not be a conference in Berlin and we decided to move to an online conference, to be held on the original dates for a registration fee of 100 euros. While we were all excited about this, we didn't know whether the online platform would work well, whether attendees would register and pay for this conference, whether speakers would be willing to pre-record their talks and whether sponsors would support us. We did, however, also see this as a great opportunity to try something new and imagined that many colleagues would long for a scientific conference after months of lockdown. In addition, the affordable online forum would allow attendees to join in from all over the world, many of whom would normally not consider joining our conference. In the end, most things indeed worked out very well: The online platform crashed for 10 minutes on day 1 but that turned out to be the only serious technological issue we had. Most attendees responded very relaxed and appreciated the online opportunities such as asking questions by chat and the possibility to see presentations at a later moment on-demand. In addition, almost all speakers pre-recorded their presentations, sponsors did support us and a record number of 5,561 people registered for our online ESHG (in comparison, in 2019 we welcomed 3,497 attendees at our meeting in Gothenburg, Sweden).

Yes, as expected, attendees most of all missed meeting each other, using the conference to catch up with old friends and colleagues, set-up new collaborations and network. That is something that an online conference cannot and will never fully replace. But at the same time there are many reasons why online conferences are here to stay. One of the major reasons is that online conferences allow people to attend who normally would not be able to do so because of cost, time, caring duties and/or distance. In these challenging times for science it may be the affordable option for many of us, and it has the added benefit of reducing our carbon footprint. In addition, the online platforms will rapidly improve and allow you to interact better with speakers and poster presenters, as well as with other attendees and sponsors.

For next year we are planning a hybrid model for our ESHG conference, with the option for people to attend the conference in Glasgow or online, ask questions in person or by chat, and look back presentations afterwards. A reduced number of physical attendees may be a consequence that is not necessarily negative for our attendees and our society, as it facilitates physical distancing and allows our conference to take place in more convention centres around Europe, not be limited to a few very large venues. We may also invite a number of top speakers to give online presentations instead of flying to Glasgow, allowing us to present work that otherwise perhaps would not be presented. Finally, the online platform also offers us new opportunities to organise and distribute our online courses, and work more closely together with other societies to offer genetics courses for various specialties.

In conclusion, the COVID19 pandemic has forced us all to rethink the organisation of scientific conferences and has allowed for new online opportunities to be integrated in exciting new hybrid meetings.

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L., Veltman, J.A. and Hui, L. (2020), Opportunities and challenges for international societies in the COVID-19 era. Prenatal Diagnosis. Accepted Author Manuscript. doi:10.1002/pd.5843

Report of the Executive Officer

***Jerome del Picchia, Executive Officer of the ESHG,* on behalf of the ESHG 2020.2 Organising Team**

Dear ESHG Members,

What a year. It has been the most unusual, unnerving and stressful time in my professional career. But then, who cannot say the same (or worse) about the past months? At the same time, we have never learned so much about online virtual meetings, new technologies, and personal limits (as you can clearly see in the change of my hair colour in the picture) in such a short time, and rarely were we forced to adapt to change as quickly.

We managed to successfully organise a virtual conference with 8 weeks notice, using unknown tools, which were, retrospectively still under development in quite some areas. Actually ESHG 2020.2 was the first virtual conference organised by an Austrian congress organiser (Vienna Medical Academy GmbH) and it was one of the first virtual meetings worldwide in this time, which did ask for a registration fee and was not offered free of charge.

The reason for asking for a flat fee of EUR 100 for a four-day conference, offering all talks and posters as on demand streams for another 14 days, was a necessity. Although we are very thankful to the *Messe and City Cube Berlin*, who were extremely cooperative in accepting to postpone the conference to 2024 without actual cancellation fees, ESHG would still have had to cover a high six-digit amount of non-refundable costs in case of a cancellation of the event. So the choice to go for an online meeting and asking a low registration fee, to attract as many delegates as possible, seemed natural, aiming to decrease the loss carried forward as much as possible.

And… it was indeed possible to scratch the black numbers, thanks to over 5,500 participants from 100 countries who registered for the conference - an unexpected high number. Thank you all for your confidence in this experiment.

It is equally time to thank the SPC for their fantastic contribution to this success, by amending the programme to a format more suitable to an online meeting on such short notice and by being available as moderators of all sessions and developing a congeniality and showing improvisation skills, which gave us all the feeling of "being there", despite some technical issues here and there.

A massive thank you also to all speakers and poster presenters who agreed to learn new presentation ways in such a short time and to deliver brilliant presentations at our meeting.

Last but not least, huge thanks to the incredible staff members Oscar Pacheco, Sarah Pirklbauer, Kristina Libova and Julian Piskula for their exemplary personal commitment in such a difficult sitation.

What have we learned from this experience? A lot I must say, and not only about how to organise a virtual meeting, but also about what you as our audience, are expecting from us for future ESHG meetings, no matter if taking place during a pandemic or not. Based on the very explicit feedback gathered via our post-conference survey, it has been decided that all future ESHG conferences will carry online/streaming elements, making them so called "hybrid" conferences, where live talks will equally be streamed as live and on-demand streams to registered participants around the globe. We have seen the potential to reach new groups of scientists, who were unable to attend our conferences in the past on a regular basis but are interested in attending online. ESHG will cater to this need.

While we can currently not yet say that we can see an end to the pandemic and a normalisation in the educational and conference sector, we can assure you that we are planning for any eventuality in the future.

Finally, for those who missed talks in previous meetings, I would also like to remind you that most Plenary and Educational Sessions from the ESHG conferences from 2014 to 2020 are still available on our YouTube channel or on the ESHG Website: www.eshg.org/index.php?id=850

In the meantime, please stay safe!

The ESHG Education Committee (EduComm) is seeking new members.

We are seeking members who express to take a role with responsibility for:

a) Informatics - e.g development of apps and other resources

b) Online resources - webinars, online courses etc

If you are interested, visit the following page for more details:

www.eshg.org/index.php?id=117

Report from the Public & Professional Policy Committee

By Francesca Forzano, Chair of the PPPC

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

It addresses these issues and provides guidance through background documents, policy statements, recommendations or other publications, and participating in the public and professional debate around these issues.

The Committee collaborates with a range of other societies and organisations such as ASHG-Social Issues Committee, ESHRE and the ELPAG group.

Activities in the pipeline

Recommendations on Opportunistic genomic screening. The final document has been produced and published on the ESHG website for membership review in April 2020. After integration of the members' comments and suggestions, it will be submitted to the Board for review and potential approval in June 2020.

The GDPR: what does it mean in practice for our genetic community?

The PPPC together with the EuroGentest Quality subcommittee is working on a document analyzing the impact of the GDPR on genetic practice and research, and hopefully offering some pragmatic guidance on the do's and don'ts. We aim to have a draft document ready by the end of 2020.

Human Gene editing

In December 2018 the WHO has set up the Expert Advisory Committee on Developing Global Standards for Governance and Oversight of Human Genome Editing to examine the implications and challenges of genome editing, both for somatic and germ cells. The Committee is in the process of developing a governance framework and to serve this aim, it has opened a consultation via an online Questionnaire, which closed on February 10th 2020. The PPPC has sent its contribution.

Post-mortem Genetics

Martina Cornel has represented PPPC and ESHG in the development of the 2020 APHRS/HRS Expert Consensus Statement on Investigation of Patients with Sudden Cardiac Arrest and of Their Families (APHRS= Asia Pacific Heart Rhythm Society; HRS= Heart Rhythm Society). The document will be released shortly.

An article was published to highlight ESHG's views on postmortem use of genetic data for research ESHG PPPC Comments on postmortem use of genetic data for research purposes. Fellmann, F., Rial-Sebbag, E., Patch, C. et al. Eur J Hum Genet 28, 144–146 (2020). https://doi.org/10.1038/s41431-019-0525-z

Future activities

Workshop Genomics and Inequity

The aim of this workshop is to take stock of the potential difficulties arising from genomics, especially where they may exacerbate existing societal problems and cause difficulty or harm for the more vulnerable members of society. We aim to establish a frame within which these potential problem areas may be examined. We will then prioritise the set of issues and - following the workshop, as part of the ongoing activities of the PPPC)- engage professional organisations and other stakeholders in debates about how to address these. The longer-term aim will be to establish a strategy for minimising the potentially harmful and/or inequitable effects of pursuing genomic medicine and enhancing its benefits for underserved and otherwise marginalised groups.

Reporting in prenatal setting

Together with the Eurogentest Committee, we aim to work on a document which will offer guidance on how best to report genetic and genomic tests in the prenatal setting.

Further topics under consideration are: Fetal personalized medicine, Contacting the family after the death of a known patient, and an update of our previous PPPC document on Testing in Minors.

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

ESHG members interested in becoming a PPPC member are requested to send their CV and a letter of interest stating their expertise and motivation for joining to the Chair.

Members of the PPPC in 2019-2020 were Angus Clarke, Christophe Cordier, Martina Cornel (co-Chair), Guido de Wert, Florence Fellmann, Francesca Forzano (Chair), Sabine Hentze, Heidi Howard, Hülya Kayserili, Béla Melegh, Alvaro Mendes, Markus Perola, Dragica Radojkovic, Emmanuelle Rial-Sebbag, Vigdis Stefánsdottir and Carla van El (Secretary-general). Dr. Fiona Ulph is currently a PPPC collaborator

Report of the Education Committee (EduComm): ESHG has a mission to promote the practice and knowledge of Human and Medical Genetics in Europe.

By William Newman, Chair of the EduComm

In January I took on the role of Chair for the ESHG Education Committee. This is a great privilege for me and I hope to build on the great work done by my predecessor, Prof Han Brunner, who has led this Committee for the past three years. Han has overseen a significant increase in the portfolio of courses that the ESHG now offers. These courses have been incredibly successful and provide a great environment for junior researchers, clinicians, counsellors and scientists to mix with experts in a range of genetic disciplines. Han, as in everything he does, has brought energy, a clear vision to the EduCom and an altruistic desire to promote genomics and the next cadre of researchers, clinicians and leaders across Europe.

The Coronavirus pandemic has meant that a number of courses that were planned for 2020 have had to be cancelled or postponed. We hope soon to be able to provide an update on the ESHG website about plans for rearranged courses. We are still keen to receive applications and ideas for future courses that will be of interest to the membership.

The pandemic also led to the cancellation of the DNA Day Essay writing competition for 2020 but we plan to open for applications for the 2021 competition on the website in September.

As we have encountered lockdown across Europe, all of us have had to become more familiar with on line resources and remote ways of learning and interacting. We are keen to explore ways that we can expand our educational offering through webinars, MOOCs or other mechanisms.

Linked to this I urge you to visit the Education Section of the ESHG website https://www.eshg.org/index.php?id=education and especially then to move to the wonderful EuroGEMS site https://www.eurogems.org that Prof Ed Tobias has invested so much energy into. This site has now been accessed by individuals from over 100 countries world wide and provides excellent links to all types of genetic information and sites. There are plans to translate some of the content of this website into other languages (initially Spanish, Russian and Ukrainian) and are keen to hear from members as to which other languages you feel would be most useful.

We are happy to be working with the Scientific Program Committee to input into the educational sessions for the Conference in Glasgow next year and future Conferences thereafter. We are keen to see how we can develop further workshops or educational meetings as satellites to the main meeting that will reduce travel expenses for members who will be attending the meeting anyway and can combine this with a specific educational activity.

Many thanks to Domenico Coviello who has stepped down from the Committee and led the work for the Schoolchildren's event which linked to the Conference and has been a real highlight. We hope to have an event in place for Glasgow 2021.

The current members of the EduCom are Inga Prokopenko and Philippos Patsalis (Teach the teacher course), Ed Tobias (teaching materials website), Christophe Cordier (DNA day essay competition), Johan den Dunnen (HGVS course organization), and Han Brunner (Course portfolio). We are currently drafting a Terms of Reference for the Committee and will be looking for additional members to contribute to our work (see website and emails for details).

We have lots of plans for developing the educational activities and profile of the ESHG and look forward to updating the membership with news of these initiatives. If you have any ideas/comments regarding the work of EduCom please contact me at william.newman@manchester.ac.uk.

EJHG Highlights 2019

by GertJan van Ommen, Editor in Chief, EJHG

Impact Factor and visibility of EJHG

In 2019 our impact factor (citations measured over 2017-2018) has gone up a bit again but not by much, from 3.636 to 3.650. The most important event for our visibility as of 2019 is the appointment in November 2018 of a Social Media Editor, Alisdair McNeill from Sheffield, who achieved the truly astonishing fact of getting ~2500 Twitter followers in the 5 first months of his activity, from December 2018-April 2019. The expectation is that this spring we will round the 4000 followers! Elsewhere in this Newsletter Alisdair will highlight his advances more in detail. But let me also stress that the best way to increase our impact remains that our frequent authors submit some of their best research to EJHG.

Reviewing and time to publication.

In 2019, we received 654 submissions, against 704 last year, once again 7% less. This continuing the ongoing trend in slowly decreasing submissions for 5 years, probably due to increased competition. Strikingly, however, we are currently witnessing a 19% upturn in the first four months, with 52, 53, 65, 74 submissions in Jan-April 2020. This may be due to more people getting tuned in by Twitter, but it may also be due to the Covid-19 lockdown: people are finally rounding up close-to-ready material which they now find the time for!

It'll be interesting to see how these fare in reviewing – and, for that matter, if perhaps our reviewers also find more time to review papers, wouldn't that be wonderful!

The geographic origin of the accepted papers is still predominantly European (79%), with USA and Canada (8%) in second, and then equal contributions of Japan and China and Southeast Asia (each 4%).

Our acceptance rate for 2019 has been 33%, like in 2018. On the other hand, the average time to first decision after review, substantially fell to 70 days from the 79 of last year. Papers which are not reviewed are on average returned within 11 days, like last year, which we should do faster but is partly due to the organizational mechanics of the EJHG. As to overall review time, I explained last year that what is often – and understandably – perceived by authors as 'THE review time', is actually a composite of the reviewer allocation - and acceptance! – time, and the actual review times. Indeed, our actual review times have kept stable over the past 5 years at 14-15 days. It just has become increasingly difficult to get 2-3 reviews in. This sometimes requires us to approach 6-10 reviewers! In this respect, the good news of 2019 was that the time between manuscript receipt and securing of the last reviewers was reduced from 30 to 20 days!

Using new procedures, online publishing time after acceptance has fallen substantially, to 19 days from 29 days in 2018! The average time to print publication, typically around 3 months, now tends to decrease a bit with the reduced submissions.

Web visibility

Website views in 2019 rose by 9% to 1,009,560 from 925,626, first time hitting a million, with full text views even rising by 16%, to 872,280 from 754,127. The top three downloads are:

1. *Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database*, by Stéphanie NGUENGANG WAKAP et al., DOI 10.1038/s41431-019-0508-0, with 6,056 views

2. *Mitochondrial DNA variability of the Polish population*, by Justyna Jarczak et al., DOI 10.1038/s41431-019-0381-x, with 4,314 views

3. *The GDPR and the research exemption: considerations on the necessary safeguards for research biobanks*, by Clara Staunton et al., DOI 10.1038/s41431-019-0386-5, with 3,694 views.

In addition, to get a sense of our broader web visibility, as of last year we are also monitoring the public's interest by the Altmetrics score. This is a qualitative and quantitative metric complementary to traditional, citation-based metrics, including (but not limited to) peer reviews on Faculty of 1000, citations on Wikipedia and in public policy documents, discussions on research blogs, mainstream media coverage, bookmarks on reference managers like Mendeley, and mentions on social networks such as Twitter and Facebook.

The three Altmetric top scoring papers of 2019 are:

1. Like at the web downloads, the highest scoring is '*Estimating cumulative point prevalence of rare diseases: analysis of the Orphanet database'* by Stéphanie NGUENGANG WAKAP et al., DOI 10.1038/s41431-019-0508-0, with an Altmetric score of no less than 577 (high scoring papers in our category usually get up to 50-80)!

2. '*People from Ibiza: an unexpected isolate in the Western Mediterranean'*, by Simone Andrea Biagini et al., DOI 10.1038/s41431-019-0361-1. AS 79.

3. '*A feline orthologue of the human MYH7 c.5647G>A (p.(Glu1883Lys)) variant causes hypertrophic cardiomyopathy in a Domestic Shorthair cat*' by Tom Schipper et al., DOI doi.org/10.1038/s41431-019-0431-4. AS 74.

Just as a reminder: in 2017/18 we upgraded the design of the EJHG web page. This is now completed. You will find several new links relaying you to 'Featured articles of the most recent issue', to the 'Editor's choice' from recent key papers, freely accessible available for one month, containing a whole slew of highly downloaded and cited papers from the last two years, and finally a link to our recent Twitter activities.

EJHG Award

As every year, EJHG, and Springer/Nature jointly offer a junior authors' high-citation award. This is given to the top-3 papers published in 2018, with citations counted in the 12 months following print publication. The 1st prize includes a € 500 award and positions 1-3 receive one year free ESHG membership, including an online EJHG subscription, and free registration for the next EJHG meeting. This year there is a shared 2nd position. Since this year there was no physical meeting, the winners of this year have been virtually honoured in the online meeting, but their free access will be valid at the next year's meeting. The winners are:

1. Eliana Marisa Ramos. Semel Institute for Neuroscience and Human Behavior, UCLA,

Department of Psychiatry and Biobehavioral Sciences. Los Angeles, CA 90095, United States.

Primary brain calcification: an international study reporting novel variants and associated phenotypes. EJHG 26-10, 10.1038/s41431-018-0185-4. 16 citations

2. Ondrej Pös. Department of Molecular Biology, Faculty of Natural Sciences, Comenius University, Bratislava, Slovakia.

Circulating cell-free nucleic acids: characteristics and applications. EJHG 26-7, 10.1038/s41431-018-0132-4. 11 citations.

2. Mélanie Rama. Centre Hospitalier Régional Universitaire de Lille, Hospital Jeanne de Flandre, Lille, France. *A decision tree for the genetic diagnosis of deficiency of adenosine deaminase 2 (DADA2): a French reference centres experience.* EJHG 26-7, 10.1038/s41431-018-0130-6. 11 citations

Report on work as Social Media Editor for EJHG December 2018 - December 2019

By Alisdair McNeill, Senior Clinical Lecturer in Genetics, the University of Sheffield.

I genuinely enjoy the role of Social Media Editor; it provides me with an opportunity to engage more deeply with the latest genomics research and make contacts with other researchers and discuss their work. I believe the management of the social media platforms is working well.

I would be very happy to continue in this role by mutual agreement with ESHG/EJHG.

Social media strategy goals 2019-2020

1. Establish a social media presence for EJHG

2. Increase "reads" of EJHG paper using altmetrics as an indicator

3. Improve engagement of readership with EJHG material

Social media successes

Since establishing the EJHG twitter profile we have gained almost 3 500 followers, and add around 100-150 followers per month.

The Twitter journal club (a discussion on a paper from a given monthly edition of EJHG) is successful in stimulating online discussion of papers and increasing the visibility of EJHG.

The adoption of our social media strategy has increased the online visibility of EJHG papers. For Dec 2017- Dec 2018 (prior to setting up a social media presence) mean altmetric score for EJHG papers was 6.5 (+/-13.3). For Dec 2018-Dec 2019 (the first 12 months of our social media strategy) the mean altmetric score was 11.8 (+/-31.1, t-test p=0.02) [the wide confidence interval reflects 2 very high scoring outliers]. Given that the Dec 2017-Dec 2018 papers have been published for longer, and therefore had longer to gather altmetric scores, this underestimates the attention being attracted by our social media strategy.

Areas for improvement

The Facebook page receives little attention but I believe this reflects the fact that this platform is used for personal rather than business purposes in general.

According to Twitter analytics the majority of our followers are from the UK/Western Europe or North America. It would be valuable to increase the number of our followers from Australasia and Asia/other "emerging" scientific economies.

We need to evaluate if there are any new social media platforms we should use, but at the moment I feel it is best to exploit our success with twitter.

We could try and produce less formal and more "engaging"/ fun content for twitter. For example, our tweet about MYH7 in feline cardiomyopathy had a strong altmetric score. We could consider tweeting around Christmas or Eid with appropriately themed free online content (e.g. 1-month free access to certain papers as a "gift")

Plan for Dec 2019 – Dec 2020

1. Continue to live tweet from conferences as this has increased our followers on twitter and interactions

2. Try to schedule tweets for times when twitter activity is highest (e.g. Saturday night)

3. Consider more formal social media audit to compare our activity with that of similar genomics journals

4. Assess impact of social media strategy on citations (e.g. any correlation between intensity of social media promotion and citation rate?)

5. We could discuss if the publisher wants to produce social media guidelines for authors (e.g. how they can help us to promote their paper by providing twitter handles, helping with video abstracts and journal clubs).

Annual report ESHG-EuroGentest Committee 2019-2020

By Gert Matthijs, Chair of EUGT

EuroGentest, that started as a project funded by the European Commission in 2005, is an integral part of ESHG since 2013. ESHG-EuroGentest focusses on quality aspects for genetic services, prepares professional guidelines related to genetic diagnostics, provides training and represent the society at international forums related to genetic services. For the time being, the work is coordinated by

a relatively small group of colleagues, but we rely on experts and representatives from different stakeholders whenever action is to be taken.

As you will read in this report, there is a lot to be done for the clinical genetics and counselling services and for the genetic laboratories. Hence, I start by launching a call for volunteers to join ESHG-EuroGentest, to allow us to further broaden the scope of our activities and gain strength and impact.

Most active is the Quality Subcommittee that directly deals with issues that affect quality and competence of the diagnostic genetic and genomic laboratories, clinical genetics and genetic counselling. Multiple workstreams have been progressed throughout the year. To promote harmonisation and push towards accreditation are the prime targets. Quality is an important element in the end to end processes of patient care and the committee works on issues that affect quality and competence of the diagnostic genetic and genomic laboratories, clinical genetics and genetic counselling clinics.

The Quality Subcommittee is preparing a Clinical Genetics Service quality questionnaire and would encourage members to complete this when available, and the annual review of laboratory performance data is underway.

The Quality Subcommittee is scheduling a 'National performance monitoring workshop for genomic & genetic testing laboratories' that will take place in September 2020 – fingers crossed against covid-19 interference of our plans! The aim of workshop is to promote the importance of regular monitoring of genomic testing laboratories performance, and to provide guidance on the implementation of national performance monitoring mechanisms to

ensure high quality genomic testing and patient safety. The latter is a delicate issue: neither the EQA providers no ESHG have a legal authority towards 'chasing' poor performance. This is a matter that belongs to the national competent authorities. ESHG-EuroGentest can highlight the importance of national surveillance. Thus, it is important to share challenges and experiences of countries with a national performance monitoring process in place, being implemented and not currently available.

ESHG-EuroGentest Quality Subcommittee also recognises that bioinformatic analysis plays an integral part in the delivery of genomic testing and would like to invite expressions of interest from individuals with expertise in bioinformatics to join one of the EuroGentest Committees.

I sincerely thank Sandi Deans and the members of the Quality Subcommittee for their engagement and effective contributions to the activities of ESHG-EuroGentest. Early last year, Luca Lovrecic has joined the Quality Subcommittee, bringing additional expertise in clinical genetics and genomic analysis. Welcome!

Reflecting the changes within genomic medicine EuroGentest has been involved in scoping the impact of the medical in vitro devices regulation (IVD-R), guidance around opportunistic screening (in collaboration with the PPPC Committee) and has identified the requirement to update the ESHG Best Practice Reporting Guidelines.

The IVD-R represents a major challenge – let's frankly call it a threat – to genetic diagnostic laboratories. The IVD-R that is or will be translated into national law, imposes the use of CE-IVD marked kits from May 2022 on. Thus, the instruction is simple: if a CE-IVD kit is available on the market, is has to be used. In other words, the use of laboratory developed tests (LDT) or non-CE-IVD marked kits is prohibited when CE-IVD kits for the same application are commercially available. This is applaudable, if it contributes to quality of testing. Also, if no such kit is available, the use of LDT is still allowed. This is also true if the laboratory proves that its LDT performs better according to state-of-art or is better suited for a specific patient population. In this situation, the laboratory has to be accredited. We are happy to mention that the latter exemption is an achievement of EuroGentest. However, the IVD-R will lead to worrisome situations. For instance, if an CE-IVD kit performs less well than an LDT, the burden to provide the evidence that the CE-IVD marked kit is not as good (or even bad) lays with the users i.e. the genetic laboratory. Or, the CE-IVD kit may not include the most representative internal standards, etc. Most importantly, the creating of new LDT and improve diagnostics is discouraged: why invest - in the genetic diagnostic lab - in development knowing that, as soon as the same application becomes commercially available as a CE-IVD marked kit, it has to replace the LDT? One could take the use of whole genome sequencing (WGS) using different 'research use only' and locally developed components or bioinformatics pipelines as an example. Vice versa, what shall happen if the CE-IVD kit disappears from the market? To try and influence the practical interpretation of the IVD-R, members of EuroGentest are active at different levels. For instance, Els Dequeker represents ESHG-EuroGentest in a European expert panel related to the implementation to the IVDR.

Under the impetus of Hans Scheffer and Gunnar Houge, the ESHG-EuroGentest Guidelines Subcommittee launched the revision of the guidelines for genomic diagnostic testing, to extend its remit to WGS applications in the clinic. To do so, EuroGentest is joining forces with experts from three European projects that feature guidelines for WGS as a deliverable: EASI Genomics, Solve-RD and B1MG (Beyond 1 Million Genomes). A workshop with the different stakeholders took place in September 2019. A first draft of the new guidelines has been written. I should not linger too long to finalize it and submit it to the ESHG membership for approval.

Finally, I want to sincerely thank Hans Scheffer for leading the ESHG-EuroGentest Committee and coordinating its activities for the past 5 years. Hans has been involved in EuroGentest since it was launched in 2005. His track record in quality in genetic testing and his generous contributions to training in laboratory genetics, EQA (External Quality Assessment) and is authoring guidelines dates back many more years. Hans has retired from his job earlier this year and thus also left the ESHG-EuroGentest Committee.

Dear Hans, we've been joining forces for so many years, our collaborations have been effective and our interactions

heart-warming. Even if you may not read this newsletter while enjoying life in your hometown near Groningen, I wish you the best on behalf of the entire team.

In 2019, I took over the lead of the ESHG-EuroGentest Committee. Admittedly, I have been a bit slow in putting this in practice. However, I am ready to strongly commit again. I start by acknowledging the work of the colleagues who have voluntarily contributed to EuroGentest-ESHG achievements and output over the last year. Like for all other activities within ESHG, we rely on the commitment of individuals that are willing to invest their time and effort for the genetic community. Please contact us if you wish to join us in this great endeavour!

Report from the European Board of Medical Genetics

by Ulf Kristoffersson, Chair of the EBMG 2019-2020

The EBMG was set 2014 up to serve the needs of patients who use genetic services in Europe through ensuing good standards of practice in three professional branches:

- Clinical laboratory geneticists (CLG)
- Genetic nurses and genetic counsellors (GNGC)
- Medical genetics and genomics (MGG)

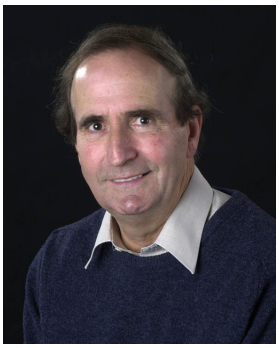
The aim of the EBMG is the establishment of professional standards of education, training and practice in human and medical genetics and genetic counselling, by developing and administering systems of certification and/or re-certification for professionals working as specialists in genetic healthcare in Europe.

The day to day work of EBMG is managed by the branches. An executive board consisting of 2 representatives from each branch, a General Secretary and a Treasurer meets monthly at a teleconference. President this year has been Ulf Kristoffersson from the MGG branch.

The CLG and GNGC have established a certification programme renewable each five years, and this year the GNGC branch offered their second renewal with 10 successful candidates and 11 new certificates issued. The medical professionals have the national licensing system for medical specialists in medical genetics. Their section within UEMS (European Union of Medical Specialists) have in collaboration with MGG launched a European specialist exam last year with a planned second round this year which, due to the circumstances, have been postponed until next year. The MGG branch has an ongoing work to launch a CME (continuous medical education) programme available for medical specialists from all countries.

ESHG and the History of Human Genetics: What Have We Achieved? What Remains to be Done?

by Peter S Harper, Institute of Medical Genetics, School of Medicine, Cardiff University, Cardiff, UK. (HarperPS@Cardiff.ac.uk)



The ESHG board has asked me to write this farewell note since, having now passed the age of 80, I have decided to finish working on the history of our field, and to hand on the responsibility for its continued documentation to the human genetics community collectively and individually. It is a pleasure for me to write this piece, since ESHG has been enthusiastically involved and supportive since the initiative to preserve and document the history of human and medical genetics started almost 20 years ago.

Beginnings

At the turn of the present century a number of us were becoming aware that as the original founders of the field were retiring or passing on, there was a very real danger that the story of how human and medical genetics began and has developed across the world was in danger of being irrevocably lost. Hence, after discussions at the 2002 ESHG meeting in Strasbourg, I decided to launch the Genetics and Medicine Historical Network, as a loose framework for bringing together and encouraging the various people interested or already involved. I am sometimes asked what exactly is the Network? The answer is that it is no more than what it does; it has never been formally constituted, yet it has managed to achieve quite a lot over the past two decades. I shall try to summarise some of this here.

International Workshops

These have formed one of the most satisfactory outcomes of the Genmedhist initiative; from the outset they have been run as satellite meetings to ESHG conferences and supported by the Society, often with financial help also from Wellcome Trust. They have helped to fulfil one of the primary aims - to bring together human geneticists, whether scientists or clinicians, with historians and other workers from the humanities, and to stimulate collaborative studies between these two very different worlds.

The first workshop was held in 2003 in Birmingham (UK), the second at Mendel's abbey in Brno in 2005 (Figure 1), and they have now reached a total of seven (Table 1), with the eighth due this year in Berlin. With hindsight it would have been good to publish the papers from each conference as separate volumes, as historians normally do, but this would have involved a lot of work and time; it might also have affected the spirit of spontaneity which has been a

hallmark of the series. Fortunately Heike Petermann has now produced a valuable book which has brought together many of the contributions from different workshops(1).

The Genmedhist Website and newsletter

Even back in 2003, when websites were not universal, it was clear that a dedicated website was the best way to link the variety of people and activities that was evolving. The www.genmedhist.org website was originally unveiled at the first Network workshop in Birmingham and, particularly since I myself have no expertise in the area, this is an appropriate point at which to thank the successive experts who have been involved in creating and maintaining it, notably Jeff Alderman in Cardiff and Stefan Foedermeyer in Vienna.

For the first 12 years the website was hosted by Cardiff University, but in 2016 it was transferred to the ESHG office, which continues to look after it (https://genmedhist.eshg.org). Now it needs more involvement from Society members to make sure that it continues to develop and to reflect recent activities in the field.

Alongside the website, the early years saw an informal electronic annual newsletter giving details of coming events and articles on a variety of topics. It was always intended that this should be absorbed into the website, but in fact it continued until 2014 (18 newsletters in all), with much valuable content edited by Judith Friedman (Canada) and others. All the newsletters - the initial ones primitive, the later ones quite sophisticated - can be viewed on the website (www.genmedhist.org/newsletters) .

Recorded interviews with older human geneticists

This is the activity of the Network that I have been most involved with personally and I am happy that it has turned out so well, despite my total inability to get any funding support for it. I managed to undertake just over 100 recorded interviews between 2003 and 2014, for almost all of which the transcripts are on the website (www.genmedhist.org/interviews). The interviews cover workers from a wide range of European countries, with an inevitable emphasis on Britain. Some North American workers are included too, though I did not try to be comprehensive here, especially as a comparable programme had started there; sadly this was discontinued after funding finished - a problem I was spared through not having had funding in the first place! I am also pleased that several initiatives are now in progress for individual countries; that for Norway is already on the website, while others for Finland, Germany and the Czech Republic are ongoing. ESHG has offered to help financially so as to allow English translations to appear on or linked to the genmedhist website. I should say here that making this series of interviews has given me great personal pleasure, as well as reinforcing my respect and admiration for those who played a large part in founding human and medical genetics across Europe. I owe a great deal to all those whom I interviewed for their generous welcome; equally I think that they valued the opportunity to tell their stories and to feel remembered.

Figure 1

Figure 2

Time moves fast though; already another generation of workers is retiring, mostly people who I considered 'too young' when I was making my series. There needs to be a rolling programme of interviews, probably organised on an individual country basis, but hopefully linked with ESHG too.

I have always looked on the interviews as a permanent resource which others can build on for more detailed historical studies; I have been pleased to hear from a number of workers that they have indeed done this and have found the transcripts useful. The original sound recordings and primary transcripts, along with much background material, are all archived as part of Cardiff University's special collections and archives. I have myself used the interviews as the core of several papers that attempt to synthesise some of the main themes arising in human genetics, and the patterns of development in different countries, including the history of ESHG itself (2-6), but there is much material that can be used by others, too. A particularly valuable link has been with the extensive series of Witness Seminars, organised and produced as a scholarly resource by Professor Tilli Tansey.

Books: the Human Genetics Historical Library

Books are no longer the chosen medium for transmitting original scientific findings, but during the earlier years of genetics the situation was very different and even today the printed book has many advantages to both reader and author for synthesising and reflecting on both broad themes and specific topics. There has been a tendency, though, throughout science, for older books to be considered worthless, in general as well as financial terms; many libraries throw them out, even when their historical significance is clear.

I had been aware of this for many years, but was fortunate to be able to form links with our Cardiff Medical School librarian, Steven Humphries, who was forming research collections; hence the Human Genetics Historical Library, Cardiff based and curated, but with contents searchable on the Web and with contributions now from different European countries, notably Finland and Sweden, as well as the complete scientific libraries of UK workers such as Paul Polani and John Edwards. Now approaching 4000 volumes, this is also a valuable repository for items such as informally published or unpublished reports, CD and

videodiscs, and similar material. We are fortunate that it has had the same librarian curator from the outset, Karen Pierce (contactable at piercek@cardiff.ac.uk). Donations of books not already in the collection remain welcome, though we may not be able to collect them as in the early years! Figure 2).

Although a number of different people have written historical books on genetics, with a few giving space to human genetics, most professional historians have preferred to focus on eugenics, so it seems reasonable to list those with which I have been associated myself (Table 2), all of which have had human and medical genetics as their remit, with much of their content based on my interview series and other activities of the Genetics and Medicine Historical Network. In them I have tried to let the workers in this community speak for themselves as far as is possible, rather than to promote any particular viewpoint of my own, but I hope that I have managed to bring together and put down simply a number of aspects of the story of our field which might otherwise have remained unknown to others, especially the younger generations.

Other activities

The existence of Genmedhist has stimulated a number of other activities, in addition to those for which it has been directly responsible. The holding of historical sessions at successive five yearly International Congresses of Human Genetics, as well as at the American Society of Human Genetics, was encouraged by the specific series of ESHG- linked workshops that I have described above. The preservation and archiving of the scientific records of key workers, many identified through the Genmedhist recorded interview programme, has encouraged other bodies, notably Wellcome Trust, to devote resources to this for the broad field of human genetics, rather than just for the Human Genome Project. Again, these records are becoming the starting point for in depth studies by historians and archivists, which would have been impossible if the original records had not been preserved.

Looking ahead

I have always felt that the post-world war 2 half century of human and medical genetics has been a 'golden age' both scientifically and medically and, like most others in the field across the world, I have felt privileged to be able to play a part in it. But preserving the collective memory of this remarkable period requires active effort from its members if its achievements are to be permanently remembered; I realised early on that one could not rely on professional historians to make this happen, but that the facts and foundations must be laid by those of us working in human and medical genetics ourselves, preferably in close contact with historians.

Now that a reasonably full, though far from complete, oral and written record has been made for these early years, a comparable effort is needed to document what people of my generation still think of as 'recent' discoveries and developments, particularly but not exclusively those associated with genomics. The approaches that have proved valuable for earlier years, such as recorded interviews with workers, the archiving of written records and close links between laboratory and clinical scientists and those in the humanities, will all remain valuable, though the preservation of digitally based information is a challenge.

Whether this challenge will be fully met will largely depend on whether the small but active community created by the Genetics and Medicine Historical Network can continue to grow and to involve younger workers; European Society of Human Genetics is well placed to continue and to strengthen its support for this, both professionally through its members and through its organisational base. The Executive has agreed to identify a person able to act as the contact point for the future. So in stepping down from my own efforts over the past 20 years, I should like not only to thank ESHG and its members, both collectively and individually, for their help and friendship during this time, but to wish it well in meeting the challenge of ensuring that the recent and future history of our field across Europe and the entire world is put fully on record for the benefit of Science, Medicine and us all.

Table 1

Genetics and Medicine Historical Network/ESHG Workshops on Genetics, Medicine and History

Year	Place	Principal Theme(s)
2003	Birmingham (UK)	Launching the Genetics and Medicine Historical Network
2005	Brno	Preserving the history of Medical Genetics
2008	Barcelona	Genetics, history and public understanding
2010	Gothenburg	Early history of human molecular genetics
2012	Nuremberg	The biological future of man: continuities and breaks on the history of human genetics, before and after 1945
2015	Glasgow	History of human gene mapping; oral history of human genetics
2017	Copenhagen/ Lund	50 years of human genetics in Europe: discoveries, challenges and the foundation of ESHG
2020	Berlin	History of eugenics; the evolution of techniques in human genetics (postponed)

Table 2

Books by Peter Harper and Colleagues Based on the Work of the Genetics and Medicine Historical Network

2004 Landmarks in Medical Genetics: Classic Papers with Commentaries. New York. Oxford University Press

2006 First Years of Human Chromosomes: the Beginnings of Human Cytogenetics. Oxford. Scion Press

2008 A Short History of Medical Genetics. New York. Oxford University Press

2010 Harper PS, Reynolds LA, Tansey EM (2019) (Eds). Clinical Genetics in Britain: Origins and Development. London; Wellcome Trust Centre for the History of Medicine, London.

2017 Petermann H, Harper PS, Doetz S. (Eds). History of Human Genetics: Aspects of its Development and Global Perspectives. Heidelberg. Springer

2020 The Evolution of Medical Genetics: a British Perspective. Oxford. CRC Press.

References

1 Petermann H, Harper PS, Doetz S (2017). History of Human Genetics: Aspects of its Development and Global Perspectives. Springer.

2 Harper PS. (2018). Conversations with French medical geneticists. A personal perspective on the origins and early years of medical genetics in France. Clin Genet. 94:115-124.

3 Harper PS (2017).Human genetics in troubled times and places. Hereditas. 155:7.

4 Harper PS (2017).Some pioneers of European human genetics. Eur J Hum Genet. 1-18

5 Harper PS (2017). The European Society of Human Genetics: beginnings, early history and development over its first 25 years. Eur J Hum Genet. doi: 10.1038/ejhg.2017.34.

6 Harper PS (2016). Recorded interviews with human and medical geneticists. Hum Genet. 2017 Feb;136(2):149-164. doi: 10.1007/s00439-016-1744-9. Epub 2016 Nov 15.

Figures

1 Participants in the Second International Workshop on Genetics, Medicine and History, held in Mendel's abbey, Brno, May 2005.

2 The Human Genetics Historical Library at an early stage: collecting books donated by the Oxford Clinical Genetics Unit.

ESHG Board Members 2019-2024

Prof. Lina Basel-Salmon, MD, PhD

I was born in Vilnius, Lithuania, and moved to Israel at the age of 24. I am a pediatrician and a medical geneticist with almost twenty years of clinical experience. My main areas of interest and research include: 1) dysmorphology; 2) identification of new gene-disease correlations; 3) carrier screening; 4) use of artificial intelligence in variant interpretation. Research in our lab so far led to the identification of more than twenty new gene-disease associations. As a result of these discoveries, preconceptional screening programs in at-risk communities with high frequency of carrier status and a high disease burden have been established.

My recent research explores the role of artificial intelligence-based platforms in the interpretation of exome sequencing results, and the role of clinical geneticists in variant interpretation of their patients. I believe that clinicians should play a more active role in variant prioritization by using next-generation genomics intelligence platforms in genetics departments.

Ana Beleza

I feel very honoured to be part of the ESHG board. My name is Ana Beleza and I am a clinical specialist in Medical Genetics since 2013, although I work in Genetics since 2003. I completed my clinical training in Portugal. However I was very fortunate to be a PhD student at the Karolinska Institute, with Profs Agneta and Magnus Nordenskjöld. These years in Sweden were very formative, and have influenced a lot, not only professionally but also very deeply on a personal level. For personal reasons, I decided not to stay in Sweden; and went on a journey that took me to London, Coimbra, Manchester and Brussels. Presently, I work at Bristol University Hospitals, although I am also supporting the Skeletal Dysplasias multidisciplinary team at the Evelina Children's Hospital in London.

It is indescribable how much one can learn by living and working in different places, having to adjust and adapt to different cultures, and put our own views into perspective. This experience, I hope, will be useful in my professional live, to my patients, who often have rare genetic conditions that mainstream clinicians do not understand very well; patients who often value our open-minded, empathic attitude towards and to them. Ultimately Clinical Genetics needs to be highly personalised to accommodate the individual patients, the local set of cultural values, and the resources of the healthcare systems.

I expect to be the voice of my patients at the ESHG board, and I am very keen to work together with my colleagues to make Medical Genetics accessible, but also delivered in an ethical and competent way to people seeking these services. Moreover, I strongly believe that the ESHG should be very active in guiding how genetic testing is regulated, ensuring high standards and quality control to all labs operating in Europe.

Dr. Cristina Rusu

Professor in the Medical Genetics Department of the University of Medicine and Pharmacy in Iasi, Romania; consultant in Medical Genetics and Pediatrics – clinical geneticist experienced in dysmorphology and diagnosis of genetic forms of intellectual disability; head of Iasi Regional Medical Genetics Centre (affiliated to ITHACA European Reference Network); coordinator Orphanet Romania.

I decided to apply for ESHG board membership because after attending the ESHG membership meetings for years, I had the impression the Eastern Europe was not really represented.

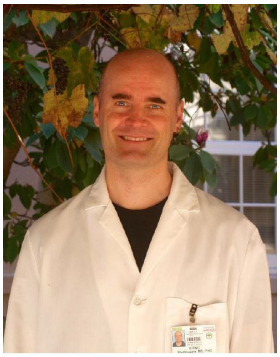
Considering my background, I would like to be involved in activities related to education, as well as those related to development of services in Eastern Europe/ Balkans.

Laura Valle

Since my teenage years I always wanted to become a researcher to help cure diseases. At the University of Navarra, while studying Biology and Biochemistry, I had the opportunity of getting in close contact with human genetics, in particular cancer genetics, thanks to an internship at the Genetics Department. From then on, I have dedicated my scientific career to the study of genetic factors that determine the risk of colon cancer, mainly focused on Mendelian cancer syndromes. After my PhD at the Spanish National Cancer Research Center (CNIO, Spain) and a postdoc at the Human Genetics Program under the supervision of Dr. Albert de la Chapelle at the Comprehensive Cancer Center of the Ohio State University (USA), in 2009 I joined the Hereditary Cancer Group of the Bellvitge Biomedical Research Institute (IDIBELL). At IDIBELL, where I hold a tenure track position, I have developed my own research lines as principal investigator, focused on the identification of new cancer predisposing genes or genetic mechanisms, and the characterization of new syndromes.

While also being a board member of the Spanish Society of Human Genetics, one of my goals at the ESHG board is to help our country get the long-time desired Laboratory and Clinical Genetics specialties, which will certainly increase the quality of patient care and diagnosis, as well as the training of professionals. In parallel, I would like to contribute to the implementation of genetics and genomics into Precision/Personalized Medicine, and to the definition of European guidelines for variant interpretation and for the management of hereditary cancer patients, in line with the goals of the ERN Genturis. It is for me a privilege to be part of the ESHG Board and I hope to meet the expectations of this position.

Kirmo Wartiovaara



Warmest greetings from somewhat cold Finnish winter and thanks for trusting me with the possibility to serve the Society as a Board Member! My wish is to help the curiosity-driven research of genetic life sciences towards having a positive effect also to the physical well-being of us all. I think that in clinical genetics we have come to the age, in which we can use our knowledge about DNA biology to develop real and effective treatments.

In my mind, we already know quite a bit about the pathogenetic mechanisms of genetic changes but also know more and more molecular biology techniques which can improve the disrupted cellular functions. In order to develop new treatments, we could now try to combine the clinical and (bio)technical advances to the societal know-how: how to make the therapeutic products safe, available, accepted and cost effective. There is still plenty of work to be done and several technical, ethical, regulatory, etc questions to be answered, but what an interesting time we are living!

In my own work I see patients regularly and supervise young bright students, who are technically superb in creating corrections to these patient cells. Our best experience is in CRISPR-Cas -based gene editing of blood and iPS stem cells, and we are interested also in the societal questions concerning gene editing in general. Part of my time goes to public awareness and education, writing popular science and medicine texts. My hobbies are literature and music, but I seem to have the interest in cells also in my free time, mainly the cellulose-walled organisms I try to modify in the garden and my basement wood workshop.

Report on the 4th Course Basics in Human Genetic Diagnostics – A course for CLGs in education

by Jelena Filipović Tričković, PhD MD, Vinča Institute of Nuclear Sciences, University of Belgrade, Serbia

The fourth course of Basics in Human Genetic Diagnostics – A course for Clinical Laboratory Geneticists in education took place in Hotel Mercure, Figueira da Foz, Portugal, and it was organized by Prof. Isabel M. Carreira and Prof. Joana B. Melo (both from University of Coimbra, Portugal) and co-advised by PD Dr. Thomas Liehr (Friedrich Schiller University Jena, Germany). Realization of this course was manageable thanks to sponsoring of European Society of Human Genetics (ESHG), ADS BIOTEC (Glasgow, UK), Agilent Technologies, MRC-Holland (Amsterdam, Netherlands), Soquímica (Portugal), ZytoVision (Bremerhaven, Germany). Apart from this generous financial support, all the organizers and speakers performed their lectures completely free of charge, and thus this course was organized inside the financial limit. Overall, 50 participants from 19 different countries (Armenia, Bahrain, Bulgaria, Cyprus, Finland, Georgia, Germany, Greece, Iceland, India, Latvia, Morocco, Netherlands, Portugal, Serbia, Spain, Thailand, Turkey and United Kingdom) attended the course. The diversity of people, including different specific area of work (diagnostic/research) and countries of origins were great for exchange of experiences, as well as for establishing new contacts and potential future collaborations.

The lectures were performed each day from the early morning to the late afternoon, whereas each day was

focused on a specific field of human genetics. Theoretical lectures were followed by workshops in a specific area and at the end of each day written exams were performed. Workshops were very interesting and dynamic with a lot of discussion. Questions on the exams were straightforward and unambiguous, and aimed to evaluate participants' interest and knowledge in a given field that was previously lectured during the day. Every day was organized in such manner that we had enough breaks to communicate with each other, as well as with the lecturers. The topics were organized in a way that all aspects of human genetics were covered; historical, current and future applications were presented with emphasis on advantages, disadvantages and possible modes of diagnostic approaches and improvements.

On the first day of course basics of human genetics and genetics counseling was discussed focussing on genomic analysis, Mendelian and non/Mendelian inheritance, including the basics of Hardy-Weinberg law, and genomic syndromes. Applications and limitations of all methods currently used for the genomic analysis were discussed. During the workshop risk-calculations in genetic counseling were introduced. We did constructions of family pedigrees and performed our own risk-calculations using the basic rules of Bayes' theorem, prior, conditional and joint probability. The second day cytogenetic and molecular cytogenetic methods were presented. Special attention was also dedicated to teratology and genetic syndromes, including presentation of databases used for dysmorphisms assessment, that help clinicians to find correct differential diagnoses in identification of genetic syndromes. Joachim Fischer from ADS Biotec Ltd presented up-to-date instruments for the automation in the cytogenetic laboratory. Workshop was focused on identifying human chromosomes and karyotyping. On the third day we learnt about molecular cytogenetic and molecular genetics methods, such as FISH, CGH and array-CGH and their application in prenatal, postnatal and tumor pathology. Reporting in cytogenetics according to current guidelines (ISCN, 2016) was elaborated. Detailed lecture about epigenetics – uniparental disomy and imprinting was provided, including mechanisms, epigenetic events, associated diseases and their diagnostics. Didier Godin from Agilent gave a comprehensive talk on array-CGH quality control and data analyses. Workshop was committed to array-CGH results confirmation and interpretation including databases. Day four was focused on molecular techniques, including MLPA (in parts presented by Marrit Rinsma from MRC-Holland), MS-MLPA, PCR techniques, Sanger sequencing and NGS. Their applications, as well as reporting according to ACMG guidelines and databases were explained in detail. Examples from diagnostics and research were presented and discussed. The final day of the course included interesting discussions about prenatal diagnosis, regarding invasive and non-invasive techniques (especially NIPT), current knowledge in gene therapy and its perspectives, as well as on future applications in NGS. Inborn errors of metabolism, cancer genetics and neurological disorders were also presented, shortly. We were introduced by Yannick Delpu in a novel next generation cytogenetics – Bionano – a remarkable technique for high-throughput optical mapping of structural variation in the human genome. At the end of this day open discussion about the overall program was done, were participants expressed their impressions and suggestions.

Summarized, this was an intense, detailed, and useful course with exceptional speakers and various participants from different scientific areas. The lectures were dynamic, interactive and hence very interesting. Non-scientific part of the course, including two dinners and one excursion to University of Coimbra, which is classified by UNESCO as World Heritage sites, was amazing and gave us the opportunity to enjoy this outstanding historical landmark. I am very grateful to organizers for excellent planning and implementation of this course, as well as to all the other speakers for sharing their knowledge with us. I would

During a lecture of Prof. Isabel M. Carreira

The participants and some of the lecturers of the course

also like to thank for providing a fellowship (overall 18 participants obtained fellowships) which allowed me to participate in this course. In conclusion, this was a wonderful experience, and will be a very useful bases for our future work and for future collaborations.

Two ESHG Board Members awarded

On May 25, 2019, former ESHG president Milan Macek Jr. , Charles University Prague, and former Chair of the EBMG Thomas Liehr, Universtiy of Jena, both distinguished ESHG members, were awarded the Gold Medal for the outstanding contribution to medical genetics by the Academic Council of the National Research Centre for medical Genetics in Moscow. Congratulations on behalf of the ESHG and its members!

Milan Macek Jr.

Thomas Liehr

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