THE EUROPEAN SOCIETY OF HUMAN GENETICS

NEWSLETTER

No. 37 - June 2022

Presidential Report

Maurizio Genuardi,
President of the ESHG 2021-2022

Dear ESHG members,

three years have elapsed since our last "real world" conference, and we are very glad to see old and new friends and colleagues in the same place and under the same roof to learn what's happening in the world of human genetics, and exchange ideas on the best strategies to improve our knowledge and ability to fight and prevent disease.

Despite the logistical constraints, the Society has been very active during these years, showing a remarkable ability to adapt to new and unexpected circumstances. Indeed, we have recorded a growth in the activities promoted by the different ESHG Committees and by its Executive Board. Attendance to the virtual conferences in 2020 and 2021 has increased over the previous years, with a record of more than 5,000 registrants in 2020 and more than 4,000 in 2021. We can assume that a number of factors have influenced the record set in 2020, such as the novelty, the low fees and overall individual expenses, and, for sure, also the lack of alternatives. The lower number, but still well above the previous meetings, in 2021 is probably physiological – the virtual conference was not a novel experience anymore – considering also the very quick rise in the offer of online scientific events, including those organized by ESHG. Of course an important reason are the excellent programs assembled by a very efficient and highly qualified Scientific Program Committee (SPC).

Based on the positive experience made with the past virtual meetings, a decision was made on organizing in Vienna the first hybrid ESHG conference. This will be a thrilling experience, and it will inform the future of our Society's meetings. The main reason why ESHG wants to pursue the dual mode is to make scientific advances available to the genetic community worldwide, facilitating the involvement of those who cannot sustain the financial burden of travel and accommodation or who cannot leave their places due to work or other commitments.

Due to the pandemic, the composition of the Executive Board has been frozen for 1 year, and my term for the Presidency of the Society has shifted from 2020-21 to 2021-22. Luckily, in many countries the restrictions adopted to face the pandemic have been eased or lifted during this year, thus allowing a swift return to a normal scientific life. However, the sense of relief is overshadowed by the dreadful events that are occurring at less than 2 hours flight from here. The Executive Committee has deplored the shameful aggression of Ukraine and has taken initiatives to support the Ukrainian genetics community. I would especially like to welcome and pay tribute to our Ukrainian colleagues who have undertaken long and difficult journeys to attend this conference n person as well as those who are attending remotely. In addition to allowing free conference registration to all Ukrainian colleagues and travel fellowships for presenters of scientific work, the Exec has approved the allocation of a fund to help recovering the Ukrainian genetic context in the post-war period. In this way we hope to provide a small but significant contribution to our Ukrainian colleagues and to the Ukrainian recovery, as a sign of our closeness to their sufferings.

War is a natural enemy to the mission of a scientific Society that aims to improve human wellbeing by reducing the burden of diseases, including those caused by members of the humankind.

As a scientific Society, ESHG has also the moral duty to alert the civil society and policy makers about the incorrect application of genetic discoveries in medicine or other domains. This has prompted the Executive and the Public and Professional Policy Committee (PPPC) to write a statement warning against the potential, and perhaps also real in some settings, misuse of polygenic risk scores (PRS) in preimplantation genetic testing. The paper, published in the European Journal of Human Genetics, has sparked interest on a controversial matter, as documented by the number of clicks on the journal website and by media coverage.

Many interesting and important tasks and initiatives have been undertaken during my presidential year. The statement on PRS is just one of the many activities undergoing within the PPPC, among which an important document on GDPR. Some of its members will also be involved in the nascent GenEthics committee, whose main mission will be to help ensure adherence to ethical standards, particularly on the matter of genetic discrimination, for manuscripts submitted to the Society's journal.

Education of genetic and non genetic professionals and trainees as well as of the lay public is an important pillar

of our mission as a scientific society dealing with human diseases. Several new ideas, including new original approaches, sprouted from the Educational Committee (EduComm), and there is a plan to increase the course portfolio. Of note, a monthly Podcast Series named Genetic Sounds was started under the EduComm umbrella. The podcasts focus on genetic and genomic topics of general interest and distinguished speakers, including members of the ESHG-Y, the ESHG Young-Committee, have been involved in brilliant conversations. I would advise those who have not had the opportunity to watch them to do so, they are easily accessible from the ESHG website. A special thank goes to Sophia Douzgou for starting and leading this important initiative. Following a proposal of the EduComm, a mentorship scheme was approved for young scientists and health professionals in genetics to take the opportunity to visit a specialist centre and to combine this with attendance at the annual conference.

I mentioned ESHG-Y, a recently established committee, that is growing, not only in the number of members, but for its contribution to the Society's activities. ESHG-Y representatives are now stably involved in the SPC and EduComm, and have made substantial contributions to the conference program and course design. A ESHG-Y member has been appointed as one of ESHG representatives in the medical branch of the European Board of Medical Genetics (BMMG). And finally, ESHG-Y representatives will also be involved in another nascent Committee, the Social Media Committee, that will work to promote ESHG activities on social media.
Much of the work performed by geneticists both in the clinical and in the research setting concerns rare diseases. Many patients affected with these conditions have suffered the stress that pandemic has imposed on our health systems. Time has come to strengthen the collaboration between ESHG and rare disease patient associations, in order to improve the perception of their needs and be able to address them in our educational programs. To this purpose, contacts were made with EURORDIS and it was agreed that representatives of their organization will be appointed as members of the PPPC and EduComm, one for each committee.

An important issue for genetic diagnosis is the implementation of the IVDR, that can have dramatic consequences on our clinical genetic laboratory system and, consequently, on patients, especially those with rare diseases. The topic is extensively discussed within the BioMed Alliance, where ESHG is represented by Gunnar Houge and Els Dequeker. An ESHG/Eurogentest webinar on this topic will take place on June 22.

This is just a brief summary of ESHG activities since June 2021. For those interested, more details can be found in this Newsletter.

In closing, I would like to express the gratitude to all those who have contributed to the ESHG life in this year: my fellow Exec members; all Committee members and chairs; Alisdair McNeill, who is leading the journal in a complex transition; all Society members, especially those who have participated in this meeting and in our educational courses. And last but not least, the Vienna Medical Academy, that, under the guidance of Jerome Del Picchia, is the infrastructure of our Society. I am sure that the life of ESHG will be characterized by important novelties in the incoming months under the guidance of Borut Peterlin, to whom I make all my very best wishes for his term of Presidency.

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 ELPAG, ESHRE and ASHG-Social Issues Committee. In 2021-2022, the PPPC has been active on the following topics.

Polygenic risk scores in pre-implantation genetic testing.
A viewpoint has been published in the EJHG on November 22nd 2021. This has attracted a huge attention on all media, including lay and scientific press (BMJ, Nature among others). A statement supporting ESHG position has been released shortly after by the ESHRE Ethic Committee on their website.

Cascade Testing: a strategy still underexploited across Europe?
Prof Guido deWert has led a working group composed by PPPC members and the alumnae Lisbeth Tranebjaerg, Hülya Kayserili and Florence Fellmann. The aim is to analyse barriers for implementation in practice and provide rec-

ommendations. The manuscript will be ready for membership review shortly.

Misuse of genetic testing for discrimination purposes
Prof Yves Moreau is leading a working group composed by PPPC members and external experts including Veronika Lipphardt; Barbara Prainsack, Gudrun Rappold, Sam Riedjik and Denise Syndercombe-Court.
Specific examples of how practices involving genomic data can lead to undesired and unethical outcomes will be discussed to focus on the lessons we can learn about these. The aim is to formulate recommendations for institutions and professional conduct, rather than giving detailed solutions.

Genomics and Inequity.
The PPPC has applied for funding to support this ambitious project and activities will be scheduled accordingly. We have identified six work packages including 1-Structural aspects of society and healthcare, 2- Data and discrimination, 3-Epigenetics, 4-Contentious applications in mental health and non-disease traits, 5-Genomicisation and individualisation *vs* public health perspectives, 6-Screening.

The GDPR: what does it mean in practice for our genetic community?
Emmanuelle Rial-Sebbag is leading a working group composed by the PPPC and EuroGentest Quality subcommittee members. Colin Mitchel, Deborah Mascalzoni and Ted Dove have joined the working group as external expert. We aim to offer some pragmatic guidance on the do's and don'ts when applying the GDPR to genetic practice and research. The draft document is in an advanced state, but completion is taking much longer than anticipated. We hope to submit the manuscript to membership for their review in summer 2022.

Survey on Expanded Preconception Carrier Screening in Assisted Reproduction.
Joint activity with ESHRE. The online questionnaire has been circulated among ESHG and ESHRE members to assess the use of EPCR across Europe. The results of this questionnaire will be published in winter 2022.

A Survey on Opportunistic genomic screening will be launched soon among membership to ascertain the impact of the ESHG recommendations on OGS. It will be clinician/scientist-based, not institution-based.
Translation in different languages will be considered. Please take some time to complete it!

PPPC members are collaborating with the Educational Committee to draft guidelines on **best practice on the use of language** when addressing patients with RD. The aim is to promote a more respectful and inclusive attitude, in collaboration with patients advocates.

PPPC members are also collaborating with EURORDIS in the Newborn Screening Working Group they are leading within the European Project **Screen4Care**. The project is funded under the IMI2 (Innovative Medicine Initiative) and it is aimed at shortening the pathway to diagnosis using advanced technologies.

Future topics on the PPPC calendar: a possible new course for the ESHG portfolio (*Things you need to know but nobody told you*); a viewpoint on Newborn Screening by WGS; a position statement on feedback of results from biobanks.

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 the 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 2021-2022 were
Angus Clarke, Christophe Cordier, Guido de Wert, Florence Fellmann, Francesca Forzano (Chair), Sabine Hentze, Heidi Howard, Milan Macek, Béla Melegh, Alvaro Mendes, Yves Moreau, Markus Perola, Dragica Radojkovic, Emmanuelle Rial-Sebbag, Edith Sky Gross, Vigdis Stefánsdottir, Fiona Ulph and Carla van El (Secretary-general). Current observers: Olga Antonova and Dr Yalda Jamshidi.

Report of the Education Committee (EduComm) 2021-2022: ESHG has a mission to promote the practice and knowledge of Human and Medical Genetics in Europe

By William Newman, Chair of the EduComm

This has been a productive and exciting year for the Education Committee as we make progress on a number of fronts. Gulcin Gumus from EURORDIS has joined EduComm to provide a patient perspective to our work. Thanks to Prof Philippos Patsalis who stepped down from the Committee this year. We appreciate all his effort and commitment to the work of EduComm.

ESHG Courses
We continue to expand our portfolio (see appendix) - including Precision Medicine, Pharmacogenetics, Bioinfor-

matics and Reproductive Medicine. Some face to face have re-started but use of hybrid and online courses is working well.
An App-based paediatric neurogenetics course has been funded through unrestricted educational grant by Illumina. Organizer: Nicola Brunetti-Pieri (Naples) with Kathleen Gorman (Dublin) and Nadia Bahi-Buisson (Paris). Twelve 15 minute segments of genetic aspects of paediatric neurology. UEMS credits will be secured. Content is being created with X-peer https://xpeer.app/ and will be available later this year.

EuroGems update - led by Ed Tobias
The ESHG educational website EuroGEMS.org has now been visited from 128 countries. A link to EURORDIS is already included in EuroGEMS. The new Spanish pages of EuroGEMS continue to function well and are already receiving visits from 29 countries. Translation into Portuguese in progress (thanks to Raquel Silva and Juliana Cerqueira) and plans for a French version next year.

Podcast Program - led by Sofia Douzgou Houge
Genetic Sounds is releasing a monthly podcast (see https://www.eshg.org/index.php?id=geneticsounds). Excellent uptake across all continents ~800 downloads already (predominantly on mobiles, via Buzzsprout, Apple and Spotify). There is a live podcast event at the ESHG conference. Plans for a second series are progressing. We believe this is a good way of interacting with a broad audience of professionals and the public and are keen for ideas regarding other topics.

International Mentorship scheme
Five successful candidates were selected in this inaugural program.

- Sharhorodska (Ukraine to visit Tartu, Estonia)
- O'Reilly (Ireland to visit Cambridge, UK)
- Yahya (Bulgaria to visit Newcastle UK)
- Castillo (Spain to visit Porto, Portugal)
- Ciuca (Romania to visit Manchester, UK)

We plan to meet the successful mentorship scheme awardees at the Vienna conference. The next round of applications will be live in November.

International Links
A recent meeting was held by members of the Canadian College of Medical Geneticists (CCMG) to discuss how the two Societies can work together.

Person First Language
Led by Prof Ramona Moldovan, with the PPPC, a program of work to explore the use of patient first language in genetics - initially within the ESHG - considering abstracts and with the European Journal of Human Genetics is being explored.
If you have any ideas/comments regarding the work of EduCom please contact me at:
william.newman@manchester.ac.uk.

Appendix

1 - Proposal for a new ESHG course by Francesca Forzano on "Things you need to know but nobody told you (tentative)". Provisionally planned for October 2022, virtual. - awaiting further details
2 - 1st European Society of Human Genetics Course on: Precision Medicine: A Focus on Clinical Utility' Virtual on 25-27 April 2022. Organizer: James O'Byrne. 320 registrants from 45 countries. 18 CMEs secured.
3 - 'Clinical Genomics and NGS Course' to be held in Bertinoro, Italy, 7-13 May 2022. Organizer: C Gillisen, A. Hoischen
4 - 6th ESHG Training Course on Cardiogenetics. Antwerp, Belgium. May 14-17 May, 2022. Organizers: Bill Newman, Bart Loeys, Johan Saenen
5 - 5th Course on Basics in human genetic diagnostics – A course for Clinical Laboratory Geneticists (CLGs) in education on September 5-9, 2022 in Figueira da Foz, Portugal
6 - ESHG Course in Hereditary Cancer' to be held in Bertinoro, Italy, 19-23 September 2022. Organizer: Nicoline Hoogerbrugge, Carla Oliveira, Hildegunn Høberg-Vetti and Elke Holinski-Feder
7 - ESHG Pharmacogenetics Course. Portorož, Slovenia. 22-24 September, 2022. Organizer: Prof Vita Dolzan.
8 - EJPRD Third Training Course Quality assurance, variant interpretation and data management in the NGS diagnostics era. TBA. Organizers: Gert Matthijs (Belgium), Domenico Tarusico (Italy), Claudio Carta (Italy)
9- ESHG GWAS course online. Organizer Inga Propopenko. 27 June-1st July 2022.
10 - ESHG Hybrid Course "Next-generation Reproductive Genetics" October 13-15, 2022 Edith Coonen (Maastricht, The Netherlands).
11 - ESHG bioinformatics course. Christian Gilissen (Nijmegen, The Netherlands). More details awaited
12 - ESHG Eye Genetics. September 14-28, 2022, Bertinoro, Italy.
13 - Plans for dysmorphology and counselling courses in 2023.

ESHG Newsletter No. 37, June 2022
Editors: Carla Oliveira, Jerome del Picchia

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Report on activities of European Journal of Human Genetics (May 2021 – May 2022)

by Alisdair McNeill, Editor in Chief EJHG, Senior Clinical Lecturer. The University of Sheffield.

Expansion of Section Editor and Editorial Board member groups

In order to increase the geographic reach of EJHG, and encourage engagement and submission, we continue to expand our editorial board.

New section editors in 2021-2022:

- Katta M Girisha, Kasturba Medical College, Manipal, India
- Ben Pode-Shakked, Tel-Aviv University, Israel
- Ramona Moldovan, University of Manchester, UK and Babeş-Bolyai University, Romania
- Andrew Lindsay, University College Cork, Ireland
- Malak Abedalthagafi, King Abdulaziz City for Science and Technology, Riyadh, Saudi Arabia

New Editorial Board members in 2021-2022:

- Dr Abdullah Al Maruf, University of Manitoba, Canada
- Dr Julia Baptista, University of Plymouth, UK
- Dr Patrick Benusiglio, Sorbonne University, Paris, France
- Dr Ryan Dhindsa, Baylor College of Medicine, USA
- Dr Jamie Ellingford, University of Manchester, UK
- Dr Zerin Hyder, Manchester University NHS Foundation Trust, UK
- Dr Rengyun Liu, Sun Yat-sen University, China
- Dr Raj Rajeshwar Malinda, Freelance, India
- Dr Belinda McClaren, Murdoch Children's Research Institute, Australia
- Prof Claire Morgan, Swansea University, UK
- Dr Eaaswarkhanth Muthukrishnan, New York University Abu Dhabi, UAE
- Dr Reuben J Pengelly, University of Southampton, UK
- Prof Jennifer Roggenbuck, The Ohio State University College of Medicine, USA
- Dr Erin Turbitt, University of Technology Sydney, Australia
- Dr Annalisa Vetro, University of Florence, Italy
- Dr Andrew Walley, St George's University of London, UK
- Dr Roddy Walsh, Amsterdam UMC, Netherlands

A Section Editor from ESHG-Y would be very welcome, my invitation still stands.

Once the ESHG-SpringerNature contract is finalised we will appoint a new Social Media Editor.

EJHG content and impact factor (4.2 in 2020)

We remain committed to scientific quality/excellence as the main criteria for paper acceptance. We aim to increase the impact factor by publishing the highest quality original research and review papers that we can.

I decided to set a very high bar for single case reports: e.g. exceptional functional validation, very important novel genomic insights, insights into disease mechanisms or treatments. These single cases are often very poorly cited and depress journal impact factor and I try to minimise this content.

We now aim to publish 4-5 editorial type pieces per month (comments, viewpoints, etc). These encourage readers to explore EJHG content in more detail and hopefully cite papers.

I also have an active program of soliciting review papers from experts in the field; can be difficult to get invited reviews but we can manage about 1 per month. Again, these are well read and highly cited.

Social Media

Twitter continues to do well. We have >6 500 followers. When we can appoint new social media editor we can restart the activities such as monthly journal clubs.

May need to change platform if Elon Musk decides to charge commercial entities for Twitter use…

Future threats to EJHG (ie move to fully open access/ online)

The main issue is the falling number of submissions. This is partly attributable to fall in submissions from China (perhaps reflecting recent ESHG statements on ethical aspects of genetics in China).

If SpringerNature want us to be a fully online journal, then we will need to increase the content we publish online year on year. To do this, we would need to lower the bar from scientific excellence for acceptance of papers to scientific validity (as PLoS One does).

The drive to publish open access may create difficulties given the cost to researchers of doing so. However, SpringerNature has a number of agreements at country and institutional level which enable a central fund to pay the open access costs rather than the individual researcher. Current APC is £2,380/ $3,380/ €2,790.

ESHG board has (I believe) agreed to pay APC for official ESHG papers/position statements after approval.

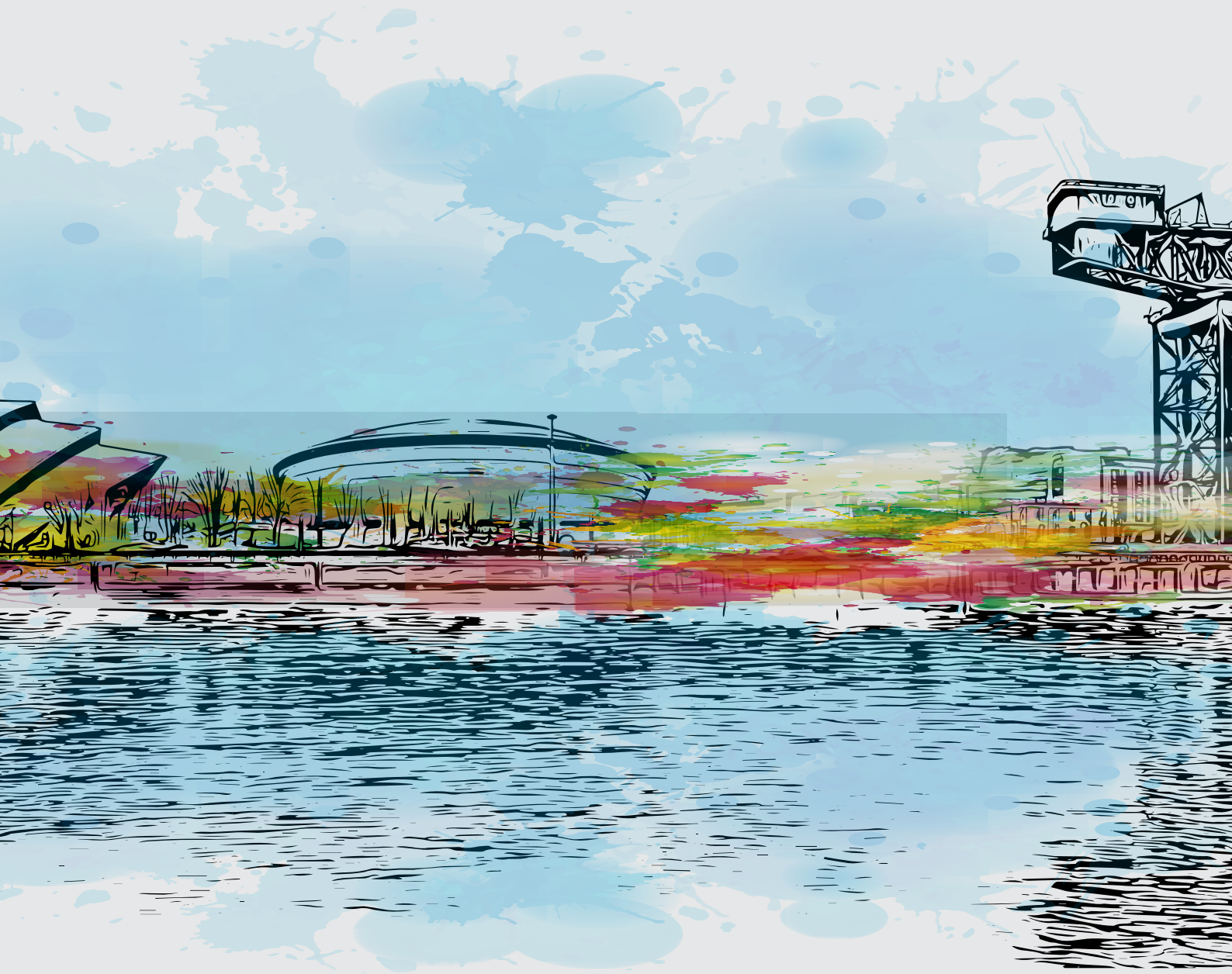
Further information on the "transformative agreements" (ie sources of APC funding paid by institutions) can be found here: https://www.springernature.com/gp/open-research/funding/articles

ESHG

EUROPEAN SOCIETY OF HUMAN GENETICS

EUROPEAN HUMAN GENETICS CONFERENCE 2023

SEC | Glasgow, Scotland, UK | June 10 – 13



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The ESHG-Y Committee - An annual activity report

by Elena Avram, Juliana XM Cerqueira, Can Ding, Mridul Johari, Ana Raquel Silva, Delia Sabau, Nuru Noor

In June 2019, the European Society of Human Genetics-Young Committee (ESHG-Y) was launched under the guidance of the EHSG by young medical geneticists who shared common goals and visions.

Its mission is to represent and support the young European human geneticists by developing strategies to enhance their interests and programs that aim for a better education. ESHG-Y's main objectives are: organize scientific events, provide equal access to educational opportunities, create a professional network, and support outstanding young geneticists to become future leaders.

Similar to 2020, in 2021 we got involved in the organizing committees of the ESHG Annual Conference and the European Dysmorphology Conference. Furthermore, in 2021 we have continued to develop our existing collaborations with outstanding organizations like ERN-Ithaca the European reference network for rare malformation syndromes and rare intellectual and neurodevelopmental disorders, UNIQUE a charity supporting families living with rare chromosome disorders or autosomal dominant single-gene disorders, and the European Joint Programme on Rare Disease (EJP RD) for the development of the "Diagnosing Rare Diseases" Course.

We established a liaison with the European Board of Medical Genetics (EBMG), by adding one of our members to the EBMG. On December 1st, 2021 we organized and coordinated a special virtual session called "Becoming European Board Certified in Medical Genetics and Genomics". This session aimed at highlighting the importance of holding the European Certification in Medical Genetics and Genomics (ECMGG) certificate and at providing young members with essential information about this exam.

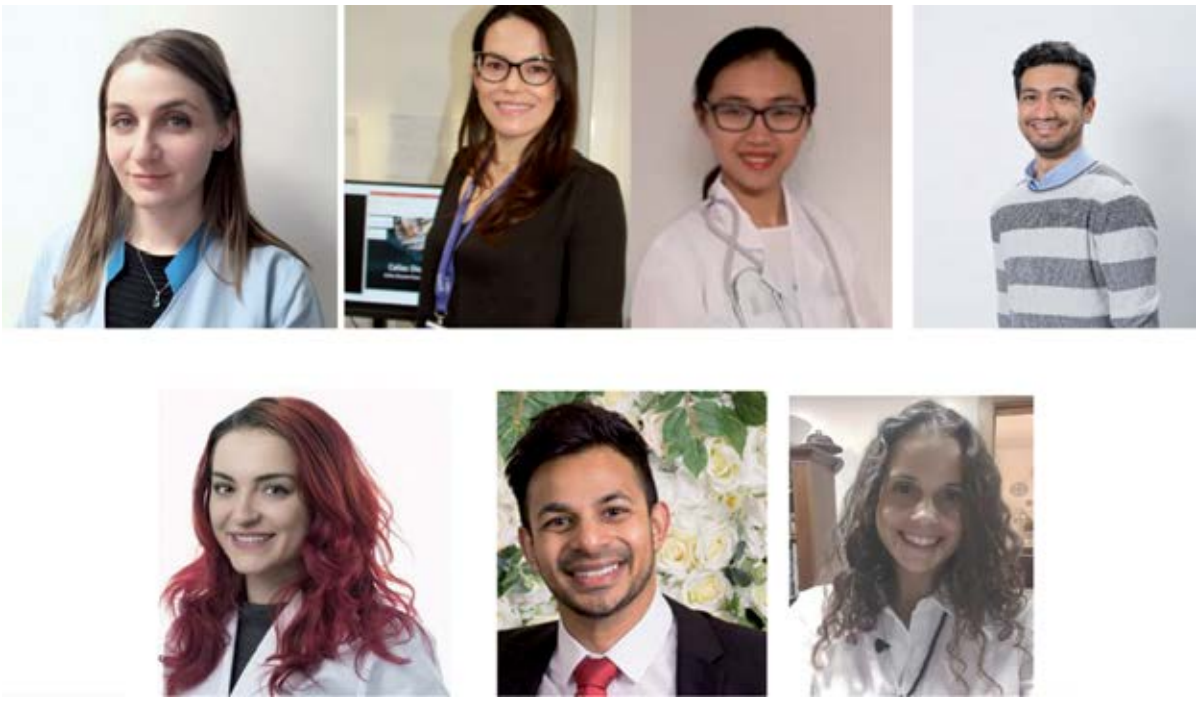
In addition we were also involved as judges for the DNA Day starting with 2021 .

In 2022 The ESHG-Y Committee helped with the EuroGEMS website translation into Portuguese, and additionally got involved in the ESHG International Mentorship Program and the ESHG's Podcast "Genetic Sounds".

During the last year we had the opportunity to publish two articles "The Young Geneticists Network and the ESHG-Young committee, a forward-looking international community" and "The Role of the European Society of Human Genetics in Delivering Genomic Education".

Furthermore the ESHG Journal has offered us a permanent section editor position.

In the last year, representatives from the ESHG-Y got involved in different committees like the ESHG Board, the



The members of the ESHG-Y Commitee: Elena Avram, Juliana XM Cerqueira, Can Ding, Mridul Johari, Delia Sabau, Nuru Noor, Ana Raquel Silva

14th International Congress of Human Genetics (ICHG 2023)

22 - 26 February 2023

Cape Town, South Africa

"Coming Home"

ICHG2023.COM

Hosted by:

AfSHG African Society of Human Genetics

SASHG Southern African Society for Human Genetics

We invite you to come home: join us in Cape Town, South Africa

IMPORTANT DATES

Abstract submission closes:
31 August 2022
Early bird registration closes:
30 November 2022
Side meetings & satellite meetings applications due by:
30 November 2022

CONFIRMED SPEAKERS:

- Francis Collins (opening)
- Fowzan Alkuraya
- Shomarka Keita
- Dennis Lo
- Mathieu Lupien
- Carina Schlebusch
- Nicole Soranzo
- Eric Topol
- Mayana Zatz

ESHG Scientific Programme Committee (SPC) and the ESHG Education Committee (EduCom).

In the future, the ESHG-Y would like to organize the "ESHG Observership for Young Geneticists" Program which aims at offering financial support for junior doctors from developing countries to visit prestigious European human genetics departments and observe specific areas of clinical care or laboratory techniques.

With the growth and expansion of Human and Medical genetics, we feel the need to inspire, interconnect and encourage the younger generation to have a major professional impact on the homogeneous development of this speciality in Europe and beyond.

European Board of Medical Genetics - Annual Report 2021-2022

by Isabel Marques Carreira, President of the EBMG, 2021-2022

We as the European Board of Medical Genetics (EBMG) have as its main purpose 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 (CLGs),
- Genetic Nurses and Genetic Counsellors (GNGC), and
- Medical Geneticists (MG).

During this year these three branches worked together pursuing our common goal of promoting the recognition of our professions at the European level. This co-work led among others to the publication of a position statement in 2022 (doi: 10.1038/s41431-022-01080-3).
In the meantime, within each branch we continued with the promotion of good standards of professional registration procedures, and a rigorous definition of training programs and professional qualification standards in order to promote not only the quality of practice but above all to ensure patients safety at clinical and diagnostic activities in the European genetics' healthcare services. At present, there are in total more than 500 professionals from 41 countries that are certified by the EBMG. During this year the EBMG Council had seven monthly meetings were current issues from each branch were presented and discussed. A strong emphasis was put on the re-registration of members as well as on the evaluation procedures of new members. In order to shared experiences between the three branches short talks were organized in alternated meetings:

- Exam experience of the Medical Branch by Ute Moog;
- Overview of the MSc assessment process by Joana Bengoa - Genetic Nurses and Counsellors Branch;
- Clinical Laboratory Genetics branch in EBMB: what we do, what we plan and where we are, by Thomas Liehr.

Also during this year, each branch, in order to regulate their activities and to comply with the statutes (https://www.ebmg.eu/fileadmin/eshg/EBMG/EBMG_statutes_GA_2021_.pdf), elaborated drafts of Standard Operation Procedures (SOPs). Those cover various aspects such as for example: purpose/aims of the branch, membership, composition of the branch, eligibility, nomination and election, duties, mandate periods, examination procedure, finances, and arbitration committee. After legal revision, these three documents will be posted in the EBMG page within each branch.

Short Reports of the 3 branches:

Clinical Laboratory Branch
The CLG branch has at present 411 registered members from ~40 countries. In 2021/22 round we had 57 recertification and 39 new certification requests; 21 were not eligible, 72 were approved and 3, each from this round and from last year-round wait for finishing the application by taking a test. The latter six candidates are using group-3-way application. Due to Covid-pandemic ~20 group 3-way applicants were finishing their application in a combination of in place written exam (most in home countries) and an online oral part with at least one committee member in place and at least one being present virtually by live video. Besides, we further are recruiting labs for the initiative Open Lab for ErCLG education and exchange (see https://www.ebmg.eu/669.0.html). All details on ErCLG registration can be found on https://www.ebmg.eu/clg.0.html. The (re-)registration process starts this year again on 15th June – see https://www.ebmg.eu/695.0.html. Here you can now also find: (i) general information, and eligibility criteria and (b) step by step instructions. Finally, the yearly course for ErCLGs in education is planned to take place this year again in Portugal (see https://www.ebmg.eu/669.0.html). Besides, it was time to organize a meeting with a Portuguese member from the European Parliament in order to draw the attention to the need of professionalization of genetics healthcare in Europe in the near future. Especially, we will discuss possibility to support the EBMG – CLG-branch to bring this issue of CLG becoming a recognized European profession on the agenda of the European Parliament.

Genetic Nurse & Genetic Counsellor Branch
During 2021, 13 genetic counsellors achieved registration for the first time and 9 genetic counsellors successfully renewed their registration; congratulations to them all. The

next cycle of registrations and renewals are under review and results will be announced in June 2022.

Regarding the assessment of Masters programs, we have moved to a digital submission platform to ensure an easy and flexible submission process.

During this year we have welcomed in the branch Alana Ward (Ireland) and Andrada Ciuca (Romania), who were approved as members at the EBMG meeting of June 2021. Next June Milena Paneque (Portugal) will finish her time in the division; we would like to thank her commitment and guidance as she has been a key member in the division. At present, Clara Serra (Spain), Joana Bengoa (France) and Sara Pasalodos (Spain) are the chairs of the division.

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

Medical Genetics Branch
Since the last General Assembly of the EBMG on September 06, 2021, the Branch of Medical Geneticists has further developed the exam for the European Certificate in Medical Genetics and Genomics (ECMGG) in close cooperation with UEMS (European Union of Medical Specialists) Section of Medical Genetics. The exam was successfully delivered online in 2021 both for the written part of 110 multiple choice questions and for six short oral assessments. The exam for the ECMGG 2022 will also be held online and has been prepared in many virtual meetings as well as a hybrid workshop in Lodz, Poland, in March 2022. More information can be found at www.uems-ecmgg.org. Negotiations in several countries are being undertaken for the adoption of the ECMGG as national specialist knowledge exam.

A working group addressing the revision of the European training requirements for medical geneticists (ETR working group) was set up. It comprises members of UEMS-SMG, EBMG-Branch of Medical Geneticists and a colleague not represented in either organisation. The group has picked up work by gathering the documents with training requirements of different countries.

Most countries have a CME register for Medical Geneticists, either mandatory or voluntary. Details and the exact number are however unknown. It is also unclear whether a European CME register would be helpful. A survey on existing CME registers was sent to European countries represented in either UEMS or ESHG to gain more information. The evaluation of results is pending.

In addition to the annual meeting, two Branch meetings were held virtually where many items have been discussed, among which the mapping of training centres in Europe and a meeting with the ESHG Young Geneticists Committee.

Finally, as Chair of our Board, I would like to thank and recognise the commitment of all the members, the positive contribution of the professional Branches of the EBMG and that of the Exec committee. It is because of everyone´s willingness that our achievements were possible. Last but not least, we would all like to thank Jerome del Picchia and Kristina van Dam from Vienna Academy for their legal and administrative support.

ESHG 2022 Hybrid Conference in Numbers

Jerome del Picchia, Executive Officer of the ESHG

The 56th European Human Genetics Conference 2022 in Vienna marks the first fully hybrid ESHG conference in ESHG'S history. After two years of virtual conferences due to the pandemic, we are finally meeting again in person in my home town. But we cannot turn back the wheels of time.

The Annual Meetings Committee (AMC) has decided quite quickly that the 2022 conference should be held in a hybrid format, so an in-person congress where all sessions would be live-streamed over the internet and to incorporate online participation in our meeting. By offering a hybrid meeting, ESHG is aiming at breaking down borders and bringing top science to geneticists all over the world through an online participation. Persons unable to attend the in-person meeting, will be able to tune in through the dedicated virtual conference and stay up to date with the latest research in the field of human genetics.

There have been discussions about the pricing of the virtual attendance, and admittedly, setting the fees many months ahead of the conference, only based on offers and being very unsure about the possible online and live participation, with a still ongoing pandemic, was just about an educated guess. Fact is that the additional costs in terms of AV, IT and personnel to make the hybrid conference possible, roughly corresponds to the rental of a conference center with basic AV equipment.

Obviously, the organisation costs increase equally due to the additional workload. So as a one off, it was decided to set the fees for online participation at the same level as live participation. And while overall participation is at a good level, it seems unlikely at this stage that the conference will be able to balance the budget.

Nevertheless, the Executive Board and the AMC will revisit the online participation fees for 2023 and we may find an alternative approach to all online educational content of ESHG.

Talking about numbers, how does the first live gathering look so far? A total of 90 sessions and 36 corporate satellites will be live streamed over four days.

2,105 abstracts from 75 countries were submitted (+100 compared to 2021, but some -250 compared to the last live meeting in Gothenburg). 168 submissions were accepted as talks in Concurrent Sessions, 1,023 as so-called hybrid posters (so paper and electronic), and 784 as e-posters.
118 invited talks will be given by scientists (41%F/59%M) from 25 different countries,

As per date of printing, 3,484 geneticists from 78 countries were registered, thereof 44% are ESHG members. 75% have indicated their live participation in Vienna.

The gender distribution is 63%F/37%M, which is in line most conferences in the last years. In addition, we will welcome over 500 delegates from exhibitors and sponsors.

On behalf of the ESHG, I would like to seize the opportunity to cordially thank our industry partners for having stayed with us through the pandemic and for their continuous support over the years, which made it possible to travel the undiscovered virtual country together.

Finally, I wish all of you attending the conference in Vienna, a wonderful time among your fellow geneticists, that you can pick up with contacts where you left them two years ago and meet some new ones. Finally I wish all of us that it will not take another two years until we meet again.

Willkommen in Wien! Welcome to Vienna!

EUROPEAN SOCIETY OF HUMAN GENETICS

AGENDA
General Assembly 2022 of the European Society of Human Genetics
Sunday, June 12, 2022, 19:00 - 20:00 hrs
Austria Center Vienna, Hall K (Level U2)

1. Opening by the President of the Society, Maurizio Genuardi
2. Approval of the Agenda and of the timely invitation of members
3. Approval of the Minutes of the last Meeting in 2021
4. Report on the Activity of the Society 2021-2022, Maurizio Genuardi
5. Financial Report of the Treasurer 2021, Gunnar Houge
6. Reports of the Auditors 2021, Helen Kingston, Lars Fredrik Engebretsen
7. Exoneration of the Treasurer
8. Exoneration of the Executive Board
9. Opening by the new President of the Society, Borut Peterlin
10. Results of the nomination for President-Elect 2022 - Confirmation by the Membership
11. Results Board Member nominations 2022 - Confirmation by the Membership
12. Thanks to leaving Board Members
13. Vote on Membership Fees 2023
14. Site of future European Human Genetics Conferences
15. Any other Business

Refereshments will be served after the General Assembly in the Foyer of Hall K