THE EUROPEAN SOCIETY OF HUMAN GENETICS

NEWSLETTER

No. 38 - June 2023

Report from the President

by Borut Peterlin,
President of the ESHG 2022-2023

Dear ESHG members,

The field of human genetics has experienced rapid evolution in recent decades. New technologies have revolutionized the diagnosis of genetic disorders and shed light on their public health impact. If we estimate the number of all human diseases to be approximately 25,000, known genetic disorders contribute to about one-fourth of that total. Considering the cumulative prevalence of genetic diseases, it is estimated that up to 400 million people worldwide could be affected. Such a substantial burden of genetic aetiology in human diseases has significant implications for the organization of healthcare systems.

However, the translation of healthcare innovations often progresses relatively slowly, and there is considerable heterogeneity in the maturity level, gaps, and barriers among these systems. Geneticists make up less than 1% of the global medical profession workforce despite the considerable burden of genetic diseases. Consequently, there is a need to expand the genetic workforce and support the development of new genetic professions in addition to clinical and laboratory geneticists. These include genetic counsellors, genetic nurses, and bioinformaticians. Furthermore, exploring new models of service provision may enhance the accessibility of and to genetic services.

Historically, genetic services have been organized at the secondary level of healthcare, with geneticists closely collaborating with other medical specialities. However, the rapid advancement of genomic medicine requires refinement in defining the role of geneticists in diagnostics and providing patient-centred care and treatment. Clinical guidelines and pathways, (co)developed by geneticists, still need to be prepared. In addition, novel models of genetic education are necessary to empower the general public and healthcare workers.

Genomic medicine is also a driving force behind personalized medicine. Hence, national and international initiatives could greatly benefit genetic professionals' participation. In addition to insights into genetic diagnostic pathways, several translational and ethical, legal, and social issues are already incorporated into genetic services.

Furthermore, scientific discoveries and clinical research have always been the foundation for subsequent implementation in clinical practice. For example, discovering new genes associated with human diseases has significantly contributed to understanding the mechanisms of diseases. However, breakthroughs in comprehending complex human disorders and public health innovations require new models of scientific collaboration, which are not adequately supported by current financing schemes.

The European Society of Human Genetics (ESHG) is Europe's primary representative of geneticists, uniquely bridging the domains of health professionals and scientists. Nevertheless, the society's activities primarily rely on the voluntary work of its structures and members.

Therefore, I would like to express my gratitude to the chairs, secretaries, and members of the ESHG committees, including the Annual Meetings Committee, Education Committee, EuroGenetest Committee, Public and Professional Policy Committee, ESHG Young Geneticists Committee and the editor of the European Journal of Human Genetics, for their invaluable contributions to the advancement of genetic science and the profession.

Additionally, the ESHG has recently established the Strategic Committee to develop the society's strategy by identifying, prioritizing, and addressing the most critical challenges related to the genetic profession and research.

Finally, I express my appreciation to the Secretary-General and her Deputy, Executive Committee and Board members, our Executive Officer and ESHG members for their unwavering support, assistance and encouragement during my tenure as the society's President. I wish my successor, Prof. Valerie Cormier-Daire, every success in continuing society's important work.

Report from the Scientific Programme Committee

By Alexandre Reymond, Chair of the SPC

The ESHG Scientific Program Committee (SPC, https://www.eshg.org/spc) primary mission is to determine the scientific contents of our annual conference. It regroups thirty-three experts of fifteen countries representing the different facets of human genetics. To prepare the 2023 Glasgow conference, the SPC also incorporated for the first time two members of the ESHG Young Geneticists Committee (ESHG-Y) (https://www.eshg.org/eshgy). Juliana Miranda Cerqueira and Mridul Johari were extremely active as first spokewoman/man of their constituency, as eight of the invited symposia/education sessions of the 2023 conference stemmed from original ideas of the ESHG-Y in conjunction with our EduCom educational committee (https://www.eshg.org/educom). The novelties do not stop there as beside the traditional award lectures, invited symposia, educational sessions, platform presentations selected from submitted abstracts and workshops, we have added "Get2gether" sessions to our Scottish program. These sessions should promote networking and professional connections in a specific area of research and/or health. In Glasgow, for example, we have scheduled Get2gether sessions to present the different European Research Networks (ERNs) and to discuss the new European *in vitro* diagnostic medical devices regulation (IVDR).

You might be surprised not to see an ESHG-ASHG "building bridges" session in our 2023 program. Together with our American partners, we decided that these sessions that aim to tackle global questions should not be restricted to participants of our respective annual conferences in the future. Starting next year and taking advantage of new streaming possibilities, the ESHG, the ASHG and the International Society of Human Genetics will organize semestrial free "building bridges" webinars showcasing speakers of all continents.

Last year in my address during the opening plenary, I mentioned that 42% of the invited speakers in Vienna were females. Striving to get even more balanced in Glasgow, we established a preliminary program last July that reached the parity between female and male speakers. Our final 2023 program will see 46% of female invited speakers presenting their research, as female PIs are slightly more likely to decline our invitation than their male counterparts. Unfortunately, this improved gender balance is paralleled by a decrease in the origin of our speakers from 24 in Vienna to 22 countries in Glasgow. We will continue to work towards organizing a conference as representative of the human genetic community as possible.

With 2487 submitted abstracts from 81 countries, we are back to pre-covid numbers of submissions on par with the conferences in Copenhagen and Milan in 2017 and 2018, respectively. To present this ongoing research in Scotland, we have selected 168 abstracts for platforms presentations and 31 for best poster pitch presentations. Importantly, and a tribute to the dynamism of our discipline and society, 56% (91 out of 168 from 27 countries) of the platform presentations will be given by early career award candidates.

Finally, I want to catch this opportunity to praise the decision of the ESHG executive committee to organize a fully hybrid meeting for the second year in a row. This allows human geneticists from around the world to participate virtually furthering and respecting the educational mission of the ESHG.

We hope that you will be as thrilled as we are to listen to the invited and selected speakers presenting their science in Glasgow. Welcome to Scotland!

www.eshg.org/spc

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Report from the Annual Meetings Committee

By Joris Veltman, Chair of the AMC

The Annual Meetings Committee (AMC) was originally established to decide on the future venues and locations of the ESHG conference. In 2022, the Annual Meetings Committee restarted with more of a focus on strategic discussions around the conference size and shape, discussing the in-person conference as well as online participation.

We see great opportunities to grow with our hybrid conference, allowing more colleagues from all over the world to join our conference. This is particularly important given the discussions around sustainability, widening participation and the flexibility to return to an online conference if ever needed again.

At the same time, we are confronted with increased costs and complexities, because of the combination of on-site and online platforms. We realize this is a developing market and have the healthy ambition to become the largest genetics conference globally in the next 10 years. In order to reach that goal, we decided to make online-only participation to our 2023 conference more affordable and stimulate all participants to become ESHG members.

We are also working with the Programme Committee to make sure the programme is attractive to both in-person and online participation, allowing for optimal participation and interaction between all participants. We are also looking at better marketing of the online participation in the coming period, as this will be essential to grow participation and make up for the lower income per participant and cover for the significant costs of the online platform. We will closely monitor the effect of these measures on both in-person and online participation, and adjust if needed.

www.eshg.org/amc

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 2022-2023, 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, a reaction was received and we have published a response on Dec 2022 (Reply to Letter by Tellier et al: Scientific refutation of ESHG statement on embryo selection | European Journal of Human Genetics (nature.com)). The interest these papers have raised continues to be very high, and we have been very busy in dissemination activities, including invitations to interviews on local televisions, talks at national and international conferences, university debates, book chapters.

The PPPC has organised a session on Privacy, consent and data sharing in the international genetic community at the ICHG 2023 conference in Cape Town on February 2023. This session has been realised with the contribution of all the full members of the IFHGS and participation from GA4GH. We have received excellent feedback, and there is current consideration for a follow-up paper with contribution of all discussants.

We have run a Survey on Expanded Preconception Carrier Screening in Assisted Reproduction, a joint activity with ESHRE. Many thanks to all the colleagues who have participated and allowed us to collect information from 299 centres from 40 different countries! The paper with the results will be published soon in Human Reproduction.

We are still finalizing the documents on Cascade Testing, a strategy still underexploited across Europe, Misuse of genetic testing for discrimination purposes and the GDPR and what does it mean in practice for our genetic community. We aim to submit at least two drafts to membership for their review after summer. The Survey on Opportunistic genomic screening, to ascertain the impact of the ESHG recommendations on OGS, and which we aimed to launch the last winter has been delayed but will now be launched soon among membership. Please watch out for it!

PPPC members are collaborating with the Educational Committee and patients advocates in a group of activities around best practice on the use of language when addressing patients with RD. The aim is to promote a more respectful and inclusive attitude. A workshop has been organised at the ESHG2023 conference (W08, Sunday June 11th).

PPPC members are also collaborating with EURORDIS in the Newborn Screening Working Group they are leading within the European Project Screen4Care, funded under the IMI2.

Our proposal for a new interactive ESHG course (*Things you need to know but nobody told you*) has been approved, and we are now finalizing the business plan. The course will be run virtually on 2 days and will be particularly addressed to young scientists and clinicians. We hope many of you will join us, as we need your feedback to understand whether it would be helpful to integrate this course in the ESHG portfolio, with or without major restructuring. We promise it will be very enjoyable!

Future topics on the PPPC calendar: a possible joint document with the EuroGentest committee on reporting in prenatal setting; a possible statement on feedback of results from biobanks; inequities in genomics; AI in genomics.

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 2022/2023 were: Angus Clarke, Christophe Cordier, Guido de Wert, Francesca Forzano (Chair), Sabine Hentze, Heidi Howard, Milan Macek, Béla Melegh, Alvaro Mendes, Yves Moreau, Markus Perola, Inga Prokopenko, 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. ESHG-Y representative: Rhys Dore.

www.eshg.org/pppc

Report from the Education Committee (EduComm) 2022-23

By William Newman, Chair of the EduComm

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

This has been a productive and exciting year for the Education Committee, as we made progress on a number of fronts. Three new members Carmen Navas, Liz Loehrer and Edith Coonen have joined our committee and are focussed especially on international links.
Thank you to Can Ding who has stepped down as one of the ESHG-Y committee members and has been replaced by Raquel Silva.

ESHG Courses
We continue to deliver a portfolio of face to face and online education and training courses. See https://www.eshg.org/courses for a current list and details of these. We are always looking for new ideas to expand our portfolio. Please feel free to nominate suggestions for consideration. Fellowships are available to support attendance for young scientists and clinicians es-

pecially from economically disadvantaged countries.
We wish to thank Nicola Brunetti-Pieri (Naples), Kathleen Gorman (Dublin) and Nadia Bahi-Buisson (Paris) who have created a free App course (delivered by X-Peer) on paediatric neurogenetics launched earlier this year. All you need to do is register for free at https://web.xpeer.app/web/en/courses/340?channelId=46. Till the end of April there have been ~8800 views, ~1760 enrolments and ~270 diplomas issued for the two modules on diagnostic approaches of genetic syndromes and inherited metabolic disorders and on developmental and epileptic encephalopathies and brain malformations has now also been launched CME points are achieved for undertaking this program.
Please visit this resource and share ideas for other app-based courses.

EuroGems update - led by Ed Tobias
The ESHG educational website https://www.eurogems.org/ has now been visited from 136 countries.
The Spanish pages have now been visited from 39 countries.
The new Portuguese pages have been visited from 12 countries.
A French translation is well advanced with launch planned for later this year.
We are delighted to see the engagement with this resource and the increased reach that it has achieved now that it is available in different languages. Please suggest others languages that would enhance the value of the site.
Many thanks to Ed and all the people who have worked with him to create this wonderful resource.

Podcast Program - led by Sofia Douzgou Houge
Genetic Sounds https://www.eshg.org/genetic-sounds has been releasing Series 2 of a monthly podcast (6 times a year) using a conversational approach to discuss broad topics related to genetics. There has been excellent uptake across all continents. For Series 1 in 2022 there were 2,342 downloads (predominantly on mobiles, via Buzzsprout, Apple and Spotify) with a big peak around the conference in June. There is a live podcast event at the ESHG conference in Glasgow - look out for it on the program. Plans for a third 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. We have also linked with the Eurordis Rare on Air podcast https://www.eurordis.org/rare-on-air/

International Mentorship and Observership schemes
We have had lots of interest in the new ESHG mentorship and observership schemes.
The mentorship scheme https://www.eshg.org/education/eshg-mentorship-programme
Offers up to five awards (up to €1500) for early career geneticists to visit another centre and attend the ESHG conference. The award will establish a long term career supportive relationship.

Awardees of the ESHG mentorship scheme for 2023 are:
Elena Avram: Romania to Strasbourg, France
Kakha Bregvadze Georgia to Manchester, UK
Laila Gallo de Souza Brazil to London, UK
Szymon Kierat Poland to London, UK
Oliver Orji Nigeria (based in UK) to Cologne, Germany

The observership scheme https://www.eshg.org/education/eshg-observership-programme offers funding (up to €2,000) to support 4 weeks for a young geneticist to visit another centre.

Awardees of the first ESHG observerships are:
- Claudia Anyigba Accra, Ghana to Surrey, UK
- Alessandro De Falco Naples, Italy to Rotterdam, The Netherlands
- Katleen Janssens Antwerp, Belgium to Utrecht, The Netherlands
- Nikolaos Marinakis Athens, Greece to Nijmegen, The Netherlands
- Ileana-Delia Sabau Bucharest, Romania to Lausanne, Switzerland

The next round of applications for these two programs will be live in August with a closing date in November 2023. One of the mentorship awards from 2024 onwards will be ring-fenced for genetic counsellors to apply for and named in honour of Professor Lauren Kerzin-Storrar who encouraged trainees to travel to broaden their experience and knowledge and is a dearly remembered colleague.

International Links
Our three new members are making great progress on working and establishing links with other international genetics societies across the world. This builds on some of the relationships established at the International Congress for Genetics in Cape Town earlier this year. We will share educational resources and develop joint educational initiatives. One of these is the endorsement of the educational guidance Rise2 Genomics (https://doi.org/10.1038/s41436-021-

ESHG

EUROPEAN SOCIETY OF HUMAN GENETICS

EUROPEAN HUMAN GENETICS CONFERENCE 2024

Messe Berlin - CityCube | Berlin – Germany | June 01-04

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https://2024.eshg.org
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01140-x) developed and led by colleagues in Australia.

Person First Language
Led by Prof Ramona Moldovan, and working with the PPPC, we have established a program of work to explore the use of patient first language in genetics. There is a workshop on this at the Conference in Glasgow which will stimulate discussion and hopefully the next steps. We will aim to develop guidance for appropriate use of terminology and appropriately sensitive language in genomics.

If you have any ideas/comments regarding the work of EduComm please contact me at william.newman@manchester.ac.uk.

www.eshg.org/educom

Annual Report ESHG-Euro-Gentest Committee 2022

by Gert Matthijs, Chair of the EuroGentest Committee

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.

The IVD-Regulation is highest on the radar and has received much attention. It presents a major challenge to genetic diagnostic laboratories, especially on the use of in house developed test when no CE-IVD kit is commercially available for the same diagnostic application. We are concerned that the regulation may badly affect the genetic laboratories, in terms of the freedom to use in house tests and the incentive to develop novel diagnostic applications.

Under the direct auspices of the Executive Board of ESHG, an ad hoc working group on IVDR has been created to promote knowledge on IVDR and participation of geneticists and ESHG members in national and international initiatives. The group is steered by Els Dequeker, Gunnar Houge, Milan Macek Jr. and Gert Matthijs. We have been able to react swiftly and effectively on preparing policy documents, e.g. at the occasion of the Czech presidency of the EU Council. Els has presented the challenges for new tests for rare diseases and the viewpoints of ESHG at the Expert Conference on Rare Diseases, organized by the Czech Minister of Health on October 25-26, 2022 in Prague.

ESHG had previously already delegated Els Dequeker to BioMed Alliance, a group of European medical societies active in lobbying at the European level, where she joined the Task Force on In Vitro Diagnostics. The focus is on reviewing and commenting the European Guidance Documents in IVDR.

The ESHG-EuroGentest IVDR working group has organized a very successful webinar on IVDR, entitled: "IVDR - beneficial or just an expensive straight-jacket?" on June 22, 2022. It was more than a webinar, as it featured 6 hours of online contributions by representatives of the different stakeholders. With over 300 attendees, it is clear that there is a need for information and guidance on IVDR!

"IVDR is a challenge for all of us, the greatest challenge is keeping 'orphan' diagnostics available." If you share that view, and if you are interested in an update on IVDR, please join the Get2gether IVDR session at this ESHG meeting, Monday June 12 from 2 to 3.30pm (Room Boisdale).

The different EuroGentest Subcommittees have focussed on multiple aspects of quality in genetic services:

The Guidelines Subcommittee has completed the revision of 'Guidelines for diagnostic next-generation sequencing' published in 2016 and extended its remit to whole genome applications in the clinic. The new guidelines were published in September 2022 in the European Journal of Human Genetics. (E Souche et al. Recommendations for whole genome sequencing in diagnostics for rare diseases. Eur J Hum Genet 30(9):1017-1021, 2022 - PMID: 35577938).

The Quality Subcommittee has updated the 'Recommendations for reporting results of diagnostic genetic testing' from 2013. The new recommendations were also published September 2022 (ZC Deans et al. Recommendations for reporting results of diagnostic genomic testing. Eur J Hum Genet. 30(9):1011-1016, 2022 - PMID: 35361922).

Thus, both were published in the same issue of the European Journal of Human Genetics, and their appearance elicited an Editorial by Dr. Alisdair McNeill entitled: "Guidelines, guidelines everywhere — and still I'm not sure what to do". A first part of the answer is, for those of us working in diagnostics: read them, and apply them. Admittedly, they cannot be comprehensive but at least they provide a reflection of what peers think about good practice in diagnostics and quality management. Of course, guidelines are not static but should evolve as new technologies and new norms for quality assessment arrive. Secondly, please realize that professional guidelines are useful as a basis for accreditation of laboratories and may even be used as a reference to good practice and standard-of-care in juridical disputes.

The Quality Subcommittee is very active in dealing with the different issues that affect quality and competence of the diagnostic genetic and genomic laboratories, clinical genetics and genetic counselling. The work includes, for instance, the annual review of the EQA Providers performance data. In close collaboration with the Public & Professional Policy Committee of ESHG (PPPC), members of the Quality Subcommittee contribute the preparations of a statement on GDPR compliance.

ESHG-EuroGentest is also involved in the European Joint Programme on Rare Diseases (EJP-RD) for training of clinicians, laboratory scientists, policy makers, patient association representatives and others, in quality assurance, variant interpretation and data management in the NGS diagnostics era. In the fall of 2022, the fourth course was organized online by Dr Holm Graessner and co-workers (Tübingen, Germany). The fifth course will organised by Dr Krystyna Chrzanowska and co-workers and will go live! in Warsaw (Poland), 18-20 October 2023. For information and registration, please check https://www.ejpraretdiseases.org/event/quality-assurance-variant-interpretation-and-data-management-in-the-ngs-diagnostics-era-2/ .

Finally, the EuroGentest Steering Committee has been slightly reshuffled, current members are Christi van Asperen, Els Dequeker, Luca Lovrecic, Isabel Maria Carreira, Mike Morris, Ales Maver and Gert Matthijs.

Upcoming activities include work on a project and paper on 'Reporting in prenatal testing', and activity which the PPPC has referred to EuroGentest. We would like to also engage on another important theme, 'Harmonization in genetic counselling'. If you are interested in participating in the ad hoc working groups, let us know!

I gratefully acknowledge the time and effort spent by the colleagues who have voluntarily contributed to EuroGentest-ESHG achievements and output over the last year. We are keen to grow and extend the activities of our committee and subcommittees. Please contact me at gert.matthijs@uzleuven.be if you wish to join us in this great endeavour.

www.eshg.org/egt

Report from the ESHG Young Committee

By Mridul Johari, Chair of ESHG-Y on behalf of all Committee Members

The European Society of Human Genetics-Young Committee (ESHG-Young) empowers young human geneticists by developing comprehensive strategies to enhance their interests and nurture their professional growth. Our core objectives include organizing cutting-edge scientific events, promoting equal access to educational opportunities, facilitating the creation of a robust professional network, and identifying and supporting exceptional young geneticists to become future leaders in this field.

Several significant milestones marked 2022-23 for the ESHG-Young. We made remarkable progress in fulfilling our mission to support and empower young human geneticists. Four new members joined our ESHG-Young board: Dr Silvia Kalantari, MSc Rita Matos, Dr Rhys Dore and Dr Magdalena Mroczek.

Our ESHG-Young Chair, Dr Mridul Johari and Consultant MSc Juliana Miranda Cerqueira continued representing us as part of the ESHG-SPC. Proposals submitted by the ESHG-Young resulted in two educational sessions, five symposia and one workshop at the ESHG2023.

In September 2022, we helped organize the 32nd Eurodysmorpho conference held in September 2022 in Barcelona. The ESHG-Young Vice-chair, Dr Ana Raquel Silva, was the Chair of the 6th session named "Syndrome delineation: neurology and central nervous system".

Dr Silvia Kalantari, Juliana Miranda Cerqueira and Dr Ana Raquel Silva represented the ESHG-Young at the ERN ITHACA Board meeting held in Budapest in December 2022. They gained a clearer view of the many projects and activities organized within the context of the ERN-ITHACA, leading to new collaboration ideas.

Our effort, led by Juliana Miranda Cerqueira, Dr Silvia Kalantari and Dr Ana Raquel Silva, along with Dr Celia Soares, managed a network of proof-readers for the Unique guides for rare disorders, allowing 27 new Unique guides to be present on the rare-chromo website. We also contributed to recruiting native French speakers to translate EuroGEMS pages. We are grateful for the work done by the volunteers and look forward to future collaborations for other translation and proofreading projects requiring the language expertise of young clinical geneticists.

In December 2022, led by Consultant Dr Elena Avram, we successfully orchestrated the "ESHG Observership for Young Geneticists" program to offer financial support for junior doctors from developing countries to visit the human genetics department in prestigious European countries and observe specific areas of clinical care or laboratory techniques.

In January 2023, for the second consecutive year, ESHG-Young Vice Chair Dr Delia Sabau led an initiative in collaboration with the European Board of Medical Genetics (EBMG) to organize a virtual session called "Becoming European Board Certified in Medical Genetics and Genomics". This session emphasized the significance of obtaining the European Certification in Medical Genetics and Genomics (ECMGG) certificate. It also provided essential information about the exam to young trainees.

In February 2023, we published an article detailing our achievements and activities in the last four years in the European Journal of Human Genetics (PMID: 36755105).

The board members of the ESHG-Young participated in the International Congress of Human Genetics in Cape Town in February 2023. We helped the African Society of Human Genetics and the Young Investigators Forum (YIF) plan to organize different sessions at the YIF forum and ICHG2023. Our work and collaborations with the YIF were widely acknowledged. We continue this partnership with the YIF by organizing a Manuscript Writing Workshop for Young Geneticists led by Rita Matos and Dr Magdalena Mroczek.

We also increased our representation in ESHG-affiliated committees. While Dr Rhys Dore was appointed an Observer in the Public and Professional Policy Committee (PPPC), Dr Delia Sabau and Dr Magdalena Mroczek joined the editorial board of the European Journal of Human Genetics as Section Editor and Editorial board member, respectively.
And finally, led by Dr Mridul Johari, we helped organize the Early Career Networking event - "Find your Genetic Match" at the ESHG2023, with approximately 400 registered attendees.

As the field of Human and Medical genetics continues to grow and expand, it is crucial to inspire and encourage the younger generation to significantly impact its development in Europe and beyond. Our goal for the next year is to continue fostering connections and providing opportunities for young trainees.

ESHG-Y are (in this order on the picture):
Mridul Johari (AU) Chair,
Delia Sabau (RO) Vice Chair,
Ana Raquel Silva (PT) Vice Chair,
Ana Rita Matos (PT) Secretary,
Silvia Kalantari (IT) Secretary,
Rhys M Dore (UK) Spokesperson,
Magda Mroczek (PL/CH) Spokesperson,

Elena Avram (RO) Consultant,
Juliana Miranda Cerqueira (FI) Consultant,
Francesca Tonini (UK) Associate Member

www.eshg.org/eshgy

The year in review at the European Journal of Human Genetics

by Alisdair McNeill, Editor in Chief EJHG

The year 2022-2023 has been a positive period for the European of Human Genetics. Our metrics continue to move in the right direction. Most importantly our impact factor rose again to 5.35 (2021 Impact Factor), and we are now ranked 35/175 in Genetics and Heredity journals. The number of submissions to our journal remains high (65 original submissions in April 2023 for example). Competition for space in the European Journal of Human Genetics is strong, with only around 25% of submissions being accepted.

We now wish to host special issues in the European Journal of Human Genetics at least once per calendar year. In 2022 we published the COVID-19 special issue. Which was well received, and highly read and cited. In 2023 we are soliciting submissions for a special issue on genomics in Low and Middle Income Countries. This should go to press in early autumn 2023.

A priority for the coming year is to revitalise the European Journal of Human Genetics social media presence. Twitter remains very strong with almost 8 000 followers. A new Social Media Editor will be appointed very soon. We will continue to focus on Twitter to promote papers being published in our journal and host monthly twitter journal clubs to discuss the significant findings of a selected paper each month. We will seek to integrate with developments in ESHG social media strategy. We believe that Social Media remains a vital tool to disseminate scientific information and permit widespread discussion of findings. I am considering whether it would be appropriate to host "social media interns" at our journal, to undertake a small amount of social media work as part of their training in genomics. For example, to help produce tweets to summarise papers and host journal clubs. I would welcome any comments - both for and against this idea.

We wish to publish the best genomics research (broadly defined) in the European Journal of Human Genetics, no matter the specialism of the authors or the geographical origin. For example, we welcome basic scientific papers, clinical work describing genomic conditions (but not single cases, unless exceptional) and papers on ethical, legal and social issues. It is important to note that we insist upon adherence to recognised ethical norms such as the declaration of Helsinki, informed consent and participant privacy.

We will have a strong presence in Glasgow for ESHG 2023. I hope geneticists of all varieties will find our Workshop on Saturday morning useful in providing ideas on how to write high quality genomics research papers. On Sunday, I will speak more formally on the paper submission and editorial process at the European Journal of Human Genetics. I hope as many of the editorial board and Section Editors will be present for the editorial board meeting as possible. Lastly, at the close of the conference I will present the Gert Jan van Ommen citation award for the top cited papers in European Journal of Human Genetics.

Of course, the success of our journal depends upon our excellent board of Section Editors and Editorial board members, along with the editorial office (Dr Shona Kirk) and our colleagues at Springer Nature. We also recognise the invaluable contribution of peer reviewers. Thanks to you all.

www.eshg.org/ejhg

European Board of Medical Genetics - Overarching Focus and Activities in 2022/2023

by
Birgitte Rode Diness, MD, PhD, President, EBMG
Clara Serra, Genetic counsellor, PhD, Incoming President
Isabel Marques Carreira, CLG, PhD, Past President

The EBMG council has throughout the year held monthly virtual meetings.

This focus for the EBMG over 2022/2023 has been on how the branches can help and support each other to succeed in the promotion of our shared goal ie. to serve the needs of patients using the genetic/genom-

ic services in Europe by developing, supporting and spreading high standards of practice.

We are pleased to report that the Standard Operating Procedures for all three branches: Genetic Counselors and Nurses (GCGN), Medical Geneticists (MG) and Clinical Laboratory Geneticists (CLG) have been finalised and published on the EBMG website. The three branches continue to work well in a regulated and efficient format on their core activities while the council has focused on the development of a strategy to increase and harness synergy between the branches.

The foundation for reporting the finances of the EBMG and the three respective branches in a more transparent way has been laid to allow reporting around income and costs to be divided and shared, when appropriate.
The visibility of EBMG has been another key issue addressed which has led to the decision of an EBMG presence on LinkedIn and a revision of the website. Please check it out! https://www.ebmg.eu

The three branches in 2022/2023

The **GCGN branch** is following their yearly cycle and has completed finalising renewals for registration for those professionals who have been registered up to 5 years previously. The branch has also called for new candidates which are now being assessed and is assessing MSc programs submitted from all over Europe for EBMG accreditation.

The **CLG branch** has processed requests for recertification (29) as well as new requests (32). The branch has also been active in the support of the French CLGs affected by legislative changes limiting their daily work. Also, all tests for candidates of Group 3 were completed for this round, independent from the ESHG meeting, by conducting the test at the institution of a CLG-committee member or a trusted registered ErCLG (eg. in India or Ireland).

The **MG branch** has, as a joint venture of EBMG, UEMS (Union of European Medical Specialists) and ESHG, once again successfully conducted a virtual exam to allow candidates the possibility to obtain the European Certificate of Medical Genetics and Genomics. Work is continuing on the revision of the European Training Requirements within the specialty. This work is also coordinated with the planned online book for trainees in Medical Genetics "APOGeE" (A Practical Online Genetics e-Education), a project initiated by ERN ITHACA.

The EBMG would like to take this opportunity to thank all members and collaborators once more for the important work you do in the branches and in your daily practice. This is the true foundation of the EBMG mission which includes the continued fostering of mutual support and respect between our professional groups, united in a common goal to provide good quality genetic care to patients.

www.ebmg.eu

Interview with Magdalena Zernicka-Goetz, ESHG Award Lecturer 2023

Magdalena Zernicka-Goetz is Bren Professor of Biology and Biological Engineering at Caltech, Pasadena, USA and Professor of Mammalian Development and Stem Cells at the University of Cambridge. She will be giving the ESHG Award Lecture on Saturday June 10 at 14.15 hrs.

Magdalena Zernicka-Goetz spent the first few years of her life living in a research institute. "My father was a neuroscientist in a Polish Academy of Sciences Institute in Warsaw. My family had lost everything, including their home, during the Second World War, so there was no money to buy an apartment. I grew up surrounded by people in white lab coats doing experiments."

At that time she found her unusual living conditions to be totally normal, as she'd never known anything else. "I was brought up partly by my father's colleagues and students, and even when we eventually moved into a normal apartment, we continued to have many scientists visit and stay with us. It might therefore have been expected for me to develop an interest in science, but in fact at first my passions were art and theatre. Art in the communist regime had a second meaning; it was a form of expressing freedom, a way to escape. Each of my paintings was conceived by an impact of the moment, to calm my heart, settle my imagination, and the stories just evolve while I am creating them. Many of our experiments evolve in the same way – not only through our logic, but also through intuition."

"In my mind, science and art are not totally apart, one from the other. As a scientist you must always be precise, logical, but also vulnerable and so honest in order to design experiments, truly see unexpected results and respond to them. Vulnerability is where the truth lies, which is also critical for art," she says. "I also spent a large part of my youth on the tennis court and in tennis competitions. It helped me to build resilience and ability to focus on the moment, as in tennis it is important to focus on each ball as it comes, rather than on planning to win the whole match. "

Her interest in science came much later. "It was a hard choice, but finally I decided to study biology and carry out my PhD work on early mammalian development. I embraced genetic approaches to understand how the embryo is built, to understand how it is possible to generate such a diversity of cell types starting from a single cell and within a very specific time window. This very beginning of our lives, the creation process per se, is what has always interested me most. "

After obtaining her PhD, Magdalena left Poland to become an EMBO post-doctoral fellow in the Wellcome Trust/Cancer Research UK Institute (now the Gurdon Institute) in Cambridge, where she started her own group in 1997 as a Senior Research Fellow of the Lister Institute and then a Senior Research Fellow of the Wellcome Trust. She became a Cambridge University professor in 2010. In 2019 she moved to establish her group at Caltech. "It was hard to leave Poland and my family and friends to start a new life in Britain, and now it is equally hard to leave Britain to start a new life in California, as I feel as British as I feel Polish by now. And it is Britain that invested in my development as a scientist. This is the main reason why I still have my lab in Cambridge too."

She is often asked how she manages to be daring and pioneering in new directions and still remain so productive, even now whilst managing labs on two different continents with an eight-hour time difference. "I feel most excited when we try to break into a new world for the first time, and we enter places that we don't understand and where we can't predict the outcome. There is fear, but also excitement. My parents, teachers, and, indeed, my husband say that I behave as if I have fire in the belly; I have an incredible drive to explore; to take risks and look deeper, and facing something I can't understand can be exhausting for me and sometimes for others around me. It could be a combination of my Polish and Jewish roots and difficult upbringing, that drives me to think about my life as an unusual gift granted for a limited time."

Interaction and mentoring are very important to her. "It is such a privilege to guide students and post-

docs along new paths. The act of discovery itself is so amazing. I think that our most striking discoveries resulted from establishing a technique for studying human embryos at stages that were previously entirely inaccessible, and in building complete mammalian embryo models by combining three distinct stem cell types representing the pluripotent epiblast and two extra-embryonic tissues."

Outside science she loves the theatre. "We go as often as possible, both in London and Los Angeles. Both of our children are artistic. Our daughter's poems melt my heart, and our son expresses himself by paining portraits (with the most vulnerable eyes)."

She will tell the conference about her fascination with what she calls the dance of life – how a fertilised egg, the foundation stone of biological form, progresses into the trillions of cells that make up an individual human being. "I will be talking about what I think is our group's biggest breakthrough in understanding – the progression of that dance; the many steps that we learned to take to be able to build embryo-like structures with the potential to develop progenitors of the brain and spinal cord and with beating hearts by combining together embryonic and extra-embryonic stem cells."

Interview with Shankar Balasubramanian, Mendel Award Lecturer 2023

Sir Shankar Balasubramanian is a Professor of Chemistry in the Yusuf Hamied Department of Chemistry at the University of Cambridge, UK, and Senior Group Leader at the Cancer Research UK Cambridge Institute. He will be giving the Mendel Lecture on Tuesday June 13 at 13.30 hrs.

There are not many world-leading researchers who are prepared to say that they went into science because they weren't good enough at football, but Shankar Balasubramanian admits it freely. "I grew up just outside Liverpool in a place called Runcorn and, as a kid, when I wasn't in the house, I'd be outside kicking a football. I grew up on that, and later was captain of my school team. Where I grew up if you played football, it was natural to dream of playing for Liverpool (in my case), and it wasn't until I was about 17 that I gave up on that dream."

He doesn't regret having become a scientist instead, though. "I don't consider what I do to be work. Like most scientists I do it because I'm driven by curiosity and the desire to explore." His curiosity started at an early age. "I can trace it back to primary school, or even earlier. I always wanted to understand how things work – whether it's machinery, devices, or the world around them. And as I learned more and more detail about the natural sciences it built an even greater intrigue around wanting to understand in considerable detail how the world works. Having inspirational teachers also played an important role in stirring my desire to be curious and to learn. As I look back, I can also see how vital it was to have parents who were supportive, while giving me the space and time to find my own path."

At school he first leaned towards mathematics, and then towards chemistry, but his chemistry teacher in his A-level years tried to persuade him to do medicine after school. "I ended up coming to Cambridge to do

a degree in Natural Sciences, which spans everything. Whilst I was leaning towards chemistry, I always had a broad interest across the natural sciences – physics, chemistry, and aspects of biology and medicine. So that probably explains why I have ended up as a researcher straddling disciplines."

Pursuing curiosity is personally fulfilling, but it isn't always clear whether what you're doing can actually make a difference to the world. "This involves luck and timing. Fundamental research can take 100 years or longer before it makes a difference in the world, so to see a practical implementation of your work in your own lifetime is hugely fulfilling. About 25 years ago, David Klenerman and I were doing some blue-sky research on how machines that replicate DNA work, by using new observation techniques. That led us to see a different way to sequence DNA as an unintended consequence of what we were doing. It set us on a pathway to develop the idea into a commercial system via a startup company, which we called Solexa. Our company and technology was later acquired by Illumina and led to the world's dominant method of sequencing genomes. The technology and the data it has generated are being used in research and also for clinical decision making. Routine human genome sequencing at scale has already had a substantial impact in cancer, rare diseases, and infectious diseases. The full impact will probably need another decade or two to be fully realised. I am fortunate to have had the opportunity to contribute to something that is having an impact on so many people and to see it happen during my lifetime."

Another major source of satisfaction is the teaching and mentoring of young people. "It's made me very happy and proud to see young people who I have mentored in my lab, or who I've taught as an undergraduate, go on to become successful in whatever they've done. Many have become hugely successful in the world of academic science or the biotech industry; several have won international prizes, and many have made major contributions to society."

Scientific research is by its nature unpredictable, he says, and short-term thinking in research can be unhelpful. "You should carefully choose a question that you think is, or could become, important and interesting, then stick at it and dig deep to ask good, probing questions. Over time, you will likely see or discover things that will add to our understanding and may make a significant difference in the long-term. It is essential that funders of scientific research take a longer-term view on evaluating progress and outcomes. Typically, it takes many years to go from an idea to building significant knowledge and understanding. The opportunity for transformative innovation is often unanticipated and can be lost by short-term research culture. I'd like to see research support that extends to ten-year horizons for certain types of projects, especially for younger researchers."

He often tells PhD students and researchers that many of the things they will try in the lab will "fail". But they won't actually be failures – that's just a perception. "Failure is often the best way to learn."

He still has the research proposal for his work with David Klenerman that led to the development of Solexa-Illumina Next Generation Sequencing. "From the proposal you could not see where this would all end up, yet it led to a method that been commercialised and is making a difference in the world. This happens in science more often than people realise, and I think we need to structure science in such a way to enable such opportunities to happen. New concepts emerge from basic research. So for applied research and commercialisation to be successful, funding agencies must support and maintain a very strong foundation of basic research."

Retirement is not on the cards, at least not just yet. "I don't think I'll ever stop being a scientist, at least not in terms of thinking about science. But there are some things I haven't done enough of, and maybe in future I will make more time to pursue them. I enjoy endurance running, but you need a lot of time to do that. I also want to spend time more widely exploring the world and its natural beauty. I took up wine tasting and wine collecting a few years ago. There's no greater pleasure than having a nice bottle with friends and chatting. Every bottle of wine has a story …"

He will be telling the conference about his journey between chemistry and genetics. "I've spent much of my 30-year career working on aspects of DNA, probing its structure and function. I'll talk about some of my adventures in the field; the primary sequence of DNA, which is genetics, and some aspects of the natural chemistry of DNA that fall under the umbrella of epigenetics. My lab has spent more than 20 years probing a four-stranded DNA structure – a quadruple helix. So, the third area I'll talk about will be about DNA structures other than the double helix. Each adventure started with chemistry and progressed towards biology and medicine."

Interview with Tara Clancy, ELPAG Lecturer 2023

Tara Clancy is an Honorary Consultant Genetic Counsellor/Senior Lecturer in Manchester, UK and is now Chair of the BSGM's Ethics and Policy Committee. She will be giving the ELPAG Award Lecture on Tuesday 13 June at 14.15 hrs.

Tara Clancy's interest in healthcare started when she was young. "My younger brother died of leukaemia when we were kids, and my dad died of gastric cancer when I was a teenager. I think it was those two events that had the most impact on my choice of career, though I also enjoyed studying biological sciences at school."

She went to university in Manchester, where she took a degree course that "was a combination of science, social science and philosophy, and I liked that it incorporated subjects beyond basic science. Afterwards, although I wasn't sure what I wanted to do in the long term, I knew that I wanted to work in the public sector, particularly the National Health Service and probably in a clinical role. So, I then trained as a nurse. I found that the training lacked teaching about counselling skills, such as how to discuss challenging issues with patients and their families. This led me to do a certificate and then a diploma in counselling theory and skills. A post was advertised in the Clinical Genetics Service in Manchester as I was completing the diploma course.

"I've often felt that I've been fortunate to be in the right place at the right time – not really in a passive way, but in situations that opened up opportunities. This was one of them, and I moved into genetic counselling, which in turn led to my deepening interest in ethics. I did an MA in healthcare ethics and then a PhD in law and ethics, both at the University of Manchester. My career evolved along with my interests, and one of the things this led to that I found especially rewarding was being appointed as a member of the Nuffield Council on Bioethics."

Mentoring students, trainees and colleagues earlier in their careers has been important to her. "Because it's a fairly small world, I have been able to follow their careers. I have tried to encourage others and facilitate their choices. I was given opportunities myself and could make the most of them, but if you don't offer others opportunities, then they can't develop and progress professionally to their full potential."

Genetic counselling is as much, if not more, about listening as it is about explaining, she says. And this doesn't apply to healthcare alone. "I think that the fact that we, as humans, often find it hard to really listen to each other, have discussions and come to agreement (or to agree to disagree) is regrettable. And the pandemic, while it didn't create this problem, seems to have exacerbated it. Too many of us focus on winning arguments without attempting to understand the other point of view. But it doesn't have to be like that."

If she could have chosen any occupation apart from healthcare, she would have liked to do something creative like glassblowing or sculpture. "But I don't think I really have any artistic talents."

Since giving up clinical work, Tara has trained as an accredited paediatric medial mediator. She is also involved in professional activities at a national level such as chairing an NHS England Policy Working Group. In terms of other interests, she is taking a pause. "I think that's a good thing to do from time to time, although I am tempted to try something more creative and artistic."

She will be telling the conference about how she sees the problem of people not listening to and hearing each other and suggesting some solutions. "I'd also like talk about the relationship and tensions between (apparently) objective scientific realities and people's real-life experiences."

EUROPEAN SOCIETY OF HUMAN GENETICS

AGENDA
General Assembly 2023
Monday, June 12, 2023, 12:15 - 13:15 hrs
Lomond Auditorium, SEC Glasgow, UK

1. Opening by the President of the Society, Borut Peterlin
2. Approval of the Agenda and of the timely invitation of members
3. Approval of the Minutes of the last Meeting in 2022
4. Report on the Activity of the Society 2022-2023, Borut Peterlin
5. Financial Report of the Treasurer 2022, Gunnar Houge
6. Reports of the Auditors 2022, 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, Valérie Cormier-Daire
10. Results of the nomination for President-Elect 2023 - Confirmation by the Membership
11. Results Board Member nominations 2023 - Confirmation by the Membership
12. Thanks to leaving Board Members
13. Vote on amendment of the statutes
14. Vote on Membership Fees 2024
15. Site of future European Human Genetics Conferences
16. Any other Business

Note the change of day and time: The GA is now taking place on Monday, June 12, 12:15 hrs