THE EUROPEAN SOCIETY OF HUMAN GENETICS
NEWSLETTER
No. 39 - May 2024

Report from the President

by Valérie Cormier Daire,
President of the ESHG 2023-2024

Dear ESHG members,

Welcome to the 57TH ESHG conference in Berlin, where the number of attendees surpasses 5300, a fully hybrid meeting for those of us who are unable to travel for various reasons.

The European Society of Human Genetics has grown significantly over the years, evolving alongside the remarkable advances in the field of genetics, genomics and precision medicine.
Our profession has also evolved. Variant interpretation has demonstrated the importance of bioinformatics tools and expertise, but also the need for multiomic approaches and the development of innovative technologies. The integration of genomic data into clinical practice has transformed the way to diagnose, treat, and manage genetic disorders but also highlighted the importance of an integrated clinical-biological approach.

Our society encompasses a wide range of expertise, spanning scientific researchers, engineers, clinical and molecular geneticists, cytogeneticists, nurses, genetic counselors, bioinformaticians, and professionals from complementary and multidisciplinary fields. This diversity is our strength, as it allows us to approach complex issues from various perspectives and to collaborate effectively towards common goals.

The contributions of boards and committees have been instrumental in shaping the trajectory of our society, addressing key aspects such as scientific expertise, policy statements, educational courses, but also involving the young generation (ESHG-Y) and creating a new strategic committee. This allows us to embrace the full spectrum of our society interests and needs, from fundamental research to translational applications, from personalized medicine to equity in healthcare access.

The link with other societies and with the European Board of medical Genetics is also an essential aspect of our society for promoting best practice of medical genetics, sharing of expertise, fostering collaboration, and harmonizing practices across different regions and disciplines.

As a clinical geneticist, working in the field of rare diseases and member of an European Reference Network, one of my goals has been to encourage formal collaborations between the ESHG and ERNs. Indeed, the link between ESHG and ERNs lies in their shared mission to advance knowledge, improve patient care, and outcomes through collaboration, expertise sharing, education and training. Shared guidelines, educational courses, and initiatives aimed at promoting multidisciplinarity may ensure that the benefits of genetic and genomic advancements reach patients and families in a more meaningful and equitable manner.

As professionals working in the field of human genetics, we hold unique positions within families and communities. Taking the time to listen to patients, understanding their needs, and involving them in decision-making processes are essential aspects of our practice. Our commitment to patient-centered care extends to partnering with patient associations, advocating for quality-of-life initiatives, and ensuring that patients remain at the heart of our concerns. By partnering with these organizations and actively involving patients and families in the research process, we can ensure that our work remains relevant, impactful, and responsive to their needs.

Finally, I would like to thank all those who have contributed during my presidency: to the society and board members, committee members and chairs, the Executive Committee, and of course, Jerome del Picchia and the Vienna Medical Academy. All my very best wishes to my successor, Professor Bill Newmann.

Report from the Scientific Programme Committee

By Alexandre Reymond, Chair of the SPC

My name is Alexandre Reymond and I am the chair of the ESHG Scientific Program Committee (SPC, https://www.eshg.org/spc). Its thirty-four members are representative of the breadth of our discipline and their main mission is to determine the scientific contents of our annual conference.

While our ESHG 2023 conference in Glasgow gathered 5,488 human geneticists (4,402 registrants and 1,086 exhibitors) we hope to see you even more numerous in Berlin. Like Glasgow, our Berlin conference will again be fully hybrid allowing geneticists who cannot attend in person to also participate. After our three hybrid conferences in Vienna 2022, Glasgow 2023 and Berlin 2024, we will assess if there is a steady demand and if streaming live serves the community well. For example, 1,090 Glasgow registrants (25%) caught the possibility of assisting remotely.

For Berlin ESHG2024, we have put together a dense program of five award lectures, forty-one invited sessions (30 symposia,10 educations sessions and one plenary) totaling 110 invited speakers, 23 workshops for a more hands-on experience and three "Get2gether" sessions to promote networking and professional connections. We also selected 192 abstracts for platform presentations and 30 for best poster pitch presentations from a total of 3153 submitted abstracts (3089 regular and 64 late-breaking abstracts), a new record!

Presented with this flurry of abstract submissions, we decided to change our setup on the fly. We will thus have up to nine concurrent sessions in parallel in Berlin. This means also that 6.1% of submitted abstracts rather than 5.3% of them will be presenting from our platforms. Is this large number of submissions associated to our German host and/or is this due to the growth of our field? We will precisely monitor this evolution in the future and act accordingly as we want our annual conference to present on-going science and continue to give center stage to early career geneticists. As a point of comparison, we should mention that 7% and 9.5% of the abstracts submitted to ESHG2023 Glasgow and ASHG2023 Washington were selected for platform presentations, respectively. Multiple and not mutually exclusive solutions should be explored, e.g. (i) change the balance between categories of sessions, (ii) have more sessions in parallel, (iii) extend the number of parallel sessions on the Saturday morning.

We are looking forward to hosting you in Berlin. Welcome to Germany!

www.eshg.org/spc

Report from the Policy and Ethics Committee

By Francesca Forzano, Chair of the PEC

In 2023, The Public and Professional Policy Committee (PPPC) has been renamed Policy and Ethics Committee (PEC). This 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, GA4GH and ASHG-Social Issues Committee.
Since 2023, the Committee took on an additional role, to provide ELSI support to the EJHG board in selected cases. An updated Terms of reference is under review and will be published soon on the PEC webpage.
In 2023-2024, the PEC has been active on the following topics:
Polygenic risk scores in pre-implantation genetic testing. PEC members continue to be involved in discussions around this topic, and have contributed

to an international workshop and conferences including the first conference on polygenic embryo screening (Boston 2023) and HUGO 2024 (Rome). Watch out for the session on Sunday June 3rd (ESHG S15 Preimplantation genetic testing for polygenic conditions)!
In 2023, we have run a Survey on Expanded Preconception Carrier Screening in Assisted Reproduction in European centres or gamete banks participating in the European IVF-monitoring consortium, a joint activity with ESHRE. The paper is in press in Human Reproduction.
We recently ran a Survey on Opportunistic genomic screening, to ascertain the impact of the ESHG recommendations on this practice in European countries – thank you to all the ESHG members who have answered! We aim to present the results by the end of 2024.
A new, interactive course has been created by PEC that will launch in October 2024: Covering The Gaps - The Course You Didn't Know You Needed. It will be a pilot version – if successful, it might be integrated in the ESHG portfolio. The course will be run virtually on 2 days and will be particularly addressed to young scientists and clinicians. The course aims to cover topics that are important for your genetics research and practice but often neglected or taken for granted. It will cover issues of quality, regulation, ethics and justice in diagnostics and research. We will look at data sharing and biobanks, at returning results to patients and reporting research.
We are 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 the ESHG membership for their review around summer 2024.
2025 will be a busy year for PEC. We will work together with the Eurogentest Committee and the International Society for Prenatal Diagnosis to draft Recommendations on reporting genetic tests in prenatal diagnosis.
We will start working on a document on AI in genomics, in collaboration with Global Alliance for Genomics and Health (GA4GH).
We have been successful in obtaining a competitive grant at the Brocher Foundation, where we will run a workshop on Rare Diseases, Genomics and Justice. This is a joint collaboration between PEC and a research team led by Ramona Moldovan funded by MRC UK. We are extremely grateful to all our distinguished invited speakers, who have demonstrated an incredible enthusiasm and support. The outcome of the workshop will be published on a special issue of the Journal of Community Genetics, including a meeting summary and the key recommendations.
And last, but not least, massive congratulations to our PEC member Prof Yves Moreau, who has been Individual Award Winner in 2024 of the Einstein Foundation Award for Promoting Quality in Research, as a recognition of his assiduous work as advocate for ethical standards in the utilization of human DNA data.
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 PEC via the Chair. ESHG members interested in becoming a PEC 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 PEC in 2023/2024 were: Angus Clarke, Christophe Cordier, Guido de Wert, Francesca Forzano (Chair), Sabine Hentze, Heidi Howard, Milan Macek, Béla Melegh, Álvaro Mendes, Yves Moreau, Markus Perola, Inga Prokopenko, Dragica Radojkovic, Emmanuelle Rial-Sebbag, Rosaline Favresse (EURORDIS), Vigdis Stefánsdottir, Fiona Ulph and Carla van El (Secretary-general). Current observers: Olga Antonova, Elena Avram, Yalda Jamshidi. ESHG-Y representative: Rhys Dore.

www.eshg.org/pec

Report from the Education Committee (EduComm) 2023-2024

By Inga Prokopenko, Chair of the EduComm

The ESHG is committed to advancing the excellence and understanding of Human and Medical Genetics across Europe.

Over the past year, the Education Committee has experienced a period of both productivity and enthusiasm, as we have made significant progress on multiple initiatives. We are pleased to welcome Rita Matos to our committee, who will focus particularly on strengthen-

ing our ties with ESHG-Young as their representative. We extend our heartfelt gratitude to Bill Newman, who served as Chair of the committee since 2020. Bill has transitioned to a new role as the ESHG president, and we are thankful for his dedicated leadership. Taking over the role of Chair from April 2024 is Inga Prokopenko.
Additionally, we commend Sofia Douzgou Houge for her exceptional leadership of the successful Genetic Sounds, the ESHG Podcast Series.

ESHG Courses
Our ongoing commitment includes a diverse portfolio of in-person and virtual educational courses. For the latest list of courses and their details, visit https://www.eshg.org/courses. We welcome your suggestions to broaden the scope of our course offerings. Additionally, ESHG provides fellowships to support young scientists and clinicians from economically disadvantaged countries in attending these programs.

Educational App
The free App course on pediatric neurogenetics, delivered by X-Peer and launched in 2023, has garnered significant attention. With monthly approximately 750 channel visitors, 1600 visualizations, 80 tests taken, and 60 diplomas obtained for the three modules covering diagnostic approaches of genetic syndromes and inherited metabolic disorders; developmental and epileptic encephalopathies and brain malformations; and the new module on movement disorders, neurodegenerative diseases and neurocutaneous syndromes. To enroll in the course, please register for free at https://web.xpeer.app/web/en/courses/340?channelId=46. CME points are available for completing this program. We encourage you to explore this valuable resource and contribute suggestions for additional app-based courses.

EuroGems update - led by Ed Tobias (Glasgow)
The ESHG educational website, https://www.eurogems.org/, is experiencing a steady rise in visitor traffic, with users from 140 countries accessing the site. We are delighted by the growing engagement and increased accessibility resulting from offering the resource in four languages. Particularly noteworthy is the surge in visits to the Spanish, Portuguese, and now professionally translated French pages. Alongside the growing number of European users, educators from countries such as Canada, Japan, Singapore, India, and Egypt have praised its utility. Ed continues to enhance the website and develop the associated free educational genomics terminology apps, available at https://www.genomicsapps.org. He welcomes comments and suggestions at edward.tobias@glasgow.ac.uk.

International Links – co-led by Edith Coonen, Liz Loehrer, and Carmen Navas
With the focus on promoting genetic education, we established links with other international genetics societies across the world. We will share educational resources and develop joint educational initiatives.

Podcast Program - curated by Sofia Douzgou Houge and in collaboration with the Whitworth group
ESHG, in collaboration with the Whitworth group and the ESHG-Young Committee, has launched Series 3 of Genetic Sounds, available at https://www.eshg.org/geneticsounds. This monthly podcast (released six times a year) delves into various genetic topics, now that genomics has taken center stage in medicine. Using a conversational approach and featuring esteemed guest speakers, the podcast covers a wide range of subjects related to genetics and genomics. Since its inception (Series 1), the podcast has consistently garnered high engagement across all continents. All episodes are accessible on multiple platforms, including Buzzsprout, Apple, and Spotify. Additionally, there will be a live podcast event at the ESHG conference in Glasgow, so be sure to watch out for it on the program. We believe this podcast is an effective way to engage with both professionals and the general public, and we welcome ideas for future topics. Furthermore, we have established a partnership with the Eurordis Rare on Air podcast, available at 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.
One of the mentorship awards from 2024 onwards is 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. We encourage genetic counsellors to apply for the Mentorship award.

Awardees of the ESHG Mentorship scheme for 2024 are:

- Onda-Tabita Calugaru (Romania)
- Robert Field (Ireland)
- Anna Hovhannisyan (Armenia)
- Deivid Souza (Brazil)
- Ivan Tourtourikov (Bulgaria)

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.
schemes.

Awardees of the ESHG Observerships scheme for 2024 are:

- Teodora Barbarii (Romania)
- Maria Chiara Baroni (Italy)
- Daniela Bohme (Chile)
- Simona Jakovchevska (Macedonia)
- Hilal Pırıl Saraçoğlu (Turkey)

The next round of applications for these two programs will be live in August with a closing date in November 2024.

DNA Day Essay and Video contest – led by Christophe Cordier

DNA Day, April 25, is commemorated internationally as a celebration of Genetics and its promises. For the 15th year, ESHG sponsored a DNA Day Essay and Video contest in high schools all over the world. Given the growing impact of Artificial Intelligence (AI) on our lives, the ESHG decided to make AI part of the contest instead of banning it. this year's question was: Ask an Artificial Intelligence chat of your choice to write a 350-word essay on the topic: "Is the human Y-chromosome vanishing in the future?" discuss the result and its consequences, should the public believe the content of the AI essay was actually true. We received the largest ever response - 220 essays and 20 videos. The winners will be announced during the ESHG meeting in Berlin.

If you have any ideas/comments regarding the work of EduComm please contact me at i.prokopenko@surrey.ac.uk .

www.eshg.org/educom

Annual Report ESHG-Euro-Gentest Committee 2023

by Gert Matthijs, Chair of the EuroGentest Committee

ESHG-EuroGentest focuses on quality aspects for genetic services, prepares professional guidelines related to genetic diagnostics, provides training and represents the society at international forums related to genetic services.

The IVD-Regulation presents a major challenge to genetic diagnostic laboratories especially on the use of in-house developed tests 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.

The Task Force on IVDR led by Els Dequeker aims to share information with diagnostic laboratories as well as to make clear to the community and regulatory authorities that there are certain concerns for genetics in relation to the implementation of the IVDR. During the ESHG 2023 conference in Glasgow, a Get2Gether meeting was organized with several international experts. Attendance was high (>100) with good interaction between the speakers and the audience.

ESHG had previously delegated Els Dequeker to BioMed Alliance, a group of European medical societies active in lobbying at the European level, where she became a member of the Task Force on In Vitro Diagnostics. Gert Matthijs has recently also joined this Task Force.

Together with the BioMed Alliance and EURORDIS, ESHG has drawn the European Commission's attention to what has been termed 'orphan diagnostics'. A letter was sent to DG Sante, expressing the concerns on the impact of certain provisions on our community and urging the European Commission to take action. The good news is that the European Commission has taken actions to ensure the availability of *in vitro* diagnostics, and will assess the impact of the legislation on devices responding to special needs, such as the 'orphan devices' and on the development of innovative devices in Europe. For

once, the Commission has also indicated that special attention shall be given to costs and administrative burden stemming from the implementation of IVDR.
During the Belgian presidency of the European Union, we had the opportunity to present the difficult and complex situation of rare diseases and 'orphan diagnostics', and the importance of innovation at a meeting of the national competent authorities; they have the task of following up the in-house tests in each country.

ESHG-EuroGentest was 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 2023, the fifth (and last) course was organised by Dr Krystyna Chrzanowska and co-workers in Warsaw (Poland).

The 2023-2024 EuroGentest committee members are Christi van Asperen, Isabel Maria Carreira, Els Dequeker, Weronika Gutowska-Ding, Milan Macek Jr, Aleš Maver, Michael Morris, Katrin Õunap, Simon Patton, Christine Saban, Nicola Wolstenholme. I gratefully acknowledge the time and effort they spent to voluntarily contribute to ESHG-EuroGentest achievements and output over the last year.

In the coming year(s), we embark on the following projects:

Harmonization in genetic counselling
This is a challenging issue, aspects of quality in clinical practice are more difficult to evaluate and impose than in the laboratory, hence a daunting task to provide guidelines. A good questionnaire for national societies would be a practical solution to get the work going. In close collaboration with the European Board of Medical Genetics (EBMG), we intend to update previous reports and try to publish about the current situation in Europe.
Poor performance in EQA Harmonisation of the definition among EQA providers is necessary: for this purpose, it is necessary to have representatives from all of them and recruiting them will be the first focus of this workgroup. The second objective will be to define how to act on performance: given that the ESHG is a professional body, and all national societies are part of it, this issue could and should be addressed.

Reporting in prenatal testing
We plan to produce recommendations on reporting genome-wide diagnostic tests in prenatal settings. This will be a joint endeavour with the International Society for Prenatal Diagnosis (ISPD) and with the Policy and Ethics Committee (PEC) of ESHG. Given that ISPD is a global society, the aim will be to develop a globally applicable guideline. The work may also incorporate recommendations on the type of tests to apply and the related indications.

Update of FMR1 guidelines
Best practice guidelines provide a reflection of what peers think about good practice in diagnostics and quality management. Professional guidelines are useful as a basis for accreditation of laboratories and may be used as a reference to good practice and standard-of-care in juridical disputes. A working group is currently being created to update the guidelines for molecular testing and genetic reporting of Fragile X syndrome (FXS). The available document dates back to 2014, and the technique that is mostly used to test the FMR1 expansion has now changed to be based on triplet repeat PCR.

If you are interested in participating in the ad hoc working groups, let us know! We also wish to recruit new volunteers for the EuroGentest committee. If you are interested, please join us on Saturday June 1st from 10am to 12noon in room M5 at the CityCube Messe (https://www.messe-berlin.de/en/organizers/our-locations/citycube/overview/), or 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) has once again demonstrated its unwavering commitment to developing and empowering young geneticists across Europe. Throughout 2023-24, ESHG-Young has been instrumental in fostering educational opportunities, networking, and professional growth, paving the way for the next generation of leaders in human genetics.

Dr Mridul Johari (Chair) and Juliana Miranda

Cerqueira (Consultant) have continued representing ESHG-Young, actively contributing to the ESHG Scientific Program Committee. Their participation has been vital in shaping an engaging and insightful program for the upcoming ESHG 2024 conference.

Dr. Ana Raquel Silva (Vice-Chair) and Dr. Silvia Kalantari (Secretary) have enhanced our collaboration with ERN-ITHACA, participating in the scientific committee for the 2023 and 2024 Eurodysmorpho conferences, attending a board meeting in Dublin (December 2023), and organising a webinar on 'noninvasive fetal autopsy techniques'. Raquel and Rita Matos (Secretary) have also been involved in EduComm, improving educational outreach.

Silvia, Raquel, and Mridul recently submitted a manuscript to the European Journal of Human Genetics (EJHG). Their paper discusses vital insights from ESHG 2023 workshop W03, «Termination of pregnancy: Legislation and ethics», shedding light on the diversity of laws on termination of pregnancy for severe fetal anomalies across Europe.

Another notable milestone was the publication of our paper, PMID: 37880422, co-authored by ESHG-Young in partnership with the African Society of Human Genetics-Young Investigator Forum. This publication underscores the success of intercontinental collaboration in advancing human genetics and highlights significant global outreach efforts. Building on this success, and assisted by Dr Magdalena Mroczek (Spokesperson), Dr Rhys Dore (Spokesperson), Mridul, Juliana and Rita, our collaboration expanded to include Jax Laboratory, resulting in an online webinar on Functional Genomics. This event drew approximately 200 researchers from diverse African and European nations, further emphasising the impact of our collaborative efforts.

Rita and Juliana recently joined the ESHG social media team, where they have significantly contributed to community engagement through innovative content and effective communication strategies, greatly enhancing society's digital presence. Meanwhile, the ESHG-Young social media team, composed of Magdalena, Juliana and Rita, continues to produce exceptional content and advertisements, enriching our outreach on various social media channels.

Rhys continued his involvement with the Policy and Ethics Committee. Magdalena and Delia continue to represent the ESHG-Young on the Editorial Board of EJHG.

We recently surveyed the topic of Young Geneticists Societies (YGS), asking ESHG trainees about their awareness of YGS and identifying ways to boost their representation. Insights from the survey have prompted us to strengthen our international ties. Mathys Weber from the Société des Internes et jeunes Généticiens de France will work on our international relations as the new Associate member.

Dr Delia Sabau (Vice-Chair) joined the ASHG Membership Engagement Committee. Mridul was appointed to the working committee of the Human Genetics Society of Australasia (Western Australia branch), advancing our intercontinental collaboration efforts.

ESHG-Young is excited to host several events at the ESHG 2024 Conference designed to foster networking and collaboration. These include a MatchMaking event, spearheaded by Mridul and Juliana, offering trainees direct interactions with recruiters, mentors, and PIs. In partnership with the German Young Geneticists Society, Junge Humangenetik, we also organise a Get2Gether session to promote exchange and unity among young genetics societies. Lastly, the 'Cruise and Connect' boat event during ESHG 2024 promises to be a highlight, providing a unique setting for ESHG trainees to build connections while cruising along the Havel.

ESHG-Young steadfastly commits to improving the representation and education of young geneticists. Intent on expanding its influence and reach, ESHG-Young is dedicated to fortifying international connections and championing essential changes in education and policy within the genetics community. We are ready to guide young geneticists toward a brighter, more inclusive future in human genetics.

ESHG-Young 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
Juliana Miranda Cerqueira (FI) Consultant

www.eshg.org/eshgy

Social Media Committee Annual report June 2024

by James Fasham, Chair of the SMC

The European Society of Human Genetics Social Media Committee plays an important role in supporting the society's engagement with social media. Our core objectives include:

1. Evaluating the Evolving Landscape
   - We assess the dynamic and ever-changing social media landscape.
   - We guide ESHG on which platforms to engage with and how to maximize our impact.
2. Expanding ESHG's Reach
   - We strive to maximize ESHG's presence across selected social media channels
   - These currently include X/Twitter, Facebook, Instagram, and LinkedIn.
3. Effective Promotion and Engagement
   - We utilize social media strategically to promote ESHG's activities and encourage active participation from our members.
   - Our focus extends to promoting ESHG membership.
4. Annual Meeting Participation
   - We actively promote attendance at the annual meeting.
   - During the event, we facilitate online engagement to enhance the overall experience.
5. Metrics and Adaptation
   - We produce and evaluate usage metrics to continually assess our performance.
   - Based on these insights, we adapt our strategies to achieve optimal results.
6. Risk Management
   - We proactively address any potential reputational risks associated with social media usage.
7. Enhancing Communication Skills
   - We provide opportunities for ESHG members to improve their communication skills through social media.
   - This is achieved by educational workshops and participation in the committee.

The ESHG Social Media Committee was founded in 2023 at the request of the Annual Meeting Committee, who appreciated the growing importance of social media as a tool to further the aims of the society to interact and engage with society members.
Dr. James Fasham and Dr. Aleena Mushtaq co-chair the committee. Working closely with Prof. Joris Veltman (Chair of the Annual Meetings Committee), Jerome del Picchia (ESHG Executive Officer), and Oscar Pachero (ESHG Administrative Officer) they convened an initial team comprising Juliana Miranda Cerqueira, Rita Matos and Dr Teodora-Maria Barbarii ahead of ESHG 2023 in Glasgow.
During ESHG2023 this group were supported by Dr Alex Cagan, Amanda Picini, Dr Danya Vears and Jose Patricio Miranda. Together they created and amplified ESHG2023 content, reporting from every corner of the meeting, and acted as the society's eyes and ears. The results were over 5,000 posts that used the #ESHG2023 hashtag and over 7,000 engagements across the meeting.
This was the first ESHG meeting where there was an ESHG Instagram presence, managed by Dr Aleena Mushtaq and focusing on short video pieces with high likely engagement.
In July the committee met again to consider its terms of reference, establish medium-term goals and specifically to draft a comprehensive Social Media Strategy. This meeting established the roles of the members and interactions with the ESHG board.
The ever-changing social media landscape posed challenges in 2023. Twitter's rebranding to X, following its acquisition by tech entrepreneur Elon Musk, led to decreased engagement and splintering of attention between competing platforms. Concurrently, Threads, a competing social network by Meta, initially gained traction but later lost market share among disenchanted Twitter users. The committee monitored and responded to this, ultimately deciding not to leave X/Twitter, but opting to diversify with Kiharu Webb joining the committee in September with the aim of investigating how we could effectively establish and utilise a presence on Linkedin.
The committee met again in April to discuss the strat-

egy ahead of ESHG2024 and to review the social media policy. We have been joined by Dr Mo Wafik, Dr Pilar Cachero, Dr Marco Vismara and Rahema Mohammad. We look forward to another highly engaged annual meeting.

Beyond this, goals for 2024-2025 include:

1. To increase the total number of posts and follower numbers for the main ESHG social media accounts.
2. To broaden the representation of the community to include those of differing ages, specialism and demographics Specifically to encourage international membership and participation, especially from outside Europe

Dr. James Fasham and Dr. Aleena Mushtaq

www.eshg.org/smc

Report from the Strategic Committee

by Borut Peterlin, Chair of the SC

The Strategic Committee (SC) has been established as a new ESHG committee in 2023 to shape the vision of the society, positioning the ESHG as the key European and international hub in genomic medicine. The increasing evidence for genetic contributions to human diseases, along with technological advancements, is namely rapidly reshaping medical practice.

The SC's mission is to analyze emerging trends, identify challenges, and recognize opportunities for translating cutting-edge genomic science into clinical practice. With a focus on advancing genetic science, diagnosis, prevention, treatment, and care for genetic and complex diseases, the committee formulates strategic initiatives and provides informed guidance to the ESHG Executive Board.

Key Objectives of the Strategic Committee:

- Developing the ESHG Strategy: Crafting a comprehensive strategy for the society to guide future actions and priorities.
- Enhancing the Role of Genetic Professionals: Defining and promoting the critical role of genetic professionals within health systems.
- Fostering Integration of Genomic Medicine: Building strategic collaborations with medical societies, patient organizations, industry partners, and other stakeholders.
- Establishing a Platform for Genomic Medicine: Creating a forum for discussing and advancing genomic medicine within EU and international health systems.
- Influencing Health Policies: Shaping health policies to support the integration and advancement of genomic medicine.
- Encouraging Public Engagement: Promoting dialogue and engagement with the public regarding the use of genomic information.
- Promoting Professional Development: Supporting continuing education and professional growth for ESHG members and other stakeholders.

These objectives represent a selection of our key initiatives, highlighting some of the critical areas of focus. In its efforts, the SC will seek advice from the ESHG board, national societies, other ESHG committees, and the entire ESHG membership. Our main priority for 2024/2025 is the development of the four-year ESHG strategy.

www.eshg.org/sc

A review of activity in the EJHG

by Alisdair McNeill, Editor in Chief EJHG

In 2023 EJHG, the official journal of the European Society of Human Genetics, continued to publish a wide spectrum of genomics research. Including work relevant to clinicians, bioinformaticians, population geneticists and social scientists. From 1st January 2023 to 31st December 2023 the European Journal of Human Genetics received 825 submissions. Our acceptance rate is 25%. In 2023, there were well over 2 million downloads of articles published in the European Journal of Human Genetics

and over 5000 social media mentions. Publications were submitted from a range of different countries - amongst accepted manuscripts the top 3 submitting regions were the United States of America, England and Germany. We continue to try and broaden our reach and welcome submissions of relevant research globally.

The latest (2022) impact factor of the European Journal of Human genetics is 5.2 (with a total of 13 086 citations), which is stable. According to the impact factor the European Journal of Human Genetics is ranked 30th out of 171 Genetics and Heredity journals (up from 35th last year). The latest (2022) citescore of the European Journal of Human Genetics has risen to 9.1, and the journal is now ranked 11th out of 90 Genetics (Clinical) journals by Scopus. This is the highest ever journal ranking for the European Journal of Human Genetics by impact factor or citescore. It is worth bearing in mind that these journal rankings also include other journals which publish only review papers, and which could thus have inflated citations. Of note, open access manuscripts are significantly more likely to be cited than those published subscription only. Publishing in the European Journal of Human Genetics remains competitive and we believe it to be a mark of prestige for your research.

We remain committed to promoting research published in the European Journal of Human Genetics by social media work. The European Journal of Human Genetics X (Twitter) account has around 7 500 followers. We are in the process of appointing a social media editor, embedded within a clinical fellow role, at the University of Sheffield. They will be tasked with growing the social media following for the journal, exploring new platforms and running promotion campaigns such as monthly online journal clubs.

In 2024-2025 the European Journal of Human Genetics will be looking to broaden its range of Section Editors, to better reflect our community. We will publish a call for this in due course. I will be at ESHG in Berlin 2024, please do let me have your thoughts on the journal in person, if you wish.

Once again, thank you to all the peer reviewers, editorial board members and section editors who make our journal function. A special mention goes to our editorial assistant Dr Shona Kirk.

www.eshg.org/ejhg

European Board of Medical Genetics - Report 2023

by Clara Serra, Genetic counsellor, PhD, President

During this year, the EBMG has focused on the improvement of our internal organization and on the dissemination of the mission of the EBMG across Europe, which is developing, supporting and spreading high standards of practice in genetic/genomic services in Europe. The collaboration among the three different branches has been based on the monthly online meetings held throughout the year.

Considering that positions on the board rotate frequently (such as the presidency every year), it became evident that it was necessary to develop documents to systematically preserve knowledge and experience each time positions changed. This strategy aims to make the transition from one person to another more effective by facilitating the transmission of information about roles and responsibilities. With this goal in mind, a document has been elaborated for each position.
In terms of spreading awareness about the EBMG mission, a comprehensive presentation has been prepared specifically for EBMG affiliates who are interested in bringing our work, including all three branches, at their respective national societies' meetings. Also, as part of the EBMG dissemination, the Counsel has worked on the website update and on the preparation of the Get2Gether session during ESHG in Berlin entitled "Medical Genetics Professionals: how can we fit future requirements?"

The three branches

The GCGN branch is progressing through its annual cycle and is currently completing assessments for first-time applicants; 12 portfolios have been evaluated. The branch is now focused on renewing registrations for professionals who were registered up to 5 years ago. Additionally, the branch is evaluating MSc programs submitted from across Europe for EBMG accreditation; currently, there are 9 master's programs accredited by the board.

The CLG branch has currently 432 registered professionals from 42 countries, with 17 applications pending. They have continued to work diligently on achieving European-level recognition for the profession, through the European Commission.
The Medical branch contributed to the revision of the European Training Requirements (ETR) for Medical Genetics, a project that spanned over 2 years and was approved and adopted by the UEMS Council in October. This document could be highly beneficial in updating national training requirements and elevating national standards. Additionally, the Medical branch organized a workshop focused on exam preparation, covering Multiple Choice Questions and Oral Assessments. The workshop attracted 23 participants from 11 countries, including a CLG from Latvia, demonstrating the close collaboration between branches.

We want to express our gratitude once again to all EBMG members and affiliates for the significant contributions you make within the branches and in your daily practice. This commitment forms the cornerstone of the EBMG mission, encompassing mutual support and respect among our professional branches dedicated to delivering high-quality genetic care. Thank you!

www.ebmg.eu

Interview with Cecilia Lindgren, ESHG Award Lecturer 2024

by Mary Rice, ESHG Press Officer

Professor Cecilia Lindgren is Professor of Genomics of Endocrinology and Metabolism at the University of Oxford, Oxford, UK. She will be giving the ESHG Award Lecture on Tuesday, June 4, at 14.15 hrs.

Cecilia Lindgren's interest in science started when she was very young. "My grandmother was a scientist and a very inspirational person who loved questions. She taught me and my brother the value of hard work and being grateful for opportunities given, but maybe more importantly to ask questions and to think them through properly." She had an idyllic childhood growing up in a location by the sea outside of Gothenburg, but then disaster struck. "My brother became very ill with a brain tumour when I was nine years old. That led to a lot of questions about why some people get sick, but others don't. Why one can rarely cure disease? That's something I've carried with me ever since and have had a real urge to find out."

As a result, she thought for a while about studying to be a medical doctor, but her parents thought she was over empathetic and would sink herself in the process of trying to help. "Though that would be great for patients, it would be too hard on me because it would be difficult for me to accept that I wouldn't always be able to help people as much as I might want to. So a career in science came naturally because that's also a way of making a difference and having an impact."

At school, she loved mathematics, chemistry and biology, so maths was her first-choice subject at university, and this led to a biomedical degree course. "I was never happier – it was like opening an Easter egg and finding that you focus and apply yourself to do what you really love." Though, at the beginning, she was worried about taking a course that didn't lead to a licence. "If you're a dentist, you obtain a licence and then you know you can get a job afterwards. You know what your job is, what the hours are, and what the outputs are. Science is much more uncertain and open-ended – you can work as much as you want all the time, and the scientific process is the product in a way that doesn't apply to jobs that are more clearly defined."

After completing her PhD in molecular genetics at the University of Lund, Sweden, during which she spent a year and a half at the Centre for Genome Research, Whitehead Institute, US, she started postdoctoral work at the Karolinska Institute. Itching to go abroad again, she joined the Wellcome Trust Centre for Human Genetics at Oxford on a Throne Holst Foundation Scholarship. She left in 2012 to join the Broad Institute, Harvard, and MIT as a Scholar in Residence, before returning Oxford in 2015 to join the Oxford Big Data Institute, which she has led as Director for the last three years.

The core belief that science is a team activity has completely permeated her entire career. "I love collaboration and believe it is a key to success in today's science eco system. I've been very lucky with the people I've worked with, who have been kind and scientifically brilliant with a really generous and collaborative work ethic. They also gave me the passion for mentoring others, and showed me how

important it is. I began supervising students when I was a PhD student myself. I prefer it to teaching – which I don't mind – but I feel that mentorship and supervision is more valuable and impactful."

A supervision that really stands out for her was at the Broad Institute, under David Altshuler, Eric Lander, Mark Daly and Joel Hirschhorn during her PhD. "While I was there, we made two seminal breakthroughs. And that gave me such a high – it's almost impossible to describe. In the first, we found that PPAR gamma was unequivocally associated with type two diabetes. At that time, association studies were cluttered with ambiguity, partly because of small sample sizes and study designs, and also a bit of over-interpretation in the field. So, finding this definitive association was fantastic."

"I was also fortunate enough to collaborate with Vamsi Motha and others at the Broad Institute in looking at gene expression studies in skeletal muscle in people with type two diabetes. We showed, for the first time, that the OXPHOS pathway was dysregulated in a concerted way in these patients."

For a PhD student to be involved in such high impact projects was "fantastic", she says. And she has followed it up with major contributions of her own, particularly in the field of obesity, where her work has made a substantial contribution to understanding its genetic landscape. "My team has been able to underline the distinction between fat distribution and overall obesity, to identify the strong sexual dimorphism involved, and to look for ways in which this is linked to female and reproductive health."

The focus on mentoring comes back again when she talks about today's scientific culture. "As scientists, we need to make sure that we provide an amazing environment for people to work in, and that they want to stay and feel they can flourish in. We all develop with the times and we have to remain nimble, not just in discovery, but in our work environment too."

Cecilia Lindgren can't imagine a life where she did not do science in some form, but that doesn't mean that she has no interests outside of it. "I love being with my family, and my friends; they are the most important thing in my life. I love reading, travelling, and all kinds of water sports. I love skiing, and being outdoors generally. When I have to retire, I will have more time for these other interests. But that's hopefully 15 years or so away, and I don't ever intend to give up mentoring people."

She will be telling the conference about her work in obesity genetics over the last 20 years, and what are the biggest lessons she and her team have learned. "We're at quite a pivotal time where we have the maps of the underlying genetics for many common complex traits. We have thousands of genetic associations, and now it's up to us to translate that into impactful outputs for patients and society. There are a number of ways in which this can be done.

"I'll also be sharing a couple of nuggets of unpublished data that point to what we should be doing in this field. And of course, I will talk about the joy of doing science and mentoring."

Interview with James R. Lupski, Mendel Award Lecturer 2024

by Mary Rice, ESHG Press Officer

Professor James R. Lupski is the Cullen Professor of Genetics and Genomics in the Departments of Molecular and Human Genetics, and of Paediatrics, at Baylor College of Medicine and Attending Physician Texas Children's Hospital, Houston, Texas, USA. He will be giving the Mendel Lecture on Tuesday, June 4, at 13.30hrs.

It's not uncommon for eminent researchers to say that their interest in science started with figuring out how things work, taking them apart, and reassembling them. But to continue by researching their own health problems is much less frequent. However, that's the path down which James Lupski travelled.

"As a child, I always liked to take things apart and put them back together, and I had a particular penchant for doing this with the neighbour's lawnmowers, making motorised bicycles that morphed into minibikes and small motorcycles. I also liked to make my own fireworks, having taught myself to make gunpowder during my teenage years, and became terribly excited by some of the explosive results," he says.

Lupski was born In Hicksville on Long Island in New York State. His grandmother's mother fled Ukraine during the Russian Revolution and arrived in New York seven months pregnant. His father's mother was born a US citizen two months later. His father was an electrician, and his mother a busy homemaker who also was a teacher of culinary arts in adult evening education classes at the high school. "They married the week dad graduated from high school and had five children before he was 25 years old."

He went to the same high school as they did, but he was home-schooled during most of period he should have spent there because he had developed a condition that caused foot deformities and difficulties in walking. "I had ten operations during high school and the first few years of college. After each, I spent considerable time in a wheelchair or on crutches while I recovered, so I spent most of high school being home-schooled. I only had tutoring for a couple of hours a day, four days a week, leaving me with plenty of free time to explore my curiosity."

His elder brother had also developed similar problems though much earlier in life, and was originally given a diagnosis of muscular dystrophy, but when James developed the same symptoms it was clear that there was an underlying familial genetic cause. Eventually this was shown to be Charcot-Marie-Tooth neuropathy (CMT).

"It was my home tutor who exposed me to DNA for the first time. The genetics that I tried to understand from the standpoint of transmission of traits with respect to my brother and myself was now the reality of a tangible molecule and the chemistry that explains the biology of the living organism. In college, I elected to study chemistry because of my fascination with the subject, but also completed biology as a major given the number of biology courses I needed to take to fulfil the requirements to apply to medical school."

After obtaining a BA from New York University in 1979, followed by a PhD and MD from the same institution, he moved to Texas with a combination of a paediatrics residency and clinical genetics training at Texas Children's Hospital. There he came across

Tom Caskey, who was a Visiting Lecturer at the NYU Medical Scientist Training Program whom Jim hosted whilst a student at NYU; Tom was in the process of forming a new Institute for Molecular and Human Genetics at Baylor College of Medicine in Houston. The Institute wanted to recruit young geneticists in all areas of genetics – bacteria, yeast, fruit fly, mouse, and human – to form a bridging department between research, clinical practice, and clinical laboratory diagnostics in clinical and human genetics. "I had been particularly intrigued by reading as a graduate student about the mapping of the Huntington's disease locus and thought it would be interesting to try to take this experimental approach in CMT disease, which although usually transmitted in an autosomal dominant manner seemed to be an autosomal recessive trait in my family," he says.
Although several dozen genes were identified by Lupski and others as possibly resulting in CMT disease, that which was responsible for the disease in his family remained elusive. It was the advent of next-generation sequencing that made personal genome sequencing feasible, both practically and financially. "The BCM Human Genome Sequencing Centre completed some of the first personal genomes, including Jim Watson's and mine. For my genome, it was extremely exciting to find the disease-causing variant alleles and the gene responsible for CMT disease in my family. Our family was particularly helpful to research efforts because all family members volunteered to participate in the studies and three generations of samples were available for study, but it was an n = 1 family. Given the size of my family and the fact that DNA samples had been collected from my grandparents 25 years earlier, the three-generation pedigree and multiple affected and unaffected siblings maximised the interpretation of per-locus sequencing data."

In addition to his work on CMT, Lupski has studied many other neurological disorders with a focus on neurodevelopment and neurodegeneration, as well as genomic disorders, eye diseases, birth defects, and more. "I use the human as a model organism and disease phenotypes to study genetics and the biology of disease," he says.

He will be telling the conference about what he calls "Biology in balance: gene dosage and genetic models as paths to characterising and mitigating disease" – a delve into the way in which the principles of biological balance and complementarity underlie all of life science, evolution, evolutionary medicine, and genomic medicine. And he won't forget to mention his feeling of privilege at being both a human subject and a direct stakeholder in these research efforts. "Not many people have the opportunity to participate in human genetics and genomics research, both as subject and physician investigator."

Interview with Angus Clarke, ELPAG Lecturer 2024

by Mary Rice, ESHG Press Officer

Angus Clarke is Emeritus Professor in Clinical Genetics at Cardiff University, Wales, UK. He will be giving the ELPAG Award lecture on Saturday, June 1, at 14.15 hrs.

Angus Clarke had 'very medical ' parents, but, he says, they would have been happy whatever career he had decided to pursue. However, he chose to follow in their footsteps, first of all hoping to become a psychiatrist. "But I found so many other things along the way that were too interesting to pass by – genetics, then, paediatrics, and then paediatric genetics," he says.

His interest in genetics started at Cambridge, where he heard talks from some of the most renowned geneticists of the time, such as Sydney Brenner and Gabby Dover. "They were hugely stimulating and encouraged me to find out more about the subject."

After obtaining a BA in genetics, he qualified in clinical medicine, working first in general medicine and then in paediatrics. As a research registrar in clinical genetics at Cardiff, he studied the clinical and molecular genetics of ectodermal dysplasia, and later developed an interest in Rett syndrome and neuromuscular disorders while working with John Burn in Newcastle upon Tyne. On his return to Cardiff in 1989, he continued these interests. "I was lucky enough to work with Peter Harper, who gave me the flexibility and control over my own activities that enabled me to pursue clinical duties and teaching obligations, while maintaining the possibility of developing my research interests.

"The combination of all three was vital to me and I thrived because of it, though it meant that research activities could only be pursued once my other tasks

had been completed, so I was often writing and pursuing research late into the night, or even in the early hours of the morning. This was not very family friendly!"

He regrets some of the changes that have taken place in UK science funding and universities since his early days. "I am unhappy about the way access to funding has been subject first to fashions in thinking in the 20 years up to 2010, and since then to across-the-board cuts. All this meant that there were lots of good projects that many researchers (not just myself) could have worked on, but which didn't get funded."

Perhaps as a result of these funding problems, careers have suffered too. "The career structure for junior post-docs today is lamentable, as is the conversion of universities into businesses."

Angus Clarke is 'retiring rather slowly'. He no longer holds regular clinics, but continues to see patients occasionally, and is still involved in research and teaching. Once he retires fully he won't be short of things to do. "There is so much going on in the world that is interesting, and so many great books to read. I'd also like to improve my languages and perhaps learn a few more." And that's not to mention trying to find ways of visiting his far-flung grandchildren. "Finding a way to visit my grandchildren without flying links, in turn, to a desire to find a way for people of my generation to be able to leave the world in a better state than we found it."

He will be telling the conference about his early clinical and research experiences, and how this engendered his interest in social science and ethics. "The time I spent talking to families across the UK for my first research post shaped what I went on to do later, and particularly my focus on the 'lifeworld' factors that influence family communication about genetic conditions and also patient decisionmaking."

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AGENDA
General Assembly 2024
Monday, June 3, 2024, 12:15 - 13:15 hrs
Room A3, City Cube Berlin, Germany

1. Welcome by the President of the Society, Valérie Cormier Daire
2. Approval of the Agenda and of the timely invitation of members
3. Approval of the Minutes of the last GA in 2023
4. Report on the Activity of the Society 2023-2024, Valérie Cormier Daire
5. Financial Report of the Treasurer 2023, Gunnar Houge
6. Reports of the Auditors 2023, 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, William Newman
10. Results of the nomination for President-Elect 2024 – Confirmation vote by the Membership
11. Vote on New Deputy Secretary General
12. Results Board Member nominations 2024 - Confirmation vote by the Membership
13. Thanks to leaving Board Members
14. Vote on Membership Fees 2025
15. Update on sites of future European Human Genetics Conferences
16. Any other business

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