THE EUROPEAN SOCIETY OF HUMAN GENETICS

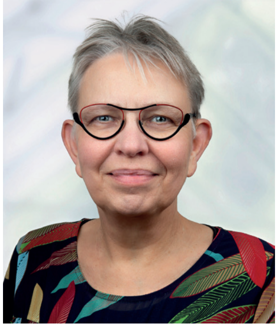
NEWSLETTER

No. 41 - June 2026

Report from the President

by Martina Cornel,
President of the ESHG 2025-2026

Dear ESHG members,



On a global scale the last year has been a difficult year with wars and disasters, and a political climate that does not always support science. Still our society is doing very well. ESHG continues to organize courses and other collective activities, the most important one being the upcoming Gothenburg conference. ESHG provides grants, awards and mentorships to individual scientists.

A new activity that started in 2025, the monthly webinar series, is still very successful. The recordings are available from the YouTube channel ESHG Webinar Series on YouTube (www.eshg.org/yt). If you missed any of the webinars, you can watch them at any moment. The first two episodes in 2026 covered genomic newborn screening, a very relevant topic which also will be covered at the conference in two sessions. Furthermore, the Journal EJHG has a special issue on DNA in public health screening programs (https://www.nature.com/collections/djigcbgbif).

The year was also used to further professionalize the society. The ESHG statutes were updated to be in line with current Austrian legislation. The support from the Vienna Medical Academy (WMA) was broadened. We now also have Oscar Pacheco Martinez as Chief Operating Officer next to Jerome del Picchia as Chief Executive Officer as well as staff with whom we have been collaborating most happily over the years.

In terms of policy development, early in the year ESHG responded to developments in Greece where genomic newborn screening was planned with lack of openness and transparency (www.eshg.org/news 2025). We also expressed concern when SRY testing was required to determine athletes' sex by several international and national sports federations. Our concern was related to the medical and psychosocial support required in the case of an unexpected test result and to the quality of testing. The Policy and Ethics committee also published the ESHG recommendation on cascade counselling and testing (https://pubmed.ncbi.nlm.nih.gov/41392321).

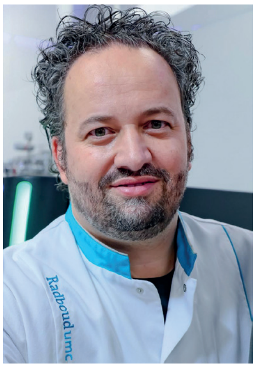
Our Strategy Committee had started discussing the ESHG Policy for the future, and to make that a shared vision answering to the demand of non-genetics experts and the patients' organisations. A co-creation process took place in the beginning of 2026, with a workshop in April 2026 in Utrecht, the Netherlands, with the Executive Board and other Committee Chairs, a delegation of the Board and WMA. The White Paper on the external strategy will be presented at the Gothenburg conference.

As Executive Committee we have attended the American Society of Human Genetics meeting in Boston in October 2025 and discussed how we can most effectively work together in protecting and enhancing genomic medicine for patient and public benefit. We look forward to the International Federation Human Genetics Societies' meeting in Guadalajara, Mexico 1-5 March 2027 International Congress of Human Genetics (https://ic.relagh.org).

My successor as President of the ESHG is Professor Angus Clarke. I wish him a very successful and enjoyable time. It has been my privilege to be in the role for the past year working with all of the very active members of the society and its Committees, and the excellent WMA support.

Report from the Scientific Programme Committee

by Alexander Hoischen & Alexandre Reymond, Co-Chairs of the SPC



We look back to a record-breaking ESHG 2025 in Milan: 6,913 participants human geneticists (4,523 registrants, 470 exhibitors and 658 industry participants from 94 countries), including 1,262 participants online.

Each year, we face the challenge of further improving the annual conference. The main task of the SPC is to showcase the most exciting science and current human genetics state-of-the-art at the annual conference. To achieve this goal, we have grown to 36 members, allowing us to better represent the breadth of topics of our specialty, as well as a larger number of countries and continents.

The ESHG conference is back in Gothenburg for a third iteration after 2010 and 2019. These points of comparison show how much our community has grown in recent years. For example, the number of submitted abstracts has increased by more than 50% since 2019 and we now start with a full program of seven parallel sessions already on Saturday morning rather than in the early afternoon.

We have invited 105 speakers to present their results at the ESHG 2026 Gothenburg conference through 4 award lectures, one plenary, 26 symposia, 7 educational sessions (with 51% invited female scientists and speakers from 26 countries across all invited sessions), 18 interactive sessions for a more hands-on experience and 5 “Get2gether” sessions to promote networking and professional connections.

These will be combined with 252 platforms presentations selected from a total of 3,171 blindly scored abstracts (3,132 regular and 39 late-breaking abstracts). Remarkably, and congruent with the ESHG mission of continuous outreach and education, they were submitted by geneticists from a record-breaking 95 different countries.

Faced with an ever-increasing number of submitted abstracts, we have intentionally slightly diminished the number of invited sessions and increased the number of concurrent sessions offering platform presentations to about 7.5% of submitted abstracts. This year, we are particularly proud to see that 52% of all abstracts selected for oral presentation will be presented by “early career scientists (ECS)”. With a growth in the number of (junior) presentations, we shall present 3 additional Early Career Awards (ECA) this year, with a total of 12 awardees for oral presentations.

This year, 72 best poster abstracts were selected to also present a “lighting talk” (https://www.nature.com/articles/d41586-021-01674-9) describing their work. These pitches, conceived and coordinated by the head of the poster jury Dr Lot Snijders Blok, will take place during the first half hour of each of the (newly increased) 6 poster sessions scheduled this year.

Finally, there are several other points ESHG supports: (i) ESHG will continue to offer childcare service on site; (ii) ESHG will sponsor a total of 231 travel fellowships in 2026; (iii) ESHG continues to support junior scientists in many levels (please also see the newsletter of the ESHG-Young Committee, and https://www.eshg.org/); (iv) ESHG shall continue to offer text captions with the on demand videos to support non-native speakers and people with auditory challenge; (v) finally, we are also excited that ESHG 2026 will host the very first “ESHG Women’s Forum”, chaired by Ana Beleza, Demetra Georgiou and Julia Baptista, bringing together professionals at different stages of their careers, to share experiences, exchange ideas, and explore practical strategies for career development, leadership, wellbeing, and equity in a demanding and evolving field.

And for those who cannot get enough of ESHG, we continue to offer three pre-ESHG Educational Courses, starting Thursday and Friday prior to the ESHG conference (https://2026.eshg.org/satellite-meetings/eshg-courses/).

This exciting program can be followed both in person and remotely, as we will, for the fifth year in a row, offer a fully hybrid format that allows geneticists from around the world to participate for a reasonable nominal fee.

We hope you will enjoy EHSG 2026 Gothenburg as much as we will. We cannot wait to meet colleagues and friends from all over the world and engage, network and learn from each other.

Welcome to Sweden and (almost) midsummer to meet old friends, make new friends and discuss science.

We are looking forward to hosting you. Varmt välkomnande,

Alex & Alex

www.eshg.org/spc

Report from the Policy and Ethics Committee

by Francesca Forzano, Chair of the PEC

The Policy and Ethics Committee (PEC) 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.

PEC 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.

The PEC also provides ELSI (Ethical, Legal and Social Implications) support to the EJHG board and the Executive Board in selected cases, as well as to other Committees of the ESHG, for instance the SPC, if and as appropriate.

In 2025-2026, the PEC has been active on the following topics:

A second edition of the virtual interactive course ‘**Covering The Gaps - The Course You Didn’t Know You Needed**’ (https://www.eshg.org/covering-the-gaps) was held in November 2025. As feedback continues to be extremely good, we are planning to make it a regular yearly course in the ESHG portfolio. A new edition will be likely offered in November 2026. The course will continue to 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.

The document on ‘**Cascade Counselling and Testing**’ (https://pubmed.ncbi.nlm.nih.gov/41392321/), a strategy still underexploited across Europe, has been published, and by the time you will read this Newsletter, the documents ‘**GDPR and what does it mean in practice for our genetic community**’ and ‘**Misuse of genetics and genomics towards group and population discrimination**’ will have been submitted for publication, while also our paper on the ‘**Survey on Opportunistic genomic screening**’, which we ran last year among the ESHG members to ascertain the impact of our previous Recommendations in practice, is being finalised.

The members of PEC together with the Eurogentest Committee and the International Society for Prenatal Diagnosis (ISPD) have drafted ‘**Recommendations on reporting genetic tests in prenatal diagnosis**’, which have been opened to ESHG members comments in March 2026. The draft integrated with the comments received has been submitted to the ISPD and ESHG boards for their final review and approval, and will likely be submitted for publication before the end of 2026. We sincerely thank all the members who have sent suggestions! But please do not relax too much now, as we are almost ready to submit for membership review another document, this time on updated ‘**Recommendations on Genetic testing in children**’.

Thanks to our PEC member Dr Sabine Hentze, we had a very fruitful meeting at the **EMBL** headquarter in Heidelberg with **BIAC** (Bioethics Internal Advisory Committee - https://www.embl.org/about/info/bioethics/) members. We discussed possible future collaborations on educational activities, as well as ethical and legal issues, which are relevant for both institutions. We were very impressed by the scale and the variety of the projects run in collaboration with EMBL, which is a testament of the dynamism and quality of scientific research across Europe.

The forthcoming two main projects on our desks relate to possible issues associated with the use of **AI in genomics** and the interplay between **Genetic testing and insurances**, which is a rapidly evolving field.

Many topics remain on our agenda. These include, but are not limited to, considerations for an international policy on gamete donation, issues related to parental access requests to trio and foetal WGS data on their foetuses and children, the creation and potential use of synthetic gametes, assessment of quality of somatic gene therapy and gene editing, and the most appropriate use of language in Genetic Counselling.
ESHG members having suggestions for new topics to work on, or being 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 PEC Chair.

Members of the PEC in 2025/2026 were: Olga Antonova, Elena Avram, Linda Battistuzzi, Christophe Cordier, Francesca Forzano (Chair), Maurizio Genuardi, Sabine Hentze, Yalda Jamshidi, Milan Macek, Deborah Mascalzoni, Alvaro Mendes, Yves Moreau, Kelly Ormond, Inga Prokopenko, Emmanuelle Rial-Sebbag, Vigdis Stefánsdottir, Fiona Ulph and Carla van El (Secretary-general). Observers: Mahsa Shabani. EURORDIS representative: Rosaline Favresse. ESHG-Y representative: Rhys Dore. Collaborators: Guido de Wert, Heidi Howard.

www.eshg.org/pec

EUROPEAN SOCIETY OF HUMAN GENETICS

ESHG Newsletter No. 41, June 2026
Editors: C. Oliveira, J. Baptista, J. del Picchia
Production Assistant: M. Seidl

c/o WMA GmbH, Alser Strasse 4
1090 Vienna, Austria
office@eshg.org - www.eshg.org

Report from the Education Committee 2025-2026

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 enjoyed a particularly productive and energising period, making meaningful progress across a range of initiatives. We are pleased to welcome to our committee Claire Vandledonck, Mridul Johari, Maria Francesca Di Feo and Magdalena Mroczek, who will focus on online courses, international relations, and links with ESHG-Young through training, visibility and outreach. We are keen for Milena Teles and Yaqoub Ashhab to join as EduCom observers to improve diversity and address disparities in genetics education around the globe. We are happy to announce that James O'Byrne will take on the Co-Chair role in June 2026 for two years. We extend our heartfelt gratitude to Gulcin Gumus, Edith Coonen, Liz Loehrer, Carmen Navas and Feliciano Ramos, who have served on the committee since 2022 and Christophe Cordier, who has served since 2016.

ESHG Courses

Our commitment to genetics education includes a growing 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 report on new successful courses "Translational Epigenetics in Precision Medicine", organised by Eka Kvaratskhelia in Telavi, Georgia in October, 2025 and "Masterclass on Skeletal Dysplasias", organised by Valérie Cormier-Daire in Paris, France in March, 2026, which brought together 28 and 29 participants from multiple countries, respectively. We introduced the online "Presentation Skills Workshop", led by Julia Baptista and aiming to support the development of communication skills for ESHG conference speakers, which was a great success for speakers and our conference, the second edition is scheduled on May 19, 2026. The second successful edition and the third planned in November 2026 of "Covering The Gaps - The Course You Didn't Know You Needed", led by Francesca Forzano and Angus Clarke, show continuing need for training in ELSI issues among our membership and beyond. We are excited to announce a new 11-13 November 2026 edition of the in person " 3rd ESHG Training Course on Pharmacogenomics", led by William Newman and John McDermott in Manchester, UK. We are looking forward to a new ESHG Course: "Tools for Communication and Competency Skills in Genetic Counselling", led by Christophe Cordier in Bertinoro, Italy on November 15-19, 2026. We continue our support for the Leena Peltonen School of Human Genetics, organised at the Wellcome Genome Campus, Hinxton, UK on 26-30 July, 2026 by Zoltan Kutalik. The committee welcomes suggestions from the membership to broaden the scope of our course offerings. The ESHG sponsored courses benefit from ESHG fellowships to young scientists and clinicians from economically disadvantaged countries to attend these programs.

Webcast Archive of Conference Talks and Webinar Programme – in collaboration with the ESHG Scientific Programme Committee (SPC)
The ESHG continues to strengthen its digital education portfolio through its comprehensive conference webcast archive (2016–2025) of Educational Sessions and Plenary Lectures on YouTube (https://tinyurl.com/mt247e6e and https://www.eshg.org/education/conference-video-webcast-archive) and the expanding webinar programme https://www.eshg.org/webinarseries and https://www.eshg.org/education/webinar-conference-library/eshg-webinar-series-archive, delivered in close collaboration with the ESHG Scientific Programme Committee (SPC). These resources provide on-demand access to a substantial library of expert-led presentations, ensuring continued learning beyond the annual conference.
The ESHG webinar series delivers regular, high-quality online events featuring leading experts and addressing timely topics across research, clinical practice and education. Complementing this, the conference video webcast archive offers a rich repository of recorded sessions spanning nearly a decade, covering key devel-opments across human genetics and genomics. Recent webinars have continued to attract strong participation, showcasing diverse perspectives and reinforcing the Society's commitment to accessible, high-quality education.

EuroGems.org update - led by Ed Tobias (Glasgow)
The ESHG educational website, at https://www.eurogems.org, continues to undergo development and regular updates. It has now attracted visitors from 150 countries across the globe, including all of Europe and numerous countries across the Americas, Asia, Africa and Australia/Oceania. We are delighted by this expanding engagement and by the increased international accessibility made possible through offering the resource in four languages. A particularly encouraging development has been the rising number of visits to the professionally translated Spanish, Portuguese, and French pages, which are being accessed from a wide range of countries both within and beyond Europe. In addition to the growing user base, educators from many regions, including Canada, Japan, Singapore, India, Egypt, South Africa, Argentina, and Mexico, have expressed strong appreciation for the site's value. Ed Tobias (who also serves as the current HUGO-International Education Committee chair and provides a valuable link to that organisation) continues to enhance and expand the website and the associated free educational genomics terminology apps, available at https://www.genomicsapps.org. Feel free to comment and suggest new educational content to edward.tobias@glasgow.ac.uk.

International Links – co-led by Edith Coonen, Liz Loehrer, Carmen Navas and Ed Tobias
With the focus on promoting genetic education, we continue to expand our world-wide connections and representation with new EduCom membership from Brazil – Milena Teles, Australia - Mridul Johari, and Arabic speaking countries - Yaqoub Ashhab. The international relations team has focused on developing an overview of common educational needs and priorities in genetics across professions, countries and regions, including guidelines for harmonising the EU Undergraduate Medical Genetics Curriculum (with contributions from Feliciano Ramos). We are strengthening connections with genetics educators and professionals to make better use of existing human genetics resources that meet their needs, while also supporting the development and promotion of new tools to address remaining gaps in genetic education.

Podcast Program - curated in collaboration with the Whitworth group
In collaboration with the Whitworth group and the ESHG-Young Committee, ESHG has delivered three successful seasons of the Genetic Sounds podcast, available at https://www.eshg.org/geneticsounds. Re-leased monthly (six episodes per year), the podcast explores a wide range of topics in genetics and genomics—now central to modern medicine—through an accessible, conversational format featuring expert guests. Since its launch, it has attracted strong global engagement, with episodes available on platforms including Buzzsprout, Apple and Spotify. Building on this success, Season Four is now in development, with an expanded scope of topics and broader international contributions. A live podcast event is also planned at the ESHG conference in Gothenburg.

This initiative continues to be an effective way to engage both professionals and the wider public, and we welcome suggestions for future topics.

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 €1,500) for early career geneticists to visit another centre and attend the ESHG conference. The award will establish a long-term career supportive relationship.
We encourage genetic counsellors to apply for the Mentorship award.
Awardees of the ESHG Mentorship scheme for 2026 are:

- Leman Binokay, from Turkey to Estonia
- Reyhaneh Dehghanzad, from Iran to UK
- Huda Farah, from Qatar to UK
- Asli Karacan Konrat, from Turkey to France
- Nikolina Tomic, from Bosnia and Herzegovina to Sweden

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 ESHG Observerships scheme for 2026 are:

- Amin Abdurrahman Abdul Rashid, from Malaysia to UK
- Emre Akbas, from Turkey to Germany
- Hanna Havriushenko, from Ukraine to UK
- Marin Ogorevc, from Croatia to Slovenia
- Marija Rozevska, from Latvia to The Netherlands

The next round of applications for these two programs will be live in August with a closing date in November 2026.

DNA Day Essay and Video contest – led by Christophe Cordier
Every year on 25 April, DNA Day celebrates the discovery of DNA's double-helix structure in 1953 and highlights the transformative role of genetics in science and medicine. For the 18th year, the ESHG is proud to sponsor the European DNA Day Essay and Video Contest for high school students. This initiative invites young people to think critically about pressing issues in genetics, deepen their understanding of the science, and reflect on its wider societal implications. The 2026 theme, "Genes and Justice: How should genetic information be used in the legal system?", challenges participants to explore how DNA is applied in forensic investigations, the ethical complexities of ancestry-based surveillance, the protection of genetic privacy, and the risks associated with the misuse of genetic data in legal contexts.
Students may submit either a 750-word essay or a short video (up to five minutes). An international jury of genetic experts will evaluate the submissions, and the winners will be announced in June 2026, with awards presented at the European Human Genetics Conference in Gothenburg.

Together, these initiatives significantly extend the reach and impact of ESHG educational activities, supporting continuous professional development and fostering international collaboration. 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 EuroGentest Committee 2025

by Gert Matthijs, Chair of the EuroGentest Committee

The ESHG-EuroGentest committee focuses on quality aspects for genetic services, preparing professional guidelines related to genetic diagnostics, providing information and representing the interests of the society at international forums related to genetic services. The EuroGentest committee also prepares the policy and training activities of ESHG in relation to IVDR.

The 2025-2026 EuroGentest committee members are Joana Barbosa Melo, Isabel Maria Carreira, Christophe Cordier, Donna Darmanin, Els Dequeker, Jenni Fairley, Judit García-Villori, Weronika Gutowska-Ding, Thomas Liehr, Milan Macek Jr, Gert Matthijs, Aleš Maver, Michael Morris, Katrin Õunap, Rebecka Ann Pestoff, Christi van Asperen and Spiros Vittas.
We gratefully acknowledge the time and effort they spent to voluntarily contribute to ESHG-EuroGentest achievements and output over the last year. Two members rotated off: Christine Vianey-Saban and Nicola Wolstenholme. We sincerely thank them for their commitment over many years.

Early in 2026, the terms of reference of the committee were updated. Members will stay on for 4 years, once renewable, and a 3-year term will be adopted for chair and co-chair. Weronika Gutowska-Ding has been elected co-chair. She will take over the chair from Gert Matthijs by the end of 2026, who has chaired the committee since EuroGentest was formally integrated into ESHG in 2012. The number of members has been set at 20, and an announcement for new members was launched early in March. The committee was specifically looking for motivated experts in quality management, newborn screening and clinical genetics, while looking for a broad geographical and professional representation. By the time you read this newsletter, new colleagues will be on board. Please check the website for the actual membership.

On April 1st 2026, **EuroGentest organized the online seminar on Updates on quality assurance, regulations and professional standards in clinical and laboratory practices**. The recordings of the presentations are freely available (registration required, see https://www.eshg.org/courses/eshg-eurogentest-webinar#c6428).

IVDR-related activities: the ESHG-EuroGentest Task Force on IVDR (Els Dequeker, Milan Macek Jr, Gunnar Houge, Thomas Liehr, Gert Matthijs, Joris Vermeesch)
The IVD Regulation poses a significant challenge for genetic diagnostic laboratories, particularly regarding the use of in-house developed tests when no CE-IVD kit is commercially available for the same diagnostic purpose. The joint ESHG-EuroGentest Task Force on IVDR aims to inform diagnostic laboratories and raise awareness within the genetics community and towards regulatory authorities, about key concerns related to the implementation of the IVDR. Leading this effort is Els Dequeker, who was previously appointed by the ESHG to represent the society in the BioMed Alliance, a coalition of European medical societies engaged in lobbying at the European level. Through this role, she became a member of the European Commission's Task Force MDCG on *In Vitro* Diagnostics and IVDR Orphan Diagnostics.

Over the past year, efforts have continued to highlight concerns regarding the loss of innovation and the availability of IVD devices for orphan diagnostics. A dedicated Medical Device Coordination Group working group on IVDR orphan devices was established and has already convened several times. Through the ESHG – BioMed Alliance, we had the opportunity to provide examples of orphan devices that currently remain unaddressed in the IVDR and its associated MDCG documents.

In response to the call for evidence on the IVDR, a survey was distributed to all ESHG members. We sincerely thank all our colleagues who have responded to the survey!
Thanks to the strong response, we were able to submit a well-documented report in March 2025, reinforcing seven key points with data from the survey to highlight the challenges genetic services face under the current IVDR framework.

The main challenges identified include:

1. Ensuring harmonization in the monitoring of IVD device safety within healthcare institutions.
2. Clarifying the distinction between CE-IVD devices and in-house IVD devices.
3. Addressing the shortage of CE-IVD devices in genetic testing while maintaining space for innovation.
4. Assessing the requirement to use CE-IVD equipment.
5. Evaluating the added value of the IVDR declaration of conformity.
6. Securing the use of in-house IVDs for orphan diagnostics.
7. Defining the legal entity status of healthcare institutions, including genetic laboratories.

We also embarked on the following projects:

Harmonization in genetic counselling (Rebecka Pestoff, Christi van Asperen, Christophe Cordier, Donna Darmanin, Katrin Õunap)
This is a challenging and timely issue to address, especially regarding the many promising initiatives to mainstream precision medicine in Europe. We strongly believe it is crucial to provide quality assurance, professional accountability and patient safety amidst the rapid developments in the field. The current published literature regarding harmonization of the profession of genetic counsellor in Europe has been undertaken by the group. The literature review provided a current evidence base, leading up to submitting an opinion piece that identifies common issues and potential solutions regarding harmonization of the profession in Europe. You can read our opinion piece in the EJHG (https://www.nature.com/articles/s41431-026-02097-8). In collaboration with the European Board of Medical Genetics (EBMG), we intend to further work on improving the current situation of "disharmony" in Europe.

Poor performance in EQA (Weronika Gutowska-Ding, Christine Saban, Els Dequeker, Nicola Wolstenholme)
This year we have focused on building a shared, practical approach to understanding and addressing poor performance across External Quality Assessment (EQA) schemes. Key progress includes:

- Bringing back joint annual performance reporting
- We have agreed to reinstate annual reporting to EuroGentest, with the aim of presenting this at the ESHG annual meeting. This will help identify common issues across schemes, support patient safety, and potentially form the basis for future joint publications.
- Agreeing on a simplified, harmonised reporting template
- We reviewed the 2020 template and simplified it so it can be used consistently by all providers. The agreed data include basic scheme information (e.g. sample type, number of labs and countries) and the percentage of poor performance. Each provider will also contribute definitions of critical errors and unsatisfactory performance, which will be included as a shared appendix to improve clarity and comparability.
- Setting common principles for interpretation
- We agreed on a pragmatic approach to interpreting the data: excluding sample failures and non-submissions, and flagging schemes with more than 10% poor performance for discussion, while being mindful of small or exceptional schemes where wider learning may still be useful.

Update of FMR1 guidelines (Weronika Gutowska-Ding)
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 has been 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.

Prenatal diagnostic genomic testing (Michael Morris, Luca Lovrečić, Lina Basel)
Recommendations for genome-wide diagnostic testing in the prenatal setting are being produced in a joint endeavour with the International Society for Prenatal Diagnosis (ISPD) and the Policy and Ethics Committee (PEC) of ESHG. Since ISPD is a global society, the aim is to develop globally applicable guidelines. The guidelines will cover clinical indications, technologies, interpretation and reporting and will be of value to genetic services and laboratories as well as to regulators. The draft manuscript has been submitted to ESHG members for review and publication is expected in 2026. The joint working group is led by Prof. Sandi Deans.

If you are interested in joining the activities of EuroGentest, let us know: eurogentest@eshg.org.

www.eshg.org/egt

Report from the ESHG Young Committee

by Silvia Kalantari, Chair of ESHG-Young on behalf of all Committee Members

The European Society of Human Genetics-Young Committee (ESHG-Young) continues to serve as a vital bridge for early-career professionals, fostering a broad community dedicated to scientific and collaborative growth. Our mission remains to empower the next generation of geneticists by providing the resources necessary to navigate the evolving landscape of human genetics.

The 2025 cycle marked a significant evolution in our leadership structure. Following the 2025 election process, we welcomed Maria Francesca di Feo, Nelson Martins and Mathys Weber to the board. They will serve a four-year mandate concluding in June 2029. We are also pleased to announce the new appointment of Teodora Barbarii as an Associate Member focusing on our collaborations with young genetic societies across Europe, while Mridul Johari continues to support our initiatives in the capacity of Consultant. As we embrace these new perspectives, we extend our sincere thanks to Juliana Miranda Cerqueira (Consultant) for her invaluable contributions as she concludes her tenure on the committee. Her role was instrumental in representing young geneticists within the ESHG Scientific Program Committee (SPC), together with Rita Barbosa-Matos (Vice-Chair). Their involvement has been key in shaping a stimulating and insightful program for the ESHG 2026 conference. Maria Francesca Di Feo will be the next young representative within the SPC, starting from June 2026.

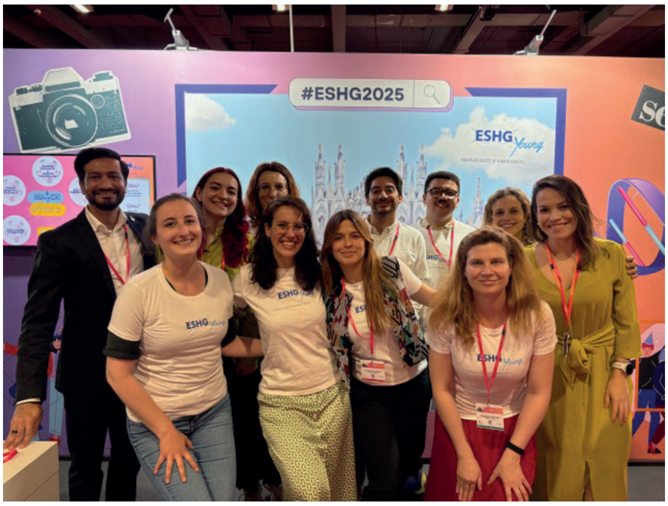
Silvia Kalantari (Chair) and Mathys Wéber (Secretary) have maintained a strong partnership with through their roles on the scientific committee for the 2026 Eurodysmorpho conference (16-19 Sept 2026, Pavia). The scientific program will feature Dr. Emilia Bijlsma, returning to delivering her practical course on Bayesian calculation, alongside sessions on the history of medical genetics by Dr. Huw Dorkins, skeletal dysplasia with Prof. Valérie Cormier Daire and new insights into cytogenetics by Prof. Elena Rossi. In addition to these preparations, Silvia and Mathys attended the society's board meeting in Bergen (December 2025).

Maria Francesca Di Feo (Spokesperson), Rita Barbosa Matos (Vice-Chair) and Magdalena Mroczek (Treasurer) continued to strengthen ties with the **EduComm** as ESHG-Young representatives. They supported the applications of early-career geneticists through the Mentorship & Observership Fellowship Programs, served as judges for DNA Day and collaborated with the Presentation Skills Workshop that took place online on the 19th May, covering foundational concepts for delivering effective presentations. They also provided feedback on proposals for ESHG courses, contributing to the development of future educational activities.

Rhys Dore (Vice-Chair) continues being the young representative on the **Policy and Ethics Committee (PEC)**, attending biannual meetings at the ESHG Conference and in Geneva while contributing to proposals and document reviews. He has also contributed to the organisation of the **EBMG** Get2Gether for the ESHG Conference 2026, which will have a focus on the European Certificate in Medical Genetics and Genomics (ECMGG) exam.

Magdalena Mroczek continues representing the young geneticists in the **EJHG Editorial Board** and to lead the African Society of Human Genetics (AfSHG) collaboration. In late 2025, several joint initiatives with the AfSHG were organized, including a Lunchtime webinar in October featuring scientific and career-focused talks, and a four-part webinar series in November in collaboration with JAX Laboratory on cell modeling for rare diseases.

The **ESHG-Young Social Media team** (Rita Barbosa-Matos, Nelson Martins, Maria Francesca Di Feo, and Mathys Wéber) has promoted content for young geneticists while supporting ESHG initiatives highlighting professional development opportunities. The committee currently manages accounts on X, Instagram, LinkedIn, and Bluesky, with a steadily increasing number of followers.



In addition to our ongoing collaborations, the 2026 ESHG Conference will feature a wide array of events organised by our committee, with the interests of the Young Geneticists in mind. The ESHG-Young **Get2Gether session, "Uniting European Young Initiatives in Human Genetics"**, is scheduled for **Saturday, June 13, 2026 (12:15–13:15)**. This year's edition introduces an innovative, interactive format in a U-shaped room, designed to facilitate a deep-dive discussion on topics recently prioritized by our community survey. The session will focus on the practical differences in clinical genetics practice, as well as a dedicated exploration of career paths in human genetics. This event is organized by Silvia Kalantari, Mathys Weber, Nelson Martins and Teodora Barbarii (Associate Member), in partnership with Rita Barbosa-Matos and Juliana Miranda Cerqueira from the SPC. The discussion will feature perspectives from representatives across Albania, Denmark, France, Germany, Italy, the Netherlands, Portugal, Romania and Turkey. The session will also host an „open floor" segment to exchange ideas on ongoing projects and future initiatives, ensuring that the needs and ideas of young geneticists remain at the heart of our international collaborative efforts.

After the success of the last two years, the ESHG-Young **Matchmaking Event** will be held in the time slot on **Sunday, June 14, 2026 (12:15-13:15)** at the shared ESHG & ESHG-Young booth. The event is designed to connect trainees, mentors, and recruiters in a dynamic setting around tables subdivided by topic (i.e. clinical careers, academia, industry). In order to make networking easier, ribbons will be used to identify participants.
This year's **Young Human Geneticists Networking**

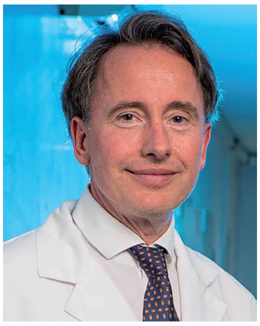
Event will take place on **Sunday, June 14, 2026 at 20:00 at Feskekörka**, a unique indoor fish market and one of the city's most iconic landmarks, and is expected to bring together 200 young geneticists. With the support of our local organizer and our toastmaster Nerges Winblad, we have designed an engaging programme inspired by Swedish Midsummer traditions. Alongside local food, participants will enjoy quizzes, bingo, and traditional songs, creating a lively environment that encourages networking, interaction, and cultural exchange.

So brace yourself for four days of events, networking, and career opportunities for young geneticists (i.e., those in training and within four years after training completion) at the 2026 ESHG Conference in Gothenburg! We warmly invite you to visit our booth during the breaks, where we will be happy to answer questions about our activities and explore future opportunities for collaboration.

www.eshg.org/eshgy

Report from the Strategic Committee

by Borut Peterlin, Chair of the SC



Over the past year, the Strategic Committee (SC) has continued its work to shape the long-term direction of the European Society of Human Genetics and to strengthen ESHG's position as a leading European and international hub in genomic medicine.

A major focus of our work has been developing the ESHG strategy. Further refined over the year, it sets out a substantive direction for the Society: supporting the integration of genomics into clinical pathways and health systems; promoting models of care in which genetic professionals work closely with other medical specialties; advancing the contribution of genomics to prevention, diagnosis, treatment and long-term care; and reinforcing the roles of research, education, policy engagement and public dialogue in achieving lasting health impact. It also highlights equitable access, secure and responsible use of genomic data, and constructive engagement with patients, policymakers, and other stakeholders. The strategy is expected to undergo public consultation as part of its further finalisation.

A broad survey on the future of genomic medicine informed this strategic work, and we are now preparing a manuscript arising from this study.
We also initiated work on a position paper on Public Health Genomics and, in this context, opened dialogue with the European Public Health Association.

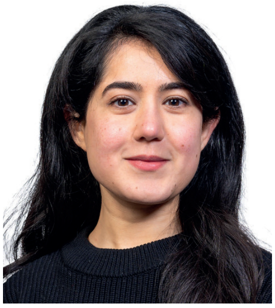
In parallel, after discussing with the ESHG Executive Committee, we are continuing work on a white paper that more directly articulates how geneticists and genomics should be embedded in healthcare systems in collaboration with other stakeholders. Our aim is to clarify for external partners what ESHG can contribute as the voice of the genetics community and how we work with other clinical, policy, and societal stakeholders. This process included a co-creation workshop in Utrecht, where we explored these questions with relevant stakeholders as part of developing the document.

I would like to sincerely thank all SC members and contributors for their commitment, insight, and collegiality.

www.eshg.org/sc

Social Media Committee Annual report June 2026

by Aleena Mushtaq Stolworthy and James Fasham, Co-Chairs of the SMC



This year the ESHG Social Media Committee advanced the society's outreach by using Buffer to schedule and manage posts across different social media platforms to ensure consistent messaging around the conference, publications from EJHG alongside other leading journals and community events. This streamlined approach has helped us to maintain a steady posting schedule.

In 2026, LinkedIn emerged as our strongest platform by reaching 5,000 followers! Bluesky grew rapidly in

popularity, gaining 744 followers as people have migrated from other traditional platforms. We have prioritised LinkedIn and Bluesky due to their high levels of engagement, with X/Twitter used selectively for major announcements.
We have taken initiatives to deepen our ties with national genetics societies, ESHG-Y and EduCom by amplifying their events and news via ESHG social media channels, fostering a more connected pan-European genetics community. Working closely with the ESHG office (Magdalena Seidl), we are exploring a dedicated Google group to centralise requests, track national society activities to ensure relevant and timely promotions.

For 2027, we aim to weave in equity, diversity, and inclusion (EDI) principles into our strategy. This means reviewing our content for inclusive language, diverse imagery, and representation of varied career stages, regions and backgrounds.

Dr. Aleena Mushtaq and Dr. James Fasham

www.eshg.org/smc

What happened in 2025 at the European Journal of Human Genetics?

by Alisdair McNeill, Editor in Chief EJHG

In 2025, the European Journal of Human Genetics (EJHG), the official journal of the European Society of Human Genetics, published a wide spectrum of genomics research in the form of original research, brief communications, and review papers. This covered the spectrum from population genetics and rare disease gene discovery through to social science aspects.

From 1st January 2025 to 31st December 2025, the European Journal of Human Genetics received 1,173 submissions (compared to 1,034 for the preceding 12-month period and 823 in the year before that). Our acceptance rate in 2025 was 17%. In 2025, there were well over 3.2 million downloads of articles published in the journal and over 1,700 Altmetric mentions. Publications were submitted from a range of different countries—amongst accepted manuscripts, the top three submitting regions were the United Kingdom, Australia, and France. We continue to try and broaden our reach and welcome submissions of relevant research globally. Acceptance for publication in the European Journal of Human Genetics remains competitive, and it remains a prestigious home for your research and review papers.

Given the rising volume of submissions, the journal faces an increasing editorial and peer review workload. To support timely manuscript processing, the ESHG executive have kindly agreed to invite board members to join our peer reviewer pool. If you felt able to volunteer, we would ask you to review 2-3 papers per anum.

The 2024 Impact Factor of the European Journal of Human Genetics is 4.6 (with a total of 12,433 citations). This was an increase from the previous Impact Factor of 3.7. According to the Impact Factor, the European Journal of Human Genetics is ranked 30th out of 191 Genetics and Heredity journals. The latest (2024) CiteScore of the European Journal of Human Genetics is 8.6, and the journal is now ranked 15th out of 100 Genetics (Clinical) journals by Scopus. Our strategy to boost the Impact Factor of EJHG centers around improved social media promotion of papers and increasing the number of review papers we publish (since these are more highly cited). We would welcome submissions of relevant review papers and suggestions for review topics.

In 2025, we further developed our social media strategy to enhance the visibility of research published in the European Journal of Human Genetics. While the EJHG X (Twitter) account currently has approximately 7,500 followers, we are shifting our strategic focus. **Subject to board approval, we intend to cease activity on X and transition our primary social media presence to LinkedIn**. Our social media editor Dr Seda Zonuzi, has already established a growing community on LinkedIn with nearly 800 followers, and we have also launched an Instagram account for the journal. Across these platforms, we will continue to promote EJHG papers through online journal clubs and brief summaries of new research as it is published. We believe this move will better align our digital outreach with professional networking and academic engagement.

In 2025, we have said farewell to colleagues who have left our editorial board (Claire Morgan, Lisenka Vissers, Mamta Muranjan, Chandana Basu, Gyaneshwer

SAVE THE
DATE!

ESHG

EUROPEAN HUMAN GENETICS CONFERENCE

Ahoy Rotterdam, **The Netherlands** | June 12-15

2027

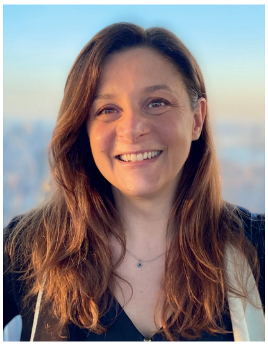
https://2027.eshg.org/ | @eshgsociety | #eshg2027

Chaubey, Ashwin Dalal, Ashraf Yahia, and Bernt Popp) and welcomed several new members. New members include: Tara Clancy, Ayse Demirkan, Alistair Pagnamenta, Ewelina Pośpiech, and Palle Duun Rohde.
I will be at ESHG 2026 in Gothenburg; please do let me have your thoughts on the journal, and ideas for its development, 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

Report from the European Board of Medical Genetics and Genomics (EBMG) 2025-2026

by Angela Peron, Clinical Geneticist, President of EBMG



The **European Board of Medical Genetics and Genomics (EBMG)** is dedicated to establishing qualification and certification standards for professionals in the field of medical genetics. It promotes the harmonization of training and certification across Europe to ensure high levels of competence in clinical genetics, laboratory genetics, and genetic counseling.

In 2025-2026 the EBMG has focused on improving its internal organization and its visibility across Europe. We have finalized and approved the documents describing each role of the Executive Board, following the work of the previous Execs, and have enhanced the collaboration between the three branches, facilitated by regular monthly online meetings. We are also in the process of amending the EBMG statute, which will be adopted before the ESHG annual conference. We have also decided to change the name from European Board of Medical Genetics to European Board of Medical Genetics and Genomics, while retaining the acronym EBMG.

Following the success of previous years, we will host another **EBMG Get2Gether** in Gothenburg during the annual ESHG conference, on **Monday June 15th 2026 at 12:15 CEST.** It will be an interactive session, where we will briefly present the three EBMG branches, and then we will focus on the the ECMGG (European Certificate of Medical Genetics and Genomics), a joint EBMG-UEMS effort with the support of the ESHG, with a live demonstration of the exam. In addition, members of the three professional branches will be available during the session to answer questions about the certification process and how to become involved with EBMG activities. **You are all invited to attend!**

The three Branches:

1. Medical Geneticists Branch:
The **Medical Geneticists (MG)** branch is, together with the UEMS section of Medical Genetics, running the exam for the ECMGG. Seventy candidates are planned to sit the exam this year (MCQ part in April and oral part on June 5th). The interest this year was overwhelming and higher than the capacity. Unfortunately, some candidates from countries not part of or affiliated with UEMS had to be turned away. The period for signing up for the 2027 exam will be shorter. Stay updated on: https://www.uems-ecmgg.org/ . Other important activities this year were the involvement with European training requirement for non-genetics medical specialists – continuously raising awareness of the importance of including genetics and rare disease in curricula of all medical specialists, and, in collaboration with other laboratory specialties, the preparation of a position paper regarding direct-to-consumer testing.

2. Clinical Laboratory Geneticist Branch:
The **Clinical Laboratory Geneticist (CLG)** branch now has 429 registered professionals from approximately 40 countries, with 20 additional applications pending. In frame of its dedicated efforts toward achieving European-level recognition for the profession through engagement with the European Commission, the Board found the following in 2025: to our knowledge, the CLG title is already state-recognized in overall 11 EU countries. The good news is that we need only 9 EU countries with state-recognized CLG titles to apply for European registration of CLGs as a regulated profession in accordance with Directive 2005-36-EC. The problem we learned about last year is that those EU member states in which the CLG title is state-recognized must register this in the database for regulated professions (RPD, https://ec.europa.eu/growth/tools-databases/regprof/professions/bycountry). However, only two of the eleven countries with state-recognized CLG titles are currently listed in the RPD for non-MD CLGs. Only when all 11

countries with national title (or at least 9 of them) are listed on RPD can the EBMG apply to national politicians for the CLG to be included as a European registered profession. We recently requested all national societies in countries that still need to be included in the RPD to contact the relevant ministry in their country that can make this inclusion.

3. Genetic and Genomic Counsellors Branch:
The **Genetic and Genomic Counsellors (GCs)** branch continues to support the development and harmonisation of the genetic counselling profession across Europe. During the current cycle, the branch is evaluating 18 new registration portfolios, and the current total number of registered genetic counsellors in Europe is 151. The branch is also undertaking the renewal of registrations for professionals reaching the five-year re-certification point. In parallel, work is ongoing to renew accreditation for several Genetic counselling MSc programmes, with 11 Master's programmes currently accredited. A major milestone this year is renewing the branch's mission and vision, which will guide future activities. The branch's vision is to promote high standards for genetic and genomic counsellor practice and education across Europe, ensuring safe, equitable and ethical patient-centred care. The branch looks forward to continuing its work in close collaboration with the MG and CLG branches of EBMG, the ESHG and other partners, and to engaging with colleagues at the ESHG conference.

The EBMG continues to work with productivity and enthusiasm, and we want to express our gratitude once again to all the EBMG members and affiliates for their significant contributions. This commitment forms the cornerstone of the EBMG mission, with mutual support and respect. Thank you!

www.ebmg.eu

ESHG
EUROPEAN SOCIETY OF HUMAN GENETICS

Interview with Tuuli Lappalainen, ESHG Award Lecturer 2026

by Mary Rice, ESHG Press Officer

Tuuli Lappalainen is Professor at the KTH Royal Institute of Technology, Stockholm, Sweden, Director, National Genomics Infrastructure & Genomics Platform, SciLifeLab and Senior Associate Faculty Member, New York Genome Center. She will be giving the ESHG Award Lecture at 13:50 hrs on Tuesday June 16.

Tuuli Lappalainen was so young when she first became interested in science that she can't remember her exact age at that time. "I think I've always just been interested in how the world works, and why things are the way they are. So looking around me and asking questions about what I'm seeing has always been part of my life."

She grew up in a family that was equally curious and questioning. "We were surrounded by books and magazines, and my parents encouraged my curiosity. On weekend mornings, I'd read the newspaper and find that my dad had done calculations on it to verify whether some of the things described made sense. So having a questioning mentality is a family characteristic."

Although she was occasionally tempted by the thought of another career when she was very young – a zookeeper was one such idea – there wasn't much doubt that she was destined for an academic career, and she didn't need any encouragement from her parents to go down that route. "I think it solidified pretty much at high school age that I would go for some kind of research career in biology, which is a subject that has always fascinated me," she says. "I was particularly interested in human biology, with its links to evolution, genetics and physiology and how these change over time through heredity. In my late teens I figured out that I was interested in genetic mechanisms and their related basic discovery research. I didn't feel that being a medical doctor was the right fit for me."

In the year 2000 she graduated from high school and went to university in Turku in South Western Finland, followed by a PhD in Helsinki. At that time there was a boom in biotechnology-related research, she says. "There was a great sense of opportunity and doors opening to allow researchers to do new things. And the sequencing of the human genome the same year as I started studying biology at university meant that I was part of the first generation of geneticists fully trained in the genome era. We had textbooks from the previous era that had become outdated basically overnight. With the genome we had a whole new toolkit in our hands."

She is proud of the data resources she's been part of building, the impact they have had on the genetics community, and the diverse forms of science that such data have enabled across the world. "When I see a paper that is doing analysis with an open resource dataset that I was part of creating as a postdoc, or with a consortium, it's rewarding to realise that I had a part in helping that science happen. Creating something that is valuable for a lot of people, whether its data resources or methods, is very satisfying. And the biggest impact we have, of course, is not via our own specific papers or datasets, but by the people we train. I've been fortunate to work with fantastic young scientists who have left my lab to start their own labs or companies. Having a role in their career path is very fulfilling."

Of course, there are the boring bits, but she accepts that there's no free lunch in the world and administrative stuff is an integral part of the system. "If you want to do ambitious work and push boundaries, you just have to get on with it." A bigger disappointment is the disintegration of the global research community. Whereas when she started her research it was easy to move to work in different countries and to collaborate across borders, new borders are starting to be erected. "It feels as though the global community that we had has been negatively affected by today's international politics, so that younger researchers don't get to live in the same open world as we did. I find that very sad, and harmful to the research community as well as to individuals."

If she hadn't been a scientist she might have become a writer. "I also love art, but I'm not an artist, so maybe managing a gallery. Retirement is a long way off, but I will probably do something related to writing when I get there, and maybe try to stay active in the research community in some way. I get bored very easily and am not good at sitting around doing nothing." Since she works on two continents at present there's not much time for inactivity. "When I get the chance, I like food-related things and just general hanging out."

She will be telling the conference about her quest to interpret molecular and cellular effects of genetic variants that capture the molecular processes driving the genetic impact on human traits and disease. "Many studies from our community have made major discoveries, but there are still many challenges ahead in figuring out what those variants actually do in cells. I'll talk about some of the roads ahead, and what kind of toolkit will help us advance on this path."

Interview with Guillaume Canaud, Mendel Award Lecturer 2026

by Mary Rice, ESHG Press Officer

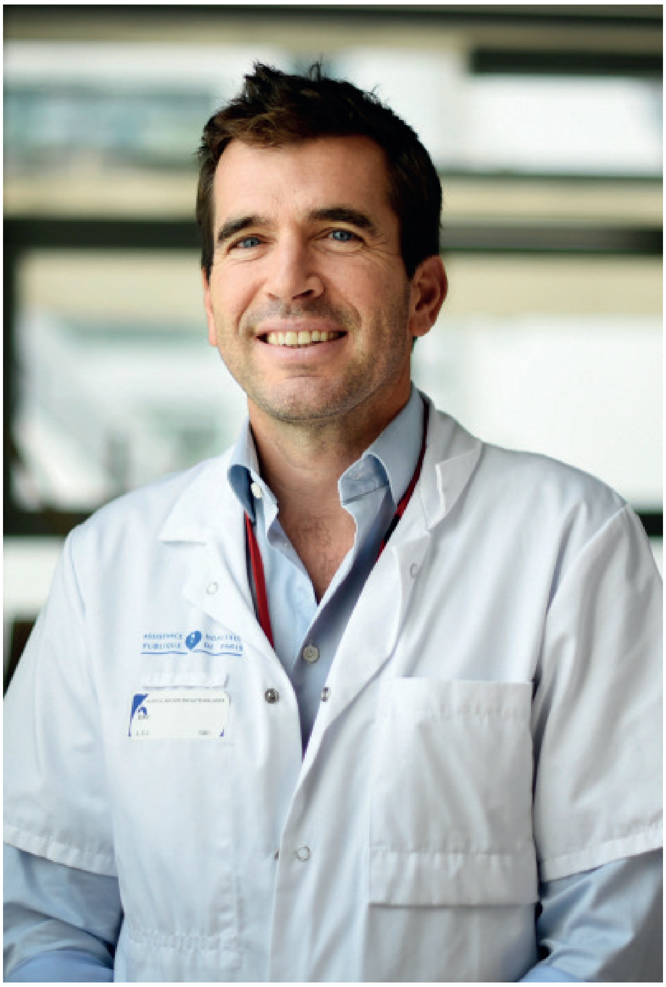
Guillaume Canaud is Professor of Translational Medicine and Targeted Therapies at the Necker–Enfants Malades Hospital in Paris. He will be giving the Mendel Award Lecture on Tuesday June 16 at 14:35 hrs.

Guillaume Canaud describes himself as a 'rather lazy and dreamy' child. "I was far more interested in my friends and life outside school than in academic work. I always enjoyed science and understanding how things functioned, but I only truly had my intellectual awakening at the age of 17, just six months before the French national examination that grants access to university."

He comes from a family of scientists. His mother was a paediatric anaesthesiologist, and his father a professor of nephrology in Montpellier, in the south of France, so medicine and science were constant topics of conversation at home. "My father is also an inventor", he says. "He developed a dialysis catheter—an access device that allows patients to undergo dialysis under optimal conditions. This catheter is now used worldwide, and this has always filled me with immense pride."

Around the age of 18, he developed what he describes as a deep fascination with understanding mechanisms. "My motivation was never to memorise and recite entire chapters of textbooks, but rather to understand in depth how things work, as this made learning both easier and more meaningful to me."

After finishing medical school in Montpellier, he initially opted to follow in his father's footsteps and chose nephrology, a subject he describes as renowned for its intellectual rigour and its breadth. But following a Master's and a PhD in science: "I developed a true intellectual love affair with basic science, and started to work on the role on intracellular signaling pathways in chronic kidney disease." This work led to publications in leading scientific journals, a postdoctoral fellowship in the United States at Harvard Medical School in Boston, and eventually his first major grant: a European ERC Starting Grant.



In 2015, a meeting with a patient changed his life profoundly. "A young man named Emmanuel, about 25 years old, was suffering from a rare mosaic genetic disease associated with kidney failure. His prognosis was devastating, with no available therapeutic options. The disease was progressive and deforming, associated with severe pain, fatigue, and multi-organ dysfunction. His story moved me deeply."

"Using the scientific and medical knowledge accumulated over the years—and a great deal of curiosity—my team and I identified a potential treatment for him. The drug was originally being developed for breast cancer. We created the first preclinical model that recapitulated the patients' symptoms and deformities, demonstrated treatment efficacy in animals, then in Emmanuel himself, and subsequently in several other patients, including children. We conducted the first real-world clinical trial, which ultimately led the U.S. Food and Drug Administration to grant marketing authorisation for this drug in this new indication."

As a result, several thousand patients worldwide now benefit from a new, less invasive therapeutic approach, with fewer surgeries and dramatically improved symptoms, he says, and this is a tremendous source of pride. The encounter with Emmanuel changed the course of his career. "I stopped practising nephrology and dedicated myself entirely to the care of adult and paediatric patients with mosaic disorders associated with overgrowth and vascular malformations. We established a genetic diagnostic platform, developed numerous preclinical models, identified several additional treatments, and became experts in drug repurposing—using medications developed for one indication to treat rare genetic diseases, thereby practicing true precision medicine."

He counts receiving authorisation to treat Emmanuel and seeing the first signs that the treatment was working as among the most memorable milestones of his career. Another – above all, he says – was the moment when a young girl he had treated and who had been paraplegic for two years took her first steps. "Today, she walks normally and has recovered fully. The joy this brought the team is beyond words."

Science is lucky not to have lost him to music. "When I was young, I would have love to have become a disc jockey." Today, outside of science, he appreciates art and reading, and is passionate about sport, particularly hiking and running. "One of my greatest pleasures, when possible, is traveling with my family. While my work allows me to travel extensively, sharing these experiences with my wife and children is something entirely different and profoundly enriching."

Retirement? "One day the question will arise, inevitably. In France, retirement comes relatively early and the opportunities to pursue a professional activity afterwards are limited. If continuing my work in France becomes impossible, I would consider moving abroad, but I would do so with sadness. I am deeply proud of my country, grateful for what it has given me, and proud to contribute—albeit modestly—to its scientific influence, even during these challenging times."

At the conference he will invite the audience to discover his unconventional career path. "Geneticists have one of the most extraordinary professions: they establish diagnoses, understand disease mechanisms, and identify potential treatments. The final step—delivering effective therapies—must now be fully embraced. We are fortunate to draw inspiration from oncology, and we must seize this opportunity and apply it to our patients. The message is clear: remain open-minded, adventurous, and ready to seize therapeutic opportunities when they arise. Patients with rare genetic diseases are waiting for this from us."

Interview with Anne Cambon-Thomsen, ELPAG Lecturer 2026

by Mary Rice, ESHG Press Officer

Dr Anne Cambon-Thomsen is Honorary Research Director, CNRS, at the INSERM Centre for Epidemiology and Research on Population Health at the University of Toulouse, France (CERPOP). She will be giving the ELPAG Award Lecture on Tuesday June 16 at 13:30 hrs.

While Anne Cambon-Thomsen's career has been varied, it has always followed a logical progression. Starting as an experimental researcher, she found herself becoming more and more involved with the ethical and legal questions surrounding genetics. "Once we knew the structure of the human genome, the logic was for people to go into functional genetics – now we have the genes, what do they do and how? But I thought that what we were going to do with this new knowledge was equally important, and that question brought me to ethics and all the societal aspects of genetics."

Born in Toulouse, France, she has lived and worked there for almost all her life. At school, she was interested in science from an early age. "Before I left school, I had decided that I wanted to study medicine and also do research. So I did both trainings in Toulouse, more or less in parallel, followed by a postdoc in Denmark."

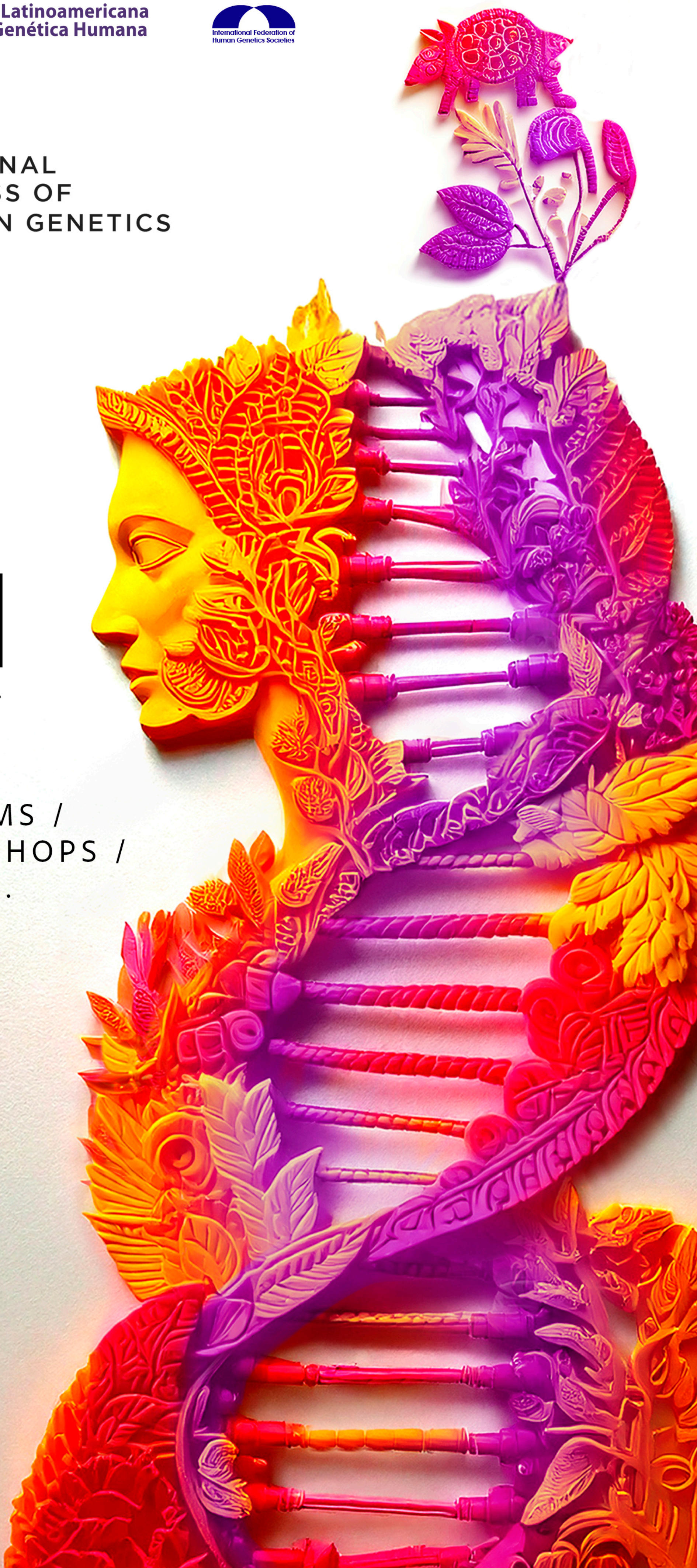
On returning to Toulouse with her Danish husband, she subsequently became the youngest female director of an Inserm research unit in France. This was followed by an invitation to also direct another unit for the CNRS, the national scientific research organisation. Directing two units at the age of 37 was 'an exceptional situation', she says, but it had to come to an end. "You can only be a unit director for a certain number of years; after that you have to restructure your unit significantly or pass the baton to someone else." It was at this stage that she started to think more deeply about the societal aspects of her work, particularly in the areas she had specialised in – im-

RELAGH
Red Latinoamericana
de Genética Humana

International Federation of
Human Genetics Societies

THE 15TH
INTERNATIONAL
CONGRESS OF
HUMAN GENETICS

ICHG 2027



SAVE THE DATE

March 1-5 2027

Guadalajara, México.

KEYNOTES / SYMPOSIUMS /
ROUNDTABLES / WORKSHOPS /
NETWORKING SESSIONS.

**From Roots
to Revelations:
Human Genetics in
Multi-Ancestry Contexts.**

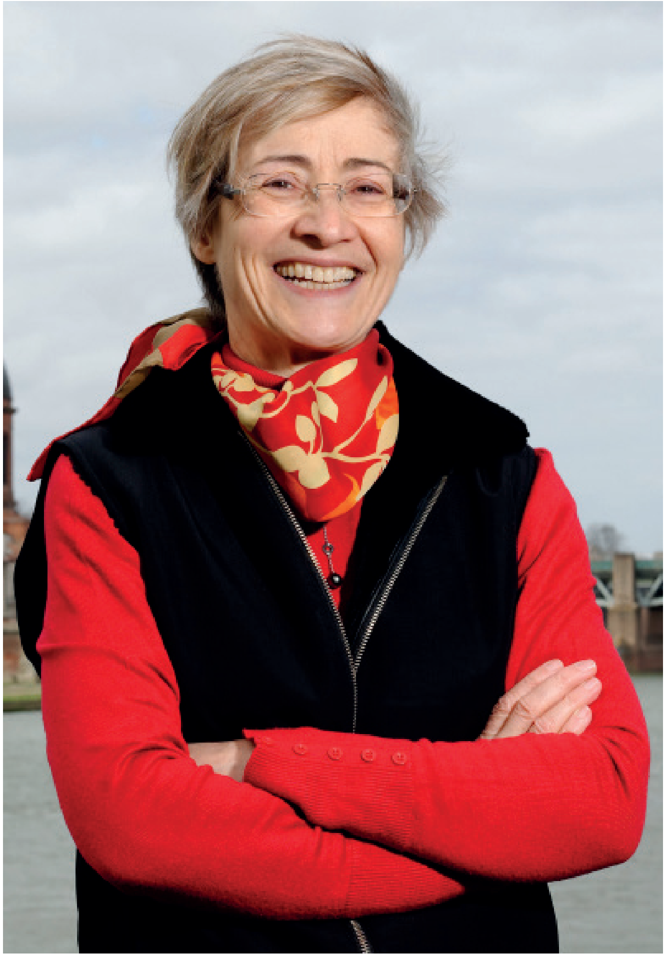
EXPO
GUADALAJARA®

munogenetics, transplantation, and population genetics.

After taking a course on the ethics of health, she approached the director of a public health unit in Toulouse with a proposal to create a new interdisciplinary team to work on an integrated approach to the subject. Many had doubts, at least at first. “Is this really research?”. “Shouldn’t expert committees be doing this rather than researchers?”. “Will young researchers in this team be able to find jobs afterwards?” were typical questions. She persisted, and was able to set up a team within a new unit of epidemiology and public health. It started with just three people. “Now, with my successor, there are forty,” she says, proudly.

In her earlier career she had been responsible for the collection of many biological samples, but the next director in the lab was not going to use these collections. “So I asked myself a practical question: what should a researcher do in these circumstances? It costs money to maintain such collections and if they are not going to be used, what happens to them?” This led her to focus her research on biobanks. “At the end of the 90s, there were a lot of collections, but no real biobanking organisation. I have published a lot on this subject, and it is for this work that I became known for in the ELSI – Ethical, Legal and Societal Implications – domain.”

Working with people from many different fields was rewarding and productive. “Coming from medicine and biology and meeting sociologists, lawyers, psychologists, and philosophers was fascinating, but also difficult to organise at first. Such a team was a novelty at the time, and finding money was difficult too.” However, she succeeded in obtaining support from the EU, who had started to introduce the idea of public accountability and ethics into research projects. “The Commission was taking seriously the idea of patient associations and public participation, so this was a good opportunity for me to advance the idea.” That also led to the creation of a societal platform, a new concept that still continues today within the Genopole of Toulouse, a service to researchers and a bridge between them and the public on ethical and societal questions.



Semi-retired now, she intends to keep her positions on some advisory boards and continue evaluating projects. “And I’ve already started some retirement projects. I have started piano lessons again, and I read to children in schools, which I really enjoy. I’m also a member of several associations, including one for the dissemination of science.”

She will be taking the conference through her career journey, giving examples of some of the ethical issues she has faced along the way. “There have been so many instances where a research project has come about because of some small event that has prompted a wider reflection. I think it’s important for people to know that what appears to be insignificant happenings can change the orientation of a career. And I’ll talk about the great pleasure that constructing and working in an interdisciplinary team of open-minded people has given me.”

EUROPEAN SOCIETY OF HUMAN GENETICS

AGENDA
General Assembly 2026
Monday, June 15, 2026, 12:15 - 13:15 hrs
Room G4, Svenska Mässan, Gothenburg, Sweden

1. Welcome by the President of the Society, Martina Cornel
2. Approval of the Agenda and of the timely invitation of members
3. Approval of the Minutes of the last GA in 2025
4. Report on the Activity of the Society 2025-2026, Martina Cornel
5. Financial Report of the Treasurer 2025, Gunnar Houge
6. Reports of the Auditors 2025, Natalia Oliva Teles, Ulf Kristoffersson
7. Exoneration of the Treasurer
8. Exoneration of the Executive Board
9. Opening by the new President of the Society, Angus Clarke
10. Results of the nomination for President-Elect 2026 – Confirmation vote by the Membership
11. Vote on new Secretary General, Julia Baptista
12. Vote on new Deputy Secretary General, Carla Oliveira
13. Vote on Auditors, Natalia Oliva Teles (2nd term) & Ulf Kristoffersson (1st term)
14. Results Board Member nominations 2026 - Confirmation vote by the Membership
15. Thanks to leaving Board Members
16. Vote on Membership Fees 2027
17. Update on sites of future European Human Genetics Conferences
18. Any other business