



17th Meeting of the European Human Genetics Societies
Sunday, June 2, 2024, 12.00 – 14.00 hrs CET
Messe Berlin, CityCube
Rooms M6+7
Zoom:

Welcome and Introduction

Milan Macek, NHGSs Liaison of the ESHG

Self-presentation of the meeting participants

Each participant introduced themselves.

ESHG highlights 2023 -2024

The ESHG President, Valérie Cormier Daire, reported on the news and highlights from 2023/2024. Detailed information can be found in the ESHG Newsletter.

European Union of Medical Specialists (UEMS) – Section of Medical Genetics

Ute Moog, President of the Clinical Genetics Section of UEMS, Heidelberg, DE

UEMS has been representing medical specialists since 1958. The section of Medical Genetics (MG) has existed since 2014; elections are planned at the ESHG 2024. Currently, there are 29 member countries. The MG specialty is recognized in all EU countries except Spain.

There is an overlap between EBMG and UEMS; all UEMS MG members are also members of the EBMG board for medical genetics.

Past /Current projects: 1. Memorandum of understanding between 3 bodies – UEMS for MG, EBMG and ESHG, 2. appraisal of the ECMGG exam (test of written and oral part, testing knowledge and communication skills and attitude) by CESMA, and 3. revision of the ETR for medical genetics (the revised version was presented and approved at UEMS in October 2023, done in collaboration between UEMS MG and Ithaca). This year there was a workshop for MCQ writing in cooperation with Ithaca.

All attendees were invited to become members of UEMS MG, promote the ECMGG exam, and include the UEMS MG link on their national society websites.

Discussion:

Question (FF): What is the value of the ECMGG for young people?

Answer (UM): Each country might decide to integrate the UEMS exam into their system. Some have adopted it as a national exam or as a prerequisite for a national exam. It is also a plus on your CV.

European Board of Medical Genetics

Clara Serra-Juhe, President of EBMG, University Pompeu Fabra, Barcelona, ES

EBMG was formed in 2012 and has three branches: the Genetic Nurses and Genetic Counsellors Branch (GCGN), Clinical Laboratory Geneticists (CLGs) branch, and Medical geneticists branch. The

activities of all three branches were presented. Please check the details in the ESHG Newsletter and the slides of the presentation.

Goals: raising awareness of EBMG by updating the webpage, better visibility at the ESHG (Get2Gether session during ESHG in Berlin entitled "Medical Genetics Professionals: how can we fit future requirements?..."), and building relationship between branches.

Discussion:

Question (GH): Are EBMG incomes from certificates only?

Answer (CSJ): Yes. This is an issue. It is difficult for us to cover the administrative costs, and we need to find a solution.

ESHG Public and Ethics Committee (PEC)

Francesca Forzano, Chairwoman of the PPPC

In 2023, the Public and Professional Policy Committee (PPPC) was renamed the Policy and Ethics Committee (PEC). Francesca Forzano presented PEC's accomplished, ongoing, and future activities. Participants are encouraged to refer to the ESHG Newsletter for detailed information. The document on Cascade testing, an underexploited strategy across Europe, will soon be submitted to the ESHG Membership for review. Additionally, a new interactive course titled "Covering The Gaps - The Course You Didn't Know You Needed" is scheduled to launch in October 2024. This pilot version will be conducted virtually over two days and is designed specifically for young scientists and clinicians. Lastly, congratulations to Yves Moreau for receiving the Einstein Foundation Award for promoting quality in Research.

Education Committee

Inga Prokopenko, Chair of Education Committee

The committee currently has 18 members, with three outgoing and four new members showing great interest in contributing to the activities.

The EuroGEMS website (the ESHG educational website) was highlighted, and everyone was invited to use the content available in four languages: Spanish, Portuguese, French, and English. The addition of new languages will be discussed on Monday, and suggestions should be sent to Ed Tobias via email (edward.tobias@glasgow.ac.uk).

The ESHG podcast has been running for several years, with 18 episodes available on multiple platforms including Buzzsprout, Apple, and Spotify. All members were encouraged to listen to this podcast.

The international membership and observership schemes were presented. New centers willing to collaborate are being sought to increase participation.

The app-based course on Paediatric Neurogenetics, sponsored by Illumina, has been extremely successful, attracting approximately 750 visitors per month.

All available courses are listed on the ESHG website, with a new course being introduced in French in Morocco.

Local activities with school children are planned for ESHG 2025 in Milan.

DNA Day 2024

Inga Prokopenko, Chair of Education Committee

ESHG sponsored a DNA Day Essay and Video contest in high schools all over the world. 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. There was the largest ever response: 220 essays from 22 countries and 20 videos from 11 countries. The essay

winner is from the UK, and the video winner is from Romania. Prizes are awarded to both the student and the teacher (mentor).

Important dates: April 25 - deadline for submission; April 25 to mid-June: evaluation.

Guest presentation 1: Towards the ESHG strategy

Borut Peterlin, Vice-President ESHG

The strategic committee (SC) was formed to address the challenges and opportunities in implementing new genetic technologies across Europe. The ESHG aims to be a hub and leader in standardizing these practices. The SC consists of chiefs from different ESHG committees and professionals from various fields. Key Objectives: developing the ESHG strategy and influencing health policy, fostering standards through the EU, particularly in smaller countries where scientists might not be heard, and encouraging public engagement in genomic medicine.

Survey Initiative: A survey initially conducted by the ESHG board last year will now be extended to all ESHG members to identify gaps, needs, and best practices within different European systems.

This aims to ensure readiness for future developments in genomics. All National Human Genetics Societies (NHGS) will be invited to voice their visions and reflect on the prepared strategy.

All participants are invited to read the detailed text in the ESHG Newsletter for more information.

Guest presentation 1: The ABC of variant classification is to ask the right questions

Gunnar Houge, Treasurer ESHG

The aim was to present the ABC variant classification system and compare it to the ACMG variant classification system. However, due to time constraints, the presentation was not given.

Participants are invited to read the recently published article (DOI: 10.1038/s41431-024-01617-8) and review the slides of the presentation.

Guest presentation 3: IVD-R: update 2024

Els Dequeker, KU Leuven, BE

An update on the IVD-R (In Vitro Diagnostic Regulation) was presented during the meeting.

Planned actions include establishing a workgroup comprising individuals with a scientific background and expertise in IVDR regulation and quality systems, who can commit time to this effort. Additionally, a survey will be launched to evaluate the national implementation of IVDR legislation and the presence of national working groups. Important deadlines associated with these initiatives were reported during the presentation. Attendees are encouraged to review further details and deadlines on the slides of the presentation.