



15th Meeting of the European Human Genetics Societies

Sunday, June 16, 2019, 11.00 – 13.00 hrs

**Swedish Exhibition and Congress Centre
Mässans Gata/Korsvägen, 412 94 Göteborg**

Room G4

Welcome and Introduction

Milan Macek, NHGSs Liaison of the ESHG

Self-presentation of the meeting participants

Number of countries represented: 31

ESHG highlights and a proposal for an update of Variant Classification

Gunnar Houge, President of the ESHG

The society is moving to Austria. This would make us easier to run the society. The transfer is expected to be finished in 1-2 years. We have strengthened our reserves in the last two years after having a big loss in Copenhagen. There is a positive development in the number of members (>3000). There is a possibility to become a member via National society. We have an educational course portfolio and give fellowships. New additional courses are expected. An ESHG fund will be created (in collaboration with Illumina). All information about the courses and policy issues are on the ESHG website.

Gunnar presented a proposal of two-dimensional variant classification system and asked all attendees to test the system and give feedback.

Online education recourses EuroGEMS.org

Edward Tobias

Edward Tobias presented an update of the EuroGEMS.org website. He received several good suggestions last year after ESHG 2018.

There are 76 documents for genetic professionals, university & students, patients & families, primary school, secondary school, and ethical, legal and social application. There are also localized/national initiatives in different languages. There were more than 31 000 visits in the 1st year from 93 countries, with an exponential increase of the returning visitors. Edward asked for help in dissemination as well as contributions from different countries and societies. Han Brunner suggested that more than translating the content of the website, it would be good to receive high quality links in different languages produced within different countries.

European Board of Medical Genetics (EBMG)

Thomas Liehr, Chair of EBMG

Thomas Liehr gave an overview of EBMG work and progress (see the Newsletter). He had handed over the chair to Ulf Kristofferson at the ESHG 2019. EBMG is structured in executive board, and three branches (medical genetics and genomics, clinical laboratory genetics, genetic nurses and genetic counsellors). They are working on establishing the fourth branch (for laboratory genetics technicians). The first goal is to recruit technicians and to find out about the existing national education schemes concerning human genetics. The next step is to set up a curriculum and eventually a certification.

The CLG-branch has approached different national governments to ask for their support in proceeding a request to EU that CLG may become EU-recognized profession.

Current status of Genetic counsellors in Europe

Christophe Cordier, EBMG, ESHG Education Committee DNA Day Coordinator

Training Programs in Genetic Counselling started in 1992 and are now implemented in at least 8 countries. A Paper has been published to highlight the collaborative nature and complementarity of the work of counsellors and geneticists. This paper also shows the relationship between these two professions. Recognition of this profession is starting to take place in many places.

DNA Day 2019

Christophe Cordier, ESHG Education Committee DNA Day Coordinator

ESHG offers the possibility to the high school students to write an essay or prepare a video and participate at the DNA Day Contest. The number of videos is still low (13), but the quality of the award-winning video was very high. The first three prizes for essay went to Turkey, Sweden, and Ireland. The title for the DNA day 2019 will be announced in September 2019.

ESHG Public and Professional Policy Committee (PPPC)

Francesca Forzano, Chair of the PPPC

Francesca Forzano reported on the activities of PPPC (post-mortem genetic testing in SCD, germline gene editing, forensic issues, opportunistic screening (in collaboration with EuroGentest; a session at the ESHG), GDPR (in collaboration with EuroGentest), SIENNA (Stakeholder-Informed Ethics for New technologies with high socio-economic and human rights impact).

Future joint activities: 1. SIC-ASHG (Agreement to organize a session at IFHGS Congress 2021.

Topics: privacy, consent and data sharing). 2. Eurogentest (Reporting in prenatal setting). 3. ESHRE (Embryo WGS).

Future topics: recommendations on IVD Regulation (comes into effect as of 2022), consent and confidentiality, Iceland experiences with Decode return of BRCA results, how genetics services, research, technologies, DTC testing could add to health inequalities, update of previous PPPC documents (pharmacogenomics, testing children, cascade screening).

Lina Basel-Salmon suggested a topic: The value of variant interpretation in the prenatal testing setting.

Education Committee and plans for a comprehensive portfolio

Han Brunner, Education Committee

Han cheered the DNA day initiative and Christophe Cordier dedication. He also thanked Edward Tobias for all the work done. Course portfolio: over the last 6-7 years, ESHG has adopted existing courses or started new ones. Currently there are 9 courses and new ones are planned (on pharmacogenomics, prenatal and PGD, bioinformatics). A proposal for pharmacogenetics course

has already been submitted by Bill Newman and will be evaluated by the board. Board has already approved a proposal for an educational workshop on variant interpretation, nomenclature, and databases that would be attached to a Balkan conference in Skopje in September 2020. Francesca Forzano suggested a course on training the trainers on quality assessment. Han Brunner commented that this course could be organised by EuroGentest. ESHG is working on the establishment of the educational fund which would finance the upcoming courses.

European Union of Medical Specialists (UEMS) activities

Bela Melegh, President of the Clinical Genetics Section of UEMS

At ESHG 2019, there was the first real exam to obtain Diploma in Medical Genetics and Genomics. Candidates were from 16 countries. European exam on rare diseases and rare adult solid cancer are foreseen.

Guest presentation 1: ESHG Young

Sara Ribeiro, Coimbra and Florence Riccardi, Marseille

The ESHG Young Network is open to medical doctors, young scientists, PhD students and Post-docs under 35-year-old or still in training. They have 420 members from 27 countries. Peer-support and sharing information on learning and training opportunities and projects is the base of this group. The board has been elected this year with 4 young women from Romania, France, Portugal, and Lithuania. They wish to have a spot in ESHG conference (to chair sessions, help organizing the ESHG meeting). They are working with the ERN ITHACA on training. They plan to improve the mode for official communication.

Guest presentation 2: 1 million EU Genomes project: overview and rare diseases

Marco Tartaglia, Rome

21 countries signed the project. France and Germany are not on the board yet. Several working groups (10) tackling different topics are active. Long-term delivery (2023-2028) – 1 million genomes.