EUROPEAN SOCIETY OF HUMAN GENETICS

13th Meeting of the European Human Genetics Societies
Sunday, June 17, 2018, 11.00 – 13.00 hrs
MiCo Milano Congressi,
Room Yellow 3

1. **Welcome and Introduction**
   Milan Macek, NHGSs Liaison of the ESHG

2. **Self-presentation of the meeting participants**

52 attendees from 31 countries

3. **ESHG highlights**

Christine Patch, President of the ESHG, gave an overview of the activities of the ESHG committees (see also Newsletter)

ESHG is growing and has more than 3000 members. This year we have the highest number of the pre-registrant, coming from 75 countries. The main work has been about the reorganisations and reconstruction of the ESHG.

4. **European Board of Medical Genetics (EBMG)**
   Angus Clarke, Chair of EBMG

Angus gave an overview of EBMG work and progress. Core tasks this year are: (i) the role of EBMG itself and how it relates to the work of the three branches, and (ii) how the constitution of EBMG should be implemented and how the Chair of EBMG should be selected. The Branch of Clinical and Medical Genetics and Genomics (CMGG) has supported the activity of the UEMS on the European Diploma in Medical Genetics and Genomics. The pilot examination was at the ESHG conference in Milan. The Branch Board for Clinical Laboratory Geneticists is aiming for CLG to be a recognized profession in EU. The CLG title is already recognized in Finland, France, Greece, Italy, Hungary, Latvia, Poland, Slovenia and UK. There are now enough countries that they could apply for EU recognition; however, this is a very difficult and time-demanding task. In some countries, the laboratory geneticists fell to be under threat by either pathologists or medical geneticists. France has a problem in the field of CLG and asked EBMG for a letter of support.
For genetic counsellors the situation is also complicated because only a small number of countries has recognised the speciality. The same applies for the genetic technicians.

5. **ESHG Public and Professional Policy Committee (PPPC)**
   Martina C. Cornel, Chair of the PPPC

Martina Cornel gave an overview of the PPPC work. Three documents on Germline gene editing appeared in EJHG. Two documents (on Post mortem genetics in sudden cardiac death and on Recontacting) were endorsed at the Board meeting (June 15, 2018). NHGS representatives are invited to disseminate the recommendations in their countries. Suggestions for new topics: i)

opportunistic screening, ii) GDPR, iii) pharmacogenomics, iv) testing children (revision), v) cascade screening/informing relatives, vi) IVD regulation (will come in force in 2020), vii) genetics and law enforcement, viii) consent and confidentiality, and ix) DTC genetics testing.
Gert Matthijs asked how additional topics could be add to the agenda and suggested the topic on legal status of broad genomic data. It could be done in collaboration with EuroGentest. Harmonised systems to share the data would be needed. There was also a discussion on new recommendations for newborn screening (last one were written in 2011). All suggestions for the topics can be send by email. The new chair of the PPPC is Francesca Forzano.

6. Education Committee, ESHG sponsored courses and "Teaching the trainers"

Inga Prokopenko, Education Committee

Philippos Patsalis and Inga Prokopenko have initiated a project Teach the Teacher and have selected a set of presentations from educational sessions from the ESHG and from the European school in Bertinoro that will be available online. Targeted group are high school teachers. NHGS that have a good contact with the teachers are requested to participate in this activity. For further information, you can contact Inga Prokopenko or sec gen.
In Italy they are going to start a new master program in genetic counselling in English.
Chris Patch reported that Ed Tobias created a website that contains a large amount of teaching materials (http://www.eurogems.org). He is collecting also material from other languages.
Germany society of human genetics has lunched the website (https://htmlexaminer.com/mediathek-humangenetik.net.html) with the material on medical genetics that addresses teachers. They would be pleased to link the website to EuroGEMS and to ESHG.

7. IFHGS

Helena Kääriäinen

ESHG is a full member of the IFHGS. We had the presidency for the last three years. Now African Society of Human Genetics (Michele Ramsay) has the presidency. The main goal of the society is to facilitate the global communication among geneticists and to organise the ICHG conferences. The next one will be in March 2021 in Cape Town.
Several national societies from the Europe are corresponding members of the ICHGS. National representatives are invited to check and update the contact details of their societies at IFHGS website.

8. DNA Day 2018

Christophe Cordier, ESHG Education Committee DNA Day Coordinator

Since 2008, ESHG offers the possibility to the high school students to write an essay and participate at the DNA Day Contest. This year 122 essays (50% less than last year) were received from 18 countries. One of the reasons for low number of participants might be late announcement of the title. The first three prices went to Bosnia and Herzegovina and Turkey. In addition, four videos were received. The first price went to France. All NHGS are invited to encourage students from their countries to write / send essays or videos and to join the assessor team at the next year's contest.

9. ESHG-EuroGentest Quality subcommittee

Ros Hastings

ESHG-EuroGentest Quality subcommittee reviewed poor performance and persistent poor performance. There are a few laboratories that repeatedly made genotyping and reporting errors. There is one lab that had poor performance for the last 7 years. Therefore, there is a need for a

regulatory body. Swiss and UK already have such regulatory bodies, but they need to be set up in other countries as well. National representatives are invited to contact Ros Hastings (ros.hastings@ouh.nhs.uk) for further information.

10. European Union of Medical Specialists (UEMS) activities

Bela Melegh, President of the Clinical Genetics Section of UEMS

Bela Melegh presented UEMS and its connections with the CESMA and EACCME. The pilot examination (written and oral part) for European Diploma in Medical Genetics and Genomics was staged on Friday, May 15, before the ESHG conference in Milan. There were eight candidates from different countries. All national representatives interested in this topic were invited to take part at the UEMS meeting on Monday, May 18 at 11:30. One of the representatives commented that it would be important to have the exam in different languages.

11. Guest presentation 1: Solve-RD.eu Horizon2020 project

Olaf Riess, Tübingen (see presentation)

Olaf Riess presented Solve-RD, a large consortium of 21 partners that has acquired a € 15 million grant to improve the diagnosis of rare diseases, to identify novel ultra-rare RD entities. Several (multi-) omics approaches will be used. The researchers will work directly with European Reference Networks (ERNs).

12. Guest presentation 2: Challenges in variant classification and reporting

Gunnar Houge, Bergen (see presentation)

Gunnar Houge commented on the problems of the current classification system (variant class 3 is not between benign and pathogenic but is a variant for which we have insufficient knowledge for grading; penetrance is not part of the system). He proposed a classification system that would have molecular grading as well as clinical grading. There was a discussion on whether to report VUS to the clinician or not. The task on updated classification system will be led by EuroGentest. A collaboration with other bodies is planned. NHGS are requested to give their feedback.

List of delegates:

- *Hans Christoph Duba, ÖGH, AT*
- *Gert Matthijs, BESMG, BE*
- *Christophe Cordier, Swiss Association of Genetic Counsellors, CH*
- *Siv Fokstuen, SSMG, CH*
- *Kyriacos Felekkis, CSHG, CY*
- *Carolina Sismani, CSHG, CY*
- *Milan Macek, Czech Society of Medical Genetics, CZ*
- *Christine Scholz, GfH, DE*
- *Elsebet Oestergaard, DSMG, DK*
- *Maris Laan, ESTONIAN Society of Human Genetics, EE*
- *Juan C. Cigudosa, Spanish Society Human Genetics, ES*
- *Feliciano Ramos, Spanish Society Human Genetics, ES*
- *Helena Kääriäinen, IFHGS, FI*
- *Sini Skarp, SCGY, FI*
- *Marie-Antoinette Voelckel, AFCG, FR*
- *Stephane Bézieau, FFGH, FR*
- *Didier Lacombe, FFGH, FR*
- *Elene Abzianidze, Georgian Society of Medical Genetics and Epigenetics, GE*

- *Eka Kvaratskhelia, Georgian Society of Medical Genetics and Epigenetics, GE*
- *Helena Fryssira , Helenic Medical Genetics Society, GR*
- *Aspasia Tsezou, Hellenic Association of Medical Geneticts, GR*
- *Istvan Balogh, Hungarian Society of Human Genetics, HU*
- *Aniko Ujfalusi, Hungarian Society of Human Genetics, HU*
- *Bela Melegh, UEMS SMG, HU*
- *Sally Ann Lynch, Irisch Society of Human Genetics, IE*
- *Vigdis Stefansdottir, Icelandic Human Genetics Society, IS*
- *Jon J. Jonsson, Icelandic Human Genetics Society, IS*
- *Maurizio Gennardi, SIGU, IT*
- *Alessandra Renieri, SIGU, IT*
- *Algirdas Utkus, LSHG, LT*
- *Zanda Daneberga, Latvian Human Genetics Association, LV*
- *Dijana Plascseska-Karanfilska, MSHG, MK*
- *Isabella Borg, Malta College of Pathologists, Human Genetics Goup, MT*
- *Hans van Bokhoven, NVHG, NL*
- *Gunnar Houge, ESHG, NO*
- *Elen Siglen, NSHG, NO*
- *Asbjorg Stray-Pedersen, NSHG, NO*
- *Posario Santos, SPGH, PT*
- *Maria Puiu, SRGM, RO*
- *Adela Chirita Emandi, SRGM, RO*
- *Ivana Novakovic , Serbian Genetic Society-Med.Gen. Section, RS*
- *Vera Izhevskaya , Russian Society of Medical Genetics, RU*
- *Karin Writzl, ESHG, SI*
- *Luca Lovrecic, Slovene Society of Medical Genetics, SI*
- *Ludevit Kadasi, SSMG, SK*
- *Natalia Gozovenko, UAEMLG, UA*
- *Nataliia Olkhovyeh, UAEMLG, UA*
- *Angus Clarke, EBMG, UK*
- *Inga Prokopenko, ESHG, UK*
- *Andrew Read, ESHG, UK*
- *Christine Patch, ESHG, UK*
- *Ros Hastings, ESHG-QSC, UK*