11th Meeting of the European Human Genetics Societies
Sunday, May 22, 2016, 11.00 – 13.00 hrs
CCIB - Centre de Convencions Internacional
Barcelona, Room 211, Floor P2

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

2. **Self-presentation of the meeting participants**
Number of countries represented: 30

3. **ESHG highlights**
President of the ESHG

Feliciano Ramos gave an overview of the activates of the ESHG committees (see also Newsletter), stressed the importance of the collaboration with other societies, specially IFHGS which is currently lead by Helena Kääriäinen. Main future challenges: official recognition of specialities in all European countries, homogenisation of training programs in Human genetics, the availability of educational recourses (physical/on line webinars), renovation of the webpage.

4. **European Board of Medical Genetics**
Heather Skirton, Chair of EBMG

Heather gave an overview of EBMG work and progress. During the past year the online system for submission of registration applications has been launched. It works already for the Clinical Laboratory Genetics (CLG) Branch and shall be introduced for Genetic Nurse and Genetics Counsellor (GC) Branch next year. CLG branch extended applications for those from countries 2 and 3. Those from countries 3 (with no national system of assessment) are required to do an exam (written and oral), and the first exam took place at the ESHG conference in Barcelona.
Associate GC registration will be offered to candidates outside Europe.

Gen-Equip project was presented. The aim is to provide genetic education for primary and secondary care. There is a website with e-material in different languages. Accreditation for CME is in the process.

5. **DNA Day 2016**
Christophe Cordier, ESHG Education Committee

In total 189 essays were received from 19 countries. Top scoring countries were Turkey, France, and Italy, contributing more than one half of all essays. The winners

were from Romania and Italy. More than 60 geneticists from 27 countries have volunteered to assist with judging these essays.

6. **European Union of Medical Specialists – Clinical Genetics**
Bela Melegh, Chair UEMS

Bela Melegh presented the structure and vision of UEMS. Section of Clinical Genetics of UEMS was established in 2013. Some of achievements and ongoing tasks: proposal to a new MJR of Rare and Undiagnosed Diseases, proposal to change the name to Section of Clinical Genetics and Genomics, preparation of syllabus and curriculum (completed and accepted).

7. **ESHG Public and Professional Policy Committee**
Martina C. Cornel, Chair of the Committee

One of the main activities last year was the paper on Expanded carrier screening recommendations, recently published in EJHG. The coming activities are: Post mortem genetics, Gene editing in cooperation with ESHRE, and Genomic Sequencing Results in Pediatric Practice and Exome sequencing in Newborns with complex severe syndromes. PPPC would like to encourage NSHG to discuss and disseminate the expanded carrier screening recommendations at the national level and to give suggestions to PPPC for future activities.

8. **Education Committee and the ESHG sponsored courses**
Han Brunner, Chair Education Committee

Han Brunner presented educational committee activates: educational courses, DNA day, school children event, and collection of resources for education. Course portfolio has been extended with new courses this year. It was stressed that courses are not organised by ESHG, however ESHG supports the courses by fellowships and a limited budget for the organisation. An additional activity was proposed: a task force on HGVS nomenclature. The aim is to provide a package of activities to improve knowledge of HGVS nomenclature. Johan den Dunnen, who currently chairs the HGVS/HVP/HUGO Sequence Variant Description Woking Group (SVD-WG), is proposed to lead the task force.

9. **Guest presentation 1: International Federation of Human Genetics Societies (IFHGS)**
Helena Kääriäinen, Helsinki

Helena Kääriäinen presented IFHGS. ESHG has a presidency of IFHGS for years 2015-2017. The challenges are: to incorporate the missing societies (like India, Iran and others), to promote genetics in countries with the biggest difficulties relating to access to genetic services and education, and to ensure that genetics is performed in an ethically sound way. An ESHG board group for IFHGS was established: Feliciano Ramos, Zeynep Tumer and Angus Clarke volunteered to think about ideas on how to promote the global genetics. All members are encouraged to send their ideas, proposals to Helena, Jerome, or ESHG board group for IFHGS.
The next IFHGS will be in Cape Town in 2021.Han Brunner proposed that the next ICHG is not just a physical but also an electronic congress.

10. **Guest presentation 2: ERN for Rare Congenital malformations and Intellectual Disability**

Jill Clayton-Smith, Manchaster

Jill Clayton-Smith presented a proposed ERN for rare congenital malformations and intellectual disability called ITHACA (Intellectual disability TeleHealth And Congenital Anomalies). The goal is to provide a patient centred network that will provide an infrastructure for diagnosis, evidence-based management and collection of patient data. Endorsement process is on track in 14 EU countries. The main challenges are to set up a network with no initial recourses, to deal with a large amount of paperwork, and to write a competitive application to get the network funded.

11. **Guest presentation 3: ERN GENTURIS**
Nicoline Hoogerbrugge

Nicoline Hoogerbrugge presented a proposed ERN for Genetic Tumour Risk Syndromes (GENTURIS). The aim of GENTURIS is to improve the identification, diagnosis and surveillance of a wide range of rare inherited syndromes predisposing for tumor development at any stage during life. The unique points: individuals are predisposed to common cancers of various systems, healthy individuals need surveillance and risk reducing treatment (prevention oriented), and relatives should be included. Endorsement has already been obtained from 8 countries, and there are some more in the process.

List of Delegates

Armenian Society of Human Genetics:	Tamara Sarkisian
Belgium Society of Human Genetics:	Paul Coucke
British Society of Genetic Medicine:	Jill Clayton-Smith
British Society of Genetic Medicine:	Joana B Melo
Bulgarian Society of Human Genetics:	Draga Ivanova Toncheva
Clinical Genetics Society:	Joana B Melo
Croatian Society for Human Genetics:	Ivana Sansovic
Czech Society of Human Genetics:	Milan Macek Jr
Danish Society for Medical Genetics:	Elsebet Östergaard
Estonian Society of Human Genetics:	Ants Kung
European Society of Human Genetics:	Gunnar Houge
European Society of Human Genetics:	Feliciano Ramos
European Society of Human Genetics:	Olaf Riess
European Society of Human Genetics - PPPC:	Martina Cornel
European Society of Human Genetics:	Karin Writzl
European Society of Human Genetics:	Jerome del Picchia
French Association of Genetic Councellors:	Christophe Cordier
Genetics Group, Malta College of Pathologists:	Isabella Borg
Georgian Society of Medical Genetics & Epigenetics:	Elena Abzianidze
Georgian Society of Medical Genetics & Epigenetics:	Eka Kvaratskhelia
German Society of Human Genetics:	Gabriele Gillessen-Kaesbach
Hellenic Society of Medical Genetics:	Aspasia Tsezou
Hungarian Society of Human Genetics:	Bela Melegh
Icelandic Human Genetics Society:	Vigdis Stefansdottir
International Federation of Human Genetics Societies:	Helena Kääriäinen
Irish Society of Human Genetics:	Sally-Ann Lynch
Israeli Society of Medical Genetics:	Lina Basel-Vanagaite
Latvian Society of Human Genetics:	Zanda Danebergh

National Board of Georgian Geneticists:	Dodo Agladze
Norwegian Society of Human Genetics:	Cystein Holla
Romanian Society of Medical Genetics:	Maria Puiu
Russian Society of Medical Genetics:	Vera Izhevskaya
Serbian Genetics Society, Medical Genetics Branch:	Ivana Novakovic
Spanish Association of Human Genetics:	Juan Cigudosa
Slovenian Society of Human Genetics:	Karin Writzl
Swedish Society of Human Genetics:	Hans Ehrencrona
Swiss Association of Genetic Concellors:	Christophe Cordier
UEMS Section Human Genetics:	Bela Melegh