



EUROPEAN SOCIETY OF HUMAN GENETICS

MINUTES from the

9th Meeting of the European Human Genetics Societies Sunday, June 9th, 2013, 11.15 – 13.15 hrs Palais de Congrès Paris, Room 342A

1) Welcome and Introduction

by Milan Macek, NHGSs liaison of ESHG

2) Self-presentation of the meeting participants

There was very good attendance, from an increasing number of CoE countries.

3) ESHG highlights

by Stanislas Lyonnet, President of the ESHG

Stan highlighted recent developments in ESHG, e.g. new membership structure, the plan to create a course portfolio, and future conferences in 2014 (Milan, May 30-June 3), 2015 (Glasgow, June 6-9) and 2016 (Istanbul). He also invited participants to suggest "sustainable" courses that could be of sufficient quality to receive ESHG endorsement and sometimes also ESHG support.

4) Establishment of the European Board of Medical Genetics (EBMG) and the new registration system for genetic nurses and counsellors *by Heather Skirton, Chair of EBMG*

Heather gave an overview of EBMG work and progress. The clinical genetics section represents a recognized medical speciality in the EU, the genetic counsellor section has just opened up for formal registration, and the laboratory genetics section has made a core curriculum but is otherwise in the progress of getting an overview of the status in different European countries. The two latter sections also await approval of the EU Professional Qualifications Directive, which takes unexpectedly long for political reasons. Heather urged medical professionals in the various countries to support genetic counsellors when they ask for recognition (www.eshg.org/ebmg.0.html).

5) Union of European Medical Specialists – Clinical Genetics

by Ulf Kristofferson, Division chair of EBMG

Recently, the Clinical genetics section (CGS; concerns only MDs) has been established at UEMS.eu This section, after being officially instituted through nominations of national delegates during Autumn 2013 (see further), will closely collaborate with EBMG. CGS will consist of 2 delegates (delegate/alternate) from each of the 34 UEMS member countries, usually selected by their respective clinical/medical specialist (professional) associations. In many countries there is only one "genetics" society comprising both the professional and scientific aspects of the field, while in other countries there is a separate professional association and a scientific society

(www.eshg.org/76.0.html). In this regard, a letter from the UEMS President was sent to the heads of the constituent national ("overarching") medical associations (or equivalent) on May 3rd, 2013 informing them about the creation of the CGS. The national medical association should have forwarded this letter to the national specialist association asking them to co-opt/elect their delegates to CGS. If you had not been informed by your national medical association please inquire directly with them. The "overarching" medical specialist body will then report the national clinical/medical genetics delegates to the UEMS secretariat and to CGS. Afterwards, CGS will form a Board that consist of members from both the professional (medical specialist) societies and the where applicable also of the "corresponding" scientific society. After elections, this board will likely "overlap" and closely collaborated with the EBMG clinical genetics section of ESHG (see attached also the letter from prof. Kristofferson from April 2, 2013 to NHGS).

6) ESHG Public and Professional Policy Committee: overview of activities
by Martina C. Cornel, Chair of the Committee

ESHG/PPPC recommendations on use of next generation sequencing in the clinic were recently published (www.eshg.org/13.0.html). Martina also touched upon Myriad Genetics policy NOT to share variant data (i.e., keep them as trade secrets), even though this company has profited from public open access data and in many instances their tests were financed by public health insurance (see recent article in EJHG, by Degan et al). Note: new developments after the NHGS meeting - www.eshg.org/141.0.html At ESHG 2013 we have a workshop on the use of genetic testing in the judicial system ("Judging our genes"). PPPC is also involved in the development of neonatal screening policies (www.nature.com/ejhg/journal/vaop/ncurrent/full/ejhg201390a.html), including other items under consideration.

7) ESHG Genetics Services Quality Committee (GSQC)
by Ros Hastings, Chair of the Committee

GSQC are currently conducting a a quality survey on genetic counselling. In addition, the GSQC deals with lab quality, especially EQAs (external quality assessments). Some European labs still do not follow OECD guidelines for reporting of genetic laboratory results. There are also several labs with persistent unsatisfactory performance, and how to deal with this is a challenge, varying from country to country and on how e.g. the accreditation bodies work. See also the Eurogentest.org web site for more information on quality assurance.

8) Guest presentation 1: European Committee of Rare Diseases Experts (EUCERD)
by Helena Kaariainen, Deputy Secretary General – Vice chair EUCERD

This is an advisory committee to the European Commission. EUCERD.eu has been very active, but the status of rare disease policies varies between different European countries. Some countries, e.g. France, Germany and Spain, have already established rare disease plans, while other countries are still within the preparatory phase. It is important that the role of geneticists is recognized in the rare disease field within NHGS constituent countries. A new EUCERD committee will start from Autumn 2013.

9) Guest presentation 2: Concerted effort in rare disease research (€ 40M):
EU projects EUrenOmics, NeurOmics and RDConnect (see www.irdirc.org)
by Olaf Riess, Tübingen, Germany

Olaf presented the RD-Connect (www.rd-connect.eu), NeurOmics (rd-neuromics.eu/) and EuRenOmics (www.eurenomics.com/) EU projects which represent an unique

concerted action of rare diseases / genetics financed by the European Union. The International rare diseases consortium (IRDIRC) is instrumental in building of transatlantic collaboration in the area of rare diseases (www.irdirc.org).

10) Guest presentation 3: IVD Directive: update of its revision and implications by David Barton, Our Lady's Hospital for Sick Children, Dublin, Ireland

10'

The In Vitro Diagnostic (IVD) directive governs the use of laboratory tests, also in genetics. The IVD directive has now been improved to better suit genetics, but the new law has not been passed yet. In general, the IVD directive mainly concerns the manufacturing of diagnostic kits, but concerning genetics, a proposal has been made that could affect clinical practise as well (e.g. requirements for genetic counselling). Eurogenetest and ESHG have published a joint statement expressing joint concerns. Providing, that you you agree please promote this view among politicians / EU parliamentarians of your own country (www.eshg.org/fileadmin/www.eshg.org/documents/ESHG/ESHG_Statement_on_IVD_Regulation.pdf).

List of Delegates

1. Armenian Society of Human Genetics: Tamara Sarkisian
2. Austrian Society of Human Genetics: Peter M. Kroisel
3. Belgian Society of Human Genetics: Thomy de Ravel
4. Clinical Genetics Society of Croatia: Nina Canki-Klain
5. Croatian Society of Human Genetics: Ivona Sansovic
6. Cyprus Society of Human Genetics: Adonis Ioannides
7. Cyprus Society of Human Genetics: Philippos Patsalis
8. Czech Society of Human Genetics: Milan Macek Jr.
9. Danish Society of Medical Genetics: Jens Michael Hertz
10. Dutch Society of Clinical Geneticists: Merleen Kets
11. Dutch Society of Laboratory Specialists Clinical Genetics: Hans Kristian Ploos van Amstel
12. Estonian Society of Human Genetics: Andres Metspalu
13. Fédération Française de Génétique Humaine: Didier Lacombe
14. Finnish Society for Medical Genetics: Carola Saloranta
15. German Society of Human Genetics: Christine Scholz
16. Hellenic Association of Medical Genetics: Manos Papadakis
17. Hellenic Society of Medical Genetics: Constantinos Pangalos
18. Hungarian Society of Human Genetics: Bela Melegh
19. Hungarian Society of Human Genetics: Marta Szell
20. Icelandic Society of Human Genetics: Reynir Arngrimsson
21. Irish Society of Medical Genetics: Sally Ann Lynch
22. Latvian Human Genetics Association: Baiba Lace
23. Macedonian Society of Human Genetics: Dijana Plaseska-Karanfilska
24. Malta College of Pathologists – Medical Genetics: Alex Felice
25. Norwegian Society for Medical Genetics: Torunn Fiskerstrand
26. Polish Society of Human Genetics: Janusz Limon
27. Polish Society of Human Genetics: Krystyna Chrzanowska
28. Polish Society of Human Genetics: Olga Haus
29. Portuguese Society of Human Genetics: Lina Ramos
30. Romanian Society of Medical Genetics: Maria Puiu
31. Russian Society of Medical Genetics: Vera Izhevskaya
32. Serbian Society of Human Genetics: Dragica Radojkovic
33. Slovak Society of Human Genetics: Ludevít Kadasi
34. Slovenian Association of Medical Genetics: Anamarija Brezigar
35. Slovenian Society of Human Genetics: Emanuela Bostjancic
36. Société Française de Génétique Humaine: Alain Bernheim
37. Spanish Society of Human Genetics: Feliciano Ramos
38. Swedish Society of Human Genetics: Hans Ehrencrona

39. Swiss Society of Medical Genetics: Dunja Niedrist
40. Turkish Society of Medical Genetics: Hakan Ulucar
41. Ukrainian Appointed Delegate: Halyna Macukh

42. Guest Speaker: Olaf Riess

43. ESHG: Stanislas Lyonnet
44. ESHG: Gunnar Houge
45. ESHG: Jörg Schmidtke
46. ESHG: Ros Hastings
47. ESHG: Tayfun Özcelik
48. ESHG: Jerome del Picchia

49. EBMG: Heather Skirton
50. EBMG: Ulf Kristoffersson