



MINUTES

10th Meeting of the European National Human Genetics Societies Sunday, June 1, 2014, 11.30 – 13.30 hrs MilanoCongressi, Room: Amber 1+2

Countries represented: 33

| | |
|---------------------|--|
| Armenia: | Tamara Sarkisian |
| Austria: | Peter M Kroisel |
| Bosnia-Herzegovina: | Naida Lojo-Kadric |
| Belgium: | Thomy JL de Ravel |
| Croatia: | Nina Canki-Klain |
| Cyprus: | Adonis Ioannides |
| Czech Republic: | Milan Macek Jr., also ESHG liaison to NHGSs |
| Denmark: | Jens Michael Hertz |
| Estonia: | Ants Kurg |
| France: | Didier Lacombe |
| Germany: | Christine Scholz |
| Greece: | Myrto Poulou |
| Hungary: | Bela Melegh |
| Iceland: | Vigdís Stefánsdóttir, Reynir Arngrímsson |
| Ireland: | Sally Ann Lynch |
| Italy: | Antonio Amaroso, Domenico Coviello |
| Latvia: | Ilze Radovica |
| Macedonia: | Dijana Plaseska-Karanfilska |
| Malta: | Isabella Borg |
| The Netherlands: | Dominique Smeets, Frank Baas, Christi van Asperen |
| Norway: | Torunn Fiskerstrand, Inga Bjørnevoll |
| Poland: | Beate Lipska-Zietkiewicz, Olga Haus, Krystyna Chrzanowska |
| Portugal: | Hildeberto Correia |
| Romania: | Maria Puiu, Christina Rusu |
| Russia: | Vera Izhevskaya |
| Serbia: | Dragica Radojkovic |
| Slovakia: | Ludevit Kadasi |
| Slovenia: | Damjan Glavac, Luca Lovrecic |
| Spain: | Juan Cruz Cigudosa |
| Sweden: | Erik Iwarsson |
| Switzerland: | Siv Fokstuen |
| Turkey: | Tayfun Özcelik |
| United Kingdom: | Angela Douglas |
| EBMG: | Heather Skirton |
| ESHG: | Han Brunner, Jerome del Picchia, Helena Kääriänen, Karen Writzl, Gunnar Houge |
| IRDIRC: | Kim Boycott |

1) Welcome and Introduction

Milan Macek, NHGSs Liaison of ESHG

2) Self-presentation of the meeting participants

No of countries represented: 33, 1-3 representatives per country

3) ESHG highlights

Han Brunner, President of the ESHG

The new status of EBMG (European Board of Medical Genetics) as a separate legal entity made by ESHG to promote high professional standards within Europe, was mentioned.

Our opposition to the inclusion of regulation of genetic counselling in the EU IVD directive was also mentioned – we do not oppose the directive, but that this directive is used to regulate clinical practise related to genetic counselling in member states – which is at best inappropriate, in reality also illegal. The regulation of such IVD devices should “stop at the hospital door”. A petition was sent around for supportive signatures.

Finally, a possible new workshop format was mentioned: A workshop with good stories from individual members – with a special reach-out to less privileged countries.

4) European Board of Medical Genetics: progress and plans for 2015

Heather Skirton, Chair of EBMG

EBMG is now a legal entity under Austrian law. Both counsellors and clinical laboratory geneticists (CLGs) await approval of the EU Professional Qualifications Directive (still unclear when/if this will be approved). In the mean time a certification system is being set up, and this work has advanced most for genetic counsellors: 18 counsellors (of 31 applicants) are now registered as counsellors approved by EBMG. Finally, Heather encouraged the attendees to promote the value of such registration in their home countries.

5) Union of European Medical Specialists – Clinical Genetics

Ulf Kristofferson, Division chair of EBMG

Each country (more precisely: the medical association of that country) appoints two members to the medical genetics section of UEMS. So far 19 countries are represented. Goals: To update the curriculum, to make an inventory of specialists in Europe, to develop a specialist exam in collaboration with EBMG (personal comment of ESHG gen sec: maybe EBMG will turn out to be the “executive arm” of the UEMS committee?), develop syllabus for the training program, and develop a course program.

Additional comment of the ESHG gen sec: maybe EBMG will turn out to be the “executive arm” of the UEMS committee? Maybe UEMS could promote some of the courses run by ESHG as good for specialist training? (e.g. the ESHG-ESMG course in medical genetics).

6) ESHG Public and Professional Policy Committee

Martina C. Cornel, Chair of the Committee

Martina presented a policy document on New Born Screening (NBS) developed together with other societies (in Australasia, US and HUGO) – see our website.

Our recommendation on Whole Genome Sequencing (WGS) for diagnostic testing was published a year ago. ESHG recommend a targeted approach – and not compulsory feedback on all findings in a list of “high risk genes”. We now also collaborate with the SIC section of ASHG on issues of common interest, e.g. the genetic testing of minors.

A document on preconception carrier screening and one on NIPT are under preparation – these documents will soon (this autumn) be put on our website for comments.

7) ESHG Genetics Services Quality Committee

Ros Hastings, Chair of the Committee

The committee has prepared reporting guidelines for genetic laboratories published in EJHG (Open Access).

A committee member is ESHG representative on the HGVS nomenclature committee.

A recurrent concern is persistent poor performance – i.e. wrong results that may give patients the wrong diagnosis and cause patient harm. A few labs do not appear to improve despite bad results year after year.

They are also modelling a EQA test for genetic counselling, currently only in English. This is a pilot – open for participation for those interested.

8) Guest presentation 1: Cross-border healthcare in genetic diagnostics

Helena Kaariainen, EUCERD.eu Joint Action

EUCERD is an EU rare disease expert group. A lot of test samples cross borders in Europe. The magnitude of this activity is unknown, and the same is the motivation. A survey was set up, and (only) 11% of labs and 17% of clinics responded. Based on this result it was estimated that maybe as many as 100 000 samples cross European borders each year. The majority of labs just sent a very small portion of their samples to other countries. The main reason was lack of test in home country.

There were several communication problems between external lab and local doctor/lab.

Often it was unclear if a test was predictive or diagnostic, often the result was misunderstood, and sometimes it was not clear if genetic counselling had been or could be offered. The main selection criteria for external labs were quality and price, on third place came reputation. Use of “brokers” (external test hubs) was not common. Reimbursement of costs was a problem not only for the external labs, but also for the local doctor/lab.

9) Guest presentation 2: International Rare Disease Consortium and the

phenomecentral.org project

Kym Boycott, University of Ottawa, Canada

Goal: By 2020 to have 200 new therapies for rare diseases approved. Present commitment by major stakeholders: > 1 billion USD worldwide. The IRDiRC diagnostics committee’s goal is to have found all the genes for rare diseases by 2020. A main challenge is to resolve the n=1 situation – i.e. a genomic matchmaker is needed. The recommended ontology of IRDiRC is HPO. Consensus on 2300 standard rare disease terms has been reached. PhenomeCentral has been developed: You add your patient using HPO terms, then you add your vcf-file from genomic sequencing, and then you may see patients that are similar to yours.

10) Guest presentation 3: Coding genetic lab tests: a proposal for a logical system

Torunn Fiskerstrand, University of Bergen, Norway

Torunn gave an orientation on the Norwegian experience of lab coding systems required by the authorities and (usually) based on ClinChem codes (NPU codes). It appears unfortunate that another speciality (Clinical Chemistry) is in charge of developing our codes – on the other hand coordination is useful, and this committee has representatives that are specialists in medical genetics.

11) DNA Day 2014

Tayfun Özçelik, Chair ESHG Education Committee

Tayfun presented the results of the 2014 DNA day. Despite a difficult question, we have three excellent winners (Italy, UK and France) and the participation was good. The feedbacks from the reviewers were also largely positive. 10-15% plagiarism was detected.