

Minutes of the 4th Meeting of National Human Genetics Societies (NHGSs)

During ESHG Conference in Barcelona, June 2nd, 2008, at 11.15-13.15

(All presentations can be viewed following the hyperlink in the title)

1. Welcome and Introduction

by vice president Pier Franco Pignatti, who was leading the meeting.

He stressed the importance of these meetings to learn to know each other and improve collaboration.

2. Self-presentation of the participants

Sarkisian, Tamara F.	Armenia
Duba, Hans-Christoph	Austria
Seneca, Sara	Belgium
Kapur, Lejla	Bosnia and Herzegovina
Barisic, Ingeborg	Croatia
Baric, Ivo	Croatia
Canki-Klain, Nina	Croatia
Georgiou, Theodoros	Cyprus
Foretova, Lenka	Czech Republic
Jensen, Thomas G.	Denmark
Aittomäki, Kristiina	Finland
Pöyhönen, Minna	Finland
Bonneau, Dominique	France
Propping, Peter	Germany
Reis, André	Germany
Papadakis, Manos	Greece
Florentin-Aveiv, Lina	Greece
Karcagi, Veronika	Hungary
Green, Andrew	Ireland
Kerem, Batsheva	Israel
Larizza, Lidia	Italy
Pronina, Nataleija	Latvia
Kucinskas, Vaidutis	Lithuania
Plaseska-Karanfilska, Dijana	Macedonia
Stefanovska, Emilija S.	Macedonia
Felice, Alex	Malta
Van Ommen, Gert Jan	The Netherlands
Heimdal, Ketil	Norway
Mazurczak, Tadeusz	Poland
Fortuna, Ana Maria	Portugal
Rusu, Cristina	Romania
Covic, Mircea	Romania
Ginter, Evgeny K.	Russia
Radoskovic, Dragica	Serbia
Brezigar, Anamarija	Slovenia
Damjan, Glavac	Slovenia
Palau, Francesc	Spain
Joncourt, Franziska	Switzerland
Ozcelik, Tayfun	Turkey
Elles, Rob	U.K.

From ESHG:

Cassiman, Jean-Jacques	President
Pignatti, Pier F.	Vice President

Kääriäinen, Helena	Secretary General
Coviello, Domenico	Education committee
Burn, John	Former president
Del Picchia, Jerome	Executive Officer
Houge, Gunnar	Secretary general elect (ref)

There were 40 national representatives and 7 from ESHG board/committees.

3. Collaboration between ESHG and NHGS

Pier Franco Pignatti (Vice President of ESHG)

ESHG has supported the EGF courses and will continue to do so. In addition, it plans to arrange European courses of its own. It is now also possible to arrange regional courses that can be supported by the ESHG. The initiative for the regional courses must come from the NHGSs. In addition, ESHG provides fellowships to ESHG Conferences via NHGSs.

The DNA Day Contest was arranged by ESHG for the first time, and there were 118 participants from different countries. Similar contest has been arranged by ASHG for some time to celebrate the description of the DNA helix. This will need help from NHGSs in the future: they have to pre-evaluate the texts and send (and translate) only the best ones to the final contest. This will be informed in detail later.

4. Membership in the ESHG and the NHGSs

Jerome del Picchia (Executive Officer of ESHG)

Jerome underlined the advantages of being a member not only of a national genetics society, but also of ESHG, e.g.

- to get the European Journal of Human Genetics
- to access to member lists/information in member area of web site

The poster prize awarded by ESHG at this conference includes a 1-year free ESHG membership.

5. The European Journal of Human Genetics

Gert Jan van Ommen (Editor in Chief of EJHG)

The Journal is doing very well both financially and scientifically. There has been a 20% increase of submissions last year, and an 8% increase in volume (i.e. also a higher rejection rate). The junior author receiving the highest number of citations 1-2 years after publication receives a 1-year free membership and a 500 Euro price.

6. Activities to be planned

Jean-Jacques Cassiman (President of ESHG)

The ESHG should be the voice of human genetics in Europe (as decided in the strategy meeting of Board that took place in Brussels in November-07). Major goals are:

- *quality* science and services - e.g. establish infrastructure for genetic testing, establish common platform for European EQAs, make guidelines for the interpretation of ISO15189 that concerns medical genetic laboratories.
- *communication* - improved web site, dedicated spokes-persons in different countries, rapid reaction group to assist in giving appropriate genetic information.
- *interaction* with e.g. Eurogentest, Orphanet, the EU scientific programs through EU-liason officer (currently Ségolène Aymé, position to be taken over by Gert Matthijs).
- *recognition of speciality in clinical genetics* by the European Union. Ulf Kristofferson gave a status report on the work promoted by him and Helen Kingston at the UEMS (the European Union of Medical Specialities) to get the speciality in clinical genetics approved by the EU. It is a positive but slow process – even slower than anticipated. It would be a great advantage if the speciality also was

promoted at the political level (e.g. by two or more ministers of EU countries). Ulf also said that there is now a system for obtaining CMEs (continuous medical education points) for clinical geneticists, e.g. by attending meetings like this.

- *information* – e.g. develop pamphlets and other patient information in collaboration with Orphanet and Eurogentest.

- stimulate *implementation of the [OECD guidelines for quality assurance in molecular genetic testing](#)* that has been ratified by most European countries.

7. Suggestions by the NHGSs representatives

The Serbian representative advertised for the 8th Balkan meeting on human genetics (to take place in Cavtat-Dubrovnik, May 14-17, 2009), and mentioned that this was a good place to promote Eurogentest and Orphanet.

Another representative would like to hear more about the organization of the more established national genetics societies: the small/new societies could possibly learn how to promote their own society.

8. [The gene patent document](#)

Gert Matthijs (Chair of ESHG Patenting and Licensing Committee)

Gert gave an overview of the document on patenting and licensing, that now has been published as a supplement to our Journal (Eur J Hum Genet 16, suppl 1, May 2008). The document has been made by the Public and Professional Policy Committee (PPPC) and Patenting and Licensing Committee (PLC) on behalf of the ESHG.

One experience made during the last years is that patent opposition in court is a very expensive and time-consuming endeavour, and that opposition through the media (to influence public opinion) can be a better approach.

9. [The susceptibility testing in common disorders document](#)

Martina Cornel (Chair of ESHG Public and Professional Policy Committee)

Geneticists should take a stand on this issue and try to influence policy in the right directions. One of the crucial issues is that susceptibility tests (as any genetic tests) should have their clinical validity and utility proven before taking them to use.

10. [A clinical genetics meeting update](#)

John Burn (ESHG Board member)

John gave an update of a meeting held in Liverpool in conjunction with a meeting of the British society of clinical geneticists in March 2008. There is an urgent need for a common (basic) clinical geneticist training program across Europe, e.g. to support countries without an established infrastructure, and to promote speciality recognition. A formal European clinical genetics diploma could also be a good idea.

To get on with this work John would like to have contact with one clinical geneticist from each European country. These representatives should make a network for common policy establishment. He also wants to establish a “clinical geneticist rapid response team” in Europe.

**The name of a clinical geneticist representing your country can be mailed to:
John.Burn@newcastle.ac.uk**

The German representative stressed the importance of having not only a clinical speciality, but also positions within the health care system for such specialists.

The Czech representative expressed great concern after the removal of clinical genetics from the

main medical speciality list one month ago. Reason: Clinical genetics was not recognized by the EU as a medical speciality.

11. **The European Network of Genetic Nurses and Counsellors**

Heather Skirton (Chair of ESHG Subcommittee for Counsellors/Nurses)

The first meeting of this network took place in Barcelona with 40 participants from 23 countries. The main goals of the network are:

- establish contact with colleagues in other countries
- establish guidelines for education of genetic nurses/counsellors
- establish professional standards for safe practise

12. **The organization of ESHG courses**

Domenico Coviello (Chair of ESHG Education Committee)

ESHG has as a new activity started to organize, in addition to yearly Conference, also European courses. Two such courses are currently being planned (see web site):

- "Training the trainees in genetic counselling"
- "Genetic epidemiology"

13. **General Discussion and Conclusions**

The Secretary General reminded the audience about BBMRI, the European collaboration of biobanks funded by EU. Information about this was sent to members short time ago. If NHGSs know about biobanks in their country, they are kindly asked to inform the biobanks about BBMRI.

The President stressed that one of the main goals of collaboration between ESHG, Orphanet, Eurogentest, etc. is to "avoid re-inventing the wheel". In depth work on important common issues done by other organisation or a member state may be adopted with or without modifications by other parties.

The Executive Officer reminded the audience that every national representative can update and expand the information on their own society by directly accessing their own pages on the ESHG web site. Username and password is provided by the Vienna office (eo@eshg.org).

It was decided that former president Pier Franco Pignatti, now vice president, will lead this meeting for (at least) one more year.