



Minutes of the 6th Meeting of the European Human Genetics Societies Sunday, June 13, 2010, 11.15 – 13.15 hrs Gothenburg Convention Centre

1. Welcome and Introduction

Milan Macek Jr., NHGSs Liaison and President Elect

Milan Macek opened the meeting, thanked Professor Pier-Franco Pignatti for initiating and establishing the meeting of National Human Genetics Societies (NHGSs), and introduced the agenda.

2. Self-presentation of the participants

Belgium	Helen A. Poirel (President)
Czech Republic	Milan Macek Jr. (President)
Bulgaria	Draga Toncheva (Secretary)
Macedonia	Dijana Plaseska-Karanfilska (Secretary)
Bosnia and Herzegovina	Lejla Kapur-Pojkic (Secretary)
Spain	Feliciano J. Ramos (President)
Portugal	Ana Maria Fortuna (Vice President)
Germany	André Reis (President)
Croatia	Nina Canki-Klain (President)
Hungary	Bela Melegh (President)
The Netherlands	Fred Petrij (Secretary)
	Nine Knoers (President)
Norway	Ketil Heimdal (President)
Poland	Tadeuz Mazurczak (President)
	Michal Witt (Member)
Romania	Christina Rusu (Vice President)
Lithuania	Vaidutis Kučinskas (President)
Finland	Kristiina Aittomäki (former President)
Turkey	Tayfun Özcelik (Member)
Denmark	Uffe Birk Jensen (President)
Latvia	Linda Piekuse (Member)
Slovenia	Karin Writzl (President)
	Mojca Stražišar
Slovakia	Ludovit Kadasi (President)
Greece	Lina Florentin (President)
Serbia	Dragica Radojkovic (President)
Cyprus	Violetta Anastasiadou (President)
Austria	Hans-Christoph Duba (President)
France	Doninique Bonneau (President)
Italy	Ludovica Volpi
	Lidia Larizza (President)

Ireland	David Barton
Israel	Karen Avraham (Treasurer)
Switzerland	Dunja Niedrist
Sweden	Niklas Dahl (President)
	Hans Ehrencrona (President)
UK	Christine Patch (Chair)

ESHG Committees:

Ad hoc Clinical genetics speciality committee/UEMS: Ulf Kristoffersson

PPPC: Martina Cornel

EC: Peter Farndon

Ad hoc Laboratory genetics speciality committee: Jacqueline Schoumans

Ad hoc Genetic counsellors and nurses speciality committee: Heather Skirton

ESHG Executive Board:

President:	Dian Donnai
Vice President:	JJ Cassiman
President Elect:	Milan Macek Jr.
Treasurer:	Andrew Read
Secretary General:	Helena Kääriäinen
Secretary General Elect:	Gunnar Houge
Executive Officer:	Jerome del Picchia

Invited:

Laurence Lwoff, Head of Bioethics Division, Council of Europe

3. ESHG highlights

Dian Donnai, President of the ESHG

As much of ESHG activities from the past year had already been presented in the ESHG Opening ceremony, Dian Donnai only mentioned several key issues. She specifically mentioned the positive development of a very strong bond between ESHG and NHGS.

Since ESHG's main role is academic promotion of science in the field of human genetics, its conferences are the most important annual "highlight". ESHG has prioritised support of young scientists for these conferences via several approaches: fellowships directly from ESHG as well as via NHGSs, lots of opportunities to present (also orally) their results, including Young Investigator Awards. In addition, ESHG has recently started to organize ESHG courses and ESHG-supported courses.

Another main function is support of professionals. There the recognition on European level of the three (main) specialties involved in human/medical genetics: clinical / medical geneticists, laboratory medical geneticists and genetic counsellors / nurses has been and will remain a priority for ESHG. The process has advanced most regarding clinical / medical geneticists and the experience drawn from this effort will help in similar work towards recognition of the two other specialties.

In general, activities of ESHG have increased and have been facilitated by the part time involvement of its Executive Officer - Jerome del Picchia, as well as by the more

active role of the ESHG Executive Committee, that has 2-3 meetings in between the yearly Board meeting at the time of the ESHG.

The achievements of respective ESHG Committees were presented only in a brief overview as they have been reported *in extenso* in the Newsletter.

The official journal of the society, European Journal of Human Genetics is doing well. Apart from the successful "Practical Genetics" series, one of its new approaches which has been developed in collaboration with the EuroGentest Network (www.eurogentest.org) is continuous publication of "Clinical Utility Gene Cards" in order to create European-wide standardized practises for genetic testing.

4. Special Guest presentation: Additional protocol to the Oviedo Convention on Genetic testing

Laurence Lwoff, Council of Europe

Laurence Lwoff explained the aim of the „Additional Protocol to the Convention on Human Rights and Biomedicine concerning Genetic Testing for Health Purposes“ (<http://conventions.coe.int/Treaty/en/Treaties/html/203.htm>), which is the first legally binding instrument on genetic testing in Europe - for further details see also the European Journal of Human Genetics 2009; Nov; 17(11): 1374-1377. The CoE considers genetic tests as having a huge benefit as well as a potentially significant risk, especially now when the capacity to analyse the human genome is much higher than the capacity to intervene based on the results of genetic testing. In creating the Additional Protocol, the working group of CoE has collaborated with ESHG experts and found PPPC reports very useful.

Implementation of the Additional Protocol in the various European legislations and practices has been slow, but has already started in several European countries. Moreover, seminars on the topic will be held in many countries, including follow up on its implementation. Laurence Lwoff expressed her hope for further collaboration with ESHG and with all other European professionals via NHGSs.

At present, many countries have not ratified the "Oviedo Convention" which would allow them to ratify its Additional protocols. In spite of that, the Additional protocol is of great importance for policy makers and professional bodies in many countries.

Milan Macek suggested that implementation of the CoE Additional protocol could happen simultaneously with implementation of EU Council Recommendation on action in the field of rare diseases (eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:C:2009:151:0007:0010:EN:PDF).

5. Current status of the recognition of the clinical / medical genetics specialty

Milan Macek Jr., NHGSs Liaison and President Elect

Milan Macek gave a brief update of the current status of the recognition of the clinical / medical genetics specialty at the EU-27 level as detailed report is available at the ESHG website (www.eshg.org/index.php?id=111). Further development will result from the pending vote of the DG Internal Market and Services Recognition committee on June 18/2010, and the result will be presented in the ESHG electronic Newsletter (Flashletter) within coming weeks.

He acknowledged the great value of the quick and efficient collaboration with NHGSs within the last year which has created a "critical mass" of supportive evidence for the deliberations of the EC Recognition committee, as compiled at the dedicated section of the ESHG website "Genetics as a Medical Specialty in Europe". It is therefore very likely that the necessary "Qualified majority" (en.wikipedia.org/wiki/Voting_in_the_Council_of_the_European_Union) will be achieved.

The majority of EU-27 countries have 4-5 years training and have recognized this specialty at the national level (22/27), except Belgium, Cyprus, Greece, Luxembourg and Spain. Hungary has only 2 years of postgraduate training since medical genetics is a subspecialty, thus far.

Experience drawn from this process will be utilised for the recognition of the two other genetic specialties and will foster national recognition of clinical / medical genetics in the countries listed above.

6. Current status of the preparations for recognition of the laboratory genetics specialty

Jacqueline Schoumans, Chair of ESHG Ad-hoc Committee on a laboratory genetics speciality. E-mail: Jacqueline.SchoumansPouw@unil.ch

Jacqueline Schoumans initially presented the members of the Ad-hoc Committee (see ESHG website – Genetics as a laboratory specialty in Europe - www.eshg.org/224.0.html). The approach to get this specialty recognised at the EU level will be similar to that applied for clinical / medical genetics – ie. to collect data a/ on the present situation in Europe, b/ on the existence and recognition of specific education programmes (nearly completed), and c/ on the existence of legal dossiers related to "official" recognition of this specialty at the national level. The suggested name of the speciality is a "*medical genetics laboratory specialist*" and the most common length of the education program appears to be 4 years, in line with requirements of the European Commission. The Ad-hoc Committee will also utilise important policy statements on quality assurance in genetic testing as stipulated by the "OECD Guidelines for Quality Assurance in Molecular Genetic testing", in particular with regards to its Section E (www.oecd.org/dataoecd/43/6/38839788.pdf), which have a bearing on the official recognition of the laboratory specialty in Europe, and beyond. Comments and suggestions are welcome, and active collaboration with NHGSs will be sought.

In the following discussion the name of medical genetics laboratory specialist was criticized by some, there were wishes to try to accomplish recognition of some or all of the genetics specialties simultaneously, with hopes that the recent progress of the adoption of the EU Directive on patients' rights in cross-border healthcare (www.consilium.europa.eu//uedocs/cms_data/docs/pressdata/en/lsa/114992.pdf) may create a momentum to pursue recognition of genetic specialties as soon as possible, including doubts that some countries do not want to have all three specialties etc.

It was noted that the EC Directive on In Vitro Diagnostic Medical Devices 98/79/EC (eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=CELEX:31998L0079:en:HTML) which forms the basis for providing CE marking on e.g. laboratory assays is under revision at the moment. Nearly all non-CE marked "in house" genetic tests are currently allowed under the "exemption" provision in this Directive. A consultation document will be sent around in order to receive support for continuation of this exemption, including a suggestion for a new provision in the updated Directive in that only ISO 15189 accredited laboratories will be allowed to use non-CE-marked genetic

tests. This measure will support quality assurance in genetic testing in line with the OECD guidelines and will also foster recognition of the laboratory specialty in Europe.

7. Current status of Genetic Nurse and Counsellor Ad Hoc Accreditation Committee

Heather Skirton – Chair of the Committee

Genetic nurses and counsellors have started a broad professional network with approximately 140 members from 23 European countries. Anyone is free to join. Many of the genetic nurses/counsellors are not members of ESHG, but they value the role of ESHG / NHGSs in advancement of their recognition at the EU level.

At present, the role of genetic nurses/counsellors in different countries/health care systems has been mapped, there is a code of practice, and a set of core competences has been accepted. The next step is that NHGSs should evaluate whether these are acceptable for the delivery of genetic services in a given country, formally approve them at the professional body level and subsequently seek their official recognition by relevant national authorities. She will send documents for evaluation and approval to NHGSs in the near future.

8. Current activities of the PPPC: Recommendations on DTC testing

Martina Cornel, Chair ESHG Professional and Public Policy Committee (PPPC)

Activities of the PPPC were summarised in the Newsletter. Martina Cornell presented the most timely topic, ie. "Statement of the ESHG on direct-to-consumer (DTC) genetic testing for health-related purposes" related to its rapid spread within Europe. This statement has been posted on the ESHG website (www.eshg.org/120.0.html) for comments, and the final approval process is ongoing. Furthermore, PPPC is preparing a "nutshell" version of the Statement, which could be used as a supporting "position paper" by NHGSs when dealing with respective national authorities regarding this topic. Future projects of PPPC will comprise genetic testing related to mental health/autism, including rapid advances in newborn screening.

9. Eurogenest: an Overview of its outcomes

Jean-Jacques Cassiman, Vice president of the ESHG

EuroGentest (EuGT; www.eurogentest.org) is a EC funded Network of Excellence which will be discontinued by the end of June 2010. However, EC will fund its successor EU Concerted Action project termed "EuGT2" (2011-2013) albeit with substantially lower budget) with the aim to achieve its long-term sustainability beyond the EC funding.

EuGT has focused especially on improvement of the quality assurance in genetic testing; development of genetic databases in collaboration with Orphanet (www.orpha.net); establishing training workshops and professional guidelines; promoted EQA-programs; collaboration with various European agencies regarding relevant policies for genetic testing; creating guidelines for genetic counselling, pregraduate and postgraduate education and services; assessing novel technologies; defining clinical utility gene cards; establishing core competences for different genetic professionals; translating very popular and informative patient information leaflets into many languages; including surveying legislation related to patient rights and genetic testing. Overall, EuroGentest became a reference source for genetic professionals in Europe and beyond.

Helena Kääriäinen also presented a web tool "Instrument for quality self-assessment in provision of genetic counselling" which is available at www.eurogentest.org/web/assessment_tool/index.xhtml

10. ESHG Quality Committee

Ros Hastings, Chair Genetics Services Quality Committee (GSQC)

Also this Committee has a comprehensive summary in the Newsletter. NHGSs could take on a role in their own countries in promoting the broad implementation of EQA-schemes in molecular genetic and cytogenetic testing.

11. DNA Day 2010 and genetic education

Peter Farndon – ESHG Education Committee

ESHG Board has decided to continue the DNA-day initiative. The plan is to develop it further and NHGSs will be informed on how they are wished to act for the next year. The role of the NHGSs probably has to be increased, to support the teachers, to advertise and to help in pre-evaluating the texts as translating all texts to English is may soon become unfeasible.

Another educational initiative is the "Educational day" at IFHGS 2011 in Montreal (www.ifhgs.org), which could help the geneticists involved in teaching to learn and share their methods etc. This issue was proposed, but has not been accepted thus far.

12. A new EU Liaison group

Strategic collaboration of NHGSs together with ESHG regarding policy matters related to the FP7/FP8 EC research framework programmes, the newly formed EUCERD committee (www.eucerd.eu), certification of clinicians, genetic infrastructure projects etc. were briefly discussed.

ESHG will start a new group/committee, the "EU-liaison group" (EULIG) which will be proactive towards the EC and with regards to EC activities in the area of genetics and rare diseases. The preparatory group will be headed by Jean-Jacques Cassiman and Martina Cornel