MINUTES

7th Meeting of the European National Human Genetics Societies
Sunday, May 29th 2011, 11.15 – 13.15
Amsterdam RAI Conference Center, room E102

1. Welcome and Introduction

Milan Macek Jr., NHGSs Liaison and ESHG President

Milan Macek opened the meeting and stated that this 7th meeting means continuing of the regular contact between ESHG and NHGSs, which ESHG has found extremely valuable.

2. Self-presentation of the participants

The participants (n = 52, listed below) usually represented Human Genetics Societies, sometimes Clinical Genetics Societies or other genetics-related societies in European countries (i.e. not only from EU27). In addition, there were participants representing ESHG and several invited speakers.

Belgium	**Helen A. Poirel**
	Jean-Jacques Cassiman
	Inge Liebaers (CoE Committee Chair)
	Gert Matthijs (EUGT2)
Czech Republic	**Milan Macek Jr. (President ESHG)**
Bulgaria	**Draga Ivanova Toncheva**
Macedonia	**Dijana Plaseska-Karanfilska**
Spain	**Feliciano J. Ramos**
Portugal	**Astrid Vicente**
Germany	**Bernhard Horsthemke**
	Jörg Schmidtke (President-Elect ESHG)
Croatia	**Ingeborg Barisic**
	Nina Canki-Klain
Hungary	**Bela Melegh**
The Netherlands	**Hans K Ploos van Chustel**
	Johan den Dunnen (Leiden Genome Technology Centre)
	Egbert Bakker (ad hoc ESHG Lab genetics committee)
	Martina Cornel (PPPC)
	Hans Scheffer (Techgene)
Iceland	**Vigdis Stefansdottir**
Norway	**Torunn Fiskerstrand**
	Gunnar Houge (General Secretary ESHG)
Romania	**Christina Rusu**
	Maria Puiu
Lithuania	**Vaidutis Kučinskas**
Finland	**Jaakko Ignatius**
	Sari Rasi
	Helena Kääriäinen (Deputy Gen Sec ESHG)
Turkey	**Tayfun Özcelik**
Latvia	**Janis Klovins**
Slovenia	**Anamarija Brezigar**
	Emanuela Bostjancic

Slovakia	**Ľudovit Kadasi**
Greece	**Lina Florentin**
	Constantinos Pangalos
Serbia	**Dragica Radojkovic**
Austria	**Peter Michael Kroisel**
Estonia	**Andres Metspalu**
France	**Alain Bernheim**
	Didier Lacombe
Russia	**Vera Izhevskaya**
Italy	**Domenico Coviello**
Malta	**Isabella Borg**
Armenia	**Tamara Sarkisian**
Ireland	**David Barton**
	Sally Ann Lynch
Sweden	**Maria Soller**
UK	**Christine Patch**
	Peter Turnpenny
	Heather Skirton (ad hoc genetic counsellors committee)
	Ros Hastings (GSQC- ESHG)
Executive Officer ESHG	**Jerome del Picchia**

3. ESHG highlights and EU-wide recognition of clinical genetics speciality

Milan Macek Jr, President of the ESHG

1) The great achievement was accepting clinical/medical genetics as an official European medical specialty in early March 2011, a detailed report by Milan Macek was published in the ESHG Newsletter 2011. This means that ESHG will end the respective *Ad Hoc* Committee, but now UEMS needs to create a section for this specialty. From now on ESHG can concentrate on recognition of European specialties for genetic nurses/counsellors and laboratory geneticists.
2) A map on the situation of European specialties was presented.
3) There was the happy news that Icelandic Human Genetics Society has been founded since the last NHGSs meeting (www.mannis.is).
4) All Societies were urged to be active in the implementation of the EU Council rare diseases recommendation by 2013 and promote the work of EUCERD.eu committee, where ESHG also has its representatives.

4. Special Guest presentation: Inge Liebaers

Last year, CoE Committee of Ministers published "Recommendation CM/Rec(2010)11 of the Committee of Ministers to member states on the impact of genetics on the organisation of health care services and training of health professionals"

Inge Liebaers (who chaired the committee) presented the background for this document. The initiative came from The Netherlands, and the work was requested by the European Parliament. Preparing a consensus text took several years and the final result, a non-binding recommendation, hopefully would stimulate the governments to recognize the importance of the quality of genetic services and its increasing impact on health care. The NHGSs are invited to promote this recommendation in their countries and actively inform about it their policy makers or health ministry officials. Inge Liebaers has also written about this recommendation in the ESHG Newsletter 2011.

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

Milan Macek Jr., NHGSs Liaison and President ESHG
Egbert Bakker, co-chair of the ESHG ad hoc lab genetics committee

Bert Bakker presented the Dutch curriculum and the respective educational program ("Genoompuzzle"). In brief, the Dutch education covers divergent functions/areas like laboratory expert, communicator, team player, manager, social skills, organisation, professional, knowledge

and science. The first two years there is strong focus on technology, the second two years further specialization into modules. Intellectual disability and oncogenetics are compulsory modules, while others are optional. The trainee accrues a training portifolio that is supervised by their trainers (supervisors). The people in charge of the program have to be trained and properly qualified, as well.

The European aim is to try to create a consensus European education programme for laboratory geneticists that could be used for molecular and chromosomal laboratory geneticists as well.

There are already several (nine at present) approved national programs in Europe/EU: the plan is to compare them and find common denominators of the required competences. Some exist only in national languages, which complicates the procedure.

ESHG is well aware that there are problems in defining what part of genetics services could be performed by MDs and what part by laboratory geneticists (while some have both specialties/competencies in several countries). ESHG however does not want to get involved in national discussions, but rather aims at finding a European level solution to the specialty of laboratory geneticists, including its recognition on both Member State and European levels. ESHG response to the Professional Qualifications Directive EC consultation is one of the ways to achieve EU-wide goals for laboratory geneticists (and genetic nurses and counsellors).

The new chair of the laboratory genetics committee is Thomas Liehr from Jena.

6. Eurogenest2 continuation of the success story: aims and objectives

Gert Matthijs, project coordinator

The major European network of excellence Eurogentest.org (EuGT) started 6 years ago and ended recently. Fortunately, its work will continue for 3 more years but within a smaller EuGT2 EC project. The aim of EuGT was to improve and harmonize the genetic testing process at all levels from indications through clinical and laboratory phases to dealing with the implications. EuGT2 will continue this work and specifically try to find a way to promote the quality in a sustainable way beyond the time limitation of EC projects.

One way towards harmonisation of the quality is via accreditation; also clinical services can and should be accredited.

EuGT2 collaborates with various EQA providers, ESHG (PPPC and GSQC), Orphanet (which registers laboratory accreditation at the European level), NHGSs etc.

EuGT2 follows closely the IVD directive (the concern is that it could restrict home-made methods in situations where these have to be used, such as in the area of rare diseases), ethical / legal issues related to DTC, fetal/prenatal testing/screening, including NIPD by utilising work performed within the previous SAFE EC project, creating clinical utility gene cards (CUGCs) and publishing them in the EJHG (and at the ESHG website).

The coming months are important in deciding in which way EuGT2 will secure the continuation of its work, a plan is to form an “Association” and in this process the support of NHGSs will be needed. EuGT2 strongly believes that both laboratory and clinical genetic services should be accredited and will work together with Techgene and ESHG PPPC on the challenges provided by introduction of NGS technology into diagnostic practice.

7. Techgene: next generation sequencing in diagnostic practice

Hans Scheffer, Project Coordinator

The Techgene.eu EC project is working in the emerging and exciting field of applying NGS into diagnostic practice and aims at training of fellows in this important field. It is not yet clear how

whole genome/exome sequencing will be used in clinical diagnostics, most likely specific informatics filters for the clinical conditions under examination will be used (i.e. MR or deafness). Issues related to unsought for (co-incidental) findings and related informed consent procedures need to be elaborated. The project will transform itself at its end into an open "Knowledge Network".

8. Databases for compilation of detected variants related to next generation sequencing: Dutch initiative

Johan den Dunnen, Leiden Genome Technology Centre (10')

The plan is to facilitate publishing data related to NGS-detected variants: i.e. to regularly publish new variants, to report them in an identical way, to propose to journals a policy that fosters inclusion of variant data into public databases as a condition for publishing submitted papers etc. Discussions were initiated with the EJHG in this regard. Finally, it is important to publish and register variants in an uniform way by using the HGVS nomenclature.
All these aims are at present being practiced by "Dutch Variant Database", in the future this kind of database/tool will be openly available. A central database is in the process of being established in Leiden and LUMC Genome Technology Centre. All variants should be shared online to facilitate their interpretation in both diagnostic and research settings..

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

Heather Skirton – Chair of committee

At present, there is an organisation/society for genetic counsellors/nurses in 5 European countries. These, other organisations and "known leaders" have been consulted with the aim to set the standard for education. There already exists a draft document which includes all the elements of counsellor education. It has been circulated for comments last year and also presented at ESHG website; no negative comments have emerged.

Next aim is to reach every individual to ask about their education and work roles. This forum was invited to help in contacting all genetics nurses/counsellors in Europe. The plan is to finish the core curriculum this year and start plans for building a process of assessment of competences and in the end achieve European recognition, similarly to the clinical / medical genetics specialty.

One of the important tasks was to comment the EC consultation on the proposed Professional Qualifications Directive.

Plan for future: The Ad Hoc committees will continue their work. Suggestions/comments from NHGSs are welcome.

Milan Macek will write an Editorial to EJHG concerning the recognition of clinical / medical genetics, its immediate consequences at the national level, and on the need for the two other specialties to be recognised in the near future.

Both items 8 and 9 aroused several comments, some are collected below:

1) What would be the names of these specialties? It would be useful to reach an European consensus on these names.

2) In some counties all three specialties have separate societies while in some other countries there exists only one society. In Spain, nurses/counsellors have recently started a society. In UK there is discussion about merging the separate societies.

3) The question whether somatic genetics should be part of the curricula of all the specialties was asked. This would create a possible overlap with pathologists, which could be sorted out by a shared part of the laboratory genetics curricula?

4) In Italy, “genetic nurse” might be a good name for professional and economical reasons.

5) ESHG was asked to inform somehow all member states/European countries about the European clinical/medical genetics specialty and about the immediate need for the other two specialities.

6) Would the specialty training be part of medical or biological faculties/schools? In the Netherlands training of laboratory geneticists must be linked to medical schools. How could PhD programs we associated with laboratory specialty training.

7) Once new specialties would have been accepted, should there be a transition period? How to inform policy makers and related professional societies (eg. pathologists and clinical chemists).

8) It should be clarified at the national level whether laboratory / genetic nurses counsellors specialties are officially (governmentally) recognised or where such recognition had been achieved at the ”learned society” level.

9) In Lithuania a new master program for genetic counsellors is starting up.

10. Current activities of the PPPC: next generation sequencing, newborn screening and behavioural genetics

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

The ESHG Statement on direct-to-consumer genetic testing for health-related purposes was published in the European Journal of Human Genetics and is available at the ESHG website.

In addition, a background document and recommendations on genetic testing for common disorders in a public health framework was developed in collaboration with EUGT1 and was published in EJHG (and is also available at the ESHG website).

Several other topics were discussed during the year 2010-2011 and are at different stages of preparation: genetic testing and mental health, whole genome sequencing and its consequences for health care, using biological samples of children in genetic research, adoption and genetic testing, neonatal screening, and preconception genetic testing.

PPPC was also involved in commenting on the IVD directive revision (background work was done by David Barton – EUGT1/2 and Stuart Hoghart), their comments are available at the ESHG website (www.eshg.org/fileadmin/www.eshg.org/documents/ESHG/ESHG-IVD-def.pdf)

11. ESHG Quality Committee

Ros Hastings, Chair Genetics Services Quality Committee (GSQC)

The main discussion in GSQC last year has been related to laboratory quality, the challenge is how to help the many laboratories who repeatedly perform poorly in EQA trials. In the future, also the quality of genetic counselling will be approached building on the work performed in EuGT1 and elsewhere.

On Friday May 26th a EHGC 2011 Conference satellite meeting “Array in daily practice: promises and pitfalls” was successfully organised and attended by approximately 120 participants.

12. DNA Day 2011

Celia de Lozier – ESHG Education Committee

The interesting DNA-day contest was shortly presented, see DNAday.eu. The question raised was: how could NHGSs promote the contest in their countries as now only some countries participate. Lithuania has been a positive example and their approach to this success was was enquired. Vaidutis Kucinskas told that the Ministry of Education has been actively involved and stimulated by the Lithuanian genetics society. In addition, local media have been very active. This could serve as a positive example for other NHGSs.

13. Other issues

There were no other issues and so Milan Macek closed the meeting.