



European Society of Human Genetics

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Report from the President

by Milan Macek

Dear colleagues and friends,

First of all I would like to cordially welcome you to our 2011 Annual European Human Genetics Conference which is being held at the RAI conference centre in Amsterdam! We are really grateful to our Dutch colleagues for their invitation and our Society returned to the same place where we held our 2006 annual conference. This proves not only the attractiveness of the venue, and of the beautiful and vibrant city of Amsterdam, but also pays tribute to high quality genetics research and services performed in The Netherlands. Our Dutch colleagues are in many areas at the forefront of genetics research, in particular in the utilisation of next-generation sequencing, which is rapidly changing current paradigms with regards to gene / mutation identification, orphan drug development and nowadays even patient treatment. This meeting will provide, yet again, an exciting overview about rapid developments in human genetics. High quality genetic research has become a central part not only in its traditional domain of rare disease research, but is also currently generating tangible progress in the area of multi-factorial disorders, where it could yield useful and clinically relevant information for individual patients. We are all looking forward to yet another memorable meeting both from the scientific and social points of view. Our thanks thus go to the local organising committee and its chair, our local host, Robert Hofstra.

We are also indebted to Brunhilde Wirth and her colleagues at the Scientific Programme Committee for preparing a great programme, which now successfully implements an informative "Educational Track" throughout the entire meeting. We also acknowledge excellent annual conference organisation by our colleagues from the Vienna Medical Academy, represented by our Executive Officer Jerome del Picchia, who combines his high professionalism with a truly enthusiastic and kind approach! We are indebted to Rose International for high quality organisation of a growing industrial exhibition, without which this conference and many other activities of our Society would not have been possible.

Long-term activities of our Society are well known and you will find reports of individual committees in other sections of this Newsletter. In this regard, I would like to acknowledge the dedicated work of the ESHG Executive

Board, Board members and all of the ESHG committees. It needs to be stressed that all reported activities are based on voluntary hard work of many people who provide their expertise and especially their valuable time to our Society! We should bare this in mind and sincerely thank them for their efforts.

Last but not least we are grateful to the Chief Editor of the European Journal of Human Genetics, GertJan van Ommen, who together with a team of Section Editors is steering our scientific journal which maintains its prominent position among genetics journals. In this respect we are pleased that the Journal not only publishes cutting edge science, but also spreads best practice through its "Practical Genetics" or "Gene Card" series, which have become very popular.

Our Society also aims at strengthening genetic services-oriented activities throughout Europe in that we are fostering European-wide recognition of various subspecialties of genetics comprising clinical-/medical geneticists, laboratory specialists and genetic nurses and counsellors. I am pleased to announce that after a long-term effort which started in late 2008, finally on March 3rd, 2011 a major milestone was reached – i.e. the Commission adopted Regulation (EU) No 213/2011 amending Annexes II and V to the "Directive 2005/36/EC of the European Parliament and of the Council on the recognition of professional qualifications". This administrative act implies that the clinical-/medical genetics specialty was officially recognised as an "EU-wide specialty". This regulation is also relevant for the so called EEA countries (Switzerland, Norway, Iceland) and will be important in the near future for many EU-Accession



countries, thereby potentially reaching a European-wide relevance, and thus facilitating transnational recognition and mobility of clinical geneticists.

ESHG would like to sincerely thank all collaborators and stakeholders who had worked in the past within the administratively and politically demanding recognition process, notably the Presidents and/or representatives of respective European national human-/clinical-/medical genetics societies, without whom this marvellous achievement would not have been possible! The facilitating role of the European Union of Medical Specialists (UEMS) in the development of a consensus curriculum is appreciated. The ESHG is particularly pleased that this successful Europe-wide initiative reflects the strength, dedication and the genuine “team spirit” of European genetics!

The ESHG would also like to encourage all stakeholders to proactively inform about the newly achieved status of clinical- / medical genetics their national medical associations, medical chambers and/or respective competent government authorities which are instrumental in the process of recognition of individual qualifications for cross-border provision of health-care, whereby rare diseases (hence predominantly genetic disorders) duly recently received a prominent status. Simultaneous recognition of medical oncology together with clinical- / medical genetics within the above mentioned Commission Directive underscores the relevance of both specialties for the field of rare diseases.

We are particularly pleased that shortly following the publication of the EC Directive there has been substantial progress in countries where the clinical specialty had not recognised, thus far. We already received encouraging news from Spain and Ireland, and these developments will be discussed at our meeting.

The ESHG wants to stress, as it had repeatedly done so in the past, that it acknowledges that the two other constituent specialties are as important as clinical- / medical genetics and that these are integral components of comprehensive provision of genetic services in Europe. Recognition of clinical- / medical genetics specialty was thus not „prioritised“ in any way, rather we utilised an unique momentum provided by the “EU Council Recommendation on an action in the field rare diseases” (adopted in June 2009), that is pertinent to the provision of clinical services.

However, there are still significant challenges in front of us in that we need to achieve the same status also for the laboratory genetics specialty. There is also tangible progress on this front - currently we are in the process of translation of the Dutch postgraduate curriculum (termed “Genoompuzzel”), which was adopted at the end of March 2011 by the Dutch society of clinical genetic laboratory specialists. This high quality structured curriculum could serve as a template for the development of the European consensus curriculum and will be presented at the ESHG 2011 conference in Amsterdam.

The Commission has recently recognised the arduous tasks particularly related to the recognition of non-medical specialties, which we have faced and that substantially

hampered our desire for a more rapid progress in the EU-wide recognition of the two „remaining“ genetics specialties. Development of consensus curricula at the transnational level is very difficult in the absence of „UEMS-like“ bodies and certain national regulatory measures have proved to be almost insurmountable, if the current DG Internal Market and Services recognition procedures were still to be applied. We are grateful that the Commission became aware of these general administrative obstacles and that they launched the “Public Consultation on the Professional Qualifications Directive”. The ESHG endorses the proposed simplification of the trans-national recognition procedures, a move which will also markedly facilitate EU-wide recognition of the laboratory specialty and of genetic nurses and counsellors. ESHG endorsed simplification of recognition procedures which, if adopted by the Commission, could significantly facilitate EU-wide recognition of the laboratory geneticists and of genetic nurses and counsellors.

Finally, the ESHG in close collaboration with the Eurogentest European Network of Excellence (that has been “reincarnated” as Eurogenest2) provided an official response of the Commission Public consultation on the revision of “Directive 98/79/ec of the European Parliament and of the Council of 27 October 1998 on *in vitro* diagnostic medical devices”, which among others endorses an important exemption for clinical genetic laboratories testing rare diseases (of which 80% have a genetic origin) and that are accredited to an appropriate international standard, such as ISO 15189.

Another part of service oriented activities in the last year comprises important publications of the Professional and Public Policy Committee on e.g. “Genetic testing and common disorders in a public health framework” or “Statement of the ESHG on direct-to-consumer genetic testing for health-related purposes”. Currently, the PPPC is concentrating on topical position statements related to the application of next-generation sequencing in clinical diagnostic practice and collaborates in this regard with colleagues from another important European project – Techgene. These documents serve as guidance for all professionals active in genetic services and are commonly used to formulate national position papers or are sometimes even “directly” used in negotiations with national regulators and/or health insurance companies.

Several ESHG Board members have been active in the “Committee of Experts on the Impact of Genetics on the Organisation of Health Care Services and Training of Health Professionals” at the Council of Europe which drafted an informative guidance document termed- “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”. We are pleased that the Chair of this Committee, Inge Liebaers, will present an outline of this important document at our Amsterdam meeting.

ESHG very much values our collaboration with 43 National Human Genetics Societies who will meet already for their 7th time at our annual meeting! Cooperation with national societies has become not only a strong tradition, but also is very fruitful – i.e. without strong support from national societies, EU-wide recognition of clinical-/human genetics

would not be possible and we plan to work with them also for the two other constitutive specialties in a similar manner. We would like to congratulate our Icelandic colleagues who established their national society last year and we have established contacts with our Ukrainian colleagues in order to get this last major European society “on board”, as well. It should also be stressed that when working with our membership and with national societies, the annual ESHG Newsletter kindly edited by Lina Florentin and the *ad hoc* ESHG electronic Flashletters proved to be optimal means of communication.

ESHG also supports on the long-term basis young students by providing them with fellowships to the medical genetics courses organised by the European Genetics Foundation. I myself am alumni of the 1990 “Sestri Levante” course where I had received a fellowship, enabling me to meet similarly minded peers (who since then have mostly assumed prominent roles in European genetics) and to meet excellent faculty. Thus, I very much understand how such “seminal” financial support is crucial for talented

people originating from the current less favoured regions in Europe, and beyond.

We are also glad that DNA Day Essay contests (in partnership with the American Society of Human Genetics) carried out at European high schools proved to be very popular! We are always pleasantly surprised and enriched by students’ unconventional and yet inspiring views on topical scientific concepts.

I am honoured to pass the presidency to Jörg Schmittke, who will invigorate our Society with a follow-up initiative to the gradual process of recognition of constituent genetic specialties, which will ensure the highest levels of competence for physicians, scientists and counsellors working in the field and to foster their mobility by the development of “portable” EU-wide qualifications. I wish him well in this challenging role!

Milan Macek
President of the ESHG

Report from the Scientific Programme Committee

by Brunhilde Wirth

A personal welcome from the program chair

On behalf of the ESHG Program Committee, I welcome you to the ESHG conference in Amsterdam.

The Scientific Programme Committee for 2010-2011 included Brunhilde Wirth (chair), Corinne Antignac, Frank Baas (Local SPC member), Antonio Baldini, Jeffrey Barrett, Alexis Brice, Han Brunner, The-Hung Bui, Manolis Dermitzakis, Robert Hofstra (Local Host), Gunnar Houge, (Secr. General elect, observer), Helena Kääriäinen, (Secretary General; ex officio observer), Stefan Mundlos, Carla Oliveria, Pete Scambler, Michael Speicher, Eduardo Tizzano, Draga Toncheva, Mikka Vikkula, Bert de Vries (Local SPC member) and Cisca Wijmenga.

The SPC met twice in order to organize the Amsterdam 2011 ESHG conference: in July 2010, to decide upon the plenary sessions and symposia and in March 2011, in Vienna at the VMA offices, to select the abstracts for oral presentations and posters.

The number of submitted abstracts was 2127 and thus the highest number ever submitted to an ESHG meeting. All abstracts have been scored on-line by 3-6 SPC members, thus allowing a reliable selection system of the top 5% for the oral presentations. Based on the topics and scores, 108 abstracts were selected for the 17 concurrent sessions and one plenary session. Among the oral presenters, 45 were Young Investigator Candidates (at least 1 in every session), reflecting the high level of contribution of young scientists to this program.

Of the 1717 posters, 73 were selected as best posters and will be marked with an ESHG star. Of these, 36 were Young Investigator Candidates and were qualified for the poster prize. 127 abstracts will be “publication only” and 152 abstracts were rejected.

2011 Meeting Highlights

The meeting will start with the first plenary session including three local stars: *Han Brunner, Cisca Wijmenga and Hans Clevers*. The “What’s new?” session will be followed by the most exciting new findings selected from submitted abstracts. The “educational sessions” throughout the program were very well attended last year, therefore the committee decided to continue with 9 educational symposia this year. These include: “Mitochondrial diseases and Neonatal screening”, “HNPCC and BRCA1 & 2”, “eQTLs & GWAS”, “Schizophrenia and Autism”, “Prenatal rapid aneuploidy detection and mosaicism”, “Marfan and EDS”, “Noonan and Neurofibromatosis”, “Copy number variations and arrays in practice”, “EJHG- How do I get my paper published?”.

On Tuesday, a third plenary session on “The personal genome” with *Aravinda Chakravarti, Carlos Bustamante and Mark McCarthy*, will give us new visions on a new era in human genetics since whole genome sequencing started.

The meeting will conclude with our distinguished speaker and 2009 Nobel Laureate *Elizabeth Blackburn* from San Francisco for her breakthrough research on telomeres and their role in aging. As usual, our second highlight of the final day of the conference will be the acceptance speech by our ESHG prize winner. This year, the ESHG award 2011 will be awarded to *Gert Jan van Ommen* from Leiden, NL, in recognition of his groundbreaking work in the development of a therapy for Duchenne Muscular Atrophy using antisense oligos and his overall contribution to the European Journal of Human Genetics.

After the Amsterdam conference, the SPC shall have to say goodbye to Han Brunner, Pete Scambler, Eduardo Tizzano, and Cisca Wijmenga. We thank them for their work and their dedication into making the meeting better. At this point I would like to particularly thank Han Brunner for being an excellent chair for many years and making this meeting an ever coming highlight of our society.

I hope that all of you will enjoy the meeting in Amsterdam.
Prof. Dr. Brunhilde Wirth



2011 Scientific Committee Program Chair, Institute of Human Genetics, University of Cologne, Germany

Report from the Secretary General

by Gunnar Houge

These are exciting times in genetics. As General Secretary of our Society, you are well positioned to follow current development in most areas of genetics, and you certainly feel how genetics has moved from the outskirts to the mainstream of medical practise. The EU-wide recognition of genetics as a clinical speciality in early March 2011 reflects this development. EU and UEMS inclusion of clinical/medical genetics was finally achieved thanks to the efforts of many, in particular John Burn, Arnold Munnich, Helen Kingston, Dian Donnai, Ulf Kristofferson and, last but not least, our current President Milan Macek. This implies that the ESHG *ad hoc* committee of clinical genetics, lead by Ulf, no longer exists because it is no longer needed. Hopefully, this will eventually be the case for our two other *ad hoc* committees as well: the one for EU recognition of medical laboratory genetics and the one for EU recognition of genetic counsellors/nurses.

Since there is no standardized system for recognition of these specialities, like there is for clinical specialities through UEMS, an alternative pathway for pan-European professional recognition must be found. The suggested EU Professional Qualifications Directive could be such

a pathway. Let us hope it gets final EU approval! Then the hurdles that must be overcome for genetic counsellors and laboratory geneticists to achieve EU recognition will be greatly reduced compared to having to move the specialities through the EU Recognition Committee, like it had to be done for clinical genetics. It is a great difference between needing support from at least 2/3 of all EU member states to maybe only 9. An idea could be to have these specialities recognized as so-called "28th regime" professions under European law, i.e. as approved European training programs with European certifications that exists in addition to national training programs. To establish a body that will deal with the three branches of professional qualifications in clinical and laboratory genetics (i.e. MDs, laboratory scientists and genetics counsellors/nurses), our next president, Jörg Schmidtke,



has suggested that a European Board of Medical Genetics under the ESHG umbrella is established.

I feel privileged to be your general secretary in a time when long-standing riddles of medicine and genetics are being solved at an increasing pace thanks to the new efficient technologies offered to us by array and high-throughput sequencing platforms. However, we all know that genetics is so much more than technology – it is how to exploit

technology to the best of our patients and how to deal with uncertainty in both the laboratory and clinical setting that distinguish our professions. These are exciting times – much of the guesswork from earlier days have been replaced by an even greater challenge: How to make sense out of an ever-increasing amount of data and to be good guardians and genomic guides for our patients and society when genetics are becoming a part of everyday medicine.

Report from the Public and Professional Policy Committee 2010-2011

by Martina Cornel

In the year 2010-2011 the Public and Professional Policy Committee (PPPC) of the ESHG mainly worked on the challenge of using genetic testing in health care in a responsible way, while avoiding unsound applications. Three published documents relate to the balancing of pros and cons of large scale applications of genetic testing for common disorders, both in health care and direct to consumer testing. A background document and recommendations on genetic testing for common disorders in a public health framework have been published in the *EJHG* (2011;19:S6-S44 and 2011;19:377-81). This was the result of a long standing collaboration with EUROAGENTEST and the IPTS: Institute for Prospective Technological Studies, Joint Research Centre, European Commission, Seville, Spain. Recommendations of the ESHG on DTC were published in the *EJHG* (2010;18:1271-3). All documents were approved by the Board after consultation of the ESHG membership. In line with the Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes, the DTC Statement wants to highlight the importance of the right to information, the quality and utility of genetic testing services, individualized medical supervision, the provision of information and genetic counselling, the protection of persons not able to consent, and the respect for private life. The first documents take the spectrum ranging from monogenic disorders on the one hand to common complex disorders on the other hand into account. It is argued that associations between genetic variants and disease risks of clinical relevance have been established, for instance for cancer and cardiovascular disorders. Although these examples relate to the monogenic sub forms of common disease, they can nevertheless be used to reflect on the possibilities and relevant obstacles when using the new genetics in public health. New possibilities for genetic

testing confront health-care workers with the question whom (not) to test and which test (not) to use. The term 'common disorders' is used to denote that the disorders are relevant from a public health point of view because of their high frequency. Common disorders are, for instance, cardiovascular disease, stroke, diabetes, cancer, dementia and depression. For a health-care practitioner – unlike a



geneticist or an epidemiologist – it is not clear, the moment a patient enters health care, whether the common disorder in this case is due to one gene with a high risk of serious disease, or due to a combination of several genes and several environmental factors. Both the lack of translation of useful applications and the availability of tests without clinical utility need, are addressed in the documents.

Several other topics were discussed during the year 2010-2011, and will be followed up later: 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. Members of the PPPC in 2010-2011 were Nurten Akarsu, Pascal Borry, Anne Cambon-Thomsen, Martina Cornel (Chair), Francesca Forzano, Shirley Hodgson, Christine Patch, Borut Peterlin, Jorge Sequeiros, Maria Soller, Aad Tibben, Lisbeth Tranebjaerg, supported by Carla van El.

Ad Hoc Genetic Nurse and Counsellor Accreditation Committee

by Heather Skirton

Co-Chairpersons: Professor Heather Skirton and Marie-Antoinette Voelckel

The focus of our committee is to ensure that genetic counsellors and genetic nurses are educationally prepared for their roles and are able to work competently with families. Ultimately, our objective is to achieve accreditation for genetic counselling as a profession in Europe.

During 2009/10 we prepared a set of standards for education and practice of genetic counsellors in Europe. These have now been published in a paper in the *Journal of Community Genetics*, where it was the most downloaded article in April, which gives us encouragement about the level of interest in this topic.

Following endorsement by the five existing national

societies for genetic counsellors and/or nurses, the standards were presented at the last meeting of the Presidents of the National Genetics Societies in Gothenburg in May, 2010. The full document was sent to all national genetics societies after that meeting and we did actively seek feedback on the acceptability of the standards in individual countries. We received very



positive comments from representatives of the national genetics societies of several countries, with no comments signifying rejection of the standards.

During this year we conducted a study of genetic counsellors and nurses in Europe, to determine the acceptance, education and legal standing of those working in these professions. We used key informants in each country to collect the data and the results of that study are being presented in poster form² at the ESHG conference in Amsterdam. We also plan to survey as many genetic nurses and counsellors in Europe as possible to ascertain their patterns of work and responsibility. The second survey is ready for distribution and we anticipate that we will be ready to publish those results in the late summer 2011. These studies are providing the basis for further activity, to ensure that we achieve European accreditation of genetic counsellors, by providing us with firm data on which to base our application. My thanks to Christophe Cordier, Debby Lambert and Ulrika Hosterey-Ugander for their hard work with respect to the surveys.

We were pleased to have an opportunity to respond to the consultation launched by the European Commission regarding the revision of the European Directive on the

recognition of professional qualifications (2005/36/EC). A copy of our response is now available on the ESHG website². It would appear that the revision to the regulations would make it a more straightforward process to protect the professional title of 'genetic counsellor' and ensure common standards of education for genetic counsellors through adoption of a curriculum-based system. It would also facilitate education and training for genetic counsellors in small countries where it would not be feasible to establish local education centres.

Finally, I would like to thank the members of the Committee, Milena Paneque, Rebecka Pestoff, Ingrid van Kessel, Vigdis Stefansdottir, Inga Bjornevoll and my Co-Chair, Marie-Antoinette Voelckel.

Heather Skirton

1. Skirton H, Patch C, Voelckel MA. Using a community of practice to develop standards of practice and education for genetic counsellors in Europe. *Journal of Community Genetics* 1(4): 169-173, DOI: 10.1007/s12687-010-0024-y.

2. <https://www.eshg.org/fileadmin/eshg/Downloads/ECCCommentsSkirtonEuropeanConsultationDocument.pdf>

Report from the ESHG Genetic Services Quality Committee

by Ros Hastings

Committee Members: David Barton; Mireille Claustres; Els Dequeker; Rob Elles; Brian Fowler; Claude Giroud; Ros Hastings (Chair); Viktor Kozich; Konstantin Miller; Cor Oosterwijk; Borut Peterlin; Conny van Ravenswaaij-Arts and Orsetta Zuffardi.

The Genetic Services Quality Committee (GSQC) meets biannually. The Committee is informally referred to as the Quality Committee and its aims are to:

- Identify gaps in quality issues within diagnostic genetic testing services;
- Identify where there can be harmonisation between the biochemical genetic, cytogenetic and molecular genetic disciplines;
- Commission and approve new documents related to quality in genetic testing;
- Give recommendations for those countries where no guidance is currently available.



Five areas of need relating to quality issues in the genetics community have been given priority and working groups with the committee will take them forward.

- Changing landscape of genetic testing;
- Laboratory performance in EQA
- Best practice Guidelines
- Rare Variants
- Newborn Screening

The application of the new genetic technologies (array CGH, whole genome sequencing), that examine the human genome, has had an impact on genetic laboratories (cytogenetic and molecular genetics) and clinical/medical genetic services. Last year the ESHG GSQC and PPPC committees coordinated an ESHG Satellite Symposium on 'Changing Landscape of Genetic Testing' prior to the ESHG conference on 11th June 2010 in Gothenburg. This year the GSQC has been involved with a further Symposium on 'Practical issues with Microarrays' that will be held on 27th May, just before the 2011 ESHG Conference.

A new revision of the ISO 15189 standards has been

circulated for a final consultation to various bodies/individuals by the ISO Standards Committee. A Quality Management System (QMS) externally verified through accreditation (ISO 15189) is the gold standard that all diagnostic genetic laboratories should attain (see also OECD guidelines). In addition, laboratories can submit their quality assurance data through the Orphanet-Eurogentest Quality Assurance database. This database enables patients, clinicians and referring laboratories to identify the nearest laboratory offering a quality service for a particular genetic disease/disorder. The database has information on the Quality Manager, EQA participation

and accreditation status.

It is an ISO requirement for an accredited laboratory to participate in EQA. External Quality Assessment (EQA) plays an important role in monitoring and improving the quality of a laboratory's service and consequently GSCQ recommends all diagnostic laboratories to participate annually in EQA for all aspects of their diagnostic service - whether or not they are accredited. The GSQC also provides a governance structure for the four European EQA schemes [CEQA - Cytogenetics, CF Network - Molecular Genetics, EMQN - Molecular Genetics and ERNDIM - Biochemical Genetics] and has reviewed their

annual management reports.

Discussions are ongoing on how the GSQC can work collaboratively with Eurogentest2 on issues around unsatisfactory (poor) performance. A working group has met to discuss the issues in more depth. In addition, the GSQC is reviewing some draft reporting guidelines applicable for diagnostic genetic laboratories.

Should you have any quality issues that need addressing, please submit them to the Chair of the Quality Committee.

A list of the Committee Members and a synopsis of the meetings are also available on the ESHG website.

Report from ESHG Education Committee April 2011

by Peter Farndon and Tayfun Oczelik (Co-chairs)

Membership and remit of the Education Committee

"The main mission is to facilitate education in genetics for health professionals to assist them in providing appropriate services for patients and families.

This will be achieved by:

- Identification of needs in educational programs in Europe with a view to support educational activities in genetics
- Supporting and encouraging the dissemination of appropriate educational material in genetics
- Promoting and encouraging the harmonization of standards in education and training in genetics for health professionals
- Liaising with relevant patient organizations and other organizations regarding education in genetics"

The ESHG executive has asked the Education Committee to reconsider the above remit. Although the mission statement remains valid conceptually, there are concerns that it may be difficult to deliver taking into account the numbers and availability of those interested in education in Europe, the constraints of professional responsibilities, and differing systems in individual countries. It has been noted that ASHG, for instance, has a fulltime education officer and supporting staff available to support education. The committee therefore felt that in the short term it was better to concentrate on developing and supporting DNA Day whilst other aims are redefined. At the Amsterdam conference, Tayfun Oczelik will take over the chair of the committee and will lead the review.

Programme during 2010-2011

DNA Day essay contest

For the fourth year, ESHG has sponsored a DNA Day Essay contest in European high schools. We have continued our partnership with the American Society of Human Genetics by using the same essay questions. Students can choose one question from the two for 2011 which are:

1. In 2010, a major discovery in genetics research found that the DNA of some modern humans contains small amounts of Neanderthal DNA. Briefly explain this finding and discuss its relevance to human ancestry and evolution.
2. A number of companies offer genetic testing directly to consumers, bypassing the involvement of physicians and genetic counsellors. Discuss whether you think this is a good idea or not. You might focus on medical, ethical, legal, or social dimensions of this issue.

Celia DeLozier, our DNA Day organiser, will be announcing the results at the European Human Genetics Conference in Amsterdam.



Educational workshops**a) European Human Genetics Conference 2011**

The committee has decided to base the education workshop held during the conference on submitted abstracts again, as this worked well last year.

b) International Congress of Human Genetics 2011

We have been involved in the conception and planning of a pre-congress workshop and a session during the conference.

i) one-day educational workshop pre-Congress on “Genetics, primary care and developing countries”

It is hoped that colleagues from twenty developing countries will consider with an expert faculty the integration of genetic medicine into mainstream clinical practice and develop general plans for improvement of genetics education and practice in each country. The structure and contents of the workshop will be tailored to meet participants’ needs. It is anticipated that workshop materials will be made available for general use via the internet.

ii) Congress invited session “Exploring International Approaches to the Evaluation of Genetics Education”

As evaluation is a key (but often neglected) component in the education cycle, this session is designed to discuss how models of evaluation can be chosen and used in a range of educational structures. This will include how to measure outcomes such as knowledge; communication; provider behaviour; and patient, social, and public health outcomes.

The ESHG programme of offering support to courses

Two workshops being held before the ESHG conference in Amsterdam are being supported through the ESHG programme. Please see the Society’s website for the procedure for applying for support for educational initiatives.

Peter Farndon, Tayfun Oczelik (co-chairs)

EJHG EDITOR'S REPORT OVER 2010

by B. van Ommen

- The major editorial change in 2010 has been the introduction of the Clinical Utility Gene Cards, a joint activity with Eurogentest, edited by Professor Joerg Schmidtke. The abstracts are published in the journal (in blocks of three to a page), and the full data are published online.
- Another improvement is that as of March 2010, authors can choose to make their paper 'Open Access', by paying a market-conform advance fee.
- During 2010, EJHG has seen a 7% drop in submissions, from 795 to 736. The number of accepted manuscripts, has in fact risen slightly, from 236 to 244, as our rejection rate fell slightly from 71 to 66%. Due to a slight change in the layout, we have, however, been able to publish this content in ca. 15% less pages, reducing the overrun of our yearly page budget from around 340 to only 37, saving cost to the society.
- Last year EJHG's impact factor fell, from 3.925 to 3.564. This implies that in the coming year the Editors have to raise the rejection rate again. Also, we wish to renew our call to our members, to consider EJHG to submit their work. We will do what is in our might to reduce the reviewing times of interesting and competitive work.
- Indeed, in the past the reviewing times have been presented in a too favorable light, because the manuscripts rejected prior to review were included, thus skewing the statistics. While it is not unusual for journals to present data this way, EJHG has deemed it more transparent to 'clean up' our statistics. About 50% of submitted manuscripts have a negative decision within 20 working days. The reviewing of the other, peer-reviewed 50% takes more time of course. After correcting for the early rejections, the mean time from submission to first decision becomes 53 working days, or about 2 ½ months. The mean time between receiving the last revision and the final decision is about three weeks. Overall, from submission to final decision, including the authors' revision time, the mean time is 97 working days or 4 ½ months. Clearly there is space for improvement in these times, and our goal is to tackle this. On the other hand, the good news for the skeptics is that amongst the peer-reviewed papers, the chance to be published is 66%.
- Once a paper is accepted, it takes, typically, only 25 days to online publication (in fact, in 7 months more than 50% of papers made it in less time, and in 9 months more than 45% of papers made this benchmark). Time to print is longer with ca. 4 months.
- As every year, EJHG has a junior authors' high-citation award, to hand out at the Amsterdam meeting. The 1st prize includes a € 500 award and places 1-3 receive one year free ESHG membership + online EJHG, and free registration for the meeting. The prizes are to be handed out at the closing session on May 31. The winners are:



1. Replication analysis identifies TYK2 as a multiple sclerosis susceptibility factor

Ban M, Goris A, Lorentzen AR, et al. EJHG 17:10.1309-1313. Oct 2009

Citations within first year of publication: 19.

2. A 15q13.3 microdeletion segregating with autism

Pagnamenta AT, Wing K, Akha ES, et al. EJHG 17:5. 687-692. May 2009

Citations within first year of publication: 14.

3. Genotype-phenotype correlations in Down syndrome identified by array CGH in 30 cases of partial trisomy and partial monosomy chromosome 21.

Lyle R, Bena F, Gagos S, et al. EJHG 17:4: 454-466: Apr 2009.

Citations within first year of publication: 14.

SPECIALTY OF GENETICS IN SPAIN FINALLY ANNOUNCED

In April 13th, 2011, the Spanish Minister of Health, Dña. Leire Pajín, announced during a session of the Spanish Parliament, (Congreso de los Diputados,) the creation of the specialty of Clinical Genetics in Spain as one of the new sanitary specialties to be included in the directory of Spain's National Health System. Ms. Pajín said that the Ministry will work with the aim of having the process ongoing before the end of the year 2011.

The Spanish Society of Human Genetics (AEGH), after almost 30 years of waiting for this announcement, is now ready to start the process for which we are already prepared, by having a draft of the specialty ready to be presented to the future National Commission of the Specialty of Clinical Genetics.

We would like to mention that we think that the inclusion of Medical Genetics in the EU Directive 2005/36/CE

concerning recognition of professional qualifications last March, supported and promoted by the ESHG, has probably influenced positively in the Ministry's decision. Finally, we hope to keep the support of the ESHG during the process.

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Brief overview of the Recommendation CM/Rec (2010)11 from the Council of Europe (September 2010)

by Inge Libaers

On September 29, 2010 the Committee of Ministers of the European Health Commission (CDSP) of the Council of Europe (CoE) approved and issued the Recommendation (2010)11 on *"The impact of genetics on the organization of health care services and training of health professionals"* to the 47 constituent member states – <https://wcd.coe.int/wcd/ViewDoc.jsp?Ref=CM/Rec%282010%2911>

The major aim of this document is to make European governments aware of the impact of the increasing knowledge in genetics and of the immediate, as well as future, benefits such progress may have on the health care in general. In order to achieve the full potential of genetics, structural and financial support should be rendered to genetic services and adequate education of professionals and of the general public in the field should be fostered. In short the CoE recommendations stipulated:

- i. to adopt policies, legislative and other measures necessary for developing a coherent and comprehensive national policy framework for genetic services;
- ii. to develop and, where appropriate, strengthen genetic services to maximize the benefits of genetic applications in health care for all patients;
- iii. to make available adequate genetic counseling in an equitable manner, whenever needed;
- iv. to be aware, while aiming at improving health by genetic applications, that the rights of patients and their families, as well as the ethical principles of human dignity and integrity, have to be respected and that social exclusion, discrimination and stigmatization have to be prevented;
- v. to ensure that consideration is given to the education of health care professionals and the public in genetics, in particular about the implications of genetic knowledge and its medical applications;
- vi. to promote research in the area of genetics and its applications within health care, including its public health implications;
- vii. to promote international networking of genetic service providers, relevant research organizations, and of other health care agencies that are active in the field, in order to share knowledge and facilitate the provision of special tests where appropriate;

- viii. to facilitate the availability of information on genetic services to citizens, patients and their families and professionals in formats and languages which they can understand;
- ix. to support the widest possible dissemination of the Recommendation and its explanatory Memorandum;
- x. to develop and adopt the measures set out in the Appendix to this Recommendation, bearing in mind respective national circumstances.

These recommendations are the result of long lasting thorough discussions that involved multiple experts from different countries, the Health Commission as well as the Ethical Commission of the CoE. They are based on written documents amended by the representatives of the member states and finally edited by the Health Commission. The main impetus for the development of these recommendations has been the rapidly expanding knowledge in human genetics as well as advances in genetic testing technologies, making respective health care applications increasingly amenable.

The fact that approximately 5% of the general population is expected to develop a genetic- or partly genetic condition (be it monogenic-, multi-factorial-, chromosomal-, mitochondrial- or imprinting-related pathology) by the age of 25 years implies that close to 30 million people in Europe suffer from a genetic disease leading to high costs for health care. Thus, genetic diseases pose a substantial challenge which governments have to properly address and that cannot be ignored.



Rapid developments in the field were also reflected by the European Society of Human Genetics (www.eshg.org) which has already issued through its PPC committee multiple guidelines on various aspects of human genetics, including guidelines on genetic services (EJHG,2003,11,suppl.2.). In addition, the ESHG has been closely collaborating with Eurogentest (a European funded Network of Excellence) that has recently addressed harmonization and improving the quality of genetic testing in its broadest sense at the European-wide level (<http://www.eurogentest.org>)

For policy matters, European countries should take into account also the Convention on Human Rights and Biomedicine (conventions.coe.int/Treaty/Commun/QueVoulezVous.asp?NT=164&CL=ENG) and its Additional protocol concerning genetic testing (www.bmj.bund.de/files/-/3146/Additional%20Protocol%20to%20the%20Convention%20on%20Human%20Rights%20and%20Biomedicine.pdf) for health purposes as well as Recommendation (1990)13 on prenatal screening, prenatal diagnosis and genetic counseling (www1.umn.edu/humanrts/instree/coerecr90-13.html) and Recommendation (92)3 on genetic testing and screening for health care purposes (www1.umn.edu/humanrts/

[instree/coerecr92-3.html](http://www1.umn.edu/humanrts/instree/coerecr92-3.html)) and Recommendation(97)5 on the protection of medical data (www1.umn.edu/humanrts/instree/coerecr97-5.html), which were basis for the current CoE document.

Members of the Committee were:

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Invitation to the

Annual Membership Meeting 2011

At the EUROPEAN HUMAN GENETICS CONFERENCE 2011

Sunday, May 29, 2011 at 7.00 – 8.00 p.m.

Room G 102-103

Amsterdam RAI, Europaplein 22, 1078 GZ Amsterdam, The Netherlands

AGENDA

Opening by the President of the Society, Professor Milan Macek Jr.

1. Activity of the Society 2010-2011
2. Financial Report of the Society 2010
3. Discharge of the Board Members for the year 2010-2011

Opening by the new President of the Society, Professor Jörg Schmidtke

4. Results of election for President-Elect
5. Results of election for Board Members
6. Membership fees 2012
7. Site of future European Human Genetics Conferences
8. Budget proposal 2012
9. Major policy questions proposed by Board
10. Future activities

Please find the minutes of the last membership meeting in Vienna 2010 in the restricted area:

<https://www.eshg.org/39.0.html>