

Report from the President

by Jörg Schmidtke

Dear colleagues,
dear guests,

Welcome to the 2012 annual conference of the European Society of Human Genetics and the 2012 European Meeting on Psychosocial Aspects of Genetics here in Nürnberg! This is the 43rd conference of our flourishing society, and the 5th time that we meet in Germany. As before, the German Society of Human Genetics has its annual meeting together with us. Our thanks thus go to *André Reis*, Chairman of the German Society of Human Genetics, and his team as our local hosts. We are looking forward to a comprehensive overview of recent developments in our fast-moving area of basic and applied research and health care services, including genetic testing and genetic counseling.

Nürnberg is a place deeply connected with the recent history of human genetics, with its misuse, but also with attempts at coming to terms with the past. This is the city where in 1935 the racial laws were set up, prohibiting relationships between "Germans" and "Jews", where after the war the International Military Tribunal tried the Nazi leaders and doctors, and where the foundations were laid for the principles of research ethics in human medicine which became cornerstones of the Geneva and Helsinki declarations in later years. A satellite meeting of our conference, the 5th International Workshop on the History of Human Genetics, organised by *Heike Petermann*, is addressing these issues.

A big conference like this requires the commitment of many highly motivated colleagues. First and foremost it is the Scientific Programme Committee, chaired by *Brunhilde Wirth*, to whom we are indebted for the scientific programme. I acknowledge with thanks the organisational contributions of our executive officer *Jerome del Picchia* and his team at



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the Vienna Medical Academy, *Rose International*, in charge of the Industrial Exhibition, the ESHG Board and all members of the Executive Board for their hard work.

According to its statutes, the European Society of Human Genetics, ESHG, has three major aims: "to promote research in basic and applied human and medical genetics, to ensure high standards in clinical practice and to facilitate contacts between all persons who share these aims, particularly those working in Europe". In the past, these aims have not received completely equal attention. In most of its 45 years' history, the society's emphasis was clearly on promoting basic and applied research and on providing a platform for scientific contacts by organising highly attractive annual meetings. However, a very active Public and Professional Policy Committee, PPPC, was established in 1997. First chaired by *Marcus Pembrey* and then by *Ségolène Aymé*, for several years now it has been under the leadership of *Martina Cornel*. The PPPC has developed general recommendations for a number of practical issues. These have included genetic screening, biobanking, insurance and workplace issues, as well as principal aspects of service provision. Nevertheless, the ESHG was for a long time not directly involved in the organisation of genetic patient care, establishment and maintenance of laboratory quality, or setting standards for the qualification of genetic staff. This restraint must be attributed to the fact that individual health care in Europe is largely the responsibility of the individual EU member states. However, with the completion of the Human Genome Project and the emergence of new powerful analytic technologies, the diagnosis of most if not all rare genetic diseases is becoming a reality. Genetic counseling for these conditions is acquiring a much improved data base, and treatments are being developed as a result of a better understanding of aetiology and patho-

genesis. Dealing with rare genetic diseases is now becoming recognised as a practical public health need, an area which is indeed seen, under the European Community Treaty, in particular Article 152, as a major focus of EU action.

While they are individually rare, due to their large number, these conditions constitute an enormous public health problem. It is estimated that some 5% of the EU population are directly affected by one or another of about 7,000 different rare genetic diseases. Moreover, the proportion of the population at increased risk of developing one of these conditions in later life or in the next generation is several-fold higher. Organising healthcare for rare diseases is a huge challenge at both member state and European level. Knowledge on any one of these diseases may exist in only a few centres in the whole EU area. For patients and health professionals alike, it is a challenge to identify the centre that would optimally respond to a particular need. The ESHG, through its members and committees, is supporting and collaborating with EU actions, including the EU Committee of Experts on Rare Diseases (EUCERD-Committee, appointed on the basis of the EU Recommendation on action in the field of rare diseases), the rare disease platform Orphanet, EURORDIS, EPPOSI, and Eurogentest (now as a Coordinated Action, EuGT2, committed to laboratory and clinical quality and harmonisation in genetic services). It will be important to find a way of incorporating EuGT2 actions into the ESHG when EuGT2 funding is terminated in 2013. One of the very successful EuGT2 projects, the "Clinical Utility Gene Cards" is already co-funded by the ESHG, and forms a section on its own in our Journal, the European Journal of Human Genetics.

A major step forward in strengthening the European dimension of genetic services has been the EU-wide recognition (also including Switzerland, Norway, and Iceland) of clinical genetics as a medical specialty, as of March 2011. We are deeply indebted to *Milan Macek Jr.*, Vice-President of the ESHG, and *Ulf Kristoffersson*, then chair of the Multidisciplinary Joint Committee Clinical Genetics of UEMS, the European Union of Medical Specialists, for their many years of hard work to accomplish this goal.

Our next aim is to achieve European-level recognition, certification and/or accreditation for the two other constituent groups of our society: genetic counsellors/nurses and laboratory geneticists. Two very active ad-hoc committees, the Ad-hoc Committee for Certification of Genetic Nurses/Counsellors (chaired by *Heather Skirton* and *Marie-Antoinette Voelckel*) and the Ad-hoc Committee for the Certifi-

cation of Laboratory Geneticists (chaired by *Thomas Liehr* and *Egbert Bakker*) have gone deeply into developing core curricula as a basis for European-level recognition. They have also contributed, by responding on behalf of ESHG, to a Consultation Paper by DG Internal Market and Services on a revision of the EU Professional Qualifications Directive. To carry these activities a step further a Working Group has been set up to work towards implementing a European Board of Medical Genetics. We will continue to work in close consultation with the presidents of the National Human/Clinical/Medical Societies in Europe.

The Genetic Services Quality Committee, chaired by *Ros Hastings*, is committed to identify and bridge quality gaps in genetic services, harmonise genetic diagnostic approaches, and develop recommendations. It currently complements and interacts with the two named ad-hoc committees and EuGT2. One of the tasks of the Working Group on implementing a European Board of Medical Genetics is to develop suggestions as to how the activities of all these contributors could be restructured in the most efficient way.

Thanks are due to the ESHG Education Committee, chaired by *Tayfun Oczelik* and *Peter Farndon*, for their involvement in organising the DNA Day contest and liaising with the European Genetics Foundation (EGF), which the ESHG continues to support by granting fellowships to participants, and to the Annual Meetings Committee, chaired by *Andrew Read*, being in charge of identifying potential venues for our future conferences many years ahead. Next time we will meet in Paris!

We further wish to thank the Chief Editor of our Journal, the European Journal of Human Genetics, *Gert-Jan van Ommen* and his team of section editors for their hard work, maintaining this Journal in a high-ranking position in genetics.

Founded in 1967, the ESHG is approaching its 50th birthday rather soon. The Executive Board realised that it was high time to become more conscious of its own history, and we are happy to announce that *Peter Harper* has agreed to start documenting our history. All members are cordially invited to contribute through archived materials and sharing their memories.

It was a great honour for me to have served the Society as its President and I am now looking forward to serving the Society as a member of the Executive Board in the years to come. I am passing my office on to *Stan Lyonnet*, and I wish him a very successful presidency.

Report from the Secretary General

by *Gunnar Houge*

It has been a good year for ESHG – despite the euro/debt-crisis and associated cut-backs that directly or indirectly have affected some of our members. Maybe this reflects the increasing importance of genetics for medical diagnostics, patient counseling and treatment. In addition, the new mass parallel sequencing techniques have put us in the middle of a new-disease-gene bonanza.



Last year's annual meeting in Amsterdam drew 2416 participants – a new record. This result was obtained thanks to our dedicated Scientific Planning Committee (SPC) lead by *Brunhilde Wirth*, the local Dutch host *Robert Hofstra*, the experienced staff of Vienna Medical Academy lead by *Mirjam Uebelhör* and our Executive Officer *Jerome del Picchia*, and our commercial manager, *Jantie de Roos* from Rose International. Also our journal, the *European Journal of Human Genetics*, is doing very well with an increase in impact (now 4.380) as well as revenues. This good result is very much thanks to the long-time dedication of our editor, *GertJan van Ommen*. Other ESHG core activities are committee work and sponsoring of various meetings, usually but not only in the form of fellowships. *European Genetics Foundation (EGF)* lead by *Giovanni Romeo* and his team has also last year been the largest recipient of fellowship support, € 25 000 in all. Our currently most active permanent committee is probably the *Public and Professional Policy Committee (PPPC)*, chaired by *Martina Cornel*, but also the *Genetic Services Quality Committee (GSQC)*, lead by *Ros Hastings*, has done a lot of work. Another important committee, the *Education Committee (EC)*, lead by *Tayfun Oczelik* and *Peter Farndon*, is arranging the *DNA day* in collaboration with *Celia DeLozier* and *ASHG* (www.dnaday.eu), with a record high number of contributions this year. The EC is also dealing with all types of educational issues, the only exception being educational sessions of the annual meeting.

The Executive Committee of the Board, usually known as the Exec, is in charge of the “daily life” of the society, and we would not have managed without the help of *Jerome del Picchia*, our EO (executive officer). The Exec has three “political” members; the coming, sitting and leaving presidents, and three “adminis-

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trative” members; the treasurer (*Andrew Read*), the deputy general secretary (*Helena Kääriäinen*) and myself. Because ESHG has grown substantially in size and activity the last years, it might be fruitful to discuss how Exec and Board structure can be optimized to reflect the interests of the members. This will be a theme for this year's Board meeting and probably also the Membership meeting, where all members are most welcome!

Gunnar Houge
ESHG General Secretary

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 Nuremberg, which is a combined meeting together with the European Meeting on Psychosocial Aspects of Genetics this year. The ESHG meeting became the second largest human genetics meeting in the world, with over 2000 scientific participants attending each year. Based on a very attractive program I hope that this year the meeting will be a highlight again, but also that you will find sufficient opportunities to meet old collaborators or find new ones, have fruitful discussions and enjoy the meeting overall.

Activities of the Scientific Programme Committee

The Scientific Programme Committee (SPC) for 2011-2012 was composed of sixteen regular SPC members: *Brunhilde Wirth* (chair, D), *Corinne Antignac* (F), *Paul de Bakker* (NL), *Jeffrey Barrett* (GB), *Alexis Brice* (F), *The-Hung Bui* (S), *David FitzPatrick* (GB), *Giovanni Neri* (I), *Francesc Palau* (E), *Anita Rauch* (CH), *Peter Robinson* (D), *Carla Oliveria* (P), *Michael Speicher* (A), *Draga Toncheva* (BG), *Joris Veltman* (NL) and *Mikka Vikkula* (B), four local SPC members from Germany: *Jürgen Kohlhase*, *Kerstin Kutsche*, *Markus Nöthen* and *André Reis* and two observers of the ESHG board *Martina Cornel* (NL) and *Gunnar Houge* (N).

The SPC met twice to organize the Nuremberg 2012 ESHG conference: in July 2011 to decide on the ple-

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nary sessions and symposia in Nuremberg and in Vienna at the VMA offices in March 2012 to select the abstracts for oral presentations and posters.

The number of submitted abstracts was 2001. All abstracts have been on-line scored by 4-12 evaluators including SPC members, ESHG board members and for the first time we included 82 external reviewers, who have been proposed by the SPC members as experts in the various topics. These new system allowed us a very solid and highly accurate evaluation. Based on topics and scores, 114 abstracts were selected for the 18 concurrent sessions and one plenary session (best abstracts). Among the oral presenters, 51 were Young Investigator Candidates (at least 1 in every session), reflecting the high level of contribution of young scientists to this program.

Of the remaining abstracts, 62 were selected as best posters and will be marked with an ESHG Rosette. Of these, 35 were Young Investigator Candidates and are qualified for the poster prize. 189 abstracts with a score <4.5 will be "published only" and 72 abstracts were rejections either due to bad quality (score < 3.0) or multiple submissions of the same first author.

The SPC felt that the actual categories/topics do no longer properly reflect our scientific spectrum in

human genetics. Therefore, please be aware of the novel division of topics and subtopics which will be introduced with next year ESHG meeting in Paris (available on the ESHG website).

After the Nuremberg conference, the SPC shall have to say goodbye to *Michael Speicher*, *Draga Toncheva*, and *Miikka Vikkula*. We thank them for their work and their dedication to making the meeting better.

2012 Meeting Highlights

The meeting will start with the first plenary session including three local stars: *Stefan Mundlos*, *Peter Lichter* and *Herbert Schunkert*. The "What's new?" session will follow with the most exciting new findings selected from submitted abstracts.

The meeting will include 16 concurrent symposia which will address topics of new insights and challenges from next generation sequencing in gene discovery, new mechanism underlying human disease, functional studies and underlying pathomechanism of various human disease groups, cancer genetics, clinical and social implications of genomics and the implementation of the new technologies in genetic testing, modeling in statistical genetics and analysis of complex datasets, challenges in clinical genetics and genetic counselling.

The "educational sessions" throughout the program were very well attended in the past years, so that the committee decided to continue with 8 educational symposia this year. These include: Complex Diseas-



es, Skin diseases, Applying family dynamics/therapy in genetic counselling (joint with EMPAG), Array CGH and next generation sequencing, Neuromuscular diseases, Next generation sequencing goes diagnostics: First experiences, Trinucleotide disorders, and How to get published in ESHG?

On Tuesday, a third plenary session on "Targeted pharmacological therapies in genetic disorder" with *Harry Dietz* (USA), *Petrus de Vries* (UK) and *Sebastien Jacquemont* (CH) will present us new therapeutic developments in Marfan syndrome, Tuberous sclerosis and Fragile X-syndrome.

The meeting will conclude with our distinguished speaker of the Mendel lecture *Evan Eichler* from Seattle who will discuss the role of genome structural variation from both an evolutionary and disease perspective, he will show how this architecture predisposes our species to recurrent copy number variants associated with developmental delay, epilepsy and autism. This year, the ESHG award 2012 will be awarded to *Peter Lichter* from Heidelberg, Germany, in recognition of his groundbreaking work in the development of molecular cytogenetics and new mechanistic findings of tumorigenesis.

I hope that all of you will enjoy the meeting in Nuremberg

Prof. Dr. Brunhilde Wirth

2012 Scientific Committee Program Chair
Institute of Human Genetics
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Report from the ESHG Ad Hoc Genetic Nurse and Counsellor Accreditation Committee

by *Heather Skirton & Marie Antoinette Voeckel*

The rationale for the formation of the Ad Hoc Genetic Nurse and Counsellor Accreditation Committee was to develop a coherent system for education and practice of genetic nurses and counsellors in Europe.

Prior to 2012, the Committee had done considerable work on development of standards of education and Code of Professional Practice for genetic counsellors in Europe (*Skirton et al*, 2010). That work was previously presented to the ESHG Board in 2010 and 2011. At the previous full Board meeting in Amsterdam, 2011, a Working Group for the proposed European Board of Medical Genetics was formed and the Committee was asked to work towards development of a European curriculum for genetic counselors and to provide a basis for a European registration system for genetic counsellors and genetic nurses.

In some countries it will be more appropriate and acceptable to utilise genetic nurses, in others genetic counsellors will be preferred. It was therefore essential to design a system that accommodates both without undue bias towards one professional group.

The Committee has undertaken three studies to inform the work. The first was a survey of genetic nurse/counselor numbers, education and legal standing in Europe. The data were published at the 2011 ESHG conference and in the *Journal of Community Genet-*



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ics (Cordier et al, 2011). In a second study, we collected data on the actual clinical practice of 216 genetic nurses or counsellors from 19 countries. Those data are the subject of a presentation at the ESHG/EMPAG conference this year (Skirton et al, 2012). And they provide a picture of the way in which these professionals are working in multi-disciplinary teams in Europe. The majority of respondents stated that they alone or with a medical colleague took responsibility for making the first contact with the family (87.9%) drawing the pedigree (85.2%), explaining a genetic test to the patient (79.5%) and providing psychological support through the testing process. Over 81% managed some cases without the input of a medical doctor.

The Committee have also used a Delphi study (using professional leaders in each country) to determine the curriculum for the Master level education of genetic counsellors and genetic nurses, based on the core competences previously agreed (Skirton et al, 2010). This curriculum was submitted to the ESHG Board for consideration in March, 2012. We used the same technique to devise a European registration system for both genetic counsellors and genetic nurses which we believe will enable practitioners to be assessed in terms of the relevant competences needed to perform the work safely. This involves an examination, case log verified by seniors colleagues, references from senior colleagues, and case studies. The final recommendations were refined at a workshop of key professional leaders in Taunton, UK in February 2012 and we are very appreciative of the support we received from the ESHG for this workshop.

The Committee has kept the genetic nurse/counsellor community in Europe well-informed of their activities through monthly email newsletters to over 200 members of our professional network and will hold a meeting for all interested colleagues during the ESHG/EMPAG meeting in Nuremberg. We look forward to discussion of the recommendations in a wider arena and further developments in our profession.

Finally, we thank all our colleagues who have served on the Committee, those who contributed by completing surveys or attending workshops and the ESHG Board for the enormous encouragement we have received to undertake this work.

Heather Skirton and Marie-Antoinette Voelckel are the Co-Chairs, ESHG Ad Hoc Genetic Nurse and Counsellor Accreditation Committee

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Skirton H, Voelckel MA, Patch C. (2010) Using a community of practice to develop standards of practice and education for genetic counsellors in Europe. *Journal of Community Genetics* 1: 169-173.

Report from the ESHG Public and Professional Policy Committee

by *Martina Cornel*

In the year 2011-2012 the Public and Professional Policy Committee (PPPC) of the ESHG continued working on **whole genome sequencing**. A publication in the EJHG (2012; advance online publication 28 March 2012; doi: 10.1038/ejhg.2012.56) on the changing landscape of genetic testing addressed the impact of techniques examining the whole genome, such as microarrays and next generation sequencing, on clinical and laboratory services and research in Europe. The paper was based on a series of workshops organized by PPPC and the Genetics Services Quality Committee at the 2010 ESHG meeting in Gothenburg. It covered challenges in handling massive amounts of data leading to difficulties in establishing clinical utility, blurring boundaries between healthcare and research, impact on genetic services and potential use in primary care, as well as ethical aspects related to for instance, incidental findings, recontacting and consent. Following these workshops and further discussion with experts, PPPC drafted Recommendations on whole genome sequencing and analysis that will be posted on the web for membership consultation.

Via PPPC, the ESHG Board endorsed an expert opinion document on **Newborn screening in Europe** [www.iss.it/binary/cnmr/cont/Expert_opinion_document_on_NBS_20120108_FINAL.pdf], prepared by *Martina Cornel, Tessel Rigter, Stephanie Weinreich, Peter Burgard, Georg F. Hoffmann, Martin Lindner, J. Gerard Loeber, Kathrin Rupp, Domenica Taruscio and Luciano Vittozzi*. This document aims to establish a joint framework for assessing technological options, weighing pros and cons, quality assessment, implementation and evaluation strategies while acknowledging local priorities and constraints.

In addition, after membership consultation PPPC helped preparing a publication on **paediatric biobanking**. It offers points to consider for researchers, clinicians and

policymakers when storing samples from minors for research. Though opinions vary in this fast evolving field, general principles, for instance concerning the increasing capability of the minor with age, may guide establishing ethically sound research facilities and responsible governance.

Several other topics were discussed during the year 2011-2012, that will be followed up later, among other things in a workshop to be organised at the ESHG meeting in Paris in 2013: genetic testing and forensics, and the new theme of prioritization in genetic testing.

Members of the PPPC in 2011-2012

Pascal Borry, Anne Cambon-Thomsen, Martina Cornel (Chair), Francesca Forzano, Shirley Hodgson, Hülya Kayserili, Christine Patch, Borut Peterlin, Wolf Rogowski, Jorge Sequeiros, Maria Soller, Aad Tibben and Lisbeth Tranebjaerg, supported by Carla van El, Heidi Howard and Florence Fellmann were attending the meetings as observers.



Report from the Genetic Services Quality Committee

by Ros Hastings

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 relating

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to quality in genetic testing;

- Give recommendations for those countries where no guidance is currently available.

Six 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
- Clinical Genetics
- 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. A summary of the workshop findings from the 2010 ESHG Satellite Symposium in Gothenburg on 'Changing Landscape of Genetic Testing' was published in the European Journal of Human Genetics in March 2012. In addition, following the 2011 ESHG Satellite Symposium in Amsterdam on 'Practical issues with Microarrays', five papers based on the workshop discussions have been published in Human Mutation. The links to these six papers will be made available on the ESHG website.

The GSQC has prepared some draft Reporting Guidelines applicable for all diagnostic genetic laboratories and these are due to be published soon.



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The following Guidelines have also been prepared by other collaborating groups. These are available (or soon to be available) for:

Cytogenetics: Generic Guidelines; Constitutional and Acquired Guidelines are due soon;

Molecular Genetics: HGVS nomenclature in CF (published in Human Mutation); Osteogenesis Imperfecta and Huntington's Disease are due soon.

The GSQC supports the quality management activities within EuroGentest2. 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. The GSQC recommends that all diagnostic laboratories participate annually in EQA for all aspects of their diagnostic service - whether or not they are accredited. The GSQC recognises there is a need for training workshops to assist laboratories with international nomenclature and interpretation of results. The possibility of a workshop is currently being discussed within EuroGentest2 and the GSQC.

The GSQC provides a governance structure for the four European EQA schemes [CEQA - Cytogenetics, CF Network - Molecular Genetics, EMQN - Molecular Genetics and ERNDIM - Biochemical Genetics] and reviews their annual management reports. The GSQC has formed a working group (in collaboration with Eurogentest 2) to discuss unsatisfactory performance identified by the EQA schemes in genetic testing.

The GSQC has prepared an online survey on Quality Assessment in Genetic Counselling. The aim of the survey is to explore the needs for a European quality assessment scheme for Genetic Counselling. The survey includes questions on whether there are any National QA Schemes for genetic counselling, the nature of these schemes if they exist, whether there is perceived need for European Genetic Counselling EQA and whether Clinical Geneticists are willing to participate in a pilot study. National Genetic Societies will be notified when this survey is available online. Following completion of the survey online, it is hoped that there will be a half day Symposium prior

to the Paris ESHG 2013 Conference to feedback the survey results and discuss a way forward.

Quality is not an act but a habit. Help us to help you by letting us know of any quality issues that need addressing. Please submit any suggestions to the Chair of the Quality Committee.

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

Report from the ad-hoc committee for Laboratory Genetics

by *Thomas Liehr and Gunnar Houge*

There is an increasing need for recognition of also a laboratory speciality in clinical genetics, in line with last year's recognition of clinical/medical genetics as an EU recognized speciality under the UEMS umbrella. As some of you may recall, the process of achieving such a clinical speciality was long and cumbersome, mainly because approval had to

be given by an EU recognition committee that required almost unanimous agreement between all 27 EU countries. To ease the bureaucracy in such certification processes, the EU has proposed to establish a professional qualifications directive scheme. Hopefully this proposal, where also ESHG has given substantial input to the process, will be passed within the year.

In order to have a laboratory genetics curriculum ready if such a proposal is passed, the ESHG ad hoc committee has been quite active the last year. A proposal for core curriculum has now been finalized, **called Clinical Laboratory Genetics as an EU-recognized specialist profession**, and it can be found on our web-page under "About ESHG/Committees" (see www.eshg.org). The curriculum has been approved by the ESHG Execu



tive Committee and awaits Board approval in Nürnberg. The curriculum has so far received support from 14 country representatives, representing 204/345 (59%) of the EU weighted (qualified majority) votes. If Board approves, the committee work will probably be followed up by the new European Board of Medical Genetics (EBMG), provided that the Board approves establishment of this new body under the ESHG umbrella.

Special Report: Medical Genetics in Belgium

by *Thomy JL de Ravel*

The practice of medical genetics is limited to eight centres in Belgium. Although genetics laboratories and clinical practices had been initiated in various centres in the mid-1960's, it was only in 1973 that the High Council for Human Genetics was founded to advise the Minister of Public Health and Family in matters related to medical genetics (Royal decree 7 November 1973). The functioning and recognition of the genetics centres were defined by the Royal decrees of 14 December 1987, 14 December 1988 and 21 April 1989. These laws facilitated further development of the centres and restricted the re-imbursment of genetics consultations and analyses to these recognized, university affiliated institutions. In October 2007, The Belgian Health Care Knowledge Centre, a body supervised by the Minister of Public Health and Social Affairs, recommended changes in the organization and financing of genetic services in Belgium (Report 65C; <https://kce.fgov.be/>). Re-imbursment will soon occur in a stratified manner (simple to complex) for both consultations and genetics laboratory analyses – the legal framework for implementation will most likely soon be put in place by the government.

The population of the federal state of Belgium is almost 11 million, of which 1 million are non-Belgian citizens (and many more of non-Belgian descent, http://www.belgium/en/about_belgium/country/Population/), thus offering diverse challenges in the field of medical genetics. As there is universal social security and mandatory health care and indemnity insurance, the whole population has access to genetics services (http://www.belgium/en/health/healthcare_costs/). In total, the federal government spends nearly 40 million euros per year on genetic testing, which corresponds to less than 4 euros per inhabitant per year. The genetic counselling and paramedical support are further subsidized by the regional governments. This combined funding, together with research funding, has resulted in inte-

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grated clinical and research medical genetics centres. Interaction and participation at European and International level has further allowed excellence in various domains.

The Belgian Health Care Knowledge Centre reported that in 2005, a total of 621 full-time staff was employed across all sectors of the 8 centres. The clinical geneticists offer counselling services not only in their centres but also at regional hospitals, institutions and day centres for the disabled, schools for specialized education and for children with special needs (over 21 000 counselling sessions in the year 2005). The genetics laboratories receive samples for analysis from throughout the country – the number of tests carried out by the centres in 2005 totalled over 200 000, of which about 123 000 were paid by the medical insurance (Report 65C).

High Council for Human Genetics

Representatives from the eight Centres forming this Council meet regularly to fulfil their mandate of advising the Minister of Public Health and Social Affairs on all medical genetics issues. As decreed in 1973, this encompasses the teaching of human genetics, the prevention of hereditary disorders, the accreditation requirements, the registration of hereditary disorders. Its role also extends to the development of clinical practice norms and matters relating to the re-imbursment for genetic services, this being done in collaboration with the BeSHG and the national medical insurance body.

The Belgian Society of Human Genetics (BeSHG)

This Society was established in 2004 and revised its statutes as published in the Belgisch Staatsblad/



The author with the patient during a genetic consultation with the family.

Moniteur Belge on 22 March 2006 (www.beshg.be). Its aims include the support and promotion of scientific research in human genetics, interchange between scientists and clinicians both within and beyond Belgium, the organisation of scientific meetings, ongoing education of its members, updating of the public, consideration of implications of new genetic technologies on the individual and society as a whole, and the formulation of a code of good clinical practice in applying advances in genetics to the individual. The members of the Board of this society are elected by its members and usually comprise representatives from all 8 genetic centres. The Board interacts closely with the High Council and, via its Board members, with all 8 genetic centres. Yearly scientific meetings, hosted by one of the 8 centres on a rotation basis, are organised for its members and the scientific community at large. This is well supported as reflected by an attendance of over 300 participants.

The BeSHG organizes an inter-university certificate course in human genetics. This credential is compulsory for all doctoral students (PhD) from the genetics centres, for recognition as a medical genetics laboratory supervisor, and is also proposed as a



Professor *Jan Duman*, retired Head of the Centre for Medical Genetics, Antwerp, next to em. Professor *Jean-Jacques Cassiman* of the Department of Human Genetics, Leuven whilst attending a course in 1990 in Leuven.

requirement for medical/ clinical genetics specialist recognition. This course is open to members and non-members of the BeSHG.

In order to address the various facets of the field, the BeSHG has a number of 'Work Groups', the members of which meet between 4 and 7 times a year. The senior scientists from the constitutional cytogenetics laboratories form BelCoCyt, those from the haematology/ oncology laboratories form BCG-HO, and those from the molecular genetics laboratories form BelMolGen. They address, amongst many other matters, the technical developments, the accreditation issues, the resolution of difficult cases and the work distribution in the country with the objective of good collaboration and harmonisation. A leading role was played by members of the BeSHG in opposing the patent on the breast cancer gene. Similarly, the laboratory technicians have recently started a forum to discuss common issues.

The Dysmorphology Club regroups the clinicians for 8 half-day meetings a year, whilst the newly-formed Good Clinical Practice Work Group has started to review and recommend best practices for various disorders, which are then passed on to the High Council for further consideration and application as local resources permit.

Medical Genetics is not yet recognised as a medical speciality in Belgium. The Work Group addressing this void has, in collaboration with the High Council for Human Genetics, submitted the necessary draft statutes to the High Council for Medical Specialities for consideration and implementation, in accordance with European Union Regulation 213/2011 of 3 March 2011.

In parallel with the European Society of Human Genetics ad hoc committee for the recognition of Medical Genetics Laboratory Scientists, a local ad hoc Work Group has formulated the statutes, leading to these individuals being recognised by a Recognition Committee of the BeSHG.

The BeSHG has initiated discussions regarding the grouping and recognition of genetic/ nurse counsellors in Belgium as they are presently not reflected in the organisation of genetic services.

The Belgian Plan for Rare Diseases

Following the European Council recommendation of 8 June 2009 on action in the field of rare diseases (2009/C 151/02), whereby its member states were advised to formulate a clear, comprehensive and integrated plan for health and social care of patients with rare diseases, the Fund for Rare Diseases and Orphan Drugs managed by the King Baudouin Foundation obtained the necessary support from the Belgian Minister of Public Health and Social Affairs for the development of "The Belgian Plan" (<http://www.kbs-frb.be/>). This committee presented in 2011 its 42 recommendations and proposed measures in order to address the needs of an additional 18000 pa-

tients with rare diseases in multidisciplinary expert centres. State budget restrictions at present make implementation very limited.

The Superior Health Council of the Ministry has also been coordinating a Working Group investigating various facets of "Direct to Consumer" genetic testing services.

The Patient Organizations

There are in Belgium about 150 organizations grouping families with rare and not so rare disorders and diseases. The Rare Diseases Organization, RaDiOrg, an umbrella organization of 90 of these, has a strong lobby presence (<http://www.radiorg.be/>).

Conclusions

The medical genetics community is small and in proportion to the size of the country. This has the advantage of facilitating regular interaction between members. However, official recognition and support remain key issues in this dynamic field.

Thomy JL de Ravel,
Chairman, The Belgian Society of Human Genetics

ESHG NEWSLETTER

The ESHG Newsletter got the opportunity to publish the summary of an ESHRE-ESHG meeting which was held on March 5-6/2012 in Brussels under the auspices of both ESHRE.com and ESHG.org. This article was originally published in the May issue of the ESHRE Focus on Reproduction newsletter

How to keep a relationship alive: The 2nd ESHRE-ESHG Expert Meeting confirms the warm feelings between reproduction and genetics

In 2004, the Public and Professional Policy Committee (PPPC) of the European Society of Human Genetics (ESHG) found it necessary to create professional recommendations on how to use IVF techniques safely and reliably from the genetic point of view. It also held important to issue guidelines on acceptable (genetic) goals of IVF treatment and on how these expensive treatments should be prioritized in the European health care systems.

ESHG teamed up with ESHRE, and together a large expert group issued from both societies, produced a paper (*Soini et al., 2006*) describing in extenso the common grounds between reproduction and genetics as well as a set of recommendations published as a policy paper in the European Journal for Human Genetics in 2006 (see below). Several meetings were necessary to produce these all-encompassing papers, the most memorable of which was at the site of the then European Commission's Joint Research Center in Seville, Spain.

Recently, *Milan Macek, Joep Geraedts* and *Joyce Harper* took the initiative to renew the expert meeting and report. The meeting was held last March in Brussels: a number of faces were familiar, but many new experts were invited demonstrating the quick pace of developments both in genetics and reproduction. While going over the first report, all present found that many points raised back in 2005 were still valid seven years later. The core aspects linking genetic testing of some description, such as genetic diagnosis of male and female infertility, PGD



Em. Professor Inge Liebaers, previous Head of the Centre for Medical Genetics, Free University of Brussels (VUB) together with Em. Professor Christine Verellen-Dumoulin, Previous Head of the Centre for Human Genetics, Institute de Pathologie et Génétique, Gosselies.

Think of...

...renewing your ESHG membership!

Visit the ESHG booth (#240) in the exhibition.

and PGS (including sex selection for social reasons), genetic testing in gamete donors and reproductive and genetic counselling, to ART are still prominently present. A more future-oriented topic is the detection of any adverse effects of ART (just think imprinting defects), and the quality and safety procedures that need to be taken to heart to prevent these. At legal and political levels, the regulation of embryo research and how this can differ in different EU member states as well as issues of public health care (including cross-border care which at that moment was emerging but is now in full bloom) were broadly discussed. The psychological issues pertaining to the way parents, and their future children born after ART, perceive their treatment was discussed separately. The future perspectives closed the paper, and it must be said that many of the predictions did come true: to name one, the development of the stem cell field was mentioned in the future perspectives and was a fully discussed topic in the follow-up meeting. No specific ethical paragraph was included although eminent ethicists in the field were involved, but ethics pervaded every sentence and every statement of the document.

During the recent expert meeting, *Luca Gianaroli* kicked off by sketching the latest developments in the field of PGD and PGS. The timing of biopsy has clearly moved away from the cleavage stage in favour of polar body biopsy, especially in countries where legally this is the only option, or blastocyst biopsy. These developments would not have been possible without the greatly improved freezing protocols that

give geneticists more time for diagnosis, but have also engendered consequences such as monozygotic twinning after the biopsy of blastocysts. This was also touched upon by *Joep Geraedts*.

The next session was devoted to stem cell research, a new kid on the block as this group is concerned. *Anna Veiga* showed an overview of the latest breakthroughs in the differentiation of human pluripotent cells (either embryo-derived human embryonic stem cells (hESC) or induced pluripotent cells, iPSC) into male and female gametes, offering a new hope for patients not able to produce their own gametes. *Karen Sermon* presented how hESC derived from embryos shown to be affected after PGD or iPSC derived from patients can be used as disease models. To close the stem cell presentations, *Claudia Spits* added a cautionary note to the clinical use of hESC or iPSC because of the frequent chromosomal – and possibly other genetic abnormalities – these cells develop in culture.

The next session was devoted to PGD and PGS. The most notable change in PGD and PGS since the last expert meeting, apart from the moment of biopsy, is of course the introduction of microarray technology allowing a comprehensive analysis of a blastomere's karyotype. This not only gives us a much broader view of what goes on in an embryo's genome than FISH ever could, but it has also unveiled new abnormalities such as segmental deletions, as illustrated by the examples shown by *Joris Vermeesch*. Needless to say, all these new developments warrant a frequent update of practice guidelines. The ESHRE PGD Consortium is working very hard to get this done.

Looking from a more molecular point of view, *Stéphane Viville* reviewed the literature on imprinting defects after ART, after an amusing diversion to *Heterocephalus glaber* or naked mole rat, the ugliest animal alive today and a fine example of how imprinting regulates development.

Inge Liebaers and *Milan Macek* discussed genetic causes of female and male infertility, but both had to conclude that the field is waiting for a breakthrough and not much has moved since 2005. Closing the wet lab part, *Michael Morris* impressed on all of us the importance of accreditation in any diagnostic laboratory, and in particular in genetic laboratories. Mike's participation highlighted the role of Eurogentest.org in this setting the scene of quality assurance in genetic testing in Europe, and beyond.



Helena Kääriäinen and Heather Skirton from the ESHG then showed how molecular findings are translated for the patients, and how these are helped with dealing with their infertility during genetic reproductive counselling.

Last but not least, the last session was devoted to societal, political, ethical and legal issues. *Pascal Borry* showed how direct to consumer genetic testing is quickly changing the way patients deal with their genetic make-up, and certainly conveyed to all of us that these developments have to be treated with the utmost caution. *Martina Cornel* demonstrated why many reports of adverse effects after ART are so difficult to pin down, because of the small numbers, while others such as iatrogenic multiple births are quite obvious and easy to avoid. *Françoise Shenfield* demonstrated the high flight of cross border reproductive care in Europe, showing that legal restrictions are the most common cause driving patients to seek help in a country other than their own. *Sirpa Soini* put this all in legal perspective, showing that Europe is a mosaic when it comes to legislation on ART treatments and embryo research.

This group representing all salient issues between genetics and reproduction is now preparing a follow-up paper to the paper of 2006. The authors look forward to being part of such an important effort, and to again provide the genetic and reproductive community with a document sketching a framework that helps practitioners of all kinds (lab workers, counsellors, doctors, ethicist, lawyers,...) in their daily work.

Karen Sermon, coordinator ESHRE Special Interest Group Stem Cells

Claudia Spits, co-deputy ESHRE Special Interest Group Reproductive Genetics

(www.eshre.eu/binarydata.aspx?type=doc&sessionId=mn212ljaypgqoy45hu5nyh45/FoRmay.pdf)

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ESHG NEWSLETTER

The Evolution of ESHG's DNA Day Activities

by **Celia DeLozier**

(ESHG- ASHG Education committee liaison)

Five years- a very short time in the eyes of evolution! But long enough for the ESHG's DNA Day Essay contest to have evolved- grown, changed, adapted. The story started about six years ago, when an idea came up at the American Society of Human Genetics' Education Committee meeting, which I was representing the ESHG. What project could the two societies share? Why not the DNA Day Essay contest, where high school students compete with their essays on one of two questions chosen by the genetics societies?

2012 was the ESHG's 5th DNA Day essay contest (see www.dnaday.eu), the 7th for the ASHG. The societies have used the same questions (this year the ASHG had only one question whereas we offered two). See the website for archives including each year's questions, resources we suggested to students, the essays chosen for prizes and for honorable mention, and memorable quotes from students' essays.

ESHG Membership Meeting

Sunday, June 25

19.00 - 20.00 hrs

Room St. Petersburg

Refreshments will be served at the entrance of the room.

Ways in which the ESHG's DNA Day activities have evolved:

- from a pilot project sponsored by the Italian society in 2008 to a contest with participation of 23 countries in 2012
- from the judging of 90 student essays in 2008 (up to 3 per class, as chosen by their teachers) to submission of 266 essays in 2012
- from submission via e-mail to web-based entries using an abstract submission system
- from one round of judging by 8 ESHG education committee judges to two rounds with the help of 21 judges this year
- from manual grading of sets of essays which I triaged and forwarded for grading by email, to this year's on-line scoring of essays
- From announcing winners and awarding and prize monies at our annual meeting (May-June) to announcing them via the website on DNA Day (April 25) – in time for classes to use their prize monies for a trip or event during the school year.
- From announcing contest winners to geneticists in attendance at our annual congress to awarding them via a live webcam to which classes can tune in.

And a totally new event species emerged this year! Drs. Marjolein Kriek and Jon Frampton each created a video presentation on one of the essay questions; these videos were available on April 25 so that classes could "spend an hour with us on DNA Day" via the ESHG website. Check out these videos by ESHG geneticists!

The Education Committee and I would like to express our thanks to the students, their teachers, the judges, and to Drs. Kreik and Frampton. The tremendous efficiency and invaluable assistance of Jerome and Manfred at the ESHG administrative offices were a driving force in the evolution of this event.

At the upcoming meeting in Nurnberg, the Education committee, ESHG Exec and the International Federation of Genetics Societies will be discussing future DNA Day activities. We would be very happy to hear from you prior to or during the Meetings ; please send your comments/suggestions for the essay contest or other potential DNA Day activities to me (cddelozier@gmail.com) or to Tayfun Ozelik (chairman, Education Committee), at tozcelik@bilkent.edu.tr

EJHG EDITOR'S REPORT OVER 2011

by *GertJan van Ommen*
(Editor in chief, EJHG)

Highlights

- Last year, EJHG has seen a quite spectacular increase in Impact Factor, from 3.56 to 4.38 (22%), causing us to rise 15 places in the ranking, from 45/144 to 30/156. Probably partly due to this, our submissions, which slightly fell in 2010, have picked up again from 736 to 760. We published 277 papers, yielding a rejection rate of 62%. This is lower than the 67% of last year, and to try to maintain the higher impact factor, we have become more strict again in the course of the year.



- The Clinical Utility Gene Cards, a joint activity with Eurogentest, edited by Professor Joerg Schmitzke, introduced in 2010, have become a strong asset of EJHG, with 37 CUGC published in 2011. The abstracts are published in the journal (in blocks of three to a page), and the full data are published online. We are already developing the first updates.

- Another key component of our portfolio, the Practical Genetics items, are still very popular as well, occupying 8/10 places in our listing of most accessed online papers.

- The 'Open Access' mode introduced in March 2010, by which authors pay a market-conform advance fee for making their paper freely available, has led to 15 Open Access papers in 2011.

Section editors sought.

Of the manuscripts going out for review, the mean time from submission to first decision has remained stable at around 50 working days, or about 2½ months. From submission to final decision (i.e. including the authors' revision time), the mean time is 97 working days or 4½ months. We are not satisfied with this and there is an active interest for expansion of EJHG section editors, notably but not only in the areas of clinical and statistical genetics. For this, we are looking for experienced mid-career scientists and clinicians. interested in advancing their field and their society - and in putting in some community-service. IF YOU HAVE AN INTEREST COME SEE US AT THE BOOTH

EJHG Award

As every year, EJHG, together with Nature Publishing Group, offers a junior authors' high-citation award. This year, this is given to the top-3 papers published in 2010 and cited in the 12 months following after publication. The 1st prize includes a € 500 award and places 1-3 receive one year free ESHG membership + online EJHG subscription, and free registration for that year's or next year's meeting.

The winners, ex aequo for the first prize, are:

1a. Specific epigenetic alterations of IGF2-H19 locus in spermatozoa from infertile men, by Boissonnas, CC; El Abdalaoui, H; Haelewyn, V; et al, EJHG 2010, 18/1, with 25 citations in the first year,

and:

1b. Gene and pathway-based second-wave analysis of genome-wide association studies, by Peng, G; Luo, L; Siu, HC; et al. EJHG 2010, 18/1, also with 25 citations in the first year,

followed by:

2. Paternally inherited microdeletion at 15q11.2 confirms a significant role for the SNORD116 C/D box snoRNA cluster in Prader-Willi syndrome, by Duker, AL; Ballif, BC; Bawle, EV; et al. EJHG 2010, 18/11, with 21 citations in the first year,

and since we did not have a no. 3 this way, we decided to hand out a special award of the top-scoring paper in the category of Practical Genetics:

* Beckwith-Wiedemann syndrome, by R Weksberg; Shuman, C; and Beckwith, JB, EJHG 2010, 18/1, with 34 citations in the first year.

The ESHG Conference „smartphone“-app

by Jerome del Picchia

(Executive Officer of the ESHG)

So-called „smartphones“ and „tablet PCs“, which have penetrated the market in the last years, are obviously very useful and can even be a short-term replacement for a laptop, but most of them have a major inconvenience: they lack a USB port to transfer data. So after having stopped printing a hard copy of the abstract book, it seemed a logical step to make use of modern tools and to develop a „conference app“. This app for iOS and Android based mobile devices will be your personal assistant during the conference.

ESHG NEWSLETTER

You are able to **navigate through the conference programme “by day” or “by session type”** and **add (and remove) presentations to your personal agenda.**

You can **add notes** to the individual presentations, **email and print them.**

Full abstract texts are available and **can be searched.** Results are displayed in papers and sessions.

The **“What’s on?” button** takes you to the presentation(s) currently on schedule.

Of course all additional information is also available (**map of the venue and exhibitors, pdf of the programme, description of session types** etc).

Should **new abstracts** become available, the automatic update function will always keep your “app”-to-date.

Your agreement provided, the app will also send you **push-messages**, notifying you of last minute changes and important information. Although very tempting from a conference organiser’s point of view, we will restrain from abusing this function and limit the use to the minimum as I agree that constant pop-up messages on your phone can be a nuisance and clearly fail the aim of the feature.

We will of course continue developing this programme for future ESHG meetings and are also planning for a “Windows Phone” version.

Together with this app, it became also necessary to slightly adapt the ESHG logo, as you probably noticed, to make it more visible on small screens.

Check your App- or Play-store for “ESHG 2012” and download the free app today. This innovation at ESHG meetings comes with another change: Wifi is now available also outside the exhibition, but still not in the lecture halls. Hence the ESHG 2012 App is obviously usable “offline” as well.

On behalf of the ESHG staff, I wish you a successful European Human Genetics Conference 2012.

Jerome del Picchia



ESHG NEWSLETTER



EUROPEAN SOCIETY OF HUMAN GENETICS

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Invitation to the

Annual Membership Meeting 2012

At the EUROPEAN HUMAN GENETICS CONFERENCE 2012

Sunday, June 24, 2012 at 7.00 – 8.00 p.m.

Room St. Petersburg
NCC Ost (Nürnberg Convention Center Ost), Grosse Strasse, Messezentrum, 90471
Nürnberg, Germany

AGENDA

Opening by the President of the Society, Professor Jörg Schmidtke

1. Activity of the Society 2011-2012
2. Financial Report of the Society 2011
3. Discharge of the Board Members for the year 2011-2012

Opening by the new President of the Society, Professor Stanislas Lyonnet

4. Results of election for President-Elect
5. Results of election for Board Members
6. Membership fees 2013
7. Site of future European Human Genetics Conferences
8. Budget proposal 2013
9. Major policy questions proposed by Board
- 10: Change of ESHG statutes concerning membership categories and non-European memberships
11. Future activities

Please find the minutes of the last membership meeting in Vienna 2011 in the restricted area:
<https://www.eshg.org/39.0.html>