European Society of Human Genetics

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Report from the President

Dear colleagues,

2009-10 has been an incredible year for Human Genetics; I know this claim has been made about several periods over the past two decades but in the last few months we have seen the massive power of technologies such as next generation sequencing to identify the genetic basis of rare disorders using samples from a handful of patients with the promise of many more discoveries to come from this technology. We are also beginning to see personalised medicine becoming a reality through personal genome sequencing as well as through our increasing knowledge of complex biological pathways in health and disease. However, before reporting the part that your society has played in moving forwards the human genetics agenda, I would like to pause and pay tribute to one of our past presidents who sadly died in March 2010 at the early age of 57. Leena Peltonen-Palotie was one of the giants of human genetics, holding senior academic positions in Finland, the United States and the United Kingdom. The sheer scale of her achievements is incredible; she published over 500 papers, supervised more than 70 PhD students and contributed enormously to science policy and progress. Her work was recognised by the award of many international honours including election as an Academician of Science in Finland. In spite of all her responsibilities and the demands on her time, Leena was an extremely warm and generous person who gave equal support to young people starting out in genetics, professional colleagues and organisations to which she belonged. The ESHG will miss her very much and sends condolences to Arno Palotie, her husband, and their two children.

Of course the high point of the ESHG year is the annual conference and we are all looking forward to Gothenburg 2010 at the invitation of our colleagues from the Scandinavian societies, which we very much appreciate. Each year we say that the scientific programme was the best ever, but the hard working SPC has created yet another fantastic programme. We are truly grateful to Han Brunner, the outgoing chairman, and to Brunhilde Wirth, the new chairman, who have led the SPC together for the 2009 and 2010 conferences. We also are very indebted to the professionalism of the Vienna Medical Academy for the superb conference organisation and to Jantie de Roos for organisation of the exhibition, which contributes a great deal to the conference.

The ESHG would not be the effective society it is without the generous amount of time and effort many, many members put into it. The ESHG Board is elected and comprises members from a wide range of European countries and specialties within genetics who work together to identify issues and initiatives for the society and its committees to consider. The Executive Board meets at the ESHG Conference and on at least two occasions during the year to ensure that the society continues to move forward its work plan, as well as dealing with society matters such as financial management, conference venues, fellowship support for a range of courses and conferences, international relationships, etc. I would also like to acknowledge the excellent work of the ESHG committees whose reports are below. The PPPC chaired by Martina Cornel

continues to produce excellent reports on topical matters which I know have been of great use for individual society members in their own countries and this year they are organising a one day symposium before the conference together with the Genetic Services Quality Committee (Chair, Ros Hastings) on the important subject of the changing landscape of genetic testing and its impact on clinical and laboratory services and research in Europe. The Education Committee, now chaired by Peter Farndon, is organising two interesting workshops in Gothenburg contributed to by members of ESHG and EMPAG, emphasising the multispecialty nature of Human Genetics.

Three ad hoc committees concerning professional status and recognition of those professions providing genetic services now report directly to the Board, reflecting the importance attached to these activities. These were previously sub-committees of the Education Committee. A great deal of progress has been made by the committee on Medical/Clinical Genetics as a medical specialty in Europe, which will be reported by Milan Macek to the society – he and colleagues including Ulf Kristofferson, Helen Kingston and John Burn have seized on opportunities presented by the system in Europe for recognition of medical specialties (UEMS, Union of Medical Specialists) and by the emphasis at EU level on services for patients with rare disorders. The experience gained by this process is already being utilised by the ad hoc committees to progress specialisation in laboratory genetics and for genetic counsellors and nurses, both of which will report to the ESHG Board in Gothenburg.

ESHG also places real importance on encouraging the careers of young people in genetics and in addition to the fellowships to allow young investigators to participate in the ESHG conference, it also supports fellowships for courses delivered by other bodies such as the European Genetics Foundation. Several other courses have also had core support from the ESHG and we were a co-sponsor of the MediMedGen Conference held in Ankara in June 2009 and hosted by Tayfun Ozcelik, which was the focus for the August 2009 Editorial in *Nature Genetics,* which made the very strong point that scientific collaborations transcend politics.

Human Genetics research is of course very much a global endeavour and it has been encouraging in recent years to note the increased prominence of the International Federation of Human Genetics Societies (IFHGS) in which the ESHG is an active participant. I would certainly recommend that members of our Society plan to attend the 12th International Congress of Human Genetics to be held in Montreal in October 2011. Within Europe, the ESHG Conference is now proud to host the annual meeting of the Presidents of Human Genetics societies of the European countries, instigated by one of our past presidents PierFranco Pignatti. The annual ESHG Newsletter expertly edited by Lina Florentin and the new ESHG Flashletters keep us in touch across the society.

Many members have received individual awards during this year but I would particularly like to congratulate GertJan van Ommen and John Burn, who each were awarded knighthoods of their respective countries in 2010. These honours reflect very well on Human Genetics. It would be good if we had photographs of them in their regalia in a future newsletter!

Finally I would like to sincerely thank the members of the Executive Board for their very hard work this year (Helena Kääriäinen, Gunnar Houge, Andrew Read, Jean-Jacques Cassiman, Milan Macek) and particularly our Executive Officer, Jerome del Picchia, who since he took on the role has moved the organisation of our society onto a wholly new and very professional level. I am very pleased to pass the presidency to Milan, who has enormous energy and vision for genetics in Europe and wish him well in the role.

Dian Donnai
President of the ESHG

Report from the Scientific Programme Committee

By Han Brunner, Co-Chair of the SPC

The Scientific Programme Committee for 2009-2010 was composed of Han Brunner and Brunhilde Wirth (co-chairs), Kristiina Aittomäki, Corinne Antignac, The-Hung Bui, Niklas Dahl (Local Host), Manolis Dermitzakis, Peter Heutink, Gunnar Houge, (Secr. General elect, observer), Thomas Jensen, Helena Kääriäinen, (Secretary General; ex officio observer), Batsheva Kerem, Mark McCarthy, Carla Oliveria, Olaf Riess, Mariano Rocchi, Pete Scambler, Michael Speicher, Eduardo Tizzano, Draga Toncheva, Mikka Vikkula, and Cisca Wijmenga.

The SPC met twice to organize the Gothenburg 2010 ESHG conference: in June 2010 to decide on the plenary sessions and symposia, and in Vienna at the VMA offices in March 2010, to select the abstracts for oral presentations and posters.

The number of submitted abstracts was 1718, and many were again of excellent quality. This allows us to keep the number of abstracts selected for oral presentation around 5% of the total number. On the first day, particularly exciting new findings are presented in a "What's new?" session from submitted abstracts.

We have followed a suggestion from the board and created an "educational thread" throughout the programme. This means that for each day of the conference participants can choose to either visit a symposium on the latest scientific findings, or to go an educational 'update" session on a specific subject. Among the subjects are: Genetic causes of infertility, Overgrowth and Undergrowth syndromes and Genome databases, to name a few.

For the final session on Tuesday 15 June, we were able this year to attract Professor Mary-Claire King from Seattle, who will speak from her tremendous experience in both the scientific and the societal aspects of human genetics.

As is 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 2010 will be awarded to Sir Alec Jeffreys from Leicester UK, in recognition of his influential and groundbreaking work which lead the way in understanding human variability at the DNA level, its causes including recombination, its consequences such as copy number variation, its role in driving genome evolution and its many applications in society.

After the Gothenburg conference, the SPC shall have to say goodbye to Mariano Rocchi, Olaf Riess, Pete Scambler, Batsheva Kerem, and Peter Heutink. We thank them for their work and their dedication to making the meeting better. Han Brunner will step down as chair of the SPC, which was about time. Starting from the Amsterdam meeting in 2011, Brunhilde Wirth will continue as the SPC chair. It is great to know that with her the meeting is in safe hands.

Report from the Public and Professional Policy Committee 2009-2010

by Martina Cornel, Chair of the PPPC

The Public and Professional Policy Committee (PPPC) of the ESHG worked on several topics during the year 2009-2010: the changing landscape of genetic testing in the age of whole genome technology, genetic testing for common disorders, genetic testing and mental health and direct-to-consumer testing (DTC). A response was sent to the UK Human Genetics Commission consultation on DTC after discussion at the PPPC meeting on 30th November 2009, as can be found at the ESHG website (under public policy). The PPPC appreciates that the Human Genetics Commission wants to set standards and principles with regard to the provision of genetic tests sold directly to the consumer and to promote their consistent use at an international level. The PPPC also applauds the fact the Human Genetics Commission has engaged in a discussion with representatives from companies offering direct-to-consumer (DTC) genetic testing services regarding these commercial activities. The principles proposed should not be limited to *genetic* issues only, but rather

to predictive health information. The governance of the fields of medication and IVDs needs to be closely linked.
Draft recommendations of the ESHG on DTC were sent to the ESHG membership for comments, and to the Board for approval at their Gothenburg meeting. In line with the Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes, this 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.
A satellite meeting on "the changing landscape of genetic testing in the age of whole genome technology" was co-organized with the Quality Committee and supported by two Netherlands Genomics Centres (CSG & CMSB). Speakers covered on the 11th of June 2010 the broad spectrum of whole genome sequencing and array technology, the challenges these techniques imply for clinical geneticists (which unintended findings to report? how to organise informed consent?), how to build databases containing the evidence needed on existing genetic variants, ethical and legal implications and the challenges for non-genetic health care professionals. Research and patient care apparently have blurred boundaries. Biobanks promise to report relevant results, while genetic diagnostic tests generate more data than needed. Regulations for the two separate fields need to be integrated.
Three members of the PPPC visited the Society for the Study of Behavioural Phenotypes (SSBP) meeting in Cambridge, 14-16th October 2009, to discuss possibilities for collaboration on genetics & mental health. Soon after the visit, an Italian appeals court reduced the sentence of a murderer after he turned out to be carrier of a few genetic variants thought to be associated with a predisposition to aggressiveness. This led to a "policy" contribution in the European Journal of Human Genetics (2010;18:519-21) of PPPC members, together with the chair of SSBP, debating several aspects of the sentence. As long as the clinical utility of these genetic susceptibility tests is not proven, use in the context of legal proceedings is questionable. People should be sentenced based on who they are, not on who they could be. The terminology that geneticists use to report on genes "responsible" for disease might at least be confusing and should better be avoided.
After public consultation on genetic testing for common disorders, suggestions were discussed and integrated in the final versions of background document and recommendations.
Members of the PPPC in 2009-2010 were Pascal Borry, Anne Cambon-Thomsen, Martina Cornel (Chair), Thoas Fioretos (till June 2010), Francesca Forzano, Shirley Hodgson, Gyorgy Kosztolanyi (till end 2009), Jan Lubinski, Christine Patch, Jorge Sequeiros, Aad Tibben, Lisbeth Tranebjearg and Veronica van Heyningen (till June 2010), supported by Carla van El.

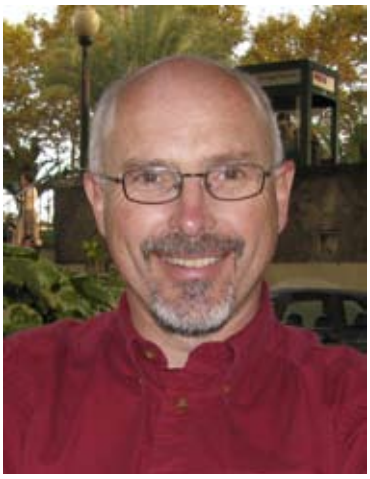
Annual report from the ad hoc Committee for Recognition of Clinical Genetics as an EU-speciality

Professor Milan Macek has been very active during the year to collect necessary information from member states on their view on the proposed European basic training guidelines and to present these to the EC Committee for Recognition of Professional Qualifications. If the required number of votes set by the EU Qualified Majority voting scheme is achieved, the specialty will be eligible for inclusion in the Directive when it is amended in 2012. Milan Macek presented his efforts at a UEMS/ESHG joint workshop in Brussels January 20, 2010. A report from that meeting is available on the ESHG website and includes a summary of a talk by Robert Newton on behalf of the UK National Genetics Education and Development Centre, where he described the UK initiative on education of non-genetic medical specialists.
The committee plans a new joint UEMS/ESHG workshop in Brussels in January 2011.

Ulf Kristoffersson
Committee chair

Helen Kingston
Committee secretary

The ad hoc committee on medical laboratory genetics has started its work



In view of the great interest expressed when this topic was raised at last year's ESHG conference in Vienna, we are happy to announce that an ESHG ad hoc committee on laboratory genetics has been established and begun its work. It is chaired by Jacqueline Schoumans, now working in Lausanne and with previous extensive work experience from Holland, Norway and Sweden. The co-chair is Bert Bakker (Leiden), and others members are David Baty (Dundee), Kim Smith (Oxford), Mireille Claustres (Montpelier), Joris Vermeesch (Leuven), Bela Melegh (Budapest) and Thomas Liehr (Jena). Gunnar Houge will serve as ESHG executive committee liaison, and the work will also be closely monitored and assisted by our next president, Milan Macek, drawing on his experience on the establishment of the clinical genetics speciality within the EU.

The committee has an about 50/50 mixture of people with main experience in either cytogenetics or molecular genetics. A common speciality for both groups of professionals will be a major goal. When a consensus on how to achieve this goal has been reached, hopefully within a short time, it is of utmost importance that the various national bodies promote the case on the national level. The national representatives must do all they can to influence and convince their national representatives at the EU Recognition Committee, where our proposal for cross-border EU recognition of laboratory genetics eventually will be voted upon. In each country, legal dossiers (if they exist) should be collected, and if not, a document/letter from the most appropriate body/organisation stating that the proposed core curriculum is in line with what is being planned or what is current status/practise should be produced.

In addition to lobbying towards EU Recognition Committee representatives, it is necessary that a few member states make an official request to the European Commission for putting the item on the agenda of the Recognition Committee. Most likely, all the work done on the clinical genetics speciality will facilitate our work and chances of recognition to a great extent.

On behalf of the committee members,
Gunnar Houge, ESHG deputy general secretary

Genetic Nurse and Counsellor Ad Hoc Accreditation Committee Report- May 2010

General

The network of genetic nurse and counsellor members currently stands at 114 members from 19 countries. The website (http://engnc.org/) is maintained so that members can obtain updates on our activities. Our thanks to Vigdis Stefansdottir, who has contributed her time again this year to maintain the website.

Standards of Education and Professional Practice

The main objective for this year was to make progress on setting standards of education and professional practice for genetic nurses and counsellors in Europe and we have made substantial progress. After preparing a draft set of standards through the use of electronic communication, we used funding from the ESHG to have a meeting to refine the standards. In November 2009, 14 participants from 11 countries met in Paris for a one-day meeting to prepare the document for dissemination to the entire group. The ESHG funding for our committee covered travel costs for the participants, plus one night accommodation and meals, but we were able to use a meeting venue arranged free of charge by genetic counsellors in Paris, and we are grateful to those colleagues for enabling us to use the venue. We finalised the draft and the membership of the network then voted overwhelmingly to approve the standards.

Following approval by the membership of the network, we sent the document to the national genetic nurse or genetic counsellor societies in Europe. It is important that the standards are commensurate with those of

individual national professional groups. We have had very positive feedback from the following:
- Dutch Association of Genetic Counsellors
- French Association of Genetic Counsellors
- Norwegian Association of Genetic Counsellors
- Association of Genetic Nurses and Counsellors (UK)

All the organisations listed above have approved the standards, with some minor suggested changes that we will discuss in Gothenburg.
We are awaiting comments from the Swedish Genetic Counsellor Association.
Our plan is to submit the standards at the ESHG conference to ask the ESHG Board to comment upon them and to eventually send them to the relevant national genetics societies.

Activities at the ESHG/EMPAG conference, Gothenburg, 2010

As there are many countries in which the number of genetic nurses and counsellors is very small, the ESHG/EMPAG meeting is an important focus for networking, as well as education, for genetic nurses and counsellors. However, many have difficulties obtaining funding to attend.
We invited members to submit an application for a Fellowship to attend the conference. All applicants had to submit an abstract (this was intended to encourage them to disseminate good practice). The standard of the applications was very high and we awarded eight Fellowships. Those awarded a fellowship were: Clara Serra, Viviane Cina, Rebecca Pestoff, Yurdugal Erdem, Nina Bosch, Beppy Caanen, Cristophe Cordier, Ramona Moldovan.
The grant was also used to fund a lunchtime meeting on Saturday 12 June from 1100hrs – 1300hrs. At this lunch we will discuss the Professional and Educational standards document and have a guest speaker.

We wish to thank Genzyme for an unrestricted grant that has enabled us to fund the Fellowships and lunchtime educational meeting.
Our workshop on the topic 'Embedding genetic counsellors into clinical genetics in Europe' will take place at the EMPAG meeting on Sunday 13 from 1500-1630hrs.

Thanks

We sincerely thank the ESHG for all the support we have received to carry out this work in the past year. In particular, we greatly appreciate the advice provided by Jerome del Picchia when we sailed into unfamiliar waters.

Heather Skirton and Marie-Antoinette Voelckel.

Editorial report for EJHG over 2009

By GertJan van Ommen, Editor in Chief, EJHG

• While last year we took the 4.0 Impact Factor Hurdle, this lead turned out to be too small to buffer fluctuations, and the slight drop of 0.078 caused us to fall just below again, to 3.925. The good news is that our ranking in Genetics and Heredity improved from 40 to 35, so let's decide that this offers us the chance to round the 4.0 cliff once more.
• Submissions increased by another 5% in 2009, a total 55% increase compared with 2006, and the acceptance rate has decreased from a several-year stable 35% to 29% to accommodate the rise in interest. EJHG published 8% more articles in 2009 than in 2008, which implies an overall 18% increase in two years.
• EJHG authorship is still predominantly European with 70% of accepted articles. US/Canadian authorship has further increased to 18%.
• Decision times have reduced further, with a median first decision of 14 working days and the median final decision time of 15 working days after submission of the last revision. Echoing last year, however, we note that these are median figures and we are aware that some manuscripts have had significantly longer processing times. Due to the increasing number of genetics journals and global budgeting, combined with rising technological possibilities, people seem to be busier than ever and soliciting reviewers (and actually have them return their

reviews in time) has become increasingly difficult. We are doing our best to address this, amongst others by a 30% extension of the editorial time commitment.
• Due to the increased submissions, time to print publication further increased in 2008, peaking at 7.2 months in October. To help deal with the backlog, 2009 pagination was increased and we ended the year 351 pages over budget. To address the increased author interest more structurally, a template change has been carried through in 2010, which should help save space and reduce the backlog.
• EJHG continued to perform well online in 2009. Eight out of twelve months we published more than 65% of articles as Advanced Online Publications within 25 working days. EJHG content accessed from PubMed averaged at 11,649 times per month (15% up). The most frequently accessed articles are quite varied, in line with our broad scope, see table.

Table 1. EJHG top-10 Pdf downloads 2009

Title	Type	Issue	Downloads
Breast cancer susceptibility: current knowledge and implications for genetic counselling	Review	17/6	1,314
Marfan syndrome: clinical diagnosis and management	Practical Genetics	15/7	883
Prader–Willi syndrome	PracticalGenetics	17/1	819
Using biological networks to search for interacting loci in genome-wide association studies	Article	17/10	710
Fragile X syndrome	Practical Genetics	16/6	689
Y-chromosomal evidence of the cultural diffusion of agriculture in southeast Europe	Article	17/6	678
Genotype–phenotype correlations in Down syndrome identified by array CGH in 30 cases of partial trisomy and partial monosomy chromosome 21	Article	17/4	574
Gene and pathway-based second-wave analysis of genome-wide association studies	Article	18/1	550
SNP frequency estimation using massively parallel sequencing of pooled DNA	Short Report	17/3	548
Smith–Lemli–Opitz syndrome: pathogenesis, diagnosis and management	Practical Genetics	16/5	546

EJHG Citation Award 2009

As every year, EJHG has a junior authors' high-citation award, to hand out at the Gothenborg meeting. In principle, 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. This year however the differences were so minimal, and the period subsequent to the counting window showed further changes, so that we decided to have three ex aequo first prize winners, each receiving € 200, a one-year online EJHG subscription and free registration:
Dr. Amy Lawson-Yuen et al. for her paper "Familial deletion within NLGN4 associated with autism and Tourette syndrome", which appeared in EJHG 16 no. 5 (2008), with 10 citations in May 2008 up to May 2009 (12 more till January 2010); Dr. Simon Heath et al. for his paper "Investigation of the fine structure of European populations with applications to disease association studies", EJHG 16 no.12 (2008) with 11 citations in December 2008 up to December 2009 (and 4 more till Jan 2010); and Dr. Beate Skinningsrud et al. for her paper "Mutation screening of PTPN22: association of the 1858T-allele with Addison's disease" EJHG 16 no. 8 (2008) with 11 citations in August 2008 up to August 2009 (and 3 more till Jan 2010).

ESHG Genetic Services Quality Committee

By Ros Hastings, Chair of the GSQC

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 relating 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 have (or will be) established to take them forward.

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

The ESHG Quality and PPPC committees have worked together to co-ordinate an ESHG Satellite Symposium on 'Changing Landscape of Genetic Testing' on 11th June 2010 in Gothenburg . The application of the new genetic technologies (array CGH, whole genome sequencing) that examine the human genome will have an impact on both laboratories (cytogenetic and molecular genetics in first instance) and clinical/medical genetic services. There is a need to discuss how best to re-structure the services logistically and determine clinical utility of genetic testing so that patients can receive appropriate advice and genetic testing. Presentations from experts will address the multiple implications of these new technologies, while the workshops will discuss possible ways forward to improve and adapt the genetic services so that patients receive accurate and relevant information. More information will follow on the outcome of this Symposium.

The GSQC recognizes the need to raise the role of its activities and promote the role of quality management; hence this article is in the ESHG newsletter. A good Quality Management System (QMS), externally verified through accreditation (ISO 15189), is the gold standard all laboratories should aspire to (see also OECD guidelines). Laboratories are also encouraged to 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. The database has information on the Quality Manager, EQA participation and accreditation status.

External Quality Assessment (EQA) plays an important role in monitoring and improving the quality of a laboratory's service. While it is a requirement for any accredited laboratory to participate continuously in relevant EQAs; EQA is equally recommended for all diagnostic laboratories. 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 components of the marking criteria for all four European schemes are similar. Given the serious impact that an inaccurate result would have on patients and their families, the number of genetic laboratories making errors in the European EQA schemes is considered to be too high by the GSQC and Scheme Organizers. Sometimes the EQA error rate is artificially raised when a rare abnormal cases is given as part of the EQA, but the analytical error rate is still of concern and the GSQC has asked the EQA providers to submit an audit of the satisfactory performance from the last three years. The GSQC has defined a satisfactory performance as 'having no critical errors' and poor performance in interpretation as either a critical interpretation error or dropping below 70% of the average mark for interpretation in an EQA. Discussions are also ongoing on the practicalities of the GSQC overseeing persistent poor performing laboratories.

The Quality Committee was asked to suggest genetic experts who could attend the WHO reference materials meeting. Mike Morris (CH) and David Barton (IE) were able to attend this meeting. Their involvement in the process resulted in the WHO approving the Prader Willi/Angelman syndrome and BCR-ABL reference materials this year.

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.

Good news: a grant proposal to continue EuroGentest has been favorably evaluated by the European Commission

In November 2009, we filed an application for a project that would allow us to continue the activities of EuroGentest, or at least to maintain the network.

Indeed, we believe that EuroGentest (www.eurogentest.org) has delivered a great deal in terms of supporting laboratories and genetic clinics towards improvement of quality in genetic testing, and that within the 5 years of its existence, it has created a momentum that should not be stopped. Some people call EuroGentest a brand or trademark, and it seems as if it has always been there and should certainly not disappear.
The new project was named EuroGentest2 (or EUGT2), since we were obliged to give it a new name and acronym. But the brand name will always be EuroGentest. The topic in the Seventh Framework call of the European Commission, to which the project was submitted, was tailored to our needs: "Harmonisation, validation and standardisation in genetic testing" (HEALTH 2010.1.2-3).
In return, we filed a project that neatly fit into the call, as can be deduced from the very positive score after evaluation: 14/15. By the end of March 2010, we received notice that it was 'acceptable for funding'.
The focus will still be on quality assurance of genetic practice, including different types of training, and the realm will also include prenatal diagnosis (PD), pre-implantation diagnosis (PGD) and non-invasive prenatal diagnosis (NIPD). Points of interest will be: External Quality Assessment (EQA) and the harmonization thereof, the validation of novel technologies and mainly the coordination thereof, and the generation of 'Clinical Utility Gene Cards', which is indeed a major endeavor. Attention will be paid to Direct-to-Consumer (DTC) testing – a feature which was very much appreciated by the reviewers – and to the recast of the In Vitro Diagnostic (IVD) Directive. In general, EuroGentest wants to actively participate in different types of workgroups, and, in close collaboration with the European Society of Human Genetics (ESHG), weigh on international decisions with regard to genetic testing. A specific, new work package will coordinate all policy activities.
The plan to create a EuroGentest foundation, which should warrant the sustainability of the network after the granting period, was applauded by reviewers, and we hope that it will be welcomed by the genetics community as well. It is not going to be another professional body, but rather a network or association of genetic centres – laboratories as well as clinics, the users of EuroGentest 'products'.
We will present EuroGentest2 at the ESHG meeting in June, at the booth, to spread the news. In addition, we plan to organize a meeting well before the start, i.e. sometime in the fall. We aim for January 1st 2011 for the project to formally begin.
EuroGentest2 is a Coordination Action. The transition for Network of Excellence to the Coordination Action, and the concomitant reduction of the budget, forces us to give in on aspects of education, ethical and legal studies, and on research per se. These aspects can better be shared with ESHG. In the meantime, the main objectives of EuroGentest will continue to obtain full attention and support.
To encompass the largest possible scope, EuroGentest will also closely collaborate with other European-funded projects and networks. For example, we consider to append the scientific meeting of EUROGENTEST back-to-back to the scientific meeting of TECHGENE ('Technological innovation of high throughput molecular diagnostics of clinically and molecularly heterogeneous genetic disorders', see www.techgene.org). We will also link to the READNA project ('REvolutionary Approaches and Devices for Nucleic Acid analysis' , www.cng.fr/READNA). The combination of these 3 projects covers all aspects from technology development to diagnostic implementation and validation to quality assurance. Other projects in which members of EuroGentest are involved are e.g. GEN2PHEN (to unify genetic variation databases, www.gen2phen.org) and NMD CHIP (designing DNA arrays to efficiently diagnose patients affected with neuromuscular diseases, www.nmd-chip.eu).

We believe that EuroGentest will continue to thrive for the benefit of the genetics services in Europe. We count on your continued support to make it work !

Gert Matthijs, Jean-Jacques Cassiman
Coordinator(s) of EuroGentest

New Programme for Genetic Counsellors in Porto, Portugal

By Jorge Sequeiros, May 2010

A new master course for education and clinical training of Genetic Counsellors was launched this past academic year, at ICBAS, University of Porto. Rhona MacLeod, genetic counsellor from Manchester, and Lavínia Schuler-Faccini, medical geneticist from Porto Alegre, Brazil, were the invited speakers at the opening ceremony. This is a fulltime, 2-year programme and the 6th professional master course in Europe. The first 6 students selected are nurses or clinical psychologists, all with some previous clinical experience, and came from mainland Portugal, the Azores, France and Brazil. The students in the photo, in a class with Milena Paneque, genetic counselor trained in Cuba and teacher at the MSc in Porto, and with Lavínia Schuler-Faccini.

Portugal, where the Medical Genetics specialty for MDs was created in 1999, has 5 genetics services in Lisbon (2), Coimbra and Porto (2), for a population of 10 million; there are 26 clinical geneticists active in the health system (though 49 are actually registered at our Medical Genetics College, some are already retired, dedicated to another specialty or involved only in research and teaching).

Programmes for non-medical Genetic Counsellors exist in the USA since 1969 (Sarah Lawrence College), but are much more recent in other countries (Canada in 1983, Australia in 1996, Israel in 1997 and Cuba in 1999). Other programmes exist at least in South Africa, Japan, China, Taiwan and Saudi Arabia.

The first European masters level training programme in genetic counselling was established in the UK in Manchester (1992) with a further course in Cardiff (2000). Master courses initiated then in Norway (Bergen, 2001), France (Marseille, 2004) and Spain (Barcelona, 2007); still others are being planned, including in Italy (Genoa) and the Netherlands (Groningen). Some of these, however, are academic rather than professionalizing courses.

In 2007, a new European Network for Genetic Counsellors and Nurses (http://engnc.org/) was initiated with the support of the ESHG.

NEWS

On April 29 Professor **GertJan van Ommen** received a Knighthood in the 'Order of the Netherlands Lion' for his internationally recognized contribution to genetic diagnostics, genomics and therapeutics, notably in the Duchenne field, and for his efforts at making geneticists in the NL and abroad work together.

Professor **John Burn**, head of the Institute of Human Genetics at Newcastle University, has been awarded the title of Knight Bachelor in the Queen's New Year Honours List for services to medicine.
He helped set up Newcastle's International Centre for Life, which was opened by the Queen in 2000.

Dian Donnai, Professor of Medical Genetics at the University of Manchester and the current President of the European Society for Human Genetics received the March of Dimes/Colonel Harland Sanders Award for achievement in genetic sciences.

Medical Genetics in Sweden, past and present

Medical Genetics in Sweden has a complex history over the last 100 years, reflecting changes in both values and knowledge. In 1910 two societies were formed: the "Swedish Mendel Society" and the "Swedish Society for Racial Biology". Both societies were influenced by contemporary ideas in eugenics (racial biology) and social Darwinism. The Swedish Parliament decided, across political borders, to establish an independent research institute of "Racial Hygienics" in 1922. Interestingly, one of the first objectives for the institute was to characterize what was believed to constitute the "Swedish race". With time and increased knowledge the research became more disease-oriented and with a focus on population health. The institute was finally transformed to a university department for medical genetics in 1958. Two years earlier, in 1956, the Swedish cytologist Albert Levan and his collaborator Joe Hin Tjio established the human diploid chromosome number to 46 (Fig. 1.). This became a fundamental starting point in human genetics and a decade later the clinical applications started to evolve at a few Swedish university hospitals. This was driven by the progress in cytogenetics and a few dedicated clinicians with different clinical backgrounds (e.g. pediatricians, gynaecologists, pathologists, psychiatrists). The hospital units for genetics were originally organized around doctors and cytogenetic laboratories and a few technicians. A milestone in medical genetics came in 1970 with the discovery of chromosome banding by Lore Zech in Torbjörn Casperssons group. The technique, still crucial for the identification of specific structural chromosome rearrangements, boosted cytogenetics in Sweden. Chromosome banding was also a prerequisite for Felix Mitelman's systematic catalogue of cytogenetic rearrangements associated with cancer. This is now accessible as the web-based "Mitelman database of chromosome aberrations".

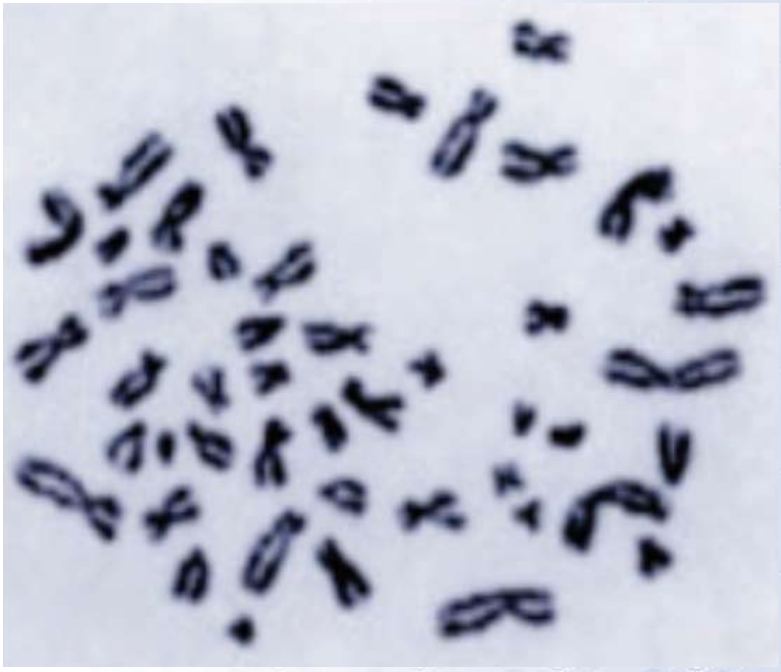


Fig 1. From: Tijo J.H. and Levan A. (1956) "The chromosome number of man". Hereditas 42, 1-6

Clinical genetics became a qualified competence in 1977 and a formal Swedish speciality in 1992. Clinical genetics is today represented at six university clinics (Göteborg, Lund, Linköping, Stockholm, Uppsala and Umeå), providing services for 1-2 million Swedes per centre. The training in clinical genetics spans a 5 year tenure-track with a defined curriculum. Most clinical geneticists are also involved part-time in research and a few are contracted by the universities. Today, 30 specialists and residents serve the Swedish population of approximately 9 million. Medical genetics is part of undergraduate studies for medical students and comprise courses equivalent to between 2 and 5 weeks, depending on the university. Other staff members such as hospital geneticists (clinical scientists), counsellors and nurses are steadily increasing in number at centres for clinical genetics. Genetic scientists today have a curriculum with 5 years of training following a master degree. The majority of scientists have a PhD background and they constitute a core group of indispensible experts in diagnostic methodologies. There are no national training programs for genetic counsellors but the Swedish Society for Medical Genetics and the counsellors' association are currently working on a formal program leading to a board exam. Bioinformaticians are still rare at the clinical units but will certainly become numerous with massive parallel sequencing around the corner.

The six clinical genetic laboratories in Sweden are today part of different quality control networks. These include e.g. EMQN, UK-NEQAS, EWALL, European Chimerism Network (molecular genetics), Labquality and CEQA (cytogenetics) and ELN (leukemias). A clinical geneticist is a board member of the Swedish malformation registry and associated with EUROCAT and ICDBSR. Some of the analyses performed at Swedish genetic laboratories are advertised through web pages (e.g. EuroGentest, EDDNAL , GeneTests and Orphanet). Sweden has a national network for quality and standards in onco-genetics with all centres as members. The network was established 15 years ago with a focus on counselling and follow-up of families with inherited predispositions to cancer.

The spectrum and incidence of disorders caused by genetic factors are similar in Sweden and other (north) western European countries, reflecting shared and recent origins. Neonatal screening of a few disorders started in the 1970s. The screening comprises PKU, galactosemia, congenital hypothyroidism, congenital adrenal hyperplasia and biotinidase deficiency. Non-invasive prenatal screening (ultrasound with or without biochemical test) is offered today to all pregnant women. The health care system in Sweden is free and mainly publicly-funded (98%). This, together with the small population of Sweden, may explain why patient organisations in Sweden are relatively weak with the exception of those for mental retardation and haemophilia. The lack of national networks for some orphan diseases as well as limited clinical experience in the care of certain rare disorders is a concern, creating difficulties for many families.

The legislation in Swedish health care is very clear about the right of self-determination. A recent law on genetic integrity takes a strong position in defending the patient's right to non-disclosure of genetic information towards e.g. employers and insurance companies. The autonomy and integrity of the individual must be considered even in cases of severe and inherited disorders involving other family members. It is also stated that the patient must be fully informed about all results obtained from genetic investigations or analyses. A biological sample deposited in a biobank after clinical diagnosis or for future research is owned by the donor. Accordingly, the specimen should be destroyed if this is a later wish from the owner. All research projects involving biological material must undergo an ethical review in Sweden.

Similar to other countries, clinical genetics in Sweden is facing exciting challenges. Scientific progress gives us the possibility to provide more and better service to the society. Improved knowledge about normal genomic variations, associations between genetic variants and phenotypes, as well as high throughput technologies, are continuously increasing the diagnostic resolution. The perspectives are fascinating but the applications may not be straightforward. The future for clinical genetics will require open discussions and continuous ethical considerations involving society and professionals, within as well as between countries. A major challenge will also be to properly communicate the possibilities and the outcome of our work with the patient.
I feel that we are not even at the beginning of the end in human genetics, but rather at the end of the beginning.

The author (right) in practice

Niklas Dahl
Chair
Swedish Society for Medical Genetics

ESHG COURSE
INTRODUCTION TO THE GENETIC EPIDEMIOLOGY OF MULTIFACTORIAL DISEASES

F Clerget- Darpoux (Paris- Sud), S Lyonnet (Paris-Descartes), P Broët (Paris- Sud)

Teaching period: **November 22 - 26, 2010**
Teaching place: CHU du Kremlin Bicêtre, Faculté de Médecine Paris-Sud, **France**

During the first three days, we will introduce **the concepts and methods of Genetic Epidemiology**. The teaching will be made at a **basic level** and mainly concerns Molecular and Medical Geneticists.

The Thursday and Friday seminars have to be considered as illustrations of the methods and approaches presented during the first three days. They will present up-to-date results on several common diseases.

For information and registration, please see the website https://www.eshg.org/courses.0.html or contact: *courses@eshg.org*

Annual Membership Meeting 2009
At the EUROPEAN HUMAN GENETICS CONFERENCE 2009, Vienna
Sunday, May 24th, at 7.00 – 8.00 p.m.

Present:
All members of Executive Board
and most Board members and Committee chairs,
about 100 members altogether.

The meeting was opened by the President of the Society,
Prof. Jean-Jaques Cassiman

1. Activity of the Society 2008-2009

Prof Cassiman updated the membership about the situation concerning recognition of clinical genetics as a medical specialty. Prof Milan Macek had been very active and the situation had been proceeding in a promising way.

New initiatives from previous year were proceeding well. There had been much more essays sent for the DNA-day and the quality had been very nice. The new fellowships offered via NHGSs had been present for the second year. The idea of ESHG courses/ESHG supported courses was also proceeding so that the plan was to have a new ESHG course in the Autumn (Genetic counselling: train the trainees) and a ESHG supported course in Vilnius in June 2009.

Short reports of the Committees were also given; more comprehensive reports had been published in Newsletter.

Regarding Education Committee, it had been decided that the former sub-committees will become ad hoc committees

- Medical/clinical genetics as a European Specialty
- Specialty issues for nurses/counsellors
- Specialty issues for laboratory genetics

2. Financial Report of the Society 2008

Prof Andrew Read, Treasurer of ESHG gave the financial report which was accepted. The financial situation of ESHG was good allowing continuation of the high number of fellowships and other new initiatives.

3. Discharge of the Board and Executive Board Members

- Pier-Franco Pignatti
- Nurten Akarsu
- Jacques Beckman
- Andres Metspalu
- Alessandra Renieri.

The second part of the meeting was opened by the President of the Society, Prof Dian Donnai

4. Results of election for President-Elect

There had been only one nomination for President elect, Prof. Milan Macek from Prague. Membership was happy to accept him as new President Elect.

5. Results of election for Board Members

There had been several nominations for Board members and an electronic voting had been organized some weeks before the Vienna conference. Domenico Coviello (Italy) and Borut Peterlin (Slovenia) had been elected. Membership happily accepted the result of the election.

6. Membership fees 2009

Membership fees were not changed.

7. Site of future European Human Genetics Conferences

Gothenburg 2010 and Amsterdam 2011 had already been decided. Annual meetings committee proposed Nurnberg or Dresden for 2012. The decision was left to the committee who was planning a site visit to see the venues.

8. Budget proposal 2009

Prof Andrew Read had drafted budget proposal for the year 2009-2010, this was accepted.

9. Major policy questions proposed by Board and future activities

Specialty issues for laboratory geneticist were discussed, it was hoped that the new ad hoc committee would actively start to work for this important but difficult goal.

It was hoped that next year's DNA-day would be announced earlier.

With encouraging experience joint memberships with national society in UK, the same has been offered to all NHGSs.

Support to EGF will continue, it was hoped that EGF and ESHG Education committee would collaborate more closely.

10. President closed the meeting

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Invitation to the

Annual Membership Meeting 2010

At the EUROPEAN HUMAN GENETICS CONFERENCE 2010
Sunday, June 13, 2010 at 7.00 – 8.00 p.m.
Room F6
The Swedish Exhibition & Congress Centre, Mässans Gata 20, Gothenburg, Sweden

AGENDA

Opening by the President of the Society, Professor Dian Donnai
1. Activity of the Society 2009-2010
2. Financial Report of the Society 2009
3. Discharge of the Board Members for the year 2009-2010

Opening by the new President of the Society, Professor Milan Macek Jr.
4. Results of election for President-Elect
5. Results of election for Board Members
6. Membership fees 2011
7. Site of future European Human Genetics Conferences
8. Budget proposal 2011
9. Major policy questions proposed by Board
10. Future activities