



European Society of Human Genetics

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Dear Colleagues and friends



*Professor Jean-Jacques
Cassiman
President of the ESHG*

The European Genetics Conference 2009 of Vienna is welcoming you with open arms. It is now a well established tradition in the European Society, that the scientific program will once more be of the highest quality and that the number of participants will again be well over 2000. This conference is without any doubt a major and successful activity of our society. Nevertheless, over the years the society has also developed a series of activities which in recent years have come more and more to the forefront and are making the society a real and important representative of Human Genetics in Europe; we are definitely becoming the "voice of human genetics in Europe".

Of course, our established activities are well known to everybody. You will find their reports further in this newsletter, but remember that all this is the result of hard work by motivated people who are giving their time and expertise in a voluntary way to the society.

As already mentioned, the Genetics Conference is a success mainly thanks to the remarkable efforts of our Science Program Committee. Nothing new there you will say, but nevertheless very important. Thanks to all members of this committee and in particular to its chair Han Brunner for their relentless efforts to provide us with such excellent scientific program, outstanding prize and award winners. With the support of the professionals, Jantje de Roos and Jerome del Picchia, we are guaranteed of a pleasant, well organized and none the least a profitable meeting.

Our European Journal is still growing in quality and number of submissions, despite getting tougher on acceptance of manuscripts. At a time when new journals of all kinds are appearing on the market, our journal has secured a good reputation and is climbing the ladder of the IFs steadily. Thanks to our chief editor Gertjan Van Ommen, the section editors and all the reviewers.

The PPPC and the ad hoc Patenting and Licensing Committee has continued its very important work of the past with new documents, guidelines and recommendations already published or in press. Their respective chairs, Martina Cornel and Gert Matthijs as well as all the members of these committees are successfully continuing a long tradition in the society.

The Educational Committee has been very instrumental in making the second DNA day a success in Europe. New courses have been started and consensus documents on the core competences for geneticists and other health professionals prepared in collaboration with the EuroGentest NoE have been drafted and approved. We are grateful to its chair, Domenico Coviello, and the committee members for making education again one of the primary responsibilities of the society.

As the society was becoming progressively recognized as an influential entity in Europe on matters pertaining to human genetics, it became necessary to set up appropriate committees to improve the professional status and recognition of all those involved in providing genetic services, whether they are clinicians, laboratory directors or counselors. As a result three new committees were set up who already achieved substantial progress. A major breakthrough was achieved by the members of the clinical/medical genetics accreditation committee in bringing the recognition of the clinical specialty close to approval at the European level. The combined efforts of Ulf Kristoffersson, John Burn and Milan Macek, resulted, with the support of the UEMS (Union of Medical Specialists) to put the recognition of the clinical specialty on the agenda of the European recognition committee. This is a major achievement. A decision from this committee is expected in June. We are crossing our fingers.

Finally, the society decided that, in view of the increasing necessity to improve the quality of the genetic services in Europe and the efforts already made in this regard by the EuroGentest NoE, that a 'Genetic Services Quality Committee' was becoming a necessity to coordinate these efforts. Representatives of all aspects of quality management of the services, including EMQN, CF Network, ERNDIM and CEQA, the different European EQA providers, are members of this committee chaired by Ros Hastings. The committee has already been quite active as you may read in its report.

In modern times communication using all possible tools is of the utmost importance. The website, under supervision of a communication committee chaired by Helena Kääriäinen, the electronic and printed newsletters with Lina Florentin as chief-editor have been instrumental in providing more regular information to our members

The meeting of the presidents of the national societies at the annual genetics conference, started at the initiative of our past president PierFranco Pignatti 5 years ago, has become one of the most important initiatives for our society. Never have the interactions with all societies been so intense and so fruitful. Indeed, the number of official members of the ESHG may be relatively limited, but the platform of presidents represents thousands of geneticists, who -through this- can voice their opinion and benefit from our activities, while increasing the impact of the society.

None of all this would have been possible without a very active board, managed by an impressively active executive board. Looking at the decisions made at the strategic meeting of the society in 2006, we can say that most if not all decisions taken there have already been realized. The executive officers usually prefer to remain in the shadow, but the number of meetings and e-mail exchanges on diverse issues has been quite impressive. I have been very fortunate to work with such dedicated people. Nevertheless, I have to single out one person, who has been very instrumental in coordinating all these activities and bringing the society progressively into a more professional structure and a way of functioning, without losing its conviviality; I mean Jerome del Picchia, our chief- executive officer.

The society is definitely on the way to become the undisputed representative of human genetics in Europe. Our colleagues from other continents have repeatedly expressed their admiration for the way we work. It is therefore with confidence that I can pass the baton on to our new president, Dian Donnai, who has all the human and scientific qualities to move the society further on the road to success.

Jean-Jacques Cassiman
President of the ESHG

Secretary General's report



*Professor Helena Kääriäinen,
Helsinki, Finland*

The ESHG Conference in Barcelona 2008 was the biggest ever: there were over 2400 participants. The 2009 Conference in Vienna is likely to be somewhat smaller in terms of number of participants (as the NHGS of Austria is much smaller when compared to its sister organisation in Spain) but in terms of the program it looks truly fascinating! All the work that European and other geneticists have been doing in the most diverse branches of genetics seems to be coming closer to each other as their shared goal, translating all this knowledge to the benefit of patients and healthcare, starting to become reality as shown by a growing number of examples. So all the things that we have been working with were not (only) weird stamp collectors' work! After all, there was sense in collecting rare families, defining phenotypes, searching for genes and mutations, building biobanks and solving the mysterious pathways from genes and to phenotypes!

While I write this, the world's economic situation has been extremely critical for already over half a year. We are threatened by a new type of influenza which has the potential of leading to a pandemic disease. The news about ecological problems, wars and violence seem to be in our newspapers every day. ESHG has to ask itself: do we have a global perspective to what we are doing? Do we understand the real priorities within genetics, medicine and human life in general?

Big changes and threats are also possibilities. In some way, they will help us to analyze our work and to better explain to ourselves and others what makes the work of European geneticists important. It may also help us to identify fields where our research is probably not so important or where our clinical work could be more efficient. We geneticists have always known the strength and joy of collaborating with colleagues all around the world but economic crisis accentuates the need and usefulness of such collaboration.

As an organisation, ESHG has become an active player in the discussion of issues around genetics and the society. To mention some of ESHG activities this year, ESHG has participated in the discussion of genetic testing of minors, over-the-counter testing, and need for counselling in genetic testing situations. Also, ESHG has actively commented on the EU draft recommendations concerning rare diseases. As ESHG has grown and as its conferences are attracting more and more geneticists, it also promotes collaboration.

ESHG has an active Board, a very hard working Executive Board, skilful and alert Executive Officer and a growing membership representing all fields of genetics. In the meeting of its membership (always on Sunday evening during the Conference!) you all have the official opportunity to tell us your ideas and ask your questions. Less officially, you can always email ESHG (see Information – Contact on www.eshg.org) and your questions or comments will be sent on to the appropriate Board or Executive member for further discussion.

Report from the Education Committee

Dear ESHG members,

The production of the first European document on “Core competence in genetics for health professional in Europe” has been the first step toward the dissemination of genetic education in the enlarged EC. During the last year different actions have been taken and others are in progress:

1) some countries have updated the national curricula starting from this document and in others, where genetics is not yet a medical specialty, has been useful to support the debate among professional. ESHG has been very active on this topics writing support letters directly to the government of specific countries and to the EC parliament;

2) the first official meeting between the Union of European Medical Specialties (UEMS, <http://www.uems.net/>) association and ESHG was held in Brussels on January 21st 2009 and Education Committee presented the work done and the possible future collaborations. The meeting was co-organized and chaired by Ulf Kristoffersson and Helen Kingston;

3) new courses were planned, entirely organized or partially supported by ESHG:

- the course on “Genetic Epidemiology of Human Diseases” (November 3-7, 2008) in Paris was very successful with 46 participants,

- in conjunction with “The MediMedGen” (Mediterranean Medical Genetics Meeting) 28-29 June 2009, Bilkent, Ankara, Turkey, will be held the Course “Medical genetics and genomic analysis in isolated and consanguineous populations” (30 June-1 July 2009),

- ESHG/LSHG courses for laboratory medical geneticists “Translating genomics into the clinics” (29 May – 1 June, 2009) will be held in Vilnius, Lithuania,

- the course “Counselling skills for genetic counselling “Training the trainers’ in a European context” (September 2009) will be held in Vienna;

4) interactions with other professional associations or other European projects were carried out:

- participation at the Meeting “New Frontiers in Evidence Based Psychology” Cluj-Napoca, Romania, 7-9 November 2008, for an invited lecture on genetic education. The interest of this group of professionals (psychologists) was on genetic counselling. At this University a master on genetic counselling will start in October 2009;

- participation at the final meeting of PHGEN project: “Public Health Genomics in Europe” (<http://www.phgen.nrw.de/>), in Istanbul, November, 26th - 28th 2008. The future interest of this group of public health professionals is on collection and production of European guidelines in this field; in September 2009 will be the first meeting of PHGEN2 project.

- partnership in the EC project EUROGENE: several meeting have been held to contribute to this project (www.eurogene.org). A document on quality criteria of education material has been produced. A more detailed description of the project is reported in this issue.

5) the European Network of Genetic Nurse and Counsellors (ENGNC) activity is progressing very well with the following action:

- Acquired a domain name for a new webpage for the ENGNC, this will be linked to ESHG website

- Been working on the database of members



*Domenico Coviello, MD, PhD
On behalf of Education
Committee*

- Collected information about current education of genetic nurses and counsellors in different countries as a basis to develop a draft proposals for recommended education of genetic nurses and counsellors in Europe

- Conducted a survey of genetic nurses and counsellor practice in 11 countries. This has been accepted as a poster for Vienna meeting of ESHG

6) The DNA Day in Europe launched last year has been quite successful! The second edition is in progress thanks the precious contribution of Celia Dawn DeLozier and Jerome del Picchia.

Finally I would like to thank all the Education Committee members and collaborating members that have made all these progress possible, and I encourage all of you, member of ESHG, to send us comments, ideas, updates from your country, including your willingness to collaborate with the Education Committee, to help us to set up priorities and to share your experience.



Group picture of participants at the Meeting "New Frontiers in Evidence Based Psychology" Cluj-Napoca, Romania, 7-9 November 2008.

ESHG-EDUCATION COMMITTEE

Members:

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Liaison members:

Alastair Kent, (Patients Organizations, GIG and EAGS), alastair@gig.org.uk
 Celia DeLozier (ASHG – Education Committee)

Sub Committees Chairpersons:

- Accreditation Committee for Clinical/Medical Geneticists (Chair Ulf Kristoffersson, ulf.kristoffersson@med.lu.se)
- Accreditation Committee Genetic Nurses/Counsellors (Co-chairs Heather Skirton, heather.skirton@plymouth.ac.uk, and Christine Patch, christine.patch@gstt.nhs.uk)
- Accreditation Committee for Laboratory Geneticists (Chair Jacques Beckmann - Switzerland, jacques.beckmann@chuv.ch)

Working Groups Coordinators

- E-learning (Francoise Clerget-Darpoux)
- Public Health Genomics/Community Genetics (Martina Cornel)
- ESHG courses (Peter Farndon)

Report from the Genetic Nurse and Counsellor Accreditation Committee

During this past year, the group has grown in numbers and enthusiasm. We currently have 81 members from 24 countries.

The interaction takes place between members via a monthly email newsletter, as well as informal contact between members on a range of issues between newsletters. This is important as one of our aims was to establish a means of supporting practitioners who have few colleagues in their own countries.

The three Working Groups have been active.

The Database group led by Vigdis Stefansdottir has established a new webpage for the European Network of Genetic Nurses and Counsellors linked to ESHG and EuroGentest webpages. This is very recent and we have to populate the page with material. We also have a means of collecting data on members online to make the database of members more robust and useful.

The Professional Standards and Educational Standards Groups have both been collecting data on the situation in various countries prior to developing draft proposals on standards for comments by all the members.

Cristophe Cordier has led a pilot survey to collect data on the practice of genetic nurses and counsellors in 11 countries and these data are being presented in a poster at the Vienna meeting.

We plan to meet at the Vienna conference of the ESHG to plan further working meetings in the next 6 months. We aim to have the draft Educational Standards and draft Professional Standards documents ready to present to the ESHG by the end of 2009.

Professor Heather Skirton, Chair, Sub-Committee for Accreditation for Genetic Nurses/Counsellors

Report of the Public and Professional Policy Committee (PPPC)

During the year 2008-2009 the members of the PPPC were: Anne Cambon-Thomsen, France; Martina Cornel, The Netherlands (chair); Thoas Fioretos, Sweden; Francesca Forzano, Italy; Shirley Hodgson, UK; Gyorgy Kosztolany, Hungary; Jan Lubinski, Poland; Christine Patch, UK; Jorge Sequeiros, Portugal; Aad Tibben, The Netherlands; Lisbeth Tranebjaerg, Denmark; Veronica van Heyningen, UK. A background document and recommendations on genetic testing in asymptomatic minors were open for discussion by the ESHG membership in the summer of 2008, approved by the board and (advance online) published in the EJHG in 2009.

Direct-to-consumer genetic testing was discussed, and a viewpoint contribution to EJHG was written by three PPPC members (Patch et al. 2009), calling for debate on strategies to balance pros and cons. If the possibility of using the discoveries from genomic science to improve health is to be realised without losing public confidence, then improvements in the evaluation and mechanisms for control of supply of tests may be as important as the science itself.



*Professor Martina Cornel
Chair of the PPPC*

A third activity on genetic testing in common disorders (susceptibility testing) progressed, especially in a workshop with genetic epidemiologists in Amsterdam in autumn 2008. Documents will become available for comments by the ESHG membership in spring 2009.

In April 2009 the European Parliament was voting on amendments to the Council recommendation on a European action in the field of rare diseases. A letter by the PPPC was distributed to all members of the European Parliament, calling for a vote against amendment 15, on the “eradication of rare disorders” by genetic counselling and pre-implantation selection of healthy embryos. The PPPC reaction reconfirmed that the professional stance on reproductive choices related to genetic disorders departs from autonomy as the central ethical principle. Non-directive counselling should help people to make choices consistent with their own values and on the basis of adequate information. “Eradicating” rare diseases by stimulating certain reproductive choices as a public health strategy is not acceptable from the professional standard of human geneticists.

Report from the Scientific Programme Committee



*Han Brunner
Chair of the Scientific Programme Committee*

The Scientific Programme Committee for 2008-2009 was composed of Han Brunner (chair), Thierry Frébourg, Peter Heutink, Raquel Seruca, Andrew Wilkie, Brunhilde Wirth, Olaf Riess, Eduardo Tizzano, Hans-Christoph Duba (Local Host), Florian Kronenberg, Draga Toncheva, Batsheva Kerem, Peter Scambler, Miikka Vikkula, Inge Liebaers, Mariano Rocchi, Mark McCarthy, and Cisca Wijmenga. Helena Kääriäinen participated as observer from the executive board.

The SPC met twice to organize the Vienna 2009 ESHG conference: in June 2009 to decide on the plenary sessions and symposia, and in Vienna at the VMA offices in March 2009, to select the abstracts for oral presentations and posters.

This year the number of outstanding abstracts has again increased and this allowed the selection of 108 abstracts for 18 concurrent sessions, as well as a "What's new session" on the first day of the meeting.

The ESHG Education award 2009 will be presented during the opening session to Dr Albert Schinzel in recognition of the enormous impact on generations of clinical geneticists and cytogeneticists from all over Europe who were given inspiration as well as information through his books, lectures and courses.

As is usual, one highlight of the final day of the conference will be the acceptance speech by our ESHG prize winner. This year, the ESHG award 2009 will be awarded to Kari Stefansson (Reykjavik) for his creation of an internationally renowned institute which has contributed greatly to elucidating the complex genetics of common human diseases, and also to the development of methods, approaches and tools that allow the scientific community in Europe and beyond to study the genetics of such diseases effectively.

For the final session on Tuesday 26 May, we were able this year to attract professor John Burn from Newcastle to give a special lecture on "The future of genetic medicine". Professor Burn has served as past president of the society. He is the creator of the Newcastle Centre for Life which serves as a model world-wide for the integration of genetic science, the application of genetic knowledge in the medical setting, and an open and interested debate with the public about the societal impact of genetic innovations, and genetic thinking.

After the Barcelona conference, the SPC shall have to say goodbye to Raquel Seruca, Andrew Wilkie, Thierry Frébourg. We thank them for their work and their dedication to making the meeting better. After the 2009 ESHG meeting, Brunhilde Wirth will serve as chairman of the Scientific Program committee. Han Brunner and Brunhilde Wirth shall share the chairmanship for the 2010 Gothenburg meeting, and from 2011 Brunhilde will be the sole chair of the committee. Brunhilde has in the past shown to be an outstanding scientist with a broad view of the entire scientific field of human genetics and excellent leadership skills. We wish her well.



ESHG Genetic Services Quality Committee

News from this new Committee



*Dr. Rosalind Hastings
Chair of the Genetic Services Quality Committee*

The Genetic Services Quality Committee held its inaugural meeting in June 2008 during the ESHG Conference in Barcelona. The Committee meets biannually in December and during the ESHG Conference. The Committee is informally referred to as the Quality Committee (QC) 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;
- To commission and approve new documents relating to quality in genetic testing;
- Give recommendations for those countries where no guidance is currently available.

The QC recognizes the need to publicize the role of quality management through the ESHG newsletter. There are several initiatives such as Eurogentest, OECD guidelines, EMQN, ERNDIM and CEQA that have publicized the need for quality management in genetic laboratories. The OECD guidelines for Quality Assurance in Molecular Genetic testing are also applicable to Cytogenetics and Biochemical genetics. The Quality Committee encourages all laboratories performing genetic testing to adhere to the OECD guidelines and ISO 15189 standards as well as to become accredited.

Since February 2008, laboratories have been able to submit their quality assurance data through the Orphanet 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. It is a requirement for any accredited laboratory to participate continuously in EQAs applicable to their diagnostic service. There are four European EQA schemes, open to all laboratories in Europe – CEQA (Cytogenetics), CF Network (Molecular Genetics), EMQN (Molecular Genetics) and ERNDIM (Biochemical Genetics). The QC has agreed to provide a governance structure for these EQA schemes and review these four EQA schemes' annual management reports. The content of the annual management report has been agreed by the QC. In addition, the scoring criteria and definitions of satisfactory performance will be discussed in future meetings of the Quality Committee.

The Quality Committee has identified nine areas of need relating to quality issues in the genetics community. Once these needs have been prioritized, the Committee hopes to establish working groups and a timetable in which to take them forward. One request already submitted to the QC was to endorse the Fragile X Reference Materials panel for use as a positive control for genetic testing or validation of a new technique. Following a letter of endorsement from the QC, the WHO approved the Fragile X Reference Panel. The QC has since written a letter of endorsement to the WHO for a second reference materials panel for Prader Willi/Angelman syndrome.

Earlier in 2008, the QC proposed that a separate annex to ISO 15189 for genetic laboratories was not required, as ISO 15189 adequately covers the genetic testing laboratories' needs. This proposal was relayed to the ISO Committee by one of the QC members and as a consequence, no separate annex for genetics has been agreed. The ISO 17043 (Conformity assessment-General requirements for proficiency testing) will be released for consultation soon and will be of interest to EQA providers.

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 last two meetings are available on the ESHG website.

Committee Members: David Barton; Jacques Beckmann; Mireille Claustres; Els Dequeker; Rob Elles; Peter Farndon; Brian Fowler; Claude Giroud; Ros Hastings (Chair); Viktor Kozich; Lidia Larizza or Konstantin Miller; Cor Oosterwijk and Orsetta Zuffardi.

Eurogentest Report - The 1st EuroGentest Symposium on Quality & Laboratory Management

EuroGentest, the EU-funded Network of Excellence, is active in many aspects of quality assurance and improvement in genetic testing.

Since 2005, EuroGentest has run interactive training workshops, addressing many topics related to quality management in diagnostic laboratories, with the aim of aiding laboratories in the process of developing and improving quality systems and working towards accreditation. The workshops have also contributed to the harmonization of the approaches to accreditation and to create a network of laboratories with a common goal – improving the quality of their services.

To complement the interactive workshops, the first EuroGentest Symposium on Quality & Laboratory Management will be held in Leuven, Belgium, on June 18-19, 2009.

Selected speakers from laboratories, accreditation bodies and companies active in quality management from 9 countries from Europe, North America and Australia will address topics such as laboratory quality management, external quality assessment and accreditation in medical laboratories. While the Symposium consists principally of formal presentations, round-table sessions will allow interaction between the speakers, experts and participants.

A EuroGentest quality award will be presented to a laboratory or organization in recognition of a particular contribution to awareness of quality management in genetic testing. All laboratories and participants are encouraged to submit a proposal concerning their experience, idea or improvement. Nominees will be able to present their proposal during the symposium and one will receive the EUGT Laboratory Quality Award 2009 and a free registration for one person to participate in a EuroGentest workshop on accreditation.

The target audience includes laboratory directors, quality managers, technicians, scientists, researchers and other people working in a genetic testing laboratory (molecular, cytogenetic and biochemical). In addition, accreditation bodies,

EQA providers and other active in laboratory quality assurance are welcome. At present, participants from over 15 countries are registered; further places are available.

Programme and registration information is available on the Symposium web site at <http://www.eurogentest.org/QualitySymposium/index.xhtml>

Prof. Elisabeth Dequeker, Sarah Berwouts, Dr Michael Morris

Report from the Clinical Genetics EU Recognition Committee



*Dr. Ulf Kristoffersson,
Chair, Lund May 2009*

The committee has been very active through the Multidisciplinary Joint Committee - Clinical Genetics (MJC) of UEMS (WWW.UEMS.net). The educational programme for medical doctors' specialisation in clinical genetics has been endorsed by the Boards and Sections' Meeting in February and finally by the UEMS Council in April this year.

In parallel, Milan Macek of Prague has been very active during the Czech presidency of EU in order to enforce a recognition, in conjunction with the Recommendations on Rare Diseases that will be decided on by the ministers later this spring. Hopefully his work will bring our efforts closer to our task which an EU recognition of Clinical Genetics as a medical speciality.

The committee will continue to work close with the MJC, who now will disseminate information about these guidelines to the national professional organisations and their section of clinical genetics in order to form a network of national contacts.

Proposal for a "EU Council recommendation on a European action in the field of rare diseases" and the amendment of the Directive 2005/26/EC with clinical-/medical genetics



*Professor Milan Macek
Jr. M.D., D.Sc*

Rare diseases and clinical- / medical genetics

EU has, together with the European Medicines Agency (<http://www.emea.europa.eu/>), defined a rare disease as one which affects fewer than 5 people per 10,000. Given the overall EU population size the number of sufferers is high, since there are over 7,000 known rare diseases. Most these diseases are due to defined genetic defects, but environmental exposure during pregnancy or later in life, often in combination with genetic susceptibility, account for another common cause. A subset of these diseases comprises also rare complications of common diseases. While first symptoms may be detected at birth or in childhood, more than 50% of rare diseases appear during adulthood, and are often life-threatening or progressive and debilitating. Usually there is no effective treatment, but early diagnosis, followed by suitable medical and social care, can improve quality of life and life expectancy of those affected.

Although clinical-/medical genetics plays a crucial role in early diagnosis and management of rare diseases it has not been included in the list of "official" EU medical specialties listed in its "Directive 2005/36/EC of the European Parliament and of the Council from September 7, 2005 on the Recognition of professional qualifications" (<http://eur-lex.europa.eu/LexUriServ/LexUriServ.do?uri=OJ:L:2005:255:0022:0142:EN:PDF>). Inclusion of a speciality in this Directive assures free mobility of respective specialists within the EU by acknowledging their qualification achieved in a given member state at the EU-wide level, thereby permitting them to work in a EU member state of their choice.

EU Council recommendation on a European action in the field of rare diseases

Rare diseases constitute a serious public health concern and are considered a priority in the EU health and research programmes (http://ec.europa.eu/health-eu/health_problems/rare_diseases/index_en.htm). Following the very successful "Public Consultation" in which amongst others many clinical-/medical geneticists have voiced their opinion on a set of questions regarding improvement of diagnosis and care for rare diseases the European Commission published on November 11 / 2008 its "Communication from the Commission to the European Parliament, the Council, the European Economic and Social Committee and the Committee of the Regions on Rare Diseases: Europe's challenges {SEC(2008)2712; SEC(2008)2713} (http://ec.europa.eu/health/ph_threats/non_com/docs/rare_com_en.pdf).

Essentially, this important document stipulates set of objectives and priorities related to a European action in the field of rare diseases which will be supported by the European Commission. According to the principles by which the EU is operating the Commission "Communication" should be accompanied by a subsequent EU Council "Recommendation", which lists priorities and objectives on which individual EU member states consensually agree upon and which ought to be practically implemented. Although a Council Recommendation is not legally binding it sets the frame for national

actions and represents a “background document” to which professional societies can refer to when pressing for national initiatives in a given area.

The Czech EU Council Presidency, for which I am serving as its scientific advisor for rare diseases, (www.eu2009.cz; January 1 – June 30/2009) has “inherited” from the previous French Presidency (www.eu2008.fr) a draft Proposal “EU Council recommendation on a European action in the field of rare diseases” (http://ec.europa.eu/health/ph_threats/non_com/docs/rare_rec_en.pdf). This document has already underwent four “examinations” of its text by EU Public Health committees at the EU Council in Brussels. It will most likely be adopted by the upcoming meeting of the Employment, Social Policy, Health and Consumer Affairs Council (EPSCO) in Luxemburg (June 8-9/2009) by EU27 health ministers. The Czech EU Council Presidency is doing its best to meet this deadline and to assure a smooth ratification process. Adoption of the Council Recommendation will be of great importance not only for rare diseases, but also for clinical-/medical genetics. This document in its Recital 15 states, inter alia, that „expertise should travel rather than patients themselves“. This statement provides the rationale for amending Directive 2005/36, since obviously our specialty is crucial for diagnosis and management of the majority of rare diseases. Furthermore, if clinical-/medical geneticists are to travel in order to provide their expertise in other EU member states, our specialty has to be included in this Directive. Thus, we are in fact in a rather unique position to achieve this otherwise politically and practically difficult goal, since this Directive has not been amended since 2005!

Union of European Medical Specialists consensus document on postgraduate training

Another critical piece of work, that has facilitated the process of amendment of Directive 2005/36, was done by prof. Ulf Kristoffersson (Lund) who has represented our field in the Joint Interdisciplinary Committee of the European Union of Medical Specialists (www.uems.net). During the last several years and after many meetings, negotiations and/or presentations of a draft clinical-/medical genetics “consensus” postgraduate curriculum to representatives of other medical disciplines the UEMS has adopted the consensus document “Description of Clinical Genetics as a medical specialty in the EU: aims and objectives for specialist training” (2009/15; on April 25/2009). This document describes the profile, entry criteria, educational goals and most importantly the consensual time frame for specialist training in our specialty of 4 years. This document was already provided to the EC - DG Internal Market and Services, that is responsible for the amendment of Directive 2005/36. This Commission department follows upon recommendations of its so called “Group of Coordinators for Recognition of professional qualifications” (or “Recognition Committee”) that convenes about 4 times per year and represents views of respective national authorities on this subject. This group mostly comprises representatives of national ministries for e.g. European affaires, Health, Research or Education who vote on the recognition of a given specialty by a proportionate vote, i.e. representatives of large EU member states have the strongest leverage. Usually, requests for amendments of Directive 2005/36 for a certain specialty are issued either by individual member states representatives or more commonly by representatives of the current EU Council Presidency. In this respect full credit goes to prof. John Burn (Newcastle) and prof. Arnold Munnich (Paris) who visited the French Minister of Health for an informal breakfast in November 2008 and requested that French EU Presidency launches an official request to the Recognition Committee to include our specialty. Arnold and John were successful (!) and the DG Internal Market has received the official document “French request for inclusion of specialty of Medical Genetics under Annex V” for its March 26 / 2009 meeting. Among others the request stated: “Concerning the specialty of Medical Genetics, the French authorities wish to address the question of its existence and of its content in the other countries of the European Union in the Committee of Directive 2005/36/EC in view of its inclusion, if necessary, in the list of those specialties which can benefit from mutual recognition, insofar as at least 2/5 of the Member States would already recognise this specialty. In France this is a speciality sanctioned by a specialised diploma (diplôme d’études spécialisées – DES), issued by the universities. You will find in Annex a sheet recapitulating the activities concerned and the duration of training“.

Czech EU Council Presidency activities aimed at EU-wide recognition of clinical- / medical genetics

The Czech EU Council Presidency followed upon this initial request in that it was up to me to prepare, together with the Czech Ministry of Education representative in the Recognition Committee Ms. L. Slobodová, an overview regarding the status of our specialty in EU27. Members of the Committee are mostly interested in a consensus EU postgraduate curriculum (ref. UEMS), consensus on the duration of training (in our case 4 years – ref. UEMS), collection of legal dossiers regarding the recognition of this specialty in individual member states (we got them all in the “pdf” format in national languages), whether clinical- / medical genetics is a primary or secondary specialty (subspecialty), including other relevant details and contacts on representatives of national professional specialties under whom our field belongs. In this respect I have contacted all presidents and/or other representatives and got thanks to their prompt response all necessary data for the March 26 meeting in Brussels. I have received a lot of supportive documents from prof. Ulf Kristofferson and from our Spanish colleagues (Drs. Feliciano J. Ramos and Ismael Ejarque Doménech) who have been struggling to have clinical-/medical genetics recognised in Spain.

The Czech delegation has presented the Recognition Committee with an overview table concerning the status of our field in the European Union. Hereby, we comply with the provision that our specialty first must be recognised in 2/5 of the EU27. With the exceptions of Belgium, Estonia, Greece and Spain where recognition process is currently under way, Cyprus and Luxemburg where medical genetics is not recognised and no initiatives to change this situation have been launched, our specialty was otherwise “officially” recognised by all national authorities. Furthermore, with the exception of Hungary our specialty is a primary specialty, which substantially increases our chance for a EU-wide recognition. Interestingly, term “clinical genetics” is used in 9 cases, “medical genetics” in 10 instances, once either term “genetics”

(Malta) or “human genetics” (Germany) are used within official legal dossiers. We had to explain to the Recognition Committee that all these terms are synonyms for the same specialty, and the UEMS in the end was more in favour of using the term “clinical genetics” for the description of our specialty. Although these differences in “semantics” may sound negligible to us, it took some effort to explain them outside of our domain and to the members of the Recognition Committee.

With respect to the duration of training eight countries require 5 year long postgraduate training, while the majority settled on 4 years of postgraduate training in our specialty. Usually the “extra” year, i.e. beyond the 4 year “consensus” adopted by UEMS, comprises one year of internship in a medical field closely related to clinical-/medical genetics. The UEMS document clearly accounts for this by stating in its last paragraph dealing with the “Time frame for specialist training, inter alia: In the longer training period, up to one year could be in another speciality of importance for clinical/medical genetics“. Thus, the main issue that needs to be resolved at the moment is to receive “endorsements” from clinical-/medical genetics professional societies, where the curriculum is set for 5 years, in that they will accept professionals from countries where the curriculum is limited to 4 years.

Currently, we are preparing these documents for the next Recognition Committee which is scheduled for June 22 /2009. I will inform you about the most recent status of this initiative at the upcoming 5th Meeting of National Human Genetics Societies at EHGC 2009 in Vienna.

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UEMS 2009 / 15

Description of Clinical Genetics as a medical specialty in EU Aims and objectives for specialist training

Endorsed by: The European Society of Human Genetics board and membership (2007) The UEMS Multidisciplinary Joint Committee for Clinical Genetics (Jan 21, 2009) The UEMS Specialist Sections & European Boards (Feb 21, 2009)

Adopted by: The UEMS Council (April 25, 2009)

Specialty Profile

Clinical Genetics describes the medical elements of Genetics Services provided to individuals and families (and sometimes populations). Other components include

laboratory genetics (cytogenetics, molecular genetics, and biochemical genetics), genetic counselling and academic genetics. The core activities of a genetic service can be defined as ‘integrated clinical and laboratory services, provided for those with/concerned about a disorder with a significant genetic component (both inherited and sporadic). Due to the sharing of genes among family members, the whole family, not only the individual, represents the core patient in clinical/medical genetics.

This document relates to medically qualified individuals intending to train in the specialty of Clinical/Medical Genetics. It recognises that there may be overlaps with training programmes for other genetic professionals (scientists and counsellors) and that there may be opportunities for joint training for periods of the course.

Entry criteria

This may vary from country to country but would generally include a specified period of general medical training to include adult +/- paediatric medicine prior to commencing specialty training in Clinical Genetics, “internship”. Some countries may have a minimum period of training to be undertaken before specialisation.

Educational goals

Knowledge and Skills

- **Theoretical genetics/Basic Science** which may include
 - understanding cellular and molecular mechanisms that underpin human inheritance,
 - understanding patterns of inheritance and methods for risk assessment,
 - genetic epidemiology and biostatistics
- **Clinical/Medical knowledge and skills**
 - Pedigree construction.

- Diagnosis, investigation and genetic management of individuals with both common and rare inherited/ genetic diseases and their families.
- Risk assessment and role in genetic testing.
- Paediatric genetics including training in Dysmorphology (knowledge of common dysmorphic syndromes, their aetiology and the use of dysmorphology databases) and investigation of learning disability in children.
- Adult genetics to include knowledge of late onset disorders and disorders with a significant genetic component presenting in adult life (including predictive testing).
- Prenatal Genetics and knowledge about fetal development and teratogens
- Population genetics, including genetic screening programmes
- Special areas of genetics including
 - Inherited metabolic disorders
 - Neuro- and neuromuscular genetics
 - Cardiovascular genetics
 - Cancer genetics
 - Neurosensory genetics (visual and hearing conditions)
 - Pharmacogenetics
 - Other subspecialties of specific interest to the trainee
- **Genetic counselling and communication skills**
 - Training in genetic counselling for all types of genetic disease and situations encountered in clinical genetic practice. This includes counselling in relation to prenatal diagnosis and for late onset such as neurogenetic and cancer genetic disorders, including predictive testing. Where applicable, training in cocounselling with other professionals such as genetic counsellors.
 - Understanding ethical issues and importance of consent and confidentiality.
 - Development of good communication skills with patients, colleagues in genetic centres and other specialists and healthcare professionals, including understanding and handling of crisis reactions.
- **Laboratory skills**
 - Thorough knowledge of principles of laboratory techniques used in diagnostic testing
 - Interpretation of results from cytogenetic, molecular genetic and biochemical genetic analyses.
 - The time spent and the practical expertise gained in laboratory work may vary between countries, but sufficient to ensure highly specialised knowledge.

Other aspects of the Training Programme

- **Maintaining Good Medical Practice**
 - Develop a commitment to lifelong learning through continuing professional development and attend relevant courses and conferences.
 - Participate in Audit and Clinical Governance
 - Adhere to established consent and confidentiality procedures
 - Understand ethical and legal issues
- **IT skills**
 - Use of information technology including online resources and databases
- **Management training**
 - Knowledge about general healthcare policy, goals and priorities
 - Understanding the organisation of genetic services
 - Opportunities to participate in departmental activities related to organizational planning, financial management, and monitoring and maintaining quality standards
 - Development of multidisciplinary team working and leadership skills
- **Teaching**
 - Develop teaching skills by participating in the education and training of various categories of staff
 - Involvement with patient groups and patient education
- **Supplementary Education and Training**
 - Subspecialty training: some trainees will elect to develop expertise in a subspecialty area such as cancer genetics, dysmorphology or neurogenetics.

Quality Assurance

- Competency-based curricula should form the basis of a training programme.
- A written agreed curriculum for the training period should be set up as a contract between the trainee and the supervisor if not otherwise determined by national regulations.

- Trainees should maintain a Training Log including details of clinical and laboratory experience, educational activities, research and publications.
- A mechanism should be in place for continuous assessment of trainees against agreed quality standards. Some countries will have a nationally prescribed system for assessment and certification.
- Specialist examination may be compulsory in some countries

Research

- Medical genetics has a rapidly changing knowledge base and during specialty training the clinical/medical geneticist should be encouraged to participate in research. Some trainees will wish to take time out from the clinical training programme to undertake an intensive period of research leading to a higher academic degree. On completion of training some academic clinical/medical geneticists will continue to lead research programmes whilst many others will collaborate with laboratory based colleagues in the genetics team.

Time frame for specialist training

- The training period should minimum 4 years full time work; part time work would extend the training period.
- An educational training programme will be agreed for each trainee according to the specialty specific curriculum • In the longer training period, up to one year could be in another speciality of importance for clinical/medical/medical genetics.
- The time spent in laboratory work may vary between countries according to national curricula.
- A period of research resulting in a PhD/other higher exam may, if appropriate, replace training for a variable period of time according to national guidelines. However, in absence of national guidelines, it is not recommended that this time period is longer than 1/3 of the total training period

Editorial Report for EJHG over 2008



*Dr. Gert-Jan van Ommen
Editor in Chief*

- An important hurdle was made in 2007. Our factor impact increased by 0.306 points, to 4.003. Its ranking in the category of Genetics and Heredity also improved by 2 points, from 40 to 42.

- Submissions increased by 12.5% in 2008, while the acceptance rate has remained stable for the past three years, at 35%. EJHG published 10% more articles in 2008 than in 2007. However, in the first months of 2009 we have already seen a further rise in submissions, most likely driven by the IF increase. To cope with this we will have to raise our acceptance bar - which of course should further increase our IF.

- EJHG authorship is still predominantly European, with 70% of accepted articles. However, US/Canadian and Asian authorship keeps increasing, to 17% and 5% in 2008, while the rest of the world contributes.

- Decision times remained low for EJHG, with a median first decision of 20 working days and the median final decision time of 18 working days after submission of the last revision. We note that these are median figures and we are aware that specific manuscripts have had much longer processing times. This is mainly due to the fact that with the increasing amount of genetics journals, it has become more difficult to solicit reviewers and actually have them return their reviews on time. We will do all that is in our powers to address this,

amongst others by a 30% extension of the editorial time commitment.

- Due to the increased submissions, time to print publication increased throughout 2008, from 3.8 months in January to 5.4 months in December, which we aim to reduce by the increased editorial office input.

- EJHG performed especially well online in 2008. Seven out of twelve months we published more than 60% of articles as Advanced Online Publication within 25 working days. The average total web page views increased by 59%, home page views by 11%, abstracts by 39% and full-text articles by 33%. The most frequently accessed article lists are quite varied, although they feature mostly recent articles. Practical genetics tend to perform well, especially in terms of PDF downloads. Four of the most cited articles in 2008 were also featured on the most accessed lists. EJHG content accessed from PubMed averaged at almost 10,000 times per month.

EJHG Top Cited Papers 2007-2008

As every year, EJHG has a junior authors' high-citation award, to hand out at the Vienna meeting. The 1st prize includes a € 500 award and places 1-3 receive one year free ESHG membership + online EJHG, and free registration for the Barcelona meeting. This year's winner is Dr. Silverberg et al. with 26 citations in all of 2007 and Jan-March of 2008, for his paper "Refined genomic localization and ethnic differences observed for the IBD5 association with Crohn's disease", which appeared in EJHG 15 no. 3 (2007). Second and third prizes go to Dr. Bronner et al. for "Progranulin mutations

in Dutch familial frontotemporal lobar degeneration”, (EJHG 15 no. 3, 2007, 22 citations), and Dr. Mayans et al. for “TCF7L2 polymorphisms are associated with type 2 diabetes in northern Sweden” (EJHG 15 no. 3, 2007, 14 citations). An EJHG Special Achievements award will also be presented to Dr. Hiekkalinna et al. for “An utter refutation of the ‘Fundamental Theorem of the HapMap’” (EJHG 14 no. 4, 2006, 32 citations).

Medical Genetics in Slovenia



Bled

Slovenia is a small country with approximately 2 million inhabitants.

The history of medical genetics in Slovenia goes back to the period between 1950 and 1960. In 1964 the first article about the Applicability of Chromosomal Analysis in Gynecology was published in the Slovene medical journal *Zdravstveni Vestnik*. In the year 1966, the cytogenetic laboratory was established at the Gynecology Clinic at the University Medical Centre (UMC) Ljubljana and this led to the foundation of the first national Division of Medical Genetics, today’s Institute of Medical Genetics at the UMC Ljubljana.

Genetic services

Today, there are two UMCs - Ljubljana and Maribor, and clinical genetics service is offered in both of them. It includes genetic counseling, and cytogenetic and molecular genetic testing at UMC Ljubljana and genetic testing at UMC Maribor. In addition, there are two diagnostic laboratories in public institutions outside UMCs and one private institution offering genetic counseling as well as one private cytogenetic laboratory.

The access to genetic services is good. The cost of genetic counseling and medically indicated genetic testing is covered by national insurance scheme. The genetic testing is either done at domestic laboratories or in collaboration with laboratories abroad.

Prenatal diagnosis for advanced maternal age is offered for women aged 37 or more at term. Each pregnant woman aged 35 to 37 at term is offered screening test, either combined test (nuchal translucency measurement and double blood test) in the first trimester or quadruple blood test carried out in the second trimester. If the result of screening test is positive, genetic counseling is mandatory before invasive prenatal diagnosis.



Hayrack Golica



Triglav

Research

Much of the research work is done at the clinical genetic departments at both UMCs, but there are also well established research groups outside UMCs, especially at the Medical University of Ljubljana. Genetic research projects are funded by the Slovenian Research agency. Our geneticists also participate in several FP6/7 and DG SANCO research projects.

Professional organizations

Human geneticists are organized through two societies: Slovenian Association of Medical Genetics and Slovenian Society of Human Genetics. Slovenian Association of Medical Genetics, which is organized as part of the Slovenian Medical Society, played a major role in the preparation of a program for specialization in Clinical Genetics.

Education issues:

Human genetics is taught at both Medical Universities. In Ljubljana, medical students have obligatory one semester course in Human Genetics and in Maribor the genetics themes are part of the Molecular Biology course. There are no clinical genetics modules.

Clinical Genetics has been recognized as a medical specialty in Slovenia. Since 2003, two residents have finished the six years program of specialization and two are in training. The program includes training in clinical genetics, cytogenetics and molecular genetics. This year the program was reorganized; it takes now five years. The recommendations for the training for the laboratory geneticists have already been prepared and we are striving to achieve the recognition of Laboratory Medical Genetics Specialty as well. There is no specific training for nurses and genetic counselors.

Quality control:

Since 2004, laboratories have to be authorized by the Ministry of Health to carry out analyses in the field of laboratory medicine (64/2004). Some laboratories take part at the Cytogenetic European Quality



Three Bridges Ljubljana

Assessment (CEQA) and European Molecular Genetics Quality Network (EMQN). The quality control will be the main theme of this year's meeting of Slovenian Association of Medical Genetics and we believe it will raise awareness of the importance of this topic.

Legislation:

Slovenia has signed and ratified the Convention on Human Rights and Biomedicine (http://www.eurogentest.org/web/info/public/unit4/ethical_legal_papers.xhtml#legal_16).



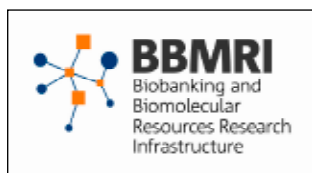
*Karin Writzl, MD, PhD,
President of Slovenian Association
of Medical Genetics*

The Board grants permission for termination of pregnancy if severe fetal abnormalities or inherited disease have been confirmed. No upper gestational limit is set according to the law on Health Measures in Exercising Freedom of Choice in Childbearing Act (11/1977). The act passed in 2000, relates to the Infertility treatment and procedures of biomedical-assisted procreation act. Preimplantation diagnosis is allowed in certain circumstances.



Piran

Biobanking and Biomolecular Resources Research Infrastructure (BBMRI)



Public website: www.bbmri.eu

BBMRI Coordinator: kurt.zatloukal@meduni-graz.at

During the preparatory phase (2008-2010) the EU-funded pan-European Biobanking and Biomolecular Resources Research Infrastructure (BBMRI) project comprises 50 partners and 182 associated organisations. The objectives are to develop the plan to integrate existing quality controlled biobanks, biomolecular resources and enabling technologies into a pan-European biomedical research infrastructure, and provide a concept for its operation and codes of conduct for European biobanks, particularly considering the different technical standards and types of integration into health care registries and databases currently available.

All work packages started their work in February 2008 although funding from the EC only became available in June 2008. This caused an obvious initial delay, however WP1 has recruited an executive manager and set up an executive management team with the coordination office. Due to the large size of the project (currently 232 partners and associated organizations) the management and organizational structure may appear complex, but has shown its effectiveness in practice. An upgraded BBMRI web site (containing public and intranet domains) has been established to facilitate external and internal communication. The other six WPs focus on one hand on the specific requirements of different biobank formats, population based biobanks (WP2) and clinical biobanks (WP3) and on biomolecular resources (WP4), and on the other hand address issues related to the whole infrastructure, such as databases and biocomputing (WP5), governance in ethical, legal and societal (ELSI) issues (WP6), operation, funding and financing (WP7). This work has been and will be performed by work package leaders and partners as well as by external expert groups.

The mission of the BBMRI is to sustainably secure access to biological resources required for health-related research and development intended to improve the prevention, diagnosis and treatment of disease as well as to promote the health of the citizens of Europe. Lack of sustained funding has been identified as a major bottleneck in long-term operation of biobanks and biological resource centres for life sciences and clinical research. Despite major investments into biobanks in various EU member states, coordination of their activities has been limited to individual projects. Thus there is an obvious need for pan-European coordination of efforts and long-term funding schemes. Fully functional distributed pan-European biobank would have a drastic impact on public health, for example, the development of prognostic biomarkers and new therapies for common diseases and their variants, and for the evaluation of the interplay of genetic, environmental and life style factors on disease susceptibility and development.

A New Additional Protocol to the Convention on Human Rights and Biomedicine

The sequencing of the human genome and the development of new technology make human genetics a very dynamic sector. The

very rapid progress in this field has prompted the Council of Europe to focus on the ethical and legal issues raised by applications of genetics, in particular genetic testing, and to draw up legal standards to protect fundamental human rights with regard to these applications. The Council of Europe Convention on Human Rights and Biomedicine (ETS No. 164) sets out a number of principles concerning genetics (Articles 11 to 14), particularly genetic testing and interventions on the human genome. In order to develop and supplement the principles set forth in the Convention, the Council of Europe Steering Committee on Bioethics (CDBI) has elaborated a new Additional Protocol to the Convention on Human Rights and Biomedicine, concerning Genetic Testing for Health Purposes which was opened for signature on 27 November 2008. For the elaboration of this Protocol, the CDBI consulted different experts and used in particular as a basis for approaching certain notions the recommendations of the European Society of Human Genetics. The Protocol covers all genetic testing carried out for health purposes, except genetic testing concerning the human embryo and foetus and that carried out for research purposes. It lays down principles concerning, in particular, the quality of genetic services, prior information and consent as well as genetic counselling. It also covers the protection of private life and the right to information obtained by means of genetic testing. Finally, it addresses the issue of genetic screening.

For the text of the Additional Protocol and its explanatory report see at:

<http://conventions.coe.int/Treaty/EN/Treaties/Html/203.htm>

Towards a consensus concerning Cystic Fibrosis carrier screening

About 20 experts from all over the world met in Garda, North Italy 20-21 March 2009 in an ECFS consensus meeting "Cystic Fibrosis Carrier Screening in Europe: Management, Development, Research", organized by Carlo Castellani, Harry Cuppens and Milan Macek. The aim was to agree upon recommendations related to possible national CF carrier screening programs.

It was generally felt that the issue was extremely complex. CF creates a heavy burden to patients, families and societies, but simultaneously treatment has been and is improving, leading to much better prognosis of the patients. There are no national CF carrier screening programs in Europe but numerous couples are screened as suggested by their gynaecologists, which leads to a non-democratic selection of those screened. Mutational background is very complex and varies between populations.

Because of all these reasons, starting population carrier screening programs is controversial. There was no clear consensus about the usefulness and feasibility of CF carrier screening programs and the decisions have to be made by national health services and based on the local situation.

Regardless, there was a strong consensus that if CF carrier screening programs are instituted, they have to be of high quality and well planned. Educating professionals and the public has to precede the program, and appropriate pre-test information has to be guaranteed to ensure free informed consent. Genetic counselling has to be available and actively offered at least to the identified carrier couples. The laboratories involved should preferably be accredited. The panel of mutations studied initially and, especially in case of spouses of carriers, has to be carefully chosen, based on the most recent research.

The group aims at completing a consensus paper by the end of this year.

National Human Genetics Societies meeting

This year in Vienna we'll be holding the 5th meeting of the National Human Genetics Societies (NHGSs) in Europe. These meetings are held in order to strengthen the collaboration between ESHG and NHGSs.

Last year in Barcelona we had a productive meeting into which 40 national representatives participated, together with 7 from the ESHG board and committees. Information on new ESHG activities was provided to the national representatives, onas how to propose courses, participate in the DNA day contest, award ESHG-sponsored fellowships to come to the European Conference, become members of the web of genetic nurses and counsellors, consult documents of interest in the website as susceptibility testing for common diseases and gene patenting, collaborate for the European recognition request of the title of specialist in clinical genetics, etc, as given in detail in the website at <http://www.eshg.org> under the heading "Genetics in Europe".

This year in Vienna we'll focus again on some of the activities of ESHG of interest to the national societies, offering an update on the above mentioned ones, and we'll ask for the national societies help in emerging issues as direct to consumer genetic testing in Europe. The current and past ESHG presidents will present the state of the Society and future directions, and the two secretaries plus the executive officer will give practical details on electronic and



*Professor Pier Franco Pignatti
(ESHG-NHGSs liaison)*

paper communication, the new committee for laboratory geneticists, course organization and double membership in the national and the European societies. ESHG will be there to take note of the wishes expressed by the National Societies, and we'll all discuss any themes of common interest to human geneticists in Europe. The Agenda of the meeting is posted on the website.

New Board Members elected:

Prof. Agnes Bloch-Zupan, Professor in Oral Biology (Oro-Dental Genetics), at Strasbourg University, Faculty of Dentistry.



I work in a Reference Centre for Oro-dental Manifestations of Rare Diseases, Dental Hospital, Hôpitaux Universitaires de Strasbourg. I participate in the ESHG Education Committee, headed by Pr Domenico Coviello (EuroGentest Unit 6 Patient and Professional Perspectives of Genetic Information/Education in

Europe), in the elaboration of the core competences document for health professionals who are generalists or specialising in a field other than genetics, such as dentists. As a member of the Scientific Editorial Committee of Orphanet for Odontology, I contribute to the enrichment of this tool for the medical/scientific community and the patients. I have also chaired the Genetic Anomalies working group of a European COST action B23 Orofacial Genetics and Regeneration. My research within IGBMC, Illkirch, focuses on the discovery of new genes involved in odontogenesis and the study of animal models displaying orodental defects and involves numerous European collaborations. I am internationally recognised as an expert in Oro-dental Genetics. At the frontier between genetics and dentistry, performing research with a strong interest in education and disseminating knowledge of rare diseases, I am delighted to serve in my competencies as a board member for ESHG.

Anne Cambon-Thomsen, MD (1978) is Director of Research in CNRS



(French National Centre for Scientific Research). She studied at the Faculty of Medicine, University of Toulouse, France and is a specialist in human immunogenetics; she holds

also a masters in Human biology and a degree in Health Ethics. After a post-doc in Denmark on human monoclonal antibodies in autoimmunity (1981-82), she directed two research units on immunogenetics and population genetics in Toulouse between 1985 and 1997. She has more than 200 scientific publications, mainly in the domains of population genetics, genetic epidemiology of autoimmune diseases (especially Type 1 diabetes, multiple sclerosis and rheumatoid arthritis), human immunogenetics and polymorphisms in the HLA system (especially the microsatellites in this genomic region), and renal and bone marrow transplantation. Besides her work in immunogenetics, she worked in recent years especially on societal aspects of

biobanks and genetic testing. Since 1998 she has developed research on the societal dimensions of genetics. She presently leads a multidisciplinary team on "Genomics and public health" involving human and social sciences as well as life sciences, in the context of research in epidemiology and public health at Inserm (National Institute for Health and Medical Research) at the Faculty of Medicine of the University of Toulouse, France (Inserm U558). She also leads a "Genetics and Society" platform at the Toulouse-Midi-Pyrénées Genopole. Her present research interests are in societal and public health implications of technological, methodological and regulatory developments in two main domains: 1) genetics and genomics of multifactorial diseases, including large scale biobanking, genetic information access and management, and 2) organisation of the transplantation and cell therapy field, especially using haematopoietic stem cells. She is involved in several EU projects in transplantation, genomic sciences, public health genomics and biobanks. She is PI on ELSI aspects in several EU projects. She has been rapporteur of an EU expert group on ethical, legal and social aspects of genetic testing; she sits in several scientific advisory boards of international projects, especially related to biobanks or genetic testing and chairs the scientific council of the French national children cohort project (ELFE). She is member of the scientific council of Inserm, of the Medicine Faculty council in Toulouse and of the board of the French Society of Human Genetics. In ESHG, she is a member of the PPC (Public and Professional Policy Committee). She is involved in several ethics committees. Former member of the CCNE (French National Advisory Bioethics Committee) and of the Toulouse hospital ethics committee, she is presently member of the European Group on ethics of science and new technologies and Chair of the Life Sciences operational ethics committee in CNRS. She participates in many actions towards various publics and to facilitate the dialogue between scientists and lay publics. She teaches in genetic epidemiology, ethics of genetics and of biotechnologies.

Dr. Tayfun Ozcelik, Department of Molecular Biology and Genetics. Bilkent University, Ankara, Turkey.



Dr. Ozcelik's research is focused on genetic mapping and identification of the molecular bases of inherited diseases. Together with his mentor Professor Uta Francke at Yale and Stanford Universities, he has determined the chromosomal localizations of approximately forty different human and mouse genes using somatic cell hybrids and recombinant DNA techniques. These mapping studies targeted mainly neuronal genes and resulted in the identification of the genes associated with Prader-Willi syndrome, Charcot-Marie-Tooth disease type 1A, and type VIII glyco-

gen storage disease. His subsequent research at Bilkent University continues to be centered on inherited diseases. He described “loss of mosaicism” for X-linked gene expression as a new candidate disease mechanism leading to autoimmune disorders in females. These diseases include scleroderma, autoimmune thyroiditis, preeclampsia and juvenile idiopathic arthritis. He showed that “somatic mosaicism” could be a survival mechanism in X-linked dominant diseases in males as exemplified by Rett syndrome. Using X-chromosome inactivation as an indicator of cell lineage, he showed that angiogenesis in multiple myeloma could be associated with clonal expansion of endothelial cells. He described homozygous mutations in MLH1 gene leading to familial hematological malignancies and neurofibromatosis. This study is one of the first examples of DNA repair genes linked with inherited hematological malignancies in humans. Most recently, he identified VLDLR mutations in a novel human phenotype characterized by quadrupedal locomotion, mental retardation and cerebellar hypoplasia. This is the first study that sheds light on the molecular mechanisms and neuronal pathways that are linked with gait in humans through the analysis of an inherited condition. Homozygosity mapping in this unique human trait revealed that there are additional loci in the human genome linked with quadrupedal locomotion. In addition, Dr. Ozcelik introduced DNA-based identification to the Turkish judiciary system in collaboration with the Ministry of Justice, Forensic Medicine Council; provided DNA based diagnostic testing services in a large number of genetic disorders; and supervised over twenty graduate students. His scholarly publications received over 2,500 citations, including those in 35 books, 23 in Victor McKusick’s “Mendelian Inheritance In Man” book and database, and in numerous review articles. He is currently a board member of the Turkish Society of Medical Genetics, after serving as the president of the society in 2005-2007. He is also a board member of the European Society of Human Genetics, as well as education committee member. He continues his research on X-inactivation and female predisposition to autoimmunity, and neurodevelopmental disorders as a faculty member and chair of Bilkent University, Department of Molecular Biology and Genetics.

Prof. Alexandre Reymond is Associate Professor at the Center for Integrative Genomics of the University of Lausanne, Switzerland. He carried out his thesis in the laboratory of Dr. Viesturs Simanis at the Swiss Institute for Experimental Cancer Research (ISREC). He studied the genetic and molecular biology of the entry into the mitotic cell cycle using fission yeast as a model. He received his Ph.D. from the University of Lausanne in 1993, before joining the laboratory of Dr Roger Brent in

the Department of Molecular Biology, Massachusetts General Hospital and in the Department of Genetics, Harvard Medical School in Boston. During this postdoctoral training he accustomed himself with new genetic technologies that allow identifying protein-protein interactions, such as the two-hybrid system. Prof. Reymond joined the medical genetics community upon moving to the Telethon Institute of Genetics and Medicine (TIGEM) in Milan in 1998

to lead a research group. During his three years in Italy, he characterized the tripartite motif-containing proteins and coined their name TRIM. The genes encoding these homomultimerizing E3-ubiquitin ligases are mutated in a large number of monogenic diseases, as well as in specific neoplasia. He then moved to the Department of Genetic Medicine and Development, University of Geneva Medical School, where he studied aneuploidies. He participated in the functional characterization of genes mapping to human chromosome 21 and the Williams-Beuren syndrome critical region, as well as in the annotation of the mouse, chicken and cow genomes. He still carries out this line of work by participating in the functional annotation of the human genome led by the ENCODE consortium. Prof. Reymond is currently studying how genome structural changes influence gene expression. He has recently shown that the level of expression of genes within Copy Number Variants (CNVs) tends to correlate with copy number changes, and that CNVs influence the expression of genes in their vicinity, an effect that extends up to hundreds of kilobases.

Prof. Jorge Sequeiros, University of Porto, Portugal,



received his MD degree in Porto, in 1975, and a PhD in 1990, and trained as an internist and as a medical geneticist (post-doc fellow at Johns Hopkins Hosp., 1982-85). He is a full professor at ICBAS, where he teaches medical genetics and clinical genetics to 3rd and 5th year medical students, and clinical molecular genetics to biochemistry students, collaborates in teaching population genetics to

1st year medical students, and is initiating a new master course to train genetic counsellors. He founded and is the group leader of UnIGENE (a neurogenetics research unit, centred mostly on triplet repeat disorders) and CGPP (a centre of genetic services to the community, including molecular genetics testing and genetic counselling in the field of neurological diseases), both at IBMC, Univ. Porto. He is the president of the Portuguese College of Medical Genetics, of the National Medical Association, and of the Human Genetics Commission, a consultant to the General-Health Directorate, and a member of CNECV (National Council for Ethics in the Life Sciences) and the ethics committee of Univ. Porto. He was among the founders of the Portuguese Society of Human Genetics (SPGH), in 1996, and served as its president and vice-president (1996-99). He is a member of the board of the ESHG since 2008, and of its Public and Professional Policies Committee (since 2002), and served on its Education Committee as well. He has been a representative at the steering groups for “Quality Assurance and Proficiency for Molecular Genetic Testing”, “Guidelines for Best Practices in Quality Assurance for Molecular Genetic Testing in OECD countries”, “Pharmacogenetics Initiative” and “Human Genetics Research Databases”, at OECD. He has also been a member of the informal network on genetic testing at the European Commission, and a representative at the NEC-Forum (national ethics committees), also at the EC. He participates in Unit 3 and in the steering group of EuroGentest (Genetic Testing in Europe), is the national representative for ORPHANET, and the scheme organizer of the external quality assessment in the spinocerebellar ataxias (SCAs) for EMQN.

He was also a member of the European network PHGEN (Public Health Genomics) and is an associate member of SAFE. Prof. Sequeiros was the secretary-general for the Ataxia Research Group, at the World Federation of Neurology, a medical genetics expert for the European Society of Neurology, and the 'contact person' for the Programme on Integrated Approaches for Functional Genomics of the ESF (European Science Foundation). He is on the editorial board of Clinical Genetics and of the Journal of Biomedicine and Biotechnology, and has participated in several prize juries and scientific review panels, including for FP7-Health calls (ethical screening) and ANR-France, INSERM (neurological and psychiatric diseases). He was the consultant for the activities of public awareness on the Human Genome of the Program "Ciência Viva". He was or is a member of the scientific councils of Euro-Ataxia (Eu-

ropean Federation of Hereditary Ataxias), the Portuguese Associations for Huntington Disease, of Inherited Ataxias and of Paramyloidosis, and of the Machado-Joseph Disease Foundation, Australia. Prof. Sequeiros is the author or co-author of more than 150 full scientific papers, including three chapters on Machado-Joseph disease or the Inherited Ataxias, in reference books on ataxia, including 100 original articles in refereed international journals; he was also the editor of a multi-authored book on MJD, the contributor for the genetics index of the 25th ed. of Stedman's medical dictionary, and to several encyclopaedias and books in the areas of genetics, neurology and bioethics. His major research interests are in genetics of rare neurological disorders (mainly the SCAs and Huntington disease), genetic testing and counselling, psychosocial genetics, and genetics and bioethics.

Minutes of the Annual Membership Meeting 2008

At the EUROPEAN HUMAN GENETICS CONFERENCE 2008

Barcelona, June 1st 2008, 19.00 – 20.00 hrs

Present: ESHG Executive Board, Several Board members, About 100 representatives of the membership

1. Activity of the European Society of Human Genetics 2007-2008

President of ESHG Pier Franco Pignatti gave activity report. He especially mentioned new issues and activities as follows:

There are new committees (Genetic Services Quality Committee, Communication Committee) and sub-committees (Subcommittee for laboratory geneticists, Subcommittee for counsellors/nurses), details on the website.

New Executive Officer Jerome del Picchia has now worked (part time) for one year.

There are new ESHG documents (on the website): Recommendations for Patenting and Licencing in Genetic Testing, Recommendations for Genetic Counselling related to Genetic Testing (from EUGT, endorsed with annotations), Core competences in Genetics for Health Professionals in Europe (collected together with EUGT, waiting for approval by ESHG); some others under preparation.

There are/will be new ESHG fellowships

-For the participation to the annual Human Genetics Conference on proposal of the NHGSs. First year: 22 nominations were received

-For short term training in Europe

-For participation in the EGF courses, awarded directly by the ESHG

There will be new ESHG Courses

-Regional: on proposal by National Human Genetics Societies

-European: on individual proposal or ESHG solicitation. 2 are being considered: "Genetic Counselling: Training the Trainers", and "Genetic Epidemiology of Common Diseases"

2008 new DNA Day contest

In collaboration with the ASHG, DNA Day was instituted 2008 with 118 participants for this first year. For details, please see the website.

New EUROGENE Project

ESHG is a partner of this EU Project for digital Education in Genetics, in several languages and quality control for sound packages. This project will establish a European reference portal for multimedia education in genetics.

New ESHG Strategy Meeting

9-10.11.2007, Brussels, a new type of strategy meeting was organized by JJ Cassiman with the participation of 27 members of the Board and past Presidents of the ESHG. After plenary and group discussions the meeting resulted in refinement of the ESHG mission for education, research, good clinical and laboratory practice, and indications for future actions

Patenting and Licencing in Genetic Testing

A press conference, Brussels, 24.4.08 was organized by G. Matthijs for ESHG-PLC with the participation of European Patent Office, DG Research of the EU Commission, Patent Attorney

The European Journal of Human Genetics

The Journal is doing very well both financially and scientifically. Some PPPC documents are on the way to be published in EJHG 2008 or 2009 (Genetic Testing in Minors, Genetic Testing in Common Disorders, Pharmacogenetics/Pharmacogenomics)

Some ESHG Collaborations

EUROGENTEST for genetic testing in clinical practice, quality assurance, education...

ORPHANET for rare diseases and orphan drugs

EGF for genetic medicine courses

ECA for quality assurance in genetic services

ASHG as a possible partner in some activities

IFHGS to promote international collaboration in human genetics

UEMS for the recognition of clinical/medical genetics as a EU specialization

EPO for patenting in genetic testing

OECD for guidelines on biobanks and genetic research databases

EMEA-PGWP for pharmacogenetics terminology to be approved by ICH and IPTS

ISE-ERA for the development of science and research in Europe

ESHG Committees:

Public and professional policy committee

Scientific Programme Committee

Publications Committee and EJHG Education Committee

and sub-committees for medical/clinical genetics, laboratory geneticists and counsellors/nurses
 Genetics Services Quality Committee
 Historical Interest Group
 Communications Committee (new)
 The committees have given their reports in Newsletter. Committee chairs were present and ready to answer questions from the membership. Especially, the Scientific Programme Committee was thanked for its excellent work.

2. Financial Report of the Society 2007

Treasurer of the society, Andrew Read, gave financial report. The financial situation of ESHG is good and there are some reserves kept for the possible situation when ESHG Conference would have a very negative balance. As the situation is good, new types of fellowships and courses have been instituted.

3. Discharge of the Board Members for the year 2006-2007

Members leaving the Board (Alexis Brice, Nicolas Levy, Gert Matthijs, Leena Peltonen) were warmly thanked for their valuable work. Also Vice-President John Burn was thanked for his work for the Society.

4. Opening by the new President of the Society

Prof. Jean-Jaques Cassiman thanked the previous Presi-

dent (now Vice-President) Pier Franco Pignatti for his excellent work.

5. Results of election for President-Elect

Prof. Dian Donnai was elected.

6. Results of election for Board Members

There had been 5 nominations for new Board members : Agnes Bloch-Zupan, Anne Cambon-Thomsen, Alexandre Reymond, Jorge Sequeiros, Tayfun Özcelik. As they were all excellent candidates, Board had suggested that they all would be elected. This was accepted.

7. Membership fees 2008

It was decided that the membership fees would be kept unchanged.

8. Budget proposal 2008

Of the future activities of the Society, especially the growing number of fellowships, new courses and collaboration with National Human Genetic Societies, are the cornerstones of the budget for the next year. In addition, the balance between attractive but however not too expensive Conference sites was discussed.

9. Closing of the meeting

As there were no other major policy questions, the meeting was closed by the President at 20.00 hrs.

Invitation to the **Annual Membership Meeting 2009**

At the EUROPEAN HUMAN GENETICS CONFERENCE 2009

Sunday, May 24, 2009 at 7.00 – 8.00 p.m.

Room F2, Austria Center Vienna, Bruno Kreisky Platz 1, 1220 Vienna, Austria

AGENDA

Opening by the President of the Society, Professor Jean Jacques Cassiman

1. Activity of the Society 2008-2009
2. Financial Report of the Society 2008
3. Discharge of the Board Members for the year 2008-2009

Opening by the new President of the Society, Professor Dian Donnai

4. Results of election for President-Elect
5. Results of election for Board Members
6. Membership fees 2010
7. Site of future European Human Genetics Conferences
8. Budget proposal 2010
9. Major policy questions proposed by Board
10. Future activities