European Society of Human Genetics

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President's Address
ESHG: the Voice of Human Genetics in Europe

Pier Franco Pignatti
President of the ESHG

Dear Colleagues,

First of all, a warm welcome to each of you to the Annual European Conference in Human Genetics. This is the 40th Conference, a well cherished meeting providing the latest developments in the field and the highest quality scientific and clinical information to our diverse audience. During these days you will be part of the Conference, and you will be able to judge directly its success, due to the hard work of the Scientific Programme Committee, the Executive Director, the Exhibition Management group, and the local Organizing Committee.
What will not be so evident during the Conference is the Society which makes all this possible, and its diverse activities. I, for one, was surprised, and even at first somewhat confused, by all that was going on "behind the scenes." Therefore I now wish to share with you some of what I have discovered, with the wish that you might find it interesting, and perhaps decide that you would like to become more directly involved. We need all the help we can get to confirm and expand the role of the Society as the coordinating organization for Human Genetics in Europe.
I will briefly indicate below some of the activities of the ESHG which I would love for you to know about.

National Human Genetics Societies
We will be having our 4th NHGSs meeting in Barcelona. This is a conference which allows us to establish contacts and better know each other from various countries, discuss, and seek common solutions to common problems. Last year in Nice we had representatives from 42 (!) different National Societies, as you can see from the nice meeting picture on this page, kindly provided by Andres Metspalu. The list of the National Societies, the minutes of the previous meetings, and the agenda with the slides of all the presentations of the 2008 meeting are available on the website (http://www.eshg.org/)

3rd Meeting of the Presidents of National Human Genetics Societies, Nice, June 2007.

I draw your attention to some recent developments of particular interest to the NHGSs: ESHG fellowships to be awarded to young researchers on the basis of scientific merit by National Societies to attend the Conference, the new call for proposals to organize regional courses, the new short term European training grants, and the DNA Day Essay Contest for schools which began this year.
The Committees
We have some very active working groups, as you can see from other articles in this Newsletter, which have been instrumental in producing a series of important documents ranging from basic, translational and clinical research to educational and social issues. You will find on the website ESHG documents on genetic services, population screening, insurance and employment, DNA banking, genetic testing, and assisted reproduction.
Other documents are being developed, and are posted on the website as drafts for which we ask for your comments, as patenting and licensing, pharmacogenetics, and core competencies.
The Journal
The European Journal of Human Genetics is an important asset to the Society and its international visibility, publishing timely and first rate research, useful editorials, commentaries, review articles, and special issues on selected scientific and social aspects.

See for example the May 2008 EJHG Supplement with the ESHG documents on Patenting and Licensing in Genetic Testing.
International Collaborations
The Society has been collaborating with several organizations on specific issues. I wish to mention here in particular two important collaborations, one with the European project EuroGenTest on several actions ranging from quality management to educational, ethical and legal issues (as described in an accompanying article in this Newsletter), and the other with Orphanet, the leading group for information on rare diseases and orphan drugs. These close collaborations are the reason why this year you will be finding a common exhibit area for the three booths of ESHG, EuroGenTest, and Orphanet.
The ESHG has also worked with several other organizations, such as ECA for a new committee on the quality of genetic services, OECD for guidelines on human genetic databases, EPO for recommendations in gene patenting, ISE-ERA for the development of science and research in Europe, EMEA for pharmacogenetics terminology, ASHG for establishing the annual DNA Day school contest in Europe, and IFHGS to promote world-wide collaboration in human genetics and organize the international congress.
Issues arising
With the help of the Committees we are working for the recognition of Clinical/Medical Genetics as an EU specialization, plus that of Laboratory Geneticists, and Genetic Nurses/Counsellors, drafting documents on susceptibility testing in common disorders, and on genetic testing in minors, organizing new ESHG international courses, and developing multimedia didactic material.
In conclusion, all of these achievements have been made possible by the dedication and spirit of collaboration of many persons, including members of the Society, the Committees, the Board, the Executive Board, and the Executive Director: to all of you, my thanks for the productive work and the pleasant experiences I have had during this year of presidency. Thanks in particular to the previous President John Burn, and best wishes to the new President Jean-Jacques Cassiman. All together now:

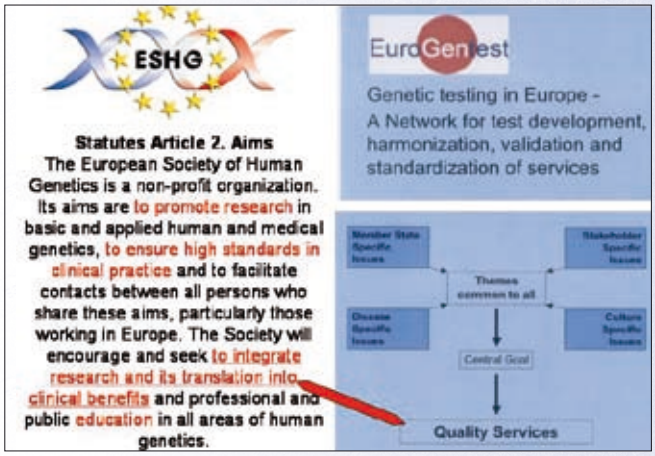
"LONG LIVE the ESHG, the VOICE of HUMAN GENETICS in EUROPE!"

Pier Franco Pignatti
President of the ESHG

ESHG and EUGT

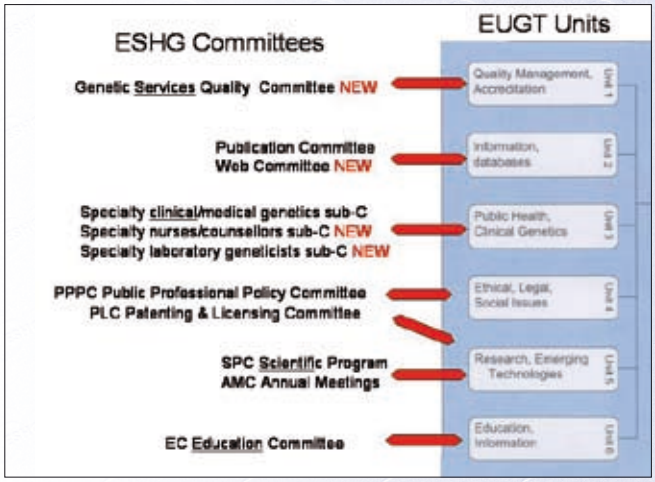
The following text is from a presentation given to the General Assembly meeting of EuroGenTest in Leuven on 22 Nov 2008.

EUGT is a FP6 Network of Excellence for the harmonization of Genetic testing in Europe. ESHG and EUGT have similar aims, as deduced from the ESHG Statutes and the EUGT project outline, and shown in Figure 1.



The aims are: 1. to promote research, 2. to ensure high standards in clinical practice, 3. to translate research into clinical benefits, and 4. to foster professional and public education, all in the fields of human genetics.

The organization of ESHG and EUGT to reach these aims is also similar, as indicated in figure 2, where different ESHG Committees are compared to EUGT Units.



The Society and the Network have groups active in: 1. research, 2. specialization of personnel, 3.service quality management, 4. ethical, legal, and social issues, 5. dissemination of information.
Many human geneticists working in Europe participate in the activities of both the ESHG and EUGT, and this obviously strengthens the collaboration between the two organizations.
EUGT is a veritable Network of Networks, as it has established links with 23 (!) different Networks, including some of particular interest to the ESHG, such as patient associations, the EU Diagnostic Manufacturers Association, and the European Federation of Biotechnology.
The ESHG is examining two documents prepared by EUGT Units, after extensive consultations and meetings, on "Recommendations for genetic counselling related to genetic testing" and on "Core competences in genetics for health professionals in Europe"
(http://www.eshg.org/ and http://www.eurogentest.org/).
The ESHG and EUGT are also collaborating for the DNA Day in Europe, the recognition of clinical/medical genetics, laboratory genetics, and genetic nurses/counsellors specialization in the EU, the EUROGENE project for digital education in human genetics, the recommendations for patenting and licensing in genetic testing, with Orphanet for rare diseases and orphan drugs, etc.
At this year's Human Genetics Conference in Barcelona, the ESHG is happy to host, as done in previous conferences, 3 different EUGT Workshops/Training courses on quality assurance in genetic testing laboratories, a pioneering and particularly successful activity of the EUGT project.

The EUGT project on apoptosis is programmed for the year 2009. We express our wish that it might survive, as it has been so useful for the progress of human genetics in Europe. In any case, the ESHG has received some EUGT stem cells, so that it is ready to save these valuable activities for the future.
We are confident that the activities will continue, and are reassured by the fact that the coordinator of EUGT is at the same time the President elect of the ESHG: best wishes Jean-Jacques for your work, from ESHG and EUGT!

Pier Franco Pignatti

Letter from Secretary General

Helena Kääriäinen
Secretary General of the ESHG

Dear ESHG members,

Being Secretary General of the ESHG is a most interesting task. According to the Statutes, the Secretary General is nominated by the Board and elected by the Membership meeting for three years and the period is renewable. In my case, after the first three years, I chose to continue for another three years as continuity in the Executive Board (Secretary General is a member of the Executive Board) helps in running the practical matters of the Society smoothly.

As Secretary General I have been asked questions concerning nearly everything. One of the most common questions is: "Can our company send advertisements to ESHG membership by email"? As a rule, the answer is "no". Instead, ESHG may send address labels to the company and they may send advertisements by ordinary mail. ESHG wants to protect its members from too many emails. Another common question is: "Can we advertise our meeting/workshop/seminar on your webpage?" The usual answer is "yes" as you know if you follow www.eshg.org and then Upcoming conferences. I suggest that you check that address every now and then –if you are not afraid of the frustration of interesting meetings which you have no time to attend!
You may have noticed that the ESHG website is improving. The reason is that ESHG now has Jerome del Picchia working as Executive Officer and he really has devoted his time in reshaping and updating the website. In addition, I in the role of Secretary General and Silke Sperling as a volunteered Board member are trying to help Jerome in creating a most interesting website. If you have suggestions for improvement, interesting links, anything…just contact us!

But actually, even more interesting than answering the several questions, is working together with the ESHG Board which represents genetics in Europe in a really broad way. And of course, to help the President and other Executive Board members in responding to the many challenges that ESHG, as a growing society, is facing.

I mean all this! In addition, I am advertising! After two years we need a new Secretary General. Just now it would be an ideal time to start practising for it as Deputy Secretary General. If you have experience of working in a genetic society at the national level or if you have been in the ESHG Board or Committees: just consider signing in as a candidate for Secretary General!

Helena Kääriäinen
ESHG Secretary General

Report from the SPC

Han Brunner
Chair of the Scientific Programme Committee

The Scientific Programme Committee for 2007-2008 was composed of Han Brunner (chair), Thierry Frébourg, Paulo Gasparini, Peter Heutink, Juha Kere, Peter Lichter, Milan Macek, Jr., Raquel Seruca, Andrew Wilkie, Brunhilde Wirth, Olaf Riess, Eduardo Tizzano (local host), Feliciano Ramos, Batsheva Kerem, Pete Scambler, and Cisca Wijmenga. Helena Kääriäinen participated as observer from the Executive Board.
The SPC met twice to organize the Barcelona 2008 ESHG conference: in Barcelona in June 2007 to decide on the plenary sessions and symposia, and in Vienna at the VMA offices in March 2008, to select the abstracts for oral presentations and posters.
The number of sessions from submitted abstracts continues to increase, and has now reached 15. This allows us to keep the number of abstracts selected for oral presentation around 5% of the total number. Secondly, we decided to keep a session on the first day that presents particularly exciting new findings in a "What's New?" session from submitted abstracts.
For the final session on Tuesday 19 June, we were able to attract Leroy Hood from the Institute for Systems Biology in Seattle to discuss "Systems Biology and Systems Medicine." Lee Hood has been one of the co-inventors of 4 key technologies used in all of human genetics today. These are the DNA gene sequencer and synthesizer, and the protein synthesizer and sequencer. His presentation is sure to keep the scientific excitement up for the entire conference, right until the end.
An innovation for this year's conference is to have awards for the best posters. The SPC has made a selection from which board members will be asked to make their choice. The prizes will be given during the closing ceremony.
As usual, our second highlight of the final day of the conference will be the acceptance speech by our ESHG prize winner. This year, the ESHG prize 2008 will be awarded to Professor Arnold Munnich (Paris) in recognition of his influential and groundbreaking work which has helped to elucidate the clinical phenotypes and the molecular basis for many monogenic diseases.
After the Barcelona conference, the SPC shall have to say goodbye to Juha Kere, Peter Lichter, Paolo Gasparini, and Milan Macek Jr. We thank them for their work and their dedication to making the meeting better.

Han Brunner

Report from the Education Committee

Domenico Coviello
Chair of the Education Committee

Dear ESHG members,

During the past year, the Education Committee has focussed its activity on several topics. Here is a summary of the work done:

1) Document on "Core Competences in Genetics for Health Professionals in Europe"

- The establishment of core competences is currently being used as a basis for health professional education in many other fields and settings (Walton & Elliott, 2006; Wold et al, 2006; Smith, 2005). The core competences have been based upon existing frameworks developed in a number of countries for a range of professional groups. These have been discussed and modified for the European context. The goal of this work is not to unify the existing genetic services accross national boundaries, but to achieve broad consensus about a coherent set of standards to guide the education of health professionals. This could provide an appropriate framework for establishing minimum standards of preparation for health care professionals in genetics across national boundaries.
- Our background document was discussed for the first time at the Porto workshop (September 23-24, 2006) and put on the web for consultation. All the comments received during the year were included and the second draft was discussed at the Milan meeting (October 27-28, 2007)
- The final document, completely revised after the Milan meeting, is now on the web for the final consultation. The document has been divided in four separate smaller documents. (http://www.eurogentest.org/web/info/public/unit6/core_competences.xhtml)

2) Partnership with the EUROGENE project (www.eurogene.eu).

- The EUROGENE project objective is to migrate towards a more efficient development of higher quality (multimedia) didactic material on genetics. This is being done by the guided editing and "assembly" of educational packages based on the IMS learning design metadata framework and the sharing of different types of "learning objects" between content owners, in 9 languages.
- The project proposes to share materials by establishing the EUROGENE web portal as a reference point for the sharing of a critical mass of educational content items and packages, offering an integrated toolset and service for content authors.

3) DNA Day (April 25th)

This event was created in the USA to commemorate the completion of the Human Genome Project in April 2003 and the discovery of the DNA double helix fifty years earlier (1953). The European Society of Human Genetics (ESHG) President, Prof. Pier Franco Pignatti, sent a letter to all Presidents of national Human Genetics Societies in Europe in order to encourage them to collaborate with local teachers' associations and promote the DNA Day in their countries. ESHG has set up a pilot contest for high school students with three prizes for the best composition sent to the ESHG office (see ESHG web site).

4) European network of Genetic Nurses/Counsellors

The first European network of genetic nurses and counsellors has been set up, and the first meeting will be held in Barcelona within the ESHG annual meeting. A survey of working roles and needs for support of the 65 current members of the network has been undertaken and is being analyzed for presentation at the ESHG meeting. Almost 30 members have already registered for the first network meeting in Barcelona.
(For info contact Heather Skirton at heather.skirton@plymouth.ac.uk)

5) Dissemination of results and interaction with other networks or scientific societies

- The Core Competences document was presented at the plenary session of the meeting of the Association for Medical Education in Europe (AMEE) in Trondheim, Norway, 27-29 August, 2007.
- Eurogentest activities were presented at the "1st Workshop in Genetics and Public Health for Developing Countries" Latin American Networks - 17th and 18th of September 2007. Guayaquil, Ecuador.
- Eurogentest activities were presented also at the plenary session at the third meeting of the European Network PHGEN - Public Health Genomics - : "From Policy Development to Assurance", in Hinxton, January 23rd-25th 2008

Domenico Coviello, MD, PhD
On behalf of Education Committee

ESHG-EDUCATION COMMITTEE
Domenico Coviello (Chair, Milan, Italy)
Martina Cornel (Amsterdam, The Netherlands)
Celia DeLozier (USA/Switzerland)
Peter Farndon (Birmingham, UK)
Peter Goetz (Czech Republic)
Shirley Hodgson (London, U.K.)
Alastair Kent, (Patients Organizations, GIG and EAGS)
Gyorgy Kosztolanyi (Pécs, Hungary)
Jorge Sequeiros (Porto, Portugal)
Heather Skirton (Plymouth, UK)

Additional collaborating members:
Jacques Beckman (Switzerland)
Agnes Bloch-Zupan (Strasbourg, France)
Francoise Clerget Darpoux (Paris, France)
Hillary Harris (Manchester, U.K.)
Vaidutis Kucinskas (Lithuania)
Taytum Ozecelik (Ankara, Turkey)
Fred Petrij (Rotterdam, The Netherlands)
Maria Soller (Lund, Sweden)
Reiner Siebert (Kiel, Germany)
Marcus Pembrey (Bristol, UK)
Jan Vejvalka (Prague, Czech Republic)

Patenting and Licensing in Genetic Testing: Recommendations Ready and Published!

Gert Matthijs, Chair of the Patenting and Licensing Committee

The Professional and Public Policy Committee (PPPC) and the Patenting and Licensing Committee (PLC) have presented the Background Document on "PATENTING AND LICENSING IN GENETIC TESTING" in 2007. The document has been published in the May issue of the European Journal of Human Genetics (EJHG), together with a set of recommendations, formulated on behalf of ESHG.

The aim of the joint work was to explore how to achieve a situation where useful tests are available at affordable costs for diagnosis of patients. The group explicitly aimed to 'go beyond the Myriad case.' Despite already available recommendations and reports on gene patenting, the ESHG found it necessary to focus further on diagnostics and public health aspects, and to define further action points to work with. With new knowledge constantly developing, such as ESTs, variants, gene expression mechanisms, genetic associations and so on, the practical framework is becoming more complicated.

This work was begun upon request by the President and the Board of the ESHG. After an initial meeting in November 2005 in Paris, and a workshop in November 2006 in Leuven, a document was prepared that would comprehensively review the background on patenting and licensing, and the current situation with a focus on genetic, diagnostic testing. During the workshops, external experts were asked for advice. The discussions and the background document have served as a basis for the generation of recommendations by the PPPC and PLC members. These recommendations have been submitted to the Board for approval, and were then finalized for publication in EJHG.

The ESHG invited the media to attend the launch of new guidelines on patenting genes at a meeting in Brussels, on April 24, 2008. The message: it is hoped that these guidelines, which are now published in the European Journal of Human Genetics, will bring to an end the long-running controversy on this subject. Despite the fact that there was a substantial amount of press coverage in response to the press release that accompanied the invitation,, no journalists attended the meeting in Brussels.

The ESHG had also invited several experts, to hear their opinion on the recommendations. This led to a very open and fruitful discussion between these experts and members of ESHG. The experts had read the recommendations with interest, and agreed with many. Some points f discussion remained, of course.

John Burn, Siobhán Yeats, Joseph Straus, William Bird

The aims of the ESHG and the recommendations were briefly presented to the audience by Prof. Pier-Franco Pignatti, president of ESHG, and by Prof. Gert Matthijs, on behalf of the PLC and PPPC (Dr. Ségolène Aymé, (former) chairperson of the PPPC, could not attend the Brussels meeting). Prof. GertJan van Ommen gave a quick overview of the state-of-the-art in genetics and DNA diagnostics.

The panel included Dr. Siobhán Yeats from the European Patent Office (EPO) (Munich, Germany), Prof. Joseph Straus, an eminent academic patent specialist from the Max-Planck-Institute for Intellectual Property, Competition and Tax Law and Munich Intellectual Property Law Center (Munich, Germany), Dr. Denis Dambois, an intellectual property expert from the European Commission (DG Research) and Dr. William Bird (Bird Goën & Co, Belgium), a patent attorney, practicing in the biotech field. Prof. John Burn from the Institute of Human Genetics (Newcastle University, UK) and former president of the ESHG represented the geneticists' view. Prof. Geertrui van Overwalle from the Centre for Intellectual Property Rights (University of Leuven, Belgium) moderated the discussion.

Other attendants from ESHG included Prof. Jean-Jacques Cassiman, president-elect of ESHG, Prof. Martina Cornel, (current) chairperson of the PPPC, Prof. Domenico Coviello, from the Education Committee, and Mrs. Sirpa Soini, who worked for the PLC-PPPC and wrote the Background Document.

The Background Document and Recommendations are freely downloadable from the EJHG website.

In brief, these are the major observations:

1. The major problems seem to be in the breadth of the claims in genetic patents, in the criteria for patentability and in the number of (overlapping) patents.
2. There is a need to improve the quality of the patents that will eventually be granted.
3. The research exemption is generally unclear, and not universal.
4. Licenses are problematic when they are exclusive. In general, licensing seems to be prohibitive, both in practical and in financial terms, partly due to the complexity of the system, and to the lack of effective guidelines.

The major recommendations that were formulated by ESHG were:

1. It could be fairly easy to prohibit patenting of individual mutations in known disease genes, for example, on the basis of an absence of novelty.
2. Establishing a link between a disease and a genetic sequence or defect is merely a discovery and therefore not patentable, unless the identification of this link includes a real conceptual innovation.

Jean Jacques Cassmiman, GertJan van Ommen, Pier Franco Pignatti

3. The ESHG proposes EPO to consider the benefit of having an ethics committee to consider issues of major interest, such as patents applied to genes.
4. Policy makers should work on the development of licensing guidelines, and effectively support those that have already been issued by international organizations such as the OECD.

Many more issues have been dealt with in the Recommendations. As suggested above, the external experts did not entirely agree with some of these standpoints. Still, it is task of ESHG to try and promote the changes that were proposed to both the patenting and licensing system., It would be worthwhile also for all ESHG members, involved in either diagnostics or research, to take a quick look at them. The Background Document is also useful as an introduction to patenting and licensing.
Finally, we thank everybody who has contributed to the generation of the Background Document and the Recommendations: the members of the PPPC and of the PLC, the external experts, and, in particular, Mrs. Sirpa Soini, whose contribution has been vital to the success of this endeavor.

Gert Matthijs
Chair of the Patenting and Licensing Committee

Report from the Editor-in-chief of the European Journal of Human Genetics

GJB van Ommen
Editor in Chief

The developments of the European Journal in 2007 have seen an impressive further growth, in readership and subscriptions as well as in submissions The submissions grew by 28%, while the size of the journal only grew ca 8%. By necessity, the rejection rate has increased, and this may have created some disappointment, and even some delays occasionally. For the latter we apologise (but meanwhile the online publication time has gone down from 7 to 6 weeks after acceptation. On the other hand, however, we want to make the EJHG count, and the best way to further increase our Impact Factor is to gradually raise the acceptation bar. That this is successful follows from last years' Impact Factor increase, from 3.25 over 2005 to 3.70 over 2006. This has placed us at nr. 42, four up from last year. The prognostics afficionados tell us that it is likely that the IF further increases over 2007.
Website attention is also steadily rising, and in collaboration with Nature Publishing Group the website is continuously further developed towards optimal utility, like recently with free links to reviews from Nature Reviews Genetics when these are deemed specifically relevant for the EJHG readership.
Another interesting development is the strong growth of the overseas attention. The US/Canadian submissions grew from 12.9% to 16.3%. This goes hand in hand with an increase of 11% in institutional subscriptions. Combined with a stable, even slightly decreasing production cost this has yielded an EJHG revenue increase of 18%.
This, we are told, is quite a unique achievement in a stabilizing if not slightly contracting market. Apparently, the EJHG still has significant development potential, which is now being realized in the wake of electronic and site-license publishing. We note that a similar development takes place with the attendance of the ESHG's meetings, notably from overseas, as this is also going from strength to strength in a stabilizing field (with a strongly increasing competition, especially in sub-field specialty meetings.
And it is hardly as if we have nothing to do either, isn't it? With the increasing interest for genetics, greatly improved diagnostic possibilities due to technological innovation, and expanding insights into biological pathways connecting rare and common diseases. The EJHG aims to continue publishing advances in all these fields, both the 'classical' human genetics areas, like clinical genetics, genetics services and societal aspects, cytogenetics and molecular genetics – the boundaries between the latter blurring ever more due to high-thoughput technologies. In parallel, there is increased attention for functional genetics/genomics studies, population- and evolutionary aspects, and bioinformatics and –statistics. The News and Commentary section is popular and will be developed further, and the same holds for the Practical Genetics series, which has greatly benefited from the collaboration with Orphanet.
As every year, EJHG has a junior authors' high-citation award, to hand out at the Barcelona 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. A. DiFonzo et al with 29 citations in all of 2007 and Jan-March of 2008, for his paper "Comprehensive analysis of the LRRK2 gene in sixty families with Parkinson's disease", which appeared in EJHG 14 no. 3 (2006). Second and third prizes go to Dr. Slavotinek et al for "Array comparative genomic hybridization in patients with congenital diaphragmatic hernia: mapping of four CDH-critical regions and sequencing of candidate genes at 15q26.1-15q26.2", (EJHG 14-9, 2007, 15 citations, and Dr. Ferec et al. for "Gross genomic rearrangements involving deletions in the CFTR gene: characterization of six new events from a large cohort of hitherto unidentified cystic fibrosis chromosomes and meta-analysis of the underlying mechanisms" (EJHG 14-5, 2007, 14 citations). Notably, our senior first authors have also done well this year: both Joe Terwilliger et al and Marcus Pembrey et al. were in the top 5, with respectively "An utter refutation of the 'Fundamental Theorem of the HapMap' (EJHG 14-4, 2007, 32 citations) and "Sex-specific, male-line transgenerational responses in humans" (EJHG 14-2, 2007, 25 citations). Our policy is to keep them /hors concours/, targeting the prizes for the junior first authors, but they do deserve an 'old hands' citation from this place.
Just recently, EJHG has published a special issue on Gene patents in genetic diagnostics, containing the ESHG's recommendations on this (also published in the regular April issue. This special was presented to the press and the EC in Brussels on April 25, DNA day. Clearly this field is in flux, as became clear from the press conference and symposium, which was attended amongst others by Professor Joseph Straus of the Max Planck Institute for Foreign and International Patent, Copyright and Competition Law in Munich, the past chair of the HUGO IPR committee and one of the world authorities in thief field, an well as Dr Siobhan Yeats from the European Patent Office.
Finally, let me stress that the Editorship of EJHG is by no means a lonely operation. While I may be the one who harvests the praise and sometimes criticism, I am greatly indebted to all the people which make EJHG tick. The Section Editors, who target reviewers and provide me with their expert assessments of the review process outcome, the Editorial assistant Jane Pleging-Vale who manages the EJHG process on a daily basis, my contacts at Nature Publishing Group: production controller Francesca Garbarini, who makes all our impossible last-minute wishes possible, Publishing manager Emma Greenwood who contributes many valuable improvements notably of the website structure, and Publishing assistant Morven Mellan who provides us with all thinkable and unthinkable statistics. The EJHG aims to present several more special issues on relevant topics to European geneticists this year.

Report of the Executive Committee Meeting of International Federation of Human Genetics Societies at The ASHG Annual Meeting in San Diego October 23, 2007

Jean-Jacques Cassiman IFHGS Liaison officer for the ESHG

Present: Judith Allanson (ASHG), Jean Jacques Cassiman (ESHG), Pornswan Wasant (APHGS), Gladys Cossio (Panama), Anne Turner (HGSA), Julie McGaughran (HGSA), Roberto Guiliani (RELAGH), Charles Rotimi (AfSHG), Minna Poyhonen (Finland), Michael Peterson (Greece), Ho Suk Saw (EAUHGS), Elaine Strass (ASHG).

The president, Jose Maria Cantu, was ill and did not attend the ASHG meeting.

There was a discussion of program development activities, and it was decided to pursue educational initiatives that will benefit countries that are just beginning to put together human genetics programs. No details were developed. Judith Hall (ASHG) will participate in a clinical genetics meeting in the Philippines in the spirit of education and sharing knowledge about clinical genetics program development especially as it relates to newborn screening programs.

The International Congress of Human Genetics, sponsored and hosted by ASHG, will take place in 2011 in Montreal. The Scientific Program Committee for that meeting is almost complete with three of the six societies having named their participants. The first SPC meeting will be at the ASHG annual meeting in Honolulu in 2009 at which time solicitations for workshops and symposia will begin. The next meeting of the Executive Committee will be at the ESHG meeting in June 2008 in Barcelona. At that time the Human Variome Project and the International Genetics Education Network will be discussed.

Giuliani announced that the RELAGH will meet 8-10 October 2008 in Cartegena, Colombia.

Liaison officer for the ESHG, Jean-Jacques Cassiman (end of term)
Next liaison officer for the ESHG Segolène Aymé

http://www.ifhgs.org

News from the Clinical Genetics Education Committee and the UEMS' MJC Genetics.

Ulf Kristoffersson, Chair Clinical Genetics Education Committee

We have tried to organise a meeting with the sections of UEMS, but have had great problems of finding suitable conditions for participation. We will try hard to have a meeting during autumn.

John Burn organised a workshop and a symposium on the topic on a common curriculum for specialisation in clinical genetics during the British Clinical Genetics Society meeting in Liverpool in March. The workshop representatives from Spain, Italy, Poland, Sweden, UK, Hungary and Romania discussed with representatives of the Society how specialisation issues are handled in the different countries, and how to reach the goal of an EC recognition.

The MJC Genetics have reached an agreement with the accreditation body of UEMS, EACCME, to validate international courses, conferences and other professional activities for CMEs which can be used by participants revealing national CMEs for their continuous educational efforts.

Ulf Kristoffersson
Chair UEMS' MJC Genetics
Clinical Genetics Education Committee

Helen Kingston
Secretary UEMS' MJC Genetics

Formal recognition of Laboratory Geneticists: A scientist's view

Lina Florentin-Arar Editor ESHG Newsletter

During the past years the ESHG is working towards the recognition of Clinical Genetics as a separate specialty that medical doctors can obtain. With Human Genetics developing rapidly across the world this has become a necessity and all of us who are working in this field strongly support it.

For the same reasons it is a necessity to have a formal recognition in the form of a specialty for Laboratory Geneticists/Clinical Scientists in Genetics. An action should be engaged towards this direction by the ESHG as the matter has become really urgent. One might argue that at the moment we are all working towards quality control and accreditation of labs, but this does not mean recognition of a specialty. Quality control and accreditation is important for either medics, scientists and nurses since this is the only way to measure and to control accurate provision of health services. Formal recognition is another issue which is another way to actually protect patients from malpractice. For example there are several European countries where micro-

biologists or clinical biochemists demand to provide laboratory genetic services just because there are no specialties or laws to stop them from doing this.

Whether we like it or not, the profession of laboratory geneticists/clinical scientists in Genetics is under question in many European countries. For the hundreds of molecular geneticists, cytogeneticists and biochemical geneticists who have a University degree and quite a few of them a Ph.D., plus many years of training in Medical Genetics, the future and the way their work will be recognized and accepted is neither obvious nor certain. It is neither obvious nor certain that they will have a profession that is respected for what it is worth and that these people can find dignified jobs and serve what they love most, Human genetics, in a most appropriate and dignified way. That is, recognized in an official way. Those of us who work in Medical Genetics have dedicated our lives to this field, worked endless hours in the labs, burned our hands in radioactivity or exposed ourselves to various dangerous chemicals, have studied and squeezed our minds to understand the nature of the disorders that we have dedicated our youth in studying, have revealed genes and explained genetic disorders, have worked along with our colleagues from the Medical discipline equally to say the least. We have helped to change the way medicine is looked upon and practiced, along with our colleagues the physicians. We are all medical geneticists and it is us who were the first to realize that this is a multidisciplinary area that cannot function, if medics and scientists specialized in this field do not collaborate and share their knowledge and love for the patient. Yet, in quite a few countries we are told that when it comes to diagnostics and providing results for patients this is a prime medical act, and that scientists do not have the knowledge to provide services or direct labs, because we do not understand the genetics behind a disorder etc. A few countries use as an excuse a law as old as the 1930s. This is the shocking part of it. Science has advanced. Medicine has advanced. However, we are still looking back at such laws to explain why medics should sign reports or direct labs, while scientists should not take up on such responsibility towards patients. But we do have a responsibility towards every patient we contact an analysis for, and we give results only when we know the reason of referral. In order to apply certain methodologies and do appropriate analyses we have to know the genetics of the disorder and we have to know all this to interpret the results. All this is linked together. This cannot be mechanically done without deep knowledge of the subject.

I strongly believe that the profession of laboratory geneticists should be supported and protected by the ESHG, both by medics and scientists, because I am more than confident that if we do not take action, we will wake up one day, only to find out that the labs are going to be emptied from inspired and hard working scientists who love what they do. Instead, in some countries at least, there will be only medics who will be directing labs and technicians who will do will do what they are told without understanding or having knowledge of the disease. This will be of no benefit either to medics or patients. Additionally, I must say that University positions or research positions are not as many that could absorb young and inspired scientists. Therefore, this is a profession that must be supported in order to provide new positions and jobs.

This is a task that has been achieved in several countries across the world, amongst them the USA and some European countries. The models are there for us to study and apply. In these countries the roles are clarified and the professions protected and consequently the patients too.

Lina Florentin-Arar, B.Sc., Ph.D.

Non-MD Medical Geneticist: a Specialty which Needs Recognition and a Contemporary System for Educating and Training

Vaidutis Kučinskas, ESHG Board Member

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Genetics services, a clinical application of the achievements of the science of medical genetics, were established in at least 29 different countries (data of the year 2000) in Europe with extreme variability in the level and structure. Genetic laboratory services (or genetic testing) are an increasingly important part of genetic services and are expected to become a routine part of healthcare. At the same time, genetic testing is apparently the area in which application of basic genetic research has the greatest impact. As a consequence, exponentially growing demands for genetic testing, on the one hand, and unique requirements for knowledge, skills and attitudes to be acquired by the specialists providing such services, on the other hand, emphasize the importance of professional education and training of such specialists, which should be maximally harmonised across countries (European Union in the first place).

*The first problem is **terminology** used to refer to the specialists providing genetic services. There is a range of terms used in different countries, most often "clinical geneticist" and "medical geneticist" being used interchangeably, not enabling to distinguish between medical and non-medical specialists providing genetic services. On the other hand, non-medical specialists performing genetic tests are in some cases referred to as laboratory scientists or clinical scientists. It would be reasonable to agree that physicians (medical doctor, MD) trained in genetics and competent to offer specialised genetic services is to be referred to as **clinical geneticist**, while laboratory non-MD specialists (with MS degree or higher) trained in human and medical genetics and competent to offer diagnostic genetic testing in a laboratory environment is to be referred to as **medical geneticist**.*

Complexity and diversity of genetic tests, implications of their results for the patient and entire family and probabilistic mode of their interpretation require that university graduates entering a diagnostic medical genetics laboratory should have adequate level of knowledge in life sciences extended with respect to human biology, human genetics and genomics, human molecular pathology, and be proficient enough in specific requirements for diagnostic genetic testing. At the same time, each medical geneticist, being a specialist who

provides a specific set of health care services (namely, genetic testing) at a more general level, belongs to the diagnostic laboratory testing system and, ultimately, is a health services professional. Therefore, the system of such specialists' education and training must also ensure adequate level of competencies specific for diagnostic laboratory testing, and competencies specific for health service professionals. Analysis of the current situation in the field in EU countries, performed by the top-level specialists under the EuroGentest, an EC FP6-supported Network of Excellence for Genetic in Europe[1] aimed at harmonising genetic testing services, has resulted in the statement that such a system of education and training is still absent in the EU. Genetic laboratory services in the EU and other countries are largely provided by a mixture of PhD scientists, MD/PhD clinician scientists and technicians with different requirements for their educational background and scientific degree. None of the existing Master studies programmes (e.g., Genetics, Molecular Biology, Biochemistry or Medical Biology) most often specified in the diploma of the non-MD specialists providing genetic testing include all the above-stated aspects essential for a medical geneticist. For example, geneticists have no background knowledge in laboratory medicine and are not familiar with specific requirements for diagnostic genetic testing, while medical biologists have inadequate knowledge and skills in human genetics. As a consequence, a number of problems emerge with a considerable variation in number and depth among countries. The most immediate problems are as follows. (1) Graduate non-MD medical geneticists must be further trained at their working place, but within limited resources of state health care most often there is no system and budget for such training. (2) System for the accreditation of health services in some countries (e.g., Lithuania) requires the presence of national standards for a health service defining professional core competencies and responsibilities of the specialists. Such standards cannot be developed for the specialty "Medical Geneticist" in the absence of the established system for specialists' education and training. As a consequence, a system for medical geneticists' certification and continued education in such countries is also absent. (3) There are persisting problems in a specialist's mobility within the EU without a certificate of EU-level qualification in medical genetics. Thus there is an urgent need to develop an adequate EU-level system for training non-MD specialists in medical genetics.

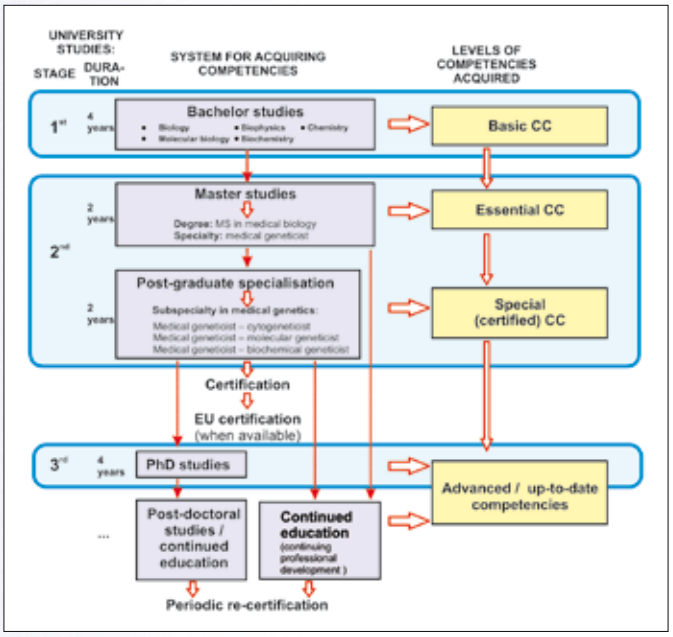

Fig. 1. A scheme of different level core competencies (CC) of non-MD medical geneticists and the system of university based education to acquire them

Outline of the system for education and training non-MD medical geneticists

The establishment of core competencies is currently being used as a basis of undergraduate and postgraduate medical training for health professionals in many countries in line with the three cycle system (Bachelors-Masters-Doctorate) for non-MD specialists. Taking this into account, a competencies-based three subsequent levels of core competencies for non-MD medical geneticists, alongside the stages of education and training to acquire them, is being suggested (see Figure) as a result of the analysis of the experience of a number of countries, discussions and collaboration under the EuroGentest Unit 6.2 "Professional Perspective"[2], ESHG Educational Committee and GenEd project[3]. A set of core competencies that could apply as an appropriate framework to geneticists responsible for laboratory services is currently under development by the experts under the EuroGentest project Unit 6.2 and ESHG Education Committee. On the other hand, a system to attain such competencies is still to be created and curricula for EU are to be set.

While the central (2nd) stage in the proposed scheme is a new Masters studies programme in Medical Genetics, admission requirements for this programme are graduating from an accredited bachelor studies programme (1st stage) in a university with life sciences orientation. Bachelor studies should result in acquiring knowledge base of fundamental principles and approaches (i.e. basic competencies) to serve as a foundation for subsequent Masters studies programme in Medical Genetics. Bachelor examinations essential to enter the master studies programme should be in genetics, cytology, molecular biology, biochemistry. If one or more relevant courses are not included in an applicant's bachelor studies curriculum (e.g. after graduating from Bachelor studies programme in chemistry), passing relevant examination(s) after completing (or without) bridge course(s) can compensate the difference.

[1] Godard B, Kaariainen H, Kristoffersson U, Tranebjaerg L, Coviello D, Ayme S: Provision of genetic services in Europe: current practices and issues. Eur J Hum Genet 2003; 11 (Suppl 2): S13– S48.

[2] http://www.eurogentest.org/

[3] http://www.medicine.manchester.ac.uk/gened/

The 2nd stage comprises two steps. ***Step 1*** is a Masters studies programme resulting in the MS degree in medical biology and acquiring the specialty "Medical Geneticist", which ensures a set of essential core competencies of a medical geneticist. Graduates of Masters studies programme in the speciality of Medical Genetics will be front-line staff qualified to be employed at the governmental and private laboratory facilities of health care institutions providing genetic services and biomedical research and educational institutions.
Step 2 is post-graduate specialisation studies programme (comparable to residency studies programme for clinical specialties in the system of integrated studies) ensuring a high level knowledge and expertise in one of the basic specialisations (subspecialties in medical genetics): molecular genetics, cytogenetics, or biochemical genetics by extending core competencies to a special level, which enables certification (see below). Principal courses of master studies should be 1) molecular genetics and DNA diagnostics in medicine, 2) clinical cytogenetics, 3) biochemical genetics, 4) bioinformatics, 5) essentials of genetic counselling. Additional courses should be basic disciplines in laboratory diagnostics. Students' exchange between EU universities (other developed countries may also be involved via Erasmus programme) should be encouraged, if not being a compulsory part of a Master studies programme.
Specialists graduating the 2nd step of the 2nd stage university studies programme and passing qualification examinations will be able to apply for certification by a recognised national certification body or national testing agency. Current certification (possibly time-limited certification) should be maintained on regular intervals (e.g., five years) by continuing education units.
Such a system is expected to be flexible enough to be revised and modified in the light of the EU level developments, changes related to new achievements in human genetics and genomics and related fields, changes in national and EU policy and legislation related to health care and education.
While a set of core competencies for non-MD medical geneticists is already under development by the EuroGentest Unit 6.2 and ESHG Education Committee, it is reasonable to initiate a project under the Erasmus programme[4] to develop a corresponding EU-level Masters studies curriculum in Medical Genetics.
Alongside with core competencies and for non-MD medical geneticists and education system to acquire them, there are some other important questions in ensuring quality, availability and accessibility of genetic testing services. In the first place, it is the status (recognition) of the specialty aimed to provide a service and the system of specialists' certification.

Recognition of medical genetics as medical specialty

Clinical/medical genetics (in the sense of a MD-based competency to offer specialised genetic services) is officially recognised as a medical specialty in many EU countries (including Lithuania), and specialist genetic physicians (MDs) can be trained through a system for post-graduate training (most often residency in clinical/medical genetics). Nevertheless, Medical Genetics is not currently a specialisation listed in Annex C of Directive 93/16/EEC[5], which denotes a medical specialty recognised at the EU level. What regards recognition of medical genetics as a non-MD laboratory-based specialty with competency to offer diagnostic genetic testing, the situation is more complicated. There are just few known cases of the recognition of the specialty on national level. Thus there is an urgent need to recognise Clinical/Medical Genetics as both a clinical specialty and a laboratory specialty on EU and national levels.

Non-MD medical geneticists' certification

Alongside with the accreditation of diagnostic laboratories, which provide genetic testing, individual accreditation, board certification, state and international registration of laboratory personnel are being acknowledged as an important instrument relevant to regulation of quality assurance in genetic services. Currently there are significant differences in the field across EU as well as in other countries.
There is a strong need for developing national and international (particularly important for relatively small countries such as Lithuania) networking for rare diseases in order to give access to reliable clinical and laboratory resources. Accordingly, an EU-level performance in genetic testing should be ensured in different countries by establishing equivalence between jurisdictions in specialists' qualification. On the other hand, mobility of non-MD medical geneticists from Member States across EU is essential. Thus it is evident that an EU system for non-MD medical geneticists' certification (e.g., similar to that applied in the U.S. for laboratory specialties of Clinical Biochemical Genetics, Clinical Cytogenetics and Clinical Molecular Genetics[6] and/or that already in action in EU for specialists in clinical chemistry and laboratory medicine[7]) is necessary. Such certification would allow entry to an EU specialist register of medical geneticists (more exactly, a special section corresponding to the specialisation). Registered specialist would be able to work in any clinical/medical genetics laboratory across EU. At the same time EU-level certification would be an official document confirming specialist's qualification in his/her native country.

Concluding remarks

It is evident that a contemporary education and training system for non-MD medical geneticists is necessary in EU countries. Such specialists could be trained according to a Masters studies programme in Medical Geneticists developed by joint activities of several countries under Erasmus programme. Those who are interested in the development of such studies programme and in the education of non-MD medical geneticists are kindly asked to contact authors of this article to join efforts in achieving the EU level recognition of non-MD medical geneticist as a health care specialist with competency to offer health care services.

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[4] http://ec.europa.eu/education/programmes/socrates/erasmus/curriculum_en.html

[5] Council of the European Communities. Council Directive 93/16/EEC of 5 April 1993 to facilitate the free movement of doctors and the mutual recognition of their diplomas, certificates and other evidence of formal qualifications. OJ L 165, 7.7.1993, p. 1–24.

[6] American Board of Medical Genetics (http://www.abmg.org/)

[7] Gurr E, Koller U, Blaton V et al.; European Communities Confederation of Clinical Chemistry and Laboratory Medicine (EC4), EC4 Register Commission. The European Register for Specialists in Clinical Chemistry and Laboratory Medicine: Guide to the Register Version 2-2003 and Procedure for Re-registration, 2003; 41(2):238–47.

The Contribution of Dentists in Human Genetics

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Human genetics is undoubtedly one of the fastest growing fields of modern science. As a human endeavor, it involves people who are making or have made things happen. One such group of specialists who are major contributors to human genetics, but not often mentioned, are the dentists. Their pioneering work was the subject of a Workshop on History of Genetics held last June in Nice, France, coordinated by Christos Yapijakis, during the ESHG meeting.

The History of Genetics workshops are designed to provide the younger geneticists with insights about the origin and adventures of thought, as well as a sense of continuity in the quest for scientific understanding of the human inheritance world. The workshop in Nice (June 18 2007) was devoted to clinical geneticists, syndromologists, and molecular geneticists, whose first degree was in Dentistry or Dental Surgery or Dental Medicine. Well-known examples of such dentists with major contributions to human genetics in the past four decades are the authors of the legendary book "Syndromes of the Head and Neck" by Robert J. Gorlin and M. Michael Cohen Jr, who have been awarded the specialty of clinical genetics in North America.

The first presentation by Euterpe Bazopoulou-Kyrkanidou (University of Athens Dental School, Greece) with the title "A Great Man, Teacher and Friend" discussed the genius pioneering work of R.J. Gorlin ("Bob" to his numerous friends) regarding the delineation of about 50 syndromes (involving mostly, but not only, head and neck), his approach to clinical and scientific work and his generous attitude towards friends, colleagues and students.

Agnès Bloch-Zupan (Louis Pasteur University of Strasbourg, Faculty of Dentistry, France) in her presentation "Contribution of Dentists in Craniofacial Syndromology and Human Genetics" reminded the audience about the several major contributions of dentists in the past thirty years. Starting with R.J. Gorlin and M.M. Cohen Jr and the syndromes named after them, she then described the various dental abnormalities (in number, shape, size; structure, color; root formation, eruption, resorption), the clinical characterization of an orofacial condition such as otodental syndrome, its linkage to a particular locus (11q13) and finally, the positional cloning of the responsible gene *FGF3*, a collaborative work she participated in with C. Gregory-Evans et al (Imperial College London, UK). The contributions of K. Storhaug (Tako Center, Norway) in ectodermal dysplasias and various other conditions, Roger K. Hall (Melbourne, Australia) in solitary median maxillary central incisor syndrome, Inger Kjaer (Copenhagen, Denmark) in normal and pathological craniofacial skeletal development, Mike Dixon (Manchester, UK) in cleft and lip palate syndromes and dentinogenesis imperfecta, Peter J.M. Crawford (Bristol, UK), Michael Aldred (Melbourne, Australia) in amelogenesis imperfecta etc as well as from other colleagues, worldwide (E. Bouvy-Berends, Netherlands and J.A. Baart, M. Hoff, E. Etty, C. Carels, Belgium) was presented. In addition, Agnès Bloch-Zupan discussed the current protocols used by geneticist-orientated dentists for rare diseases, as well as the role of the dental practitioner in diagnosis, advice, management, prevention, and referral of genetic disorders with oro-cranio-facial manifestations. The specialist dental expert who is able to recognize orodental anomalies is an indispensable partner of the multidisciplinary team working with a particular patient. In Europe, some of these professionals belong to networks linking clinical diagnosis centres and research laboratories [European Cooperation in the field of Scientific and Technical Research, COST Action B23 Oro-Facial Development and Regeneration; in France, INSERM (French Medical Research Agency) "Réseau de Recherche Clinique et Réseau de Recherche en Santé des populations 2003"; GIS maladies rares, Odontogenetics network; French National Rare Disease Plan 2004-2008 certificating in 2006 a Reference Centre for Orodental Manifestations of Rare Diseases in Strasbourg (M.C. Maniere) and in 2007 a Reference Centre for Rare Malformations of the Face and the Oral Cavity in Paris (M.P. Vazquez)].

In his presentation "Dentists and Human Genetics in Europe", Christos Yapijakis (University of Athens Medical School, Greece) discussed the obtained data from a European survey on the dentists who are working in various fields of human genetics. As the only member of the ESHG Board with a dental degree, Yapijakis prepared a questionnaire that was sent to several dental specialists all over Europe. Twenty-nine doctors in Dental Surgery or Dental Medicine from five countries (Belgium, Denmark, France, Greece and Norway) responded. The majority of them (93%) had a PhD, while a further 3% were PhD candidates. One fifth of them (21%) had two additional degrees, i.e. a Masters and a degree in Medicine or Biology. Most of the surveyed dentists (60%) had an academic position (Assistant, Associate and Full Professors), mostly in Dental Schools (57%) but also in Medical Schools (3%). Regarding their current specialization, 41% were specialized in pediatric dentistry, 31% in orthodontics, 10% in oral medicine and oral pathology, 10% in oral developmental biology and 8% in head and neck syndromology. With regards to their experience in clinical genetics, 10% had more than 20 years, 49% had 11-20 years, 10% had 4-10 years and 31% had up to three years. All of them were involved in human genetic research and were interested in obtaining an "orofacial clinical genetics" specialty. In conclusion, European dentists who are involved in almost all aspects of orofacial human genetics are well educated, with about 2/3 of them having experience in clinical genetics and capable of contributing a great deal in human genetics if they are given the chance.

The ESHG Education Committee headed by Domenico Coviello will pursue the establishment of parallel European specialties of clinical genetics for physicians and orofacial clinical genetics for dentists. Agnès Bloch-Zupan and Christos Yapijakis are participating in this Committee.

ESHG and Advanced Training in Europe

Alessandra Renieri
ESHG Board Member

One of the most important missions of a Scientific Society is to encourage advanced training. The ESHG has sponsored since the early 90' the European School of Genetic Medicine whose courses were held initially in Sestri Levante (Genoa) and later on in in Bertinoro and Ronzano, two new venues of the School in the proximity of Bologna. The courses of this School are taught by some of the most famous geneticists including Victor McKusick and Mario Capecchi, Nobel Prize awarded for Medicine in 2007 (Fig.1) .

The School was started in 1988 by Giovanni Romeo with the first one week of an intensive course in Medical Genetics which was characterized by continuous student-teaching interactions not only during formal morning lectures and afternoon workshops but also during lunch and dinner-time discussions (not to mention the inspiring individual or group meditations on the splendid beaches of Sestri Levante…). Starting from 1997, the main course in Medical Genetics was flanked by other courses in specific fields of Genetic Medicine such as Cancer Genetics, Statistical Genetic Analysis, Genetic Counselling, Molecular Cytogenetics, Bioinformatics, etc., thus fulfilling the mission implicit in the definition of the European School of Genetic Medicine (see: www.eurogene.org). Every year about 500 students from all over Europe and from countries of the Mediterranean region and the Middle East attend these courses. In addition, most of these courses can be followed via Internet by Remote Training Centers organized by the School in different parts of the world and the "remote" students can ask questions to the speakers by e-mail and have answers in real time. Usually the students in Remote Training Centers outnumber those attending the "real" course by a factor of two or three thus bringing the total number of attendees to 1000-1500 attendees per year. The ESHG grants a variable number of fellowships per year (5-20) encouraging participation from developing countries to the "real" courses which give to students the advantage of a direct and continuous interaction with faculties, in the spirit of the original course started 20 years ago in Sestri Levante.

Beside the promotion of the European School of Genetic Medicine, the ESHG endorses an increasing number of advanced training initiatives in the two main fields of Human Genetics: Clinical and Laboratory Genetics. The ESHG endorses initiatives on the basis of the following rules: i) the Organizer should be a member of the ESHG; ii) the Board members should include non-local teachers; iii) the ESHG Education Committee should be involved in the process of endorsement.

For Laboratory Genetics, Eurogentest produces very valuable teaching materials (www.eurogentest.org). One of the recent initiatives in Clinical Genetics deserved to MD graduate students is the inter-University Master in Clinical Genetics:
Multiple Congenital Anomalies. The Master lasts one year and students are actively involved one week per month. It is held in several Medical Genetics Units spread all over Italy: Benevento, Bologna, Genova, Milano, Padova, Roma and Siena (Fig.2). During the week, students have very few formal lessons (in English) and a number of scheduled external clinics and in-ward clinics. They have discussions with their tutor before and after each clinic, and they are involved in writing the family report. Two things make this Master very innovative and appreciated by students: the itinerant shape and the practical approach (real patients and real consultations). In order to preserve this last characteristic admitted students are only 5 per year.

In addition to the endorsement of the education initiatives all over Europe, the ESHG itself may organize or co-organize courses. The proposal should be submitted to the Education Committee who after a positive evaluation asks the Executive Board for the final approval and financing.

Alessandra Renieri, M.D., Ph.D.
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Fig. 1: Group picture taken on the occasion of the 20th Course in Medical Genetics organized by the European School of Genetic Medicine (left). Mario Renato Capecchi and Rettore Pier Ugo Calzolari on the occasion of the Laurea ad honorem in Medical Biotechologies awarded by the Alma Mater Studiorum-University of Bologna on May12, 2007 in Bologna, Italy (right)

Fig. 2: Teaching board of the Master in Clinical Genetics.
From left to right: Livia Garavelli, Han Brunner, Francesca Faravelli, Alessandra Renieri, Bruno Dallapiccola, Dian Donnai, Domenico Coviello, Heather Skirton, Manuela Priolo, Faustina Lalatta, Maurizio Clementi, Romano Tenconi, Gianni Romeo, Marco Seri, Gioacchino Scarano.

Medical Genetics in Serbia

Jelena Milasin

In 2008 the Serbian Medical Genetics Section will celebrate 40 years. The Serbian Genetic Society, as part of the Yugoslav Genetic Society, was founded in 1968. It presently consists of five Sections: 1) Breeding Organisms, 2) Population and Evolutionary Genetics, 3) Medical Genetics, 4) Mutagenesis and Genotoxicology, and 5) Molecular Genetics. At this time the Medical Genetics Section counts 100 members. Even before the establishment of the Section, medical genetics and more specifically medical cytogenetics started to develop very rapidly as the result of a successful collaboration between medical doctors and biologists. Most of the laboratories dedicated to cytogenetic investigations were created in our country in the period between 1960 and 1970. The very first cytogenetic laboratory was launched at the Military Medical Academy in 1960 by Professor M. Kicic. In the early sixties, tissue culture techniques were introduced for the first time, aimed at obtaining chromosome preparations.

One of the pioneers in this field was Professor B. Garzicic, later on tutor of many cytogeneticists in the west Balkan region. In 1964, Professor Slavka Moric-Petrovic inaugurated the first genetic counseling at the Institute of Mental Health. First amniocenteses were initiated in 1979, and from 1980 amniocentesis is a routinely performed prenatal diagnostic procedure. Choriocentesis and cordocentesis appeared in 1987. Today the biggest centers studying genetic diseases, both at the molecular and cytogenetic level, from a scientific as well as diagnostic point of view, are located in Belgrade. These are: Military Medical Academy, Institute of Mental Health, Mother and Child Health Institute, Clinical Center Zvezdara, Institute of Nuclear Sciences, Institute of Molecular Genetics and Genetic Engineering, Institute of Oncology and Radiology, etc. Well developed and organized medical genetics services connected to apposite Medical Schools exist also in other big cities of Serbia such as Novi Sad, Nis and Kragujevac. In smaller towns such as Cuprija, Jagodina, Kraljevo, and Subotica, etc. cytogenetic laboratories are also spreading out in recent years.

Fortress of Golubac

Zlatibor

Education:

Undergraduate studies: Human Genetics is an obligatory, one semester course at several Schools of Medicine and Dentistry in Serbia (the Universities of Belgrade, Novi Sad, Kragujevac and Pristina). At the Medical School of the University of Nis, first year students are trained in Molecular and Human Genetics. The School of Pharmacy has also an obligatory 1st year subject-Biology and Human Genetics and the School for Special Education and Rehabilitation has an obligatory course in Medical Genetic. Optional courses in Medical, Clinical, Human and Behavioural Genetics exist at the School of Biology (Belgrade), Medicine (Novi Sad) and Psychology (Belgrade), respectively.

Postgraduate studies: There is a possibility to acquire a master degree in Medical Genetics at the School of Medicine in Belgrade (2- year programme), whilst the School of Biology proposes a master programme in Human Genetics.

PhD studies: Several courses related to the genetics of men are included in PhD programmes, for instance in Molecular Medicine (Schools of Medicine, at the Universities of Belgrade, Novi Sad and Nis), Basic and Clinical Studies (School of Dentistry, University of Belgrade), but there is no exclusively Human Genetics PhD curriculum.

Specialization: There is a 2 year specialization in Genetics for biologists working in hospitals, organized at the School of Biology, University of Belgrade. The School of Medicine (University of Belgrade) offers a subspecialization in Clinical Genetics, lasting 12 months, for medical doctors, following an obligatory 4-year specialization. Although there is no specialty in Clinical or Medical Genetics, most of the specialties have in their curricula courses in Human and Medical Genetics adapted to specific needs and profiles.

Kapaonik

Research: The research in human and medical genetics in Serbia has been always tightly connected to everyday practice. Since the foundation of the first laboratories, until the nineties of the last century, the research was predominantly focused on chromosomal aberrations in various pathological conditions. Culture techniques for lymphocytes, fibroblasts, bone marrow cells, solid tumors, and pleural effusion cells in cancer patients have been rapidly developed. For 20 years, GTG banding was the main technique applied to cytogenetic studies in chromosome syndromes, leukemias, solid tumors, sterility, etc.

River Uvac

It is worth noting that the Institute of Mental Health has the biggest repository of Down syndromes in the Balkans. Subsequently other techniques and tests have been introduced, such as FISH, Ag NOR, sister chromatide exchange, etc. Fifteen years ago, molecular genetics started its expansion and many laboratories brought in novel PCR-based techniques for molecular diagnostics, while maintaining their work in the field of cytogenetics. In molecular genetics, the tie between diagnostic procedures and research is even stronger and various publications are emerging (for instance "Deletion and duplication screening in the DMD gene using MLPA" by Lalic T. et al., Eur J Hum Genet). Molecular tests for the most frequent monogenic diseases are routinely applied (cystic fibrosis, Duchenne and Becker muscular dystrophies, neurofibromatosis, neurodegenerative disorders specially those related to trinucleotide expansion, hemoglobinopathies, etc.). Sometimes, several laboratories are doing the same analyses while sometimes there is exclusivity in the tests. The research in the field of cancer genetics is also very important. Mutation screenings in cancer genes have been done in leukemias, solid tumors and myelodysplastic syndromes. New mutations have been found in the latter group. Most recently, large scale screenings of various gene polymorphisms (apo E, MTHFR, GSTM, CYP etc.) have been undertaken and pharmacogenetics is taking its first steps. Members of our section are engaged also in forensic medicine.

Quality control: There are no systematic quality assessments, although some laboratories undergo sporadically external quality control by the European Molecular Genetics Quality Network (EMQN) or similar institutions. For instance, the performance of the genetic laboratory of "Mother and Child Health Institute" is controlled by the Cystic Fibrosis European Network.

Legislation: Apart from the law on genetically modified organisms, genetics in general does not have its place in the Serbian laws yet. Serbia has signed the *Convention for the protection of human rights and dignity of human beings with regard to the application of biology and medicine* of the Council of Europe. There is also a national bioethical committee that works according to guidelines from UNESCO (Guide no1 on servicing bioethical committee).

Belgrade

A Geneticist -Professor Veronica van Heyningen- elected a Fellow of the Royal Society

Veronica van Heyningen
Former President of the ESHG

The Royal Society elected on 17 May 2007 forty-four new Fellows and eight Foreign Members from the fields of science, engineering and technology. Among them is Professor Veronica van Heyningen, a long time member of our Society who has served as member of the Board and President of ESHG. Her election brings great honor to our Society.

Veronica van Heyningen began human gene mapping in 1970 during her PhD studies with Walter Bodmer. Later disease-gene identification led to her odyssey around the PAX6 eye and brain development gene implicated in aniridia. Identification of other major transcription factors, SOX2 and OTX2, followed. Detailed study of PAX6 mutations led to a broader interest in long range regulation of gene expression, while the non-Mendelian patterns of inheritance often seen for reduced or absent eyes pointed to a study of how cryptic mutations may be uncovered and modulated by environmental factors that require stress response gene activity. Veronica has progressed from postdoc to Head of the Medical and Developmental Genetics Section, MRC Human Genetics Unit over the past 30 years. She has been honoured in several ways, and among these she was the proud recipient of the ESHG prize in 2006. This year she was elected a Fellow of the Royal Society, which is often considered the ultimate accolade for UK scientists.

The Royal Society's foundation (which dates back to the 28th of November 1660) is its Fellowship, which is made up of the most eminent scientists, engineers and technologists from the UK and the Commonwealth. The history of science since 1660 is closely intertwined with the story of the Royal Society. According to the Society's statutes, candidates for election to the Fellowship must have made "a substantial contribution to the improvement of natural knowledge, including mathematics, engineering science and medical science". Each year, the Fellows elect 44 new Fellows and six new Foreign Members, chosen for their scientific achievements - an honour that is regarded as the highest accolade a scientist can receive next to a Nobel Prize. There are currently 1317 Fellows, only 66 of whom (5%) are women. Over the last 7 years, 10% of new Fellows elected to the Royal Society have been women (33 of the 344 Fellows elected).

Welcome to the new board members

Professor Gerry Evers-Kiebooms, PhD Psychology, started working in genetics in 1977 and is currently head of the Psychosocial Genetics Unit in the Genetic Clinic of the Center for Human Genetics of the University of Leuven, Belgium. She has been trained as a mathematical psychologist and has a strong interest in methodology and psychological decision making.

The three major objectives of her unit are: 1. Psychological counseling and psychosocial support in the context of a multidisciplinary team approach to predictive and diagnostic testing for autosomal dominant late onset diseases (neurodegenerative disease, hereditary cancer and hereditary heart disease) 2. Research about psychological aspects of genetic risk perception, decision making involving genetic risk and longitudinal studies evaluating the psychological impact of predictive testing and (3) Genetic education including presentations, leaflets, etc. for the public and directed to specific target groups. She has a long list of publications in peer reviewed international journals (see www.kuleuven.be/Psychogen.) She was the coordinator or promoter of several national, European and international projects, mainly focused on predictive testing and prenatal testing for late-onset disease. Over the years she organized several national, European and international meetings that usually resulted in proceedings published by international publishers. She was an invited speaker or invited expert in many meetings and commissions in Europe.

She is a member of the editorial committee of some European journals, a regular reviewer of papers as well as scientific proposals and a member of the scientific programme committee of meetings focused on the psychosocial aspects of genetics. Over the years genetic testing for Huntington's disease has been a major focus in the activities of the Psychosocial Genetics group. Recently she has taken the initiative to start a Working Group on Genetic Testing for HD within the European Huntington disease network (Euro-HD).

She is a member of the Public and Professional Policy Committee of the ESHG from the start of its activities about 10 years ago. At a European level she has promoted the role of the psychosocial staff in genetic centres and she realized the conjunction between the European Meeting on Pychosocial Aspects of Genetics (EMPAG) and the European Conference of the ESHG since the meeting in Strasbourg. This successful conjunction was and is supported by several ESHG-members with a key role in the society, including Ségolène Aymé.

Over the years her interest in the ethical aspects of genetics has increased steadily. She is a member of the Belgian Advisory Committee on Bio-ethics and in this context co-president of many commissions preparing advice about genetics-related topics. More recently she was designated as a member of the Belgian Federal Commission for the Research on embryos in vitro.

Professor Giuseppe Novelli is Full Professor and Chair of the Medical Genetics Laboratory at the Tor Vergata University of Rome and Adjunct Professor, Division of Cardiology, University of Arkansas, Little Rock, (USA). Dr. Novelli received his B.S. degree from Urbino University in Italy and then went on to do his Ph.D. in Medical Genetics at the "La Sapienza" University in Rome. Dr. Novelli performed his postdoctoral training at the INSERM U73, Paris (France). Dr. Novelli has worked towards deciphering the molecular basis of disease and disorders, using genetic, developmental, biochemical, and cell biological tools. The research in his laboratory today focuses on understanding the genomic basis of laminophaties and complex diseases such as psoriasis and atherosclerosis. His research led to the identification of different genes, responsible for human rare diseases. The knowledge acquired about how mutations lead to phenotype and information about the proteins these genes encode is being applied towards the development of therapeutic techniques for the treatment of laminopathies.
Professor Novelli is member of the Pharmacogenetics working party at the EMEA, the European European Medicines Agency and member of the Board of the American Society of Gene Therapy (ASGT) and member of the Editorial Board of Clinical Genetics, Acta Myologica, Expert Opinion on Pharmacotherapy, and BMC Medical Genetics.

Maria Soller, M.D., Ph.D. studied medicine at the Lund Medical Faculty and started her PhD project at the Department of Clinical Genetics in 1989. Professor Felix Mitelnam and Professor Sverre Heim supervised her work in the characterization of cytogenetic aberrations in lung tumors. After her PhD she completed her medical studies and moved to The Department of Clinical Genetics at Karolinska Hospital, Stockholm under the direction of Prof Magnus Nordenskjöld to begin her residence in Clinical Genetics. In 2000 she moved back to the Lund University Hospital to join the clinical staff at the Department of Clinical Genetics. Since then her main interest has been clinical work and education, in addition to research involving both constitutional and acquired genetics. Maria Soller is currently Director of Medical Interns at the Lund University Hospital and senior consultant and the Vice Head of the Department of Clinical Genetics.

New Executive Officer of the ESHG

The ESHG has appointed **Mr. Jerome del Picchia** as Executive Officer in order to improve, facilitate and ease the internal workflow within the Society. His tasks will include the following.

1. Uphold the guiding principles of the Society in working to maintain the highest standards of ethical practice and academic excel lence in the area of human genetics and its clinically related areas.
2. Seek to ensure decisions taken by the elected officers and by the Board of the ESHG are acted upon in an effective and timely manner.
3. To this end, the Executive Officer shall
   a. Ensure, in close collaboration with the General Secretary, that an agenda and minutes are circulated to the ESHG Board for all scheduled meetings.
   b. ensure that all major meetings are properly documented.
   c. maintain an accurate record of the contact details of all elected representatives and maintain regular contact with them.
   d. maintain an accessible record of key European organisations, especially an up-to-date list of the officers of national human and medical genetics societies.
   e. facilitate the production of published reports, newsletters, etc.
   f. maintain an up-to-date website.
   g. attend in person all meetings of the Board and Executive Board.
   h. meet in person with the General Secretary at least once each year other than at meetings of the Executive Board.
   i. ensure that new Board members and elected officers are provided with an induction pack. containing details of all committees and working parties, any recent publications of the Society, a copy of the constitution and the expected contribution of the individual concerned.
   j. Provide administrative support to committees.
4. The Executive Officer will be accountable to the Board through the Secretary General.

Jerome del Picchia, 38, born in Vienna, father of 2 daughters, is a graduate of the Lycée Français de Vienne. Due to this bilingual education, his knowledge of French and German is excellent and hence a great asset for his later professional endeavours. After studying economics at the University of Vienna, he worked in the Austria Center Vienna from 1987-1989. His career in the Vienna Medical Academy began in 1989, being a conference manager. In 2004, after 15 years of continuous success in the international conference industry, he was appointed Executive Director of the VMA and shares this position's responsibilities with Ms. Romana König. His first contact with the ESHG was during the 10th ICHG meeting in Vienna in 2001. His ESHG activities are currently covering general organisational matters and handling of abstracts at the annual meeting, and more recently, the position of the Executive Officer. Mr. del Picchia's email address is eo@eshg.org.

Jerome del Picchia (center, seating, and the VMA team)

E-RARE Consortium Activities and Featured Actions

Nurten Akarsu
Board Member of the ESHG

Funds for the work on rare disorders are as rare as the disorders themselves, especially when compared with common diseases. Despite the fact that they are rare, they represent a considerable burden on health care systems especially in emerging countries. Therefore, they are excellent candidates bridging the gaps in the European Research Area. Although their nature perfectly fits to multidisciplinary and transnational research activities, they did not take much attention in previous framework programmes and network type of actions. Fortunately, last year an ERA-NET action in the Sixth Framework programme (FP6) announced a call, specifically for rare disorders named E-RARE (ERA-Net for research programs on rare diseases). Nine full partners (Belgium, France, Germany, Israel , Italy, Spain, The Netherlands, and Turkey) and two affiliated partners (Region of Lombardy, Italy and Russian Federation) currently participate in the consortium. Over 150 applications were received and a total of 13 projects have been selected for funding (for details: http://www.e-rare.eu/cgi-bin/index.php).

ERA-NET scheme aims to build a transnational network of research councils between the Member or Associated States through the networking of research programmes. Programme "owners", such as ministries/governments, or programme "managers", such as research councils or funding agencies, are eligible to be partners of the consortium, whereas research organisations or scientists are not. Therefore, this scheme is an excellent opportunity to draw attention of decision makers to rare disorders. One major question is whether or not a follow-up proposal will be considered in the continuation of E-RARE projects. Four steps of actions are desired in ERA-NETs: (1) information exchange; (2) definition and preparation of joint activities; (3) implementation of joint activities; and (4) funding of joint transnational research actions. It seems that a follow-up proposal under FP7 could be available for ERA-NETs launched under FP6, if they propose a strong coordination of action directly starting with steps three or four. As researchers, we thank the E-RARE consortium for their initiative effort, which will hopefully lead to a continuation under FP7, constructing a common strategy for funding, common calls for proposals, and a multinational evaluation system focusing on rare disorders.

We are pleased to announce that principal investigators of four funded projects of the E-RARE consortium are ESHG members and brief summaries of their projects are included below.

Nurten Akarsu, Ankara 2008

EuroRETT : European Network on Rett Syndrome

Dr. Laurent Villard,
EuroRETT Coordinator
INSERM U910
Faculte de Medicine
La Timone
Marseille, FRANCE

Rett syndrome (RS) is a severe neurological disorder primarily affecting girls, with an incidence of about 1/10,000 female births. 250 girls affected by RS are born each year in the European Union. Rett syndrome is caused by mutations in the methyl-CpG binding protein 2 (*MeCP2*) gene or the *CDKL5* gene.
MeCP2 encodes two closely related proteins who act as transcriptional repressors. *CDKL5* encodes a kinase which mediates MeCP2 phosphorylation. However, the mechanisms leading to the severe, progressive and specific neuronal dysfunction when these genes are mutated are currently unknown. Several mouse models of RS have been generated. These models were used to show that the phenotype was due to Mecp2 dysfunction in post-mitotic neurons. Recent key experiments demonstrated that re-expressing Mecp2 in the knock-out mouse displaying overt symptoms was able to reverse disease progression. This possible reversibility fully justify the development of therapeutic approaches for this disorder, especially pharmacological interventions. European teams are strongly involved in this research area.

Our network joins the forces of 10 research groups in Italy (Milano and Roma), France (Marseille and Paris), Spain (Madrid), Germany (Berlin and Göttingen) and Israel (Tel Hashomer). Our objectives are:
1- to improve phenotype-genotype correlations (building a large European cohort of patients)
2- to study chromatin organization and identify MeCP2 and CDKL5 targets and interactors,
3- to understand neuronal dysfunction in RS,
4- to develop therapeutic approaches for RS.

Rett syndrome is a model for autism-spectrum disorders. It is a severe phenotype for which there is currently no efficient treatment but that could be reversible. The strong commitment of parent associations to support research has generated a huge interest for the condition. The EuroRETT E-RARE network gives us the opportunity to organize research efforts at the European level to fight against this devastating neurological disease.

EUROSPA: European and Mediterranean network on spastic paraplegias

Pr Alexis Brice
EUROSPA Coordinator
INSERM U679
La Salpêtrière Hospital
Paris, France

The E-Rare EUROSPA project is a European and Mediterranean network dedicated to research on Hereditary Spastic Paraplegias (HSP). HSPs are a clinically and genetically heterogeneous group of rare diseases in which affected corticospinal tracts provoke a functional gait handicap frequently associated with various other neurological symptoms. Very few symptomatic treatments exist against these disabling diseases, leading to the social isolation of the patients and their families. Improving this situation is the key objective of the EUROSPA project, with the strategy to focus on unravelling the genetic basis of the clinical heterogeneity of HSPs and providing a sharp phenotypic description of each genetic entity.
This transnational project is based on the networking of four reference clinical and research teams led by Pr Alexis Brice (Paris, France), EUROSPA coordinator, Pr Ludger Schöls (Tübingen, Germany), Pr Filippo Santorelli and Pr Enrico Bertini (Rome, Italy) and Pr Alexander Lossos (Jerusalem, Israel). The force of EUROSPA is to combine the biological resources, manpower, knowledge and expertise gained by those four member teams within two recognized networks, SPATAX and GeNeMove, devoted respectively to spinocerebellar degenerations and movement disorders. The added value of EUROSPA is its focus on HSPs and its extended geographical coverage including Europe, North Africa and the Middle East, with the largest collection of recruited HSP families known worldwide (currently 1300). Based on these assets, the main following aims will be pursued: (1) to enlarge the clinical and molecular spectrum of known HSP genes through wide genetic testing, thanks to the development of a resequencing microarray (Tübingen); (2) to identify new loci and responsible genes in pure dominant HSP forms (Tübingen), as well as in recessive pure and complex forms (Paris), specifically in spastic ataxia (Rome) and (3) to establish genotype-phenotype correlations in the largest number of sampled families and improve the nosology of these complex disorders.

The results of these studies will offer to HSP patients and their families an increased possibility for a molecular diagnosis. They will also provide new clues to understand the deleterious cellular mechanisms involved in the pathology, from which to gain the fundamental knowledge needed for future curative treatments.

HSCR - Genetics and molecular bases of oligogenic Hirschsprung disease: from transcriptome to modifier genes

International Hirschsprung Disease Concortium
E-Rare Coordinator: Pr Stanislas Lyonnet
INSERM U-78, Paris Descartes University, Hôpital Necker-Enfants Malades, Paris, France

In complex disorders with multifactorial inheritance, common predisposing genetic variants such as SNPs are combined to result in the disease phenotype, and to modify the recurrence risk. This is the case for Hirschsprung disease (HSCR, 1/4-5,000 live births), characterized by the absence of an enteric nervous system. The goal of our Consortium is to study the molecular bases of the oligogenic predisposition to HSCR, and, thereby, to identify genes involved in the development of the neural crest. The rationale for this study is twofold: i) among orphan disorders, HSCR could be regarded as a model for multigenic inheritance of malformation, as it is clearly inherited as a sex-modified multifactorial trait, ii) HSCR is a typical anomaly of neural crest development, whose elucidation may certainly benefit to less frequent neurocristopathies.

Our research programme involves:
- Extensive mutation and polymorphism screening at the major locus *(RET)*;
- Association and linkage studies in sporadic and familial cases, and analyses of candidate genes in the chromosomal regions where *RET* modifier loci have been mapped;
- A transcriptome approach of the highly multipotent developing neural crest cells in human to generate novel candidate genes for HSCR;
- The design of novel statistical genetic methods to deconstruct the complex inheritance of HSCR.

Our Consortium involves 6 groups on 3 continents: i) in Europe: Italy (Dr Isabella Ceccherini, Genoa), The Netherlands (Pr Robert Hofstra, Groningen), Spain (Dr Salud Borrego, Barcelona), and France (Pr Stanislas Lyonnet, Paris); ii) the USA (Pr Aravinda Chakravarti, Baltimore); and China (Pr Paul Tam, Hong-Kong). The European groups have been funded by an ERaNet programme for the eligible countries (Italy, Spain and France

CRANIRARE: An integrated clinical and scientific approach for craniofacial malformations

Dr. Bernd Wollnik
CRANIRARE Coordinator
University of Cologne
Germany

CRANIRARE aims to analyse the molecular and pathophysiological mechanisms of rare craniofacial malformations and to transmit the results into improved patient care. CRANIRARE will work on the molecular basis and gene identification of 12 selected rare craniofacial malformations (e.g. frontofacial dysostosis [MIM 229400], frontonasal dysplasia [MIM 136760], Manituba oculotrichoanal syndrome [MIM 248450], mandibulofacial dysostosis including Treacher Collins syndrome [MIM 154500], Genee-Wiedemann syndrome [MIM 263750], and Nager syndrome [MIM 154400], Pierre Robin sequence [MIM 261800], Moebius syndrome [MIM 157900], syndromic and isolated craniosynostosis [MIM 218500 and 123100], nasopalpebral-lipoma coloboma syndrome [MIM 167730], and LADD syndrome [MIM 149730]). Novel clinical entities will be also described and included.
Standardization and harmonization of clinical and molecular diagnosis, genetic counselling, and therapeutic strategies will be done using the information on over 850 patients with these diseases collected within this consortium. Several large and unique families are available for gene identification studies. We are using a combination of different up-to-date methodologies for gene identification, such as genome-wide array technologies for mapping, analysis of copy number changes, gene expression studies, and systematic candidate gene approaches based on functional analysis of signalling pathways. The pathophysiological effects of identified genes and proteins will be analyzed, and we will also search for modifying factors of clinical variability of phenotypic expression in some of these disorders.
To improve future therapeutic options for patients with craniofacial malformations, we aim to develop novel technologies for local delivery of bioactive agents to induce bone tissue regeneration and test these technologies in cell systems and animal models. We have chosen to conduct CRANIRARE by integrating partners from different specialities and countries into a multidisciplinary approach. Our network consists of six partners, three from Turkey (Nurten Akarsu, Ankara; Hülya Kayserili, Istanbul, Erhan Piskin, Ankara), two from Germany (Dagmar Wieczorek and Dietmar Lohmann, Essen; Bernd Wollnik, Cologne), and one from France (Stanislas Lyonnet, Paris).

Minutes of the Annual Membership Meeting 2007

at the EUROPEAN HUMAN GENETICS CONFERENCE 2007
Nice, France, Sunday, June 17, 2007, 19.00 – 20.00.

The President of the society, Professor John Burn, opened the Annual Membership Meeting with a welcome address. About 85 members were present.

The Secretary General, Helena Kääriäinen, summarized the activities of the society 2006-2007 and some of the committees gave their activity reports, however, detailed reports had already been distributed to the membership via the pre-conference newsletter.

Ségolène Aymé resigned as Chair of PPPC and Vice-chair Martina Cornell became new Chair. The assembly expressed warm thanks to Ségolène Aymé.

The Treasurer, Andrew Read, gave the financial report 2006 of the society which was accepted. The society's financial situation of is very good and allows a greater number of fellowships to the yearly ESHG conferences as well as increasing support to the European Genetic Foundation.
President John Burn announced the resignation of four board members, i.e. Thoas Fioretos, Milan Macek Jr., Veronica van Heyningen and Christos Yapijakis, after their five years service on the board. Also, he announced the resignation of Vice-President Professor Andres Metspalu from the Executive Board; he will serve as board member the next two years.

Thereafter, the new President, Professor Pier Franco Pignatti, was asked to chair the meeting.

Professor Pier Franco Pignatti expressed his thanks to the leaving board members, Vice-President and President (who became now Vice-President for the coming year). He reported that Professor Jean-Jaques Cassiman was confirmed President-elect and that Gerry Evers-Kiebooms (Belgium), Giuseppe Novelli (Italy), Maria Soller (Sweden) became new board members. Since there were no nominations besides the above, no voting procedure was necessary.

It was suggested that the membership fees 2008 remain unchanged for the next year. The meeting accepted.
There was a lively discussion about the sites of future European Human Genetics conferences. The 2008 conference will take place in Barcelona. Istanbul, Scandinavian countries and Vienna were proposed for the following years, however, not all details were available and the years/dates were not decided during the meeting
The proposed budget 2008 was accepted.

Future activities of the society, especially the growing collaboration with National Human Genetic Societies, were discussed and seen as a very positive new development.

President Pier Franco Pignatti closed the meeting and wished the members a fruitful year.

The ESHG Board 2007-2008

Executive Board

President: Prof. Pier Franco Pignatti
President-Elect: Prof. Jean Jacques Cassiman
Vice-President: Prof. John Burn
Secretary-general : Prof. Helena Kääriäinen
Treasurer: Prof. Andrew Read

Board Members

Ass.Prof. Nurten Akarsu, Ankara
Prof. Karen B. Avraham, Tel Aviv
Prof. Jacques Beckmann, Lausanne
Prof. Alexis Brice, Paris
Prof. Francoise Clerget-Darpoux , Villejuif
Prof. Dian Donnai, Manchester
Prof. Gerry Evers-Kiebooms, Leuven
Prof. Peter Heutink, Amsterdam
Ass.Prof. Klaus W. Kjaer, Copenhagen
Prof. Vaidutis Kucinskas, Vilnius
Prof. Nicolas Levy, Marseille
Prof. Jan Lubinski, Szczecin
Prof. Gert Matthijs, Leuven
Prof. Andres Metspalu, Tartu
Prof. Giuseppe Novelli, Rome
Prof. Leena Peltonen, Helsinki
Prof. Alessandra Renieri, Siena
Dr. Maria Soller, Lund
Dr. Silke Sperling, Berlin

Liaison Members,

Prof. Martina Cornel, Chair, PPPC
Prof. Han Brunner, Chair, SPC
Prof. Jean-Jacques Cassiman, IFHGS
Dr. Domenico Coviello, Chair, Education Committee

Editor's Letter

Lina Florentin-Arar
Editor ESHG Newsletter

Dear All,

It is almost 4 years now that I have the privilege to act as the Editor of the Newsletter of the ESHG, a task which I have thoroughly enjoyed. I have tried so far to choose and include articles that are of interest to all of you, either by continuing the series of articles on Medical Genetics of each country, professional issues which are at the moment ongoing and "burning", quality control matters, presentation of scientific groups, activities of the Society, profiles of eminent geneticists in Europe etc.

For several reasons, some of them personal as well, it was not possible to have two newsletters this year but I hope that this issue which comes as 2 issues in 1 will compensate for the delay. I would like to thank Karen Abraham and Jerome del Picchia who have worked very hard for the current issue.

I have welcomed you in the past and I welcome you once again to submit articles to our Newsletter. Do submit articles. Give us ideas. Europe is very large and diverse and Genetics is a field of continuous development. Do not imagine that our colleagues find no interest in local news. We all do.
Finally I would like to say that the ESHG newsletter has now an Editorial Board and therefore I would like from this position to welcome the two members Drs. Karen Abraham and Maria Soller.

Lina Florentin-Arar,B.Sc.,Ph.D.
Editor of the ESHG Newsletter

Invitation to the Annual Membership Meeting 2008

At the EUROPEAN HUMAN GENETICS CONFERENCE 2008

Sunday, June 1, 2008 at 7.00 – 8.00 p.m.
Room 113

CCIB, centre convenicions internacional barcelona,
Rambla Prim 1-17, 08019 Barcelona, Spain

AGENDA

Opening by the President of the Society, Professor Pier Franco Pignatti
1. Activity of the Society 2007-2008
2. Financial Report of the Society 2007
3. Discharge of the Board Members for the year 2007-2008

Opening by the new President of the Society, Professor Jean Jacques Cassiman
4. Results of election for President-Elect
5. Results of election for Board Members
6. Membership fees 2009
7. Site of future European Human Genetics Conferences
8. Budget proposal 2009
9. Major policy questions proposed by Board
10. Future activities