



# European Society of Human Genetics

November 2006 / Newsletter No. 15

The Society's Administrative Office  
ESHG c/o Vienna Medical Academy  
Alserstrasse 4, 1090 Vienna, Austria  
Tel: +43 1 405 13 83 20  
Fax: +43 1 405 13 83 23  
Website: [www.eshg.org](http://www.eshg.org)

Email Inquiries: [office@eshg.org](mailto:office@eshg.org)  
Membership Inquiries: [membership@eshg.org](mailto:membership@eshg.org)  
Online Access EJHG: <http://members.eshg.org>

## Medical Genetics in Romania

A history of medical genetics in Romania has to start by mentioning Alexandru Caratzali (1905-1980) who was among the first to recognize the Down Syndrome as a chromosomal disease, he belonged to the group led by Raymond Turpin. The same direction was later undertaken by Constantin Maximilian, (1928-1997) who was Professor of Genetics and Bioethics at the Faculty of Medicine in Bucharest, an outstanding personality with many appreciated books and articles published on eugenics in Romania. The Medical Students' Group of Bioethics in Bucharest was named after him. Contemporary representative's names are those of Professor Mircea Covic in clinical genetics and Professor Dragos Stefanescu in radiocytogenetics. The preoccupations for sharing information and experience concerning the rapidly progressing, dynamic field of medical genetics in Romania, are expressed through the increasing number of scientific meetings on that subject over the last decade.



*Professor Ioan Victor Pop, MD, PhD  
President Executive of the Romanian Society of  
Medical Genetics*

### **The Romanian Society for Medical Genetics**

was founded on the occasion of the symposium "Metabolic and Endocrine Diseases in Children" organized by Prof. Paula Grigorescu-Sido in Cluj-Napoca on the 27th October 1995. The organization reunites professionals with interest in genetics, has juridical autonomy, functions by observing statutes which are close to statutes of other similar organizations in Europe. The actual number of members is 173, forming 10 groups spreading throughout the country, most of them around university centers. The objectives of the society are:

- ◆ to develop top quality fundamental and clinical research in Romania,

- ◆ to ensure the formation of specialists at different university degrees,
- ◆ to elaborate programs for the development of a national network of diagnosis, investigation and prevention in Centers of Medical Genetics,
- ◆ to organize scientific meetings and forums,
- ◆ to issue specific publications,
- ◆ to collaborate with other national and international Societies/Associations with interests in Medical Genetics,
- ◆ to promote collaboration with associations of people with genetic/malformative diseases.



*Certeze, Village, NW Romania*

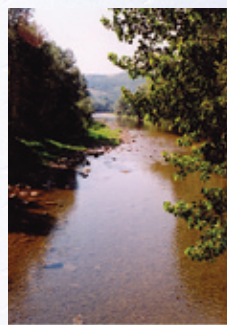


*Landscape near Sibiu, Central Romania*





*Spring Feast Oas Country, NW Romania*



*Barsana River, N Romania*

### **Research and Centers of Medical Genetics in Romania**

There are several Romanian universities that have their own research projects financed through national/international grants, there are fruitful collaborations and exchanges with universities from different EU countries. Research is focused mainly on clinical genetics and is performed either in universities or in some specialized Centers of Medical Genetics where prenatal and postnatal cytogenetic analysis is performed, there is one center specialized in lisosomal diseases, two in cystic fibrosis. The quality control is not standardized on a national level, each laboratory has its internal quality control.



Geographical distribution of the Centers of Medical Genetics.

### **Education issues:**

- ◆ **University Courses:** All Medical Universities in Romania offer Medical Genetics included in their curriculum, usually it is studied for only one semester or some universities have an additional clinical genetics module in the fifth year.
- ◆ **Residency:** In 1997 the medical specialty of Medical Genetics was officially included among the clinical specialties. Approximately 5 residents per year were admitted for a five year specialization in Romania and were sent to university hospitals up to this year when this specialization, along with others like oncology, haematology, was abolished in August 2006 by the Ministry of Health “due to non conformity to the nomenclature of accredited medical specialties in the EU”. This is contrary to the EU request for the curriculum of the residency in medical genetics in Romania, the curriculum that was positively appreciated by an Italian committee. The Romanian Society of Medical Genetics and its board is making great efforts to persuade the ministry that medical genetics should not be abolished in Romania due to the inclusion of the country in the EU as this specialty is successfully functioning in 23 out of 25 EU countries. In this respect, a common official complaint was already submitted to the authorities in charge.



- ◆ in turn, post degree courses are organized each year by each university.
- ◆ Symposiums are organized at least yearly by each university.
- ◆ The National Congress is held every three years, the last one, with international participation - more than 10 foreign invited speakers from European countries and more than 300 participants, was held in Cluj-Napoca, in September 2006.
- ◆ Summer schools were organized in 2001, 2003, 2005 by Professor Marius Bembea from the University of Oradea.



### **Bioethics and legislation**

Romania observes the Convention of Human Rights and Biomedicine signed in Oviedo on the 4th of April 1997 and the additional Protocol of the European Convention for the protection of human rights signed in Paris, on the 12th of January 1998, the Romanian law on the above mentioned issues is number 17/22nd February 2001.

### **Organizations of people with genetic diseases**

There is a strong collaboration of medical professionals with interest and specialization in Medical Genetics with the Associations of people with genetic diseases in Romania, we mention the most active:

- ◆ HEMOPHILIA ASSOCIATION – ROMANIA, PLOIESTI
- ◆ DOWN ASSOCIATION ORADEA ROMANIA, ADOR
- ◆ HUMANITARIAN FOUNDATION FOR SOCIAL ORIENTATION AND INTEGRATION HAND-ROM
- ◆ MYASTHENIA GRAVIS NATIONAL ASSOCIATION
- ◆ ROMANIAN MUCOVISCIDOSIS ASSOCIATION
- ◆ SAVE THE CHILDREN ORGANIZATION - TIMIS
- ◆ PERSONAL AID SYNDICATE FROM AIUD – children with Down and Turner syndrome
- ◆ The Association of parents of deaf and blind children in Romania – UNICUL SENS
- ◆ WILLIAMS SYNDROME ASSOCIATION
- ◆ PRADER WILLI ASSOCIATION IN ROMANIA
- ◆ STAR OF HOPE - IASI (Down syndrome).

We hope the above presentation will be a good incentive for future collaboration with colleagues from abroad, with readers of the European Journal of Human Genetics.

Professor Ioan Victor Pop, MD, PhD

President Executive of the Romanian Society of Medical Genetics

## **Minutes of the ESHG General Assembly**

Sunday, 7th May 2006, 18:45-19:45 hrs

Amsterdam, RAI Congress Center, Auditorium

Present: more than 100 members

The President of the Society, Professor Andres Metspalu, opened the meeting.

1. The society's activities during the past year were presented as follows:

ESHG Board had 3 meetings in May 2005 (in Prague), in October 2005 (Salt Lake City) and in 2006 (Amsterdam)

There were two excellent ESHG conferences: in Prague and ongoing in Amsterdam.

According to the initiative of Professor Pier Franco Pignatti, ESHG organized a meeting of the presidents of National Societies of Human Genetics in September 2005 (Brussels Airport); contact with National Societies of Human Genetic will be regular in the future.

The long project to update the statutes to reflect the present organisation better and the work of the society is finished, including electronic voting (134 "accept", 9 "refrain", 0 "not accept"); only the final acceptance by the ESHG General Assembly was missing at that point (see item 9).

There were 2 issues of the Newsletter (Editor: Lina Florentin-Arar, lflorentin@leto.gr ); 2 (or 3) issues are planned for next year.

A plan started to invest more resources to update and develop the website (website assistant Paula Koivumäki, [paula.koivumaki@utu.fi](mailto:paula.koivumaki@utu.fi)).

ESHG committees have been working on their projects. Reports of Scientific Program Committee (SPC, Chair: Han



Brunner), Publications Committee (Chair: Jean-Jacques Cassiman), and Public and Professional Policy Committee (PPPC; Chair: Segolene Aymé) are in the Newsletter May 2006/No. 14. Patenting and Licensing Committee (PLC, Chair Gert Matthijs) is organizing a workshop with PPPC. Education Committee (Chair: Domenico Coviello) is collaborating closely with Eurogentest.

EJHG had another good year (report by the Editor: GertJan Van Ommen is in the Newsletter May 2006/No. 14). Practical matters of the society were taken care of excellently by Karin Knob at Vienna Medical Academy.

2. Financial Report 2005 of the Society was presented by Professor Andrew Read. The economy of the society is sound and ESHG Board has decided to give more support to young researchers in the form of fellowships to different courses/conferences.

3. Board Members leaving the Board are Karen Brondum-Nielsen, Thierry Frebourg, Eduardo Tizzano and Gert-Jan van Ommen. Leena Peltonen's term in the Executive Board is also over, but she continues two more years as a Board member. The new President of the Society is Professor John Burn; he started to chair the meeting at this point.

4. Board had suggested Professor Pier-Franco Pignatti as the new President-Elect; this was accepted by the membership.

5. Board had made two suggestions for new Board Members: Professor Jan Lubinski (Poland) and Dr. Assistant Professor, Klaus Kjaer (Denmark). The ESHG general assembly suggested two more new Board members: Professor Karen B. Avraham (Israel) and Professor Peter Heutink (The Netherlands). All four were accepted.

6. Membership fees for 2007 were decided to remain the same.

7. The membership was invited to make suggestions for sites for future ESHG conferences. The 2007 conference will be in Nice, France, June 16-19, 2007.

8. Professor Andrew Read presented a budget proposal for 2007 which was accepted.

9. The revised statutes were accepted by the meeting.

10. President John Burn closed the meeting.



*Professor Helena Kääriäinen,  
Secretary General*

## **Professor John Burn MD FRCP FRCPCH FRCOG FMedSci**

John Burn is Professor of Clinical Genetics and head of Newcastle University's Institute of Human Genetics. He trained in medicine in Newcastle. In 1972 he took a year out to complete an honours degree in genetics. He spent his student elective under Victor McKusick at Johns Hopkins and decided then to become a consultant in clinical genetics. After training in medicine and paediatrics he was appointed as a clinical scientific officer in the Medical Research Council Genetics Unit at Great Ormond Street



*Professor John Burn,  
new President*

Hospital London. In 1984 he returned north to become the first consultant in clinical genetics. He became head of the Northern Genetics Service in 1989, responsible for all clinical and laboratory diagnostic testing for

three million people. When he stepped down from the role of clinical director, there were over 120 staff in the service.

The University Institute he leads now has 27 research group leaders including 15 professors. With his colleague Professor Tom Strachan, Scientific Director, he has built a research team which has the highest research rating of 5\*\*.

Professor Burn has a wide range of research interests but is particularly interested in the genetics of colorectal cancers. He is chief investigator on CAPP2, a worldwide trial of aspirin and resistant starch as preventive agents in carriers of hereditary cancer. He is Executive Director of the Life Knowledge Park, one of six 'genetic knowledge parks' funded by the UK government to promote the development of genetics and healthcare. Professor Burn was founding chair of the International Society for Gastrointestinal Hereditary Tumours (InSiGHT) in 2003-5. He has been a government advisor on the White Paper on Genetics and from 2000 to 2005 he was a member of the UK Human Genetics Commission.



## Pecking order

John Burn

### Pecking order

1. A scale of ascendancy noticeably operating in a flock of poultry
2. Any social hierarchy in animals or humans, or system of ranks.

Being elected president of the European Society of Human Genetics was a very proud moment but also an anxious one. It wasn't the thought of public speaking, or chairing difficult board meetings. The big problem I foresaw was all those European Greetings. I must confess that despite many years of working and travelling in Europe I remain confused and bewildered by the methods of simply saying hello.

### peck<sup>1</sup>

1. Said of a bird: to strike, nip or pick at it with the beak.
2. To kiss someone or something in a quick or perfunctory way.

When I grew up in Northern England there were no problems with greetings. These involved a brief meeting of eyes coupled with a smile and a nod, sometimes accompanied by a muttered "alright?" or "how y' doin". When rare formal meetings occurred we would shake hands. Now I regularly kiss my female relatives on the cheek while my son and my son-in law receive a bear hug. Handshakes are everywhere. Slowly the insular British are learning more tactile forms of acknowledgement. But I still keep getting it wrong. Last month I greeted the Israeli wife of a friend and kissed her on her right cheek. I then turned to show them in as she offered her left cheek and almost lost her balance.

Even when I remember to kiss both cheeks it goes wrong; I will forget that Dutch friends expect three kisses and French inflation has now reached four! Is there an unstable repeat at work?

But there's an even bigger problem. I can never remember if I'm supposed to start with the right cheek or the left cheek which can result in some difficult moments of meeting in the middle. This fades into insignificance compared to the experience of my colleague Judith Goodship. Aware of the American tradition of firm handshakes she went to greet her American collaborator at their first meeting at the ASHG. Meanwhile the tall American decided that inclusion of the UK in the European Union must mean we all did the kissing cheek thing. Combine a tall American stepping forward to kiss a short European who is, at that moment, trying to shake hands and a unique and unusually friendly form of contact is the result.

I discussed this with our secretary Helena Kaariainen who agreed that maybe we should try a new approach and adopt the traditional Eskimo greeting of rubbing noses.



*Professor Helena Kääriäinen,  
Professor John Burn*

## New Board Members

**Dr. Heutink** has been trained as a molecular biologist at the University of Amsterdam and started his PhD project in 1988 at the Erasmus University in Rotterdam, The Netherlands, where he worked with Drs. Oostra and Sandkuijl on developing strategies for the mapping of genetic risk factors for complex diseases. After his PhD, Dr. Heutink started his own research group and worked on numerous gene mapping and identification studies at the department of Clinical Genetics of the Erasmus University. Over the years he located many disease genes, and by positional cloning strategies identified pathogenic mutations in genes for frontal temporal dementia, hereditary hemochromatosis,

benign hereditary chorea, autosomal dominant ataxia, hereditary Porencephaly and early onset Parkinsons disease..

In 2003 Dr. Heutink moved to the VU University Medical Center in Amsterdam where he became head of the newly founded section of Medical Genomics of the Department of Human Genetics that is part of the Center for Neurogenomics and Cognitive Research (CNCR). The main lines of research are to identify and characterize genetic risk factors for complex neurological diseases and phenotypes. This includes neurodegenerative disorders such as dementia and parkinsonism but also behavioral traits such as cognition, attention and depression. Dr. Heutink is currently chairman of the Dutch Anthropogenetic Society and coordinator of Epidemiology for a Dutch national center of excellence in genomics: The Center for Medical Systems Biology (CMSB).





**Professor Karen Avraham** is Associate Professor and Chair of the Department of Human Molecular Genetics and Biochemistry at the Sackler School of Medicine, Tel Aviv University. Karen was born in Canada and moved to the US at a young age. Dr. Avraham received her B.A. degree from Washington University in St. Louis, MO, USA and



then went on to do her Ph.D. at the Weizmann Institute of Science in Rehovot, Israel, when she immigrated to Israel. Dr. Avraham performed her postdoctoral training at the National Cancer Institute in Frederick, MD, USA. Since joining the faculty at the Sackler School of Medicine in 1996, Dr. Avraham has worked towards deciphering the molecular basis of disease and disorders, using genetic, developmental, biochemical, and cell biological tools. The research in her laboratory today focuses on understanding how hearing is maintained

through the delicate balance of protein expression in sensory hair cells. Much of her work focuses on mouse models whose contribution to our understanding of ear function has been dramatic.

Her research led to the identification of several genes, in particular in the Israeli Jewish population, responsible for human hearing loss. The knowledge acquired about how mutations lead to hair cell loss and information about the proteins these genes encode is being applied towards the development of therapeutic techniques for the treatment of deafness.

Professor Avraham is an elected member of EMBO, the European Molecular Biology Organization, and the international Collegium ORLAS, for scientists who have contributed to research in inner ear research. She was also awarded the Burt Evans Young Investigator Award from the US-based National Organization for Hearing Research (NOHR) and the Sir Bernard Katz Prize. She is currently co-coordinator of EuroHear, a European Consortium for Advances in Hearing Science: from Functional Genomics to Therapies.

**Klaus Wilbrandt Kjaer**, 37, (M.D., Ph.D, Assistant Professor at the Institute of Medical Biochemistry and Genetics, University of Copenhagen) is primarily focusing on human malformations and the molecular study of processes leading to formation of normal or abnormal body parts. This also covers studies of species homology and



the evolutionary processes that let such homology emerge. His current position implies 25% teaching of undergraduate students, 25% clinical work at the Clinic for Medical Genetics which is located within the University of Copenhagen, and 50% research. He was a board member of the Danish Society for Reproduction and Fetal Development for five years (including four years as chairman). Beside his work he plays the violin and the viola, has three beloved daughters aged 4, 7, and 9, and is married to Sigrid Kjaer who is a Lutheran Pastor. He founded the Danish semi-professional string orchestra Copenhagen Young Strings, and the Danish organization Youth for Chamber Music.

**Prof. J. Lubinski** is the national consultant for clinical genetics in Poland since 1998. He is head of International Hereditary Cancer Center, established in Poland in 1992. The most important divisions of this center are:



- a network of cancer genetic outpatient clinics covering almost the entire country
- a cancer bio-bank with

biological samples and clinical data from 135,000 cancer cases and appropriate controls including registries of thousands of mutation carriers such as a registry of 3,500 of BRCA1 carriers.

J. Lubinski is the author of around 200 papers on clinical and molecular genetics of cancers and author and coordinator of a few EU projects.

J. Lubinski is the editor of a journal published under the auspices of UICC – Hereditary Cancer in Clinical Practice. In 2005 he received the title of Doctor Honoris Causa of Riga Stradins University, Riga, Latvia.

## **Clinical/Medical Genetics as an EC recognised speciality. Report from the ad hoc committee**

In Europe, our medical speciality has many different names: human genetics, medical genetics or clinical genetics - being the most common. I will use the term "clinical genetics" to describe the speciality in this report.

In the European Union there is a free trade of goods and free movement of workers between the 25 countries.

This is one of the fundamentals of the community, but for some types of professionals there are restrictions. Whereas a medical license to practice as a medical doctor is valid within the whole community, there are restrictions in the mutual recognition of medical specialists. In a 1994 directive, updated 2005, the EC listed a number of medical specialities that are mutually



recognised. However, clinical genetics is not amongst them. Nevertheless, in 2006 it was recognised in all 25 EC countries but Belgium, Greece and Spain.

The issue of clinical genetics to be mutually recognised in the EU countries was approached by the ESHG board some years ago, see statement from 2001 on the web site, and, in 2004. I, together with Dian Donnai (Manchester) and Didier Lacombe (Bordeaux), were commissioned to form an ad hoc committee for specialisation in clinical genetics and to establish a contact with UEMS to find ways to have our speciality recognised within the EU.

UEMS (Union Européenne de Médecine Spécialistes) is an independent organisation of medical specialities recognised in the EU. The UEMS consists of the national societies and their respective specialist organisations. These specialist societies have issued recommendations for education and training of medical specialists within their speciality, which are available on the website ([www.UEMS.net](http://www.UEMS.net)). Many of these societies also offer European specialist exams. As we are not recognised we cannot, by definition, be members of UEMS. However, within the UEMS there is a possibility to form a JMC, joint multidisciplinary committee, for specific purposes.

We met with representatives of the UEMS and they approved to form a JMC for clinical genetics. Members of this JMC will be representatives from us and from those specialist organisations that are interested. The JMC for clinical genetics will meet during autumn to discuss a common minimum curriculum education and training of medical specialists in clinical genetics. With the help of the UEMS we will then address the Commission to have clinical genetics mutually recognised on a European level. However, this must be expected to be a slow, in part political, process.

In order to be able to have this meeting we have drafted a common core curriculum that should form the backbone for the discussion. This draft has been approved by the board.

Not only the name of the speciality differs, also the tasks carried out by the specialist in clinical genetics vary between the different health care systems in the member states. In some countries clinical geneticists are both, laboratory supervisors and the clinicians seeing patients, whereas in other countries these are separated. Therefore, finding a common core curriculum might be complicated. It is not possible in these guidelines for education and training to give detailed recommendations on e.g. the duration of specific parts of the training. However, this is not unique for our speciality; several of the recognised specialities have similar differences in training and tasks for specialists in the different member states.

Initiated by the ESHG board several implications follow. If we have a common European speciality and issue guidelines for training and education and organise

other activities related to clinical professional activities, there is a need for a governing board to oversee the work. These curators must come from the profession itself and ESHG is a scientific organisation composed of scientists spanning the whole field of human genetics and with members of various educational backgrounds. Therefore it cannot be a task of the ESHG board to be responsible for the governors. Thus, there will be a need for a European specialists' organisation – a European college of clinical genetics.

The UEMS is only interested in the professional issues regarding medical doctors. As it is well known to all of us, the clinical staff in a clinical genetics unit does not only consist of medical doctors. Biologists with a PhD (hospital geneticists, clinical scientists, etc) as well as, in many countries, genetic counsellors and genetic nurses, and other specialists are important collaborators in the clinical department. Education and training of these groups of specialists is as important as the training of the medical specialist. Therefore there is also a need to find consensus on a minimum criteria for core competences of these specialists. A European college of clinical genetics must take this into account to avoid conflicts.

At present, there are some other organisations seeking to find guidelines for training and education in clinical genetics. Within the OECD, the Working Party for Biotechnology had an expert group drafting guidelines for quality assessment in molecular genetics testing laboratories, including a suggested core curriculum for laboratory staff with different training background. A document is at present open for public consultation, and will hopefully end in recommendations from the OECD. EUROAGENTEST ([www.eurogentest.org](http://www.eurogentest.org)), a network of excellence sponsored by DG Research, has a unit for education and training, co-chaired by Domenico Coviello (Milano), also chair of the education committee of ESHG and Alastair Kent (London) leader of Genetic Interest Group, GIG, a UK patient's organisation. This unit has as one of its goals to create guidelines for education and training of clinical staff members in a genetic counselling unit, irrespective of their profession.

In summary, we are slowly moving towards EU recognition of our speciality, but on the way there is much we can, and have, to do, to be prepared when that day comes. The most important may be to have an organisation to be the curators and governors in order to form the speciality.



*Ulf Kristoffersson,  
associate professor,  
senior consultant  
(Lund, Sweden)*

Ulf Kristoffersson (Lund)  
[Ulf.Kristoffersson@med.lu.se](mailto:Ulf.Kristoffersson@med.lu.se)



## Quality Assurance in Molecular Genetic Testing International Guidelines from the Organisation for Economic Co-operation and Development

The Paris based OECD is a club of governments of 30 developed countries recognised for high level analysis and comparative studies of finance and economies. Less well known is that the OECD supports policy development in many areas including science and has had an active interest in Clinical Molecular Genetic Testing dating to an international workshop in Vienna in 2000. Following the meeting the OECD Biotechnology Division brought together an expert group to consider issues in the quality of Molecular Genetic Testing. Their main effort; to develop an evidence base through a survey of over 800 Molecular Genetic Testing Laboratory Directors from 18 countries was completed in 2003 and published in 2005 (<http://www.oecd.org/dataoecd/25/12/34779945.pdf>). The results confirmed that Diagnostic Molecular Genetic Testing for rare single gene disorders is a truly international and networked activity with over 60% of Molecular Genetic Testing laboratories reporting that they either received or sent cases across national borders. This significant and unusual mechanism for providing healthcare confirmed that questions around harmonisation of practice and quality assurance should be addressed through the development of guidelines for Quality Assurance in Molecular Genetic Testing.

A further two-year effort by a guideline group drew on experts and government representatives from most OECD member and affiliated countries and drew heavily on European initiatives including EuroGentest, the European Molecular Genetics Quality Network and Certified Reference Materials projects and the ESHG's Public and Professional Policy Committee.

Draft guidelines which were posted on the web for public consultation this summer

(<http://www.oecd.org/dataoecd/43/26/37103271.pdf>).

The expert group are currently working with the OECD Secretariat to collate over 90 comments received from patient groups, governments, professional organisations and individuals. An amended text will form the basis of

discussion amongst OECD governments and if all goes well the guidelines will be adopted and published by OECD in early to mid 2007.

The guidelines are divided into five chapters formatted into statements of principle (addressed to governments and regulators) and of best practice (most relevant to professional bodies and practitioners).

They describe the General Features of Molecular Genetic Testing, Quality Assurance systems (emphasising the role of laboratory accreditation and recognising the unique and valuable role of research laboratories in testing for rare genetic conditions) and monitoring laboratory performance through Proficiency Testing. In addition the guidelines address in detail test reporting and call for adoption of standard terminology and nomenclature and evidence-based interpretation of results. Finally the guidelines address Education and Training Standards of Molecular Genetic Testing laboratory personnel.

The expert group hope that the guidelines represent a consensus of current best practice. As such they may be adopted across all OECD jurisdictions, help form policy and come to carry authority amongst professionals and regulators as Molecular Genetic Testing becomes a more widespread activity both in the developed and developing world and begins to enter mainstream medical practice.

Rob Elles, National Genetics  
Reference Laboratory,  
Manchester UK,  
Chair – expert group.



*Dr. Rob Elles*



## ESHG-Education Committee

Domenico Coviello (Chair, Milan, Italy), [coviello@unige.it](mailto:coviello@unige.it)  
 Martina Cornel (Amsterdam, The Netherlands), [mc.cornel@vumc.nl](mailto:mc.cornel@vumc.nl)  
 Celia DeLozier (USA/Switzerland), [cddelozier@comcast.net](mailto:cddelozier@comcast.net)  
 Peter Farndon (Birmingham, UK), [p.a.farndon@bham.ac.uk](mailto:p.a.farndon@bham.ac.uk)  
 Peter Goetz (Czech Republic), [petr.goetz@fnmotol.cz](mailto:petr.goetz@fnmotol.cz)  
 Shirley Hodgson (London, U.K.), [shodgson@sgul.ac.uk](mailto:shodgson@sgul.ac.uk)  
 Alastair Kent, (Patients Organizations, GIG and EAGS), [alastair@gig.org.uk](mailto:alastair@gig.org.uk)  
 Gyorgy Kosztolanyi (Pécs, Hungary), [Gyorgy.Kosztolanyi@aok.pte.hu](mailto:Gyorgy.Kosztolanyi@aok.pte.hu)  
 Jorge Sequeiros (Porto, Portugal), [jsequeir@ibmc.up.pt](mailto:jsequeir@ibmc.up.pt)  
 Heather Skirton (Plymouth, UK), [heather.skirton@plymouth.ac.uk](mailto:heather.skirton@plymouth.ac.uk)

## Additional collaborating members:

Agnes Bloch-Zupan (Strasbourg, France), [Agnes.Bloch-Zupan@dentaire-ulp.u-strasbg.fr](mailto:Agnes.Bloch-Zupan@dentaire-ulp.u-strasbg.fr)  
 Rodney Harris (Manchester, U.K.), [rodney.harris@man.ac.uk](mailto:rodney.harris@man.ac.uk)  
 Vaidutis Kucinskas (Lithuania), [Vaidutis.Kucinskas@santa.lt](mailto:Vaidutis.Kucinskas@santa.lt)  
 Taytum Ozelcelik (Ankara, Turkey), [tozelcelik@fen.bilkent.edu.tr](mailto:tozelcelik@fen.bilkent.edu.tr)  
 Fred Petrij (Rotterdam, The Netherlands), [f.petrij@erasmusmc.nl](mailto:f.petrij@erasmusmc.nl)  
 Maria Soller (Lund, Sweden), [Maria.Soller@med.lu.se](mailto:Maria.Soller@med.lu.se)

## Dear ESHG members,

The Education Committee this year has increased its activity and here I am presenting the results achieved and/or the events that myself and/or other members have attended, in chronological order:

- 1) The draft document of the work performed last year "Patient and Professional Perspectives of Genetic Information/Education in Europe" in conjunction with Eurogentest Unit 6 is completed and is now on the web site for consultation ([www.eurogentest.org](http://www.eurogentest.org)). Anyone (patients and professionals) who is interested in genetic services and genetic tests is welcome to send comments for integration into this document.
- 2) On 8th of March 2006, Prof. Peter Farndon, director the NHS- National Genetics Education and Development Centre ([www.geneticseducation.nhs.uk](http://www.geneticseducation.nhs.uk)), organized a meeting in Birmingham (UK) titled "Supporting Genetics Education for Health". It was a very productive initiative; most of the research groups involved in genetic education in the UK were present with stands and education material on display. A list of activities can be found on the web site [[www.eurogentest.org](http://www.eurogentest.org)]. During this meeting I had the opportunity to establish two new contacts, one with the genetic counsellor network and another with the Association for Medical Education in Europe (AMEE, [www.amee.org](http://www.amee.org)).
- 3) On 8th of May 2006, the Education Committee met in Amsterdam during our ESHG conference. The work in progress was discussed and priorities were decided:
  - a) Update the list of courses available in Europe and set up a new web tool to enable users to search for courses on line.
  - b) Continue the work to complete a draft document on Core Competence in Genetics for Health Professionals.
  - c) Organize a workshop (Porto September 23-24) to discuss the document.
  - d) Liaise with other International Organization to share ideas, tools, initiatives:
    - ASHG Liaison (Celia DeLozier, CD) -- Celia has been in contact with Susanne Haga, Chairman of the ASHG Education Committee. The ASHG is pleased to have the opportunity of cooperation and it was suggested that CD attend the ASHG education committee meeting in October in New Orleans to discuss further steps. In the meantime, Susanne Haga proposed a specific activity that could be carried out together: DNA Day, which commemorates the sequencing of the human genome (see website [www.ashg.org](http://www.ashg.org), section on educational activities). The ASHG had a number of activities this year on April 25, targeting pupils from Kindergarten-12th grade, which included: essay contest, chat page, web-based presentations, mentoring. Celia DeLozier summarized the activities and agreed to make a proposal to the committee about what activities might be shared in 2007. Committee members noted that some European countries already have a DNA or a similar genetics day, including Holland and perhaps Spain. Turkey is currently hosting an exhibit from the Cold Spring Harbor Symposium, targeting high school students (the six sections include advocacy and genetics and lessons for life). The Education Committee is considering whether similar events could take place on the same date in Europe next year. CD will contact Education Committee members by e-mail to present the ASHG project and get feedback about similar public education initiatives in Europe.
    - Nurses in Genetics Liaison (ISONG). Heather Skirton is the current President of the International Society of Nurses in Genetics. ISONG is keen to involve more nurses from European countries in its activities and would be willing to work closely with the Education Committee. ISONG holds its annual Educational and



Scientific conference in Europe for the first time in Bristol (UK) from 18-21 May 2007 and wishes to liaise with ESHG to ensure that ESHG members have the opportunity to be involved.

- International Bioethics Committee (UNESCO): George Kosztolanyi is on the committee and works in Informed Consent and Public Health Initiatives section. The ESHG Education Committee would like him to be our formal liaison person with the IBC.
  - HGSA liaison – Domenico Coviello was in contact with Sylvia Metcalfe, head of Education Committee of Human Genetic Society of Australasia. Sylvia and Domenico met in Genoa in June to further discuss the possible interaction initiatives. Sylvia was also part of the organizing committee of the ICHG in Brisbane, where a meeting of the heads of the Education Committee participating to the International Federation Human Genetics Societies (IFHGS) took place.
- 4) On the 8th of May, 2006, in Amsterdam, again during our ESHG conference, Prof. Pier Franco Pignatti has organized the 2nd meeting of presidents of national societies of Human Genetics. Three members of the Education Committee presented the work in progress and the question if a European network with contact persons in each EC country interested in genetic education could be established.
  - 5) May 15-17, 2006, an International meeting took place in Manchester (UK) titled “Genetic Counseling Education: “Connecting the Global Community”. All directors of Genetic Counselling courses worldwide and representatives of several organizations connected with genetic education were invited to this meeting. Experiences were compared and a “Transnational Alliance of Genetic Counsellor” (TAGC) was proposed as an umbrella organization under the initial coordination of Prof. Janice G. Edwards, from University of South Carolina, USA. (<http://igce.med.sc.edu/conference%20overview.htm>).
  - 6) On the 10th of August, 2006, in Brisbane (AU), during the International Congress of Human Genetics (ICHG), Sylvia Metcalfe organized the first meeting of representative of chairmen of education committees of societies part of IFHGS (<http://www.ifhgs.org/>) and from other institutions involved in genetic education. The “International Genetics Education Network” (IGEN) was launched. The overall goal is to provide a sharing of expertise based on agreed education principles. These principles would then be applied appropriately in relation to different contexts of different countries, e.g. professional standards, culture, etc. A mentor network similar to the ASHG mentor network was proposed. Experts in each country would assist in developing programs. The focus would be on genetics education for healthcare professionals with a clear focus on conceptual and practical education, not clinical or laboratory issues.
  - 7) September 23-24, 2006, The Working Party for Biotechnology, had an expert group drafting guidelines for quality assessment in molecular genetics testing laboratories, including a suggested core curriculum for laboratory staff with different training backgrounds. The Eurogentest Unit 6 (Education) workshop took place in Porto, where the background paper on genetic core competences for health professionals was discussed. The document is under revision and the final draft will be on the website for consultation from January 2007 on. The document emphasizes the need for a common minimal standard of education and training for all health professionals in Europe, to equip them for appropriate practice. It is obvious that the level of knowledge, skills and attitudes related to genetic healthcare will vary according to the profession of the practitioner, the setting in which he or she works and the relevance of genetics to his or her area of practice. This document will be useful to each single state to verify and update the curricula on national levels.

All of you are invited to send us comments, ideas, updates from your country, and anything else you want to let us know (including willingness to collaborate with the education committee), to help us setting up priorities, to harmonize education and training, and to share experience or tools available.

On behalf of the Education Committee,

Domenico Coviello, MD, PhD



*Domenico Coviello  
Unit 6*



*Steering committee Unit 6  
all the participants:  
Celine Lewis, Heather Skirton,  
Domenico Coviello, Alastair Kent*



*working group in Porto*



## ***Towards a Network of Human Genetics Societies in Europe***

### ***The purpose***

In order to foster Human Genetics progress in Europe, it was proposed to stimulate reciprocal knowledge and to strengthen the relations between the European Society of Human Genetics (ESHG) and the different National Human Genetics Societies (NHGSs).

### ***The decision***

At the 2005 European Human Genetics Conference in Prague, the board of the ESHG has discussed the idea, has agreed that it was important to proceed to build stronger links among the Societies. This would increase collaboration by discussing themes of common interest, establish a forum for the confrontation of national politics in transnational matters arising, and finally, augment the visibility of Human Genetics in Europe. The board therefore gave me the task to convene an ESHG-NHGSs meeting and allocated a budget for inviting the presidents of the various societies to the meeting prior to the 2006 Conference.

### ***The first meeting***

The names and addresses of various representatives were collected with the important help of the EUROAGENTEST project which provided a list of the societies to be contacted, and helped with the local arrangements at the venue at Brussels airport, where the meeting was held on 12 September, 2005.

The agenda included a short presentation by the various national societies' representatives, and one by the president of the ESHG, a review of the possible subjects of common interest and proposals on how the societies could coordinate some activities and stimulate reciprocal knowledge. An update was given on the progress of the request for an EU medical/clinical genetics specialization, the EU project on genetic testing was presented, continuing medical education and human genetic education issues in the E U were discussed. Website links among the societies and a common English format for all websites were suggested.

At the end it was felt that the meeting was successful and it was proposed to have regular meetings of the representatives which should be nominated by the societies during the annual conferences.

The proceedings of the meeting are available on the ESHG website ([www.eshg.org](http://www.eshg.org), under the opening page headline "Genetics in Europe").

### ***The second meeting***

A second meeting of the ESHG and the NHGSs was therefore held during the 2006 European Human Genetics Conference in Amsterdam on the 8th of May, 2006.

An effort was made to reach other societies of which no contact was available, and this was achieved: 31 NHGSs were represented at the meeting, compared with 23 societies represented at the first meeting. Also, the ESHG was very well represented at the meeting, with almost the complete executive board, again showing the appreciation for the networking activities. Updates on the EU Specialty request, EUROAGENTEST activities, education in Genetics for geneticists and for non-geneticists were presented and discussed.

It was proposed that the persons representing the different nation's societies should be in charge for a long period of time (3-5 years) in order to facilitate reciprocal knowledge and relations. It was decided that the ESHG would put a link on its website to the websites of the NHGSs to keep information updated, and the minimal form in English for the various societies websites was approved and shown in Table I.

The proceedings of the second meeting can also be found on the ESHG website ([www.eshg.org](http://www.eshg.org), Genetics in Europe).

### ***Programme***

The network of the societies is now established and is being used by more and more geneticists to disseminate documents, and to request information and advice.

This is helped by the continuous updating of the list of contact persons for the NHGSs, which is shown, in its September 30, 2006 version, in Table II. We still need more information on missing societies' contacts and on their websites.

For suggestions, corrections, and updates, please contact me at the following address: [pierfranco.pignatti@univr.it](mailto:pierfranco.pignatti@univr.it), and I'll be glad to help.

And please make a note to participate in the third meeting which will be held during the 2007 European Human Genetics Conference in Nice, 16-19 June 2007 !

I will send an agenda and details of the meeting to all on the above list as soon as we'll be assigned a room and time slot by the conference organizing committee.

Pier Franco Pignatti,  
ESHG contact person for the NHGSs  
President-elect, ESHG  
30 September, 2006



*Professor Pier Franco Pignatti,  
President elect*



## **Contact persons, National Societies of Human Genetics (NSHG) in Europe**

### **Austrian Society of Human Genetics**

[www.oegh.at](http://www.oegh.at)

Hans-Christoph Duba, M.D. (president)

Tel. + 43 50 554 63 29600

Fax + 43 50 554 63 29604

[Hans-christoph.duba@gespag.at](mailto:Hans-christoph.duba@gespag.at)

### **Belgian Society of Human Genetics**

[www.beshg.be](http://www.beshg.be)

Miikka Vikkula (president)

Tel + 32 2 764 74 90

Fax + 32 2 764 74 60

[vikkula@bchm.ucl.ac.be](mailto:vikkula@bchm.ucl.ac.be)

### **British Society of Human Genetics**

Andrew Read (contact person)

Tel +44 161 276 6276

[andrew.read@man.ac.uk](mailto:andrew.read@man.ac.uk)

### **Bulgarian Society of Human Genetics**

Stoyan Lalchev (president)

Tel/Fax + 359 2 91-72-713

Mobile: + 359 888 83-12-15

[slalchev@yahoo.com](mailto:slalchev@yahoo.com)

### **Croatian Society of Human Genetics**

Ingeborg Barisic (board member)

Tel. + 385 1 4600107

[ingeborg.barisic@kdb.hr](mailto:ingeborg.barisic@kdb.hr)

### **Czech Society of Medical Genetics**

Petr Goetz (chairman)

Tel.: + 420 2 2443 3503

Fax + 420 2 2443 3520

[petr.goetz@lfmotol.cuni.cz](mailto:petr.goetz@lfmotol.cuni.cz)

### **Danish Society of Medical Genetics**

Kirsten Rasmussen, MD (president)

tel: + 45 6541 1605

fax: + 45 6541 1911

[kirsten.rasmussen@ouh.fyns-amt.dk](mailto:kirsten.rasmussen@ouh.fyns-amt.dk)

### **Estonian Society of Human Genetics**

[www.estshg.ee](http://www.estshg.ee)

Andres Metspalu (representative)

[esthg@ebe.ee](mailto:esthg@ebe.ee)

### **Finnish Society of Medical Genetics**

Minna Poyhonen (president)

tel: + 358 9 19125091, mobile: + 358 503828948

Fax + 358919125105

[minna.poyhonen@helsinki.fi](mailto:minna.poyhonen@helsinki.fi)

### **French Society of Human Genetics**

Segolène Aymé (president)

Tel 33 4 67 41 53 60

Fax 33 4 67 41 53 65

[ayme@orpha.net](mailto:ayme@orpha.net)



**German Society of Human Genetics**

[www.gfhev.de](http://www.gfhev.de)

André Reis (vice president)

Tel: 49 9131 8522318

Fax: 49 9131 209297

[reis@gfhev.de](mailto:reis@gfhev.de)

**Hellenic Association of Medical Geneticists**

Zoe Kosmaïdou-Aravidou, MD, PhD (president)

Tel./Fax +302107777716

[garavidis@yahoo.gr](mailto:garavidis@yahoo.gr)

**Hungarian Society of Human Genetics**

G. Kosztolanyi (representative for president Istvan Rasko)

Tel. 36-62-599-681

Fax 36-62-433-503

[rasko@nucleus.szbk.u-szeged.hu](mailto:rasko@nucleus.szbk.u-szeged.hu)

**Italian Society of Human Genetics**

[www.sigu.net](http://www.sigu.net)

Franca Dagna Bricarelli (president)

Tel. + 39 010 5634371 / mobile + 39 328 1503310

Fax + 39 010 5634381

[dagna@galliera.it](mailto:dagna@galliera.it)

**Latvian Society of Medical Genetics**

Natalia Pronina (representative)

Tel + 371 7514618 / 371 7536840

Fax + 371 7536515

[biochem@gc.bsg.lv](mailto:biochem@gc.bsg.lv)

[rita@gc.bsg.lv](mailto:rita@gc.bsg.lv)

**Lithuanian Society of Human Genetics**

Vaidutis Kucinskas (president)

Tel. + 370-5-2365195

Fax + 370-5-2365199

[vaidutis.kucinskas@santa.lt](mailto:vaidutis.kucinskas@santa.lt)

**Macedonian Academy of sciences and Arts, research center for genetic engineering and biotechnology**

Dr. Dijana Plaseska-Karanfilska (secretary)

Tel + 389 2 3235410

[dijana@manu.edu.mk](mailto:dijana@manu.edu.mk)

**Norwegian Society of Medical Genetics**

Geir Julius Broathen (board member)

Tel: + 47 6792 7820

Fax: + 47 6792 7803

[g.j.broathen@medisin.uio.no](mailto:g.j.broathen@medisin.uio.no)

**Polish Society of Human Genetics**

Tadeusz Mazurczak (president)

Tel. (+48 22) 32 77 361

Fax (+48 22) 32 77 151

[tadmaz@imid.med.pl](mailto:tadmaz@imid.med.pl)

(second representative indicated:

Pawel Stankiewicz MD, PhD

[pstankiewicz@imid.med.pl](mailto:pstankiewicz@imid.med.pl) or [sekgen@imid.med.pl](mailto:sekgen@imid.med.pl) )



**Portuguese Society of Human Genetics**

[www.spgh.net](http://www.spgh.net)

Isabel Carreira (president)

Tel + 351 239838866

[hgp@utad.pt](mailto:hgp@utad.pt)

Ana Maria Fortuna (representative)

Tel.: +351-226070300

[ana.fortuna@igm.min-saude.pt](mailto:ana.fortuna@igm.min-saude.pt)

**Romanian Society of Human Genetics**

Cristina Skrypnyk (representative)

Tel + 40 722413662

[cristinaskrypnyk@yahoo.com](mailto:cristinaskrypnyk@yahoo.com)

**Serbian Society of Genetics**

Dragica Radojkovic (contact person)

[dada@eunet.yu](mailto:dada@eunet.yu)

**Slovenia:****- Slovenian Association of Human Genetics**

Borut Peterlin MD, PhD (president)

T/F: + 386 1 5401 137

[borut.peterlin@guest.arnes.si](mailto:borut.peterlin@guest.arnes.si)

**- Slovenian Society of Human Genetics**

Damjan Glavac, PhD

Tel + 386 1 543 7195

Fax + 386 1 543 7181

[damjan.glavac@mf.uni-lj.si](mailto:damjan.glavac@mf.uni-lj.si)

**Spanish Association of Human Genetics**

[www.aegh.es](http://www.aegh.es)

Feliciano J. Ramos, MD PhD (president)

Tel: + 34 976 761739

FAX + 34 976 761726

[framos@unizar.es](mailto:framos@unizar.es)

**Swedish Society of Medical Genetics**

[www.suls2.se/sektioner/mg](http://www.suls2.se/sektioner/mg)

Thoas Fioretos (president)

tel + 46-46-17-33-62

fax + 46-46-13 10 61

[thoas.fioretos@med.lu.se](mailto:thoas.fioretos@med.lu.se)

**Swiss Society of Medical Genetics**

Albert Schinzel (representative)

Phone: + 41 1 6557051

Fax.: + 41 1 6557220

[info@medgen.ch](mailto:info@medgen.ch)

[schinzel@medgen.unizh.ch](mailto:schinzel@medgen.unizh.ch)

**The Netherlands:****- The Netherlands Society of Clinical Genetics**

[www.vkgr.nl](http://www.vkgr.nl)

M.H. Breuning (president)

Tel. + 31 71 526 6090

Fax + 31 71 526 6749

[m.h.breuning@lumc.nl](mailto:m.h.breuning@lumc.nl)



**- Dutch Anthropogenetics Society**

www.nav-vkgn.nl  
 Peter Heutink (president)  
 Tel + 31 205godggr  
[p.heutink@umc.nl](mailto:p.heutink@umc.nl)

**- Dutch Clinical Molecular Genetics Society**

Hans Scheffer, PhD (president)  
 Tel + 31 3613799  
[h.scheffer@antrg.umca.nl](mailto:h.scheffer@antrg.umca.nl)

**Turkish Society of Medical Genetics**

www.tamgen.org  
 Tayfun Ozcelik, MD (president)  
 Tel: 90-312-290-2139  
 Fax: 90-312-266-5097  
[tozcelik@fen.bilkent.edu.tr](mailto:tozcelik@fen.bilkent.edu.tr)

(update Sept. 6, 2006)

**On behalf of The European Society of Human Genetics**

## **PROPOSAL FOR a "MINIMAL FORM" (written in English) that could describe the activities of National Societies of Genetics in Europe**

**1. Home (Name and identity of the society)**

including the followings :  
 -complete postal and electronic addresses for contact  
 -Web site  
 -composition of the Executive Board or Council or Commission  
 -updated number of Members

**2. Summary of Statutes**  
(including private and/or public)**3. Aims****4. Background****5. Activities****6. Symposiums****7. Publications, Reports, Recommendations, Guidelines and/or Policies****8. Newsletters****9. Fellowships and Job Opportunities****10. Legislation of activities in the country****11. Careers in Human Genetics in the country****12. Related links and useful websites**





# Structure Organisation

# 2006

## THE BOARD OF THE SOCIETY (May 2006-May 2007)

Term as board member ends at the general assembly in the year shown in brackets

### EXECUTIVE BOARD

President	Prof. John Burn (2010)
President-Elect	Prof. Pier Franco Pignatti (2008)
Vice-President	Prof. Andres Metspalu (2009)
Secretary-general	Prof. Helena Kääriäinen (2007)
Deputy secretary-general	Prof. Thomas Meitinger (2007)
Treasurer	Prof. Andrew Read (2007)

### BOARD MEMBERS

Ass.Prof. Nurten Akarsu	Ankara (2009)	Prof. Nicolas Levy	Marseille (2008)
Prof. Karen B. Avraham	Tel Aviv (2011)	Prof. Jan Lubinski	Szczecin (2011)
Prof. Jacques Beckmann	Lausanne (2009)	Prof. Milan Macek Jr.	Prague (2007)
Prof. Alexis Brice	Paris (2008)	Prof. Gert Matthijs	Leuven (2008)
Prof. Francoise Clerget-Darpoux	Villejuif (2010)	Prof. Leena Peltonen	Helsinki (2008)
Prof. Dian Donnai	Manchester (2009)	Ass.Prof. Alessandra Renieri	Siena (2009)
Ass.Prof. Thoas Fioretos	Lund (2007)	Dr. Silke Sperling	Berlin (2010)
Prof. Peter Heutink	Amsterdam (2011)	Prof. Veronica van Heyningen	Edinburgh (2007)
Ass.Prof. Klaus W. Kjaer	Copenhagen (2011)	Dr. Christos Yapijakis	Athens (2007)
Prof. Vaidutis Kucinskas	Vilnius (2010)		

### LIAISON MEMBERS

Dr. Ségolène Aymé	Chair, PPPC
Prof. Han Brunner	Chair, SPC
Prof. Jean-Jacques Cassiman	IFHGS
Dr. Domenico Coviello	Chair, Education Com.

## Election of President-Elect & Board Members 2007

The Society Board intends that the President-Elect and three Board members will again be elected by postal ballot of all the members, rather than by voting in person at the Annual General Meeting, to increase participation of all members. Members of the society who wish to stand for election are requested to read the job descriptions of the duties of the President and Board Members detailed on the back of the enclosed nomination form. The Statutes of the Society state that the Board should be broadly representative of nationalities and specialties. A list of current Board members is on the Society's web site [www.eshg.org](http://www.eshg.org) (Term of office ends at the general assembly in the year shown in brackets)

The ballot form showing the names of members wishing to stand for election will be sent to eligible voting members in the next mailing and a date given by which it should be returned. The result will be announced at the Annual General Meeting.



## Membership Renewal - Information for 2007

It is now time to ask you to renew your subscription for 2007. You will shortly be receiving a form to renew your membership, showing the different categories available. Please note, that if you take out a full (regular) membership you may have an electronic subscription to the Journal as well as a paper copy. The Journal will not be delivered to you from December 2006 until we receive your renewal form and payment, so please do not delay when you receive your renewal request.