

European Society of Human Genetics

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Vice-President: Professor Gerd Utermann

President-Elect: Professor Gert-Jan van Ommen

Secretary-General: Professor Peter Farndon

Deputy Secretary-General: Dr Célia DeLozier-Blanchet

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EUROPEAN PARLIAMENT REPORT ON THE ETHICAL, LEGAL, ECONOMIC AND SOCIAL IMPLICATIONS OF HUMAN GENETICS

*Temporary Committee on Human Genetics and Other New Technologies in
Modern Medicine Rapporteur: Francesco Fiori*

In December 2000, the European Parliament decided to establish a Temporary Committee to examine recent developments in the field of human genetics. A dedicated page has been set up on their website www.europarl.org.uk to facilitate access to the work of the Committee, and to the views expressed on the issue by European and UK Institutions. The page will be updated from time to time to reflect major developments in this area. Human Genetics dedicated page <http://www.europarl.org.uk/infocus/focusgenmain.htm>

The president and past president, Stylianos Antonarakis and Jean-Louis Mandel, attended the hearing of the committee in July. Professor Antonarakis presented the views of the Society and the problem of diagnosis of rare genetic disorders at the European level. Professor Mandel presented suggestions for an EU sponsored network of diagnostic laboratories for rare genetic diseases. These papers and the policy statements from our PPPC were placed on the Temporary Committee website http://www.europarl.eu.int/comparl/tempcom/genetics/contributions/contri_eshg_en.pdf

The European Parliament was due to vote on the report on 29th November 2001. The Society had concerns, that if adopted unchanged, the report would harm the future interests of patients and families with genetic disorders.

The Board of the Society has produced three documents which are currently available on the ESHG website (downloadable as pdf files). They are:

1. A letter to members of the European Parliament
2. Changes to the Fiori Report recommended by the ESHG
3. Overview comments about the report and its history so far.

Professor Segolene Ayme, chair of the PPPC, wrote to members of the European Parliament on behalf of the Society. Many colleagues in National Societies also wrote directly. At the time of printing the outcome of the vote in the European Parliament is not known.

VIENNA 2001

The International congress of Human Genetics was organized in Vienna, from May 15 to May 19, 2001 by the European Society of Human Genetics, on behalf of the International Federation of Human Genetic Societies.

An important aim of the congress was to provide young geneticists with the opportunity to attend this unique event in human genetics i.e. a world meeting on human genetics at a time when the human genome sequence is practically completely known and when research and also diagnoses based on this knowledge receive an unprecedented stimulus. For this purpose, the ESHG decided to make a special effort to keep registration and housing costs as low as possible while also providing fellowships for non-EU young scientists.

Under the Patronage of the Federal President of the Republic of Austria, the congress was a true international and worldwide event. UNESCO, EU commissioner Busquin, and Europabio lent their auspices to the congress. Moreover Unesco provided a series of plenary speakers and discussants to the congress. The OECD, at its own request, was involved in the organization of sessions on genetic testing. Dr Manuel Hallen, Head of the 'Genetic Research and Infrastructure' Unit, addressed the participants as representative of EU commissioner Busquin.

3295 participants, representing 86 countries, attended the meeting. Of these, 687 were young scientists (less than 35y old) from all over the world; 181 of them, from 29 different non-EU countries, benefited from EU fellowships to attend the congress.

In addition to the 20 plenary lectures by invited speakers, the scientific program offered 60 invited lectures in 20 concurrent symposia, 108 oral presentations of submitted abstracts in 20 concurrent sessions and 1677 posters divided over 17 topics.

The press room generated a series of summaries for the newspapers and many invited speakers were interviewed by the press and the Austrian TV.

The congress website was hit by 52.000 individual visitors (no "refresh" counted) since November 15, 2000.

A very successful and well attended exhibition provided information from 83 different companies.

The expenses of the meeting amounted to EUR 977.289, yet to be finalized, and even with the possibility of registering at uniquely low rates, there is a good chance that a small amount of money will be available for the ESHG for future scientific activities.

The response of the participants was very positive at the time of the meeting. The 181 fellows wrote reports full of positive comments. Nevertheless let us wait for the results of the written evaluations, which will be available soon, to see whether our biased positive impressions are confirmed.

Many people deserve credit for this success, the students, the professionals, the selected and invited speakers, the attendants, the sponsors and many others. Allow me to mention specially the joint chiefs of staff immortalized in a picture on my wall: Jerome del Picchia, Charles Buys, Gert Utermann, Jantje De Roos, John Hodgson, Peter Farndon and your servant JJ Cassiman.

Jean-Jacques Cassiman

STRASBOURG 2002



The next ESHG meeting taking place in Strasbourg May 25-28 looks very exciting. For the first time, it will be organised in conjunction with the European Meeting on Psychosocial Aspects of Genetics (EMPAG). Each meeting will keep its own identity, but there will be common sessions on topics of shared interest including (neonatal screening and genetic testing in minors, behavioural aspects of genetic syndromes) and registered participants at the ESHG meeting will be able to participate in sessions of EMPAG and vice-versa. The scientific

program of the ESHG meeting will cater for varied tastes, from the latest in human genome analysis, to sessions on cancer, molecular karyotyping, various types of genetic pathologies, complex diseases and approaches to therapy. Almost all invited speakers have accepted, which is an excellent indication of the interest in our meeting.

The social program will be very attractive. The welcome reception will take place in the ample space of the newly built Museum of Modern and Contemporary Art, that offers magnificent views on the old city and Strasbourg cathedral, and hosts very interesting collections spanning a century of painting and sculptures. The conference party will be held in the The Palais Universitaire, a monumental neo-renaissance building, beautifully located in the late 19th century "Imperial quarter" of the city. Its elegant Aula will host a buffet dinner, allowing sampling of delicious Alsatian food and wines (while EMPAG participants will have their own cruise dinner). Finally, an organ and choir concert will feature an award winning organist playing on a famous 18th century instrument, in an exquisite church setting. Many hotels located in the city centre, within 10-15 minutes of the Congress Centre, have very moderate prices, in the 50-70 euros/night range, which will allow attendance of even the most budget-conscious geneticists.

Dont forget the abstract deadline, January 14, 2002, at www.eshg.org/eshg2002 (for both ESHG and EMPAG participants). We look forward to an exciting meeting.

Exhibition Bookings for Strasbourg

22 companies have now signed up for the exhibition at Strasbourg. They are:

- Agowa
- Applied Imaging
- Blackwell Science
- Bruker Daltonik
- Cytocell
- Genevac
- Ingenuity International
- S. Karger
- Lark Technologies
- LI-COR Biosciences
- MetaSystems
- Nature Publishing Group
- Orchid Bio Sciences
- P.A.L.M. Microlaser Technologies
- Pyrosequencing
- Qbiogene
- Schleicher & Schuell
- Science
- Sequenom
- Vysis
- Whatman BioScience
- Carl Zeiss



Of these two of the companies signed up at ICHG 2001 - Ingenuity International and Qbiogene



MEMBERSHIP RENEWAL INFORMATION FOR 2002

It is now time to ask you to renew your subscription for 2002. You will shortly be receiving a form to renew your membership, showing the different classes 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 2001 until we receive your renewal form so please do not delay when you receive your renewal request.

GENETICS IN EUROPE:

MEDICAL GENETICS IN NORWAY

Norway has a population of 4.5 million. The population was considered to be rather homogeneous, but an increasing heterogeneity is now seen, due to a large number of immigrants, mainly of ethnic Pakistanis. Consanguinity is rare except in some immigrant groups. A mandatory national insurance scheme covers health care for the population. In-patient treatment in public hospitals is free of charge, whereas out-patient consultations are subject to a minor fee paid by the patient.

Norway is not a member of the European Union.

Education

Medical genetics evolved from the Institute of Medical Genetics at the University of Oslo, and was established as a medical specialty as early as 1971. This was very different from the situation in Sweden and Denmark, where medical genetics was only recently recognized as a specialty. In the first years medical doctors had their training in medical genetics as part of research programmes. A system for postgraduate residency was later established at the Departments of Medical Genetics at the University Hospitals in Oslo, Bergen and Tromsø. Training includes a minimum of 4 years at an approved department of medical genetics with 2 years of genetic counselling covering a variety of genetic disorders, one year of cytogenetics and one year of molecular genetics. Approval of satisfactory training is given by the National Board of Medical Genetics, which is a committee of the Norwegian Medical Association. A qualifying examination is not required.

The number of approved specialists is about 30. However, only about 15 of these are currently working in Departments of medical genetics.

Genetic counselling has so far mainly been given by medical doctors. Due to the increasing demand for counselling, especially in cancer genetics, a two years Masters degree programme in Genetic Counselling was initiated at the University of Bergen for 5 students in 2001.

Genetic services

Regional centres provide comprehensive services in clinical genetics, cytogenetics, and molecular genetics at the University hospitals in Oslo, Bergen and Tromsø. The University hospital in Trondheim has established a Department of Medical Genetics, but no medical specialist is so far employed by that service. The Norwegian Radium Hospital in Oslo has a department of Medical Genetics which delivers cytogenetic and clinical services in cancer genetics. A cytogenetics laboratory is associated with a Research laboratory for Occupational and Environmental Genetics at Telemark County Hospital in Skien.

Medical genetics services are covered by the national insurance scheme.



Prenatal diagnosis

Prenatal genetic services are regulated by the Biotechnology Act (see below). Genetic counselling is mandatory before invasive prenatal diagnosis. Prenatal diagnosis for advanced maternal age is, by a political decision, offered for women aged 38 or more at term. Chorionic villus sampling is rarely used, and mainly for high-risk pregnancies. The proportion of women requesting prenatal diagnosis is lower in Norway than in most European countries and is currently performed in about 2.5% of pregnancies. This percentage has been the same for many years.

There are no serum screening programmes. Ultrasound examination at one regional hospital is offered at weeks 17-19 to all pregnant females, and an increasing number of invasive prenatal diagnostic tests are performed after ultrasound examination.

Legislation

An Act passed in August 1994 relating to the application of biotechnology regulates the delivery of medical genetic services. (For English version see: www.helsetilsynet.no/bioweb/05_publicasjoner/bio_act)

It is divided into sections pertaining to assisted reproduction, research on fertilized eggs, preimplantation diagnosis, prenatal diagnosis, genetic investigations after birth, and gene therapy. Donation of eggs, and extraction of gonadal cells from the testis for assisted reproduction are not permitted. Furthermore, genetic information can not be asked for in connection with insurance.

Genetic counselling is required for presymptomatic, predictive and carrier testing, and testing is limited to individuals above the age of 16 years.

The Act is currently being reviewed.

An Act regulating the use of Biobanks is currently being prepared.

Dr. Karen Helen Orstavik

ESHG EDUCATION COMMITTEE

It is in the nature of our profession to teach, and all of us are involved to one degree or another in assisting undergraduates, graduates or the general public to explore the fascinating field of human genetics. But with the technical advances and resulting explosion of knowledge human genetics has undergone over the past decade, we must do more. In the public eye, medical genetics has passed from a marginal discipline to one with a dominant place in the future of medicine.

Until recently, and for 34 years now, our educational efforts as a Society were concentrated on holding a stimulating and successful annual meeting, for initial training or continuing education of members of our profession. But no matter how successful, training of ourselves is not enough.

For the past three years, the ESHG has lent our support to the courses of the European School of Genetic Medicine, which the European Genetics Foundation (www.eurogene.org) has held nearly 15 years now. Our association with the EGF is also manifest financially; in 2002 the ESHG will offer 5 scholarships to students attending one of three EGF courses. But again, we are talking about the training of individuals already motivated to learn and practice genetics.

This, then, is not enough either. Shouldn't we be doing things like proposing a core European curriculum for medical students? Setting up a system of Continuing Education which would be accredited Europe-wide? Setting out to better inform the general public, beginning in grade school or high school? And perhaps even more essential, short-term, work with that majority of health care professionals whose formal training hardly included genetics, so they will understand the goals, power and limitations of medical genetics as practiced today?

On the other hand, the ESHG as a Society has only a part-time administrative staff, and almost all of us are already overextended professionally. What to do?

First, as a Society, set some priorities for genetics education. Concurrently, identify pre-existing courses, resources and research initiatives in education which can be made available to the wider scientific community. And next, through a network of interested individuals to whom the Society can provide an infrastructure, decide on which fronts, or in what manner, the Society can promote education: pre-medical, medical, pre-doctoral, doctoral, post-doctoral, post-graduate, continuing professional education, genetics for the public at large.

The Board has taken a small step in that direction, in deciding to make the "ad hoc" education group a permanent committee of the ESHG. There is room on this committee for dedicated individuals! And the committee would like to begin setting up a network for educational matters.

With this introduction, the Board is asking for your help in two ways:

If you are personally interested in working on the ESHG education committee, or being part of the "educational resource network," please contact me (Celia DeLozier, e-mail: delozier@cmu.unige.ch) with your ideas and suggestions! The Board will formalize the organization of the education committee at our May meeting.

Please tell us (myself or the Administrative office: eshg@eshg.org) of existing sources (e.g. nationally-organized courses open to those from other countries) and ongoing projects (such as the survey concerning non-medical genetics education curricula which Rodney Harris is leading with the help of the EU), so that we can combine our efforts!

Célia DeLozier-Blanchet

Geneva, November 2001

MAKE A MAJOR CONTRIBUTION TO THE SOCIETY – AS PRESIDENT OR BOARD MEMBER

The Society Board intends that the President-Elect and two 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)

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.

EUROPEAN GENETICS: NATIONAL NEWS AND VIEWS

ITALY

The 4th meeting of the Italian Society of Human Genetics (S.I.G.U.) will be held in Orvieto on the 28-30th November 2001. Sixty eight abstracts have been selected for oral presentation among 339 scientific contributions, and have been distributed in parallel sessions of: Molecular Genetics, Cytogenetics, Neurogenetics, Clinical Genetics, Complex Traits and Cancer Genetics. The sessions will be chaired by Italian experts in the specific fields. The program of the meeting schedules also a symposium on Ionic Channelopathies, an updating of Continuing Medical Education in Genetics, a round table on Postgenomic Medicine, and two lectures, on Genetics of male Infertility (in collaboration with Italian Society of Medical Andrology) and on the Impact of Internet on Genetic Diseases, as experienced by Orphanet" (S. Aymé; Paris).

THE EUROPEAN GENETICS FOUNDATION HAS MOVED!

Please note our new address:

European Genetics Foundation, Pad.23 c/o direzione Clinica Oculistica, Policlinico S.Orsola, Via Massarenti 9, 40128 Bologna, Italy.

Tel: +39 051 306171 email: egf@eurogene.org website: www.eurogene.org



DEPUTY SECRETARY-GENERAL

Célia DeLozer-Blanchet wishes to relinquish the post of deputy secretary-general from May 2002. Any members who are interested in helping the Society in this way are invited to contact the ESHG administrative office in Birmingham, UK.

The Society has been very grateful for all the hard work which Linda Bates, who was an assistant in the office in Birmingham, performed on behalf of the Society. Linda was probably the person who you dealt with for your subscription enquiries. She has now moved on to University – she is reading English Literature at Cambridge University. We all wish her well for the future.



CALENDAR OF NATIONAL AND INTERNATIONAL MEETINGS

DECEMBER 2001

41st Annual meeting of the American Society for Cell Biology

8-12 December 2001

www.ascb.org or ascbinfo@ascb.org

European School of Genetic Medicine: 3rd course in Molecular Cytogenetics and DNA Arrays

12-15 December; Bertinoro Conference Centre, ITALY

organised by EGF: www.eurogene.org or courses@eurogene.org

JANUARY 2002

Diabetes Mellitus: Molecular Mechanisms, Genetics and New Therapies

10-16 January; Keystone, Colorado, USA

organised by Keystone Symposia: www.keystonesymposia.org or info@keystonesymposia.org

FEBRUARY 2002

Genotype to Phenotype: Focus on Disease

19-24 February; Santa Fe, New Mexico, USA

organised by Keystone Symposia: www.keystonesymposia.org or info@keystonesymposia.org

MARCH 2002

Annual Clinical Genetics Meeting (American College of Medical Genetics)

13-17 March; New Orleans, Louisiana, USA

ACMG Administrative office: www.faseb.org/genetics or gcantrel@genetics.faseb.org

European School of Genetic Medicine: XV Course in Medical Genetics

17-23 March; Bertinoro Conference Centre, ITALY

organised by EGF: www.eurogene.org or courses@eurogene.org

ESGM: 1st Course in Comparative and Functional Genomics

24-26 March; Bertinoro Conference Centre, ITALY

organised by EGF: www.eurogene.org or courses@eurogene.org

APRIL 2002

Third International meeting on the Genetic Epidemiology of Complex Traits

4-6 April; Cambridge, UNITED KINGDOM

Twin Research and Genetic Epidemiology Unit, St Thomas' Hospital, London, United Kingdom

Cambridg2002@twin-research.ac.uk

IV International Symposium on Preimplantation Genetics

10-13 April 2002; Limassol, CYPRUS

www.IVFonline.com

Human Gene Mapping 2002

14-17 April; Shanghai, CHINA

organised by HUGO; www.hugo-international.org/hugo/ or www.hgm2002.mrc.ac.uk

American Cytogenetics Conference

25-28 April; Santa Fe, New Mexico, USA

Laurel.Estabrooks@Gensyme.com or Sue.Berend@Gensyme.com

MAY 2002

European Human Genetics Conference 2002

25-28 May; Strasbourg, FRANCE

European Society of Human Genetics: www.eshg.org or eshg@medacad.org

SEPTEMBER 2002

British Human Genetics Conference

23-25 September 2002; University of York, UNITED KINGDOM

www.bshg.org.uk

CONTRIBUTIONS

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or Mrs. Ruth Cole (eshg@eshg.org)