

European Society of Human Genetics

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ICHG: VIENNA IN MAY WILL BE THE PLACE TO BE!

Dear colleagues and friends,

2001 will be an historic year for the ESHG. Indeed, the International Congress of Human Genetics will be hosted by our society in Vienna and it will be a high level scientific event. Moreover, chances are that we will not be able to repeat this for another 20 years or more. The congress will provide a unique opportunity for all our members to meet in the unique historic setting of Vienna and to intensely share scientific, cultural and social activities with our colleagues from all over the world.

The different congress organizing committees have been working very hard to prepare this meeting to the best of their abilities. As a result, abstracts are flowing in from all over the world through the net; numerous applications for the EU funded fellowships have been received from non-EU scientists and the exhibition space is about to be sold out. In January the final selection of abstracts for oral presentation will be made by the ISPC to finalize an already impressive and attractive scientific program. In addition, a series of satellite meetings and workshops before, during and after the congress are being organized. All that is left is the arrival of a massive number of participants in good spirits to make this congress a unique and unforgettable event in the annals of the society and of the International Federation of Human Genetic Societies. Please, block these dates in your calendar and check the website for all further information. Stimulate your collaborators, in particular the younger ones, to leave their benches or patients for a few days. For many of us it will be a unique opportunity of a lifetime. We expect all of you, without exception from **May 15 to May 19, 2001** in Vienna.

JJ Cassiman

(Chairman, Congress Committee)

Visit <http://www.ichg2001.org> for the latest information

INTERNATIONAL RECOGNITION FOR ESHG GENETIC SCREENING REPORT

The activities of the Public and Professional Policy Committee in the Year 2000

During the year 2000, the Public and Professional Policy Committee organized three workshops on topics of concern in the field of Human Genetics. The purpose of the workshops was to identify, from a professional viewpoint, the most important/pressing/burning ethical issues relating to the topic debated. On the average, two experts per European country were invited. These experts were representatives of the seven following sectors:

- | | |
|--------------------------------------|-------------------------------------|
| 1 - Medical Genetics | 5 - Biotechnology / Pharmaceuticals |
| 2 - Human Genetics Societies | 6 - Insurance / Employment |
| 3 - Ethical, Legal and Social Issues | 7 - European Union Institutions |
| 4 - Support Groups | |

After each workshop, the PPPC wrote up a final report and issued recommendations, including all the comments made during the workshop and sent prior to it. These documents have been put on the ESHG website for public consultation and discussion.

The recommendations will be endorsed at the Vienna ESHG annual meeting in May 2001 by the ESHG members and the International Federation of Human Genetics Society (IFHGS).

The recommendations on *Population Genetic Screening Programmes* (Workshop 1 held in Amsterdam in 1999) have been endorsed at the Amsterdam ESHG annual meeting in May 2000 by the ESHG members; they have been also endorsed by the Board of Directors of the American Society of Human Genetics. The recommendations have been published in the EJHG (vol. 8, n. 9, Sept. 2000). The recommendations and the report are available on the ESHG website.

Workshop 2: Genetic Information and Testing in Insurance and Employment: Technical, Social and Ethical Issues

This topic was discussed during a workshop in Manchester (UK), February, 25-27, 2000 where 47 experts from 14 European countries were invited. The workshop was organized by Martin Bobrow, Peter Farndon, Marcus Pembrey, and Sandy Raeburn.

The report reviews the technical, social, economical, and ethical aspects of advances in genetics and the concerns of parties who are involved, i.e. the insurers, the employers and the public. Issues debated by these parties are reported as well as the existing guidelines and legislation on this topic. The report was reviewed critically by a wide range of people and organizations for comments.

The recommendations and the report are available on the ESHG website for consultation until December 31st 2000.

Workshop 3: Data Storage and DNA Banking for Biomedical Research: Informed Consent, Confidentiality, Quality Issues, Ownership, Return of Benefits - A Professional Perspective

This topic was discussed during a workshop in Paris (France), April, 07-08, 2000 where 50 experts from 12 European countries were invited. The workshop was organized by Ségolène Aymé, Jean-Jacques Cassiman and Joerg Schmidtke.

The report reviews the existing professional guidelines, legal frameworks and other documents related to the data storage and DNA banking practices in public and private sectors in Europe. The report also defines questions which need debate, in particular the consent requirements for banking and further use of samples, the control of banked samples and quality issues, the ownership of banked samples and the return of benefits to the community. The following case-studies are described: the ALSPAC study, deCODE genetics and the Act on a Health Sector Database, and Guthrie cards. The report was reviewed critically by a wide range of people and organizations for comments.

The recommendations and the report will be available on the ESHG website very soon.

Workshop 4: Provision of genetic services in Europe - Current Practices and Issues

This topic was discussed during a workshop in Helsinki (FIN), September, 08-09, 2000 where 43 experts from 17 European countries were invited. The workshop was organized by Helena Kääriäinen, Domenico Coviello, Ulf Kristoffersson and Lisbeth Tranebjærg.

The report reviews the national and international regulatory frameworks of genetic services in Europe and elsewhere. It also defines questions which need debate, in particular the organisation, quality assessment, public education, partnership with support groups and the multicultural aspects of genetic services. The report has been sent out to a wide range of people and organizations for comments.



The recommendations and the report will be available on the ESHG website very soon.

*Members of the PPC after a committee meeting in Helsinki discussing their report on Provision of Genetic Services in Europe - Current Practices and Issues
Back row: Ulf Kristoffersson, Joerg Schmidtko, Marcus Pembrey, Lisbeth Tranebjærg, Peter Farndon, Gerry Evers-Kiebooms, Domenico Coviello and Leo ten Kate
Front row: Beatrice Goddard, Segolene Ayme and Helena Kaariainen*



MEMBERSHIP RENEWAL **INFORMATION FOR 2001**

Over the past year, the Society's own office in Birmingham, UK has been working hard to set up a new system for membership administration with the result that we now have over 1000 members again. And now it is time to ask you to renew your subscription for 2001! Please find a form enclosed 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 2000 until we receive your renewal form so please do not delay. Return your form today - we have enclosed an addressed envelope to make it even easier!



We are also pleased to offer reduced subscription to two journals published by Current Trends, part of Elsevier Science - 'Current Opinion in Genetics & Development' and 'Trends in Genetics'. Please see the enclosed flier and membership renewal letter for further details.

EUROPEAN HUMAN GENETICS CONFERENCE GOES FROM STRENGTH TO STRENGTH

The ESHG's annual meeting, which was held in Amsterdam during May 2000, was attended by representatives of many countries both European and worldwide.

There were over 1000 international participants, over 650 posters and 59 spoken presentations from submitted abstracts. The young Investigator awards were won by Alessandra Maugeri from the Netherlands and Roman Chrast from Switzerland.

The symposia and plenary sessions generated exciting scientific discussions and the efforts of the local organising team ensured that the social programme was enjoyed by all.



Exhibition Bookings for ICHG 2001

At the ESHG exhibition in Amsterdam, which was a great success and had over 50 companies represented, the 3 following companies were the first to book their exhibit space for the ICHG 2001 Exhibition in Vienna.

Their enthusiasm and quick decision is of course highly appreciated and is therefore rewarded with a short description of their company and picture of their ESHG 2000 booth.

Their initiative is followed by many other companies, making the ICHG 2001 Exhibition the most important international platform for the industry in the field of human genetics!



ACADEMIC PRESS

Email: ap@acad.com

Website: www.academicpress.com

Academic Press announced the new journal *Molecular Therapy* in addition to *Genomics*, *Molecular Genetics and Metabolism* and *Journal of Molecular Biology*. Featured book titles included: Bishop: *Genetic Databases*; Ashcroft: *Ion Channels and Disease*; Huang: *Nonviral Vectors for Gene Therapy*; and Lechler: *HLA in Health and Disease*.

INGENY

Email: info@ingeny.com

Website: www.ingeny.com

Ingeny is a manufacturer of equipment for DGGE. DGGE-primers are available for a growing number of genes (e.g. CFTR, BRCA1, BRCA2,

MLH1, MSH2, MSH6, p53). These primers, together with Ingeny's equipment, yield a virtually 100% mutation detection. Other techniques that will readily run in Ingeny's equipment are SSCP, CDGE, TGGE and PTT.



GENTRA SYSTEMS, INC.

Fax: +1 763 5430699

Website: www.gentra.com

Gentra Systems, a leading manufacturer of nucleic acid purification products for the clinical molecular diagnostic and research markets, exhibited its PUREGENE® DNA Isolation Kits, PURESCRIPT® RNA Isolation Kits, and the GENERATION® Purification System. In addition, Gentra introduced its new automated nucleic acid purification system.

GENETICS IN EUROPE: MEDICAL GENETICS IN HUNGARY

At the end of an animated decade devoted to a shift from communism towards democracy, Hungary is a country full of economic, social, legal, scientific and educational issues to be addressed and changed. The Republic of Hungary is a member of the Council of Europe, and intends to enter the EU in the coming years. The territory is small (93,032 km²) with a population of 10 million (including 0.5 million Gypsies or Romas, the only numerically significant ethnic group). Demographic trends are unfavourable to population growth, since the birth-rate is low (except among the Romas) and death-rate relatively high. The rate of consanguinity is low (except, again, among the Romas). Frequencies of the more common Mendelian disorders and of congenital anomalies correspond well to those reported in international surveys (rates of cleft palate, cardiovascular defects and club foot among Gypsies slightly exceed the overall Hungarian figures).



At present there are 13 registered genetic counselling clinics in Hungary. Eight of them are functioning in university hospitals, the others in various institutions. The head of each clinic is a medical doctor (mainly pediatricians and gynecologists) with a 'human geneticist' sub-specialty. The genetics clinics are prepared to deal with the most frequent genetic disorders, some of them being specialized in specific pathologies. Most of the counselling clinics have a laboratory set-up for routine chromosome analysis, some also for molecular genetic techniques. Similar to the tendency in medical genetics in the rest of the world, facilities for DNA analysis (mostly PCR) emerged also in Hungary in many non-genetic diagnostic laboratories. They mostly do research on somatic genetics issues; however, genetic testing and counselling have also been initiated by these laboratories. A process to accredit these labs and to create by-laws to control their functions is in preparation. These are a few genetics research groups outside the university clinical genetics units. Of these, the best-equipped and most productive institute is the Biological Research Centre in Szeged.

Although there are some labs whose facilities for mutation analysis developed significantly in recent years, the capacities are far behind those of developed Western countries, and for many pathologies Western European or overseas collaborations are needed (and realized, mainly thanks to personal contacts).

As an important element of the social restructuring, a dramatic transformation has been going on also in higher education over recent years. The four independent medical schools (Budapest, Debrecen, Pécs, Szeged) merged with other institutions into large universities. Medical genetics departments exist only in the medical faculties of Pécs and Szeged.

Medical education in Hungary takes six years. Basic elements of molecular genetics are taught in the first two years as a part of biology and biochemistry courses, while medical genetics is part of the fifth year curriculum. In the two universities with independent departments a final exam is required, in the two others the course is included in pediatrics or obstetrics.

Until recently, qualification for 'human genetics' as a medical specialty was awarded by individualized judgement and without a qualifying examination, based on MD diploma, clinical genetics practice (minimum of 4 years), two recommendations and a list of publications. At present there are 34 board-certified human geneticists.

A new system for postgraduate medical training (residency) was introduced in 1999, fitting into the EU expectations. This means that primary specialization is possible in 33 medical disciplines. Unfortunately, the lack of medical/clinical genetics in the approved list of the *Union Européenne des Médecins Spécialistes* made it harder to have genetics approved as a registered specialty by the Hungarian health authority. However, following steady argumentation, it seems finally that clinical genetics will be enrolled as a secondary specialty. Guidelines for appropriate training are in preparation. There is, as yet, no special status or training for genetics nurses, counsellors or lab associates.

Medical genetics in Hungary is coordinated by the *Hungarian Society of Human Genetics*, created in 1972, which now includes about 240 members. The *College of Medical Genetics*, a board under direct subordination of the Ministry of Health, which was withdrawn after a thoughtless decision in 1993, is to be re-established in 2001.

The health care sector in Hungary suffers from underfinancing and shortages (surprising?). Needs and running costs of genetic services funded partly by state budget and partly by social insurance exceed the resources. Technical facilities have lagged behind the rapid development of our discipline. These gaps cause difficulties for both medical services and research.

There is no specific regulation of genetic testing in laboratories. In 1999 an *ad hoc* committee was called together in order to elaborate guidelines and recommendations on ethical, legal, social issues, and on quality control and accreditation for both public and private labs. By-laws might be expected by early next year.

- G. Kosztolanyi, Pécs, Hungary

EDUCATIONAL ACTIVITIES

The link between the European Society of Human Genetics and the European Genetics Foundation, which has as its primary goal and activity the development of genetics education in Europe, continues to be strengthened. Currently, some financial support comes from the ESHG, in the form of two fellowships (currently ear-marked for individuals from Eastern European countries) to the post-graduate genetics course held each March. Other national societies, including the German and Swiss genetics societies, are also offering scholarships to EGF courses.

For more information on EGF courses, see the Foundation's website (<http://www.eurogene.org>) or follow the link from the ESHG's home page. Other courses held this fall included the first course in "Genetics and Renal Disease" (15-18 November), first course in Bioinformatics (22-26 October) and the second course in Molecular Cytogenetics (19-22 November 2000). A more detailed summary of two of this fall's courses is given below.

5th Gaslini-IARC-Menarini Course in 'Cancer Genetics and Pediatric Oncology' Sestri Levante (Italy), Sept. 27 - Oct 1, 2000.

This year Sestri Levante Course in Cancer Genetics was attended by some 70 students from all over Europe including Russia and various East European countries, but also from the USA, Canada, Cuba and Argentina. A total of 15 EC, 4 NIH and 2 EGF fellowships were assigned to all the requesting students.

The course was structured according to the "Sestri Levante scheme", i.e. with conventional lectures in the morning sessions and more interactive workshops in the afternoon. Moreover, three evening research seminars were included in the program to allow the students to attend more research-oriented lectures from selected faculties. Particular attention was devoted to the informal interaction between students and faculty members during lunches, workshops, and, last but certainly not least, in the evening beer-drinking sessions!

Each day of the course has been devoted to a specific theme ("Cancer as a Genetic Disease", "Familial Cancer Syndromes", "Cancer Biology", and "The Molecular Basis of Cancer") around which both morning lectures and afternoon workshops were centered. Faculty members from Europe and the United States included leading experts of their respective fields such as Jerry Shay (apoptosis and senescence), Carmen Sapienza (genetic imprinting), René Bernards (cell cycle), Gerard Evan (c-myc), and many others. Also, particular attention was devoted to the issue of "Pediatric Oncology" and hereditary syndromes like Neuroblastoma, Wilms Tumor, Retinoblastoma and Leukemia.

The course was enthusiastically evaluated by the vast majority of the students.

(Dr. Riccardo Fodde, PhD, Dept. of Human and Clinical Genetics, Leiden University Medical Center, Leiden, The Netherlands)

2nd annual EGF course 'Genetic Counselling in Practice',

Sestri Levante (Italy), October 15-21, 2000.

This short course in principles and practice of genetic counseling was organized for the second time this year by the European Genetics Foundation and held in the new permanent venue of the EGF in a transformed 15th century Dominican monastery in Sestri Levante, Italy. Given that only two European countries (Great Britain and The Netherlands) currently offer degree programs in genetic counseling, this course intends to help fill knowledge gaps for those coming from other specialities and disciplines-nurses, midwives, laboratory geneticists, primary care physicians who find themselves called to do genetic counseling. The genetic counselor will be increasingly called to serve as an interface between the patient, his or her physician and the laboratory; one of the goals of the course is to clarify and prepare (future) genetic counselors for this task.

The forty-three "students", who came from 13 countries (including several from Eastern Europe) also included several experienced genetic counselors, as well as a number of physicians relatively new to their genetics departments. The "traditional" format of Sestri courses includes lectures in the morning and small-group workshops in the afternoon; the moderate size and the collective experience of this year's group fostered discussion and exchange of ideas, providing a learning experience for the faculty as well as students. New this year was an "Edushare" forum for exchanging educational material, brochures, Web sites, etc.

Eleven students received partial or full fellowships to attend the course and it is hoped that financial assistance of this type can be offered in the future.

Given numerous requests from Italian colleagues for a similar course in their language, this second course (in English) overlapped with a 4-day genetic counseling course in Italian, which "shared" some faculty as well as a few students.

The third annual course in Genetic Counseling in Practice is planned for late September 2001.

(Dr. Célia DeLozier-Blanchet, Division of Medical Genetics, Geneva University Medical School and Hospital, Switzerland)

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.

HONORARY DEGREE AWARDED TO LEADING GENETICIST

The Medical Faculty, University of Essen presented the honorary degree Medical Doctor (M.D.) (Dr. med. h.c.) to Professor James German, III., M.D., New York City on Friday, July 7, 2000

James L. German, III., M.D. has been a witness to the development of human genetics during the past 35 years. He has seen and contributed to perhaps the most fascinating years in the history of medicine and biology, especially in human genetics.

The close relationship between Professor James German and the Medical Faculty of Essen, especially the Department of Human Genetics through personal contacts with Eberhard Passarge, MD, as well as his extraordinary and internationally recognized contributions on the field of human genetics are the main reasons for bestowing this honorary degree on Professor German

For more than four decades Professor German has made important scientific discoveries: the identification of individual chromosomes by means of new methods (autoradiography, 1962), the evidence that an exchange of chromosomes occur during mitosis in somatic cells (somatic recombination, 1964), the evidence of instability of chromosomes in a rare and genetically determined disease (Bloom syndrome, 1965). His main scientific work has been the systematic analysis that lead to the understanding of important developmental processes, for example the development of carcinoma.

Professor German is author of more than 300 scientific works in leading scientific journals and books.

He has published four books on his special field cancer, genes, and chromosomes.

HUMAN GENETICS IN THE NEWS

BioNews is a free, weekly email newsletter covering the latest advances in human genetics and assisted reproduction. Sent to subscribers each Monday, BioNews provides summaries of the week's news, commentary on the leading story and recommendations on relevant articles, books, conferences and other events.

As well providing an update of the latest scientific, social, legal and ethical issues in areas such as human genome research, preimplantation diagnosis, gene patenting, embryo research and cloning, BioNews also aims to reflect the coverage they receive in the UK newspapers. You can sign up to BioNews and view the latest issue at: <http://www.progress.org.uk/News/>

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CALENDAR OF NATIONAL AND INTERNATIONAL MEETINGS

(see also <http://www.eshg.org/Noticeboard>)

FEBRUARY 2001

DNA Analysis 2001

23 February 2001, Florence - dna2001@medway.ch

MARCH 2001

European School of Genetic Medicine's 14th course in Medical Genetics

25-31 March, 2001; Sestri Levante, Italy - <http://www.eurogene.org> or eurgef@tin.it

Annual Clinical Genetics Meeting (American College of Medical Genetics)

March 1-4, 2001; Miami, Florida - contact: Gerry Gurvitch (gurvitch@faseb.org)

The Society for Inherited Metabolic Disorders Annual Meeting

March 4-7, 2001; Miami, Florida - Contact: Leslie Lublink (lublink@ohsu.edu)

First Euroconference on Animal Models of Human Diseases: Modelling Human Cancer in the Mouse (European School of Hematology)

9-12 March, 2001; Sesimbra, Portugal - e-mail: ghyslaine@eshg.org or www.eshg.org

APRIL 2001

II International conference on Chromosome 21 and Medical Research on Down Syndrome

Fundacio Catalana Sindrome de Down and Down Syndrome Medical Interest Group

April 6-7, 2001; Barcelona, Spain - www.fcsd.org or Bonnie Patterson (pattbO@chmcc.org)

Human Genome Meeting 2001 (HUGO)

April 19-22, 2001; Edinburgh, Scotland - <http://hgm2001.hgu.mrc.ac.uk>

MAY 2001

10th International Congress of Human Genetics

15-19 May, 2001; Vienna, Austria - www.ichg2001.org

3rd World Congress of Pediatric Cardiology and Cardiac Surgery

Toronto, Canada, May 2001

Prior to the main meeting The Fetal Centre is organising the satellite symposium - Advances in Fetal and Perinatal Cardiology - for details see <http://www.sickkids.on.ca/fetalcentre/AdvancesInFetalPerinatalCardiology.asp>

JUNE

Eleventh International Clinical Genetics Seminar on 'Genetics of Diabetes Mellitus'

Heraklion, Crete, 9-14th June 2001

The Seminar will deal with genetic and etiopathogenesis aspects of all inherited types of diabetes mellitus.

Faculty includes: Graeme Bell, Stefan Fajans, Philippe Froguel, Leif Groop, Jorma Ilonen, Cecile Julier, Mikael

Knip, J.A.Maasen, Jørn Nerup, Alan Permutt, Flemming Pociot, Anders Green, Hans Åkerblom and others.

Further information from : Dr C Bartsocas, PO Box 17177, GR-10024 Athens, Greece. (Fax: +30 1 779 6461, email:

cbartsok@cc.uoa.gr) or the congress secretariat web site: www.triaenatours.gr.

AUGUST

9th Biennial Southern African society of Human Genetics Congress

12-15 August, 2001; contact Philip Venter (philipve@mweb.co.za) or Lynn Luttig (luttig@unin.unorth.ac.za)

SEPTEMBER 2001

British Human Genetics Conference

10-12 September 2001; York, United Kingdom - www.bshg.org.uk

OCTOBER

51st Annual meeting of the American Society of Human Genetics

12-16 October, 2001; San Diego, California

*Appropriate announcements are published free of charge here and on the ESHG website.
See below for contact details.*

CONTRIBUTIONS

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