European Society of Human Genetics

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What's in this issue

This issue contains the agenda and reports for the annual membership meeting on Monday 27th May 18:15 in Room Tivoli, Palais de la Musique et des Congrès, Strasbourg

Please bring it with you.

BOARD MEMBERS

New President Elect

Statement of Professor Veronica van Heyningen, President Elect 2002/2003

The European Society of Human Genetics has played a major role in fostering collaborative research interactions in Europe and internationally, with a strong focus on the societal and ethical aspects of the work. I would like to have the opportunity to make a contribution to this endeavour. I have long promoted the concept that the study of human variation and disease provides major insight into biological mechanisms, which in turn can be used to ameliorate the human condition; to understand and manage human health, particularly as the age spectrum shifts world-wide towards more elderly people. Turning the information from the human genome project into useful, realistic knowledge will require not only imaginative research from scientists and clinicians, but also informed endorsement from society. For this, we need to renew efforts at communicating the complexities of genetics and biology. There is a need for new initiatives in genetic education from an early age. Meaningful dialogue on complex ethical issues must be built on solid understanding of the principles. My work as a research scientist over three decades, and, more recently, insights gained from membership of the UK Human Genetics Commission, provides me with a strong background and enthusiasm for this task.

New Board Members

Nominations from the ESHG members

Nominations were requested from the ESHG membership for two members of the Board. Two nominations were received. They are Thoas Fioretos and Christos Yapijakis. Their statements follow.

Thoas Fioretos (Sweden) nominated by Gerry Evers-Kiebooms and Euterpe Bazopoulou-Kyrkanidou

Thoas Fioretos, born in 1962, received his MD in 1988 at the Medical Faculty of Lund University, Sweden. He obtained his Ph.D. at the Department of Clinical Genetics, Lund University Hospital, in 1996 and became a board certified clinical geneticist in 1999. Currently, he is an associate professor of clinical genetics, working as a senior consultant and an active researcher

Christos Yapijakis (Greece) nominated by Jorg Schmidtke and Ulf Kristoffersson

I consider an honor my nomination for election as Board Member. I am a full member of ESHG since 1990, and participated in 8 annual conferences. Please allow me to introduce myself. My training both as a basic scientist and clinician involved degress in Biology and Dental Medicine, and graduate degress in Molecular Biology and Human Genetics at the Universities of Athens (Greece) and California (USA), receiving scholarships for the entire study period. I also took the Medical Genetics course (European School, Italy). Since 1993, I have been at the Neurology Department (University of Athens), where I contributed in the establishment of a (unique in Greece) Neurogenetic Unit specialized on research (14 programs including 4 BIOMEd) and diagnostics. Beside molecular work, I am involved in clinical diagnosis of craniofacial syndromes and genetic counselling. I was President of the Greek Biological Society and Board Member of the European Countries Biologists' Association (1993-2000). I am currently Board Member of the Hellenic Association of Medical Geneticists. I will try my very best to contribute to the Board in this sensitive period in which ESHG will attempt to establish the specialty of Medical Genetics in a European level. I believe that my experience in European and National Boards, and my combined clinical/molecular perspective may be beneficial to the Board, in which South and East European countries are under-represented.

Nominations from the ESHG Board

The Board of the Society, according to the Statutes may nominate up to two members of the Board, for approval by the Society. The Executive Board is proposing one nomination.

Milan Macek (Czech Republic)

I am an Associate Professor of Medical Genetics at Charles University 2. School of Medicine, Institute of Biology and Medical Genetics - Cystic Fibrosis Centre, Prague, Czech Republic. Recently, I also obtained a D.Sc. degree in general genetics. Shortly after medical school I started my career in pediatric / molecular genetics: initially as a fellow at the Institute of Human Genetics, Free University Berlin (now Humboldt University) and then during the early nineties I completed my scientific training within my second postgraduate fellowship at the Center for Medical Genetics, Johns Hopkins University School of Medicine, Baltimore (USA). My research interest focuses on molecular genetic / population genetic studies in cystic fibrosis and respective genotype phenotype correlations. I have also been involved in the organization of scientific workshops, such as the 1997 HUGO Mutation Detection Workshop at the Mendel´s Monastery in Brno and the EC 1996 PECO and 1997 INCO-BIOMED symposia in Prague. Furthermore, I have been teaching Czech and foreign medical students in medical and molecular genetics. I would like to utilize my participation in the ESHG board to promote up-to-date research in human genetics and to harmonize provision of molecular genetic services in Central and Eastern European countries with the EU standards.

Annual Membership Meeting 2002: Agenda

Monday 27th May at 18:15 in Room Tivoli, Palais de la Musique et des Congrès,

Opening by the President of the Society, Professor Stylianos Antonarakis

1. Activity report of the Society 2001-2002
2. Financial report of the Society 2001
3. Discharge of the board members for the year 2001-2002

Opening by the new President of the Society, Professor Gert-Jan B van Ommen

4. Results of election for president-elect
5. Results of election for board members
6. Membership fee 2003
7. Sites of future European Human Genetics Conferences
8. Budget proposal 2003
9. Major policy questions proposed by Board

EUROPEAN SOCIETY OF HUMAN GENETICS

MINUTES OF MEMBERSHIP MEETING HELD ON THURSDAY 17TH MAY 2001, VIENNA

The meeting was opened at 18h35 by Gerd Utermann, who welcomed the new President, Stylianos Antonarakis. Prof. Utermann was thanked for his year as President, and also as host of the current meeting. 71 members were present at 19h00.

I. ACTIVITY REPORT

(Peter Farndon, Secretary General):

- Newsletter No 4 (April 2001) contains complete reports of committee activities
- The ESHG membership is growing with 200 new members joining in the last year. The Secretary General reported that the Society is thriving, as is the Website; the activities of the PPPC having a major influence on this growth.
- The membership accepted the Board's proposal to prolong the mandates of several individuals :
  - Gert Jan van Ommen as Journal Editor (3 years),
  - Segolene Ayme as chairman of the PPPC (3 years),
  - Andrew Read as chairman of the Scientific Program Committee (2 years),
  - Celia DeLozier as Deputy-Secretary General (1 or 2 years).

II. 10th INTERNATIONAL CONGRESS OF HUMAN GENETICS *(Jean-Jacques Cassiman)* :

- About 3000 people present, from 85 different countries (including invited speakers and exhibitors). 1800 posters accepted. The EC funded 181 fellowships
- Financially, the meeting should at least break even.

III. REPORTS FROM PERMANENT COMMITTEES

Professional and Public Policy Committee (PPPC) *(Segolène Aymé):*

- Three individuals have resigned from the PPPC (Peter Farndon, Martin Bobrow and Sandy Raeburn) The Board approved three new members: Georgi Kostolanyi, Violetta Anastasiodou, Shirley Hodgson.
- PPPC membership will be increased by the "presidential team" (President, Vice-President and President-Elect) acting as associate members, as of this year. PPPC membership is otherwise on a rotating basis.
- PPPC/ESHG policy recommendations will be updated every 3 years.
- The recommendations paper will be published in EJHG and a book may be produced to cover the background papers
- Two documents ratified by the PPPC now appear on the Website and will be published in the European Journal of Human Genetics: Population Screening and Genetics and Insurance.
- Members were encouraged to comment during the following two months on two further reports on the website: Databases and DNA Banking, and Genetic Services Provision.
- Topics being considered for future position papers: Pre-implantation diagnosis; Genetic contribution to multifactorial diseases; From Good Science to Sound Clinical Practice. Prof C Buys suggested another topic from the floor: Orphan genotypes.

International Federation of Human Genetics Societies *(Marcus Pembrey)* :

- The development of this group, formed in 1996, was reviewed. The meeting in Vienna is the first international meeting since the IFHGS was founded. American Society of Human Genetics, European Society of Human Genetics, and Human Genetics Society of Australasia were the founding members. There are strict criteria for membership in order to serve WHO and international organizations. Currently there were four full members and 40 affiliated members.
- Marcus Pembrey reported that his term of office would be completed at the end of 2001.The Board has approved Jean-Jacques Cassimann as the ESHG's next liaison officer for the IFHGS for a 5-year term.
- Malcolm Ferguson-Smith reported that it was possible that the previous international association which organized international meetings has not been formally deconstituted., and he asked about the monetary assets of the former Permanant Committee. Marcus Pembrey agreed to determine the current position.

IV. EUROPEAN JOURNAL OF HUMAN GENETICS *(GJ van Ommen)* :

- The citation index in 2000 was 3.18. Average time from submission to response is 7.2 weeks, revision to final answer is less than 1 week and acceptance to print is 3-4 months. A few papers are rejected immediately.
- Coverage: several new categories, including Genetics in Practice (disease profiles, diagnosis, and summary of genotype-phenotype correlations) are being developed.

- The Editor-in-Chief encouraged members to submit manuscripts.
- The contract with our publisher, Nature Publishing Group, is due for review in 2002. A mandate was sought from the membership to consider bids from companies. ESHG has been approached by several (including Karger, Elsevier, Blackwell) and some proposals are very attractive. It was noted that the Journal is self-supporting, but our dealings with Nature Publishing Group in some areas (e.g. electronic publishing options) have not been as satisfactory as the Society would wish. The membership endorsed a mandate to allow the Publications Committee to take over these negotiations.

V. BUDGET *(Peter Farndon):*

- The ESHG is in a steady financial state; there was a surplus of income over expenditure in 2000, income coming from membership subscriptions, the Journal, industrial partners and bank interest. There is a continuing need to increase the Society reserves because the ESHG is responsible for pre-financing our future conferences.
- The membership ratified the proposal that membership fees remain the same in 2002.
- The surplus income over expenditure in 2000 will be reserved for producing a membership directory and financing PPPC activities.
- The accounts were approved by the Membership.

VI. ANNUAL MEETINGS COMMITTEE
(Peter Farndon):

- The European Human Genetics Conference in 2002 will be in Strasbourg and will be organised by the Vienna Medical Academy. The organisation of the 2003 meeting (Birmingham) is under review.
- The current contract with Rose International ends with the 2003 meeting. It is expected that a bidding process will be opened for congress organiser and exhibitions organiser for the 2004 Munich meeting..
- The ESHG Executive has determined that the Society's current and planned administrative support would not be sufficient to organize the annual meetings itself.
- The ESHG will be pleased to receive invitations from National Societies for their countries to host the European Human Genetics Conference from 2005.

VII. MAJOR POLICY MATTERS TAKEN UP BY THE BOARD:

Medical Genetics as a specialty in Europe

(Gerd Utermann):

The Board has adopted a statement concerning the importance of recognizing Medical Genetics as a specialty. This statement intends to serve as a baseline text for the various national societies, and to help longterm in obtaining recognition on the European level.

— Professor Antonarakis presented the document, which was discussed. The need for recognizing laboratory specialties in medical genetics was highlighted. The statement was to placed on the website for 8 weeks and comments from the membership invited.

Committee for EU policies

(Jean-Louis Mandel):

The ESHG has formed a new committee which is to interact with the European Parliament. Members : Jean-Louis Mandel, Jean-Jacques Cassiman, Rob Elles, Stylianos Antonarakis.

Education committee:

The Board has created a permanent education committee extending its original mandate which was liaison with the European Genetics Foundation. Celia DeLozier will propose members and goals of the committee, the first task of which will be to evaluate genetics courses already available on various levels.

By-laws revision:

Marcus Pembrey is the chairman of an ad hoc committee (with Ulf Kristoffersen and Celia DeLozier) to propose modifications of the By-Laws to the Board. The Board has not yet had the opportunity to discuss the proposed changes before submitting them to the membership.

The meeting was closed by Stylianos Antonarakis at 20h00.

FUTURE ESHG CONFERENCES

European Human Genetics Conference

Birmingham, England May 3rd-6th, 2003

Munich, Germany June 11-15th, 2004

Activity Reports 2001-2002 (Pending approval by the board)

Secretary-General's report -

Peter Farndon

During the past year the Society has continued to develop its activities in several areas, as detailed more fully in the reports which follow.

The Society was very pleased to have hosted the 10th International Congress of Human Genetics in Vienna last May. It was a great credit to many people in our Society who had worked so hard in its organisation and our professional Conference Organiser and Exhibition Organiser, the Vienna Medical Academy and Rose International respectively.

Membership of the Society continues to grow, as does the impact of the European Journal of Human Genetics. As you will see from the following report, our publishing contract with Nature Publishing Group is due for renewal, and several other Publishers have expressed an interest in publishing the Journal with us. The latest position will be presented at the membership meeting in Strasbourg.

The Professional and Public Policy Committee has completed its first round of research. The reports are highly respected internationally, and the Committee, as you will read, is deciding which topics to consider next.

The Society is increasingly developing links with other European organisations, as well as continuing to play an important role in the International Federation of Human Genetics Societies. One particular initiative is the setting up of a Committee for EU Policies, also described below. The ESHG was instrumental in informing members of the European Parliament about the concerns of the genetics community over certain parts of the Fiori report, which included the banning of research on embryonic stem cells. Recognising the importance of professional standards, training and education the Society has set up its own Education Committee.

There has been a thorough review by the Executive Committee of many aspects of the role of the Society and its administration. We had planned to present revisions to the Society's Statues to the membership meeting in Strasbourg, but this may need to be postponed until all the changes required can be identified and incorporated.

We are actively seeking sites for future European Human Genetics Conferences, and the European Society encourages national societies to bring to our attention potential conference sites for the years 2005 onwards.

Professional and Public Policy Committee -

The PPPC has welcomed new members: Gyorgy Kosztolanyi, Violetta Anastasiadou and Shirley Hodgson. One meeting took place in Brussels in September 2001 where the topics to be addressed in the coming months were discussed. We selected the two following:

- Ethical, social and legal issues raised by genetic variation in drug response and disease risk.
- The interface between medically assisted reproduction and genetics: technical, psycho-social and ethico-legal issues.

The objective is to issue policy statements, guidelines and reports on ethical and social issues raised by these topics Reports will be produced as a result of working parties organised as follows : A background document will be drafted, based on the medical and scientific literature. About 150 experts will be asked to review it and to prepare their answers to defined questions which will be debated later, during a two day workshop of about 50 participants. The PPPC will finalise the document which will be put on the web site of the ESHG for public discussion for 2 months.

The PPPC will then draft recommendations. When finalised, both the documents and the recommendations will be published and widely disseminated. They will contribute to the debate among the community and will serve as references for policy makers and at the European Union level. An application has been introduced at the last call of FP5, in January 2002. Results are pending.

Ségolène Aymé

Scientific Programme Committee -

Andrew Read (Chairman)

The work of the SPC follows a fixed cycle. A few weeks after the ESHG conference we spend a weekend developing the framework for the next conference - this was done in Strasbourg in June 2001. Much email activity follows, and we take advantage of the presence of many SPC members at the American Society of Human Genetics conference to review progress - understandably, this year's meeting in San Diego was rather smaller than usual. The major work of reviewing and selecting abstracts takes place in January, culminating in a very hectic weekend at the Vienna Medical Academy offices to finalise the selections and award fellowships. Because this year's conference is being held in conjunction with the European Meeting on Psychosocial Aspects of Genetics, two EMPAG committee members (Gerry Evers-Kiebooms and Lauren Kerzin-Storrar) were involved in all stages of the process, and it is a pleasure to report that there was light and harmony at every stage.

Over 1300 abstracts were submitted for the Strasbourg conference. This all time high is gratifying evidence of increasing participation in the Society's programmes - but it does make the selection process harder! Jerome Del Picchia and his staff have our heartfelt thanks for being unfailingly helpful and well organised - they deserve much of the credit for making the whole process run smoothly. We can select only around 60 of the 1300 abstracts for oral presentation, which inevitably leaves many people disappointed. Scientific quality is the primary criterion for selection, but we also consider whether the data might be better suited for presentation on a poster, and we try to assemble a programme for each concurrent session that has some thematic unity. We have noticed that a few institutions or groups send in a very large number of abstracts. Probably they imagine that this will increase their chances of acceptance or selection for a fellowship. We would like to point out that in fact it achieves precisely the opposite effect. Where an institution has a large and diverse programme of high quality work, then of course we would like to see all of it - but "salami submission" simply dilutes the quality of each individual abstract. Given the pressure on poster space, we are having to be a little more stringent in our selection, and salami abstracts are the first to go.

Stylianos Antonarakis and Thomas Meitinger rotated off the SPC, and I thank them for their many positive contributions to our work. We welcome Karl-Heinz Grzeschik, Gudrun Rappold and Niels Tommerup as new members. My own time as chairman is coming to an end. Han Brunner has very nobly agreed to take on this task, at the invitation of the ESHG Board. We will work together on the programme for next year's meeting in Birmingham, after which I will leave the committee in his safe hands.

The International Federation of Human Genetics Societies

ESHG liaison officer: JJ Cassiman (M Pembrey until October 2001)

JJ Cassiman

In October 2001, Marcus Pembrey's mandate as liaison officer for the ESHG to the IFHGS came to an end. We are grateful to Marcus for the excellent job he did in keeping the momentum going in the IFHGS, in particular by stimulating and facilitating the creation of a South American continental society, the RELAGH. The three founding continental societies of the IFHGS do now have a fourth partner. M Pembrey attended the meeting of the Colombian Society for Human Genetics in an attempt to further strengthen the cohesion between the different founding societies of RELAGH. The four recommendation documents drafted by the PPPC of the ESHG during the Eurogapp project were also submitted to the IFHGS members for approval: the results of this poll will be reviewed at the next Board meeting. At the Vienna board meeting an exchange with V Boulisjenkov of WHO, allowed the IFHGS to inform the organization of its aims and plans. Collaboration in the future between both organizations was agreed. Marcus Pembrey initiated contacts in the East, which should lead to the creation of additional continental societies there. This is where my task will start and a first meeting in Thailand is planned for October 2002. Due to unforeseen events, Marcus nor I were able to attend the IFHGS board meeting in San Diego, which delayed somewhat the smooth transition of power, but this has since been remediated. The IFHGS board will meet in Strasbourg.

M Pembrey

European Journal of Human Genetics -

Gert-Jan van Ommen (Editor in Chief)

The European Journal has been going steady for most of the past year. In the spring of 2002 a temporary fall of submissions suggested that most clinical geneticists were too busy with the ever-expanding opportunities to provide services, or applying for money in the Genomics programmes emerging in many countries, to come around to writing papers. However, the rate of submissions has picked up after the summer. Indeed, the year 2002 started with a bang, with, thus far, a steady flow of over 30 manuscripts per month.

In the year 2001, around 1070 pages have been published, with an acceptation rate of manuscripts of somewhat under 50%. The citation index of the EJHG shows a small but steady increase and in 2000 was 3.18

An additional reason for submitting to the EJHG lies in the steep decline of publication delay in the middle of 2001, down now to a publication date, on average, of 11-12 weeks after acceptation.

'Genetics in Practice' - As of January 2002, a new series has been started up, called 'Genetics in Practice', in which experts have been asked to review all current, practical aspects of a given disorder in a few pages, typically providing a map of the gene concerned, the mutations found with genotype-phenotype relationship if any, and, if possible, a clinical picture. As with all fledgling series in journals, we still only have a modest supply, so the Editors would enthusiastically welcome additional contributions to this series from experts amongst our readership. The contributions are undergoing regular peer review, with more emphasis placed on further improvement than on selection. Any new developments in your field, or need to put a record straight(er), then this is your chance.

Finally, with the Publishing contract of the ESHG with Nature Publishing Group running out, the Publications Committee has been soliciting bids from various publishers during the past year. Next to NPG itself, three candidates have made it to the final consideration, with discussions ongoing between the Publications Committee and the ESHG Executive Board. A full report will be made to the ESHG membership meeting in Strasbourg.

The Publications Committee -

members JL Mandel, S Antonarakis, GJ Van Ommen, T Meitinger, P Farndon, JJ Cassiman.

The committee had two missions this year: (1) to supervise the policy aspects of the relations with our present publisher, the Nature Publishing group; (2) to select a new publisher for the Journal of the society, since our present contract with NPG ends in 2002. The issues pertaining to the journal were discussed with the representative of NPG at the Vienna meeting. The problems identified by the committee, such as increasing the circulation of the journal, delays in solving practical problems, the cost for the publication of supplements by committees of the society, were reviewed and suggestions made by the publisher to solve these problems were agreed upon. A review of the implementation of the promises will be undertaken at the Strasbourg meeting.

In addition, a call for proposals to publish the EJHG was sent to 7 different publishing houses: Karger, Wiley, Blackwell, Elsevier, Springer, Radcliffe and NPG. The proposals of the first 6 were reviewed and based on the outcome of this, a meeting was set up in Amsterdam on November 28, 2001 with representatives of Wiley, Blackwell, Springer and Elsevier. The four invited publishing houses gave a very convincing exposé, which left the committee with the difficult task of selecting the best candidate. After the meeting, the selection was further reduced to Elsevier, Blackwell and Springer. Based on the information received from the other publishers a series of conditions were drafted, which were then sent to NPG to allow them to formulate a really competitive proposal. Additional meetings were held by some members of the committee with individual candidates to obtain more detailed information on specific issues. The committee will propose different alternatives to the board and the membership in Strasbourg. A very difficult decision will have to be made based on this information.

The committee for EU policies,

members S Aymé, JL Mandel, U Kristoffersson, R Elles, S Antonarakis, JJ Cassiman.

The committee has not had formal meetings during the past year. This does not mean that no activities were deployed. First there was the Fiori rapport, which contained all the information obtained by the Temporary Committee on Human Genetics of the EU parliament. Different members of our committee presented papers at the hearings of the EU committee. While the report contained a series of excellent recommendations about the provision of Genetic Services in Europe, it also recommended the banning of all research on embryonic stemcells. Mainly for the latter reason, in collaboration with the PPPC, and based on the criticisms drafted by its chair S. Aymé, a pan-european action was undertaken to inform the members of parliament of all countries about our position and to convince them of the drawbacks of the approval of this report. As a result of the action, the Fiori report was rejected by parliament by a large majority. Since the report also contained many positive recommendations, efforts are now underway, in collaboration with MEPs from different parties, to define a strategy to revive the good parts of the Fiori report. A meeting with MEPs and members of our committee will take place in the coming weeks.

Education Committee -

Celia DeLozier Blanchet

As explained in our last Newsletter , the Board at last year's annual meeting created a permanent committee for educational issues. Célia DeLozier was asked to propose members and an agenda of activities to the Board for approval . The committee, which will be composed of individuals representing the medical, paramedical and laboratory aspects of genetics, and coming from various parts of Europe, will meet formally for the first time in Strasbourg.

One of the priorities of the committee, on which work has already begun, is to create a database (which might be available on-line through the ESHG site) of genetic training programs available in Europe. The chairmen of Human Genetics departments across Europe will shortly be receiving a questionnaire on current programs, and we hope to have preliminary information to report to the Membership in Strasbourg.

European Science Foundation (ESF)

The European Science Foundation (ESF) is funding a programme to facilitate interdisciplinary European collaboration in all aspects of functional genomics. The programme is supported by research councils and academies from 20 different European countries and operates workshops, training events and exchange grants.

The programme is currently accepting proposals for workshops and training courses to be held in 2003. For more information, see http://www.functionalgenomics.org.uk/sections/News/programme.htm.

Also available are a number of fellowships for scientists wishing to work in another European country for a period of up to 6 months. Exchange Grants are primarily intended for young scientists, however applications from more senior scientists with permanent positions will be accepted. More information can be found at http://www.functionalgenomics.org.uk/sections/activities/grants.htm.

Attendance at all workshops and training courses is free of any registration fee. For more information, please go to http://www.functionalgenomics.org.uk.

The following events are being run in 2002. If you are interested in attending any one, please look at the web site for details.

Events in 2002

Workshops

- Modelling of molecular networks - June 12-14th- Granada, Spain
- The future of biobanks: Practical and ethical issues - September 12-13th - Uppsala, Sweden
- Ontology for biology - November 7-8th - Heidelberg, Germany

Training courses

- Genomics: Structural, functional, comparative and evolutionary approaches - July 15-27th - Corsica, France *
- Molecular interactions: New frontiers for computational methods - July 20-25th - Verona, Italy
- APEX on DNA microarrays: Application in SNP analysis, mutation detection and DNA resequencing- August 26th - September 1st - Tartu, Estonia

ORPHAN MEDICINAL PRODUCTS

Report on the EMEA workshop with Health Professionals and Academia on Orphan Medicinal products held in London on 24 January 2002, where Ségolène Aymé was representing the ESHG

The EMEA is the European agency for the evaluation of medicinal products. It is a decentralised administration, not part of the Commission. It is a pan-European networking system.

In 2000, a new EU regulation (141/2000 article 3 b) on Orphan medicinal products was adopted. This regulation aims at boosting the development of therapies for rare diseases, most of them being genetic diseases. Companies developing orphan drugs get a market exclusivity of 10 years and reduction of registration fees. To be designated as orphan, a product should be targeted at treating a life threatening or chronically debilitating disease, affecting no more than 5 in 10,000 persons in Europe.

The EMEA committee in charge of the designation of orphan drugs is the COMP (Committee of Orphan Medicinal Products) which was the organizer of the workshop. It is composed of 21 members: 1 representative per EU country, plus 3 patients representatives and 3 EMEA delegates.

The COMP is a very open structure which works closely with all possible partners. It had already organized 2 similar workshops, one with the Industry and one with the patients organizations. This time the scientific and medical societies were invited to discuss issues of mutual interest.

Up to now the COMP has received 155 applications of which 88 received a positive COMP opinion, 5 a negative one, the others being withdrawn. Three products have already received a marketing authorization.

The first topic discussed was the role of scientists and health professionals in orphan medicinal product development. It was seen as essential in:

- basic and clinical research in the field of rare diseases
- drug development: development of methods to limit invasiveness, development of appropriate end-points, choice of the appropriate design, development of methods to facilitate the recruitment of patients, development of tools to monitor cohorts in phase IV studies.
- Protocol assistance: necessary tests and trials required for market approval, ensure adequate design to show efficacy and safety even with small numbers of patients, determine primary end points

Institutional funding is necessary to support these activities, especially for phase I and II studies.The second topic discussed was the identification of experts to assist the COMP in its tasks. All societies present agreed to respond to specific requests. The ESHG has confirmed that is fully open to collaborations.

EUROPEAN HUMAN GENETICS CONFERENCE - STRASBOURG 2002 - Jean-Louis Mandel

Welcome to Strasbourg

The ESHG 2002 Conference will soon take place in Strasbourg (May 25-28). Following on the huge success of the 10th International Congress organised by ESHG in Vienna last year, we can expect an excellent and stimulating meeting. For the first time, our Conference will be held in conjunction with the European Meeting on Psychosocial Aspects of Genetics (EMPAG), which will allow fruitful interactions on problems that are at the heart of human genetics practice. 1200 colleagues have already registered at the time of writing, a record attendance. This is paralleled by the record submission of 1284 abstracts (and this is not including the EMPAG participants and abstracts). The scientific program will thus allow sampling between 140 oral presentations, and more than 1000 posters (including EMPAG). This will span all aspects of human genetics, from the latest developments in disease gene identification and analysis of pathogenic mechanisms, to molecular karyotyping, cancer genetics, complex diseases, issues in genetic testing in newborns and minors, progresses in therapeutic approaches. The ever more complete and accurate human genome sequence, the rapidly progressing mouse genome sequencing, offers unprecedented prospects for the analysis of gene function in normal and diseased states. In a city that houses the European Parliament and many other european institutions, the increasing impact of such institutions on research and practice of human genetics will naturally be a topic of discussion. And the commercial exhibit will also be the largest ever at an ESHG Conference, offering a view on the latest technological developments and their applications.

To those readers of the newsletter who will attend the meeting, I wish you an exciting meeting, that will bring you novel ideas, useful informations for your research or clinical and laboratory practice, for the improved care of patients and their families, and profitable encounters with colleagues. I hope you will also have time to enjoy the artistic and architectural treasures of our beautiful city and of the picturesque surrounding region of Alsace, and to sample some of its gastronomic delights.

Genetics in Europe (5): Human Genetics in France

Research in Human Genetics

Research in Human Genetics is actively supported in France through main institutions like CNRS, INSERM, Universititesand Charities. It is organized through a National programme. In 1997 a National Sequencing Center (www.genoscope.cns.fr) was created. The main activities of the centre are divided into internal and external projects. The external projects are collaborations with laboratories in the public sector, or under contract, with those of the private sector. This center is located in a large campus, the Genopole of Evry (www.genopole.com) which includes in addition the National Genotyping Center (www.cng.fr) which is producing 6 million genotypes per year, the Genethon laboratories (www.genethon.fr), the national bioinformatics institute (www.infobiogen.fr) and many biotech companies dedicated to genetics, genomics and their associated disciplines (scientific instrumentation, nanotechnology, robotics, bioinformatics, etc). Its annual budget is 16.6 million Euro. The Genopole of Evry is part of a larger network including six other regional genopoles located in Lille, Lyon-Grenoble, Marseille, Montpellier, Strasbourg and Toulouse.

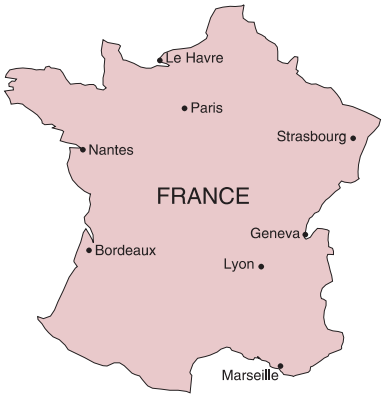
In 2002, the Ministry of Research created the Institute of Rare diseases which is going to coordinate the research funds targeted to rare disorders, mainly genetic disorders. (For more information: www.recherche.gouv.fr).

Health Care Delivery System

France has a population of about 60 million people. Primary medical care is provided both by general practitioners and independent specialists working from their own premises. General practitioners represent 54% of all physicians and do the follow-up of 40% of the pregnancies during the first two trimesters. The French health care system is an attempt to reconcile solidarity and liberalism by means of a combination of collective financing and a public and private sector for the delivery of health care. The government has control of the health insurance schemes, it determines the level of doctors' fees and the prices of pharmaceuticals and investigative procedures. This system is not very well adapted to quick changes such as the sudden availability of new diagnostic tools which may stay in the research domain for quite a long period before the regulatory authorities consider them for financial support.

Services in Genetics

There are 110 general genetic counselling clinics run by 135 geneticists. In addition there also more specialized counselling clinics in the field of cytogenetics (45), cardiology (2), ophthalmology (5), deafness (4), oncology (45) and dermatology (10) for instance. Some clinics are organized to manage patients' diagnosis and care and also provide counselling at the same time.



There are 57 different types of such clinics in France. There are 59 dysmorphology clinics. Regional genetic centres, including clinical, chromosomal and molecular services, were not originally individualized but are now getting organized in teaching hospitals.

Activities in biochemical genetics and molecular genetics were developed mainly by research teams as in most countries. Many tests are still not recognized as routine investigations reimbursed by the national health care insurance. They are financed by the global budget of the institutions hosting the activity which is not adapted to an activity serving a very large community, most often national, and sometimes even international. Some laboratories have difficulties offering their expertise to the community as their budget is limited to a regional activity. This is currently changing as specific financing from the Ministry of Health is in progress for some subgroups of diseases. Currently financial support is foreseen for cystic fibrosis, neurogenetics and oncogenetics.

The total number of laboratories performing molecular biology tests for genetic conditions is 53. There are 80 cytogenetics laboratories. There are also 45 biochemistry laboratories performing tests for genetic diseases. A test is available for more than 500 different diseases. For some, a large number of laboratories offer testing. For instance 43 laboratories provide tests for cystic fibrosis, 17 for Duchenne muscular dystrophy and 15 for familial breast cancer. The directory of services is accessible at the address www.orpha.net

Up to now, the development of genetic services has not been planned at a central level. It is only regulated. The recent creation of a National Advisory Board for Clinical Genetics which is advisory to the Minister of Health, may change this situation.

Accreditation/authorization of laboratories

Since 1988, laboratories have to be authorized by the Ministry of Health to perform any prenatal diagnostic test. The non observance of the regime of authorization is punished by jail since 1995. Labs have to apply separately for each type of activity: cytogenetics, molecular biology, biochemistry, serum screening. They are authorized for five years and have to produce an annual report on their activity. The advisory committee to the Minister for the authorizations is composed of qualified professionals, of representatives of professional organizations, of lay associations and

different bodies like the National Advisory Ethics Committee and the INSERM, and of representatives of the ministries of Health and Justice. A similar organization was set-up in 2001 for pre-symptomatic testing. The regime of authorization is not an accreditation system. The laboratories have to fulfil the criteria, which are defined in scheme law. There is no quality control attached to this authorization process. A quality control is going to be organized and financed for a few common tests.

Training for professionals in medical genetics

Medical Genetics was recognized as a specialty in 1995. Before, Genetics was considered as a sub-specialty since 1983 but a University Professorship in Medical Genetics was created as early as 1981. At the request of the National Board of Physicians (Conseil National de l'Ordre de Médecins) a competency certificate in medical genetics was created by law on February 10, 1981. Review boards were only legally established on July 30, 1990 and physicians were able to get qualified in medical genetics as a sub-specialty by the year 1990 for the first time.

Specialist status in Medical Genetics is acquired after a four-year training, which includes practical training (running residency of six month in clinical genetics, cytogenetics, molecular biology and paediatrics) and formal lectures.

Genetics is taught to undergraduate medical students, sometimes by instructors from the Science Faculty, in the first year (approximately 30 hours). Masters programs in cytogenetics or in human genetics were the only degrees available until 1988. From legislation passed on April 29, 1988, two sub-specialty diplomas were created: a diploma in molecular biology, and a diploma in human cytogenetics. The latt er, available to those already specialists in medical biology, gynaecology-obstetrics, haematology, internal medicine, oncology or paediatrics.

There is no training in genetics for nurses, mid-wives and psychologists and the profession of genetic counsellors does not exist. This is due to the opposition of the medical geneticists to the principle of having non-MDs advising patients in genetics clinics. In some centres, psychologists or psychoanalysts do work in collaboration with geneticists but do not provide genetic counselling per se.

Professional organisations

Human genetics is organised through several societies: « Société Française de Génétique », « Société Française de Génétique Humaine », « Association des Cytogénéticiens de Langue Française », « Club de Conseil Génétique de Langue Française », « Asociation Nationale des Praticiens de Génétique Moléculaire », "Collège National des Enseignants et Praticiens de Génétique Médicale". For the first time in 2002, they have organized a common scientific meeting. Up to now only concurrent meetings took place.

Quality assessment and quality control

No professional guidelines have been produced about teaching, training, practicing and quality control by the geneticists themselves. There is no quality-assessment network in cytogenetics, nor in molecular biology or in genetics biochemistry. The regulation of practices is exclusively done through legislation.

Legislation

A « Bioethics » law was passed on July 29, 1994. It is regulating several aspects of the genetics services. Since May 1995, all the laboratories involved in prenatal diagnosis have to apply to the Ministry of Health to be authorized for their specific activity. The authorization is given for 5 years and an activity report is due every year. This should contribute to the improvement of the services. The same authorization procedure applies to pre-implantation diagnosis and pre-symptomatic testing since 2001.

The law states that genetic study of a person's characteristics may only be undertaken for medical or scientific research purposes. Before such a study is undertaken the person's consent must be secured. Strict penalties are provided if consent has not been obtained or if the genetic study is carried out for non medical or non scientific purposes. The restriction of genetic testing for medical or scientific purposes has been reaffirmed in the law. The decrees delineate 5 conditions for prescribing and implementing genetic testing for medical purposes: 1) Condition of prescription; 2) Condition of approval from appropriate authorities both for clinicians and laboratories; 3) Conditions of reporting results; 4) Conditions of medical record protection; and 5) Approval from the National Consultative Commission created for this purpose. Physicians responsible for this genetic analysis must be qualified in medical biology or biology pharmacology. Exceptionally, a senior scientist (non-MD) may be responsible for these genetic analyses only if he/she is experienced in cytogenetics or molecular biology. A consultative Commission must be asked to rule on the necessity of such procedures and on their implementation.

Preimplantation diagnosis is only allowed in certain circumstances and can only be performed by centers which are approved for this activity..

Patients organizations

Many patient organisations exist in France. The largest one is l'Association Française contre les Myopathies (A.F.M) which supports scientific research and diffuses information to patients and physicians. In most of these associations medical geneticists are board members.

Most of the patients support groups are organized through an umbrella organization, the "Alliance Maladies rares" which is running a toll-free telephone line delivering information on genetic diseases and services to both patients and physicians. It is financially supported by the Ministry of Health and the A.F.M.

Ségolène Aymé and Didier Lacombe

EUROPEAN GENETICS: NATIONAL NEWS AND VIEWS

United Kingdom

Abstracts from a Speech by Rt Hon Alan Milburn MP, Secretary of State for Health at an international conference Genetics and Health - a Decade of Opportunity 16 January 2002

In addition to a £30 million package of new investment in NHS genetic services announced in April 2001, the Secretary of State for Health announced the establishment of two new National Genetics Reference Laboratories in Manchester and Salisbury, as part of the existing Genetic Centres in the North Western and Wessex regions. The national reference labs will help the NHS to keep abreast of scientific and technological discoveries in genetics and develop new and improved genetic testing. They will explore how better ways of working in laboratories such as automation or the more effective use of junior staff can reduce reporting times and increase cost effectiveness. And they will train NHS staff in the application of new genetic tests and technologies; and support the national NHS genetic testing network.

He also announced the establishment of six Genetics Knowledge Parks with funding from the Department for Trade and Industry and the Department of Health. They will consist of consortia of institutions based in Newcastle, London, Oxford, Cambridge, the North West and Wales. The Knowledge Parks will bring together clinicians, scientists, academics and industrial researchers seeking to improve the diagnosis, treatment and counselling of patients, and genetic technologies. The Genetics Knowledge Park network will have a role in improving public engagement and education about medical genetics.

Calendar of National and International Meetings

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

MAY 2002

Impact of Genomics on Medicine (6th annual)
13-14 May, 2002, Munich, GERMANY
www.beyondgenome.com

Pharmacogenomics/Pharmacoproteomics Europe
15-16 May 2002, Munich, GERMANY
www.healthtech.com

European Genetics Conference 2002
European Society of Human Genetics:
25-28 May 2002, Strasbourg, FRANCE
www.eshg.org or eshg@medacad.org

JUNE 2002

Beyond Genome 2002—Cambridge Healthtech Institute
2-7 June 2002, San Diego, California USA
www.beyondgenome.com

AUGUST 2002

Clinical Biochemistry and Molecular Medicine 2002
XXVIII Nordic Congress in Clinical Chemistry
August 10-13, 2002, Reykjavik, ICELAND
www.iii.is or e-mail: mail@iii.is

XVI th World Congress of Edipemiology
International Epidemiological Association
August 18-22, 2002, Montreal, CANADA
www.iea2002.com

SEPTEMBER 2002

3rd International DNA Sampling Conference
5-8 September 2002, Montreal, CANADA
www.humgen.umontreal.ca or e-mail: conference ADN@droit.umontreal.ca

British Human Genetics Conference
23-25 September 2002; University of York, UNITED KINGDOM
www.bshg.org.uk

First Aegean "Biologie Prospective" Santorini Conference
25-28 September 2002; Nomikos Conference Center, Thira Santorini - GREECE

German Society of Human Genetics
13th Annual Meeting
29 September-2 October 2002, Leipzig, GERMANY
www.SFK-Hamburg.de or e-mail: info@SFK-Hamburg.de

II World Conference on Bioethics
International Society of Bioethics
30 September-4 October, 2002, Gijon, SPAIN
www.sibi.org

OCTOBER 2002

Clinical complications in pregnancy
International Federation of Placenta Associations 8th Meeting
6-10 October 2002
www.ifpa8.com

4th course Genetic Counselling in Practice
European Genetics Foundation
6-11 October, 2002, Bertinoro, ITALY
www.eurogene.org

AFC Cytométrie 2002
8-11 October, 2002, Bordeaux, FRANCE
e-mail: afc2002@alphavisa.com

XIIth World Congress on Psychiatric Genetics
8-12 October 2002, Brussels, BELGIUM
www.wcpg2002.be

American Society Human Genetics 52nd Annual Meeting
15-19 October, 2002, Baltimore, MD, USA
American Society Human Genetics: www.ashg.org/

4th HUGO Pacific Meeting in 5th Asia-Pacific
Conference on Human Genetics
27-30 October 2002, Pattaya, Chonburi, THAILAND
www.mu-st.net/hugothai/

Symposium on Genetics in Health and Disease
Saudi School of Molecular Genetics
27-29 October and 2-4 November, 2002, Riyadh, SAUDI ARABIA
ichr@erc-iran.com or mohsen@ksu.edu.sa