ESHG

European Society of Human Genetics

April 2003 / Newsletter No. 8

The Society's administrative office:
European Society of Human Genetics, Clinical Genetics Unit, Birmingham Women's Hospital,
Edgbaston, BIRMINGHAM B15 2TG, United Kingdom
Tel/Fax: +44 121 623 6830
Website: http://www.eshg.org
Email: eshg@eshg.org

This issue contains the Agenda and Reports for the Annual Membership Meeting, which will take place on Monday, 5 May 2003 at 19.15 in Hall 9 at the International Convention Centre, Birmingham, United Kingdom.

Please bring it with you

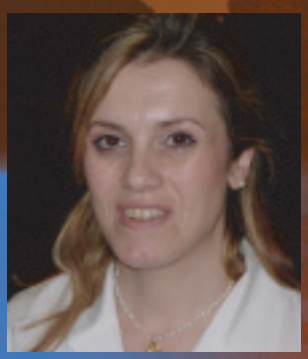
MEDICAL GENETICS IN GREECE

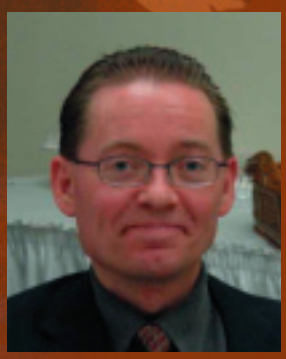
History

The first genetics units appeared in Athens in the early 1960s in the "Aghia Sophia" Children's Hospital and the "Alexandra" Maternity Hospital. Cytogenetic laboratories were the first to offer their services. In 1965, the Institute of Child Health in Athens started the investigation of aminoacidopathies and newborn screening started in 1973 initially for PKU and later for hypothyroidism and G6PD deficiency. This programme is well functioning, is centralised to this Institute and is based on the voluntary participation of all national and private maternity units. Prenatal diagnosis of chromosomal disorders by amniocentesis was initiated by the First Department of Pediatrics of the University of Athens and the "Alexandra" Maternity Hospital in 1976, followed by prenatal diagnosis of hemoglobinopathies by the First Departments of Internal Medicine and Obstetrics and Gynecology of the University of Athens. The new abortion law (1977) offered an excellent opportunity for the development of prenatal diagnosis centres. The Ministry of Health supported the establishment of prenatal diagnosis laboratories by providing funds for the purchase of necessary equipment and hiring of personnel. Chorionic villus sampling for prenatal diagnosis has been performed since 1983. The major developments in molecular genetics in the 1980s and 1990s were applied also in Greece, initially mostly by the same centres, offering diagnosis, carrier detection and prenatal diagnosis of several monogenic diseases.

Health Care Delivery System

Greece has a population of 10.450.000 and there are about 40 doctors and 49 hospital beds per 10.000 people. Since 1985 Greece has got a National Health System under which patient care is covered, depending on the type of care, entirely or at minimal cost by health insurance. Despite this, many individuals choose to have also private insurance, which allows them a wider use of medical services in Greece, also in private hospitals and clinics. Coverage for overseas medical care or diagnosis has to be approved by special committees. Almost 99% of the population have insurance coverage. Private hospitals exist throughout the country, with quality varying from average to excellent.


Dr Lina Florentin-Arar


Michael B Petersen

Medical Genetics in Greece continued

Genetic Services

A recent survey has shown that there are at least 24 public institutions and at least 5 private laboratories offering a variety of genetic services in Greece. About 20 of them are offering cytogenetic services (some of them including molecular cytogenetic services) and 14 of them are offering molecular genetic services, i.e. for hemoglobinopathies, hemophilias, cystic fibrosis, neuromuscular disorders, mental retardation syndromes, mitochondrial disorders, cancer genetics, deafness etc. There are official referral centres for hemoglobinopathies with a well organized network of public centres across the country, offering patient treatment, carrier screening and prenatal diagnosis. Biochemical genetic services are mainly offered in one institute. Overall the following are covered at the level of metabolite analysis, enzyme assays and DNA analysis: lysosomal storage diseases, peroxisomal disorders, aminoacidopathies etc. Several well-trained geneticists work in Greece and offer important services in clinical genetics, cytogenetics, molecular genetics, biochemical genetics, as well as genetic counselling. However, the country lacks a sufficient number of formal genetics centres and the number of physicians in full-time clinical genetics is extremely limited. At the same time there is a large number of very well trained clinical scientists in genetics who cannot be absorbed despite the growing need for genetic services. The departments are usually understaffed and underpaid because of the lack of budgets for trained personnel. There are no registries concerning genetic disorders, hereditary cancer, cytogenetic abnormalities, nor congenital malformations. Contacts and collaborations with many foreign centres allow continuity and support for services not fully offered locally. Despite the difficulties, for almost 30 years genetic departments have maintained high standards and have contributed to the prevention of birth of babies with chromosomal abnormalities and other genetic disorders.

Authorisation/Quality Control

The development of genetic services has not yet been sufficiently planned at a central level and thus there is no official authorisation of laboratories to perform diagnostic genetic tests. There are no professional guidelines concerning quality assessment of genetic services and thus no accreditation. However, gradually many molecular genetic laboratories have joined several quality assessment schemes for genetic disorders organised by EMQN. At least one cytogenetics laboratory has joined the UK NEQAS in Clinical Cytogenetics.

Professional Organisations

The Hellenic Association of Medical Geneticists (Syndesmos Iatrikon Genetiston Elladas or SIGE) was founded in 1982 and is the only active professional organisation. SIGE counts about 180 members, half of them physicians, mostly pediatricians, neurologists and dentists trained in genetics and those involved in hematologic diseases (hemoglobinopathies, hemophilias), and half of them laboratory geneticists (cytogeneticists, molecular geneticists, and biochemical geneticists). The official scientific organ of SIGE is the Hellenic Journal of Human Genetics, a biannual journal in Greek language, which so far has been issued 4 times and at the moment has stopped due to financial difficulties. In collaboration with the Aristotle University of Thessaloniki, SIGE organised the 1st Balkan Meeting on Human Genetics in Thessaloniki in 1994 and proposed the foundation of the Balkan Association of Human Genetics. Greece has hosted several international conferences and seminars related to medical genetics.

Speciality

After many years of efforts by the Hellenic Association of Medical Geneticists, a special committee in Genetics authorized by the Ministry of Health reached a unanimous decision and issued a statement with which they propose the following: an interscientific speciality with two different directions one for clinical geneticists (includes those with a degree in medicine or dentistry) and one for laboratory geneticists (includes also those with a degree in biology, biochemistry and pharmacy) and a subspecialization in genetic counselling which includes also nurses, psychologists and social workers. This proposal is also supported by the Ministry of Health and is at the moment under consideration by the Greek Central Board of Health.

Education

Many personnel are well educated because they support themselves to further education or because they receive scholarships from the State Scholarship Foundation, National Health System, Onassis Foundation, Bodosaki Foundation, Fulbright Foundation, or the British Council, allowing access to excellent training at the best Greek and foreign centres. There are no programs for continuous education at the moment but most of the geneticists (clinical and laboratory) are trying very hard to educate themselves by attending as many conferences as possible. During the recent years a few universities are offering graduate programs leading to a Master degree in biomedicine.

Research

Research in human genetics in Greece is usually supported by funds from the General Secretariat for Research and Technology, the Central Health Council, and from the E.U. and is performed in the major University Departments of the Schools of Basic Sciences and Medicine in the Universities of Athens, Thessaloniki, Patra, Ioannina, Alexandroupoli, Larisa, and Crete. Also in the National Foundation for Research, National Center for Scientific Research "Dimokritos", Institute of Child Health, Institute of Technology and Research, and in at least 4 private institutions that join into research projects together with public institutes. Funding for research in general in Greece and especially for basic research is not enough to sustain and promote major projects, so most of the projects that are financed are those with direct applicability to patients (applied research). Limited funding for both clinical and laboratory work is an important inhibitor, and financial difficulties are a constant barrier to obtaining new equipment and promoting original research. Nonetheless, either through public or private grants, some laboratories manage to be well equipped.

Legislation

An abortion law issued in 1977 allowed termination of pregnancy for medical reasons up to 24th week. In 1993 a decision was taken by the Central Board of Health regarding the formal structure and organisation as well as the number of Genetics Centres in Greece. A law concerning IVF is underway at the moment.

Patients Organisations

There are several patients organisations, which include an MDA organisation for neuromuscular disorders, organisations for the handicaps, for children that suffer from pediatric cancers, Down syndrome etc. All these organisations try to raise money in order to support and counsel patients and families in the first place and to support research in the second place.

Lina Florentin-Arar and Michael B. Petersen

Annual Membership Meeting 2003

Monday, 5 May 2003 at 19:15 in Hall 9 at the International Convention Centre, Birmingham, United Kingdom

AGENDA

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

1. Activity of the Society 2002-2003
2. Financial Report of the Society 2002
3. Discharge of the Board Members for the year 2002-2003

Opening by the new President of the Society, Prof Veronica van Heyningen

4. Results of election for President-Elect
5. Results of election for Board Members
6. Membership fees 2004
7. Site of future European Human Genetics Conferences
8. Budget proposal 2004
9. Major policy questions proposed by Board
10. Review of Society Statutes

NOTE: Minutes of Membership Meeting held on Monday, 27th May 2002 at 18.15, Strasbourg will be available at the next Membership Meeting.

Activity Repor

(Pending approv

Secretary General's Report

Prof Peter Farndon

During the year we said "Good Bye" and "Thank You" to Celia Delozier-Blanchet who ended her term of office as Deputy Secretary-General. Along with the Society's thanks, I would also like to extend my personal thanks to Celia who worked so hard behind the scenes on behalf of the Society. I am very pleased to say that Celia has agreed to be the Chairman of our Education Committee. We are pleased to welcome to the Executive team, Prof Lisbeth Tranebjaerg as Deputy Secretary-General.

We also said "Good Bye" to Ruth Cole who was our part-time administrator and welcomed Patricia Wright as the Society's part-time administrator based in Birmingham, United Kingdom.

The attached reports make very interesting reading and you will see that the society is continuing to initiate new developments. The number of members is continuing to grow, particularly with members who are taking out subscriptions to access online our journal, the European Journal of Human Genetics. The European Society of Human Genetics has played a major role in encouraging new members to join the International Federation of Human Genetics Societies; there have been exciting developments in our Journal with papers featured on the Nature web site and prizes for the top cited papers. Amongst its activities, our Committee for European Affairs is actively seeking recognition of medical genetics as a European-wide medical specialty.

At last year's Annual General Meeting we had intended to discuss new statutes for the Society but these were held in abeyance because of proposed changes to the Society's administration, in particular, for the future publishing of our journal. We will now need to return to the review of the Society's statutes and the future organisation of the European Human Genetics conferences.

I am responsible as Secretary-General for the Society's finances and administration. Many people ask me whether it is possible to reduce the costs of our annual conferences. I can assure everyone that the costs are continually reviewed. Because of the success of the conferences, numbers attending are now too large for the accommodation provided by universities. We, therefore, have no choice but to visit commercial conference centres. The cost of the centre is a fixed cost irrespective of the number of delegates attending. The numbers attending our conferences are too large for university accommodation but not large enough to reduce the cost per person of the fixed conference centre fees. If anyone has any alternative suggestions for mounting our conferences, we would be very pleased to receive them.

Prof Peter Farndon

Professional and Public Policy Committee

The latest report from this committee was published in Newsletter Number 7 in December 2002.

Chair: Dr Ségolène Aymé

rts 2002 - 2003

val by the Board)

Scientific Programme Committe

Prof Andrew Read

The Strasbourg meeting will be a hard act to follow, but we have been working hard to produce an attractive programme for the Birmingham meeting. The outlines were agreed at a weekend meeting of SPC members in Birmingham in July. Most of the invited speakers were arranged over the next two or three months, and progress was reviewed at a meeting (convened by Han Brunner as I was not there) at the ASHG conference in Baltimore. Our main difficulty has been with stem cells and RNAi – the leading personalities in these areas are no doubt deluged with requests to speak at meetings, and they probably don't see themselves primarily as human geneticists, so that we don't come at the top of their priority lists. It is frustrating that we have such difficulty in getting these cutting-edge topics properly represented at our conference. Apart from this problem, I am well pleased with the programme of invited talks.

Just over 1,000 abstracts were received by the January 22nd 2003 extended deadline. This is a small decrease on the Strasbourg number, but only small once the EMPAG abstracts are removed from the Strasbourg total. Submitted abstracts were sent to SPC members for review soon after the deadline, and we met in Vienna over the weekend of 8th-9th February to make the necessary decisions. Thanks to Jerome del Picchia's excellent organisation and a good-natured willingness of all the SPC members to work hard and flexibly, the process seemed to run very smoothly. Only 54 of the 1000+ abstracts could be selected for oral presentation, inevitably disappointing the many hundreds of submitters requesting this form of presentation. I do sometimes wonder why so many people see posters as second best – after all, surely most of us feel better informed when we have read something rather than seen it on TV. For the selection process, it is time-consuming but relatively easy to make up a short-list, based on review of each abstract by at least 3 SPC members; the real work is in trying to devise a coherent themed programme for each concurrent session, with something for everybody in the overall programme, while minimising the inevitable conflicts between parallel sessions. Thus although the primary criterion for oral presentation is scientific quality, other considerations do enter into the decisions.

This year all the conference fellowships (44 in all) are being awarded to participants from Central and Eastern Europe, with no "West" fellowships. Whatever we do, we inevitably have to disappoint the majority of applicants. We try to ensure that at least one person from each eligible country receives a fellowship, hoping that this will be the most efficient way of developing relationships and encouraging scientific progress in countries where it is difficult to do good work.

We welcomed three new members this year, Cornelia van Duijn, Andres Metspalu and Andre Reis, and we said goodbye to Marcella Devoto, Veronica van Heyningen and Jacek Zaremba. All three of the retiring members have made great contributions to the ESHG through their work, and on behalf of the whole SPC and Board, I thank them very much for their work. This is my last year as chair of the SPC, and after the Birmingham meeting I will be leaving it in the safe hands of Han Brunner, who has been acting as deputy chair this year. I wish him luck.

Chair: Prof Andrew Read

Education Committee

The latest report from this committee was published in Newsletter Number 7 in December 2002.

Chair: Dr Celia Delozier-Blanchet

Activity Reports 2002 - 2003 continued

Report from the Committee on European Union Affairs

Members: JJ Cassiman, chair; JL Mandel vice chair; S Antonarakis; S Aymé; R Elles; U Kristoffersson; G Utermann.

The committee had set three major objectives for the past year:

- to actively pursue the recognition of medical genetics as a European medical specialty.
- to revive the sections on Human Genetics of the Fiori report from the ad hoc committee of the European parliament and to bring these again before the EU parliament for approval.
- to continue the close relationship with the European Platform for Patient Organisations, Science and Industry (EPPOSI) of which S Aymé and JJ Cassiman are board members.

For the first objective, the European Union of Medical Specialists was approached. Based on the information we provided and the fact that most EU countries have a speciality in Clinical or Medical Genetics, the secretary general of the Union, Dr Maillet, agreed to propose to the board of the EUMS the creation of a commission for clinical genetics. This commission would be equal to an official recognition of the speciality and would make further steps towards full recognition at the EU level almost automatic. We are awaiting their decision.

For the second objective, limited progress has been made, except that the awareness in the parliament and the EU commission about genetics has drastically increased. The commission funded a special survey on Genetic Testing in Europe, the ESTO project, and set up a committee on Ethical and legal issues of Genetic testing, the Strata group. Members of the ESHG are represented in those groups. The EU parliament produced a provisional recommendation to the commission about the need to regulate genetic testing. The council of Europe produced a 'Working document on the applications of genetics for health purposes'. This document is under review in the committee.

EPPOSI was recognised as an International Foundation and is supporting the initiative of the European Society of Human Genetics' Professional and Public Policy Committee to jointly organise consensus meetings on the topics proposed earlier.

The International Federation of Human Genetics Societies

Prof J J Cassiman

The membership of the executive committee of the IFHGS has been extended to representatives of the 5 continental societies and includes now: representatives of the European Society of Human Genetics: Jean-Jacques Cassiman and GertJan Van Ommen; the representatives of the American Society of Human Genetics: David Valle, Judith Allanson and Elaine Strass; the representatives of the Human Genetics Society of Australasia: Agnes Bankier and Eric Haan; the representative of the Latin American Network of Human Genetics Societies: Roberto Giugliani; and the representative of the East Asian Union of Human Genetic Societies: Takehiko Sasazuki.
In addition, an application is expected from the Asia-Pacific Society of Human Genetics, which discussed its affiliation with me at the occasion of my visit to the 5th Asia-Pacific conference on Human Genetics in Pattaya, last October.

The situation in India and Pakistan has not yet become clear, but remains difficult notwithstanding the efforts of the local health professionals.

The list of National societies, who have become a member, is steadily increasing but an effort will be made to contact those who have not joined yet.

The opportunity has been created for corresponding members to publish information on their society in an international column of the ACMG journal 'Genetics in Medicine'.

The contacts with WHO are being reactivated in order for the IFHGS to become an official advisor on Genetic issues.

The IFHGS plans to hold a workshop on genetic services in developing countries at the July 2003 meeting of the AASHG in Melbourne, where also the next executive meeting is planned.

The IFHGS is looking forward to the next International congress of Human Genetics to be held in Brisbane, August 6 –10, 2006.

JJ Cassiman
Liaison Officer for the ESHG

Activity Reports 2002 - 2003 continued

Report from the Publications Committee

Members: S. Antonarakis, G.J. Van Ommen, T. Meintinger, J.J. Cassiman (chair), G. Utermann, P. Farndon, Observer, Nature Publishing.

After having received the approval of the Board to extend the contract with the Nature Publishing Group (NPG) for one year, in order to evaluate the performance of the newly appointed NPG staff, the committee informed the other candidate publishers of this decision. Some of these candidates kept in close contact during the past year.

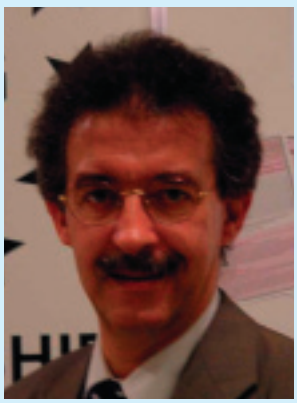
In the mean time numerous improvements were observed in the management of the journal by NPG. The editor-in-chief and the chair of the committee were informed regularly of the changes made, such as the highlighting on the NPG website on genetics of articles published in the European Journal of Human Genetics, the creation of a best cited paper of the year prize, and the implementation of the electronic submission and review system (EJP).

At a meeting in Leiden, in February, GJ Van Ommen, M.Osuch (Managing Editor for NPG) and JJ Cassiman reviewed the achievements and suggestions for improvement in preparation of the Editorial Board meeting in Birmingham.

The Board of the European Society of Human Genetics will have to decide in Birmingham, based on the evolution of the relations with NPG, whether the search for a new publisher is still on, or whether a new contract can be signed with NPG.

Further information on the journal can be found in the report of the editor-in-chief.

Editor's Report - European Journal of Human Genetics



Prof Gert-Jan van Ommen

- The major change of this year is that the European Journal of Human Genetics has gone over to Web-based submission and reviewing, using the E-Journal Press system. This has been preceeded by several months of preparation: entering our database and letters and adapting the system to our procedures (our thanks to Trevor Barton and Denise Taylor). So far all has gone amazingly well. Authors and reviewers (and editors) have reacted very positively on the increased speed and transparency.
- As announced in the Editorial of the October 2002 issue, the ESHG in collaboration with the Nature Publishing Group will be rewarding the top 3 cited papers of the previous year. Last year's and this year's top three citations (in 2001) will all be celebrated in Birmingham and receive a prize and a free one year membership (EJHG 2002: Tan Q et al: 10: 119-224; Adato A et al: 10: 339-350; Wutz K et al: 10: 449-456; Kronenberg F et al.: 10: 539-546; Gasparini P et al: 8: 19-23; Dobson-Stone et al: 8: 24-32; Schiller S et al: 8: 54-62) and free conference admission.
- Submissions are quite steady at around 30 manuscripts per month. Due to a changeover of typesetters, we had a temporary shrinkage of the first few issues in 2003. This will be made up in later issues.
- A very successful new service is that nearly every month one EJHG manuscript is highlighted on the Nature.com website. This has caused significant extra attention, culminating in the worldwide press coverage of the indications for multi-generational epigenetic inheritance described by Kaati G. et al. and commented on by Pembrey (vol. 10 no.11, 2002, pp 682-688 and pp 669)

Gert-Jan B van Ommen

GENETICS, MEDICINE AND HISTORY

FIRST INTERNATIONAL WORKSHOP

(Supported by Wellcome Trust)

Saturday 3rd May, International Convention Centre,Birmingham, UK
(In conjunction with the European Society of Human Genetics Annual Congress)

PROGRAMME

10.00 Coffee and registration
Professor Maj Hulten. Cytogenetic Milestones in Human Genetics. How to count to 46.
Professor John Edwards. 200 years of genetics in Birmingham
Professor Peter Harper. Julia Bell and the Treasury of Human Inheritance

Buffet Lunch

1.00 – 2.30 The Genetics and Medicine Historical Network
Introduction
Presentation of web-site
Further development
General discussion

The meeting is open free of charge to all interested, whether geneticists, historians or others. Anyone wishing to attend should contact Professor Peter Harper (HarperPS@Cardiff.ac.uk)

Future European Society of Human Genetics Conferences:-

Birmingham, England, May 3rd – 6th 2003
Munich, Germany, June 12th – 15th 2004

(European Meeting on Psychosocial Aspects of Genetics will be held in conjunction with the ESHG conference in Munich.)

Calendar of Events

To mark the 50th anniversary of Crick and Watson's famous paper on the structure of DNA, The Institute of Ideas in association with Pfizer is to host a weekend long Genes and Society Festival at the Battersea Arts Centre, London, UK 26 –27 April 2003 www.instituteofideas.com

Genetics of Obesity – Satellite Meeting at the 12th European Congress on Obesity, University of Kuopio, Finland, 27 – 28 May 2003. www.uku.fi/Genetics_of_Obesity

June 16 – 17 2003 2nd International Congress on Shwachman-Diamond Syndrome, Toronto, Canada www.cme.utoronto.ca/sds

679th Biochemical Society Meeting "Stress, Signalling and Control" 2 – 4 July 2003. www.biochemistry.org

European Conference on Rare Disorders and Disabilities, 16 – 17 October 2003 in Paris. www.rare-paris-2003.org

International Congress on Twin Studies 2004 will be held at the University of Southern Denmark, Odense, Denmark from 2 – 4 July 2004. www.icts2004.sdu.dk

European Skeletal Dysplasia Network newsletters available on www.ESDN.org

Contributions

We welcome contributions for future newsletters which should be sent to Patricia Wright email eshg@eshg.org for consideration.

Patricia Wright