European Society of Human Genetics

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Are we there yet?

No doubt this phrase will evoke the memory of a long car journey in many who read it. Maybe you were the bored child or the stressed parent. The answer was usually "soon" or "not yet", avoiding the obvious fact that we were still a very long way from the end of the journey. My year as President has coincided with the end of the five year funding for our genetic knowledge park, a government initiative to advance genetic health care. Perhaps it is just in my head, but I sense that many inside the field of genetics are a little anxious that our world continues to change this quickly while those outside are frustrated that it doesn't seem to be changing fast enough. Where, we are asked by various authorities, are the health gains promised by the Human Genome Project? When will genetic testing really start to make a difference? "Are we there yet?"

Executive Committee of the ESHG

The response tends to be to focus on the massive increase in our understanding of monogenic disorders and the rapid pace of development of laboratory diagnostics but, like the harassed parent, we know it's not enough. An expectation has developed that genetics will make a big difference in the care of people with common disorders like diabetes, coronary heart disease and dementia. Geneticists understand the challenge and must remain calm. Real advances are being made in our understanding of common diseases; CARD15, a factor in inflammatory bowel disease and fillagrin in eczema are important discoveries which have changed our view of underlying mechanisms and will probably change therapeutic research, but they are unlikely to lead to large scale population testing any time soon. We can see the challenges at clinical and laboratory level but the primary restraint in genetic testing for common disorders is the fact that having only half or even less of the story makes it difficult to offer a person information that will enable them to change the future of their health.

As our capacity to test gene variants expands rapidly and the pieces of the genetic jigsaw fall into place, we can foresee a time when a quick mouth swab could lead to a useful dialogue in relation to a wide range of chronic diseases, but not for a while. Some of you will have seen my photograph of Jamie, our 25 year old son standing by a horse chestnut tree behind our house. It reached about 12 metres in height last year when we were forced to curtail its upward growth. Jamie had planted the seed, or conker as we call it, when he was 5 years old. I saw him doing it and asked why. "I want to grow a tree so I can build a tree house", was the reply. The logic was perfect but his grasp of the time scale was weak.

We need to convince those in power that medical genetics is here to stay and will steadily grow in influence. Given the profound influence of the European Union it is essential that we, as a professional community, have a clear voice at the European level and it is here that the ESHG has a major role, one set to grow in coming years. We need to continue to exert influence in areas of policy such as gene patenting, work more closely with Eurogentest to increase the coordination and quality of laboratory services and press for the recognition of Clinical Genetics as a full

medical specialty. The recent decision of the Romanian government to downgrade their Genetics specialty is a major wakeup call.

We have made great strides as an academic society; the programme for Nice organised by Han Brunner and his team is of the highest quality and reflects to stature of the ESHG. But academic excellence alone is not enough. We need to expand our political influence. My successor, Pier Franco Pignatti, is pressing forward with his excellent work to build links with the national genetics societies across Europe. This is an essential development but adds to the ever growing burden of administration.

It became apparent in our discussions last year that we needed to invest more in our infrastructure. Thanks to Jantie de Roos who runs our commercial sponsorship and Gert Jan van Ommen at the Journal, coupled with the prudent management of our treasurer Andrew Reed, our finances are in very good order. We have decided, therefore, to appoint a part time executive officer to provide administrative support to our General Secretary Helena Kääriäinen and the rest of the Board. Impressed by the way the Vienna Medical Academy have run our meetings in recent years, the executive team opened successful negotiations to hire Jerome del Picchia to fill the role of Executive Officer. Many of you will know Jerome from his work at our conferences. I am delighted to say that the full Board have given an overwhelmingly positive response to this development which, I am confident, will enhance the impact of ESHG.

So next time you're asked, the answer is "no, we're not there yet, but we are well on our way"

John Burn
President ESHG

ESHG Secretary General's Report

Helena Kääriäinen
ESHG Secretary General

If some of you ESHG members feel confused about the organisation of ESHG, please visit our excellent website. There the organisation is nicely presented, including a brief history, the Statutes and names of Board members and officers. Whenever you get ideas on how to improve our Society, please contact one of us!

After having said this, I must confess that the work load of the ESHG officers and committees (especially Scientific Program Committee) has grown, at the same time as the work load in everybody's real job seems to be growing as well. This has led to a situation where the excellent ideas that many of us have simply never come true. For this reason, the ESHG Executive Board has developed a proposal to employ an Executive Officer for the Society. This will structure our work and enable the ESHG to achieve new goals. We will report about this new arrangement at the ESHG Membership meeting in Nice.

One of the most important tasks of the ESHG is to organize the scientific conference every year. This task has, in spite of everyone being overwhelmed with work, been carried through successfully every year. Again in 2007 in Nice, there will be a scientifically excellent program, combined with beautiful surroundings and the wonderful opportunity to meet colleagues from all parts of Europe and beyond.

During every ESHG Conference there is always the official meeting of the membership. This is an opportunity to join the discussion about the future of the ESHG. What do we really want our Society to aim at? In addition to the excellent conferences, should we be more active players in health and research politics in Europe? Should the Society give more concrete help to small national societies? Should this Newsletter get an even more important role among European geneticists? How should the website be developed to better serve the membership? On Sunday, June 17, 2007 at 19.00 at Nice Acropolis, Room Hermès, the General Assembly and Membership Meeting of the ESHG takes place. Let's meet there!

Helsinki April 25, 2007

Helena Kääriäinen
ESHG Secretary General

EJHG Report 2006

2006 has been a very good year for the EJHG. Its citation index increased significantly, from 2.74 up to 3.25, moving up the journal from position 51 up to 46 in the Genetics and Heredity category. The steady increase in institutional online subscriptions has continued (up to 153 from 132 in 2005), causing an increased readers' interest as judged from a 43% increase in online pageviews. Editorially, 2006 is the year in which we finally launched the Practical Genetics Series, under the editorship of Phil Beales, in collaboration with Orphanet. In addition, we published a much-acclaimed special issue on Psychiatric Genetics, co-edited by Associate Editor Tony Monaco and Guest Editor Jonathan Flint, which has received major interest in extra pageviews and downloads. The News and Commentary category continues to raise interest, as well as pageviews and downloads. Proposals from the membership for special issues of broad interest and N&C topics are welcome; please contact the editorial office at ejhg@lumc.nl.

The first half of 2006 saw a slight decrease in submissions, 512 against 557 in 2005. Possibly our authors are very aware of the movements of our citation index, but then the good news is that this has picked up again later in 2006, with the last quarter scoring 9% better than in 2005 again. In addition, we may have become clearer about our editorial policy, since our acceptation rate also increased from 31 to 35%. Notably in the field of association studies we have had to become stricter, requiring an independent validation study by other means of a second cohort. Processwise, the average time to final decision has decreased further, by 8 days from 36 to 28 days, but we still want to apologise for the occasional manuscript taking much longer due to factors often not really in our control, like the holiday/travel season and the incidental difficulty in finding enough reviewers.

In 2006, the Journal has again made a solid contribution to the revenue of the ESHG, which is partly used – as in the previous years – to publish 100 more pages than we could have done otherwise, and furthermore to help financing other ESHG activities like the PPPC meetings and reports as well as scholarships for students to, e.g., EGF activities. Finally, like in the previous years, we have been monitoring the short term citation success of our content, and the first or first junior author of the three top scoring papers published in 2005 and cited in 2006 and the first months of 2007, will receive a year's ESHG membership including EJHG online subscription, free access to our Nice meeting, while the no. 1 paper will receive an additional money prize of 500€ .

G.J.B. van Ommen,

Editor in Chief

International Federation of Human Genetics Societies

J.J. Cassiman Chair publications committee
http://www.ifhgs.org

Liaison officer for the ESHG, Jean-Jacques Cassiman

The Executive committee of the IFHGS met in Brisbane on August 6, 2006. People present: Jose Maria Cantu, president, Pornswan Wasant, APSHG; John MacMillan, John Christodoulou, HGSA; Yoichi Matsubara, Ho Suk Saw, Yusuke Nakamura, EAUHGS; Jean-Jacques Cassiman, ESHG; Roberto Giuliani, RELAGH; Judith Allanson, ASHG; Elaine Strass, executive secretary; and guests Judith Hall, Ysbrand Poortman, Alastair Kent, Dianne Petrie, Arnold Christiansen and Richard Cotton.

Future meetings

Plans are now underway for the 2011 – 12th ICHG in Montreal. Affiliated groups will be asked to participate in the planning process: the Canadian College of Medical Genetics (CCMG), the Canadian Association of Genetic Counsellors (CAGC), the National Society of Genetic Counselors (NSGC), the Genetic Alliance (GA), the International Genetic Alliance, (IGA), the Genetics Society of America (GSA) and the American College of Medical Genetics (ACMG) as co-hosts or sponsors. Details will be worked out.

Future members and associate members

The Executive voted to welcome the APSHG as its sixth full member society.

The **Asia Pacific Society of Human Genetics** (APSHG) has individual members, not groups. Singapore was accepted by the network of geneticists as

the home base for the society. The society has a meeting, a Web site, bylaws and an election of officers. The members include representatives from the following countries: Thailand, Philippines, Hong Kong and Singapore. China is not represented in this group or in the EAUHGS. The Chinese Association of Medical Genetics will be invited to join with the EAUHGS.

Proposed bylaws were provided by Rajkumar Ramesar of the **African Society of Human Genetics**. It was not clear which countries were represented and how many members were now active in the society. Strass will find out more about the situation and report back.

An application for membership from the **Canadian Association of Genetic** Counselors was discussed. As the CCMG is the primary member from Canada, and as the CAGC is a specialized group, they were granted affiliate member status.

Human variome project (hvp)
Richard Cotton visited the executive to report on the HVP. The goal of the project is to devise a database that would include all known human mutations, with a worldwide registry scheme to support it.
Two motions were carried by the executive:

"The Executive of the IFHGS endorsed the concept of the Human Variome Project leading to the integration of data bases and harmonization of nomenclature related to human variation to facilitate research, knowledge and applications related to human variation and health."

"The Executive of the IFHGS wishes to facilitate communication about the Human Variome Project to its members when information is provided. "

Medical genetics in corresponding member states
There is a need for the Federation to support geneticists in countries without medical genetics programs. The Spanish Society has no programs within the country. Cantu wrote a letter to the Queen and asked for preparedness in this area. Cantu will survey the membership to find out what other countries would benefit from this initiative.

International genetic alliance and the world alliance
The Global Awareness of Genetics campaign has been launched. This will involve getting the word out about genetics services for developing countries. Of utmost concern is alerting health professionals of the availability of newborn screening programs and methods to identify infants with birth defects. The message the group wants to give to Federation members is to contact regional groups of the IGA and invite them to annual scientific meetings dealing with human genetics research and medical genetics programs.
The Federation will add a link to the Web site that shows the agreement between the IGA and the Federation. In addition, the link will be sent as part of an e-mail message to all Federation member societies to encourage them to invite local IGA representatives to their annual meetings.

In connection with upcoming genetics meetings, the IGS needs to propose different levels of support to various societies. A cafeteria approach might add appeal when they ask for inclusion at the meeting. For example, a large meeting can support the costs of a large meeting room, a free exhibit booth, AV services, free badges and a catered lunch. A small meeting might only be able to offer a free exhibit booth and a few badges. But if the list were available it would focus the attention of the host society and encourage them to offer something of value.

Report of the Scientific Programme Committee 2006-2007


Han Brunner

The Scientific Programme Committee for 2006-2007 was composed of Han Brunner (chair), Thierry Frébourg, Paulo Gasparini, Peter Heutink, Juha Kere, Peter Lichter, Stanislas Lyonnet, Milan Macek, Jr. , Raquel Seruca, Andrew Wilkie, Brunhilde Wirth, Olaf Riess, Nicholas Levy, Eduardo Tizzano, Véronique Paquis (Local Host), and Batsheva Kerem. John Burn and Helena Kääriäinen participated as observers from the executive board.
The SPC met twice to organize the Nice 2007 ESHG conference: in Nice in June 2006 to decide on the plenary sessions and symposia, and in Vienna at the VMA officies in March 2007, to select the abstracts for oral presentations and posters.
The number of sessions from submitted abstracts continues to increase and has now reached 14. This

allows us to keep the number of abstracts selected for oral presentation at around 5% of the total number. Secondly, we decided to hold a session on the first day that presents particularly exciting new findings in a "What's new?" session from submitted abstracts.
For the final session on Tuesday 19 June, we were able to attract the Nobel Prize winner of 2004, Professor Aaaron Ciechanover this year to discuss "The ubiquitin proteolytic system and pathogenesis of human diseases". This will hopefully keep the scientific excitement up for the entire conference, right until the end. A similar session last year with Sydney Brenner was very successful.
As is usual, our second highlight of the final day of the conference will be the acceptance speech by our ESHG prize winner. This year, the ESHG prize 2007 will be awarded to Professor Andrea Ballabio (Naples) in recognition of his work which demonstrates how modern-day genetics can address questions regarding genetic causation through functional genomics and pathogenesis right up to therapy.
Another highlight will be the presentation of the ESHG 2007 Education Award to Tom Strachan and Andrew Read for their wonderful book Human Molecular Genetics.
After the Nice conference, the SPC will have to say goodbye to Stanislas Lyonnet and to local organizers Veronique Paquis as well as Nicholas Levy. We thank them for their work and their dedication to improving the meeting.

Activity Report of the PPPC - May 07

Ségolène Aymé

Composition of the PPPC
The PPPC is currently composed of Violetta Anastasiadou , Ségolène Aymé (Chair), Suzanne Braga, Martina Cornel (Vice Chair), Domenico Coviello, Gerry Evers-Kiebooms, Veronica van Heyningen, Shirley Hodgson, Helena Kääriäinen (secretary), Gyorgy Kosztolanyi, Ulf Kristoffersson (Deputy-chair), Christine Patch, Jorge Sequeiros, Lisbeth Tranebjaerg.

Activities
During the past year, the PPPC dedicated its activities
- to the preparation of a document on « Patenting and licensing in genetic testing: Ethical, Legal and Social Issues"
- to planning the next document to be produced.

The Committee met twice: at the last annual ESHG meeting in May 2006 and in Amsterdam, on 7 February 2007.

1- "Patenting and licensing in genetic testing: Ethical, Legal and Social Issues" Report

The PPPC was requested, at the 05 annual meeting, to look at the issue of gene patenting in relation with genetic testing, jointly with the Patenting and Licensing Committee. Two preparatory meetings took place in October 05 and in November 05. Sirpa Soini was appointed secretary of the two committees for this project. During the past year she drafted a background document. The methods used to produce this document were to systematically review the contemporary literature and articles on patenting and human genes in the major journals. Furthermore, the recent reports and reviews produced by the international organisations (OECD, Council of Europe, UNESCO) were studied. A first draft of the document was produced by the first author of the paper and sent to approximately 90 experts for review, of which half responded. The list of experts was put up by asking each of the PPPC and of the PLC members to nominate experts in their country: geneticists having been involved in the public debates around gene patenting, attorneys, technology transfer officers, academic lawyers, and experts of NGOs in the field, and of the European Patent Office (EPO). Their comments were incorporated to produce a second version which was sent to a smaller group of experts who were invited to attend a workshop in Leuven, held on November 2006. The participants were asked to give five personal statements or recommendations they consider as crucial to improve the patenting process in the field of genetic testing. These statements and recommendations were incorporated in the document, together with the product of the discussion during the workshop, to produce the third version. This third version is now open for discussion on the ESHG website. When finalised, this document will be published in the European Journal of Human Genetics.

2- Report in preparation: "Genetic susceptibility testing"
The topic of the next document and workshop will be "genetic susceptibility testing". All activities will be organised as a joined project with Eurogentest, Unit 3.
The goals of this project are to define ways to establish the clinical utility of susceptibility testing for multi-factorial diseases and to give decision-making tools to heath care planners. The role of appropri-

ate counselling services to be put in place will be reviewed. The project will be based on some tests currently used: Factor V Leiden, hemochromatosis, high and low risk cancer genes. An input of clinicians in other fields (internists, haematologists) is expected. The workshop will be organised jointly with IPTS in Sevilla in October 07.

Report of the ESHG-Education Committee

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Celia DeLozier (USA/Switzerland), cddelozier@comcast.net
Peter Farndon (Birmingham, UK), p.a.farndon@bham.ac.uk
Peter Goetz (Czech Republic), petr.goetz@fnmotol.cz
Shirley Hodgson (London, U.K.), shodgson@sgul.ac.uk
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Maria Soller (Lund, Sweden), Maria.Soller@med.lu.se

Dear ESHG members,

During the past winter, the Education Committee has focussed its activity on completing the draft document "Core competences in genetics for health professionals in Europe". As you may know, all members of the Committee are working in Unit 6 of the EuroGentest project to harmonise genetic testing in Europe, charged with addressing the professional and patient education issues connected with genetic testing. After the Porto meeting (September 2006) the document received input from experts from several European countries (see list of contributors) and is now open for consultation on the web site of our society (see news). The aim of the document is to describe and agree, by consensus, on a set of core competences that could apply to health professionals in Europe, whatever their national setting. This could provide an appropriate framework for establishing minimum standards of preparation for health care professionals in genetics across national boundaries. While core competences in genetics have been described for some health professionals in some countries, a set of competences that could be applied across Europe to the range of health professionals involved in provision of genetic healthcare does not exist.

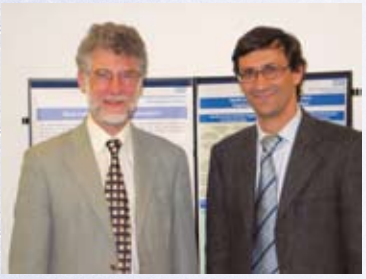
We would welcome your comments as to the relevance and applicability of this competence framework to your own country and profession, however extensive or brief. We will also have the chance to comment on it at the Education workshop at the ESHG meeting in Nice.

On 27 April 2007, I visited the NHS National Genetics Education and Development Centre in Birmingham (UK) (www.geneticseducation.nhs.uk), which is working with a range of groups throughout the UK to facilitate the integration of genetics education into all levels of education and training for NHS health professionals. The Centre is currently working with dieticians, medical professionals, nursing professionals and pharmacists. An educational competence framework for nursing professionals and learning outcomes for medical students, specialty trainees and GP trainees has been agreed. Additionally, the Centre, working with Skills for Health, has developed a set of nine competences in genetics for non-genetics healthcare professionals. This competence framework specifies which tasks need to be completed in a patient pathway but does not specify who should carry out the tasks. The Centre is now focusing on implementing the genetics competences and on developing resources to support the competence frameworks and learning outcomes in clinical practice. Evaluating the outcomes of the education programmes is a key component of the Centre's work. I believe that in the near future a joint initiative between the Centre in the UK and the ESHG could be very productive to establish an educational framework in Europe.

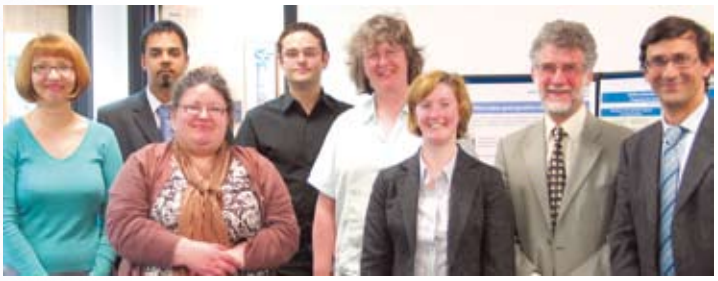
As usual all of you are invited to send us comments, ideas, updates from your country, including your willingness to collaborate with the Education Committee, to help us to set up priorities and to share experience or new tools available.

On behalf of Education Committee,

Domenico Coviello, MD, PhD



Prof. Peter Farndon, Director of NHS National Genetics Education and Development Centre in Birmingham (UK)
Dr. Domenico Coviello, Chairman of Education Committee of European Society of Human Genetics



Dr. Domenico Coviello and staff members of NHS National Genetics Education and Development Centre in Birmingham (UK)

EGAN
European Genetic Alliances' Network

It was in spring 1991 that some representatives of parent and patient organizations were invited to attend the annual meeting of the European Society of Human Genetics (ESHG) in Leuven.
With great interest they followed the presentations and highly valued content and environment.
A year later, at the occasion of the ESHG annual meeting in Helsingor, they founded the European Alliance of Genetic Support Groups (EAGS).
Ever since, this alliance has organized its annual meeting in conjunction with the annual congresses of the ESHG.
The genetic alliance and the ESHG share interests in research and in networks of genetic services meeting high standards regarding quality and ethics.
In 2000 in Amsterdam it was decided to rename and refocus the EAGS to: European Alliance for Genetic Services and the Innovation of Medicine. The focus was on genetics, genomics and biotechnology with a strong output in terms of projects and on a direct or indirect influence in the political, scientific and industrial arena. For these reasons relationships, collaborations and partnerships were established with scientific societies and industrial associations while contacts with officials of the various EU/DGs were maintained.
In Prague in 2005, the name was amended again: European Genetic Alliances' Network, EGAN.

In 2001 in Vienna at the annual ESHG- congress and the 2nd Congress of the International Federation of Human Genetic Societies (IFHGS) it was decided to found an International Genetic Alliance of parent and patient organisations (IGA). IGA meets bi-annually in conjunction with the World Life Sciences' Forum BioVision in Lyon.

EGAN is an alliance of national genetic alliances and European disease specific patient groups with a special interest in innovative medicine and in genetics, genomics and biotechnology.
Moreover, they are highly supportive for networks of genetic services.

EGAN works with a rather low profile. The Dutch and the British genetic interest groups, (having 165 and 63 member organisations, respectively) are the driving forces behind EGAN, followed by other more modest national networks in Austria, Poland and Spain. In other countries there are contacts or more vague networks.
The Central and East European Genetic Network

(CEEGN) is developing fast and represents groups in 10 countries. Various European disease specific organisations are members of EGAN.
Representation from south European countries is rather weak while the need for the activities of a genetic alliance is probably high.

EGAN serves to cater for the common aims of individuals and families with genetic disorders and to promote the provision of services which meet the needs of these individuals and families.
EGAN's main activities are advocacy via the issuing of position papers and statements on topics, representing the members in other European and international joint ventures and alliances, empowering its members, the collaboration with science and industry on issues of joint interest.
Position papers cover subjects like diagnosis, testing, screening, stem cells, reproductive choice, gene-, cell- and tissue therapy, paediatric medicines, bio banks, patenting and intellectual property and animal experimentation.

EGAN also initiated the publication of various books and organised expert meetings on subjects of their interest.
It was EGAN who took the initiative to found the European Platform for Patients' Organisations, Science and Industry. (EPPOSI)
EGAN closely works together with the European Federation of Good Clinical Practice (EFGCP) and shares offices with them in Brussels. With the European Federation of Biotechnology, a science oriented organisation, with EuropaBio, the association for bio industries, and with EFPIA the European Federation of Pharmaceutical Industries and Associations.
EGAN participates in a number of major EU projects such as GENED, EUROGENTEST, CONSERT (gene therapy) EUROGENGUIDE and CAPABILITY.
EUROGENGUIDE focuses on the development of genetic information for parents and patients.

In general: genetic disorders are very common, incurable, repetitive and often seriously burdening in physical, psycho social and economical respect.
Families involved in genetic/chronic/serious disease speak the same emotional language all over the world. This is why they find each other, understand each other, support each other.
Parents and patients getting confronted with disease basically have the same questions: where can we find the very best doctor, where can we get the best treatment.
In the second phase the questions are: what about the future, what about the implications for the family, could we have prevented it.
In the third phase it is realised that the healthcare system is a system for the care of the sick and not yet ready to face the new options given by genetics, genomics and biotechnology. It is realised that society is not yet ready for real healthcare focussing on prevention and minimising the burden of disease by timely testing, accurate diagnosis, genetic counselling, well balanced decision making, and early and focussed treatment.
These questions underpin the relevance of parents/patients organisations.


Ysbrand Poortman

Parent and patient organisations are - generally speaking - very well organised, they work together efficiently and increasingly get the credits they deserve (with the exception of financial governmental support). Although they deliver adequate services to healthcare, they are in no way rewarded on the level they should.
Most work is voluntary and dependent on motivation, experienced problems and envisaged solutions, on horizon and vision. In contrast with some other activities like Greenpeace and animal protection groups, the members of parent/patient organisations never chose for this activity for free, nor have the time, money and energy for it.

Parent/patient organisations have developed into a well coordinated, collaborative and efficient worldwide network of services and are structured on the basis of specific interests and priorities, on experienced problems and needs, own talents and capacities. These organisations are disease or subject specifically organised on the national, on the regional/continental or global level.
The leaders of these groups meet several times a year at various occasions such as in informal think-tank meetings and in EPPOSI- workshops.

A science writer recently interviewed 12 patient leaders all active in EGAN and published the reports in "It is MY life". (egan@egan.eu) These portraits show that patients are a serious partner in the dialogue with clinicians, politicians, researchers and industry. Patient organizations have great expertise about their diseases and play an important role in prevention, therapy development and disease management.

Ysbrand Poortman
Founder, Past President and Advisor EGAN, Vice President WAO (World Alliance of Organizations for Prevention and Treatment of Genetic & Congenital Conditions)

Subject specific patient organisations:

Rare diseases:
EURORDIS, European Organisation for Rare Diseases.
Focus: Advocacy and EU health policy making, information on rare diseases
Members: organisations involved in rare diseases
Paris, France

Public health and health advocacy
EPF, European Patient Forum
Founded as a reaction to a direct response to calls by the European Commission to have one pan-European patient body to address and consult on issues of interest to patients in the European healthcare debate.
Members: European umbrella organisations
Brussels, Belgium

Research and health policy related to prevention and treatment of genetic disease
EGAN, European Genetic Alliances' Network
Members: organisations with a specific interest in R&D, in innovative medicine and networks of genetic services.
Vredehofstraat 31
3761 HA Soestdijk
The Netherlands

Healthcare policies towards treatment & prevention of serious disease
EPPOSI; Patient organisations, science and industry
Exchange of information and discussion of policies in EU human healthcare.

European Disease Specific Organisations
See: www.patient-view.com/directories.htm

Relevant websites:
www.egan.eu
www.eurordis.org
www.eu-patient.eu
www.epposi.org

Orphanet: 10 Years of Service to the Rare Disease Community

Orphanet held a 1-day conference on 15 February to celebrate its 10th birthday, at which its chief collaborators, healthcare professionals and patient organizations presented their accounts, appreciations and comments on the creation of the European database, its evolution and its current place on the international internet scene. The day was punctuated by presentations from the major institutions which have supported Orphanet over the years: the French Health Ministry (DGS), the French National Institute for Health and Medical Research (Inserm) and the European Commission, represented by John Ryan from the Commission's Directorate of Public Health and Risk Assessment. All were present to retrace and relive the determining events between 1997 and 2007. Their enthusiastic eye witness accounts and presentations also took into account the difficulties related to the challenge of managing such a project. It is not so easy to fund the activity of over 30 professionals on soft money and to convince hundreds of professionals to collaborate.

Within the initial Working Group set up in 1996 to determine the creation of Orphanet, it was said that three exceptional ladies were to play a determining role in the Orphanet venture. The first is Simone Veil, former French Minister of Health and first woman President of the European Parliament, famous for her active role in European politics. The second, Annie Wolf of the Mission des Médicaments Orphelins, was just back from a fact-finding mission in the USA where at the time the US Orphan Drug Act had already created orphan drug legislation and where she had met key international figures Abbey Meyers, President of NORD and Marlene Haffner of the FDA. The third grande dame is, of course, the future Director of Orphanet herself, Ségolène Aymé, who had already been singled out for her expertise and energy as an experienced geneticist in the rare disease field. Together, these three women had a remarkable impact on events leading towards the creation of a totally innovatory European database of rare diseases.

Today Orphanet is widely regarded as a "model" in its field and is the world's first website on rare diseases and orphan drugs. It is visited daily by 20,000 users (300 000 hits/day) from roughly 160 countries. Half of them are regular users, while the others discover the site using a search engine. The website offers an encyclopaedia of rare diseases in six languages (English, French, German, Italian, Spanish, and Portuguese), which is written by experts and peer-reviewed. Lists of expert resources are attached to over 2,000 diseases. This includes lists of clinical laboratories, expert clinics, registries, research projects, clinical trials, and patient support groups collected in 22 countries. The data collection is in progress in another 13 countries. The data collection is done at country level. If you have not registered with Orphanet yet, please do so. The forms are accessible from the front page of the website.

Orphanet has established many strong partnerships. Together with EuroGenTest, information on clinical genetic tests will be soon complemented with information on quality assurance schemes of the labs. Together with Genatlas and GeneTests, Orphanet will soon be able to provide information on genes associated with the listed diseases, an attractive feature for those users who would like to query the database by gene. Most recently, Orphanet was invited by the WHO to help revise the International Classification of Diseases to take into account the need for specific codes for genetic conditions, even those that are very rare. Many more new features are expected on the Orphanet website in the coming year.

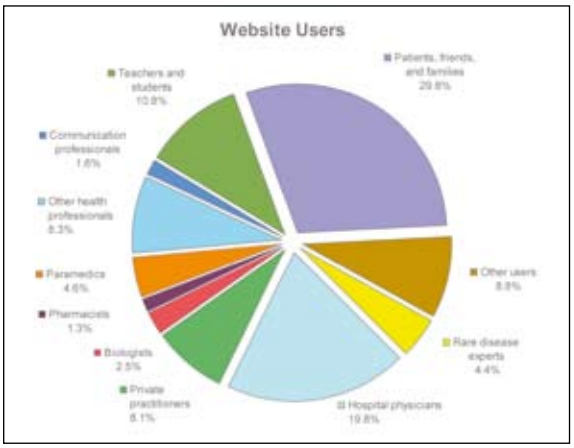


Figure 1. Orphanet User Characteristics

Fig 2 : Evolution of the number of accesses to the Orphanet website

EuroGentest – Year 3 Sees Major Progress

Eurogentest is a Network of Excellence funded by the EU to achieve its goal throughout the EU and associated states by harmonizing and improving standards. Now half way through its initial 5 years, the project is more than meeting expectations. At the recent AGM in Leuven, all 6 Units reported major progress in their allocated work packages. Groundbreaking actions ranged from oversubscribed accreditation workshops and evaluations of new technologies to surveys on patient rights and guidelines for patient information materials. Two countries with established health systems and the newer member states have embraced the project as a prime example of how a European project can deliver real results. Examples of other highlights are:

Quality underpins all activities

Unit 1 deals with quality management (accreditation, certification and licensing). Having charted the situation the Unit has been organizing a series of Expert Workshops on Laboratory Accreditation, which were oversubscribed. This work continues in 2007 with a **further two workshops and Round-table sessions** at the European Society of Human Genetics on specific, quality related, topics.

In **Molecular Genetics**, work has focussed on harmonizing EQA schemes. A draft document on best practice in EQA scheme provision and a Directory of Guidelines for Molecular Genetic Testing have been produced. In 2007 there is a new focus on developing a mechanism to **draft, review and update consensus guidelines.**

In **Cytogenetics**, having initiated the creation of the first European Cytogenetic labs register, a further breakthrough was made when the Unit demonstrated the feasibility and viability of creating On-line EQA assessment. To be called **CEQA, the second pilot will occur in 2007** featuring an EQA Management system available to participants with on line registration.

In **Biochemical Genetics** scoping best practice in Purine and Pyrimidine analysis and Lysosomal storage diseases and novel disorders has led to an interim report entitled Biochemical Genetic Testing in Europe: deficits and needs and EQA.
Furthermore, EuroGentest has also facilitated a directory of laboratories being put on the European Research Network for evaluation and improvement of screening, diagnosis and treatment of Inherited disorders of Metabolism (ERNDIM) website.

Quality management
Taking a wider view, EuroGentest has also initiated discussions on **quality management of EQA Schemes.**

Reference materials
EuroGentest first International Reference Materials Symposium resulted in the initial production of 3 CRMs for prothrombin (incorporating PT for rare mutations). In 2007 **a second RM symposium is being held**, guidelines for commutability and use of RMs in GT drawn up and further trials of CF multiplex RMs held. The Unit will also start to look at RMs for Cytogenetics.

Validation and SOPS
Unit 5 also assists Unit I in the validation of existing technologies and creation of SOPs. Ongoing validation activities include DNA extraction methods for large volumes of blood, Multiplex Ligation-dependent Probe Amplification (MLPA) and Diagnostic use of commercial kits for Cystic Fibrosis.

Generating the world's most comprehensive database
The information gathered by Unit 1 on the quality assurance status (QAu) of genetic testing in Europe is being fed into Unit 2 and Orphanet. During 2007 this data will be used to provide the first comprehensive, independent survey of services, current practice and QAu status.

Towards tighter definitions
Unit 3 has been organizing a series of expert meetings. At the Porto meeting in late 2006 there were 39 persons from all over Europe as well as from US, Canada, South-Africa, Argentina and Ecuador. To date the Unit has completed a survey of National Regulations on Genetic Counselling which has thrown up 49 different variables in those 56 guidelines. Further analysis is ongoing and is intended to lead to draft **guidelines on the minimal criteria for genetic counselling and a definition of genetic testing.**

Tackling legal aspects
Genetic testing throws up a variety of controversial legal aspects regarding patient rights. Unit 4 has concentrated on three main topics - the status of the European Convention on Human Rights and Biomedicine (1997). The **first two reports on patient's rights** – covering Denmark and the Czech Republic – were published in March.

Evaluating new technologies
EuroGentest launched a **technology evaluation service** offering access to its 100 plus member laboratories across Europe. The results of the evaluations are being presented at a satellite meeting at the EHSG meeting in Nice in June 2007. A second call has already been issued aiming to cover technologies such as improvement of detection of DNA changes such as mutations, deletions, insertions, duplications, translocation, methylation etc, as well as the improvement of technical aspects (approaches such as arrays) that are related to the detection of these genetic changes.

Ensuring patients get the right information
Last but not least, Unit 6 looks at educational aspects of genetic testing and has been working to assess whether information currently available to patients and families is sufficient and effective. Following further consultation with patient interest groups, Unit 6 has now published generic information leaflets for patients and families covering key issues related to genetics. These are being translated into a number of European langu-

ages where there are significant gaps at present. In addition, Unit 6 is working to identify **the minimum set of skills required by any health professiona**l that provides genetic counselling in the context of genetic testing. The elements of this set of skills will be discussed by both patient groups and professionals.

Jean-Jacques Cassiman
Coordinator of the EuroGentest NoE
On behalf of the participants of the NoE

Encouraging Quality Management and Accreditation in Genetic Testing Laboratories: Workshops on Accreditation and Quality Assurance Database

Sarah Berwouts[1,2], Anniek Corveleyn[1,2], Nick Nagels[1,2], Michael A. Morris[1,3], Elisabeth Dequeker[1,2]
[1]EuroGentest Network of Excellence. EU Contract no: FP6-512148.
[2]Department for Human Genetics, University of Leuven, Belgium
[3]Service of Genetic Medicine, Geneva University Hospitals, Switzerland.

The demand for genetic testing and the number of laboratories involved is increasing throughout Europe. In this context, and particularly given the potential medical, personal and societal impact of genetic test results, effective quality assurance (QAu) is essential to guarantee that patients receive the most accurate results possible. The implementation of quality standards in genetics laboratories and the growing interest in accreditation have led to a need for a better understanding of quality management (QM) and QAu.

The EU project EuroGentest1 has the aims of improving and harmonizing the overall quality of genetic services, and two EuroGentest initiatives are specifically addressing facilitating and encouraging uptake of QAu in laboratories.

Accreditation workshops. In a focus on training and education, key parameters in improving quality, EuroGentest has developed specialized workshops to help laboratories in their processes of developing QM systems and working towards accreditation.

To date six workshops have been held since 2005, attended by 123 participants from 66 institutions in 20 different countries. The team consists of a director of an accredited laboratory (Dr Michael Morris), a lab quality manager who conducts accreditation audits for the Belgian Organisation for Accreditation (BELAC) and who is a member of ISO Technical Committee 212 (Prof. Dr Elisabeth Dequeker), a EuroGentest scientist (Sarah Berwouts), and collaborators of EuroGentest partner 'Management, Consulting & Research' (MCR) Leuven, specialists in the "human side of change processes" (Mieke Gielis and An Wynants).

The participants come from laboratories that are already accredited, that are working actively towards accreditation, or that are early in the process of developing and planning their QM system. They have included laboratory directors, scientists, technicians and quality managers, from cytogenetic, biochemical and molecular genetic laboratories, as well as from other fields such as forensic genetics and clinical chemistry.

The workshops are designed to cover all aspects of QAu and to be very interactive, based around case-studies and targeted group discussions, with complementary presentations on specific topics. Feedback from the participants reveals that the format is highly appreciated for illustrating the theory of QM in practical, real-life situations in the laboratory.

The most recent workshop took place in Athens, Greece, on April 12-13 2007. Thanks to the warm welcome and organization of the local host, Dr Lina Florentin-Arar, as well as to the enthusiasm and involvement of the 32 participants, the workshop was very successful. Amongst its topics were: interpreting accreditation standards; comparing the different standards in Europe2; implementing and living with quality systems; and case-studies of concrete situations related to quality processes in laboratories.

Two further workshops will be held this year, first the ESHG congress in Nice on 15-16 June 2007, and in Leuven on 20-21 September 2007. The workshop in Nice will follow the same format as Athens, in contrast to the workshop in Leuven, which will be specifically on performing internal audits. A mixture of video clips and role-playing will give the participants an overview of all aspects of auditing from preparing

1 EuroGentest Network of Excellence, an EU-funded project. EU Contract no: FP6-512148. Website: www.eurogentest.org

2 Accreditation is a recognition that a lab works according to a good quality management system and is technically competent. Standards for accreditation in Europe include ISO 15189, ISO 17025, CCKL and CPA

and reporting the audit to the communication and behaviour skills of the auditor. It is planned to organize a workshop each year just in advance of the ESHG.

Quality assurance database. Although a number of public websites provide lists of medical genetic testing laboratories and of available tests, reliable public information about QAu is sparse or even intentionally absent. EuroGentest is surveying QAu in European genetics laboratories, in collaboration with Orphanet:

- to determine the current state of QAu in genetic testing laboratories in Europe;
- to follow the changes in QAu during the period of activity of EuroGentest;
- and to make available reliable public information about QAu, to facilitate the choice of testing laboratories for "consumers" (medical doctors and patients), to encourage laboratories to invest in QAu, and to educate the general public and policy-makers about the importance given to QAu by the medical genetics profession.

By compiling data provided by many collaborators (including Orphanet, EQA providers, and national genetics societies), over 1000 laboratories in Europe offering some form of genetic testing were identified. In 2006, they received an online survey addressing critical aspects of QAu, including the presence of a designated quality manager, accreditation, certification and/or licensing status, and regular participation in external quality assessment (EQA) programmes (performance in EQA is considered as private to the laboratories, and is not evaluated).

To ensure the highest possible quality of the data, which might vary according to the identity of the respondent in a laboratory, replies were peer-reviewed and then validated by comparison with EQA providers and accreditation bodies. After validation by EuroGentest, data is returned to the laboratories for permission to disseminate the data from the European QAu database, which will be available via the EuroGentest and Orphanet web sites. Data is shared between EuroGentest and Orphanet to ensure coherence and to avoid contacting laboratories unnecessarily.

To date, 350 labs from 32 countries have provided complete replies and their data have been validated. This initial group cannot be considered representative of the whole field, but would be expected to be biased towards laboratories that are currently more advanced in the development of their QAu systems. Even within this group, however, the implementation of formal quality management systems is currently the exception rather than the rule: less than 20% of laboratories are fully accredited and only half have designated quality managers; approximately half laboratories participate in EQA schemes for genetic tests.

The first version of the database will be released publicly in spring 2007; it will be available initially through www.eurogentest.org and later in the year also through www.orpha.net.

With the new awareness of the central role of QAu, making this information available will benefit consumers, by facilitating informed choice of laboratory partners for performing tests, and genetics services, by facilitating selection of reliable partners for referral of tests which cannot be performed locally and by valorizing their efforts and investment in QAu. This study provides the first overview of the status of QAu in European genetics laboratories.

More information, activity reports and details on how to participate in the accreditation workshops and in the QAu database can be found at http://www.eurogentest.org/unit1/.

Through these and many other initiatives, the quality management unit of EuroGentest is working intensively to achieve its stated aims: to measurably improve the quality of management and provision of genetic laboratory services, for the benefit of patients; and for laboratory accreditation to be considered as the norm.

Acknowledgements

We are very grateful to Lieve Desmet and Romy Gentens for their expert assistance.

Athens Workshop at Lito Maternity Hospital and AlfaLab Molecular Biology and Cytogenetics Center

Announcement 3rd Meeting, National Human Genetics Societies (NHGSs) Monday 18 June 2007, 11.15 to 13.15, Room Eutherpe (top floor), Nice Acropolis

This is to announce the third meeting of the European Human Genetics Societies in Nice on June 18.
The tentative agenda is as follows.
A questionnaire has been distributed - see below.
A high participation from all European human genetics and related societies or from those interested in establishing a new society in their country if a society is not yet present is encouraged. Please send your mail of participation, reply to the questionnaire, or enquiry, to the following address: pierfranco.pignatti@univr.it

Best wishes,
Pier Franco Pignatti

TENTATIVE AGENDA

11.15 Welcome and Introduction

11.20 Self-presentation of the participants

11.30 The ESHG activities (John Burn)

11.40 Report on the questionnaires received from the NHGSs, and proposals on how to increase ties among NHGSs and the ESHG (Pier Franco Pignatti)

11.50 Quality Assurance (QAu) for genetic testing, a QAu-database, and laboratory accreditation in Europe (Mike Morris - Els Dequeker)

10.10 Medical/Clinical Genetics recognition as a EU Specialty: an update (Ulf Kristofferson)

20.20 The education of genetics counsellors and who can do genetic counselling. Can we have one model in Europe? (Helena Kääriäinen)

30.30 European exchange of candidates for medical genetics specialization (Albert Schinzel)

40.40 Other topics, General discussion and Conclusions

15.15 End of meeting

(lunch buffet or lunch boxes will be provided)

Human Genetics Societies in Europe Questionnaire

The Society:
Name:
Web-site:
Documents available on the web-site (please indicate titles):
Number of Members:
What is the (annual) membership fee (in Euros please)?
Name of president or contact person:
Address, Tel, Fax, E-mail of president or contact person:

Meetings of the Society:
Frequency? (e.g. annual, biannual)
Composition? (e.g. the Society alone/with other societies/ regional or interest group)
How many persons attended last meeting?

Is Medical/Clinical Genetics officially recognized as a specialization in your country?
What is the name of the specialization?
How many years does it last?
Is the specialization for MDs only or for PhDs and other graduates as well?
In that case, are the curricula (partly) different?

Information on other human genetics related societies in your country
(e.g. Medical, Cytogenetics, Clinical):
Is there a federation? (If so please indicate)
E-mail of president or contact person:

Comments: (free)

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European Society of Human Genetics

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Email: office@eshg.org

Invitation to the

Annual Membership Meeting 2007

At the EUROPEAN HUMAN GENETICS CONFERENCE 2007

Sunday, June 17, 2007 at 7.00 – 8.00 p.m.

Room Hermès

Nice Acropolis, 1, Esplanade Kennedy, BP 4083, 06302 Nice Cedex 4, FRANCE

AGENDA

Opening by the President of the Society, Professor John Burn

1. Activity of the Society 2006-2007
2. Financial Report of the Society 2006
3. Discharge of the Board Members for the year 2006-2007

Opening by the new President of the Society, Professor Pier Franco Pignatti

4. Results of election for President-Elect
5. Results of election for Board Members
6. Membership fees 2008
7. Site of future European Human Genetics Conferences
8. Budget proposal 2008
9. Major policy questions proposed by Board
10. Future activities