



European Society of Human Genetics

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The Society's administrative office:

European Society of Human Genetics, c/o Vienna Medical Academy
Alser Strasse 4, A-1090 Vienna, Austria

Tel: +43 1405 13 83 22

Fax: +43 1 407 82 74

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, 14 June 2004 at 18.45hrs to 19.30hrs in Room 1 at the International Congress Centre, Munich, Germany.

Please bring it with you

Medical Genetics in Lithuania



Vaidutis KUCINSKAS

Clinical genetic services are now established in at least 29 different countries in Europe (Godard et al, 2003), although a series of papers resulting from the Concerted Action on Genetic Services in Europe (1997) had revealed extreme variability in the level of available services. There has been a powerful medical influence on the practice of genetic counsellors in Europe, as doctors lead clinical genetic services.

In Lithuania, genetic services are provided exclusively by medically trained staff, with very little input from other professions such as nursing. However, other non-medical professionals are employed in the team, e.g. geneticists, biochemists, medical biologists, and psychologist.

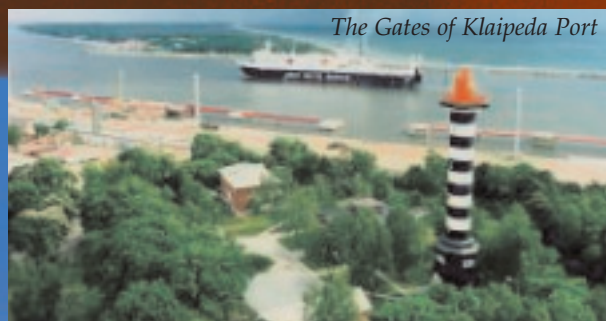
Genetic services in Lithuania, similarly to a number of other countries, have been developed as multidisciplinary regional centres with strong links with academic human genetics. Considerable expertise has been built up in the delivery of clinical and laboratory services for a wide range of genetic disorders in Lithuania. Effective working relationships have been established with various clinical specialists as well as with those in primary care.

Legislation The provision of genetic services is specifically legislated in Lithuania in different ways. According to Act No. 136/1991 of the Ministry of Health the Human Genetics Centre (Centre for Medical Genetics from 2003) was established in Santariskiu Clinics of Vilnius University Hospital. Clinical genetics became a recognised medical specialty in Lithuania by the Act No. 199/1991 of the Ministry of Health. The Act No. 74/1992 of the Ministry of Health regulates the registration of congenital anomalies in Lithuania. Any medical doctor who has diagnosed congenital anomaly/anomalies at birth must notify it/them to the Lithuanian Registry of Congenital Anomalies (LIRECA) at the Centre for Medical Genetics at Santariskiu Clinics of Vilnius University Hospital. The Act No. 706/1997 followed by the Act No. V-220/2003 of the Ministry of Health regulate national standards for genetic counselling and professional responsibilities of clinical geneticists. The main activities of the Centre for Medical Genetics at Santariskiu

Clinics of Vilnius University Hospital are being performed according to the national health programme "The Structure, Defects and Protection of Gene Pool of the Lithuanian Population" (1994-2003) and "Genomics in Prevention, Diagnostics and Treatment of Children Diseases" (2004-2007). The Act No. VIII-1679/2000 of the Parliament of the Republic of Lithuania has passed a Law on Bioethics, which regulates genetic testing in humans. Genetic testing in Lithuania can only be carried out for medical, scientific and forensic purposes. Research projects dealing with genetic testing of individuals from Lithuania must be approved by the Lithuanian National Committee on Biomedical Ethics. Essential precondition for genetic testing for diagnostic and/or research purposes is a written informed consent obtained from the individual or his/her legal representative.

Institutions At present the main institution for medical genetics in Lithuania is the Department of Human and Medical Genetics (DHMG) of the Faculty of Medicine of Vilnius University (<http://www.geneticalhumana.lt/VU/engl/home.htm>) together with its clinical, training and experimental base in the Centre for Medical Genetics at Santariskiu Clinics of Vilnius University Hospital.

DHMG traces its roots back to the first genetic counselling office in Lithuania established in 1971. In the present form it was founded in 1991 as the Human Genetics Centre of the Faculty of Medicine of Vilnius University. Since then a number of organisational changes took place. Finally, in September, 2002 the Human Genetics Centre was changed to the Department of Human and Medical Genetics, while the Human Genetics Centre at Santariskiu Clinics of Vilnius University Hospital was in parallel reorganised to the Centre for Medical Genetics. In 2003 a research unit - Human Genome Research Centre - was formed under the auspices of the DHMG.



The Gates of Klaipėda Port

Medical Genetics in Lithuania (cont'd)

Kaunas Academical Clinics are also engaged in genetic services, training and research in the field of human and medical genetics, although on a smaller scale. Some genetic services are provided in Siauliai Regional Hospital and Panevezys Regional Hospital.

Lithuanian Society of Human Genetics (<http://www.geneticahumana.lt/LZGD/>) is a non-profit organisation, which provides a forum for the Lithuanian specialists working/interested in the field of human and medical genetics.

Research Investigations in the field of human and clinical genetics were started in Lithuania in the third decade of the 20th century. They were virtually stopped in post-World War II Soviet occupation period (so-called Lysenkoism period (1935-1965), when a totalitarian state had been used to suppress all research in genetics) and started to recover just in the 7th decade. Restoration of the independence of the Republic of Lithuania in 1990 gave a new impulse to the development of research in the field of human and medical genetics and implementation of its achievements into clinical practice in Lithuania by eliminating strong negative ideological pressure and opening doors for extensive collaborative contacts with leading European and world research, clinical and educational centres. The Department was engaged in a number of national and international research and health programmes. The main fields of research interest are (1) application of DNA-based diagnosis to patient care and (2) human genome diversity (mtDNA, Y chromosome and nuclear DNA polymorphisms) in human populations. For more information and a list of publications visit the website of the Department. (<http://www.geneticahumana.lt/VU/engl/research.htm>)

The Cathedral and Bell Tower, Vilnius



Training and education Courses of lectures on genetics and human genetics were started at Vilnius University in the 7th decade of the 20th century. Systematic courses on human and clinical genetics were introduced at the Faculty of Medicine of Vilnius University in 1990. Approximately the same type of teaching was introduced in Kaunas University of Medicine, another locality for medical training in Lithuania.

Special training was introduced for medical genetics after clinical genetics had been recognised as a medical specialty in Lithuania. Initially, six years of undergraduate training were followed by one year of compulsory primary residency, then secondary specialist residency. Finally special training in clinical genetics took place in a two year course of the tertiary residency under the auspices of the Human Genetics Centre of the Faculty of Medicine of Vilnius University (till 2002). In 2003 the system of residency in Lithuania was reorganised according to the EU recommendations. At present, a six year programme of the undergraduate training in medicine is followed by a one year internship with a subsequent programme of four years residency training in clinical genetics.

Genetics services At present the Centre for Medical Genetics (CMG) at Santariskiu Clinics of Vilnius University Hospital is the main institution (and the single specialized one) providing genetic services for the population of Lithuania. Genetic services are also provided at Kaunas Academical Clinics (one clinical geneticist, M.D.), Siauliai Regional Hospital (one clinical geneticist, M.D.), and Panevezys Regional Hospital (one clinical geneticist, M.D. and one cytogeneticist).

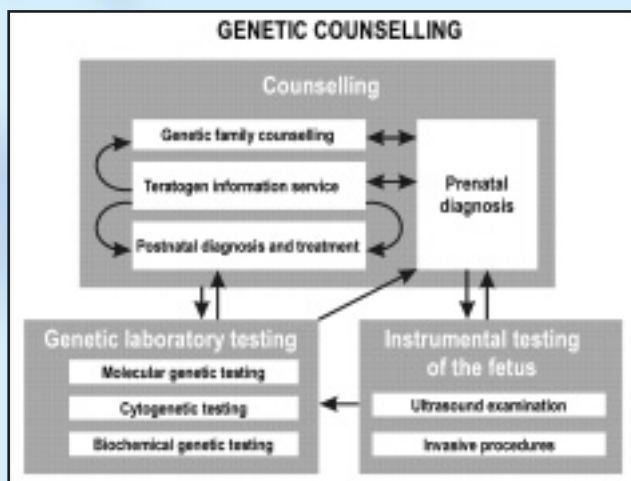
Centre for Medical Genetics at Santariskiu Clinics of Vilnius University Hospital - CMG was established in 1991. It originated from the first genetic counselling office in Lithuania, which had been founded in 1971. The main activities of the CMG are focussed on the prevention of congenital anomalies (inherited diseases and congenital malformations) in Lithuania and are organised in four major blocks: (1) genetic counselling, (2) prenatal diagnosis, (3) nationwide neonatal screening for phenylketonuria and congenital hypothyroidism, (4) Lithuanian Registry of Congenital Anomalies (LIRECA). Activities of the CMG are closely interrelated with the activities of the Department of Human and Medical Genetics of the Faculty of Medicine of Vilnius University and deal with scientific research as well as graduate and post-graduate specialists' training in human and medical genetics. At present, nearly 40 specialists are employed in CMG.

About 6000 genetic consultations are performed per year in the CMG. Of these, 1500 include laboratory testing performed in the laboratory unit of the CMG (cytogenetics, molecular genetics and biochemical genetics laboratories) as well as non-invasive (ultrasound examination) and invasive (amniocentesis, chorionic villi sampling) procedures for prenatal diagnosis (Fig. 1). HLA-based tissue typing for organ transplantation and biological paternity testing are also performed in the

Medical Genetics in Lithuania (cont'd)

CMG. Quality of diagnostic laboratory testing in the laboratories of the CMG is ensured by the system of internal and external quality assurance.

Figure 1. Scheme of genetic counselling services provided by the Centre for Medical Genetics at Santariskiu Clinics of Vilnius University



Other sources of information about medical genetics in Lithuania:

- 1) websites of the Department of Human and Medical Genetics, Centre for Medical Genetics, Lithuanian Society of Human Genetics
- 2) previously published papers (Kucinskas, 1997; Kucinskas, Steponaviciute, 1999; Steponaviciute, Kucinskas, 2001).

Conclusions

1. Republic of Lithuania with 3.5×10^6 population and 30,000 annual birth rate has one adequately developed centre providing genetic services for the population, namely the Centre for Medical Genetics at Santariskiu Clinics of Vilnius University Hospital. In Kaunas, which is the second largest city of Lithuania, genetic counselling is under development. Some clinical genetics services are provided by clinical geneticists in the cities of Siauliai and Panevezys.
2. The main research and education centre in the field of human and medical genetics in Lithuania is the Department of Human and Medical Genetics of the Faculty of Medicine of Vilnius University. Courses on human and clinical genetics are included in the graduate studies programme for medical students of Vilnius University. The Department provides residency training programme in genetics, Ph.D. studies programme in clinical genetics and post-graduate training courses for physicians on important aspects of clinical genetics. Activities of the Department of Human and Medical Genetics are closely interrelated with the Centre of Medical Genetics at Santariskiu Clinics of Vilnius University Hospital, which is the clinical and laboratory basis of the Department.
3. Nation-wide neonatal screening programme for phenylketonuria and congenital hypothyroidism is in action

in Lithuania since 1975 and 1993, respectively. It is performed by the Centre for Medical Genetics at Santariskiu Clinics of Vilnius University Hospital. Patients diagnosed in the course of the programme implementation are treated by the specialists of the Centre for Medical Genetics.



4. Lithuanian Registry of Congenital Anomalies (LIRECA) is being carried out in the Centre for Medical Genetics at Santariskiu Clinics of Vilnius University Hospital.

5. At present (March, 2004) master studies programme for non-medical medical geneticists in the Faculty of Medicine of Vilnius University is at the initial stage of development by the specialists of the Department of Human and Medical Genetics of the Faculty of Medicine of Vilnius University. Assistance of the specialists from West European clinical, research and education centres is of great importance.

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Annual Membership Meeting

Annual Meeting 2004 - Monday 14 June 2004, Room 1, International Congress Centre,
Munich, 18.45hrs to 19.30hrs.

AGENDA

Opening by the President of the Society, Professor Veronica van Heyningen

1. Activity of the Society 2003-2004
2. Financial Report of the Society 2003
3. Discharge of the Board Members for the year 2003-2004

Opening by the new President of the Society, Professor Leena Peltonen

4. Results of election for President-Elect
5. Results of election for Board Members
6. Membership fees 2005
7. Site of future European Human Genetics Conferences
8. Budget proposal 2005
9. Major policy questions proposed by Board
10. Review of Society Statutes

Minutes of the European Society of Human Genetics Membership Meeting, Birmingham, May 6th 2003

The meeting was opened by Gert-Jan van Ommen 19.25 hours. 79 members were present.

1) ACTIVITY REPORT (Peter Farndon)

Complete reports of committees' activities were published in the last Newsletter. At the end of 2002, the ESHG had 1025 paid up members. There has been an increase in membership, particularly in members requesting online access to the Journal rather than the printed version.

2) FINANCIAL REPORT (Peter Farndon)

The completed accounts for 2002 were presented. The Society has operating accounts in sterling and in euros.

- 2.1 At the end of 2002, the sterling account contained about £25,000 of which £17,000 is earmarked for printing the Professional and Public Policy Committee reports in the European Journal of Human Genetics.
- 2.2 Income into this account is from membership subscriptions and the ESHG share of the yearly proceeds from the European Journal of Human Genetics.
- 2.3 Expenditure is for journal costs to our publishers for our members, for web space, and for printing and distributing newsletters and pre-financing future European Human Genetic conferences.
- 2.4 There remained an excess of income in the Euro account of about €140,700.
- 2.5 The Society needs to keep a substantial reserve because of the high costs of deposits for hire of conference centres for conferences.

3) DISCHARGE OF PRESIDENT AND BOARD MEMBERS FOR THE YEAR 2002-2003

Gert-Jan van Ommen turned over the meeting to the new President, Veronica van Heyningen. Prof Gert-Jan van Ommen was thanked for his year as President and for his future year as Vice-President. Three retiring Board members were thanked for their years of service, Jean-Louis Mandel, Bruno Dallapiccola, and Philippos Patsalis.

4) WELCOME TO NEW BOARD MEMBERS/LIAISON MEMBERS:

Leena Peltonen Palotie, (Helsinki) is the new President-Elect (unable to be present). New Board Members are: Pier Franco Pignatti (Verona), Alexis Brice (Paris), Gert Matthijs (Leuven), Nicolas Levy (Marseille). The members present briefly introduced themselves. The new members were welcomed by the meeting.

THE NEW PRESIDENT OF THE SOCIETY, Veronica van Heyningen was welcomed, and took-over the meeting.

5) BIRMINGHAM MEETING REPORT

Members present congratulated the Scientific Programme Committee for an excellent programme and thanked the local hosts, particularly Fiona Macdonald, for their hospitality.

6) FUTURE MEETINGS

The Annual Meetings Committee has recommended that the 2005 meeting will take place from 7th - 10th May in Prague. Milan Macek will be the local host. The members approved this decision.

There was a discussion as to whether the meeting should take place over the weekend period or during weekdays. The Secretary-General explained that members had previously requested a weekend conference because of the cost of airfares midweek; also conference centre hire was often considerably cheaper at certain times of the week. A question on preferences for future conferences will be included in the on-line evaluation for the Birmingham meeting.

7) EUROPEAN JOURNAL OF HUMAN GENETICS

- 7.1 The Board accepted the Publications Committee recommendation that the contract to publish the European Journal of Human Genetics remain with the Nature Publishing Group on a yearly basis for 2004.
- 7.2 The Editor noted that 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 the closing ceremony of this conference 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 to the next European Human Genetics Conference.

8) MAJOR POLICY MATTERS PROPOSED BY THE BOARD

- 8.1 *Administrative support to Society*
Peter Farndon explained that the roles of the Secretary-General post had increased greatly over the last four years, particularly in relation to liaison between the Society, its professional conference organisers and its journal publisher. The Society taking over its own membership administration has proved very beneficial to the ESHG but this is another area the Secretary-General has overseen. He confirmed that because of other commitments, he did not feel able to continue to undertake all these roles. The Board discussed Peter Farndon's suggestion that the Secretary-General responsibilities were redistributed into three posts.

The Board recommended discussion with the Vienna Medical Academy of the possibility of their taking over the administration of the Society. At last year's membership meeting the membership had given the Secretary-General a mandate to enter into such discussions but these had not been finalised. In view of the increasing workload and Peter Farndon's wish to step down, urgent discussions will now be held with the Vienna Medical Academy.

Peter Farndon agreed to continue for another year to oversee the transition of the administration to the Vienna Medical Academy. Helena Kääriäinen has agreed and is welcomed by the membership meeting to become the new Secretary-General. The Deputy Secretary-General is willing to step down from her job if relevant for the re-organised secretarial functions.

- 8.2 *Nominations Committee*
The board recommended setting-up a Nominations Committee to co-ordinate the nomination and election procedure for new Board members, and particularly to seek expressions of interest. The members of the committee will be those members rotating off the board (not officers) and serve for a term of two years. The first committee will consist of Jean-Louis Mandel (Chair), Philippos Patsalis, and Bruno Dallapiccola, Chair of the Scientific Programme Committee or nominee (Han Brunner), Chair of the Professional and Public Policy Committee or nominee (Ségolène Aymé), the Secretary-General, (Peter Farndon) and the President (Veronica van Heyningen).
- 8.3 *Review of the Statutes*
There is a need to update our Statutes to incorporate current practice. The Board held long discussions over the proposed modifications. When the proposals have been edited, members' approval will be sought for ratification in accordance with the legal rules. Following a period of consultation on the ESHG website, a final printed version of the Statutes will be sent by post to allow voting on their adoption.
- 8.4 *Newsletter - Editors*
The board has appointed two editors of the Society's Newsletter. These are Lina Florentin and Lisbeth Tranebjærg. Members were invited to contribute articles, particularly about genetics in their country.

9) REPORTS FROM PERMANENT COMMITTEES:

Full reports are published in the society's Newsletter, No. 8.

Professional and Public Policy Committee (Ségolène Aymé)

Position papers from previous workshops will be published in a special issue of our journal.

Two workshops are being organised in 2004.

The topics are 1) Pharmacogenomics - Professor Jörg Schmidtke, Hannover, Germany, is Chairman of the organising committee and 2) Genetics and Assisted Reproduction - Dr Helena Kääriäinen, Helsinki, Finland, is Chairman of the organising committee.

Other topics, which have been discussed for future work, are:

- 1) Behaviour genetics and
- 2) Provision of genetic services in countries with low income.

The members of the present Professional and Public Policy committee are all continuing for the next period, 2003-2006.

10) MEMBERSHIP FEES AND BUDGET:

After three years without an increase in membership fees and the fact that the Board has approved major administrative changes in the roles for the Secretary-General's function, it was therefore recommended to raise fees by about €8 (eight euros) per membership category. Approved by hand raising of the members.

The membership agreed the budget proposals for 2004.

11) CLOSE OF MEETING

The meeting was closed by Veronica van Heyningen at 20.00 hrs.

Minutes taken by Deputy Secretary-General, Lisbeth Tranebjærg, May 2003.

Activity Reports 2003 - 2004

President's Report



Being President of ESHG is not a job for the power-hungry, though I am sure the post could, and perhaps should, be developed in that direction. Currently, the President is predominantly a figurehead, and all the hard work is done by others. Apart from ceremonial duties at the annual European Human Genetics

Congress, the major role is as a facilitator, moving forward society business, taking final organisational decisions on the basis of extensive discussions at the level of the Board and the executive group.

Over the past 5 years Peter Farndon as Secretary-General has really carried the major burden of running most aspects of ESHG. In addition to the duties of Secretary-General, he ran the membership office in Birmingham, with expert and dedicated support from Ruth Cole and Patricia Wright, and he also carried the job of Treasurer, in addition to primary Secretarial tasks of gathering nominations, developing agendas and convening Board meetings. These tasks inevitably and relentlessly grow in a successful society like ESHG, until at the 2003 Congress in Birmingham the Board realised that a redistribution of

responsibilities was essential. Following very successful interactions for three years with the Vienna Medical Academy as our conference organisers, we decided to approach Jerome del Picchia and his staff to negotiate the transfer of the ESHG membership office to the VMA. We were delighted that Andrew Read (Manchester) agreed to take on the job of Treasurer, to help oversee financial activities. In his usual buoyant and selfless way, Peter agreed to stay on for a bridging year to transfer activities to the VMA and to ensure that he can pass on complete archives of all ESHG activities to his successors. Helena Kääriäinen (Turku) has been Secretary-General elect over the past year, gradually taking on the full set of tasks for the revamped post.

Ex officio, the President is a member of most of the Society's subcommittees, but we have been most fortunate in the committee chairs and memberships so these groups have made splendid effective contributions without much input from me. The Scientific Programme Committee under the knowledgeable guidance of Han Brunner (Nijmegen) has continued to deliver wonderful meeting schedules, that I feel very privileged to participate in. The Professional and Public Policy Committee, headed by Ségolène Aymé (Paris) is another ESHG institution to acknowledge with pride. They have conducted consultations and produced important reports, on topics of public and societal significance, which are available on the web site and were published in December 2003 as a supplement to the Society's journal, the European Journal of Human Genetics (EJHG). EJHG is going from strength

to strength, enhancing our image to the outside world, under the imaginative guidance of the knowledgeable, enthusiastic and hard working Editor in Chief, Gert-Jan van Ommen (Leiden). The Society's role is further fostered by the European Union Affairs Committee led by Jean-Jacques Cassiman (Leuven), who has an enviable ability to keep up with proceedings in this arena and ensures that ESHG's views are represented in important EU discussions. The Education Committee, under Celia Delozier (Geneva and California) has continued to gather information, for example on the available postgraduate genetics programmes in member countries. As some of you may recall, I suggested the development of educational tools, mainly for public education purposes as a major aim for my presidency, but we have not made much corporate progress in that direction. I hope, however, this will be carried forward, possibly by a more broadly based reconstituted Education Committee. Communicating with people is of major importance, and in this context we are proud and delighted with the ESHG Newsletter that has been evolving so successfully under the editorship of Lina Florentin.

These days, Societies like ESHG cannot function without very significant professional input into the smooth running of meetings and publications. I have already mentioned the new role of Jerome del Picchia and his team in running the new general secretariat, but the VMA had already fulfilled successfully the role of organising secretariat for the last three meetings. Rose International, under the leadership of Jantie de Roos, has managed the organisation of the commercial exhibitions at our annual conferences very effectively for a number of years. Recently we entered into an initial three-year agreement

with them to develop a general sponsorship programme to allow for more advantageous and stable arrangements. The Publications Committee, who meet with the Nature Publishing Group regularly about the optimal ways to present and promote EJHG, are considering some longer-term arrangements with NPG.

So the Presidential year has flown past, enriching my mailbox with over 450 emails. I want to thank all the people who have worked so hard to enable ESHG to make such good progress. No one will, I hope, object if I single out Peter Farndon's enormous contributions for special mention. My input has been very limited, for example ensuring Society support for the enthusiastic efforts of a recently recruited young member of the Board, Gert Matthijs, and his colleagues, to improve the European patenting position. The Nominations Committee, headed by Jean-Louis Mandel (Strasbourg), would welcome more strong proposals from young ESHG members for Board membership - self nominations are regarded as a sign of strong commitment and are very welcome, though they need to be seconded by other members.

I look forward to an excellent meeting in Munich, jointly with EMPAG (the European Meeting on Psychosocial Aspects of Genetics) and with the German, Austrian and Swiss Human/Medical Genetics Societies. Thomas Meitinger has been a very effective local organiser. Each annual meeting is the glorious crowning climax for the outgoing President, and the dawn of a new era as the new President is installed, this year the highly respected Leena Peltonen from Helsinki.

Prof Veronica van Heyningen

Secretary-General's Report



The Society again has had a very busy and successful year as evidenced by the activity reports in this newsletter.

Our President, Veronica van Heyningen, has included in her report many of the highlights of the past year. The membership administration of the ESHG has been transferred to the capable hands of the Vienna Medical Academy, who have an instinctive understanding of the needs of a professional society. We are fortunate to work so closely with them and with Rose International (our exhibition organisers) who also always puts the needs of the Society first.

My term of office is ending at the AGM in Munich. I welcome Helena Kaariainen as Secretary-General and Andrew Read as Society Treasurer and I am sure they will enjoy the "ESHG experience" as much as I have. There have been challenges, particularly over stabilizing the Society's finances so that the Board should now be able to plan major initiatives.

I would like to thank all the members for their help and support to me and the Society, particularly the local hosts of past meetings and members of the ESHG committees.

Thank you!

Professor Peter Farndon

Activity Reports 2003 - 2004

(Pending Approval by the Board)

Professional and Public Policy Committee (PPPC)



The PPPC is currently composed of Violetta Anastasiadou, Ségolène Aymé (chair), Suzanne Braga, Jean-Jacques Cassiman, Domenico Coviello, Gerry Evers-Kiebooms, Helena Kääriäinen (secretary), Gyorgy Kosztolanyi, Ulf Kristofferson (Deputy-chair), Joerg Schmidtke, Jorge Sequeiros, Leo ten Kate and Lisbeth Tranebjaerg. As Leo ten

Kate is willing to step down after many years serving the committee, candidates are invited to contact PPPC members before the Munich meeting.

The PPPC dedicated its activities to the preparation of a document on pharmacogenetics. The work was conducted using the same scheme, which has been operated successfully in the past. The PPPC members met once to define the work plan and to identify the experts to be invited to comment (around 150 experts).

A background document, summarising the professional and societal issues to be debated, was drafted by a genetic counsellor working with Segolene Ayme, based on the medical and scientific literature and on any other relevant document. The experts were asked to review the

background document. A sub-committee was then invited to a two-day workshop of 50 participants (March 04) in Seville. This workshop was jointly organised with the Institute for Prospective Technological Studies, which is an EC joint research centre, and with EPPOSI the European Platform of Patients organisations, Science and Industry. Experts from all disciplines actively took part in the exercise and agreed on conclusions. The findings of this workshop formed the basis of a report that will be published soon as a joint ESHG/EPPOSI document. The discussions during the workshop have been incorporated in the background document, which has been sent for review to the 150 experts in the field. The main conclusions will be presented at the ESHG membership meeting in Munich. The final document and the ESHG recommendations derived from it will be put on the ESHG website for public consultation in June and July. The final document will be published as a supplement issue of the EJHG.

The PPPC is considering doing the same exercise soon to explore the societal issues around "reproductive medicine and genetic testing". The workshop will be organised in March-April 2005.

The next topics to be explored are still in discussion, however, one of the topics under consideration is "Behavioural genetics".

Ségolène Aymé

Education Committee



Celia D. DeLozier and Domenico Coviello (for the ESHG Education Committee).

In an effort to foster and coordinate the harmonious development of educational programmes in Human Genetics in Europe, the ESHG created a standing Education Committee, which met for the first time in Strasbourg (2002).

The committee began its work by strengthening ties with other groups having similar goals, either through the committee's initiatives or by relying on committee members directly involved in these projects as liaisons, specifically:

- the Professional and Public Policy Committee of the ESHG (<http://www.eshg.org/PPPC.htm>)
- the European Genetics Foundation (www.eurogene.org), which offers 7-8 courses a year from its new venue in Bertinoro, Italy; the ESHG funded a total of five scholarships to three of these courses
- the EC-funded survey of genetics education for non-geneticist health care providers, GenEd (<http://www.medicine.man.ac.uk/gened/default.htm>)
- the ORPHANET database, dedicated to providing scientific and medical information on rare diseases (<http://www.orpha.net/>)

Education Committee (cont'd)

- EDDNAL (European Directory of diagnostic DNA laboratories), a non-profit registry specifically intended for professional use (www.eddnal.com)
- the European Platform of Patient Organizations Science and Industry (EPPOSI) (<http://www.epposi.org/>)
- the Institute for Prospective Technological Studies (IPTS) (<http://www.jrc.es/home/index.html>)
- the European Cytogenetics Association (ECA) (<http://www.biologia.uniba.it/eca/>).
- more recently, with the new EC-promoted Networks of Excellence project EUROAGENTEST, an Integrated Network for test development and harmonization of quality of genetic testing services in Europe. Within this framework, a directory of organizations that produce and provide quality educational material for health care providers and the public will be established.

The committee is also working on its first independent project, development of an on-line directory of specialty training programmes in human/medical genetics. A questionnaire was developed and sent to presidents of the national societies to assess post-graduate specialty programmes in genetics for physicians, other health care professionals and scientists. Information has thus far been collected on 45 programmes from 13 countries. The Directory, which provide summaries and contact information will be accessed through the ESHG website.

The committee welcomes suggestions from members of the ESHG concerning its future work.

Scientific Programme Committee



Following the successful Birmingham meeting, the SPC met once to agree on invited speakers and plenary sessions for the Munich meeting. It was decided to have 12 concurrent sessions rather than the usual 9, because a larger number of attendees were expected this year. The reason is that

this is going to be a joint meeting with the German, Swiss, and Austrian societies. A similar arrangement was made with EMPAG. A joint plenary session and symposium were agreed upon, entitled: "Regional differences in genetic testing and genetic counselling in Europe" and "Models for service delivery" respectively. The SPC met again in February 2004 to review 1300 abstracts and to choose the 72 that were most suitable for oral presentation.

It was decided to accept the offer by Professor Gert-Jan van Ommen to create a special award for the best presentation from submitted abstracts at the annual conference in the field of complex and statistical disease

genetics. This award is in memory of the late Lodewijk Sandkuyl who died in December 2002.

The joint format with the German, Swiss, and Austrian societies further allows us to have additional educational sessions at the start of the conference. If these are deemed successful, we shall try to make similar arrangements for next year's meeting. Another addition to the programme is an evening poster viewing session. The organization of the posters has been reviewed as they were too spread out in Birmingham. This will be improved in Munich.

Members of the SPC for the meeting in Munich 2004 included Han Brunner (Chair), Thomas Meitingner (local host), Françoise Clerget-Darpoux, Cornelia van Duijn, Karl-Heinz Grzeschik, Stanislas Lyonnet, Jean-Louis Mandel, Andres Metspalu, Gudrun A. Rappold, Andrew P Read, Niels Tommerup, Gert Jan van Ommen and Orsetta Zuffardi. Representing the German, Swiss, and Austrian societies were Claus Bartram, Bernhard Weber, Brunhilde Wirth, and Gerd Utermann joined the SPC to create a joint programme for this meeting. Helena Kaariainen joined us in her function as Secretary-General elect of the ESHG.

Following the Munich meeting, Orsetta and Françoise will leave the SPC. We thank them for being there always, and bringing their many ideas and insights to the committee. We welcome as new SPC members for the Prague meeting 2005 Peter Lichter, Juha Kere, Paulo Gasparini, and, local host, Milan Macek jr.

Han G. Brunner, chair of the SPC

Report from the Publication Committee

Members: G.J. van Ommen, T. Meitinger, J.J. Cassiman (chair), Helena Kaäriäinen, P. Farndon, Veronica van Heyningen



The contract with the Nature Publishing Group (NPG) was maintained as a yearly renewable contract as decided by the board. In the meantime 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 EJHG, 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 Amsterdam in February 2004, the committee, together with M.O'Such (managing editor for NPG) Ruth Kirby and Sue Deeley (NPG), reviewed the achievements and suggestions for improvement of the journal in preparation of the editorial board meeting in Munich. Particular attention was given to further developments of the NPG website. A suggestion to propose to the board of the ESHG to formalise a longer contract period with NPG was discussed. Further information on the journal can be found in the report of the editor-in-chief.

European Journal of Human Genetics - Editor's Report

The last year has seen major improvements in the service and information to readers and authors. The *online submission* system has now been fully operational for more than a year. Submitted manuscripts and illustrations are converted into pdf files on-the-fly and entered directly into the reviewing process and, if accepted, into the typesetting process. Authors can follow the status of the reviewing process step by step (at the same time as the Editor is finding out), and communications between editors and the office are streamlined and time-consuming couriering is a thing of the past.

The next major upgrade, as of last September, has been the introduction of *Advance online publication (AOP)*. Within 4 weeks after acceptance of the proofs the manuscripts are visible to ESHG members and other subscribers on-line.

The location of the *EJHG under the Nature.com pages* has tremendously increased its visibility. We are noticing this by, amongst others, a gradual rise of submissions from the Americas. The visibility of our content is further enhanced by the monthly scanning of its contents by the NPG staff for newsworthy topics, which are then highlighted on the Genetics page of the Nature.com website, and, occasionally, even on the Nature.com home page. The full content of the paper in question is then made freely available to non-EJHG subscribers. So far, every month one of the EJHG's author teams has received an email indicating that their paper has thus been brought under the attention of a much wider audience. Also, a special issue has been devoted to the ESHG *Public and Professional Policy Committee's recommendations* and background documents on Genetic Services, Genetic Screening, Genetic Testing and Sample Banking. This entire issue is freely accessible and the documents can be (and are intensively) downloaded as pdf files by professionals and policy makers throughout Europe.

In terms of content development, the *'Practical Genetics'*-series of focused descriptions of genetic disorders, with the current status of insights and diagnostic possibilities, is gradually picking up, but we still would wish to encourage authors to give their favourite clinical entity a try.

The Society, in association with Nature Publishing Group, is delighted to announce this year's winners of the *annual award for most highly cited articles*. The winning authors receive one year's subscription to the journal and a free registration at the European Human Genetics Conference 2004 in Munich. The first prize winner also receives a cash award. The winning papers are:

- 1st place:** Christoph Plass and Paul D Soloway, DNA methylation, imprinting and cancer, EJHG 2002; 10: 6-16.
- 2nd place:** Marie-Pierre Audrézet, Jian-Min Chen; Cedric Le Maréchal et al.
Determination of the relative contribution of three genes to the etiology of idiopathic chronic pancreatitis, EJHG 2002; 10: 100-106
- 3rd place:** Florent Soubrier, Sabrina Martin, Amalia Alonso et al.
High-resolution genetic mapping of the ACE-linked QTL influencing circulating ACE activity
EJHG 2002; 10: 553-561

Finally, a novel activity is a *'News and Commentary'* item, aimed at the description of a specific advance in the field of Human Genetics from the recently published literature. Typically we signal the topics and solicit an expert, but here too, our readers are more than welcome to approach us in case they would wish to alert their colleagues to a major advance in their area of expertise.

Prof Gert-Jan van Ommen - Leiden

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:

- (1) to actively pursue the recognition of medical genetics as a European medical specialty.
- (2) to increase the visibility of Human Genetics and its practice in the European Union.
- (3) to continue the close relationship with the European Platform for

Patient Organizations, Science and Industry (EPPOSI) of which S Aymé and JJ Cassiman are board members.

The European Union of Medical Specialists, which last year proposed to the board of the EUMS the creation of a commission for clinical genetics, had to retract its generous offer a bit. They asked us to contact the presidents and secretaries of the Gynaecology and Paediatric commissions to ask for support to create this commission for Genetics. After contacting them these people promised their active support, which should be formalized at their March meeting this year. The next step will be to set up a mixed committee with Human Geneticists (to be appointed by the board of the ESHG) to determine the minimal European criteria for the specialty.

This commission would lead to an official recognition of the specialty 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. The ESTO report is available on:

<http://www.jrc.es/home/publications/publication.cfm?pub=1124>

The STRATA report and recommendations will be available soon.

EPPOSI continued its activities as an International foundation and organized another successful 'Rare Disease Therapy' workshop in The Hague, 2003, as well as a number of smaller workshops. In collaboration with the ESHG Professional & Public Policy Committee, EPPOSI will participate in the organization of the two planned workshops on pharmacogenetics and genetics and infertility. The next EPPOSI Therapy workshop will be held in Berlin, November 23-24, 2004.

*Prof JJ Cassiman
Belgium*

Activities of the International Federation of Human Genetics Societies

Liaison officer for the ESHG: JJ Cassiman

The IFHGS now represents 6 continental societies and regional societies represented by: the European Society of Human Genetics: Veronica van Heyningen and JJ Cassiman; the representatives of the American Society of Human Genetics: Robert Nussbaum, 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 Jose Maria Cantu, the representative of the East Asian Union of Human Genetic Societies: Takehiko Sasazuki; the Asia-Pacific Society of Human Genetics: Sangkot Marzuki. The situation in India and Pakistan is still not clear, but remains difficult notwithstanding the efforts of the local health professionals. In Bangladesh a Society of Human Genetics, Bangladesh was founded, the Academy of Science of Guatemala applied for membership. The IFHGS has 49 members (5 full members, 3 affiliate members and 41 corresponding members) and the number is steadily increasing. Efforts will be made to contact those who have not joined yet.

At the last executive board meeting in Melbourne, August 2003, the preparations for the International congress 2006 were presented. The international SPC will be composed soon.

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 chaired by Victor Penchaszadeh at the International congress of Human Genetics to be held in Brisbane, August 6 - 10, 2006.

More information on: <http://www.ifhgs.org/>

Professor Robin Michael Winter: 1950-2004



Robin Winter, who made major contributions to Medical Genetics in Europe and beyond, has died at the age of 53 years of oesophageal cancer. Robin's main medical interest was Dysmorphology and he used his unique skills in clinical observation to delineate many previously unrecognised syndromes and, as

technology developed, initiated programmes of research to identify the genes and pathways involved.

Robin undertook his pre-clinical studies, and an intercalated BSc Course in Genetics in the Galton Laboratory, at University College London where his fascination with genetics and patterns of malformations began. He was also a Post-Doctoral Fellow with Walter Nance at the Department of Human Genetics, Medical College of Virginia.

In 1978 he returned to the UK and took up one of the first three training posts in Clinical Genetics in the UK. In 1992 he moved to the Institute of Child Health, and Hospitals for Sick Children, Great Ormond Street, London where he spent the rest of his professional life and in 1994 was appointed Professor of Dysmorphology and Clinical Genetics.

Robin published widely he was as much 'at home' with scientists as clinicians and was respected by both groups.

A major theme in his research was the homology between mouse and human at clinical and molecular levels.

The London Dysmorphology Database series that he developed with Michael Baraitser are essential tools for experts and trainees, particularly in Dysmorphology but increasingly for other disciplines. (www.lmdatabases.com). With Michael Baraitser and me he founded and edited the journal *Clinical Dysmorphology*. He served on the Scientific Programme Committee of the European Society of Human Genetics and was honoured by the Society with the award of the Baschirotto Prize in 2001. He was also awarded the International Querci Prize for Paediatric Research for 1997-2000. The respect of his colleagues in the UK was reflected in his election as President of the Clinical Genetics Society.

Colleagues from all over the world sought Robin's opinion and he made major contributions to teaching and training in Europe particularly at the European Genetics Foundation courses at Sestri Levante and Bertinoro. He and I have organised Syndrome Identification Workshops at the ESHG Annual meetings for many years. He was much in demand as a mentor; numerous visiting trainees from the UK and overseas benefited from spending time in his department.

In spite of his many achievements Robin Winter was a modest man with a clever, rather understated, sense of humour. It is difficult for many of us to imagine professional life without him but the messages sent from colleagues around the world following his death, bear testament to his enduring legacy for the profession.

Dian Donnai,
(Dian.Donnai@cmmc.nhs.uk)

Genetics education: Improving non-genetics health professionals' understanding of genetic testing (GenEd)



In 1985 Hilary Harris, a general practitioner, and I wrote, "The clinical use of genetics should begin with general practitioners, backed by specialists in medical genetics. Primary care teams will need further education, supported by specialist advice, if they are to bring the benefits of genetic advances to their patients. The jargon of risks and probabilities

makes medical genetics seem complex and confusing, so

the task is challenging". (BMJ 1995;311:579-580)

To assess the way in which non-geneticist clinicians actually managed genetic patients' problems, the Department of Health (DoH) in UK funded a series of Confidential Enquiries (**summarised in BMJ 2001; 322:1061**) into the clinical management by non-geneticist clinicians of Downs, thalassaemia, neural tube defects, cystic fibrosis or multiple endocrine neoplasia type II. We found that;

"Clinical records were unacceptably poor, rarely showing whether genetic counselling had been offered or stating the reasons for accepting or rejecting an abortion. Less than half of cases known in advance to be at high genetic risk were referred to a clinical geneticist" .

If clinicians in UK were poorly equipped to deal with genetic problems how well might the speciality of clinical genetics in other European countries cope with a rapid influx from other specialities suddenly alarmed by the ubiquitousness of such problems and the complexity revealed by advances in molecular genetics. Data from Germany, for instance showed a marked increase (>44%) in the utilisation of genetic diagnosis in prenatal care in the 90s and a steady increase in the amount of molecular diagnosis paid for by the sickness funds. (Nippert I, Nippert RP, Horst J, Schmidtke J: Die medizinisch-genetische Versorgung in Deutschland. In: Medizinische Genetik 2 (1997) 188-205).

To answer these questions the EU Concerted Action was set up to see how well clinical geneticists were organised in Europe. The result of this 5-year programme of international co-operation gave a unique overview of the availability, access and quality of health services (Genetic Services in Europe. European Journal of Human Genetics 5 (suppl 2) 1-220 1997). It recognised the increasing participation by many medical specialities for whom education and training in genetics were clearly very important to allow them to deal with the routine genetic problems that arise in their practices

As a consequence under the 5th Framework the EU Accompanying Measure GenEd was funded. The explicit aims were "To conduct an empirical assessment of educational needs and priority topics for education in genetics among primary care providers and other non-genetics health professionals". Partners were drawn from France (Claire Julian-Reynier), Germany (Irma Nippert and Joerg Schmidtke), The Netherlands (Leo P ten Kate), Sweden (Ulf Kristofersson) and UK (Hilary Harris and Kirsty Challen) with observers from Greece, Hungary, Italy, Lithuania, Poland, and Spain.

The first phase of GenEd is now complete and has assessed the current structures and policies for the provision of the genetic education needs of non-genetic healthcare providers in the participating countries. The GenEd partner in each country used both published (paper or website) curricula, survey instruments (written or telephone) and personal contacts to establish individuals responsible for genetic education in the relevant institutions and to ascertain the topics, timeframe and assessment criteria for genetics in each educational programme. Details have been collected of undergraduate medical education (including how many institutions are involved, how long the undergraduate course lasts, if it assessed locally or nationally and by whom, is there a national curriculum and, if so, who is responsible for setting it and how much genetics is taught).

Procedures have been documented for specialist medical education in training between qualification as a doctor and recognition of the ability to practise as an

independent specialist.) The information collected included how many specialties are recognised in each country and is genetics one of these? How long is specialist training, who organises it, is there formal assessment during or after specialist training and who regulates this?

Continuing medical education (CME) is often required for re-accreditation as a specialist and information has been systematically collated on whether in each country CME is compulsory, how is it regulated and by whom and is genetics well represented in CME? The results of the phase I project have been presented at the ESHGC and the ASHGC and a series of papers are in preparation. (Benjamin CM et al. Phase 1 results of the GenEd Project. Spoken presentation at the European Society of Human Genetics, Birmingham UK, May 5th 2003), GenEd - Genetic education for non-genetic health professionals across Europe Benjamin CM et al November 2003, ASHG).

Phase II of GenEd has now commenced with a confidential postal questionnaire survey of randomly selected paediatricians, obstetricians & gynaecologists, midwives, and general practitioners. It is emphasised that the survey is a **Call for advice** from experienced clinicians and is not designed to reveal deficiencies in the practitioners' knowledge. Questions include; demographics, training, experience and opinion relating to training needs in genetics for their speciality. Each speciality is asked to comment on management involved in a clinical scenario relevant for their speciality namely paediatricians (multiple familial polyposis coli), obstetricians & gynaecologists (myotonic dystrophy), midwives (sickle cell disease), and general practitioners (sudden death in young people).

Phase II is scheduled for completion in January 2005 with an open conference in April/May 2005 Although designed to assess genetics in the education of non-geneticist practitioners GenEd has broadly compared European medical education in general. In collaboration with many European organisations and drawing upon the advice received from respondents to GenEd the next steps will encourage effective programmes for education and training.

Rodney Harris April 2004

Copies of the questionnaires are obtainable from Dr Caroline Benjamin RGN MSc PhD, Project Manager, GenEd coordinating office, Department of Medicine, Manchester Royal Infirmary, Manchester, UK. M13 9WL

Tel +44 (0)161 276 8546 (secretary) or +44 (0)161 276 5048 (office) & Mobile 07720 461060 Fax:+44 (0)161 274 4833

Gene patenting and licensing - on and beyond the BRCA case



The least one could say about the BRCA case, is that it has caused a stir in the field of genes and patenting. In contrast to the colleagues in the US, the geneticists in Europe had never worried about, nor considered, the possibility that genes and genetic tests could become

the exclusive property of a laboratory or company. In January 2001, the first European patent on BRCA1 was granted to Myriad Genetics, a US-based company. In the meantime, this company owns 4 European patents on the BRCA1 and BRCA2 genes. The problem is that Myriad Genetics has chosen to exert its monopoly rights, which allows it to halt other diagnostic laboratories from offering the BRCA testing. This has upset the genetic community and the public, and caused protest. Many professionals, mostly those who are familiar with gene patenting, including the experts at the European Patent Office (EPO), have commented that the problem is in licensing, not in patenting. Indeed, under the existing legislation, based on the European Patent Convention (EPC) and supported by the European 'Biodirective' 98/44/EC, genes and genetic sequences are patentable. Nevertheless, several national genetic societies, research institutes, and a few other organisations have opted to also question the patentability of these genes. The BRCA1 and BRCA2 loci have been identified and fine mapped as a result of a large, international collaborative effort. The BRCA1 gene was eventually identified by Myriad Genetics in 1994. The BRCA2 gene was sequenced in 1995, first at the Sanger Institute, and almost at the same time, by Myriad Genetics. Thus, as is often the case with the identification of novel genes, Myriad Genetics has heavily relied on the data generated through other, mostly publicly funded, research activities. Because the EPC allows a democratic control on patenting via an opposition procedure, several oppositions have thus been filed against Myriad's European BRCA1 and BRCA2 patents. The fact that EPC - and therefore also EPO - does not recognise the specific nature of DNA as a carrier of genetic and private information, which makes it different from the classical chemical compounds, and the observation that broad patents are commonly granted on genes and 'genetic inventions', has incited the opponents to also attack the current practice of patenting. In 2002, the ESHG issued a statement on gene patenting, thereby expressing its concern about the very broad scope of gene patents (see back page). We should not hide that the genetic community is also divided about patenting and genes. Several colleagues have - or have applied for - patents

on genes, genetic sequences and genetic tests. Many other geneticists reject their patentability on principle. In the meantime, Cancer Research UK (CRUK), who were recently granted a competing patent on the BRCA2 gene, has announced that it will allow public laboratories throughout Europe to use the patent for free. This does not entirely solve the problem.

The only way out is to define pragmatic solutions, that respect the rights of the patent owners and the users, and at the same time, warrant the patient's right and access to affordable health care provisions. I believe that the ESHG could and should play a prominent role in the shaping of a new patenting and licensing policy for genes, sequences and genetic tests.

Patents are meant to reward the inventors who have undertaken research as well as to encourage innovation. Gene patents typically cover the clinical applications of mutation analysis, as well as the use of the gene sequences for the development of therapies. Under current patent law, naturally occurring substances can be patented if their isolation from their natural environment involves a technical step. It is under these premises that genes and genetic sequences have been patented in Europe and in the US. The European 'Biodirective' literally states that 'an element isolated from the human body or otherwise produced by means of a technical process, including the sequence or partial sequence of a gene, may constitute a patentable invention, even if the structure of that element is identical to that of a natural element' (Directive 98/44/EC of the European Parliament and of the Council 6 July 1998). This is obviously a controversial matter.

In the specific case of the BRCA1 patents, one can argue about 'novelty' and 'inventive step', 2 of the criteria that are used to determine whether an invention is patentable. By 1994, the BRCA1 gene had been mapped to chromosome 17q with a LOD score over 6, and the candidate region had been reduced to less than one Mb. The effort to reach an obvious goal does not constitute a patentable act in the sense of patent law, even if it is costly and laborious.

Together with Dicky Halley, I have previously summarised the main arguments from the opponents (Eur. J. Hum. Genet 2002; 10:783-784). It is now time to move beyond the BRCA case, and thoroughly consider the different problems with patenting and licensing, at a global level.

There is evidence that the costs for the tests will increase. In a survey about the effect of the patent on the hereditary haemochromatosis (HFE) gene, J.F. Merz and co-workers (Nature 2002; 415:577-9) have reported that the present patent owner requests up-front payments of the order of \$25,000 per laboratory plus a per test fee of about \$20. I strongly believe that a royalty fee of \$20 (roughly €16 or £12) for a simple diagnostic test is outrageous. Merz et al. have also shown that many laboratories in the US have already refrained from offering testing for hereditary hemochromatosis, because

Gene patenting and licensing - on and beyond the BRCA case (cont'd)

of the licensing policy of Bio-Rad Laboratories. We know that companies with an interest in marketing kits for genetic diagnosis also refrained from developing kits for this disease. In the long run, patents will thus slow down biomedical innovation and delay the availability of cheaper tests. At this point, no-one has even suggested what the licensing cost for the mutation screening of a large gene could or should be. It is foreseeable that, for 'multiple gene' assays - which will be available with chip-based technology - stacking of royalties will make the tests prohibitively expensive. Thus, we are afraid that the societal aim of the patenting systems, which is the promotion of progress through the generation of useful new products - will not be met in the case of patents on gene-based diagnostic methods.

One reason why the market system does not always operate properly in the case of patents on genes is because genes and genetic sequences are different from classical chemical compounds. Genes and genetic sequences have an informational content. There is also no way of 'inventing around' the sequence if it is patented, because each gene and each gene sequence is unique in its kind. When the uniqueness of the genetic code is combined with the exclusive rights of patents, a true monopoly arises.

Thus, at first glance, this problem could practically be circumvented by royalty-free licenses, like the one offered to the European public laboratories by Cancer Research UK (CRUK) on BRCA2. However, this will be criticised by patent owners and supporters of the patenting system because it seemingly disregards the efforts and funds that the grantees have invested. Of course, billions have been spent on patent applications in the last 20 to 25 years, and it is not realistic to make all these patents and applications voidable.

The geneticists who oppose against patenting of genes and genetic tests on principle have found support in the discussion paper on 'The ethics of patenting DNA', issued in 2002 by The Nuffield Council on Bioethics (http://www.nuffieldbioethics.org/publications/pp_000000014.asp). In this important document, it was suggested that 'the description of an association between a gene and a disease amounts to little more than a discovery'. According to patent law, discoveries cannot be patented. I believe that a claim to exclude gene-based diagnostics from patenting is also legitimate from the standpoint that the patenting system should not interfere with the availability of a genetic test for the patient. The basis for such an exemption has been laid years ago: Art. 52(4) of the European Patent Convention of 1973 excludes 'diagnostic methods, practised on the human body' from patenting (<http://www3.european-patent-office.org/legal/epc/index.html>). This definition dates back to the days when people did not want surgical methods and other medical practices to be patented. This could be updated to include genetic testing.

This is a way to avoid monopolies on the use of a genetic sequence for diagnostic purposes in humans, without interfering with patent protection of other genetic applications, including the development of therapeutics. I acknowledge that the latter requires huge investments, but the first does not: once the relationship between a gene and a disease is established, diagnostic tests can be developed with relatively low costs. Patents and licenses are more a burden than a protection in this respect.

Ardent supporters of the patent system have publicly advised us to shut up. They do not see what the fuss is all about, as, in 15 years or so, the problem will be solved because all patents on genes will have expired. This is dumb. First, it fully disregards the current need of the patients. Second, patents on genes or combinations of genetic variants will be granted for several years on. In a moderate vision, patent offices and courts, left on their own, will find the appropriate equilibrium. But this will be a lengthy process, and this is why we have to push a little. Otherwise, I fear that, by the time it all settles, the public genetic services, and even the health care system, will have succumbed under exaggerated licensing fees and unbreakable monopolies.

As mentioned in the introduction, patent specialists and patents offices claim that the problem is in licensing, not in patenting. Indeed, Europe lacks a system that ensures guidance and surveillance of licensing of genetic inventions. The compulsory licensing system is the traditional safeguard against excesses in licensing. It is a system whereby a (national) committee or court obliges a patentee to grant a license to a user, at a fair expense. Strangely, no-one has ever applied for a compulsory license for gene-based diagnostics in Europe, so its effectiveness has yet to be proven. Experts in the field have now proposed the 'clearing house' model. The idea is reminiscent of the 'radio-song' principle, whereby a songwriter is remunerated for every time his song is broadcast. However, the necessary organisations for collecting the incomes from gene licensing are not in place, and it will not be easy to install them in practice.

I strongly believe that we should dare and devise better and new rules for patenting genes and for licensing, and that ESHG has a responsibility in this matter. My worry is that most of the discussions will be held at highly pitched, mostly economical forums where, unfortunately, the medical and genetic community, the social health care services and the patients are not strongly represented.

That is why I cordially invite everybody to assist to the special session on 'Genes and Patents' which will be organised on June 14, at the European Conference on Human Genetics in Munich, and where the different views and the novel models will be presented by the different stakeholders.

Gert Matthijs
Belgium



European Society of Human Genetics Statement on gene patenting

May 2002, revised January 2003

The ESHG wishes to highlight the legal, social and ethical issues surrounding the patenting of genes, as exemplified by the problems arising from the BRCA1 patent owned by Myriad Genetics.

While some hold the view that genes cannot be patented, others regard patenting as ultimately beneficial for genetic services and treatment. This represents a classical ethical dilemma, where there is a tension between two valid positions.

In reality, the PPPC notes that there is a continuum from initial research and discovery through to practical application. The issue becomes one of when in this process, it becomes reasonable to protect intellectual property.

The ESHG believes the BRCA1 debate highlights the need for the European Patent Office to revise its current practice to accept patent claims that are relatively early in the R&D process and very broad in scope.

A mismatch between where the Patent Office draws the line in the R&D process and the practical consequences for genetic services will lead to an unworkable situation, where compliance with international patenting law will be undermined.

The ESHG, through its Public and Professional Policy Committee, urges the European Patent Office to review its position in the light of the recent developments, as discussed thoroughly in the report "The ethics of patenting DNA" of the Nuffield Council for Bioethics (July 2002).

Contributions

We welcome articles for future newsletters and these should be sent to the lead editor, Dr Lina Florentin at the following email address: alab@leto.gr

Dr Lina Florentin



For Membership enquiries please contact:

Ms Karen Knob at the Society's administrative office:
European Society of Human Genetics, c/o Vienna Medical Academy
Alser Strasse 4, A-1090 Vienna, Austria
Tel: +43 1405 13 83 22
Fax: +43 1 407 82 74
Website: <http://www.eshg.org>
Email: eshg@eshg.org