



European Society of Human Genetics

December 2002 / Newsletter No. 7

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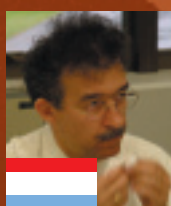
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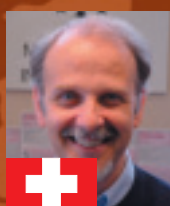
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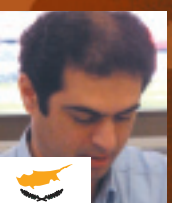
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For further information see the Society's Website: <http://www.eshg.org>



Statement on Gene Patenting

European Society of Human Genetics, 2002

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 ESHG Public and Professional Policy Committee 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).

Election of President & Board Members 2003

The Society Board intends that the President-Elect and two Board Members will again be elected by postal ballot of all the members, rather than by voting in person at the Annual General Meeting, to increase participation of all members. Members of the Society who wish to stand for election are requested to read the job descriptions of the duties of the President and Board Members detailed on the back of the enclosed nomination form. The Statutes of the Society state that the Board should be broadly representative of nationalities and specialties. A list of

current Board members is on the Society's web site (www.eshg.org)

(Term of office ends at the general assembly in the year shown in brackets by board members photographs on front cover.)

The ballot form showing the names of members wishing to stand for election will be sent to eligible voting members in the next mailing and a date given by which it should be returned. The result will be announced at the Annual General Meeting.

Free Listing of European Genetics Clinics in the GeneTests-GeneClinics Clinic Directory

Roberta A Pagon, Medical Director, would like to extend an invitation to members of the ESHG to list your genetics clinic in the GeneTests-GeneClinics Clinic Directory, a voluntary listing of US and international genetics clinics that provide genetic evaluation and genetic counseling. There is no cost to list your clinic. Listings include information on staff genetic board certification, services provided, specialty clinics and appointment scheduling. Links are provided to clinic Web sites as available.

The Web site consists of:

- **GeneReviews (formerly GeneClinics)**
An online publication of expert-authored disease reviews containing descriptions of inherited disorders and the genetic testing used for diagnosis, management, and genetic counselling of patients and families with those conditions.
- **Laboratory Directory**
An online international directory of genetic testing laboratories
- **Clinic Directory**
An online international directory of genetics and prenatal diagnosis clinics
- **Educational Materials**
An introduction to genetic testing and counselling concepts, glossary and teaching tools

For further details contact www.genetests.org

If you have any questions contact Monica Smersh at msmers@chmc.org

Society Website: www.eshg.org

ESHG Education Committee

Progress Report October 2002

Members: ESHG: Celia DeLozier, Rodney Harris, Shirley Hodgson, George Kosztolanyi, Thomas Meitinger
Associates: Heather Skirton (Genetic Counsellor), Antonio Savorelli (for the European Genetics Foundation)

The committee met for the first time in Strasbourg. Three priorities were set for the first year, and work begun on those, as follows:

- Associate the ESHG to some European Community Expressions of Interest with the goal of fostering the formation of networks interested in genetics education, counselling and testing issues. Two such expressions of interest papers were submitted with the ESHG Education committee as a partner:
 - Genetic Testing in Europe - Integrated Network for test development and harmonization of quality of genetic testing services (JJ Cassiman and Rob Elles are among the partners)
 - European Coalition of Health Professionals Education in Genetics: ECHPEG (Rodney Harris and Irma Nippert are among the partners)
- ESHG Web site: add a section of links about genetics education: sites, audio-visual and other material already existing and judged as helpful in individual education or training programs (intended for health care professionals). The Education committee has been collecting such sites. **All ESHG members are asked to contribute sites via an e-mail to Celia DeLozier, before 1st January 2003 (email cddelozier@yahoo.com)**
- Establish an on-line directory of genetics training programs in Europe, along the model of the one used by the American Society. The Committee is working on a one-page form to be sent to heads of all genetics departments in Europe; the document will be sent to ESHG Board members as well, our initial goal being to catalogue programs which are already set up and are open to others (either nationally or internationally). Included would be programs aimed at pre- and post-graduate medical training, genetic counsellors and for laboratory geneticists.

The ESHG continues its partnership with the European Genetics Foundation, which offered 8 genetics courses in 2002, most of them in the new centre in Bertinoro. The ESHG funded five scholarships this year, three to the March Medical Genetics course and 1 each to Cancer Genetics and to Genetic Counselling in Practice (the course in October, had 55 attendees, including 15 from southern Mediterranean countries, funded by the EUROMEDIS grant).

C. DeLozier, 10/2002

ESHG Committee on European Union Affairs

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

The mission of the committee is to liaise between the ESHG and European Institutions or organizations on all aspects of human genetic services or research in Europe, which require a common European approach and to find ways to facilitate efficient and timely solutions for problems identified in these issues.

As a first objective, the committee will actively pursue the recognition of medical genetics as a European medical specialty. For this purpose, contact will be made with the appropriate commission on medical specialties and the document approved by the board will be submitted to the members of this committee as a first step in this process.

A second objective is to revive the sections on Human Genetics of the Fiori report from the ad hoc committee of the European parliament and to bring these again before the EU parliament for approval. In a first approach, a select group of EU MPs will be invited to determine the best strategy to achieve this aim. Based on the outcome of this meeting, further steps will be undertaken to bring this issue to a

satisfactory conclusion. Of course one will have to take into account that elections for parliament are scheduled within the next two years. Support for this strategy has already been obtained from an MP of the Christian democrats and from the biotech industry (Europabio and in particular the European affairs delegate of Smith Kline Glaxo).

A third objective is to continue the close relationship with the European Platform for Patient Organizations, Science and Industry (EPPOSI). S Aymé and JJ Cassiman are board members of this organization and the Professional and Public Policy Committee (PPPC) has obtained the support of EPPOSI for the organization of the two planned workshops on genetic aspects of infertility and pharmacogenomics.

A final and more **general objective** is to follow closely evolutions on the European scene on matters, which might affect Human Genetic practice e.g. the creation of a European platform for Laboratory medicine (ELM), the European Life Scientist Organization etc. and to report to the society when issues of importance for the society emerge.

European Human Genetics Conference 2003

The next ESHG meeting takes place in Birmingham May 3 - 6.

A warm welcome awaits visitors to Birmingham, Britain's friendly second city, located in the heart of England. Birmingham is excellently served by motorways, rail, and an international airport. The conference is being held in the ICC, which is in the centre of the city, with most conference hotels within a few minutes walk.

Join us for an excellent scientific programme in the heart of a vibrant and exciting city. If you can spare a few extra days, visit Shakespeare's birthplace just down the road at Stratford or the Cotswolds or the Peak district. We look forward to seeing you in 2003 - or as a native Brummie would say "Taa Raa a bit!"

Fiona Macdonald

3rd - 6th May 2003

International Convention Centre,
Birmingham, United Kingdom

Scientific Programme

Plenary sessions:

- Low penetrance genes and cancer susceptibility
- Life, sex and WT1
- DNA banks
- Genetic epilepsies
- Congenital muscular dystrophies
- Limb development
- Polydactyly
- Brachydactyly
- Late breaking research

Workshops:

- Syndrome identification
- Cytogenetics population
- Issues in counselling
- Genotyping and mutation detection arrays
- Quality control
- Prenatal cytogenetics

Symposia:

- Bioinformatics
- Stem cells
- Sensory genetics
- SNPs and haplotypes
- Chromosomes in genetic disease
- Genetics and endocrine problems
- Alternative splicing
- Cancer genetics
- Alzheimer disease

Abstract Deadline:

Submission will be via world wide web.
Closing date 13th January 2003

A printed brochure is being mailed to all members of the ESHG and further details are available at www.eshg.org/eshg2003

Birmingham



Society Website: www.eshg.org

European Human Genetics Conference Birmingham 2003 - Exhibitors

"The Trade Exhibition goes from strength to strength"

The European Society of Human Genetics Annual Conference is considered a high profile event by trade exhibitors. Two companies immediately booked for next year's conference after the 2002 European Society of Genetics Conference in Strasbourg.

One of these companies was Progeny who reported, "European Human Genetics Conference has been a wonderful show with great opportunities to meet with prospects as well as existing clients. The show in Strasbourg was a pleasant experience and proved to be a great venue for mixing business with some free time to visit a beautiful city. For our company Progeny Software, providers of pedigree software technology, ESHG has been a great place to showcase the latest enhancements and forthcoming versions of our product to the European market. The opportunity to meet face-to-face with clients and prospects has been truly rewarding and we're looking forward to the show in Birmingham."

The other company to sign-up for Birmingham 2003, was Affymetrix:

"Affymetrix is the market leader in creating breakthrough tools that are driving the genomic revolution. By applying the principles of semiconductor technology to the life sciences, Affymetrix develops and commercializes systems that help scientists alleviate human suffering and improve the quality of life.

Affymetrix offers an expanding portfolio of integrated products and services including a new line of DNA analysis tools. With GeneChip(R) arrays, one can perform highly paralleled interrogation of DNA; enabling rapid, accurate, and cost effective genetic analysis for both resequencing and genotyping applications. CustomSeq(TM) allows resequencing of up to 30Kb in a single experiment. Our next generation mapping array has 10,000 SNPs with an easy to use, scalable assay that does not depend on locus specific PCR. Affymetrix brings the power of whole genome analysis to the bench top.

2003 promises to be a very good year for Affymetrix and we look forward to exhibiting at the European Human Genetics Conference in Birmingham."

List of the Exhibitors booked for the European Human Genetics Conference, Birmingham, 2003.

Affymetrix

Agowa

Amersham Biosciences Benelux

Applied Biosystems

Applied Imaging International

Asper Biotech

Biocomputing Finland

Blackwell Publishing

Bruker Daltonics

Cytocell

ECACC - European Collection of Cell Cultures

Euroclone

Gentra Systems

Genzyme Europe

GMP Companies

Innogenetics

Invitrogen

Metachem Diagnostics

MetaSystems

P.A.L.M. Microlaser Technologies

Progeny Software

Qbiogene

Savyon Diagnostics

Science, c/o GTP Management Services Intl

Syngene

TKT- Transkaryotic Therapies Europe 5S

Transgenomic

Wisepress Bookshop



Membership Renewal Information for 2003

It is now time to ask you to renew your subscription for 2003. You will shortly be receiving a form to renew your membership, showing the different classes available. Please note that if you take out a full (Regular) membership you may have an electronic subscription to the Journal as well as a paper copy. The Journal will not be delivered to you from December 2002 until we receive your renewal form so please do not delay when you receive your renewal request. We are pleased to report that subscription rates have not been increased for 2003 and remain at the same level as 2002.



The Asia-Pacific Society of Human Genetics and the International Federation of Human Genetics Societies

At the occasion of the 5th Asia-Pacific conference and the 4th HUGO pacific meeting on Human Genetics, October 27 - 30 in Pattaya, Thailand, a breakfast meeting took place with some of the founding fathers of this loose network of human geneticists.

The representatives of the Asia-Pacific society were: Sangkot Marzuki (Indonesia), President of the society, Suthat Fucharoen (Thailand), secretary general of the conference, Pornswan Wasant (Thailand), Nizam Isa (Malaysia) and Carmencita Padilla (Philippines).

The network of human geneticists started in 1994 with a 1st Asia-Pacific conference in Bangkok, followed by Djakarta in 1995. In 1996 in Kuala Lumpur the Society was created and the name 'Asia-Pacific conference on Human Genetics' was chosen. Then followed Shanghai in 2000 together with the 3rd HUGO pacific conference. At this occasion a draft constitution (written by G. Sutherland), was approved by a group of geneticists from Australia, Indonesia, Japan, Malaysia, the Philippines, Singapore, Thailand and China. Indian geneticist were interested and wanted to be kept informed. The draft constitution contained the following

elements: membership was for individuals interested in the genetics of the AP region, not for national societies; a board of 10 members would be constituted with a mandate for 5 years and a president, president elect, past president, secretary general and treasurer would be chosen. This draft was not formalized and the society therefore continued to function as a loose network.

At this meeting, the members present decided to officially create the Asia-Pacific Society with Singapore as legal office and a treasurer from that country. The different steps to put the society officially on the map were defined and spelled out clearly.

In conclusion: the Asia-Pacific society has a fairly long tradition, has many outstanding scientists from a large number of countries in the region and is eager to join the IFHGS.

The 6th AP conference and 5th HUGO meeting will be held in Singapore 2004.

JJ Cassiman

News

- Dr Segolene Ayme will represent the ESHG at a Joint Meeting with all interested parties on "A Continuity Policy for Orphan Medicinal Products in the European Union" in December 2002 in London. The invitation was from the European Agency for the Evaluation of Medicinal Products.
- Professor Giovanni Romeo has advised us that the three scholarships awarded by the ESHG for the 15th Course in Medical Genetics have been awarded to:

Cesare Rossi, Italy Vladimir Strelnikov, Russia
Christian Windpassinger, Austria

Nature.com

Nature.com - now the platform for all Nature Publishing Group's journals - including the European Journal of Human Genetics. The content of all NPG's journals is now hosted on the same platform, www.nature.com, strongly enhancing the value of the site. Anyone can now search across the complete range of NPG journals, or search all titles in a particular subject area. The subject areas themselves provide a 'shop window' into the titles Nature Publishing Group publishes in that field, and the articles selected each week reflect the most important papers published. Nature View, on the nature.com home page, highlights weekly the best papers from across the whole of NPG's publishing. There is also a society publishing page with links to both European Journal of Human Genetics and European Society of Human Genetics.

Launch of European Union Framework Programme 6:

Fundamental knowledge and basic tools for Functional Genomics in all organisms

Summary of Programme

- Fundamental knowledge and basic tools for functional genomics in all organisms to foster the basic understanding of genomic information, by developing the knowledge base, tools and resources needed to decipher the function of genes and gene products relevant to human health and to explore their interactions with each other and with their environment.

Research topics:

- Gene expression and proteomics
- Structural genomics
- Comparative genomics and population genetics
- Bioinformatics
- Multidisciplinary functional genomics approaches to basic biological processes

Web Sites for Information

For up to date information on Calls for Proposals, Work Programmes, Guides, etc. :

<http://www.cordis.lu>

For information on Genomics, see Pages 11-12 of Specific Programme for RTD for FP6

http://europa.eu.int/comm/research/fp6/pdf/11385-2_en.pdf

FP6 Genomics and biotechnology for health

<http://www.cordis.lu/rt2002/fp-activities/genomics.htm>

Information: FP6 & Expressions of Interest:

<http://www.cordis.lu/fp6/>

Help and Contact Information

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e-mail: research@cec.eu.int

Society Website: www.eshg.org

New incentives to submit your science to the European Journal of Human Genetics

Papers from our European Journal of Human Genetics are being highlighted on the Nature.com news pages, with topics all indicating the broad scope of the EJHG

- centenarian research, by Tan et al. ¹
- the Usher3 gene, by Adato et al. ²
- night blindness, by Wutz et al.³
- a serum bilirubin genome scan for anti-atherogenic effects, by Kronenberg et al. ⁴
- human handedness, by Van Agtmael et al.⁸

The European Society of Human Genetics and Nature Publishing Group wish to reward very high-impact EJHG papers. We will keep a tab on the yearly citations of research published in EJHG during the preceding year. The first authors of the top three papers will win free ESHG membership for one year, including online and hardcopy EJHG subscription. In addition, the first author of the number one paper will receive a modest financial prize. These awards will be presented to the winners at the annual European Human Genetics Conference - as an additional bonus, their registration fee to this meeting will be waived.

The top 3 papers of 2000, cited in 2001, whose authors will be honoured together with the 2002 winners, at next year's European Human Genetics Conference in Birmingham are

- Gasparini et al. ⁵, about the high frequency of the 35delG GJB2 mutation in DFNB1 deafness (19 citations),
- Dobson-Stone et al.⁶, pitting fluorescent SSCP against DHPLC (17 citations, showing that technology papers can be highly cited, too), and
- Schiller et al. ⁷ describing phenotypic variation and genetic heterogeneity in Leri-Weill Syndrome (also 17 citations).

Our congratulations to these authors, and also our thanks, as they contribute significantly to the impact of the EJHG. We plan to keep the scope of the Journal as broad as human genetics is becoming - mirrored in our subtitle of 'International coverage of all aspects of human genetics'. We have added a few specific categories to which we welcome your contributions: Medical genomics, Statistical and computational genetics and Bioinformatics.

Gertjan B van Ommen - Editor

References:

1. Tan, Q. et al. EJHG, 2002, Vol. 10, pp. 119 - 124
2. Adato A. et al. EJHG, 2002, Vol. 10, pp.339-350
3. Wutz K et al. EJHG, 2002, Vol. 10, pp.449-456
4. Kronenberg F et al. EJHG, 2002, Vol. 10, pp. 539-546
5. Gasparini P et al. EJHG, 2000, Vol 8, pp. 19-23
6. Dobson-Stone et al. EJHG, 2000, Vol 8, pp. 24-32
7. Schiller S et al. EJHG, 2000, Vol 8, pp. 54 -62
8. Van Agtmael et al. EJHG, 2002, Vol 10, pp. 623-630

Journals cited by European Journal of Human Genetics

Top 5 cited articles published in EJHG in each year 1998-2001 (as at 9/10/2002) Papers listed in alphabetical order by author.

YEAR	FIRST AUTHOR	TITLE
1998	Calafell F	Short tandem repeat polymorphism evolution in humans
1998	Devoto M	First-stage autosomal genome screen in extended pedigrees suggests genes predisposing to low bone mineral density on chromosomes 1p, 2p and 4q
1998	Dewalle M	Phenotype-genotype correlation in Jewish patients suffering from familial Mediterranean fever (FMF)
1998	Rozet JM	Spectrum of ABCR gene mutations in autosomal recessive macular dystrophies
1998	Taylor JE	Correlation of SMN1 and SMN2 gene copy number with age of onset and survival in spinal muscular atrophy
1999	Annese V	Genetic analysis in Italian families with inflammatory bowel disease supports linkage to the IBD1 locus - A GISC study
1999	Correa-Cerro L	CAGnCAA and GGN repeats in the human androgen receptor gene are not associated with prostate cancer in a French-German population
1999	De Benedictis G	Gene/longevity association studies at four autosomal loci (REN, THO, PARP, SOD2)
1999	Shohat M	Phenotype-genotype correlation in familial Mediterranean fever: evidence for an association between Met694Val and amyloidosis
1999	Tanke HJ	New strategy for multi-colour fluorescence in situ hybridisation: COBRA: COMbined Binary RAtio labelling
2000	De Bona C	Preserved speech variant is allelic of classic Rett syndrome
2000	Dobson-Stone C	Comparison of fluorescent single-strand conformation polymorphism analysis and denaturing high-performance liquid chromatography for detection of EXT1 and EXT2 mutations in hereditary multiple exostoses
2000	Gasparini P	High carrier frequency of the 35delG deafness mutation in European populations
2000	Mirzayans F	Axenfeld-Rieger syndrome resulting from mutation of the FKHL7 gene on chromosome 6p25
2000	Schiller S	Phenotypic variation and genetic heterogeneity in Leri-Weill syndrome
2001	Cuisset L	Molecular analysis of MVK mutations and enzymatic activity in hyper-IgD and periodic fever syndrome
2001	Horvath S	The family based association test method: strategies for studying general genotype-phenotype associations
2001	Matsumoto N	Mutation analysis of the DCX gene and genotype/phenotype correlation in subcortical band heterotopia
2001	Nielsen JB	MECP2 mutations in Danish patients with Rett syndrome: High frequency of mutations but no consistent correlations with clinical severity or with the X chromosome inactivation pattern
2001	Prince JA	Lack of replication of association findings in complex disease: an analysis of 15 polymorphisms in prior candidate genes for sporadic Alzheimer's disease

These papers encompass a wide range of subject areas, covering topics as diverse as association studies, population genetics, linkage studies and mutation detection.

