October 2008

For members and interested parties

International Genetic Alliance

"seeks a world where genetic conditions are understood, prevented, treated, ameliorated, and cured"

O3

INTRODUCTION

This third newsletter will inform you about events, progress in member countries, publications and projects. It serves to provide information and to facilitate communication and also to encourage emerging alliances. Your contributions for the last Newsletter of this year are welcomed before the 8th of December 2008.

UPCOMING EVENTS

4th International Conference on Birth Defects and Disabilities in Developing Countries, India, New Delhi, 4 – 7 October 2009.
"Translating Research into Care and Prevention."

This conference will cover many aspects of birth defects such as surveillance programs and registries; major risk and preventive factors; individual and community-level prevention strategies, including micronutrient fortification and immunization programs and preconception health and healthcare; care and counseling of common birth defects and genetic disorders; rehabilitation and rights of the disabled and legal frameworks for their protection.

Plenary sessions will present the important advances and successful models of care and prevention in this field. These will be followed by symposia, workshops and free paper sessions. "Meet the Experts" sessions addressing such topics as neural tube defects, oral facial clefts, hemoglobinopathies, muscular dystrophy, haemophilia, mental retardation, cerebral palsy and autism will be organized by the interested parties and presented prior to the Conference. Website: www.icbd2009.com

PROJECTS

The PatientPartner project - Identifying the needs for patients partnering in the clinical trial context.

PatientPartner is a project aiming at enforcing the role of patient organisations in clinical trials. The Dutch Genetic Alliance VSOP, the Genetic Interest Group (GIG, UK), the European Genetic Alliances' Network (EGAN) and the European Forum for Good Clinical Practice (EFGCP) have joined forces as partners in the project.

PatientPartner is based on the belief that involving patient organisations as equal partners at all stages of clinical research, contributes to research that is better adjusted to the real needs of patients. Furthermore, this will contribute to speeding up the research, improving its quality, giving more insight into the possible benefits and disadvantages and finally reaching better healthcare.

The projects' main goals are:
- To make an inventory of the needs of patient organisations regarding their involvement in clinical research.
- To identify and realize common points of action amongst all stakeholders through engaging in an active dialogue with patient organisations.
- To establish an European Network for Patients Partnering in Research (ENPCR) which will support the greater involvement of patient organisations in clinical research.
- The realization of patient centred guiding tools and recommendations on how to create

a successful partnership in the clinical trials context. The inventory will also consist of a survey to identify the current status of patient organisations' involvement in clinical research in Europe. Furthermore, patient organisations are currently asked to supply examples of known best practices of partnerships between patient organisations and the other stakeholders in clinical research, both in relation to the major diseases as well as rare diseases.
The results of this inventory will be discussed in five workshops in different regions of Europe to better understand how patients and patient organisations could be more efficiently partnering in the clinical research process. These workshops will give rise to guidelines and recommendations on the portrayed role of patient organisations in clinical research. In 2011, the recommendations shall be presented to the patient organisations, to European and national policy makers, scientists and the biopharmaceutical industry.

For the success of the project the cooperation of as many European patient organisations as possible is essential for its success. If you want to get involved in the project or have information on (best) practices concerning participation of patient organisations in clinical research, we would love to hear from you. To do so or to obtain more information about the project, feel free to contact Ms. Kim Wever, Project Officer PatientPartner (info@patientpartner-europe.eu).

The project website, www.patientpartner-europe.eu is currently under construction.
PatientPartner is funded by the 7th Framework Programme (FP7) of the European Commission.

The EuroGen guide Project

EuroGenGuide is a three-year project funded by the European Commission for Life Sciences and Biotechnology, as part of the Sixth Framework Programme for Research and Technological Development, and co-ordinated from London by the Genetic Interest Group. It started in January 2007 and will last for three years. The aim of EuroGenGuide is to provide accurate, high-quality information on the Internet about genetic testing and research in Europe, for use by all those living in the continent to whom information about genetics is relevant, whether as a patient, a relative of a patient, or a health professional interested in providing genetic services to their patients.

The EuroGenGuide team is comprised of thirteen different member organisations from around Europe, from across the spectrum of genetics in healthcare. The participants were invited to take part in the project due to the specific areas of professional knowledge and knowledge that they can bring to EuroGenGuide, in order to create material relevant to as many different people as possible. Made up of a diverse mix of patient support groups and doctors from a range of different European countries, you can find out a little about everyone working to develop EuroGenGuide at the team page.

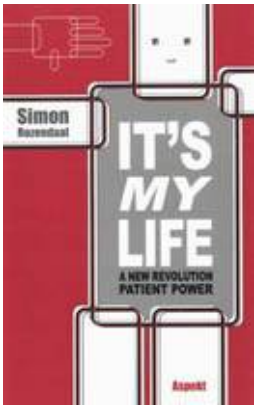
This summer of 2008 the project has reached the halfway stage and the website has gone online for the first time. The core information on the site has been divided into a section for the public and patients, and a section for health professionals.
EuroGenGuide particularly wishes to be able to provide knowledge and information to those regions and countries in Europe where knowledge of genetics and available treatment is still limited. In raising the public visibility and awareness of genetic research and what it may be able to achieve in future, EuroGenGuide hopes to be able to contribute to maintaining the pace of this research and thus ensure that an increasing number of people affected by genetic disease can get the help and treatment they need, wherever in Europe they might be.

The EuroGenGuide website contains information about genetic testing, counseling and research across Europe. The information is intended as a resource for both patients and their families, and health professionals alike. You might be concerned that you or a relative may be affected by a genetic condition, or your doctor may have asked you if you would be prepared to

donate biological samples to a DNA bank for use in research. Alternatively you may be a doctor with a patient who might have a genetic condition and requires a test, or you might be a genetic researcher carrying out a study. If any situations such as this apply to you, then EuroGenGuide can help. Within the website you will be able to find comprehensive information for patients and the public, and also guidelines for health professionals.
You can find EuroGenGuide at the following websites: www.eurogenguide.eu
and www.eurogenguide.org.uk

IT'S MY LIFE

The book 'It's My Life' portrays a revolution that is taking place now! Twelve patients with a rare hereditary disorder all over the world have been interviewed. Their stories show a determination to take charge, to make a real difference. They want to get involved in the process of decision making. About medicines and treatments. It's their body, it's their life. These people embody a new energy a new promise to change medical science, the pharmaceutical industry and health care. We much welcome your assistance and support to advertise for this special book and to inspire other patients and patient organisations. This book can be ordered at any local bookstore. Copies ordered via the **European Genetics Alliances' Network** (EGAN) will bring a donation for each copy towards patient support activities. Your order can be send to egan@egan.eu
The price for each book will be € 15 (± US $ 20) excluding shipping costs.



PUBLICATIONS AND REPORTS

IGA Executive Committee meeting at the ESHG / EMPAG AGM, May 31 – June 4 2008, Barcelona, Spain

t the occasion of the AGM's of the European Society of Human Genetics (ESHG) and the European Meeting Psychosocial Aspects of Genetics (EMPAG), IGA had its executive committee meeting in the International Convention Centre in Barcelona. We discussed the implications of the fast expanding alliance especially in the developing countries, the options for a virtual patient academy and for a global multi media awareness marathon. Moreover the procedures for voicing the interests of the members and the presenting statements on topics were subject of exchange of ideas. Other subjects were the management of the extensive IGA network, the various major projects, better visibility, programs for the various coming events and internal communication. For reports about the AGM's see: www.eshg.org/eshg2008

BIO, June 17 – 20 2008, San Diego, USA

BIO is the global event for biotechnology. Over 20.000 people engaged in biotechnology attended this most impressive event. IGA participated in the Patient & Health Advocacy Display. Patrick and Sharon Terry of the Genetic Alliance were speakers in different sessions.

The IGA Display (left) at the BIO global event in San Diego

1st Central and Eastern European Summit on Preconception Health and Prevention of Birth Defects, August 27 – 30 2008, Budapest, Hungary

The Research and Prevention for Families Foundation (RPFF) in Hungary together with the Hungarian Congenital Abnormality Registry organised an impressive and successful summit with internationally well-

Dr. Melinda Medgyaszai, Secretary of State for Health Policy and Dr. Ferenc Falus, Chief Medical Office of the Republic of Hungary.

known speakers and chaired by Dr. Csaba Siffel in Budapest, Hungary. Dr. Melinda Medgyaszai representative of the Ministry of Health in Hungary was the main patron, highlighting the importance of the summit. Participants from 21 countries including 10 Central and Eastern European countries reviewed and discussed the promotion of women's health before, during and beyond pregnancy, and the role of preconception health and health care in the prevention of birth defects in the Central and Eastern European region.

Folic acid was one of the key topics of the summit. Besides plenary sessions, several breakout sessions were held including one on the role of NGOs and networks in prevention of birth defects and genetic conditions. There was also a workshop organized to discuss the EuroGenGuide project. Susan Szendro (RPFF) coordinated the initial step to establish the Hungarian Genetic Network during the summit.

Dr. Adolfo Correa (right) congratulates the conference chair Dr. Csaba Siffel (left) with the successful conference.

During the small group discussions participants discussed: 1) what countries of the region should do to promote preconception health and prevention of birth defects, how to monitor improvements, and what networks, communications and resources are needed; 2) the current situation in countries of the region; and 3) what needs to be done (recommendations, guidelines, implementations).

The summit was co-sponsored by the Centers for Disease Control and Prevention (USA), University of South Alabama (USA), March of Dimes Foundation (USA); and the Ministry of Health, the National Public Health and Medical Officer Service, and the National Institute of Child Health in Hungary. The WAO, IGA, EGAN, and CEEGN were among the several other international, European, and national partners of the summit. The organizing committee is currently working on recommendations to be published from this Summit.
By: Susan Szendrö and Dr. Csaba Siffel

European Health Forum Gastein (EHFG), October 1 - 4 2008, Hofgastein, Austria is the most important health policy event in the European Union. Leading experts from business and industry, science and academia, patient organisations/NGOs as well as numerous prominent decision makers in health policy presented new ideas and use the EHFG as a platform for the exchange of experiences and opinions at the international level. A major feature of the European Health Forum is its broad range of topics: issues such as the improvement of local health infrastructure just as current international challenges such as increasing migration within the health professions. Thus, the EHFG is a meeting place for experts from different sections of the health system and is fundamentally different from events which are specialised in highly restricted areas. This year the conference took place from 1 – 4 October 2008. At the beginning of this conference the think tank of the Thrombosis Research International (TRI) also met. Alastair Kent, Ysbrand Poortman and Rod Mitchell participated in this high level multi stakeholder meeting. This meeting was organised in collaboration with the International Alliance Patient Organizations (IAPO). Reason was the burden of disease caused by thrombo embolism (TE). TE is highly undervalued and so are the many options for prevention. In Europe 400.000 deaths occur as a result of TE. It is the second common medical complication. So far TE is related to post surgery. There are no data about the prevalence of TE in home bound chronic patients The genetic risks were tabled and discussed and that what needs to be done in health care to prevent TE.

EuroBio, 6 - 9 October 2008, Paris, France
EuroBio 2008 aims at "bridging the gap between the pro-innovation rhetoric and the reality of the struggle to create and maintain life science businesses". This major event motto was: "Make change happen". It attracted about 5000 people, including delegates and visitors from the whole of Europe, from Asia and United States. EuroBiO has increased its political importance and the congress has become a recognized European Presidency Event.

Alastair Kent of the European Genetic Alliance (EGAN) spoke in the plenary health session on the future of medicine and had the lead in a plenary session called “House of Commons” debate. Ysbrand Poortman and Alastair Kent are members of the EuroBio International Steering Committee. For more information please look at: www.eurobio2008.com/steering_committee.php

Dr. Françoise Barré-Sinoussi received the Nobel Prize for Medicine 2008 for her discovery of the human immunodeficiency virus. Immediately after this good news she visited the plenary health session at EuroBio. Alastair Kent was on the panel and congratulated Dr. Barré-Sinoussi on behalf of IGA & EGAN.

Dr. Françoise Barré-Sinoussi (right) is being congratulated with her Nobel Prize by Alastair Kent at EuroBio.

Series of brochures on genetics

EuroGentest has recently developed a series of information leaflets for patients and families about genetics and genetic testing. They are available in numerous European and ethnic minority languages. They are freely available to download from www.eurogentest.org/patients

The titles that are currently available are:
- Amniocentesis Test
- Chromosome Translocations
- Chromosome Changes
- Chorionic Villus Sampling (CVS) Test
- Dominant Inheritance
- Frequently Asked Questions
- Genetic Glossary
- Recessive Inheritance
- Some Information About Your Genetic Appointment *(to be provided before the appointment)*
- What is a Genetic Test?
- X Linked Inheritance

The EGAN/IGA & Roche Collaboration has lead to various outcomes and one of them is a series of brochures about subjects related to patient involvement in clinical trials. More and more patients get involved in clinical trials and would like to have more detailed information on what clinical research is all about, why and how to contribute. These user-friendly brochures are free available and can be downloaded from IGA’s website:
http://www.internationalgeneticalliance.org/english/View.asp?x=1432

Titles currently available are:
- Frequent asked questions
- Glossary of terms
- Biobanks *(in production)*

INSIDE INFORMATION FROM MEMBERS

Ms Zhu Changqing representative of IGA and MDA China delivers the Paralympics torch relay

Ms. Zhu Changqing represented China on various IGA events and she is founder of the MDA China. The purpose of MDA China is to improve the quality of life, diagnostics, treatments, research and the prevention of neuromuscular diseases. These diseases are one of the five most common death-threatening diseases according to the WHO. Examples of neuromuscular diseases that are life threatening are Amyotrophic Lateral Sclerosis (ALS), Duchenne Muscular Dystrophy and Spinal Muscular Atrophy (SMA). In China about 4,500.000 million patients are diagnosed with a neuromuscular

Torchbearer Wang Seng lights the torch of Zhu Changqing during the Beijng 2008 Paralympics torch relay in Changhai (Xinhua / Xu Liang)

disease. Ms Zhu Changqing also has a neuromuscular disease and is associate professor of Shanghai Polytechnic University. She has devoted herself to the improvement of patients lives and raising awareness in society. MDA China has been honoured with several awards. Among them the 'Innovation Fund Award 2008' of NIKE. On September 1st, Zhu Changqing was the 17th deliverer in the Paralympics torch delivering in Shanghai. She kept holding the torch using a stand made for her on her electronic wheelchair. She told the reporter of a Chinese Women's paper: 'The most meaningful thing is to encourage people with neuromuscular diseases and let people know this group. They can overcome themselves and participate in society. We all can get something when we really take part in it. We do not need sympathy, but we just need proper help and understanding. The most important is to be equal with other people'.

Zhu Changqing carrying the torch

Genetics in healthcare system Brazil

There is a huge gap between theory and practice. The Brazilian Constitution states that health is a right of all and that the state has the duty to provide health services. Is SUS – Sistema Único de Saúde (Unified Health System), the healthcare system in Brazil, ready to provide healthcare for all? No. And when it comes to genetics, the mission to offer health services becomes even more challenging. Definitely, SUS authorities are not paying attention to genetics. Genetic counseling? Genetic tests? Treatments for rare diseases? It was once in 2004 that the Ministry of Health signalled to collaborate to implement a genetic public policy project. A committee was formed with representatives from the government and from the Brazilian Society of Medical Genetics to elaborate a proposal of "Public Policy and Clinical Genetics". A few months later the PPCG proposal was handed to the Ministry of Health but, believe it or not, nothing has happened since then. This is a precarious situation since 85% of the population is totally dependent upon SUS, which means that, roughly speaking, 150,000,000 people are directly affected either by genetic public policies or by the lack of genetic public policies. Now, willing to find a reasonable way to grant our rights, and to accelerate the process of turning theory into practice, the Brazilian Society of Medical Genetics (BSMG), in conjunction with the Brazilian Genetic Alliance (BGA), decided to make people acutely aware of the existence of a PPCG proposal that has been stuck in the Ministry of Health for exactly 4 years. On October 28, BSMG & BGA will hold a public nationwide act to mobilize public opinion to pressure the government to consider the PPCG proposal of inclusion of genetics in SUS. Having the proposal considered and accepted will be the first step to bridge the huge gap between theory and practice in the healthcare system.

By: Martha Carvalho, Brazilian Genetic Alliance, www.abg.org.br
Email: abg@abg.org.br

Genetic Alliance of the USA

New events in Genetics and Health—USA

In the IGA Newsletter of Q2 we informed you that on May 21, 2008, US President George W. Bush signed the Genetic Information Non-discrimination Act (GINA) into law.

Genetic Alliance continues to celebrate this monumental event as the culmination of thirteen years of perseverance and dedication from the entire genetics community, led by the Coalition for Genetic Fairness, and more than 500 Congressional offices on Capitol Hill. As the first civil rights legislation since the new millennium, GINA provides every individual in the nation with federal protection against genetic discrimination in both health insurance and employment. "Now that GINA has been

approved and signed into federal law by the President, American healthcare consumers and employees will no longer have to fear the adverse effects of being tested to

US President Bush signing GINA into law

determine their risk status for genetic diseases," said Joann Boughman, Ph.D., Executive Vice President of the American Society of Human Genetics and a member of the Coalition for Genetic Fairness executive committee. "Once this legislation has taken effect, clinicians will be able to order genetic tests for patients and their families in a manner that ensures the full realization of the advantages of personalized medicine, while also easing patients' concerns about the risk of discrimination by insurance companies and employers based on this data." "With this assurance, the promise of genetic testing and disease management and prevention can be realized more fully," stated Sharon Terry, President of the Coalition and CEO of Genetic Alliance. "It is now our responsibility to make sure the public knows that these new protections are in place." In addition to a main priority of providing resources and education on the impact of the legislation, Genetic Alliance will play an active role in the formation of regulations between GINA's passage and the full implementation of the law in November 2009. The Genetic Information Non-discrimination Act will usher in a new era for healthcare, one where the obstacles and limitations to the proactive management of health are eliminated. Every individual in the nation will benefit from the protections afforded by GINA.

To prepare Americans for GINA the Coalition of Genetic Fairness launched a guide at: www.geneticfairness.org/ginaresource.html Here everyone can learn about history of legislation, hypothetical situations of genetic discrimination, and key examples and definitions. This interactive guide includes information for the general public, clinicians, health care providers, employers, health insurers, researchers and state officials.
By: Andria Cornell, Public Policy Program Assistant and Sharon F. Terry, President and CEO, Genetic Alliance.

BECOME A MEMBER OF

The international multidisciplinary community genetics network. It's aim is to facilitate communication between all those working in the field of community genetics (and genomics). To become a member you only need to send an e-mail mentioning this desire to commgennet@gmail.com As a member you are invited to stimulate your colleagues to become a member, or to send me the e-mail address(es) of (the) person(s) you propose.

Policy Bulletin of Genetic Alliance.
The public policy department at Genetic Alliance keeps a close eye on international, federal, and regional policy news. As debates are conducted and votes are held, we keep our stakeholders informed via the weekly GA Policy Bulletin sent to our MemberForum listserv. To subscribe to MemberForum, send an email to membership@geneticalliance.org

Genetic Alliance Weekly Bulletin.
To subscribe, send an email to membership@geneticalliance.org

International Alliance of Patients' Organisations (IAPO) is a unique global alliance representing patients of all nationalities across all disease areas and promoting patient-centred health care around the world. If you like to receive IAPO's newsletter, please go to this website: http://www.patientsorganizations.org/index.pl?n=210;section=3 to submit your request.

RELEVANT WEBSITES

ORGANISATIONS

BIO – a biotechnology industry organisation whose members are involved in research and development of innovative healthcare
Website: www.bio.org/healthcare/issues/

EGAN – focuses on genetics, genomics and medical biotechnology and their implications.
Website: www.egan.eu

International Alliance Patient Organizations (IAPO) - global alliance

representing patients worldwide across all disease areas
Website: www.patientsorganizations.org

International Genetic Alliance (IGA)
Website:
www.internationalgeneticalliance.org

March of Dimes – is an organisation for pregnancy and baby health
Website: www.marchofdimes.com

UNESCO – gives information on life sciences and the implications on all species.
Website: www.unesco.org/shs/bioethics

WAO – for the prevention and treatment of genetic and congenital conditions.
Website: www.world-alliance.org *(under reconstruction)*
WHO – its Genomic Resource Centre provides information and raises awareness on human genetics and human genomics.
Website: www.who.int/genomics/en

MAJOR PROJECTS with International dimension and genetic alliances' engagement:

CAPABILITY – Capacity Building for the Transfer of Genetic Knowledge into Practice and Prevention
Website: www.capabilitynet.eu

PATIENTPARTNER – to promote the role of organisations in the clinical trials context.
Website: www.patientpartner-europe.eu

EuroGenGuide – contains information about genetic testing, counseling and research across Europe.
Website: www.eurogenguide.eu

EuroGenTest – harmonizing genetic testing across Europe.
Website: www.eurogentest.org

Human Genome Project Information – informs about project facts, education, medicine and the new genetics.
Website: www.genomics.energy.gov

RARE DISORDERS:

Orphanet – the portal for rare diseases and orphan drugs
Website: www.orpha.net

International Birth Defects Information Systems (IBIS) – to promote better care and prevention of birth defects through information sharing
Website: www.ibis-birthdefects.org

IGA MEMBERS:

Europe: European Genetic Alliances' Network (EGAN), Website: www.egan.eu
Central & Eastern Europe: Central & Eastern European Genetic Network (CEE GN), Website: www.ceegn.org
Asia: Indian Genetic Society
Africa: Southern African Inherited Disorders Association (SAIDA), Website: www.saida.org
Latin America: Brazilian Genetic Alliance, Website: www.abg.org.br
North America & International: Genetic Alliance, Website: www.geneticalliance.org
Middle East: Iranian Genetic Alliance of Parent/Patient Organisations & Gulf State Alliance
Australia and New Zealand: Association of Genetic Support of Australasia (AGSA), Website: www.agsa-geneticsupport.org.au & New Zealand Organisation for Rare Disorders (NZORD), Website: www.nzord.org.nz

UPCOMING INTERNATIONAL EVENTS with IGA - involvement and partnering

25 - 29 January 2008, Prague, Czech Republic
Meetings of the European Forum for Good Clinical Practice; Central & Eastern European Genetic Network; European Genetic Alliances Network; Patients Roadmap to Treatment

8 – 11 March 2009, Lyon, France
BioVision 2009, Life Sciences as a Vector for Change and Sustainability

4 – 7 October 2009, New Delhi, India
4th International Congress Birth Defects & Disabilities in a Developing World (ICBDD – DW)

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