International Genetic Alliance

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

May 2008

For members and interested parties

O2

INTRODUCTION

This second 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. You will find reports of activities of young member organisations and from senior alliances. Your contributions for the 3rd newsletter will be welcome before 25th of August 2008.

UPCOMING EVENTS

Cluster of annual and EU project meetings in Barcelona, Spain, Barcelona, 31 May – 2 June 2008.
The European Genetic Alliances Network (EGAN) has its assembly in Barcelona in conjunction with the annual congress of the European Society of Human Genetics. The International Genetic Alliance and the World Alliance of Organisations for Prevention and Treatment of Genetic and Congenital Conditions also have their annual meetings there. One of the meetings of EGAN concerns the participation in major European Projects. Recently the Dutch Genetic Alliance (VSOP) in collaboration with the UK Genetic Alliance (GIG) and the European Forum Good Clinical Practice (EFGCP) achieved a major grant covering the costs of the project "Patient Partner in Clinical Research". EGAN already participates in and contributes to the EuroGenGuide Project (Genetic Interest Group), EuroGenTest (University of Louvain), Capability (University of Münster and World Alliance). EGAN has also started a collaboration with the European Forum of Good Clinical Practice (EFGCP) to facilitate the contribution of patients groups to the research and development process. The EFGCP has much professional expertise and a broad network. EGAN works already for some time with various scientific associations and major industries on specific subjects on the road to treatment. These collaborations have already led to various publications such as a glossary of terms, frequent asked questions and answers, workshops on biobanks and animal research. A patient manual and a special website are some of the various plans. In Barcelona, 2nd of June the EGAN/ EFGCP working party will meet together with members of the board of EGAN and IGA.

BIO International Convention / IGA, USA, San Diego, 17 - 20 June 2008
BIO is the global event for biotechnology (www.bio2008.org). Biotechnology is one of IGA's constitutional areas of interest; another area is multi stakeholder collaboration. That's why BIO / San Diego is considered as a great opportunity to promote IGA's objectives. A consortium of industries (a.o. Amgen, Genzyme Corporation, Human Genome Sciences) has organised a "patient advocacy and health display" (on ground floor F) during BIO. Genzyme enabled IGA to participate in this. About 40 patient organisations will present. The Genetic Alliance based in Washington will be there as well. The IGA Executive Committee meeting – originally scheduled in San Diego has moved to Barcelona and will take place the 2nd of June.

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

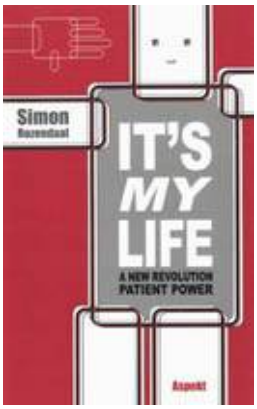
This Summit will be hosted by the Research and Prevention for Families Foundation and the Hungarian Congenital Abnormality Registry. The aim is to provide a platform for review, analysis and discussion of the

promotion of women's health before, during and beyond pregnancy, the role of preconception health and health care in the prevention of birth defects in the Central Eastern European region. For more information see: http://www.diamond-congress.hu/birthdefects2008/

PROJECT

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

Oman and genetics

The first National Workshop took place at the conference on The Prevention of Birth Defects and Genetic Conditions in Muscat in 2007. Support and consultation was provided by the World Alliance of Organizations for Prevention and Treatment of Genetic and Congenital Conditions (WAO) represented by Prof. Irma Nippert and Mr Ysbrand Poortman. Since then the support and assistance in technology transfer was facilitated by WAO allies to ensure successful implementation of the plan to develop comprehensive Genetic Services in The Sultanate of Oman. Two meetings were held with the Minister of Health who showed great interest and involvement in genetics, its

Prof. Giordano from Leiden University, Dr Anna Rajab, Prof. Irma Nippert from WAO and Mr. Ysbrand Poortman from IGA speaking with the Minister of Health of Oman.

opportunities and implications. There were also various meetings with patient/family groups and expected is that they will soon play their role in the international genetic community. Patient support groups have achieved a recognized place in the Oman society. On a visit to the Sultanate of Oman

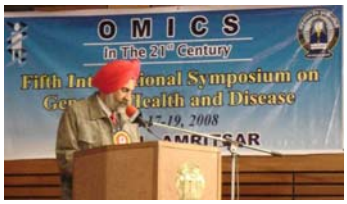
In front of the Dutch embassy in Oman. IGA and WAO board members support Oman to increase genetic services. Dr Anna Rajab (in white), is the driving force in Oman and of great help for the many families involved in genetic disorders and for family support groups.

of the President of the WAO and the Secretary General of the IGA, the Dutch ambassador hosted a dinner for the stake-

holders at her residence on 11th February. Prof. Piero Giordano and Mr Sander Kneppers from the department of Human Genetics and Haematology from Leiden University Medical Centre were honoured guests because of their support and advice to the fast developing genetic networks in Oman. Prof. Irmgard Nippert presented the outcome of a Report from the workshop the Ministry of Health of Oman organised in collaboration with the WAO in Muscat last year. Dr Anna Rajab from the Ministry of Health in Muscat is the driving force of the development of genetic services in Oman. Dr Rajab had organized various workshops and conferences to contribute to building competent genetic services meeting the needs of the families involved.

Amritsar - 5th International Symposium on Genetics, Health and Disease

Prof. Jai Rup Singh and his group of the Guru Nanak Dev University had organized an impressive and very well attended congress in Amritsar. Many countries from all over the world were represented. The prominent participation of young people was remarkable and promising for the future. The congress very well met its target: to share knowledge, to facilitate interactions between the various disciplines and to build collaborations the world over.



Prof. Jai Rup Singh presenting at the well attended congress in Amritsar

There was also a joint IGA / Indian Genetic Alliance session on the subject "International Networking in the OMICS age". Experts by experience reported about their activities in the field of mental disabilities, autism and breast cancer. Memorable was the presentation of Mr Akhil Mehta who has Down syndrome. His performance included a musical underpinning of his message. Mr Chawala gave an impressive presentation on his activities for prevention of breast cancer by public awareness. The Indian Genetic Alliance is in the process of innovation. Prof. Jai Rup Singh is president and Mr Jagdish Mehta the coordinator. Mr Mehta is the former secretary general of Paviaar, the national foundation of parents for Down syndrome, cerebral palsy and autism. Some other active organisations are the Indian Muscular Dystrophy Association and the Innocent World Charitable Society. The alliance had been active and successful in convincing the Indian Ministry of Social Affairs to from a National Trust to provide structurally appropriate social security and other welfare measures for people involved. Unfortunately, in spite of good intentions of the Government of India other pressing problems facing the overpopulated country has taken away the major portion of revenue leaving behind these categories who lack any lobby, force or votes to influence the politicians of the day. Therefore most of cherished dreams of patients/parents such as independent living for their wards, providing for social security and mainstreaming with normal society and most important the eternal question for us as parents "Who cares after us?" remain unfulfilled. There is still much to do for the parents in India.

Mr. Ysbrand Poortman and Prof. Irma Nippert at the IGA/ Indian Genetic Alliance meeting

INSIDE INFORMATION FROM MEMBERS

Saida in South Africa

In developing countries, up to 8% of children are born with a birth defect, many of which lead to disability or death. And many are completely preventable. The South African Inherited Disorders Associations (Saida) is one of very few bodies that extends the reach of primary genetic healthcare in Southern Africa. We see greatest responsibility as being among those who are beyond the reach of even the most rudimentary healthcare. In South Africa we inherited a great disparity in healthcare services. We have skilled health workers and technologically advanced hospitals. But these are those – particularly in the rural areas – who have no clinic nearby. So while we may lobby medical aids to pay for expensive drugs and treatments, most of our energy goes into extending

Jo- Anne Richards, secretary Saida

primary genetic care into the most far-flung areas of the country. This primary healthcare system carries the burden of diagnosing, basic treatment, advising, counselling and preventing. To aid this process, we have written a handbook for training community and parent councillors - in language suitable for anyone, from rural communities with little access to further education, to tertiary-level healthcare councillors. We have also translated information pamphlets on a specific genetic disorder into eight different languages. In each language there were different cultural sensitivities, formal and informal speech patterns and regional variations to consider. We are also writing a comprehensive genetic textbook - the first of its kind in Africa - written in the simplest possible language. It's aimed at doctors, nurses and primary health care practitioners, whose first language is not English. This is a major difficulty for training. Saida is made up of all the support groups that exist for inherited and genetic disorders, the Department of Human Genetics at the University of the Witwatersrand, and representatives from the Department of Health. We work closely with the Department, which recognises our work, but lacks the funds and manpower to undertake it themselves.

Brazilian Genetic Alliance

The Brazilian Genetic Alliance (BGA) is a non-profit organisation founded in 2005, with the aim of bringing together advocacy organisations to better support those with genetic diseases and their families. Working as an umbrella organisation BGA has main focus on spreading the word about genetic conditions, identifying solutions to emerging problems, and making genetic support groups stronger in Brazil. As a middle - income country, Brazil strives for greater efficiency in education and health care, and its regions vary in their capacities in medical genetics. Some may not have the resources to set up appropriate genetic services, and most of the time there is a lack of genetic centres. Meanwhile, others provide excellent genetic services but need assistance to improve equality of access to their services. As a result, diagnoses of genetic conditions are often delayed, preventing the possibility of early intervention services and family planning. BGA is in contact with organisations from all over the country providing them useful sources of information on genetics and on advocacy. These contacts have led to productive discussions about the latest issues in genetics. BGA serves as a contact centre for genetic advocacy organisations. 31 organisations had joined BGA, which means that more than 600,000 individuals affected by genetic diseases have had their voices leveraged in Brazil. BGA's surveys provide guidance to move forward with a better sense of what the strength, the weakness, the needs, and the priorities of affected families are.

Martha Carvalho, President Brazilian Genetic Alliance and IGA board member

BGA is engaged in promoting public and professional awareness regarding a wide range of topics, from educational issues to fair access to health services, medications and genetic counselling. BGA has given lectures in a variety of meetings, interviews to newspapers and TV broadcast, and has extensively distributed pamphlets and brochures from affiliated organisations. BGA wrote a chapter on patients and parents organisations in a medical reference book and held a well attended meeting in conjunction with IGA in Rio de Janeiro.

BGA has always been supported by donations from its board members and never received money from the Brazilian government. During almost two years BGA was stuck on bureaucracy struggling to get documents and registers that are crucial for fundraising in Brazil. With registered documents and official status, it will be possible for BGA to apply for grants, to establish partnerships and to obtain financial support from stakeholders. BGA is currently seeking funding partners to implement a minimum structure to build up a better organisation. BGA needs and welcomes partners!

Malaysian Rare Disorder Society (MRDS)

Malaysian Rare Disorders Society

Founded in 2004, the Malaysian Rare Disorders Society (MRDS) began as an informal group for families with rare genetic and metabolic conditions treated at the

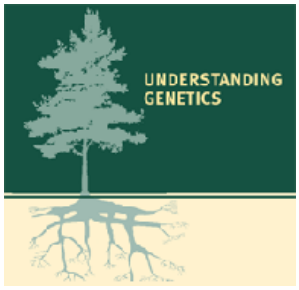
Genetics Unit of the Department of Paediatrics at the University of Malaya Medical Centre in Kuala Lumpur. In 2007, MRDS was registered as a non-profit organisation that acts as a support network for individuals and families affected with rare disorders in Malaysia. MRDS has almost 30 families registered. The main objectives of MRDS are to create a network among individuals and families with rare disorders, to serve as a contact and resource centre and to promote awareness on rare disorders among the Malaysian community. Our past activities included organizing a Family Day for MRDS members, 1st MRDS Annual General Meeting together with talks presented by invited speakers on special education and disability benefits. As a new support group, we look forward to network with other rare disorders societies to learn the ropes and exchange ideas. Mrs Dato' Hatijah Ayob is the president. She is assisted by her committee members. Professor Dr. Thong Meow-Keong, a clinical geneticist who proposed the setting up of the society, is the advisor of MRDS. www.mrds.org.my

Genetic Alliance of the USA

For over 20 years, Genetic Alliance has focused on "what matters": health. To this end, we have developed several tools and resources available via our website: www.geneticalliance.org

Genetic Alliance provides the community with several e-communications, including *Advocacy in Genetics.* This quarterly e-newsletter features upcoming events, spotlights organisations, and keeps our stakeholders informed about legislation before Congress. Through our *Weekly Bulletin,* Genetic Alliance keeps the genetics and advocacy community up-to-date on events and activities. In addition, Genetic Alliance keeps a close eye on international, federal and regional policy news, highlights of which are published in our weekly *Policy Bulletin*. As debates are conducted, votes held and strategic events occur, we keep our stakeholders Informed via these newsletters, sent through our *Member Forum* and *Announcements* listservs. Beyond our e-communications, we exchange information, create discussion, and engage communities through our inclusive *Webinar Series*. *Webinars* focus on three categories – Strategies for Success, Meet your Neighbours, and Hot Topics. At our website you can view current and past issues of *Advocacy in Genetics, Weekly Bulletins and* upcoming sessions of the *Webinar Series.* Genetic Alliance also provides several other tools directly through our website like: *Disease InfoSearch*, a constantly evolving online search tool and database of advocacy organisations and resources for genetic conditions. *Understanding Genetics: A Guide for Patients and Health Professionals* covers basic genetics concepts and in-depth information about receiving a diagnosis of a genetic condition, newborn screening, family health gathering, genetic counselling, genetic testing and its applications. The *Resource Repository* is a robust document repository that aggregates the combined resources of advocates, healthcare professionals, government agencies, think tanks and other contributors. These resources cover a wide range of topics such as fundraising, FDA genetic testing guidance's, advocacy at the state and federal level as well as media strategies for organisations.

Sharon Terry, President Genetic Alliance



A Guide for Patients and Health Professionals on Genetics

WikiAdvocacy and *WikiGenetics* are public online encyclopaedias. *WikiAdvocacy* (www.wikiadvocacy.org) is a compilation of the wisdom of the advocacy community, who continually add and refine the tips and tools offered through this resource. W*ikiGenetics (*www.wikigenetics.org) is an open source, user generated encyclopaedia on human genetics information. To ensure credibility, *WikiGenetics* requires references for all contributions and is reviewed by advisory and editorial boards comprised of experts in genetics. Genetic Alliance extends the reach of our successful resources by translating materials, such as *Understanding Genetics: A Guide for Patients and Professionals*, both linguistically and culturally. As countries across North Africa and the Middle East quickly strive to strengthen newborn screening programs, Genetic Alliance hopes to begin translating materials into Arabic in 2008.

NEWS

NZORD: Impact of smoking for rare diseases

The impact of smoking on the health of the population is huge. It's one of the most common avoidable causes of health problems in society. A recent released Public Health Bill in New Zealand seeks strategies for reducing non-communicable diseases, such as cardiovascular disease, cancer, respiratory disease, diabetes and others. The New Zealand Organisation for Rare Diseases (NZORD) supports this initiative, but also notes that smoking is the number one contributor to these kind of diseases and on the total cost to the health sector and society. In 2003 the National Health Committee produced a report on Molecular Genetic Testing that concluded that the services were not well sourced and well short of international standards. The Committee recommended an urgent upgrade of these genetic services that provide a crucial basic necessity for diagnosis and treatment of rare diseases. Unfortunately this report wasn't implemented by the Ministry of Health because these genetic services were not a priority and preventable diseases caused by smoking are. This demonstrates the disadvantage to NZORD's interests, as a large sum of the Health budget goes to these (preventable) types of diseases. Because of lack of priority investments such as newborn metabolic screening are lagged behind. Opportunities to prevent or treat some rare diseases have therefore been lost in New Zealand. In a country like New Zealand with a tightly budgeted health system, reduction in tobacco driven diseases has the potential to free up significant resources and provide a realistic opportunity for rare diseases to get a more equitable share of health services and research. www.nzord.org.nz

How we convinced the Alberta Government in Canada to pay for treatment in a rare genetic disease

Linda Paré: My son Trevor has Pompe disease, a rare genetic disease of the muscles. He participated in a clinical study to show efficacy of enzyme replacement therapy. Trevor did well on this treatment and improved. We started to get our Government to fund this treatment about five years ago when we learned that once the clinical trial was over, Trevor could only keep getting the treatment when it would be reimbursed. As a start we got to know as many politicians as we could possibly know. Members of Parliament are all elected, so they will listen to the people if you have enough people to support you. We started this battle with a letter writing campaign and asked everyone to fan it out to all their friends. If we heard that one of the politicians was going to be at the mall or picnic we were there. I gave them information on Pompe Disease and ask for their help. We took pictures, with their permission of course and always sent out a thank you with one of the pictures attached, so they would remember us. One of Trevor's school teachers knew someone that worked for Canadian television (CTV) and we knew someone at our local paper. This is how we got our story heard. If someone wanted to give money I asked them to write a letter instead to our Premier, The Health Minister, and as many politicians that they knew. When the story was first aired on CTV the other TV stations all contacted us. We told our story to any media that would listen and we never quit. A key factor was the support of Trevor's doctors they kept writing letters and filling out what ever form came their way. A lady in Edmonton read our story in the newspaper and started a face book group to help Trevor. Another lady started an online petition. We had support from all over the world. The people that heard or read Trevor's story became Trevor's supporters. When we were told "NO" that meant we needed to fight harder. We never took no for an answer. When I received letters from our government that said, "They did not have time to meet with me" I let the public know and they stepped in to help me fight harder. This was my baby's life. We kept in touch with every media person that we were in contact with.

Trevor Paré is one of the 13 Canadians with the rare, genetic Pompe disease.

On Mother's Day (May 11th 2008) the Minister of Health of the state of Alberta in Canada called me telling me that the Alberta government will fund the treatment for Trevor through the Blue Cross Plan. This was the best Mother's Day gift I could get. I still don't know what to say.
For more information look at: www.pompecanada.com and www.cord.ca

President Bush signs Genetic Nondiscrimination Information Act into Law

On May 21, President George W. Bush signed into law the first civil rights legislation of the new millennium - the Genetic Information Nondiscrimination Act (GINA). GINA provides protections against discrimination based on an individual's genetic information in both the health insurance and employment settings. Representative Louise Slaughter (D-NY) says: "Since we all are predisposed to at least a few genetic-based disorders, we are all potential victims of genetic discrimination". Sharon Terry, president of the Coalition of Genetic Fairness and Genetic Alliance, stated: "Individuals no longer have to worry about being discriminated against on basis of their genetic information, and with this assurance, genetic testing, disease management and prevention can be realised more fully". The health insurance provisions of the law will take effect in 12 months and the employment protections will take effect in 18 months. See: www.geneticfairness.org

RELEVANT WEBSITES

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

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

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

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

IGA – International Genetic Alliance
Website: *under construction*

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

UPCOMING EVENTS

31 May – 02 June 2008 Barcelona, Spain
ESHG/EMPAG, EGAN board, EGAN EU projects, Board meetings IGA & WAO

17 – 20 June 2008 San Diego, USA
BIO Conference

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

07 – 10 October 2008 Paris, France
EuroBio

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

October 2009 New Delhi, India
4th International Congress Birth Defects & Disabilities in a Developing World

IGA CONTACT DETAILS

International Genetic Alliance (IGA),
Helios 130, 2592 CV The Hague, The Netherlands
Phone: +31 70 3855170 or +31 35 6831920
Fax: +31 35 6831891 or +31 35 6027440
E-mail: landfort@tiscali.nl
RABO bank account number: 1389.71.099
IBAN: NL26 RABO 0138 9710 99
BIC or Swift Code: RABONL 2U

Editor IGA Newsletter:
Maryze Schoneveld van der Linde
Contact: maryze@pacesworld.com

The edition of this newsletter is sponsored by an unrestricted educational grant of Genzyme