International Genetic Alliance

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

2010

For members and interested parties

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EDITORIAL

Written by: Martha Carvalho of the Brazilian Genetic Alliance and International Genetic Alliance

Diversity in Cultural Backgrounds and the acceptance of Genetic Diseases

Despite the substantial progress in the genetic field, it is not rare to see people who are not taking full advantage of advances in prevention and treatment of genetic diseases. This unevenness came clear when considering cultural aspects among families affected by genetic conditions.

Diversity in cultural backgrounds can make it difficult for families to accept genetic advances as something that have beneficial effect on people with birth defect, disabilities and genetic diseases.

Just making the service available is not enough, it is vital to build trust and to protect families from additional distress.

Genetic diseases have emotional, psycho social, medical and economical implications, and their consequences can be devastating for individuals, their families and also for the society. In the developing countries the impact of those conditions is dramatically aggravated by the lack of education, resources and appropriate public policies.

IGA values cultural competence to interact effectively with people of different cultural backgrounds. It is crucial to provide information at appropriate comprehension and literacy levels, and in the context of each family's cultural beliefs and practices.

The perception of illness and disease, and their causes, varies according to culture. Culture shapes values and attitudes, including perceptions about what works and what doesn't work, what is helpful and what is not, what makes sense and what does not. Culture also influences how people seek health care, and how they behave toward health care providers. It is important to focus on socio-cultural issues which can interfere in the interpersonal approach, the delivery of genetic information and the adherence to necessary procedures to provide strategies to address them.

Mixed and conflicting feelings are reported from families we are in contact with in Brazil. A classic example is a family who had her 9-year old daughter diagnosed with fragile X syndrome. Parents felt reluctant and scared of participating in genetic counseling sessions since they are supposed to be told what to do or what not to do concerning future pregnancies. On the one hand, as strict Catholics, they do believe it is not for them dare to interfere in natural course of conception and they resigned themselves to the fact that they had been chosen to raise a special child, or special children if it happens. On the other hand they searched the Web for fragile X and parent & patient organizations and became aware about possible impacts of the genetic condition in their daughter's life, and they felt confused.

As there is no right or wrong reaction, it is important to refrain from judgment, it is paramount to be ready to listen and to provide accurate information. Families should be confident to make informed decisions by themselves.

Genetic diseases affect families all over the world, regardless of their race, ethnicity, or cultural background. If we seek a world where genetic diseases are better understood, prevented, treated, and eventually cured, we should learn about the people we care for to have a framework that allows us to provide appropriate care for any family, anywhere.

IGA plays an important role in promoting and facilitating interaction among patients, families and genetic organizations. Collaborative efforts raise awareness about genetic conditions and contribute to make easier for individuals and families to get diagnosis, genetic counseling and treatment. With commitment to be respectful and

sensitive to cultural diversity, IGA has succeeded in clarifying genetic concepts and procedures, helping families to make informed choices concerning prevention and treatment of genetic diseases.

UPCOMING EVENTS

World Orphan Drug Congress, Geneva, Switzerland, 29 November – 1 December 2010

The World Orphan Drug Congress is the World's first industry-led Orphan Drug conference tackling global trends, market access and Big Pharma impact in rare diseases. At this congress you can get a critical understanding of the future of the orphan drugs landscape from the National Institute of Health, European Commission, French Ministry of Health etc. You can learn from industry approaches to improve orphan drug access and understand how patient partnerships are driving the successful development programmes of some industries and discuss the best-practice strategies for developing patient partnerships with EURORDIS and NORD.
You can find more information at: www.terrapinn.com/2010/orphandrugs

9th Asia-Pacific Conference on Human Genetics, Hong Kong, China, 30 November – 3 December 1010

In recent years, significant advances in genetics and genomics have been made at the scientific front. With the development of new technology and understanding of human diseases, these advances have paved way for major impacts in healthcare. The application of knowledge gained at the laboratory bench and clinical settings have been successfully transferred to diagnostic, therapeutic and preventive care. This trend is not only found in specific regions of the world, where medical and scientific development is more advanced. Instead, it has become a pervasive phenomenon throughout the whole world. Genetic and Genomic Medicine has come of age. The Asia-Pacific Region has not been left behind in this tide. For the past couple of decades, countries and areas in this Region have taken on the challenge to develop human genetics and its application. A network of collaboration has also been formed among academic centres, professional societies and various types of organizations in this Region. Every two to three years, the Asian-Pacific Society of Human Genetics organizes a conference for all stake-holders in this field to share knowledge and development opportunities for collaboration. This will be the ninth conference organized by this society.
For more information visit: http://hksmg.org/conference

Human Genome Meeting – Genomics of Human Diversity & Hereditable Disorders, Dubai, United Arab Emirates, 14 – 17 March 2011

In March 2011, the Human Genome Organisation (HUGO) will convene its 15th annual HGM meeting in Dubai, with a main focus on genomics and hereditable disorders. This event represents a new milestone in the mission of HUGO to be a leading international scientific forum with an important role to play especially with the rise of scientific capabilities in the emerging and developing countries. Inherited disorders remain as a mounting global health burden with greater impact in emerging countries. Genomic technologies are providing unheralded precision in identifying the causative genetic mutations, and uncovering new mechanisms of mutagenesis. Deciphering the molecular etiology of these disorders is a key priority and will drive healthcare delivery into new dimensions. The publication of human and other genome sequences offers us a great opportunity to perform large-scale comparative analysis of disease states. The interaction of international scientists with a larger cadre of regional investigators and clinicians from the Middle East will also undoubtedly expand deep collaborations and generate new ideas in human genetics. For more information, please visit: www.hgm2011.org

The 5th International Conference on Birth Defects and Disabilities in the Developing World, Łódź, Poland, 24 – 27 September 2011

The conference is being organised under the sponsorship of the March of Dimes Foundation, Centres for Disease Control and Prevention, USA, National Institutes of Health, USA, World Health Organisation, Medical University of Łódź and the Łódź Voivodship.

The primary theme of the conference will be economics of healthcare and methods for establishing sustainable financial resources to implement programs of value to health and assure access to care. The venue of this conference was selected for a specific reason. The countries of Eastern Europe experienced economic difficulties during the times of their communist regimes and, once their political system changed, managed to reorganise their economies and establish sound functioning economic and public health systems. These countries offer examples applicable in or adaptable to the developing countries. Please visit the conference website www.icbd2011.com for further information.

From Australia

Finally diagnosed – so where to from here?

Dianne Petrie, Director, Association of Genetic Support of Australasia

The Association of Genetic Support of Australasia (AGSA) was established in l988 as an umbrella group for genetic conditions so rare that there is no support group. AGSA provides peer support, information and contact with others with the same or similar condition. AGSA has an extensive Database/Contact Register representing over 850 genetic conditions in 2200 families. AGSA is sandwiched between science and affected families

With the development of micro array testing, AGSA experienced an increase in enquiries regarding chromosomal abnormalities. There is also a significant increase in callers requesting information on who is researching their particular genetic condition, how to get up-to-date research information, who is the clinical expert on their condition and what are the treatment options. Unfortunately science is ahead of the support capabilities. Communication and collaboration with support groups is lacking.

In Australia, there are poorly coordinated services. Those affected by rare conditions are part of a fragmented system lacking any holistic approach. In Europe, Eurordis established a rare disease task force and was successful in changing European legislation to protect the rights of 30 million people with rare diseases and lobby for clinical centres of expertise throughout Europe. In Australia, AGSA, is in a strong position to work with other parties to achieve similar outcomes for the 1.5 million people with rare diseases of which 300,000 are children. AGSA is currently collaborating with the Australian Paediatric Surveillance Unit, Steve Waugh Foundation and SMILE to research the impact of rare diseases in Australia.

You can find more information on AGSA at: www.agsa-geneticsupport.org.au

Genetic Disorders Awareness Week

AGSA's Genetic Disorders Awareness Week seeks to educate the public about the impact of genetic conditions on individuals and families. There are over 6,000 known hereditary single gene disorders and hundreds of syndromes known to be due to chromosomal abnormality. The number of genetic conditions identified due to an interaction of a genetic predisposition and environmental factors, many of which cause common health problems, increases every year e.g. cardiovascular, neurological, cancer, metabolic and mental illness. About 50% of the Australasian population will, during their lifetime, be adversely affected by a condition with a genetic basis. It is estimated that in the first twenty years of life, about 5% of the Australasian population will be affected by an illness, impairment or disability either wholly or partly due to their inherited information. Approximately 30% of patients in paediatric hospitals have a genetic component to their illness and at least 28% of all infant deaths result from genetic factors. At least 50% of all miscarriages are caused by chromosomal abnormalities.

AGSA provides support and information for individuals and families affected by genetic conditions. This is especially important for the more rare conditions, which may not have support groups. AGSA runs specialised seminars on genetic conditions bringing together families, individuals and professionals in an informal setting.

AGSA has been making the right connections for people affected by genetic disorders since 1988.

From the United States

Global Campaign to find all children with Progeria

Location of children living with Progeria around the World.

Begun last year in October 2009, the 'Find the other 150' campaign is a global effort to find all children with Progeria throughout the world.

The campaign was launched by the Progeria Research Foundation (PRF) and has already managed to identify 14 new children with this rare genetic disease. At this moment 68 people with Progeria are known in the world. With a frequency of 1 in 4 – 8 million, it is estimated that there must be around 200 children with Progeria in the world. The most recent new identified children with Progeria live in Brazil, India, Japan, Peru, Turkey, The Philippines, Portugal, South Africa and the United States.

Progeria is an extremely rare genetic condition that causes aging to start at a very early age. Children affected by Progeria usually have small, fragile bodies, like those of very old people. They also have very thin skin, baldness, small faces and pinched noses, which makes them easily recognisable.
"Most of the children we recently made contact with would have remained isolated and would have missed the opportunity to better care and participate in research activities," argues Audrey Gordon, President and Executive Director of the Progeria Research Foundation.

At this moment multiple clinical trials are being reviewed or ongoing from which more can be discovered about Progeria and potential treatments. The recent children with Progeria identified through the 'Find the other 150' campaign, will potentially benefit from these life-changing treatments and will be able to connect with other families of children with Progeria.

PRF has set up a special website with Progeria fact sheets, photo galleries and a collection of podcasts in 6 languages to help put a face to Progeria. Please visit www.findtheother150.org and help spread the word.
This article was derived from the EURORDIS Newsletter of November 2010.

NEWS

The Necessity To Ban Gene Patents

Thousands of cancer patients in Australia could be protected from commercial exploitation by gene monopolies if the Australian federal Parliament supports a motion to amend the Patent Act proposed by the Member of Parliament, Melissa Parke.

Cancer Council Australia and the Clinical Oncological Society of Australia said Ms Parke had shown global leadership by issuing the notice, which calls for an amendment to the Patents Act (1990) to "ensure that patents cannot be granted over

any biological materials which are identical or substantially identical to what exists in nature".

Cancer Council Australia Chief Executive Officer and medical oncologist, Professor Ian Olver, said amending the Patents Act on the basis of Ms Parke's notice would eliminate much of the ambiguity in Australia's outdated patent laws.

"There is a clear principle that human biological materials are not inventions and therefore should not be patented and monopolised by commercial interests," Professor Olver said. In an article written by Nobel Prize winners Professor Joseph Stiglitz and Professor John Sulston published in the Wall Street Journal it is stated that genes and human genetic sequences are naturally occurring things, not inventions. They are part of all our bodies and contain the most fundamental information about humanity – information that should be available to everyone. The researchers and private companies that applied for these gene patents did not invent the genes; they only identified what was already there. There is no objection to corporations that have invented in research to develop a new diagnostic test or vaccine or medicine being granted patents. However, there is a very large objection to corporations being granted patents over the underlying biological materials (genes and proteins), because by having the patent on them corporations prevent others from carrying out clinical tests or undertaking research using those biological materials.

"The amendment put on notice by Ms Parke would uphold this principle – thereby protecting cancer patients today and into the future from the sort of monopoly that almost occurred in 2008 when a company sought to stop public laboratories from testing for genetic risk of breast and ovarian cancer.
Professor Bruce Mann, a surgeon who treats women at genetic risk of breast cancer, said a court in the US had already invalidated one of the patents for breast and ovarian cancer risk, expressly on the same principle underpinning Ms Parke's proposal.
"At present there are only a handful of genetic tests that indicate cancer risk, yet we are already seeing significant problems caused by outdated patent laws," Professor Mann said. "If we do not update patent laws now according to the principles summarised in Ms Parke's proposal, there may be enormous inequities when genetic testing becomes commonplace, if control of human genes is monopolised by commercial interests." In 2008 a company in Melbourne, Genetic Technologies, ordered Australian hospitals and clinical laboratories to stop testing for breast cancer, claiming it had the exclusive right to control access to the relevant gene under the licence it had obtained from Myriad Genetics, a company in the USA.

It is likely that banning gene patents will actually accelerate innovative competition in the biotechnology sector, because the raw materials for new diagnostics, treatments and medicines will be freely available and unencumbered.

This motion to oppose the granting of gene patents is supported by Cancer Council Australia, the Breast Cancer Foundation of Australia, the Breast Cancer Foundation of Australia, the Royal Australian College of Pathologists, the Human Genetics Society of Australia and the Australian Medical Association.

A part of this article has been derived from the Cancer Council Australia. More information on this topic can be found at the website of The Association of Genetic Support of Australasia: http://www.agsa-geneticsupport.org.au

BECOME A MEMBER OF

The international multidisciplinary community genetics network. It facilitates 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.

The **Genetic Alliance Network** offers a wide array of programmes, resources, and events for all the individuals and organisations in our network, including: - Annual Conference, - Genetic Alliance Webinars, - Discussion listserv, - Weekly Bulletin, - Policy Bulletin, and quarterly newsletter, - WikiAdvocacy and WikiGenetics, - Disease InfoSearch
To join the network, please visit: http://www.geneticalliance.org/join

International Alliance of Patients' Organisations (IAPO) represents patients of all nationalities across all disease areas and promotes patient-centred health care around the world. 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.intga.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

Genetic Alliance UK – is a national alliance of patient organisations of genetic disorders.
Website: www.geneticalliance.org.uk

Erfocentrum – Dutch national genetic resource and information centre.
Website: www.erfocentrum.nl/english.php

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, counselling 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

29 November – 3 December 2010, Geneva, Switzerland
World Orphan Drug Congress

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30 November – 3 December 2010, Hong-Kong, China
9th Asia-Pacific Conference on Human Genetics

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14 – 17 March 2011, Dubai, United Arab Emirates
Human genome Meeting – Genomics of Human Diversity & Hereditable Disorders

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28 – 29 April 2011, Amsterdam, The Netherlands
12th International Meeting on Psychological Aspects of Hereditary Cancer (IMPAHC)

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21 – 23 May 2011, Tokyo, Japan
7th International Conferences for Rare Diseases and Orphan Drugs

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23 – 25 June 2011, Washington DC, USA
2011 Genetic Alliance Annual Conference – 25 Years of Innovation

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24 – 27 September 2011, Łódź, Poland
The 5th International Conference on Birth Defects and Disabilities in the Developing World

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