International Genetic Alliance

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

2011

For members and interested parties

 # 11

EDITORIAL

Written by: Alastair Kent, Genetic Alliance UK and International Genetic Alliance

As you are all aware, IGA has achieved much in terms of raising the profile of genetic disorders and of the role of patient organisations in supporting individuals and families affected by, or at risk from these conditions. It has done this with very little by way of tangible resources, relying on the good will and voluntary efforts of us all, supported by small amounts of cash and/or in kind facilitation to enable us to meet together from time to time to share ideas and agree on actions and priorities.

IGA's activities

One of IGA's strengths has been in its ability to develop locally relevant interpretations of themes and issues that have a wider international or global significance for those living with the consequences of a genetic condition in their family. At a Board meeting in Amsterdam last year the decision was taken to focus IGA's activities around three broad themes:

1) Screening, from pre-conception to the neonatal period.
2) Access to services and support for those affected or at risk.
3) Promoting Research & Development of responses to unmet medical needs and their application in the delivery of care and support.

The past months progress has been made, notably with regard to pre-conception screening and care. The recent Biovision Conference in Lyon, France saw the launch of a programme of action in partnership with a number of major nutraceutical partners to promote awareness of the importance of diet in the pre-conception period as well as in pregnancy and after birth for maternal and child health, and an expert conference on pre-conception care is planned for later this year. The IGA has written an IGA Newsletter Special on this topic.

IGA has also been represented at recent European Union conferences on sustainable innovation in healthcare, and through participation in a needs assessment and service planning tool kit under development by the Public Health Genomics Foundation for use in middle income developing countries.

The IGA has been actively involved in the Preparing for Life Initiative. Preparing for Life is a worldwide joint venture of patients/ parent support groups (IGA), science (academia) and service (Rotary) and open to international organisations, governmental bodies, non-governmental organisations, academics, industry and committed individuals. The aim is to promote international and regional awareness and the development of programmes for preconception care, particularly in middle and low-income countries and so contribute to safe motherhood and the birth of healthy infants with the expectation of healthy longevity.

Throughout the world every day some 1400 women die from pregnancy and childbirth related causes and almost 30 000 children under age of 5. The majority of these deaths occur in middle- and low income countries. Many of the maternal and childhood deaths, and other adverse pregnancy outcomes, are preventable with preconception care.

Preparing for Life follows the UN millennium development goals in striving to reduce child mortality by 50% and to improve mother and child health.

Helping to Improve Quality of Care through e-Health

Rod Mitchell is a Board member of EGAN and Acting Treasurer IGA. He has contributed this report which was written with support from IAPO for the Health First Europe Newsletter. This article below is an abbreviated version of the original.

E-health represents a significant avenue to empower patients and their carers as well as aid other healthcare stakeholders in providing truly patient-centred healthcare. Many of the benefits of e-health have been stated in this report, however it is worth reiterating the opportunities that innovation in information and health technologies can bring.

As Europe and the world face the double challenge of an ageing population and an increase in non-communicable diseases there is a growing need to adapt health systems in a cost-effective way that meets the needs and preferences of patients. E-health systems can aid patient choice, improve patient-health professional communication, assist in the day-to-day management of long-term conditions, improve diagnostics and prevention, and connect people in remote areas to healthcare providers. These benefits cannot be underestimated, however there are challenges that need to be met such as data security and interoperability before they can be realised.

Many of us working within the patient community to improve the quality of care of people with rare, chronic and other conditions believe that until the major issues surrounding the interoperability of e-health systems are resolved, within and across the different health related structures of each European Union (EU) member state, there is a fear that many of the already reported positive e-health initiatives being put in place by the EU and others will have little impact in improving the e-health arena and consequently the lives of the European citizens affected by ill health.

It is important to remember that for many citizens across Europe and beyond the security of e-health IT systems is a major concern as they do not believe that their most personal (ill) health information/records will be secure when held within a transportable/shared IT system. Many are concerned that reported access abuses of the are seen by people not authorised to have the information. Besides the security also the money consuming image of e-systems due to inefficient use, serious delays of commissioning dates, makes that EU citizens are critical.

What we now need is a Europe wide initiative involving citizens of all ages though first piloting the use of those “volunteers” who understand and use IT in their daily lives. This would actively involve European citizens and their friends across the world who already communicate with one another in huge numbers using mobile phones and the world wide web who could then transfer their IT skills to the accredited teaching about the benefits of e-Health and how they can assist in resolving the uncertainties that exist in people’s minds. On-line toolkits also have a role to play but we must not forget the many people without access to the latest technologies in their everyday lives.

The EU is based upon solidarity among its people and health is at the top of its agenda so we must encourage our leaders and politicians at all levels to involve their citizens, as many will also have constructive ideas to share.

We know already that e-health systems can successfully provide us with opportunities for regular monitoring, on line contact with physicians allowing variation in treatment programmes for those with long-term conditions and a shared care facility and the collection of data. In scaling up these systems we should reduce costs.

We have learnt from projects like the WHO Patient Safety initiative with its Patient Safety Champions and the consequent introduction of similar projects in Europe, that the patient community has a role to play in reducing medical errors so improving patient safety. Just moving from illegible hand written records to easily understood electronic records shows what benefits can quickly follow when we work together, most importantly building trust among one another too!

While the majority of European citizens hold a bank account and trust their bank to hold their personal financial information in a secure IT environment and increasing numbers undertaking their transactions via the web connected bank services, very many remain unwilling to accept that the same level of high security systems are available for their health records. So it is essential that before we invest huge public sums in developing IT e-health systems further we must overcome this reluctance.

It is important to work together to constructively harness the benefits of e-Health to meet the health challenges of the future and deliver patient -centered healthcare for all.

Genetics finally recognised as a medical speciality in the EU

This article was first published in the June 2011 issue of the EURORDIS newsletter by Nathacha Appanah, It is an abbreviated version of the original.

In many European countries hospitals do have a a genetics department. The geneticists in such a department undertakes tests, interpret them and make a diagnosis; they also provide counselling for patients and their families. But in countries such as Spain, Greece and Belgium, genetics has been an unrecognised specialty: that is until the Commission finally recognised medical genetics as a speciality in the European Union on 3 March 2011.

Milan Macek, President of the European Society of Human Genetics (ESHG), a major stakeholder in this process, is very satisfied and proud of this adoption that comes after years of advocacy. This was one of the top priorities on his agenda. In the EU countries where this medical speciality was not recognised, genetic testing was done by a variety of medical departments. In Spain, for example, standard laboratories would undertake the tests and the patient would benefit from no genetic consultation or genetic counselling. “Lack of knowledge of genetic diseases would lead to needless tests and wrong genetic advice,” says Rosa Sanchez, who is a former President of the Spanish National Alliance, FEDER and is now Vice-President of EURORDIS.

The regulation states that medical genetics is a speciality that responds to the rapid development of knowledge in the field of genetics and its implication in numerous specialised fields, such as oncology, foetal medicine, paediatrics and chronic diseases. “Genetic disorders have a global implication: when a patient is diagnosed with a genetic disease, geneticists have to explain the origin of the disease, discuss how it can affect the family at large, present and future, and arrange for a complete and multi-disciplinary medical support,” explains Ségolène Aymé, director of Orphanet and a geneticist herself.

Saying that medical genetics is crucial for rare diseases is an understatement. Without this field, many rare diseases would still go undiagnosed. The knowledge geneticists have of rare diseases aids the entire medical and scientific community. “When the Human Genome project started, geneticists were the first to come forward with their studies, their cases and their data. They dealt with rarity, with rare diseases and these rare diseases would help understand our DNA,” says Ségolène Aymé.

Genetics and geneticists have modified the way research and medicine work. Geneticists elaborated the first databases; they were pioneers in making their literature accessible, their holistic approach is well regarded, and they are keen to share their data.

Genetic counselling is a specific field and requires special skills and knowledge.

In countries where genetics have not previously been considered a medical speciality, many doctors and medical specialists believed that they could give genetic counselling in their own field. “This is not true. Genetics is different and a specific field. I would always say in my lectures advocating for this regulation that it is unethical to deny training for geneticists,” states Ségolène Aymé. .

Still, Milan Macek states that there is much more to do. “There are significant challenges in front of us, in that we need to achieve the same status also for laboratory genetics speciality and for genetic nurses/counsellors. The ESHG wants to stress that it acknowledges that these specialities are as important as clinical/medical genetics and that these constitute integral components of comprehensive provision of genetic services.”

BECOME A MEMBER OF

The **Genetic Alliance Network** offers a wide array of programmes, resources, and events for all the individuals and organisations in our network.
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.

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.

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 – Patients Network for Medical Research & Health.
Website: www.egan.eu

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

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

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

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

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

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

RARE DISORDERS:

EURORDIS – is a non-governmental patient-driven alliance of patient organisations representing more than 479 rare diseases in 45 countries
Website: www.eurordis.org

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

National Organization for Rare Disorders (NORD) - dedicated to helping the nearly 30 million Americans with rare diseases through programmes, research etc.
Website: www.rarediseases.org

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

Rare Disease Foundation – to change the lives of children and families affected by rare and undiagnosed diseases
Website: http://rarediseasefoundation.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

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

11—15 October 2011, Montreal, Canada
The 12th International Congress of Human Genetics (ICHG).

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