International Genetic Alliance

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

2012

For members and interested parties

 # 15

Preparing for Life aims to promote awareness and contribute to the development of programs for preconception care for all women of childbearing ages and their partners, particularly in middle and low-income countries.

The ultimate goal for Preparing for Life is to reduce maternal and childhood mortality and morbidity and so to contribute to safe motherhood and the birth of healthy infants with the expectation of healthy longevity.

In the scientific consortium of the Preparing for Life Initiative are experts from, National Center of Birth Defects & Developmental Disabilities and the Centers for Disease Control and Prevention in the USA, International Clearinghouse for Birth Defects Surveillance and Research in Italy, March of Dimes in the USA, the Universities of Witwatersrand, Rotterdam, Amsterdam, Münster and Beijing, World Alliance of Organizations for Prevention and Treatment of Genetic Conditions, patients and families (International Genetic Alliance) and service clubs (Rotary and Zonta).

If you wish to join or to support the Initiative, contact the Preparing for Life secretariat.

History of Preparing for Life

Preparing for Life is an initiative that started in the Netherlands and is rooted in the seventies.

TimeLine Preparing for Life	
1975	Foundation of the Dutch Genetic Alliance (VSOP)
1994	International Conferences on Genetic Diseases
2000	1st WHO International Conference in The Hague (organised by VSOP)
2007	Conference on 'The Hidden Toll of Birth Defects' in Geneva
2008	'Preconception Care, a good beginning' report of Dutch Health Council, The Hague
2010	International Expert Meeting 'Preparing for Life', launching the Plan and Program in The Hague
2011	Preparing for Life started in collaboration with WHO
2012	WHO and Preparing for Life organised international and consensus and implementation meeting in Geneva

Preparing for Life at this moment

The initiative was taken in the Netherlands and it is this country that is currently in the driving seat. The Dutch initiators are working together with the International Genetic Alliance. To be able to achieve the Preparing for Life goals, timed action, free and well informed decision making, awareness and good networks are important. The Preparing for Life joint venture includes international expert and consensus meetings, regional policy and implementation meetings and meetings to integrate the preparing for life concept in existing primary healthcare services in all countries.

The regions in the world are divided among the partners depending on already existing networks and experiences. IGA is involved in Africa, Europe and West Pacific. WAO will be involved in the eastern Mediterranean. IGA and WAO both

will be involved in China. By the various networks and collaborations excellent initiatives are developed such as Born Healthy.

Preparing for Life in the future
The goal is that all babies born in the world are wanted and healthy.

Imagine if you could transform the lives of almost 6 million children each year?

We believe that together we can!

Born Healthy is a community portal developed by the UK-based Foundation for Human Genomics and Population Health (PHG Foundation) in response to the need for developing countries to focus on reducing and treating birth defects in their populations. The PHG Foundation, at the 2010 Clinton Global Initiative Meeting, made a pledge to aid developing countries fight birth defects.

Born Healthy has two principle elements: a toolkit for gathering knowledge on birth defects and the measures that can prevent or treat them; and a global community that can work together supporting national and regional initiatives in this area. The toolkit, for undertaking a health needs assessment, is freely available and to date includes 7 topics:

- Down syndrome;
- Neural tube defects;
- Orofacial clefts;
- Health services;
- Preconception care and screening;
- Prenatal services; and Newborn screening.

Mr. Ysbrand Poortman and Mr. Alastair Kent both also closely involved in IGA are members of the steering committee of Born Healthy. A fruitful interaction between IGA and its activities and Born Healthy is therefore guaranteed.

For more information, please look at: www.bornhealthy.org

The Chinese Genetic Alliance

IGA is active in membership outreach in the African and in the Asia Pacific Area. Both areas with huge problems, huge development and opportunities. Last year, 1st September an Asian Pacific Genetic Alliance (APGA) was established in Hong Kong in conjunction with the 11th Annual Meeting of the Asia Pacific Society of Human Genetic (APGA) (see photo).

On November 6 2011 a group was formed in Beijing aiming at setting up a Chinese Genetic Alliance. Thanks to the liaison and support of prof. Nanbert Zhong and prof. Xinliang Zhao both of the center of medical genetics of the Peking, representatives of a variety of interest met under the chairmanship of Mrs Ma Jia Nian on 6th November in Beijing. After an animated discussion about common interests, objectives, priorities and funding, then and there was decided to establish a Chinese Genetic Alliance (CGA). CGA will seek membership of the APGA. Because of the major problems in and importance of China the CGA will have a direct communication with the board of IGA.

Mrs Ma Jia Nian is the mother of a pre term child and president of the Chinese parent organisation for pre term born babies. This organisation is quite powerful in China and one of the leading parent organisations. The CGA will actively partner in the Preparing for Life Initiative, the Roadmap to Treatment and in the Nutrition projects.

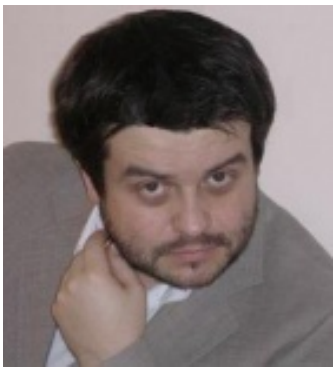
In September next year an Asian Pacific conference is scheduled in Bangkok, Thailand and in 2013 the global conference of the 6th International Congress of Birth defects & Disabilities in a developing World will be held in Manila, the Philippines.

Members of the recently founded Chinese Alliance of Parent and Patient Organisations.

Welcome to Central & Eastern European Genetic Network (CEE GN)

During a working conference held by EGAN and Roche from January 12 – 13 2012 in Basel, also a meeting with patient organizations' representatives from the Eastern Europe took place.

The aim of the meeting was to plan and coordinate the activities for 2012-2014 in the Eastern part of Europe. The actions taken so far do not enable the proper diagnosis regarding rare diseases in this part of Europe, where the level of performance of the patient organizations appears to be insufficient (Poland, Bulgaria, the Czech Republic). On the map of countries that still need help there are, e.g., Russia including the former Republics as well as Slovenia, Croatia, Serbia, Montenegro, Bosnia and Herzegovina. The lack of data regarding both diagnosing and treatment in the Ukraine and Belarus creates a major issue in evaluating the scale of incidence of rare diseases.



Mr. Krzysztof Swacha, the new Executive Director of CEEGN.

With the objective to build and coordinate the structures of CEEGN organization following the model of the existing umbrella organizations such as EGAN, Mr Krzysztof Swacha (Poland) has been appointed to the position of Executive Director. Developing genetic networking together by the scientific circles and the industry in the Central and Eastern European Countries seems to be indispensible.

"The lack of education in society, the lack of sufficient resources to support research in this area and the lack of joint projects, such as conferences, makes the situation even more complicated," says Krzysztof Swacha, CEE GN. "On the other hand, developing national plans for rare diseases in each of these countries will enable a dialogue with the governments, as with government support the patients affected by such diseases will be offered more efficient help. We do hope that the actions that have been taken will soon result in a significant improvement of the situation of people and patient organizations involved in rare diseases also in this region."

CALL - Your participation in paediatric research

Children do not respond to medications in the same way as adults. However, the majority of drugs prescribed for children have not been tested in paediatric populations. Without adequate data from such testing, prescribing drugs appropriately becomes challenging for clinicians treating children, from infancy through to adolescence. Not only does this create a challenge for health professionals but it also places the health of children at stake as available treatments are not always as effective as possible.

To address the current barriers present in paediatrics research, the EU has funded a project called Global Research in Paediatrics GRIP (www.grip-network.org). The main aim of the project is to implement an infrastructure to stimulate and facilitate the development and safe use of medicine in children. GRIP also aims to create consensus on international standards and methodologies, and tools for paediatric research. GRIP will develop guidelines, training programmes, and paediatric formulations etc. Patient groups will be able to contribute to these processes with evidence from their experiences.

GRIP involves more than 20 institutions as partners and 16 major networks that represent hundreds of clinical sites and a total of more than 1000 researchers across Europe, the US and Asia. The WHO and the European Medicines Agency (EMA) are also involved.

The GRIP consortium wants to work closely with patient organisations and affected families, to provide children with safe and effective medicines. On behalf of EGAN (www.egan.eu), the Dutch Genetic Alliance VSOP (www.vsop.nl) coordinates the interaction between patient organisations and the other consortium members.

EGAN/VSOP is looking for interested patient representatives of European or international patient organisations, to be consulted on issues that the project is dealing with. Your active input and review as a patient or member of an affected family can make a difference in how research in children will be performed in the future.

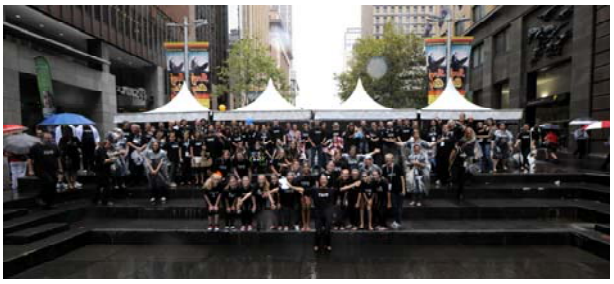
Please send your contact details to grip@egan.eu if you are interested in participating in the GRIP Patient Network to improve research and clinical trials in children.

THE RARE DISEASE MOVEMENT – An Australian Perspective

The Association of Genetic Support of Australasia (AGSA) was established in 1988 to provide peer support and information to people isolated by the experience of living with a genetic condition and to lobby the Government for more Genetic Services. In this role, AGSA supports professionals and families and has a database of over 1200 genetic conditions / rare diseases. AGSA offers a wide range of support options to ensure that access to our service is equitable and meaningful.

Background to the Rare Disease Movement:
EURORDIS aims to improve the quality of life of people living with rare diseases in Europe through advocacy at the European level, support for research and drug development, networking patient groups and raising awareness of the impact of rare diseases. In 2003, EURORDIS held the first **Rare Disease Day** to raise awareness of the issues surrounding rare diseases and the need for an united approach.

The Australian approach:
As more support groups/patient organizations become more aware of the coordinated



Raising awareness on Rare Disease Day in Martins Place in Sydney.

international movement for Rare Diseases and aware of how far Australia has yet to come, many ran their own activities in recognition of Rare Disease Day or collaborated with others. For Rare Disease Day 2012, AGSA participated with the Steve Waugh Foundation, the SMILE Foundation, The Centre for Genetics Education and the Association for the Wellbeing of Children in Hospitals in an awareness raising activity in Martin Place, Sydney. The event included volunteers handing out free bottled water with an informative newsletter wrapped around it directing people to the rare disease website for more information.

On 17-20 April 2011, the *Awakening Australia to Rare Diseases: Global perspectives on establishing a coordinated approach to a national plan seminar* was held in Fremantle, Western Australia. The symposium was designed to bring together patients, support groups, academics and policy makers to discuss the development of a National Plan for rare diseases. The aims of the symposium were as follows: *to improve the welfare and wellbeing of all Australian people affected by rare diseases and their families, through better knowledge, research, treatment, care, information, education and awareness of rare diseases.*

The main topics for discussion included:
• Development of national plans for rare diseases;
• Patient empowerment;
• Patient care, support and management;
• Research and translation;
• International and national networks;
• Networks, partnerships and collaboration.

AGSA attended the symposium to participate as the key issues are areas we have been working on, with limited funding and resources, for over 24 years. AGSA has the largest registry in the region with which we have established national and international networks and collaborations, facilitated contact between individuals and families and participated in research. AGSA has provided a united voice on behalf of support groups relating to government policy and advocates for families when requested. Our support activities focus on strategies to enable and empower individuals and families in all aspects of their rare disease / genetic condition.

AGSA has embarked on a partnership with the Australian Paediatric Surveillance Unit, SMILE Foundation and the Steve Waugh Foundation Royal Australasian College of Physicians, Children's Hospital at Westmead plus **Student involvement** PhD Scholar - Psychology, (University of Sydney) , PhD Scholar - (Health Economics, University of Sydney), Advanced Medical trainee, Royal Australasian College of Physicians, to research over three years the Psychosocial and Economic Impacts of Rare Diseases on Australian Children, Families and Health Professionals

Although we support the drive for a national plan, we are finding it increasingly important to ensure that there is no wasting of resources, of existing well developed and respected services and the many volunteers who have the capability, skills and experience in many of these areas. AGSA is calling for the newly formed Rare Voices Australia to thoroughly audited what already exists and to consult with key members of the field such as genetic services throughout Australia.

IAPO Latin America Regional Network Meeting – Mexico City, Mexico – 6-8 June 2012

In June, IAPO will hold a series of meetings in Latin America to bring together IAPO members and other patient groups from different countries and disease areas across Latin America and the Caribbean to establish communications and objectives for an active and empowered network. Through a day and a half of workshops a defined shortlist of practical objectives for the IAPO Latin American regional network, as agreed by workshop participants, will be developed. IAPO will also hold a multi-stakeholder seminar on the afternoon of 7 June which will focus on the two key issues of non-communicable diseases and patient safety.

More info can be seen by visiting: www.patientsorganizations.org and scolling down the home page.

BECOME A MEMBER OF

The **Genetic Alliance Network** offers programmes, resources, and events for everyone in our network: http://www.geneticalliance.org/join

International Alliance of Patients' Organisations (IAPO) global alliance representing patients worldwide across all disease areas: http://www.patientsorganizations.org/index.pl?n=210;section=3

International Multidisciplinary Community Genetics Network. Facilitates communication between those working in community genetics (and genomics). To become a member send an e-mail to commgennet@gmail.com.

RELEVANT WEBSITES

ORGANISATIONS:

BIO – a biotechnology industry organisation 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 – 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.geneticalliance.org.uk/projects/eurogenetictesting.htm

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 patientdriven 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) - 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

September 2013, Bangkok, Thailand
Asian Pacific Conference

2013, Manila, The Philippines
The 6th International Conference on Birth Defects and Disabilities in the Developing World

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