International Genetic Alliance

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

2010

For members and interested parties

 # 8

INTRODUCTION

Newsletter #8 is the first edition in 2010. In our last Newsletters # 6 and #7 we raised your attention to the Declaration on Genetic & Congenital Disorders. You can read the Declaration at: http://www.gig.org.uk/docs/policy/igadeclaration_09.pdf If your organisation is interested to sign this Declaration, please send an email to maryze@pacesworld.com

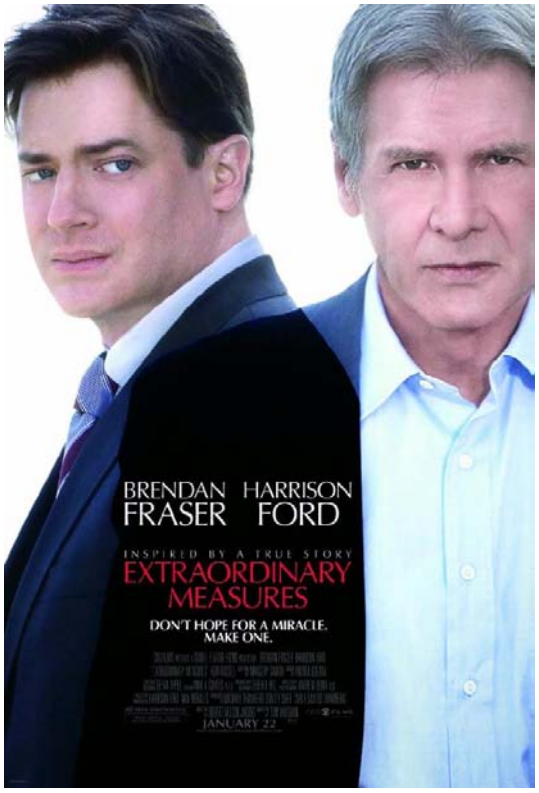
EDITORIAL

Written by: Maryze Schoneveld van der Linde, Advisor International Pompe Association

Hollywood movie on a rare genetic disease: Pompe disease

Genetic diseases are not generally understood in the general public. Many people still don't know that having a healthy child is more a matter of luck than a common right. Even when a child appears to be healthy at birth it can still have a genetic disease that manifests later in life. When a genetic disease is rare often not much is known about it. One must then be lucky to have a dedicated physician who is willing to spend time to gain knowledge on the disease. Research on a specific rare genetic disease is scarce because financial resources are lacking.
The past years, however, changes can be seen. New technologies and new insights in molecular biology and biochemistry bring new perspectives. The Orphan Drug Act, designated by President Ronald Reagan in 1983, brought opportunities never thought possible before. Thanks to this Act and the new technologies, rare genetic diseases, have a better chance than ever. Right now one third of all new drugs developed are for rare (genetic) diseases. One of these rare genetic diseases is Pompe disease.

People with Pompe disease, lack the enzyme alpha-glucosidase and their muscles deteriorate so much that they need breathing support and a wheelchair etc. After many years of research and dedication of physicians, scientists, patients and industry an enzyme replacement therapy to treat this disease entered the market in March 2006. This is an extraordinary achievement for Pompe patients and their families. In 2006, Geeta Anand, wrote the book *The Cure*, the story of John Crowley, a father with two children with Pompe disease.



This book forms the basis of the Hollywood film *Extraordinary Measures*, in which a desperate father and a biochemist race to develop a cure for Pompe disease while his children are deteriorating fast and the doctors wring their hands, Crowley, a drug company marketing executive, played by actor Brendan Fraser, decides to take matters into his own hands. In partnership with biochemist Robert Stonehill, played by Harrison Ford, Crowley launches a biotech company aimed at bringing a lifesaving Pompe drug into clinical trials. In the end, the

drug treatment is just in time to save his children.

In real life, however, the treatments his children receive didn't come from his company, but from the American biotech company Genzyme. Crowley's drug treatment, developed by his start-up company, never entered clinical trials. A lot can be said about this movie. It is a Hollywood movie on a rare genetic disease that was lucky enough to get a treatment, but that certainly must not be regarded as the whole story on Pompe disease.

Many people in the world who participated in and contributed to the drug development process to treat Pompe disease, were left out and didn't get the credits they should get. *Extraordinary Measures*, however is a good movie to raise awareness of Pompe disease and other genetic diseases. It shows there are still people in need of treatment and much can be done to get these treatments. *Extraordinary Measures* is an excellent opportunity for us all to raise awareness. The trailer of Extraordinary Measures can be viewed at: http://extraordinarymeasuresthemovie.com

If you are interested in reading more information about the story of Pompe disease, you can read the Pompe blog of Kevin O'Donnell, advisor to the International Pompe Association. You can find it at: http://pompestory.blogspot.com

UPCOMING EVENTS

International Federation of Human Genetics Societies (ICHG/ASHG 2011), Montréal, Quebéc, Canada, 11-15 October 2011

The International Federation of Human Genetics Societies (IFHGS) was founded in 1996. It is an international collaboration of professional human genetics societies. The IFHGS ensures that an International Congress of Human Genetics will take place every five years and that a credible forum for the sharing of ideas about human genetics is provided. The purpose of The International Federation of Human Genetics Societies is to provide a forum for organised groups dedicated to all aspects of human genetics, including research, clinical practice, and professional and lay education. The Federation will enable communication between its member groups and encourage interaction between workers in genetic fields and related sciences and will make itself available to promote meetings and publications and other forums which support human genetic research and practice."
For more information visit:
www.ichg2011.org

2010 International Symposium on MPS and Related Diseases, Adelaide, Australia, 23 – 27 June 2010

11TH INTERNATIONAL SYMPOSIUM
ON MUCOPOLYSACCHARIDE AND RELATED DISEASES

"Translating Research into Clinical Reality" The Australian Mucopolysaccharidosis Society, the Lysosomal Diseases Australia and Lysosomal Diseases New Zealand is hosting the 2010 International Symposium on MPS and Related Diseases in Adelaide. The conference will include scientific and family programmes and is an unprecedented opportunity for Australian MPS families to have access to a wide range of leading scientific and medical experts in one place at one time. Please look for more information at: www.mps2010.com.au

BioVision Alexandria 2010 – New Life Sciences: Future Prospects, Alexandria, Egypt, 11 – 15 April 2010

The Bibliotheca Alexandria is organising its Fifth International Biennial Conference. The theme of BioVision Alexandria 2010 will be New Life Sciences: Future Prospects. It aims to identify and explore new frontiers and new areas in life sciences that will vastly serve humanity and provide hope for solving the world's most pressing issues. The conference will focus on three major themes: Health, Food and Agriculture and

Environment. Each of these themes will be addressed by representatives of the greatest minds in industry, science, policy-makers and civil society fields. For information look at: www.bibalex.org/bva2010

SUPPORT NEEDED

Support the enactment of Rare Disease Act of the Philippines

Philippine Society for Orphan Disorders, Inc.

The Philippine Society for Orphan Disorders, Inc needs 500,000 signatures. Please join the campaign to increase awareness on rare diseases and to rally law makers in passing Senate Bill 3087 or the Rare Diseases Act of the Philippines. The Bill will help provide people with rare diseases better access to medical care, health information, and health care products needed to treat their conditions. You can be part of the campaign via an online petition: http://www.thepetitionsite.com/2/rare-diseases-act-of-the-philippines

From South Africa

The Achievements and Challenges on Genetic Disease in South Africa

The South African Inherited Disorders Association formed in 1973 as an umbrella body for 22 support groups. Originally, SAIDA was intended to support and co-ordinate the efforts of individual patient / parent groups.

When SAIDA was formed, medical genetic services were limited. And what there was served mainly middle class whites. There were 20 genetic nurses, who saw mainly white patients.

In 1985, there were 14 genetic counselling clinics. They saw only 4500 patients – but 63% were from the cities in which the clinics were situated. Only 18% were black.

The SA Society of Human Genetics, made up of doctors and scientists, was formed only in 1986 – 13 years after the patient / parent body, SAIDA. The first medical geneticist qualified only in 2002.

It fell to support groups to provide affected families with information.

When my own child was diagnosed with Turner Syndrome in 1989, the genetic clinic could tell me all about karyotypes (the number and appearance of chromosomes) and genetic tests, but could answer none of my questions about what her condition would mean for us. They advised me to join the support group for answers.

I joined Turner Syndrome Contact Group, a member of SAIDA. Like other support groups, our main task in those days was to provide psycho-social support for affected families and provide information.

As a support group we raised the funds to attend international conferences to stay abreast of research. We produced report-back booklets, containing full reports of papers and research, which were then sent to doctors.

We gave talks to medical students, to groups of GPs, to nurses and to teachers. We supported research, by encouraging our members to be part of any research projects.

We arranged national conferences, sometimes with international speakers. Our members underwent training in counselling so that we could provide counselling support.

After 1994, post apartheid, the new government recognised the immense importance of birth defects to child health. They also recognised the need to extend genetic services to disadvantaged areas.

But these included both urban areas, and huge rural areas with little or no primary health care … This is still the situation my country faces today.

Recognising an existing need, many groups have tried to initiate projects in economically disadvantaged, black communities, But this is more difficult than at first appears.

The legacy of apartheid means communities in SA remain divided. Access to far-flung rural areas is just about impossible for volunteers – most of whom have jobs and families of their own.

South Africa has 11 official languages, which makes communication and spreading of information difficult. Families in disadvantaged areas often can't contribute financially. And diminishing funds in middle-class areas restricts what can be achieved.

Cultural differences play a role, as does the learned helplessness created by apartheid and poverty. It's rare to see disadvantaged families pro-actively organising groups - and most existing groups lack the human and financial resources to carry the whole burden.

Larger groups, with secure funding, have made this transition – to a limited extent. And many smaller groups have undertaken projects of benefit to disadvantaged communities. For example, the Turner Syndrome group has written and produced an information pamphlet, and had it translated into eight local languages, for use in clinics and hospitals.

But the greatest role for all support groups has become their support of the projects of their umbrella body. SAIDA now initiates projects significant in their own right, rather than just acting as a supportive umbrella.

There are 10 trained clinical geneticists in the country – in urban academic centres. In vast areas of rural South Africa, a few "outreach" clinics still reach only limited numbers.

In this situation, SAIDA has taken on a role which, in more developed countries, would be carried by state.

SAIDA is made up of lay volunteers, the University of the Witwatersrand's human genetics department and by the Department of Health, who bring moral and some financial support, as well as their distribution and networks.

SAIDA has initiated a genetic training programme to give nurses a primary health role in the care and prevention of genetic conditions. It has also developed a community training programme, providing training in basic counselling and psycho-social support.

SAIDA is part of a collaboration of North-South partners aimed at sharing knowledge, research and experiences and, in this way, developing appropriate genetic services for developing countries and promoting capacity building.

For the future: SAIDA is in the process of producing a textbook, containing the information and concepts needed by doctors and medical students, but written in "plain" language. It is intended for use by doctors whose first language is not English, and even primary health workers without medical training.

It would help in standardising skills across the country – and possibly across Africa.

SAIDA has an ambitious plan to train trainers, who will train rural community members to become primary genetic health care providers. SAIDA lacks the funding to undertake this project yet. But it remains a great need. By: Jo-Anne Richards, SAIDA, http://www.saida.org.za/

NEWS

biomedinvo4all
Roadmap to Treatment

Biomedinvo4all.com provides independent information on innovative medical research fields to support the active involvement of patients / patient organisations, relatives, users of health care and participants in health research.

Being actively involved in research and therapy development means much more than being a 'subject' of the research. It means participation in setting the research agenda, in the way research is carried out and how it should be implemented. Your personal experiences of living with a certain illness, combined with those of others, are very relevant to this.
This website informs you on several topics involved in the Roadmap to Treatment such

as biobanks, medical genetics, animal research, clinical trials, pharmacogenetics, gene therapy, stem cell therapy. Biomedinvo4all.com also works on getting information on future themes such as medical genomics, metabolomics, nutrigenomics, personalised medicine, regenerative medicine and perinatal care (preconception, prenatal and neonatal care). If you are interested you can read information on these topics at: http://www.biomedinvo4all.com

Personal look at genes locates disease causes

Children inherit fewer gene mutations from their parents than was previously thought, say researchers in the USA who are the first to sequence the entire genome of a family. The blueprint for life, called DNA, contains about 22,000 genes, and researchers calculated the number of changes by analysing the genes of mother, father and children.

Researchers at the Institute of Systems Biology in Seattle studied a family in which the parents had no genetic abnormalities, but each carried recessive genes that resulted in their son and daughter being born with two extremely rare conditions, Miller Syndrome and primary ciliary dyskinesia (PCD). Miller Syndrome, which causes facial and limb malformations, has been diagnosed in only two families in the world.

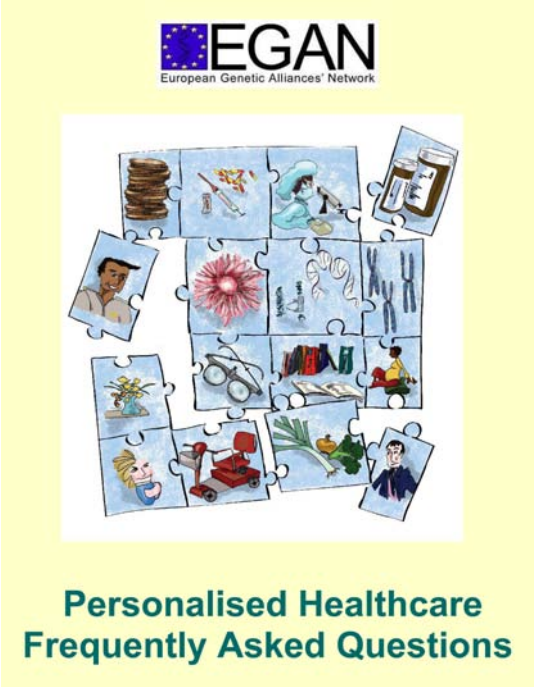
PCD is a condition in which the tiny hair-like structures that are supposed to move mucus out of airways in the lungs do not function. The chances of having PCD are estimated to be 1 : 10,000.

Comparing the genomes (genetic makeup) between the parents and their children allowed researchers to identify the gene responsible for Miller Syndrome, which afflicts both children. Experts have long known that Miller Syndrome is genetic. But they had never been able to pinpoint the gene.

Having genomes from all the family members allowed the Seattle team to focus on identical genetic sequences in the two children. It helped to shrink the genetic haystack from thousands of genes to four likely targets. The analysis of the four family members (the parents, daughter and son) also revealed that each parent passes about 30 mutations to their children. It had long been believed that each parent passes 75 gene mutations to their children. The mutation rate was less than half of what was thought. Most gene mutations passed from parents have no effect on children's health. But it's important to know the number of parent-to-child gene mutations. The rate at which things change is considered critical, helping explain the gradual development of changes. The actual rate of gene mutations each parent gives a child will vary depending on the age of the parents (particularly the father) when a child is conceived. Genomic studies like this can help researchers find ways to identify individual genes or mutations that can lead to inherited disease such as Alzheimer's disease and Diabetes. Looking at the genomes of families will give a real boost to the power to identify the various genetic contributions to disease.

BOOKS

In one sense, personalised healthcare is nothing new. It is what doctors have aimed to provide for their patients through the exercise of their clinical judgement, backed



up by specialist knowledge and the use of appropriate diagnostic procedures. However, as research advances and we begin to understand more about the complexity of common diseases, it is clear that a standard approach to the treatment of cancer, heart

diseases and other common physical and psychological disorders will not yield the results patients need. Increasingly therapies are being selected based on an understanding of the underlying genetic components of a condition and how these interact. This has come to be known as Personalised Healthcare. Properly applied and adjunct to the clinical skills of doctors, it will open up new opportunities for preventing, treating or curing many currently intractable diseases. It is an area where there has been much hype (both positive and negative) about possibilities for progress. We hope this booklet steers a middle way between these extremes and gives a balanced view of Personalised Healthcare as it is now and for the immediately foreseeable future.
It is the result of a collaboration between the patient group EGAN (European Genetic Alliances' Network) and Roche, with EGAN collecting commonly asked questions and Roche experts formulating answers that have then been checked by independent specialists.
You can download this booklet at: http://www.gig.org.uk/docs/egan_personalisedhealthcare.pdf

THE VOICE OF 12,000 PATIENTS
This book on the experiences and expectations of rare disease patients on diagnosis and care in Europe can be downloaded fully or in sections from the Eurordis website at: http://www.eurordis.org/article.php3?id_article=1960

IT'S MY LIFE
The book portrays a revolution that is taking place now! Twelve patients with a rare hereditary disorders all over the world have been interviewed. Their stories show a determination to take charge, to make a real difference. 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.

BECOME A MEMBER OF

The international multidisciplinary community genetics network. Its 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 encourage your colleagues to become members, or to send me the e-mail address(es) of anyone you propose.

The **Genetic Alliance Network**. Genetic Alliance offers a wide array of programmes, resources, and events for all the individuals and organisations in our network, including:
- Annual Conference, - Genetic Alliance Webinars, - Member Forum, - 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) 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.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 Interest Group (GIG) – is a national alliance of patient organisations of genetic disorders.
Website: www.gig.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

11 – 15 April 2010, Alexandria, Egypt
BioVision Alexandria 2010 – New Life Sciences: Future Prospects

3 – 6 May 2010, Chicago, USA
BIO International Convention

11 – 15 October 2011, Montréal, Canada
International Federation of Human Genetics Societies (ICHG/ASHG)

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