

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

INTRODUCTION

This newsletter will inform you about events, about progress in member countries, publications and projects. It serves to provide information and to report on activities which could serve as a model for others.

Your contributions for the next Newsletter will be welcome but should arrive no later than the 28th of August.

UPCOMING EVENTS

**World Health Summit,
Germany, Berlin, 15 – 18 October 2009**



IGA has been invited to participate in the World Health Summit which will take place from 14 - 18 October 2009. This forum - under patronage of Angela Merkel and Nicolas Sarkozy - will bring together an international array of eminent researchers, physicians, and representatives of government, industry, health care systems and patient organisations.

As a global forum, the World Health Summit will address those health related issues which challenge us as physicians, scientists, political leaders and those employed in the health care industry. We need cogent and timely responses to the urgent questions arising from world-wide demographic trends, the financing of medical progress and innovation, the understanding of the potentials of medical research and health economy, and to emerging medical issues. Among these are health-related consequences of climate change, international pandemic strategies, and the impact of the financial crisis on global health and health care.

The forum will set the broad agenda for future medical research and for structures of health care for all. For more information visit: <http://www.worldhealthsummit.org/>

**4th International Conference on Birth Defects and Disabilities in Developing Countries,
India, New Delhi, 4 – 7 October 2009.
“Translating Research into Care and Prevention.”**



The theme of the conference is ‘Establishing effective services for the care and prevention of birth defects’.

The conference will cover many aspects such as surveillance programs and registries; major risk and preventive factors; individual and community-level prevention and strategies, including micronutrient fortification and immunization programs and preconception health and health care; counselling and care of common birth disorders and genetic conditions; rehabilitation and rights of disabled and legal frameworks for their protection.

IGA is a partner in organising and promoting these conferences. Together with Thalasseemics India (an exemplary patient/parent organisation in India, see: www.thalasseemicsindia.org), IGA and 4th ICBDD are organising a plenary session on “The Important Role of Patient/Parent Organisations in the Care and prevention”. Patients and parents from India, Europe, Latin America and Middle East will present from personal experience, table the situation and needs in their region and speak about their association’s contribution to support the families and mention their efforts and results in improving the healthcare system.

Visit: <http://www.4icbddd2009.com>

Pre conferences on 4th October 2009 in Delhi (co-) organised by related organisations are:

- World Alliance NeuroMuscular Disorder Associations and the Indian Association for Muscular Dystrophy will hold a session on "Roadmap To Treatment" .

-Thalassemia and Sickle cell disease will hold a session on "the burden of Thalassemia and sickle cell disease around the world".

- Cerebral Palsy and Autism will hold a session on: "Diagnosis and management".

- Parivaar, the Indian Alliance of associations involved in mental retardation will also held a session.

TREAT-NMD/National Institute of Health (NIH) international conference on 'Bringing down the Barriers - Translational Medicine in Inherited Neuromuscular Diseases', 17 - 19 November 2009, Brussels, Belgium



TREAT-NMD is a major project sponsored by the European Union to bring together many experts and expert centres on neuromuscular disorders. This conference is the first international meeting and is organised in collaboration with the National Institute of Health in the USA. The aim of the meeting is to share progress in the area of translational medicine in inherited neuromuscular diseases and set the future collaborative agenda. This conference will build on achievements of the NIH and TREAT-NMD. It will be a highly interactive meeting with a strong focus on the key issues surrounding "trial readiness" in the neuromuscular field. The conference will be held at the Crowne Plaza Hotel in Brussels City Centre in Belgium.

Visit: <http://www.treat-mdconference2009.eu/>

SUPPORT NEEDED

"Metabolic myopathies"- research project Message from Croatia

Ivo Barić, Ksenija Fumić, Danijela Petković Ramadža, Mario Ćuk, Oliver Vugrek from Department of Pediatrics and Clinical Institute for Laboratory Diagnosis, School of Medicine, University Hospital Center, Zagreb, Croatia, Department of Molecular Medicine, Institute "Ruđer Bošković", Zagreb, Croatia

A research project of the study of metabolic myopathies in Croatia has been initiated following the discovery of a novel inherited metabolic disease, the S-adenosylhomocysteine hydrolase (SAHH) deficiency, which has subsequently showed association with congenital myopathy in all confirmed cases. At any phase, depending on the results, additional, for instance confirmatory enzyme or gene tests can be done. To include as many patients as possible, all colleagues dealing with neuromuscular diseases in Croatia and patients' groups have been informed. Lectures were given in all university centres. Participation of patients and physicians outside Croatia is desired.

As you can see we are looking for more patients and collaboration with other physicians and scientists. In this "hard time for science in Croatia" we are looking also for some finances. For more information please contact: Ksenija Fumić, PhD Email: kfumic@kbc-zagreb.hr

PUBLICATIONS AND REPORTS

From Europe

The Central and Eastern European Genetic Alliance (CEEGN) had a meeting on 24th – 25th January 2009 in Prague on the initiative of IGA in collaboration with European Genetic Alliances' Network (EGAN), European Forum Good Clinical Practice and the World Alliance Neuromuscular Disorder Associations (WANDA). This meeting was attended by 35 participants from 15 countries. Among them were CEEGN Board members, representatives from patient organizations, and guests representing Projects, and other organizations. During the meeting the challenges in CEE countries were identified:

- There is a need for more transparency over reimbursement decisions and reimbursement programs.
- Lack of information on genetic and rare disease and lack of infrastructure for follow up of diagnosed patients. Therefore, the invitation for membership to the CEEGN Medical & Scientific Committee was distributed to ten nominated physicians in the CEE.
- Due to the language barrier, there is a need for websites and all other information tools to be translated in different eastern European languages. Google translate can be used as a tool although translation is not perfect and requires check up.
- Collaboration with journalists and media was highlighted as very important to prevent irresponsible behaviour of the media and providing false information.

To tackle these problems participants of the CEE-GN meetings agreed upon priority projects for 2009. One of them is Preconception care- folic acid prevention in CEE countries: The objective of the CEEGN Pre-conception Workgroup is 1) to provide the inventory of current situation in the countries, 2) prepare an action plan and 3) endorse activities on folic acid prevention in the CEE countries where this is needed. Another project is Neonatal screening in CEE countries: The CEEGN should prepare the statement on the topic of neonatal screening, including the notice that we intend to extend the neonatal program and screening services to Central and Eastern European countries. The third priority is to Improve awareness and information in CEE countries: There is a reported lack of information about the disease, diagnosis, available treatments. There is no guidance for patients after diagnosis, patients do not know where to go, whom to contact etc. Some (if not many) patients get lost for follow up, there is usually no tracking system in place. In Bulgaria, the information centre called "Information Centre for Rare Diseases and Orphan Drugs (ICRDOD)" has been established under the roof of Bulgarian government and functions successfully since several years. One of the main goals of this centre is to facilitate the access of patients with rare diseases to information about the disease, and provide medical professionals with quality information about rare diseases. The centre is financed by the government (see www.raredis.org). CEEGN encourages activities for information provision and the

building national information centres in countries of Central and Eastern Europe.



Information Centre for Rare Diseases and Orphan Drugs (ICRDOD) in Plovdiv, Bulgaria.

In the context of the program of the 4th Eastern European Conference for Rare Diseases and Orphan Drugs, Plovdiv, Bulgaria, 13 - 14 June 2009, two sessions were organised in collaboration with CEEGN. If you are interested in membership of the CEEGN, please look at: www.ceegn.org

BIOVision 2009 in association with IGA
IGA's contribution - as a partner of BioVision 2009 - to the bi annual BioVision- event was the organisation of a debate dinner session on "Genetic citizenship in the age of biomedicine, biotechnology and genomics". The session took place in the prestigious building of the Chambre de Commerce et de l' Industry in Lyon, France, 9 March 2009. The well attended meeting was moderated by Ysbrand Poortman, secretary general of IGA; speakers were a.o. Alastair Kent of the European Genetic Alliances Network and Irmgard Nippert of the World Alliance of Organizations for Prevention and Treatment of Genetic Conditions. Prof. Jean-Michel Roy from Lyon participated on behalf of BioVision. Prof. Richard Roberts, Nobel Prize winner (physiology of medicine) from USA, was one of the active discussants in the meeting. Remarkably many young people attended and showed their interest for the subject. See www.biovision.org/dinnerdebates

TREAT-NMD project

TREAT-NMD is a network for people with neuromuscular diseases and professionals working in the field. This sponsored EU project could serve as a model for many other diseases. It aims to advance diagnosis and care and develop new treatments for neuromuscular diseases.

The TREAT-NMD network has 22 partners covering 11 European countries. Those partners have committed themselves to



perform specific tasks within the network. Several of those specific tasks are:

- Training and education on neuromuscular diseases (NMDs) worldwide.

- Care and Trials Site Registry. As neuromuscular diseases are very rare, clinical trials must recruit patients at multiple centres so as to achieve large enough sample sizes and have sufficient statistical power to address important clinical questions. If you know an institution in your country that would be interested in hosting potential future clinical trials for NMDs or has a population of neuromuscular disease patients that would benefit from TREAT-NMDs' care standards information, please look at: <http://www.treat-nmd.eu/patients/clinical-trials/care-and-trial-site-registry/>

- Global Patient Registry. New therapeutic strategies for NMDs are being developed and, for some of these treatments, plans for large studies involving patients from more than one country are already in place. Several new therapeutic strategies target specific genetic defects. When a clinical trial is being planned, it is important that patients suitable for that trial can be found and contacted quickly. If your organisation is interested to participate, please look at: <http://www.treat-nmd.eu/patients/patient-registries/toolkit/>

- Cataloguing existing Outcome Measures (OM). Outcome Measures are tools used to assess change in a patient over time. They measure change in a way that informs collaborative decisions about treatment. By cataloguing these outcome measures researchers can more easily find existing OM's and more rapidly assess what they do and how they are administered. It may also increase harmonisation in the selection of outcome measures for clinical trials. The

Registry is available on-line at www.researchchrom.com For help finding an outcome measure or to use the Registry, please contact the Registry Co-ordinator Joanne Auld at: Joanne.Auld@iop.kcl.ac.uk

- Biobanks. The basic scientific research being carried out on neuromuscular diseases relies on the availability of high-quality biomaterials (DNA, cells and tissue), while clinical trials rely on the availability of suitable patient cohorts. For new treatments to make their way into clinical practice for patients affected with neuromuscular disorders, it is essential that access to biomaterials is facilitated. The improvement of supranational biobanks and the introduction of European patient databases for DMD, SMA and other neuromuscular diseases are therefore major goals of this integrating activity within TREAT-NMD. The unique network of twelve biobanks from seven EU countries stores and distributes quality DNA, cell and tissue samples for scientists conducting research on rare diseases, including neuromuscular disorders. It was set up by two patient organisations, now partners of TREAT-NMD, Eurordis (European Organisation for Rare Diseases) and AFM (Association Française contre les Myopathies), You can find more information on the Biobank website: <http://www.eurobiobank.org/en/services/CatalogueHome.jsp.html> This information is available in German, English, Spanish, French, Italian, Slovenian and Hungarian.

- TREAT-NMD Newsletter. If you would like subscribe to the Newsletter that appears every two weeks in a digital format, you can go to the TREAT-NMD website: www.treat-nmd.eu You can find the subscription link at the bottom of the homepage.

- TREAT-NMD membership. When you are involved in neuromuscular diseases you can become member of TREAT-NMD free of charge by filling in the membership application form at: http://www.treat-nmd.eu/userfiles/file/general/TREAT-NMD_Membership_Application_Form.doc and to send it to Stephen Lynn to: Stephen.lynn@treat-nmd.eu or Stephen.lynn@ncl.ac.uk

From North and South America **BIO Atlanta, USA**

A contribution by Martha Carvalho; Brazilian Genetic Association

To attend BIO Atlanta was a unique opportunity for me. I represented IGA, BGA /

LAGA and AXFRA. After these days at the BIO Patient & Health Advocacy Display, immersed in a crowd of exhibitors, attendees, speakers, colleagues



From left to right: Martha Carvalho (Brazilian Genetic Association), Tricia Brooks (BIO Patient & Health Advocacy Display), James Greenwood (President & CEO Biotechnology Industry Organization) and Rod Mitchell (IGA).

etc. I feel really overwhelmed, but ideas, opportunities and challenges are quite clear on my mind to shape next steps in building / developing networks to benefit our advocacy organizations. A report on this is in preparation.

Some sessions were very good, others were "Greek" for me. Most of them seemed to be tailored for investors rather than patients. Anyway, there were lesson to be learned... This is the overview of some sessions:

- Biobriefing: Biotechnology for the Non-Science Professional
- Creating New Markets Where Doctors Don't Want Them
- Stem Cells: Translating Exciting Preclinical Data into Human Clinical Trials
- Is Venture Philanthropy the New Venture Capital?
- Diagnostics: When Disease Information Enables Treatment Decisions
- Competing Clinical Trials: When Patients Have a Choice, How Do They Decide?
- Crossing Borders and Barriers to Health Care in Developing Countries
- Anti-Biopiracy Restrictions on Patenting and Use of Genetic Resources

NEWS

Genetic Salons

The Genetic Alliance in USA informs us about a new and also an old idea to achieve (some of) our goals.

As a means of transformative visioning, Genetic Alliance in USA regularly holds salons, similar to the 17th - 19th century gatherings of intellectuals and other eminent individuals. In our case, individuals are not chosen for their expertise or the expansiveness of their resumes, but for their characters and their reputations as open-minded, curious, and willing to explore all sides of a given issue. Salons are fuelled by the mechanism of open space, allowing unfettered thought and discussion; we leave our certitude and intellectual property at the door. Through our conversation, we realize more than any one of us could have done alone. This is our chief aim: transformative knowledge through conversation with others. We have held several of these salons. The salons are an increasingly useful way to practice openness and transformation; dynamic interaction around rigorous topics inevitably leads to disruptive and exciting solutions.

Genetic citizenship

Genetics, genomics and biotechnology are rapidly unravelling the genetic background of life and are providing tools for early detection, accurate diagnosis, effective targeted treatments and for prevention of disease. Many new disciplines emerge such as predictive medicine, personalized healthcare and many -omics such as genetics, genomics, metabolomics, pharmaco genomics, nutri genomics. These developments give options for new effective strategies and tools for combating health threats also such as classical and emerging infectious diseases, viral pandemics and drug resistant pathogens.

These developments also shape a new kind of citizenship as individual and collective genetic make-ups become knowledgeable and repairable.

Every individual is in several ways involved in these developments. As a consumer of medicines produced by DNA/RNA-technology, as a decision maker regarding issues towards his/her own health and off spring, as an opinion maker on the applications and implications of new technologies such as stem cells, as a partner in trials, as a potential driver of healthcare.

In the BioVision / IGA's debate dinner session of the World Life Sciences' Forum (9 March 2009, Lyon) various aspects of

genetic citizenship were discussed such as solidarity, responsibility, human rights and duties.

Genetic citizenship implies awareness of genetic health risks and the options and the competence to handle these; implies solidarity, protection against discrimination, confidentiality of his/her health oriented information.

Investments are needed to establish awareness about genetics in society and facilities for education and empowerment of the citizens to exercise their rights and duties; moreover competent networks of genetic services integrated in primary healthcare will be more than necessary. It is in these areas that genetic alliances are active and expanding fast.

BOOKS

THE VOICE OF 12,000 PATIENTS – Experiences and Expectations of Rare Disease Patients on Diagnosis and Care in Europe

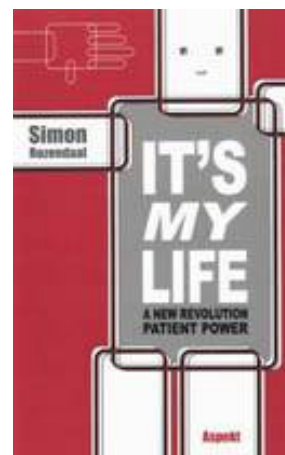
Rare diseases, of which many are also genetic diseases, are often chronic, progressive, degenerative, life-threatening and disabling diseases. Many rare disease



patients are denied their right to the highest attainable standard of health and continue to advocate their need to overcome common obstacles. Through the publication of this book, 'The Voice of 12,000 Patients', the patient's perspective can go beyond patients' anecdotes and be additionally represented by the analysis of data collected through two surveys. These surveys investigated patients' experiences and expectations regarding access to diagnosis and to health services, for a variety of significantly relevant rare diseases across Europe. This book can be downloaded fully or by sections from the Eurordis website at: http://www.eurordis.org/article.php3?id_article=1960

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.

BECOME A MEMBER OF

The international multidisciplinary community genetics network. It's 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 stimulate your colleagues to become a member, or to send me the e-mail address(es) of (the) person(s) you propose.

The **Genetic Alliance Network**. Genetic Alliance offers a wide array of programs, resources, and events for all the individuals and organizations in our network, including:

- Annual Conference, - Genetic Alliance Webinars, - MemberForum, - 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.internationalgeneticalliance.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**

17 – 19 July 2009, Washington DC, USA

Discovering Openness in Health Systems -
the annual conference of Genetic Alliance

4 – 7 October 2009, New Delhi, India

4th International Congress Birth Defects &
Disabilities in a Developing World
(ICBDD – DW)

17–19 November 2009, Brussels, Belgium

TREAT-NMD/National Institute of
Health (NIH) international conference

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