



CDG INTERNATIONAL NEWSLETTER

(January 2012)

EDITORIAL MESSAGE

Welcome to the **CDG INTERNATIONAL NEWSLETTER** presented to you on behalf of the CDG families' organizations from worldwide. Our goal is to share initiatives promoted and coordinated by nonprofit organizations, researchers and physicians that are related with CDG. We should therefore use it to raise visibility related with research, medical and advocacy issues. We encourage you to disseminate this newsletter amongst your key stakeholders.

This year, we will celebrate the **5th Rare Disease Day, on 29th February 2012**, and there are several important events worldwide. This special day for the rare disease community is the perfect occasion to announce key events that are big steps forward in our mission to raise the visibility of rare diseases and make them a public health priority both at national and International levels. Thus this newsletter is focus on several important events.

On February 24th, the annual **Sanford-Burnham Rare Disease Symposium organized by Dr Freeze** will focus on glycosylation-based disorders.

On **February 29, the first fundraising Gala Dinner** organized by the European

Organization for Rare Diseases (EURORDIS) will be held in Brussels, Belgium: capital city of the European Union. The net proceeds collected from the Gala will support community building initiatives for rare disease patients and families in Europe, help to raise public awareness about rare diseases and stimulate research by assisting patients to launch or get involved in research projects.

Finally, on **May 23-25, the European Conference on Rare Diseases and Orphan Products (ECRD 2012 Brussels)** will join the main key stakeholders in the field.

I hope you will enjoy this newsletter with such clear focus on CDG and Rare Disease developments at the International level.

Thanks. Once again, the Managing Editors wish to thank families and their organizations, researchers and physicians, who sent us their texts in order to complete this newsletter.

Vanessa Ferreira (CDG patients' voice) and Sandra Pereira Pinto (Communication & Patients Affairs Assessor, APCDG-DMR).

1. How important is the annual Sanford-Burnham Rare Disease Symposium from a research perspective?

A memorable and successful Symposium can be measured by how many new ideas are presented. It is important to present new ideas, new concepts, issues or topics to get people thinking. It is also important to present new breakthroughs in research and expand and share the understanding of any new concepts, therapies and/or data.

With this year's Symposium, we have many new topics that will be addressed. We are delighted to have a variety of distinguished speakers who will be presenting and sharing breakthrough concepts and therapies.

Another important goal for the Symposium is to prepare a program that follows a theme and presents topics and discussions that complement each other. This year's Symposium has been organized in such a way as to draw the attention and interest of the scientific community and will include all of the above key material points.

2. From your perspective, what have been the highlights and benefits achieved from prior Symposiums?

The keynote speakers have always been a highlight from prior Symposiums. Last year's keynote is actually going to be speaking at an event in conjunction with Rare Disease Day in Sioux Falls, Iowa this year. Another benefit of prior Symposiums is that everyone is a stakeholder. Although coming from different perspectives and specialties, the Symposium allows everyone to contribute and take away important and applicable information for their field.

Family organizations have also been a significant avenue of support for the Symposium, both financially, as well as in attendance. We anticipate that this year's Symposium will have the highest number of parent and families attending to date. It is impressive that family organizations recognize and support the connection between basic science and medical progress. I like to describe our non-physician Symposium speakers as "patient-concerned" scientists.

3. The 3rd annual Sanford-Burnham Rare Disease Symposium brings international researchers from around the world. What criteria were used to select the panel of speakers for this year's event? Can you share an overview of the topics that will be discussed?

The program for the Symposia is self-generated; I do not have a committee that guides the programming process. For this year's event, I wanted to focus on glycosylation disorders. The theme will encompass Congenital Disorders of Glycosylation, but other related disorders as well.

Over the past six months, I have been seeking pertinent topics and ideas, and have identified different works and therapies within the field of rare disease to include in this year's program. The program includes CDG presentations where therapies will be discussed and new findings will be revealed.

We will also have an exciting update on therapy that was reported last year from works performed at the Sanford Burnham Institute. We are finding connections between therapies that we work on for rare diseases and how they may be able to be applied to pathological processes that occur

across a much broader population. This translation means that basic science is not just affecting the 15 people who live with a particular rare disease, but that our efforts could have major implications toward affecting therapies for a broad range of more common ailments and treatments.

I am very excited about the program for this year's event. Our distinguished speakers include Dr. Esko, who will be discussing the diagnosis and treatment of mucopolysaccharidoses. Dr. Esko will be presenting exciting new methods to look for in patients who are unable to turn over glyco-molecules.

Dr. Green, director of the National Human Genome Research Institute at the NIH will be presenting a big picture talk of the Human Genomics Landscape. An additional highlight is that we have Dr. Cole, from Washington University in St. Louis joining us to talk about the technical process of how we go about discovering new genes. We are collaborating with Dr. Cole and working on research directly related to CDG.



Figure 1. Dr Hudson Freeze laboratory sent us a photo in which we can see all lab members that do research work on Congenital Disorders of Glycosylation (CDG) at the Sanford Burnham Medical Research Institute, La Jolla CA. **THANKS!**

4. Several CDG families will be attending this year's Symposium. What role will they have and can you identify specific speakers or topics that will be of particular benefit for them?

We do not have any specific role for families during the event, but are excited that this year we anticipate more families to attend which will provide them an opportunity to be in the presence of all of these exciting things happening in the research of glycosylation-based disorders. Sessions that may be of particular interest and benefit to families include Dr. Green's Keynote Address, Dr. Marquardt's presentation of new therapies specific to CDG, my presentation regarding the collaborations we have made in the realm of glycosylation disorders, and Dr. Yashari's talk, "When the Disease is Only Half the Battle".

Dr. Yashari presents a unique perspective in that she has a rare glycosylation disorder... she will talk about what it is like being a physician, being a patient, and the decisions that she is faced with regarding whether she should accept therapy now, or wait until additional studies and outcomes are available. Additionally, with this rare disorder, there is the high probability that she and others with this disorder will have children with the defect, thus leading to many additional questions... considerations to be made individually, made in concern for your children and even for future generations.

5. What are the expectations and goals for the 3rd Annual Sanford-Burnham Rare Disease Symposium?

Our primary goal is to raise awareness of glycosylation disorders and to provide an opportunity for stakeholders to gather and exchange information. For those who may be unable to attend, we will be videotaping each session and will put these videos on our website, so that they can be viewed after the event by anyone interested in a particular topic or therapy.

We will also have a Question and Answer session during the lunch hour where Dr. Marquardt and myself will be taking questions and responding to issues that are raised from the mornings sessions. We think that this will be very successful and help tie in and introduce the topics for the afternoon sessions.

Lastly, we expect to receive local press coverage for our Symposium this year. We hope that by bringing in distinguished speakers to share these new breakthroughs and therapies in rare disease, we will be able to raise awareness and impact practices and treatments far beyond that of those attending the event.

NOTE: The program of this event is available at the end of the Newsletter.

CDG AUSTRALIA: FUNDRAISING EVENT TO RAISE AWARENESS AND MONEY FOR RESEARCH ORGANISED by Remiko Prosser

Text from: Guardian Messenger newspaper



Figure 2. COURAGE AND GRACE: Allira Prosser is defying the limitations of her disease, buoyed by the love of family and friends led by her mum Remiko Prosser Picture: Roger Wyman H0370662

LITTLE Allira Prosser was not expected to live to see her second birthday. Born on April 8, 2009, Allira was only one when diagnosed with the rare and incurable disease congenital disorder of glycosylation (CDG).

“At present Allira is unable to talk, walk, crawl, sit or eat, yet she has an amazing personality and no matter what, Allira’s smiles and giggles always light up the room,” her mother, Remiko Prosser, says. Allira is already beating the odds and in June this year she was discharged from palliative care. The doctors are uncertain of her new life expectancy. There are only 10 children in Australia with CDG and 700 cases worldwide. Mrs Prosser and seven friends formed Allira’s Angels Foundation to raise awareness and money for more research. “We started Allira’s Angels nine months ago to promote awareness of CDG and to communicate Allira’s grace and strength amongst the struggle,” Mrs Prosser says. The Allira’s Angels Foundation held its first fundraising event on November 11, raising \$14,500. Half of the money will go to supporting Allira and the rest to the Women’s and Children’s Hospital’s palliative care section.



Figure 3: Banner from Allira’s Angels Foundation event. **Follow Allira’s Angels on Facebook at www.facebook.com/pages/Alliras-Angels**

**INTERVIEW WITH ROBERT PLETICHA,
ONLINE PATIENT COMMUNITIES MANAGER AT EURORDIS by
Vanessa Ferreira**



1. What is the main goal of the Rare connect community?

The main goal of RareConnect: the Rare Disease Communities project is to create disease specific social networks in partnership with leading international patient organisations so that a vibrant international movement towards improving the lives of people living with these diseases can develop. In order to facilitate international networking patients, caregivers, and patient groups can exchange messages on RareConnect and have their messages translated by humans.

2. Which role do associations have or can have?

Associations play the key role of creating, moderating, and maintaining the community along with support staff from EURORDIS and NORD. Associations are the experts in their diseases and RareConnect wants to make sure that patients are finding and in communication with these associations and getting the quality information they need to make difficult decisions from the correct source.

3. Which are the advantages of this online social network compared with others such as facebook, for example?

Facebook has changed the way we all spend time online. But it has its limitations. Anyone can create a group on Facebook and begin to exchange potentially dangerous advice. Many groups for various rare diseases are only available in English. Advertisements can sometimes be insensitive and intrusive.

A RareConnect community is only created in partnership with leading international patient groups that are trained in moderation and ensure quality advice or information is being exchanged on the forums. The site is in 5 languages: French, English, Spanish, Italian, and German so that everyone can join the global conversation. There are no advertisements allowed and the Community should be a place where people feel safe.

4. How would you describe your role as Online Patient Communities Manager?

As the Online Patient Communities Manager, I work on aspects of the project such as: support of existing communities, creation of new content, outreach to new communities, presentation of the project to patient groups at conferences, facilitation of translation services, moderation of new messages, and trying to help network people so that they can find the information they are looking for.

5. Which are the short-term plans for the rare connect platform?

We will change the URL from rarediseasecommunities.org to rareconnect.org this Spring. As part of that, we are in the process of creating a new portal homepage and a new logo. This project is always looking for user feedback and to better serve the people affected by rare diseases and their families. Any ideas or suggestions on improving the CDG Community can be sent to robert.pleticha@eurordis.org

CDG Community:

Several videos from Dr Belén Pérez Dueñas, Dr Rafael Artuch and Dr Mercedes Serrano are now available, please check:

<http://www.rarediseasecommunities.org/en/community/cdg/article/new-videos-on-cdg>

CDG AT THE EUROPEAN LEVEL: AWARDS TO CDG RESEARCHERS AND MEDICAL DOCTORS



We are pleased to announce that CDG families voted for two categories of awards that EURORDIS will announce during the **Eurordis Gala dinner 2012**. One category is the **European Rare Disease Leadership Award** (An individual having demonstrated remarkable leadership on a European level in the domain of rare diseases) and the other is the **Scientific Award** (A distinguished researcher whose career represents a unique blend of scientific excellence and support of the patient community).

The event will be held in Brussels and the operating costs to organise and host the EURORDIS Gala Dinner will be covered by corporate philanthropy. The net proceeds collected from the Gala will be used to support or initiate the following three actions:

- Breaking the isolation of rare disease patients and their families through the development of European networking and international patient communities;
- Empowering leaders of rare disease communities through training, capacity-building activities and seed money to foster their research and therapeutic development activities;
- Increasing public awareness through our international communication campaign, Rare Disease Day, which takes place every year on the last day of February.*

*Text from: <http://galadinner.eurordis.org/>

FRENCH ASSOCIATION FOR CDG: THE FRENCH VERSION FOR THE "CDG AWARENESS AND DISSEMINATION KIT" IS NOW AVAILABLE!

Traduction du CDG kit de dissémination en français

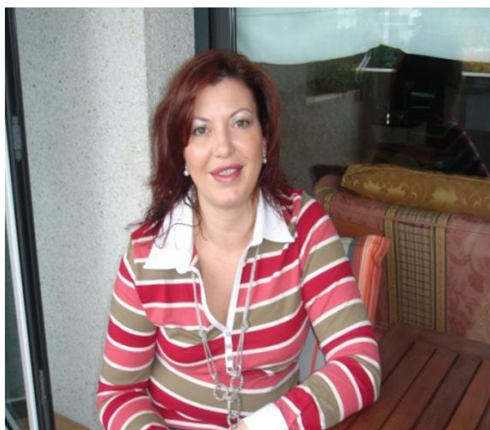
"L'objectif de la toute jeune association française "Les P'tits CDG" est simple : jouer la complémentarité avec le tissu associatif existant, principalement orienté vers la recherche, pour permettre aux familles françaises concernées par le CDG Syndrome de se rencontrer, d'échanger leur expérience et d'organiser des événements pour les enfants atteints. Nous relaierons également des informations sur l'avancée médicale et les actions des associations-amies (spécifiquement dédiée au CDG Syndrome ou, plus largement, aux syndromes cérébelleux).

L'une de nos premières actions a été d'ailleurs de favoriser la diffusion de la connaissance du CDG Syndrome en assurant la traduction du kit de dissémination en français qui est téléchargeable sur notre site (<http://www.lesptitscdg.org>) ou sur [rarediseasecommunities.org](http://www.rarediseasecommunities.org)."

On behalf of CDG patient's voice, we are grateful to Nathalie Saint-Alme ("Les P'tits CDG"), who was the person responsible for translating the "CDG AWARENESS AND DISSEMINATION" Kit in French. **THANKS!**

Rachel Levillain, President for "Les P'tits CDG" shares her experience as mother at the www.rarediseasecommunities.org/en/community/

SPANISH ASSOCIATION FOR CDG:
Yolanda Scott was elected president for the AESCDG
(Asociación Española Síndrome CDG)



Yolanda Scott lives in Barcelona (Spain) and recently she was elected President for the AESCDG. Fundraising initiatives are one of the main goals that she wants to implement in the near future. THANKS!

The **AESCDG** association is a non-profit organizations founded by parents seeking information and support for Congenital Disorders of Glycosylation. Some goals are:

- To offer practical help, promoting the exchange of information between families and physicians;
- To provide high quality up to date information about best practice in treatment and care;
- To promote greater understanding and awareness of CDG syndrome amongst the general public and amongst medical professionals.

For more information:

<http://webs.ono.com/aescdg/>

Facebook: AESCDG

PORTUGUESE ASSOCIATION FOR CDG: New members of the Communication and Patients affairs Department



Sandra Pinto, lives in between Porto (Portugal) and Madrid (Spain) and she is an active member in the Portuguese and Spanish Associations for CDG. Recently she joined our working team.

She participated in several events at EMA and Eurordis as CDG patient representative. In addition she organised several communication and dissemination activities. THANKS!

Sandra Pinto is now the Communication Assessor and Patient Affairs at the Portuguese Association for CDG and other Rare Metabolic Diseases.

Sandra will play an essential role to all CDG community. She will develop an overall communications and public affairs strategy targeted to our community.

Generally, she will lead all communications activities including corporate communications/media relations, sponsorships and events, community and philanthropic activities. She will also support the government affairs initiatives at the International level. Furthermore, she is responsible for orchestrating communications and public affairs activities.

Finally, she will intervene, assist, manage and fight for our community needs. This task will be performed in collaboration with Rosália Félix (Advocate for Patients with Rare Metabolic Diseases).

Contact: communicationapcdg@gmail.com



Rosália Félix, lives in Lisbon (Portugal). She is vice-president for the Portuguese Association for CDG and other Rare Metabolic Diseases. She is mother of a CDG adult which allows her to have the perfectly knowledge concerning the needs of our community. THANKS!

Rosália Félix is now Advocate for Patients with Rare Metabolic Diseases

Some of her tasks are:

- Coordinate communication between patients, family members, medical staff, administrative staff, or regulatory agencies.
- To identify problems relating to care.
- To maintain knowledge of community services and resources available to patients.
- To Refer patients to appropriate health care services or resources.
- To investigate and direct patient inquiries or complaints to appropriate medical staff members and follow up to ensure satisfactory resolution.

Contact: defesadopacienteapcdg@gmail.com

Please feel free to suggest any topics or ideas that you might like to discuss with Sandra Pinto (Portuguese, Spanish and English) or Rosália Félix (Portuguese, French and Spanish).

RESEARCH:

The International Rare Disease Research Consortium (IRDiRC)



Integrating national funding for rare disease research within common international goals and policies.

IRDiRC Goal's:

IRDiRC will team up researchers and funding agencies in order to achieve two main objectives by the year 2020, namely to deliver 200 new therapies for rare diseases and diagnostic tools for most rare diseases.

A number of grand challenges will need to be addressed through collaborative actions to reach these 2020 goals such as: establishing and providing access to harmonised data and samples, performing the molecular and clinical characterisation of rare diseases, boosting translational, preclinical and clinical research, and streamlining ethical and regulatory procedures.*

More information:

*http://ec.europa.eu/research/health/medical-research/rare-diseases/irdirc_en.html

PATIENT ADVOCACY: IMPROVING CARE FOR PATIENTS WITH CDG



The Portuguese Association for CDG and other Rare Metabolic Diseases (APCDG-DMR) identified a barrier when patients with a chronic disease, such as CDG, want to access their Family Medical Doctor in some Portuguese Healthcare Centers: basically priority was not given in several emergency Healthcare Center appointments. A meeting with the "Citizen Office" Department (Gabinete do Cidadão) took place February, 2nd to further discuss this subject.

The law in Portugal related to priority in the healthcare centers is the following:

Decreto-Lei n.º 135/99 de 22 de Abril

O presente diploma aplica-se a todos os serviços da administração central, regional e local, bem como aos institutos públicos nas modalidades de serviços personalizados do Estado ou de fundos públicos.

Artigo 9.º Prioridades no atendimento

1 – Deve ser dada prioridade ao atendimento dos idosos, doentes, grávidas, pessoas com deficiência ou acompanhadas de crianças.

CDG ARTICLES ON PUBMED JANUARY 2012

5-Thiomannosides Block the Biosynthesis of Dolichol-Linked Oligosaccharides and Mimic Class I Congenital Disorders of Glycosylation.

Zandberg WF, Gao N, Kumarasamy J, Lehrman MA, Seidah NG, Pinto BM.
Chembiochem. 2012 Jan 19. doi: 10.1002/cbic.201100647. [Epub ahead of print]

SRD5A3-CDG: A patient with a novel mutation.

Kasapkara CS, Tümer L, Ezgü FS, Hasanoğlu A, Race V, Matthijs G, Jaeken J.
Eur J Paediatr Neurol. 2012 Jan 10. [Epub ahead of print]

If you are interested to receive these articles send an email to: sindromecdg@gmail.com

**ARTICLE RECOMMEND BY DR DIRK LEFEBER,
Nijmegen Center for Disorders of Glycosylation in Netherlands**



Dr Dirk Lefeber is Clinical Biochemical Geneticist at the Nijmegen Center for Disorders of Glycosylation in Netherlands.

In CDG ONLINE COMMUNITY you can find Dr Dirk's contribution about the Exome sequencing as diagnostic tool.

<http://www.rarediseasecommunities.org/en/community/cdg>

<http://www.nijmegencdg.nl/>

Autosomal Recessive Dilated Cardiomyopathy due to *DOLK* Mutations Results from Abnormal Dystroglycan O-Mannosylation

Dirk J. Lefeber^{1,2,3*}, Arjan P. M. de Brouwer^{3,4}, Eva Morava⁴, Moniek Riemersma¹, Janneke H. M. Schuurs-Hoeijmakers³, Birgit Absmanner⁵, Kiek Verrijp⁶, Willem M. R. van den Akker³, Karin Huijben², Gerry Steenbergen², Jeroen van Reeuwijk³, Adam Jozwiak⁷, Nili Zucker⁸, Avraham Lorber⁹, Martin Lammens⁶, Carlos Knopf⁹, Hans van Bokhoven³, Stephanie Grünwald¹⁰, Ludwig Lehle⁵, Livia Kapusta¹¹, Hanna Mandel⁹†, Ron A. Wevers^{2†}

1 Department of Neurology, Institute for Genetic and Metabolic Disease, Radboud University Nijmegen Medical Centre, Nijmegen, The Netherlands, **2** Department of Laboratory Medicine, Institute for Genetic and Metabolic Disease, Radboud University Nijmegen Medical Centre, Nijmegen, The Netherlands, **3** Department of Human Genetics, Nijmegen Centre for Molecular Life Sciences, Donders Institute for Brain, Cognition, and Behaviour, Nijmegen, The Netherlands, **4** Department of Pediatrics, Institute for Genetic and Metabolic Disease, Radboud University Nijmegen Medical Centre, Nijmegen, The Netherlands, **5** Department of Cell Biology and Plant Biochemistry, University of Regensburg, Regensburg, Germany, **6** Department of Pathology, Radboud University Nijmegen Medical Centre, Nijmegen, The Netherlands, **7** Institute of Biochemistry and Biophysics, Polish Academy of Sciences, Warsaw, Poland, **8** Heart Institute, Schneider Children's Medical Centre of Israel, Petach Tikva, Israel, **9** Metabolic Unit and Unit of Pediatric Cardiology, Meyer Children's Hospital, Rambam Medical Center, Technion-Israel Institute of Technology, Haifa, Israel, **10** Great Ormond Street Hospital, UCL Institute of Child Health, University College London, London, United Kingdom, **11** Pediatric Cardiology Unit, Edith Wolfson Medical Center, Holon, Israel

UPCOMING MEETINGS, WORKSHOPS AND CONFERENCES

RARE DISEASE DAY, 29 FEBRUARY

<http://www.rarediseaseday.org/>

There you can find out about activities going on in your country, upload a photo or video, and tell your story. Give it a Like on Facebook or Like the [Rare Disease Day Facebook page](#) to share it with your contacts.

Send this link to your webmaster for other patient group's webpage, on adding a **RDD banner** to your website:

<http://www.rarediseaseday.org/article/place-ribbon-rare-disease-day-on-your-blog-website>

3rd Annual Sanford-Burnham Rare Disease Symposium, February 24, 2012

Sanford | Burnham
Medical Research Institute

3rd Annual Sanford-Burnham
Rare Disease
Symposium
10905 Road to the
Cure
San Diego, CA
92121
858-646-3100

Identifying and Treating Rare Disorders*

*Please note that this is a preliminary program – presentation titles and speaking order are subject to change.

8:00 – 9:00	REGISTRATION	
9:00-1:00	Session 1: Science, Medicine and Therapy-I	
	9:00-9:05	Introduction and Welcome Hudson Freeze, Ph.D., Sanford-Burnham Medical Research Institute
	9:05-9:10	Opening Remarks John C. Reed, Ph.D., Sanford-Burnham Medical Research Institute
	9:10-9:45	KEYNOTE ADDRESS The Human Genomics Landscape: From Base Pairs to Clinical Applications Eric Green, M.D., Ph.D., Director, National Human Genome Research Institute, NIH
	9:45-10:15	N-acetylmannosamine (ManNAc) or Sialic Acid as Therapy for Disorders of Hyposialylation Marjan Huizing, Ph.D., National Human Genome Research Institute
	10:15-10:45	The Sugar Pill – Treatment of Congenital Disorders of Glycosylation Thorsten Marquardt, M.D., University of Münster, Germany
	10:45-11:15	“Disease in a Dish” Drug Screens for Dystroglycanopathies Michael Jackson, Ph.D., Sanford-Burnham Medical Research Institute
	11:15-11:30	BREAK
	11:30-12:00	Exome Sequencing for Novel Disease Gene Discovery in Families with Rare Mendelian Phenotypes Sessions Cole, M.D., Washington University in St. Louis
	12:00-12:30	Novel Rare Defects of the Human Immune System – from Genes to Therapy Christoph Klein, Ph.D., Ludwig-Maximilians-University, Munich, Germany
	12:30-1:00	Common Pathways in Neurodegeneration: Focus on Lysosomes Dimitri Krainc, M.D., Ph.D., Massachusetts General Hospital, Boston
1:00-2:00	LUNCH	
2:00-3:45	Session 2: Science Medicine and Therapy-II	
	2:00-2:30	Diagnosis and Treatment of Mucopolysaccharidoses Jeffrey Esko, Ph.D., University of California, San Diego
	2:30-3:00	Heparan Sulfate, Hereditary Multiple Exostoses, and Autism: Insight from a Rare Disease for a Common Disease Yu Yamaguchi, M.D., Ph.D., Sanford-Burnham Medical Research Institute
	3:00-3:30	The La Jolla Collaboration on Glycosylation Disorders Hudson Freeze, Ph.D., Sanford-Burnham Medical Research Institute
	3:30-3:45	Updates on Successful Treatment of Hypophosphatasia Enobia Pharma
	3:45-4:00	BREAK
4:00-5:00	Session 3: Advocates, Funding, and Philanthropy	
	4:00-4:30	2-15 Minute Presentations** Neuromuscular Disease Foundation
	4:30-5:00	Panel Discussion: Philanthropy in Rare Disease Research Participants TBD

The 6th European Conference on Rare Diseases and Orphan Products (ECD 2012)
will be held on **23 – 25 May 2012** at the **MCE Conference Centre** in **Brussels, Belgium**.

The European Conference on Rare Diseases & Orphan Products is the unique platform/forum across all rare diseases, across all European countries, bringing together all stakeholders - academics, health care professionals, industry, policy makers, patients' representatives.

It is an annual event, providing the state-of-the-art of the rare disease environment, monitoring and benchmarking initiatives. It covers research, development of new treatments, health care, social care, information, public health and support at European, national and regional levels. It is synergistic with national and regional conferences, enhancing efforts of all stakeholders. There is no competition with them, but efforts are complementary, fully respecting initiatives of all. *

* <http://www.rare-diseases.eu/2012/6th-European-Conference-on-Rare>

CDG Family Network Summer Conference!
June 22, 23 & 24
Chicago, IL

Save the Date for the CDG Family Network Summer Conference!

June 22, 23 & 24

Chicago, IL



And finally:

CONGRATULATIONS TO.....

Hall of fame - Prof Dr. Gert Matthijs

Dr. Matthijs has a PhD in pharmaceutical sciences and is a certified specialist in clinical molecular genetics. He is the head of the Laboratory for Molecular Diagnostics at the Center for Human Genetics in Leuven, Belgium, and a professor at the University of Leuven. His research interest is in CDG. The gene for CDG-Ia was first cloned in his laboratory, back in 1996, and his group has been involved in the elucidation of different other types of CDG. He is also the Coordinator of EUROGLYCANET, a European Network for the systematic study of CDG and related diseases (www.euroglycanet.org). The network offers expert testing for CDG, and is of course also interested in novel types of the disease. Leuven acts as a reference centre in the context of the network. The Laboratory for Molecular Diagnostics is offering genetic testing for the different types of CDG, including prenatal diagnosis.

Hall of fame - Prof Dr Jaak Jaeken

Dr. Jaak Jaeken is a full Professor at the Faculty of Medicine at the University of Leuven, Belgium. He studied Pediatric Medicine at the University of Leuven (Katholieke Universiteit Leuven) where he received his MD and PhD. After completing his Fellowship at the University of Zurich he became Director of the Center for Metabolic Diseases at the University of Leuven. In 1980 Dr. Jaeken was the very first doctor to publish his description of CDG. Today Dr. Jaeken continues to collect and examine new cases of this rapidly expanding group of genetic diseases and test new methods of diagnosis and therapy on patients when they are available.



The CDG Patient's Voice

Links from different associations:

Portugal: <http://sindromecdg.orgfree.com/>

Spain: http://webs.ono.com/aescdg/SINDROME_CDG/Bienvenidos.html

France: <http://www.lesptitscdg.org/>

USA: <http://www.cdgfamilynetwork.org/>

Canada: <http://www.thefog.ca/>
<http://thelittlefightersfoundation.com/>

Germany: <https://www.cdg-syndrom.de/>

Denmark: <http://www.cdgforeningen.dk/>

Sweden: <http://www.cdgs.se/>



CDG UNITED

Grupo cerrado — glycosylation@groups.facebook.com

227 miembros 212 fotos 1 documento