

***“Conference on Congenital Disorders of
Glycosylation for Families and Professionals:
a booming story of sugar trees:
a unique event!”***



***31 August 2013:
Informal Families Meeting***

***1 and 2 September 2013:
Satellite Symposia
Centro Cívico la Sedeta,
Barcelona, Spain***

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This summary is available at Metabolic Guide- CDG section:

<http://www.guiametabolica.org/subhome-enfermedad/defectos-congenitos-de-la-glicosilacion-cdg>

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During the first week of September 2013, three important conferences / meetings on diseases classified as Inborn Errors of Metabolism (IEM) have been held in Barcelona.

The ICIEM2013 gathered over 2300 metabolic specialists from all over the world. More than 90 countries were represented. During this 4-days conference more than 900 communications in oral or poster format were presented.

In the next weeks, the Metabolic Guide will publish several articles on the different sessions, highlighting new developments for each disease group.

We begin this week with the **“The 1st World Conference on Congenital Disorders of Glycosylation for Families and Professionals: a booming story of sugar trees”**, followed by the **“4th International Symposium on Urea Cycle Disorders: Catalyzing New Therapeutic Approaches”**.



The first World Informal CDG families meeting

August 31, was an unforgettable day. Many families met for the first time other CDG families. Others knew each other through social networks. Together they have walked the streets of the emblematic neighborhood Gràcia until Plaza Catalunya. The day finished with a wonderful typical Catalan dinner. It was an emotional and enriching moment!

Satellite Symposia: “1st World Conference on Congenital Disorders of Glycosylation for Families and Professionals”

“The 1st World Conference on Congenital Disorders of Glycosylation for Families and Professionals: a booming story of sugar trees” was fruitfully designed in a collaboration with leading experts in the field of CDG: families and professionals worked together to exchange knowledge, experiences, needs and perspectives. The coordination and overall operations have been coordinated by the Portuguese Association for CDG and related Rare Metabolic Diseases (APCDG-DMR). A big “thank you” must go to all the volunteers who dedicated their precious time before, during and after the conference. Their time, their willingness to participate, encouragement and advice were very significant to the overall organization of this event. This symposium had four main objectives:

- (1) strengthening global awareness of these diseases,
- (2) fighting geographic isolation that impacts families and professionals,
- (3) sharing knowledge and experiences,
- (4) potentiating visibility of the work performed by patient representatives and professionals and
- (5) identifying areas in which international collaboration can have an immediate impact on the life quality of these families.

This event was very special since it was organized by a committee consisting of worldwide patient representatives and professionals. It should be noted that this congress was a satellite meeting of the 12th International Congress of Inborn Errors of Metabolism.

The **“1st World Conference on Congenital Disorders of Glycosylation for Families and Professionals”** has been held on 1 and 2 September 2013, in Barcelona. It were interesting days where families and professionals, (clinicians, clinical and basic researchers, biologists, chemists and pharmacologists ... had the opportunity to share their knowledge and experiences. The participants consisted of:

- 43 families from 17 countries: Australia, Belgium, Brazil, Canada, Czech Republic, Denmark, England, Finland, France, Germany, Italy, Israel, Norway, the Netherlands, Portugal, Spain, Sweden and USA.
- 71 professionals from all continents and from 25 countries.

Outreaching professionals: total 71	
Europe	
France, Spain, Sweden, Germany, Italy, UK, Portugal, Serbia, Czech Republic, Netherlands, Belgium, Slovakia, Bulgaria	
South America	North and Central America
Brazil, Uruguay, Argentina, Chile, Colombia	Canada, USA and México
Asia	Middle East
Hong Kong	Qatar, Turkey
Australia	
25 countries	

CDG Participatory Medicine

During the conference, it became clear that joining forces between patients, physicians and basic researchers, represents an invaluable added value in the fight against these diseases.

CDG-ePACIBARD (ePatients Generating Collective Intelligence to Advance Biomedical Rare Diseases) will be officially launched on the International Rare Disease Day (28 February 2014): on this day, families will start to complete surveys aimed at expanding the current knowledge about CDG (clinical description, education, rehabilitation, and other disease management). Currently, families, patient organizations and professionals are filling in the scientific content, and are carrying out various actions to collect funds for this project. **This is a large project that aims to benefit patients and their families.** Invitations amongst main stakeholders are currently being sent out. International collaborations are urgently needed to achieve our goals in particular disseminating knowledge, and stimulating research on all aspects of these diseases in order to improve patients' and families' quality of life. If you want to become a collaborator please contact sindromecdg@gmail.com

"CDG Gala Awards: Hope and Dreams: represented by Tenacity, Love, Motivation and Fight"



"CDG Gala Awards: Hope and Dreams" were the first-ever CDG awards honouring persons who have made significant contributions to CDG worldwide by improving CDG knowledge and by basic and applied research hence improving CDG families' quality of life.

The audience heard a poem written by Julia Boonnak (CDG UK). Julia was the winner of a poetry contest Global Genes Project in June 2013. Her poem focuses on perseverance and can be read at: <http://globalgenes.org/poetry-contest-winners-announced/>. Julia also writes her own blog focused on CDG awareness <http://cantdogymnastics.blogspot.fr/>

The categories and assigned people have been:

1. **CDG medical care award** (recognizes breakthrough work done by a medical doctor in the field of CDG). : Dr Eva Morava.
2. **CDG research award** (recognizes an individual for excellent CDG research, international collaboration, and support of the patient community): Dr Hudson Freeze.
3. **CDG Lifetime Award** (for lifelong dedication and commitment to CDG (this award is already assigned): Peleman family.
4. **CDG inspirational award** (celebrates the achievements of (a) remarkable person(s) who inspire those around them): the CDG Families.



Awards were attributed to Dr. E. Morava (USA), Dr. H Freeze (USA), the Peleman family (whose daughters were the first to be diagnosed with the disease, Belgium), and to the families in general (Dr. Vanessa Ferreira received this award on their behalf). In addition, special awards were given to Drs. J. Jaeken (Leuven, Belgium), M-A. Vilaseca (Barcelona, Spain) and P. Briones (Barcelona, Spain) for their dedication and passion during their professional careers for this group of diseases.

Dr Mercedes Serrano dedicated the following words to Dr Vilaseca:

“She took care of patients and always ensured that she would be close to them. She remembers every single name, issues about their families, their lives... She has always thought about patients’ daily care, their difficulties in life and their quality of life.

Throughout her career she has been brilliant from the scientific and professional point of view, but particularly, she has been brilliant as a person.

3 years ago she founded Guía Metabólica, once more trying to help patients, trying to solve their needs and difficulties, and nowadays she is the most “2.0 doctor” in the hospital! It is our great pleasure to give this recognition to our loved María Antona Vilaseca (MAV)”. Text by Dr Mercedes Serrano.

Unique speakers, unique moments:

The most rewarding and emotional moment was the opportunity to hear oral presentations from families and patients, focused to share their experiences.

Regarding the contributions of families and patients, Morgan Liddle (Australia), a CDG patient, aged 22, told us her expectations about life and her right to dream. A presentation where optimism and strength impressed all attendees. Next, Bas Holten (The Netherlands), Duncan Webster (Canada), Sandra Pereira Pinto (Spain), Noelle Schmitz (The Netherlands), Eric Jerman (USA) and Vanessa Ferreira (Portugal) explained their contributions to the daily life of children from a personal, familial or professional point of view. Examples are the design of adapted playgrounds, electronic tablet apps to promote the neurodevelopment of children, the establishment of a support group on social networks for CDG families. It was also demonstrated how the active participation of families in the field of basic and clinical research, through their possible collaboration as coordinators, managers, source of inspiration, funders, and others, can positively catalyze research projects based on the immediate needs and concerns of families.

At the end of the afternoon there was also time to talk about collaboration between patients using RareConnect platform (a EURORDIS and NORD initiative) or through Metabolic Guide as pioneer initiatives that use new technologies to advance clinical knowledge of these pathologies.

The first day closed with an emotional video made by Merell Liddle (CDG Australia) in collaboration with Vanessa Ferreira (CDG Portugal). It included more than 80 pictures of CDG children. The video transmitted the thoughts of the families at the beginning of their diagnosis and their current feelings and desires. It was accompanied by a folk song composed by the parents of two CDG girls (Australia) entitled "Ordinary Life" (to listen it: <http://www.youtube.com/watch?v=g9W-FyodPo4>).

The image shows a composite of two web pages. On the left is the 'TeamMorgan' website, featuring the logo 'Morgan TEAM' and the name 'Morgan Webb Liddle, Para-Equestrian'. It includes navigation links like 'HOME | OTHER LINKS | CONTACT US' and sections for 'LATEST FACEBOOK NEWS' and 'LATEST NEWS'. The Facebook news section shows two posts about Morgan Webb Liddle and her supporters, TeamMORGAN, with one post mentioning 'Cross training on the bike with Taylor' and an image of Morgan with a horse. On the right is a Facebook page for 'MORGAN', showing a profile picture of Morgan Webb Liddle smiling next to a white horse. The page has tabs for 'MORGAN', 'ATHLETE', 'COMPETITOR', and 'SUPPORT'. Below the tabs are two small video thumbnails and a larger video player titled 'VIDEO NEWS' showing Morgan and a horse. A quote is visible: 'Despite all my challenges, my goal is to represent my country.'

Together we can help Morgan achieving her dream! Please visit the following links and if you are interested donations can be done through her webpage:

Webpage: <http://www.teammorgan.com.au/>

Facebook : <https://www.facebook.com/pages/Morgan-Webb-Liddle-Para-Equestrian-and-her-supporters-TeamMORGAN/145308438889087?fref=ts>

Youtube channel: <https://www.youtube.com/user/TEAMmorgan28?feature=mhee>

The group BandSambant closed this unforgettable day with a Batucada Show!



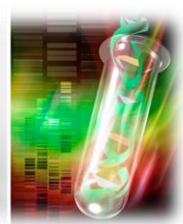
Morgan Liddle (Australia) with Elisa Balducci from BandSambant group.

Last news related to the Scientific and Medical content:

As to the medical content, the first presentation was given by Dr. Jaak Jaeken (Belgium), who discovered the first congenital disorder of glycosylation, and several others. Dr Jaeken, gave a short history on the first 20 years of CDG, and a concise overview of this rapidly expanding field, comprising actually more than 70 subtypes.

Subsequently, representatives from Argentina (Dr Carla Asteggiano), Brazil (Dr Charles Lourenço), Portugal (Dr Esmeralda Rodrigues), Spain (Dr Belén Pérez-Dueñas), France (Dr Nathelie Seta), Qatar (Dr Tawfeg Ben-Omran), Italy (Dr Rita Barone and Dr Ágata Fiumara), Bulgaria (Dr Malina Stancheva) and the Czech Republic (Dr Thomas Honzik) described their experiences. These talks nicely illustrated that CDG can affect every organ, particularly the nervous system, and that the disease severity comprises a very broad spectrum (from very mildly affected adults to prenatal death).

Dr. Patterson's (USA) talked about the neurological aspects and evolution of CDG, and in particular about the cerebellar involvement in PMM2-CDG. This involvement is most probably a combination of hypoplasia (poor formation) and atrophy (destruction of the tissue after formation). In particular the Purkinje cells, the most important neurons in the cerebellum, are very vulnerable to injury. In its presentation Dr Patterson also noted that there is an alteration not only at the cerebellar and cerebral level (which could account for the increased risk of epilepsy in this group of patients), but also at the level of the peripheral nerves in the form of demyelinating neuropathy (loss of the nerve sheath that protects these neurons and makes them "drive" with greater speed and efficiency).



Dr. Daisy Rymen (Belgium) explained the different ocular manifestations of PMM2-CDG (CDG-Ia), mainly strabismus, nystagmus (abnormal horizontal eye movements), retinitis pigmentosa (degeneration of the retina, with deposition of pigment and loss of photoreceptors that are neurons able to capture the light signals). Dr Rymen also spoke about myopia, cataracts (usually present in other CDG than PMM2-CDG (CDG-Ia), coloboma (structural defect in any of the ocular structures: iris, retina, optic nerve) and hypoplasia (lack of formation) of the optic nerve.

Dr. Dulce Quelhas (Portugal) explained in a non-specialized language the Isoelectrofocusing (IEF) of serum transferrin, a technique widely used for CDG diagnosis. Dr Hudson Freeze (USA) described the CDG with recessive or X-linked inheritance. He highlighted new CDG with therapeutic possibilities by dietary treatment with fucose or galactose. During his talk he emphasized the need for collaboration between patients, doctors and researchers who, like him, work in a basic research laboratory.

Dr. Lefeber (The Netherlands) presented methods that are currently being used to improve CDG diagnosis and that have enabled the discovery of new types of CDG. An important goal is to ensure an early diagnosis (ideally less than three months).

The Drs. Corral and Morena Barrio (Spain), explained their studies of coagulation and hemostasis in patients with PMM2-CDG (CDG Ia). The coagulation factors need to be properly glycosylated to correctly perform their function. Coagulation requires a delicate balance between procoagulant and anticoagulant factors. PMM2-CDG patients are at risk of vascular events (mostly thrombosis) in situations such as prolonged fever, dehydration or immobilization. Studies in this laboratory not only imply alterations in the patients themselves, but also in the family members who are carriers.

Dr M Mohamed (Netherlands) presented a scale for clinical assessment of patients. This tool is of great importance to assess the development of the disease or the evaluation of new therapies.

Dr Paula Videira (Portugal) reviewed the importance of glycosylation for proper functioning of the normal immune system. In generally these patients have more bacterial infections, particularly in the early years of life. She stressed the need to follow a regular vaccination schedule, if permitted by the patient's condition.

Drs Eva Morava (USA) and Thorsten Marquardt (Germany) spoke about therapeutic studies. Dr Morava reported on a treatment with galactose of a novel CDG namely PGM1-CDG. Dr Marquardt discussed his preliminary studies with mannose and oral hypoglycemic agents (metformin) or disulfiram in PMM2-CDG (CDG-Ia) patients. He found evidence for a reduction in non-glycosylated proteins during two years of treatment with both drugs in two patients, but there were no clinical changes. No long term studies have been performed yet with mannose only or with both drugs simultaneously. A number of questions were asked by the parents about this therapy. Together with Dr. Stephanie Grünwald (England) and Dr. Eva Morava (USA), both international CDG experts, a joint recommendation was reached: we must waiting for conclusive results and be cautious.

At this moment, there is no scientific evidence in favor of this therapy in children with the PMM2-CDG (CDG-Ia). On the other hand, we have also to consider risks of anti-diabetic drug in children with problems such as nausea, anorexia, diarrhea, and lactic acidosis.



Dr Marc Martinell (MINORYX, Spain) and John Evans (Agiros Pharmaceuticals, USA) presented the progress of the pharmaceutical industry in the development of drugs to combat these diseases, with the emphasis on the possibilities of chaperones. Dr Körner (Germany) closed the last day of the symposium by sharing his experience in the field of animal models mimicking PMM2-CDG (CDG-Ia). They are important for the study of physiological and anatomical processes and for pharmaceutical drug development, and for other purposes.

Finally, new and pioneer communication, education, awareness and empowerment tools available in the reference platform for this group of diseases, named METABOLIC GUIDE, have been shared with the audience. The section dedicated to CDG is:

<http://www.guiametabolica.org/subhome-enfermedad/defectos-congenitos-de-la-glicosilacion-cdg>

Some examples are:

- (1) The story dedicated to CDG and now available in **11 languages** can be downloaded at the following link <http://www.guiametabolica.org/aprender-jugando/glicolandia-y-las-antenas-de-colores>
- (2) Brochures in four languages: http://www.guiametabolica.org/sites/default/files/CDG_EN_DIP.pdf or
- (3) The practical guide for CDG families: http://www.guiametabolica.org/sites/default/files/guia_practica_familias_CDG.pdf

The first world conference on CDG was an exciting event both from a scientific and human perspective. This experience proved how limited resources (human and economic) can result in a conference with a high level of professionalism. **This conference was dedicated to all CDG families that in their daily life have to face challenges resulting from a disease with major medical needs. Family love, tenacity and bravery made coordinators believe in the power of dreams.** It was a milestone that will motivate many other associations of patients with rare diseases. In addition, it prompted the interest of several families in Brazil, England and Italy to form their own associations and consequently to pool efforts and resources together within organizations and CDG representatives worldwide.

Foundation Glycosylation (FoG) is the official sponsor of the videos targeted to the “The 1st World Conference on Congenital Disorders of Glycosylation for Families and Professionals”:



The organisers are pleased to announce that the Foundation Glycosylation (FoG) founded by Duncan Webster (Canada), is the official sponsor of the videos that are currently being prepared. This material will be available in the future Youtube channel dedicated to the **“The 1st World Conference on Congenital Disorders of Glycosylation for Families and Professionals: a booming story of sugar trees”**. Duncan Webster is the father of Maria Webster and President of this organization. For more information about the work of this organization which is focused on research to ALG9 -CDG (CDG -1L), visit the following link: <http://www.thefog.ca/main.html>

Youtube Channel World CDG Conference was created:

http://www.youtube.com/channel/UCvGr7WnYaylJgfmyL_xQgWw?guided_help_flow=3

Currently, oral presentations are being adapted and prepared. We will inform all community as soon as videos are posted on Youtube Channel World CDG Conference.

Important message about the access to PowerPoint presentations:

Please sign up until **12th October 2013** to receive PowerPoint presentations by writing an email to (specify name, country and if you are a family member or a professional): access.worldcdgconference2013@gmail.com

The book of photographs dedicated to the event it is available:

The official photographer of the event, Marisa de Andrés, completed a book of photographs dedicated to the event. Please find it at: <http://blur.by/1c8XyPD>

Please note that images in the printing format have high quality.

On behalf of all participants, thanks Marisa for your friendship and time dedicated to CDG families and professionals. It is very rewarding!

“The Second World Conference on Congenital Disorders of Glycosylation for Families and Professionals: a booming story of sugar trees” will be held in...

Many families and professionals have shown their interest in coordinating **“The Second World Conference on Congenital Disorders of Glycosylation for Families and Professionals: a booming story of sugar trees”**. The idea would be to organize this in connection with the SSIEM 2015 in Lyon (France). Currently, different stakeholders are exploring and setting up a robust operational strategy for the organization of this event. A working group will be announced in December 2013.

Finally, our thanks to the patient representatives, organizations and volunteers for supporting us in such interesting Satellite Symposia.

For more information:

ICIEM: <http://www.iciem2013.com/index.php/programme/official-satellite-symposia>

RareConnect : <https://www.rareconnect.org/en/community/cdg>

CDG parent associations:

Portugal: <http://sindrome-cdg.orgfree.com/>

Spain : <http://www.aescdg.com>

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

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

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

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

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

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

For more information on organizations and representatives:

<https://www.rareconnect.org/en/community/cdg>

Follow CDG organizations on Facebook:

- “CDG Family Network”: <https://www.facebook.com/groups/129138224432/?fref=ts>
- “CDG United”: <https://www.facebook.com/groups/glycosylation/>
- “CDG Spain”: <https://www.facebook.com/groups/47286836357/>
- “CDG Brasil”: <https://www.facebook.com/sindrome-cdg.brasil>
- “Les Ptits CDG-France”: <https://www.facebook.com/lesptitscdg?fref=ts>
- “CDG Denmark”: Our Louise Rimmen is figuring out why is not working at this moment!
- “CDG Portugal”: <https://www.facebook.com/pages/SINDROME-CDG/135220796488836>

Thanks to:



- Family Committee:** Merell Liddle (CDG Australia and mother), Rosália Félix (CDG Portugal and mother), Noelle Schmitz (CDG Netherlands and mother), Júlia González (CDG Spain and mother), Murielle da Silva (CDG France and mother), Andrea Berarducci (CDG USA and mother), LaRae Mercer (CDG USA and mother), Pierre Morandat (CDG France and father), Cristina Navas (CDG Spain and mother), Yoland Scott (CDG Spain and mother) & Team of highly motivated translators: Begoña Cano (CDG Spain and mother), Jorge Palomero (CDG Spain and father), Sandra Pereira Pinto (CDG Spain and mother), Ana Sánchez Cáceres (CDG Spain and mother), Luciana Zorzoli (CDG Argentina and mother), Louise Skov Rikken (CDG Denmark and mother), Adriana Tito Maciel (CDG Brasil and mother), Les P'tits CDG (France), Rachel Levillain (CDG France and mother) and Tim and Christine Jardine (CDG UK and parents)
- Scientific and Medical Committee:** Maria Antonia Vilaseca Busca (Spain), Eva Morava (USA), Hudson Freeze (USA), Erik A Eklund (Sweden), Thierry Hennet (Switzerland), Elisa Leão Teles (Portugal), Carla G. Asteggiano (Argentina), Sílvia Sequeira (Portugal), Rita Barone (Italy), Paula Videira (Portugal), Belén Pérez Dueñas (Spain), Mercedes Serrano (Spain), Rafael Artuch (Spain), Paz Briones (Spain) and Célia Pérez-Cerdá (Spain)

**“Overcoming challenges, Increasing knowledge, Doing excellence, Sharing experiences, in definitive:
BOOSTING PATIENT-FOCUSED RESEARCH”**



“2nd World Conference on Congenital Disorders of Glycosylation for Families and Professionals”



ISNS
International Society for Neonatal Screening

HOME NEWS **AGENDA** MEMBERSHIP

Agenda

1 September, 2015 - 4 September, 2015
SSIEM, Lyon, France

Details to follow.

See www.ssiem.org

See you all in 2015 (France)!

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