1st World Conference on Congenital Disorders of Glycosylation for Families and Professionals: "A booming story of sugar trees"

31st August 2013
Informal families Meeting

1st and 2nd September 2013
Satellite Symposia

Centro Cívico la SEDETA
BARCELONA - SPAIN

WELCOME MESSAGE

This conference is dedicated to all CDG families who in their daily life share the passion that only a loving and concerned parent or family member can have to face challenges resulting from a disease with high unmet medical needs.
Family love, tenacity and bravery make us believe in the power of dreams. THANKS!

WELCOME MESSAGE

We are happy to announce that the "**1st World Conference on Congenital Disorders of Glycosylation for Families and Professionals: a booming story of sugar trees"** is well underway!

We have an amazing lineup of speakers and events. You can check them out at:
http://www.iciem2013.com/index.php/programme/official-satellite-symposia

Dear families, professionals, colleagues and friends:

It is with great pleasure that we invite you to the Satellite Symposium named "**1st World Conference on Congenital Disorders of Glycosylation for Families and Professionals: a booming story of sugar trees"** to be held in the beautiful city of Barcelona on the 1st – 2nd of September 2013.

This conference will take place in a unique year: The Spanish Minister of Health, Social Services and Equality, Ana Mato, declared 2013 the Spanish Year for Rare Diseases.

This Satellite Symposium will provide a program which will highlight important information in the clinical and research area, as we progress to our goal of better care and improvement of the quality of life of people living with CDG and related metabolic rare diseases.

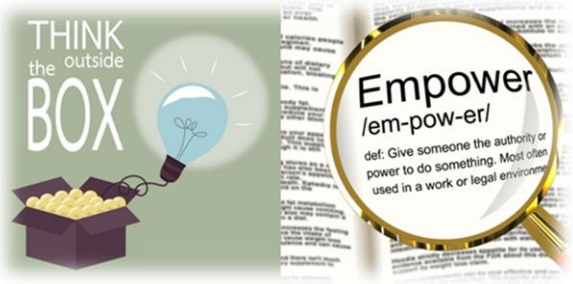
The Scientific-Medical and Family organizing committee is sure that this unique event will be unforgettable and look forward to welcoming you to this First World CDG conference.

We would like to express our thanks to all experts, professionals and families, for their outstanding contributions and in particular to the members of the committees for their contribution and helpful support. Likewise we would also like to express our appreciation to the speakers, as well as to the invited chairs for their careful preparation of the invited sessions.

CDG Parents and family members together with professionals can make the difference! Laying the groundwork for future stages of research and development based on family's needs is the most rewarding aspect of this Satellite Symposia–boosting translational research will be a dream come true!

We are looking forward to seeing you in Barcelona!
On behalf of the organizing committee,

Vanessa Ferreira (sibling to CDG patient) and Rosália Félix (mother to CDG patient).



1. ORGANISATION

The conference is being organized jointly by the Family Committee and the Scientific and Medical Committee. The **Family Committee** is involved in disseminating information worldwide, and in helping families in the organization of events aimed at securing funds that will guarantee families attendance to this unique event.

The Scientific and Medical Committee is made up of 14 recognized CDG experts. The Family Committee, coordinated by the Portuguese CDG Association and Other Rare Metabolic Diseases (APCDG-DMR) has announced country organizing Delegates and task forces with families representatives from around the world.

The list of the organizers that are involved in the coordination of this event is available at http://www.iciem2013.com/index.php/programme/official-satellite-symposia

2. INTRODUCTION

In a unique partnership, researchers and practitioners will meet together with CDG patients and families from all around the world at the Satellite Symposium named "**The 1st World Conference on Congenital Disorders of Glycosylation for Families and Professionals: a booming story of sugar trees**", to be held 1st and 2nd September 2013 in Barcelona (Spain) and is part of the ICIEM program.

This event is coordinated by the Portuguese CDG Association and Other Rare Metabolic Diseases (APCDG-DMR).

An exciting **Draft Agenda** includes experts with the most up-to-date medical and scientific knowledge in this emerging field together as partners with family and patients who will meet in a combination of presentations and round table discussions. For the first time, several CDG parents and patients will give talks. Families and health professionals are equal partners in this unique conference designed to educate and empower CDG families. It aims to create valuable partnerships within worldwide CDG experts and families. A complete list of Family, Scientific-Medical Committee members and Draft Agenda are available at the ICIEM website (http://www.iciem2013.com/index.php/programme/official-satellite-symposia).

All families and patients affected by CDG together with professionals on metabolic disease are encouraged to attend this conference. Learning more about researchers and physicians achievements and knowledge will benefit us all, and boosting a frank discussion will help to clarify the key challenges and will potentiate translational research.

3. GENERAL INFORMATION

Registration

The registration forms are available at:
http://www.iciem2013.com/index.php/programme/official-satellite-symposia

Terms and conditions (It is of great importance to read all the content):

A. Registration Process and Payment Methods

1. The **registration fee is a donation that will support the overall conference**, the organization, logistics for the event, as well as the social program targeted for children and adults and so forth. In addition, the Registration fee includes Conference participation, information material and networking sessions.
2. **Conference registrations will not be confirmed until the completed form and the correct payment are received and processed by the Registration Office** (cdgawareness@gmail.com). We reserve the right to refuse admission if payment is not received on time.
3. Registrants should obtain confirmation from the Registration Office (please be sure your spam filters will allow mail from Registration Office (cdgawareness@gmail.com) and the conference coordinator.
4. The deadline for registration is 1 July 2013.
5. **Accommodation**, travel and meal costs are not included in the conference registration fees. Attendees are responsible for making their own lodging arrangements. The meals are paid the 1th September 2013 at the registration desk (9.00 am to 10.00 am).
6. Capacity is limited to 300 attendees. Registrations will be handled on a first-come, first-served basis.

B. Cancellation Policy

Conference registration can be cancelled only in writing by mail seven (7) weeks before the Conference is to be held. Thereafter, the full Conference fee is non-refundable.

C. Personal insurance

APCDG-DMR cannot be responsible for healthcare, dental and ambulance services during the Satellite Symposium "1st World Conference on Congenital Disorders of Glycosylation for Families and Professionals: a booming story of sugar trees". **The APCDG-DMR strongly recommends that participants take out comprehensive medical and travel insurance, which should cover the possibility of flight cancellation due to strikes and other causes**. Therefore, APCDG-DMR and its local co-organizers do not accept responsibility of any nature at this level.

D. Privacy

In registering for the conference, relevant details will be incorporated into a participant list. All this information will be treated in a confidential way. The Portuguese Association for CDG and related Rare Metabolic Diseases (APCDG-DMR) may use these details to inform current participants of Conference updates or future conferences via email. **Participants are responsible for advising the Portuguese Association for CDG and related Rare Metabolic Diseases if they do not wish to have their email addresses included in the conference participant list or APCDG-DMR distribution list for future events.**

E. Copyright

All intellectual property rights in all materials produced or distributed by the Coordination in connection with this Conference is expressly reserved and any unauthorized duplication, publication or distribution is prohibited.

F. Conference Program

Conference program is subjected to change and is available at:
http://www.iciem2013.com/index.php/programme/official-satellite-symposia

Registration desk

Participants can pick up their personal Conference material at the registration desk which will be open at the venue (Centro Cívico La Sedeta). The Conference secretariat will be available to assist you during the Conference at the Centro Cívico La Sedeta.

Registration desk hour

Sunday 1th September 9.00 am to 10.00 am

Conference language

The official Satellite Symposium language is English. Currently, we putting a lot of efforts in order to guarantee simultaneous interpretation into Spanish and French are being considered.

Lunch, Breaks and Dinners

Meals will be at the restaurant of the Centro Cívico La Sedeta.

Name Tags

Please wear your name tag at all times during the conference, including the breaks and CDG Gala dinner. You may be asked to present your nametag.

Note: Our Children and adults will have a specific identification: parents name and phone contact will be included to facilitate possible actions of the volunteering service.

Note to speakers

If you are schedule to present, **please ensure your PowerPoint is loaded well in advance of your presentation time**. A **central computer and technician** are available and well identified at the **Registration desk** where you may upload your presentation, which in turn will be uploaded on the computer in the appropriate room. Please visit the registration desk if you have any questions or for further details.

Adhering to this will help to ensure your presentation is available when needed, and it should also prevent confusion when loading the presentation files on to the main computer. The Day and Time for your presentation can be found in the final program, which is available, via:

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

Should you have any questions related to your presentation, please do not hesitate to contact the conference general inquiries via cdgawareness@gmail.com

Mobile Phones, Pagers and Laptop sound

As a courtesy to presenters and colleagues, please ensure that all mobile phones, pagers and sound of your laptop **are switched off during the satellite symposium sessions**.

Currency and credit cards

Spain's unit of currency is euro. Foreign currency can easily be exchanged at banks and money changer booths located at the international airport and throughout Barcelona. Major credit cards are accepted throughout Barcelona.

Dress code

1 st September 2013: **Chic & Happy!**

4. IMPORTANT DATES

Important dates:

Official Announcement	10 th November 2012
Official invitations	15th November 2012
Details concerning the conference	12th January 2012
Launch Registration	28 th February 2013
Conference surprise deadline	30 th March 2013
Biographies and titles of presentations deadline	20th April 2013
Booklet with full information official presentation	15th June 2013
Launch Promotional video	15th June 2013
Launch CDG Awards nominations	25 th of July 2013
Registration deadline (mandatory to all participants)	**1 th of July 2013**
Instructions for formal oral presentations	15th of July 2013
CDG Awards nominations deadline	25th of August 2013
Skype meeting to support inspiring speakers presentations deadline	1th-5th August
1st WORLD CDG INFORMAL FAMILY MEETING	**31th (Saturday) August 2013**
THE SATELLITE SYMPOSIUM: The 1st World Conference on Congenital Disorders of Glycosylation for Families and Professionals: a booming story of sugar trees	**1st (Sunday) and 2nd (Monday) September 2013**

5. IMPORTANT MEETING DAYS

5.1. 1st WORLD CDG INFORMAL FAMILY MEETING

- **31st (Saturday afternoon) August 2013**

Targeted only to CDG families. More details will be available once all registrations have been received. For further details, please write to: cdgawareness@gmail.com

5.2. THE SATELLITE SYMPOSIUM

"The 1st World Conference on Congenital Disorders of Glycosylation for Families and Professionals: a booming story of sugar trees"

- **1st (Sunday) and 2nd (Monday) September 2013**

Reasons to attend

- Engage in meaningful discussions with the ones that experience CDG in their daily lives
- Knowledge sharing
- Empowerment
- Gain insights into new breakthroughs in the CDG field
- To forge partnerships with main stakeholders who addressed the same concerns
- Being surrounded by inspiring and courageous people
- Shaping basic and applied research based on CDG community concerns and needs
- Networking Promotion
- Identify and assess current CDG challenges
- To attend presentations from several relevant International CDG professionals
- To raise awareness of CDG in an International scope
- To encourage global CDG community union
- To promote translational research

Who will be there?

Families, key researchers, physicians, educationalists and other practitioners.

Type of sessions

- Plenary session
- Round tables (short communications)
- Lectures

Themes

1. *The worldwide CDG state-of-the-art*
2. *Congenital Disorders of Glycosylation Thoughts on Clinical* Care
3. *Biochemical and genetic diagnosis of CDG*
4. *Therapeutic approaches*
5. *Animal models*

The Satellite Symposium program at a glance

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

6. PREPARING FOR THE MEETING

Preparation is key to getting the most out of the meeting. Listed here are multiple tips that will guarantee your successful attendance:

1. **Register for the meeting**: **Registration** is **MANDATORY to everybody**. **DEADLINE 1st July 2013**. Be sure to register very early in advance for the meeting (a separate registration form targeted, in one hand to families and on the other hand to professionals are available at http://www.iciem2013.com/index.php/programme/official-satellite-symposia)

2. **Make travel arrangements early**: Flights and hotel rooms will be in demand during the dates of the Satellite Symposium.

3. **Plan your schedule.**

4. **Download and print the program:** we do not have available programs

5. **Prepare to network:** read carefully the program and select the relevant key stakeholders to whom you wish to interact. You will have the opportunity to meet well-known individuals that will be accessible during all the conference. Breaks and meals will also be suitable times to meet and network with people that you do not normally get to see.

6. If it is the case, **Practice your formal oral presentation** (instructions and templates will be sent to all inspiring speakers)

7. **Taking notes:** it helps you to focus your listening; it will be a record for later reference and helps to promote retention.

8. **Asking questions**: we encourage you to ask questions at the sessions you attend. Asking good question is an important skill that can be developed. To become a more active meeting participant, we suggest you to try **to have at least three good questions**, and write these questions in your notebook-whether or not you intend to ask them. **Please, work up to asking your question(s) preferentially in public or after the session.**


7. INSTRUCTIONS FOR FORMAL ORAL PRESENTATIONS AT THE SATELLITE SYMPOSIUM

All speakers must comply with their time for presentation.

7.1. FOR RESEARCHERS AND MEDICAL DOCTORS

Please inform the organizers immediately if, for some reason, you are NOT able to give the presentation at the "1st World Conference on Congenital Disorders of Glycosylation for Families and Professionals: a booming story of sugar trees" emailing at cgdawareness@gmail.com

The main audience will be composed by Families and Professionals. We kindly request you to prepare presentations in a non specialized, family-friendly language.

The purpose of your presentation can be found in the letter of invitation previously sent to you. All speakers are asked to arrive 10 minutes early to meet the chairpersons and the conference assistant.

7.2. CHAIRS

Thank you for accepting to be a chairperson at one of the sessions at the upcoming "**1st World Conference on Congenital Disorders of Glycosylation for Families and Professionals: a booming story of sugar trees",** 1th and 2th September 2013, at the Centro Cívico La Sedeta, Barcelona, Spain.

If you are unable to act as chairperson for a session at the session agreed, please contact the Conference Secretariat as soon as possible at cdgawareness@gmail.com

Key roles of chairpersons are to:

- Ensure **best use of the limited time for presentations and discussions**
- During discussion share your knowledge with speakers and audience
- Encourage as much audience participation within the limited time available. If discussion does not occur spontaneously, you may want to ask the audience a few questions.
- Refocus discussion when needed

Please can you:

- Arrive 10-15 minutes early to meet your conference assistant and the speakers
- One of the chairs per session will perform a short opening statement about the session
- A short presentation about speaker will be done (using the biography available in this booklet)
- Close the meeting when all questions have been addressed during the available time for that purpose

7.3. FOR INSPIRING SPEAKERS

We are grateful that you accepted to be one of our inspiring speakers. The presentations must follow a template that will be available via cdgawareness@gmail.com

We will also distribute material focused on communication and dissemination skills. Skype Meetings to support you during the preparation will be scheduled in advance via cdgawareness@gmail.com

8. Program Targeted to our children and adults

8.1. Volunteer Service provided by Orange

About Solidarity Orange: The "Solidarity Orange" emerged in 2009 in response to the concern of many employees who wanted to participate in charitable activities and did not know how. Through this initiative, employees, their families and friends can help, share and collaborate with the most needed groups in a very different environment to the office and reinforce their sense of community.

The range of activities is varied: working with children and the elderly, activities with disabled people, help in soup kitchens, collaboration in natural disaster (organizing fundraising activities) or reforestation. In this way, each employee can choose the initiative that makes him/her feel comfortable.

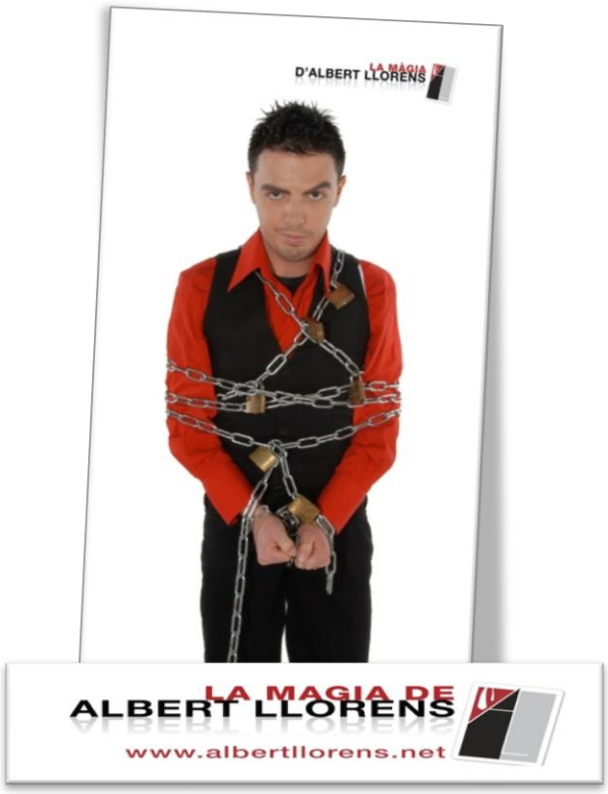
The volunteer service will watch and entertain children's and adults meanwhile parents and family members are attending the conference.


8.2. Activities

Merche Ochoa and Magician Albert Llorens

1st September 2013, Sunday
10.40-11.50: Merche Ochoa: Clown Show "Por dentro y por fuera"
15.00-16.00: Magician Albert Llorens

2nd September 2013, Monday
10.00-11.30: Merche Ochoa: Clown Show "Por dentro y por fuera"



http://www.guiametabolica.org/noticia-articulo/exito-de-dins-i-fora-un-programa-para-explicar-cuentos-sobre-ecm-en-las-bibliotecas
http://www.merche8a.com/
http://vimeo.com/49751446

www.albertllorens.net

Les P'tits CDG

We want to express our sincere gratitude to the CDG French Association" Les P'tits CDG" for supporting the Clown Show "Por dentro y por fuera".
THANKS! http://www.lesptitscdg.org/

9. SOCIAL EVENTS

9.1. "First World CDG Gala dinner"

We are delighted to announce the **"First World CDG Gala dinner"** shaping up to be a truly spectacular evening of entertainment, Spanish food and awards.
This event will take place at **19.30** on **1st September 2013** in Barcelona, Spain at the Centro Cívico La Sedeta.

At **20.00** a **Batucada show** will be performed by la BandSambant at the Centro Cívico La Sedeta.

Website:

http://www.labandsambant.com/

The organizers would like to express their deep gratitude to all members from "la BandSambant" for joining us.

9.2. "First World CDG Gala Awards: Hope and Dreams"

"CDG Gala Awards: **Hope and Dreams**" will be the first-ever CDG awards honoring those who have made significant contributions to CDG worldwide. The awards will be presented at our CDG Gala Dinner on the evening of 1st September at the Centro Cívico la Sedeta.

The purpose is to recognize exceptional achievements and contributions in the field of CDG, improving CDG knowledge, basic and applied research and consequently improving CDG families' quality of life.

Instructions: Nominees will be announced and all CDG Community (family members, volunteers, scientists and medical doctors amongst others) vote by sending an email to: cdgawareness@gmail.com

The CDG Award Board makes the final decision based in the following criteria:

- Publications;
- citations of the work performed;
- Since how long are the nominee is active in the CDG field.

The Portuguese Association for CDG and related rare metabolic diseases in collaboration with all CDG community will be presenting 5 awards for excellence and leading work in the field of CDG, at the CDG Gala dinner that will be held on 1th September at the Centro Cívico la Sedeta.

The following categories are purposed:

1. ***CDG medical care award*:** recognizing impressive breakthrough work done by a medical doctor in the field of CDG. It aims at recognizing an individual who goes above and beyond in clinical care and treatment for a CDG patient, family, or the community at large.
2. ***CDG research award*:** recognizing an individual for **overall scientific excellence, promotion of CDG research International collaboration,** and **support of the patient community**
3. ***CDG Lifetime Award*:** lifelong dedication and commitment to addressing CDG (this award is already assigned)
4. ***CDG inspirational award*:** celebrate the achievements of a remarkable person who inspire those around them.

Joana Demestre, Sculptor for the "CDG Gala Awards: Hope and Dreams"

In 2011, Joana Demestre was responsible for a special gift dedicated to Professor Jaeken: "The wise Grasshopper, which is the main character of the fairy-tale, named "Glycoland and the colored antennas". She is pleased to participate again in an activity targeted to CDG community. She accepted the challenge of inventing the **"CDG Gala Awards**: **Hope and Dreams**" that will represent: **Tenacity, Love, Motivation and Fight.**

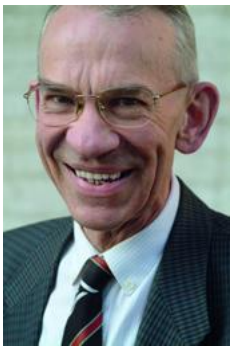
"The wise Grasshopper"

For further information:
http://www.joanademestre.com/ca/index.htm

The sculptures are sponsored by Agios Pharmaceuticals

10. SPEAKER BIOGRAPHIES (it follows the order from the agenda)

10.1. Plenary session



Professor Jaak Jaeken

Jaak Jaeken is an Emeritus Professor at the Faculty of Medicine, 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. He was involved in the description of many other diseases. He is member of the the Society for the Study of Inborn Errors of Metabolism (SSIEM) (since 1978) and the International Child Neurology Association (since 1989). He has about 517, including 304 peer reviewed international publications, 2 books and 32 book chapters; guest editor of 2 special volumes on CDG.

He received the following Awards and Honors:
- Cross of Knight in the Crown-Order, 1984
- European Science Award of the Körber foundation, 2004 (shared by six winners)
- Honorary member of the SSIEM, 2007

Today Dr. Jaeken continues to collect and examine new cases of the rapidly expanding group named CDG and test new methods of diagnosis and therapy on patients when they are available.

10.2. Theme 1: The worldwide CDG state-of-the-art

10.3. Round table I: CDG Latin America and Iberian countries

Dr Carla Asteggiano

Carla G. Asteggiano received her PhD degree in Science and a postdoctoral position in human genetic diseases from the National University of Cordoba. She is currently teaching in the School of Medicine at the Catholic University of Cordoba. She has been a strong advocate for genetic metabolic diseases from the beginning of her professional career. She has been working on the field of Congenital Disorder of Glycosylation (CDG) as a Scientific Researcher at National Council for Scientific and Technological Research (CONICET) since 2004. She involves her fellow doctorates in different lines of investigation to acquire thorough knowledge of biochemical and molecular mechanisms present in CDG diseases. For the last few years, they have focused on skeletal dysplasia due to CDG (Mainly EXT1/EXT2- CDG).

Her current projects include:
- Biochemical and molecular bases of Congenital Disorders of glycosylation.
- New approach in the expression and glycosylation status of NCX1 and NCKX1 exchangers in human platelets and megakaryocytes cell lines: a putative role of these exchangers in the thrombus-hemorrhagic events associated with Congenital Disorders of Glycosylation.
- A broad spectrum of genomic changes in EXT1/EXT2-CDG patients with a severe phenotype of multiple osteocondromatosis.

- Glycobiology in skeletal dysplasia associated with Congenital Disorders of O-glycosylation: GALNT3-CDG (familial tumoral calcinosis); LFNG-CDG (spondylocostal dysostosis); SLC35D1-CDG (Schneckenbecken dysplasia); B3GALTL-CDG (Peter-Plus syndrome) and B4GALT7-CDG (Progeroid type of Ehlers-Danlos syndrome).

In addition, she has served as a Scientific Researcher at the National Council for Scientific and Technological Research (CONICET) since 2004 and has been the head of the CDG Research Group in the Center for the study of congenital metabolopaties (CEMECO) and Assistant Professor, Chair of Biological Chemistry, Faculty of Medicine, Catholic University of Córdoba, since 2008. She has been a member at the Ethics in Human Research Committee (CIEIS) under the Ministry of Health in Argentina, since May 2007. She is a member of different scientific associations related to human genetic field, as well.

Her future purpose is to continue collaborating with referential CDG international colleagues, sharing projects that will allow the training and research of different aspects related with her CDG studies, as well as to increase the diagnosis of human glycosylation disorders in a *CDG Latin American Net* establishing common goals with Latin-American scientists and clinicians.

Dr Charles M. Lourenço

Neurogenetics Unit, Medical Genetics Division, University of Sao Paulo, Ribeirão Preto, Brazil.

Charles M. Lourenço is a clinical biochemical geneticist and a consultant physician at the Neurogenetics Clinic of the Hospital of Ribeirão Preto, Brazil, where he is also a member of the Lysosomal Unit in charge of the infusion centre treating patients with lysosomal storage disorders. His PhD thesis in neurogenetics focused on spinocerebellar ataxia of early onset, especially on a subset of patients with ataxia and hypogonadism. Dr Lourenço's interests include the clinical and molecular aspects of leukodystrophies, hereditary spastic parapareses, hereditary spinocerebellar ataxias, lysosomal disorders of the brain (neurolipidoses) and inborn errors of metabolism with adult presentation.

Dr Elisa Leão-Teles / Dr Esmeralda Rodrigues

(Not available in the moment of edition. Soon will be online)

Dr Belén Pérez-Dueñas

Belén Pérez-Dueñas, Doctor in Medicine from the University of Barcelona, expert paediatric neurologist in rare diseases and inborn errors of metabolism. Consultant in the Pediatric Neurology Department of the Hospital Sant Joan de Deu, Barcelona and Researcher from the Centre of Biomedical Investigation in Rare Diseases (CIBERER), ISCIII, Barcelona, Spain.

Dr Nathalie Seta

Nathalie Seta received her PhD in Biochemistry from university Paris Sud and is currently teaching Public Health at University Paris Descartes. She involves her students and collaborators in studying congenital disorders of glycosylation (CDG and alpha-dystroglycanopathies). Her current projects include phenotype-genotype relation in PMM2-CDG. She was recently honoured with the Price Maurice-Louis GIRARD (Académie Nationale de Pharmacie) for her contributions to rare diseases.

10.4. Round table II: CDG worldwide

Dr Tawfeg Ben-Omran

Tawfeg Ben-Omran received his speciality training in clinical and metabolic genetics at the Hospital for Sick Children, University of Toronto, Toronto-Canada. He obtained degree of FRCPS and FCCMG in medical genetics in 2006. He completed MBCHB and M.D. in Pediatrics from Al-Arab Medical University, Faculty of Medicine, Benghazi, Libya. Currently, he in is an Asst. Prof at Weill Cornell Medical College in Qatar and New York-USA.

Tawfeg Ben-Omran actively and consistently contributes to the body of published knowledge in the field of clinical and metabolic genetics, with over 30 published articles in peer reviewed journals and chapters in text books. In addition to publishing in peer reviewed journals, his expertise is acknowledged as an invited reviewer for many of the journals in the field of clinical genetics.

Dr. Ben-Omran is also recognized as an expert in disorders of the Arab population. Through observation of Dr. Ben-Omran's earned awards and invited guest lecture status, his national and international presence is clear. Just recently in February 2013, Dr. Ben-Omran received the "Princess Aljawhara Center Award for The Best Research in Basic Genetics". This is one of the most competitive and prestigious awards, where researchers from outside and within Saudi Arabia included in this competition and present their research work. The title of his research project was: "The use of whole exome sequencing to unravel disease genes causing autosomal recessive disorders in Qatari population.

In 2011, he received Research Award from Medical Research Center-Hamad Medical Corporation (Subject: Homocystinuria, a common but treatable inborn error of metabolism in Qatar, Treatment and Pathophysiology. In addition he was awarded Stars of Excellence Award 2011 for Pioneering Newborn Screening and Stars of Excellence Award 2011 for specialized care of Genetic Diseases in the Middle East.

Within Qatar, Dr. Ben-Omran is active as the Director of Qatar Medical Genetic Center and Member of the Child Health Research Committee at Hamad Medical Corporation, Doha-Qatar.

He has memberships in many societies including: American Society of Human Genetics, European Society of Human Genetics, Society for the Study of Inborn Errors of Metabolism, Middle East Metabolic Genetic Group, Congenital Hypothyroidism Training Working Group, the Middle East and North Africa (MENA) Newborn Screening Initiative, Middle Eastern Lysosomal Storage Diseases Expert Council Advisory Board, Child Health Research Advisory Committee, International Society for Prenatal Diagnosis, Chairman of Middle East Metabolic Dieticians Group and Founder Member and Regional Representative of SSIEM Adult Metabolic Physicians Group.

Dr Rita Barone

Researcher of Child Neurology and Psychiatry, School of Medicine, University of Catania, Italy.
Department of Medical Sciences and Pediatrics Neuropsychiatry Unit Policlinic Medical Faculty of the University of Catania.

Rita Barone was the first to offer laboratory diagnosis for CDG in Italy since 1996 and contributed to a better knowledge of these disorders by studying clinical, biochemical and genetic features of patients with CDG.
Her major clinical interests include: (1) diagnosis and characterization of patients with neurogenetic diseases, including novel diseases of the central nervous system (2) Clinical trials with orphan drugs for rare diseases affecting the central nervous system (3) Development of novel methods in omic sciences: glycomics of neurological diseases and (4) Assessment of communication and rehabilitation tools' development in Rett syndrome and Autism Spectrum Disorders.

She is a member of the national scientific committee of the Italian Association for Mucopolysaccharidosis. Dr. Barone is author of several scientific articles on diagnosis and characterization of Congenital Disorders of Glycosylation.

Dr Agata Fiumara

Researcher of Child Neurology and Psychiatry, School of Medicine, University of Catania, Italy.
Department of Medical Sciences and Pediatrics Neuropsychiatry Unit Policlinic Medical Faculty of the University of Catania.

Agata Fiumara currently is Associate Professor of Paediatrics(MED38) in the University of Catania. She is is head of the Specialization School of Child Neurology and Neuropsychiatry since 2011.

She is coordinator and member of several research projects approved by MURST (i.e. autism and other behaviour anomalies in metabolic diseases, cerebellar anomalies in metabolic disorders), European Community (European group for investigation of glycosylation defects, EUROGLYCAN 1999-2003 and EUROGLYCANET 2004-2008), Health Ministry (multicentric national project on epidemiology, natural history, therapeutic opportunities for lysosomal disorders) and therapeutic trials for lysosomal diseases (Enzyme Replacement therapy for MPS VI, BioMarine). She is author and co-author of more than 350

reports, 70 of which on international journals, Speaker at national (155) and international (75). Congresses.

Dr Malina Kirilova Stancheva-Ivanova

Malina Kirilova Stancheva-Ivanova MD, PhD, specialist paediatrician, post-graduate student of Paediatric Neurology in Multiprofile Hospital for active treatment in Neurology and Psychiatry "St.Naum", Clinic for neurological diseases in children, Medical University – Sofia. She attended the "Protein Glycosylation in health and disease course" in Paris, France, 2007. Short specializations in Assistance hopitaux publique de Paris, Necker Enfants Malades, Paris, France in 2008 and in the Center for Metabolic diseases, Katolieke Universiteit, Leuven, 2009 in congenital disorders of glycosylation and classical galactosemia.

She participated in Euroglycanet from 2006-2009. She was principal investigator in Bulgarian Project "Screening for CDG with IEF of serum transferrin" from 2009-2010. In 2013 she defended thesis "Screening for congenital disorders of glycosylation in children with mental retardation". Her professional interests focus on metabolic and neuromuscular diseases. Finally, she is a member of Bulgarian Scientific Society of Pediatric Neurology, Psychiatry and Developmental Psychology.

Dr. Honzík Tomáš

Tomas Honzik received his MD in the field of General Medicine, with consultancy exams from Pediatrics and Clinical Genetics, and his Ph.D. in the field of biochemistry and pathobiochemistry from First Faculty of Medicine, Charles University in Prague. He currently teaches Pediatrics and Metabolic Medicine at that University. His professional interests focus primarily on disorders of glycosylation and mitochondrial diseases. He published 48 research articles in international and domestic journals. He is a member of the Czech Medical Association of J.E.Purkyně – Czech association for Pediatrics, Czech Medical Chamber and Society for Study of Inborn Errors of Metabolism.

He was honored by the Ministry for Health of the Czech Republic for excellent contribution to research project. He currently participates in two research projects dedicated to CDG: 1) study of the basis of Congenital disorders of glycosylation supported by International Grant Agency (IGA) and 2) study of the biochemical and molecular basis of glycoprotein biosynthesis disorders supported by Grant Agency of Charles University (GAUK). Other collaborators involved in the research are MSc. Hana Hansikova, Ph.D., MSc. Nina Ondruskova, MSc. Marketa Tesarova, Ph.D., MSc. Katka Vesela, Ph.D. and prof. Jiri Zeman, MD., Ph.D.

10.5. Highlighting sessions

10.6. Round table III: Inspiring speakers 1

Morgan Liddle

Morgan Liddle was diagnosed with CDG 1A eight years ago. Now 21-years-old, she is an Australian Champion Para-Equestrian and competes at National and International Level in Dressage. When she is not riding or involved in fitness training, Morgan loves music and dance. Her determination not to let her disabilities get in the way, has led her to write and speak extensively on communication, dance and riding. Morgan has a bright, bubbly personality and loves to encourage others to live their dreams.

Bas Holten

Bas and Esther Holten are the parents of an almost 6-year-old son with Congenital Disorders of Glycosylation (CDG). He was diagnosed at 2 months (November 2007). Both parents are supporters and advocates for rare diseases-specifically CDG. Both are active members of the VKS, the Dutch Metabolic Disease Association for children and adults.

In addition, Bas gives presentations on the life of Tijmen, living his life to the maximum with CDG. He aims to create more awareness of the daily life of parents who with all their love and power are taking care of their child.

Trying to get people to see through 'those eyes' and in addition utilizing of all the experience and knowledge parents have, is something they strive for. With his family's daily love and energy, Tijmen is now at this moment in time enjoying life to the max!!

Dr Duncan Webster

Duncan Webster is the father of a six year old daughter with Congenital Disorder of Glycosylation (CDG). His daughter, Maria, was diagnosed at the age of 6 months. Duncan is a supporter and advocate for rare diseases-specifically CDG. He is a founder and trustee of Foundation Glycosylation (the FoG), established in order to support research for the development of therapies targeting CDG, to help raise awareness of the disorder and to advocate for individuals living with this enzyme deficiency.

Duncan Webster received his MD at Dalhousie University in Halifax, Nova Scotia, Canada. He received specialty training in Internal Medicine and Infectious Diseases at the University of Alberta before returning to Dalhousie University where he received further fellowship training in Medical Microbiology. He is currently providing clinical service and is active in education and research at the Saint John Regional Hospital located in the Atlantic Canadian city of Saint John, New Brunswick.

Through collaboration with researchers at Dalhousie University and the University of New Brunswick, Duncan is actively involved in the development of disease models for ALG9-CDG and ALG12-CDG. The aim of this research is to gain further understanding of the cellular processes in these rare enzyme deficiencies and ultimately to develop therapeutic strategies.

Sandra Pereira Pinto

Sandra Pinto is the mother of a 4-year-old son with Congenital Disorders of Glycosylation CDG). He was diagnosed at 6 months. Sandra is a supporter and advocate for rare diseases-specifically CDG. She is responsible for the Accessibility Department of the APCDG-DMR (Portuguese CDG Association and other Rare Metabolic Disease), an active member of the AESCDG (Spanish Association of CDG), a Spanish moderator at Eurordis-Rareconnect as well as the representative for FEDER (Spanish Rare Disease Federation) at the Universal Accessibility group of CERMI (Spanish Committee for People with Disabilities).

In addition, Sandra Pereira Pinto is an architect dedicated to Universal Accessibility and Design for All Area in partnership with Jorge Palomero at their office *eCapaz* and currently holds a scholarship for a Masters in Domotic and Digital Home at the UPM.

Noelle Schmitz

Noëlle Schmitz was born in Amsterdam, Holland. In her younger years Noelle worked as a nurse. She later became a manager in the field of music, and more recently a manager in International trade involving several countries including Sweden, France, Germany, Canada, England and the Netherlands where she also has worked as a manager for a well known temporary job agency. Noelle has studied in marketing and Commerce as well.

Noelle is also a mother of three children. Her oldest daughter, 20-year-old Zanne, is aspiring to become a fashion photographer. Noelle's second child, her son, Liam is now 8-years-old. Her youngest child, her daughter, Eline is four.
Noelle's journey with CDG began with Eline. After she was born, Eline had difficulty with feeding. She couldn't eat and developed hydrocephalus and at two months of age Eline underwent surgery as a result. Eline's feeding problems continued and within a matter of weeks, Eline was back in hospital as she was continuously losing weight. Noelle comforted her baby girl as she struggled for her life in intensive care on numerous occasions during her first year of life.

Eline was diagnosed with Cdg when she was 9-months-old. Although Noelle now had an answer for her daughter's issues, Eline's CDG subtype remained a mystery until she a year and a half old when she was given the subtype 1C. By this time in Eline's young life, she had practically resided in the hospital due to her complex medical problems.

Noelle decided that raising three children, the youngest with a diagnosis of CDG that would prove to be the beginning of a life-long journey that would include constant doctor’s visits, therapies and more, would not leave her enough time to dedicate to her career. Noelle now works full time at home taking care of Eline and her other children.

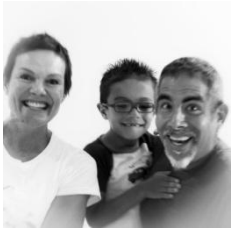
Eline enjoys writing, painting and is a strong advocate for CDG. Eline is an active member and admin of the well-known online CDG support group for parents and CDG experts called CDG United.

10.7. Round table IV: Inspiring speakers 2

Eric Jerman

Eric Jerman is a father of a five-year old boy, named Jake, who has been diagnosed with CDG 1A. Jake is legally blind, has Cortical Vision Impairment (CVI), is nonverbal, non-ambulatory, and has multiple disabilities. Eric received his Masters Degree in special education from the University of Massachusetts and he works as an Orientation and Mobility instructor at the Perkins School for the Blind, in Watertown, Massachusetts, USA. In the past three years, Eric has given two-dozen presentations on use of iPads with children who have vision impairments as well as additional multiple disabilities.

Eric has presented to the special education department of a city school district; to early intervention staff; to chaplains at Children's Hospital, Boston; at a regional conference of educators of children with low vision (AER); at a national conference of parents of children with vision impairments (NAPVI); at the Helen Keller National Center in New York City; at a province-wide conference of educators of deaf-blind in Toronto, Canada; and at a conference for families with children diagnosed with the rare metabolic disorder CDG.



Claudia and Nick Vasquez

Claudia and Nick Vasquez are the parents of 6 year old Oliver Vasquez. Born July 7, 2007 and diagnosed with CDG-1a in March of 2009.

A rare disease brings rare challenges and an uncertain future. The role of special needs parents is a heavy burden that can seem daunting. But the love of one's child brings strength.

Living in Benicia California, they will give a glimpse of their experience with Healthcare and other services available to them, and how they manage and negotiate the logistics of raising Oliver.
Claudia and Nick have brought CDG awareness to their community through their and Oliver's participation in local events. Hoping to one day create their own annual event for research and awareness.

Dr Vanessa Ferreira

Vanessa Ferreira received a bachelor's degree in Biological Sciences from Badajoz University (Spain). She holds a PhD (Sc.D.) in Cell and Developmental Biology from the Center for Genomic Regulation, University of Pompeu Fabra, Biomedical Research Park in Barcelona (PRBB). Currently, she is Patient Affairs Director in a Company focused in developing therapies in the field of rare diseases.

She founded the Portuguese CDG Association and other Rare Metabolic Disorders (APCDG-DMR) in 2010. Vanessa approaches Congenital Disorders of Glycosylation (CDG) from her unique perspective of being Cell Biologist, as well as the sister of a patient with CDG. Vanessa's personal journey with her sister, which began more than 30 years ago, was the impetus for APCDG-DMR focus on (1) boosting scientific breakthroughs, (2) building awareness among the community on the value and impact the association's participation can have on research (3) and fostering translational research in full collaboration with worldwide CDG professionals.

The APCDG-DMR coordinates and implements education and communication activities, and supports strategies to increase awareness and understanding targeted to CDG and related diseases. In addition, builds and enhances ongoing relationships with other patient community organizations and leaders in the rare disease space. The aim of these activities is to establish a relationship between scientists and society, to contribute to scientific debate and to increase media coverage of different aspects of rare diseases. Since 2010, Vanessa has participated as speaker by direct invitation/selection in more than 20 sessions at major International conference/symposia sessions; she actively organized 7 conferences. She wrote scientific projects focused to CDG and she coordinated several patient-friendly resources.

She is an active member for the Spanish CDG organization, EURORDIS and moderator at RARECONNECT. She is the author of the chapter of the book named "Congenital Disorders of Glycosylation (CDG): From glycoproteins to patient care", Royal Society of Chemistry (2012).

Julia Boonnak

Julia Boonnak is the mother of a 3 year old son with Congenital Disorders of Glycosylation (CDG). He was diagnosed at 1 year. She is a supporter and advocate for rare diseases-specifically CDG.

She is the Head of English as an Additional Language at Bromsgrove School Thailand and has recently moved to the UK to take the position of Head of English as An Additional Language at Bromsgrove School in the UK. She is a member of the team of writers for Globalgenes.org, and also writes her own CDG awareness blog at cantdogymnastics.blogspot.com.

In addition, Julia Boonnak received an MEd in Bilingualism in Education from the University of Birmingham and is currently taking a second degree in Natural Sciences with the Open University in the UK.

Julia was the winner of the Global Genes Project June 2013 Poetry Contest! Julie's poem (which you can read in the link below) was a great poem displaying the theme of perseverance.

http://globalgenes.org/poetry-contest-winners-announced/

10.8. Highlighting projects: Promoting collaboration within families and professionals

Robert Pleticha

Robert Pleticha joined EURORDIS in 2010 as the Online Patient Communities Coordinator later becoming RareConnect Project Manager.
Robert (Rob) is part of the Communication team where he works with patients and patient groups to develop and maintain social networks for specific rare diseases. In addition he actively promotes volunteer opportunities for the Therapeutic Recreation Programs.

Previously, Rob has worked as a respite care provider, creating volunteer networks and organising capacity building workshops for patient representatives. He first worked with Developmental Services Center in Champaign, Illinois supporting adults with disabilities in a sheltered workshop environment.

Following this, he became a United States Peace Corps volunteer in Zalau, Romania working with Dorica Dan and the Romanian Prader Willi Association, and the Romanian National Alliance for Rare Diseases to create the Pilot Reference Center for Rare Diseases in Romania in partnership with Frambu.

Rob has a Bachelor of Science in Psychology from the University of Illinois, Champaign-Urbana.

He is a native English speaker who also speaks Romanian and French.

Dr Mercedes Serrano

Mercedes Serrano holds a degree in Medicine from the University Miguel Hernández (Alicante), specializing in Pediatrics and specific areas in the Hospital Universitario La Paz in Madrid. Doctor of Medicine from the University of Barcelona (Barcelona). Master in Neuroscience and Behavioral Biology at the University Pablo de Olavide (Sevilla). Master in Pediatric Neurology at the University of Barcelona (Barcelona).

Since 2007, she has worked as a researcher for the CIBER-ER (Consortium for Biomedical Network Research on Rare Diseases), Instituto de Salud Carlos III, attached to the Metabolic Diseases Unit of the Hospital Sant Joan de Deu (Barcelona, Spain).

10.9. Theme 2: Congenital Disorders of Glycosylation Thoughts on Clinical Care

Dr Marc C. Patterson

Marc C. Patterson studies Niemann-Pick disease type C and other lysosomal diseases, congenital disorders of glycosylation, and pediatric multiple sclerosis.
Dr. Patterson's research activities have included laboratory studies in neuropharmacology at the University of Queensland and in lysosomal diseases at the National Institutes of Health (NIH).

He has also participated in clinical trials and natural history studies at NIH, Columbia University and Mayo Clinic. Dr. Patterson's research has been funded by the NIH, industry, the National MS Society and other not-for-profit foundations.

Pr Javier Corral

Javier Corral holds a Ph.D. in Biochemistry and Molecular Biology from the University of Salamanca 1992 (Spain). He was a Post-doctoral Fellowship at the Medical Research Council from the Laboratory of Molecular Biology (MRC-LMB) in Cambridge (UK). In addition, he was scientific visitor of the Welcome Trust Centre at the CIMR in the University of Cambridge (UK). From 2006, he is Professor at the University of Murcia. Department of Medicine. Spain.

He is author of 150 articles in peer-reviewed journals with more than 3500 citations. Hirsch's index 26. His major research focus is related to antithrombin and genetic risk factors of thrombosis. He is vice-President of the Spanish Society of Thrombosis and Haemostasis (SETH).

Dr Maria Eugenia de la Morena Barrio

Maria Eugenia Barrio holds a Ph.D. in Medicine at the University of Murcia 2013 (Spain). She is the author of 11 articles in peer-reviewed journals. Her work is focused on antithrombin and genetic risk factors of thrombosis.

In the department of Internal Medicine at the University of Murcia, the group that is now working on Congenital Disorders of Glycosylation is formed by Prof. Javier Corral de la Calle and Dr. Mª Eugenia de la Morena Barrio. Prof. Corral has directed Eugenia PhD thesis in which one of the main aims was to unravel the mechanism underlying the high incidence of hemostatic problems in CDG patients. Some results are published in the Journal of Thrombosis and Haemostasis.

Dr Daisy Rymen

Daisy Rymen received her MD from the University of Leuven (Belgium) in 2010. Currently she combines her internship in Pediatrics with research. She is a PhD student in the lab of Professor Gert Matthijs and Professor Jaak Jaeken. Her main field of interest is Congenital Disorders of Glycosylation.

Dr Miski Mohamed

Miski Mohamed received her medical degree from the Radboud University Nijmegen (Netherlands) and is currently obtaining a PhD at the department of Pediatrics. Miski Mohamed has special interest in diagnosis and treatment of common but complex symptoms in CDG.

Furthermore she is a strong advocate for bringing clinical research closer to patients and parents and encouragement of interaction between these two. To reach these goals she is one of the organizers of the CDG patient and parent annual meeting in Nijmegen. She is active in the Dutch online CDG community where in an interactive environment patients and parents can ask questions and be informed by professionals.

For information purposes she helped develop the Nijmegen CDG website.

10.10. Theme 3: Biochemical and genetic diagnosis of CDG

Dr Dulce Quelhas

Dulce Quelhas received her MSc in Human Genetics from Porto University Sciences Faculty and is currently working at the Unidate de Bioquimica Genética, Centro de Genética Médica (Centro Hospitalar do Porto, Portugal).

After finishing her training period and her MSc in Human Genetics in 1998, she was trained in the Congenital Disorders of Glycosylation in the Metabolic Department of University Hospital Gasthuisberg in Leuven, Belgium, by Prof. Jaak Jaeken.

In 2000 she was trained in the molecular diagnostic of Congenital Disorders of Glycosylation in the Laboratory for Molecular Diagnosis of the Department of Human Genetics of KU Leuven, Belgium, by Prof. Gert Matthijs. Since 1998 she actively participated in research projects dedicated to CDG. Amongst them, the “Screening Project for CDGS”, financed by “Comissão de Fomento de Investigação em Cuidados de Saúde” (portuguese health ministery).

In 2000 in the “Hepatic presentation of CDGS”, financed by “Comissão de Fomento de Investigação em Cuidados de Saúde (portuguese health ministery). From 2000 to 2003 participation in the E.U. Project “EUROGLYCAN” as member of the Porto satellite Center. And later she actively participated in the project “EUROGLYCANET”.

Dr Hudson H. Freeze

Hudson Freeze received his Ph.D. in Biology from the University of California, San Diego in 1976 and is currently both Director of the Genetic Disease Program and Professor of Glycobiology at Sanford-Burnham Medical Research Institute in La Jolla, CA. Dr. Freeze has worked in Glycobiology for over 37 years – the last 17 focused primarily on the identification and understanding of human glycosylation disorders. His early work in the late 1970's centered on understanding lysosomal enzyme targeting related to human I-cell disease, and later (1997-on) refocused on Congenital Disorders of Glycosylation (CDG).

The Freeze lab has a strong and well-demonstrated history of success in basic science and in patient-centered, translational and therapeutic applications. Dr. Freeze and his collaborators discovered the first patient with an inherited deficiency in phosphomannose isomerase (CDG-Ib) and successfully treated him with oral mannose supplements Alone or in collaborations; the Freeze lab has now discovered 17 human glycosylation disorders.

At present the lab is the only one in the United States primarily devoted to studying CDG. Close collaborations with physicians and family organizations have enabled the lab to (research) diagnose approximately 140 CDG patients and carry out research on another 60 with unknown glycosylation defects. To understand the basic nature of these defects, researchers in the Freeze lab combine genetic and biochemical analysis with cell biology.

Recently, researchers in the lab have also turned to whole exome sequencing—this technology has allowed the Freeze lab to identify 7 new glycosylation disorders. Motivated by these patients and their new defects, the lab is poised to expand the fundamental understanding of monosaccharide metabolism, monosaccharide and nucleotide sugar transport and N-glycan transfer to proteins.

Dr. Freeze is a leader in the Glycobiology field, and a sought-after speaker on CDG issues. In recent years, Dr. Freeze served as the President for the Society of Glycobiology (2012), co-chaired an ASBMB/SFG co-meeting (November 2012), and chaired the 2011 Glycobiology Gordon Research Conference. He is the Vice President Elect for Science Policy of FASEB, a US-based organization representing over 100,000 medical research scientists.

Dr Dirk Lefeber

Radboud University Medical Center, Department of Neurology, Laboratory for Genetic, Endocrine and Metabolic disease, Nijmegen, The Netherlands

Dirk Lefeber received his PhD degree in Chemistry from Utrecht University, studying the chemical and analytical aspects of glycans. After a post-doctoral visit to the Eijkman-Winkler Institute for microbiology to work on glycans in the immune system, he followed a 4-year training to become a registered Clinical Biochemical Geneticist and hold a staff position on Glycosylation Disorders at the Radboud University Medical Center.

His professional interests focus on elucidation of novel human genetic disorders of glycosylation, development of novel methods to facilitate CDG diagnostics and understanding of the pathophysiology in CDG. In addition, he serves as scientific expert on CDG diagnostics in the scientific advisory board of ERNDIM, by organizing a quality control scheme for CDG screening with >60 world-wide participating centers.

Dr Paula Videira

Faculdade de Ciências Médicas, Universidade Nova de Lisboa, Portugal Chronic Diseases Research Center (CEDOC)
Paula was born in 1972, graduated in Biochemistry at the Coimbra University (1995), and got a Ph.D. degree in Biotechnology at Instituto Superior Técnico, Lisbon (2002).

She is presently an Assistant Professor at Faculdade de Ciências Médicas (FCM), from Nova University of Lisbon (UNL), Portugal and invited researcher at Brigham and Women's Hospital - Harvard Institutes of Medicine, in Boston USA. She belongs to the Chronic Diseases Research Center (CEDOC), where she coordinates the Glycoimmunology research group.

Paula is engaged in teaching and research activities. She coordinates disciplines such as Immunology within the first and PhD degree courses. In 2013 she is launching the international *e-learning* post graduate course in Glycobiology and Glycochemistry, that includes topics related with Congenital Disorders of Glycosylation. She has regular activity as peer-reviewer in international journals in the field of Immunology and Glycobiology. Paula is also the scientific coordinator of the Cell Biology Platform @ CEDOC and her team has developed unique skills in the development and maintenance of primary cultures of immune cells.

Her scientific activity is devoted to dendritic cell (DC), one of the most important cells of the immune system. She presently supervises 8 PhD students and the main goal of her research team concerns the exploitation of DC-based immunotherapy, and to understand the role of specific glycan structures. This knowledge may give cues to fine tune the DC ability to encounter T cell niches when administered intravenously and to activate T cell. This could result in the establishment of improved immune-based treatments. She has been awarded by international institutions such as the EMBO (2011), the Fulbright Commission (2013 as Fulbright Professor).

Her articles in international scientific periodicals with referees
http://www.ncbi.nlm.nih.gov/pubmed/?term=videira%20p

Paula and her team have been strongly interacting with the Portuguese Association CDG and other Metabolic Rare Diseases. This collaboration aims to contribute to a better understanding of the causes and mechanisms of CDG and to potentiate research projects (such as meta-analysis) in order to create CDG teaching resources, and to raise awareness amongst society, clinicians and researchers.

10.11. Theme 4: Overview of therapeutic approaches

Dr Eva Morava-Kozicz

Eva Morava was born in 1966 in Budapest, Hungary. She completed her medical studies in 1990 at the University of Pecs, in Hungary. She specialized in pediatrics in 1994 and from that time on she worked as a staff member at the Department of Pediatrics, and later at the Department of Human Genetics at the University of Pecs, Hungary. Between 1996 and 1998 she participated in the fellowship training program in neonatology and biochemical genetics at the Tulane University Medical Center at the Department of Pediatrics and at Hayward Genetics Center in New Orleans, LA. She specialized in human genetics in 1999.

She defended her PhD thesis on *Molecular cytogenetic investigations in mental retardation syndromes* in 2000 in Pecs, Hungary, where she worked as a clinical geneticist until 2002. She did her further year of training in metabolic pediatrics from December 2002 at the Radboud University Nijmegen Medical Center (RUNMC) and worked as a staff member and metabolic pediatrician at the RUNMC since 2004. Since 2012 she became faculty at the Tulane University Medical Center, at the Hayward Genetics Center, as a biochemical geneticist.

Eva Morava is a member of the national and international committees and scientific advice groups. Her list of publications includes more than 150 peer reviewed scientific papers. Her research group focuses on syndromic forms on inborn errors of metabolism. Her special interest lies in research on mitochondrial disorders and research on congenital disorders of glycosylation (CDG). She has a strong collaboration with the Institute of Genetic and Metabolic Disease at the Radboud University Medical School and she established the Nijmegen Center for CDG (www.nijmegencdg.nl). She is a member of the editorial board of the Journal of Inherited Metabolic Disorders. Eva Morava is currently a full professor of pediatrics at the Hayward Genetics Center at Tulane University, New Orleans.

Information about Research on Congenital Disorders of Glycosylation is available at**:**
http://tulanehealthcare.com/physicians/profile/Eva-Morava-Kozicz-MD

Dr Thorsten Marquardt

Dr. Thorsten Marquardt is Professor of Pediatrics and Head of the Department for Inherited Metabolic Diseases at the Children's Hospital of the University in Münster, Germany. It is one of the largest metabolic disease units in Germany taking care of more than 1000 children with inherited metabolic diseases imcluding a laboratory offering a a broad spectrum of biochemical and molecular analyses.

He has a longstanding research interest in CDG and his group discovered several new subtypes beginning with CDG-Ib in 1998 and ending with PGM1-CDG in 2013. Treatments for CDG-Ib with mannose, for CDG-IIc with fucose, and PGM1-CDG with galactose were developed. The current focus is on the development of a specific treatment for CDG-Ia.

Awards:
1998 Horst-Bickel-Award (for the treatment of CDG-Ib)
2003 Adalbert-Czerny-Award

Dr Marc Martinell

Marc Martinell received a PhD in chemistry from the University of Barcelona focused on molecular recognition. His work was awarded with University's extraordinary award as one of the best thesis from 2004. The same year Marc joined Crystax Pharmaceuticals where he became Head of Biophysics with the responsibility of setting up a platform for drug discovery based on fragment screening. Later on, Marc became Head of Project Generation at Oryzon Genomics working on epigenetics.

At Oryzon, Marc was responsible of the team in charge of structural biology, biophysics, target ID and assay development, and actively participated in the identification of first-in-class LSD1 inhibitors. In late 2011, Marc, together with Juan Aymamí and Xavier Barril, founded Minoryx Therapeutics, a company committed to finding innovative treatments for life threatening rare diseases. Minoryx's focus is on neurometabolic diseases of genetic origin, and its approach is mainly based on the identification of pharmacological chaperons through a proprietary technological platform named SEE-Tx (Site-directed Enzyme Enhancement Therapy). As a complementary approach, Minoryx also pursues drug repositioning based projects. Marc co-authored 12 patents and several publications.

Minoryx is collaborating with Dra. Belen Pérez, from Universidad Autónoma de Madrid, in the discovery of pharmacological chaperones for the treatment of CDG's.

John Evans

John Evans joined Agios in September 2009 and has over 10 years of biopharmaceuticals industry experience across a wide range of business functions.
At Agios, John is head of business development and new product planning.

Agios is a biopharmaceutical company passionately committed to applying its scientific leadership in the field of cellular metabolism to transform the lives of patients with cancer and inborn errors of metabolism (IEMs), which are a subset of orphan genetic metabolic diseases.

Prior to joining Agios, John served as Director of Product Development at Infinity Pharmaceuticals where he led the company's most advanced oncology programs, the Hsp90 inhibitors IPI-504 and IPI-493. He also established Infinity's program management, new product planning, and investor relations functions.
Before Infinity, Mr. Evans held positions in the Pharmaceuticals practice of McKinsey & Company, at MedImmune, and at Bayer Pharmaceuticals. John earned an MBA in Healthcare Management from Wharton, a Masters in Biotechnology from the University of Pennsylvania, and a BA from Yale University.

10.12. Theme 5: Animal models

Dr Christian Körner

Christian Körner received his PhD on `Interactions of the M6P/IGF-II-receptor with heterotrimeric GTP-binding proteins´ in the `Institute for Biochemistry II´ of the Georg-August-University Göttingen and is currently teaching at `Inherited endocrine and metabolic diseases´ in the `Center for Child and Adolescent Medicine´ of the University of Heidelberg. Since March 2013 he is Chairman of the state doctorate supervisory board I of the Medical Faculty at the University Heidelberg. Congenital Disorders of Glycosylation (CDG) is one of his major research focuses. He actively writes articles and chapters for books focused on CDG. He is strongly involved in improving diagnosis and in expanding therapeutic options targeted to this rare metabolic disease. He also participates in the events organized by the CDG German association, named Glycokids. Some of his publications:

Schneider A, Thiel C, Rindermann J, DeRossi C, Popovici D, Hoffmann GF, Gröne HJ, Körner C. Successful prenatal mannose treatment for congenital disorder of glycosylation-Ia in mice. *Nature Medicine* 18:71-73 (2011)

Thiel. C., Körner C., Mouse models for congenital disorders of glycosylation. Journal of Inherited Metabolic Diseases 34: 879-889 (2011)

11. SATELLITE SYMPOSIUM VENUE

Information about the venue:

Centro Cívico la Sedeta
Sicília, 321
08025 , BARCELONA
SPAIN

Website: http://www.bcn.cat/cclasedeta/ca/welcome.htm

"La Sedeta" was the most important textile plant in Gracia and, maybe, in Barcelona because it was one of the first textile plants in Europe who manufactured silk. Now it is a civic building, where there are classes for adults, concerts and conferences.

Walking around it, we can see that this is a very quiet neighborhood. It has the structure of the Eixample, but it's quieter. In the streets we can't see a lot of people staying and talking, maybe because there aren't many green areas. The only gardens are inside the interior patio shared by the entire block, like "jardins d'Antoni Puigverd", "jardins de Caterina Albert" or "La Sedeta". The centre of the activities is passeig Sant Joan, where there are a lot of shops and one of the only sites of the neighborhood where we can see people (generally old people) sitting on the benches and talking.

We are very grateful to Centro Cívico la Sedeta for allowing us use their facilities during the Satellite Symposia.

12. GENERAL INQUIERIES, PUBLIC AND MEDIA INQUIRIES

Contact of Conference Coordination Office at:

Rosália Félix (CDG mother, Portugal)
Portuguese Association for CDG and related Rare Metabolic Diseases.

Website: http://sindromecdg.orgfree.com/

Phone: 0033 641 84 25 55

Email: cdgawareness@gmail.com

13. COMMUNICATION AND INFORMATION CHANNELS

Please follow us on our different communication channels to stay update with news and announcements related to this Satellite Symposia:

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

- RareConnect
  https://www.rareconnect.org/en/community/cdg

- Facebook

  Please follow:

"CDG United"	https://www.facebook.com/groups/glycosylation/
"CDG Family Network"	CDG Family Network
"Sindrome CDG Portugal"	https://www.facebook.com/sindrome.portugal
"AESCDG Spain"	https://www.facebook.com/aescdg.cdgsindrome
"Syndrome CDG Family France"	Syndrome CDG Family : plus jamais seul !!!

14. OTHER ADDITIONAL INFORMATION

14.1. Advice if you are travelling with children and adults

Please be sure that you have all documentation in case that you need medical assistance.

Emergency phone numbers

- National Police: 091
- Local Police: 092
- Firefighters: 080
- Medical emergency: 061

Medical urgency of a patient with inborn errors

In case of a medical urgency of a patient with inborn errors of metabolism the hospitals are:

- Pediatric Hospital:
  Hospital Sant Joan de Déu
  Passeig Sant Joan de Déu, 2
  08950 Esplugues de Llobregat,
  Barcelona,
  Espanha
  +34 932 53 21 00

- Unity for Adult with Inborn Errors of Metabolism Hospital:
  Hospital Clínic de Barcelona
  Unitat de Malalties Metabòliques, Hospital Clínic de Barcelona, carrer Villarroel 170
  C.P. 08036
  Phone: +34 932 27 54 00

Medication

Please inform your medical doctor about the trip to Barcelona and collect advices and tips directly from your CDG professional.

We want to stress that Families should not forget to take their home medication (and enough!) with them!

14.2. Transport Barcelona and the airport and back

The airport is located 16 kilometers south of the city and is also known as El Prat, the town where it is located.

Connections between Barcelona and the airport and back:

- Aerobús (A1 and A2): adapted to disabled people
- Train (RENFE)
- Bus number 46 (TMB): adapted to disabled people
- Night bus (NITBUS): adapted to disabled people

Complete and reliable information available in spanish, catalán, english and french is available at:
http://www.barcelona-access.com/?idioma=1

14.3. Accommodation

A list of hotels is provided by the Portuguese Association for CDG and related Rare Metabolic Diseases. Special rates to some hotels have been negotiated**.** Please make your reservation as soon as possible to avoid any inconveniences. The list is available at cdgawareness@gmail.com

14.4. After conference hours

Turisme de Barcelona's website for disabled people

In the following website there is important and useful information available in in spanish, catalán, english and french: http://www.barcelona-access.com/?idioma=1

- A search engine for accessible places of interest for each type of disability:
  Museums, parks, beaches, unique buildings, monuments and World Heritage sites.
- Lists accessible transport facilities as well as obstacles, to help you get to Barcelona and get around the city, either by metro, bus, tram or the Barcelona Bus Turístic.
- Here you'll find tourist information offices, options for visits and tours or adapted sports, as well as specialised travel agencies or orthopaedic shops.

More interesting information about Accessible Tourism in Barcelona and surroundings cities

http://www.turismeperatothom.com/en/
http://barcelona.de/en/barcelona-disabled-persons.html

Attractions that are fully accessible

An overview of the attractions and accessibility restrictions can be found on the following site:
http://www.vienaeditorial.com/barcelonaaccesible/angles/ind_lugares.htm

Restaurants

Full information is found at: http://www.timeout.com
Although a search was done and refined by**:**
Types of venues: Restaurants and cafés, Search by: Critics' choice: 497 matches found.
We have selected few based in our own experience (these are recommendations, you must check for detailed information):

Taberna el Glop	La tavern del Born
Taberna la Llesca	Los Cachitos
Can Maño	Fonda Espanya
7 Portes	Quimet i Quimet
Casa Leopoldo	Fresco
L'Antic Magatzem	Els Quatre Cats
Attic	Mubufet
Citrus	Txapela
La Fonda Port Olímpic	Café de l'Ópera
Buenas Migas	Tinta Roja (Cabaret-bar)

The weather forecast

http://fr.weather.com/

15. Official Photographer: Marisa de Andrés

About the Official Photographer:

Marisa de Andrés received BSc in Biology from Universitat de Barcelona and is currently doing her PhD in Hematopoiesis Biology at the CRG. Her professional interests focus on the field of leukemia and hematopoietic stem cell biology, and in her current project she's investigating the role of Msi2 protein in hematopoiesis. In her spare time she is doing a degree in Humanities and she is a passionate photographer.

Marisa de Andrés was interviewed and shared her perspective about her participation as official photographer in this event.

Q&A with Marisa de Andrés:

Q: Two years ago you were the official photographer at the "FIRST CDG IBERIAN CONFERENCE". Can you tell us more about what you experienced during that conference?

MA: I enjoyed very much the time that I spent with the families and the researchers. I think it was an extraordinary event where the families had the opportunity to talk with the experts and ask for advice. I think it was a unique experience!

Q: This year you repeat the experience at the "1st World Conference on Congenital Disorders of Glycosylation for Families and Professionals: a booming story of sugar trees". What are your main expectations?

MA: I'm completely sure that it's going to be a success and a unique opportunity for CDG patients, families and researchers.

"Marisa's de Andrés Art"

16. Video recording conference

The meeting organizers and presenters will enable recording the meeting and allow making it available on the Youtube channel of the future Portuguese Association for CDG and related Rare Metabolic Diseases. The recorded presentations will include the slides shown at the meeting.

The video recording will be especially useful for people who cannot attend the meeting and might want to view it at convenient time. In addition, these videos will be an important educational and awareness material amongst the general public.

BIOcomuniCA'T is in charge of video recording the Satellite Symposia dedicated to CDG. To learn more about their organization:

CAT:
BIOcomuniCA'T és un grup d'investigadors internacionals amb inquietuds divulgatives que promou l'apropament de la ciència i la societat a través de l'organització d'activitats científiques.
Oferim a les institucions científiques recursos per mostrar la seva recerca al públic. Organitzem i realitzem a activitats educatives a tot tipus de centres educatius. Estimulem el debat entre científics i societat a través de cafès i tertúlies.

ES:
BIOcomuniCA'T es un grupo de investigadores internacionales con inquietudes divulgativas que promueve el acercamiento de la ciencia y la sociedad a través de la organización de actividades científicas.
Ofrecemos a las instituciones científicas recursos para mostrar sus investigaciones al público. Organizamos y realizamos actividades educativas en todo tipo de centros educativos. Estimulamos el debate científico entre científicos y sociedad a través de cafés científicos y charlas.

EN:
BIOcomuniCA'T is a group of international researchers interested in science dissemination among the science through the organization of all kind of scientific activities.
We provide resources to scientific institutions to transmit their research to the public. We organize and carry out educational activities for all types of schools. We stimulate scientific discussions through Scientific Cafes.

Website:

http://www.biocomunicat.com/

"Good, loving care is the most important parents can give to their child with a CDG" , Interview with Pr. Dr. Jaak Jaeken,"First Iberian Conference 2011, Barcelona, Spain". Guía Metabólica. Sant Joan de Déu Hospital.Barcelona. Spain.

Portuguese Association for CDG and related Rare Metabolic Diseases disclaimer

The organizers for the "**1st World Conference on Congenital Disorders of Glycosylation for Families and Professionals: a booming story of sugar trees"** have made every effort to ensure that the conference achieves its goal of disseminating the last advance and breakthrough information related to **Congenital Disorders of Glycosylation**. Furthermore, the organizers have made every effort to ensure that all participants remain comfortable and enjoy the experience of the conference. However, the organizers do not take any responsibility for any damage, loss or inconvenience participants may incur or experience in connection with the conference. In addition, the organizers cannot be held responsible for the correctness or appropriateness of the talks, papers, panels, tutorials and demonstrations included in the conference.

Moreover, in the event of industrial disruption or other unforeseen circumstances, the organizers accept no responsibility for loss of monies incurred by delegates. The organizers accept no liability for injuries/losses of whatever nature incurred by participants and/or accompanying person, nor for loss or damage to their luggage and/or personal belongings. Participants are expected to make their own arrangements with respect to personal insurance.

About the Portuguese Association for CDG and related Rare Metabolic Diseases:
The Portuguese Association for CDG and other Rare Metabolic Diseases (APCDG-DMR) is a non-profit organization founded in 2010 by families affected by Congenital Disorders of Glycosylation (CDG) and related Rare Metabolic Diseases.

©**Author**: Vanessa Ferreira (Portuguese Association for CDG and related Rare Metabolic Diseases).
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