

**SSIEM Official Satellite Symposia
Second World Conference on
Congenital Disorders of Glycosylation (CDG)
for Families and Professionals:
a challenging story of sugars trees**

28 August 2015 to 30 August 2015 - Lyon (France)



**Congenital Disorders
of Glycosylation**

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The “SSIEM Official Satellite Symposia – Second World Conference on Congenital Disorders of Glycosylation (CDG): a challenging story of sugar trees” aims to raise awareness about Congenital Disorders of Glycosylation (CDG) around the world and to foster an exceptional collaborative model involving patients, family members, researchers and physicians.

This conference is organized by the Portuguese Association for CDG (APCDG), in partnership with several associations and/or country CDG patient advocates: CDG Australia, CDG Brazil, CDG Czech Republic, CDG Denmark, Foundation Glycosylation (the FoG) Canada, CDG Italy/Ireland, CDG Israel, Les ptits CDG France, CDG Spain, CDG Sweden, CDG USA, CDG UK charity and CDG Netherlands.

Acknowledgements

We wish to thank our talented speakers who kindly accepted our invitation. Without their willingness to share their expertise, this conference would not be possible.

We would also particularly like to thank Professors Pascale de Lonlay (France), Nathalie Seta (France), Maria Antonia Vilaseca (Spain), Jaak Jaeken (Belgium) and Christine Vianey-Saban (France), for providing superb brainstorming and advices that will make this conference a reality.

We are also indebted to all volunteers who have read early drafts of the conference materials and offered their feedback and corrections: Merell Liddle (CDG Australia), Sandra Pereira Pinto (CDG Portugal and Spain) and Pierre Morandat (CDG France).

On behalf of the organisers,
Vanessa Ferreira, president and founder APCDG

Foundation Glycosylation (FoG) is the official sponsor of the videos targeted to the “SSIEM Official Satellite Symposia – Second World Conference on Congenital Disorders of Glycosylation (CDG): a challenging story of sugar trees”:



The organisers are pleased to announce that the Foundation Glycosylation (FoG) founded by Duncan Webster (Canada), is the official sponsor of the videos of all oral session that will be given during the conference. This material will be available in the Youtube channel dedicated to “SSIEM Official Satellite Symposia – Second World Conference on Congenital Disorders of Glycosylation (CDG): a challenging 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>

The CDG French Association” Les P’tits CDG” supports one activity targeted to our children and adults during the kindergarten service.

”Les P’tits CDG” is formed by parents of children with CDG Syndrome. The association stimulates the meeting of families concerned by this disease, the sharing of experiences and the integration of all disabled children in society.

Thank you all for your kindness, which will provide an exceptional moment for our children and adults. For more information about Les P’tits CDG: <http://www.lesptitscdg.org/>



The official website to disseminate information and materials elaborated to the “Second World Conference on Congenital Disorders of Glycosylation (CDG): a challenging story of sugar trees” is:

<http://www.ssiem2015.org/>

We are not responsible for any information posted in other websites.

Pre-Program at a glance

The different sessions, initial program, invited speakers and chairs are listed below. Last minute changes may occur:

28 August 2015, Friday

14.00	Informal gather together (visit Lyon)
19.00	Dinner in a Brasserie (an authentic and historical place in Lyon's gastronomy)

29 August 2015, Saturday

08.00-9.00	Registration, Welcome and gather together Kindergarten receives your child/adult
09.00-09.10	Welcome Jaak Jaeken, Centre for Metabolic Diseases, University Hospital Gasthuisberg Leuven, Belgium

Topic 1	Clinical presentations of CDG (I) Session Chair: Jaak Jaeken (Belgium) and Begoña Cano (CDG patient advocate, Spain)
09.10-09.30	<u>Unraveling neurologic aspects from a cross-sectional study.</u> Mercedes Serrano & Belén Pérez Dueñas, Child Neurologist and CIBERER researcher at the San Juan de Déu Children's hospital in Barcelona, Spain
09.30-09.50	<u>Intellectual disability in CDG.</u> Marc Patterson, Chair of the Division of Child and Adolescent Neurology at Mayo Clinic and Professor of Neurology, Pediatrics and Medical Genetics at Mayo Clinic College of Medicine, USA
08.50-10.15	<u>The cerebellar involvement in CDG.</u> Rita Barone, Department of Pediatrics, University of Catania, Catania, Sicily, Italy
10.15-10.45	Round table: Session Chair: Mercedes Serrano (Spain). The panel that will answer to questions is formed by: Rita Barone (Italy), Marc Patterson (USA), Stephanie Grunewald (UK), Belén Pérez Dueñas (Spain), Eva Morava (USA & Belgium) and Ágata Fiumara (Italy)
	Session Chair: Heta Pontela (CDG advocate, Finland) and Marketa Tesarova (Charles University in Prague, First Faculty of Medicine and General

	University Hospital in Prague)
10.45-11.05	<u>Out of the Undiagnosed Diseases program: Early Results from the Clinical and Basic Investigations into Known and Unknown CDG's, the USA experience.</u> Lynne A. Wolfe, MS, CRNP Associate Investigator Undiagnosed Diseases Network, USA
11.05-11.30	Coffee break 2 & networking
11.30-11.50	<u>The major clinical findings concerning the skin manifestations in CDG patients.</u> Daisy Rymen, Centre for Human Genetics, University of Leuven, Leuven, Belgium
11.50-12.10	<u>Endocrine aspects in PMM2-CDG: diagnostic approach and proposed management.</u> Miski Mohamed, Institute for Genetic and Metabolic Disease, Radboud University Nijmegen Medical Centre, Nijmegen, Netherlands
12.10-12.20	Round of Questions & Answers
Topic 2	Diagnosis of CDG Session Chair: Andrea Berarducci (CDG Family Network, USA), Sandra Pereira Pinto (AESCDG & APCDG, Spain) and Hana Hansikova (Department of pediatrics and adolescent medicine, Charles University - First faculty of medicine)
12.20-12.40	<u>CDG genetics.</u> Dulce Quelhas, Centro de Genética Médica Jacinto de Magalhães, Porto, Portugal
12.40-13.00	<u>CDG diagnosis: strengths, weaknesses, and the road forward.</u> Dirk Lefeber, Institute for Genetic and Metabolic Disease, Radboud University Nijmegen Medical Centre, Nijmegen, Netherlands
13.00-13.20	<u>How can a genetic counselor help CDG families? Lessons from the Undiagnosed Diseases program.</u> Ellen MacNamara, MS, GC Associate Investigator Undiagnosed Diseases Network, USA
13.20-13.30	Round of Questions & Answers
13.30-15.00	Networking and sharing lunch 1
Topic 3	Day- to- day Live Session Chair: Esther Holten (CDG patient Advocate, Netherlands)

	and Dylan Mordaunt (SA Pathology, University of Adelaide, University of Queensland, Australia)
15.00-15.20	<u>Socio-emotional Problems in Children with CDG.</u> Charles Lourenço, Clinical Geneticist Hospital das Clínicas - Faculdade de Medicina da USP de Ribeirão Preto, Brasil)
15.20-15.40	<u>Helping siblings when a brother or sister's has special needs.</u> Regine Scelles, Psychology department, Université de Rouen, France
15.40-16.00	<u>Professional and parental perspectives on living with a rare disease.</u> TBC
16.00-16.30	Coffee break 2 & networking
Topic 4	From patients to professionals Session Chair: Bas Holten (CDG advocate, Netherlands) and Kimberley Walsh (CDG advocate, Australia)
16.30-16.45	<u>From diagnosis to action.</u> Julia Boonnak, CDG charity representative & mother, UK
16.45-17.00	<u>Managing CDG daily live.</u> Jill Ash, CDG charity representative & mother, UK
17.00-17.15	<u>How liver transplantation changed my life?</u> Fiona W., CDG Netherlands
17.15-17.30	<u>CDG Challenges and choices.</u> Barbara Vulso, Ireland& Italy CDG patient advocate & mother
17.30-17.45	<u>News from CDG USA patient group!</u> Andrea Berarducci and LaRae Mercer, CDG USA patient advocates and mothers
17.45-18.00	Round of Questions & Answers
	Session Chair: Esther Holten (CDG patient Advocate, Netherlands) and TBC
18.00-18.15	<u>Partageons l'expérience de notre vie ! (Share the experience of our lives!)</u> François-Xavier Levillain and all family, France
18.15-18.30	<u>How Salon Gianna can help in the research of CDG?</u> David Dragotto, Gianna's father, USA
18.30-18.45	<u>Ed's journey and how we helped in the discovery of SLC35A2-CDG.</u> Tim Jardine, CDG charity representative & father, UK

18.45-19.00	<u>NGLY1-CDDG: actions from a non-CDG organisation.</u> Matthew Might, NGLY Foundation, USA
19.00-19.15	Round of Questions & Answers
19.15-22.30	Networking and sharing dinner 1

30 August 2015, Sunday

Topic 5	Clinical presentations of CDG (II) Session Chair: Belén Pérez-Dueñas (Hospital Sant Joan de Déu, Barcelona, Spain)
09.00-09.20	<u>Orthopedics, The musculoskeletal system in CDG.</u> David Coman, Medical Director of Paediatrics, The Wesley Hospital, Brisbane, Australia and Academic Lead for Paediatrics, UnitingCare Health Clinical School
09.20-09.40	<u>The management of major gastrointestinal findings in CDG.</u> Stephanie Grunewald, Consultant Metabolic Medicine at Great Ormond Street Hospital for Children NHS Foundation Trust, London, United Kingdom
09.40-10.00	Round of Questions & Answers
10.00-10.20	<u>Which parameters must to be checked in order to avoid thrombotic complications in CDG patients?</u> Maria Eugenia de la Morena-Barrio, Centro Regional de Hemodonación, Ronda de Garay, Universidad de Murcia, Spain
10.20-10.50	<u>Clinical manifestations and management in adults with congenital disorders of glycosylation.</u> Ágata Fiumara, Associate professor, pediatric clinic, University of catania, Italy
10.50-11.00	Round of Questions & Answers
11.00-11.30	Coffee break 3 & networking
Topic 6	Collaboration and dissemination Session Chair: Ana Sánchez (CDG advocate, Spain) and Merell Liddle (CDG Australia)
11.30-11.45	<u>Argentinean experience with CDG.</u> Carla Asteggiano, CONICET-Centro de Estudio de las Metabolopatías Congénitas (CEMECO) , Facultad de Ciencias Médicas, Universidad Nacional de Córdoba (UNC) , Argentina

11.45-12.00	<u>Portuguese Research Network of Professionals and Patients Association for CDG: start small, think big.</u> Dr Esmeralda Martins, Unidade de Doenças Metabólicas, Departamento da Infância e Adolescência, Centro Hospitalar do Porto, Portugal and Dr Paula Videira, Assistant Professor, Medical Sciences Faculty, Chronic Diseases Research Center, Lisbon, Portugal
12.00-12.15	<u>Many are rare: a reference campaign for rare diseases.</u> Dafne D. Horovitz, Medical genetics at Instituto Fernandes Figueira, Brasil
Topic 7	Research in CDG Session Chair: TBC
12.15-12.40	<u>Unraveling COGs: clinical, biochemical and molecular highlights.</u> François Foulquier, Unité de glycobiochimie structurale et fonctionnelle (UGSF), CNRS/Université Lille, France.
12.40-13.00	<u>Role of the Conserved Oligomeric Complex (COG) and its partners in glycosylation in human cells.</u> Vladimir V. Lupashin, University of Arkansas for Medical Sciences Department of Physiology and Biophysics, USA
13.00-13.20	<u>The Undiagnosed Diseases program: Rarer CDG's Major clinical findings and management.</u> Lynne A. Wolfe, MS, CRNP Associate Investigator Undiagnosed Diseases Network, USA <u>Note:</u> this session will be defined under families requests
13.20-13.30	Round of Questions & Answers
13.30-15.30	Networking and sharing Lunch 2

Topic 8	Patient registries and biobanks Session Chair: TBC
15.30-15.50	<u>Understanding CDG patient registries and biobanks.</u> TBC
Topic 9	Animal models Session Chair: TBC
15.50-16.20	<u>The importance of animal models in CDG research progress.</u> Hudson Freeze, Director, Genetic Disease Program; Professor of Glycobiology, Sanford-Burnham Medical Research Institute, USA
Topic 10	Therapies

	Session Chair: TBC
16.20-16.50	<u>Novel therapeutic approach on Congenital Disorders of Glycosylation.</u> Thorsten Marquardt, Professor Pediatric Metabolic Diseases, Centrum für seltene Erkrankungen (ZSE) Münster, Universitätsklinikum Münster, Germany
16.50-17.00	Round of Questions & Answers
17.00-17.30	Coffee break 4 & networking
17.30-18.00	<u>Metabolomics and CDG.</u> Antonio Pineda-Lucena, Structural Biochemistry Laboratory, Centro de Investigación Príncipe Felipe, Valencia, Spain
18.00-18.20	<u>Learning about human iPSC models for glycosylation-related disease.</u> Stephen Dalton, Department of Biochemistry and Molecular Biology, University of Georgia, USA
18.20-18.35	<u>From discrete dilated cardiomyopathy to successful cardiac transplantation in congenital disorders of glycosylation.</u> TBC
18.35-18.50	<u>PMM2-CDG and chaperones.</u> TBC
18.50-19.20	<u>Successful dietary therapy in three CDG types and outlook for future therapies.</u> Eva Morava, Professor in pediatrics, Tulane University Medical School, USA
19.20-19.40	Round of Questions & Answers
19.40-19.50	Miscellaneous session TBC
Topic 11	Workshop
19.50	<u>Dancing at the hands of Morgan Webb Liddle.</u> Morgan Liddle and her followers (Grade 1A Para-Equestrian, Australia)
20.00	<u>Closing remarks</u>
20.15	Networking and sharing dinner 2

BACKGROUND:



The 1st World Conference on Congenital Disorders of Glycosylation for Families and Professionals: a booming story of sugar trees” (2013) 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. Our “First World CDG Conference (2013)” has welcomed more than 200 participants, formed by:

- 42 CDG families from 18 countries,
- 70 professionals from all continents and 23 countries.

The oral presentations are available at:

<https://www.youtube.com/playlist?list=PLa3xsZodHVEDRyAXVPP4imaqKKwrW79ld>



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<http://www.lyon-france.com/html/mvonlylyon/>