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 and chairs 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, PhD, MBA
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

NGLY1.org is the proud sponsor for the kindergarten nursing care available on 29 and 30 August 2015.

NGLY1.org is a patient-run, non-profit organization registered as the NGLY1 Foundation with the mission to eliminate the challenges of N-glycanase deficiency through research, awareness, and support. For more information about NGLY1.org visit: http://www.ngly1.org/

The CDG French Association" Les P'tits CDG" supports one activity on 30 August 2015 at the kindergarten service targeted to our children and adults.

"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/

Minoryx Therapeutics supports activity for the CDG kindergarten

Minoryx Therapeutics is focused on Inborn Errors of Metabolism, a group of rare diseases of genetic origin with high unmet medical need.

The company develop a new generation of pharmacological chaperones, a small molecule drug class aimed to restore protein functionality and which offer one of the most promising approaches to the treatment of genetic diseases severely affecting the central nervous system. As a complementary approach, Minoryx is also involved on developing small molecule drugs which counteract the cellular dysfunctions originated by the corresponding protein deficiency.

The program for the PMM2-CDG is at the early stages of discovery.

Source: http://www.minoryx.com/

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. If you wish to be updated we advise you to write to **worldconferencecdg2015@gmail.com**

Pre-Program at a glance

The following Family and Scientific program was elaborated by Vanessa Ferreira (Portuguese Association for CDG, APCDG) in collaboration with major CDG KOLs. All speakers and chairs will contribute in an altruistic manner.

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 (activity to be determined)**
18.30	**Dinner** in a Brasserie (an authentic and historical place in Lyon's gastronomy)

29 August 2015, Saturday

08.00-08.30	**Registration, Welcome and gather together** Kindergarten receives your child/adult
08.40-08.50	**Welcome** Jaak Jaeken, Centre for Metabolic Diseases, University Hospital Gasthuisberg Leuven, Belgium

Topic 1	**Clinical presentations of CDG (I)** Session Chair: Begoña Cano (CDG patient advocate, Spain) and Malina Stancheva-Ivanova (Bulgaria)
08.50-09.10	***Unraveling neurologic aspects from a cross-sectional study.*** Mercedes Serrano, Child Neurologist and CIBERER researcher at the San Juan de Déu Children's hospital in Barcelona, Spain
09.10-09.30	***Intellectual disability in CDG.*** 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
09.30-09.50	***The cerebellar involvement in CDG.*** Rita Barone, Department of Pediatrics - Child Neurology University of Catania, Catania - Italy
09.50-10.05	**Round table:** Session Chair: Mercedes Serrano (Spain). The panel that will answer to questions is formed by: Rita Barone (Italy), Marc Patterson (USA), Belén Pérez Dueñas (Spain), Stephanie Grunewald (UK), Eva Morava (USA & Belgium)

	and Ágata Fiumara (Italy)
	Session Chairs: Jaak Jaeken (Belgium) and TBC
10.05-10.25	***Out of the Undiagnosed Diseases program: Early Results from the Clinical and Basic Investigations into Known and Unknown CDG's, the USA experience.*** Lynne A. Wolfe, MS, CRNP Associate Investigator Undiagnosed Diseases Network, USA
10.25-10.40	***The major clinical findings concerning the skin manifestations in CDG patients.*** Daisy Rymen, Centre for Human Genetics, University of Leuven, Leuven, Belgium
10.40-10.55	***Endocrine aspects in PMM2-CDG: diagnostic approach and proposed management.*** Miski Mohamed, Institute for Genetic and Metabolic Disease, Radboud University Nijmegen Medical Centre, Nijmegen, Netherlands
10.55-11.05	Round of Questions & Answers
11.05-11.30	Coffee break 1 & networking
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, Czech Republic)
11.30-11.45	***CDG genetics.*** Dulce Quelhas, Centro de Genética Médica Jacinto de Magalhães, Porto, Portugal
11.45-12.00	***CDG diagnosis: strengths, weaknesses, and the road forward.*** Monique van Scherpenzeel, Institute for Genetic and Metabolic Disease, Radboud University Nijmegen Medical Centre, Nijmegen, Netherlands
12.00-12.15	***Towards the structural analysis of aberrant glycosylation: Focus on CDGs.*** Zuzana Pakanova, Institute of Chemistry, Department of Glycobiology, Central Analytical Laboratory, Slovak Academy of Sciences, Slovakia
12.15-12.30	***How can a genetic counselor help CDG families? Lessons from the Undiagnosed Diseases program.*** Ellen Macnamara, ScM, GC Associate Investigator Undiagnosed

	Diseases Network, USA
12.30-12.45	***How GlyMAP can boost discovery of new CDGs?*** Lars Hansen, Faculty of Health Sciences, University of Copenhagen, Denmark
12.45-12.55	Round of Questions & Answers
12.55-15.00	Networking and sharing lunch 1
Topic 3	**Day- to- day Live** Session Chair: Duncan Webster (Foundation Glycosylation (FoG) & CDG Canadian patient representative) and Dylan Mordaunt (SA Pathology, University of Adelaide**,** University of Queensland, Australia)
15.00-15.20	***Argentinean experience with CDG.*** Carla Asteggiano, CONICET-Centro de Estudio de las Metabolopatías Congénitas (CEMECO) , Facultad de Ciencias Médicas, Universidad Nacional de Córdoba (UNC) , Argentina
15.20-15.40	***Socio-emotional Problems in Children with CDG.*** Charles Lourenço, Clinical Geneticist Hospital das Clínicas - Faculdade de Medicina da USP de Ribeirão Preto, Brasil
15.40-16.00	***Helping siblings when a brother or sister's has special needs.*** Regine Scelles, Psychology department, Université de Rouen, France
Topic 4	**From patients to professionals** Session Chair: Kimiyo Raymond (USA) and Kimberley Walsh (CDG advocate, Australia)
16.00-16.15	***From diagnosis to action.*** Julia Boonnak, CDG charity representative & mother, UK
16.15-16.30	***How liver transplantation changed my life?*** Fiona W., CDG Netherlands
16.30-16.45	***CDG Challenges and choices.*** Barbara Vulso, Ireland& Italy CDG patient advocate & mother
16.45-17.00	***News from CDG USA patient group!*** Andrea Berarducci and LaRae Mercer, CDG USA patient advocates and mothers
17.00-17.15	Round of Questions & Answers
17.15-17.45	Coffee break 2 & networking

	Session Chair: David Cassiman (Belgium)
17.45-18.00	***How Salon Gianna can help in the research of CDG?*** David Dragotto, Gianna's father, USA
18.00-18.15	***Ed's journey and how we helped in the discovery of SLC35A2-CDG.*** Tim Jardine, CDG charity representative & father, UK
18.15-18.30	***NGLY1-CDDG: actions from a non-CDG organisation.*** Matthew Might, NGLY Foundation and father to Bertrand, USA
18.30-19.00	Round of Questions & Answers
19.00-19.30	***Surprise reserved for the Second World Conference on CDG (children and adults should attend)***
20.00-22.30	Networking and sharing dinner 1

30 August 2015, Sunday

Topic 5	**Clinical presentations of CDG (II)** Session Chair: Luísa Diogo (Portugal) and David Cassiman (Belgium)
08.40-09.00	***Orthopedics, The musculoskeletal system in CDG.*** David Coman, Medical Director of Paediatrics, The Wesley Hospital, Brisbane, Australia and Academic Lead for Paediatrics, Uniting Care Health Clinical School, Australia
09.00-09.20	***The management of major gastrointestinal findings in CDG.*** Stephanie Grunewald, Consultant Metabolic Medicine at Great Ormond Street Hospital for Children NHS Foundation Trust, London, United Kingdom
09.20-09.40	***Which parameters must to be checked in order to avoid thrombotic complications in CDG patients?*** Maria Eugenia de la Morena-Barrio, Centro Regional de Hemodonación, Ronda de Garay, Universidad de Múrcia, Spain
09.40-10.00	***Clinical manifestations and management in adults with congenital disorders of glycosylation.*** Ágata Fiumara, Associate professor, pediatric clinic, University of catania, Italy
Topic 6	**Collaboration and dissemination** Session Chair: Ana Sánchez (CDG Spain) and Merell Liddle (CDG Australia)

10.00-10.15	***Portuguese Research Network of Professionals and Patients Association for CDG: start small, think big.*** Esmeralda Martins, Unidade de Doenças Metabólicas, Departamento da Infância e Adolescência, Centro Hospitalar do Porto, Portugal and Paula Videira, Assistant Professor, Medical Sciences Faculty, Chronic Diseases Research Center, Lisbon, Portugal
10.15-10.30	***Many are rare: a reference campaign for rare diseases.*** Dafne D. Horovitz, Medical genetics at Instituto Fernandes Figueira, Brasil
10.30-10.45	***Learning about human iPSC models for glycosylation-related disease.*** Stephen Dalton, Department of Biochemistry and Molecular Biology, University of Georgia, USA
10.45-11.00	***Project occurrence of elevated cholesterol and heart disease CDG*** M van den Boogert, Vascular Medicine, University of Amsterdam, Academic Medicine Center, Amsterdam, Netherlands
11.00-11.10	Round of Questions & Answers
11.10-11.40	Coffee break 3 & networking
Topic 7	**Research in CDG** Session Chair: TBC
11.40-12.00	***TMEM165 a new player in CDG.*** François Foulquier, Unité de glycobiologie structural et fonctionnelle (UGSF), CNRS/Université Lille, France.
12.00-12.20	***Role of the Conserved Oligomeric Complex (COG) and its partners in glycosylation in human cells.*** Vladimir V. Lupashin, University of Arkansas for Medical Sciences Department of Physiology and Biophysics, USA
12.20-12.40	***The Undiagnosed Diseases program: Rarer CDG's Major clinical findings and management.*** Lynne A. Wolfe, MS, CRNP Associate Investigator Undiagnosed Diseases Network, USA Note: this session will be defined under families requests
12.40-12.50	Round of Questions & Answers
12.50-13.10	***CDG needs and opportunities*** Sandra Pereira Pinto & Vanessa Ferreira, Portuguese Association for CDG and related rare metabolic diseases (CDG patient advocates, APCDG-DMR, Portugal), Mercedes Serrano and Belen

	Perez Dueñas Mercedes Serrano (Child Neurologists and CIBERER researchers at the San Juan de Déu Children's hospital in Barcelona, Spain)
13.10-15.30	Networking and sharing Lunch 2

Topic 8	**Insights on breakthrough innovative research models** Session Chair: TBC
15.30-15.50	***The importance of animal models in CDG research progress.*** Hudson Freeze, Director, Genetic Disease Program; Professor of Glycobiology, Sanford-Burnham Medical Research Institute, USA
15.50-16.10	***Synapse Glycosylation Drives CDG Neurological Outcomes: Insights from an Animal Model*** Patricia Jumbo Lucioni, Postdoctoral Research Scholar, Department of Biological Sciences, Vanderbilt University, Nashville, TN, USA
Topic 9	**CDG Therapies** Session Chair: Elisa Leão-Teles (Portugal) and Patrícia Janeiro (Portugal)
16.10-16.30	***Novel therapeutic approach on Congenital Disorders of Glycosylation.*** Thorsten Marquardt, Professor Pediatric Metabolic Diseases, Centrum für seltene Erkrankungen (ZSE) Münster, Universitätsklinikum Münster, Germany
16.30-16.50	***Metabolomics and CDG.*** Antonio Pineda-Lucena, Structural Biochemistry Laboratory, Centro de Investigación Príncipe Felipe, Valencia, Spain
16.50-17.00	Round of Questions & Answers
17.00-17.30	Coffee break 4 & networking
17.30-18.00	***Metabolomics and CDG.*** Antonio Pineda-Lucena, Structural Biochemistry Laboratory, Centro de Investigación Príncipe Felipe, Valencia, Spain
18.20-18.35	***Glycomine: unravelling a new therapeutic approach for PMM2-CDG.*** Agnes Rafalko, Glycomine, USA
18.35-19.00	***Successful dietary therapy in three CDG types and outlook for future therapies.*** Eva Morava, Professor in pediatrics, Tulane University Medical School, USA
19.00-19.20	***A foster plan for the orphans.*** David Cassiman (Dept of Hepatology and Metabolic Center University of Leuven, Belgium)

19.20-19.30	***Closing remarks*** Jaak Jaeken, Centre for Metabolic Diseases, University Hospital Gasthuisberg Leuven, Belgium
19.30-20.00	***Dancing at the hands of Morgan Webb Liddle***. Morgan Liddle and her followers (Grade 1A Para-Equestrian, Australia)
20.00-22.30	Networking and sharing dinner 2

KINDERGARTEN ACTIVITIES

29 August 2015, Saturday

08.00-08.30	**Kindergarten receives your child/adult**
08.30-09.00	**Welcome and gather together** **Kindergarten volunteer leader:** Zuzana Pakanova, Institute of Chemistry, Department of Glycobiology, Central Analytical Laboratory, Slovak Academy of Sciences, Slovakia
09.00-10.50	***Dancing at the hands of Morgan Webb Liddle***. Part I Morgan Liddle and her followers (Grade 1A Para-Equestrian, Australia)
10.50-11.00	Parents bring children and adults to coffee break 1
11.00-11.30	Coffee break 1
11.30-12.45	***Dancing at the hands of Morgan Webb Liddle***. Part II Morgan Liddle and her followers (Grade 1A Para-Equestrian, Australia)
12.55-14.45	Parents bring children and adults to Networking and sharing lunch 1
14.40-14.55	**Kindergarten receives your child/adult**
14.55-16.00	Painting faces! **Kindergarten volunteer leader:** Zuzana Pakanova, Institute of Chemistry, Department of Glycobiology, Central Analytical Laboratory, Slovak Academy of Sciences, Slovakia
16.00-17.00	Activity will be announced soon
17.00-17.15	Parents bring children and adults to coffee break 2
17.15-17.45	Coffee break 2
17.35-17.45	**Kindergarten receives your child/adult**
17.45-18.45	Film projection

18.50-19.00	Parents bring children and adults to amphitheater
20.00-22.30	Networking and sharing dinner 1

30 August 2015, Sunday

08.00-08.30	**Kindergarten receives your child/adult**
08.30-09.00	**Welcome and gather together** **Kindergarten volunteer leader:** Zuzana Pakanova, Institute of Chemistry, Department of Glycobiology, Central Analytical Laboratory, Slovak Academy of Sciences, Slovakia
09.00-10.50	Activity to be determined
11.00-11.10	Parents bring children and adults to coffee break 3
11.10-11.40	Coffee break 3
11.30-12.45	***Dancing at the hands of Morgan Webb Liddle***. Last instructions and meeting Morgan Liddle and her followers (Grade 1A Para-Equestrian, Australia)
13.10-15.15	Parents bring children and adults to Networking and sharing Lunch 2
15.15-15.30	**Kindergarten receives your child/adult**
15.45-16.45	Clown show
16.45-17.00	Parents bring children and adults to coffee break 4
17.00-17.20	Coffee break 4
17.20-17.30	**Kindergarten receives your child/adult**
17.45-18.45	General activity
19.15-19.30	Parents bring children and adults to amphitheater ***Dancing at the hands of Morgan Webb Liddle presentation*** Morgan Liddle and her followers (Grade 1A Para-Equestrian, Australia)
20.00-22.30	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=PLa3xsZodHVEDRyAXVPP4imaqKKwrW79Id

Images used to perform this document:
Sources and acknowledgements:
http://fr.wikipedia.org/wiki/Lyon
http://www.lyon-france.com/html/myonlylyon/