CONGENITAL DISORDERS OF GLYCOSYLATION WORLD CONFERENCE

The power of advancing patient-oriented research united

FAMILIES AND PROFESSIONALS

3RD WORLD CONFERENCE ON CDG FOR FAMILIES AND PROFESSIONALS

"UNITED SHAPING THE FUTURE FOR CDG"

15-16TH July 2017, Leuven – Belgium

Speakers' Biographies

CDG & Allies – PPAIN

CDG & Allies - Professionals and Patient Associations International Network

PORTUGUESE ASSOCIATION CDG

SPEAKER BIOGRAPHIES (it follows the order from the agenda)

1.1. CDG history and clinical presentation

Prof 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 degrees.
In 1980 Dr. Jaeken was the very first doctor to publish the diagnosis of CDG. He was involved in the description of many other diseases. He is a 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 of metabolic diseases named CDG and to test new methods of diagnosis and therapies.
Indeed, Dr. Jaeken has been an active collaborator of CDG & Allies – Professionals and Patient Associations International Network (CDG & Allies – PPAIN,for more information visit HERE). Dr. Jaeken is a member of the Advisory Committee, and gives his constant and unvaluable scientific guidance and support.

1.2. Biochemical aspects

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.

1.3. Diagnostics Tools

Dr Christian Thiel

Dr. Thiel is working in the field of glycosylation deficiencies since 1999 and identified several new CDG defects. In 2002 he received his Ph.D. in Goettingen for the identification of the ALG2-CDG index patient. After completion of his degree he was appointed as a Faculty Fellow in the Department of Biochemistry II at the University of Goettingen till 2004. Since 2004 Dr. Thiel is working at the Children´s Hospital in Heidelberg as person in charge for the routine and advanced CDG diagnostics. His interests are focused on the identification of known and new CDG types. Besides, his laboratory is currently working on understanding pathophysiological mechanisms in CDG by using cellular as well as animal models (mouse, frog and fish). He aims to contribute for CDG lives by testing therapeutic approaches in the different model systems. Dr. Thiel has a long-standing and close link to the German family association `GlycoKids e.V.´ and serves as a consultant for glycobiochemical questions.

1.4. Gastrointestinal Problems

Dr Stephanie Grünewald

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

1.5. CDG Neurology:What you need to know

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).

1.6. The role of the cerebellum in cognition and behavior:beyond coordination in the Central Nervous System

Dr Marc 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.

1.7. CDG Adults: main symptoms and management

Dr David Cassiman

Dr. Cassiman received his Ph.D. in 2001 awarded by the Katholieke Universiteit (KU) Leuven. Prior to that he had completed his MD degree in 1996 at KU Leuven. Since the completion of his doctorate he has had a highly diversified career.

Last year, he was assigned to the post of Full Professor at KU Leuven. Furthermore, since 2012 he also teaches at University of Hasselt. Moreover, at KU Leuven Dr Cassiman is head of the Hepatology Unit. Alongside his teaching career, Dr Cassiman continues to be a Medical Doctor at UZ Leuven, with focus on liver diseases and metabolic diseases.

As far as his career as a Researcher is concerned, Dr Cassiman has been a half-time fundamental clinical researcher at Research Foundation Flanders (FWO) Vlaanderen for ten years now. His research interests include metabolic liver diseases (haemochromatosis), Wilson's disease, rare inborn errors of metabolism, mitochondrial dysfunction, orphan diseases, orphan drugs and orphan drug regulations. Consequently, he has an extensive and remarkable list of publications in several relevant scientific magazines. He hopes his knowledge and work will ultimately make a difference in the lives of CDG patients.
He is currently Working Group leader of CDG & Liver. To know more about the ongoing projects of this group visit HERE.

1.8. Genetic aspects of CDG

Dr Gert Matthijs

Gert MATTHIJS is head of the Laboratory for Molecular Diagnostics at the Center for Human Genetics in Leuven and Professor at the Katholieke Universiteit Leuven, Belgium. He received his Ph.D. in 1989 from this university and has been involved in the diagnostics of inherited diseases since 1994. His major research interest is in Congenital Disorders of Glycosylation (CDG). He was the coordinator of 2 major European projects: EUROGLYCANET and EUROGENTEST2. The former focusing on the identification of novel defects and the generation of models for CDG, and the latter focusing on the development and standardization of genetic testing.Currently, he is the coordinator of EURO-CDG-2, a European research network dedicated to improving diagnosis and treatment of inborn errors of glycosylation. He was a Board member and chaired the Patenting and Licensing Committee of the European Society of Human Genetics (ESHG), and played an important role in the European opposition

against the BRCA patents. He is a member of the Scientific Diagnostic Committee of IRDiRC and an Alternate member of the Commission Expert Group on Rare Diseases.
At the national level, he has been a thriving force for a revision of the reimbursement system for genetic tests. He has coordinated a workgroup on legal, ethical and societal aspects of total genome analysis (Metaforum). Additionnaly, he organized an exposition on cartoons and genetics in the summer of 2014, and published a book about genetics for the public ('Iedereen GENiaal'), that was translated in English in 2016 ('the Human Recipe').

1.9. Treatment

Dr. Eva Morava

Dr. Morava graduated as a Medical Doctor by the University of Pecs, Hungary. She specialized in pediatrics in 1994. She specialized in human genetics in 1999.

Dr. Morava defended her PhD thesis on Molecular cytogenetic investigations in mental retardation syndromes in 2000, and worked as a clinical geneticist until 2002. Since 2012 she has beenfull professor at the Tulane University Medical Center, at the Hayward Genetics Center, as a biochemical geneticist. Since 2015 she is also faculty at the UZLeuven, in Belgium. Eva Morava is a member of national and international committees and scientific advice groups.

Her list of publications includes more than 200 peer reviewed scientific papers. Her special research interests are 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 established the Nijmegen Center for CDG (www.nijmegencdg.nl). She is the chief editor of the Journal of Inherited Metabolic Disorders.

Dr. Morava is a collaborator of the international network CDG & Allies – PPAIN (for more information visit HERE). Dr.Morava shares her expertise an knowledge as a member of the Advisory Committee and of the Working Group CDG & Glycoimmunology. To know more about the ongoing projects of this group visit HERE.

1.10. Physical Therapy

Dr. Chantal Verhille

Dr Chantal Verhille concluded her MSc's degree in physiotherapy motor revalidation in 1986. Following the completion of her MA she did several specialization courses, namely in pediatrics and Bobath. In addition to these specializations, Dr. Verhille has also gained experience in various disciplines, such as child neurology, mentally and physical retarded children, muco, neuromuscular diseases, oncologic, psychosomatic, gastro-entero, nefrologic, and general physio treatments for children. With over 30 years of experience in phsysiotherapy, mainly focused on pediatric revalidation, for the last 15 years Dr Verhille has been predominately dedicated to metabolic diseases, like osteogenis imperfecta and CDG. Currently and since 1992, she has been working at UZ Gasthuisberg, Department Pediatry, Leuven, Belgium. Dr Verhille hopes to keep contributing to the motor development and improvement of CDG patients.

1.11. Psychomotor Therapy and Body Experience

Dr. Dolores Viegas Gamito

Dr Dolores Viegas Gamito received her BA Special Education and Rehabilitation from the Faculty of Human Motricity, University of Lisboa. After the completion of her degree, she did a specialization course in psichomoticity. She also did other training courses, namely on topics such as parental education and psycho-pedagogic and artistic inter-relation. With an active professional career since 1998, Dr Viegas Gamito has occupied several positions in different institutions and organizations, although she has always remained as a psychomotricist at Centre Dr. João dos Santos – Casa da Praia. She has participated in very diverse projects, worked with children with special educational needs and accompanied them in various contexts, from school to home environment. Dr Viegas Gamito has implemented several ludic-therapeutic strategies, intervened at the level of prevention and health promotion and was also a teacher of adapted swimming and of adaptation to the aquatic environment. Also, Dr Viegas Gamito has experience as supervisor and as team coordinator. Having a very rich and diversified professional career, Dr Viegas Gamito aims to apply her knowledge and expertise to improve the lives of CDG patients.

1.12. Educational digital resources for CDG

Joana Peixinho

Joana Peixinho has a BA in Basic Education, a Post Grad in Special Educational Needs: Both cognitive and motor, a MSc's degree in Education Sciences –specializing in evalutions, and is currently doing her PhD in Multimedia in Education. Her current research interests are mainly related to developing digital tools, namely school books, for educational purposes.

She has occupied various teaching positions since 2006 ,when she concluded her BA degree, having great experience in communication. Dr. Peixinho has had her worked published on both national and international journals and she has also presented her studies in several conferences and meetings.Joana Peixinho has been involved with CDG,as a Patient mother and advocate and she is committed to improving the lives of all CDG patients, particularly focusing on educational needs and challenges.

1.13. Overall strategies to improve communication in CDG

Merell Liddle

Merell Liddle has a BA in Politics and Economics and a Post Grad Dip in Securities Markets. She spent 15 years working in the areas of merchant banking, money market and takeovers regulation. She then completed a degree in Psychology and a Post Grad Dip in Health Psychology whilst having a family that included a child with complex disabilities who was later diagnosed as having CDG. Ms Liddle has a particular interest in speech and communication disabilities and has been active in the area of Augmentative and Alternative Communication. She has been extensively involved in CDG patient advocacy,support, and education and is committed to collaborative research to develop safe, effective therapies and treatments for CDG.

Currently, Merell is a member of two CDG & Allies – PPAIN (for more information visit HERE) Working Groups: CDG & Health Psychology (more information HERE) and CDG Patient Reported Outcomes (PROMs) (more information HERE).

1.14. Tips for home adaptation and accessibility

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.

Sandra Pereira Pinto is a member of the Advisory Committee at the CDG & Allies – PPAIN (for more information visit HERE).

1.15. Emotional Impact and Coping Strategies in Families of Children with Congenital Disorders of Glycosylation (CDG)

Prof Luísa Barros

Prof. Luísa Barros received her Ph.D. in Clinical Psychology from the University of Lisbon, Portugal (1992). After completion of her degree she was appointed as a Faculty Fellow in the Department of Psychology at the University of Lisbon. She is currently a Full Professor of Clinical and Health Psychology at the University of Lisbon and coordinates the Research Group on Health and Clinical Psychology – Familial and Individual Adaptation Processes. Her interests are focused on clinical and pediatric psychology. In particular her laboratory is currently working on understanding how parental processes are related to and affect children's health and adaptation, both in healthy and chronically ill populations and developing measures of parenting processes and family adaptation.

She is currently Working Group leader of CDG & Health Psychology. To know more about the ongoing projects of this group visit HERE.

1.16. Patient Reported Outcomes for CDG

Prof Liz Forbat

Dr. Forbat received her Ph.D. in the psychology of care relationships from the Open University, UK, in 2001. After completion of her doctorate she worked as a post-doc at the Open University for two years and then the University of Edinburgh for three years. Dr. Forbat then served as senior research fellow at the University of Stirling for four years, before being appointed as co-director of the Cancer Care Research Centre at Stirling.
Dr. Forbat joined the Faculty of Health Sciences at Australian Catholic University in 2015, as professor of palliative care. Her interests as a research psychologist and family psychotherapist are focused on: (i) care relationships, (ii) care pathways and (iii) quality clinical care. In particular her team are currently working on understanding the outcomes of family meetings in palliative care, conflict between clinicians and families, and avoidable presentations to emergency departments.

Dr. Forbat currently serves on the Editorial Board of the British Journal of Learning Disabilities and The Journal of Family Therapy. She is a member of the Australian Capital Territory palliative care clinical network.
She is currently Working Group leader of CDG Patient Reported Outcomes (PROMs). To know more about the ongoing projects of this group visit HERE.

1.17. Advocacy tool kit to integrate rare diseases into social services and policies

Dr Raquel Castro

Raquel holds a Bachelor of Arts in Communication as well as a post graduate degree in Project Management at the Lisbon School of Economics and Management. A Portuguese national, Raquel is also fluent in English, French and Spanish. With over ten years of experience working in not-for-profit organisations, Raquel has previously been involved in humanitarian and development projects in Europe and in Asia.

Raquel coordinated the Portuguese Help Line for rare diseases (Linha Rara), obtaining a broad experience in dealing with rare disease patients' and families' needs. In 2012, Raquel Castro joined EURORDIS. Based in Paris, she is part of the Operations Team and is responsible for managing EURORDIS' activities related to the integration of rare diseases into social services and policies.

Raquel is currently in charge of INNOVCare project (for more information visit HERE), focused on promoting integrated care and on improving social services for people living with

a rare disease. She is also involved in the RD-Action, the new Joint Action for rare diseases promoted by the European Union and Member States.

1.18. Natural History of CDG, and envisioning the future

Lynne Wolfe

Lynne has been a Nurse for over 30 years and a Nurse Practitioner working with children and adults who have all types of Inborn Errors of Metabolism and Mitochondrial diseases for 25 years. Lynne is a Senior Nurse Practitioner, Associate-Investigator, and the Undiagnosed Diseases Network NIH-UDP Site Coordinator. She is also an Associate-Investigator and Study Coordinator for the Epi-743 Investigational Drug trial and the Congenital Disorders of Glycosylation Natural History study at National Human Genome Research Institute. Her areas of research include: Congenital Disorders of Glycosylation, Mitochondrial disease/dysfunction, treatment of rare diseases, nutrition and supplement support for metabolic and mitochondrial diseases, and transitional care, all areas she has also published in. She is speaks frequently to professional and family support groups around the world.

Prof 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. 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).

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. 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. He is the Vice President Elect for Science Policy of FASEB, a US-based organization representing over 100,000 medical research scientists.