



Thursday January 13

- 08.00 Opening registration desk
- 08.30 - 09.00 **Opening of the meeting - welcome**
(Jaak JAEKEN, Leuven, Belgium; Gert MATTHIJS, Leuven, Belgium)
- 09.00 - 09.30 **Jaak JAEKEN (University Hospital, Dept Paediatrics, Metabolic Diseases, Leuven, Belgium):
What's new in CDG ?**
- 09.30 - 13.00 CDG: new diseases, novel models**
Chair: Jaak JAEKEN, Nathalie SETA
- 09.30 - 10.10 **Andrea SUPERTI-FURGA (Centre Hospitalier Universitaire Vaudois, Lausanne, Switzerland):
Bone, connective tissue and glycosylation**
- 10.10 - 11.10 Short presentations:
1. Christian THIEL (Center for Child and Adolescent Medicine, Center for Metabolic Diseases, Heidelberg, Germany): Congenital Disorder of Glycosylation-Ip (ALG11-CDG): Molecular defect and identification of new patients.
 2. François FOULQUIER (Université des Sciences et Technologies de Lille, CNRS, Lab de Glycobiologie Structurale et Fonctionnelle, Lille, France): TPARL- towards understanding of its physiological role in pH homeostasis.
 3. Hudson FREEZE (The Burnham Institute, La Jolla, U.S.A.): CDG collaborations in The United States
- 11.10 - 11.30 **Coffee break**
- 11.30 - 12.15 Selected presentations:
1. Eva MORAVA (Radboud University Medical Centre, Department of Pediatrics, Nijmegen, The Netherlands): A novel cerebello-ocular syndrome with abnormal Glycosylation due to abnormalities in dolichol metabolism.
 2. Thierry DUPRE (AP-HP, Laboratoire de Biochimie, Groupe Hospitalier Bichat-Claude Bernard, Paris, France) : News about dolichol deficiency and CDG
 3. Byron LAM (Bascom Palmer Eye Institute, Miami, U.S.A.): Retinitis pigmentosa (RP) caused by the K42E mutation in DHDDS (dehydrodolichol diphosphate synthase)
- 12.15 - 13.00 **David STEPHENS (University of Bristol, Dept of Biochemistry, Bristol, U.K.):
COPII-dependent trafficking - implications for morphogenesis and development**
- 13.00 - 15.00 **Lunch + Poster session**
- 15.00 - 17.00 Analysis of Glycosylation**
Chair: Dirk LEFEBER, Eva MORAVA
- 15.00 - 15.40 **David SMITH (Emory University School of Medicine, Dept of Biochemistry, Atlanta, Georgia, USA):
Glycomics and Glycan Microarrays**
- 15.40 - 17.00 Selected presentations:
1. Maily GUILLET (Radboud University Medical Centre, Nijmegen, The Netherlands):
A new B4GALT1-CDG patient identified by serum N-glycan profiling by mass spectrometry.
 2. Wendy HEYWOOD (Institute of Child Health, London, U.K.): Proteomic analysis reveals potential alternative marker proteins for the diagnosis of Congenital disorders of Glycosylation types I and II.
 3. Hana HANSIKOVA (First Faculty of Medicine, Charles University, Prague, Czech Republic):
Activities of lysosomal enzymes in patients with CDG syndrome.
 4. Stuart HASLAM (Imperial College, Molecular Biosciences, London, U.K.): G6PC3 mutations are associated with a major defect of Glycosylation: a novel mechanism for neutrophil dysfunction.
 5. Riet BAMMENS (K.U.Leuven, Center for Human Genetics, Leuven, Belgium):
A new inborn error of Glycosylation due to DPM2 deficiency.
- 19.30 - 22.30 **Cocktail/Dinner**
in the Faculty Club, Groot Begijnhof, Leuven



Friday January 14

09.00 - 12.30

CDG: clinical aspects and therapy

Chair: Gert MATTHIJS, Belen PEREZ-DUEÑAS

09.00 - 09.40

Luc VAN ROMPAEY (Galapagos nv, Mechelen, Belgium):

Chemical compound screening: From target to the clinic

09.40 - 10.40

Short presentations:

1. Vandana SHARMA (Sanford-Burnham Medical Research Institute, La Jolla, California, U.S.A.): Screening small molecules as therapy for Congenital Disorders of Glycosylation, CDG-Ia (PMM2-CDG)
2. Christian KÖRNER (Center for Child and Adolescent Medicine, Center for Metabolic Diseases, Heidelberg, Germany): Prenatal Mannose treatment prevents embryonic lethality in a mouse model for CDG-Ia (PMM2-CDG).
3. Martin WILD (Max Planck Institute for Molecular Biomedicine, Münster, Germany): A human defect in alpha2,3-sialylation causing a severe bleeding disorder

10.40 - 11.00

Coffee break

11.00 - 12.30

Selected presentations

1. Samira ACHOUITAR (Radboud University Medical Centre, Genetic and Metabolic Disease, Nijmegen, The Netherlands): Nijmegen paediatric CDG rating scale: a novel tool to document disease progression.
2. Agata FIUMARA (University of Catania, Dept of Pediatrics, Italy): Clinical and neuro-imaging findings in a dysmorphic patient with CDG-Ix.
3. Rafaël ARTUCH (Hospital Sant Joan de Déu, Clinical Biochemistry, Barcelona, Spain): Mild clinical and biochemical phenotypes in 2 patients with CDG-Ia (PMM2-CDG).
4. Celia PEREZ-CERDA (Universidad Autónoma de Madrid, Centro de Diagnóstico de Enfermedades Moleculares, Madrid, Spain): Screening for Congenital Disorders of Glycosylation in foetus and newborn samples with congenital malformations.
5. Miski MOHAMED (Radboud University Medical Centre, Institute Genetic and Metabolic Diseases, Nijmegen, The Netherlands): Neurologic involvement and diagnostic approach in Congenital Disorders of Glycosylation type II-x.
6. Jaimé BRUM (The SARAH Network of Rehabilitation Hospitals, Brazil): Congenital Disorders of Glycosylation (CDG) in Brazil

12.30 - 13.00

General assembly: Creation of a CDG Foundation

13.00 - 14.30

Lunch + Poster session

14.30 - 16.30

Cell biology, novel findings

Chair: Christian KÖRNER, François FOULQUIER

14.30 - 14.50

Weston STRUWE (University College Dublin, Conway Institute, NIBRT, Dublin, Ireland)

Modeling a congenital disorder of glycosylation type I in *C. elegans*: a genome-wide RNAi screen for N-glycosylation-dependent loci

14.50 - 15.15

Richard STEET (Carbohydrate Research Center, Athens, Georgia, U.S.A.): Modelling of CDG in Zebrafish.

15.15 - 16.15

Selected presentations

1. Sabrina ROSA (Mount Sinai School of Medicine, New York, U.S.A.): Zebrafish as a model of Congenital Disorders of Glycosylation, CDG-Ib (MPI-CDG).
2. Vladimir LUPASHIN (University of Arkansas for Medical Sciences, Little Rock, U.S.A.): The COG complex functions in trafficking of glycosyltransferases through the Golgi.
3. Florence HABAROU (AP-HP, Groupe d'étude des glycopathies, Hôpital Bichat, Paris, France): Studies on involvement of microtubules and kinesin 1 in disturbed Golgi retrograde trafficking coupled to glycosylation abnormalities
4. Dirk LEFEBER (Radboud University, Medical Centre, Neurology, Nijmegen, The Netherlands): Gene identification by Whole-Exome-Sequencing in Congenital Disorders of Glycosylation.

16.15 - 17.00

Final discussion and farewell drink