SSIEM Official Satellite Symposia
“The 2nd World Conference on Congenital Disorders of Glycosylation (WCCDG) for Families and Professionals: a challenging story of sugar trees”, 28 to 30 August 2015, in Lyon (France)

This conference is part of the Educational Program of Excellence on CDG created by the Portuguese Association for CDG (APCDG, www.apcdg.com).

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

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This and other resources available at:
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“CDG diagnosis: strengths, weaknesses, and the road forward”

Monique van Scherpenzeel, PhD
Postdoc scientist Glycomics
Radboud University Medical Center
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“Translational Research”
from fundamental biochemical mechanisms into the clinics and back
“Increase number of identified subtypes”

<table>
<thead>
<tr>
<th>CDG-I</th>
<th>CDG-II</th>
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<td>2005: ~60% solved</td>
<td>2005: 15% solved</td>
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<tr>
<td>2015: &gt;95% solved (120 patients)</td>
<td>2015: 75% solved (85 Patients)</td>
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“CDG type I subtype identification”

Whole exome sequencing (WES)

CDG-I

WES

Clinical symptoms

QTOF

CDG-I bioinformatics filter

Sanger sequencing

~65% solved within 6 months

PGM1-CDG/ALG1-CDG

Timal S et al. *Hum Mol Genet* 2012
“Do CMS symptoms occur in other CDG-I defects?”

Our protocol:
- Genetically solved CDG-I patients are seen by our neuromuscular doctor
- On indication: EMG, single fiber EMG, etc.
- If CMS symptoms are present: medication trial with Mestinon or 3,4-DAP

2 PMM2-CDG patients seem to benefit from CMS medication

Dr. Corrie Erasmus

If you observe such complaints, discuss with your local CDG clinical expert and contact us so that we can share our protocol and experience
“Use of a balance for glycans in CDG”

More accurate detection of your weight

Highly accurate detection of glycan weights
Disease monitoring

Mannose therapy

After liver transplantation

“Successful liver transplantation in CDG-Ib”

Janssen MC et al. *Pediatrics* 2014
“CDG type II subtype identification”

23 patients PGM1-CDG

5 patients SLC35A2-CDG

12 patients Man1B1-CDG

Largely improved through advances in technology

Patient diagnosed!
“Application to PGM1-CDG”

CDG screening: type II profile

- First combined CDG type I & II
- Lack of galactose

Loss of glycans: CDG-I

Truncated glycans: CDG-II

Truncated glycans: CDG-II
“Improved glycosylation on galactose therapy”

"Understanding mechanisms of disease"
Through dynamic analysis of the glycan building blocks

Protein glycosylation

Galactose + Glucose → Energy

Ultimate goal: (optimized) Therapy
- Mechanism based: treatment with (sugar) supplements
- Symptom based: treatment of individual disease symptoms
“Dynamic analysis of sugar metabolism”

- Ion pair- Liquid chromatography & Mass spectrometry
- Stable isotope tracing

CMP-sialic acid
UDP-galactose
UDP-glucose
UDP-GalNAc
UDP-GlcNAc
GDP-mannose
UDP-GlcA
GDP-fucose
ADP-glucose
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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” at:

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](http://www.thefog.ca/main.html)

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