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.

©Foundation of Glycosylation and Portuguese Association for CDG and related rare metabolic diseases (APCDG-DMR, Portugal). 2015. All rights reserved.
This and other resources available at: www.apcdg.com
Portuguese Association for CDG (APCDG): addressing the community needs

Presentation by

a Graphic Designer Diogo sampaio
http://www.diogosampaio.pt/

a Sibling Vanessa Ferreira, PhD, MBA
APCDG President and Founder
Diagnosis

Princess Lili (6 years old), cerebral palsy diagnosis
First reactions
To be diagnosed with PMM2-CDG

- Misdiagnosis
- Chronic
- Life-threatening
- Disabling
- Understanding
- Broad diversity of symptoms to manage
- No cure
- Few interest from industry
- Government funding
- Specialized treatment and care
- Financial Challenges
- Find other families
- Explain to our family
- Awareness
Immediate needs

- Information
- Community building and outreach
- Advocacy
- Research

10 March 2010
Our single, urgent goal:
Advancing progress of medical science to ensure the development of expeditious therapies for CDG in our lifetime.

“To boldly go where no man has gone before”, Star trek
First decisions
Networking, Networking, Networking

- Professionals
- CDG and related Patient Advocacy Groups
- Societies of knowledge
Outcomes from Networking

• Patient representatives at International level
  ✓ Attendance 17 conferences
  ✓ Active participation 20 conferences

• Conference organisers
  ✓ 5 CDG-specific conferences
  ✓ 5 Society

• Involvement in projects as volunteers
  ✓ Resources
  ✓ Guia Metabólica
  ✓ Co-researchers
  ✓ Reviewers
  ✓ Advisors
  ✓ Driving force

• Starting our own ideas, projects

Visibility
Trust
Reliability

100 volunteers contributed to our projects in an altruistic manner !!!!
Facing a Host of Unique Challenges

Lack Specialised care centres
Educational Roadmap, delay diagnosis
Integration in Society, lack of economic support, accessibility and symptomatic treatment

Adult phase

Treatment

Professional integration, psychologic support, genetic counseling and future

Treatment and sibling reaction

Isolated, Lack of information

Social integration

Lack Specialised care centres, support therapies
1-2% of our genes is involved in glycosylation. Many CDG still to be discovered!
Addressing the current needs of CDG community
Addressing the current needs of CDG community:
For you always: One CD=One live CDG

Guia Metabolica = 1200 €
Liliana Summer Research Scholarship 2015 = 2365 €
Glycoimmunology project = 300 €/patient sample analysis
Think tank CDG 2015 = Dr. Cláudia de Freitas
Graphic designer = TBD

9000 €
THINK VISUAL

10% People remember of what they hear

20% People remember of what they read

80% People remember what they see & do
The correct sugar transfer to proteins or lipids is essential for their biological function.

This disruption leads to:
- A great variety of more or less severe symptoms;
- Involvement of nearly every organ.
PATIENT FRIENDLY

POSSIBLE INTERVENTION
BY A PHYSICIAN

NASOGASTRIC TUBE OR GASTROSTOMY TUBE FEEDING. IN CASE OF CHRONIC DIARRHOEA, LACTOSE-FREE OR ELEMENTARY DIETARY FORMULA INSTEAD OF MILK PRODUCTS

GLASSES, PATCHING, BOTULINUM TOXIN OR SURGERY

IS IT A STROKE-LIKE EPISODE, EPILEPSY OR A VASCULAR EVENT?

CONSULTATION WITH A SPEECH THERAPIST/GASTROENTEROLOGIST/NUTRITIONIST

SURGICAL TREATMENT IN SEVERE FORMS

CORTICOSTEROIDS AND SALICYLIC ACID, PERICARDIAL DRAINAGE

L-TYROIDINE SUPPLEMENTATION

CLINICAL FEATURE

FAILURE TO THRIVE (+/- ENTEROPATHY, HYPOGLYCAEMIA)

STRABISMUS

ACUTE MOTOR EVENT AND/OR LOSS OF CONSCIENCE

ORAL MOTOR DYSFUNCTION

SCOLIOSIS/KYPHOSIS

PERICARDIAL EFFUSION

TRUE HYPOTHYROIDISM
THE POWER OF ADVANCING PATIENT-ORIENTED RESEARCH UNITED

FAMILIES AND PROFESSIONALS

01 CDG IMPACT ON FAMILIES
02 RESEARCH
03 GOAL: CURE CDG
04 UNITED TO OVERCOME CHALLENGES
05 WHY YOU SHOULD ATTEND WORLD CONFERENCE ON CDG (WCDDG)
06 OUTCOMES FROM THE CONFERENCE?
07 HOPE FOR PATIENTS AND THEIR FAMILIES

CDG

WORLD CONFERENCE CONGENITAL DISORDERS OF GLYCOSYLATION

www.apcdg.com
Congenital Disorders of Glycosylation (CDG)

CDG should be considered in every patient with an unexplained syndrome.

CDG is a group of disorders caused by the absence or abnormality of enzymes that modify sugars on proteins or lipids.

1. CDG is a rare disease, with only a few cases occurring in each family.

2. CDG is caused by the absence or abnormality of enzymes that modify sugars on proteins or lipids.

3. NMO-CDG (neurological) is the most common form of CDG, with a prevalence of 1:20,000.

4. CDG can affect many organs, including the brain, liver, heart, and kidneys.

5. Symptoms can range from mild to severe, and may include developmental delay, seizures, and organ damage.

6. CDG is a lifelong condition, and patients will require ongoing medical care.

7. International collaboration between affected families, researchers, and healthcare professionals is essential to improve understanding and treatment options.

www.apcdg.com
- Seizures
- Stroke-like episodes
- Cerebellar hypoplasia
- Dystonia
- Ataxia

- Abnormal subcutaneous fat pattern
- Absence of pubertal development
- Inverted nipples

- Scoliosis

- Strabismus
- Nystagmus

- Pericardial effusion

- Abnormal hematological and endocrine parameters
  - Low serum concentration of factors XI, antithrombin III, protein C and/or proteins
  - Low total serum thyroxine with mostly normal free serum thyroxine

- Hepatomegaly

- Abnormal biochemical parameters
  - Increased serum transaminases
  - Low serum cholesterol

- Osteopenia

- Failure to thrive
MANY PEOPLE INVOLVED

Jaak Jaeken | Paz Briones | David Cassiman
Eva Morava | Paula Videira | Mercedes Serrano | Donna Krasnewich | Belén P Dueñas | Maria Antonia Vilaseca
Etienne Barbier | Vanessa Ferreira | Michaela Špeciánová
Barbara Vulso | Maria Monticelli | Anna Lund and Anne Kristin
Rana Atwi | Fiona Wadell | Sibu Mundiyanaipurath | Malina Stancheva | Nathalie Seta | Hana Hansikova | Rita Barone
Ammi Grahn | Miski Mohamed
Lessons learned

You’re Not Superwoman...

And That’s Okay!
Lessons learned

- CDG specificities
- Multasking requirements
- Finding the money
- Strategic planning/setting priorities
- Technology
- Board-related issues
- Professionalization
- Members
- Competition
- Staffing/Few volunteers
  - Fundraising
  - Media and public relations
  - Administration
“It is only with the heart that one can see rightly; what is essential is invisible to the eye.”

― Antoine de Saint-Exupéry, The Little Prince

Believe, Challenge yourself, Audacity, tenacity, Dream big, Ask for help, Positive thinking

Advices to run non-profit representing CDG

Positive thinking
Change is possible. The more united we are, the stronger we are. Don’t give up.
The future
Clinical trial for PMM2-CDG

KEEP CALM + UNITED
AND
FOCUS ON
PHARMACOLOGICAL CHAPERONES

BEPOSSIBLE
### CDG clinical trials

#### Challenges
- Small sample size
- Lack of outcome assessment
- Ethics
- Research involving mostly children
- Funding

#### Our Solutions
- Information
- Community-building and outreach
- Advocacy
- Research
"You never change things by fighting existing reality. To change something, build a new model that makes the existing model obsolete." Buckminster Fuller

Families + Researchers + Physicians + Industry + Regulatory agencies

Partnership is the Key: "Coming together is a beginning; keeping together is progress; working together is success." ... Henry Ford
The future: Plan of action 2015-2017

• Information
  ✓ 1st World Educational program on CDG 2015-2017
  ✓ Website

• Community building and outreach
  ✓ 3rd World Conference on CDG 2017

• Advocacy
  ✓ World Congenital Disorders of Glycosylation (CDG) Day recognition
  ✓ National CDG Disease Awareness Day (Portugal), 29 January
  ✓ 1st Think Tank on CDG 2015
  ✓ Publications
  ✓ Fairy-tale
  ✓ Advancing CDG Research through Awareness Resources campaign

• Research
  ✓ CDG Professionals and Patient Associations Research Consortium (CDG-PPARC)
  ✓ Liliana Summer Research Scholarship 2015
  ✓ Fundraising One CDG One live CDG
CDG community: uniquely and differently united

“If you fight for your dream, one day the dream will fight for you, too!”
Where we stand today?

Typical Alentejo landscape, Montado, Évora, Portugal
ACKNOWLEDGMENTS

To my always Maria Antonia Vilaseca

Thanks

for transforming the small steps in humble achievements!
To encourage us to follow the good way with infinite enthusiasm and smile
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 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.

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)

©Foundation of Glycosylation [http://www.thefog.ca/main.html](http://www.thefog.ca/main.html) and Portuguese Association for CDG and related rare metabolic diseases (APCDG-DMR, [www.apcdg.com](http://www.apcdg.com), Portugal). 2015. All rights reserved.
ACKNOWLEDGMENTS

• All speakers
• All participants
• Associations
• Organizations
• Domaine Saint Joseph
• All Volunteers
• Pf Pascale de Lonlay and Nathalie Seta
• Dr Maria A Vilaseca and Pf Jaeken
• Biocommunicat