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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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 (WCCDG)
06 OUTCOMES FROM THE CONFERENCE?
07 HOPE FOR PATIENTS AND THEIR FAMILIES

WORLD CONFERENCE CONGENITAL DISORDERS OF GLYCOSYLATION

This and other resources available at:
www.apcdg.com
Skin manifestations in CDG

D. Rymen, MD
What is Skin all about?

- 3 layers with a myriad of functions

1. Epidermis: A Protective Barrier
What is Skin all about?

- 3 layers with a myriad of functions

2. Dermis: An Elastic Cushion
What is Skin all about?

- 3 layers with a myriad of functions

3. Hypodermis: An Insulating Layer
What is Skin all about?

- 3 layers with a myriad of functions...

... plus some interesting appendages:

1. Hair
2. Nails
3. Eccrine sweat glands
4. Apocrine or “odor” sweat glands
5. Sebum producing glands
What does skin do?

- Sensation
  - Skin feels...
    - The difference between pain, touch and pressure
    - The difference between warm and cold
What does skin do?

- Protection
  - Water resistance
  - Barrier to fluid loss
What does skin do?

- **Protection**
  - Water resistance
  - Barrier to fluid loss

1. Keratinocytes producing keratin *(bricks)*
2. Lipids *(cement)*
What does skin do?

- Absorption and excretion
What does skin do?

- Protection
  - Barrier against pathogens and mechanical damage

A Though Layer of Keratinocytes

0,1 mm
What does skin do?

- Protection
  - Barrier against pathogens and mechanical damage

Langerhans cell

A Local Defense System
What does skin do?

- Protection
  - Barrier against UV-radiation

Melanocyte

Little umbrellas of melanin protecting our genetic material
What does skin do?

- Protection
  - UV-radiation vs vitamin D synthesis
What does skin do?

- Protection
  - UV-radiation vs vitamin D synthesis
What does skin do?

- Heat regulation
  - When it’s too hot...
    - Hairs lay down
    - Sweating
    - Vasodilatation

Evaporation

Heat
What does skin do?

- **Heat regulation**
  - When it’s too cold...
    - Hairs straight up
    - Goosebumps
    - Vasoconstriction
Skin and CDG

Different topics

- Fat pads
- Orange peel skin
- Inverted nipples
- Ichthyosis
- Cutis laxa
- Tumoral calcinosis
- Abnormal pigmentation
- Adams-Oliver syndrome
- Hypohidrosis
The CDG Trio

- The most frequently cited abnormalities
  - Fat pads
  - Orange peel skin
  - Inverted nipples
Ichthyosis

- Fish scaled skin
Ichthyosis

- Fish scaled skin

A Defective Barrier
Ichthyosis

- Fish scaled skin

A Defective Barrier
Cutis Laxa – Wrinkly Skin

- A problem of the elastic fibers

ALG8 (CDG-Ih) – ATP6V0A2 (CDG-II) – COG5 (CDG-Ili) – COG7 (CDG-IIe) – MAN1B1 (CDG-II)
Tumoral Calcinosis

- Deposition of sodium-phosphate crystals in soft tissue

Pi (re)absorption ↑

Ectopic calcifications
Abnormal Pigmentation

Salt and pepper syndrome

Dowling Degos disease
Adams-Oliver Syndrome

Aplasia cutis congenita

Transverse limb defects
Hypohidrosis

± Insensitivity to pain

± palmoplantar hyperkeratosis

± Immunodeficiency

± teeth problems
Acknowledgments

Center for Human Genetics, Leuven, Belgium

Gert Matthijs, PhD
Jaak Jaeken, MD, PhD
Romain Péanne, PhD
Erika Souche, PhD
Valérie Race, PhD
Liesbeth Keldermans
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. 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

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