CDG Professionals and Patient Associations Research Consortium (CDG-PPARC) :
start small, think big
Rethink Translational Medicine meaning

Disease Mechanism

Therapeutic target

Benefit for patients

Drug development

Clinics Treatments

Patients / families
CDG Professionals and Patient Associations Research Consortium (CDG-PPARC):

- Encourage collaborations
- Increase knowledge
- Promote faster diagnosis
- Develop relevant basic research
- Boost collaborative clinical research
- Generate public resources
- Increase training opportunities

✓ improve the lives of affected patients
CDG in Portugal

CDG patients - 58

- PMM2: 53%
- CDG-X: 29%
- Unknown: 25%
- COG1: 2%
- COG4: 2%
- MAN1B1: 3%
- SRD5A3: 3%
- ATP6V0A2: 3%
- GALE: 2%
- ALG1: 2%
- RFT1: 2%
CDG in Portugal

Clinical diagnosis and follow-up

Clinical Metabolic Units
- 2 North (Porto)
- 1 Centre (Coimbra)
- 2 South (Lisboa)
- Others ( Açores, Madeira )

Metabolic Units
- Composed by professionals experienced in the diagnosis and management of CDG patients
- Following a multidisciplinary approach
- Organize educational pre and post graduate activities
CDG in Portugal

Biochemical/molecular diagnosis

National Laboratory at CGM Jacinto Magalhães: CH Porto

- Collaborative work with international partners

Basic research

- Immunology
- In vitro models
- Glycan analysis
CDG in Portugal

Collaboration with the Patient’s Association

- Organization of meetings
- Fund raising / join projects
- Awareness (panflets, articles)
CDG-PPARC

CDG Professionals and Patient Associations Research Consortium
**CDG-PPARC Project Plan - Work Packages**

**Work Package 1:** Paula Videira & Vanessa Ferreira
Overall management of project activities

**Work Package 2:**
Diagnosis.
Lead: Dulce Quelhas

**WP2:**
Partners: Vanessa Ferreira

**Work Package 3:**
Biology to guide therapy development.
Lead: Paula Videira

**WP3:**
Partners: all collaborators

**Work Package 4:**
Clinical research in CDG:
Lead: Esmeralda Martins
Co-lead: Esmeralda Rodrigues

**WP4:**
Partners: Luisa Diogo, Paula Garcia, Sívia Sequeira ...
...TBC

**Work Package 5:**
Educational, Dissemination and training material for healthcare professionals, for patients, families and general public
CDG Professionals and Patient Associations Research Consortium (CDG-PPARC)

**WP1: Management**
- development and execution of the project plan,
- day to day management,
- communication, coordination, preparation and delivery of progress reports.

**WP2: Diagnosis**
- Awareness campaign for professionals
  - Increase number of samples suspicious of CDG received at
- Centro de Genética Médica jacinto de Magalhães

**WP3: Biology**
- Project focused immunology
- Research models
- Other areas
  - Metabolomics
  - Cell biology
  - etc
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WP4: Clinical research
- Platform for knowledge sharing
- Increase publications from Portuguese experts (case reports, case series etc)
- Facilitate exchange of biological samples
- Identify priority areas for studies
- To facilitate the ability to develop common approaches and definitions to characterize CDG

WP5: Educational, Dissemination and training material
- Awareness campaign for professionals and families
- To compile resources easily applied to CDG
- To increase opportunities to partners to speak about CDG or consortium at high level conferences
- Public website targeting patient community, doctors, professors, researchers, educators, etc
CDG Professionals and Patient Associations Research Consortium (CDG-PPARC)

WP1:
✓ Collaborative research agreement
✓ Invitation Call to be partner
✓ press releases

WP2:
✓ List of Portuguese Societies of knowledge for further dissemination
✓ Infographic

WP3 & WP4:
✓ Collaboration

WP5:
✓ Practical guide targeted to CDG families
✓ Fairy-tale: "Glycoland and the coloured antennas" available in 11 languages
✓ CDG awareness and dissemination tool
✓ Brochures and leaflets
✓ Quarterly eNewsletter
✓ First World CDG Conference (Youtube channel)
✓ Identification & selection of articles and news of interest for overall community.
✓ Creation and/or participation in events targeted to the Rare Disease Day
How Defects in sialylated glycans alters immunity

Higher ability to PRIME T cells

Higher Expression of CYTOKINES

Affected capacity to UPTAKE antigens

Higher maturation

Less migration abilities

**Cabral MG, et al (2012)** The phagocytic capacity and immunological potency of human dendritic cells is improved by α2,6-sialic acid deficiency. *Immunology.*


**Crespo et al (2009)** Effect of sialic acid loss on dendritic cell maturation *Immunology* 128:e621-31

- Clinical, biochemical, imaging data, molecular characterization and CDG patients registration

- National / International collaboration in translation research projects to understand the pathogenesis of the disease

- Identify, collect and develop guides and resources of existing technical guidelines for the provision of care health CDG
WP 4 – Clinical research in CDG

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CDG-PPARC
WP 4 – Clinical research in CDG

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Partnership: Families, researchers & professionals

Patient’s Association & Families initiative

Fundraising

Liliana Summer Research Scholarships 2015

Resources for basic & clinical research

CDG awareness

Maria Monticelli
Conclusions:

The key ingredients to progress in combating CDG

- Consortium
- Information & resources sharing,
- collaboration,
- and community building among researchers, doctors, and patients are critical to CDG research