

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

start small, think big



PORTUGUESE ASSOCIATION

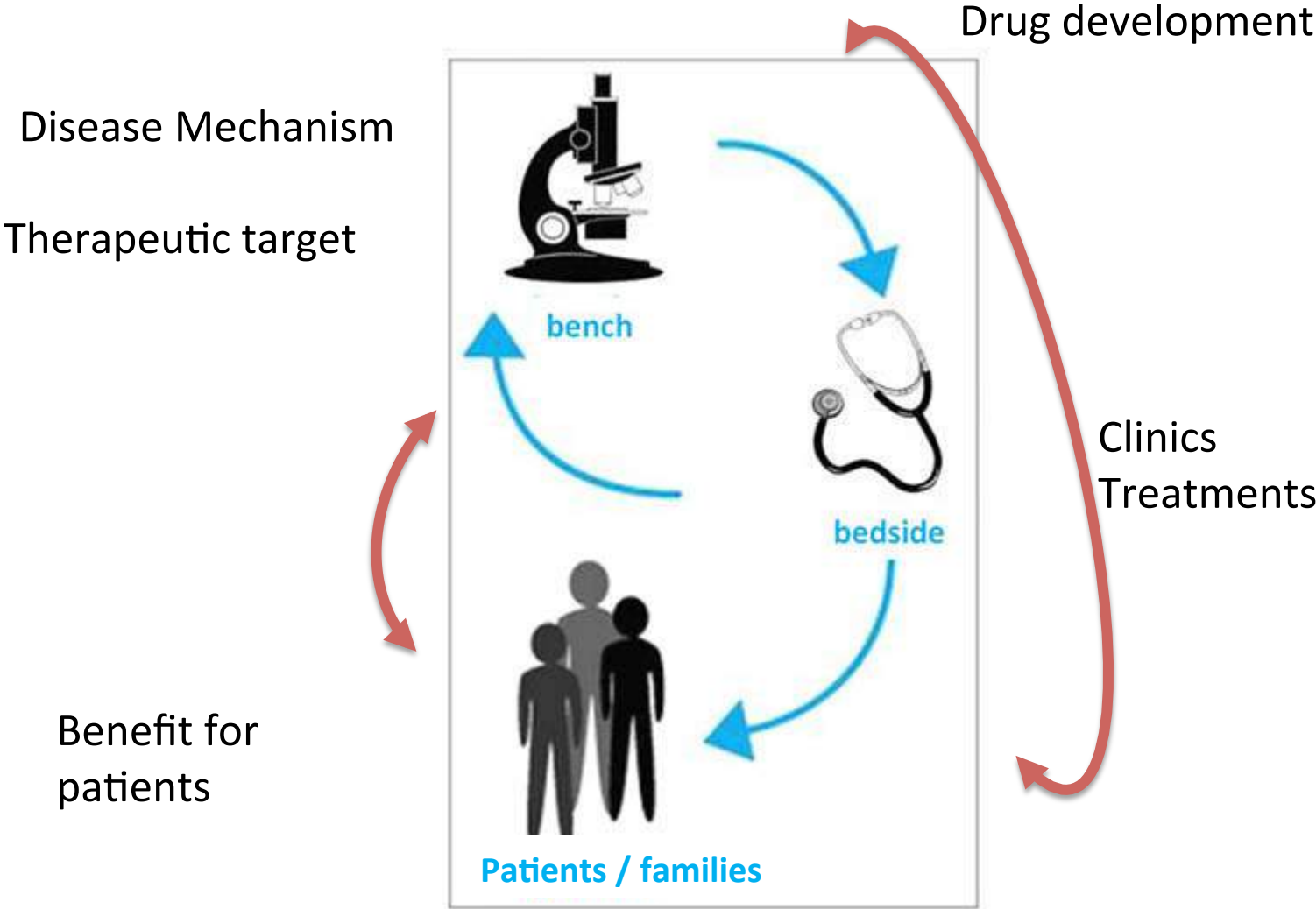


CONGENITAL DISORDERS OF GLYCOSYLATION WORLD CONFERENCE

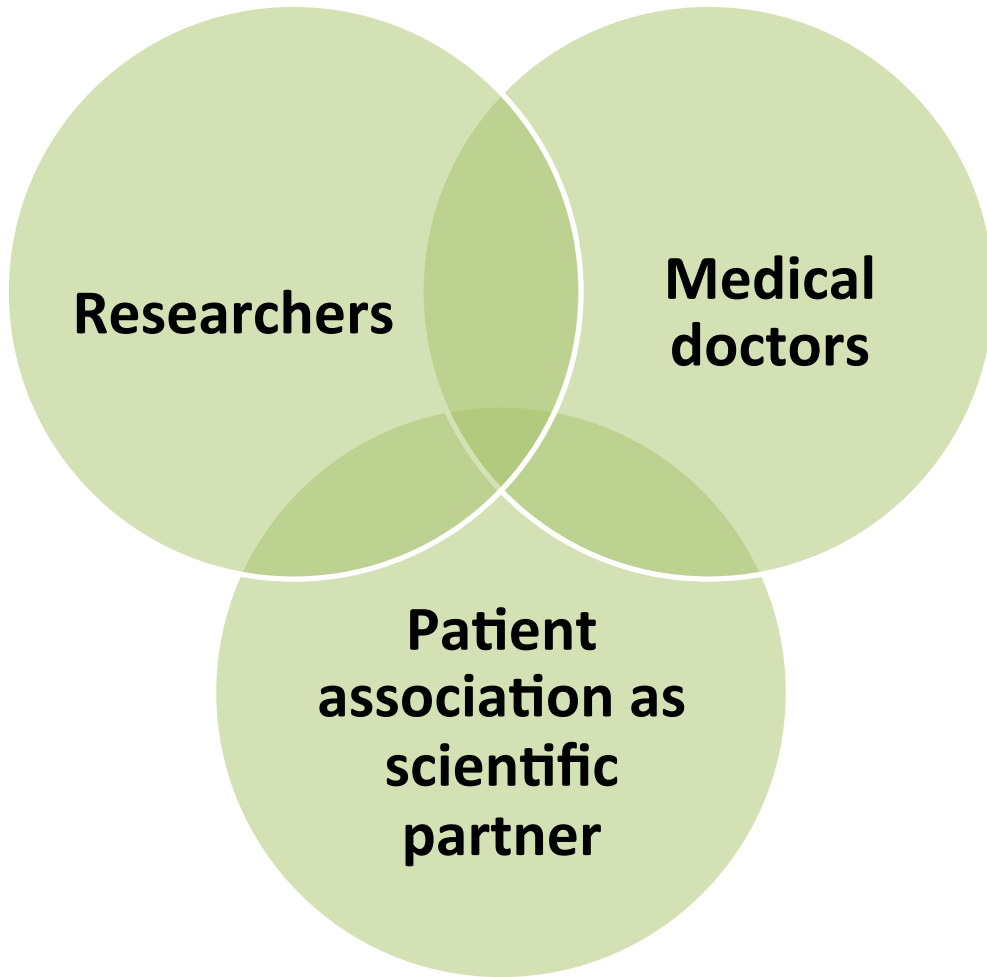
The power of advancing patient-oriented research united FAMILIES AND PROFESSIONALS



Rethink Translational Medicine meaning



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

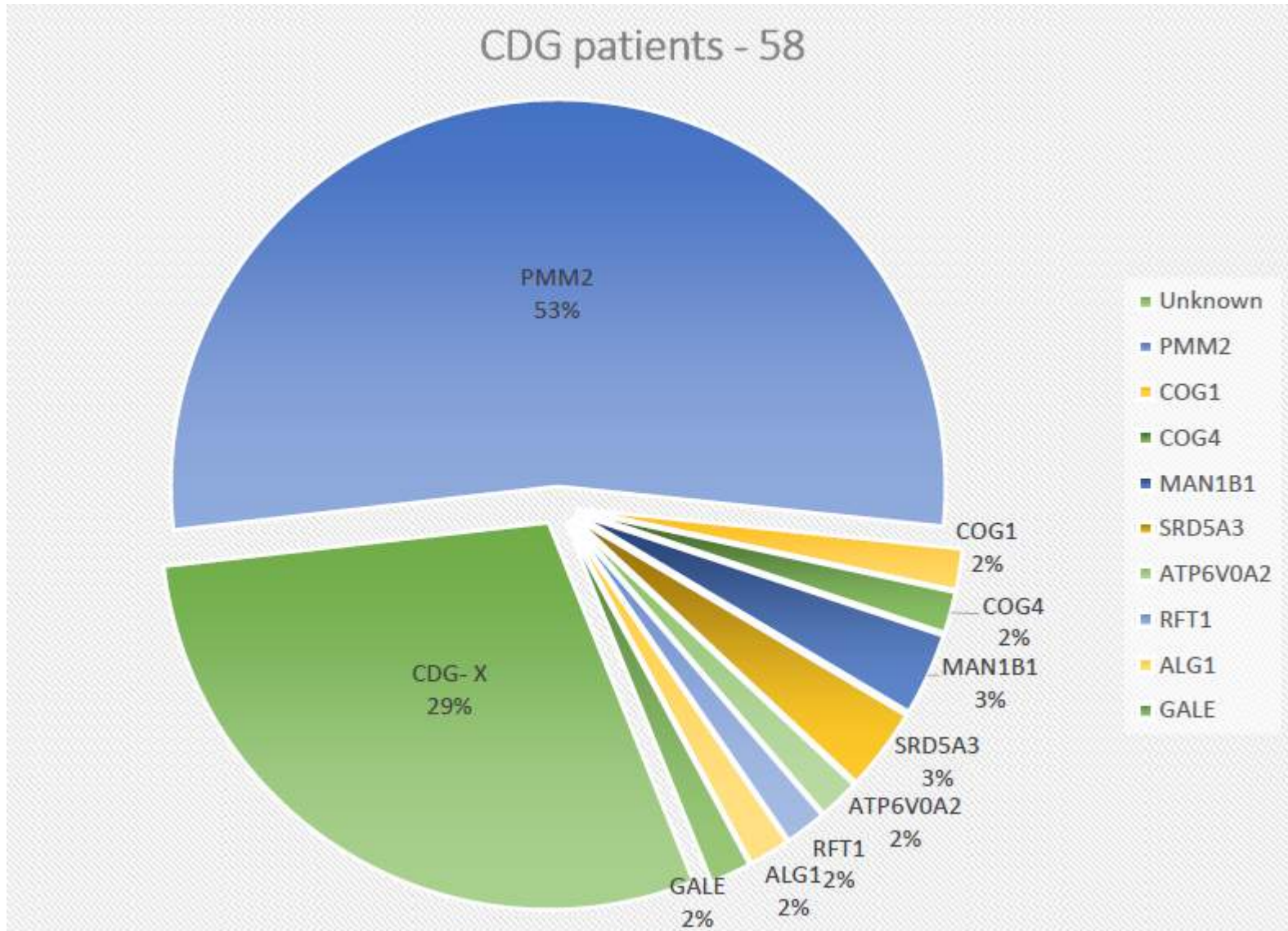


- ✓ Encourage collaborations
- ✓ Increase knowledge
- ✓ Promote faster diagnosis
- ✓ Develop relevant basic research
- ✓ Boost collaborative clinical research
- ✓ Generate public resources
- ✓ Increase training opportunities



✓ **improve the lives of affected patients**

CDG in Portugal



CDG in Portugal



Clinical diagnosis and follow-up

Clinical Metabolic Units

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

Metabolic Units

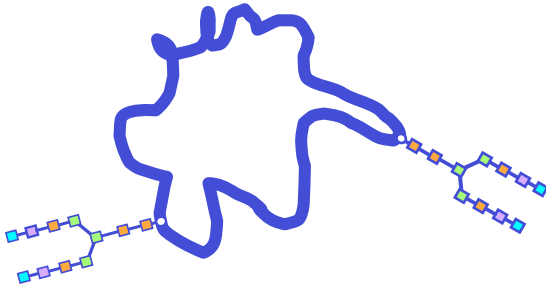
- Composed by professionals experienced in the **diagnosis** and **management** of CDG patients
- Following a **multidisciplinary** approach
- Organize **educational pre and post graduate activities**

CDG in Portugal



Biochemical/molecular diagnosis

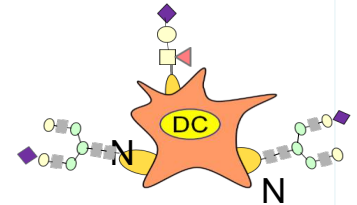
National Laboratory at CGM Jacinto Magalhães: CH Porto



- 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)



PORTUGUESE
ASSOCIATION

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

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

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

WP2:

Partners: Vanessa Ferreira

WP3:

Partners: all collaborators

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

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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

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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

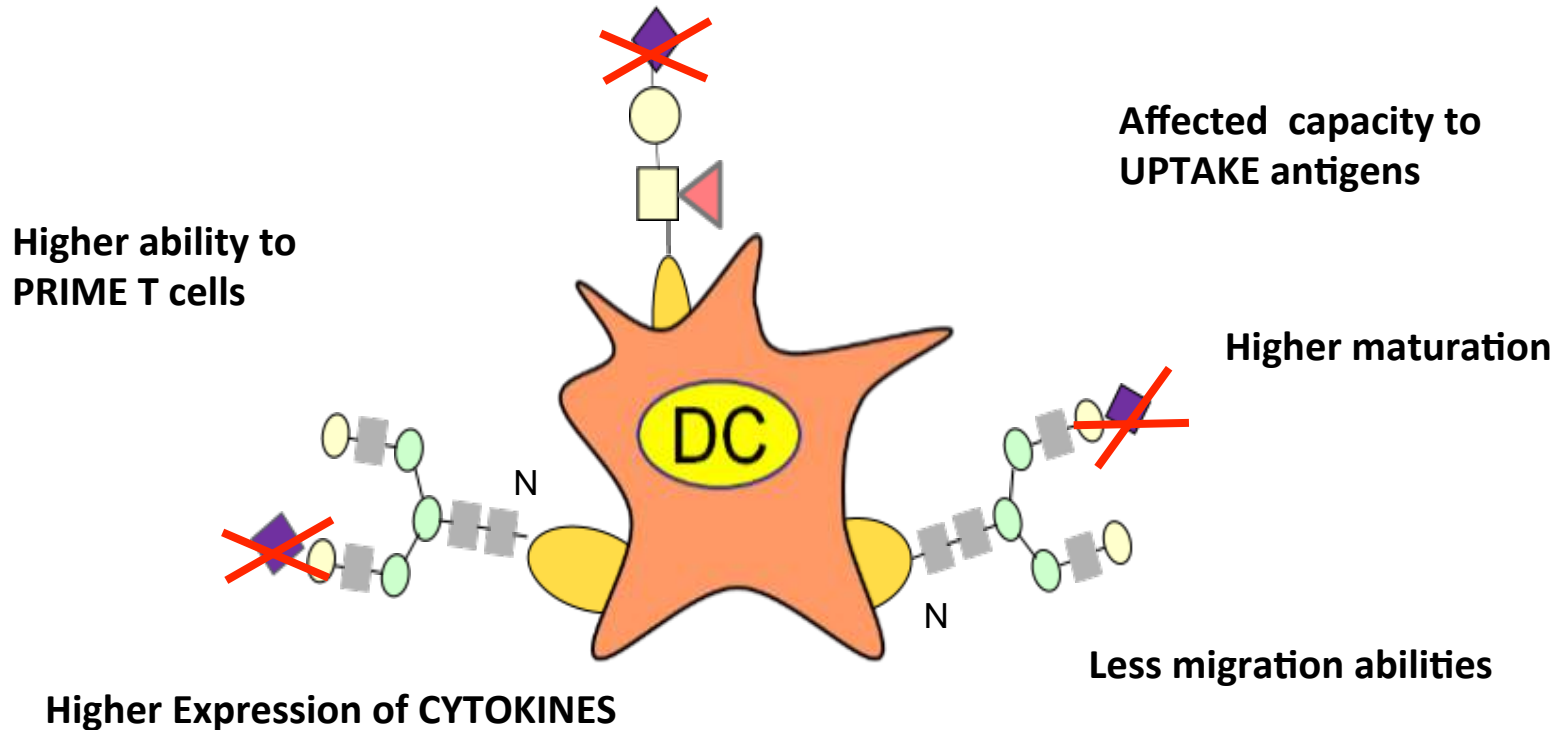
WP5:

- ✓ 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
- ✓ Writing articles (Congenital Disorders of Glycosylation (CDG): from glycoproteins to patient care . Royal Society of Chemistry. 2012)

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WP 3 – Biology

How Defects in sialylated glycans alters immunity



Cabral MG, et al (2012) The phagocytic capacity and immunological potency of human dendritic cells is improved by α 2,6-sialic acid deficiency. **Immunology**.

Videira et al (2008) Surface α 2-3- and α 2-6-sialylation of human monocytes and derived dendritic cells and its influence on endocytosis **Glycoconj. J.** 25:259-268;

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

Silva et al (2011) Sialyl Lewisx-dependent binding of human monocyte-derived dendritic cells to selectins **Biochem. Biophys. Res. Commun.**, 409, 459-464

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WP 4 – Clinical research in CDG

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

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

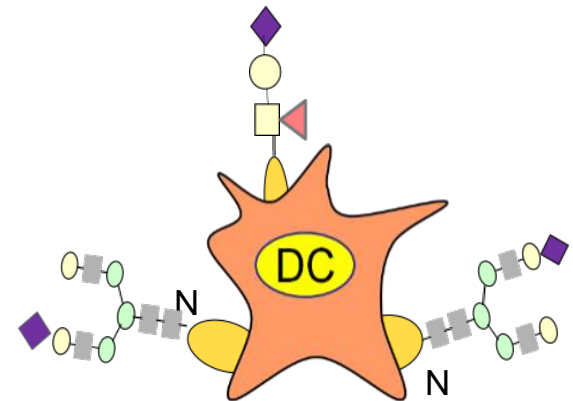
- Identify, collect and develop guides and resources of existing technical guidelines for the provision of care health CDG

General data	Patient ID
	Gender
	Born date
Classification	CDG type
	%CDTs
	Transferrin isoelectric focusing
	Molecular diagnosis
	Intellectual disability family history
	Consanguinity
	Gestational events
	Gestation length
	Newborn weight
	Newborn length
	Newborn head circumference
	Newborn period events
	Age at the onset of symptoms (months)
	Initial symptoms
	Age at diagnosis
	Multiorgan involvement
	Fatal event/death
	Current eight
	Current stature
	Current head circumference
	Micro/macrocephaly
	Psychomotor delay /intellectual disability
	Epilepsy
	Age at onset of epilepsy
	Seizure classification
	Number of antiepileptic
	EEG
	Ataxia
	Hypotonia
	Abnormal ocular movements
	Stereotypies
	Other movement abnormalities
	Polyneuropathy
	Stroke like episodes
	Head MRI
	Dysmorphic features
	Ophthalmologic abnormalities
	Orthopaedic abnormalities
	Growth failure
	Endocrinology abnormalities
	Heart abnormalities
	Gastro abnormalities
	Blood/coagulation abnormalities
	Kidney abnormalities
Other organs and systems	

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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