

CDG Natural History Study sponsored by Glycomine

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Conflict of interest statement:

I'm the main PI for the natural history study sponsored by Glycomine

I have no financial disclosure and I'm not an advisor for Glycomine

What are Natural History Studies (NHS)?

In medicine, a **natural history study** is a **study** that follows a group of people over time who have a specific medical condition or disease.

A **natural history study** collects health information over time in order to understand how the medical condition or disease develops and to give insight into how it might be treated.

Why do we need them?

There is an unmet need: we do not have prospective information on the natural history of CDG

1. What is normal variation?
2. What is natural improvement?
3. Do certain labs vary (go up and down), or reliable?
4. How can we develop therapies if we don't know what would happen to the patient **without** therapy
5. What should we measure on follow up during therapy to know, if the therapy works (biomarkers and end points)?

NHS: Where do we stand in CDG?

We only have retrospective natural history data in PMM2-CDG and NGLY1

The NIH conducts a prospective NHS and still enrolls CDG patients for an extensive 1 week workup

No publication on NHS in any other CDG

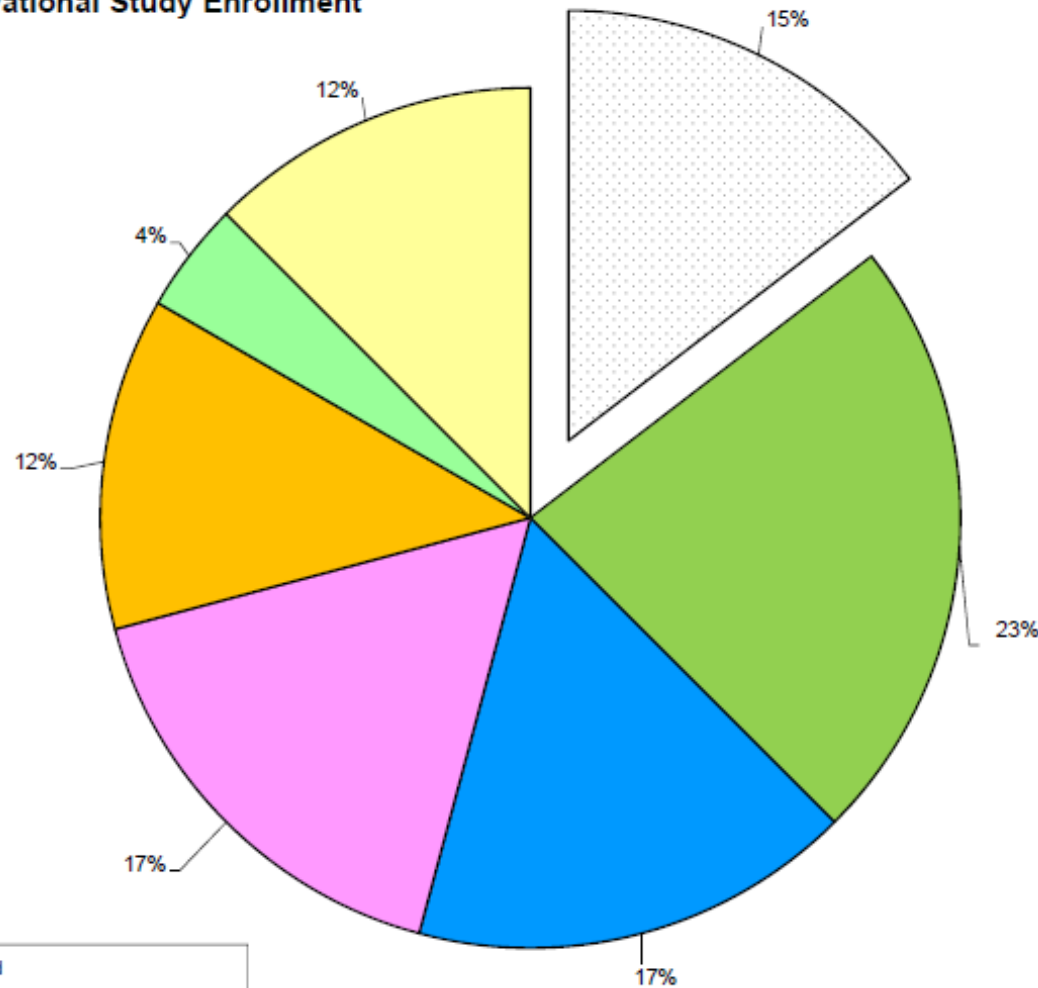


CDG NHS by Glycomine

- For whom is it: **only PMM2-CDG**
- How can families be involved? **Patients have to be followed at one of the study centers**
- Inclusion/exclusion criteria: **confirmed genetic diagnosis of PMM2**
- What are the main goals?

To define the natural outcome, clinical symptoms, laboratory findings and glycosylation abnormalities in PMM2-CDG patients

GLY-000 Observational Study Enrollment



- Not yet enrolled
- Mayo Clinic Rochester
- University Hospital Leuven
- Children's Hospital of Philadelphia
- Seattle Children's
- Tulane University
- Necker Hospital Paris

Planned enrollment: 48 patients
 Current enrollment: 41 patients
 as of July 05, 2019

Where do we stand so far?

	Enrollment	Percentage
Not yet enrolled	7	14.6%
Mayo Clinic Rochester	11	22.9%
University Hospital Leuven	8	16.7%
Children's Hospital of Philadelphia	8	16.7%
Seattle Children's	6	12.5%
Tulane University	2	4.2%
Necker Hospital Paris	6	12.5%
Current Total	41	85.4%

Still enrolling

Future perspectives for CDG NHS

Collaborative grant with the support of CDG Allies PPAIN and CDG CARE

Goal is to enroll 100 patients with all kind of CDG

Use unified follow up protocol to collect and share results

