# SYMPTOMATIC MANAGEMENT OF PMM2-CDG (CDG-Ia)


Jaak Jaeken

## WHAT IS CDG?

**Congenital Disorders of Glycosylation (CDG) are a growing group of diseases among the 8000 known rare diseases.**

They are caused by the incorrect or absent synthesis of sugar antennas (glycans) on proteins and lipids.

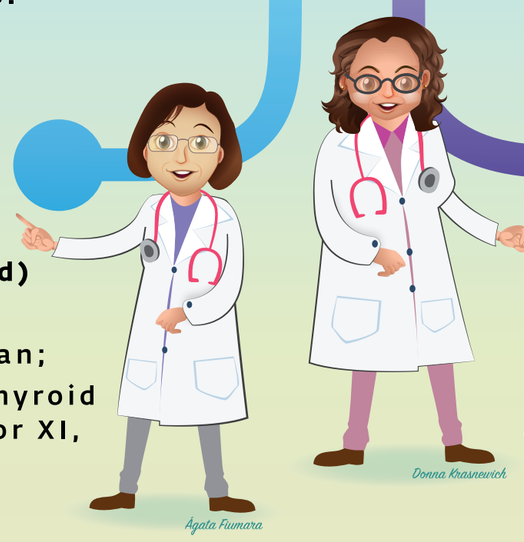
## HOW MANY PMM2-CDG PATIENTS ARE KNOWN?

Since many cases are unrecognized or misdiagnosed it is difficult to determine the real number of patients.

The prevalence may be as high as:

**1 in 20.000**


Marc C. Patterson


Ágata Fiumara

Donna Krasnewich

## RECOMMENDED ANNUAL SURVEILLANCE

**(or more frequently when indicated)**

- Eye examination;
- Complete clinical assessment by a physician;
- Blood examination: liver function tests, thyroid function tests, hematological factors (factor XI, protein C, protein S, antithrombin...)...

## WHAT ABOUT PMM2-CDG TREATMENT?

Currently, there is no specific treatment for PMM2-CDG but research towards this goal is going on in several centers.

## SYMPTOMATIC TREATMENTS

The scheme drawn below summarizes symptomatic treatments for PMM2-CDG. In addition, physical, speech and occupational therapy are very important.
**Note:** before surgery the consultation of a haematologist, familiar with CDG, is indicated.


Princess Lili (at age of 6 years old, PMM2-CDG)

| CLINICAL FEATURE | POSSIBLE INTERVENTION BY A PHYSICIAN |
|---|---|
| FAILURE TO THRIVE (+/- ENTEROPATHY, HYPOGLYCAEMIA) | NASOGASTRIC TUBE OR GASTROSTOMY TUBE FEEDING.[1] IN CASE OF CHRONIC DIARRHOEA, LACTOSE-FREE OR ELEMENTARY DIETARY FORMULA INSTEAD OF MILK PRODUCTS |
| STRABISMUS | GLASSES, PATCHING, BOTULINUM TOXIN OR SURGERY [2] |
| ACUTE MOTOR EVENT AND/OR LOSS OF CONSCIENCE | IS IT A STROKE-LIKE EPISODE, EPILEPSY OR A VASCULAR EVENT? |
| ORAL MOTOR DYSFUNCTION | CONSULTATION WITH A SPEECH THERAPIST/GASTROENTEROLOGIST/NUTRITIONIST [1][3] |
| SCOLIOSIS/KYPHOSIS | SURGICAL TREATMENT IN SEVERE FORMS [1] |
| PERICARDIAL EFFUSION | CORTICOSTEROIDS AND SALICYLIC ACID, PERICARDIAL DRAINAGE [4] |
| TRUE HYPOTHYROIDISM | L-THYROXINE SUPPLEMENTATION [1] |


1. S. E. Sparks and D. M. Krasnewich. GeneReviews. 2011.
2. D. Coman, M. Irving, P. Kannu, J. Jaeken and R. Savarirayan, Clin Genet, 2008, 73, 507-515.
3. D. Coman, D. Bostock, M. Hunter, P. Kannu, M. Irving, V. Mayne, M. Fietz, J. Jaeken and R. Savarirayan, Am J Med Genet A, 2008, 146, 389-392.
4. B. J. Feldman and D. Rosenthal, Pediatr Cardiol, 2002, 23, 469-471.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**THIS RESOURCE IS BROUGHT TO YOU BY THE PORTUGUESE ASSOCIATION FOR CDG AND RELATED RARE METABOLIC DISEASES (APCDG).**



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