SYMPTOMATIC MANAGEMENT OF PMM2-CDG (CDG-1a)

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.

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 ongoing 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.

POSSIBLE INTERVENTION BY A PHYSICIAN
Nasogastric tube or gastrostomy tube feeding in case of chronic diarrhoea, lactose-free or elementary dietary formula instead of milk products.

CLINICAL FEATURE
FAILURE TO THRIVE (-/- ENTEROPATHY, HYPOGLYCAEMIA)

CDG FAMILIES AND PROFESSIONALS UNITED TO BOOST RESEARCH AND ACHIEVE THERAPIES

This resource is brought to you by the Portuguese Association for CDG and Related Rare Metabolic Diseases (APCDG).

Authors: Vânia Ferreira (Portuguese Association for CDG - vaniaferreira@gmail.com), Ken Meres (Tufts University Medical School, USA)
Mercedes Serrano & Dali Pérez-DonCut (San Juan de Dios Children's Hospital, Spain) & Rita Barreira (CHU Hospital University of Canterbury, New Zealand),
Recipients: Joã Da Jersey (Centre for Metabolic Diseases, University Hospital of Galveston, Galveston, Texas, USA)

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