MAJOR SIGNS
AND SYMPTOMS OF
PMM2-CDG (CDG-IA)

GLYCOSYLATION

Glycosylation is the synthesis of sugar trees (glycans) and their attachment to proteins and lipids. It is a major post-translational modification that affects the functions of proteins such as enzymes, carriers of hormones and vitamins, receptor proteins, etc.

WHAT IS CDG?

Congenital Disorders of Glycosylation (CDG) is a growing group of diseases among the 8,000 known rare diseases.

Patients range from neonatal to adult age. Symptoms vary from patient to patient and there are severe and milder phenotypes.

NO CDG VS CDG

IMPORTANCE
OF GLYCOSYLATION

Glycosylation is the most important and complex form of post-translational modification. Approximately 1-2% of the total number of human genes (thus about 200-400 genes) are involved in this process.

CDG SHOULD BE CONSIDERED IN EVERY PATIENT WITH AN UNEXPLAINED SYNDROME

- Seizures
- Stroke-like episodes
- Cerebellar hypoplasia
- Dysmetria
- Ataxia
- Scoliosis
- Strabismus
- Nystagmus
- Pericardial effusion
- Abnormal hematological and endocrine parameters
- Low serum concentration of factors XI, Antithrombin III, Protein C and/or Protein S
- Low total serum thyroxine with mostly normal free serum thyroxine
- Abnormal biochemical parameters
- Increased serum transaminases
- Low serum cholesterol
- Failure to thrive
- Osteopenia

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).

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