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# MAJOR SIGNS AND SYMPTOMS OF PMM2-CDG (CDG-1A)

## 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.

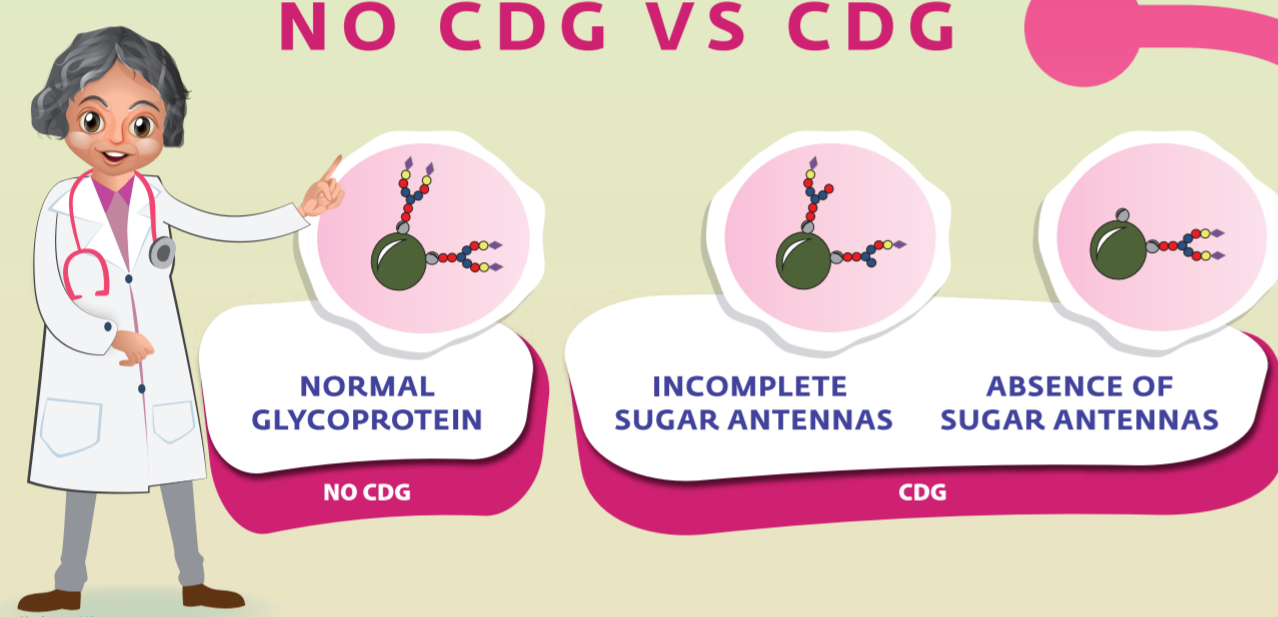
## WHAT IS CDG?

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

H. Schachter and H. H. Freeze, Biochim Biophys Acta, 2009, 1792, 925-930

## CDG SHOULD BE CONSIDERED IN EVERY PATIENT WITH AN UNEXPLAINED SYNDROME



- SEIZURES
- STROKE-LIKE EPISODES
- CEREBELLAR HYPOPLASIA
- DYSMETRIA
- ATAXIA



- SCOLIOSIS



- STRABISMUS
- NYSTAGMUS

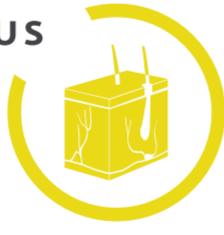


- PERICARDIAL EFFUSION



- 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 SUBCUTANEOUS FAT PATTERN
- ABSENCE OF PUBERTAL DEVELOPMENT
- INVERTED NIPPLES



- HEPATOMEGALY



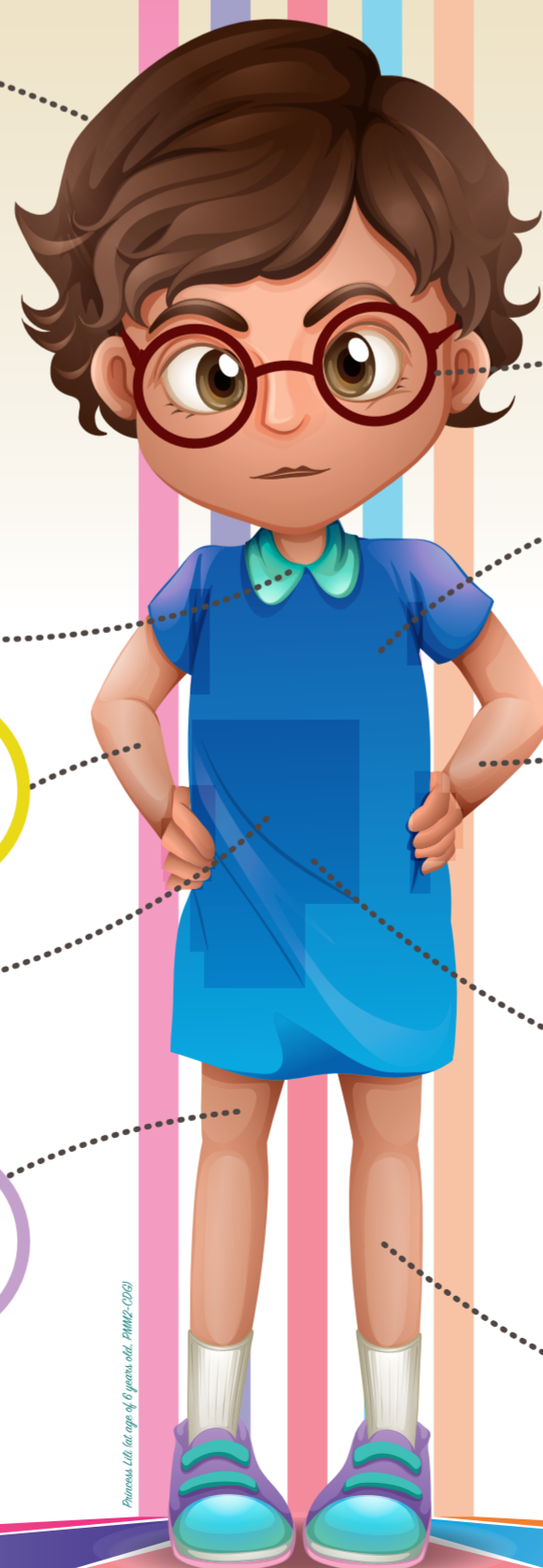
- ABNORMAL BIOCHEMICAL PARAMETERS
- INCREASED SERUM TRANSAMINASES
- LOW SERUM CHOLESTEROL



- FAILURE TO THRIVE



- OSTEOPENIA



**CONGENITAL DISORDERS OF GLYCOSYLATION WORLD CONFERENCE**  
 The power of advancing patient-oriented research united families and professionals

**CDG FAMILIES AND PROFESSIONALS UNITED TO BOOST RESEARCH AND ACHIEVE THERAPIES**

PORTUGUESE ASSOCIATION

