CDG DIAGNOSIS

A simplified guide for different stakeholders

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INTRODUCTION

Congenital disorders of glycosylation (CDG) are inherited metabolic diseases that cause defects in glycosylation. CDG diagnosis is most of the times challenging due to the high and growing number of CDGs (~140) but also due to heterogeneous phenotypes and variable severity. The classical screening test is serum transferrin isoelectrofocusing. However, it cannot be used for every CDG and can generate false negatives.

AIM

To address the challenges of CDG diagnosis by representing an overview of the different approaches that help CDG diagnosis.

METHODOLOGY

Literature review to identify the CDGs’ clinical and biochemical hallmarks as well as the biochemical and genetic testing available.

RESULTS

01. Clinical manifestations reported in CDG per organ/system

02. Biochemical and laboratory findings reported in CDG per organ/system

03. Clinical features suggestive of distinct CDG

04. Diagnostic decision tree in face of a patient with a suspicion of CDG

CONCLUSIONS

This work provides:

- Guidance for health care providers on how to diagnose CDG.
- A list of clinical and biochemical hallmarks per CDG which can help direct the suspicion of a specific CDG diagnosis.
- Raises awareness for CDG.

REFERENCES


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Figure 1 – Flowchart for CDG diagnosis. Schematic flowchart for CDG diagnostics.

Legend: ALP – Alkaline phosphatase, Age-C – Age at presentation, CDG – Congenital dysglycosylation, MCAD – Methylcrotonic acidemia, MNGIE – Mitochondrial neurogastrointestinal encephalomyopathy, OTC – Ornithine transcarbamylase, PKU – Phenylketonuria, T1/2 – Median survival time, T2 – Median survival time, WES – Whole exome sequencing, WGS – Whole genome sequencing.