Clinical and Basic Investigations into Congenital Disorders of Glycosylation

Status: Recruiting

Eligibility Criteria

Sex: All
Age: Not specified
This study is NOT accepting healthy volunteers

Inclusion Criteria:

• Patients diagnosed with congenital disorders of glycosylation based on genetic confirmatory testing

Exclusion Criteria:

• Patients without congenital disorders of glycosylation

Conditions & Interventions

Conditions:
Congenital Disorders of Glycosylation
Keywords:
CDG, CDDG, Congenital Disorders of Glycosylation, Congenital Disorders of Deglycosylation, ALG1, ALG3, ALG6, ALG12, ALG13, COG6, DPAGT1, DPM1, EDEM3, MAN1B1, MPDU1, MPI, NGLY1, PGAP3, PGM1, PIGA, PIGG, PIGN, PIGS, PIGT, PMM2, SLC35A2, SLC35C1, SLC39A8, SRD5A3, SSR4, FUT8, GALNT2, MAN2B2, VMA21

More Information

Description: Define natural history, validate patient reported outcome and share knowledge on congenital disorders of glycosylation. We will recruit and enroll patients with CDG in this study evaluating clinical variation and natural history when a patient is being seen as part of routine clinical care.
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