Unexplained developmental delay, multisystemic disease with or without a neurologic component, multiple unexplained abnormalities of basic laboratory results

Order both of the following:
- CDGN / Congenital Disorders of N-Glycosylation, Serum
- OLGU / Oligosaccharide Screen, Random, Urine

Order CDGN - consistent with type I CDG (congenital disorder of glycosylation)
- OLGU - Normal

Order PMMIL / Phosphomannomutase and Phosphomannose Isomerase, Leukocytes

Consider either
- NGLY1 Gene List ID: IEMCP-2GS7X1
- MOGS Gene List ID: IEMCP-KLLWC2


Consider CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing, Varies
Indicate either
- NGLY1 Gene List ID: IEMCP-2GS7X1
- MOGS Gene List ID: IEMCP-KLLWC2

Deficient phosphomannomutase
- CGPH and indicate PMM2 Gene List ID: IEMCP-F6CKG8 on the request
- Referral to a genetics specialist

Deficient phosphomannose isomerase
- CGPH and indicate MPI Gene List ID: IEMCP-DJHNFJ on the request
- Referral to a genetics specialist

Normal activity

Consider CDGN - consistent with mixed type CDG
- OLGU - Normal


CDG confirmed
- Consider referral to a genetics specialist

POSITIVE

STOP or consider referral to a genetics specialist for additional clinical or research testing

NEGATIVE

Rule out secondary causes

**CDG presentations are variable and often not recognizable on clinical grounds alone**

**One or more of the following: elevated ALP, reduced clotting factors, reduced blood/urine manganese, endocrine abnormalities, proteinuria**

**Frontiers in Congenital Disorders of Glycosylation (FCDGC)**

**Secondary causes include alcohol abuse, liver dysfunction**