Newborn screen result: decreased sphingomyelinase with or without abnormal 2nd-tier testing

- ASMW / Acid Sphingomyelinase, Leukocytes
- OXYBS / Oxysterols, Blood Spots

Decreased acid sphingomyelinase (ASM)
Elevated lyso-sphingomyelin (LSM)*

Niemann-Pick Disease (NPD) confirmed**

Referral to Genetics Specialist

Optional

Decreased ASM
Normal oxysterols

Consider

NPABZ / Niemann-Pick Disease, Types A and B, Full Gene Analysis, Varies

NPD confirmed*

None or 1 mutation identified

SMPD1 deletion/duplication analysis should be considered if sequencing is not informative

Normal ASM
Normal oxysterols

Not NPD (Consult with Genetics Specialist if clinical suspicion for NPD is high)

* Cholestan-3 beta, 5 alpha, 6 beta-triol (COT), and/or 7-ketocholesterol (7-KC) may also be elevated

** Limited genotype-phenotype correlation is known. Clinical correlation is required to make a diagnosis of type A versus type B