Newborn Screen Follow-up for Niemann Pick Type A and B

Decreased sphingomyelinase with or without abnormal 2nd-tier testing

- PLSD / Lysosomal and Peroxisomal Storage Disorders Screen, Blood Spot
- OXYBS / Oxysterols, Blood Spots

Decreased ASM
Elevated LSM*

NPD confirmed**

Referral to Genetics Specialist

Optional

Decreased ASM
Normal oxysterols

Consider

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

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)

ASM=acid sphingomyelinase
LSM=lyso-sphingomyelin
NPD=Niemann-Pick Disease

* Cholestane-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
*** Consider targeted mutation analysis (NPABP / Niemann-Pick Disease, Types A and B, Mutation Analysis) if the patient is of Ashkenazi Jewish descent or from the Maghreb region of North Africa, Chile, Saudi Arabia, or Turkey