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

- **ASMW** / Acid Sphingomyelinase, Leukocytes
- **OXYBS** / Oxysterols, Blood Spot

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

#### Niemann-Pick Disease (NPD) confirmed**
- Referral to Genetics Specialist (optional)

#### NPD confirmed*

#### None or 1 variant identified
- **SMPD1** deletion/duplication analysis should be considered if sequencing is not informative

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

* 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