If deficient, MLD confirmed

One of the following suspected:
- Aspartylglucosaminuria
- α-Mannosidosis
- β-Mannosidosis
- Pompe disease
- Sandhoff disease
- Sialidosis
- Galactosialidosis*
- α-L-fucosidosis*
- Mucolipidosis II/III*
- Morquio A & B*

Consider: LSDGP / Lysosomal Storage Disease Gene Panel, Varies


Clinical information:
- Developmental delay/Cognitive impairment
- Coarse features/Organomegaly
- Dysostosis multiplex
- Neurodegeneration/Behavioral changes
- Ichthyosis
- Hearing defects/loss

LSDS / Lysosomal Storage Disorders Screen, Random, Urine
Testing includes:
- Mucopolysaccharides (MPS)
- Oligosaccharides (OLIGO)
- Ceramide trihexosides (CT)
- Sulfatides (S)

Order 1 of the following:
- ARSAW / Arylsulfatase A, Leukocytes
- ARSU / Arylsulfatase A, 24 Hour, Urine

Order BOTH of the following:
- ARSAW
- I2SW / Iduronate-2- Sulfatase, Blood

For recommended diagnostic workup, see Fabry Disease Diagnostic Testing Algorithm

* These conditions may also have elevated MPS