Newborn Screen Follow-up for Mucopolysaccharidosis Type I

Decreased alpha-L-iduronidase (IDUA) and abnormal 2nd-tier testing

- IDSBS / Alpha-L-Iduronidase, Blood Spot
- IDSWB / Alpha-L-Iduronidase, Blood
- MPSSC / Mucopolysaccharides (MPS) Screen, Urine*

IDUA activity – decreased
Urine MPS – abnormal

MPS I confirmed

Referral to Genetics Specialist
optional

MPS1Z / Hurler Syndrome, Full Gene Analysis***

IDUA activity – decreased
Urine MPS – normal

IDUA pseudodeficiency likely

IDUA activity – normal
Urine MPS – normal or abnormal

Not MPS I**

MPS I = Mucopolysaccharidosis type I
IDUA = Alpha-L-iduronidase
MPS = Mucopolysaccharides

* Mucopolysaccharides are also known as glycosaminoglycans
** Consult with Genetics Specialist if clinical suspicion for MPS I or other condition is high
*** IDUA deletion/duplication testing should be considered if sequencing is not informative