Newborn Screen Follow-up for Mucopolysaccharidosis Type I

Newborn screen result: decreased alpha-L-iduronidase (IDUA)

Order 1 from each of the following groups:
- IDUAW / Alpha-L-Iduronidase, Blood
- MPSBS / Mucopolysaccharidosis, Blood Spot
- MPSWB / Mucopolysaccharidosis, Blood

- IDUA activity – decreased
- Mucopolysaccharidosis (MPS) – elevated dermatan and heparan sulfate

Mucopolysaccharidosis I (MPS I) confirmed

Referral to Genetics Specialist

Order: MPS1Z / Hurler Syndrome, Full Gene Analysis, Varies
Consider IDUA deletion/ duplication analysis if sequencing is uninformative

- IDUA activity – decreased
  - MPS – negative

IDUA pseudodeficiency or carrier status likely

- IDUA activity – normal
  - MPS – negative or positive

MPS I unlikely
Consider consultation with a Genetics Specialist if clinical suspicion for MPS I or other condition is high

optional