

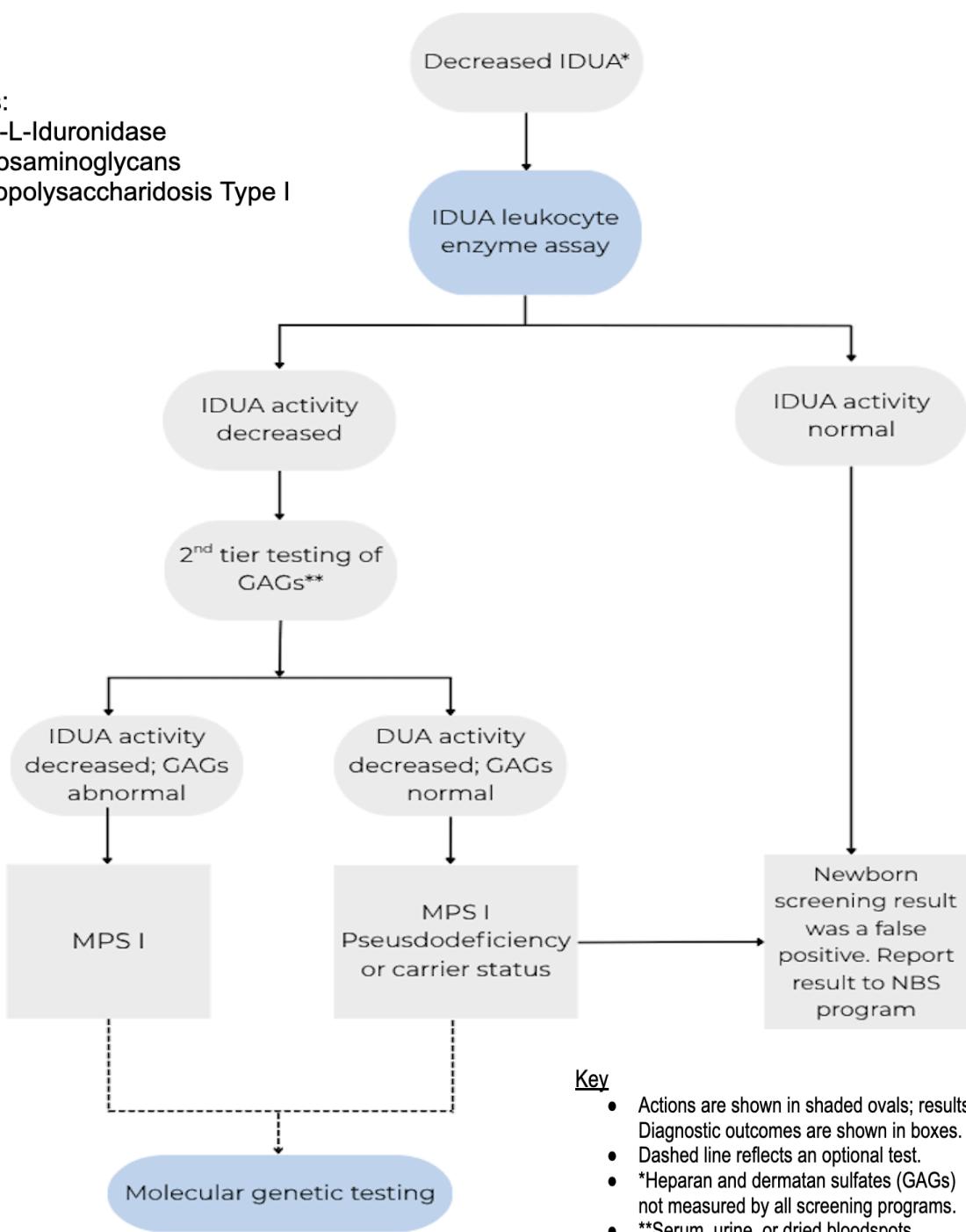
MPS I: Decreased Alpha-L-Iduronidase; Elevated Dermatan and Heparan Sulfates

Abbreviations:

IDUA = alpha-L-Iduronidase

GAGs = glycosaminoglycans

MPS I = Mucopolysaccharidosis Type I



Disclaimer: This practice resource is designed primarily as an educational resource for medical geneticists and other clinicians to help them provide quality medical services. Adherence to this practice resource is completely voluntary and does not necessarily assure a successful medical outcome. This practice resource should not be considered inclusive of all proper procedures and tests or exclusive of other procedures and tests that are reasonably directed to obtaining the same results. In determining the propriety of any specific procedure or test, the clinician should apply his or her own professional judgment to the specific clinical circumstances presented by the individual patient or specimen. Clinicians are encouraged to document the reasons for the use of a particular procedure or test, whether or not it is in conformance with this practice resource. Clinicians also are advised to take notice of the date this practice resource was adopted, and to consider other medical and scientific information that becomes available after that date. It also would be prudent to consider whether intellectual property interests may restrict the performance of certain tests and other procedures. Abbreviations: IDUA = alpha-L-Iduronidase GAGs = glycosaminoglycans MPS I = Mucopolysaccharidosis Type I IDUA activity decreased GAGs elevated consistent with MPS I IDUA leukocyte enzyme assay Blood or urine GAGs** Molecular genetic testing Key - Actions are shown in shaded ovals; results are in the unshaded ovals. Diagnostic outcomes are shown in boxes. - Dashed line reflects an optional test. - *Heparan and dermatan sulfates (GAGs) not measured by all screening programs. - **Serum, urine, or dried bloodspots.