Glycosaminoglycans in Urine

Specimen Type	Urine
Specimen Volume	20 mL
Collection	Urine collection cup. Freeze immediately. Include the patient age with the requisition.
Minimum Volume	10 mL
Handling	Ship frozen on dry ice.
Rejection Criteria	Unfrozen specimens. Specimens outside of listed stability. Samples submitted without two unique identifiers and date of collection.
Stability	3 freeze-thaw cycles. Frozen at -20°C for 8 weeks. Frozen at -80°C for 8 weeks.
Methodology	Colorimetric (1,9-dimethyl-methylene blue) dye binding

Reference Range

Age	**mg GAGs/mmol creatinine**
0-1 year	<36.0 mg GAGs/mmol creatinine
2-3 years	<17.8 mg GAGs/mmol creatinine
4-5 years	<14.8 mg GAGs/mmol creatinine
6-11 years	<12.4 mg GAGs/mmol creatinine
12-13 years	<9.5 mg GAGs/mmol creatinine
14-17 years	<5.1 mg GAGs/mmol creatinine
>17 years	<3.1 mg GAGs/mmol creatinine

Please Note: The reference range year accounts for the full calendar year.

Turnaround Time	Up to 7 business days.
CPT Code	83864

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Clinical Significance	The mucopolysaccharidoses (MPSs) are a family of inheritable disorders caused by a deficiency of lysomal enzymes required to degrade mucopolysaccharides, also known as glycosaminoglycans (GAGs). The un-degraded or partially degraded GAGs are stored in lysosomes and excreted in the urine. The quantity of excreted urinary GAGs is age-dependent. Infants secrete more GAGs than adults. Normal urine contains primarily chondroitin sulfate with small quantities of heparin sulfate and dermatan sulfate. Once mucopolysaccharidoses is diagnosed by total GAG analysis, a differential diagnosis based upon the abnormal distribution of sulfated GAGs in urine must be performed. Differential diagnosis is a requirement because many of the various enzyme deficiencies share similar clinical features. These features include a chronic and progressive course, multi-system involvement and organomegaly. Hearing, vision, cardiovascular function and joint mobility are affected. Profound mental retardation is found in the Hurler, Hunter and San Filippo syndromes (MPS types I, II and III), but normal intellectual functioning is retained in other MPSs and some mildly affected Hunter patients.
Principle	The urine samples are pre-treated to concentrate the glycosaminoglycans and remove any background, interfering components. The samples, controls and standards are incubated with a dye label (1,9-dimethyl-methylene blue). The dye binds specifically with the sulfated, polysaccharide component of proteoglycans and protein-free, sulfated, glycosaminoglycan chains. The dye-GAG complexes precipitate within 10 minutes. The samples, controls and standards are centrifuged and the supernatant decanted. The samples are re-dissolved in a disassociation reagent. After an incubation step, the absorbance of the samples are measured at 656nm. The absorbance of the samples are compared to known standards and the concentrations of the samples are determined. Finally, the GAG concentration is normalized against creatinine in order to control for variation in urine flow rates.