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Total Glycosaminoglycans (Mucopolysaccharides), quantitative, random urine.

Useful For

1. Screening test for diagnosis of mucopolysaccharidosis
2. Follow-up testing for treatment of mucopolysaccharidosis

Methodology

Spectrophotometry using dimethylmethylene blue (DMMB) method

Clinical Information

Early diagnosis of mucopolysaccharidosis (MPS) in the asymptomatic stage may be effective at preserving organic function and improving outcomes. When a mucopolysaccharidosis (MPS) is suspected on the basis of screening results, additional tests may be necessary. Finally, an enzyme test is required to establish a diagnosis. MPSs are a group of rare lysosomal storage disorders with multi-organic and severe symptoms. These group disorders caused by the deficiency of any of the enzymes involved in the stepwise degradation of dermatan sulfate, heparan sulfate, keratan sulfate, or chondroitin sulfate (glycosaminoglycans: GAGs). Undegraded or partially degraded GAGs (also called mucopolysaccharides) are stored in lysosomes and excreted in the urine. Accumulation of GAGs in lysosomes interferes with normal functioning of cells, tissues, and organs resulting in the clinical features observed in MPS disorders. There are 11 known enzyme deficiencies that result in MPS. Clinical features can be different and depending on the specific enzyme deficiency, and include coarse facial features, neurological symptoms, cognitive retardation, hernias, kyphoscoliosis, corneal clouding, hepatosplenomegaly *etc.*

Necessary Information

Test order form
Clinical background of patient
Payment document
The sample should be delivered cold or preferably frozen to the Mega-Lab, laboratory wing, ground floor on week days between the hours 08:00-15:00.
Instruct patient: The patient's first morning urine is not to be collected. Then collect a random second morning urine in a collection container without any

preservative. Early-morning urine **should be discarded** because GAG excretion referred to creatinine concentration is increased during the night.

Patient Preparation: Do not administer low-molecular weight heparin prior to collection

Specimen Volume : 2 ml

Specimen Stability Information
Room temperature: Unacceptable
Refrigerated: 2 days
Frozen (preferred): 90 days at -20 ^{0}C

Interpretation
Total GAG quantification can aid biochemical diagnosis.
An abnormally elevated excretion of GAG is characteristic of mucopolysaccharidoses.
However, an abnormal mucopolysaccharide analysis is not sufficient to conclusively establish a specific diagnosis. Results of this test should be interpreted in the context of relevant clinical and family history and physical examination findings. It is strongly recommended to seek confirmation by an independent method, typically in vitro enzyme assay (available in either blood or cultured fibroblasts from a skin biopsy) or molecular analysis.
GAG levels may normalize or remain elevated in patients who have undergone bone marrow transplants or are receiving enzyme replacement therapy. Therefore, GAG measurements can be advisable for monitoring in these cases.
False positive results are common, particularly in young infants, patients with excessive connective tissue destruction (disseminated lupus erythematosus, some patients with rheumatoid arthritis), bladder disease, or bone disease. In addition, heparin and acrylic acid polymer used in paper diapers, and glue from collecting bags will interfere with the assay.

Clinical Reference

The total GAG concentration for DMMB assay is determined as Heparan Sulfate concentration and expressed in mg/mmol creatinine. Heparane sulfate is used as marker for MPS types I, II, III, VI and VII. in this assay.
Excretion of urinary GAGs is related to the age of the patients.

Age Groups	Urine MPS reference values (mg/mmol creatinine) Low – High
0-5 m	15.2 – 52
6-12 m	15.1 – 32

1Y	9.1 – 30
2-3Y	7.7 – 21
4-5Y	7.6 – 14
6-7Y	5.7 – 13
8-9Y	5.2 – 12
10-14Y	3.4 – 11
15-19Y	1.5 – 7
>20Y	1.5 – 5

Turnaround time: 30 working days

Ministry of Health code: 83866

References:

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