Mucopolysaccharidosis Type VII (Sly Syndrome): Beta-Glucuronidase Enzyme Activity, Leukocytes

Test Code: LQ
Turnaround time: 7 days - 10 days
CPT Codes: 82657 x1

Condition Description

MPS VII is a disease that affects the breakdown of dermatan sulfate and heparan sulfate. Like other lysosomal storage disorders, severity varies among individuals. Onset may be in the prenatal period with hydrops fetalis. Clinical symptoms include postnatal growth deficiency, macrocephaly, coarse facies, hearing loss, variable corneal opacities, valvular heart disease, hepatosplenomegaly, dysostosis multiplex, hydrocephalus and hirsutism. Development is usually slowed by 1-3 years of age followed by progressive neurodegeneration until death.

For further information about lysosomal storage diseases, please call the Emory Lysosomal Storage Disease Center at (404) 778-8565 or (800) 200-1524. For general questions, call EGL Genetics at 470-378-2200.

Click here for the GeneReviews summary on this condition.

Indications

This test is indicated for children or adults with symptoms of MPS VII (β-glucuronidase deficiency).

Methodology

Flurometric Enzyme Assay using artificial 4-MU substrate. Beta-glucuronidase is evaluated to confirm a diagnosis of MPS VII.

Specimen Requirements

Submit only 1 of the following specimen types

Type: Whole Blood (Sodium Heparin)

Specimen Requirements:
Sodium Heparin (Green Top)
3-5 ml

Specimen Collection and Shipping:
Ship sample at room temperature for receipt at EGL within 24 hours of collection. Do not refrigerate or freeze. Not accepted on Saturday. (Late Friday collections may be stored at room temperature over the weekend for Monday receipt.)

Related Tests

- Mucopolysaccharide screen, GAG's (GA)
- Lysosomal enzyme screening panel (LS)