Mucopolysaccharidosis Type IIIA: SGS\textit{H} Gene Deletion/Duplication

**Test Code:** HX  
**Turnaround time:** 2 weeks  
**CPT Codes:** 81228 x1

### Condition Description

Mucopolysaccharidosis type IIIA (MPS IIIA, Sanfilippo syndrome type A), is a member of a group of inherited metabolic disorders collectively termed mucopolysaccharidoses (MPSs). The MPSs are caused by a deficiency of lysosomal enzymes required for the degradation of mucopolysaccharides or glycosaminoglycans (GAGs) within the lysosome. When functioning normally, the lysosomal enzymes break down these GAGs, however when the enzyme is deficient, the GAGs build up in the lysosomes causing damage to the body's tissues. The MPSs share a chronic progressive course with multisystem involvement, characteristic physical features, laboratory findings, and radiographic abnormalities. MPS IIIA is an autosomal recessive condition caused by a deficiency of the enzyme heparan N-sulfatase.

Clinical features of MPS IIIA include hyperactivity, aggressiveness, and developmental delays in childhood. Mental abilities decline as the disease progresses. Involvement of other organ systems tends to be mild and dysmorphic features are subtle than those observed in other type of mucopolysaccharidosis [1].

MPS IIIA is caused by mutations in the SGS\textit{H} gene, but is clinically indistinguishable from MPS IIIB, MPS IIIC, and MPS IIID, which are caused by mutations in other genes. All four forms of MPS III result in buildup of the same GAG, heparin sulfate. Diagnostic sequencing analysis of the SGS\textit{H} gene coding region is available for MPS type IIIA patients and their at-risk relatives on a clinical basis.

For questions about testing for MPS IIID, call EGL Genetics at 470-378-2200. For further clinical information about lysosomal storage diseases, including management and treatment, call the Emory Lysosomal Storage Disease Center at (404) 778-8565 or (800) 200-1524.


### References:
Older Children & Adults: 5-10 ml

Specimen Collection and Shipping: Refrigerate until time of shipment. Ship sample within 5 days of collection at room temperature with overnight delivery.

**Type: Saliva**

Specimen Requirements:

Oragene™ Saliva Collection kit (available through EGL) used according to manufacturer instructions.

Specimen Collection and Shipping: Store sample at room temperature. Ship sample within 5 days of collection at room temperature with overnight delivery.

**Special Instructions**

Submit copies of diagnostic biochemical test results with the sample. Sequence analysis is required before deletion/duplication analysis by targeted CGH array. If sequencing is performed outside EGL Genetics, please submit a copy of the sequencing report with the test requisition. Contact the laboratory if further information is needed.

**Related Tests**

- Mucopolysaccharide screen (urine GAG) (GA)
- Gene sequencing for MPS IIIB (BB) and MPS IIID (BH)
- Known mutation analysis (Custom Diagnostics) is available to test family members.
- Prenatal testing is available for known familial mutations only. Please call the Laboratory Genetic Counselor for specific requirements for prenatal testing before collecting a fetal sample.