Mucopolysaccharidosis Type IVA: GALNS Gene Sequencing

Test Code: BU
Turnaround time: 4 weeks
CPT Codes: 81479 x1

Condition Description

Mucopolysaccharidosis type IV A (Morquio syndrome, MPS IV A) is a member of a group of inherited metabolic disorders collectively termed mucopolysaccharidoses (MPSs). The MPS's are caused by a deficiency of lysosomal enzymes required for the degradation of mucopolysaccharides or glycosaminoglycans (GAGs). Morquio syndrome type IVA is caused by deficiency of galactosamine-6-sulfatase (N-acetyl-galactosamine-6-sulfate sulfatase deficiency). Deficiency of this enzyme leads to the accumulation of the GAG, keratan sulfate, in the lysosomes.

Symptoms of Morquio syndrome include the excretion of specific urinary glycosaminoglycans and skeletal abnormalities. Most individuals affected by Morquio syndrome do not have coarse facial features or mental retardation. Skeletal manifestations of Morquio syndrome include: odontoid hypoplasia, a striking short trunk dwarfism, and genu valgus. Compared to other patients with MPS, those with Morquio syndrome tend to have greater spine involvement with scoliosis, kyphosis, and severe gibbus, as well as platyspondyly, rib flaring, and ligamentous laxity. In earlier clinical descriptions, MPS Type IVA was considered to have more severe manifestations than type IVB. However, with the ability to differentiate between types A and B by enzyme analysis, it is understood that significant variability in clinical expression exists within both groups. No clear clinical differentiation between Morquio syndrome type IVA and IVB exists.

Mutations to the GALNS gene cause deficiency of galactosamine-6-sulfatase. Diagnostic sequencing analysis of the GALNS gene coding region is available for MPS IV A patients and their at-risk relatives on a clinical basis.

For patients with mutations not identified by full gene sequencing, a separate deletion/duplication assay is available using a targeted CGH array (LX).

For questions about testing for MPS IV A, call the Emory Genetics Laboratory at (404) 778-8499 or (855) 831-7447. 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:
Submit only 1 of the following specimen types

* Preferred specimen type: Whole Blood

**Type: Whole Blood**

Specimen Requirements:

In EDTA (purple top) or ACD (yellow top) tube:
- Infants (2 years): 3-5 ml
- 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 of Emory Genetics Laboratory, 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)
- Known mutation analysis (Custom Diagnostics) is available to test family members.
- Deletion/duplication analysis for the GALNS gene is available separately for individuals where mutations are not identified by sequence analysis.
- 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.