Mucopolysaccharidosis Type I: \textit{IDUA} Gene Deletion/Duplication

\textbf{Test Code:} LL  
\textbf{Turnaround time:} 2 weeks  
\textbf{CPT Codes:} 81228 x1

\section*{Condition Description}

Mucopolysaccharidosis type I or Hurler Syndrome (MPS I) is a progressive multisystem disorder with features that range over a continuum from mild to severe. MPS I is an autosomal recessive progressive disorder that results from the body’s inability to make lysosomal alpha-L-iduronate, an enzyme that helps break down mucopolysaccharides. The enzyme deficiency found in MPS type I causes mucopolysaccharides to build up in the body, causing damage to many tissues and organs in the body.

MPS Type I is divided into three subtypes, but there is no clear distinction between the groups. Therefore, a classification based on disease severity has been suggested: Hurler as severe MPS I, Hurler-Scheie as intermediate MPS I, and Scheie as mild MPS I. Treatment is available through hematopoietic stem cell/bone marrow transplantation or enzyme replacement therapy.

MPS I is caused by mutations in the \textit{IDUA} gene and the diagnosis relies on the demonstration of deficient activity of the lysosomal enzyme alpha-L-iduronidase in peripheral blood leukocytes or cultured fibroblasts. Diagnostic sequencing analysis of the \textit{IDUA} gene coding region is now available for MPS type I patients and their at-risk relatives on a clinical basis.

For questions about testing for MPS I, call EGL Genetics at (470) 378-2200 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.

\section*{References}


\section*{Indications}

- Confirmation of a clinical diagnosis of MPS I Disease  
- Prenatal testing for known familial mutations.  
- Assessment of carrier status in high risk family members known mutation analysis.

\section*{Methodology}

DNA isolated from peripheral blood is hybridized to a CGH array to detect deletions and duplications.

The targeted CGH array has overlapping probes which cover the entire genomic region. Please note that a “backbone” of probes across the entire genome are included on the array for analytical and quality control purposes. Rarely, off-target copy number variants causative of disease may be identified that may or may not be related to the patient’s phenotype. Only known pathogenic off-target copy number variants will be reported. Off-target copy number variants of unknown clinical significance will not be reported.

\section*{Specimen Requirements}

Submit only 1 of the following specimen types

\* Preferred specimen type: Whole Blood

\section*{Type: Whole Blood}

Specimen Requirements:

- In EDTA (purple 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
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)
- 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.