Mucopolysaccharidosis Type VI (Maroteaux Lamy Syndrome): Arylsulfatase B Enzyme Activity, Leukocytes

Test Code: BMPS6
Turnaround time: 7 days - 10 days
CPT Codes: 82657 x1, 84155 x1, 84311 x1

Condition Description

Mucopolysaccharidosis type VI (MPS VI), also known as Maroteaux-Lamy Syndrome, is a lysosomal storage disorder caused by absence or dysfunction of the enzyme arylsulfatase B (N-acetylgalactosamine 4-sulfatase). This enzyme is one of a group responsible for the degradation of dermatan sulfate, a glycosaminoglycan (GAG) normally broken down in the lysosomes. In MPS VI, insufficient enzyme activity is available and the degradation of dermatan sulfate is blocked, leading to accumulation of this substrate in the lysosomes of several tissues.

The clinical presentation can vary from mild to severe. The major clinical manifestations are corneal clouding, joint stiffness, and a skeletal dysplasia known as dysostosis multiplex. Unlike most lysosomal storage disorders, intelligence is unaffected. Macrocephaly and sternal abnormalities can be present at birth, and inguinal/umbilical hernias are common. Restriction of joint movement develops sometime in the first few years of life, and a typical crouched posture is assumed. Hepatomegaly, corneal clouding, claw-hand deformities, cardiac valve involvement, decreased pulmonary function, and sleep apnea become evident as the child ages. Respiratory infections are common. Growth in height is usually less than normal, but variable with the severity of disease. Facial features become more coarse with age, and individuals with MPS VI often resemble one another. Deafness, both sensorineural or conductive, is seen in all types of mucopolysaccharidoses, including MPS VI. Spinal cord compression is a typical complication in older children and adults. Carpal tunnel syndrome and nerve compression is also seen in older children and adults. Enzyme replacement therapy (ERT) for MPS VI has been approved by the FDA and is available for treatment of this disorder.

Indications

Corneal clouding, joint stiffness, dysostosis multiplex, short stature, coarse features, normal intellect

Methodology

Enzymatic reaction detected by spectrophotometric assay. Arylsulfatase B activity is evaluated to confirm diagnosis of Maroteaux Lamy.

Detection

The vast majority of affected patients will have a detectable deficiency.

Specimen Requirements

Type: Whole Blood

Specimen Requirements:
In sodium heparin (green top) tube: 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.

Special Instructions

Related Tests

Lysosomal Enzyme Screening Panel (LS)
Mucopolysaccharide screen (urinary GAGs) (GA)
Full gene sequencing, for known affected or for carrier screening
A deletion/duplication assay is available separately for individuals where mutations are not identified by sequence analysis. Refer to the test requisition or contact the laboratory for more information
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