






Iduronate 2-Sulfatase Enzyme Analysis

	Test Code	B0014
	Test Summary	This test detects iduronate 2-sulfatase activity.
	Turn-Around-Time (TAT)*	3 days
	Acceptable Sample Types	Dried Blood Spots Whole Blood (EDTA)
	Acceptable Billing Types	Self (patient) Payment Institutional Billing

Indications for Testing

This test may be appropriate for individuals with a clinical suspicion of mucopolysaccharidosis type II and/or individuals with a family history of mucopolysaccharidosis type II.

Test Description

This test detects iduronate 2-sulfatase enzyme activity that has been associated with mucopolysaccharidosis type II.

Condition Description

Mucopolysaccharidosis type II is a disease associated with the accumulation of complex carbohydrates in various tissues caused by a lysosomal storage disorder. The disease mainly affects males with age of onset occurring between 2 to 4 years of age. Symptoms usually include full lips, rounded cheeks, a broad nose, enlarged tongue, deep and hoarse voice, sleep apnea, a large head, hydrocephalus, hepatosplenomegaly, umbilical and inguinal hernias, thick skin, skin growths, hearing loss, light sensitivity, reduced vision, weakness in the hands and fingers, spinal stenosis, heart valve abnormalities, short stature, delayed growth, contractures, and thickening of the long bones. More severe forms of the disease can also include symptoms of developmental regression, intellectual decline, rapid disease progression, and a life expectancy of 10-20 years of age. Milder forms of the disease typically do not have affected intelligence but do have a shortened life expectancy in adulthood. The incidence of mucopolysaccharidosis type II is estimated to be ~ 1 in 100,000

Test Methods and Limitations

Tandem mass spectrometry analysis can be coupled with liquid chromatography, a technique referred to as LC-MS/MS. In this methodology, chromatography is used to separate analytes of interest prior to measurement by the mass spectrometer. This separation reduces interference from matrix components and can also allow for the measurement of analytes that have the same mass (isobaric species), thereby improving analytical specificity compared to MS/MS.

Detailed Sample Requirements

Dried Blood Spots

Whole Blood (EDTA)

Collection Container(s):



EDTA (purple top)

Collection:

Infants (< 2-years): 2 to 3 mL; Children (>2-years): 3 to 5 mL; Older children and adults: Minimum 5mL. The blood tube should be inverted several times immediately after blood collection to prevent coagulation.

Sample Condition: Store at ambient temperature. Do not refrigerate or freeze.

Shipping: Ship overnight at ambient temperature ensuring receipt within 5-days of collection.

SPECIAL INSTRUCTIONS: Clotted or hemolyzed samples are not accepted.