






## Glucocerebrosidase (Glucosylceramidase) Enzyme Analysis

	<b>Test Code</b>	B0010
	<b>Test Summary</b>	This test detects glucosylceramidase activity.
	<b>Turn-Around-Time (TAT)*</b>	3 days
	<b>Acceptable Sample Types</b>	Dried Blood Spots Whole Blood (EDTA)
	<b>Acceptable Billing Types</b>	Self (patient) Payment Institutional Billing

### Indications for Testing

This test may be appropriate for individuals with a clinical suspicion of Gaucher disease and/or individuals with a family history of Gaucher disease.

### Test Description

This test detects glucocerebrosidase (glucosylceramidase) enzyme activity that has been associated with Gaucher disease.

### Condition Description

Gaucher disease is associated with accumulation of harmful lipids throughout the body. There are 5 types of Gaucher disease. Type 1 Gaucher disease is the most common form, with a variable age of onset and symptom severity. Symptoms typically include hepatosplenomegaly, anemia, easy bruising, lung disease, and bone abnormalities. Type 2 and Type 3 Gaucher disease both affect the central nervous system and symptoms typically include the symptoms of Type 1 Gaucher disease plus abnormal eye movements, seizures, and brain damage. Type 2 Gaucher disease typically onsets in infancy and rapidly progresses, while Type 3 Gaucher disease typically progresses slower. The perinatal lethal form of Gaucher disease has symptom onset during pregnancy or in early infancy. Symptoms include hydrops fetalis, ichthyosis, skin abnormalities, hepatosplenomegaly, distinct facial features, and severe neurological problems. Death usually occurs a few days after birth. The cardiovascular form of Gaucher disease typically show symptoms of calcified heart valves, eye abnormalities, bone disease, and splenomegaly. The incidence of Gaucher disease is estimated to be ~ 1 in 50,000.

### 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.