






## Psychosine Biochemical Assay

	<b>Test Code</b>	B0028
	<b>Test Summary</b>	This test detects plasma psychosine.
	<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 diagnosing and monitoring disease progression and/or treatment response in individuals with Krabbe disease. Of note, this test can also be used to diagnose and monitor individuals with saposin A cofactor deficiency. Those with saposin A cofactor deficiency will have normal galactocerebrosidase (GALC) activity.

### Test Description

This test detects plasma psychosine activity that has been associated with Krabbe disease. Of note, psychosine is also elevated in saposin A cofactor deficiency which has a similar clinical phenotype to Krabbe disease.

### Condition Description

Krabbe disease is associated with the demyelination of the nervous system and abnormal globoid cells in the brain. Age of onset is typically in infancy, although other forms occur later in life. The symptoms usually include muscle weakness, irritability, feeding difficulties, fever without infection, developmental delay, stiff posture, vision loss, and seizures. The infantile form of the disease causes death by age 2, and life expectancy is decreased in the later onset forms. The incidence of Krabbe disease 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.