






## X-Linked Adrenoleukodystrophy - C26:0 Lysophosphatidylcholine

	<b>Test Code</b>	B0023
	<b>Test Summary</b>	This biochemical test analyzes the level of C26:0-lysophosphatidylcholine
	<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

Individuals with a clinical suspicion of X-linked adrenoleukodystrophy. Individuals with a family history of X-linked adrenoleukodystrophy.

### Test Description

Elevated levels of C26:0-lysophosphatidylcholine is associated with X-linked adrenoleukodystrophy.

### Condition Description

X-linked adrenoleukodystrophy is a disease that primarily affects the nervous system and adrenal glands. There are 3 forms of X-linked adrenoleukodystrophy and the disease occurs mainly in males. The cerebral form of X-linked adrenoleukodystrophy typically has an age of onset in childhood with symptoms of learning difficulty, behavioral problems, vision problems, difficulty swallowing, poor coordination, and impaired adrenal function. Death usually occurs a few years after symptoms begin. The adrenomyeloneuropathy form of X-link adrenoleukodystrophy typically has an age of onset in adulthood with symptoms of progressive stiffness and weakness in the legs, urinary and genital tract disorders, behavioral changes, and adrenocortical insufficiency. In severely affected individuals, the disease can lead to an early death. The Addison disease form of X-linked adrenoleukodystrophy has a variable age of onset with the symptom of adrenocortical insufficiency being the hallmark feature. The disease gradually progresses, and by middle age the symptoms of the adrenomyeloneuropathy form are present. In severely affected individuals, the disease can lead to an early death. It is estimated that the incidence of X-linked adrenoleukodystrophy is ~ 1 in 20,000

### 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.