



Spinal Muscular Atrophy (SMA) Carrier Screening

What is Spinal Muscular Atrophy?

Spinal Muscular Atrophy or SMA is a genetic disorder in which an abnormal gene is passed from parents to their children. It is an illness that does not have a cure, though treatment options are available. SMA affects the part of the nervous system that affects voluntary muscle movements, such as breathing, swallowing, and motor function. It can be a progressive disease leading to loss of muscle strength and possible paralysis. ACOG recommends screening all women considering pregnancy or are currently pregnant.

What is SMA carrier screening?

SMA is a recessive trait, which means that people can carry the gene that causes SMA without actually having the illness. These people are called “carriers”. Genetic screening can be done to rule out being a carrier with specialized blood tests. Carrier frequency is 1/47 Caucasians, 1/67 Ashkenazi Jewish, and 1/59 Asian. SMA is second to Cystic Fibrosis as a cause of death from autosomal recessive gene.

Is testing covered by my insurance?

Although this simple test is highly recommended, insurance coverage varies greatly. For this reason, it is recommended that patients check with their insurance company to see if this test is covered. To check coverage, call the customer service number on the back of your card and give the representative the following information:

Diagnosis Code:

Z31.430

Test Code:

81329

What if my test is negative?

A negative test indicates that your risk of being a SMA carrier is extremely small. No additional testing is recommended for you or the baby’s father.

What if my test is positive?

A positive result indicates that you are a carrier of the abnormal gene that causes SMA. If your test is positive, your doctor will recommend that the baby’s father undergo the same test.

What if I’ve been tested in the past?

If you have had a SMA carrier test in the past, please tell us! Your result will not change, so repeating the test is not necessary. We will simply add your result into your medical record.