Fragile X Syndrome Carrier Screening

What is Fragile X Syndrome?
Fragile X Syndrome 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. Fragile X Syndrome can cause developmental delays, learning disabilities, and social /behavioral problems. ACOG recommends offering this test based on clinical indicators. This testing is not routinely performed; but if requested can be ordered.

What is Fragile X Syndrome carrier screening?
Fragile X can be tested by bloodwork, genetic testing, that looks at your DNA. Males and Females may be carriers. Genetic screening can be done to rule out being a carrier. Screening is indicated for family and personal history of unexplained autism or related disorders such as ADD, autism spectrum disorders, seizures, language disorders, intellectual disability, unexplained early ovarian failure, or insufficiency. Occurrence is 1/4000 males and 1/8000 females.

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:

-If you are not pregnant: Z31.430. (Fragile X screening)
-If you are pregnant: O99.89 (Other specified diseases or conditions complicating pregnancy, childbirth, and puerperium) AND Z13.430 (Fragile X screening)

Test Code: 81243, 81244

What if my test is negative?
A negative test indicates that your risk of being a Fragile X 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 Fragile X. 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 Fragile X carrier screen 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.