# Fragile X Syndrome (Amniotic Fluid)

## Does this test have other names?

FMR1 test, molecular Fragile X diagnosis, Fragile X DNA test

## What is this test?

This test checks a sample of your amniotic fluid to find out if your unborn baby may have fragile X syndrome, a rare genetic disorder.

Fragile X syndrome often causes intellectual disability, behavior and learning challenges, and autistic disorders. This prenatal test isn't commonly done. But you may need this test if you or your partner is a known carrier of the gene mutation that causes it. You may also need this test if you or your partner have the condition. DNA analysis is done on the sample to look for a change to the FMR1 gene, which causes fragile X syndrome.

If there's a chance your child has fragile X syndrome, nothing can be done to prevent it. Knowing that your unborn baby has fragile X syndrome can help you plan accordingly to care for a child with special needs.

A blood test that's similar to this test may be used to help diagnose people with symptoms of fragile X syndrome, or a family history of fragile X syndrome.

## Why do I need this test?

You may need this test if you are pregnant, and you are at risk of having a child with fragile X syndrome. Your child is at risk if you have the syndrome, or you or your partner is a known carrier of the gene that causes it.

## What other tests might I have along with this test?

You may also need other prenatal tests to look for other risk factors.

## What do my test results mean?

Test results may vary depending on your age, gender, health history, and other things. Your test results may be different depending on the lab used. They may not mean you have a problem. Ask your healthcare provider what your test results mean for you.

Normal results are negative, meaning that no gene mutation was found, and your child does not have fragile X syndrome.

A positive result means that changes to the gene were found and that it's likely your unborn baby has the syndrome.

## How is this test done?

This test is done with a sample of amniotic fluid. The sample is collected through a procedure called amniocentesis. The healthcare provider will insert a thin needle through your belly and uterus and into the fluid-filled sac that surrounds the baby. A small sample of amniotic fluid will be removed from the sac.

## Does this test pose any risks?

Amniocentesis carries a slight risk for miscarriage. It also poses a slight possibility that it will cause birth defects if done too early in the pregnancy. You may have minimal cramping during or after the test.

You may need to rest for the remainder of the day after the procedure. You should be able to resume normal activities the next day.

## What might affect my test results?

Other factors aren't likely to affect your results.

## How do I get ready for this test?

You don't need to prepare for this test. Tell your healthcare provider knows about all medicines, herbs, vitamins, and supplements you are taking. This includes medicines that don't need a prescription and any illegal drugs you may use.