

UNDERSTANDING PRENATAL GENETIC TESTING

Most babies are born healthy; however, there is a 3-5% risk in every pregnancy for health problems. There are several genetic tests that will be offered to you and your family during your pregnancy. These tests are offered to try to find some, but not all, health problems that the baby could have.

Understanding these tests can be complicated. The following may guide you as you consider prenatal genetic testing.

What is a prenatal genetic screening test?

A screening test is offered to ALL pregnant women in our practice. Screening tests are **OPTIONAL**; it is up to you whether or not these tests are done. You may choose to do **some, all, or none** of these tests.

Screening tests give a risk, or chance, of a medical condition for the pregnancy. A negative result means that there is a low risk of your baby having the problem being tested. If the result of a screening test is positive, this means that there is a higher chance of a problem with the pregnancy. In this situation, you would be offered further, more certain testing (diagnostic testing). Diagnostic testing gives a “yes” or “no” answer about the baby having a specific problem.

If a woman is considered high risk, such as a woman 35 or older or a woman with a family history of a genetic problem, she may be offered a more certain test (diagnostic test) without first undergoing the screening test.

Which screening tests are offered to all pregnant women in our practice?

All patients in our office are offered screening for the following: Cystic Fibrosis, Fragile X, Spinal Muscular Atrophy (SMA), trisomies 13, 18, and 21 (Down Syndrome). Some women may be screened for sickle cell trait and/or thalassemia depending on their ethnic background.

Does it matter if there is a history of these problems in my or my partner’s family?

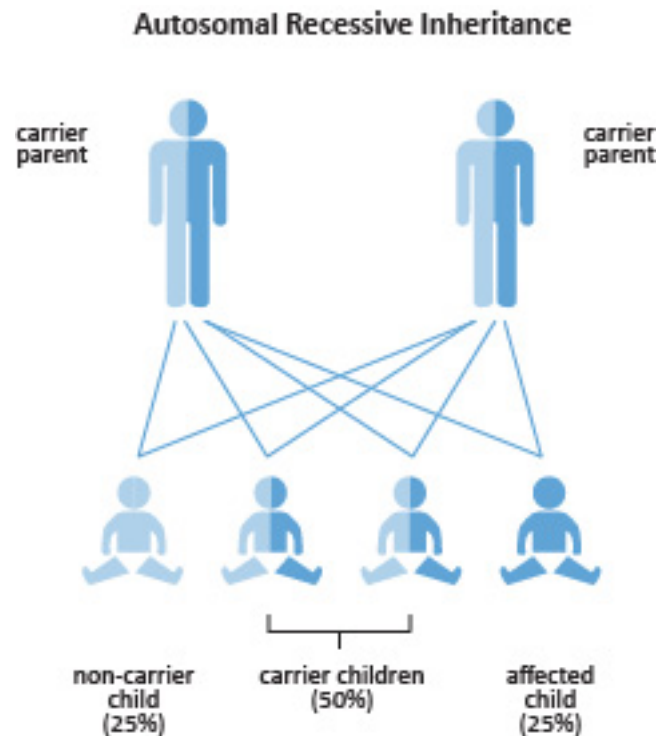
Some problems in babies are considered random, or spontaneous, and not predictable. These problems can happen whether or not there has been someone else in the family with the problem. Spontaneous problems include trisomies 13, 18, and 21 (Down Syndrome).

Other problems are carried in the parents’ genes, and are more likely to happen in certain families. If someone in the family has an inherited disease, the baby may be at higher risk. If there is no family history of the problem, there could still be a possibility of the baby having the disease. Cystic Fibrosis, SMA, and Fragile X are inherited diseases.

What is “carrier” testing?

Cystic Fibrosis, Spinal Muscular Atrophy (SMA), and Fragile X are problems that are almost always caused by one OR both parents having a gene mutation.

Individuals with one gene mutation (carriers) typically do not have symptoms. This person would not know that they have a gene mutation. For diseases such as Cystic Fibrosis and SMA, if both parents have a gene mutation, there is a one in four chance of having an affected child.



I have heard these tests called “non-invasive” what does that mean?

“Non-invasive” means that the test itself does NOT create a risk of losing your pregnancy. Most non-invasive screening tests are a blood test taken from the mother (a simple “blood draw” or stick of the finger).

More certain testing (diagnostic testing such as amniocentesis or chorionic villi sampling) carries a small chance of losing the pregnancy. These tests are considered “invasive” and typically are only done if a patient is at high risk of a genetic problem based on family history, age, or screening test results.

How will the results of genetics testing affect my outlook during pregnancy?

For some women, results of genetic screening tests can decrease anxiety in pregnancy, whether the results are negative or positive. Information obtained may help a family to prepare for after the baby’s birth. Diagnosis of a medical condition with the pregnancy may impact healthcare, childcare, or work decisions for the family.

For some families, results of genetic testing may affect decisions to continue the pregnancy. For these families, genetic testing should be done as early as possible in pregnancy.

For other families, abnormal genetic results can create anxiety and may impact a woman and her family in a negative way. These families may prefer to decline genetic screening tests in pregnancy.

Are there medical treatments that can be offered if I find out my baby has a problem before my baby is born? Will knowing my baby has a problem change my medical care during pregnancy?

Genetic testing results may alter pregnancy medical care, depending on the medical issue. Some medical conditions, such as Cystic Fibrosis, are not treatable prior to the baby being born. For other conditions, surgeries may be done while the baby is still inside the mother, although these surgeries are very rare. In some situations, such as in the case of Down's syndrome, medical monitoring and testing will be offered to decrease risks in the pregnancy.

When can these tests be done?

Some of these tests can only be done at a certain time in pregnancy, while others can be done at any time. **MOST TESTS ARE TYPICALLY DONE WITH YOUR INITIAL BLOODWORK, IF YOU DECLINE THE TESTS AT THIS VISIT IT IS YOUR RESPONSIBILITY TO ASK FOR THE TESTS TO BE DONE AT A LATER TIME.**

Does my insurance pay for these screening tests in pregnancy?

Prenatal genetics screening tests may or may not be paid for by your insurance plan. These tests are not considered "routine"; whether or not these tests are paid depends on your individual insurance plan.

If you are worried about the costs of these tests, it is important that you contact your insurance company about coverage. Information about cost is included in the following pages.

I have a friend who was told her screening test was abnormal, but her baby was fine. I don't want to worry and then have everything be okay.

Screening tests cannot say with 100% certainty whether or not the baby has a problem; however, these tests are much more accurate today than they were a few years ago. If these tests come back showing a chance of a problem with the baby, you will need to decide if you will do further testing (diagnostic testing), which has a small risk of pregnancy loss.

But I thought my ultrasound at 20 weeks would find any problems with the baby.

Some problems can't be seen on ultrasound. Ultrasound will detect about 30% of problems with babies.

What if there is a history of a genetic disease in my family or my partner's family?

Depending on the disease, you may be referred for further counseling with a genetics counselor or high-risk specialist. It is important you complete our genetics screening form, so that we are aware of any problems in your and your partner's families.

If I have had these tests done in a previous pregnancy, do I need to repeat the screening tests?

Certain tests, such as Cystic Fibrosis carrier testing, does not need to be repeated if it was negative in a previous pregnancy. Other testing, such as cell free DNA testing, has a result that only gives information about that specific pregnancy. These screening tests need to be done with each pregnancy.

EVEN IF ALL GENETIC SCREENING TESTS ARE NORMAL, THERE IS STILL A SMALL CHANCE THAT YOUR BABY COULD HAVE GENETIC/CHROMOSOMAL PROBLEMS. THESE TESTS CANNOT RULE OUT EVERY GENETIC PROBLEM. OUR PRACTICE OFFERS SCREENING TESTS FOR THE MOST COMMON GENETIC PROBLEMS. IF YOU WOULD LIKE TO DISCUSS ALL AVAILABLE GENETIC TESTS, PLEASE REQUEST A CONSULTATION WITH A GENETICS SPECIALIST.