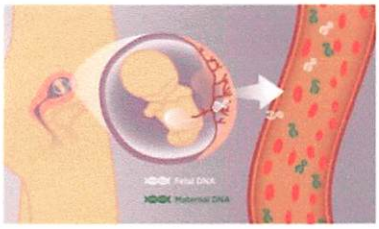
Chromosome Testing at BWH
Center for Fetal Medicine

What is a chromosome screening test?

• Chromosomes are made up of DNA and DNA in specific patterns are called genes. All women are offered the chance to determine if their pregnancy has a different chromosome makeup. One example is Down syndrome, which is due to an extra chromosome number 21.
• Usually chromosome conditions occur by chance and are not inherited. There usually is no family history.
• Chromosome *screening* tests give you an estimate of your risk but not a diagnosis.
•Detecting pregnancies with chromosome conditions lets families be informed and help plan their care. Not every woman chooses to screen or diagnose a chromosome condition during her pregnancy.

What chromosome *screening* tests are available?

• **First Trimester Screening or "First Look"** – combines an early ultrasound with maternal blood studies in the first trimester.
• **Sequential Screening** – combines an early ultrasound with maternal blood studies in the first AND second trimester.
• **Cell-free DNA (cfDNA)** – detects pieces of the genetic code, or DNA, from the placenta in the mother's blood after 10 weeks of pregnancy. This test is most appropriate for women when there is a higher chance of a chromosome condition.



What chromosome diagnostic tests are available?

• **Chorionic villus sampling (CVS) -** a small piece of the placenta (chorionic villi) is removed and sent for testing.
•**Amniocentesis -** a small sample of the fluid around the baby (amniotic fluid) is removed and sent for testing.

Which test is right for me?

You and your provider will discuss the testing that is right for you.

Some details to think about are:
- How soon do you want to know the screening results?
- How far along in your pregnancy are you currently?
- Are you 35 years of age or older?
- Do you have a twin pregnancy?
- Do you, your partner or families have a history of a chromosome condition?

What conditions are part of the screening ?

- Down Syndrome or Trisomy 21
- Edwards Syndrome or Trisomy 18
- Finding other chromosome changes is possible but less accurate.

What if my chromosome screening test is positive?

None of the screening tests can tell for sure that a chromosome condition is present. If a screen is positive, a genetic counselor can review the results and discuss options for more testing.

What if my chromosome screening test is negative?

A negative result lowers, but does not remove the chance to have a child with a chromosome condition.

How much do these tests cost?

In many instances, chromosome screening is covered by insurance. We suggest you check with your insurance to see if preapproval is needed.

A cost-estimator is available: www.counsyl.com/price.

We recommend you view a short, but helpful, explanation of chromosome screening prior to testing:
https://www.counsyl.com/services/informed-pregnancy-screen/noninvasive-prenatal-screening/

To discuss chromosome testing in more detail, genetic counselors are available through Brigham and Women's Hospital.
Appointments for genetic counseling (617) 732-4208

Genetic Carrier Testing at BWH

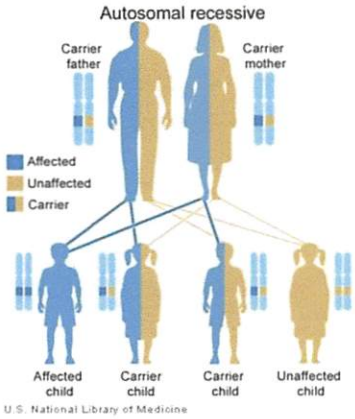
Center for Fetal Medicine

What is genetic carrier testing?

• Genetic carrier testing identifies couples at risk for passing specific genetic conditions to their children.
• A negative test result lowers, but does not eliminate, the chance of being a carrier and having a child with the genetic condition. Changes may be found that are not currently known to cause the genetic condition or testing may not detect all genetic changes.

How are these conditions inherited?

• Genes are the instructions to make our bodies work. We have two copies of each gene in every cell of our body.
• A person is a "carrier" of a recessive genetic condition when there is one working and one changed copy of the same gene. When each parent passes on the changed copy to their child, the child will have the genetic condition.
• Most often carriers have no symptoms of the genetic condition.
• These conditions affect both males and females.



What if I am a carrier?

If you are a carrier for one of these genetic conditions, your doctor or healthcare provider will contact you to go over your results. Genetic counseling is available to discuss your test results, provide information about the condition and arrange carrier testing for your partner, if desired.

Which genetic conditions are commonly screened?

You and your doctor or healthcare provider will decide on the genetic carrier testing that is right for you. Testing is offered based on your race, personal and family history and desire for testing.

Information about you or your partner which is important to share with your provider includes:

❑African, Caribbean, Mediterranean or Asian ancestry
❑Ashkenazi Jewish ancestry
❑French Canadian ancestry
❑Personal or family history of a genetic condition
❑Personal or family history of birth defects (such as cleft lip, clubfeet, heart defects)
❑Personal or family history of intellectual disability, autism or premature ovarian failure/insufficiency
❑You and your partner are related by blood
❑You or your partner are adopted

What about expanded genetic carrier testing?

You can be tested to see if you are a carrier for a larger panel of inherited recessive conditions, referred to as expanded genetic carrier testing. With expanded genetic carrier testing, the chance you are identified as a carrier increases with the number of conditions tested. Expanded genetic carrier testing still only covers a portion of the numerous inherited conditions possible. Expanded genetic carrier panels are not routine.

How much do these tests cost?

In many instances, the cost of genetic carrier testing is reimbursed by insurance. We suggest you confirm your individual coverage or need for preapproval with your insurance carrier.

The laboratory provides a cost-estimator on their website: www.counsyl.com/price.

Prior to testing, we suggest a short, but helpful, explanation of genetic carrier testing:
https://www.counsyl.com/services/family-prep-screen/carrier-screening/

To discuss genetic carrier testing in more detail, genetic counselors are available through Brigham and Women's Hospital.
Appointments for genetic counseling: (617) 732-4208

National Society of Genetic Counselors

Prenatal Cell-Free DNA Screening

Screening for genetic conditions and birth defects is offered to all pregnant women. Some screening methods are routine, such as an ultrasound. Other screening tests are optional, such as blood tests for Down syndrome. Decisions about screening are very personal and should be based on your values and needs. You may find it helpful to discuss your thoughts and feelings about prenatal screening with your obstetric provider. You may also want to talk about how you might use the results. To help guide this discussion, we have outlined some questions to consider.

Should I undergo prenatal screening for genetic conditions?

Based on your needs and values, you may choose whether or not you want these screening tests. Some benefits of prenatal screening might include preparing for raising a child with a health issue or disability. Other benefits might include learning more about the condition or making birth plans. Some families might also want to start treatments as soon as possible after birth or, in some cases, prepare for a baby who may not survive. Some might opt to terminate a pregnancy or create an adoption plan for a child with a disability. Some patients might not want any prenatal screening. They may feel it would not be helpful or that it would cause more stress and worry.

What is cell-free DNA screening?

You may be offered cell-free DNA screening (cfDNA) as a way to screen for some genetic conditions. cfDNA screening might also be called: non-invasive prenatal testing (NIPT), non-invasive prenatal screening (NIPS), or other specific brand names. cfDNA screening looks at a blood sample from a pregnant woman and poses no risk of miscarriage. While this blood test is often accurate, it does not give a definite answer. cfDNA screening can miss pregnancies when the baby does actually have the condition (false negatives). On the other hand, these tests can sometimes show that a baby has high chances for a condition and be wrong (false positives). Therefore, diagnostic testing is recommended for those who want to know for sure. In addition, cfDNA screening does not find all genetic conditions or risk factors possible in a pregnancy. The results should be reviewed with you by a medical professional.

Several other blood tests for Down syndrome may also be offered to you, such as a first trimester screen, second trimester screen, sequential screen, or integrated screen. You should not have more than one type of Down syndrome screening test performed at the same time. Your obstetric provider can discuss the benefits and drawbacks of each option. For questions about these screening tests, please contact your obstetric provider.

What is diagnostic testing, and how is it different from screening?

Diagnostic testing is available to any woman who wants the most accurate testing option. Diagnostic testing is over 99% accurate and can be used to confirm cfDNA screening results. These tests can also detect other genetic conditions not found by screening tests. Depending on the timing during your pregnancy, two diagnostic testing options may be available: chorionic villus sampling (CVS) or amniocentesis. These are invasive tests in which a small sample of tissue from the placenta or amniotic fluid is used to study the baby's chromosomes. Because these procedures are invasive, there is a risk, likely less than 1%, for a miscarriage.

What conditions are screened for by cfDNA screening?

cfDNA almost always screens for Down syndrome, trisomy 18, and trisomy 13. Screening for gender, sex chromosome conditions, and other conditions may also be a part of a cfDNA screen. cfDNA screening is most effective at screening for Down syndrome even though the results are still not certain. However, the accuracy of results for the other conditions is still being studied. The screening options are constantly changing to include

more conditions, so you may want to talk to your medical provider about what is included in the different testing options.

People with chromosome conditions can have a broad range of outcomes. For example, babies with trisomy 13 and 18 often have major health and cognitive issues with only about 10% living past the first year. People with Down syndrome commonly have mild to moderate developmental disabilities and some treatable health issues, with a minority having more severe issues. They are usually active members of their communities and live an average of 60 years. An extra or missing sex chromosome can sometimes cause learning delays and health issues, but can also be so mild that it goes undiagnosed.

How long do results take for cfDNA screening?
cfDNA results are typically available in 5-10 days. You can ask your obstetric provider or genetic counselor how and when you will be getting your results.

What do cfDNA screening results mean?
A "negative" cfDNA result means that the chances your baby has the genetic conditions on the screening panel is low. It does not eliminate the chance and cfDNA does not screen for all conditions. A "positive" cfDNA result greatly increases the chances your baby has a specific condition. Your chances after a "positive" screen depends on many factors: the condition itself, your age, timing during pregnancy, family history, and ultrasound results. Sometimes, a cfDNA result cannot be reported for a number of reasons. When a result cannot be given by cfDNA, you can discuss what this could mean with your genetic counselor or obstetric provider.

How do I get information and support if my screening test comes back "positive" or "high risk" for a genetic condition?
While prenatal screening offers more information about the pregnancy, it can also lead to many more questions. What do these screening results or this diagnosis mean? What kind of life does a person with this diagnosis live? Where can I find reliable information?

Sometimes you might find incorrect or out-of-date information when trying to learn about different conditions. The outcomes and attitudes about many disabilities have changed greatly in recent years. This means expectant parents need current updates about genetic conditions so that they can make informed choices about the pregnancy and find any needed services, resources, and support. Your obstetric provider can show you accurate and trusted resources.

If you would like more information, you can ask for a referral to a genetic counselor. Genetic counselors are health care professionals with training in prenatal genetics. Genetic counselors can help you understand your options and discuss your thoughts about testing. They can also give you accurate information about your test results. You can find a genetic counselor through your obstetric provider or by using the "Find a Genetic Counselor" link on the www.nsgc.org website.

Where can I find more information about prenatal screening and testing?
You can find more information about prenatal screening, testing, and chromosome conditions at www.lettercase.org/prenataltesting/. This pamphlet is an introduction to your prenatal testing options, and you can continue the discussion further with your genetic counselor or obstetric provider.