What conditions can I be screened for?

Insight Medical Genetics currently offers carrier screening for genetic conditions associated with 165 genes. Most of these disorders can be passed on to children only when you and your partner are both carriers of the same disorder. However, 12 of the disorders on the carrier screening panel are typically passed from a healthy carrier mother to her sons. For these disorders only, the father of a baby's carrier status does not matter.

How do I get carrier screening?

The first step is to meet with a genetic counselor to discuss your carrier screening options. The results of the carrier screening you choose to undergo will be available approximately three weeks after your blood draw. You will be contacted with your results and a copy of the results will be sent to you and your doctor.

Call 312-981-4400 to schedule an appointment at one of our Chicagoland locations.

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What are Prenatal Chromosome Screening & Carrier Screening?

Genetic screening is a testing process that provides you with information regarding your risk to have a child with a genetic disorder. The screening process typically involves a consultation with a genetic counselor to discuss screening options, a blood draw, and sometimes an ultrasound.

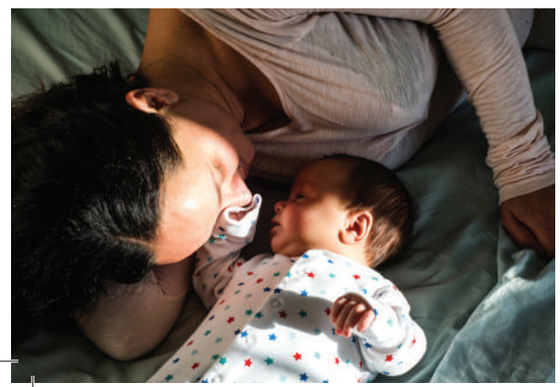
This brochure describes two types of genetic screening – **prenatal chromosome screening, which focuses on the genetic health of a pregnancy, and carrier screening, which focuses on you and your partner.**

Prenatal Chromosome Screening

Prenatal chromosome screening is a blood test (sometimes with an ultrasound) that determines the risk for chromosome abnormalities in a pregnancy. Chromosome abnormalities are genetic changes that typically occur at the moment of conception and are usually sporadic, not inherited. The most common of the chromosome abnormalities is Down syndrome. Insight Medical Genetics offers two different prenatal screening tests that both assesses a pregnancy's risk for chromosome abnormalities – first trimester screening and cell-free DNA (cfDNA) screening. First trimester screening is typically for women under the age of 35 at the time of delivery. cfDNA screening is typically for women 35 or older at the time of delivery, but it is available to women of any age.

Who is screening for?

Because every pregnancy is at-risk for having a chromosome abnormality, regardless of family history or a woman's age, prenatal chromosome screening is available to all pregnant women. In fact, several leading medical professional organizations recommend that prenatal chromosome screening be offered to all pregnant women.



What are first trimester screening and cell-free DNA screening?

First trimester screening combines information from a blood test and ultrasound to provide you with specific risks to your pregnancy for three chromosome abnormalities – Down syndrome (or trisomy 21), trisomy 18, and trisomy 13. The blood test and ultrasound must be performed at approximately 11-13 weeks of pregnancy.

cfDNA screening (sometimes called non-invasive prenatal testing or NIPT) is a blood test that evaluates small fragments of genetic material (DNA) present in your blood. cfDNA screening can tell you whether your pregnancy is at low risk or high risk for many different chromosome abnormalities, including Down syndrome, trisomy 18, trisomy 13, and sex chromosome abnormalities, such as Turner syndrome. This screen can also predict the sex of your baby with 95% accuracy. Although cfDNA screening does not specifically incorporate ultrasound measurements, a first trimester ultrasound is recommended and is typically performed in conjunction with cfDNA screening. cfDNA screening can be performed any time after 10 weeks of pregnancy.

If your prenatal chromosome screening results indicate an increased risk for a chromosome abnormality, you will be offered diagnostic testing to confirm whether or not a chromosome abnormality is present. It is important to remember that screening tests can have both false-positive and false-negative results.

What should I expect at a prenatal chromosome screening appointment?

During your appointment, you will typically view an informational presentation, have an ultrasound, and then meet with a genetic counselor. Your genetic counselor will review all of your screening and diagnostic testing options with you. If you choose to have prenatal chromosome screening, you will then have your blood drawn.

You will be contacted with your results approximately 7-10 days after your blood draw. Your doctor will also receive a copy of your results.

Carrier Screening

Carrier screening is a blood test that determines if you or your partner carry certain changes in your genes (called mutations) that could cause a genetic disorder to be passed on to your children. Carrier screening is ideally done before pregnancy, however, it can also be performed during pregnancy.

Who should have carrier screening?

All people are carriers of multiple genetic disorders. Being a "carrier" of a genetic disorder means that you do not have the disorder, but that you can pass the disorder on to your children if your partner is also a carrier of the same disorder.

Many people feel that if they have no one in their families with a genetic disorder, they do not need to consider carrier screening. However, it is important to know that most babies born with the genetic disorders for which carrier screening is available are the first babies in their families with the disorder. If you or your partner do have a family history of a genetic disorder, or have a family member who is a known carrier of a genetic disorder, the chance of being a carrier of that disorder is increased.

The American Congress of Obstetricians and Gynecologists (ACOG) and the American College of Medical Genetics and Genomics (ACMG) recommend that all couples planning a pregnancy or those already pregnant be offered carrier screening.

Call to schedule an appointment with a genetic counselor to discuss all the details of both screening approaches.