Three Main Categories of Tests

Carrier Screening - blood or saliva test that assesses the chance to pass on select inherited genetic conditions

Screening for chromosomal conditions- blood test that either assesses placental DNA or hormones produced by the placenta to predict the chance for certain chromosomal conditions.

Diagnostic testing - requires either a chorionic villus sampling (CVS) or amniocentesis procedure to be performed, depending on the stage of pregnancy. The procedure involves obtaining a sample of either placenta or amniotic fluid from the pregnancy. With that sample, diagnostic tests can be performed and assess for chromosomal conditions and/or other genetic conditions.

Who We Are

Modern Reproduction's mission is to facilitate the discussion on the current technologies and genetic testing options available prior and during pregnancy by educating, empowering, and embracing modern reproduction. This handout is not medical advice. Please refer to Modern Reproduction's Disclaimer and Terms and Service on their website.

Find Us

www.modernreproduction.org

Follow Us

Instagram

@modernreproduction

Genetic Testing

During Pregnancy

Modern Reproduction

CARRIER SCREENING

Carrier screening is a test that helps individuals or couples understand if they have a higher chance of having a child with an inherited genetic condition. This screening checks specific genes using a blood or saliva sample. When these genes don't work as they should, they can cause a genetic condition. If someone is identified as a carrier, it means they may have a 25% or 50% chance of having a child with that condition, but they themselves are not expected to have the condition.

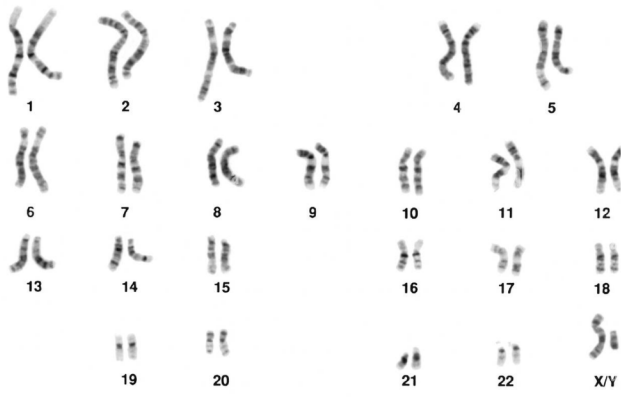
Diagnostic testing is another type of test that can be done during pregnancy or after the baby is born. It checks for the specific condition in the pregnancy.

There are various genetic conditions that can be assessed through carrier screening. Some common examples are Cystic Fibrosis, Sickle Cell Disease, and Spinal Muscular Atrophy. Healthcare providers can guide individuals in deciding which conditions to test for.

CHROMOSOMAL CONDITIONS

Our bodies rely on about 20,000 genes to guide our growth and development. These genes are packed into structures called chromosomes within our cells. Each chromosome carries around 500-1000 genes. Most people have a total of 46 chromosomes. However, sometimes there can be a missing or extra chromosome, which can cause extra or missing genes. This imbalance may lead to conditions like heart defects or intellectual disabilities.

Chromosomal conditions occur by chance during conception, meaning anyone who conceives a pregnancy could have a baby with a chromosomal condition. To assess this possibility, there are screening and diagnostic tests available.



SCREENING

There are two types of screens, both require a blood sample from the pregnant person. Multiple marker screens assess hormones produced by the placenta and evaluate for Down Syndrome, Trisomy 18, and Open Neural Tube defects. Cell-free DNA screening assesses placental DNA for the chance of Down syndrome, Trisomy 13, Trisomy 18, sex chromosomal conditions, and possibly other chromosomal conditions. Screening tests can only estimate the chance of having a certain condition. They cannot provide a definitive diagnosis, so diagnostic testing would be offered.

DIAGNOSTIC TESTING

Certain procedures (CVS or amniocentesis) are performed to obtain a sample from the pregnancy. With that sample, a karyotype or microarray can be performed which provides a more accurate chromosomal evaluation than the screen. If there is a known risk of an inherited genetic condition or if ultrasound identified a birth defect, then select genetic conditions can also be assessed. It's important to note that these procedures carry some risks, which will be discussed by your healthcare provider. Testing after birth is available.