Genetic screening tests explained

What genetic screening means

Genetic screening is a structured way to estimate whether a person, pregnancy, fetus, embryo, or newborn has an increased chance of a genetic condition. In pregnancy care, screening is often offered to people who have no symptoms and no known family history. This is what makes screening different from targeted genetic testing, which is more often used when there is already a specific concern, such as a known familial variant, an abnormal ultrasound, recurrent pregnancy loss, or a previous child with an inherited condition.

Genetic testing is an umbrella term. Depending on the situation, a test may examine DNA sequence, chromosomes, specific gene variants, proteins, enzyme activity, or biochemical markers. Genetic screening is one part of that larger field. Its goal is not to provide certainty in every case, but to sort risk: who appears at lower risk and who may benefit from more detailed discussion, diagnostic testing, or specialist care.

In prenatal care, genetic screening may include carrier screening before pregnancy or early in pregnancy, first-trimester or second-trimester serum screening, ultrasound markers, and Non-invasive prenatal testing \(NIPT\). Some families also encounter preimplantation genetic testing during in vitro

fertilization, although that is performed before transfer of an embryo rather than as routine prenatal screening.

Screening versus diagnostic testing

The most important concept is screening versus diagnostic testing. A screening test gives a probability estimate. A diagnostic test is designed to determine whether a specific condition is present or absent with much greater certainty. In pregnancy, diagnostic tests usually involve obtaining fetal or placental cells through chorionic villus sampling or amniocentesis, followed by laboratory analysis such as karyotype, chromosomal microarray, or targeted molecular testing.

A screening result may be described as low risk, screen negative, increased risk, high risk, positive, no call, or inconclusive. These words can sound more definitive than they are. A high-risk screening result does not mean the fetus definitely has the condition. A low-risk result does not exclude every genetic condition, chromosome change, birth defect, or developmental problem.

False positives and false negatives occur because screening tests infer risk from indirect signals or selected DNA fragments rather than examining every fetal cell. For example, cell-free DNA screening analyzes small DNA fragments in maternal blood, most of which come from the placenta. Because placental and fetal genetic findings are usually but not always identical, results can occasionally be affected by confined placental mosaicism, maternal chromosomal variation, vanishing twin, transplant history, malignancy, or technical limitations. This is why prenatal genetic counseling can be so helpful after an unexpected or complex result.

Carrier screening before or during pregnancy

Carrier screening asks whether a person carries a gene variant associated with an inherited condition, usually one that follows autosomal recessive or X-linked inheritance. Carriers are typically healthy because they have one working copy of the relevant gene, but if both reproductive partners carry variants in the same autosomal recessive gene, each pregnancy may have an increased chance of being affected. For some X-linked conditions, a carrier pregnant person may have a higher chance of having an affected male fetus,

depending on the condition and fetal sex.

Carrier screening may be offered as targeted screening based on ancestry or family history, or as expanded carrier screening that includes many conditions. Examples commonly discussed in clinical practice include cystic fibrosis, spinal muscular atrophy, hemoglobinopathies, and fragile X-related carrier testing in selected situations. The exact panel varies by laboratory and clinical guideline.

Carrier screening is ideally discussed before conception because it allows more time to consider options, including natural conception with prenatal diagnostic testing options, donor gametes, adoption, or IVF with embryo testing in some circumstances. However, many people first encounter carrier screening during prenatal care. If one partner is found to be a carrier, testing the other reproductive partner is often the next step. If both are carriers for the same recessive condition, a genetics professional can explain recurrence risk, test accuracy, and reproductive choices without pressuring the family toward one decision.

Prenatal screening for chromosomal conditions

Prenatal screening for chromosomal conditions estimates the chance of conditions such as trisomy 21, trisomy 18, trisomy 13, and sometimes sex chromosome aneuploidies or selected microdeletions, depending on the test. Traditional first-trimester screening combines maternal age, ultrasound measurement of nuchal translucency, and maternal blood markers. Second-trimester screening may include serum analytes such as alpha-fetoprotein, human chorionic gonadotropin, estriol, and inhibin A. Some approaches combine first- and second-trimester information.

Cell-free DNA screening, often called NIPT, is a blood test usually performed from around 10 weeks of gestation onward, depending on local practice and laboratory requirements. It generally has higher sensitivity and specificity for common trisomies than traditional serum screening, but it remains a screening test. It is not a full fetal genome analysis and does not rule out all chromosome abnormalities or single-gene disorders.

Ultrasound is also part of prenatal risk assessment. A first-trimester scan may

clarify gestational age and identify some major concerns. A second trimester anatomy ultrasound can evaluate fetal structures in detail. Structural anomalies or soft markers may change the interpretation of screening results and may prompt discussion of diagnostic testing. Importantly, normal ultrasound findings do not exclude all genetic conditions, and abnormal findings do not automatically mean a genetic diagnosis is present.

How to interpret results without panic

Genetic screening results are best understood through absolute risk rather than only labels. For example, "high risk" may mean a 1 in 5 chance for one condition in one clinical context, but a 1 in 100 chance in another. The positive predictive value of a test depends on the condition, the person’s baseline risk, gestational age, test performance, and the population being tested. This is why two people with the same laboratory wording may have different real-world probabilities.

Low-risk results can be emotionally reassuring, but they should be interpreted within the test’s scope. A screening panel only addresses the conditions it was designed to assess. It may not detect balanced rearrangements, many copy-number changes, single-gene disorders, imprinting conditions, multifactorial birth defects, or conditions that develop later in pregnancy. Conversely, a high-risk or abnormal result deserves prompt but calm follow-up. In many situations, the next step is not immediate decision-making but confirmatory discussion, review of gestational age, ultrasound evaluation, and consideration of chorionic villus sampling and amniocentesis.

Inconclusive results also happen. A "no call" NIPT result may be related to low fetal fraction, early gestational age, higher maternal body weight, medication or medical factors, fetal or placental chromosome differences, or laboratory issues. Repeating the test may be reasonable in some circumstances, while diagnostic testing may be discussed in others. Your clinician can help determine which path fits the clinical picture.

Who may benefit from genetic counseling

Genetic counseling is not only for families with a known problem. It can support anyone who wants help understanding risk, uncertainty, and choices. A

genetic counselor or clinician with genetics expertise can take a detailed family history, explain inheritance patterns, clarify what a test can and cannot detect, and help interpret results in the context of your values.

Referral is especially useful when there is a family history of inherited disease, a previous pregnancy or child with a genetic condition, consanguinity, recurrent miscarriage, abnormal ultrasound findings, a high-risk screening result, a known parental chromosome rearrangement, or uncertainty about which test is appropriate. Counseling is also valuable when results reveal unexpected information, such as nonpaternity, donor-related findings, maternal chromosomal variation, or a variant whose significance is unclear.

Good counseling should feel non-directive. The purpose is to help you make an informed decision, not to tell you what you should do. Some parents want every available piece of information. Others prefer only screening that would change pregnancy management or newborn planning. Both approaches can be reasonable when made with clear information and professional support.

Questions to ask before choosing a test

Before consenting to screening, it is reasonable to ask exactly what the test includes and excludes. This can prevent surprise later, especially with broad panels or add-on options. Helpful questions include: What conditions are being screened for? Is this a screening or diagnostic test? What are the detection rate and false-positive rate? What happens if the result is high risk, low risk, or inconclusive? Will the result affect pregnancy care, delivery planning, neonatal treatment, or family decision-making?

Ask about timing. Some tests are only informative after a certain gestational age. Others require partner testing, fetal sex information, or ultrasound confirmation of dating and number of fetuses. In twin or higher-order pregnancies, interpretation can be more complex. Ask whether insurance coverage, out-of-pocket cost, turnaround time, and laboratory policies might influence your options.

It is also worth considering emotional readiness. Screening can provide useful information, but it can also create a period of anxiety while waiting for results or confirmation. If you know that uncertain information is particularly

stressful for you, mention this to your care team. They can help you choose a testing strategy that balances medical usefulness with emotional well-being.