Genetic screening before pregnancy

What genetic screening before pregnancy means

Genetic screening before pregnancy usually refers to carrier screening, a blood or saliva test that checks whether a person carries specific pathogenic or likely pathogenic variants associated with inherited conditions. A carrier typically has one working copy and one altered copy of a gene. For many autosomal recessive conditions, carriers have no symptoms because the working copy provides enough gene function.

The clinical significance becomes greater when both egg and sperm contributors carry pathogenic variants in the same autosomal recessive gene. In that situation, each pregnancy generally has a 25% chance of being affected, a 50% chance of resulting in a child who is an unaffected carrier, and a 25% chance of a child who inherits neither familial variant. These probabilities apply to each pregnancy independently; they do not "even out" over time.

Carrier screening can also identify some X-linked conditions, where a variant on the X chromosome may pose particular risks depending on the sex chromosomes inherited by the embryo. In some cases, the person carrying the variant may have mild or variable symptoms themselves. Because inheritance patterns differ, interpreting results with a clinician or genetic counselor is important.

Why screening before conception can be helpful

Timing matters. When carrier screening is done before conception, people often have more space to absorb the information, ask questions, involve a partner if relevant, and consider options without the immediate time constraints of pregnancy. This can be particularly meaningful for conditions that are severe, childhood-onset, life-limiting, or associated with major medical care needs.

Preconception screening may support informed decision-making in several ways:

It can identify carrier couples before pregnancy begins.
It can guide whether a reproductive partner should be tested after one person receives a positive carrier result.
It can help determine whether genetic counseling is appropriate before trying to conceive.
It can inform planning for in vitro fertilization, preimplantation genetic testing, donor sperm, donor eggs, prenatal diagnostic testing, or pediatric specialty care.
It can reduce uncertainty for people with known family histories of genetic disease.

Importantly, screening is optional. Some people want as much information as possible before trying to conceive; others prefer limited testing or choose not to test. A good preconception discussion respects both medical evidence and personal values.

Who should consider carrier screening

Professional guidance commonly supports offering carrier screening to people who are pregnant or planning pregnancy. Testing may be discussed at a preconception visit, fertility consultation, primary care appointment, or obstetric-gynecologic visit. It is not reserved only for people with known risk factors; many carriers have no family history because recessive variants can be silent for generations.

Screening may be especially relevant if:

You or your partner have a family history of an inherited disorder, intellectual disability of unknown cause, congenital anomalies, early childhood deaths, or neuromuscular disease.
You have previously had a child or pregnancy affected by a genetic condition.
You and your partner share ancestry associated with higher carrier frequencies for specific conditions.
You and your partner are biologically related, such as cousins, because shared ancestry can increase the chance of carrying the same rare variant.
You are using donor eggs, donor sperm, or embryos and want to understand genetic compatibility or residual risk.
You are planning IVF and want to know whether preimplantation genetic testing for a monogenic condition may be relevant.

Ethnicity-based screening has historically been used for certain conditions, such as Tay-Sachs disease in people with Ashkenazi Jewish ancestry or hemoglobinopathies in people with ancestry from regions where malaria has been common. However, expanded carrier screening panels are increasingly used because ancestry may be mixed, unknown, or not fully predictive of carrier status.

Types of tests and conditions commonly included

Carrier screening may be targeted or expanded. Targeted screening focuses on specific conditions based on ancestry, family history, or known familial variants. Expanded carrier screening uses a panel that can include dozens to hundreds of genes, often across many ancestry groups. The most appropriate approach depends on clinical context, test availability, insurance coverage, and personal preference.

Conditions often discussed in carrier screening include cystic fibrosis, spinal muscular atrophy, fragile X-related conditions, hemoglobinopathies such as sickle cell disease and thalassemias, and certain metabolic or neurodegenerative disorders. Not every panel includes the same genes, and not every condition has the same severity, onset, treatment options, or predictability.

Before testing, it is reasonable to ask what the panel includes, whether it evaluates only common variants or uses broader sequencing, how variants are classified, whether variants of uncertain significance are reported, and what residual risk remains after a negative result. A medically literate patient may also want to know whether copy number variants, repeat expansions, or pseudogene-related regions are adequately assessed for conditions where those technical issues matter.

How the testing process usually works

The practical steps are usually straightforward. A clinician orders the test, a blood or saliva sample is collected, and a laboratory analyzes selected genes. Results may take several days to a few weeks depending on the laboratory and panel complexity.

There are two common testing strategies. In sequential screening, one partner is tested first. If that person is a carrier for an autosomal recessive condition, the other partner is offered testing for the same gene. In simultaneous screening, both partners are tested at the same time, which can be useful when pregnancy is planned soon or when faster answers are desired.

Results usually fall into broad categories: negative for the variants tested, carrier for one or more conditions, or occasionally a finding that has implications for the tested person's own health. A positive carrier result is not a diagnosis of disease in a future child. It is a risk marker that needs interpretation in relation to the other genetic contributor, the inheritance pattern, and sometimes the exact variants identified.

If one or both partners are carriers

If only one partner is a carrier for an autosomal recessive condition and the other partner tests negative for that same condition, the chance of an affected child is usually substantially reduced, though not zero. This remaining possibility is called residual risk and exists because testing may not detect all pathogenic variants.

If both partners are carriers for the same autosomal recessive condition, or if a significant X-linked risk is identified, the next step is usually genetic counseling. A genetic counselor or specialist can explain the condition, inheritance pattern, variant-specific considerations, reproductive risk, test limitations, and available options.

Possible paths may include:

Trying to conceive without assisted reproductive technology, with or without prenatal diagnostic testing such as chorionic villus sampling or amniocentesis.
Using IVF with preimplantation genetic testing for a specific monogenic condition, sometimes called PGT-M, to help select embryos that are not affected by the condition being tested.
Using donor sperm, donor eggs, or donor embryos to reduce or avoid a specific inherited risk.
Considering adoption or choosing not to pursue pregnancy.
Planning for early neonatal evaluation or specialty care if the couple chooses to conceive and accepts the identified risk.

There is no single "right" decision. The best decision is one made with accurate information, compassionate counseling, and respect for the person’s or couple’s values.

Limitations, uncertainty, and emotional considerations

Genetic screening is powerful, but it is not complete. It does not screen for every genetic disorder, every chromosomal abnormality, every birth defect, or every cause of miscarriage, infertility, neurodevelopmental difference, or childhood disease. Some conditions arise from new, or de novo, variants that are not inherited from either parent. Others involve multifactorial inheritance, where many genes and environmental factors contribute.

A negative result can be reassuring but should not be interpreted as "no genetic risk." A positive result can feel alarming, even when it only indicates carrier status and not illness. People may also feel guilt, blame, grief, confusion, or disagreement with a partner about next steps. These reactions are common and deserve support.

Privacy and insurance questions may also arise. Genetic information can have implications for biological relatives, and regulations vary by country. Before testing, ask how results will be stored, who can access them, whether they become part of the medical record, and whether the laboratory has policies on data sharing or recontact if variant classifications change.

How to prepare for a preconception genetics discussion

Before an appointment, gather as much family history as possible from both sides, ideally across three generations. Note known diagnoses, unexplained infant or childhood deaths, recurrent pregnancy losses, congenital anomalies, intellectual disability, autism with additional medical features, early-onset cancers, neuromuscular conditions, and consanguinity. If a relative has had genetic testing, a copy of the report is more useful than a verbal description of the condition.

Useful questions to bring include:

Which carrier screening approach do you recommend for my situation: targeted, expanded, or both?
Should my partner and I be tested sequentially or simultaneously?
What conditions are included, and how severe or treatable are they?
What does a negative result mean for residual risk?
If we are both carriers, how quickly can we meet with a genetic counselor?
Would IVF with embryo testing, prenatal diagnostic testing, or donor gametes be relevant options for us?

Genetic screening fits best within broader preconception care, including review of medications, chronic conditions, immunization status, folic acid intake, infectious disease risks, and reproductive history. Genetics is one part of preparation, not the whole picture.