Module

GCS_V2_US

Display

Intro to Carrier Screening

Backend Name: GCS_V2_US_01_Intro

Source Video:

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Genetic carrier screening is a test that can be performed prior to pregnancy to assess your risk of having a child with a genetic disease (Moved from spot 4). When you think about genetic disease, you may think about conditions that are passed down from one generation to the next impacting multiple people within a family. These are known as **dominant conditions**. However, there are some types of genetic disease, called **recessive conditions,** that may be present in a family's DNA without any of the family members knowing or being affected.. Carrier screening looks at your DNA, as well as the DNA of your reproductive partner to determine if you have changes in your genes that could put you at an increased risk of having a child with a genetic disease. With this information, you can take action towards having a healthy child.. To better explain how this works, let's review some basic genetics. Our bodies are made up of trillions of tiny cells, and inside these cells are chromosomes. Chromosomes are the structures that contain our DNA -- the hereditary material that provides important instructions for the human body. We each have 23 pairs of chromosomes within our cells; one chromosome in each pair is inherited from the egg and the other from the sperm. Pairs 1-22 are called autosomes. .
The 23rd pair are the sex chromosomes X and Y. Most women have two X chromosomes while most men have one X and one Y chromosome. . Within our chromosomes, certain stretches of DNA are organized into genes . Since we have two copies of every chromosome, we also have two copies of every gene. Changes within these genes are called "variants. " Variants can be inherited or can occur spontaneously. Some variants cause harmless changes, like differences in hair and eye color, but other variants, called mutations, can cause a gene to stop working and may lead to genetic disease.

How Diseases are Inherited

Backend Name: GCS_V2_US_02_Diseases

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Genetic carrier screening specifically looks at recessive conditions. If a person has one working copy and one non-working copy of a gene, they are called a "carrier," which is where the term "carrier screening" comes from.

Carriers typically do not exhibit any symptoms of the condition they carry, and their carrier status typically has no impact on their health or well-being. But when fertilization occurs, the chromosomes and genes from the sperm and egg can combine in four possible variations, each with a 25% likelihood. If the same autosomal recessive condition is present in both the egg and the sperm, there is a 25% chance of having a child who is affected by the condition, meaning they will have a genetic condition that may require a lifetime of specialized care. There is a 50% chance of having a child who is unaffected but is a carrier, so their children may be affected, and a 25% chance of having an unaffected child who neither carries the condition nor is affected by it. Examples of autosomal recessive conditions include: cystic fibrosis, sickle cell anemia, Spinal muscular atrophy, and thalassemia. Genetic Carrier Screening has the potential to determine your carrier status for hundreds of genetic diseases, all from a single DNA sample. . . . .

X Linked Conditions

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There are two types of recessive conditions. Autosomal recessive conditions are caused by changes in genes located on chromosomes 1-22, and these conditions are not found on the sex chromosomes X or Y. X-linked genetic conditions, on the other hand, are caused by changes in genes located on the X chromosome. For a person to be affected with an X-linked condition, they must have no working copies of the affected gene. Because most men only have a single X chromosome, if the gene on that chromosome is non-functional, they do not have a working copy of the gene to compensate and would be affected by the condition. This means that if someone with XY chromosomes has an X-linked genetic disease, it would present with symptoms and they would be aware of the condition. Only someone with two- X ~~XX~~ chromosomes could be an unknowing carrier, which is why carrier screening for X-linked conditions is only performed on people with two-X chromosomes.. . . If you are a carrier of an X-linked condition, your children have a 50% chance of being unaffected, a 25% chance of being an unaffected carrier and a 25% chance of being affected by the condition.. . . .

Examples of X-linked conditions include Fragile X syndrome and Duchenne Muscular Dystrophy.

It's important to note that if you are a carrier of an X-linked condition, it is possible to have issues related to the non-working copy of the gene. For this reason, genetic counseling is very important if you determine that you are a carrier.

How Carrier Screening Works

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To perform carrier screening, a small blood or saliva sample is collected and sent to a specialized genetic testing laboratory for analysis. After a few weeks, your results will be released to your healthcare provider. Your carrier screening results will identify you as a carrier or a non-carrier for each condition tested. If both reproductive partners are identified as carriers for the same autosomal recessive condition,. there is a 25% chance, or 1 in 4, of having a child affected with that condition. Some people opt to test **both** reproductive partners at the same time. This allows more time for decision-making if high risk results are identified, and can reduce anxiety about test results. While others simply test one partner, and then decide about additional testing once those results are received.. This can result in cost savings if additional testing is not necessary, but can increase the time to results and patient anxiety if partner testing is indicated. Your healthcare provider can discuss these options with you in more detail. If you are using an egg and/or sperm donor, be sure to ask whether the donor has undergone carrier screening.

Options for High Risk Results

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If genetic testing identifies you or your reproductive partner as a carrier of the same ~~a~~ genetic disease, you are considered high risk.. One way to manage this risk is with a genetic test called Preimplantation genetic testing for monogenic diseases, or PGT-M. . . . By testing each embryo with PGT-M, you can determine which embryos have the condition in question and may choose to transfer an unaffected embryo. This enables high risk couples to have biological children while significantly reducing their risk of having an affected child. PGT-M is one of many options for those with high-risk results. Your healthcare provider or genetic counselor can discuss all of your options in detail after your carrier screening results are delivered.

While carrier screening technology has advanced significantly, there are still risks that you should be aware of. Carrier screening is not diagnostic but instead is designed to assess risk. A negative result on carrier screening can reduce but not

completely eliminate your risk of being a carrier of a genetic variant. There is always a risk that you may have a disease-causing variant that the test does not find. This risk is known as your “residual risk” after testing. Some residual risk is inevitable with all carrier screening tests as there are some diseases with many mutations, and some mutations may still be unknown. Typically, the most serious and common conditions are included. However, carrier screens do not test for all possible genetic diseases or all possible variants. . Utilizing genetic screening will only reveal those conditions and variants that are tested for, which will vary depending on your testing provider. As with all pregnancies, there is always a risk that one or both reproductive partners have very rare genetic mutation that will not be identified during screening.

Additionally, carrier screens do not test for chromosomal abnormalities like Down syndrome or Turner Syndrome or Multi factorial conditions such as autism or cancer, which can be caused by multiple genetic and non-genetic factors.

The field of genetics and our understanding of variants are continuously evolving as new discoveries are made. For this reason, the information obtained from genetic testing today may vary from what is available in the future. We encourage you to reach out to your healthcare provider if you plan to use information from older genetic testing results in your current reproductive decision-making.

While some residual risk exists, carrier screening technology is an important and effective tool in understanding your likelihood of having a child with an inherited genetic disease, and taking action if necessary. Your medical team and genetic counselors are available to answer any outstanding questions to help you pursue or decline genetic carrier screening. . .