My results were normal/negative. What does that mean?

When it comes to genetic carrier screening, a negative result significantly reduces, but does not eliminate, the chance to be a carrier of those genetic conditions. The chance to be a carrier for these genetic conditions after a negative genetic test is called the residual risk. The residual risk will vary depending on the laboratory that runs the test and the type of testing that was performed. Your provider or genetic counselor can help you determine what your residual risk is after having a negative genetic carrier screening test.

My results were positive, or said that I'm a carrier for a genetic condition. Now what?

To be a carrier for a genetic condition generally means that you have one working and one non-working copy of a gene. Most of these genes follow autosomal recessive inheritance patterns, which mean that someone would have to inherit two non-working copies of a gene in order to develop that genetic condition. For example, if someone inherits a non-working copy of the CFTR gene from both parents, it causes cystic fibrosis. However, if someone carries one non-working copy of a gene, they generally do not have any symptoms. However, they do have a higher chance to have a child that may have that genetic condition.

It is not uncommon to be a carrier for a genetic condition. We have over 20,000 genes, so everyone is likely a carrier for something. The next steps somewhat depend on what condition you were found to be a carrier for. If you were a carrier for a condition with autosomal recessive inheritance, the next step would be to consider carrier testing for your partner to see if they are also a carrier for the same genetic condition. If you and your partner are both found to be carriers for the same autosomal recessive genetic condition, there is a 25% chance for both parents to pass down the non-working copy of the gene, meaning that the baby would have that genetic condition.

If you are found to be a carrier for an X-linked condition, for women the chance to have an affected baby depends on whether the baby is a boy or a girl. If a woman is a carrier for an X-linked condition and is having a boy, there is a 50% chance to pass down the X chromosome with the nonworking gene, which would result in the baby having the genetic condition. There is also a 50% chance to pass down the X chromosome that has the working copy of the gene, which would result in a baby boy that does not have the genetic condition.

If the woman is a carrier for an X-linked condition and is having a girl, there is a 50% chance to pass down the X chromosome with the nonworking gene, which would mean that the baby girl would be a carrier for that genetic condition just like her mom. There is also a

50% chance to pass down the X chromosome that has the working copy of the gene, which would result in a baby girl that is not affected with the genetic condition, and is also not a carrier for the genetic condition.

Both my partner and I are carriers of the same autosomal recessive genetic condition; what does this mean for our baby/potential baby and what are our options?

If you and your partner are both found to be carriers for the same genetic condition, there is a 25% (or 1 in 4) chance that the baby will inherit a nonworking copy of the gene from both parents. That also means that there is a 50% chance that the baby will inherit one working and one non-working copy of the gene (which would make them carriers, just like their parents). There would also be a 25% chance that the baby will inherit a working copy of the gene from both parents.

Finding out that you and your partner are both carriers for the same autosomal recessive condition can be stressful. There are a couple of different options moving forward: either have prenatal diagnostic testing to find out of the baby is affected, or wait until the baby is born and then revisit the option of genetic testing.

Prenatal diagnostic testing, such as chorionic villus sampling (CVS) and aminocentesis, comes with some risk (up to a 1 in 200 (0.5%) to 1 in 300 (0.03%) chance for a complication that could lead to a miscarriage from the procedure), but can provide a 'yes' or 'no' answer as to whether the baby is affected or not. For most genetic conditions, diagnostic testing can be done after the baby is born. The decision of whether to move forward with prenatal diagnostic testing is very personal, and should be done after a talking through the risks, benefits, and limitations with your provider.

I found out that my partner and I are both carriers for the same recessive genetic condition and are concerned about having a child with a genetic condition. What are my options?

The answer to this question may depend on many things, including how severe the condition is, and what your personal thoughts are around having a child with a genetic condition. If you find that you and your partner are carriers for the same recessive genetic condition, there are several options:

- Move forward with pregnancy (or future pregnancy) as planned, and decline further genetic testing

- Move forward with pregnancy (or future pregnancy) and undergo diagnostic testing, such as CVS or amniocentesis, to find out during the pregnancy if the baby has the genetic condition. Some people want to pursue prenatal genetic diagnosis to be prepared for when the baby arrives, while other people want prenatal diagnosis because they would end a pregnancy that was affected. Other people would make plans for adoption if they found out the baby had a genetic condition. No matter the reason, it is important to discuss the risks, benefits, and limitations of diagnostic testing with your provider.
- Pursue in-vitro fertilization (IVF) with preimplantation genetic diagnosis (PGD). IVF with PGD involves fertilizing an embryo in a the lab, and then testing it to see if it has that genetic condition. You would then have the option to only transfer embryos that were not affected with the genetic condition. This process can be expensive, but some insurances may cover part or all of it.
- Consider using a sperm/egg donor
- Consider adoption

These options are not all easy, some of them cost a lot of money, and some of them may not fit with your personal beliefs and values. In terms of emotional support, it may be helpful to connect with another couple who has gone through this process; your provider or genetic counselor may be able to help facilitate this.

Click here to learn more about scheduling a genetic counseling appointment for pregnancy-related questions.

Click here to learn more about scheduling a genetic counseling appointment for infertility or preconception questions.

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