NBS
Newborn
Screening

Igenomix®
WITH SCIENCE ON YOUR SIDE

CARRIER RESULT

What does a carrier result mean?

The test has detected a pathogenic or likely pathogenic variant in one of the genes studied. Generally, we have two copies of each gene. Conditions with an autosomal recessive inheritance present with symptoms when both copies of the gene do not work properly. A person who is a carrier of a recessively inherited condition, or a female that is a carrier of an X-linked condition, have a copy of the gene that does not work properly due to presence of the genetic variant and has a copy of the gene that works normally, which is why that person only "carries the condition" but is not affected by it. In some cases, females who are carriers of an X-linked condition may develop mild symptoms of the condition.

It is important to highlight that when a carrier status of a genetic condition with recessive inheritance is detected, in some cases there may exist a second variant in the gene that has not been detected by our test. This could be due to lack of clinical evidence of this variant, due to it been located in cryptic regions of the genome or due to the limitations of the technology used. In these cases, a residual risk exists of developing the condition, even though the person has only been found to be a carrier. We recommend you share these results with your child's doctor in order to determine if further evaluations are required.

What should I do now?

It is important that you share these results with your child's doctor. Your doctor will inform you if there are any other tests that should be considered.

What implications do these results have on my relatives?

Since the test has detected the presence of a pathogenic or likely pathogenic variant in carrier status in the baby, his or her parents and other relatives may also be carriers of the variants detected. We recommend you share these test results with your relatives and your doctors so that personal risks can be discussed. We recommend genetic counseling and the consideration of genetic studies for any interested relatives.

Where can I get some help?

Genetic counseling is a useful tool to understand genetic test results and their implications. Your doctor may provide genetic counseling or refer you to an independent genetic counsellor. We have genetic counsellors available at Igenomix if you wish to discuss your test results.

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NEGATIVE RESULT

What does a Negative Result mean?

The test has not identified any pathogenic or likely pathogenic variants in the analyzed genes that could be associated with the development of a genetic condition based on its inheritance pattern. The risk to be affected by one of the conditions included in the test is very low, although not zero, due to the possibility of the presence of other variants not yet classified as pathogenic or likely pathogenic. A negative test result does not rule out the possibility of other genetic or nongenetic conditions that are not included in this study and that may present with symptoms in the neonatal period.

A carrier status of pathogenic or likely pathogenic variants in autosomally recesive or X linked inherited conditions will be considered a negative result. If carrier status reporting has been requested, this type of result will be reported as a carrier result.

What should I do now?

We recommend you share these test results with your baby's doctor. The risk of developing a disease is not solely based on our genetic information, therefore it is important for you to develop an appropriate care plan with your doctor.

How can these negative results affect my relatives?

The test results are negative, therefore the risk is not increased for family members.

Where can I get some help?

Even though the test results are negative, you may still have questions. You can contact us at Igenomix or you can speak with your healthcare professional.

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POSITIVE RESULT

What does a positive result mean?

The test has identified one or more pathogenic or likely pathogenic variants in one (or more) of the genes analyzed. The presence of these types of variants, which potentially affect the function of the gene, could be related to the disease or genetic condition associated with this gene, based on its pattern of inheritance. Its possible clinical implications need to be evaluated by a medical professional. The carrier status of pathogenic or likely pathogenic variants in recessively inherited or X linked conditions will not be considered a positive test result.

What should I do now?

It is very important that you share these test results with your baby´s doctor. Your doctor should inform you of the studies and treatments that need to be considered and will prepare a care plan for your child.

What implications do these results have on my relatives?

The baby's family members may also be carriers of the variant(s) that have been detected. We recommend you share these test results with your relatives and your doctors so that personal risks can be discussed. We recommend genetic counseling and the consideration of genetic studies for any interested relatives.

Where can I get some help?

Genetic counseling is a useful tool to understand genetic test results and their implications. Your doctor may provide genetic counseling or refer you to an independent genetic counsellor. We have genetic counsellors available at Igenomix if you wish to discuss your test results.