

Understanding Your ECS Results

Your results are in. Here's how to read them.

Expanded Carrier Screening (ECS) looks at your DNA to see if you are a carrier of any non-working genes associated with recessive genetic conditions. Being a carrier is not the same as having the condition. However, if your partner is also a carrier of the same non-working gene, you will have an increased chance to have a child with a genetic condition.

TYPES OF RESULTS

Negative Result



No disease-causing variants were found in the genes tested. This result is reassuring and reduces, but does not fully eliminate your risk of being a carrier.

Carrier of X-linked Condition



The female partner is a carrier of an X-linked condition. She can pass her variant on to her children, with sons being primarily affected. In rare cases, a female carrier can have mild or atypical symptoms of the condition.

Carrier of Recessive Condition



You are a carrier of a recessive genetic condition. You are not affected by the condition, but you could pass the gene to a child. Your result matters most if your partner is also a carrier of the same condition. Then there would be a 1 in 4 chance to have a child affected with this condition.

Both Partners are Carriers



There is an increased chance to have an affected child with each future pregnancy.

- 25% (1 in 4) chance of an affected child
- 50% (1 in 2) chance of a carrier child
- 25% (1 in 4) chance of an unaffected, non-carrier child

Your Own Health



In some cases, the test may identify a genetic condition that you have, even if you are not currently showing symptoms. It is also worth knowing, although rare, that some carriers do experience mild symptoms.

WHAT THIS MEANS



FOR YOU:

- A carrier result is not a diagnosis. You carry one non-working copy of a gene alongside one working copy, and this combination typically does not cause the condition or require any medical attention.
- Many carriers go through life without ever knowing their carrier status, unless they have genetic testing like this done.



FOR YOUR FAMILY:

- Your parents, siblings, and other blood relatives may also carry the same variant, since it was inherited.
- Sharing your result with close relatives gives them the option to get tested too, especially if they are planning a pregnancy.



FOR YOUR FUTURE CHILDREN:

- If both parents are carriers, your children could be affected (25%), carriers like you (50%), or unaffected and not a carrier (25%).
- If only one parent is a carrier, your child has a 50% chance of being a carrier as well.
- When your children are older, they can choose to have genetic testing to learn more about their carrier status.

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Expanded Carrier Screening (ECS) looks at your genes to see whether you carry a nonworking copy of a gene associated with genetic conditions that can occur in a child when both parents are carriers and the child receives both non-working copies of the gene (recessive), OR when a mother is a carrier of a non working gene on the X chromosome (X-linked). ECS focuses on carrier status or parents-to-be, and is not designed to diagnose genetic disease. Most people who get ECS are healthy and simply want more information to help plan for a future pregnancy.

TYPES OF RESULTS

Negative Result

No disease-causing variants were found in the genes tested.

This result is reassuring and greatly reduces, but does not fully eliminate your risk of being a carrier. Rare variants that the test isn't designed to find may still exist.

Carrier of X-linked Condition



The report identifies the female partner as a carrier for an X-linked condition. She can pass this on to her children, with sons being primarily affected.

Carrier of Recessive Condition



You carry one variant copy of a gene. You are most likely completely healthy.

The report identifies a recessive gene you carry. You are not affected by the condition, but you could pass the gene to a child. Your result matters most if your partner is also a carrier of the same condition. Then there is 1 in 4 chance to have a child affected with this condition.

Both Partners are Carriers



Both partners carry one variant copy of a gene for the same recessive disorder.

- 25% (1 in 4) chance of an affected child
- 50% (1 in 2) chance of a carrier child
- 25% (1 in 4) chance of an unaffected, non-carrier child

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FOR YOUR FAMILY:

- Your parents, siblings, and other blood relatives may also carry the same variant, since it was inherited.
- Sharing your result with close relatives gives them the option to get tested too, especially if they are planning a pregnancy.



FOR YOUR FUTURE CHILDREN:

- Whether this result affects your children's risk depends on your partner's carrier status for the same condition, except for X-linked conditions, where risk depends only on the mother's carrier status.
- Even if only one parent is a carrier, the child has a 50% chance of being a carrier as well.
- These chances apply the same way to every pregnancy, they don't add up or change.