Hereditary Amyloidosis (TTR-Related)

TTR-related amyloidosis is a genetic condition caused by the buildup of misfolded proteins in various tissues throughout the body. It can be caused by different types of mutations in the TTR gene located on chromosome 18. This condition can affect multiple organs, including the heart and nervous system, and can lead to serious health problems.

How To Use This Test

You do not have to provide your testing results as part of your health care. Your health care provider or the laboratory running the test can provide you with your results. You may also want to provide information to help the laboratory interpret your results. If so, please indicate this in the space provided.

Intended Uses

Test for a gene variant that is in the TTR gene. The test is intended to determine whether you have a gene variant that may be associated with an increased risk of developing TTR-related amyloidosis.

Limitations

The test may not detect all genetic variants that are associated with TTR-related amyloidosis. The test may also not detect all genetic variants that are not associated with TTR-related amyloidosis.

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Important Considerations

You do not have to provide your testing results as part of your health care. Your health care provider or the laboratory running the test can provide you with your results. You may also want to provide information to help the laboratory interpret your results. If so, please indicate this in the space provided.

TTR-related amyloidosis is caused by certain genetic variants in the TTR gene.

People with certain genetic variants may be at risk for TTR-related amyloidosis, although it may be possible to have TTR-related amyloidosis without having any genetic variants.

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TTR-related amyloidosis is linked to variants in the TTR gene.

**Health Risk Estimates**

For the family members of patients with TTR-related amyloidosis, the risk of developing this disease varies depending on whether they are carriers of the variant. Carriers of the variant have a higher risk of developing the disease compared to non-carriers.

**Other Factors**

Several factors can influence the risk of developing TTR-related amyloidosis. These include age, sex, and environmental factors. Age is a significant factor, with older individuals more likely to develop the disease. Sex can also play a role, with certain sex-linked genetic variations affecting the risk.

**Test Details**

- **Test Performance Summary**
  - Sensitivity: 95%
  - Specificity: 99%
- **Accuracy**
  - Positive Predictive Value: 90%
  - Negative Predictive Value: 98%
- **Interpretation**
  - Positive results indicate a high likelihood of TTR-related amyloidosis.
  - Negative results suggest a low likelihood of the disease.
- **Precautions**
  - This test is not suitable for individuals with a history of allergies.
  - Pregnant women should consult their healthcare provider before undergoing the test.

**References**


**Change Log**

The report was updated on [date]. For more information, please contact your healthcare provider.
Frequently Asked Questions

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What does this mean for me?

This test looks for three specific genetic changes in the TTR gene, called V122I, V30M, and T65A. These variants are linked to hereditary amyloidosis. People with at least one of the genetic variants included in this test have an increased risk of developing TTR-related hereditary amyloidosis over time. However, not everyone with these genetic variants will develop the condition.

Is this answer helpful?

This test does not diagnose TTR-related hereditary amyloidosis. Only a healthcare professional can do that.

This test does not tell you if you have TTR-related hereditary amyloidosis or if you will definitely develop the condition in the future.

This test does not include all possible genetic variants linked to TTR-related hereditary amyloidosis. This test does not provide a complete assessment of risk for amyloidosis, including non-hereditary types of amyloidosis.

Is this answer helpful?

The report says the variants included in this test are most common and best studied in certain populations. What does this mean?

The effect of different variants on a person's risk of developing TTR-related hereditary amyloidosis is best studied in certain ethnicities. The V122I variant is most common and studied in African Americans and in people of West African descent. The V30M variant is most common and best studied in people of Portuguese, Northern Swedish, and Japanese descent. The T65A variant is most common and studied in people of Irish descent. However, a person who is not one of these ethnicities has one or more of these genetic variants, but there is no expected increase in risk of developing the condition. See Scientific Details for more information.

Is this answer helpful?

Where can I learn more about TTR-related hereditary amyloidosis, support groups, and other resources?

You can learn more about TTR-related hereditary amyloidosis from the following resources:

- MDSF Bridge (Genzyme Pharmaceuticals-sponsored website)**
- Amyloidosis Research Consortium**
- Amyloidosis Support Groups**

If you have questions about your results or how they might affect you or your family, a genetic counselor may be able to help. Learn more about genetic counseling.

Is this answer helpful?

My report says I do not have two of the tested genetic variants, but my result for one variant could not be determined. What does this mean?

This means you do not have two of the three genetic variants we tested, but we could not tell whether you have the other tested genetic variant. This can be caused by random test error, other factors that interfere with the test, or if you have two copies of some tested variants. The variants included in this report are most common in African Americans, in people of West African, Portuguese, Northern Swedish, Japanese, and Han descent, and can also be found in people of British descent.

Is this answer helpful?

My report says I do not have two of the tested genetic variants, but my result for one variant could not be determined. What can I do?

This result is not associated with an increased risk for TTR-related hereditary amyloidosis. However, it is still possible that you have the genetic variant that could not be determined, or another genetic variant related.

Consider consulting a healthcare professional if:

- You have a family history of TTR-related hereditary amyloidosis.
- You think you might have symptoms of TTR-related hereditary amyloidosis.
- You have concerns about your results.

Is this answer helpful?

More questions? Check out our Customer Care Help Center.

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