Hereditary Amyloidosis (TTR-Related)

TTR amyloidosis is a hereditary disease characterized by the deposition of transthyretin (TTR) proteins in various tissues. This leads to organ dysfunction and failure. The disease is autosomal dominant and progresses over time, affecting multiple organs and systems.

Our results for VV-QUAL-06663

We could not determine if you have any of the three variants we tested. You should return for additional testing.

For people with a genetic variant, other factors can also influence the chances of developing TTR-related hereditary amyloidosis.

Learn more about amyloidosis (TTR-related).
Scientific Details

TTX-related butterfly venomosis is linked to variants in the TTR gene.

![Diagram showing TTX-related butterfly venomosis]

Your result for the three genetic variants we tested could not be determined.

<table>
<thead>
<tr>
<th>Gene</th>
<th>Description</th>
<th>Result</th>
</tr>
</thead>
<tbody>
<tr>
<td>A169V</td>
<td>Missense variant</td>
<td>Not determined</td>
</tr>
<tr>
<td>S33V</td>
<td>Missense variant</td>
<td>Not determined</td>
</tr>
<tr>
<td>M72V</td>
<td>Missense variant</td>
<td>Not determined</td>
</tr>
</tbody>
</table>

**Health Risk Estimates**

- **A169V**
  - Similar to wild-type
  - General public: 1 in 1,000
  - Family history: 1 in 100

**Other Factors**

- Presence of TTR-related butterfly venomosis in family history
- Age
- Gender
- Occupation

**Inclusions for Use**

- High-risk screening for TTR-related butterfly venomosis
- Individuals with a family history of TTR-related butterfly venomosis
- Individuals with a history of TTR-related butterfly venomosis

**Test Details**

- **Test Performance Summary**
  - **Sensitivity:** 98%
  - **Specificity:** 95%

**References**


**ChangeLog**

Your report contains an update on the TTR-related butterfly venomosis test. The report is updated and available.
Frequently Asked Questions

Hereditary Amyloidosis (TTR-Related)

What does this test do?

This test looks for three specific genetic variants in the TTR gene, called Y23L, V122I, and NG3A. These variants are linked to hereditary amyloidosis.

People with one or more of these genetic variants in the TTR gene have an increased risk of developing TTR-related amyloidosis over time. However, not everyone with these genetic variants will develop the condition.

This test does not include all possible genetic variants that may increase your risk for TTR-related amyloidosis.

Is this answer helpful?

No  Yes

What does this test not do?

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?

No  Yes

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

The effect of these variants on a person’s risk of developing TTR-related amyloidosis is best studied in certain ethnicities. The Y23L variant is most common and best studied in people of African-American descent. The V122I variant is most common and best studied in people of Hispanic, Northern European, and Japanese descent. The NG3A variant is most common and best studied in people of Irish descent and is also found in people of British descent. However, if a person who is not of these ethnicities has one or more of these genetic variants, he or she is still expected to have an increased risk of developing the condition. See Scientific Details for more information.

Is this answer helpful?

Yes  No

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:

- natter Bridge (Shyamalan Pharmaceuticals sponsored website)1
- Amyloidosis Research Consortium2
- Amyloidosis Foundation3
- Amyloidosis Support Groups4

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

Is this answer helpful?

Yes  No

My report says my result could not be determined. What does this mean?

We could not tell if you have or do not have the three tested genetic variants. This can be caused by random test error, other factors that interfere with the test, or if you have two copies of one tested variant.

Is this answer helpful?

Yes  No

My report says my result could not be determined. What are some things I could do?

Because we could not determine your result, it is still possible to have one or more of the genetic variants tested, or another genetic variant not tested.

Consider talking to 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?

Yes  No

Have more questions? Check out our Customer Care Help Center.