Two Y402H Two A69S Overview Page

Age-Related Macular Degeneration

Age-related macular degeneration (AMD) is the most common cause of irreversible vision loss among older adults. The disease results in damage to the central part of the retina (the macula), impairing vision needed for reading, driving, or even recognizing faces. This test includes the two most common variants associated with an increased risk of developing the condition.

Basic, you have **both** of the genetic variants we tested.

People with this result have an increased risk of developing AMD. Lifestyle and other factors can also affect your risk.

Variants detected

Two copies of each variant detected in both the CFH and ARMS2 genes.

How To Use This Test

This test does not diagnose AMD or any other health conditions.

Please talk to a healthcare professional if this condition runs in your family, you think you might have this condition, or you have any concerns about your results.

Review the Genetic Health Risk tutorial
See Scientific Details
See Frequently Asked Questions

Intended Uses

- Tests for the **Y402H** variant in the CFH gene and the **A69S** variant in the ARMS2 gene associated with an increased risk of developing AMD.

Limitations

- Does **not** test for all possible variants associated with an increased risk of developing AMD.
- Does **not** test for variants in other genes associated with an increased risk of developing AMD.

Important Ethnicities

- The variants included in this test are common in many ethnicities, but are best studied in people of **European** descent.

You have an **increased risk** of developing AMD based on your genetic result.

It is important to discuss this result with a healthcare professional.

We detected two copies of the Y402H variant in the CFH gene and two copies of the A69S variant in the ARMS2 gene.
See Scientific Details

People with these variants are more likely to develop AMD than people without these variants.
This doesn't mean you will definitely develop AMD. Lifestyle and genetic factors not covered by this test can also affect your chances of developing AMD.
See Scientific Details

Since you share DNA with your family members, they may also be interested in this result.
Both of your parents and each of your children likely have both of these variants. Your siblings may also have one or both of these variants.

Lifestyle and other factors can also influence the chances of developing AMD.

Consult with a healthcare professional before making any major lifestyle changes.

Smoking

Smoking is associated with higher risk of developing AMD. If you smoke, quitting is one of the easiest ways to reduce your risk.

See Scientific Details for more information

Smoking | Age | Family history | Ethnicity | Diet

About Age-Related Macular Degeneration

Also known as: Age-related maculopathy, AMD, ARMD

When it develops

AMD is rarely diagnosed in people under the age of 50. Vision loss related to AMD usually becomes noticeable in a person's 60s or 70s and tends to worsen over time.

How common is the condition?

AMD affects about 2% of individuals over the age of 50 in the U.S. Approximately 2 million Americans are currently living with AMD.

Typical signs and symptoms

- Blurred or distorted vision
- Vision loss
- Yellow fatty deposits in the retina called "drusen"
- Blood or fluid leakage in the retina

How it's treated

There is currently no known prevention or cure for AMD. Having regular eye exams can help detect early signs of the condition. Progression of AMD can be slowed with the use of certain treatments and medications.

Read more at: National Eye Institute | NCBI: Age-Related Macular Degeneration | Cleveland Clinic | Genetics Home Reference

It is important to discuss this result with a healthcare professional.

If you have a family history of this condition or think you have symptoms, consult with a healthcare professional.
Print report

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

See our Frequently Asked Questions for more information.
FAQs

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Two Y402H Two A69S Scientific Details Page

Age-Related Macular Degeneration

Scientific Details

Age-related macular degeneration (AMD) is the most common cause of irreversible vision loss among older adults. The disease results in damage to the central part of the retina (the macula), impairing vision needed for reading, driving, or even recognizing faces. This test includes the two most common variants associated with an increased risk of developing the condition.

AMD is associated with variants in many genes.

This report includes one variant in the CFH gene and one variant in the ARMS2 gene. This test does not cover variants in other genes associated with AMD.

CFH | ARMS2

The CFH gene provides instructions for making a protein called complement factor H. This protein is part of the immune system that helps the body fight foreign invaders such as bacteria and viruses. It is important for the body to regulate this system so that healthy cells are not destroyed unnecessarily. Complement factor H, together with other related proteins, helps regulate this system by turning it off when it is not needed.

Read more at Genetics Home Reference

Chromosome 1 | Gene: CFH

You have two copies of both genetic variants we tested.

Variants Detected | View All Tested Markers

Marker Tested	Genotype*		Additional Information
Y402H Gene: CFH Marker: rs1061170	**C** Variant copy from one of your parents	**C** Variant copy from your other parent	Biological explanation; Typical vs. variant DNA sequence(s); Percent of 23andMe customers with variant; References [5, 7, 10, 14, 23, 24, 25, 31, 32] | ClinVar
A69S Gene: ARMS2 Marker: rs10490924	**T** Variant copy from one of your parents	**T** Variant copy from your other parent	Biological explanation; Typical vs. variant DNA sequence(s); Percent of 23andMe customers with variant; References [5, 10, 11, 19, 23, 24, 25] | ClinVar

*This test cannot distinguish which copy you received from which parent. This test also cannot determine whether multiple variants, if detected, were inherited from only one parent or from both parents. This may impact how these variants are passed down.

23andMe always reports genotypes based on the "positive" strand of the human genome reference sequence (build 37). Other sources sometimes report genotypes using the opposite strand.

Test Interpretation

This report provides risk estimates for people of European descent. Estimates for other ethnicities are not currently available.

Health Risk Estimates

Risk estimates are based on clinical studies that identify an association between a genotype and a health condition.

Consider talking to a healthcare professional if you have any concerns about your results.

References [23]

Likelihood ratios | Odds ratios

A "likelihood ratio" estimates how the test result affects the chances of a condition, compared to the chances of the condition prior to testing. In the table below, values greater than 1 mean that the chances of developing AMD are higher based on the test result. Values less than 1 mean that the chances are lower based on the test result. Values close to 1 mean that the chances of developing AMD have not changed significantly.

These values are calculated by 23andMe using data from Rivera et al. (2005).

Genotype	Likelihood ratio	95% confidence interval
No variants detected	0.23	0.17 - 0.30
One copy of Y402H variant	0.50	0.42 - 0.59
One copy of A69S variant	0.67	0.51 - 0.88
Two copies of Y402H variant	1.64	1.25 - 2.14
Two copies of A69S variant	1.99	1.18 - 3.38
One copy of Y402H and one copy of A69S variant	1.24	1.03 - 1.50
One copy of Y402H and two copies of A69S variant	4.12	2.60 - 6.53
Two copies of Y402H and one copy of A69S variant	4.49	3.18 - 6.33
Two copies of Y402H and two copies of A69S variant	21.70	6.87 - 68.50

Other Factors

Other factors besides the variants included in this test can influence your chances of developing AMD.

This is not a complete list of other factors.

People with several risk factors, including having multiple genetic variants, may have a higher risk of developing AMD.

Consult with a healthcare professional before making any major lifestyle changes.

Other Factors	References
Smoking In general, smoking increases the risk of developing AMD. For people who have multiple risk variants, smoking might further increase their risk. Professional guidelines recommend quitting smoking to reduce AMD risk.	[1, 4, 27, 29]
Age The risk of developing AMD increases greatly as a person ages. About 1% of the general U.S. population between the ages of 55 and 70 have AMD. Over the age of 80, 2-14% of people have AMD, depending on ethnicity.	[20]
Family history First-degree relatives of a person with AMD have a higher chance of developing AMD themselves. This may primarily be explained by genetic factors, but could also be related to family members sharing a similar lifestyle.	[12, 13, 28]
Ethnicity People of European descent are more likely to develop AMD than people of other ethnicities. In the US, 2.5% of people of European descent over age 50 have AMD. By comparison, less than 1% of people of African American, Hispanic, and Asian descent over age 50 have the condition.	[20]
Diet Understanding the effects of diet on the risk of AMD is an active area of research. The American Association of Ophthalmology advises individuals to eat healthy foods that have also been shown to benefit eye health. A healthy diet for the eyes emphasizes the consumption of dark green leafy vegetables, citrus fruits, nuts, and whole grains. Consuming healthy fats -- found in fish, nuts, and olive oil -- and minimizing saturated and trans fats are also important.	[3, 6]
Sunlight exposure The effect of sunlight exposure on the risk for AMD is still an active area of research. However, for general eye health, professional organizations recommend wearing sunglasses when outdoors to protect the eyes from harmful exposure to the sun.	[8, 21, 26, 30]
Other genes There are other genes and variants that have been linked to AMD. However, many of these variants may have only a small effect on risk on their own.	[22]

Test Details

Indications for Use

The 23andMe PGS Genetic Health Risk Report for Age-Related Macular Degeneration (AMD) is indicated for reporting of the Y402H variant in the CFH gene and the A69S variant in the ARMS2 gene. This report describes if a person's genetic result is associated with an increased risk of developing AMD, but does not describe a person's overall risk of developing AMD. This report is most relevant for people of European descent.

Special Considerations

- Genetic testing for AMD is not currently recommended by any healthcare professional organizations.

Test Performance Summary

Clinical Performance [9, 27]

- The Y402H variant in the CFH gene is expected to be responsible for approximately 43% of all cases of AMD in older adults.
- The A69S variant in the ARMS2 gene is expected to be responsible for approximately 36% of all cases of AMD in older adults.

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing. All test results were correct. While unlikely, this test may provide false positive or false negative results. For more details on the analytical performance of this test, refer to the package insert.

Warnings and Limitations

- This test does not cover all variants that could cause this condition.*
- This test does not diagnose any health conditions.
- Share results with your healthcare professional for any medical purposes.
- If you are concerned about your results, consult with a healthcare professional.

See the Package Insert for more details on use and performance of this test.

* Variants not included in this test may be very rare, may not be available on our genotyping platform, or may not pass our testing standards.

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Change Log

Your report may occasionally be updated based on new information. This Change Log describes updates and revisions to this report.

Date	Change
July 25, 2017	Age-Related Macular Degeneration report created.

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Two Y402H Two A69S FAQ Page

Age-Related Macular Degeneration

Frequently Asked Questions

Age-related macular degeneration (AMD) is the most common cause of irreversible vision loss among older adults. The disease results in damage to the central part of the retina (the macula), impairing vision needed for reading, driving, or even recognizing faces. This test includes the two most common variants associated with an increased risk of developing the condition.

Age-Related Macular Degeneration

I read that there are two types of AMD, the "dry" type and the "wet" type. Do the risks associated with these variants apply to both types of AMD?

In this report, we do not distinguish the two types of AMD, the "dry" type and the "wet" type. Dry AMD is caused by the buildup of yellow fatty deposits beneath the retina called "drusen." Dry AMD is associated with vision loss that worsens slowly over time. This type of AMD accounts for about 90% of all AMD cases. The remaining 10% of cases are wet AMD, which involves the growth of abnormal blood vessels underneath the macula. When these vessels leak blood and fluid, it damages the macula and makes central vision appear blurry, resulting in severe vision loss that can worsen rapidly.

This test includes the two most common variants associated with an increased risk of developing both "dry" and "wet" AMD. Besides the two variants included in this test, lifestyle and genetic factors not covered by this test can also affect your chances of developing AMD.

You can learn more about AMD from the following resources:

- American Macular Degeneration Foundation
- American Optometric Association (AOA)
- AMD Alliance
- Macular Degeneration Partnership
- Bright Focus

Is this answer helpful? Yes No

What does this test do?

This test looks for two specific genetic variants associated with AMD, one in the CFH gene called Y402H and one in the ARMS2 gene called A69S.

People with multiple genetic variants included in this test are more likely to develop AMD. 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 of developing AMD.

Is this answer helpful? Yes No

What does this test not do?

This test does not diagnose AMD. Only a healthcare professional can do that.

This test does not tell you if you have AMD or if you will definitely develop the condition in the future.

This test does not take into account other risk factors for AMD, such as personal and family health history and smoking history. Thus, this test cannot provide a complete assessment of your risk of developing the condition.

This test does not include all possible genetic variants that may increase your risk of developing AMD.

This test does not include any genetic variants or genes linked to early-onset or juvenile macular degeneration.

Is this answer helpful? Yes No

The report says the variants included in this test are best studied in people of European descent. What if I'm not of European descent?

The tested variants are common in many ethnicities.

However, the effect of these variants on a person's risk of developing AMD is best studied in people of European descent. If a person who is not of European descent has multiple genetic variants, he or she is still expected to have a higher risk of developing AMD, but the exact risk numbers are not well understood. See Scientific Details for more information.

Is this answer helpful? Yes No

Where can I learn more about AMD, support groups, and other resources?

You can learn more about AMD from the following resources:

- American Macular Degeneration Foundation
- American Optometric Association (AOA)
- AMD Alliance
- Macular Degeneration Partnership
- Bright Focus

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? Yes No

My report says variants were detected. What does this mean?

This means you have two copies of each of the two genetic variants we tested.

People with this result are expected to have an increased risk of developing AMD compared to people without either of the tested variants. However, this does not mean you have developed or definitely will develop AMD. Other risk factors such as family history, lifestyle, and other genetic variants not covered by this test can also affect your risk of developing AMD. Learn more about other factors.

Is this answer helpful? Yes No

My report says variants were detected. What are some things I could do?

This genetic result is associated with an increased risk of developing AMD. Consider setting up an appointment with a healthcare professional, such as a physician, optometrist, or a genetic counselor, to discuss this result. This is especially important if you have other family members who have had AMD or if you have other risk factors, such as smoking, that may increase your chances of developing the condition. Learn more about other factors.

The American Academy of Ophthalmology recommends maintaining a healthy lifestyle and following healthy eye habits for protecting your vision. This can include having regular eye exams, wearing sunglasses to protect your eyes from UV rays, avoiding smoking, and eating a healthy diet.

Is this answer helpful? Yes No

What does increased risk mean?

An "increased risk" means that, based on your genetic result for the test, your chances of developing AMD are higher than average. See Scientific Details for more information.

It is important to discuss this result with a healthcare professional, especially if you have other risk factors, such as a family history of AMD or if you smoke.

Genetic variants not covered by this test and non-genetic factors may also influence your risk of developing AMD. Learn more about other factors.

Is this answer helpful? Yes No

How could my result affect my family?

Since you share DNA with your family members, they may also be interested in your result. If you are thinking about talking to family members about your results, see this article for a discussion of things to consider before having the conversation.

Because you have two copies of the Y402H variant and two copies of the A69S variant, it is expected that:

- Each of your children will inherit both of these variants from you.
- Each of your parents has both of these variants.
- Each of your siblings may have one or both of these variants.

Is this answer helpful? Yes No

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

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