Chronic Kidney Disease (APOL1-Related)

Chronic kidney disease is a condition in which the kidneys stop working properly over time. Because the kidneys serve as filters for our blood, chronic kidney disease can cause excess fluid and waste from the blood to build up in the body. This can lead to health problems including bone damage, heart disease, and stroke. This test includes two variants in the APOL1 gene that can increase the risk of developing chronic kidney disease. These variants are most common in people of African descent.

Overview Scientific Details Frequently Asked Questions

Jamie, you have **one** of the two genetic variants we tested.

People with only one variant are not likely at increased risk of developing APOL1-related chronic kidney disease. However, most cases of chronic kidney disease are not caused by the variants included in this test. Your risk for chronic kidney disease also depends on other factors like lifestyle, family history, and genetic variants not covered by this test.

If you have a personal or family history of chronic kidney disease, it's important to talk with a healthcare professional.

How To Use This Test

This test does not diagnose chronic kidney disease 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 S342G and N388_Y389del variants in the APOL1 gene, which are used to define the **G1** and **G2** haplotypes, respectively. These haplotypes are linked to an increased risk for chronic kidney disease.

Limitations

- Does **not** test for variants in other genes linked to chronic kidney disease.
- Does **not** provide information on diabetes-related chronic kidney disease.
- Does **not** account for non-genetic factors, such as lifestyle and family history, that influence the risk for chronic kidney disease.

Ethnicity Considerations

- The variants included in this test are most common and best studied in people of **African** descent.
- These variants are also found in people with African ancestry, including people of **Hispanic** or **Latino** descent.

You are **not likely at increased risk** of developing APOL1-related chronic kidney disease based on your genetic result.

However, people with your result are still at risk for other types of chronic kidney disease due to other factors.

We detected the G1 variant in the APOL1 gene.

However, people typically need two variants or two copies of a variant to have an increased risk for APOL1-related chronic kidney disease.

See Scientific Details

It's estimated that at least 40% of adults in the U.S. will develop chronic kidney disease during their lifetime.

Most cases of chronic kidney disease are not caused by the APOL1 genetic variants in this report. Your risk is also influenced by many other factors, including lifestyle, family history, and other genetic factors.

See Scientific Details

Since you share DNA with your family members, they may also be interested in this result.

Your parents and siblings may also have this variant, and each of your children has a 50% chance of inheriting this variant from you. If your partner also has one or more of the APOL1 variants included in this test, your children could inherit a combination of variants that puts them at increased risk for chronic kidney disease.

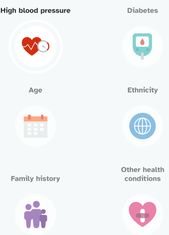
Lifestyle and other factors can also influence the chances of developing chronic kidney disease.

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

High blood pressure

High blood pressure (also called hypertension) is one of the biggest risk factors for chronic kidney disease in adults. High blood pressure can damage the blood vessels inside the kidneys, which can prevent the kidneys from filtering the blood properly.

See Scientific Details for more information



About Chronic Kidney Disease

The information here applies to APOL1-related chronic kidney disease as well as chronic kidney disease due to other factors.

When it develops

In general, the risk of developing chronic kidney disease increases with age. Most cases of chronic kidney disease are diagnosed in people age 65 and over. However, people with APOL1-related chronic kidney disease tend to develop the condition at an earlier age. Their kidney function also tends to decline more quickly than people whose chronic kidney disease is due to other factors.

Typical signs and symptoms

Chronic kidney disease often has no symptoms at first. Early chronic kidney disease is often diagnosed using blood and urine tests that look for loss of kidney function (called reduced glomerular filtration rate) and the presence of protein in the urine (called albuminuria).

- **Symptoms** such as nausea, fatigue, high blood pressure, and swelling of the feet and ankles (called edema) can occur as the condition progresses.
- **Complications** of chronic kidney disease include heart disease or stroke, anemia, bone problems, a weakened immune system, and kidney failure.

How common is the condition?

It's estimated that at least 40% of adults in the U.S. will develop chronic kidney disease during their lifetime. Most people with the condition don't know they have it.

About 13% of African Americans have two APOL1 risk variants, which puts them at an even greater risk for chronic kidney disease.

How it's treated

Treatment for chronic kidney disease depends on the severity of the condition.

- When detected early, chronic kidney disease may be treated in part through lifestyle changes to slow progression. Medications may also be prescribed to treat symptoms.
- If the condition progresses to end-stage kidney disease, patients may require ongoing dialysis or a kidney transplant.

Read more at: National Institute of Diabetes and Digestive and Kidney Diseases* Centers for Disease Control and Prevention* Mayo Clinic*

Learn more about APOL1-related chronic kidney disease.

See our Frequently Asked Questions for more information.

FAQs

If you have a family history of chronic kidney disease 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.

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Chronic Kidney Disease (APOL1-Related)

Chronic kidney disease is a condition in which the kidneys stop working properly over time. Because the kidneys serve as filters for our blood, chronic kidney disease can cause excess fluid and waste from the blood to build up in the body. This can lead to health problems including bone damage, heart disease, and stroke. This test includes two variants in the APOL1 gene that can increase the risk of developing chronic kidney disease. These variants are most common in people of African descent.

Overview | **Scientific Details** | **Frequently Asked Questions**

APOL1-related chronic kidney disease is linked to the G1 and G2 haplotypes.

These haplotypes are defined by variants in the APOL1 gene.

APOL1

The APOL1 gene contains instructions for making a protein that is found in the blood and in multiple organs, including the kidneys. The APOL1 protein plays a role in the immune system, helping the body fight certain parasites called trypanosomes. Although the mechanism is still unknown, certain variants in the APOL1 gene increase the risk for chronic kidney disease, perhaps by causing injury to kidney cells called podocytes.

Read more at NCBI.

You have one of the two genetic variants we tested.

Variants Detected | View All Tested Markers

Marker Tested	Genotype*		Additional Information
G1 **Gene:** APOL1 **Marker** rs73885319	**A** Typical copy from one of your parents	**G** Variant copy from your other parent	✓ **Biological explanation** ✓ **Typical vs. variant DNA sequence(s)** ✓ **Percent of 23andMe customers with variant** ✓ **References** [5, 12, 17, 18, 28, 36, 44, 53, 57] | 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 information about risk for chronic kidney disease in people who have two variants or two copies of a variant included in this test.

Health Risk Estimates

Risk estimates for APOL1-related chronic kidney disease are based on clinical studies that identify an association between a genotype and a health condition.

For reference, information about chronic kidney disease and end-stage kidney disease in the general population is provided.

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

References [2, 9, 11, 12, 13, 14, 15, 17, 18, 25, 28, 39, 34, 39, 46, 47, 48, 52]

General population chronic kidney disease risk | APOL1-related chronic kidney disease risk

About 15% of adults in the U.S. currently have **chronic kidney disease.** It's estimated that at least 49% of adults in the U.S. will develop chronic kidney disease during their lifetime.

Chronic kidney disease can progress to end-stage kidney disease (also called kidney failure). The percentage of people in the U.S. who develop **end-stage kidney disease** during their lifetime varies by ethnicity:

Race/Ethnicity	Male	Female
Non-Hispanic White	3.86%	2.93%
Non-Hispanic Black	8.85%	6.78%
Non-Hispanic Native American	3.69%	3.63%
Non-Hispanic Asian/Pacific Islander	5.95%	3.77%
Hispanic	6.23%	4.32%
Overall	3.96%	2.84%

Other Factors

Other factors besides the genetic variants in this report can influence your chances of developing chronic kidney disease.

This is not a complete list of other factors.

People with multiple risk factors may have a higher risk of developing chronic kidney disease.

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

Other Factors	References
High blood pressure High blood pressure (also called hypertension) is one of the biggest risk factors for chronic kidney disease in adults. High blood pressure can damage the blood vessels inside the kidneys. This can prevent the filtration units inside the kidneys (called nephrons) from functioning properly.	[3, 9, 37, 38, 56]
Diabetes Diabetes is also one of the biggest risk factors for chronic kidney disease in adults and is the most common cause of end-stage kidney disease. Having too much sugar in the blood makes the kidneys work harder to filter the blood, which can damage them over time. Variants in the APOL1 gene do not increase the risk of developing diabetes-related chronic kidney disease. However, people with two APOL1 risk variants are still at risk for this type of chronic kidney disease.	[4, 9, 37, 38, 51, 56]
Age In general, the risk of developing chronic kidney disease increases as a person ages. About 38% of adults age 65 and older have chronic kidney disease, compared to about 7% of adults under the age of 45. As we age, our kidneys stop filtering blood as well. In addition, older adults are more likely to have other risk factors for chronic kidney disease, such as high blood pressure and diabetes. People with APOL1-related chronic kidney disease tend to develop chronic kidney disease at an earlier age than people whose chronic kidney disease is due to other factors. They also tend to progress more quickly from early- to late-stage kidney disease.	[9, 22, 24, 28, 38, 46, 50, 54]
Ethnicity The rates of early chronic kidney disease are similar in most ethnicities in the U.S. However, end-stage kidney disease is more common in certain ethnicities. For example, African Americans are about three times as likely to develop end-stage kidney disease as Americans of European descent. End-stage kidney disease is also more common in Native Americans, Native Hawaiians/Pacific Islanders, and people of Hispanic or Latino descent. Although genetic factors may play a role in explaining these differences, other factors are important, too. For example, differences in the rates of other risk factors for chronic kidney disease — such as high blood pressure and diabetes — likely play a role. Differences in access to healthcare may also contribute. The two APOL1 variants in this report are thought to account for a large proportion of the excess risk for end-stage kidney disease among African Americans.	[2, 8, 9, 11, 16, 38, 41, 43, 49]
Family history People with a family history of chronic kidney disease are more likely to develop the condition themselves. This may be due to shared genetic factors that increase chronic kidney disease risk. In addition, risk factors like diabetes and high blood pressure tend to run in families, and shared lifestyle factors may also contribute.	[1, 7, 19, 38]
Other genetic factors Genetic variants in about 598 different genes have been linked to chronic kidney disease. Many of these variants cause kidney disease in childhood, whereas others are linked to adult-onset kidney disease. In adults, about 15% of end-stage kidney disease cases are thought to be caused by inherited genetic variants.	[10, 19]
Obesity Being overweight or obese increases the risk for chronic kidney disease. This is partly because obesity increases the risk for other important risk factors for chronic kidney disease, like diabetes and high blood pressure. Obesity may also force the kidneys to work too hard, increasing the pressure inside the kidneys and causing damage over time.	[28, 31, 37, 38]
Other health conditions Certain health conditions — such as sickle cell disease, lupus, and HIV — can increase the risk of developing chronic kidney disease. In addition, people who develop heart disease (in particular, heart failure) are more likely to develop end-stage kidney disease. Abnormal kidney structure and a history of acute kidney injury (caused by infection or other environmental factors) can also increase the risk for chronic kidney disease.	[16, 23, 37, 38, 40]
Smoking Smoking cigarettes is associated with an increased risk of developing chronic kidney disease. This may be partly because smoking increases blood pressure, which can damage the blood vessels inside the kidneys. Smoking also increases inflammation and creates molecules called free radicals that can damage the kidneys. The risk is highest in people who smoke many cigarettes and have smoked for a longer time, and risk appears to go down with time after quitting.	[9, 21, 38, 45, 59, 69]
Sex In the U.S., females are more likely to have chronic kidney disease, but males are more likely to develop end-stage kidney disease (also called kidney failure). Chronic kidney disease may be more common in females because they have a longer life expectancy. Hormonal and lifestyle differences may explain why males progress to end-stage kidney disease more quickly than females.	[6, 9, 42]

Test Details

Indications for Use

The 23andME PGS Genetic Health Risk Report for Chronic Kidney Disease (APOL1-Related) is indicated for reporting of the S342G and N388_Y389del variants in the APOL1 gene. These variants define the G1 and G2 haplotypes, respectively. This report describes if a person's genetic result is associated with an increased risk of developing chronic kidney disease, but it does not describe a person's overall risk of developing chronic kidney disease. This report is most relevant for people of African descent.

Special Considerations

- This report does not include the I384M variant in the APOL1 gene, which is part of the G1 haplotype. However, the S342G variant included in this report is often used to define the G1 haplotype in clinical studies and genetic tests. S342G is sufficient to increase risk for chronic kidney disease.
- Genetic testing for APOL1 variants in the general population is not currently recommended by any healthcare professional organizations.

Test Performance Summary

Clinical Performance [11, 16, 49]

- The two variants in this report are thought to account for a large proportion of the excess risk for end-stage kidney disease among African Americans.
- Among African Americans, an estimated 68% of FSGS cases, 68% of HIV-associated nephropathy (HIVAN) cases, and 52% of HIv.ESKD cases can be attributed to having two APOL1 risk variants.
- The G1 and G2 haplotypes are the only APOL1 variants that have been linked to an increased risk for chronic kidney disease.

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing. Greater than 99% of 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 chronic kidney disease.*
- 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 rare, may not be available on our genotyping platform, or may not pass our testing standards.

References

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3. **American Heart Association. "How High Blood Pressure Can Lead to Kidney Damage or Failure." Retrieved Apr 19, 2020, from https://www.heart.org/en/health-topics/high-blood-pressure/health-threats-from-high-blood-pressure/how-high-blood-pressure-can-lead-to-kidney-damage-or-failure.** ^
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7. **Cahalan-Garre M et al. (2019). "Genetic Susceptibility to Chronic Kidney Disease - Some More Pieces for the Heritability Puzzle." Front Genet. 10:453.** ^
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10. **Connaughton DM et al. (2019). "Monogenic causes of chronic kidney disease in adults." Kidney Int. 95(4):914-928.** ^

See all references

Change Log

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

Date	Change
Aug. 5, 2020	Chronic Kidney Disease (APOL1-Related) report created.

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Chronic Kidney Disease (APOL1-Related)

How is APOL1-related chronic kidney disease different from other types of chronic kidney disease?

What does this test do?

What does this test **not** do?

The report says the variants included in this test are most common and best studied in people of **African** descent. What if I'm not of African descent?

Where can I learn more about chronic kidney disease, support groups, and other resources?

My report says **one variant** was detected. What does this mean?

What does **not likely at increased risk** mean?

My report says **one variant** was detected. What are some things I could do?

How could my result affect my family?

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

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