MUTYH-Associated Polyposis

MUTYH-associated polyposis (MAP) is one of the three main hereditary colorectal cancer syndromes. People with two variants or two copies of a variant in the MUTYH gene tend to develop colon and rectal polyps and have an increased risk of developing colorectal cancer. They may also have a slightly increased risk of developing certain other cancers. This test includes two genetic variants in the MUTYH gene that are most common and best studied in people of Northern European descent.

map2_two_G396D, you have an **increased risk** of developing colorectal cancer.

You have two copies of a genetic variant we tested.
People with this result have a higher than average risk of developing colorectal cancer. Risk for certain other cancers may also be slightly increased.

Please share your result with a healthcare professional. It is important to confirm this result in a clinical setting before taking any medical action.

How To Use This Test

This test does not diagnose cancer or any other health conditions and should not be used to make medical decisions. Results should be confirmed in a clinical setting before taking any medical action.

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

Review the MUTYH-Associated Polyposis tutorial

See Frequently Asked Questions

See Scientific Details for complete Indications for Use statement and full list of Warnings, Precautions, and Limitations

Intended Uses

- Tests for the the **Y179C** and **G396D** variants in the MUTYH gene. These variants are linked to MAP, which increases a person's risk of developing certain cancers.
- Provides information on whether a person's genetic result is associated with an increased risk for colorectal cancer and may be associated with a slightly increased risk for certain other cancers.

Limitations

- Does **not** test for all possible variants in the MUTYH gene. More than 100 variants in the MUTYH gene are known to increase colorectal cancer risk. Only two of those variants are included in this test.
- Does **not** test for variants in other genes linked to hereditary colorectal cancer syndromes, such as Lynch syndrome and familial adenomatous polyposis (FAP).
- Does **not** account for non-genetic factors, such as environment and lifestyle, that influence overall cancer risk.

Important Ethnicities

- The variants included in this test are most common and best studied in people of **Northern European** descent. However, these two variants have also been found in other ethnicities.

You have an **increased risk** of developing colorectal cancer based on your result.

It is important to talk with a healthcare professional about options for screening and prevention. It is also important to confirm this result in a clinical setting before taking any medical action.

We detected two copies of the G396D variant in the MUTYH gene.

See Scientific Details

Studies suggest that people with this result have a 43-100% chance of developing colorectal cancer in their lifetime without appropriate surveillance.

However, there are options for screening and prevention that may reduce colorectal cancer risk.

See Scientific Details

Since you share DNA with your family members, they may also wish to learn more about their colorectal cancer risk.

Both of your parents and each of your children likely have this variant. **Your siblings may also be at risk for MAP. If your partner has a variant linked to MAP**, each child has a **50% chance** of having this condition.

If you are thinking about sharing your results with family members, **see this article** for a discussion about things to consider before having the conversation. **Genetic counselors** can help your adult family members decide about genetic testing.

There are things you can do to reduce your risk for colorectal cancer.

People with two variants or two copies of a variant in the MUTYH gene tend to develop colon and rectal polyps and have an increased risk of developing colorectal cancer. They may also have a slightly increased risk for certain other cancers. However, there are options to help manage your risk, so it's important to talk with your doctor about your result. Genetic counseling can also help you understand your results and options. For more information and possible next steps, see this **help article***.

Understand your options for screening and prevention

For people with your genetic result, professional guidelines recommend earlier and more frequent screening for colorectal cancer, along with surveillance for small bowel cancer. This can help identify cancers and precancerous polyps at an early stage. It's important to discuss your result with a healthcare professional to help develop a screening strategy that is best for you.

Maintain a healthy lifestyle

In general, eating a healthy diet, maintaining a healthy weight, engaging in physical activity, limiting alcohol consumption, and avoiding smoking can reduce the risk for colorectal cancer. However, more research is needed to fully understand the impact of these and other lifestyle factors on cancer risk in people with your genetic result.

It is important to confirm your result in a clinical setting before taking any medical action.

See Scientific Details for more information

About MUTYH-Associated Polyposis

MUTYH-associated polyposis (MAP) is a genetic condition where having two variants or two copies of a variant in the MUTYH gene increases a person's chance of developing colorectal cancer. This is because individuals with these variants are prone to developing colon and rectal polyps that, over time, can become cancerous. Variants in the MUTYH gene may also be associated with a slightly increased risk for certain other cancers.

When it develops

Most colorectal cancers start as abnormal growths on the inner lining of the colon or rectum, called polyps. People with MAP tend to develop between ten and a hundred polyps by age 50. These polyps can become cancerous. However, some people with MAP may develop colorectal cancer in the absence of colon or rectal polyps.

Lifetime cancer risks

- Studies suggest that people with MAP have a 43-100% chance of developing colorectal cancer in their lifetime without appropriate surveillance. These individuals may also have a slightly increased risk for certain other cancers.
- See Scientific Details to learn more about these risks

How common is MAP?

MAP accounts for about 1% of all colorectal cancer cases. Between 1 in 10,000 and 1 in 40,000 people of Northern European descent are expected to have MAP.

Screening and prevention

Professional guidelines recommend that individuals with two variants or two copies of a variant in the MUTYH gene should be screened for colon and rectal polyps earlier and more often, and undergo surveillance for small bowel polyps.

Current U.S. guidelines recommend that individuals with one MUTYH variant follow colorectal screening recommendations for the general population. However, for people who have had a first-degree relative with colorectal cancer and people who have a personal history of colorectal polyps (regardless of whether they have a MUTYH variant), these guidelines have different recommendations, which may include screening earlier and more often than the general population.

Read more at: National Cancer Institute* GeneReviews*

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

It's important to consult with a healthcare professional to confirm your result and discuss options for cancer screening and prevention.

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

MUTYH-Associated Polyposis

Scientific Details

MUTYH associated polyposis (MAP) is one of the three main hereditary colorectal cancer syndromes. People with two variants or two copies of a variant in the MUTYH gene tend to develop colon and rectal polyps and have an increased risk of developing colorectal cancer. They may also have a slightly increased risk of developing certain other cancers. This test includes two genetic variants in the MUTYH gene that are most common and best studied in people of Northern European descent.

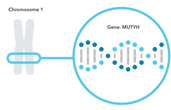
MAP is caused by variants in the MUTYH gene.

This report includes two variants in the MUTYH gene linked to MAP. These two variants account for the majority of the MUTYH variants in people of Northern European descent. However, more than 100 variants in this gene are known to be linked to MAP.

MUTYH

The MUTYH gene contains instructions for making a protein that helps repair damaged DNA. Certain variants in the MUTYH gene disrupt the protein's function. This can lead to a buildup of DNA errors and can cause normal cells to become cancer cells.

Read more at Genetics Home Reference.



You have two copies of a genetic variant we tested.

Variants Detected | View All Tested Markers

Marker Tested	Genotype*			Additional Information
G396D Gene: MUTYH Marker: rs36053993	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 [1, 11, 14, 18, 21, 25, 33, 34, 40] | ClinVar.

* The percent of 23andMe customers with a variant may not be representative of the general population.

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 several cancers associated with MAP. These estimates represent a general risk for individuals with two MUTYH variants or two copies of a MUTYH variant, including but not limited to the two variants included in this report. This test does not take into account non-genetic factors that influence a person's overall risk for these cancers.

Health Risk Estimates

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

Scientists are uncertain as to how having one MUTYH variant may affect the risk of developing colorectal cancer. Some studies suggest that people with one MUTYH variant may have a slightly increased risk, particularly if they have a family history of colorectal cancer. However, more research is needed to understand cancer risks for people with this result.

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

References [6, 25, 28, 32, 37, 39, 40, 41]

Lifetime risk | Risk by age

The risk estimates for colon/rectal and small bowel cancers shown below represent the proportion of people expected to develop these cancers during their lifetime. Estimates for the general population are based on observed cancers among people in the United States. Estimates for people with MUTYH variants are based primarily on studies of people of European descent.

Some studies have also found an association between MUTYH variants and certain other cancers.

Cancer type	General population	Two MUTYH variants	One MUTYH variant
Colon/rectal	4.2%	43-100%	Uncertain to slightly increased risk
Small bowel (duodenal)	<1%	Increased risk	Not available

Other Factors

This is not a complete list of other factors. Except for age, the effects of these factors on cancer risk in people with your genetic result are not as well understood.

People with multiple risk factors may have a higher risk of developing colorectal cancer.

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

Other Factors	References
Age In general, the chances of developing colorectal cancer increase with age. However, for people with MAP, colorectal cancer may develop at an earlier age.	[26, 32]
Family history In the general population, people with a first-degree relative who has been diagnosed with colorectal cancer are about twice as likely to develop colorectal cancer themselves. The effect of family history on colorectal cancer risk in people with MAP is not as well understood.	[4, 9, 16, 40]
Inflammatory bowel disease In the general population, having a chronic inflammatory bowel disease (IBD), such as ulcerative colitis or Crohn's disease, increases a person's chances of developing colorectal cancer. IBD is a condition in which the tissue of the digestive tract is inflamed over a long period of time. Colorectal cancer risk increases with the duration and severity of the inflammation.	[4, 22, 30, 43]
Ethnicity In general, African Americans have a higher risk of developing and dying from colorectal cancer compared to many other ethnic groups. The reasons for this are not well understood, but may be due to a combination of genetic, lifestyle, and socioeconomic factors.	[3, 38]
Obesity In the general population, being overweight increases a person's chances of developing colorectal cancer. This association seems to be stronger in men than in women.	[4, 17, 20, 23]
Type 2 diabetes In general, people with type 2 diabetes may have an increased risk for colorectal cancer. This may be because type 2 diabetes directly influences colorectal cancer risk or because diabetes and colorectal cancer share some of the same risk factors, such as being overweight and having a sedentary lifestyle.	[4, 13, 15, 19, 35]
Physical activity In general, people who regularly engage in physical activity have a lower risk of developing colon cancer than people who rarely or never do. Current U.S. guidelines recommend at least 150 minutes a week of moderate exercise for a healthier life. Learn more from the U.S. Department of Health and Human Services.	[4, 8, 29, 31, 36, 42]
Alcohol consumption In the general population, moderate to heavy alcohol use (more than one drink a day) increases the chances that a person will develop colorectal cancer. This association seems to be stronger in men than in women. The risk also increases with greater alcohol consumption and does not seem to vary by type of alcohol consumed.	[2, 4, 5, 12, 42]
Smoking In the general population, smoking is associated with an increased risk for colorectal cancer. The association seems to be stronger for rectal cancer than colon cancer.	[4, 7]
Diet In the general population, dietary patterns have been found to influence a person's risk for colorectal cancer. Eating a diet high in red meat or processed meat has been associated with an increased risk for colorectal cancer. In contrast, eating a diet with plenty of green leafy vegetables, fruits, whole grains, and healthy fats such as those found in fish, nuts, and olive oil has been associated with many health benefits, including a possible reduction in cancer risk.	[4, 10, 27, 42]

Cancer Screening and Prevention Guidelines

Screening can help detect cancers at an earlier stage, when they may be more treatable. Screening can also detect precancerous polyps, which may allow them to be removed before they become cancerous. The guidelines below contain recommendations for people with **two MUTYH variants** or **two copies of a MUTYH variant**. These guidelines may help you and your doctor create a screening plan that's right for you.

Current guidelines recommend that people with MAP should be screened for colorectal cancer earlier and more often than the general population, using a procedure called colonoscopy. Guidelines also recommend these individuals be screened for small bowel cancer. Your doctor can advise you on the best screening and prevention plan. Learn more from GeneReviews.

The guidelines above represent the colorectal cancer screening recommendations for people with two variants or two copies of a variant in the MUTYH gene. Note that guidelines from different healthcare professional organizations may differ slightly in their recommendations.

For more information and possible next steps, see this **help article**.

Test Details

Indications for Use

The 23andMe Personal Genome Service (PGS) uses qualitative genotyping to detect select clinically relevant variants in genomic DNA isolated from human saliva collected from individuals ≥18 years with the Oragene Dx model OGD500.001 for the purpose of reporting and interpreting genetic health risks, including the 23andMe PGS Genetic Health Risk Report for MUTYH-Associated Polyposis. The 23andMe PGS Genetic Health Risk Report for MUTYH Associated Polyposis is indicated for reporting of the Y179C and the G396D variants in the MUTYH gene. The report describes if a person is at increased risk of developing colorectal cancer. The two variants included in this report are most common and best studied in people of Northern European descent and may not represent the majority of the MUTYH variants found in people of other ethnicities. The test report does not describe a person's overall risk of developing any type of cancer, and the absence of a variant tested does not rule out the presence of other variants that may be cancer-related. This test is not a substitute for visits to a healthcare provider for recommended screenings or appropriate follow-up and should not be used to determine any treatments.

Special Considerations

- Genetic testing for MUTYH variants in the general population is not currently recommended by any healthcare professional organizations.
- Cancer risk associated with MUTYH variants varies from person to person. Overall risk depends on family history and other factors.

Test Performance Summary

Clinical Performance [11, 25, 40]

The two variants included in this report are linked to MAP, which increases a person's risk of developing colorectal cancer and may be associated with a slightly increased risk for certain other cancers. However, most cases of these cancers are not caused by inherited genetic variants.

- Approximately 1% of colorectal cancer are caused by inherited variants in the MUTYH gene.
- The two variants in this report account for 80-90% of cancer-related MUTYH variants among individuals of Northern European descent.
- About 1-2% of the general Northern European population have one of the two variants in this report, which means that between 1 in 10,000 and 1 in 40,000 people of Northern European descent are expected to have MAP. These two variants have also been observed in people of other ethnicities.

Analytical Performance

Accuracy was determined by comparing results from this test with results from sequencing. Greater than 99% of test results were correct. The comprehensive 95% confidence interval for the total number of samples tested was 97.4% to 100%. While unlikely, this test may provide false positive or false negative results. It is possible that the presence of certain mutations in your sample may interfere with the performance of this test. The effects of the interfering mutations on the performance of this test have not been studied. For more details on the analytical performance of this test, refer to the package insert.

Warnings, Precautions, and Limitations

- This test does not diagnose cancer or any other health conditions and cannot determine your overall risk of developing cancer in the future.
- This test should not be used to make medical decisions. Results should be confirmed in a clinical setting before taking any medical action.
- This test does not cover all variants that could increase risk for cancer.* The absence of a variant tested does not rule out the presence of other genetic variants that may impact cancer risk.
- Other factors, such as environmental and lifestyle risk factors, may affect your risk of developing cancer. This test does not account for those factors, and does not test for variants in other genes linked to hereditary cancers.
- Your ethnicity may affect how relevant this test is for you.
- This test is intended to provide you with genetic information to inform conversations with your doctor or other healthcare professional.
- This device is not intended for prenatal testing.
- This test should not be used to assess the presence of genetic variants that may impact response to medications.
- This test is not intended to detect the presence of deterministic variants in autosomal dominant diseases or conditions.
- This test is not a substitute for visits to a healthcare professional for recommended screenings. Consult with a healthcare professional if you have any questions or concerns about your results or your current state of health.
- Some people feel a little anxious after getting genetic health risk results. This is normal. If you feel very anxious, you should speak to your doctor or a genetic counselor.

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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10. Chan DS et al. (2011). "Red and processed meat and colorectal cancer incidence: meta-analysis of prospective studies." PLoS One. 6(6):e20456. *

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
June 27, 2019	MUTYH-Associated Polyposis report created.

MUTYH-Associated Polyposis

Frequently Asked Questions

MUTYH-associated polyposis (MAP) is one of the three main hereditary colorectal cancer syndromes. People with two variants or two copies of a variant in the MUTYH gene tend to develop colon and rectal polyps and have an increased risk of developing colorectal cancer. They may also have a slightly increased risk of developing certain other cancers. This test includes two genetic variants in the MUTYH gene that are most common and best studied in people of Northern European descent.

MUTYH-Associated Polyposis

What does this test do?

This test looks for two specific genetic variants in the MUTYH gene, called Y179C and G396D. These variants are linked to MAP, which increases a person's risk of developing colorectal cancer.

This test provides information on whether a person's genetic result is associated with an increased risk for colorectal cancer and may also be associated with a slightly increased risk for certain other cancers.

This test does not include all possible variants in the MUTYH gene that may increase a person's risk of developing colorectal cancer.

This test does not include variants in other genes that are linked to other hereditary colorectal cancer syndromes, such as Lynch syndrome and familial adenomatous polyposis (FAP).

Is this answer helpful? Yes No

What does this test **not** do?

This test does not diagnose any type of cancer or any other health conditions. Only a healthcare professional can do that.

This test should not be used to make medical decisions. Results should be confirmed in a clinical setting before taking any medical action.

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

This test does not take into account other risk factors for colorectal cancer, such as personal and family health history. Thus, this test does not provide a complete assessment of your overall risk of developing colorectal cancer.

This test does not include all possible variants in the MUTYH gene that may increase a person's risk of developing colorectal cancer.

This test does not include variants in other genes that are linked to other hereditary colorectal cancer syndromes, such as Lynch syndrome and familial adenomatous polyposis (FAP).

Is this answer helpful? Yes No

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

Even though these two variants are most common in people of Northern European descent, they have also been observed in people of other ethnicities.

Similarly, even though the effect of these variants on a person's risk of developing colorectal cancer is best understood in people of Northern European descent, the effect is expected to be similar in people of other ethnicities. For example, if a person who is not of Northern European descent has both of the variants included in this report, he/she is still expected to have a similar elevated risk of developing colorectal cancer. **See Scientific Details for more information.**

Is this answer helpful? Yes No

Where can I learn more about MAP and colorectal cancer, support groups, and other resources?

You can learn more about **MAP** from the following resources:

- **Cancer.net (American Society of Clinical Oncology)***

You can learn more about **colorectal cancer** from the following resources:

- **American Cancer Society***
- **Colorectal Cancer Alliance***
- **Fight Colorectal Cancer***

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.**

You can review the MUTYH-Associated Polyposis tutorial **here***.

Is this answer helpful? Yes No

My report says **two copies of a variant** were detected. What does this mean?

This means you have two copies of a genetic variant we tested.

People with this result have an increased risk of developing colorectal cancer and may have a slightly increased risk for certain other cancers. However, there are options for screening and prevention that may reduce cancer risk.

It is important to discuss this result with a healthcare professional, who can provide information about options for screening and prevention. It is also important to confirm this result in a clinical setting before taking any medical action.

Is this answer helpful? Yes No

What does **increased risk** mean?

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

People with two MUTYH variants or two copies of a MUTYH variant tend to develop colon and rectal polyps, which can become cancerous. Studies suggest that people with this result have a 43-100% chance of developing colorectal cancer during their lifetime without appropriate surveillance. Risk for certain other cancers may also be slightly increased. However, there are options for screening and prevention that may reduce colorectal cancer risk.

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

Is this answer helpful? Yes No

My report says **two copies of a variant** detected. What are some things I could do?

This result is associated with an increased risk of developing colorectal cancer. It is important to share this result with a healthcare professional, such as a doctor or a genetic counselor.

Professional guidelines recommend that people with MAP should be screened for colorectal cancer earlier and more often using a procedure called colonoscopy. Guidelines also recommend these individuals be screened for small bowel cancer. Your doctor can advise you on the best screening and prevention plan. Learn more from **GeneReviews***.

It is important to discuss your result with a healthcare professional. **Results should be confirmed in a clinical setting before taking any medical action.**

For more information and possible next steps, see this **help article***.

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 a variant, it is expected that:

- Each of your children will inherit this variant from you. If your partner has a variant linked to MAP, each child has a 50% chance of having this condition.
- Each of your parents has at least one copy of this variant.
- Each of your siblings has at least a 25% chance of having two copies of this variant, which means they would have an increased risk of developing colorectal cancer.

Because the variant we detected is associated with an increased risk for colorectal cancer, your adult family members may wish to learn more about their own colorectal cancer risk. They can talk with a healthcare professional, such as a doctor or genetic counselor, to help them decide if genetic testing is right for them. **Learn more about genetic counseling.**

Is this answer helpful? Yes No

I have questions about my results. Who should I talk to?

It's normal to have questions or concerns after viewing this report. Some people feel anxious, upset, or worried about their risk or risk for their family members. Others simply want to understand their results better or talk to someone about what they can do. Genetic counselors can help. Genetic counselors are healthcare professionals with special training in genetics and genetic testing. **Learn more about genetic counseling.**

For more information and possible next steps, see this **help article***.

Because you have two copies of a variant detected, it is also important to talk with a healthcare professional about your result and options.

Is this answer helpful? Yes No