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_total_nocall, we could **not determine** if you have the two genetic variants we tested.

This test is intended to detect two variants in the MUTYH gene, but your result could not be determined.

This can be caused by random test error or other factors that interfere with the test.
If you have a personal or family history of colorectal cancer or colorectal polyps, you should talk to a healthcare professional, who may recommend additional screening or genetic testing options for you.

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.

We could **not determine** if you have either of the two variants we tested linked to MAP.

If you have a personal or family history of colorectal cancer, consider talking with a healthcare professional about additional testing.

We could not rule out either of the two variants we tested.

These variants are most common and best studied in people of **Northern European** descent and may not account for the majority of MUTYH variants in people of other ethnicities.

See Scientific Details

In the general population, about **1 in 25** people will be diagnosed with colorectal cancer during their lifetime.

Less than 1% of colorectal cancer cases are caused by the two genetic variants in this report. Your risk is influenced by many other factors, including lifestyle, family history, and other genetic factors.

See Scientific Details

If you have a personal or family history of colorectal cancer or multiple colorectal polyps, talk to a healthcare professional about other testing options.

A genetic counselor can help you assess your overall cancer risk. **Learn more about genetic counseling.**

Lifestyle, family history, and other factors can also influence the chances of developing colorectal cancer.

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

Ethnicity

African Americans have a higher risk of developing colorectal cancer compared to many other ethnic groups.

See Scientific Details for more information

Age
Family history
Other genetic variants
Inflammatory bowel disease
Ethnicity
Lifestyle

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*

Learn more about MAP and colorectal cancer.

See our Frequently Asked Questions for more information.

FAQs

If you have a personal or family history of colorectal cancer or colorectal polyps, consult with a healthcare professional.

Print report

Learn more about colorectal cancer screening to help you and your doctor create a screening plan that's right for you.

Learn more

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.

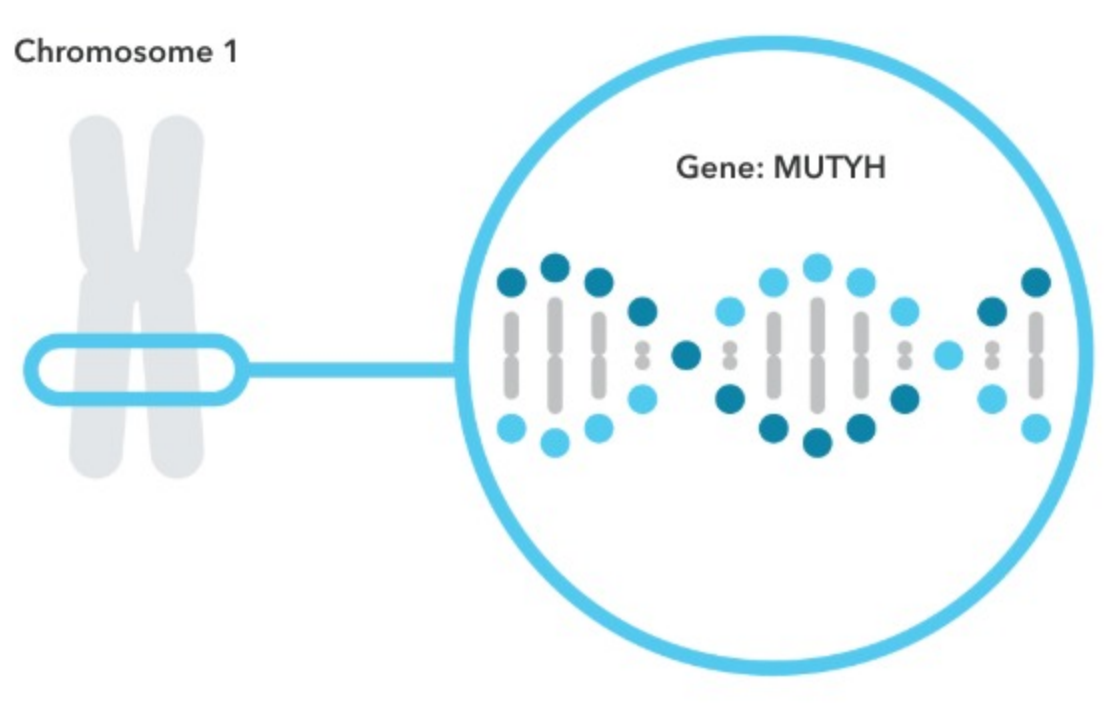
MAP is caused by variants in the MUTYH gene.

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



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

Variants Detected | View All Tested Markers

Marker Tested	Your Genotype*	Additional Information
Y179C Gene: MUTYH Marker: rs34612342	**Not determined**	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
G396D Gene: MUTYH Marker: rs36053993	**Not determined**	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, 33, 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

Having two genetic variants or two copies of a variant in this report is associated with an increased risk for colorectal cancer. However, other factors besides the genetic variants in this report can influence your chances of developing colorectal cancer.

This is not a complete list of other factors.

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 Like most cancers, the risk of developing colorectal cancer generally increases with age. For the average person in the U.S., the risk of developing colorectal cancer is about 1 in 300 by age 50. That number rises to about 1 in 25 by age 80.	[26, 32]
Family history People with a first-degree relative who has been diagnosed with colorectal cancer are about twice as likely to develop colorectal cancer themselves, compared to the general population. The risk is even higher if that relative was diagnosed before the age of 45 or if there are two or more first-degree relatives who have developed colorectal cancer.	[4, 9, 16, 40]
Other genetic variants Many studies have identified additional variants in the MUTYH gene linked to MAP. In addition, variants in other genes can also increase colorectal cancer risk, including genes linked to Lynch syndrome and familial adenomatous polyposis (FAP).	[4, 24, 25]
Inflammatory bowel disease 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 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 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 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 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 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 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 general, 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 Guidelines

Screening can help detect cancers at an earlier stage, when they may be more treatable. Colorectal cancer screening can also detect precancerous polyps, which may allow them to be removed before they become cancerous. The guidelines below contain recommendations for people in the **general population**. These guidelines may help you and your doctor create a screening plan that's right for you.

Current U.S. guidelines recommend that people in the general population should start regular colorectal cancer screening at age 50. Screening can be done through a visual examination that looks at the colon and rectum, such as a colonoscopy exam, or through a stool-based test that looks for signs of cancer. Depending on the test, screening should be repeated every 1 to 10 years. Learn more from the **U.S. Preventive Services Task Force**.

Consider talking to a doctor if you have a family history of colorectal cancer or a personal history of colorectal polyps. Your doctor may have specific screening recommendations for you, such as earlier or more frequent screening.

The guidelines above represent colorectal cancer screening recommendations for the general population. Note that guidelines from different healthcare professional organizations may differ slightly in their recommendations.

Keep in mind that you could still have a variant in the MUTYH gene or other genes not included in this report that could affect your colorectal cancer risk. In that case, different screening and preventive actions may be recommended. Consult with a healthcare professional to learn more.

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

1. Ali M et al. (2008). "Characterization of mutant MUTYH proteins associated with familial colorectal cancer." Gastroenterology. 135(2):499-507.
2. Allen NE et al. (2009). "Moderate alcohol intake and cancer incidence in women." J Natl Cancer Inst. 101(5):296-305.
3. American Cancer Society. "Cancer Facts & Figures for African Americans 2016-2018." Atlanta: American Cancer Society, 2016. Retrieved Mar 12, 2019, from https://www.cancer.org/content/dam/cancer-org/research/cancer-facts-and-statistics/cancer-facts-and-figures-for-african-americans/cancer-facts-and-figures-for-african-americans-2016-2018.pdf
4. American Cancer Society. "Colorectal Cancer Facts & Figures 2017-2019." Atlanta: American Cancer Society, 2017. Retrieved Aug 20, 2018, from https://www.cancer.org/content/dam/cancer-org/research/cancer-facts-and-statistics/colorectal-cancer-facts-and-figures/colorectal-cancer-facts-and-figures-2017-2019.pdf
5. Bagnardi V et al. (2015). "Alcohol consumption and site-specific cancer risk: a comprehensive dose-response meta-analysis." Br J Cancer. 112(3):580-93.
6. Barnetson RA et al. (2007). "Germline mutation prevalence in the base excision repair gene, MYH, in patients with endometrial cancer." Clin Genet. 72(6):551-5.
7. Botteri E et al. (2008). "Smoking and colorectal cancer: a meta-analysis." JAMA. 300(23):2765-78.
8. Boyle T et al. (2012). "Physical activity and risks of proximal and distal colon cancers: a systematic review and meta-analysis." J Natl Cancer Inst. 104(20):1548-61.
9. Butterworth AS et al. (2006). "Relative and absolute risk of colorectal cancer for individuals with a family history: a meta-analysis." Eur J Cancer. 42(2):216-27.
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
[redacted]	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 my result could not be determined. What does this mean?

This means we could not tell if you have or do not have the two genetic variants we tested. This can be caused by random test error or other factors that interfere with the test.

More than 100 variants in the MUTYH gene have been linked to MAP, which increases the risk for colorectal cancer. This report only includes two of those variants. In addition, this test does not include variants in other genes that can also increase colorectal cancer risk, including genes linked to Lynch syndrome and familial adenomatous polyposis (FAP). So you could still have a variant that is not included in this test.

About 1 in 25 people will be diagnosed with colorectal cancer during their lifetime, and the majority of these cases are not caused by inherited genetic variants. This means that other factors such as lifestyle, age, and family history are also important. For example, the risk for colorectal cancer is higher in people with a family history of colorectal cancer. **Learn more about other factors.**

Is this answer helpful? Yes No

My report says my result could not be determined, but I have a personal or family history of colorectal cancer. What does this mean for me?

People with a family history of colorectal cancer have a higher risk of developing colorectal cancer themselves. Specifically, people with a first-degree relative who has been diagnosed with colorectal cancer are about twice as likely to develop colorectal cancer themselves, compared to the general population. The risk is even higher if that relative was diagnosed before the age of 45 or if there are two or more first-degree relatives who have developed colorectal cancer.

We could not determine whether you have the two tested variants, so we can't tell you whether these variants are contributing to your family history. More than 100 variants in the MUTYH gene have been linked to MAP, which increases the risk for colorectal cancer. This report only includes two of those variants. In addition, this test does not include variants in other genes that can also increase colorectal cancer risk, including genes linked to Lynch syndrome and familial adenomatous polyposis (FAP).

Other non-genetic factors may also influence your risk of developing colorectal cancer, even if you do not have any genetic variants. **Learn more about other factors.**

It is important to discuss your personal or family history of cancer with a healthcare professional, who can help you determine if additional genetic testing is appropriate. Genetic counseling can also help you understand your results and your options for additional testing. **Learn more about genetic counseling.**

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 both of the genetic variants tested or another genetic variant not tested. So your result doesn't give you any new information about your risk for colorectal cancer.

There are many other genetic and non-genetic factors that can affect your risk, which this test does not take into account. **Learn more about other factors.**

It is important to continue with any colorectal cancer screenings your healthcare provider recommends. Current U.S guidelines recommend that people in the general population should start regular screening at age 50. Learn more from the **U.S. Preventive Services Task Force.**

If you have a family history of colorectal cancer or a personal history of colorectal polyps, consider discussing your result with a healthcare professional to learn more about options for screening and prevention. Your doctor may have specific screening recommendations for you, such as earlier or more frequent screening.

Talk to a healthcare professional if:

- You have a personal or family history of cancer.
- You think you might have colorectal polyps, colorectal cancer, or any other type of cancer.
- You have questions about other risk factors you may have.

Is this answer helpful? Yes No

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