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.

Overview Scientific Details Frequently Asked Questions

Jamie, you do not have the two genetic variants we tested.

However, this test does not include the majority of variants known to increase colorectal cancer risk. In addition, most cases of colorectal cancer are not caused by inherited variants, so people without a variant are still at risk of developing colorectal cancer. It's important to continue with any cancer screenings your healthcare provider recommends.

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 199 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 **do not have** the two variants we tested linked to MAP.

However, people without these variants are still at risk for colorectal cancer because most cases of colorectal cancer are caused by other factors.

You could still have a variant not included in this test.

More than 199 variants in the MUTYH gene have been linked to MAP, which increases the risk for colorectal cancer. This test only includes two of those variants. In addition, this test does not include variants in other genes which can also increase colorectal cancer risk, including genes linked to Lynch syndrome and familial adenomatous polyposis (FAP).

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.

Age

The risk of developing colorectal cancer generally increases with age. The majority of colorectal cancer cases are diagnosed in people over the age of 59.

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 59. 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 43-199% 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 19,999 and 1 in 49,999 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

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Print

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 cancer and 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.

Overview | Scientific Details | Frequently Asked Questions

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 166 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 MedlinePlus®

Chromosome 1

Gene: MUTYH

You do not have the two genetic variants we tested.

Variants Detected | View All Tested Markers

Marker Tested	Your Genotype*		Additional Information
Y179C Gene: MUTYH Marker: rs34612342	**T** Typical copy from one of your parents	**T** Typical copy from your other parent	➡ **Biological explanation** ➡ **Typical vs. variant DNA sequence(s)** ➡ **Percent of 23andMe customers with variant** **References** [1, 14, 19, 22, 26, 33, 34, 48]	ClinVar™
G396D Gene: MUTYH Marker: rs36053993	**C** Typical copy from one of your parents	**C** Typical copy from your other parent	➡ **Biological explanation** ➡ **Typical vs. variant DNA sequence(s)** ➡ **Percent of 23andMe customers with variant** **References** [1, 11, 14, 19, 22, 26, 33, 34, 48]	ClinVar™

* The percent of 23andMe customers with a certain may not be representative of the 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, 26, 28, 33, 37, 39, 48, 49]

Lifetime risk | Risk by age

The risk estimates for colorectal 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 cancer 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
Colorectal	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 259 by age 50. That number rises to about 1 in 25 by age 69.	[15, 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, 17, 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, 25, 26]
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, 23, 38, 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. Differences in access to healthcare may also contribute.	[3, 36]
Obesity Being overweight increases a person's chances of developing colorectal cancer. This association seems to be stronger in males than in females.	[4, 16, 21, 24]
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, 16, 20, 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 males than in females. 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 fruits, vegetables, 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, 18, 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 provide 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 45. Screening can be done through a variety of methods 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 DG505(MR) kit 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 cause 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, 26, 48]

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 89-98% 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 10,666 and 1 in 49,666 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 99.9%. 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 that 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 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 cancer.
- 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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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.

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Overview Scientific Details Frequently Asked Questions

MUTYH-Associated Polyposis

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 **Northern European** descent. What if I'm not of Northern European descent?

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

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

My report says **zero variants** were detected. Does this mean I'm not at risk of developing colorectal cancer?

My report says **zero variants** were detected, but I have a personal or family history of colorectal cancer. What does this mean for me?

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

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

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