This patient is at an ABOVE AVERAGE lifetime risk of colorectal cancer

Based on the information provided combined with the polygenic risk score, the patient's estimated risk for developing colorectal cancer over their lifetime (to age 90) is 19.72%. This is higher than the average risk of 4.54% for a patient of the same age, sex and race / ethnicity from the general US population.

The risk scores are patient-specific and cannot be used to estimate risk in relatives. These results should be interpreted by a healthcare provider in the context of the patient's full clinical history particularly if any SNPs are undetermined.

*The Polygenic Risk Score is a relative risk calculated as the multiplicative product of the patient's risk alleles weighted according to ethnicity-specific allele frequencies and odds ratios.
Clinical Responses

Does the patient have a medical history of colorectal cancer, Hereditary Non-Polyposis Colorectal Cancer (HNPPC)/Lynch syndrome, or Familial Adenomatous Polyposis (FAP)?

NO

Does the patient have a mutation in the MLH1, MSH2, MSH6, MUTYH, PMS2, or APC gene, or a diagnosis of a genetic syndrome that may be associated with an elevated risk of colorectal cancer?

NO

What is the patient’s age?

60

What is the patient’s sex?

Male

What is the patient’s race/ethnicity?

Caucasian

Is the patient aware of any first-degree relatives who have had colorectal cancer? (father / mother / brother / sister / son / daughter)

NO

About the Test

GeneType for Colorectal Cancer incorporates clinical responses with an analysis of the genetic markers known to be associated with colorectal cancer. The test is intended to help patients and their healthcare providers make informed decisions regarding colorectal cancer screening and prevention options.

The patient’s genotypes are used to generate a Polygenic Risk Score. The Polygenic Risk Score is calculated using a multiplicative model of colorectal cancer susceptibility. The risk model incorporates clinical risk factors (see Clinical Responses for a full list) and polygenic risk, combined with incidence and mortality data for colorectal cancer derived from the Surveillance, Epidemiology, and End Results Program (SEER), in a proprietary algorithm to provide an absolute estimate of the 5 year, 10 year and remaining lifetime risk of developing colorectal cancer.

Indication: GeneType for Colorectal Cancer is a colorectal cancer risk assessment test.

Population Risk: In the US, the average lifetime risk of developing colorectal cancer is 1 in 22 (4.55%) for men and 1 in 24 for women (4.15%).

Validation: GeneType for Colorectal Cancer is currently validated in Caucasian men and women between the ages of 30 and 80 and relies on the patient correctly reporting their ethnicity. This risk model also incorporates population incidence data for patients of African American and Hispanic American descent derived from the Surveillance, Epidemiology, and End Results Program (SEER), however, the model has not been validated in these populations as yet. Other ethnicities are under investigation but not yet available.

Limitations: GeneType for Colorectal Cancer is a colorectal cancer risk prediction test only. An increased risk score does not mean that a patient will definitely develop colorectal cancer. A low risk score does not mean that a patient will definitely not develop colorectal cancer. GeneType for Colorectal Cancer provides an estimate as to the likelihood that a person will develop disease at some stage in the future. Cancer is a multifactorial disease and it is not possible to incorporate all potential risk factors into a risk prediction model. Test results should be interpreted by a healthcare provider in the context of the patient’s full clinical history. Medical management and decision-making for colorectal cancer screening and prevention practices should not rely solely on a patient’s GeneType for Colorectal Cancer results.

Measurement of Uncertainty: Estimated potential variation in the polygenic risk score for varying numbers of undetermined SNPs are as follows: 1 = ± 7.40%, 2 = ± 11.06%. For patients that are very close to an average risk value, such variation may move the patient just above or just below average, however the magnitude of the change is small and their overall risk remains as “close to average”. Genotypes which are undetermined are assigned a risk score of 1.00.

Test Methodology: GeneType for Colorectal Cancer uses PCR arrays to determine the genotype of 45 polymorphic colorectal cancer susceptibility loci. Genomic DNA is extracted from buccal swab samples using standard DNA extraction methods. SNPs are genotyped using Taqman® chemistry on a customized OpenArray™ system using a QuantStudio™ 12K Flex Real Time PCR platform. The Polygenic Risk Score is calculated using a multiplicative model of colorectal cancer susceptibility. The risk model incorporates family history-based risk and the polygenic risk, combined with incidence and mortality data for colorectal cancer derived from the Surveillance, Epidemiology, and End Results Program (SEER), in a proprietary algorithm to provide an absolute estimate of the 5-year, 10-year, and remaining lifetime risk of colorectal cancer.

This test was developed, and its performance characteristics determined by Genetic Technologies’ Phenogen Sciences Laboratory. This test has not been cleared or approved by the United States Food and Drug Administration (FDA). The FDA does not require this test to go through premarket FDA review. This test is used for clinical purposes. It should not be regarded as investigational or for research.

References and Resources


NHMRC Clinical practice guidelines for the prevention, early detection and management of colorectal cancer Updated 2018.

For a full list of references supporting the GeneType for Colorectal Cancer risk assessment test, please visit www.genetype.com
Your Risk

Your risk of developing colorectal cancer is **ABOVE AVERAGE** for your age.

Your risk over the **rest of your life** is **19.72%**. The risk for an average woman of your age is **4.54%**. Any lifetime risk score greater than is **4.54%** is considered above average risk.

Your risk over the **next 5 years** is **2.41%**. The risk for an average woman your age is **0.55%**.

Your risk over the **next 10 years** is **4.9%**. The risk for an average woman your age is **1.25%**.

This test is designed for use by health care providers. The test results are to be discussed with your healthcare provider.

A very low risk result does not mean that you definitely will not develop colorectal cancer. A very high risk result does not mean that you definitely will develop colorectal cancer.

Some patients who do not develop colorectal cancer have higher risk estimates than patients who do develop colorectal cancer.

Understanding Colorectal Cancer Risk Factors

Your likelihood of developing colorectal cancer will be determined by a variety of risk factors. Some of these risk factors can be modified by making different lifestyle choices, while others are beyond your control.

**Colorectal Risk Factors You Cannot Change**

**Age:** Increasing age is the strongest risk factor for colorectal cancer. Most colorectal cancers occur in people over the age of 50. It is important to maintain regular health checks as you get older.

**Polygenic Risk:** You are born with a set of genetic markers called Single Nucleotide Polymorphisms (SNPs). This test looks at your SNPs to help determine your risk of developing colorectal cancer.

**Personal History:** Some people develop adenomatous polyps in their colorectal tract. These are growths that can develop into cancer. The presence of these polyps indicates an increased risk of colorectal cancer, especially if you have many of them or if they are large.

Inflammatory Bowel Disease can also increase your risk of developing colorectal cancer. Irritable Bowel Syndrome, or IBS, is not the same as Inflammatory Bowel Disease. IBS is not associated with an increased risk of colorectal cancer.

**Family History:** If other family members have been diagnosed with colorectal cancer or have had adenomatous polyps, this will increase your risk of developing the disease. Your risk will be even higher if there are multiple family members affected, if the family members are closely related to you (parent, sibling or child), or if they were diagnosed when they were younger than 45 years old.

Along with age, family history is one of the strongest risk factors for colorectal cancer. It’s important to talk to your relatives about their colorectal health and to share information about any diagnoses you may have received. Understanding your family history will help your healthcare provider develop an appropriate screening and prevention plan.

**Inherited Syndromes:** Certain inherited genetic conditions have been linked to an increased risk of colorectal cancer. Lynch Syndrome is the most common condition associated with colon cancer. Familial adenomatous polyposis, or FAP, also increases your risk. Talk to your doctor about whether you should be tested for these or other inherited genetic conditions that may increase your risk of cancer.

**Racial / Ethnic Background:** Some racial and ethnic groups have a higher incidence of colorectal cancer. African Americans and Jews of Eastern European descent are known to have a high risk of colorectal cancer. Further research is required to fully understand why these groups experience higher incidence rates and higher mortality rates.

**Type 2 Diabetes:** If you have type 2 diabetes, you will also have an increased risk of colorectal cancer. Some risk factors for type 2 diabetes are risk factors for colorectal cancer, such as obesity and a sedentary lifestyle. Even if you are managing your diabetes through healthy eating, exercise and other lifestyle changes, you will still have an increased risk of colorectal cancer.
Understanding Colorectal Cancer Risk Factors

Colorectal Risk Factors You Can Change

**Weight:** Obesity is associated with an increased risk of colorectal cancer in both men and women. Maintaining a healthy weight can reduce your risk, especially if you carry excess weight around your waistline.

**Physical Activity:** A sedentary lifestyle can contribute to a higher risk of colorectal cancer. Regular exercise can help to reduce your risk. As you increase the time you spend engaging in moderate physical activity, such as walking or cycling, your risk of colorectal cancer will reduce even further.

**Diet:** Chemicals in some food may increase your risk of colorectal cancer. Processed meats have been linked to an increased risk of the disease. Consuming large amounts of red meat or meat that has been cooked at high temperatures (grilled or fried) can also increase your risk.

Foods that are high in fibre help keep your colon healthy. Fruits and vegetables, whole-grains and legumes, such as lentils or chickpeas, can reduce your risk of colorectal cancer.

**Smoking:** Smoking is linked to an increased risk of many types of cancer, including colorectal cancer. Quitting smoking is one of the most important actions you can take to reduce your risk.

**Alcohol:** Alcohol is also linked to many different types of cancer. Your risk of colorectal cancer will increase if your consumption of alcohol is moderate or heavy. In general, consumption should be limited to 2 drinks per day for men and 1 drink per day for women. Talk to your doctor about the alcohol guidelines that will provide the most health benefits to you.

What You Can Do

The United States Preventive Services Taskforce (USPSTF) have published a recommendation titled, “Aspirin use to prevent cardiovascular disease and colorectal cancer: preventative medication”. Discuss your colorectal cancer prevention options, including the risks and benefits of risk-reducing medication, with your healthcare provider.

The USPSTF recommends an initial colonoscopy at age 50, with a re-screen in 10 years or an annual stool-based iFOBT. These screening recommendations may vary based on your ethnicity and your personal medical history. Some individuals will warrant initiation of screening at an earlier or later age depending on their additional risk factors that may not be considered as part of the GeneType for Colorectal Cancer risk assessment test.

Talk to your healthcare provider about a screening plan that best suits your needs.

Maintaining a healthy lifestyle is a simple way to reduce your risk of colorectal cancer.