Ambry Genetics®

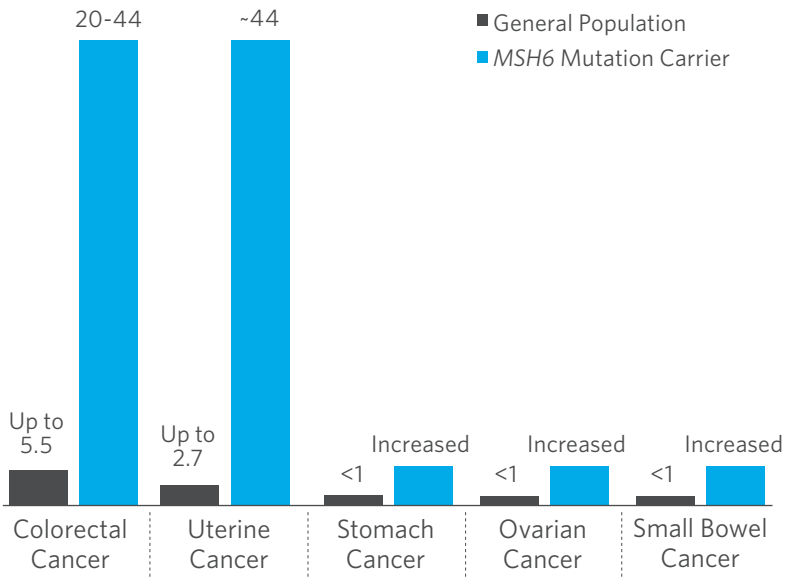
Understanding Your Positive *MSH6* Genetic Test Result

INFORMATION FOR PATIENTS WITH A **PATHOGENIC MUTATION** OR **VARIANT, LIKELY PATHOGENIC**

5 THINGS TO KNOW

1	*MSH6* mutation	Your testing shows that you have a pathogenic mutation or a variant that is likely pathogenic in the *MSH6* gene.
2	Lynch syndrome	People with *MSH6* mutations have Lynch syndrome, previously known as hereditary non-polyposis colorectal cancer (HNPCC).
3	Cancer risks	You have an increased chance to develop colorectal, endometrial/ uterine, stomach, ovarian, small bowel, and other types of cancer.
4	What you can do	There are risk management options to detect cancer early or lower the risk to develop cancer. It is important to discuss these options with your doctor, and decide on a plan that best manages your cancer risks.
5	Family	Family members may also be at risk – they can get tested for the *MSH6* mutation that was identified in you.

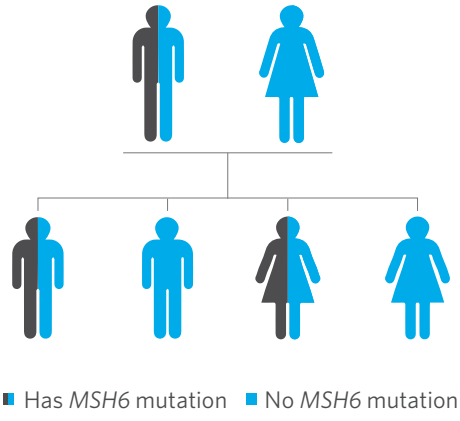
MSH6 MUTATION LIFETIME CANCER RISKS (%)*



*The above cancer risks represent the typical range for individuals with a mutation in this gene. If available, cancer risks specific to the mutation found in you will be provided in your results report.

MSH6 MUTATIONS IN THE FAMILY

There is a 50/50 random chance to pass on a genetic mutation in *MSH6* to your sons and daughters. The image below shows that both men and women can carry and pass on these mutations.



Understanding Your Positive *MSH6* Genetic Test Result

INFORMATION FOR PATIENTS WITH A **PATHOGENIC MUTATION** OR **VARIANT, LIKELY PATHOGENIC**

RESULT	MUTATION	Your testing shows that you have a pathogenic mutation (a disease-causing change in the gene, like a spelling mistake) or a variant that is likely pathogenic in the *MSH6* gene. Both of these results should be considered positive.
GENE	*MSH6*	Everyone has two copies of the *MSH6* gene, which we randomly inherit from each of our parents. Mutations in one copy of the *MSH6* gene can increase the chance for you to develop certain types of cancer in your lifetime.
CONDITION	LYNCH SYNDROME	People with *MSH6* mutations have Lynch syndrome, previously known as hereditary non-polyposis colorectal cancer (HNPCC).
CANCER RISKS	INCREASED	You have an increased chance to develop colorectal, endometrial/uterine, stomach, ovarian, small bowel, hepatobiliary tract, upper urinary tract, brain, sebaceous, prostate, and possibly other types of cancer.
SCREENING OPTIONS	FOR WOMEN	Options for early detection and prevention for uterine and/or ovarian cancer may include: random endometrial biopsies, transvaginal ultrasounds, CA-125 blood test, and options for preventive surgeries. Talk to your doctor about which options may be right for you.
SCREENING OPTIONS	FOR MEN & WOMEN	Options for early detection and prevention for men and women include: colonoscopy, upper endoscopy, and urinalysis (a test of your urine). Talk to your doctor about which options may be right for you.
RISK MANAGEMENT	VARIES	Risk management decisions are very personal, and the best option depends on many factors. Screening typically begins earlier than the general population and is often more frequently performed. It is important to discuss these options with your doctor.
FAMILY MEMBERS	50/50 CHANCE	Your close relatives (like your parents, brothers, sisters, children) have a 50/50 random chance of inheriting the *MSH6* mutation that you carry, and other family members (like your aunts, uncles, cousins) may also inherit it. Your relatives can be tested for this same mutation. Depending on the family history, those who DO NOT have it may not have an increased chance (above the general population) to develop cancer.
NEXT STEPS	DISCUSS	It is recommended that you share this information with family members so they can learn more and discuss this with their healthcare providers.
REACH OUT	RESOURCES	• Ambry's Hereditary Cancer Site for Families patients.ambrygen.com/cancer • Hereditary Colon Cancer Foundation hcctakesguts.org • I Have Lynch Syndrome ihavelynchsyndrome.com • Lynch Syndrome International lynchcancers.com • Genetic Information Nondiscrimination Act (GINA) ginahelp.org • National Society of Genetic Counselors nsgc.org • Canadian Association of Genetic Counsellors cagc-accg.ca • The HEROIC patient registry aliveandkickn.org

Please discuss this information with your healthcare provider. The cancer genetics field is continuously evolving, so updates related to your *MSH6* result, medical recommendations, and/or potential treatments may be available over time. This information is not meant to replace a discussion with a healthcare provider, and should not be considered or interpreted as medical advice.

WATCH THE VIDEO

Scan or visit our website to watch a short video with additional information to help you understand your results.
ambrygen.com/cancer/uyr/msh6

7 Argonaut, Aliso Viejo, CA 92656 USA Toll Free +1 866 262 7943 Fax +1 949 900 5501 ambrygen.com