Center for Genomic Health

MLH1 Fact Sheet

Mount Sinai

Cancer Risk Estimates

Cancer type	Average lifetime risk	Risk by age 70 with *MLH1* variant*
Colorectal	6%	Male: 27 – 74% Female: 22 – 53%
Endometrial	3%	14 – 54%
Gastric	<1%	0.2 – 13%
Ovarian	2%	11 – 20%
Small Bowel	<1%	4 – 12%
Liver	<1%	0.2 – 4%
Urinary Tract	<1%	0.2 – 25%
Brain	<1%	1 – 4%
Sebaceous (skin)	<1%	1 – 9%
Pancreas	1.5%	0.4 – 4%
Prostate	16%	9 – 30%
Breast	12%	5 – 18%

*Risk estimates are from GeneReviews, and may vary amongst different studies.

Medical Recommendations

Medical recommendations based on your genomic result include:

Colorectal: Colonoscopy every 1-2 years beginning at age 20-25

Gynecologic:

- Pelvic exam every year and consider yearly endometrial biopsy and transvaginal ultrasound beginning at age 30-35
- Consider risk-reducing removal of uterus (hysterectomy) and ovaries and fallopian tubes (salpingo-oophorectomy)

Stomach/Small Bowel: Consider upper endoscopy with biopsy of stomach every 2-3 years beginning at age 30-35

Kidney and Bladder: Consider urinalysis every year beginning at age 30-35

Skin: Annual skin exam by a dermatologist

Recommendations may be individualized based on your personal and family history of these cancer types. Routine screening for other cancers is also advised. Speak to your doctor about what options are right for you.

Individuals with a variant in both copies of *MLH1* have a rare childhood cancer syndrome known as constitutional mismatch repair deficiency syndrome (CMMRD). If you plan to have children, genetic counseling is recommended to discuss the risk to have a child with CMMRD.

For More Information

- General information and support resources: www.lynchcancers.com
- Genetic Information Nondiscrimination: www.ginahelp.org
- Find a Genetic Counselor: www.nsgc.org/page/find-a-gc-search