Lynch Syndrome: What You Need to Know

What is Lynch syndrome?
Lynch syndrome, also known as Hereditary Non-Polyposis Colorectal Cancer (HNPCC) syndrome, is a hereditary cancer predisposition condition caused by mutations in the MLH1, MSH2, MSH6, and PMS2.

What is my risk for cancer if I have an MLH1, MSH2, MSH6, or PMS2 gene mutation?
If you have an MLH1, MSH2, MSH6, or PMS2 gene mutation, you have an increased risk of getting certain types of cancer. However, not everyone who has a mutated gene will develop cancer.

- A person with an MLH1, MSH2, MSH6, or PMS2 gene mutation has a 50-80% chance of developing colon cancer in their lifetime. The average age of diagnosis is between 45 and 60 years old. In comparison, a person in the general population has a 5-6% risk of developing colon cancer in their lifetime.
- Women who have an MLH1, MSH2, MSH6, or PMS2 gene mutation have a 30-60% chance of developing uterine (endometrial) cancer. In comparison, a woman in the general population has a 2-3% chance of developing uterine cancer in their lifetime.
- There is also a small increase in the risk of developing gastric (stomach), ovarian, hepatobiliary (liver/gallbladder), small bowel, urinary tract, skin and central nervous system (brain) cancers in people with an MLH1, MSH2, MSH6, or PMS2 gene mutation.
- Finally, people with an MLH1, MSH2, MSH6, or PMS2 gene mutation have higher chances of developing more than one type of cancer over their lifetime.

It is important to note that these risks are based on individuals who did not have regular screening and/or other treatments such as preventative surgery. There is data that suggests that people with an MLH1, MSH2, MSH6, or PMS2 gene mutation can significantly decrease their risk of developing cancer by careful medical and surgical follow-up.

What is the chance that I will pass an MLH1, MSH2, MSH6, or PMS2 mutation to my children?
Both men and women who have an MLH1, MSH2, MSH6, or PMS2 gene mutation have a 50% chance of passing it on to each of their children. There is no evidence that suggests that being born with an MLH1, MSH2, MSH6, or PMS2 gene mutation increases the risk for childhood cancers.

How can I get tested for Lynch Syndrome?
There are 2 different ways a person can get tested for Lynch Syndrome:

1. Tumor testing: A portion of your cancer or polyp is tested for two characteristics. Microsatellite instability (MSI) and immunohistochemistry (IHC). If either of these tests come back abnormal, it is more likely that you have Lynch syndrome and additional genetic testing (on a blood sample) is recommended. If MSI and IHC both are normal, it is very unlikely that you have Lynch syndrome and no further testing is recommended.

2. Germline (blood) testing: A sample of your blood is analyzed to determine if a mutation in MLH1, MSH2, MSH6 or PMS2 is detectable.