

MSI and IHC: Colorectal Tumor Screening Studies for Lynch Syndrome

M. D. Anderson Cancer Center offers several screening tests that help identify patients who may have Lynch syndrome, also called hereditary nonpolyposis colorectal cancer (HNPCC for short). Lynch syndrome is a genetic condition, which means it runs in families and is passed from parents to children.

People with Lynch syndrome have a much higher chance to develop colon, rectal and other cancers than people in the general population. For someone who has cancer, the diagnosis of Lynch syndrome may mean that the person has a higher risk of developing a new cancer in the future. Following the early detection and prevention guidelines that have been established for Lynch syndrome can reduce these cancer risks. For more information about Lynch syndrome, please ask your nurse or doctor for a copy of the brochure “Lynch Syndrome (Hereditary Nonpolyposis Colorectal Cancer).”

MSI and IHC are two screening tests that can help identify patients who may have Lynch syndrome. The MSI and IHC tests are usually performed on tissue that has already been obtained from previous procedures. The details of these tests are explained below.

MSI Test

DNA is the genetic makeup of the millions of cells that form the body. DNA has all of the instructions that tell the cells of our bodies what to do. Usually, certain regions of the DNA are all the same length in each person. But sometimes, changes happen in the DNA that make these regions have many different lengths. This is known as microsatellite instability (MSI).

The MSI test compares the DNA of normal tissue to the DNA of tumor tissue (tissue in the body that contains cancer). It checks the two samples to see if the regions of DNA are the same length or different lengths.

MSI Test Results

About 85 percent of the time, the MSI test result is stable or low. This means that most of the tested samples showed DNA regions of the same length. If this is your test result, then you probably do not have Lynch syndrome.

About 15 percent of the time, the MSI test result is high. This means that most of the tested samples showed DNA regions of different lengths. If this is your test result, then you may have Lynch syndrome. More tests can be performed on tumor tissue to better determine if you may have Lynch syndrome.

IHC Test

IHC stands for immunohistochemistry. The IHC test uses special staining to look for four specific proteins in colon and rectal tumors. The four proteins are MLH1, MSH2, MSH6 and PMS2. These proteins play an important role in helping to keep the body healthy. All four of these proteins are usually found in normal colon and rectal tissue. Sometimes, in people with Lynch syndrome, one or more of these proteins are missing in colon or rectal tumors.

IHC Test Results

About 80 percent of the time, the test shows that all four proteins are in the tumor. If this is your test result, then you probably do not have Lynch syndrome.

About 20 percent of the time, one or more of the proteins are missing from the tumor. If this is your test result, then you may have Lynch syndrome. More tests can be performed on tumor tissue to better determine if you may have Lynch syndrome.

Receiving the Test Results

If you have questions about the testing, please talk with your doctor. Test results are usually available about 10-14 business days after your surgery. If you do not have a chance to discuss these results with your doctor at your post-operative visit, please contact your doctor to review the results.

You may be referred to meet with a genetic counselor if:

- The MSI test result is high **and/or**
- One or more of the proteins is missing in the IHC test

This means that you may have Lynch syndrome. You should meet with a genetic counselor to talk about these results in greater detail.

If you will be coming back to M. D. Anderson, an appointment can be scheduled for you with the Clinical Cancer Genetics Program. This meeting will help you learn more about the test results, and you can talk about additional testing options. If you will not be coming back to M. D. Anderson, we can provide you with contact information for a genetics clinic in your area.

To speak with a Genetic Counselor for additional information regarding these tests, please call 713-745-7391 or toll-free call 1-877-641-0979.