Yale NewHaven Health
Smilow Cancer Hospital

Yale CANCER CENTER
A Comprehensive Cancer Center Designated by the National Cancer Institute

Genetics of Lynch Syndrome
Information for patients

Cancer and hereditary factors
Most cancer cases are not hereditary. However, cancer can be inherited in some families. Many factors can increase the likelihood that the cancers in a family may be hereditary. These include early onset of cancer (colon cancer or uterine cancer in people less than 50 years of age); more than one primary (new) cancer in an individual; the same cancer in two or more close relatives on the same side of the family; unusual cancers; and related cancers (such as colon/uterine/ovarian/other gastrointestinal cancers) found in the same family. In addition, abnormal results on screening tests performed on a tumor such as microsatellite instability (MSI) or immunohistochemistry (IHC) may also suggest a higher likelihood that the tumor is due to a hereditary cause.

Colon cancer genes
Several gene changes (mutations) have been discovered that put individuals at greater risk for colon cancer. The most common form of hereditary colorectal cancer is called Lynch syndrome. Genetic testing of the genes that cause Lynch syndrome is available, including *MLH1, MSH2, MSH6, EPCAM* and *PMS2*.

The cancer risks and management recommendations can vary greatly between the different genes associated with Lynch syndrome. People identified as having Lynch syndrome should receive genetic counseling to be sure they understand their specific cancer risks.

Lynch syndrome is passed down in families in a pattern called autosomal dominant. A parent who has Lynch syndrome has a 50 percent chance of passing on that mutation to each of their children. If a person has Lynch syndrome their siblings also have a 50 percent chance of having the same mutation.

People with Lynch syndrome are at increased risk for several types of cancer. Both males and females with Lynch syndrome have a 10 - 60 percent chance of developing colorectal cancer versus the four to five percent risk for colorectal cancer in the general population. People with Lynch syndrome are also at increased risk for a second primary colon cancer. Females with Lynch syndrome have a 16- 57 percent chance of developing uterine cancer versus the three percent risk in the general population, and a 4 - 38 percent lifetime risk of ovarian cancer versus one to two percent in the general population. People with Lynch syndrome may also have an increased risk for other types of cancers, including stomach, small bowel, pancreas, urinary tract, prostate, sebaceous skin cancers and brain tumors. The risks for cancers can vary greatly between the different genes associated with Lynch syndrome.

Testing and screening

Individuals who learn they have Lynch syndrome are offered special screening as well as options to reduce their risks of developing cancer. Genetic testing for Lynch syndrome is available beginning at age 18. High-risk screening typically begins at age 25.

Contact information

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