Lynch Syndrome
Fact Sheet for Healthcare Professionals

Colorectal cancer is the second leading cause of cancer death after lung cancer.

Lynch Syndrome is an inherited condition that increases a person’s risk of developing colorectal and other cancers.

About 3% of patients with colorectal cancer have Lynch syndrome.

It is crucial to identify patients with Lynch Syndrome and their relatives to allow them to take advantage of interventions that can significantly reduce their risk of cancer in the future.

It is estimated that up to 98% of Lynch syndrome cases with and without colorectal cancer are undiagnosed.

Lifetime Cancer Risks Associated with Lynch Syndrome:

- 10-80% risk of colorectal cancer
- 15-60% risk of endometrial cancer
- 1-13% risk of stomach cancer
- 4-24% risk of ovarian cancer
- 2-4% risk of cancers of the hepatobiliary tract
- 1-4% risk of cancers of the urinary tract

Increased risks for cancers of small bowel, brain, and skin

Evidence-Based Clinical Recommendation for Lynch Syndrome Universal Tumor Screening

Several organizations recommend universal tumor screening to identify patients at risk for LS.

The Multi-Society Task Force on Colorectal Cancer (2014)
American College of Gastroenterology, American Gastroenterological Association, American Society of Colon and Rectal Surgeons, American Society of Gastrointestinal Endoscopy

The NCCN recommends that all patients under the age of 70 years with colorectal cancer be tested for Lynch syndrome.

The Evaluation of Genomic Applications in Practice and Prevention (EGAPP) Recommendation (2009)
The EGAPP Working Group found sufficient evidence to recommend offering tumor screening and genetic testing for Lynch syndrome to individuals with newly diagnosed colorectal cancer.

Cascade Screening
Increasing Identification of Unaffected Individuals at Risk for Lynch Syndrome

For those patients who screen positive for Lynch syndrome on universal tumor screening, genetic testing can be performed to diagnose Lynch syndrome by identifying the patient’s specific mutation. Other family members can then be tested for that mutation. This process is known as cascade screening.

Potential Benefits of Genetic Testing

If a mutation is found, increased tumor surveillance can be implemented to detect cancer in the early stages or prophylactic surgery can be utilized to reduce risk.

Provide a patient’s relatives with useful information, including which mutation relatives should be tested for.

For relatives, a negative test result may provide a sense of relief and will help them avoid unneeded screening and surgeries.

An employer may never discriminate against someone with respect to employment based on genetic testing information.
Adapted from the Lynch Syndrome: Fact Sheet for Healthcare Providers as part of the Tier 1 Genomic Applications Toolkit for Public Health Departments by the Centers for Disease Control and Prevention, Public Health Genomics in the U.S. Department of Health and Human Services. For people with disabilities, this document is available on request in other formats. To submit a request, please call 1-800-525-0127 (TDD/TTY call 711).

For IHC testing schematics visit: https://www.lynchscreening.net/wp-content/uploads/2012/04/IHC-methylBRAF.jpg; For MSI testing schematics visit: https://www.lynchscreening.net/wp-content/uploads/2012/04/MSI-flowchart.jpg

References