Lynch Syndrome

A Guide for Patients and Their Families

What is Lynch Syndrome?

Lynch Syndrome, also known as hereditary nonpolyposis colorectal cancer (HNPCC), is an inherited condition.

Lynch Syndrome is associated with a higher-than-normal chance of developing certain types of cancer before the age of 50, including:

- Colorectal
- Endometrial (uterine)
- Stomach
- Pancreatic
- Prostate
- Ovarian
- Urinary Tract
- Liver
- Kidney
- Bone
- Brain
- Skin

Lynch syndrome can be passed down in families from generation to generation through changes (genetic mutations) that occur in one of the following major mismatch repair genes (MMR): **MLH1, MSH2, MHS6, PMS2 and EPCAM**[1].

These genes are involved in repairing DNA (a long molecule containing our unique genetic code), therefore inheriting an alteration in one of these genes increases a person's risk of developing colorectal cancer and/or other related cancers.

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1. Stjepanovic, N. et al. Hereditary gastrointestinal cancers: ESMO Clinical Practice Guidelines for diagnosis, treatment and follow-up. Annals of Oncology vol. 30 1558–1571 (2019).

Diagnosing Lynch Syndrome

A blood test can determine if a person carries a change (mutation) in these genes and confirm Lynch Syndrome.

Screening for the likelihood of Lynch Syndrome can be performed on tumour tissue from patients with an existing **colorectal** or **endometrial cancer diagnosis.**

There are **two types** of Lynch Syndrome tumour screenings:

Immunohistochemistry (IHC): A screening test for the proteins made by the MMR genes; if the genes are working properly, the proteins will be present. The absence of MMR proteins means an increased likelihood of Lynch Syndrome.

Microsatellite Instability (MSI): A DNA analysis test checks if the MMR genes are working properly by looking at the stability of parts of the DNA, called microsatellites. A high MSI status means a high likelihood of Lynch Syndrome.

When necessary, further genetic tests and counselling may be recommended by your doctor based on your screening results and family's history of cancer.

What does it mean to live with Lynch Syndrome?

If you have Lynch Syndrome, you have an increased risk of:

Developing **colorectal cancer** before **age 50**

Developing **endometrial cancer** before **age 50**

Developing a **second cancer** such as gastric, ovarian, small bowel, urinary tract, pancreatic, brain and skin cancer

It does not mean you **will** develop one of these cancers, but it means **you may**. So, living with Lynch Syndrome means extra steps need to be taken to reduce your cancer risk. This includes earlier, more frequent, and additional screenings for cancers associated with Lynch Syndrome.

Colonoscopy every **one to two years,** beginning between the **ages of 20 to 25** or **five years younger** than the earliest age at diagnosis in the family

Upper endoscopy every **three to five years,** in addition to testing for **Helicobacter pylori infection** at an initial medical exam with treatment if positive

For women: yearly **pelvic examination, pelvic ultrasound, endometrial biopsy,** from **age 30 to 35**. Women who are past childbearing age may want to consider having preventive surgery to remove the uterus and ovaries

Recommendations and possible questions for your healthcare team

Talk to your family members to understand your family's history of cancer, and your doctor if you think you might be at risk of Lynch Syndrome. Feel free to raise any concerns you might have with your health care team. You might consider asking them the following questions:

1. What are my risks of developing colorectal cancer, endometrial cancer or other Lynch Syndrome associated cancers?
2. When should I be tested for Lynch Syndrome?
3. How can I reduce my risk of developing cancer with my Lynch Syndrome diagnosis?
4. What are my cancer screening options?
5. How does my diagnosis affect my family?
6. For those already diagnosed with an associated cancer - have immunohistochemistry (IHC) or microsatellite instability (MSI) tests been done on my tumour tissue?

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