Lynch Syndrome: Know Your Risk, Protect Your Future

What is Lynch Syndrome?

Lynch syndrome is a hereditary condition that increases the risk of certain cancers, especially colorectal and endometrial cancer. It is caused by inherited mutations in DNA mismatch repair (MMR) genes and is also known as hereditary nonpolyposis colorectal cancer (HNPCC).

What Causes Lynch Syndrome?

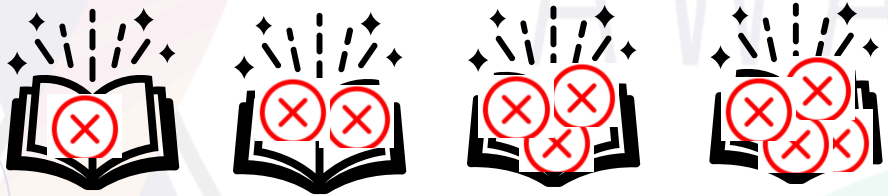
- Mutations in MMR genes (MLH1, MSH2, MSH6, PMS2, EPCAM) prevent proper DNA repair, increasing cancer risk.
- Normally, MMR genes fix errors that occur during DNA replication. In Lynch syndrome, mutated MMR genes fail to correct these errors.
- This leads to microsatellite instability (MSI), where DNA mutations accumulate unchecked.
- Over time, these mutations increase the risk of uncontrolled cell growth and cancer development.

Imagine This....

- Your DNA is like a book being copied over and over.
- Normally, the MMR genes act as skilled editors, scanning each new copy for typos and fixing them before the book is published.

Imagine This....

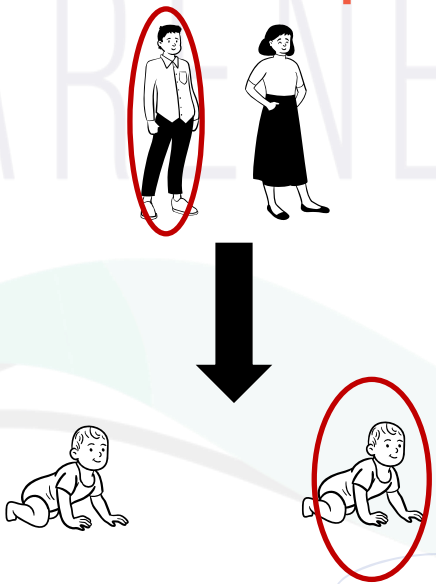
- But with Lynch syndrome, these editors are missing or malfunctioning.
- As a result, small typos (mutations) begin to appear in each new edition.



- Over time, these errors build up, changing the meaning of important passages. Eventually, the mistakes become so severe that they cause chaos in the story—leading to cancer as the cells lose control over their normal functions.

How is Lynch Syndrome Inherited?

- Passed down from parent to child (autosomal dominant inheritance).
- Chance of passing on mutation: 50% if one parent or both parents carry the mutation.



How is Lynch Syndrome Inherited?

- There is a slight chance (around 1 in a million) that a child could inherit mutated genes from both parents, leading to a more severe condition called constitutional mismatch repair deficiency (CMMRD).

- This can cause an extremely high cancer risk starting in childhood----but it is very rare.

Cancer Risks by Lynch Syndrome MMR Gene Mutation

MMR Gene	Colorectal Cancer Risk	Endometrial Cancer Risk	Other Associated Cancers
MLH1	Up to 80%	40–60%	Stomach, ovarian, urinary tract, bile duct, brain
MSH2	Up to 80%	40–60%	Stomach, ovarian, small intestine, urinary tract, brain
MSH6	10–44%	16–71%	Ovarian, small intestine, urinary tract
PMS2	15–20%	15%	Brain, endometrial, ovarian
EPCAM	Increased (via MSH2 silencing)	Increased (via MSH2 silencing)	Biliary tract, brain

What are the Signs and Symptoms of Lynch Syndrome?

Lynch syndrome itself does not cause symptoms, but certain patterns may indicate an increased risk of related cancers:

- Early-Onset Cancer: Colorectal or endometrial cancer diagnosed before age 50.
- Family History: Multiple relatives with Lynch-associated cancers, often across generations.

What are the Signs and Symptoms of Lynch Syndrome?

- Multiple Cancers: The same individual develops more than one Lynch-associated cancer over their lifetime.

- Fewer Polyps: Unlike other hereditary colorectal cancer syndromes, Lynch syndrome patients develop fewer polyps before cancer forms

What are the Signs and Symptoms of Lynch Syndrome?

- Rapid Cancer Progression: Tumors in Lynch syndrome patients often grow and spread faster than sporadic cancers.

- Other Cancer Signs: Symptoms related to affected organs, such as rectal bleeding, abdominal pain, unexplained weight loss, changes in bowel habits, or abnormal vaginal bleeding.

How is Lynch Syndrome Diagnosed?

Lynch syndrome diagnosis typically involves a combination of genetic testing, tumor analysis, and clinical evaluation:

- Genetic Testing: A blood or saliva test detects mutations in the MMR genes (MLH1, MSH2, MSH6, PMS2, or EPCAM). A positive result confirms the presence of Lynch syndrome.

How is Lynch Syndrome Diagnosed?

- Tumor Testing: If cancer is present, the tumor tissue is analyzed for microsatellite instability (MSI) and immunohistochemistry (IHC) testing to check for loss of MMR protein expression, which suggests Lynch syndrome.

- Family History Evaluation: Doctors use clinical criteria like the Amsterdam Criteria and Bethesda Guidelines to assess risk based on family cancer patterns.

How is Lynch Syndrome Diagnosed?

- Cascade Testing: If Lynch syndrome is confirmed in a family, relatives are advised to undergo genetic testing to determine their risk and need for early screening.

What Does Screening and Prevention Look Like?

Gene	Colorectal Cancer	Endometrial Cancer	Other Cancer Screenings
MLH1	Colonoscopy every 1-2 years starting at age 20-25	Annual transvaginal ultrasound & endometrial biopsy starting at age 30-35	Consider upper endoscopy every 1-3 years; Annual urinalysis
MSH2	Colonoscopy every 1-2 years starting at age 20-25	Annual transvaginal ultrasound & endometrial biopsy starting at age 30-35	Consider upper endoscopy every 1-3 years; Annual urinalysis
MSH6	Colonoscopy every 1-2 years starting at age 30-35	Annual transvaginal ultrasound & endometrial biopsy starting at age 40	Consider upper endoscopy every 1-3 years; Annual urinalysis

What Does Screening and Prevention Look Like?

Gene	Colorectal Cancer	Endometrial Cancer	Other Cancer Screenings
PMS2	Colonoscopy every 1-2 years starting at age 35	Consider endometrial screening based on family history	Consider upper endoscopy every 1-3 years; Annual urinalysis
EPCAM	Same as MSH2 guidelines (due to EPCAM deletions silencing MSH2)	Same as MSH2 guidelines	Same as MSH2 guidelines

What are Some Other Prevention Strategies?

- Prophylactic Surgery: Some individuals at high risk may consider preventive surgeries, such as hysterectomy or colectomy, to reduce cancer risk.

- Healthy Lifestyle Choices: Maintain a balanced diet focusing on healthy choices such as lean protein, fruits, and vegetables; avoid processed foods and red meat.

What are Some Other Prevention Strategies?

- Exercise & Weight Management: Regular physical activity helps reduce cancer risk and overall health improvement.

- Avoid Smoking & Alcohol: Reducing tobacco and alcohol consumption lowers cancer risks associated with Lynch syndrome.

What are Some Other Prevention Strategies?

- Aspirin Therapy: Some studies suggest daily aspirin use may lower colorectal cancer risk; discuss with a doctor before starting.

- Genetic Counseling: Family members should be informed and tested to determine their risk and necessary preventive measures.

Living With Lynch Syndrome

Lynch syndrome is not a death sentence. With early detection, proper medical care, and proactive lifestyle choices, individuals with Lynch syndrome can live long, healthy lives. By staying vigilant with screenings and maintaining a strong relationship with healthcare providers, many cancers linked to Lynch syndrome can be caught early or even prevented!

Living With Lynch Syndrome

- Stay up-to-date with screenings and follow medical advice.
- Seek genetic counseling for yourself and family members.
- Join support groups for guidance and emotional support.
- Stay informed about new treatments and preventive strategies through trusted medical sources.

Living With Lynch Syndrome

Many people with Lynch syndrome never develop cancer, and for those who do, early intervention significantly improves outcomes. A proactive approach to health makes all the difference!

Take ACTION!

Being diagnosed with Lynch syndrome can be overwhelming, but taking proactive steps can make a significant difference in your health and well-being. Here's what you can do:

•Know Your Family History: Talk to your relatives and gather information about any history of colorectal, endometrial, or other Lynch-associated cancers. Share this information with your doctor.

Take ACTION!

- Discuss Genetic Testing: If Lynch syndrome runs in your family, genetic testing can provide clarity on your risk and help guide medical decisions.

- Encourage Relatives to Get Tested: If you have Lynch syndrome, your close family members (parents, siblings, and children) have a 50% chance of having it too. Encourage them to undergo genetic testing and screenings.

Take ACTION!

- Stay Proactive with Screenings: Early detection is key to preventing or catching cancer at an early, treatable stage. Stick to the recommended screening guidelines and work closely with your healthcare provider.

- Adopt a Healthy Lifestyle: Regular exercise, a nutritious diet, and avoiding smoking and alcohol can help lower cancer risks.

Take ACTION!

- Join Support Groups: Connecting with others who have Lynch syndrome can provide emotional support, shared experiences, and valuable resources.

- Stay Informed: Medical research is continually advancing. Stay updated on new treatments, clinical trials, and preventive strategies through reliable sources like the Centers for Disease Control and Prevention (CDC), National Institutes of Health (NIH), and Lynch Syndrome International.

Take ACTION!

Remember: Early detection saves lives. Being informed and proactive empowers you to manage your health effectively and live a full, healthy life despite having Lynch syndrome.

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