

Your next steps



→ Find a specialist

Consult a gastroenterologist, gynaecologist and genetic specialist.



→ Follow medical guidelines

Regular specialist checkup: colonoscopy and gynaecological check-ups for women are advised to be scheduled.



→ Monitor symptoms

Keep track of new or changing health issues



→ Consider treatment options

Discuss available therapies and potential surgical interventions.



→ Stay updated on Lynch syndrome research

Clinical trials may offer new treatment possibilities.



→ Find a patient organisation

They can provide guidance and support.

Visit www.genturis.eu for medical guidelines and Lynch syndrome treatment centres.

About ERN GENTURIS

ERN GENTURIS is a European expert network for hereditary tumour syndromes. While we do not offer direct treatment, we provide patients and healthcare professionals with trusted information, guidance, and connections to specialists.

More info and contact

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Patient information

Lynch syndrome: what this diagnosis means for your health and next steps

If you or a family member has been diagnosed with Lynch syndrome, this guide provides clear medical information and practical next steps.



What is Lynch syndrome?

Lynch syndrome is one of the most common genetic tumour risk syndromes. About 2-3 % of all colorectal and about 4-5 % of all endometrial cancers occur in the context of this syndrome. Individuals with Lynch syndrome have a particularly increased risk of colorectal cancer and, in women, endometrial cancer. However, other tumours (such as ovarian, stomach, and urothelial cancers) are also part of the spectrum.

How is Lynch syndrome inherited?

Lynch syndrome is inherited in an autosomal dominant manner. This means that a mutation in one of two copies of the causative gene is sufficient to increase cancer risk. A child always inherits one of the two gene copies from each parent, the chances of also having a Lynch syndrome carrier is usually 50 %. Tumour risk in individuals with Lynch syndrome is significantly increased, and cancers tend to occur earlier in life than in the general population.

What causes Lynch syndrome?

Variants in four different genes (*MLH1*, *MSH2*, *MSH6* and *PMS2*) can cause Lynch syndrome.

All of these are DNA repair genes that help prevent DNA changes (mutations) and therefore cancer in the cells. The associated tumour risks differ between the four genes; therefore, experts increasingly refer to four distinct Lynch syndrome subtypes. Lynch syndrome should be suspected in individuals or families with a clustering of associated tumours or with a very early age of onset. Genetic testing can also be offered to family members to identify individuals who may be at increased risk of developing cancer.

What are the diagnostic/testing options?

Lynch syndrome should be suspected in a patient or family with a clustering of associated tumours or a very young age of onset in patients. Also, typical signs in tumour tissue indicating a DNA repair defect can indicate Lynch syndrome. But still germline genetic testing is needed to confirm a diagnosis. Genetic testing can be offered to relatives to identify persons who are also at increased risk for tumours.

What are the surveillance recommendations?

There are specific surveillance recommendations for Lynch syndrome patients, typically including regular colonoscopies starting in early adulthood and routine gynaecological examinations for women.

How this diagnosis affects you?

Receiving a genetic diagnosis requires careful medical supervision. Here are key points to consider:

How will Lynch syndrome and the associated tumour risk impact your health?

- The condition varies from person to person; regular medical evaluations are crucial.
- You may need a specialist evaluation over time.
- Since Lynch syndrome is genetic, family members should also consider genetic testing and surveillance.

What medical steps should I take?

- **Step 1:** Schedule an appointment with a specialist in Lynch syndrome.
- **Step 2:** Discuss options for genetic testing of relatives
- **Step 3:** Develop a personalised long-term surveillance plan with your doctor.

ERN GENTURIS provides information and resources to help you understand this condition and guide your medical team toward the best possible care.

More information on **Lynch syndrome**:

