National Cancer Control Programme

Bowel (colorectal) cancer and genetic testing for Lynch syndrome

Information for people who have received a diagnosis of bowel (colorectal) cancer

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Contents

About this Leaflet

This leaflet will help you understand what happens now that you have been diagnosed with bowel (colorectal) cancer. It starts by explaining why we first need to try to find out what caused your cancer. We need to do a test to find this out. We explain a little about inherited cancer and genes below. We look at what Lynch syndrome is and next steps.

What happens now?

It is important for you and your family, and for us, to find out what caused your cancer. It may have been caused by chance, or you may have inherited an increased risk. To understand this more, we need to do some genetic testing.

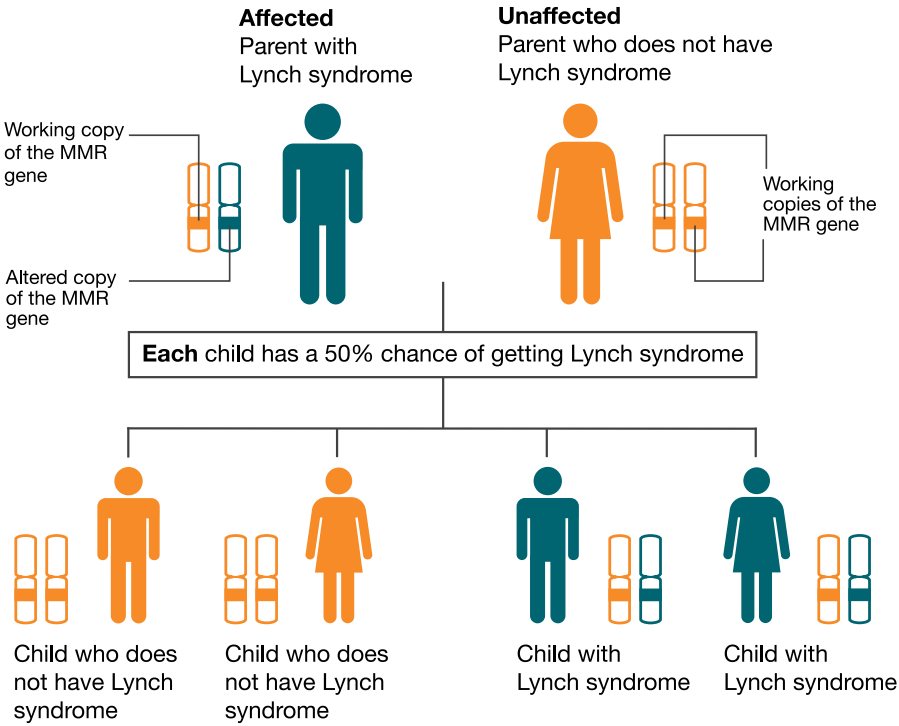
What is inherited cancer?

Our bodies are made up of small building blocks called cells. Genes are found in most of the cells in the body. A gene is a section of DNA, which usually makes one particular thing that contributes to how we grow and function. When something goes wrong in one or more of the genes in the cell, the cell may keep growing out of the body's control and this can lead to cancer.

If you inherit an increased risk of bowel cancer, this likely means that a change (in a gene) was passed on from your parent to you. Lynch syndrome is an inherited risk of many different types of cancer.

How is Lynch syndrome inherited?

You inherit genes in pairs. You inherit one gene from your biological father and one from your biological mother. If someone has Lynch syndrome, one copy of a mismatch repair (MMR) gene is altered and is not working properly. A mismatch repair gene is a gene involved in making repairs to errors in DNA. If one parent has Lynch syndrome, there is a 1 in 2 (or a 50%) chance of passing on the altered gene (Lynch syndrome) each time they have a child. This is called autosomal dominant pattern of inheritance.



Autosomal dominant pattern of inheritance

How is Lynch syndrome caused?

Lynch syndrome is caused by a change (called a 'pathogenic variant') in a gene. Lynch syndrome is caused by changes in one of the following genes:

- MLH1
- MSH2
- MSH6
- PMS2
- EPCAM

MLH1, MSH2, MSH6 and PMS2 are called 'mismatch repair genes'. EPCAM is not a mismatch repair gene, but it can stop MSH2 working as it should.

If you have Lynch syndrome, your risk of developing cancer depends on the gene affected. People with Lynch syndrome have a higher risk of bowel cancer.

How do the MMR genes work in the cell?

Errors in the DNA can happen when cells are dividing to make copies of themselves. Cells do this to replace old and damaged cells.

The mismatch repair (MMR) genes make proteins that repair errors in the DNA. If you have Lynch syndrome, the particular MMR gene affected cannot make the corresponding protein. This in turn means errors can build up over time and cause an increased risk of cancer.

Testing for Lynch sydrome

When you are first diagnosed with bowel cancer, your cancer cells are checked using a test called immunohistochemistry (IHC).

This is a standard test that looks at whether the mismatch repair (MMR) proteins are present or not in your tumour sample.

The results will determine what happens next. Ask your doctor about your test result.

This test is a cancer tumour test. It will likely show one of two results:

Result 1

All Mismatch Repair Proteins **Present**

Result 2

Mismatch Repair Proteins **Missing**

Result 1 - All Mismatch Repair Proteins **Present**

All mismatch repair proteins are present.

You are **unlikely to have Lynch syndrome**, and your cancer is unlikely to be an inherited cancer associated with Lynch syndrome.

Despite this result, you may still need to be referred to a Specialist Cancer Genetics Service. Your doctor will discuss this with you. It will also depend on your family and clinical history.

It is important to let your doctor know if you have a family history of cancer.

OR

Result 2 - Mismatch Repair Proteins **Missing**

A result that shows that the mismatch repair proteins are missing.

This means **you may have Lynch syndrome**.

The laboratory will likely do further molecular test(s). While these do not confirm a Lynch syndrome diagnosis, the results provide further useful information.

Depending on the results of these molecular tests you may be offered genetic testing for Lynch syndrome. Your doctor will help you decide on this.

Your consent is needed for this test.

What is genetic testing?

The main test for Lynch syndrome is a genetic test on a sample of your blood or saliva. Genetic tests look for pathogenic variants (gene change) in someone's DNA.

If you decide to have a genetic test, the results can help your healthcare team:

- confirm you have Lynch syndrome
- plan your treatment
- check for a clinical trial (new medical treatments being tested) that might be suitable

What your test results mean?

If you decide to have a genetic test for Lynch syndrome your Lynch syndrome test will show if:

1. A gene change that causes Lynch syndrome **was found**

If your result is **"pathogenic variant (a change in a gene) or likely pathogenic variant identified"**, this means that **you have a gene change that causes Lynch syndrome**.

If this happens:

- you will have a **higher risk** of certain cancers but it does not mean that you will definitely get cancer
- your relatives can be offered similar genetic testing called cascade testing
- your healthcare team will give you advice on ways to reduce your risk of specific cancers

2. A gene change that causes Lynch syndrome **was not found**

If your result is **"no germline pathogenic variant identified"**, this means that **a gene change for Lynch syndrome was not found.**

If this happens:

- you **likely do not have Lynch syndrome**
- your healthcare team may recommend another test on your tumour sample, called a somatic test.

  A somatic test is a genetic test that looks for gene changes in your tumour cells that are not inherited. Your healthcare team will explain what the results mean.

3. A gene change of **uncertain significance**

If your result is **"a variant of uncertain significance identified"**, this means that **you have a gene change but it's not clear if it causes cancer or not.**

If this happens:

- we will ask you to contact your genetic counsellor or healthcare team to check if there are any updates on this variant
- your family members will not have genetic testing
- we will offer you advice on how to reduce the risk of cancers that you have a family history of. You can also follow general advice on how to reduce your risk of cancer.

Final word

We hope you found this leaflet useful. If you have any further questions, please ask your doctor or genetics healthcare professional.

Acknowledgements

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Murphy Print & Design 064 6634650

National Cancer Control Programme,
King's Inns House,
200 Parnell Street,
Dublin 1.
D01 A3Y8

Tel: **01 8287100**
Email: **info@cancercontrol.ie**

www.hse.ie/cancer
Twitter: **@hseNCCP**

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