Feedback
We appreciate and encourage feedback. If you need advice or are concerned about any aspect of care or treatment please speak to a member of staff or contact the Patient Advice and Liaison Service (PALS):

**Freephone:** 0800 183 0204
**From a mobile or abroad:** 0115 924 9924 ext 65412 or 62301
**E-mail:** pals@nuh.nhs.uk
**Letter:** NUH NHS Trust, c/o PALS, Freepost NEA 14614, Nottingham NG7 1BR

[www.nuh.nhs.uk](http://www.nuh.nhs.uk)

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**Lynch Syndrome**

Information for patients

**Clinical Genetics**

This document can be provided in different languages and formats. For more information please contact:

Nottingham Regional Clinical Genetics Service
City Hospital
The Gables, Gate 3
Hucknall Road
Nottingham NG5 1PB
Tel: 0115 962 7728
Email: nuhnt.clinicalgenetics@nhs.net

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If you require a full list of references for this leaflet please email patientinformation@nuh.nhs.uk or phone 0115 924 9924 ext. 67184.

The Trust endeavours to ensure that the information given here is accurate and impartial.

Adapted from leaflet by Clinical Genetics, University Hospital of Southampton NHS Foundation Trust.

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Rebecca Collier, Genetics Department © December 2018. All rights reserved. Nottingham University Hospitals NHS Trust. Review December 2020. Ref: 1965/v2/1218/AH.
This booklet has been written for people with a strong family history of bowel cancer (also known as colorectal cancer), which may be due to an inherited tendency. It is for use with a clinical genetics appointment and should answer some of the common questions you may have about Lynch syndrome, (previously known as hereditary non-polyposis colorectal cancer or HNPCC).

Is bowel cancer inherited?
Bowel cancer is common in the general population. About 1 in 14 men and 1 in 19 women living in the UK will develop bowel cancer in their lifetime.

In about 5 to 10% of people with bowel cancer, the development of the cancer is thought to be due to a genetic cause.

Several different genes have been identified that are associated with a significantly increased risk of developing bowel cancer.

What is Lynch syndrome?
Lynch syndrome is a genetic (inherited) condition that causes an increased chance of developing certain cancers, particularly bowel cancer, endometrial cancer (cancer of the lining of the womb) and ovarian cancer.

It is caused by an alteration (mutation) in one of a number of genes. There are five genes known to be associated with Lynch syndrome, but there are also likely to be other genetic factors linked to an increased risk of bowel cancer still to be discovered.

What about life insurance?
People with a Lynch syndrome associated mutation that have been affected with a Lynch syndrome related cancer.
If buying new policies insurance companies are entitled to ask if you have had a genetic test just as they may request other information about your treatment or surgery.

People with a Lynch syndrome associated mutation who have not had a Lynch syndrome related cancer.
Currently insurance companies are not allowed to ask about predictive test results, sometimes people will choose to disclose a Lynch syndrome negative genetic test result to try to lower the premium of their policy which may be increased due to their family history of cancer.

The Genetic Alliance have a patient leaflet relating to all aspects of insurance questions: www.geneticalliance.org.uk/information/living-with-a-genetic-condition/insurance-and-genetic-conditions/

The team involved in your care are:

Genetic Counsellor: ..........................................................
Tel no: .................................................................
Your family reference number is: ..................................

Sources of further information
Lynch Syndrome UK
www.lynch-syndrome-uk.org/

Macmillan Cancer Support
Tel: 0808 808 0000
www.macmillan.org.uk/
Pancreatic cancer screening can be considered if there is a history of this in a first degree relative. Your clinician will discuss this with you further if relevant.

**Do lifestyle and diet make any difference to cancer risk?**

Currently there is no scientific evidence that there are specific lifestyle or dietary factors that reduce or increase the incidence of bowel polyps/bowel cancer in Lynch syndrome. However, there is evidence to suggest that a healthy lifestyle including not smoking, limiting alcohol consumption, maintaining a healthy weight and being physically active can reduce the overall risk of developing cancer in the general population.

**I've heard of research studies involving people with a family history of cancer, how can I find out more?**

There are several research studies that you may be able to take part in if you wish. It is important to remember that research studies may not benefit you directly, but may help future generations. Whether you decide to participate in a research study or not will not affect your future care in the NHS.

Studies have shown that taking aspirin reduces the chance of developing bowel cancer in people with Lynch syndrome. Current research is investigating what is the best dose of aspirin to take. If you would be interested in considering this please ask your Genetic Counsellor for further information.

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**How is Lynch syndrome inherited?**

Genes are the instructions that tell our bodies how to grow and function. All of our genes come in pairs and we inherit one copy from our mother and the other copy from our father.

A person with Lynch syndrome has a mutation (gene change) in one copy of a Lynch syndrome genes, they will also have a second working copy of the gene. Therefore, each of his or her children has a one in two (50%) chance of inheriting the mutation. If a person has not inherited the mutation, then they cannot pass it on to their own children.

The genes associated with Lynch syndrome are usually involved in repairing damage to our DNA. When a mutation is present in one of these genes, the gene does not work properly. If damage accumulates, the risk of developing cancer increases.
How do I know whether Lynch syndrome is the cause of the cancers in my family?

If you have several closely related family members (such as sisters, brothers, a parent, grandparent, aunts/uncles who are blood relatives) with bowel or endometrial cancer, and if their cancers occurred at a younger age than average, this may be suggestive of Lynch syndrome. However, even if this is the case, you may not have inherited it.

Can I have a test to see if the cancers in my family are due to Lynch syndrome?

Initial studies for Lynch syndrome in a family are often performed on tumour samples, at the time of surgery or at a later date. The results of these studies will indicate whether further testing to look at the Lynch syndrome genes would be helpful.

A person’s genetic material (DNA) can be looked at in a blood sample. However, because only a small proportion of people with bowel cancer will have a Lynch syndrome mutation, the test is currently only offered to people with a personal and strong family history of certain cancers, or following the results of tumour testing indicating the possibility of Lynch syndrome.

Usually testing is a two-stage process

Usually the first step, the ‘diagnostic test’ looks for a mutation in a blood sample from a person who has had cancer. This ‘diagnostic test’ currently takes 2-3 months.

If we find a mutation in a person with cancer that we feel is responsible for the family history of cancer, we can then offer testing to unaffected relatives to see whether or not they have the same mutation. This is called a ‘predictive genetic test’.

The hospital will give you details about what you need to do when you are given your appointment. We recommend that bowel screening continues every 18 months to 2 years from age 25. Bowel screening can sometimes be started a bit later than this, depending on the Lynch gene involved in your family. Your clinician will discuss whether this is appropriate for you.

Bowel screening greatly reduces the chance of developing cancer, but occasionally cancers can develop between colonoscopies. If you develop any concerning symptoms such as bleeding, passing mucus, unexpected weight loss or a persistent change in bowel habit, this should be discussed promptly with your GP.

Sometimes the Gastroenterologist may suggest preventative bowel surgery to manage any recurring or changing polyps.

At present, we do not know of any effective screening to detect the early signs of endometrial or ovarian cancers. Symptoms can include bleeding between periods or at any time post-menopause, persistent pelvic and abdominal pain, persistent bloatedness and feelings of fullness. Women should discuss any concerns promptly with their GP.

Many women will consider surgery to remove their ovaries and womb in their late 30s to early 40s. The advantages and disadvantages of this will be discussed with you, taking into account the specific Lynch syndrome gene in your family. Women found to have Lynch syndrome will also be referred to a Gynaecologist to discuss the management of their gynaecological cancer risks.

Currently we do not routinely offer upper endoscopy screening to look for signs of stomach or small bowel cancers. If there is a history of these in your family, your clinician will discuss this with you further. Gastric symptoms to be aware of include abdominal pain, bloating or reflux and persistent indigestion.
Does everyone who inherits a mutation associated with Lynch syndrome get cancer?

The risk of developing bowel and/or endometrial cancer is high if you inherit a Lynch syndrome associated mutation, but it is not inevitable. Some people who have a mutation in one of the Lynch syndrome genes never develop cancer, whereas other people with the same mutation may develop multiple cancers.

It is not possible to predict whether you will or will not develop cancer or at what age. It is also important to remember that the risk of developing cancer is not the same as the risk of dying from cancer. If a cancer develops, there is a good chance that the disease can be managed if detected and treated early.

If I have not inherited Lynch syndrome can I still get cancer?

Yes, there is still a chance of developing cancer, because cancer can occur for other reasons. People who have not inherited Lynch syndrome have a similar chance of developing cancer to someone in the general population.

Is Lynch syndrome the only condition associated with hereditary bowel cancer?

No, there are some other genes that increase the chance of developing bowel cancer. Your Genetic Counsellor/Doctor will discuss whether testing for other genes is relevant to you.

What can be done to manage the increased risk of developing cancer in Lynch syndrome?

Bowel screening is effective and is offered by a procedure called a colonoscopy. This is where a long flexible tube is passed up the back passage to look at the inside of the bowel. If polyps are found they can be removed there and then. It is necessary to prepare the bowel by emptying it before the test.

What are the possible results of ‘diagnostic genetic testing’?

Testing can reveal one of three results:

1. No gene changes (or mutations) are found

   If cancer tissue studies suggestive of Lynch syndrome:
   It is likely your personal and family history is due to Lynch syndrome but the specific genetic cause in your family cannot be found.

   You and at risk family members will be offered additional screening for Lynch syndrome but no genetic testing will be available for other relatives.

2. A cancer causing gene change (mutation) is found

   This confirms that the inherited tendency to bowel cancer in the family is caused by Lynch syndrome and other members of the family can have a ‘predictive’ genetic test to see if they have inherited the same mutation.

3. An ‘unclassified variant’ is found

   This is a gene change of unknown significance which may be entirely harmless. We would still offer regular bowel screening because of the family history, but no genetic testing is available for other family members. Sometimes research into the significance of the unclassified variant is possible. This will be discussed if appropriate.
If we cannot find a mutation you may wish to seek an update from the genetics service in the future, particularly if there are further cases of cancer diagnosed in the family.

**What is a predictive genetic test for Lynch syndrome?**

If someone in your family is known to have a mutation causing Lynch syndrome, you can have a genetic test to see if you have inherited the same mutation. We call this a predictive test and it involves taking a blood sample.

Before we offer you a predictive test, we will discuss with you what the test could mean for you and your family. We will give you the opportunity to ask questions. Your doctor or genetic counsellor will help you to consider:

- How this test result could affect you and your future
- If this could impact on your employment or insurance (currently insurance companies are not allowed to ask about the results of such genetic tests)
- Who you plan to talk to about this test and result
- Who you would like us to share your result with
- How this test result could impact on your family, children and relationships

Predictive test results are usually given in person, and further appointments can be arranged. Results usually take 4 weeks.

**Is there an alternative to genetic testing?**

You may decide not to have genetic testing but if you are not tested, you may still be eligible for additional screening and can be referred for this.

**What are the chances of developing cancer with a Lynch syndrome mutation?**

Mutations in the Lynch syndrome genes are primarily associated with an increased risk of bowel cancer. Women who carry a mutation in a Lynch syndrome gene have an increased risk of developing cancer of the lining of the womb (endometrium) and a slightly increased risk of developing ovarian cancer.

The chances are slightly different depending on which Lynch syndrome gene is involved. Lifetime (up to age 70) risks for specific cancers is detailed in the table below.

<table>
<thead>
<tr>
<th>Disorder</th>
<th>MLH1</th>
<th>MSH2</th>
<th>MSH6</th>
<th>PMS2</th>
<th>EPCAM</th>
<th>Population Risk</th>
</tr>
</thead>
<tbody>
<tr>
<td><strong>Bowel cancer</strong></td>
<td>35-65%</td>
<td>35-75%</td>
<td>20-70% (men)</td>
<td>15-20%</td>
<td>75%</td>
<td>6%</td>
</tr>
<tr>
<td><strong>Endometrial (womb) cancer</strong></td>
<td>20-25%</td>
<td>30-40%</td>
<td>25-70%</td>
<td>15%</td>
<td>15%</td>
<td>2%</td>
</tr>
<tr>
<td><strong>Ovarian cancer</strong></td>
<td>10-15%</td>
<td>10%</td>
<td>Similar to population</td>
<td>Similar to population</td>
<td>10%</td>
<td>1-2%</td>
</tr>
</tbody>
</table>

**Lifetime (up to age 70) risks for specific cancers with different Lynch gene mutations**

Figures from The Institute of Cancer Research

There is also a slightly increased risk over the general population risk for stomach, urinary, small bowel, and brain cancers. We would not recommend any specific screening for these cancers but encourage symptom awareness. Cancer risk figures will vary in studies looking at this data. Recent research suggests that the risks of endometrial cancer in MLH1 and MSH2 carriers may be higher than in the table above.