RM Partners
West London Cancer Alliance
Hosted by The Royal Marsden NHS Foundation Trust

St MARK'S
THE NATIONAL BOWEL HOSPITAL

NHS

Information for Patients with Endometrial Cancer

Your genetics appointment and testing for Lynch syndrome

You have been given this leaflet because of your diagnosis of endometrial cancer (also known as womb cancer). Initial testing of your cancer (tumour) suggests it might be due to an inherited condition called Lynch syndrome. We would like to offer you a genetic blood test to help determine whether you have Lynch syndrome.

This leaflet aims to answer some of the most commonly asked questions about Lynch syndrome. If you would like more information, you can visit:
https://rmpartners.nhs.uk/lynch-syndrome-early-diagnosis-pathway/patient-information/

Lynch syndrome

Womb cancer is the fourth most common cancer in women in the UK, and most diagnoses are due to older age, an unhealthy lifestyle or being overweight or obese. However, a small proportion of womb cancer is caused by inherited or genetic conditions; one of these conditions is called Lynch syndrome.

Most women with Lynch syndrome are well, but a women living with Lynch syndrome has a higher chance of developing bowel and endometrial (womb) cancer. Both men and women with Lynch syndrome have a slight increased risk of developing cancers in other parts of the body than people in the general population.

People with Lynch syndrome are monitored through colonoscopic surveillance to reduce the chances of bowel cancer developing.

Genes and DNA

To understand genetic testing for Lynch syndrome and what it means for you, we need to look at your DNA and genes.

DNA is the code our bodies use to make genes. Genes are the instructions that tell our body how to grow and develop and each have their own job to perform. Some of our genes determine what hair and eye color we have and some are responsible for protecting us against diseases like cancer.

Inherited conditions are due to an alteration (also known as a variant or genetic change) in a particular gene, which can be passed on in a family. An altered gene may change the level of protection a family has against disease such as cancer.

The Lynch syndrome genes are genes that protect us against cancer by repairing DNA mistakes that can occur when our cells are made. The Lynch syndrome genes are like police officers in our body, checking everything is working properly and protecting us against cancer. If these genes aren't working properly, then mistakes can occur in our DNA code, because we have less police officers protecting us against cancer.

Inheritance pattern

When we are conceived, we inherit 2 copies of every gene, 1 copy from our mother and 1 copy from our father. If one of our parents has Lynch syndrome, at the point of conception, they have a 50% chance of passing on their altered copy and 50% chance of passing on their unaltered copy. If a person inherits the altered copy, they will have Lynch syndrome, this is also known as an autosomal dominant inheritance pattern. This also means a person with Lynch syndrome has a 50% chance of passing on their altered gene each time they have a child.

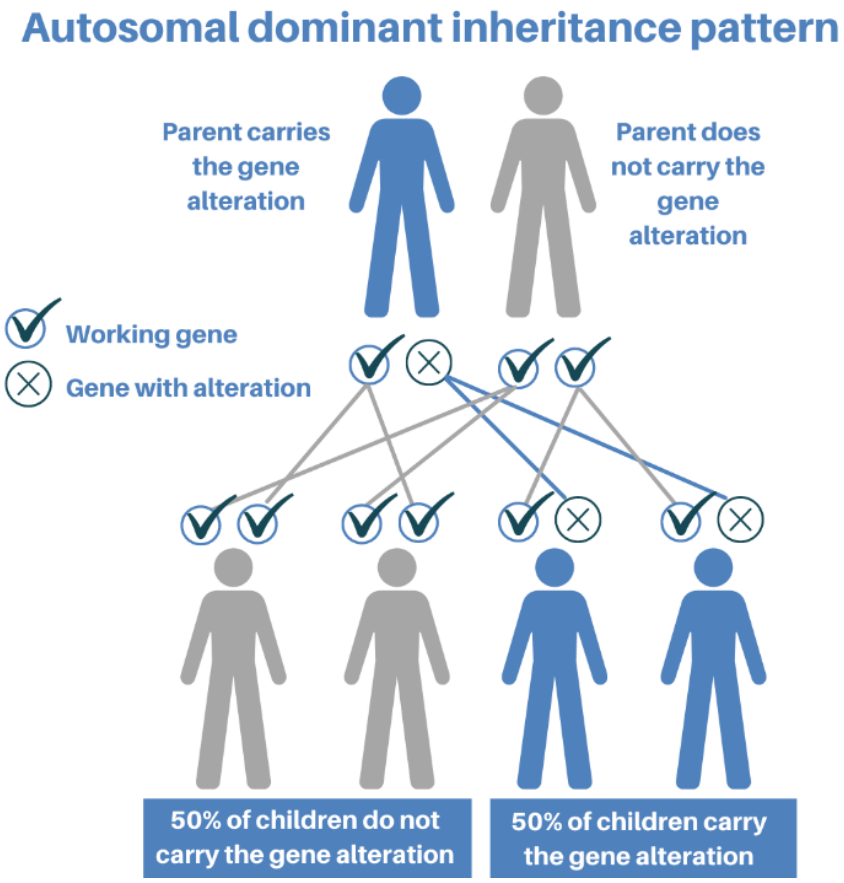

Figure created by North East Thames Regional Cancer Genetics Service

Genetic testing

Genetic testing for Lynch syndrome will involve having a blood test. DNA will be extracted from your blood to look for gene alterations (variants or genetic changes) that cause disease or increase your risk of developing a disease. This testing takes approximately 3 months to complete.

Genetic testing for Lynch syndrome helps us to understand you and your family's risk of developing cancer in the future. Positive test results will also help medical professionals understand the treatments you may require. There are many therapies available such as immunotherapy that can help to tailor your personal care.

Having genetic testing is optional. Your decision will not affect the standard of care you receive. However, knowing you have Lynch syndrome will give you and your family access to personalised treatments, advice and tailored surveillance programs. It will also enable testing of other family members to ensure they are kept safe and reduce the chance of further cancers developing in the family.

Genetic results

Genetic test results do not always give a clear answer. This is because the science of genetics is still in its infancy. For this reason, and for clarity, your results will be classified as: *positive*, *uncertain* or *negative*.

Positive: a genetic change (alteration) for Lynch syndrome is identified

This result would confirm the diagnosis of Lynch syndrome and provide an explanation for why you developed womb cancer. Your cancer team will use this information in their management decisions and will discuss this with you further. You might be considered for additional therapies. We will offer predictive genetic testing to your first degree relatives (parents, siblings, and children) as they might carry the same genetic change (alteration).

Uncertain: genetic change (alteration) of unknown clinical significance is found

A genetic change is found, however we cannot be certain it is the cause of your womb cancer. If appropriate, there may be further analysis of your tumour or other family studies recommended. Unfortunately, genetic testing cannot be offered to your family members at this time but we will still recommend cancer surveillance for you and your family.

Negative: We do not find any genetic changes (or alterations)

This result **doesn't rule out an inherited condition**. It might be that the limited knowledge we have about genetics means we are unable find a genetic change currently. If appropriate, there may be further analysis of your tumour recommended, to try to clarify if you have Lynch syndrome. Unfortunately, genetic testing cannot be offered to your family members but we will still recommend cancer surveillance for you and your family.

Your results will be given by the oncology team, who will also refer you to your local clinical genetics department. You will receive an appointment in clinical genetics if your result is positive. If your result is uncertain, your local clinical genetics department may contact you, either after your results or in the future, if there is any further testing that might clarify your results, or any further recommendations for your family members.