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Gynaecology Department

Genetic testing following a diagnosis of endometrial cancer

There is a 2020 national guideline [(1)] which states that endometrial cancers (womb cancers) should be tested to identify patients who may be at high risk for Lynch Syndrome.

What is Lynch Syndrome?
It is an inherited genetic condition that can significantly increase the risk of an affected individual developing endometrial cancer (womb), bowel cancer (colorectal) and some other cancers including ovarian cancer. It is also sometimes known as **HNPCC**, which stands for **Hereditary Non-Polyposis Colorectal Cancer.**

What causes Lynch Syndrome?
It is caused by a fault in an individual's genes, which should usually work to *prevent* cancer. This group of genes are called **Mismatch Repair (MMR) genes**, and include abnormalities called MLH1, MSH2, MSH6 and PMS2. The MMR genes usually work to fix mistakes in our cells' DNA, but when there is a fault in one of them, the DNA mistakes are not corrected, and this can lead to developing cancer. We all have two copies of the MMR gene in every cell of our bodies, but with Lynch Syndrome, one of the MMR genes in every cell is abnormal. Each time an affected individual has a child, there is a 1 in 2 chance that the child will inherit the altered copy of the gene and have a diagnosis of Lynch Syndrome too. This is known as "dominant inheritance".

What tests have been done for me so far?
All endometrial cancer samples are tested in the laboratory with a special staining process. This is called immunohistochemistry (IHC), and is used to identify an abnormality in the MMR gene.
Your cancer cells in your tumour have been identified to have an abnormality, and require further testing. However, so far, only the cancer cells have been tested. Cancers can be shown to have these genetic abnormalities; whilst all of the body's other non-cancer cells *do not* have the abnormality.
This initial test *does not mean* you definitely have Lynch syndrome, but it does mean we recommend referral to the genetics department at Addenbrookes hospital for further testing.

What further testing do I need?
The tests performed so far have confirmed the abnormality in the MMR genes in your cancer cells. Further tests need to clarify whether these changes are present in just the cancer, or the whole body.
The next step may require either of the following:

1. the tumour cells need further testing in the laboratory
2. you need a blood test to do further genetic tests (also known as germline testing)

Both of these options require a formal referral to the genetics department at Addenbrookes hospital, and with your permission, your consultant will write to their team.

How likely am I to have Lynch Syndrome given these initial tests?
Only **1 in 10** of the patients referred to the genetics department with these initial results go on to have a confirmed diagnosis of Lynch Syndrome.

What are the implications if I am found to have Lynch Syndrome?

If you are found to have Lynch Syndrome, this information can be used to help both you and your family.

- Your risk of developing bowel cancer can be reduced through surveillance and screening; you will be referred to the colorectal surgical team for regular monitoring.
- Your family (siblings, parents, children) can be referred to their local genetics department for testing, and if they are found to carry the genetic alteration too, they can be helped to reduce their risk of Lynch Syndrome-associated cancers with monitoring, or they could consider risk-reducing surgery e.g. hysterectomy.

Reference:

1. www.nice.org.uk/guidance/dg42. Testing strategies for Lynch Syndrome in people with endometrial cancer. October 2020.

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