Patient Information Leaflet

York and Scarborough Teaching Hospitals NHS Foundation Trust

Lynch Syndrome and Mismatch Repair Testing for Patients Diagnosed with Endometrial Cancer

Information for patients, relatives and carers

ⓘ For more information, please contact: Mr C Brewer, Consultant Gynaecologist at York Hospital, Wigginton Road, York, YO31 8HE or telephone 01904 725545.

Introduction

In most people cancer occurs by chance or due to the effects of certain risk factors. In a minority of people with Endometrial Cancer (about 3% or three out of every hundred), cancer occurs because they have a condition called Lynch Syndrome.

Lynch Syndrome is an inherited condition, which causes an increased risk of certain cancers. It is caused by a mutation (genetic change) in one of five different genes. Genes are pieces of the DNA code that each have a specific job or function that helps our bodies grow and function normally. We inherit our genes from our parents. We have two copies of each gene-one copy is inherited from our mother and one from our father. Some genes work to protect against cancer by correcting damage that can occur in the DNA when the cells in our bodies divide and grow.

In Lynch Syndrome there is mutation in a type of gene called a tumour suppressor gene. This by itself does not cause cancer to occur. It puts an individual at greater risk of developing cancer because their cells' ability to repair damaged DNA may be affected. This is sometimes called mis-match repair. A build-up of DNA damage can cause a cell to change into a cancerous cell. For reasons that are not yet fully understood the changes

seen in Lynch Syndrome give a risk of colorectal cancer and gynaecological cancers (such as Endometrial Cancer) rather than other types of cancer. There are four main genes which, when mutated, can cause Lynch Syndrome. These genes are called MLH1, MSH2, MSH6 and PMS6. There is also a gene called EPCAM which can cause Lynch Syndrome, but this is rare. These genes make proteins which help repair DNA, the mismatch repair proteins.

The risk of cancer in an individual with Lynch Syndrome varies due to a number of factors:

- The specific gene that is affected.
- Age
- Gender
- Family history of cancer
- Previous cancer diagnoses
- Previous surgery
- Diet and lifestyle

If an individual is found to have Lynch Syndrome it provides them and their doctor with information to help reduce the risk of future cancer by screening, for example colonoscopy (a telescope test looking at the bowels) to identify cancer changes in the bowels early when it is more easily treated.

Since LS is an inherited condition it can provide information for relatives about their risks of cancer. Children have a one in two (50%) chance of inheriting LS from an affected parent. It also means close relatives like brothers or sisters have a 50 percent chance of having Lynch Syndrome.

Individuals who are at risk of having Lynch Syndrome will be referred to specialists in gene problems, geneticists or genetic counsellors. These specialists will discuss how to test for Lynch Syndrome (usually a blood test) and what the result would mean for that individual. They will also discuss what it would mean for that individual's relatives.

How do you identify who is at risk of Lynch Syndrome?

When an individual is diagnosed with Endometrial Cancer the cancer cells will be tested for the presence or absence of the so called mismatch repair proteins. These proteins are absent in about one quarter of cases of endometrial cancer. If these proteins are absent, it might be a sporadic event (one off event only present in the cancer cells but not the rest of the body) or it might be due to Lynch Syndrome. If it is found that one or more of the mismatch repair proteins are absent, also called mismatch repair deficiency, the individual will be offered referral to the specialist genetics team for further discussions about and testing.

What happens once the cancer cell MMR protein deficiency results are available?

At present the results of MMR protein deficiency testing does not affect what treatment is offered, and the result might not be available until after treatment has been completed. This is because some of the tests have to be sent to a specialist laboratory to be performed. Your doctor will inform you of the MMR protein deficiency results once they are available.

If the results **do not** show any MMR protein deficiency, there is no risk of so referral to the clinical genetics specialists is not required.

If the results **do** show an MMR protein deficiency, then you will be given the option of being referred to the clinical genetics specialists. The presence of an MMR protein deficiency does not mean that you have, it means that you **might** be affected. As such, further testing will confirm whether or not you actually have.

If you do not wish to be referred to the clinical genetics specialists at this time we will inform your GP so that they are aware and have the necessary information should you change your mind and wish to be referred in the future.

If you do wish to be referred to the clinical genetics specialists, then your doctor will refer you to the clinical genetics specialists to discuss in detail:

- The implications of testing for you.
- Your options for future screening if found to be affected by.
- The implications of testing for your family.

At that stage only after detailed discussion with the clinical genetics specialists and only with your specific consent will you be tested for.

Further information

A Beginners Guide to Lynch Syndrome. The Royal Marsden NHS Foundation Trust.
Testing Strategies for Lynch Syndrome in people with Endometrial Cancer (DG42). NICE Guideline.

Tell us what you think of this leaflet

We hope that you found this leaflet helpful. If you would like to tell us what you think, please contact: Mr C Brewer, Consultant Gynaecologist at York Hospital, Wigginton Road, York, YO31 8HE or telephone 01904 725545.

Patient Advice and Liaison Service (PALS)

PALS offers impartial advice and assistance to patients, their relatives, friends and carers. We can listen to feedback (positive or negative), answer questions and help resolve any concerns about Trust services.

PALS can be contacted on 01904 726262, or email yhs-tr.patientexperienceteam@nhs.net

An answer phone is available out of hours.

Teaching, training and research

Our Trust is committed to teaching, training and research to support the development of health and healthcare in our community. Healthcare students may observe consultations for this purpose. You can opt out if you do not want students to observe. We may also ask you if you would like to be involved in our research.

Leaflets in alternative languages or formats

If you would like this information in a different format, including braille or easy read, or translated into a different language, please speak to a member of staff in the ward or department providing your care.

Patient Information Leaflets can be accessed via the Trust's Patient Information Leaflet website: www.yorkhospitals.nhs.uk/your-visit/patient-information-leaflets/

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