Liverpool Women's
NHS Foundation Trust

Tissue Studies for Lynch Syndrome
Information for People with A Personal or Family History of Bowel Cancer or Bowel Polyps

Introduction

Lynch syndrome is a hereditary condition that causes people to have an increased risk of developing certain types of cancer. People with Lynch syndrome have an increased risk of developing colorectal cancer, which is a cancer of the large bowel (colon) and rectum. Women who have Lynch Syndrome also have an increased risk of developing cancer of the womb (uterus) and ovaries.

How common is colorectal, womb and ovarian cancer in the general population?

The estimated lifetime risk of being diagnosed with bowel cancer is 1 in 15 (7%) for males, and 1 in 18 (6%) for females born after 1960 in the UK.[1]

The estimated lifetime risk of being diagnosed with uterine cancer is 1 in 36 (3%) for females born after 1960 in the UK.[1] The estimated lifetime risk of being diagnosed with ovarian cancer is 1 in 50 (2%) for females born after 1960 in the UK.[1]

How common is Lynch Syndrome

Lynch Syndrome is rare: Approximately 1-3% of people diagnosed with bowel cancer will have Lynch Syndrome.[2]

Why should I have this test?

Finding out your cancer/bowel polyps are due to Lynch Syndrome will mean we can test other family members to see if they also have Lynch Syndrome. We can then offer screening and/or surgery to you and family members with Lynch Syndrome to detect cancers at a very early stage or even help prevent certain cancers developing in the first place.

What causes Lynch Syndrome?

Lynch Syndrome is caused by an alteration (also called a spelling mistake/error) in one of 4 genes. These genes are called MLH1, MSH2, MSH6 and PMS2.

What do Tissue Studies involve?

Tissue studies are tests on samples of a cancer or polyp material removed during surgery or bowel screening. These tests may help to identify if the cancers and/or bowel polyps which have occurred in the family are likely to be due Lynch Syndrome. It is important to remember we can only perform tissue testing on certain types of bowel polyps.

What test is performed?

The test we would like to perform is called **I**mmuno**H**isto**C**hemistry (IHC) testing.

IHC testing checks if the 4 genes associated with Lynch Syndrome are working properly. Genes make proteins and IHC tests to see if the MLH1, MSH2, MSH6 and PMS2 proteins are present in your tumor sample or bowel polyp.

What happens if any abnormalities are identified?

If testing shows that one or more of the proteins is missing this could indicate that the tumor/polyp was due to Lynch Syndrome. **However, it is important to remember that further testing will be needed to confirm this**. You will be offered a follow-up appointment to discuss in more detail should this be the case.

What happens if NO abnormalities are identified?

If the IHC test is normal it is much less likely that bowel cancer and the cancers in the family are due to Lynch Syndrome. In these circumstances screening advice will be offered to family members in relation to the family history of cancer.

What about the results?

The test is complicated and the results can take up to six months. We will contact the person who has consented to the tests to give them the results. We will also update our advice about screening and whether any further testing could be useful. Someone from the genetics team will contact you to discuss this further if appropriate.

What if no tissue is available?

Sometimes we cannot get a tissue sample. However, it may be possible to get one from another relative who has had a related cancer. If we cannot get tissue from any relatives, we will give advice based on the information we already have.

What do I need to do?

To go ahead with these tissue studies we need permission from the individual who has had cancer or bowel polyps. If this is not possible because the person with the bowel cancer or bowel polyps has passed away, consent can be obtained from one of the following people: [3]

1. Spouse or partner (including civil or same sex partner). The Human Tissue Act states that, for these purposes, a person is another person's partner if the two of them (whether of different sexes or the same sex) live as partners in an enduring family relationship.
2. Parent or child (in this context a child may be of any age and means a biological or adopted child)
3. Brother or sister
4. Grandparent or grandchild
5. Niece or nephew
6. Stepfather or stepmother
7. Half-brother or half-sister
8. Friend of long standing.

If you want us to go ahead with these tissue tests, you need to complete and return the appropriate consent form. If you do not have a consent form and would like one, please contact the department. If you are consenting to tests on tissue of a deceased relative we would encourage you to discuss the decision with other family members.

If you do not want us to go ahead with the tests please let us know so we can provide advice based on the family history you have given without delay.

If you need more advice about any aspect of bowel cancer, you are welcome to contact:

Liverpool Centre for Genomic Medicine
Liverpool Women's Hospital NHS Foundation Trust
Crown Street
Liverpool
L8 7SS
Telephone: 0151 802 5001 or 5008 Email: lwft.clingen@nhs.net

Further information

Liverpool Women's Hospital Lynch Syndrome information leaflet

Cancer Research UK (http://www.cancerresearchuk.org/)

Macmillan (http://www.macmillan.org.uk/Home.aspx)

Human Tissue Authority (HTA) Code of Practice – 1 Consent. Version 14.0 Updated: July 2014 Scheduled review date: July 2016 www.hta.gov.uk.

Consent and confidentiality in clinical genetic practice: Guidance on genetic testing and shearing genetic information A report of the Joint Committee on Medical Genetics Royal College of Physicians and Royal College of Pathologists 2011.

http://www.Lynch-syndrome-uk.org

https://www.bowelcanceruk.org.uk/

https://www.nhs.uk/conditions/Cancer-of-the-colon-rectum-or-bowel

https://www.genomicseducation.hee.nhs.uk/resources/genetic-conditions-factsheets/item/81-lynch-syndrome

References

[1]Lifetime risk estimates calculated by the Statistical Information Team at Cancer Research UK. Based on Office for National Statistics (ONS) 2016-based Life expectancies and population projections. Accessed December 2017, and Smittenaar CR, Petersen KA, Stewart K, Moitt N. Cancer Incidence and Mortality Projections in the UK Until 2035. Brit J Cancer 2016.

[2]Cunningham JM, Kim CY, Christensen ER, Tester DJ, Parc Y, Burgart LJ, Halling KC, McDonnell SK, Schaid DJ, Walsh Vockley C, Kubly V, Nelson H, Michels VV, Thibodeau SN. The frequency of **hereditary** defective mismatch repair in a prospective series of unselected **colorectal** carcinomas. Am J Hum Genet. 2001;69:780–90.

[3]Human Tissue Act 2004, chapter 30. www.legislation.gov.uk/ukpga/2004/30/contents.

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This leaflet can be made available in different formats on request. If you would like to make any suggestions or comments about the content of this leaflet, then please contact the Patient Experience Team on 0151 702 4353 or by email at pals@lwh.nhs.uk

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