Lynch Syndrome

Information for people with either a MLH1, MSH2 or EPCAM gene change (variant)

This leaflet is for people with Lynch Syndrome. It is intended to provide a general summary of what Lynch Syndrome is and how it can be managed. Any information specific to you that differs from this will be outlined in a separate letter from Clinical Genetics.

What is Lynch Syndrome?

People with a clinically significant change (or variant) in any of a group of genes called ‘mismatch repair’ genes are said to have Lynch Syndrome. These genes include: *MLH1, MSH2, MSH6, EPCAM* & *PMS2*. You have been given this leaflet because your genetic test showed that you have a variant in either the MLH1, MSH2 or EPCAM gene.

Lynch Syndrome is associated with an increased risk of developing cancer. There are several different genes that cause Lynch Syndrome, and the associated cancer risks and management options vary between these. In women with a MLH1, MSH2 or EPCAM gene change the main cancer risks are in the colon (large bowel/colorectal), endometrium (womb/uterus) and ovaries. In men the main cancer risks are in the colon and prostate. In both women and men, there are other cancers associated with this gene change including urinary tract, skin, upper gastrointestinal tract cancers including stomach, small bowel and pancreatic cancer.

Managing increased risk of colorectal cancer

Colonoscopy Screening

Screening for individuals at significantly increased risk of colon cancer is by colonoscopy every 2 years from the age of **25 until 75**. This should be reviewed with your GP at the age of 75 if you wish to continue.

Colonoscopy is a camera test that looks directly at the lining of the bowel from the inside. The aim is to detect polyps (small growths) or cancer early when it is more treatable. It is not uncommon to develop polyps and they can often be easily removed during the procedure, **reducing the risk of cancer developing**. Colonoscopy is usually done in the hospital outpatient department. For most people a colonoscopy is a straightforward procedure, but in rare cases there can be complications.

For people who have had colon cancer diagnosed recently:
You will be followed up as is routine by your colorectal team for at least 3 years after diagnosis. Following that, it is important that your remaining bowel is checked every 2 years. Your colorectal team should arrange this for you. If this does not happen, you can ask your GP or contact us.

For people who have not had colon cancer:
If you are above (or approaching) the age to begin your colonoscopy screening, your Clinical Genetics team will refer you to your local screening centre directly.

If this is some time away for you, you should arrange an appointment with your GP when you are aged 24, to ask them to refer you for your colonoscopy screening. This will not happen automatically if you don't request it.

You can find more information about having a colonoscopy here:

www.macmillan.org.uk/cancer-information-and-support/diagnostic-tests/colonoscopy.

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It is important that you are always proactive about your colonoscopy screening and contact the endoscopy department if it is approaching 2 years since your last one and you have not been sent an appointment. Patients can contact their local Endoscopy Waiting List Office directly: NHS Lothian on 0131 536 4162, NHS Fife on 01592 729245, NHS Borders on 01896 826440.

Aspirin and colon cancer risk reduction

Clinical studies show that **taking aspirin regularly can half the colon cancer risk** in people with Lynch Syndrome. They should therefore consider taking low-dose aspirin (75mg) daily for at least 5 years after being diagnosed with Lynch Syndrome (**between age 20 to 65 yrs**). The protective effects of aspirin remain for at least 10 years and so it is suggested that people take an "aspirin holiday(s)" for perhaps 5 years after that before restarting again and so on; especially if they suffer from any side effects such as stomach irritation or bruising.

People with a BMI ≥30 may need a higher dose to gain the same benefit. They can discuss with their GP whether it would be appropriate for them to consider taking 150mg of aspirin per day rather than the lower dose.

It is recommended **to stop taking aspirin at 70 years** because of an increased risk of side effects.

Before starting aspirin, you should consult your GP to ensure there are no medical reasons for you to avoid taking it. It is important that you have blood pressure within the normal range before starting aspirin. If you find you are not able to tolerate aspirin, speak with your GP again who may consider medication to help with this.

Symptom awareness – colon cancer

You should be 'bowel aware' and know what is normal for you. Although some symptoms can be due to other causes, you should see your GP if you are concerned at all. When seeking advice about symptoms, you should always remind them that you have Lynch Syndrome.

The symptoms of colon cancer can include:

- blood in/on your poo (stools), or bleeding from the back passage
- a change in your normal bowel habit that happens for no obvious reason, such as needing to go more often, looser stools or constipation
- losing weight when you haven't been trying to
- pain in your tummy (abdomen) and/or back passage
- feeling that you have not emptied your bowel properly after you poo
- unexplained tiredness, dizziness or breathlessness
- an increasing amount of mucous in your stools.

You can find out more on the website www.nhsinform.scot/illnesses-and-conditions/cancer/cancer-types-in-adults/bowel-cancer

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Managing an increased risk of gynaecological cancer

This section is for women who still have their womb and/or ovaries and/or fallopian tubes.

The effectiveness of screening for gynaecological (womb/endometrial and ovarian) cancers has not been proven and so we do not currently recommend it.

Women with a gene change can consider having **risk reducing surgery** (removal of their womb, ovaries and fallopian tubes). This **reduces the risk of these cancers by over 90%**. In general, this should be once they have completed their families and not before 35-40 years of age, although any advice about timing may also depend on other risk factors and family history. The disadvantages of surgery include the small risks involved in the procedure itself but more importantly the implications of loss of fertility and the onset of the menopause. To manage menopausal symptoms, women should consider taking HRT, until 51 years of age (or until they wish to stop, in consultation with their specialist). Women who have had cancer in the past should ensure their specialist is aware of this so they can take that into consideration when advising about HRT.

Following your Clinical Genetics appointment, you may already have decided to be referred to our colleagues in Gynaecology. However, if not, or if you are too young, you can contact us when you are approaching the age of 35 to review the current advice and arrange appropriate referrals for you. This does not happen automatically.

You should continue to participate in routine cervical screening via the NHS Cervical Cancer Screening Programme.

Symptom awareness – endometrial and ovarian cancer

The main symptoms of cancer of the endometrium (womb lining) and/or ovarian cancer include:

- bleeding or spotting from the vagina after the menopause
- heavy periods that are unusual for you
- vaginal bleeding between your periods
- a change to your vaginal discharge
- a lump or swelling in your tummy or between your hip bones (pelvis)
- pain in your lower back or between your hip bones (pelvis)
- pain during sex
- blood in your pee
- needing to pass urine more frequently than usual
- increased abdominal size and persistent bloating (not bloating that comes and goes)
- persistent pelvic and abdominal pain
- unintentional weight loss
- difficulty eating and feeling full quickly, or feeling nauseous

You should speak to your GP if you develop any symptoms. We would suggest that your GP has a lower threshold than usual to refer you to a Gynaecologist for investigation of symptoms.

Stomach (gastric) cancer

The effectiveness of screening for stomach cancer has not been proven and is not currently offered.

***Helicobacter pylori (H.pylori)* testing**: *H.*pylori is a bacterium commonly found in the stomach. Having *H.pylori* can slightly increase the chance of developing stomach cancer and other stomach issues. It is therefore recommended that you are tested for *H.pylori* as it is easily treated if you have it. **This is especially important if you decide to take aspirin and you should be tested before starting it**. You should ask your GP to arrange *H.pylori* testing for you. Although there is limited guidance, it would be reasonable for this to be done every 5 years.

Prostate cancer

Men with Lynch Syndrome have an increased risk of developing prostate cancer. There is currently no proven screening programme for prostate cancer in men with Lynch Syndrome. However, they may wish to consider PSA testing from the age of 40. PSA is a blood test which looks for the levels of Prostate Specific Antigen (PSA). You should arrange to discuss this with your GP around that time. This may be particularly relevant in men with a family history of prostate cancer and/or those with a MSH2 gene change.

You should seek advice from your GP if you experience symptoms. Symptoms can include:

- needing to urinate more frequently, often during the night
- needing to rush to the toilet
- difficulty in starting to pee (hesitancy)
- straining or taking a long time while urinating
- weak flow
- feeling that your bladder has not emptied fully
- bone and back pain
- loss of appetite
- pain in the testicles
- unexplained weight loss.

Other cancers

Currently, there is no national recommended screening for the other cancers associated with Lynch Syndrome. We would suggest that you speak to your GP about any symptoms that concern you and remind them that you have Lynch Syndrome. For example:

- Blood in your urine or stool
- Urine infection which don’t respond to antibiotics.
- Abdominal mass (lump), persistent bloating, feeling of fullness
- Abdominal or loin pain (pain in your side or back)
- Fatigue
- Unexplained weight loss
- Change in bowel habit
- Reduced appetite

- Feeling sick

It is important to know what is normal for you and your body. If you notice a change in how you feel or how your body works, it is safer to get it checked. A change usually does not mean you have cancer, but it could still be something that needs treatment. If it is cancer, the earlier it is found, the more likely it is that it can be treated successfully. If it is nothing serious, your GP can reassure you.

You may find it helpful to read more about what to be aware of on the Macmillan website:

www.macmillan.org.uk/cancer-information-and-support/worried-about-cancer/signs-and-symptoms-of-cancer

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Some people with Lynch Syndrome develop a rare type of skin cancer, called ‘sebaceous’ tumours. These are most commonly found on the upper eyelid but can occur anywhere on the body. You should see your GP about any lumps you may have or develop.

Pancreatic cancer

There is no proven screening programme for pancreatic cancer. However, if you have Lynch Syndrome and have a family history of pancreatic cancer, you are eligible to take part in a research programme called the ‘EUROPAC’ trial. This is run by the University of Liverpool. They keep a registry of people at increased risk of pancreatic cancer. There is also a screening part of the study aimed at developing a reliable way of screening those who are at increased risk, aiming to diagnose a developing pancreatic cancer at a stage where treatment can be offered. You can find out more on their website www.europactrial.com and contact the research team directly, if you wish, by email europac@liverpool.ac.uk or phone 0151 795 1256.

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Diet and lifestyle

There are some steps you can take with your general diet and lifestyle to help to reduce your risk of cancer.

A **daily dietary fibre intake of 30-35 grams per day is recommended**. Research shows that “resistant starch” reduces the risk of the other (non-colonic/bowel) cancers associated with Lynch Syndrome, especially those of the upper gastrointestinal tract. Sources of resistant starch include seeds, beans peas, lentils, sweetcorn and green tipped bananas. It is also found in certain foods that are cooked and cooled (they can be reheated) for example, rice, potatoes and pasta.

You can find more information using the following website:

www.ukcgg.org/information-education/patient-resources

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If you have had bowel surgery, you should speak to your surgical team to ensure tolerance of dietary fibre.

The following are some things you can do to reduce your risk of cancer:

- **Eat a healthy diet**: plenty of fibre (such as beans & oatmeal), fruit and vegetables. Avoid processed meat and reduce the amount of red meat, fat and salt you eat.
- **Regular exercise**: try to be physically active every day. Aim for a mix of moderate aerobic activity and strength exercises every week. These could include fast walking, running, cycling or swimming. See website for further guidance for your age-group and circumstances:

  www.nhsinform.scot/healthy-living/keeping-active

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- **Maintain a healthy weight**. If you need help to lose weight you can speak to your GP. You may find the resources available here helpful:

  www.nhsinform.scot/healthy-living/weight-loss/

  www.nhs.uk/better-health/lose-weigh

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- **Stop smoking**: smoking increases your risk of many cancers including colon cancer. If you smoke, giving up is an important thing you can do for your health. If you want to give up smoking, you should ask your GP about the services available to you. There is also an online resource called Quit Your Way Scotland available which you may find helpful:

  www.nhsinform.scot/care-support-and-rights/nhs-services/helplines/quit-your-way-scotland

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- **Limit how much alcohol you drink**: it is safest not to drink more than 14 units of alcohol a week on a regular basis. If you do regularly drink this amount, it is best to spread your drinking evenly over 3 or more days and have several alcohol-free days every week. 14 units is the equivalent of 6 pints of beer (4% ABV), 6 medium size glasses of wine (13.4% ABV) or 7 double measures of spirits (40%).
- Look at the website for more information & resources:

  www.nhsinform.scot/healthy-living

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Family planning

People with Lynch Syndrome may have questions about the options available for having children in the future. Each of their children has a 50:50 (1 in 2) chance of inheriting the gene change. This chance is the same for each pregnancy. There are several reproductive options available for people with Lynch Syndrome. This is a personal decision and there is no right or wrong decision. The options include:

- No testing: their children can seek testing for the gene change when they are adults.
- Preimplantation Genetic Testing (PGT): PGT is a process that involves '*in vitro* fertilisation' (IVF) to create embryos from the couple in the laboratory, which are then tested at an early stage for the familial gene change. One unaffected embryo would then be replaced into the womb, with the hope that a pregnancy would result. The current success rate for PGT is approximately 40%. There are certain criteria to be eligible for PGT, including that both partners must be non-smokers, the female must be less than 39 years old and that her body mass index (BMI) must be less than 30.
- Tests in pregnancy: there are tests that can be done during pregnancy to find out whether the baby has inherited the gene change. These include Chorionic Villus Sampling (CVS) at 11-14 weeks or Amniocentesis from 15 weeks. There is a small associated risk of miscarriage caused by these procedures.
- Adoption

- Sperm or egg donation
- Not having children.

You can request an appointment to discuss your options with a Genetic Counsellor.

Your family members

Your relatives are at risk of having inherited the gene change and so may also have an increased risk of developing cancer. A definitive genetic test is available for them, usually from the age of 18. This allows them to find out whether they have the gene change and access appropriate screening, risk reducing measures and discuss reproductive options. It also allows them to find out whether there is a chance they may have passed/pass it on to their children.

Each of your children has a 50:50 (1 in 2) chance of having inherited the gene change from you. **Your relatives will not be contacted directly by us and so it is important that you let them know**. There is some helpful information about talking to children about this at different ages in the Beginner's Guide to Lynch Syndrome (see other sources of information).

Your Clinical Genetics team will provide information for you to pass on to your relatives. They should ask their GP to refer them to their local Clinical Genetics Service.

Future questions

We understand that you may have different questions in the future, depending on your stage in life and what is happening in your family. You are welcome to contact us at any time in the future should you have any questions at all. Times that may be particularly important may include coming up to an age where you are considering risk reducing gynaecological surgery, coming up to the age where your colonoscopy screening should begin, talking to your children about genetic testing, thinking about having children and struggling to tell your relatives about Lynch Syndrome.

Our contact details

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More information and support

Lynch Syndrome UK:

www.lynch-syndrome-uk.org

A beginner's guide to Lynch Syndrome:

https://patientinfolibrary.royalmarsden.nhs.uk/lynchsyndrome

Lynch Choices:

https://canchoose.org.uk/

The EVE Appeal:

https://eveappeal.org.uk/inherited-risks/lynch-syndrome/

Macmillan Cancer Support:

https://www.macmillan.org.uk/cancer-information-and-support/worried-about-cancer/causes-and-risk-factors/lynch-syndrome-ls

Maggie’s, Edinburgh:

www.maggies.org/our-centres/maggies-edinburgh

Bowel Cancer UK:

www.bowelcanceruk.org.uk