



Heredity Nonpolyposis Colorectal Cancer (HNPCC) / Lynch syndrome

This factsheet gives information on a condition that runs in families called hereditary nonpolyposis colorectal cancer (HNPCC). It is not a cancer as such but it can increase your risk of getting certain cancers. The factsheet describes what HNPCC is, how it is passed on and what happens if you have it. If you have any further queries or concerns about it, please contact the **National Cancer Helpline 1800 200 700** or speak to your GP.

Some families are advised to have regular screening for colorectal (bowel) cancer. This can happen when a family member has been diagnosed with colorectal cancer at a young age but does not have HNPCC. This factsheet does not concern this group of people.

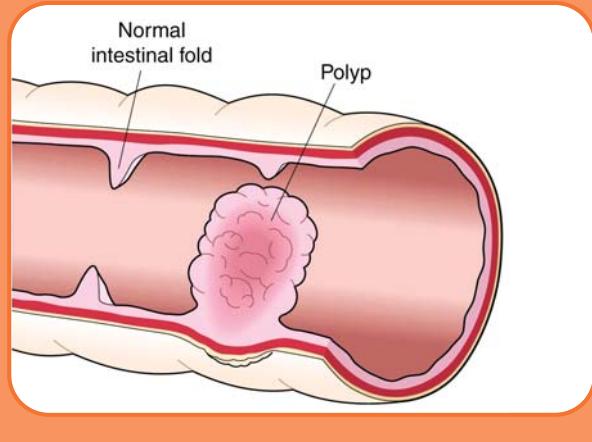
What is hereditary nonpolyposis colorectal cancer?

Hereditary nonpolyposis colorectal cancer (HNPCC) is a genetic condition that is passed on between families (inherited). It is also known as Lynch syndrome. A syndrome is a group of signs and symptoms that occur together and point to a particular condition. Colorectal cancer is also known as bowel cancer.

People with HNPCC tend to get colorectal cancer at a younger age than the rest of the population. They can also get cancers outside the bowel. These can include cancers of the stomach, small intestine, liver, gallbladder, urinary tract (ureter), brain, skin and prostate. Women with this condition also have a higher risk of womb and ovarian cancer. A tiny amount of those who get colorectal cancers will have HNPCC (about 2–5%).

What is a polyp?

A polyp is an abnormal growth of tissue in the lining of your bowel. They can vary in size from small to large. Colon polyps are very common and most are harmless. But if they are left untreated, they can lead to cancer.



You might think the word 'nonpolyposis' means polyps are not present, but those with HNPCC are at risk of getting polyps. The word 'nonpolyposis' is used to tell the difference between HNPCC and another type of bowel cancer called familial adenomatous polyposis (FAP). This also is an inherited syndrome. In FAP, thousands of polyps develop in the bowel.

How is HNPCC inherited?

Genes are found in all the cells that make up your body. They hold your genetic material (DNA) and are responsible for many things like the colour of your eyes or how tall you are. They also contain information that tells your body how to work and grow. Genes are passed on from your parents, which means that you inherit them.

Sometimes a gene develops a fault. Faults are also known as mutations. The faulty gene can lead to the abnormal growth of cells that becomes a cancer. If you inherit a faulty gene at birth, it means that cells throughout your body are more likely to develop changes that can lead to cancer.

The genes involved in HNPCC are MLH1, MSH2, MSH6 and PMS2. Faults in these genes and other genes yet to be identified are found in some people with HNPCC. However, not everyone who inherits these gene faults will get cancer.

Who might be at risk of having HNPCC?

Doctors have put guidelines together to help them find out who may have HNPCC and which families may be at risk. These are called the Bethesda/Amsterdam criteria. Doctors can find out who is at high risk and send them for more tests, if needed. Those at high risk are:

- a. **If you have three family members with colorectal cancer:** One must be a first-degree relative of the other two; and two generations in a row of the same family must be affected by cancer; and one of these relatives must be diagnosed with cancer before the age of 50.
Note: A first-degree relative means your parents, brother or sister, or your child.
- b. **If you have two HNPCC-related cancers:** These cancers include colorectal, ovarian, womb, ureter or kidney cancer.
- c. **If you have colorectal cancer and a first-degree relative with colorectal cancer and/or a HNPCC-related cancer:** These cancers include ovarian, womb, ureter or kidney and/or colorectal adenoma; with one of these cancers diagnosed under the age of 45 years and the adenoma under the age of 40 years.

Note: Colorectal adenoma is a tumour in the glands of the bowel. Although not cancer, adenomas may develop into colorectal cancer over time.

What should I do if I have a strong family history?

If you are concerned about your family history, it is important to discuss these concerns with your GP. He or she may then refer you to a family risk assessment clinic.

At this clinic, your family history will be looked at more closely and a family tree done. This family tree is known as a pedigree. If HNPCC is suspected, a test is done on the tumour of a family member with cancer. This happens only with the person's consent. Based on this, you may be referred for genetic testing to the National Centre for Medical Genetics at Our Lady's Children's Hospital in Crumlin, Dublin.

What is genetic testing?

A number of genetic changes may lead to HNPCC, unlike other conditions, e.g. FAP, where a single gene mutation is the cause of the condition. Some of these mutations may not yet be found. As a result, genetic testing is only possible if a mutation has been found in a family member.

Genetic testing involves taking a blood sample. This is sent to a special laboratory to try and find the particular fault in the gene. It may be many weeks before you get the results of these tests.

If you need genetic testing, you will be given information and counselling before the test takes place as well as after you get the results. At present, genetic testing is only suitable for people with a strong family history.

Who can be tested?

There are two stages to genetic testing within a family. As mentioned above, a family member with cancer gives their consent to have their tumour tested. Based on this result, they may be offered genetic testing to find the fault on the gene.

If a fault is found in that family member, then genetic testing can be offered to other members of the family. It is up to each family member to decide if they wish to be tested or not.

What happens if a family member has HNPCC?

If either of your parents has HNPCC, you have a 1 in 2 chance (50%) of inheriting the gene. In this case, you will be invited to go for genetic testing.

If you inherit a gene, it does not mean you will definitely get cancer. HNPCC is a syndrome not a cancer. If you have this syndrome, it increases your risk of getting cancer. Your chance of getting colorectal (bowel) cancer in your lifetime would be 4 in 5 (80%). For females with HNPCC, their lifetime risk of getting womb cancer would be 3 in 5 (60%) and they would have a 1 in 10 chance (10%) of getting ovarian cancer.

What happens if I have a gene for HNPCC?

As mentioned above, if you have a gene for HNPCC it increases your risk of getting some cancers. As a result, you will be encouraged to go for regular screening. This includes:

- Colon (large bowel) screening
- Womb and ovarian screening
- Stomach screening

Colon (large bowel) screening

This involves a test called a colonoscopy, where a camera is used to examine the bowel for any abnormal changes. You will be advised to have this every 1–2 years from the age of 25 years onwards. It is also important to be aware of any new bowel symptoms, including bleeding from your back passage, weight loss or a change in bowel habit. If you get any of these symptoms in between screening, have them checked out by your doctor.

Womb and ovarian screening

Women who are carriers of the HNPCC gene will also be advised to have regular screening as they have a higher risk of getting womb and ovarian cancer. Screening involves pelvic exams, ultrasounds and blood tests done at least every 2–3 years. Screening should start from the age of 35 years. You should also be aware of the symptoms of womb problems. These include heavy periods, bleeding between periods, bleeding after sex and bleeding after the menopause. If you get any of these symptoms between screenings, visit your doctor to have them checked out.

Some women with a high risk of getting cancer and who have completed their families can decide to have risk-reducing surgery. This involves removing the womb and ovaries.

Stomach screening

This involves a test called a gastroscopy. Here a camera is used to examine the stomach for any abnormal changes. This should be done every 2 years from the age of 25–30 years. It is also important to be aware of new symptoms including weight loss, feeling sick, loss of appetite, bloating or difficulty in swallowing. If you get any of these symptoms between screening, have them checked out by your doctor.

What happens if I do not inherit the gene?

If you have not inherited an abnormal gene, you will not need any special screening. This is because your risk of getting cancer is the same as the rest of the population. But you should continue to be aware of bowel changes and report any new symptoms to your doctor. This can include bleeding from the back passage, weight loss or a change in bowel habit.

It is also best to eat a healthy diet, which is low in fat and high in fibre, be a healthy weight, drink alcohol in moderation and get regular exercise to reduce your risk of getting cancer.

Support

Genetic testing and an inherited cancer can be hard for families to deal with. It is important to get support during this time. You may get this help from other family members, from friends or from your genetic counsellor/nurse. You can also speak to a nurse on the **National Cancer Helpline 1800 200 700**.



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Further information

If you are concerned about any aspect of your health or family history, you should contact your family doctor (GP). He or she will discuss your concerns and your family history and refer you to a specialist if needed.

For more information on HNPCC or about cancer in general, call the **National Cancer Helpline Freephone**

1800 200 700

(Monday–Thursday, 9am–7pm,
Friday 9am–5pm)

or email helpline@irishcancer.ie
for confidential advice from our
cancer nurse specialist.

Irish Cancer Society
43/45 Northumberland Road, Dublin 4

Tel: **01 231 0500**
Fax: **01 231 0555**
Email: helpline@irishcancer.ie
Website: www.cancer.ie

Useful organisations and websites

American Cancer Society

Website: www.cancer.org

Citizens Information Board

7th Floor, Hume House
Ballsbridge, Dublin 4
Tel: 01 605 9000
Email: information@ciboard.ie
Website: www.citizensinformation.ie

The National Centre for Medical Genetics

Our Lady's Children's Hospital
Crumlin, Dublin 12
Tel: 01 409 6793
Website: www.genetics.ie

The Johns Hopkins Guide for Patients and Families: Hereditary Nonpolyposis Colorectal Cancer

Website: www.macgn.org/cc_hnpcc1.html

National Cancer Helpline: **1800 200 700**