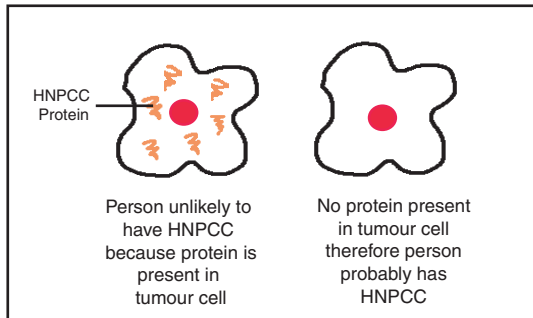


The immunohistochemistry test looks at proteins within tumour cells. If one of the HNPCC genes is faulty, there will be an absence of proteins within the tumour cells. If these proteins are absent or faulty, it means that there is **likely** to be a genetic cause for the tumour.

We are currently able to test 3 of the 5 HNPCC genes in this way, accounting for 90% of HNPCC families.



How long do the tests on tumour cells take?

It often takes many months to collect tumour samples from hospital laboratories. Once the samples are collected, the testing process usually takes around six months up to one year.

What happens after the tumour cell tests are completed?

If the tumour cell tests indicate that the cancers in a family are likely to be genetic, we are sometimes then able to offer a blood test to someone in the family who has already had cancer. This blood test is designed to look for a specific genetic mistake in the HNPCC genes. If a mistake is found in the blood, we know that the cancer in the family is inherited. Healthy family members can then be tested to see whether or not they carry the faulty

gene. If a family member does carry a faulty gene, they are more likely to develop colorectal cancer, cancer of the uterus, and a few other rarer cancers. If a family member does not carry a faulty gene, they do not have any increased risk of developing cancer.

For more information:

If you need more information about inherited cancer please contact:

Clinical Genetics Departments

Northern Scotland (main base Aberdeen)

Tel: 01224 552120 Fax: 01224 559390

(Aberdeenshire, Moray, Highland, Western & Northern Isles)

Tayside (main base Dundee)

Tel: 01382 632035 Fax: 01382 645731

(Perth & Kinross, Angus, North East Fife)

South East Scotland (main base Edinburgh)

Tel: 0131 651 1012 Fax: 0131 651 1013

(Borders, Falkirk, South West Fife)

West of Scotland (main base Glasgow)

Tel: 0141 201 0808 Fax: 0141 201 0361

(Glasgow, Argyll & Bute, Argyshire, Dumfries & Galloway, Stirling, Lanarkshire, Lothian)

If you need more advice about cancer and support groups, please contact:

www.maggiescentres.org

Maggies Highlands Tel: 01463 706302

Maggies Dundee Tel: 01382 496384

Maggies Edinburgh Tel: 0131 5373131

Maggies Glasgow Tel: 0141 330 3311

This leaflet was written by Guy's & St. Thomas' Clinical Genetics.

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Family history of colorectal cancer?



Tests that can be carried out to look at whether the cancers are inherited

Seen in clinic by.....

Introduction

This leaflet is designed to explain some of the tests that can be done to help decide if there is a genetic cause for bowel cancer in a family. Bowel cancer is also known as colorectal cancer (CRC).

Can colorectal cancer be inherited?

In most families colorectal cancer is not an inherited condition - it is not caused by a gene that is running in the family. In a small number of families, however, there does seem to be a tendency for people to develop colorectal cancer. We estimate that only about 5-10% of colorectal cancers are due to an inherited cause.

How can you tell if colorectal cancer is inherited?

The family tree and family history sometimes indicate that colorectal cancer is likely to be inherited. When the family history strongly suggests an inherited cause for cancer, we may be able to offer a genetic blood test to look for specific gene changes that are known to cause cancer.

If the family tree is less suggestive of an inherited cause of cancer, we can do another type of test. It is sometimes useful to look at tumour cells from people in the family who have had cancer. These cells can give us a better idea of whether the cancers in a family are likely to be inherited.

Inherited colorectal cancer

The most common type of inherited colorectal cancer is known as HNPCC (hereditary non-polyposis colon cancer). In families with HNPCC there are usually at least three closely related people who have had

colorectal cancer. HNPCC is more likely if the cancers occur under the age of 50. In HNPCC families, there are sometimes other cancers, such as cancer of the uterus (womb).

Why do people with HNPCC get cancer?

Families with HNPCC have a fault in one of five genes. These genes are known as the HNPCC genes. When one of these genes is faulty, the body is unable to repair the mistakes that sometimes occur in the genetic code. If a mistake is not properly repaired, a cancer can develop.

What can be learned from looking at tumour cells?

Most of the cells in the body are constantly dividing - old cells are being replaced by new ones. When a new cell is made, the DNA in the centre of the cell needs to be copied. Mistakes can occur as the DNA is copied from one cell to another. The body is usually able to correct these mistakes, with the help of the HNPCC genes. But if one of these genes is faulty, the mistakes cannot be corrected. By looking at tumour cells, we can see if more mistakes than usual are present.

Where can tumour cells be found?

When someone has an operation to remove a cancerous tumour, a small piece of the tumour is usually preserved in the hospital laboratory. This will be saved in the laboratory for many years. This sample contains valuable DNA which can be studied to look for genetic mistakes. If a tumour sample from someone who has had cancer in a family is

available, it can contain valuable clues that may reveal if the cancer is likely to have been caused by a fault in one of the HNPCC genes.

What tests can be done on tumour cells?

There are two tests that can be done on tumour cells to look for genetic clues. One is called the MSI test. MSI stands for microsatellite instability. The other is called the immunohistochemistry test.

The MSI test looks at the DNA in tumour cells. This is shown in the picture below.

