Hereditary Mixed Polyposis Syndrome

Introduction
Cancer is unfortunately very common, with more than 1 in 3 people developing cancer during their lifetime. Colorectal cancer, affecting the large bowel or rectum, occurs in about 1 in 20 people, but usually at older ages. Most colorectal cancers are not inherited, but are due to environmental factors. However about 15-30% of colorectal cancers are thought to be due to inherited genetic factors.

What is Hereditary Mixed Polyposis Syndrome?
Hereditary Mixed Polyposis Syndrome (HMPS) is a very rare inherited condition that increases the risk of developing multiple colorectal nodules known as polyps which can develop into cancer. Depending on their characteristics polyps can be classified into different types or groups. However HMPS patients can develop more than one type of polyp and sometimes single polyps have characteristics from more than one group, hence the name “mixed” polyposis syndrome.

If the polyps are not removed there is a high risk they will develop into a cancer.

What are genes?
We have about 25,000 genes in every cell of our body and genes are arranged in pairs, therefore there are two copies of every gene. Genes are units of hereditary material and contain the instructions for our bodies to function and maintain health. The genetic code can be thought of as a long sentence with thousands of letters. Changes in this code can interfere with the normal working of a gene and are called mutations. Changes in specific genes can be associated with certain types of cancer.

HMPS is caused by the duplication of a small section at the end of the SCG5 gene located on chromosome 15 which is thought to cause over production of an adjoining gene called GREM1. This is a newly described genetic alteration. All of the families identified in a recently published study were of Ashkenazi Jewish origin and it is thought that they share a single distant ancestor.

What is the risk of passing a genetic mutation on to children?
Children can develop HMPS as it is an inherited condition in an autosomal dominant way. If an affected parent has HMPS they have an altered (mutated) copy of the gene and a normal copy of the gene. When they have children they will either pass on the mutated copy or the normal copy of the gene. The children therefore have a 50% (1 in 2) chance of inheriting the mutated gene from their parent. This chance is the same for both male and female children and is . This is illustrated in the diagram below.
Bowel screening

At present there are insufficient data to give definite screening guidelines. Currently bowel screening by colonoscopy for patients at risk of HMPS is started at the age of 25 years because the polyps do not appear to develop at a particularly early age. The interval for screening will depend on the findings at the first colonoscopy. If polyps are found they can be easily removed and the risk of developing cancer is greatly reduced.

As far as we know, patients with HMPS are not at an increased risk of developing other types of cancer.

Family members who have not inherited the gene alteration are not at increased risk of developing polyps and will not require any additional bowel screening other than that offered by the NHS Bowel Cancer Screening Programme.

General lifestyle advice

Eating a healthy diet (high-fibre, low fat diet with plenty of fruit and vegetables), limiting your alcohol intake, not smoking and doing regular exercise are beneficial for general health reasons and have a role in preventing the development of cancer.

We recommend families to develop bowel awareness, and for people to see their GP if they have concerns about changes in their bowel habits or bleeding.

For more information please contact your local Genetics Service or St Mark’s Hospital Family Cancer Clinic on 020 8235 4266.

Reference

Jaeger E et al Nature Genetics 2012 May 6;44(6) 699-703 doi:10.1038/ng.2263
PALS is a confidential service for people who would like information, help or advice about the services provided by any of our hospitals. Please call 0800 783 4372 between 9am and 5pm or e-mail pals@nwlh.nhs.uk.

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