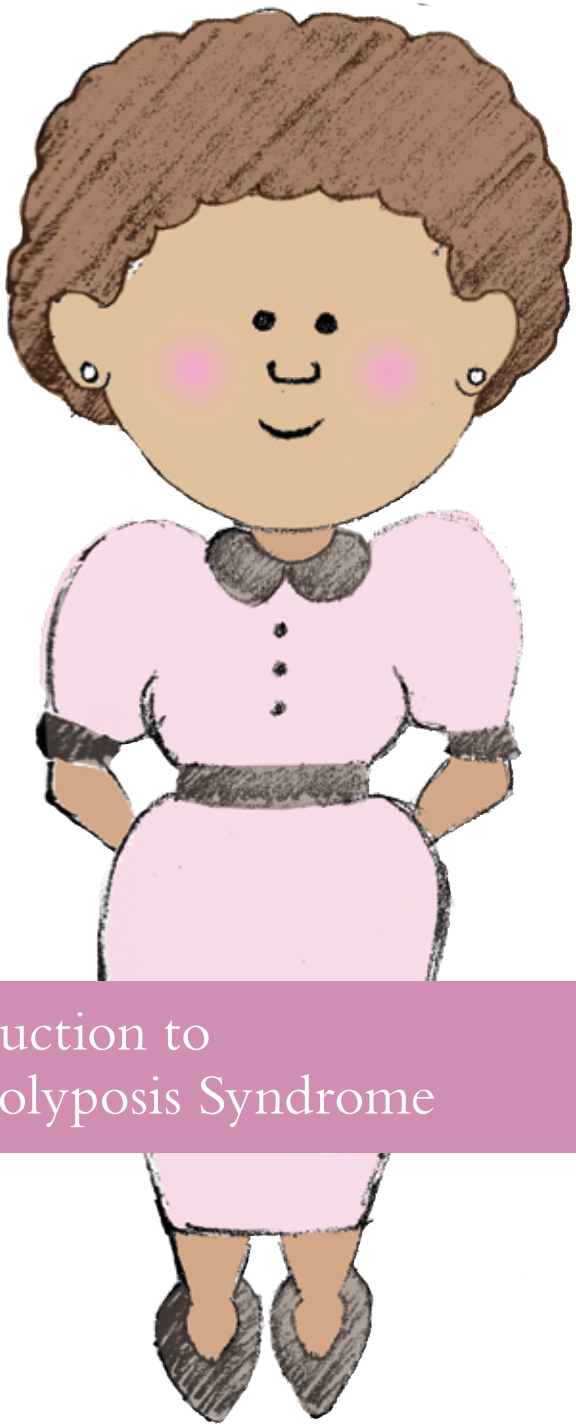

An Introduction to Juvenile Polyposis Syndrome

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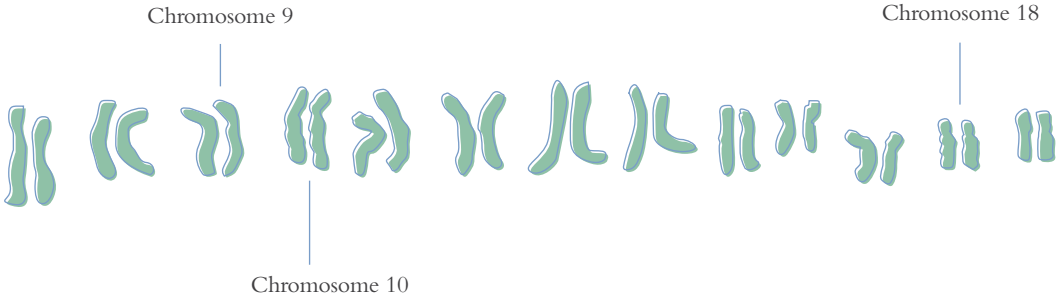
What is Juvenile Polyposis Syndrome (JPS)?

JPS is an inherited condition which mainly affects the stomach and large intestine (also known as the large bowel or colon and rectum). A diagram of the intestine can be found later in this booklet on page 9.

People with JPS develop polyps, which are like small cherries on stalks, inside their stomach, colon and rectum. There are many types of polyps but these are called juvenile polyps. The term "juvenile" refers to the type of polyp rather than to the age of the patient when the polyps develop. Most people with JPS have some polyps by the time they are 20 years old. Some may only have three or four polyps over their lifetime, while others may have hundreds. Juvenile polyps often bleed and if they are left untreated may cause anaemia (iron deficiency in the blood). Most juvenile polyps are non-cancerous, however may become cancerous if left untreated.



What causes JPS?

JPS is a genetic condition, which means it is caused by an altered gene. There are many thousands of genes carried on our chromosomes which determine our physical characteristics (our hair and eye colour, the shape of our nose, our blood type and the number of fingers and toes we have). Genes carry the information required to determine these characteristics as a chemical code in the form of DNA.

Sometimes mistakes occur in the DNA code so that genes carry wrong information. JPS occurs when the genetic recipe is wrong.

For example, consider a recipe. For a ham sandwich you need bread, butter and ham. If, when writing the recipe, the H in ham is changed to a J the end result would be a jam sandwich. Just a minor change, but the end result is very different.

When the JPS gene carries the correct information, the colon is protected from developing polyps. When the JPS gene is altered (so that incorrect information is carried) this protection is lost and the polyps develop in the bowel and sometimes in the stomach as well.

JPS results when there is an alteration on one of three genes: the SMAD 4 gene on chromosome 18, the BMPR1A gene on chromosome 10 or the ENG 1 gene on chromosome 9.

What is the chance of inheriting JPS?

JPS is usually inherited from a parent who has the condition. Each child born to a person with JPS has a 50:50 chance of inheriting the altered gene that causes it. A 50:50 chance is the same as the chance of getting heads or tails when you toss a coin. This is known as an autosomal dominant mode of inheritance.

If a person has not inherited the altered gene that causes JPS then that person's children will not be at any increased risk of getting polyposis.

Sometimes a person may be affected even though both parents have unaltered genes. This is because alterations can occasionally occur in the DNA during formation of the embryo.

In cases where this appears to have happened it is important that parents, brothers and sisters are screened to be sure. The children of a new person with JPS will have a 50:50 chance of inheriting the altered gene.

How would you know if you have JPS?

You might not have any signs or symptoms. Because early diagnosis makes such a difference to people with JPS, anyone who thinks they might be at risk should not wait for symptoms to develop, but should seek advice from their GP. They should ask their GP to refer them to St Mark's Hospital or their local genetics centre.

There are 2 ways of determining if a person has JPS:

::: DNA analysis

::: Bowel screening with histological confirmation

DNA Analysis

There have been many developments in this area.

Because it is known that JPS is caused by an altered gene, it is possible to take a blood sample, extract DNA, and then test to see if one of these genes is affected.

The alteration in the gene varies from family to family. Searching for the genetic alteration in someone with a clinical diagnosis of JPS can be time consuming. Once the genetic alteration is found in someone with JPS, other individuals in that family can be tested to see if they have the same genetic alteration (predictive testing). This is quite straightforward and will provide a definitive answer. In some families, however, the gene alteration cannot be found, in which case predictive genetic testing is not possible.

For example, suppose there is a suspected fault in a train and it is known that one of the bolts may be missing from under the back seat, on the left, in the fifth carriage. An engineer would soon be able to tell if there was a fault or not. This is like checking for a fault in a gene, in a family where the position of the fault has already been identified. But suppose nobody knows where about in the train the fault is. In this case every carriage of the train would have to be examined thoroughly in order to find it. This is what has to be done when the faulty gene in a family has not yet been identified.

Before blood is taken for DNA analysis, a person will be offered genetic counselling. The counsellor will be able to explain the situation in more detail.

Bowel Screening and histological confirmation

If you are at risk of inheriting JPS but the genetic alteration in the family has not been found, you should be advised to have bowel screening to look for polyps. The check-up is straightforward. The doctor will ask questions about your general health, feel your abdomen and arrange for you to have a colonoscopy. A colonoscopy involves passing a flexible telescope into the bottom so that the doctor can have a look to see if there are any polyps in the colon. Some people find the examination a bit embarrassing and uncomfortable but you will be sedated and it should not hurt.

Polyps can appear in people at different ages and if nothing is seen during the examination you may be advised to take part in a screening programme.

If during the examination, the doctor sees polyps a small sample (called a biopsy) will be taken. This is not painful and cannot usually be felt. The sample is sent to the laboratory for analysis to confirm that it is a juvenile polyp.

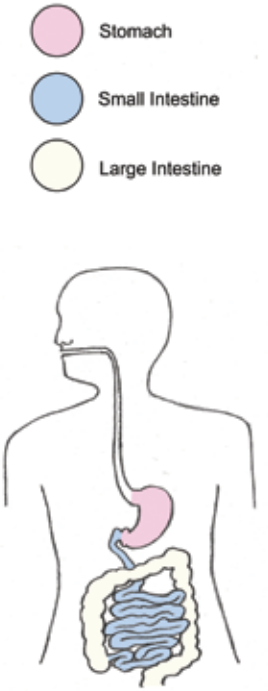
The gastro-intestinal tract

The intestine is divided into 3 parts:

The first part is the stomach. When food is swallowed it goes into the stomach where it is churned around and mixed with gastric juice.

The second part is the small intestine or small bowel. The blended food passes from the stomach into the small bowel where nutrients from food are absorbed.

The third part is called the large intestine. In the diagram you will see that the large intestine is itself divided into three parts, the colon, the rectum and the anus. The main function of the colon is to absorb fluid. The remains are passed into the rectum (bottom) which acts like a storage area before the faeces ("poo") are passed when going to the lavatory.

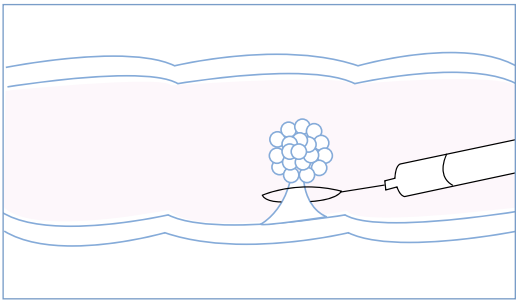


How is JPS treated?

The aim of treatment is to identify and remove polyps before they become too numerous or large enough to cause problems or become cancerous.

Polyps in the colon and rectum can be monitored by a regular colonoscopy. This involves passing a colonoscopy (flexible telescope) into the bottom to examine the colon. Polyps can be removed painlessly during this procedure.

A few people have such a large number of polyps in the colon that it is impossible to control them using the colonoscope. If this happens the doctor may advise them to have an operation to remove the colon. This is rare, but should it be necessary, the surgeon would discuss choices and provide detailed information.



Polyp Removal

Polyps in the stomach can be monitored by oesophageal gastroduodenoscopy (OGD). This involves passing a different endoscope in through the mouth to examine the stomach and first part of the small bowel (the duodenum). If necessary, polyps can be removed.

Once again there are a few people who develop such a large number of polyps in the stomach that an operation is recommended. This is very rare but should it be necessary, the doctor would discuss the situation and detailed information will be given.

These examinations will be repeated at one to three year intervals according to how many polyps develop over time.

What else is a person with JPS more likely to get?

People with JPS, who have an alteration in the SMAD4 gene, have been found to be more likely to develop Hereditary Haemorrhagic Telangectasia (HHT). People with HHT have a tendency to nose bleeds and they are also more likely to have abnormal blood vessels. If surgery is to take place, it is important that the doctors are informed about JPS so that a chest X-ray, echocardiograph and ECG is done prior to the general anaesthetic.

Where can I get advice?

The St Mark's Hospital Polyposis Registry provides a helpline for patients, their relatives and other healthcare professionals.

Please contact us on:
Telephone: 020 8235 4270
Email: nwlh-tr.PolyposisRegistry@nhs.net

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