



An Introduction to
Peutz Jeghers Syndrome



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Contents

What is Peutz Jeghers Syndrome?	2
What causes Peutz Jeghers Syndrome?	3
How do I know if I have Peutz Jeghers Syndrome?	4
At what age should my children be checked?	5
How is Peutz Jeghers Syndrome treated?	7
Will I get cancer?	11
Glossary	13
Where can I get advice?	14

What is Peutz-Jeghers Syndrome?

Peutz-Jeghers Syndrome, often called PJS, is a rare condition that is usually inherited, although it can occur spontaneously.

People who have PJS develop polyps, which occur most commonly in the gastrointestinal tract. Polyps are growths that look like little bumps or warts but which grow to look a bit like cherries on stalks; these polyps can grow quite large if left untreated.

There are many different types of polyp. The polyps in PJS are a unique type of polyp and are called Peutz-Jeghers polyps.



What causes Peutz-Jeghers Syndrome?

Characteristics, such as hair colour, eye colour and the number of fingers and toes we have, are determined by the thousands of genes that we inherit from our parents. These genes are carried on the twenty three pairs of chromosomes that we have in each of our cells. PJS is caused by a mutation, or change, in a gene known as the STK 11 gene (also known as LKB1) on chromosome 19.

PJS cannot be caught like a cold or cough, but is passed through families from generation to generation. Each child, boy or girl, born to a person with PJS has a 50:50 chance of inheriting the faulty gene that causes it.

In every family there will be a person who was the first to develop PJS and whose parents did not have it. In this person the mutation, or change, in the gene occurred by chance at a very early stage of development. These people can also pass PJS on to their children.

Chromosome 19

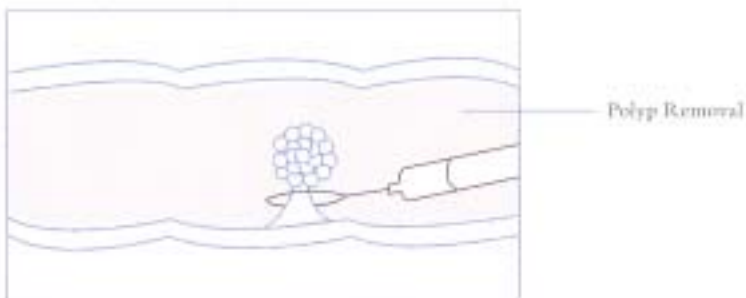


How do I know if I have Peutz-Jeghers Syndrome?

A feature of Peutz-Jeghers Syndrome is freckling, which is distinctive from the freckles of fair skinned people. People with PJS have darker freckles, often found on the lips, inside the mouth and on the fingers and toes. These freckles tend to occur around the age of two or three.

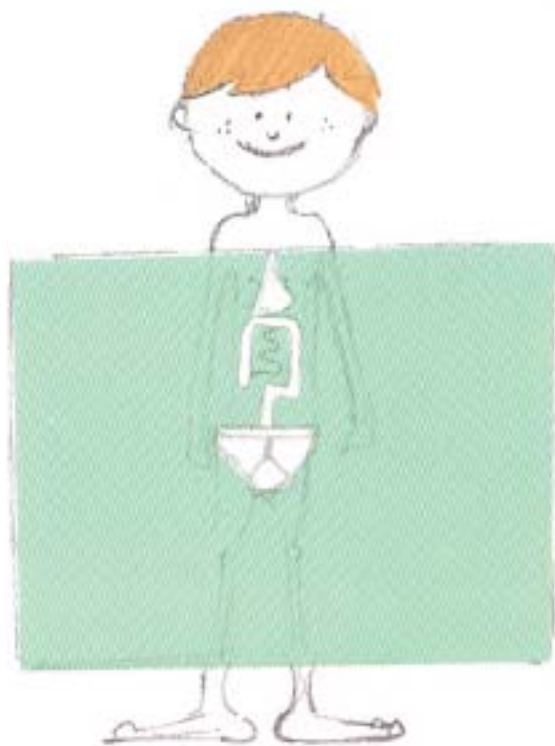
The other main feature of PJS is the polyps mentioned earlier. These polyps can occur throughout the intestine (virtually anywhere from mouth to anus) but can only be seen by a doctor using an endoscope (a kind of flexible telescope) and by special x-rays. Because different illnesses are associated with different types of polyps, the doctor will remove a polyp for the pathologist to confirm that the polyps in your case are the typical type as seen in PJS.

People with PJS may have symptoms common to other bowel conditions, such as diarrhoea, constipation, bloating, loss of weight, rectal bleeding and/or abdominal pain and possibly anaemia. A diagnosis of PJS, however, cannot be confirmed until after examination by a doctor.



At what age should my children be checked?

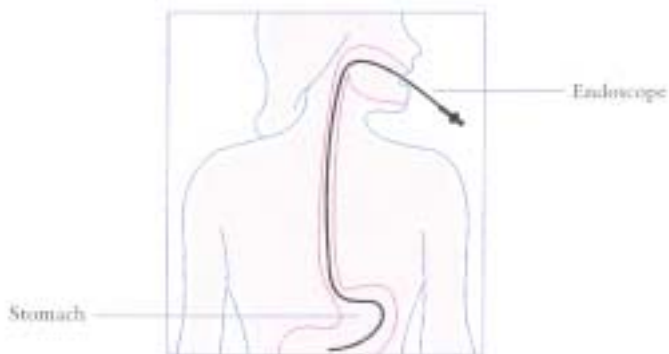
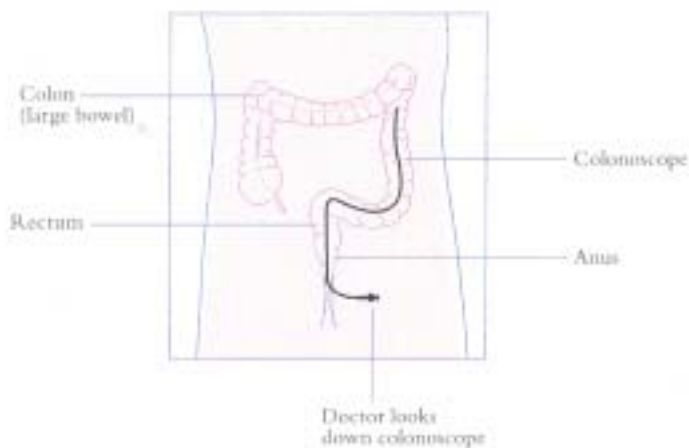
Young children frequently get colic (e.g gripe water is commonly used by parents). Little children with PJS are just as likely to get this sort of colic as are babies and toddlers. In addition from time to time the bowel tries to expel a polyp, leading to pain and vomiting (a condition known as intussusception). Because of this, it is sensible for new babies to be seen by a specialist paediatrician so that in the event of an emergency, the child is known to the hospital department and an emergency appointment can be arranged without any delay.



How is Peutz-Jeghers Syndrome treated?

Abdominal pain, caused by polyps in the bowel, is the most common complaint of children and young people with PJS. If left untreated the body may try to expel the polyps. This can cause pain and the polyp may become traumatised and bleed. There may be visible bleeding or there may be anaemia. In addition, since the polyp is anchored to the wall of the bowel, when the body tries to expel a polyp it succeeds in expelling the piece of bowel where the polyp is attached. This causes the bowel to slide inside itself (much like a naval telescope does when it is closed). This is called intussusception and causes the bowel to become blocked requiring emergency surgery. In an emergency the patient will be taken to the nearest hospital where the polyp that has caused the blockage will be removed and the bowel pulled back into its normal state (called de-intussuscepted). By careful management of patients with PJS we try to avoid emergency surgery and intervene with planned endoscopy and, when necessary, a planned surgical procedure. This will reduce the number of operations for each person.

In order to identify and remove polyps before they become large enough to cause problems, in adults we carry out the following tests every three years:



1

Colonoscopy

This involves passing an colonoscope (flexible telescope) into the bottom to examine the colon. Polyps can be removed painlessly during this procedure.

2

Gastroscopy

This involves passing a different endoscope in through the mouth to examine the stomach and the first part of the small bowel (the duodenum). Once again polyps can be removed.

3

Capsule endoscopy

This involves swallowing a small camera, just 2cm in size. This takes regular pictures as it passes through the 'small bowel'. It passes naturally in the faeces and does not need another examination to retrieve it.





**If capsule endoscopy is not available
barium follow-through may be used.**

This involves drinking a chalky drink which will allow the doctors to examine the small bowel between the duodenum and the colon on an X-ray.

If a large polyp is seen in this middle bit of the bowel, an operation, known as a laparotomy, can be planned to remove it. The reason for doing this is to avoid the need for future, unplanned emergency surgery, perhaps when on holiday overseas or performed by a surgeon not that familiar with PJS. Most importantly, at the same time as removing the large polyp the surgeon can use an endoscope, during the operation, to examine the rest of the small bowel and remove other smaller polyps. This will reduce the chance of future surgery, either elective or as an emergency.

If a single polyp, or maybe just two or three are seen in the middle bit of the small bowel it may be possible to remove these by using a procedure known as a 'double balloon' endoscopy. This will avoid the need for surgery.

Will I get cancer?

From studying the lives of people with PJS it has become apparent that cancer arises more frequently in people with PJS than in the general population. But not everyone with PJS will get cancer and those who do cannot predict where it will occur. Understandably people with PJS sometimes worry about this possibility and want to do everything possible to detect cancer at the earliest possible stage so that it can be treated when there is a good chance of a cure.

We recommend:

- ::: Bowel screening as described previously.
- ::: Breast screening as per the national screening programme.
- ::: Cervical smear tests as per the national screening programme.
- ::: Haemoglobin check every year.
- ::: Self examination of breasts and testes to check for unusual lumps.



We also recommend that any unusual lump or bump on the body should be taken seriously and not just be allowed to be considered 'one of those things'. These should be reported to the specialist doctor as soon as possible. Patients cared for at St Mark's should contact the Polyposis Registry so that an appointment can be arranged.

We do not, however, recommend:

- ::: Pelvic or testicular ultrasound scans.
- ::: CT scans of the pancreas.
- ::: The use of tumour markers.

The reason why we do not recommend these scans and tests is that they are not accurate screening tools. For example, all young women develop small cystic areas on their ovaries to do with their monthly cycle. We have found that the frequent interventions caused by the scans of the pelvis caused more harm than good. Likewise, CT scans supply radiation to the body (which might help induce cancer) and do not reliably pick up early cancers.

In conclusion, there is an increased risk of cancer in people with PJS. But many of these cancers can be diagnosed and treated early by testing for anaemia and sensible reporting of unusual lumps and bumps.



Glossary

Anus

the sphincter muscle at the very end of the gastrointestinal tract

Autosomal dominant

50:50 chance

Biopsy

a piece of tissue taken for examination under the microscope

Capsule endoscopy

examination of the bowel by a small pill camera that is swallowed

Chromosome

a length of coiled DNA

DNA

deoxyribonucleic acid, a complex chemical that contains the genetic code. Passed from one generation to the next

Elective

planned in advance

Gastrointestinal tract

the guts from mouth to anus

Gene

a genetic building block

Intussusception

telescoping of the bowel in on itself causing a blockage

Mutation

a change that has occurred in a gene

Polyp

a small swelling, arising from the lining of the bowel

Symptoms

what is complained about

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: kneale3@btconnect.com

Write to:

Professor RKS Phillips FRCS
or Miss Sue Clark FRCS

The Polyposis Registry
St Mark's Hospital
Watford Road
Harrow Middlesex
HA1 3UJ

