

## Who should I contact for further advice?

The first person you should ask if you have any health problems is your GP. It may be that he/she is not very familiar with this problem, so you should take along this fact sheet. If anyone in your family has PJS, then they will have a surgeon, and you could ask them for advice. Every region in the UK has a genetics service and they can be contacted for help via your GP or directly for advice.

## For more information:

If you need more advice about any aspect of PJS, you are welcome to 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, Lothian, South West Fife)

West of Scotland (main base Glasgow)

Tel: 0141 201 0808 Fax: 0141 201 0361

(Glasgow, Argyll & Bute, Ayrshire, Dumfries & Galloway, Stirling, Lanarkshire, Falkirk)

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

[www.maggiescentres.org](http://www.maggiescentres.org)

Maggies Highlands Tel: 01463 706302

Maggies Dundee Tel: 01382 496384

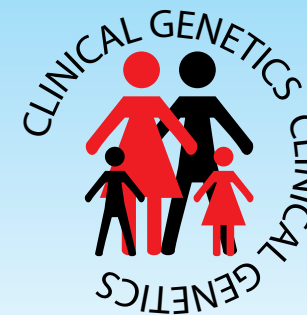
Maggies Edinburgh Tel: 0131 5373131

Maggies Glasgow Tel: 0141 330 3311

## Seen in clinic by.....

This leaflet was written by Clinical Genetics, Guys Hospital. Updated by Genetic Interest Group Scotland Updated June 2002

# Peutz-Jeghers Syndrome (PJS)



Information booklet

## What is Peutz–Jeghers Syndrome (PJS)?

PJS is a condition which runs in families. It causes little lumps called polyps to grow inside the bowel, especially the small bowel. These polyps do not usually cause any harm. However, if they are left inside the bowel for a long time (i.e. many years) one might become cancerous. They may occasionally block the bowel and cause pain. They can also cause bleeding and require surgery.

In people with PJS, small black flecks may be seen on the lips, fingers, toes and genital areas, although they are not always present and may fade in adult life.

## What is a polyp?

It is a little mushroom-like growth inside the bowel wall. They are benign growths which means they are NOT cancerous.

Screening the ovaries by vaginal ultrasound is therefore also offered. Men can develop a tumour of the testis and so regular testicular examinations should be carried out.

## Is there a cure?

The gene for PJS has now been found and researchers are trying to work out what job it does. This may help us to develop different ways of treating the condition. There is no way at present of correcting or replacing the faulty gene.

## Explanation of unfamiliar words?

<b>FAMILIAL</b>	Something which runs in families.
<b>POLYP</b>	A non-cancerous lump on the bowel wall.
<b>SMALL BOWEL</b>	The part of the bowel between the stomach and the large bowel, where most food digestion occurs.
<b>LARGE BOWEL</b>	The end section of the intestine or food pipe made up of colon and rectum.
<b>GENE</b>	One of the chemical recipes which control the working of the body.
<b>DOMINANT</b>	Only one of the pair of genes is faulty.
<b>COLONOSCOPY</b>	A short tube with a light at the end is passed into the rectum and the colon to look for polyps.

## Looking for polyps

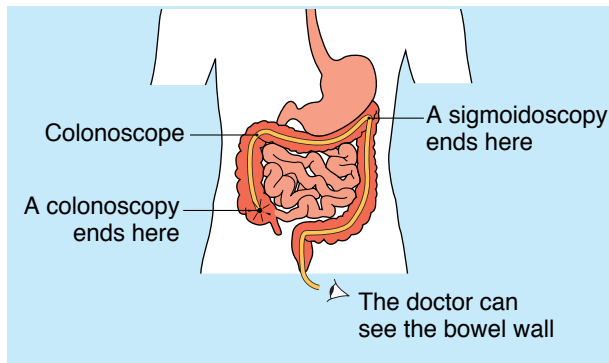
In some people, the polyps may cause bleeding or a painful blockage in the bowel. However, they do not always cause symptoms, so screening for them is important too.

A long, flexible tube can be used to examine the large bowel, and this is called a **colonoscope** (shown in the following picture).

Similar, examination of the stomach and upper part of the small bowel using an endoscope can screen for polyps.

An upper gastrointestinal endoscopy is performed by inserting a tube usually through the mouth, down through the stomach into the upper part of the small bowel. A barium meal and follow-through (x-ray after drinking a special fluid) is also offered.

### How the bowel is examined by colonoscopy and sigmoidoscopy



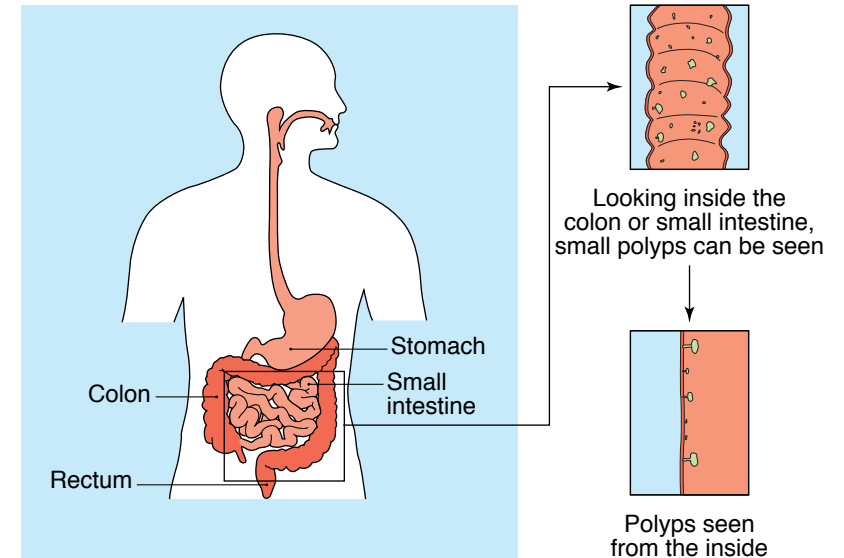
## What happens if polyps are found?

These may be removed during the endoscopy or colonoscopy, or may require surgery.

## Checking for another problem associated with PJS

Breast screening should be offered to adult women because breast cancer is a little more common in PJS. An unusual form of ovarian tumour can develop.

## The bowel



**Polyps may develop in any part of the small or large bowel (colon and rectum)**

## How does PJS run in families?

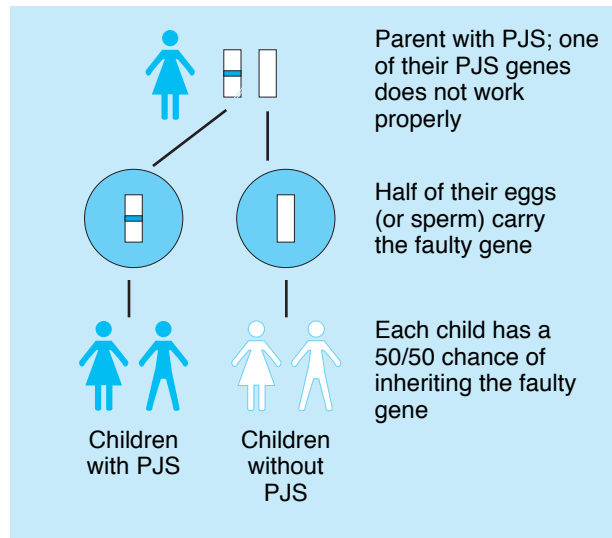
PJS is caused by a change in a gene. Genes are messages which control the working of the body and decide things like the colour of our eyes. Think of genes as recipes. The recipes allow the body to make chemical tools and building blocks. If part of the recipe is missing or has not been copied correctly, that part of the body will not be able to do its job properly.

Most genes come in matching pairs, with a copy of each gene coming from the mother's egg and the father's sperm.

It only takes one copy of the faulty gene to cause PJS.

## How PJS is passed from one generation to the next

If someone has PJS it means that they have a working copy and a faulty copy of the gene which causes the condition. Each time they have a child, there is a fifty-fifty chance of passing on the working copy or the faulty one.



How PJS is passed from one generation to the next

## Gene tests

If we think of a gene as a long word carrying a message, then the PJS gene has nearly 9000 letters in it. In different families it is a different letter or group of letters which have been wrongly copied which causes the gene not to work. The genetics team will try to work this out for each family, and if we find the "spelling" mistake we can offer a test to the rest of the family to find out who has it and who has not.

Blood cells contain copies of all your genes, so we can test the PJS gene from a blood sample.

## If the gene test shows:

- that someone does not have the faulty copy of the gene then they do not need bowel screening, and they cannot pass PJS onto their children.
- that someone does have the faulty copy, they need to continue with regular screening.

For some families we can offer a gene test, but not for all. This may be because we have not yet found the fault in the gene in that family, or it may be that no one is available, who has PJS, to test.

If a gene test cannot be done, you may be advised to carry on with regular screening.