Feedback

We appreciate and encourage feedback. If you need advice or are concerned about any aspect of care or treatment please speak to a member of staff or contact the Patient Advice and Liaison Service (PALS):

Freephone: 0800 183 0204
From a mobile or abroad: 0115 924 9924 ext 65412 or 62301
E-mail: pals@nuh.nhs.uk
Letter: NUH NHS Trust, c/o PALS, Freepost NEA 14614, Nottingham NG7 1BR

www.nuh.nhs.uk

Peutz-Jeghers Syndrome (PJS)

Information for patients

Clinical Genetics

This document can be provided in different languages and formats. For more information please contact:

Clinical Genetics Service
City Hospital Campus
The Gables, Gate 3
Hucknall Road
Nottingham NG5 1PB

Tel: 0115 962 7728
Email: nhu nt.clinicalgenetics@nhs.net

Rachel Harrison, Clinical Genetics © August 2018. All rights reserved. Nottingham University Hospitals NHS Trust. Review August 2020. Ref: 1017/v3/0818/AH.
What is Peutz-Jeghers Syndrome (PJS)?

Peutz-Jeghers Syndrome is a condition which runs in families. In people with PJS, small brown-coloured freckles may be seen on the lips, fingers, toes and genital areas, although they are not always present and may fade in adult life.

These are caused by areas of colour change in the skin and are not cancerous. PJS also causes polyps to develop in the gastrointestinal tract. Polyps are growths that look like lumps or bumps, but can grow to look a bit like cherries on a stalk.

These can start to develop from childhood or adolescence onwards and can grow anywhere throughout the intestines. There are many different types of polyps. The ones found in PJS are unique and are called Peutz-Jeghers polyps.

Is there a cure?

Researchers are trying to understand what the function of the STK11 is in the body and how this is changed in PJS. 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 some unfamiliar words

Familial - Something that runs in families.

Polyp - A non-cancerous lump on the bowel wall.

Small bowel - The part of the bowel between the stomach and the large bowel, where most food digestion occurs.

Large bowel - The end section of the intestine made up of colon and rectum.

Colonoscopy - Procedure in which a short tube with a light at the end is passed into the rectum and the colon to look for polyps.

Mutation - Change in a gene that stops it working properly.

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 they are not very familiar with this condition, so you can take along this leaflet.

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 and advice directly or via your GP.

Further information

www.polyposisregistry.org.uk/syndromes/peutz-jeghers/
Gene tests

Genetic testing uses a small sample of blood. Blood cells contain copies of all your genes. The genetic information (or ‘DNA’) is extracted from the blood cells and is tested to look for the genetic alteration for PJS. The inherited instruction in each gene is carried in a chemical code a little like an alphabet. The instruction in the PJS gene has nearly 9000 letters in it. If a letter or group of letters in the code are altered, this stops the gene working and is described as a mutation.

Different families affected with PJS can have different changes in the same gene. When someone is diagnosed with PJS they can have a genetic test to see if the gene change causing PJS in them can be detected. Often, but not always, the gene change can be identified. If a gene change is found then other family members can then be tested to see if they have also inherited PJS.

Genetic testing for the family

In families where the gene change causing PJS has been found other family members can have a genetic test.

- If a relative does not have the familial gene change then they have not inherited PJS. They do not need bowel screening and they cannot pass PJS onto their own children.
- If a relative does have the familial gene change then they do have PJS. They need to continue with regular screening of the bowel. Their own children may also inherit PJS.
- In some families a genetic test for relatives is not possible. This may be because we have not yet found the genetic spelling mistake in the family. If a gene test cannot be offered, relatives may be advised to carry on with regular screening.

Can polyps cause any problems?

Occasionally polyps can cause sliding and blocking of the bowel (intussusception) resulting in pain and possibly bleeding. This may need treatment in hospital and sometimes an operation. Sometimes people with PJS may have other symptoms such as diarrhoea or constipation, or they may develop anaemia (low red blood cell count).

People with PJS are offered regular checks of the stomach and intestines to look for polyps, so that they can hopefully be removed before they cause these problems.

Will I get cancer?

Studies have suggested that cancers of the gastrointestinal tract including the oesophagus, large bowel (also called colon and rectum), stomach and pancreas appear to be more common in adults with PJS than in the general population, particularly over the age of 50. Regular checks of the stomach and bowel in adults with PJS aim to detect any problems early, as well as looking for polyps. It is also recommended that people with PJS have their blood count checked each year.

Breast cancer is also thought to be more common in PJS. Adult women with PJS are currently eligible for extra breast screening from the age of 40 with a mammogram (X-ray of the breast) each year until the age of 59 when they can transfer to the National Breast Screening Programme. Women with PJS are also recommended to take part in the NHS cervical screening programme.

Research is continuing in this area and you should ask for current advice. Any unusual symptoms in an individual with PJS should be discussed with your GP so that they can be assessed appropriately.
Looking for polyps

Polyps do not always cause symptoms, so screening for them is important. Recommendations on the timing of screening vary and are altered when new information becomes available. This can be discussed in more detail with your gastroenterologist (doctor who specialises in managing bowel conditions) but the different techniques are described here.

A video capsule endoscopy can be done by swallowing a camera the size of a small coin or jelly baby. It takes pictures of the small bowel and transmits them to a box that you wear on a belt. The camera is passed in the stool (poo) and is not used again. This test is first offered at the age of 8. Sometimes a barium meal and follow-through (x-ray after drinking a special fluid) may be offered instead.

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

Similarly, an upper gastrointestinal endoscopy can be performed by inserting a tube, usually through the mouth, down through the stomach and into the upper part of the small bowel.

If polyps are found they may be removed during the endoscopy or colonoscopy, or may require surgery depending on their size.

How does PJS occur?

PJS is caused by a change in a gene called STK11. Genes are the unique instructions which control the workings of the body and make each of us an individual. If the instruction in the STK11 gene is altered (or ‘mutated’), it no longer makes sense and cannot perform its normal job properly.

We all have two copies of the STK11 gene. One copy is inherited from each parent. If one copy of the gene is altered, this is enough to cause PJS, even though the other copy is normal.

How is PJS passed from one generation to the next?

When we have children, we pass on one copy of each of our genes. Someone with PJS has one working copy and an altered copy of the STKL11 gene that causes the condition. Each time they have a child there is a 50% chance of passing on the working copy, and a 50% chance of passing on the altered copy of the gene.

This means that each of their children has a 50%, or one in two chance of inheriting the altered gene and being affected by PJS. This is the same for each child, regardless of their sex.