CONDITION

Peutz-jeghers syndrome

Peutz-Jeghers syndrome is an inherited condition causing digestive-tract polyps, skin pigmentation, and increased cancer risks.

WHAT IT IS
Inherited syndrome causing digestive polyps and increased cancer risks

BODY SYSTEM
Digestive system, skin, and several organs

MAIN SYMPTOMS
Dark skin spots, abdominal pain, bleeding, bowel blockage

WHAT CAUSES IT
Inherited change in the STK11 gene

HOW IT IS DIAGNOSED
Clinical findings, polyp examination, and genetic testing

MAIN TREATMENT
Polyp removal and lifelong cancer surveillance

CAN IT BE CURED?
No - the inherited gene change remains

IS IT AN EMERGENCY?
Sometimes - severe pain, vomiting, or heavy bleeding

LONG-TERM OUTLOOK
Often manageable with regular surveillance and timely treatment

Overview

Peutz-Jeghers syndrome, often shortened to PJS, is a rare inherited condition. It mainly causes distinctive dark spots on the skin and lips and a type of growth called a hamartomatous polyp in the digestive tract.

A hamartomatous polyp is an overgrowth of tissue normally found in that part of the body. These polyps are usually not cancer. However, they can bleed, cause anaemia, or block the bowel. They can also make one section of bowel slide inside another, a problem called intussusception.

People with PJS also have an increased lifetime risk of several cancers. These include cancers of the bowel, stomach, pancreas, breast, ovaries, cervix and testicles. This does not mean that everyone with PJS will develop cancer. It does mean that planned, lifelong screening is

important.

PJS affects people differently, even within the same family. Some people have symptoms during childhood, while others are diagnosed after a relative is found to have the condition.

Symptoms

Small, flat, dark blue, brown or black spots are often one of the earliest signs. They commonly appear during childhood around the lips, inside the mouth, near the nose and eyes, and on the fingers or toes.

Spots on the face may fade after puberty. Those inside the mouth often remain. The spots are harmless and do not become skin cancer.

Polyps can grow in the stomach, small bowel and large bowel. They occur most often in the small bowel. Possible symptoms include:

Anaemia means there are not enough healthy red blood cells to carry oxygen. In PJS, it can develop slowly because a polyp is bleeding.

Sudden, severe abdominal pain may be caused by intussusception or another bowel blockage. Young children may draw their knees toward their chest, become unusually sleepy, vomit, or have episodes of intense crying.

Some people have no obvious digestive symptoms between screening appointments. Lack of symptoms does not mean that polyps are absent.

- Cramping or repeated abdominal pain
- Nausea or vomiting
- Blood in the stool or black, tar-like stool
- Tiredness, weakness or breathlessness caused by anaemia
- Constipation, bloating or difficulty passing gas
- A polyp protruding from the rectum

Causes and risk factors

PJS is usually caused by a harmful change in a gene called STK11 , also known as LKB1 . This gene normally helps control how cells grow and divide.

The condition follows an autosomal dominant inheritance pattern. This means a person with PJS has a 1 in 2 chance of passing the altered gene to each biological child. The chance is the

same for children of any sex.

Some people inherit the gene change from a parent. In others, it begins as a new genetic change, so there may be no previous family history.

A person can sometimes meet the clinical criteria for PJS even when genetic testing does not find an STK11 change. Current tests may not identify every type of genetic alteration.

Lifestyle does not cause PJS. Diet, stress and everyday activities do not create the underlying gene change.

Diagnosis

Diagnosis is based on your medical history, family history, pigmentation, digestive polyps and, when available, genetic test results.

A removed polyp is examined under a microscope. PJS polyps have a characteristic branching pattern of smooth muscle within them. This helps distinguish them from other types of bowel polyp.

Clinical diagnosis may be possible when there are:

Genetic testing is usually performed on a blood or saliva sample. Finding a disease-causing STK11 change can confirm the diagnosis and allows relatives to be tested for that specific change.

Tests used to assess the digestive tract may include an upper endoscopy, colonoscopy, capsule endoscopy or magnetic resonance imaging of the small bowel. Capsule endoscopy involves swallowing a small camera. It may not be suitable if a bowel blockage is suspected.

Children and adults with a possible diagnosis are often referred to a genetics or hereditary cancer service. Genetic counselling can help explain test results, inheritance and what the findings may mean for relatives.

- Two or more confirmed PJS-type polyps
- PJS-type polyps in someone with a close relative who has the syndrome
- Characteristic pigmentation in someone with an affected close relative
- Both characteristic pigmentation and a PJS-type polyp

Treatment and management

There is no treatment that removes the underlying inherited tendency. Management focuses

on finding and removing significant polyps, treating complications, and screening for cancer.

Larger polyps are often removed during an endoscopy. An endoscope is a flexible camera passed through the mouth or rectum. Specialised endoscopy can reach farther into the small bowel.

Surgery may be needed when a polyp cannot be removed safely by endoscopy or when there is intussusception, bowel blockage, tissue damage or uncontrolled bleeding. When possible, treatment aims to preserve bowel length because repeated operations can lead to long-term digestive problems.

Iron replacement may be used when ongoing blood loss has caused iron-deficiency anaemia. Severe bleeding may require additional hospital treatment.

Lifelong surveillance usually includes planned examinations of the stomach, small bowel and colon. The timing depends on age, earlier findings, symptoms and the number and size of polyps.

Cancer surveillance may also cover the breasts, pancreas and reproductive organs. Depending on age and anatomy, this can involve breast imaging, pancreatic imaging, gynaecological assessment or examination of the testicles. Pancreatic screening is generally organised through a centre experienced in inherited cancer risk.

Surveillance plans should be recorded and reviewed rather than relying only on symptoms. Recommendations can differ between specialist guidelines and may change as new evidence appears.

Complications

The most common serious digestive complication is intussusception. A polyp can pull one part of the intestine into the next, much like sections of a telescope. This can block the bowel and may interrupt its blood supply.

Other possible complications include:

Short bowel syndrome occurs when too little working small intestine remains to absorb enough fluid and nutrients. Modern surveillance and endoscopic polyp removal aim to reduce the need for repeated bowel surgery.

The cancer risk is lifelong and affects several organs. A new symptom is not automatically cancer, but persistent changes should be assessed rather than left until the next planned screening appointment.

- Bleeding from a polyp

- Iron-deficiency anaemia
- Complete or partial bowel blockage
- Repeated abdominal operations
- Short bowel syndrome if large amounts of intestine are removed
- Cancer in one of the organs associated with PJS

Prevention

PJS itself cannot be prevented because it is caused by an inherited or newly occurring gene change.

Regular surveillance cannot remove the genetic risk, but it may prevent some bowel emergencies by finding larger polyps before they cause blockage or intussusception. Screening also aims to detect cancer at an earlier, more treatable stage.

If an STK11 change has been identified in a family, adult relatives can choose predictive genetic testing. A relative who has not inherited that specific change will usually not need PJS surveillance.

People planning a pregnancy may choose to discuss reproductive options with a genetics service. These can include testing during pregnancy or testing embryos created through in vitro fertilisation. These are personal choices, not required steps.

General measures such as avoiding tobacco, limiting alcohol and maintaining a balanced diet support overall health. They do not eliminate the cancer risks caused by PJS and should not replace specialist surveillance.

Living with it and what to expect

PJS is lifelong, but symptoms often come and go. Pigmented facial spots may become less noticeable with age, while the tendency to develop polyps and the increased cancer risks remain.

During childhood and adolescence, care often focuses on detecting bowel polyps and preventing intussusception. Some children need endoscopic procedures or surgery. Others have few symptoms but still need scheduled checks.

In adulthood, surveillance expands to address cancer risks in several organs. Appointments and tests can take time, especially when different specialists are involved. A written

surveillance plan can help keep track of what is due.

Many people continue with school, work, family life, exercise and travel. Planning may be needed around procedures or episodes of abdominal symptoms. It can be useful to carry a brief medical summary explaining the diagnosis and the risk of intussusception, especially when travelling.

Repeated testing and uncertainty about cancer can cause anxiety. Genetics services, hereditary cancer clinics, counsellors and patient support organisations may help. Family members may also have different feelings about whether to pursue genetic testing.

The course cannot be predicted for one person from another relative's experience. Some people need frequent polyp removal, while others need fewer procedures. Regular follow-up allows the plan to respond to new findings over months and years.

When to get medical help

Arrange routine medical follow-up if you have PJS and:

These symptoms have several possible causes, but they should be assessed rather than saved for a distant screening visit.

Seek urgent medical care for worsening abdominal pain, repeated vomiting, marked abdominal swelling, or inability to pass stool or gas. These may indicate a bowel blockage.

Get emergency help for sudden severe abdominal pain, fainting, vomiting blood, heavy rectal bleeding, confusion, or signs of shock such as pale, cold or clammy skin. A child with severe episodic pain, repeated vomiting, unusual sleepiness or bloody stool also needs emergency assessment.

- Your surveillance appointments are overdue
- Abdominal discomfort keeps returning
- You notice small amounts of blood in your stool
- You develop increasing tiredness or breathlessness
- A new breast, testicular or abdominal lump appears
- Your bowel habits change and do not return to normal

Common questions

Is Peutz-Jeghers syndrome cancer?

No. PJS is an inherited condition, not a cancer diagnosis. Most PJS polyps are non-cancerous. However, people with the syndrome have increased risks of several cancers and need lifelong surveillance.

Do all Peutz-Jeghers polyps need to be removed?

Not necessarily. Specialists consider each polyp's size, position, growth and likelihood of causing bleeding or blockage. Larger or troublesome polyps are more likely to be removed.

Can the dark spots be removed?

The spots are harmless and often do not need treatment. Facial spots may fade naturally. Cosmetic laser treatment may be possible, but pigmentation can return and treatment does not affect internal polyps or cancer risk.

Can I pass PJS to my children?

If you have a disease-causing STK11 change, each biological child has a 1 in 2 chance of inheriting it. The outcome of one pregnancy does not change the chance for another.

Can I have PJS if my genetic test is negative?

Yes. A negative test does not always rule it out. Some people meet clinical criteria based on their polyps, pigmentation and family history even when current testing does not identify an STK11 change.

What foods should I avoid with Peutz-Jeghers syndrome?

There is no specific diet proven to stop PJS polyps from forming. Food choices may need temporary adjustment after a procedure or if narrowing of the bowel causes symptoms. A specialist or dietitian can advise if this applies to you.

Sources

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