



Family Education Sheet

Peutz-Jeghers Syndrome (PJS)

What is Peutz-Jeghers Syndrome (PJS)?

- PJS is a gastrointestinal (GI) syndrome characterized by the development of multiple polyps (abnormal growths or tumors) in the body. In PJS, the polyps are called hamartomas.
- Polyps in PJS can be found anywhere in the GI tract – from the stomach to the rectum – but they are mostly found in the small intestine.
- Patients with PJS may have dark-blue or black spots (called pigmentation) on the lips and inside the mouth. These spots are less frequently found on the hands and feet.

What is a polyp?

- A gastrointestinal polyp is an abnormal growth of tissue that is either attached to the intestinal wall by a stalk (pedunculated) or growing directly from the wall (sessile).

How common is PJS?

- PJS occurs in approximately 1 in 150,000 people and affects both males and females equally.

What causes PJS?

- PJS is usually caused by a mutation of the serine threonine kinase (*STK11*) gene (also called the *LKB1* gene).
- The gene mutation can happen in two ways:
 1. It can be passed down from a parent.
 2. It can be a new mutation in the *STK11/LKB1* gene.

What are the symptoms and possible complications of PJS?

- Individuals may begin to experience symptoms of PJS within the first ten to fifteen years of life. Abnormal signs and symptoms that may develop in the course of the disease include:
 - Abdominal pain
 - Rectal bleeding
 - Diarrhea
 - Anemia
- PJS patients have an increased risk of developing GI and non-GI related cancers during adulthood. These types of cancers are listed below in order from most common to least common in PJS:
 - **Gastrointestinal (GI) cancers**
 - Small intestine
 - Stomach
 - Large intestine
 - Esophagus

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➤ **Non-gastrointestinal cancers**

- Breast
 - Pancreas
 - Ovary
 - Lung
 - Uterus
- The greatest threat in PJS is GI cancer and breast cancer.
 - Dark-blue or black spots on the skin are a common sign of PJS (found in more than 95% of cases) and can be seen within the first two years of birth, long before polyps are found. These spots may be found in the following areas of the body:
 - Lips
 - Inside the mouth (buccal mucosa)
 - Hands
 - Feet
 - It is possible for the pigmentation to fade as the child grows into an adult.

Who should be screened for PJS?

- You should be screened for PJS if you have:
 - A family history of PJS or any intestinal cancer at a young age, or
 - Any of the unusual signs listed above.
- An initial screening should be done:
 - By 10-12 years of age, or
 - At the time of first symptoms.

How are patients screened for PJS?

- Patients are screened for PJS by performing a colonoscopy. A doctor uses a long, flexible tube with a light and camera on the end to look inside the rectum and up into the large intestine. An upper GI endoscopy is often done at the same time. An upper GI endoscopy also uses a long, flexible tube but looks down into the esophagus, stomach, and small intestine.
- Another way to screen for PJS is by genetic testing. Identifying the PJS-mutation in the *STK11* gene is very accurate, with a detection rate of more than 90%.
- Other screening studies may include radiology imaging.

How is PJS treated?

- Most polyps are treated by removing them with an endoscope, a procedure known as a polypectomy. However, if the polyps are very large or if they present a risk for cancer, then surgery may be necessary.
- There is no cure for PJS. Treatment is directed to reduce complications, such as abdominal pain, bowel obstruction, GI bleeding, and cancer.

A ***Spanish*** version of this is available from your provider

Send comments or questions to: Familyed@childrens.harvard.edu
