Children's Hospital Boston

Family Education Sheet

Juvenile Polyposis Syndrome (JPS)

What is Juvenile Polyposis Syndrome?

- JPS is a gastrointestinal (GI) syndrome characterized by the development of multiple polyps (abnormal growths or tumors) in the body. In JPS, the polyps are called juvenile or inflammatory polyps.
- The term "juvenile" refers to the type of polyp, not the age of when the polyps occur. However, most individuals with JPS have some polyps by the age of 20.
- The number of polyps will vary for each individual. Some people may have only four or five polyps, whereas others may have more than a hundred.
- Polyps in JPS can be found anywhere in the GI tract – from the stomach to the rectum - but they are mostly found in the large intestine.

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 JPS?

- JPS occurs in approximately 1 in 100,000 people and affects both males and females equally.

What causes JPS?

- JPS is usually caused by a mutation in one of two possible genes: *SMAD4* or *BMPR1A* gene.
- The gene mutation can happen in two ways:
  1. It can be passed down from a parent (75% of cases).
  2. It can be a new mutation in the *SMAD4* or *BMPR1A* gene (25% of cases).

What are the symptoms and possible complications of JPS?

- Individuals may begin to experience symptoms of JPS 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
  - Bowel obstruction/Constipation
  - Visible rectal polyp
  - Anemia
- Most polyps in JPS are non-cancerous (benign). However, polyps can change and become cancerous.
- Colon cancer is the most serious risk of JPS, with up to a 50% chance of developing the disease during a patient's lifetime.

- JPS patients have an increased risk of developing other GI and non-GI related cancers during adulthood. These include: stomach, small intestine, esophagus, and pancreatic cancer.

Who should be screened for JPS?

- You should be screened for JPS if you have:
  - A family history of JPS or colon cancer at a young age, or
  - Any of the unusual conditions listed above.
- An initial screening should be done:
  - By 8-10 years of age for a high-risk individual, or
  - By 15 years of age for an average-risk individual, or
  - At the time of first symptoms.
- Average and high-risk individuals are determined based on family history and/or known high-risk gene mutations.

How are patients screened for JPS?

- Patients are screened for JPS 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 JPS is by genetic testing of the *SMAD4* or *BMPR1A* gene.
- Other screening studies may include radiology imaging (X-rays, CAT scan, MRI, ultrasound, etc.).

How is JPS 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 JPS. 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***