Cleveland Clinic
Genomic Medicine Institute

Juvenile polyposis syndrome (JPS)

Juvenile polyposis syndrome (JPS) is a condition associated with the development of juvenile polyps in the intestinal tract and an increased risk of bowel cancer. JPS can usually be distinguished from other conditions by the number, location, age of onset, and pathology of the polyps. Approximately 75% of individuals with JPS will have a family history of the condition. The remaining 25% of individuals are the first person in their family to develop JPS.

The polyps in JPS are usually benign and develop during the first or second decade of life. The most common presenting symptom is painless rectal bleeding. Other warning signs include polyps protruding in the rectum, anemia, diarrhea, and chronic weight loss. Patients may have anywhere from 5 to 500 juvenile polyps, mostly in the large bowel and rectum. Juvenile polyps can also be found in the stomach and small intestine. The polyps have a distinctive stalk-like appearance and are round with a normal, *smooth* outer surface. There appears to be a high risk for these polyps to reappear after surgical removal and an increased risk of bowel cancer. Individuals with JPS have a 9-68% lifetime risk for the development of colorectal cancer and have been reported to have an increased risk for gastric (stomach), duodenal (part of the small intestine), and pancreatic cancers. Birth defects such as congenital heart defects, cleft lip or palate, and malrotations (abnormal placement of internal organs) have been described in 11-20% of individuals with JPS.

Individuals with JPS due to *SMAD4* mutations are also at risk to have hereditary hemorrhagic telangiectasia syndrome a condition associated with nosebleeds; skin and mucosal telangiectases; pulmonary, cerebral, and hepatic arteriovenous malformations (AVMs); and increased risk of hemorrhage due to these malformations.

AVMs are blood vessels that lack capillaries. This means that the artery connects directly to the vein. Small AVMs are called telangiectases and can occur on the lips, fingers, nose, tongue, and gastrointestinal tract. Telangiectases have thin walls, which allow them to rupture and bleed more easily. Large AVMs typically occur in the brain, lungs, and liver and can cause catastrophic events such as thrombosis or embolus. Symptoms of lung AVMs may include migraine headache and digital clubbing. Often the first symptom of HHT is recurrent nosebleeds beginning around 12 years of age. Telangiectases often do not develop until the early 30s. If an individual has gastrointestinal bleeding, it is often difficult to determine if the cause of the bleed is a juvenile polyp or telangiectasia. Anemia may result from GI bleeding.

The Center for Personalized Genetic Healthcare