

Juvenile Polyposis Syndrome

This section has been reviewed and approved by the [Cancer.Net Editorial Board \[1\]](#), May / 2012

Overview

What is juvenile polyposis syndrome?

Juvenile polyposis syndrome (JPS) is a hereditary condition that is characterized by the presence of hamartomatous polyps in the digestive tract. Hamartomas are noncancerous (benign) masses of normal tissue that build up in the intestines or other places. These masses are called polyps if they develop inside a body structure, such as the intestines. The term juvenile polyposis refers to the type of polyp (juvenile polyp) that is found after examination of the polyp under a microscope, not the age at which people are diagnosed with JPS.

Polyps may frequently develop in a person with JPS by age 20. The number of polyps a person has during his or her lifetime can range from around five to more than 100. Most juvenile polyps are benign (noncancerous), but there is an increased risk of cancer of the digestive tract ([stomach \[2\]](#), [small intestine \[3\]](#), [colon \[4\]](#), and [rectum \[4\]](#)) in families with JPS.

JPS is suspected when a person's symptoms and family history fit one of the following categories:

- More than five juvenile polyps of the colon and/or rectum
- Multiple juvenile polyps throughout the digestive tract
- Any number of juvenile polyps and a family history of juvenile polyps

What causes JPS?

JPS is a genetic condition. This means that the risk for polyps and cancer can be passed from generation to generation in a family. Two genes have been linked to JPS. They are called *BMPR1A* and *SMAD4*. A mutation (alteration) in either the *BMPR1A* gene or the *SMAD4* gene makes a person more likely to develop juvenile polyps and cancer of the digestive tract over his or her lifetime. Not all families that have JPS will have mutations in *BMPR1A* or *SMAD4*. Other genes are being studied regarding their link to JPS.

How is JPS inherited?

Normally, every cell has two copies of each gene: one inherited from the mother and one inherited from the father. JPS follows an autosomal dominant inheritance pattern, in which a mutation needs to happen in only one copy of the gene for the person to have an increased risk of getting that disease. This means that a parent with a gene mutation may pass along a copy of their normal gene or a copy of the gene with the mutation. Therefore, a child who has a parent with a mutation has a 50% chance of inheriting that mutation. A brother, sister, or parent of a person who has a mutation also has a 50% chance of having the same mutation.

How common is JPS?

It is estimated that between one in 16,000 and one in 100,000 people has JPS.

How is JPS diagnosed?

A diagnosis of JPS is assumed if a person's symptoms and family history fits one of the three categories listed above. People who have JPS can have a blood test to look for a mutation in the *BMPR1A* gene or the *SMAD4* gene. If a specific gene mutation is found, other family members may be diagnosed with JPS if they are tested and have the same gene mutation.

It appears likely that there are other genes associated with JPS that have not yet been identified, so a blood test result that comes back as ?negative? (meaning a gene mutation cannot be found) does not necessarily mean that a person does not have JPS. Therefore, meeting with a health professional who specializes in genetics, such as a genetic counselor or medical geneticist (a doctor with training in genetic diseases and conditions), is recommended for people who have a family history or symptoms that suggests JPS.

What are the estimated cancer risks associated with JPS?

People with JPS are considered to be at an increased risk for [colorectal](#) [4], [stomach](#) [2], [small intestine](#) [3], and [pancreatic cancers](#) [5]. The overall estimated cancer risk associated with JPS is 9% to 50%, but the risks for each specific type of cancer have not been determined.

What are the screening options for JPS?

It is important to discuss with your doctor the following screening options, as each individual is different:

- Any signs of rectal bleeding, anemia, abdominal pain, constipation, diarrhea, or other changes in the stool should be brought to the attention of a doctor and evaluated.
- A complete blood count (CBC [6], a blood test), [colonoscopy](#) [7], and [upper endoscopy](#) [8] should be done at age 15 (or earlier if there are symptoms); if tests are normal, they should be repeated every three years.
- Screening should be done every year. If polyps are found, they should be removed.
- Individuals who develop large numbers of polyps may need to have surgery to remove part of the colon or stomach.

Screening options may change over time as new technologies are developed and more is learned about JPS. It is important to talk with your doctor about appropriate screening tests.

Learn more about [what to expect when having common tests, procedures, and scans](#) [9].

Questions to ask the doctor

If you are concerned about your risk of [colorectal cancer](#) [4] or other types of cancer, talk with your doctor. Consider asking the following questions of your doctor:

- What is my risk of developing cancer in the digestive tract?
- How many colon polyps have I had in total?
- What type of colon polyps have I had? (The two most common types are hyperplastic polyps [noncancerous growths in the lining of the colon] and adenomatous polyps [growths in the lining of the colon that can become cancerous].)
- What can I do to reduce my risk of cancer?
- What are my options for cancer screening?

If you are concerned about your family history and think your family may have JPS, consider asking the following questions:

- Does my family history increase my risk for [colorectal cancer](#) [4] or other types of cancer?
- Should I meet with a genetic counselor?
- Should I consider [genetic testing](#) [10]?

Additional resources

[Guide to Colorectal Cancer](#) [4]

[The Genetics of Colorectal Cancer](#) [11]

[Guide to Stomach Cancer](#) [2]

[Guide to Small Bowel Cancer](#) [3]

[Guide to Pancreatic Cancer](#) [5]

[What to Expect When You Meet With a Genetic Counselor](#) [12]

Colon Cancer Alliance

www.ccalliance.org [13]

C3: Colorectal Cancer Coalition

www.fightcolorectalcancer.org [14]

To find a genetic counselor in your area, ask your doctor or visit the following websites:

National Society of Genetic Counselors

www.nsgc.org [15]

National Cancer Institute: Cancer Genetics Services Directory

www.cancer.gov/cancertopics/genetics/directory [16]

Links:

[1] <http://www.cancer.net/about-us>

[2] <http://www.cancer.net/node/19645>

[3] <http://www.cancer.net/node/19632>

[4] <http://www.cancer.net/node/18701>

[5] <http://www.cancer.net/node/19495>

[6] <http://www.cancer.net/node/24716>

[7] <http://www.cancer.net/node/24481>

[8] <http://www.cancer.net/node/24731>

[9] <http://www.cancer.net/node/24959>

[10] <http://www.cancer.net/node/24895>

[11] <http://www.cancer.net/node/24898>

[12] <http://www.cancer.net/node/24907>

[13] <http://www.ccalliance.org>

[14] <http://www.fightcolorectalcaner.org/>

[15] <http://www.nsgc.org>

[16] <http://www.cancer.gov/cancertopics/genetics/directory>