UT Southwestern Harold C. Simmons Comprehensive Cancer Center | NCI Designated Comprehensive Cancer Center

Juvenile polyposis syndrome (JPS)

What You Should Know About Juvenile polyposis syndrome (JPS)

Individuals with a mutation in the *BMPR1A* gene or the *SMAD4* gene have juvenile polyposis syndrome (JPS), a condition characterized by increased risks for polyps in the gastrointestinal (GI) tract. The term "juvenile" refers to the type of polyp rather than to the age of onset of polyps. Most juvenile polyps are benign; however, cancer can occur. If polyps are left untreated, they may cause bleeding and anemia. Individuals with *SMAD4* mutations also have hereditary hemorrhagic telangiectasia (HHT), which can cause malformed blood vessels in many organs of the body.

Cancer Risks Associated with Juvenile polyposis syndrome (JPS)

- Most individuals with JPS will have polyps by age 20. The number of polyps in one's lifetime can very between less than 10 to greater than 100. Individuals in the same family may have varying numbers of polyps.
- Individuals with JPS are at a 40-50% lifetime risk to develop colon cancer, and a 21% lifetime risk to develop stomach cancer if they have multiple stomach polyps.
- An increased risk of small intestine and pancreatic cancer has been suggested for individuals with JPS; however, exact lifetime risks are unknown
- Individuals with *SMAD4* mutations are at increased risk to develop HHT, a condition that causes abnormal blood vessel growth in the skin, lungs, liver, brain, and other organs.

Risks to Family Members

Juvenile Polyposis Syndrome (JPS) is inherited in an autosomal dominant fashion, and is caused by mutations in either the *BMPR1A* gene or *SMAD4* gene. This means that children, brothers, sisters, and parents of individuals with a *BMPR1A*/*SMAD4* mutation have a 1 in 2 (50%) chance of having the mutation as well. Individuals with a *BMPR1A*/*SMAD4* mutation may develop colon polyps, one cancer, more than one cancer, or none at all. Both males and females can inherit a familial *BMPR1A*/*SMAD4* mutation and can pass it on to their children.

Managing Cancer Risks

The following surveillance is recommended by the National Comprehensive Cancer Network (v3.2019):

- Colonoscopy and upper endoscopy beginning at age ~15 years
    - Repeat annually if polyps are found
    - Repeat every 2-3 years if no polyps are found
- No recommendations have been made for screening of the small intestine or pancreas. Individuals with JPS should discuss their personal and family history with their healthcare provider to determine an appropriate screening regimen.
- In individuals with *SMAD4* mutations, screening for vascular lesions associated with HHT should being within the first 6 months of life.

Last updated 1/9/2020

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