

Peutz-Jeghers syndrome is caused by inherited mutations in the STK11 gene. This condition is associated with increased risk for several types of cancer, as well as other characteristic features.

Mutations in STK11 are inherited in an [autosomal dominant](#) pattern, meaning that children of a mutation carrier each have a 50% risk to inherit the mutation and associated cancer risks. Notably, women and men both have the STK11 gene and have the same chances to inherit and pass down mutations in this gene. Therefore, both sides of the family are important when assessing inherited risk. However, the associated cancers and lifetime risks are different between men and women.

The cancer risks in Peutz-Jeghers syndrome include:

<b>Cancer Type</b>	<b>Risk in the general population</b>	<b>Risk in STK11 mutation carriers</b>
Female Breast	12%	45-50%
Colon	5.5%	39%
Stomach	0.8%	29%
Small intestine	0.3%	13%
Pancreas	1.6%	11-36%
Ovarian	1-2%	18-21%
Cervical	0.6%	10%
Uterine (Endometrial)	2.8%	9%
Lung	6.4%	15-17%

Other characteristics of Peutz-Jeghers syndrome include:

- Polyps (hamartomatous type) in the small intestine
- Freckles in unusual places (in/around the mouth, nose, eyes, fingers, groin)

### **How is Peutz-Jeghers syndrome diagnosed?**

Peutz-Jeghers syndrome is diagnosed if your personal and family history meets established clinical criteria (based on medical history, physical examination, and family history). Currently, the criteria for diagnosis, as determined by the [National Comprehensive Cancer Network \(NCCN\)](#), includes confirmation of two or more of the following in an individual:

- At least two Peutz-Jeghers-type hamartomatous polyps in the small intestine

- Mucocutaneous hyperpigmentation (freckles) of the mouth, lips, nose, eyes, genitals, or fingers
- Family history of Peutz-Jeghers syndrome

This condition can also be diagnosed by identification of a mutation in the STK11 gene through genetic testing.

### Genetic Testing for Peutz-Jeghers syndrome

A diagnosis of Peutz-Jeghers syndrome may also be made by identification of a mutation in the STK11 gene. Genetic testing can be ordered with different approaches, depending on the history and available information:

- [Single site analysis](#): Testing specific to a known mutation in the family
- [Full gene sequencing and rearrangement analysis](#): Comprehensive testing to search for all currently detectable mutations in the genes. If past testing included full gene sequencing but not the rearrangement analysis, this additional testing can be ordered to evaluate for these types of mutations that would have been missed on older testing.
- [Gene panels](#): Newer, more broadly based gene tests that would include not only the STK11 gene, but other genes known or suspected to be associated with increased cancer risks

### Cancer screening and risk management for Peutz-Jeghers syndrome

If genetic testing confirms a known mutation in the STK11 gene, it is recommended to discuss your management plan with your healthcare team, and if available, to seek consultation through a specialized high risk clinic. General recommendations are included here based on the updated guidelines of the ([NCCN](#)), but may be tailored to your specific medical and family history.

*Breast cancer*: starting at age 25, women should have annual mammograms and breast MRIs, and should have a clinical breast exam every 6 months.

*Colon cancer*: starting in the late teens, men and women should have colonoscopy screening every 2-3 years

*Stomach cancer*: starting in the late teens, men and women should have upper endoscopy screening every 2-3 years

*Small intestine cancer*: starting at age 8-10, boys and girls should have small bowel

screening (CT, MRI or video capsule endoscopy), with follow up by age 18 (or sooner if first screen found something), then repeat every 2-3 years

*Pancreatic cancer:* starting at age 30-35, men and women should have MRI or endoscopic ultrasound every 1-2 years

*Ovarian, cervical, uterine cancers:* starting at age 18-20, women should have annual pelvic exam and Pap smear; should also consider transvaginal ultrasound

*Testicular cancer:* starting at age 10, boys should have annual testicular exam; should also observe for feminizing changes (e.g. breast growth)

*Lung cancer:* Education on symptoms and smoking cessation