

Peutz-Jeghers Syndrome (*STK11* mutations)

What You Should Know About Peutz-Jeghers Syndrome

Individuals who inherit a mutation in the *STK11* gene have a diagnosis of Peutz-Jeghers Syndrome (PJS). *STK11* mutations are associated with an increased lifetime risk for developing gastrointestinal polyps as well as breast (female), colon, stomach, small bowel, pancreatic, gynecologic (ovarian, cervical, uterine), testicular, and lung cancers. Individuals with PJS can also have hyperpigmentation (freckling) on the fingers, around the mouth, eyes, nose, and perianal area. The freckling is typically more pronounced in childhood, and begins to fade in puberty and adulthood.

Cancer Risks Associated with Peutz-Jeghers Syndrome (*STK11* mutations)

Cancer Type	<i>STK11</i> Mutation Carrier Cancer Risks	General Population Lifetime Cancer Risks
Female Breast	32-54%	12.4%
Colon	39%	4.5%
Stomach	29%	<1%
Small Intestine	13%	<1%
Pancreas	11-36%	<1%
Gynecologic	Ovarian: 18-21%	1-2%
	Cervical: 10%	<1%
	Uterine: 9%	2.7%
Testes (sex cord/Sertoli cell tumors)	9%	<1%
Lung	7-17%	6%

Risks to Family Members

Mutations in the *STK11* gene are inherited in an autosomal dominant fashion. This means that children, brothers, sisters, and parents of individuals with a *STK11* mutation have a 1 in 2 (50%) chance of having the mutation as well. Individuals with a *STK11* mutation may develop one cancer, more than one cancer, or none at all. Both males and females can inherit a familial *STK11* mutation and can pass that it on to their children.

Managing the Risks

The following recommendations are based on the NCCN Colorectal Cancer Guidelines version 1.2020

Breast Cancer

- Clinical breast exams every 6 months beginning at age 25.
- Annual mammography and breast MRI beginning at age 25

- Risk reducing options including prophylactic mastectomy or chemoprevention medication are not indicated based on *STK11* mutation alone, but can be considered based on personal risk factors and family history.

Colon, Stomach, Small Intestine Polyps/ Cancers

- Begin colonoscopy screening at age 8 and repeat every 2-3 years if polyps are found; if no polyps found in childhood begin screening at age 18 and repeat every 2-3 years
- Upper endoscopy every 2-3 years starting in late teens (or age 8 years)
- Small bowel visualization (CT or MRI enterography or video capsule endoscopy) every 2-3 years starting between ages 8-10

Pancreatic Cancer

- Annual screening (using MRI/Magnetic Resonance cholangiopancreatography with contrast and/or endoscopic ultrasound) beginning at age 30-35 (or 10 years younger than the earliest diagnosis in the family).

Ovarian, Cervical, Uterine Cancers

- Annual pelvic examination and Pap smear beginning at age 18-20.

Testicular Cancer

- Annual testicular exam beginning at age 10.

Lung Cancer

- Avoid smoking; education regarding symptoms

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