Ambry Genetics®

Clinician Management Resource for *STK11* (Peutz-Jeghers syndrome)

This overview of clinical management guidelines is based on this patient's positive test result for a *STK11* gene mutation. Unless otherwise stated, medical management guidelines used here are limited to those issued by the National Comprehensive Cancer Network® (NCCN®)[1,2] in the U.S. Please consult the referenced guideline for complete details and further information.

Clinical correlation with the patient's past medical history, treatments, surgeries and family history may lead to changes in clinical management decisions; therefore, other management recommendations may be considered. Genetic testing results and medical society guidelines help inform medical management decisions but do not constitute formal recommendations. Discussions of medical management decisions and individualized treatment plans should be made in consultation between each patient and his or her healthcare provider, and may change over time.

SCREENING/SURGICAL CONSIDERATIONS*[*,1,2]	AGE TO START	FREQUENCY
Breast Cancer		
Clinical Breast Exam	25 years old	Every 6 months
Breast Screening • Mammogram and breast MRI	25 years old	Every 12 months
For consideration of risk-reducing mastectomy, manage based on family history	Individualized	N/A
Colorectal Cancer		
Colonoscopy	Late teens	Every 2-3 years
Gastric Cancer		
Upper endoscopy	Late teens	Every 2-3 years
Intestinal Cancer		
Small bowel visualization via CT or MRI enterography or video capsule endoscopy	8-10 years old (baseline) By age 18 at regular intervals	Every 2-3 years
Pancreatic Cancer		
Magnetic resonance cholangiopancreatography with contrast or endoscopic ultrasound	30-35 years old	Every 1-2 years
Ovarian, Cervical, and Uterine Cancer		
Pelvic exam and Pap smear	18-20 years old	Every 12 months
Consider transvaginal ultrasound	18-20 years old	Individualized
Testicular Cancer		
Testicular exam and observation for feminizing changes	10 years old	Every 12 months
Lung Cancer		
Provide education about symptoms and smoking cessation.	Individualized	N/A

** Due to the rarity of the syndrome and complexities of diagnosing and managing individuals with Peutz-Jeghers syndrome, referral to a specialized team is recommended.*

1. NCCN Clinical Practice Guidelines in Oncology®. Genetic/Familial High-Risk Assessment: Colorectal. V.3.2017. Available at nccn.org.

2. NCCN Clinical Practice Guidelines in Oncology®. Genetic/Familial High-Risk Assessment: Breast and Ovarian. V1.2018. Available at nccn.org.

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