

Clinician Management Resource for *SMAD4* (Juvenile polyposis syndrome)

This overview of clinical management guidelines is based on this patient's positive test result for a *SMAD4* gene mutation. Unless otherwise stated, medical management guidelines used here are limited to those issued by the National Comprehensive Cancer Network® (NCCN®)¹ 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}	AGE TO START	FREQUENCY
Colorectal Cancer		
Colonoscopy**	15 years old	Individualized if polyps are found Every 2-3 years if no polyps found
Gastric Cancer		
Upper endoscopy**	15 years old	Individualized if polyps are found Every 2-3 years if no polyps found
Intestinal Cancer		
No specific screening guidelines exist at this time	N/A	N/A
Pancreatic Cancer		
No specific screening guidelines exist at this time	N/A	N/A
Hereditary Hemorrhagic Teleangiectasia		
Screen for vascular lesions associated with HHT	Within first 6 months of life	Individualized

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

** Gastrectomy and/or colectomy should be considered if polyp burden or polyp-related symptoms (ie, anemia) cannot be controlled endoscopically or prevent optimal surveillance for cancer.

1. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Genetic/Familial High-Risk Assessment: Colorectal. V1.2020. © National Comprehensive Cancer Network, Inc. 2020. All rights reserved. Accessed August 20, 2020. To view the most recent and complete version of the guideline, go online to NCCN.org. NCCN makes no warranties of any kind whatsoever regarding their content, use or application and disclaims any responsibility for their application or use in any way.