

Genetic Syndromes (COLON-17)

Cancer genetics summaries focus on the genetics of specific cancers that are inherited cancer syndromes.

The goal and benefit of cancer surveillance is to identify a genetics-predisposed neoplastic process earlier in the course than standard procedure surveillance/screening recommendations in the absence of a genetic syndrome.

The genetics of specific cancers include syndrome-specific information on the risk implications of a family history of cancer, the prevalence and characteristics of cancer-predisposing variants, known modifiers of genetic risk, opportunities for genetic testing, outcomes of genetic counseling and testing, and interventions available for people with increased cancer risk resulting from an inherited predisposition.

Endoscopy and colonoscopy surveillance recommendations are based on the recommended surveillance intervals for the specific genetic defect when possible, or presence of a genetic-associated specific neoplasm in the absence of genetic assessment.

If a positive neoplastic finding is identified via surveillance, subsequent endoscopic testing is based on the shorter interval of either the genetic syndrome specific surveillance guideline or specific tumor follow-up Oncology recommendations.

As this is a rapidly changing field, clinical judgment in conjunction with recommended guidelines for specific genetic tumor syndromes are considered when supported by national guidelines.

References

PDQ® Cancer Genetics Editorial Board. PDQ Cancer Genetics Overview. Bethesda, MD: National Cancer Institute. Updated 08/23/2024. Available at: <https://www.cancer.gov/publications/pdq/information-summaries/genetics/overview-hp-pdq>. [PMID: 26389204]

Ness RM, Llor X, Abbass MA, et al. National Comprehensive Cancer Network (NCCN) Guidelines Version 1.2024 – February 27, 2024. Colorectal cancer screening, available at: https://www.nccn.org/professionals/physician_gls/pdf/colorectal_screening.pdf.

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Gupta S, Weiss JM, Axell L, et al. National Comprehensive Cancer Network (NCCN) Guidelines Version 3.2024 – October 31, 2024. Genetic/Familial high-risk assessment: colorectal, endometrial, and gastric, available at: https://www.nccn.org/professionals/physician_gls/pdf/genetics_ceg.pdf. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines™) for Genetic/Familial high-risk assessment: colorectal, endometrial, and gastric V3.2024 – October 31, 2024. ©2024 National Comprehensive Cancer Network, Inc. All rights reserved. The NCCN Guidelines™ and illustrations herein may not be reproduced in any form for any purpose without the express written permission of the NCCN. To view the most recent and complete version of the NCCN Guidelines™, go online to NCCN.org.