

GENE(S)	ASSOCIATED CANCERS**									
	Breast	Ovarian	Uterine	Colorectal & Polyposis	Pancreatic	Prostate	Renal	Gastric	Other	Recessive Disease Association
APC				●				●	●	
ATM	●	●			●	●				Ataxia telangiectasia
AXIN2				●						
BAP1							●		●	
BARD1	●									
BMPR1A, SMAD4				●				●		
BRCA1, BRCA2	●	●			●	●			●	Fanconi anemia
BRIP1		●								Fanconi anemia
CDH1	●							●		
CDKN2A					●				●	
CHEK2	●						●			
FH							●		●	
FLCN							●			
GREM1, POLD1, POLE				●						
HOXB13						●				
MBD4				●					●	
MET							●			
MLH1, MSH2, MSH6, PMS2, EPCAM		●	●	●	●	●	●	●	●	CMMRD††
MSH3				●†						Polyposis
MUTYH				●†					●	Polyposis
NF1	●								●	
NTHL1				●†						Polyposis
PALB2	●	●			●					Fanconi anemia
PTEN	●		●	●			●		●	
RAD51C, RAD51D	●	●								Fanconi anemia
RPS20				●						
STK11	●	●	●	●	●			●	●	
TP53	●			●					●	
TSC1							●		●	
TSC2							●		●	
VHL							●		●	

* Ambry Clinician Management Resource (CMR) included with test report and available at <https://www.ambrygen.com/providers/resources/clinical-materials>.

** This figure depicts primary cancer associations and may not specify all gene-disease associations. Gene-disease associations and risk estimates vary from study to study, and data are rapidly evolving.

† Biallelic/Autosomal recessive colorectal cancer risk only

†† CMMRD = constitutional mismatch repair deficiency

REFERENCES

1. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Genetic/Familial High-Risk Assessment: Breast, Ovarian, and Pancreatic. V1.2025. ©National Comprehensive Cancer Network, Inc. 2024. All rights reserved. Accessed September 13, 2024. 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.
2. Referenced with permission from the NCCN Clinical Practice Guidelines in Oncology (NCCN Guidelines®) for Genetic/Familial High-Risk Assessment: Colorectal, Endometrial, and Gastric. V2.2024 ©National Comprehensive Cancer Network, Inc. 2024. All rights reserved. Accessed October 8, 2024. 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.
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