
 78 - 90 gene hereditary cancer test

 Medical management guidelines available for most genes



*Ambry Clinician Management Resources (CMRs) are included with test reports and are available on our website.

GENE(S)	ASSOCIATED CANCERS**													Recessive Disease Associated
	Breast	Ovarian	Uterine	Colorectal & Polyposis	Pancreatic	Prostate	Gastric	Renal/Urothelial	Endocrine	Central Nervous System	Melanoma	Heme Malignancy	Other	
<i>AIP</i>										●				
<i>ALK</i>										●				
<i>APC*</i>				●			●		●	●			●	
<i>ATM*</i>	●	●			●	●								Ataxia telangiectasia
<i>AXIN2*</i>				●										
<i>BAP1*</i>								●			●		●	
<i>BARD1*</i>	●													
<i>BMPR1A*</i>				●			●							
<i>BRCA1*</i>	●	●			●	●								Fanconi anemia
<i>BRCA2*</i>	●	●			●	●					●			Fanconi anemia
<i>BRIP1*</i>		●												Fanconi anemia
<i>CDC73</i>									●				●	
<i>CDH1*</i>	●						●							
<i>CDK4</i>											●			
<i>CDKN1B*</i>									●	●				
<i>CDKN2A*</i>					●					●	●			
<i>CEBPA*</i>												●		
<i>CHEK2*</i>	●							●						
<i>CTNNA1</i>							●							
<i>DDX41*</i>												●		
<i>DICER1</i>		●								●				
<i>EGFR</i>													●	
<i>EPCAM*</i>		●	●	●	●	●	●	●		●			●	CMMRD††
<i>ETV6*</i>												●		
<i>FH*</i>								●	●				●	FH deficiency
<i>FLCN*</i>								●						
<i>GATA2*</i>												●		
<i>GREM1*</i>				●										
<i>HOXB13*</i>						●								
<i>KIT</i>													●	
<i>LZTR1</i>										●				
<i>MAX*</i>									●					
<i>MBD4*</i>				●								●		
<i>MEN1*</i>										●			●	
<i>MET*</i>								●						
<i>MITF</i>											●			
<i>MLH1*</i>		●	●	●	●	●	●	●		●			●	CMMRD††
<i>MLH3*</i>				●†										
<i>MSH2*</i>		●	●	●	●	●	●	●		●			●	CMMRD††
<i>MSH3*</i>				●†										Polyposis
<i>MSH6*</i>		●	●	●	●	●	●	●		●			●	CMMRD††
<i>MUTYH*</i>				●†									●	Polyposis

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NF1*	●								●	●			●	
NF2										●				
NTHL1*				●†										Polyposis
PALB2*	●	●			●									Fanconi anemia
PDGFRA													●	
PHOX2B										●				
PMS2*		●	●	●	●	●	●	●		●			●	CMMRD††
POLD1*				●										
POLE*				●										POLE-deficiency syndrome
POT1											●	●	●	
PRKAR1A									●	●			●	
PTCH1										●			●	
PTEN*	●		●	●				●	●	●	●			
RAD51C*	●	●												Fanconi anemia
RAD51D*	●	●												
RB1										●	●		●	
RET*									●					
RPS20*				●										
RUNX1*												●		
SDHA*								●	●				●	Mitochondrial deficiency syndrome
SDHAF2*									●					
SDHB*								●	●				●	Mitochondrial deficiency syndrome
SDHC*								●	●				●	
SDHD*								●	●				●	Mitochondrial deficiency syndrome
SMAD4*				●			●							
SMARCA4		●								●			●	
SMARCB1										●			●	
SMARCE1										●				
STK11*	●	●	●	●	●		●						●	
SUFU										●			●	
TMEM127*									●					
TP53*	●			●						●		●		
TSC1*								●		●			●	
TSC2*								●		●			●	
VHL*								●	●	●				Polycythemia
WT1								●					●	

Optional CancerNext-Expanded Add-ons:

Limited evidence genes (7): *ATRIP, EGLN1, KIF1B, PALLD, RAD51B, RNF43, TERT*

There is limited evidence to support a causal role for these genes in association with cancer predisposition.

Pancreatitis genes (5): *CFTR, CPA1, CTRC, PRSS1, SPINK1*

** 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