

THE MOST RELEVANT GENES FOR COMMON HEREDITARY CANCERS

GENE	BREAST	OVARIAN	UTERINE	COLORECTAL	MELANOMA	PANCREATIC	STOMACH	PROSTATE*
BRCA1	●	●				●		●
BRCA2	●	●			●	●		●
MLH1		●	●	●		●	●	●
MSH2		●	●	●		●	●	●
MSH6		●	●	●			●	●
PMS2*		●	●	●				●
EPCAM*		●	●	●		●	●	●
APC				●		●	●	
MUTYH*				●				
MITF*					●			
BAP1					●			
CDKN2A					●	●		
CDK4*					●			
TP53	●	●	●	●	●	●	●	●
PTEN	●		●	●	●			
STK11	●	●	●	●		●	●	
CDH1	●						●	
BMPRIA				●		●	●	
SMAD4				●		●	●	
GREM1*				●				
POLD1*				●				
POLE*				●				
PALB2	●	●				●		
CHEK2	●			●				●
ATM	●					●		●
BARD1	●							
BRIP1	●	●						
RAD51C	●	●						
RAD51D	●	●						

* CDK4: analysis is limited to chr12:g.58145429-58145431 (codon 24). EPCAM: analysis is limited to deletions that minimally encompass the 3' end of the gene including exons 8 and/or 9. GREM1: analysis is limited to duplications that overlap the upstream regulatory region. MITF: analysis is limited to chr3:g.70014091 (including c.952G>A). MUTYH: single heterozygous pathogenic variants, likely pathogenic variants, and variants of uncertain significance are not reported. PMS2: variants of uncertain significance are not reported for exons 12-15. Analysis excludes three variants commonly observed in the pseudogene PMS2CL: c.2182_2184delinsG, c.2243_2246delAGAA and deletion of exons 13-14 (chr7:g.6015768_6018727del). POLD1: analysis is limited to chr19:g.50909713 (including c.1433G>A). POLE: analysis is limited to chr12:g.133250250 (including c.1270C>G).