Hereditary cancer risk testing reference guide

Clear guidance for important decisions. Because a test result is only as good as the action it inspires.

Quest Advanced® Oncology

Test codes[b]

Gene	Comprehensive Hereditary Cancer	38611 Guideline-Based Hereditary Cancer	38621 Hereditary Breast Cancer	38631 Hereditary Colorectal Cancer	38641 Hereditary Endocrine Cancer	92587 BRCA Panel Plus	91863 BRCA Panel	91461 Lynch Syndrome	Additional tests available[a]	Associated cancers[c]: Brain/Central nervous system (CNS)	Breast	Colorectal/Gastrointestinal (GI)	Thyroid	Hereditary paraganglioma-pheochromocytoma (PGL/PCC)	Endocrine other	Gastric	Melanoma	Ovarian	Pancreatic	Prostate	Renal	Uterine / Endometrial	Other cancers	Non-cancerous findings
APC	✔	✔		✔					✔	●		●[d]	●				●			●				●	●
ATM	✔	✔	✔						✔		●	○					○		●	●	○				
AXIN2	✔	✔		✔								●[d]													●
BAP1	✔								✔							●					●			●	●
BARD1	✔	✔	✔								●														
BLM	✔								✔		○	○													
BMPR1A	✔	✔		✔					✔			●[d]							●		●				●
BRCA1	✔	✔	✔			✔	✔		✔		●							●	●	●	●		○		
BRCA2	✔	✔	✔			✔	✔		✔		●						●	●	●	●	●		○		
BRIP1	✔	✔																●							
CDH1	✔	✔	✔	✔		✔			✔		●	●													
CDK4	✔	✔															●		○						
CDKN1B	✔													●											●
CDKN2A	✔	✔							✔								●		●						
CHEK2	✔	✔	✔	✔					✔		●	○					○				●			○	
DICER1	✔												●						●					●	●
EGFR	✔																							●	
EPCAM	✔	✔	✔	✔				✔	✔	●		●					●		●	●	●	●	●	●	●
FANCA	✔																			●					
FANCC	✔										●								●						
FANCM	✔										●														
FH	✔				✔				✔				○								●			●	●
FLCN	✔								✔												●				●
GALNT12	✔											●[d]													
GREM1	✔	✔		✔								●[d]													
HOXB13	✔	✔							✔											●					
MAX[e]	✔				✔								●												
MEN1	✔				✔				✔					●											●
MET	✔																				●				
MITF	✔								✔								●				●				
MLH1	✔	✔	✔	✔				✔	✔	○		●							●	●			●	●	●
MRE11A	✔										○														
MSH2	✔	✔	✔	✔				✔	✔	●		●							●	●	●	●	●	●	●
MSH3[f]	✔	✔		✔								●[d]													
MSH6	✔	✔	✔	✔				✔	✔	○	○	●							●	●	○			●	●
MUTYH[f]	✔	✔		✔					✔			●[d]												○	●
NBN	✔																			○				○	
NF1	✔	✔	✔		✔				✔	●	●			●										●	●
NTHL1[f]	✔	✔		✔							●	●[d]											●		
PALB2	✔	✔	✔			✔			✔		●								●						
PMS2	✔	✔	✔	✔				✔	✔	○		●						○				●	●	○	●
POLD1	✔	✔		✔								●[d]											○		
POLE	✔	✔		✔								●[d]													
POT1	✔									●							●								
PTCH1	✔								✔	●														●	●
PTEN	✔	✔	✔	✔		✔			✔		●	●	●				●				●	●			●
RAD50	✔										●														
RAD51C	✔	✔	✔								●								●						
RAD51D	✔	✔	✔								○								●						
RECQL	✔										●														
RET	✔				✔				✔				●	●	●										●
SDHA	✔				✔										●							○			●
SDHAF2[e]	✔				✔										●										
SDHB	✔				✔										●							●			●
SDHC	✔				✔										●										●
SDHD[e]	✔				✔										●							○			○
SMAD4	✔	✔		✔					✔			●[d]							●			○			●
SMARCA4	✔								✔	○									●			○			
STK11	✔	✔	✔	✔		✔			✔		●	●[d]							●	●			●	●	●
SUFU	✔								✔	●														●	●
TMEM127	✔				✔									●											
TP53	✔	✔	✔	✔		✔			✔	●	●	●	●	●	●	●	●	●	●	●	●	●	●	●	
TSC1	✔								✔	●												●			●
TSC2	✔								✔	●												●			●
VHL	✔				✔				✔	●				●	●					●		●			●
XRCC2	✔										○														

● Literature supports association ○ Literature suggests possible association

[a] Call for more information. Single-site testing available for all genes under test code 93945

[b] Tests selected based on ordering patterns. See page 2 for a complete selection of tests available

[c] Gene association based on guidelines and internal data

[d] Colorectal cancer risk with polyposis phenotype

[e] Gene exhibits a parent-of-original effect

[f] Table reflects risks associated with biallelic pathogenic variants

Please contact **Quest Genomics Client Services at 1.866.GENE.INFO (1.866.436.3463) or visit QuestHereditaryCancer.com** with questions

Quest Advanced Oncology

Hereditary cancer complete genetic testing menu

Comprehensive and guideline-based hereditary cancer panels

Test code	Test name	Genes
38600	**Comprehensive Hereditary Cancer Panel (66 genes)**	*APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN1B, CDKN2A* (p16, p14), *CHEK2, DICER1, EGFR, EPCAM, FANCA, FANCC, FANCM, FH, FLCN, GALNT12, GREM1, HOXB13, MAX, MEN1, MET, MITF, MLH1, MRE11 (MRE11A), MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PMS2, POLD1, POLE, POT1, PTCH1, PTEN, RAD50, RAD51C, RAD51D, RECQL, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL, XRCC2*
38611	**Guideline-Based Hereditary Cancer Panel (32 genes)**	*APC, ATM, AXIN2, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A* (p16,p14), *CHEK2, EPCAM, GREM1, HOXB13, MLH1, MSH2, MSH3, MSH6, MUTYH, NF1, NTHL1, PALB2, PMS2, POLD1, POLE, PTEN, RAD51C, RAD51D, SMAD4, STK11, TP53*

Additional hereditary cancer risk tests

Test code	Test name	Genes
38621	**Hereditary Breast Cancer Panel (18 genes)**	*ATM, BARD1, BRCA1, BRCA2, CDH1, CHEK2, EPCAM, MLH1, MSH2, MSH6, NF1, PALB2, PMS2, PTEN, RAD51C, RAD51D, STK11, TP53*
38631	**Hereditary Colorectal Cancer Panel (20 genes)**	*APC, AXIN2, BMPR1A, CDH1, CHEK2, EPCAM, GREM1, MLH1, MSH2, MSH3, MSH6, MUTYH, NTHL1, PMS2, POLD1, POLE, PTEN, SMAD4, STK11, TP53*
38641	**Hereditary Endocrine Cancer Panel (12 genes)**	*FH, MAX, MEN1, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL*
91461	**Lynch Syndrome Panel (5 genes)**	*EPCAM, MLH1, MSH2, MSH6, PMS2*
38651	**Nevoid Basal Cell Carcinoma (NBCCS) (Gorlin) Syndrome Panel (*PTCH1*, *SUFU*)**	*PTCH1, SUFU*
38661	**Tuberous Sclerosis Complex Panel (*TSC1*, *TSC2*)**	*TSC1, TSC2*
94053	**Juvenile Polyposis Panel (*BMPR1A* and *SMAD4*)**	*BMPR1A, SMAD4*
93945	**Hereditary Cancer Single Site(s)**	*APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN1B, CDKN2A* (p16, p14), *CHEK2, DICER1, EGFR, EPCAM, FANCA, FANCC, FANCM, FH, FLCN, GALNT12, GREM1, HOXB13, MAX, MEN1, MET, MITF, MLH1, MRE11 (MRE11A), MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NTHL1, PALB2, PMS2, POLD1, POLE, POT1, PTCH1, PTEN, RAD50, RAD51C, RAD51D, RECQL, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, STK11, SUFU, TMEM127, TP53, TSC1, TSC2, VHL, XRCC2*
See genes column	**Single-gene tests**	*APC* – 93797, *ATM* – 38802, *BAP1* – 38803, *BLM* – 38804, *CDH1* – 92568, *CDKN2A* – 93939, *CHEK2* – 93940, *EPCAM/MSH2* – 91471, *FH* – 38805, *FLCN* – 38806, *HOXB13* – 38807, *MEN1* – 93942, *MITF* – 38808, *MLH1* – 91460, *MSH6* – 91458, *MUTYH* – 93944, *NF1* – 93941, *PALB2* – 92571, *PMS2* – 91457, *PTEN* – 92566, *RET* – 93796, *SMARCA4* – 38809, *STK11* – 92565, *TP53* – 92560, *VHL* – 93943

Components of panels may be ordered separately.

Please contact **Quest Genomics Client Services at 1.866.GENE.INFO (1.866.436.3463) or visit QuestHereditaryCancer.com** with questions

Test codes may vary by location. Please contact your local laboratory for more information.

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