

# Hereditary cancer risk testing reference guide

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

Gene	Test codes <sup>b</sup>								Associated cancers <sup>c</sup>																	Other cancers	Non-cancerous findings
	38611	38621	38631	38641	92587	91863	91461		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						
APC	✓	✓	✓	✓				✓	●		● <sup>d</sup>	●			●		●						●	●			
ATM	✓	✓	✓					✓		●	○				○		●		○					●			
AXIN2	✓	✓	✓					✓			● <sup>d</sup>													●			
BAP1	✓	✓	✓					✓							●						●		●	●			
BARD1	✓	✓	✓					✓		●														●			
BLM	✓	✓	✓					✓		○	○													●			
BMPR1A	✓	✓	✓					✓			● <sup>d</sup>				●									●			
BRCA1	✓	✓	✓		✓			✓	●							●		●		●		○					
BRCA2	✓	✓	✓		✓			✓	●						●		●		●		○						
BRIP1	✓	✓	✓					✓																			
CDH1	✓	✓	✓		✓			✓	●						●												
CDK4	✓	✓	✓					✓							●												
CDKN1B	✓	✓	✓					✓							●									●			
CDKN2A	✓	✓	✓					✓							●									●			
CHEK2	✓	✓	✓		✓			✓	●		○				○				●				○				
DICER1	✓	✓	✓					✓															○	●			
EGFR	✓	✓	✓					✓															○	●			
EPCAM	✓	✓	✓					✓	●		●				●				●		●		●	●			
FANCA	✓	✓	✓					✓																●			
FANCC	✓	✓	✓					✓	●															●			
FANCM	✓	✓	✓					✓	●															●			
FH	✓	✓	✓		✓			✓							○								○	●			
FLCN	✓	✓	✓					✓															○	●			
GALNT12	✓	✓	✓					✓			● <sup>d</sup>													●			
GREM1	✓	✓	✓		✓			✓			● <sup>d</sup>													●			
HOXB13	✓	✓	✓					✓																●			
MAX <sup>e</sup>	✓	✓	✓		✓			✓							●									●			
MEN1	✓	✓	✓		✓			✓							●									●			
MET	✓	✓	✓					✓																●			
MITF	✓	✓	✓					✓																●			
MLH1	✓	✓	✓		✓			✓	○		●				●								○	●			
MRE11A	✓	✓	✓					✓			○													●			
MSH2	✓	✓	✓		✓			✓	●		●				●								○	●			
MSH3 <sup>f</sup>	✓	✓	✓		✓			✓			● <sup>d</sup>													●			
MSH6	✓	✓	✓		✓			✓	○		●				●								○	●			
MUTYH <sup>f</sup>	✓	✓	✓		✓			✓			● <sup>d</sup>												○	●			
NBN	✓	✓	✓					✓															○	●			
NF1	✓	✓	✓		✓			✓	●						●									●			
NTHL1 <sup>f</sup>	✓	✓	✓		✓			✓	●		● <sup>d</sup>													●			
PALB2	✓	✓	✓		✓			✓	●															●			
PMS2	✓	✓	✓		✓			✓	○		●				○								○	●			
POLD1	✓	✓	✓		✓			✓			● <sup>d</sup>												○	●			
POLE	✓	✓	✓		✓			✓			● <sup>d</sup>													●			
POT1	✓	✓	✓					✓	●															●			
PTCH1	✓	✓	✓					✓	●															●			
PTEN	✓	✓	✓		✓			✓	●		●													●			
RAD50	✓	✓	✓					✓	●															●			
RAD51C	✓	✓	✓		✓			✓	●															●			
RAD51D	✓	✓	✓		✓			✓	○															●			
RECQL	✓	✓	✓					✓	●															●			
RET	✓	✓	✓		✓			✓							●									●			
SDHA	✓	✓	✓		✓			✓							●									●			
SDHAF2 <sup>e</sup>	✓	✓	✓		✓			✓							●									●			
SDHB	✓	✓	✓		✓			✓							●									●			
SDHC	✓	✓	✓		✓			✓							●									●			
SDHD <sup>e</sup>	✓	✓	✓		✓			✓							●								○	●			
SMAD4	✓	✓	✓		✓			✓			● <sup>d</sup>				●								○	●			
SMARCA4	✓	✓	✓					✓	○														○	●			
STK11	✓	✓	✓		✓			✓	●		● <sup>d</sup>				●									●			
SUFU	✓	✓	✓					✓	●						●									●			
TMEM127	✓	✓	✓		✓			✓							●									●			
TP53	✓	✓	✓		✓			✓	●		●				●									●			
TSC1	✓	✓	✓					✓	●															●			
TSC2	✓	✓	✓					✓	●															●			
VHL	✓	✓	✓		✓			✓	●						●									●			
XRCC2	✓	✓	✓					✓	○															●			

● Literature supports association ○ Literature suggests possible association

<sup>a</sup> Call for more information. Single-site testing available for all genes under test code 93945

<sup>b</sup> Tests selected based on ordering patterns. See page 2 for a complete selection of tests available

<sup>c</sup> Gene association based on guidelines and internal data

<sup>d</sup> Colorectal cancer risk with polyposis phenotype

<sup>e</sup> Gene exhibits a parent-of-origin effect

<sup>f</sup> Table reflects risks associated with biallelic pathogenic variants

## 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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