

# VistaSeq Hereditary Cancer Panels

Gene	Transcript Reference	Sequencing Analysis	Deletion/Duplication Analysis	Special Notes
<i>ALK</i>	NM_004304	✓	✓	
<i>APC</i>	NM_000038	✓	✓	Analysis includes 1A & 1B promoter deletions
<i>ATM</i>	NM_000051	✓	✓	
<i>AXIN2</i>	NM_004655	✓	✓	
<i>BARD1</i>	NM_000465	✓	✓	
<i>BLM</i>	NM_000057	✓	✓	
<i>BMPR1A</i>	NM_004329	✓	✓	
<i>BRCA1</i>	NM_007294	✓	✓	
<i>BRCA2</i>	NM_000059	✓	✓	
<i>BRIP1</i>	NM_032043	✓	✓	
<i>CDC73</i>	NM_024529	✓	✓	
<i>CDH1</i>	NM_004360	✓	✓	
<i>CDKN2A</i>	NM_000077	✓	✓	
<i>CDK4</i>	NM_000075	✓	✓	
<i>CHEK2</i>	NM_007194	✓	✓	
<i>EPCAM</i>	NM_002354		✓	Analysis is limited to deletion/duplication only
<i>FAM175A</i>	NM_139076	✓	✓	
<i>FANCC</i>	NM_000136	✓	✓	
<i>FH</i>	NM_000143	✓	✓	
<i>FLCN</i>	NM_144997	✓	✓	
<i>GPC3</i>	NM_004484	✓	✓	
<i>HOXB13</i>	NM_006361	✓		Analysis is limited to sequencing only
<i>MAX</i>	NM_002382	✓	✓	
<i>MEN1</i>	NM_130799	✓	✓	
<i>MET</i>	NM_001127500	✓	✓	
<i>MITF</i>	NM_000248			Analysis is limited to the c.952G>A variant only
<i>MLH1</i>	NM_000249	✓	✓	
<i>MRE11A</i>	NM_005591	✓	✓	
<i>MSH2</i>	NM_000251	✓	✓	
<i>MSH6</i>	NM_000179	✓	✓	
<i>MUTYH</i>	NM_001128425	✓	✓	
<i>NBN</i>	NM_002485	✓	✓	
<i>NF1</i>	NM_000267	✓	✓	
<i>NF2</i>	NM_000268	✓	✓	

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<i>PALB2</i>	NM_024675	✓	✓	
<i>PHOX2B</i>	NM_003924	✓	✓	
<i>PMS2</i>	NM_000535	✓	✓	
<i>POLD1</i>	NM_002691	✓		Analysis is limited to sequencing only
<i>POLE</i>	NM_006231	✓		Analysis is limited to sequencing only
<i>PRKAR1A</i>	NM_002734	✓	✓	
<i>PTCH1</i>	NM_000264	✓	✓	
<i>PTEN</i>	NM_000314	✓	✓	Analysis includes promoter variant c.-1300_-750
<i>RAD50</i>	NM_005732	✓	✓	
<i>RAD51C</i>	NM_058216	✓	✓	
<i>RAD51D</i>	NM_002878	✓	✓	
<i>RB1</i>	NM_000321	✓	✓	
<i>RET</i>	NM_020975	✓	✓	
<i>SDHB</i>	NM_003000	✓	✓	
<i>SDHC</i>	NM_003001	✓	✓	
<i>SDHD</i>	NM_003002	✓	✓	
<i>SMAD4</i>	NM_005359	✓	✓	
<i>SMARCB1</i>	NM_003073	✓	✓	
<i>STK11</i>	NM_000455	✓	✓	
<i>SUFU</i>	NM_016169	✓	✓	
<i>TMEM127</i>	NM_017849	✓	✓	
<i>TP53</i>	NM_000546	✓	✓	
<i>TSC1</i>	NM_000368	✓	✓	
<i>TSC2</i>	NM_000548	✓	✓	
<i>VHL</i>	NM_000551	✓	✓	
<i>WT1</i>	NM_024426	✓	✓	

- Candidate genes and testing methodology are selected from published literature and market review to target genes and/or regions of genes that are associated with clinical utility and clear evidence of pathogenicity. This remains current through regular review.
- Next generation sequencing is used to examine the entire gene coding regions, as well as flanking non-coding regions, of genes known to be involved in the development, progression, and susceptibility of cancer. Flanking regions for the *BRCA1* and *BRCA2* genes include +/- 20bp and +/-10bp for all other genes.
- Copy number variations are assessed by microarray or multiple-ligation-probe amplification assay (MLPA) to detect gross deletions and duplications.
- Special considerations for individual genes on the VistaSeq panels are listed in the table above.