ASSAY DETAILS

VistaSeq Hereditary Cancer Panels

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

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

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