Comprehensive Hereditary Cancer Panel

Targeted Genes, Next-Generation Sequencing (NGS)

This Next-Generation Sequencing test is performed to evaluate for the presence of variants in coding regions and extending to +/- 20 base pairs of adjacent intronic sequence on either side of the coding exons of the 154 genes analyzed. In addition, the analysis will cover select noncoding variants. Confirmation of select reportable variants may be performed by alternate methodologies (Multiplex ligation-dependent probe amplification (MLPA), long-range polymerase chain reaction, etc.) based on internal laboratory criteria.

This is the current from April 2026 to the present. This document is intended to highlight the regions of interest in the genes tested, additional evaluations, and the technical limitations. However, this document does not comprehensively reflect all genomic regions covered by this test.

For additional questions, contact the laboratory at +971 2 493 0400.

Genome Build GRCh37 (hg19) – In Alphabetical Order

Gene	Transcript ID	Exons Covered	Additional Evaluations	Technical Limitations
ACD	NM_001082486.2	Exons 1-12		
AIP	NM_003977.4	Exons 1-6		
ALK	NM_004304.5	Exons 1-29		
ANAPC1	NM_022662.4	Exons 2-48		
ANKRD26	NM_014915.3	Exons 1-34	5’ UTR up to c.-134	
APC	NM_000038.6, NM_001127511.3	Exons 2-16	Promoter region (c.-195 to c.-125) in NM_001127511.3 and deep intronic mutation c.933+829A>G	
ATM	NM_000051.4	Exons 2-63	Deep intronic mutations c.497-2661A>G, c.1236-404C>T, c.2839-579_2839-576del, c.2639-384A>G, c.3994-159A>G, and c.5763-1050A>G	
AXIN2	NM_004655.4	Exons 2-11		
BAP1	NM_004656.4	Exons 1-17		
BARD1	NM_000465.4	Exons 1-11		
BLM	NM_000057.4	Exons 2-22	Deep intronic mutation c.3020-258A>G	
BMPR1A	NM_004329.3	Exons 3-13		
BRCA1	NM_007294.4	Exons 2-23	Promoter mutation c.-107A>T and deep intronic/splice region mutations c.442-22_442-13del, c.4185+4105C>T, c.4358-31_4358-27del, c.4358-33T>G, c.5333-36_5333-22del, c.5407-25T>A and c.5468-40T>A	
BRCA2	NM_000059.4	Exons 2-27		
BRIP1	NM_032043.3	Exons 2-20	Deep intronic mutation c.1629-498A>T	
BTK	NM_000061.3	Exons 2-19		
BUB1B	NM_001211.6	Exons 1-23		
BUB3	NM_004725.4	Exons 2-8		
CBL	NM_005188.4	Exons 1-16		

Gene	Transcript ID	Exons Covered	Additional Evaluations	Technical Limitations
CDC73	NM_024529.5	Exons 1-17		
CDH1	NM_004360.5	Exons 1-16		
CDK4	NM_000075.4	Exons 2-8		
CDKN1B	NM_004064.5	Exons 1-2		
CDKN1C	NM_001122630.2	Exons 2-3		Exon 2 with low coverage (bases can have minimum coverage <10X)
CDKN2A	NM_000077.5	Exons 1-3	Promoter mutation c.-34G>T and deep intronic mutation c.458-105A>G	
CEBPA	NM_004364.5	Exon 1		Exon 1 with low coverage (bases can have minimum coverage <10X)
CEP57	NM_014679.5	Exons 1-11		
CHEK2	NM_007194.4	Exons 2-15		
CTNNA1	NM_001903.5	Exons 2-18		
CYLD	NM_001378743.1	Exons 3-19		
DDB2	NM_000107.3	Exons 1-10		
DDX41	NM_016222.4	Exons 1-17		
DGCR8	NM_022720.7	Exons 2-14		
DICER1	NM_177438.3	Exons 2-27	Deep intronic mutation c.5364+1187T>G	
DIS3L2	NM_152383.5	Exons 2-21		
DKC1	NM_001363.5	Exons 1-15		
EGFR	NM_005228.5	Exons 1-28		
ELP1	NM_003640.5	Exons 2-37		
EPCAM	NM_002354.3	Exons 1-9		
ERCC2	NM_000400.4	Exons 1-23		
ERCC3	NM_000122.2	Exons 1-15		
ERCC4	NM_005236.3	Exons 1-11		
ERCC5	NM_000123.4	Exons 1-15		
ETV6	NM_001987.5	Exons 1-8		
EXT1	NM_000127.3	Exons 1-11		
EXT2	NM_207122.2	Exons 2-14		
FAH	NM_000137.4	Exons 1-14		
FANCA	NM_000135.4	Exons 1-43		
FANCC	NM_000136.3	Exons 2-15		
FANCD2	NM_001018115.3	Exons 2-44		
FANCE	NM_021922.3	Exons 1-10		
FANCF	NM_022725.4	exon 1		
FANCG	NM_004629.2	Exons 1-14		
FANCM	NM_020937.4	Exons 1-23		
FGFR1	NM_023110.3	Exons 2-18		
FH	NM_000143.4	Exons 1-10		

Gene	Transcript ID	Exons Covered	Additional Evaluations	Technical Limitations
FLCN	NM_144997.7	Exons 4-14		CNV in noncoding exons 1-3 not tested
FOXR2	NM_198451.4	exon 1		
GALNT12	NM_024642.5	Exons 1-10		Exon 1 with low coverage (bases can have minimum coverage <10X)
GATA2	NM_032638.5	Exons 2-6		
GNAQ	NM_002072.5	Exons 1-7		
GPC3	NM_004484.4	Exons 1-8		
GPR161	NM_001375883.1	Exons 2-6		
GREM1	NM_013372.7	exon 2		CNV duplication of upstream enhancer region not analysed
HOXB13	NM_006361.6	Exons 1-2		
HRAS	NM_005343.4	Exons 2-5		
IDH1	NM_005896.4	Exons 3-10		
IDH2	NM_002168.4	Exons 1-11		
IGF2	NM_000612.6	Exons 2-4		
IKZF1	NM_006060.6	Exons 2-8		
KIT	NM_000222.3	Exons 1-21		
KRAS	NM_004985.5	Exons 2-5		
LZTR1	NM_006767.4	Exons 1-21		
MAFA	NM_201589.4	Exon 1		
MAX	NM_002382.5	Exons 1-5		
MBD4	NM_001276270.2	Exons 1-8		
MC1R	NM_002386.4	Exon 1		
MEN1	NM_000244.4	Exons 2-10	Splice region mutation c.-23-22C>A & 3' UTR mutation c.*412G>A	
MET	NM_000245.4	Exons 2-21		
MITF	NM_001354604.2	Exons 1-10		
MLH1	NM_000249.4	Exons 1-19	Promoter mutation c.-27C>A and deep intronic mutation c.1732-264A>T	
MLH3	NM_001040108.2	Exons 2-13		
MSH2	NM_000251.3	Exons 1-16	Promoter mutation c.-78_-77del and deep intronic mutation c.2635-24A>G	10 Mb inversion of exons 1-7 not tested
MSH3	NM_002439.5	Exons 1-24		
MSH6	NM_000179.3	Exons 1-10		
MUTYH	NM_001048174.2	Exons 2-16	Splice region mutation c.420+19_420+31del	
MYT1	NM_004535.3	Exons 3-23		
NBN	NM_002485.5	Exons 1-16		
NF1	NM_000267.3	Exons 1-58	Promoter mutations c.-272G>A and c.-273A>C; deep intronic mutations c.61-7486G>T,	

Gene	Transcript ID	Exons Covered	Additional Evaluations	Technical Limitations
			c.888+651T>A, c.888+744A>G, c.888+789A>G, c.1260+1604A>G, c.1527+1159C>T, c.1642-449A>G, c.2490-1398A>G, c.2490-1398A>G, c.3198-314G>A, c.3974+260T>G, c.4110+945A>G, c.5749+267A>G, c.5749+332A>G, c.5750-279A>G, c.5750-177A>C, c.7908-321C>G, and c.8050+86A>G; splice region mutations c.889-21C>A, c.1261-21T>G, c.1722-26T>C, c.2252-33_2252-8del, c.4515-21T>C/G, c.5206-38A>G, c.6579+26C>G, c.6579+31T>G, c.7807-24_7807-19delinsTTTTAG, and c.8050+25A>T	
NF2	NM_000268.4	Exons 1-16		
NTHL1	NM_002528.7	Exons 1-6		
PALB2	NM_024675.4	Exons 1-13		
PAX5	NM_016734.3	Exons 1-10		
PDE11A	NM_016953.4	Exons 1-20		
PDE8B	NM_003719.5	Exons 1-22		
PDGFRA	NM_006206.6	Exons 2-23		
PHOX2B	NM_003924.4	Exons 1-3		Exon 3 with low coverage (bases can have minimum coverage <10X)
PIK3CA	NM_006218.4	Exons 2-21		
PMS2	NM_000535.7	Exons 1-15		
POLD1	NM_002691.4	Exons 2-27		
POLE	NM_006231.4	Exons 1-49	Splice region mutation c.1686+32C>G	
POLH	NM_006502.3	Exons 2-11		
POT1	NM_015450.3	Exons 5-19		
PRKAR1A	NM_002734.5	Exons 2-11		
PTCH1	NM_000264.5	Exons 1-23		
PTEN	NM_000314.8	Exons 1-9	Promoter region c.-589 to c.-1247 (c.-1239A>G, c.-1178C>T, c.-1111A>G, c.-1001T>C)	
PTPN11	NM_002834.5	Exons 1-15		
RABL3	NM_173825.5	Exons 1-8		
RAD50	NM_005732.4	Exons 1-25		
RAD51B	NM_133510.4	Exons 2-11		

Gene	Transcript ID	Exons Covered	Additional Evaluations	Technical Limitations
RAD51C	NM_058216.3	Exons 1-9		
RAD51D	NM_002878.4	Exons 1-10		
RB1	NM_000321.3	Exons 1-27	Deep intronic/splice region mutations c.2490-1398A>G, c.2490-28T>G/C, c.2490-26A>C/G/T & c.1215+63T>G	
RECQL	NM_002907.4	Exons 2-15		
RECQL4	NM_004260.4	Exons 1-22		Exons 14-16 with low coverage (bases can have minimum coverage <10X)
REST	NM_005612.5	Exons 2-4		
RET	NM_020975.6	Exons 1-20		
RHBDF2	NM_001005498.4	Exons 3-19		
RNF43	NM_017763.6	Exons 2-10		
RPS20	NM_001023.4	Exons 1-4		
RUNX1	NM_001754.5	Exons 2-9		
SAMD9	NM_017654.4	exon 3		
SAMD9L	NM_152703.5	exon 5		
SBDS	NM_016038.4	Exons 1-5		
SDHA	NM_004168.4	Exons 1-15		
SDHAF2	NM_017841.4	Exons 1-4		
SDHB	NM_003000.3	Exons 1-8	Splice region mutation c.73-22T>A	
SDHC	NM_003001.5	Exons 1-6	Deep intronic mutation c.20+74A>G	
SDHD	NM_003002.4	Exons 1-4		
SF3B2	NM_006842.3	Exons 1-22		
SMAD4	NM_005359.6	Exons 2-12		
SMARCA4	NM_003072.5	Exons 2-35		
SMARCB1	NM_003073.5	Exons 1-9		
SMARCE1	NM_003079.5	Exons 2-11		
SMO	NM_005631.5	Exons 1-12		
SOS1	NM_005633.4	Exons 1-23		
STK11	NM_000455.5	Exons 1-9		
SUFU	NM_016169.4	Exons 1-12		
TBXT	NM_001366285.2	Exons 1-8		
TERC	NR_001566.3	Exon 1		
TERF2IP	NM_018975.4	Exons 1-3		
TERT	NM_198253.3	Exons 1-16		
TMEM127	NM_017849.4	Exons 2-4		
TP53	NM_000546.6	Exons 2-11	Splice region mutation c.673-39G>A	
TRIM28	NM_005762.3	Exons 1-17		Exon 1 with low coverage (bases can have minimum coverage <10X)

Gene	Transcript ID	Exons Covered	Additional Evaluations	Technical Limitations
TRIP13	NM_004237.4	Exons 1-13		
TSC1	NM_000368.5	Exons 3-23	Deep intronic mutations c.363+668G>A & c.2625+367A>G	
TSC2	NM_000548.5	Exons 2-42	Deep intronic mutations c.600-145C>T, c.848+281C>T, c.2838-122G>A	
VHL	NM_000551.4	Exons 1-3		
WRN	NM_000553.6	Exons 2-35		
WT1	NM_024426.6	Exons 1-10		
XPA	NM_000380.4	Exons 1-6		
XPC	NM_004628.5	Exons 1-16		
XRCC2	NM_005431.2	Exons 1-3		

Unless otherwise indicated, coverage of the intronic regions flanking the exons is ±20 bp. For all genes, MANE Select transcripts are used.

Effective date	Version	Update
April, 2026	V1	NA