

PreSENTIA HEREDITARY CANCER

GENES TESTED

The following table indicates the genes tested per panel.

TYPE OF PANEL	SNVs/INDELS, CNAs
BREAST & GYNECOLOGICAL (26 Genes)	ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, DICER1, EPCAM [†] , MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, POLD1, POLE, PTEN, RAD50, RAD51C, RAD51D, SMARCA4, STK11, TP53
BREAST/ GYNECOLOGICAL GUIDELINES-BASED (19 Genes)	ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM [†] , MLH1, MSH2, MSH6, NBN, PALB2, PMS2, PTEN, RAD51C, RAD51D, TP53, STK11
BREAST HIGH RISK (7 Genes)	BRCA1, BRCA2, CDH1, PALB2, PTEN, STK11, TP53
BRCA1 / BRCA2 (2 Genes)	BRCA1, BRCA2
COLORECTAL (17 Genes)	APC, BMPR1A, CDH1, CHEK2, EPCAM [†] , GREM1 [‡] , MLH1, MSH2, MSH6, MUTYH, PMS2, POLD1, POLE, PTEN, SMAD4, STK11, TP53
COLORECTAL HIGH-RISK (10 Genes)	APC, BMPR1A, EPCAM [†] , MLH1, MSH2, MSH6, MUTYH, PMS2, SMAD4, STK11
COLORECTAL NON-POLYPOSIS (5 Genes)	EPCAM [†] , MLH1, MSH2, MSH6, PMS2
COLORECTAL POLYPOSIS SYNDROME (7 Genes)	APC, BMPR1A, MUTYH, POLD1, POLE, SMAD4, STK11
MYELODYSPLASTIC SYNDROME LEUKEMIA (24 Genes)	ATM, BRCA1, BRCA2, BRIP1, EPCAM [†] , ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, MLH1, MSH2, MSH6, PALB2, PMS2, RAD51C, SLX4, TP53
GASTRIC (14 Genes)	APC, BMPR1A, CDH1, EPCAM [†] , MLH1, MSH2, MSH6, PMS2, SDHB, SDHC, SDHD, SMAD4, STK11, TP53
PROSTATE (15 Genes)	ATM, BRCA1, BRCA2, CHEK2, EPCAM [†] , HOXB13 [‡] , MLH1, MSH2, MSH6, NBN, PALB2, PMS2, PTEN, RAD51D, TP53
PANCREATIC (17 Genes)	APC, ATM, BRCA1, BRCA2, BMPR1A, CDK4, CDKN2A, (CDKN2A ^{p16(INK4A)} , CDKN2A ^{p14(ARF)}), EPCAM [†] , MEN1, MLH1, MSH2, MSH6, PALB2, PMS2, SMAD4, STK11, TP53
RENAL (13 Genes)	BAP1, EPCAM [†] , MLH1, MSH2, MSH6, PMS2, PTEN, SDHAF2, SDHB, SDHC, SDHD, TP53, VHL
SKIN (XP-ASSOCIATED) (9 Genes)	DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA, XPC
FAMILIAL MELANOMA (7 Genes)	BAP1, BRCA2, CDK4, CDKN2A (CDKN2A ^{p16(INK4A)} , CDKN2A ^{p14(ARF)}), PTEN, RB1, TP53
PARAGANGLIOMA/ PHEOCHROMOCYTOMA (6 Genes)	RET, SDHAF2, SDHB, SDHC, SDHD, VHL
PARATHYROID (1 Gene)	MEN1
THYROID (1 Gene)	RET
PAN-CANCER (62 Genes)	APC, ATM, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, (CDKN2A ^{p16(INK4A)} , CDKN2A ^{p14(ARF)}), CHEK2, DDB2, DICER1, EPCAM [†] , ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, GREM1 [‡] , HOXB13 [‡] , MEN1, MLH1, MRE11, MSH2, MSH6, MUTYH, NBN, PALB2, PMS2, POLD1, POLE, POLH, PTEN, RAD50, RAD51C, RAD51D, RB1, RET, SDHAF2, SDHB, SDHC, SDHD, SLX4, SMAD4, SMARCA4, STK11, TP53, VHL, XPA, XPC

[†] SNVs/INDELS are not covered [‡] CNAs are not covered

SNVs: Single Nucleotide Variants

INDELS: Insertions and Deletions

CNAs: Copy Number Alterations