

Hereditary Cancer Panel (154 cancer-related genes)

Genes:

ABRAXAS1, AIP, AKT1, ALK, ANKRD26, APC, ATM, ATR, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, BUB1B, CASR, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CEBPA, CEP57, CHEK2, CTC1, CTNNA1, CTRC, CYLD, DDB2, DDX41, DICER1, DIS3L2, DKC1, EGFR, EGLN1, ELANE, ENG, EPCAM, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, EXO1, EXT1, EXT2, EZH2, FAN1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, FLCN, GALNT12, GATA2, GEN1, GPC3, GREM1, HOXB13, HRAS, IKZF1, KIF1B, KIT, LZTR1, MAX, MBD4, MEN1, MET, MITF, MLH1, MLH3, MRE11, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NHP2, NOP10, NTHL1, PALB2, PALLD, PAX5, PDGFRA, PHOX2B, PIK3CA, PMS2, POLD1, POLE, POLH, POT1, PRKAR1A, PRSS1, PTCH1, PTCH2, PTEN, RAD50, RAD51C, RAD51D, RB1, RECQL, RECQL4, REST, RET, RHBDF2, RNF43, RPS20, RUNX1, SAMD9L, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLC45A2, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, SPINK1, SRP72, STK11, SUFU, TERC, TERT, TGFBR1, TINF2, TMEM127, TP53, TRIP13, TSC1, TSC2, TYR, VHL, WRAP53, WRN, WT1, XPA, XPC, XRCC2 (154 genes)

Gene Specifics:

Gene	Notes
GALNT12	Due to high GC content, copy-number variants located in exon 1 of the GALNT12 gene (NM_024642.4) cannot be reliably detected and will not be reported.
MSH2	Inversion of MSH2 exons 1-7 ("Boland" inversion) is assessed for Lynch Syndrome, Colorectal, Endometrial, and all other hereditary cancer panel testing.
PIK3CA	Because the vast majority of PIK3CA pathogenic variants arise postzygotic and are thus mosaic, more than one tissue may need to be tested. Failure to detect a PIK3CA pathogenic variant does not exclude a clinical diagnosis of the PIK3CA-associated segmental overgrowth disorders in individuals with suggestive features (PubMed: 23946963).