

PORTFOLIO DE ESTUDIOS GENÉTICOS PARA DIAGNÓSTICO DE CÁNCER HEREDITARIO.

Incluye desde una variante familiar, hasta paneles específicos diseñados en base a las guías clínicas nacionales e internacionales.

TECNOLOGÍA

NGS (Next Generation Sequencing) NextSeq550 Dx y NovaSeq 6000, de Illumina.

PANELES

MAMA	ATM,BARD1, BRCA1, BRCA2, CDH1, CHEK2, NBN, NF1, PALB2, PTEN, RAD51C, RAD51D, STK11, TP53
GINECO MAMA	ATM,BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, DICER1, EPCAM, MLH1, MSH2, MSH6, PMS2, MUTYH, NBN, NF1, PALB2, POLE, POLD1, PTEN, RAD51C, RAD51D, SAMD4, SMARCA4, STK11, TP53
COLON	APC, ATM, BLM, BMPR1A, CHEK2, GALNT12, GREM1, MLH1, MSH2, MSH6, PMS2, EPCAM, MUTYH, NBN, NTHL1, POLE, POLD1, PTEN, SMAD4, STK11, TP53
PRÓSTATA	ATM, BRCA1, BRCA2, BRIP1, CHEK2, EPCAM, HOXB13, MLH1, MSH2, MSH6, PMS2, PALB2, RAD51C, RAD51D, STK11, TP53
MELANOMA	BAP1, BRCA1, BRCA2, CDK4, CDKN2A, LZTR1, MC1R, MITF, NF1, NF2, POT1, PTCH1, PTEN, RB1, SMARCB1, SUFU TER, TP53
30 GENES	APC, ATM, BAP1, BARD1, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDKN2A, CHEK2, EPCAM, GREM1, HOXB13, MLH1, MSH2, MSH6, MUTYH, NBN, NTHL1, PALB2, PMS2, POLE, POLD1, PTEN, RAD41C, RAD51D, SMAD4, STK11, TP53
50 GENES	APC, ATM, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CDH1, CDK4, CDKN2A, CHEK2, DICER1, EPCAM, FANCA, FANCC, FANCD2, FH, FLCN, GALNT12, GREM1, HOXB13, KIT, MEN1, MET, MITF, MLH1, MSH2, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PMS2, POLD1, POLD1, POLE, POT1, PTEN, RAD51C, RAD51D, RET, SMAD4, STK11, TP53, VHL, WT1
100 GENES	AIP, APC, ATM, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CASR, CDC73, CDH1, CDK4, CDKN1B, CDKN2A, CEBPA, CHEK2, DICER1, DIS3L2, EPCAM, FAM175A/ABRAXAS1, FANCA, FANCB, FANCD2, FANCC, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FH, FLCN, GALNT12, GATA2, GPC3, GREM1, HOXB13, KIF1B, KIT, LZTR1, MAX, MEN1, MET, MITF, MLH1, MRE11A, MSH2, MSH3, MSH6, MUTYH, NBN, NF1, NF2, NTHL1, PALB2, PDGFRA, PHOX2B, PIK3CA, PMS2, POLD1, POLE, POT1, PRKAR1A, PTCH1, PTEN, RAD50, RAD51B, RAD51C, RAD51D, RB1, RECQL4, RET, RHBDF2, RINT1, RUNX1, SDHA, SDHAF2, SDHB, SDHC, SDHD, SLX4, SMAD4, SMARCA4, SMARCB1, SMARCE1, SPINK1, SPRED1, STK11, SUFU, TERF2IP, TERT, TMEM127, TP53, TSC1, TSC2, VHL, WT1, XRCC2

