

G-ESSENCE

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

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