

**Gen canónico:** gen estándar debido a una función bien definida, conservación evolutiva y una sólida evidencia experimental que confirma su importancia en enfermedades.

**Gen candidato:** gen que, según se cree, puede estar relacionado con un rasgo determinado, como una enfermedad o un atributo físico. Estos genes pueden aportar información relevante sobre distintos tipos de cánceres.

### Panel Oncología 1

Este panel analiza los genes canónicos y candidatos exclusivamente de la **región codificante**.

#### Genes canónicos (región codificante)

A1CF, ABI1, ABL1, ABL2, ACKR3, ACVR1, ACVR1B, ACVR2A, AFDN, AFF3, AKT1, AKT2, AKT3, ALK, AMER1, ANK1, APOBEC3B, AR, ARAF, ARHGAP26, ARHGAP5, ARHGEF10, ARHGEF10L, ARHGEF12, ARID1A, ARID1B, ARID2, ARNT, ASXL1, ATF1, ATP1A1, ATP2B3, ATR, ATRX, AXIN2, BAP1, BARD1, BAX, BAZ1A, BCL10, BCL11A, BCL11B, BCL2L12, BCL3, BCL9, BCL9L, BCOR, BCORL1, BIRC6, BLM, BMP5, BMPR1A, BRAF, BRCA1, BRCA2, BRD3, BRD4, BRIP1, BTG1, BTK, BUB1B, CACNA1D, CALR, CARD11, CARS1, CASP3, CASP8, CASP9, CBFA2T3, CBL, CBLB, CBLC, CCDC6, CCNB1IP1, CCNC, CCND1, CCND2, CCNE1, CCR4, CCR7, CD209, CD274, CD28, CD74, CD79A, CDC73, CDH1, CDH11, CDH17, CDK12, CDK4, CDK6, CDKN1A, CDKN1B, CDKN2A, CDKN2C, CDX2, CEBPA, CHD2, CHD4, CHEK2, CHST11, CIC, CLTC, CLTCL1, CNBD1, CNBP, COL2A1, CPEB3, CREB1, CREB3L1, CREB3L2, CRLF2, CRNKL1, CRTC1, CSF1R, CSF3R, CTCF, CTNNA2, CTNND1, CTNND2, CUL3, CUX1, CYLD, CYP2C8, CYSLTR2, DAXX, DCAF12L2, DCC, DDB2, DDR2, DDX10, DDX5, DDX6, DEK, DGCR8, DICER1, DNMT2, DNMT3A, DROSHA, EBF1, ECT2L, EED, EGFR, EIF1AX, EIF3E, ELF3, ELF4, ELK4, ELL, EP300, EPAS1, EPHA3, EPHA7, EPS15, ERBB2, ERBB3, ERBB4, ERCC2, ERCC3, ERCC4, ERCC5, ERG, ESR1, ETNK1, ETV1, ETV4, ETV5, EWSR1, EXT1, EXT2, EZH2, FAM135B, FAM47C, FANCA, FANCC, FANCD2, FANCE, FANCF, FANCG, FAS, FAT1, FAT3, FAT4, FBLN2, FBXW7, FCGR2B, FCRL4, FEN1, FES, FEV, FGFR1, FGFR2, FGFR3, FGFR4, FH, FHIT, FKBP9, FLCN, FLNA, FLT3, FLT4, FOXL2, FOXO3, FOXO4, FOXP1, FOXR1, FSTL3, FUBP1, FUS, GATA1, GATA2, GLI1, GNA11, GNAQ, GNAS, GPC3, GPC5, GRIN2A, GRM3, H3-3A, H3-3B, H3C2, HEY1, HIF1A, HIP1, HLF, HMGA1, HMGA2, HNF1A, HNRNPA2B1, HOXA11, HOXA13, HOXA9, HOXC11, HOXC13, HOXD11, HOXD13, HRAS, IDH1, IDH2, IGF2BP2, IKBKB, IL6ST, IL7R, IRF4, IRS4, ISX, ITGAV, JAK1, JAK2, JAK3, JUN, KAT6A, KAT6B, KAT7, KCNJ5, KDM5C, KDR, KEAP1, KIT, KLF4, KLF6, KMT2A, KMT2C, KNL1, KNSTRN, KRAS, LARP4B, LATS1, LATS2, LCK, LEF1, LMO1, LMO2, LRIG3, LRP1B, LYL1, LZTR1, MACC1, MAF, MAFB, MALT1, MAML2, MAP2K2, MAP3K1, MAP3K13, MAPK1, MAX, MDM2, MDM4, MECOM, MED12, MET, MGMT, MITF, MLF1, MLH1, MLLT10, MN1, MPL, MRTFA, MSH2, MSH6, MSI2, MTCP1, MTOR, MUTYH, MYB, MYCL, MYCN, MYD88, MYH9, MYOD1, N4BP2, NAB2, NBEA, NBN, NCOA2, NCOA4, NCOR2, NDRG1, NF2, NFATC2, NFE2L2, NFKBIE, NKX2-1, NOTCH1, NOTCH2, NPM1, NR4A3, NRAS, NRG1, NSD2, NSD3, NT5C2, NTHL1, NTRK1, NTRK3, NUP98, NUTM1, OLIG2, P2RY8, PABPC1, PALB2, PATZ1, PAX3, PBX1, PCBP1, PDCD1LG2, PDGFB, PDGFRA, PDGFRB, PER1, PHF6, PHOX2B, PIK3CA, PIK3CB, PIK3R2, PLAG1, PLCG1, PML, PMS1, PMS2, POLD1, POLE, POLG, POLQ, POT1, POU2AF1, POU5F1, PPARG, PPM1D, PPP2R1A, PPP6C, PRDM1, PRDM16, PRDM2, PREX2, PRF1, PRKACA, PRKAR1A, PRKCB, PRPF40B, PSIP1, PTCH1, PTK6, PTPN11, PTPN13, PTPN6, PTPRB, PTPRC, PTPRD, PTPRK, PTPRT, QKI, RAC1, RAD17, RAD21, RAD51B, RAF1, RANBP2, RAP1GDS1, RARA, RBM10, RECQL4, REL, RET, RFWD3, RGPD3, RGS7, RIT1, RMI2, ROBO2, ROS1, RPL10, RPL22, RPL5, RSPO2, RSPO3, RUNX1, RUNX1T1, SALL4, SBDS, SDHA, SDHAF2, SDHB, SDHC, SET, SETBP1, SETD1B, SETD2, SETDB1, SF3B1, SFRP4,

SH2B3, SH3GL1, SIRPA, SIX1, SIX2, SKI, SLC34A2, SMAD2, SMAD3, SMARCB1, SMARCD1, SMARCE1, SMC1A, SMO, SND1, SOCS1, SOX2, SOX21, SOX9, SPEN, SPOP, SRC, SRSF2, SRSF3, SSX1, SSX2, SSX4, STAG1, STAG2, STAT3, STAT5B, STAT6, STIL, STK11, SUFU, SUZ12, SYK, TAF15, TAL1, TAL2, TBL1XR1, TBX3, TCF7L2, TEC, TENT5C, TET1, TET2, TFE3, TFEB, TGFBR2, TLX1, TLX3, TMEM127, TNC, TNFAIP3, TNFRSF14, TNFRSF17, TPM3, TRAF7, TRIM24, TRIM27, TRIM33, TRRAP, TSC1, TSC2, TSHR, U2AF1, UBR5, USP44, USP6, USP8, VAV1, WAS, WIF1, WNK2, WRN, WT1, WWTR1, XPA, XPC, XPO1, YWHAE, ZBTB16, ZEB1, ZFH3X, ZMYM3, ZNF331, ZNF429, ZNF479, ZNF521, ZNRF3, ZRSR2

### Genes candidatos (región codificante)

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**Panel Oncología 2**

Este panel analiza los genes canónicos y candidatos tanto en la **región codificante** como en la **no codificante**.

**Genes región codificante y no codificante****I. Genes región codificante (mismos que en el 'Panel 1')****II. Genes en la región no codificante****a. Genes canónicos****Mutaciones germinales con asociación a cáncer**

APC, ASXL2, ATM, AXIN1, BCL2, BCL6, BCLAF1, BIRC3, CAMTA1, CBFB, CCNB1IP, CDH10, CIITA, CNOT3, CXCR4, DDIT3, DDX3X, ETV6, FUBP1, GATA3, KDM5A, KDM6A, LEPROTL1, MAP2K1, MAP2K4, MEN1, MYC, NCOR1, NF1, NFKB2, PAX5, PBRM1, PIK3R1, PIM1, PTEN, RB1, RHOH, RNF43, S100A7, SDHD, SFPQ, SGK1, STCL1A, TERT, TP53, TP63, VHL

**Vejiga**

LEPROTL1, TERT.

**Linfoma de Burkitt**

ARID1A, BCL6, CCND3, CD79B, CREBBP, CXCR4, DDX3X, DNMT1, FBXO11, FOXO1, ID3, KMT2D, MYC, PIK3R1, PTEN, RHOA, SMARCA4, TCF3, TCL1A, TP53

**Linfoma Folicular y DLBCL**

B2M, BCL11A, BCL2, BCL6, BIRC3, CXCR4, FLI1, IKZF1, LPP, PAX5, RHOH, SGK1

**Cáncer de Mama**

CTNNB1, FOXA1, LEPROTL1

**Melanoma y melanoma mucoso.**

TERT

**Colangiocarcinoma, Carcinoma hepatocelular.**

AFF4, BCL6, TERT

**Cáncer de células de Hürthle**

TERT

**b. Genes candidatos****Mutaciones germinales con asociación a cáncer**

ACSM4, ADAMTS17, ADCY2, ADD2, ADGRG6, ADRB3, AHSA1, AKR1D1, ALB, ALOX12B, ALYREF, ANG, ANKRD16, ANKRD30BL, ANKRD50, ANXA2, ANXA8L1, AP2A1, AP3D1, API5, APMAP, AQP7, ARHGEF18, ARHGEF3, ASPSCR1, ASTN2, ASZ1, ATAD3B, ATG4B, ATP10D, ATP5MPL, ATP5PD, B3GALT6, BACH2, BAG4, BAGE2, BCL7A, BORA, BRDT, BUD23, C10orf90, C11orf49, C16orf91, C19orf53, C1orf159, C1orf185, C1orf194, C1QTNF1, C6orf62, C7, C8orf44-SGK3, CA10, CACNG6, CADM2, CALCB, CALCOCO1, CAST, CATIP, CBX6, CCDC107, CCDC124, CCDC66, CCDC77, CCK, CCL26, CD1A, CD2BP2, CD86, CDC20, CDC23, CDC37, CDC37L1, CDH8, CEP83, CGB2, CGB5, CHAF1B, CHD6, CHDH, CHKB, CHRM5, CILK1, CIPC, CLDN20, CLIC5, CLUAP1, CNDP2, COL21A1, COL4A2, COQ10B, COX7C, CPNE5, CR1, CRBN, CRELD2, CRP, DCAF15, DDX58, DEFB129, DEPDC4, DERL1, DGKB, DGKI, DHRS1, DHX16, EIF3G, ELOVL3, EN1, EPB41, EPB41L3, EPS8L1, ERGIC3, EVI2A, EXOC7, FAHD2B, FAM221B, FAM234A, FBF1, FBXO9, FCGR3A, FDX1, FDXR, FEM1A, FGB, FN3KRP, FOXP2, FRG1, FRG2B, FTH1, FUBP3, FXYD3, FSHR, FSTL5, G2E3, GABRA1, GABRA2, GALNTL5, GALNTL6, GATM, GDF5, GFRA1, GFRAL, GIMAP4, GLYAT, GNA15, GNAI1, GNG7, GOLGA2, GRAMD1B, GSK3A, GSTA4, GSTM5, H2AC17, H2AC6, H2BC5, HAPLN1, HDAC1, HDAC9, HES1, HLA-DQA1, HLA-DRB1, HLA-DRB5, HLA-F, HM13, HNRNPC, HOXC6, HSBP1, ICAM3, IFI44L, IFITM2, IFITM3, IGLL5, IQGAP1, IRAG2, ISCA2, KATNB1, KCNH5, KCNK1, KCNN2, KCNQ3, KCTD2, KDELR1, KDM1B, KLHL10, KLHL11, KLHL17, LAPTM4A, LDHAL6A, LEPR, LGSN, LILRB2, LIMD2, LMNTD2, LPAR3, LRRC45, LRRC75B, LRRN3, LRTM1, LRWD1, LY6K, LYPD8, MACROD2, MAGEC1, MALSU1, MAN2A1, MAP1S, MAP3K19, MAPRE3, MBD6, MBOAT2, MC1R, MCM5, MED16, MED31, MEF2C, METTL26, MFSD10, MKRN1, MOCS3, MORN1, MROH1, MROH2A, MRPL20, MRPL36, MRPS14, MRPS31, MRPS33, MTG2, MTRF1, MUC12, MUC3A, MUC4, MUSK, MYO5B, MYT1L, NANP, NBPFF1, NBPFF8, NDEL1, NDUFAF8, NDUFB9, NEUROG2, NFKBIZ, NME8, NOC2L, NOP53, NOS1, NOX4, NPAS3, NR1D2, NR1I2, NR5A1, NUMB, OBI1, ODF4, ONECUT2, OR10H2, OR1L1, OR1M1, OR2F2, OR4C5, OR4K17, OR51F2, OR7G3, OR8K5, OR9G1, OR9K2, OS9, OSBPL8, OXNAD1, PAAF1, PAPOLB, PARP2, PAX6, PCDH10, PCDH15, PCDH9, PCDHGA8, PCF11, PCMTD1, PDCD11, PDCD2, PDP2, PDZRN4, PELI3, PES1, PEX16, PHAX, PHF21A, PHYHD1, PIGC, PLCB2, PLEKHS1, POLR2D, POLR3E, PPP1R9B, PPP2R3A, PPP4C, PRAMEF4, PRKAG1, PRKCZ, PRKG1, PRMT5, PROZ, PRRC2B, PRRT3, PRSS3, PSMC3, PSMG4, PTDSS1, PTX3, QPRT, RAB11A, RAB14, RABEPK, RALY, RANBP10, RAVR1, RBFOX1, RCC1, REG1A, RESP18, RFLNB, RGCC, RGS18, RINL, RNASE11, RNASE4, RNF168, RNF185, RNF216, RNF26, RNF4, RNF40, RPE, RPIA, RPL13A, RPL18A, RPL34, RPS27, RPS27A, RRAS2, RS1, S100A7A, S100A8, SCYL2, SDCCAG8, SDF4, SEC14L1, SEC16A, SEMA3A, SEMA6A, SERPINB7, SESN2, SGCZ, SHF, SI, SIK3, SIRPD, SIRPG, SLAMF1, SLC12A5, SLC22A11, SLC22A2, SLC24A5, SLC25A17, SLC25A45, SLC30A6, SLC35G1, SLC35G2, SLC44A5, SLC66A1L, SLC6A9, SMUG1, SNRPD2, SNRPN, SNX16, SPAM1, SPANXN1, SPATA19, SPATA5L1, SPATC1L, SPI1, SPN, SQOR, SRSF9, SSBP1, ST6GAL1, STAG3, STIM2, SULT2B1, SUPT7L, SYF2, TAF11, TBC1D12, TBC1D22A, TBC1D29P, TBL2, TCAF1, TCF21, TCF4, TDRD5, TEDC2, TIGIT, TIPIN, TM4SF18, TMBIM6, TMC3, TMEM116, TMEM121B, TMEM128, TMEM160, TMEM179, TMEM208, TMEM210, TMEM63A, TMPRSS4, TMSB4X, TOB1, TOR1B, TOX3, TPRG1L, TPSAB1, TPTE, TRAF3IP3, TRAI, TRAPPC2L, TRMT10C, TTC4, TUBA1C,

## PANELES ONCOLOGÍA

TYW1B, UBAC1, UBE2J2, UBXN7, UMPS, URB1, VAMP4, VCPIP1, VIPAS39, VIT, VPS51, VPS53, VSX2, WDR74, WLS, WNT5B, WRAP73, XCL2, XRCC5, YAE1, ZC3H18, ZC4H2, ZCCHC2, ZFP14, ZFP36L1, ZFPM1, ZKSCAN3, ZNF165, ZNF180, ZNF253, ZNF324, ZNF503, ZNF558, ZNF578, ZNF677, ZNF711, ZNF717, ZNF728, ZNF814, ZSCAN31, ZSCAN4, ZSWIM6

### **Cáncer de mama**

ALDOA, CITED2, IRF2BP2, PIGY, PLEKHS1, TBC1D12, WDR74, ZNF143

### **Vejiga**

ADGRG7, KCTD7, NDUFS7, PLEKHS1, PLXDC1, RABGEF1, TBC1D12, TSPAN18, WDR74

### **Linfoma de Burkitt**

ADGRG7, BACH2, BCL7A, BMP7, BTG2, CD70, CHD8, DTX1, ETS1, FZD3, GNA13, GNAI2, H1-4, H2AC11, H2AC16, H2AC17, H2BC12, H4C11, HNRNPU, IGLL5, IKZF3, IRF8, KCNK10, MCL1, MME, PABPC4L, PCDHA10, RFX7, RNF144B, SIN3A, SNTB2, TFAP4, TMSB4X, USP7, WDR7, ZFP36L1

### **Carcinoma (cáncer) papilar tiroideo**

FANK1, RHPN2

### **Colangiocarcinoma, Carcinoma hepatocelular.**

C22orf35, GRIA2, LUC7L, MED16, MYOZ3, PRSS38, R3HDM4, SLC22A1, STUB1, TNFRSF8, TRPC6, VPS45, WDR74

### **Melanoma**

BLCAP, CANX, CCDC9, CCN6, CCP110, CYP4A11, CYS1, ERGIC3, FIZ1, HRH4, IMPG2, KBTBD8, MED31, NSUN6, NUDT10, NUP50, PNPLA2, PSMD8, PSMF1, PTPA, RALY, RNF185, RPL29, RPS27, TBC1D13, TEDC2, TEX22, TGS1, UAP1, UBAC2, ZNF778, ZWINT

### **Adenocarcinoma de esófago**

ABCG3, FTO, H2AC17, H2BC17, MMP24, MTERF3, MTG2, OPALIN, PTDSS1, RPGRIP1L, SDCCAG8, WDR74

### **Linfoma Folicular y DLBCL**

ACTB, ADIPOQ, ARID5B, ATG3, BLK, BOD1, BTG2, BTLA, CCDC102B, CD200, CNTRL, COL28A1, CPEB4, CSMD3, DAAM1, DCK, DDX54, DISP1, DMXL1, DNAJB9, DTWD2, EAF2, EIF3G, EIF4A2, ERI1, ETS1, FAM167A, FAM177B, FDX1, FIGNL1, FLII, FRMD8, GATA4, GRHPR, GRSF1, GSDMA, H2BC11, H2BC3, HS2ST1, IGLL5, IKZF3, IMM2L, INPP4B, IQCB1, IRF2BP2, IRF8, KRCC1, LRRC32, METTL7A, MFHAS1, MIOS, MOB1B, MRPL12, MS4A6E, NABP1, NAP1L1, NUBP1, PCDHB1, PLSCR2, PLXND1, PNPLA8, PNRC1, POLDIP3, POLM, RFC4, RFTN1, RPIA, RTTN, SCIN, SEC61G, SH3GLB1, SLC25A45,

SLC35A5, SLC9C1, SLITRK6, SPIB, ST6GAL1, THAP5, TIGD3, TMX3, TNFAIP8, TPRG1, TRAF1, ZBTB5, ZBPB2

## Anexo I- Bibliografía

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