

**NGS alapú vizsgálat paraffinos blokkból:
„Oncomine Comprehensive Plus” panel, 517 gén**

517 gén vizsgálata, mikroszatellita instabilitás (MSI), tumor mutációs terhelés (TMB) és célzott terápiához releváns mutációk, amplifikációk, fúziók kimutatására új generációs szekvenálással (NGS).

A panel géntartalmának listája:

- Kódoló szekvencia (CDS) egyedi génekben:

CALR, CIITA, CYP2D6, ERCC5, FAS, ID3, KLHL13, MTUS2, PSMB10, PSMB8, PSMB9, RNASEH2C, RPL22, RPL5, RUNX1T1, SDHC, SOCS1, STAT1, TMEM132D, UGT1A1, ZBTB20

- Elsődleges „hotspot” gének:

ACVR1, ATP1A1, BCR, BMP5, BTK, CACNA1D, CD79B, CSF1R, CTNNA1, CUL1, CYSLTR2, DGCR8, DROSHA, E2F1, EPAS1, FGF7, FOXL2, FOXO1, GLI1, GNA11, GNAQ, HIF1A, HIST1H2BD, HIST1H3B, HRAS, IDH1, IL6ST, IRF4, IRS4, KLF4, KNSTRN, MAP2K2, MED12, MYOD1, NSD2, NT5C2, NTRK2, NUP93, PAX5, PIK3CD, PIK3CG, PTPRD, RGS7, RHOA, RPL10, SIX1, SIX2, SNCAIP, SOS1, SOX2, SRSF2, STAT5B, TAF1, TGFBR1, TRRAP, TSHR, WAS

- Elsődleges „hotspot” gének és kópiaszám variációk (CNV) :

ABL1, ABL2, AKT1, AKT2, AKT3, ALK, AR, ARAF, AURKA, AURKC, AXL, BCL2, BCL2L12, BCL6, BRAF, CARD11, CBL, CCND1, CCND2, CCND3, CCNE1, CDK4, CDK6, CHD4, DDR2, EGFR, EIF1AX, ERBB2, ERBB3, ERBB4, ESR1, EZH2, FAM135B, FGFR1, FGFR2, FGFR3, FGFR4, FLT3, FLT4, FOXA1, GATA2, GNAS, H3F3A, H3F3B, IDH2, IKBKB, IL7R, KDR, KIT, KLF5, KRAS, MAGOH, MAP2K1, MAPK1, MAX, MDM4, MECOM, MEF2B, MET, MITF, MPL, MTOR, MYC, MYCN, MYD88, NFE2L2, NRAS, NTRK1, NTRK3, PCBP1, PDGFRA, PDGFRB, PIK3C2B, PIK3CA, PIK3CB, PIK3R2, PIM1, PLCG1, PPP2R1A, PPP6C, PRKACA, PTPN11, PXDNL, RAC1, RAF1, RARA, RET, RHEB, RICTOR, RIT1, ROS1, SETBP1, SF3B1, SLCO1B3, SMC1A, SMO, SPOP, SRC, STAT3, STAT6, TERT, TOP1, TPMT, U2AF1, USP8, XPO1, ZNF217, ZNF429

- CNV nyerő gének:

ABCB1, CTNND2, DDR1, EMSY, FGF19, FGF23, FGF3, FGF4, FGF9, FYN, GLI3,

IGF1R, MCL1,
MDM2, MYCL, RPS6KB1, RPTOR, YAP1, YES1

- CNV vesztő gének és kódoló szekvencia (CDS):

ABRAXAS1, ACVR1B, ACVR2A, ADAMTS12, ADAMTS2, AMER1, APC,
ARHGAP35, ARID1A,
ARID1B, ARID2, ARID5B, ASXL1, ASXL2, ATM, ATR, ATRX, AXIN1, AXIN2, B2M,
BAP1, BARD1,
BCOR, BLM, BMPR2, BRCA1, BRCA2, BRIP1, CASP8, CBF3, CD274, CD276,
CDC73, CDH1,
CDH10, CDK12, CDKN1A, CDKN1B, CDKN2A, CDKN2B, CDKN2C, CHEK1,
CHEK2, CIC,
CREBBP, CSMD3, CTCF, CTLA4, CUL3, CUL4A, CUL4B, CYLD, CYP2C9, DAXX,
DDX3X,
DICER1, DNMT3A, DOCK3, DPYD, DSC1, DSC3, ELF3, ENO1, EP300, EPCAM,
EPHA2, ERAP1,
ERAP2, ERCC2, ERCC4, ERFF1, ETV6, FANCA, FANCC, FANCD2, FANCE,
FANCF, FANCG,
FANCI, FANCL, FANCM, FAT1, FBXW7, FUBP1, GATA3, GNA13, GPS2, HDAC2,
HDAC9, HLA-A,
HLA-B, HNF1A, INPP4B, JAK1, JAK2, JAK3, KDM5C, KDM6A, KEAP1, KMT2A,
KMT2B, KMT2C,
KMT2D, LARP4B, LATS1, LATS2, MAP2K4, MAP2K7, MAP3K1, MAP3K4, MAPK8,
MEN1, MGA,
MLH1, MLH3, MRE11, MSH2, MSH3, MSH6, MTAP, MUTYH, NBN, NCOR1, NF1,
NF2,
NOTCH1, NOTCH2, NOTCH3, NOTCH4, PALB2, PARP1, PARP2, PARP3, PARP4,
PBRM1,
PDCD1, PDCD1LG2, PDIA3, PGD, PHF6, PIK3R1, PMS1, PMS2, POLD1, POLE,
POT1, PPM1D,
PPP2R2A, PRDM1, PRDM9, PRKAR1A, PTCH1, PTEN, PTPRT, RAD50, RAD51,
RAD51B,
RAD51C, RAD51D, RAD52, RAD54L, RASA1, RASA2, RB1, RBM10, RECQL4,
RNASEH2A,
RNASEH2B, RNF43, RPA1, RUNX1, SDHA, SDHB, SDHD, SETD2, SLX4, SMAD2,
SMAD4,
SMARCA4, SMARCB1, SOX9, SPEN, STAG2, STK11, SUFU, TAP1, TAP2, TBX3,
TCF7L2, TET2,
TGFB2, TNFAIP3, TNFRSF14, TP53, TP63, TPP2, TSC1, TSC2, USP9X, VHL, WT1,
XRCC2,
XRCC3, ZFH3, ZMYM3, ZRSR2

- Gén fúziók:

AKT2, ALK, AR, AXL, BRAF, BRCA1, BRCA2, CDKN2A, EGFR, ERBB2, ERBB4,
ERG, ESR1, ETV1,
ETV4, ETV5, FGFR1, FGFR2, FGFR3, FGR, FLT3, JAK2, KRAS, MDM4, MET, MYB,
MYBL1, NF1,
NOTCH1, NOTCH4, NRG1, NTRK1, NTRK2, NTRK3, NUTM1, PDGFRA, PDGFRB,
PIK3CA,
PPARG, PRKACA, PRKACB, PTEN, RAD51B, RAF1, RB1, RELA, RET, ROS1,

RSPO2, RSPO3,
TERT

- Tumor mutációs terhelés (TMB) meghatározásához vizsgált gének:

A1CF, ACSM2B, ADAM18, ANO4, ARMC4, BRINP3, C6, C8A, C8B, CANX, CASR, CD163,
CNTN6, CNTNAP4, CNTNAP5, COL11A1, DCAF4L2, DCDC1, GALNT17, GPR158, GRID2,
HCN1, HLA-C, KCND2, KCNH7, KEL, KIR3DL1, KRTAP2-1, KRTAP6-2, LRRC7, MARCO,
NLRC5, NOL4, NRXN1, NYAP2, OR10G8, OR2G6, OR2L13, OR2L2, OR2L8, OR2M3, OR2T3,
OR2T33, OR2T4, OR2W3, OR4A15, OR4C15, OR4C6, OR4M1, OR4M2, OR5D18, OR5F1,
OR5L1, OR5L2, OR6F1, OR8H2, OR8I2, OR8U1, ORC4, PAK5, PCDH17, PDE1A, PDE1C,
PLXDC2, POM121L12, PPFIA2, RBP3, REG1A, REG1B, REG3A, REG3G, RPTN, RUND3B,
SH3RF2, SLC15A2, SLC8A1, SYT10, SYT16, TAPBP, TPTE, TRHDE, TRIM48, TRIM51, ZIM3,
ZNF479, ZNF536