

Cytopeniák és veleszületett anemiák (Cytopenias and congenital anaemias) génpanel v:2025-05-22

Panel tartalma 176 gén:

ABCB7, ABCG5, ABCG8, ACD, ACKR1, ACTN1, ADA2, ADAMTS13, AK1, AK2, ALAS2, ALDOA, AMN, ANK1, ANKRD26, AP3B1, ATRX, BLM, BRCA1, BRCA2, BRIP1, CBL, CD59, CDAN1, CDIN1, CEBPA, CLPB, COX4I2, CTC1, CUBN, CXCR2, CXCR4, CYCS, CSF3R, DDX41, DHFR, DKC1, DNAJC21, DUT, EFL1, ELANE, EPB41, EPB42, ERCC4, ERCC6L2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FLI1, FLT3, FYB1, G6PC3, G6PD, GATA1, GATA2, GCLC, GFII, GLRX5, GP1BA, GP1BB, GP9, GPI, GSS, HAX1, HBA1, HBA2, HBB, HBD, HK1, HOXA11, IKZF1, JAGN1, KIF23, KLF1, LAT, LPIN2, MAD2L2, MASTL, MECOM, MPL, MTR, MTRR, MYH9, MYSM1, NBEAL2, NBN, NHP2, NOP10, NPM1, NT5C3A, PALB2, PARN, PFKM, PIEZO1, PKLR, PUS1, RAC2, RAD51, RAD51C, RAP1B, RHAG, RMRP, RPA1, RPL11, RPL15, RPL18, RPL27, RPL31, RPL35A, RPL5, RPL9, RPS10, RPS17, RPS19, RPS20, RPS24, RPS26, RPS28, RPS29, RPS7, RRAS, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SEC23B, SEC61A1, SLC11A2, SLC19A2, SLC25A38, SLC2A1, SLC4A1, SLFN14, SLX4, SPTA1, SPTB, SRC, SRP19, SRP54, SRP72, SRPRA, STIM1, STN1, TCIRG1, TCN2, TERC, TERT, TF, THPO, TINF2, TMPRSS6, TOP3A, TPI1, TSR2, TUBA8, TUBB1, UBE2T, UROS, USB1, VPS45, WAS, WIPF1, WRAP53, WWTR1, WWTR1, XK, XRCC2, YARS2

Forrás:

Cytopenias and congenital anaemias (Version 1.118)

<https://panelapp.genomicsengland.co.uk/panels/159/>

Confirmed Fanconi anaemia or Bloom syndrome (Version 2.6)

<https://panelapp.genomicsengland.co.uk/panels/508/>

Cytopenia - NOT Fanconi anaemia (Version 4.1)

<https://panelapp.genomicsengland.co.uk/panels/519/>

(Megtekintve: 2025-05-22)