

Seznam genů zahrnutých ve virtuálním panelu – Holoprosencefalie, mikrocefalie a kraniosynostózy (FocusedExome)

Virtuální panel zahrnuje analýzu 445 genů:

ABCC9, ADAMTSL4, ADARB1, AGMO, ALPL, ALX1, ALX3, ALX4, AP4B1, AP4E1, AP4M1, AP4S1, ARSB, ASPM, ASXL1, ATP1A2, ATR, ATRX, AXIN2, B3GAT3, BBS9, BLM, BMP4, BRAF, BRCA2, BRIP1, BUB1B, CASK, CCBE1, CD96, CDC6, CDK5RAP2, CDON, CDT1, CENPJ, CEP120, CEP135, CEP152, CEP57, CEP63, COASY, COLEC11, CREBBP, CRTAP, CTCF, CTNNB1, CTSK, CYP26B1, DHCR7, DHRS3, DIAPH1, DISP1, DLL1, DNA2, DNMT3A, DPP6, DYRK1A, EDNRB, EFNA4, EFNB1, EFTUD2, EOMES, ERCC4, ERCC5, ERCC6, ERCC8, ERF, ESCO2, FAM20C, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FBN1, FGF3, FGF8, FGF9, FGFR1, FGFR2, FGFR3, FLNA, FLNB, FREM1, GLI2, GLI3, GNAS, GNPTAB, GPC3, HDAC8, HUWE1, CHST3, IDS, IDUA, IER3IP1, IFT122, IFT140, IFT43, IGF1, IGF1R, IHH, IL11RA, IRS4, IRX5, IYD, JAG1, KANSL1, KAT6B, KCNAB2, KCNJ1, KCNJ10, KCNJ11, KCNJ5, KCNQ1OT1, KDM6A, KIF11, KISS1R, KLB, KLLN, KMT2D, KRAS, LARP7, LARS2, LEP, LEPR, LHCGR, LHX3, LHX4, LIFR, LIG4, LIMK1, LIPE, LMNA, LMNB1, LMNB2, LMX1B, LRBA, LRP4, LRP5, LZTFL1, MAGEL2, MAMLD1, MAP2K1, MAP2K2, MAP3K1, MASP1, MAX, MC2R, MC4R, MCM4, MCPH1, MEN1, MFSD2A, MINPP1, MKKS, MKS1, MLH1, MNX1, MRAP, MS4A1, MSH2, MSH4, MSH5, MSH6, MSMO1, MSX2, MYCN, MYT1L, NAA10, NBN, NCAPD2, NDE1, NEBL, NEUROD1, NEUROG3, NF1, NFIA, NFIX, NFKB1, NFKB2, NHEJ1, NIN, NIPBL, NKX2-1, NKX2-5, NKX2-6, NNT, NOBOX, NODAL, NOG, NPHP1, NPHS1, NPPA, NR0B1, NR3C1, NR3C2, NR5A1, NRAS, NSD1, NSMF, NTRK2, OBSL1, OCRL, ORC1, ORC4, ORC6, OSTM1, OTX2, P4HB, PALB2, PAX3, PAX6, PAX8, PBX1, PCBD1, PCNT, PCSK1, PCYT1A, PDE11A, PDE3A, PDE4D, PDHA1, PDX1, PGRMC1, PHEX, PHF6, PHIP, PIK3CA, PIK3R1, PITX2, PLIN1, PMM2, PMS2, PNKP, PNPLA6, POC1A, POF1B, POLD1, POLE, POLG, POLR3H, POMC, POR, POU1F1, POU5F1, PPARG, PQBP1, PRDM16, PRKAR1A, PRKCD, PRL, PROK2, PROKR2, PROP1, PRRT2, PRRX1, PRSS8, PSMC3IP, PTEN, PTF1A, PTH, PTHLH, PTCH1, PTPN11, PTPRD, PTRH2, RAB23, RAD21, RAD50, RAD51C, RAF1, RAG1, RAG2, RAI1, RBBP8, RECQL4, REN, RET, RFC2, RFX6, RIT1, RMI1, RMRP, RNF168, RNU4ATAC, ROR2, RPL10, RPS6KA3, RSPO1, RTTN, RUNX2, SALL1, SAMD9, SCARF2, SCN4A, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SDHA, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SECISBP2, SEMA3E, SERPINA6, SETBP1, SH2B1, SH3PXD2B, SHH, SHOC2, SIM1, SIX1, SIX3, SKI, SLC12A1, SLC12A3, SLC16A1, SLC16A2, SLC19A2, SLC20A2, SLC25A19, SLC26A3, SLC26A4, SLC29A3, SLC2A2, SLC3A1, SLC40A1, SLC52A2, SLC5A5, SLC7A9, SLC9A6, SLX4, SMAD6, SMC1A, SMC3, SMO, SOHLH1, SOS1, SOX10, SOX2, SOX3, SOX6, SOX9, SPECC1L, SPRY4, SRCAP, SRD5A2, SRY, SST, STAMBp, STAR, STAT1, STAT3, STAT5B, STIL, STX16, SUFU, SULT2A1, SYP, TAC3, TACR3, TBCE, TBL1X, TBX1, TBX19, TBX2, TBX3, TCF12, TCOF1, TDGF1, TF, TFAP2B, TFR2, TG, TGFB1, TGFB2, TGIF1, THRA, THRB, TMCO1, TMEM127, TNFRSF13B, TNFRSF13C, TNFSF12, TOE1, TP53, TP63, TPO, TRAF3IP2, TRAPPc9, TRH, TRHR, TRIM32, TRIM37, TRMT1, TRPM6, TSC1, TSC2, TSHB, TSHR, TSPYL1, TTC8, TTR, TUBB1, TUBGCP4, TUBGCP6, TWIST1, TWIST2, UBE3A, UBR1, UCP2, VHL, VIPAS39, VPS13B, VPS33B, WDFY3, WDPCP, WDR11, WDR19, WDR35, WDR4, WDR62, WFS1, WNK1, WNK4, WRN, WT1, XRCC4, ZEB2, ZFP57, ZFPM2, ZIC1, ZIC2, ZMPSTE24, ZNF335, ZNF526