



Seznam genů zahrnutých ve virtuálním panelu – Závažná dětská onemocnění (FocusedExome)

Virtuální panel zahrnuje analýzu 2161 genů:

AAAS, AARS1, AARS2, AASS, ABAT, ABCA1, ABCA12, ABCA3, ABCA4, ABCB11, ABCB4, ABCB7, ABCC2, ABCC6, ABCC8, ABCC9, ABCD1, ABCD4, ABCG5, ABCG8, ABHD12, ABHD5, ABL1, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAN, ACAT1, ACE, ACO2, ACOX1, ACP5, ACSF3, ACSL4, ACTA1, ACTA2, ACTB, ACTC1, ACTG1, ACTN4, ACVR1, ACVR2B, ACVRL1, ACY1, ADA, ADAM17, ADAMTS10, ADAMTS13, ADAMTS17, ADAMTSL2, ADAR, ADCY5, ADK, ADNP, ADSL, AFF2, AFG3L2, AGA, AGK, AGL, AGPS, AGRN, AGT, AGTR1, AGXT, AHI1, AICDA, AIFM1, AIMP1, AIPL1, AIRE, AK1, AK2, AKR1D1, AKT1, AKT3, ALAD, ALAS2, ALB, ALDH18A1, ALDH1A3, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG3, ALG6, ALG8, ALG9, ALMS1, ALOX12B, ALOXE3, ALPL, ALS2, ALX1, ALX3, ALX4, AMACR, AMELX, AMER1, AMN, AMT, ANK1, ANKH, ANKRD11, ANKRD26, ANKS6, ANO10, ANO3, ANO5, ANO6, ANOS1, ANTXR1, ANTXR2, AP1S1, AP1S2, AP3B1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APC, APOA1, APOA5, APOB, APOC2, APOE, APOL1, APRT, APTX, AR, ARFGF2, ARG1, ARHGAP31, ARHGDI, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARL6, ARSA, ARSB, ARSL, ARX, ASAH1, ASCC1, ASH1L, ASL, ASNS, ASPA, ASPM, ASS1, ASXL1, ASXL3, ATCAY, ATIC, ATL1, ATM, ATN1, ATP13A2, ATP1A2, ATP1A3, ATP2A1, ATP6AP2, ATP6V0A2, ATP6V1B1, ATP7A, ATP7B, ATP8A2, ATP8B1, ATPAF2, ATR, ATRX, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, AUH, AUTS2, AVPR2, B2M, B3GALNT2, B3GALT6, B3GAT3, B4GALNT1, B4GALT1, B4GALT7, B9D2, BAAT, BAG3, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCKDHA, BCKDHB, BCKDK, BCL10, BCL11A, BCL11B, BCOR, BCS1L, BEAN1, BFSP2, BHLHA9, BICD2, BIN1, BLM, BLNK, BLOC1S3, BLOC1S6, BMP1, BMP2, BMP4, BMPER, BMPR1A, BMPR1B, BNC2, BOLA3, BPNT2, BRAF, BRAT1, BRCA1, BRCA2, BRIP1, BRSK2, BRWD3, BSCL2, BSND, BTB, BTK, BUB1B, C12orf57, C19orf12, C1QA, C1QB, C1QC, C1R, C1S, C2, C3, C4A, C4B, C5, C6, C7, C8A, C8B, C8G, C9, C9orf72, CA2, CA5A, CA8, CACNA1A, CACNA1C, CACNA1D, CACNA1E, CACNA1F, CACNA1G, CACNA1S, CACNB4, CAMTA1, CANT1, CAPN3, CARD11, CARD14, CARD9, CARS2, CASK, CASP10, CASP8, CASQ2, CASR, CAT, CAV3, CBL, CBLIF, CBS, CC2D1A, CC2D2A, CCBE1, CCDC103, CCDC22, CCDC39, CCDC40, CCDC65, CCDC8, CCDC88C, CCN6, CCNO, CCNQ, CCT5, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD55, CD59, CD79A, CD79B, CD81, CD8A, CDAN1, CDC6, CDH1, CDH15, CDH23, CDH3, CDK5RAP2, CDKL5, CDKN1C, CDON, CDSN, CDT1, CEBPE, CENPJ, CEP120, CEP135, CEP152, CEP164, CEP290, CEP41, CEP57, CEP63, CERT1, CFAP410, CFAP418, CFB, CFD, CFH, CFHR5, CFI, CFL2, CFP, CFTR, CIB2, CIC, CIITA, CILK1, CISD2, CLCN2, CLCN4, CLCN5, CLCN7, CLDN1, CLDN14, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CLPP, CLRN1, CNBP, CNGB3, CNKSR2, CNNM2, CNOT3, CNPY3, CNTNAP2, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COCH, COL10A1, COL11A1, COL11A2, COL12A1, COL17A1, COL18A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL4A2, COL4A3, COL4A4, COL4A5, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, COL7A1, COL9A1, COL9A2, COL9A3, COLEC11, COLQ, COMP, COPA, COQ2, COQ4, COQ6, COQ7, COQ9, CORO1A, COX10, COX14, COX15, COX20, COX6A1, COX6B1, COX7B, CP, CPA6, CPLANE1, CPOX, CPS1, CPT1A, CPT2, CR2, CRADD, CRB1, CREBBP, CRLF1, CRPPA, CRTAP, CRX, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYGD, CSF1R, CSF2RB, CSF3R, CSPP1, CSTB, CTC1, CTCF, CTDP1, CTH, CTLA4, CTNNA1, CTNND1, CTNS, CTSA, CTSC, CTSD, CTSK, CUBN, CUL4B, CUL7, CUX2, CXCR4, CY5R3, CYBA, CYBB, CYC1, CYCS, CYP11A1, CYP11B1, CYP17A1, CYP1B1, CYP21A2, CYP27A1, CYP27B1, CYP2U1, CYP4F22, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DBH, DBT, DCAF17, DCC, DCDC2, DCLRE1C, DCTN1, DCX, DDB2, DDC, DDHD1, DDHD2, DDOST, DDR2, DDX59, DEAF1, DES, DGKE, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHODH, DHTKD1, DCHS1, DIAPH1, DIS3L2, DISP1, DKC1, DLAT, DLD, DLG3, DLG4, DLL3, DLX3, DLX5, DMD, DMP1, DMPK, DNA2, DNAAF1, DNAAF11, DNAAF3, DNAH11, DNAH5, DNAH9, DNAI1, DNAJB6, DNAJC19, DNAJC5, DNASE2, DNM1, DNM1L, DNM2, DNMT1, DNMT3A, DNMT3B, DOCK3, DOCK6, DOCK8, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPP6, DPYD, DPYS, DSP, DSPP, DSTYK, DTNBP1, DUOX2, DYM, DYNC1H1, DYNC2H1, DYNC2I1, DYNC2I2, DYRK1A, DYSF, EARS2, EBP, ECEL1, EDA, EDAR, EDARADD, EDNRA, EDNRB, EEF1A2, EFHC1, EFN1, EFTUD2, EGR2, EHMT1, ECHS1, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, ELAC2, ELANE, ELN, ELOVL4, ELP2, EMC1, EMD, EMX2, ENG, ENO3, ENPP1, ENTPD1, EP300, EPB41, EPB42, EPG5, EPM2A, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ERF, ERLIN2, ESCO2, ESRRB, ETFA, ETFB, ETV6, ETHE1, ETV6, EVC, EVC2, EXOSC3, EXT1, EXT2, EXTL3, EYA1, EYA4, EZH2, F10, F11, F12, F13A1, F13B, F2, F5, F7, F8, F9, FA2H, FADD, FAH, FAM111A, FAM111B, FAM126A, FAM161A, FAM20A, FAM20C, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FARS2, FAS, FASLG, FASTKD2, FAT4, FBLN5, FBN1, FBN2, FBP1, FBXL4, FBXO11, FBXO7, FCGR3A, FCGR3B, FCN3, FECH, FERMT3,



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Seznam genů zahrnutých ve virtuálním panelu – Závažná dětská onemocnění (FocusedExome)

UNC119, UNC13D, UNC80, UNC93B1, UNG, UPF3B, UQCRB, UROC1, UROD, UROS, USB1, USH1C, USH1G, USH2A, USP9X, UVSSA, VAMP1, VARS2, VCAN, VCP, VDR, VHL, VIPAS39, VKORC1, VLDLR, VMA21, VPS13A, VPS13B, VPS33B, VPS45, VRK1, VSX2, VWF, WAC, WARS2, WAS, WDPCP, WDR11, WDR19, WDR35, WDR4, WDR45, WDR45B, WDR62, WDR81, WFS1, WIPF1, WNT10A, WNT10B, WNT3, WNT5A, WNT7A, WRAP53, WRN, WT1, WWOX, XDH, XIAP, XPA, XPC, XRCC4, XYLT1, XYLT2, YARS2, YY1, ZAP70, ZBTB18, ZBTB24, ZC4H2, ZDHHC9, ZEB2, ZFP57, ZFYVE26, ZIC1, ZIC2, ZIC3, ZMPSTE24, ZMYND11, ZNF469, ZNF711