

## CENTOGENE's Curated Gene List of Pathogenic and Likely Pathogenic Variants Indicated in Severe and Early-Onset Disorders

VALID FROM 18.11.2020

AAAS, AARS1, AARS2, ABAT, ABCA12, ABCA3, ABCB11, ABCB4, ABCB7, ABCC6, ABCC8, ABCC9, ABCD1, ABCD4, ABHD12, ABHD5, ACACA, ACAD9, ACADM, ACADS, ACADVL, ACAN, ACAT1, ACE, ACO2, ACOX1, ACP5, ACSL4, ACTA1, ACTA2, ACTB, ACTG1, ACTL6B, ACVR2B, ACVRL1, ACY1, ADA, ADAM17, ADAM22, ADAMTS19, ADAMTS2, ADAMTSL2, ADAR, ADARB1, ADAT3, ADCY5, ADGRG1, ADGRG6, ADGRV1, ADK, ADNP, ADPRHL2, ADSL, AFF2, AFG3L2, AGA, AGK, AGL, AGPAT2, AGPS, AGR2, AGRN, AGT, AGTPBP1, AGTR1, AGXT, AHCY, AHDC1, AHI1, AIFM1, AIMP1, AIPL1, AIRE, AK2, AKR1D1, AKT2, AKT3, ALAD, ALDH18A1, ALDH1A3, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALOX12B, ALPL, ALX3, ALX4, AMACR, AMER1, AMN, AMPD1, AMPD2, AMT, ANK2, ANK3, ANKH, ANKRD11, ANKS6, ANO10, ANO5, ANOS1, ANTXR1, ANTXR2, AP1B1, AP1S1, AP1S2, AP3B1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APC2, APTX, AR, ARCN1, ARFGF2, ARG1, ARHGAP31, ARHGDI, ARHGEF9, ARID1A, ARID1B, ARID2, ARL13B, ARL3, ARL6, ARL6IP1, ARMC4, ARMC9, ARNT2, ARSA, ARSB, ARSL, ARV1, ARX, ASAH1, ASCC1, ASH1L, ASL, ASNS, ASPA, ASPH, ASPM, ASS1, ASXL1, ASXL2, ASXL3, ATAD3A, ATCAY, ATIC, ATL1, ATOH7, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2B3, ATP5MD, ATP6AP2, ATP6V0A2, ATP6V0A4, ATP6V1A, ATP6V1B1, ATP6V1B2, ATP7A, ATP7B, ATP8A2, ATP8B1, ATRX, AUH, AUTS2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT7, B4GAT1, B9D1, B9D2, BANF1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCKDHA, BCKDHB, BCKDK, BCL11A, BCOR, BCS1L, BGN, BHLHA9, BICD2, BIN1, BLM, BLNK, BMP1, BMP2, BMP4, BMPER, BMPR1B, BOLA3, BPTF, BRAT1, BRD4, BRPF1, BRWD3, BSCL2, BSND, BTB, BTK, C12orf57, C12orf65, C15orf41, C19orf12, C1QBP, C2CD3, C8orf37, CA2, CA5A, CA8, CACNA1A, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CAD, CAMK2A, CAMK2B, CAMTA1, CANT1, CARD11, CARS2, CASK, CASR, CAV1, CAVIN1, CBL, CBS, CC2D1A, CC2D2A, CCB1, CCDC103, CCDC114, CCDC115, CCDC151, CCDC22, CCDC39, CCDC40, CCDC47, CCDC65, CCDC78, CCDC8, CCDC88A, CCDC88C, CCND2, CCNK, CCNO, CCNQ, CCT5, CD19, CD247, CD2AP, CD3D, CD3E, CD3G, CD40, CD40LG, CD79A, CD79B, CD96, CDC45, CDH2, CDH23, CDH3, CDK10, CDK13, CDK19, CDK19, CDK5RAP2, CDKL5, CDKN1C, CDON, CDSN, CDT1, CENPF, CENPJ, CEP104, CEP135, CEP152, CEP164, CEP290, CEP41, CEP57, CEP63, CEP83, CERS1, CERS3, CERT1, CFAP298, CFAP300, CFAP410, CFC1, CFL2, CFTR, CHAMP1, CHAT, CHD2, CHD3, CHD4, CHD7, CHD8, CHKB, CHM, CHMP1A, CHRDL1, CHRNA1, CHRNA2, CHRNA4, CHRN1, CHRN2, CHRNE, CHRNA3, CHST14, CHST3, CHSY1, CHUK, CIB2, CILK1, CISD2, CIT, CKAP2L, CLCN1, CLCN2, CLCN4, CLCN5, CLCN7, CLCNKA, CLCNKB, CLDN1, CLDN16, CLDN19, CLMP, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLRN1, CLTC, CNKSR2, CNNM2, CNOT1, CNOT3, CNPY3, CNTNAP1, CNTNAP2, COA6, COA8, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL10A1, COL11A1, COL11A2, COL17A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A1, COL4A2, COL4A3, COL4A4, COL4A5, COL6A1, COL6A2, COL6A3, COL7A1, COL9A1, COL9A2, COL9A3, COLEC10, COLEC11, COLQ, COMP, COQ2, COQ4, COQ6, COQ8A, COQ8B, COQ9, CORO1A, COX10, COX15, COX20, COX6B1, COX7B, CPLANE1, CPS1, CPT1A, CPT2, CRADD, CRB1, CRB2, CRBN, CREB3L1, CREBBP, CRELD1, CRIPT, CRKL, CRLF1, CRPPA, CRTAP, CRX, CRYAA, CSF1R, CSF2RB, CSF3R, CSNK1E, CSNK2A1, CSNK2B, CSPP1, CSTA, CSTB, CTC1, CTCF, CTD1P1, CTNNA2, CTNNA1, CTNND1, CTNS, CTSP1, CTSA, CTSD, CTSK, CTU2, CUL3, CUL4B, CUL7, CWC27, CWF19L1, CXCR4, Cxorf56, CYB5R3, CYBA, CYBB, CYC1, CYP11A1, CYP11B1, CYP11B2, CYP17A1, CYP21A2, CYP24A1, CYP27A1, CYP27B1, CYP2U1, CYP4F22, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DBT, DCAF17, DCDC2, DCHS1, DCLRE1C, DCX, DDB2, DDC, DDHD1, DDHD2, DDR2, DDX11, DDX3X, DDX59, DDX6, DEAF1, DENND5A, DEPDC5, DGAT1, DGKE, DGUOK, DHCR24, DHCR7, DHDDS, DHH, DHODH, DHTKD1, DHX16, DHX30, DHX37, DIAPH1, DIS3L2, DKC1, DLAT, DLD, DLG3, DLG4, DLL1, DLL3, DLL4, DLX5, DMD, DMP1, DMPK, DMXL2, DNA2, DNAAF3, DNAAF4, DNAAF5, DNAH5, DNAH9, DNAJC12, DNAJC19, DNAJC3, DNAJC5, DNM1, DNM1L, DNM2, DNMT3A, DNMT3B, DOCK6, DOCK7, DOCK8, DOK7, DOLK, DPAGT1, DPF2, DPM1, DPM2, DPYD, DRC1, DSE, DSG1, DSP, DST, DSTYK, DTNA, DUOX2, DUOX2A, DVL1, DVL3, DYM, DYNC1H1, DYNC2H1, DYRK1A, DYSF, EARS2, EBF3, EBP, ECEL1, ECHS1, EDA, EDAR, EDN3, EDNRA, EDNRB, EED, EEF1A2, EFEMP2, EFN1, EFTUD2, EGR2, EHMT1, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF4A3, ELANE, ELMO2, ELN, ELOVL4, ELP1, ELP2, EMC1, EMC10, EMD, EMG1, EMX2, ENG, ENPP1, ENTPD1, EOGT, EPB42, EPCAM, EPG5, EPHB4, EPM2A, EPRS1, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6L2, ERCC8, ERF, ERLIN1, ERLIN2, ESCO2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOC3L2, EXOSC3, EXOSC8, EXOSC9, EXPH5, EXT1, EXT2, EXTL3, EYA1, EZH2, F10, F13A1, F2, F7, F8, F9, FA2H, FADD, FAH, FAM111A, FAM126A, FAM149B1, FAM161A, FAM20A, FAM20C, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAR1, FARS2, FASTKD2, FAT4, FBLN5, FBN1, FBN2, FBP1, FBXL4, FBXO11, FBXW11, FBXW4, FCSK, FEZF1, FGA, FGB, FGD1, FGD4, FGF10, FGF12, FGF3, FGF8, FGF9, FGFR1, FGG, FHL1, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLNA, FLNB, FLT4, FLVCR1, FLVCR2, FMN2, FMR1, FN1, FOLR1, FOXC1, FOXC2, FOXE3, FOXF1, FOXG1, FOXL2, FOXN1, FOXP1, FOXP3, FOXRED1, FRAS1, FREM1, FREM2, FRMPD4, FRRS1L, FTCD, FTL, FTSJ1, FUCA1, FUT8, FXN, G6PC, G6PC3, GAA, GABBR2, GABRA1, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GALT, GALE, GALK1, GALNS, GALT, GAMT, GAN, GAS8, GATA1, GATA2, GATA3, GATA4, GATA6, GATAD2B, GATM, GBA, GBA2, GBE1, GCDH, GCH1, GCK, GDAP1, GDF1, GDF3, GDF5, GDF6, GDI1, GEMIN4, GFAP, GFER, GFM1, GFM2, GFPT1, GHR, GJA1, GJA8, GJB2, GJB3, GJC2, GK, GLA, GLB1, GLDC, GLDN, GLE1, GLI2, GLI3, GLIS2, GLIS3, GLMN, GLRX5, GLUD1, GLUL, GLYCTK, GM2A, GMNN, GMPA, GMPB, GNA11, GNAI3, GNAO1, GNAQ, GNAS, GNB1, GNB5, GNPAT, GNPTAB, GNPTG, GNS, GOLGA2, GORAB, GOSR2, GOT2, GPAA1, GPC3, GPC6, GPHN, GPSM2, GPT2, GPX4, GREB1L, GRHL2, GRHL3, GRIA2, GRIA3, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRIP1, GRM1, GRM7, GSS, GTF2H5, GTPBP1, GTPBP3, GUCY2C, GUF1, GUSB, GYS1, GZF1, H1-4, HACD1, HACE1, HADH, HADHA, HADHB, HARS1, HAX1, HBA1, HBA2, HBB, HCCS, HCFC1, HCN1, HDAC4, HDAC8, HECW2,



TMEM107, TMEM126B, TMEM138, TMEM165, TMEM199, TMEM216, TMEM231, TMEM237, TMEM67, TMEM70, TMEM94, TMPRSS6, TMTC3, TMX2, TNFRSF11B, TNFRSF13B, TNFSF11, TNNT1, TOE1, TOP3A, TP63, TPI1, TPK1, TPM2, TPO, TPP1, TRAPPC11, TRAPPC2, TRAPPC4, TRAPPC9, TREX1, TRIM2, TRIM32, TRIM37, TRIO, TRIP11, TRIP12, TRIP13, TRIP4, TRIT1, TRMT1, TRMT10A, TRMU, TRNT1, TRPM1, TRPM6, TRPS1, TRPV3, TRPV4, TRPV6, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN54, TSFM, TSHB, TSHR, TSPAN7, TSPYL1, TTC19, TTC21B, TTC26, TTC37, TTC7A, TTC8, TTN, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUBGCP2, TUBGCP6, TUFM, TUSC3, TWIST1, TWIST2, TWNK, TXNL4A, TYMP, TYR, TYRP1, UBA1, UBA5, UBE2A, UBE2T, UBE3A, UBE3B, UBR1, UBTF, UFC1, UFM1, UGDH, UGP2, UGT1A1, UMPS, UNC13D, UNC80, UPB1, UPF3B, UQCC2, UQCRC2, UROC1, UROS, USB1, USH1C, USH1G, USH2A, USP18, USP53, USP9X, UVSSA, VAMP1, VAMP2, VANGL1, VARS2, VDR, VIPAS39, VLDLR, VPS13B, VPS33B, VPS37A, VPS41, VPS45, VPS53, VRK1, VSX2, WAC, WAS, WASF1, WDFY3, WDPCP, WDR11, WDR19, WDR26, WDR34, WDR35, WDR37, WDR45, WDR45B, WDR60, WDR62, WDR73, WDR81, WHRN, WNK1, WNT1, WNT10A, WNT10B, WNT3, WNT4, WNT5A, WNT7A, WRAP53, WT1, WWOX, XIAP, XPA, XPC, XRCC4, XYLT1, XYLT2, YAP1, YARS2, YIF1B, YWHAG, YY1, ZAP70, ZBTB18, ZBTB20, ZBTB24, ZC3H14, ZC4H2, ZDHHC9, ZEB2, ZFP57, ZFPM2, ZFYVE26, ZIC1, ZIC2, ZIC3, ZMIZ1, ZMPSTE24, ZMYM6, ZMYND10, ZMYND11, ZNF335, ZNF423, ZNF462, ZNF711, ZNHIT3

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