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## ENDOCRINOLOGY DISORDER PANEL (226 GENES)

ABCA1, ABCC8, ABCG5, ABCG8, ACAT1, ACSF3, ADCY3, AGL, AIRE, ALDOA, ALDOB, ALMS1, AMH, AMHR2, ANOS1, AP2S1, APOA1, APOA5, APOB, APOC2, APOC3, APOE, AR, ARL6, ARMC5, ARX, ATRX, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BLK, BMP15, BSND, CASR, CDC73, CDKN1A, CDKN1B, CDKN1C, CDKN2B, CDKN2C, CEP290, CEP41, CHD7, CLCNKB, CLDN16, CLDN19, CNNM2, CNNM4, CREB3L3, CREBBP, CUL4B, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DUOX2, DUOXA2, DYNC2H1, DYRK1B, EGF, EIF2AK3, ENO3, EPM2A, ERCC3, FAM111A, FBP1, FGF8, FGFR1, FIG4, FOXE1, FOXL2, FOXP3, FRAS1, FSHR, FXD2, GAA, GALT, GATA4, GATA6, GBE1, GCK, GCM2, GLIS3, GLUD1, GNA11, GNAS, GNRHR, GPIHBP1, GYG1, GYS1, GYS2, HADH, HESX1, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HSD17B3, HSD3B2, IGSF1, IL17RD, INS, INSR, IRF6, KCNA1, KCNJ10, KCNJ11, KISS1R, KLF11, KSR2, LAMP2, LDHA, LDLR, LDLRAP1, LEP, LEPR, LHCGR, LIPA, LMF1, LMNA, LPL, MAGEL2, MAGT1, MAMLD1, MAP3K1, MC2R, MC3R, MC4R, MEN1, MKKS, MKRN3, MKS1, MPV17, MRAP, NEUROD1, NEUROG3, NHLRC1, NIPA2, NKX2-1, NKX2-5, NNT, NOBOX, NROB1, NROB2, NR3C1, NR5A1, NTRK2, OXCT1, PAX4, PAX8, PC, PCBD1, PCK1, PCSK1, PCSK9, PDX1, PFKM, PGAM2, PGK1, PGM1, PHF6, PHKA1, PHKA2, PHKB, PHKG2, POLG, POMC, POR, POU1F1, PPARG, PRKAG2, PRKAG3, PRKAR1A, PROK2, PROKR2, PROP1, PTF1A, PTH, PYGL, PYGM, RBCK1, RET, RFX6, RSP01, SARS2, SDCCAG8, SECISBP2, SIM1, SLC12A3, SLC16A1, SLC16A2, SLC26A4, SLC2A2, SLC37A4, SLC5A5, SOX9, SRD5A2, SRY, STAR, TACR3, TG, THRA, THRB, TPO, TRPM6, TSHB, TSHR, TTC8, UCP2, UCP3, VPS13B, WDPCP, WFS1, WT1, ZFP57, ZFPM2.

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## **GASTROENTEROLOGY PANEL (146 GENES)**

LIPA, GPIHBP1, BBS1, BBS2, MKKS, BDNF, TCTN2, AHI1, B9D2, NPC2, CEP290, CEP41, PHOX2B, ANKS6, DCDC2, ATP8B1, IL10RA, IL10, DGAT1, BBS9, PEX26, BBS4, ABCB11, MKS1, IL21, PEX1, NPHP1, CREB3L3, ZEB2, CLMP, SLC5A1, MITF, EDN3, SOX2, CC2D2A, STX3, APOC2, IQCB1, XIAP, MYO5B, BAAT, WDR19, RET, EFTUD2, MVK, TRMU, SPINT2, NPHP3, PAX3, ARL6, TMEM231, ARL13B, PKHD1, ZNF423, CPA1, NPC1, DHCR7, NEUROG3, INVS, CYP7B1, INPP5E, SEC63, SI, LRP5, NPHP4, SLC26A3, JAG1, L1CAM, TTC8, HSD3B7, VIPAS39, SERPINA1, GLI3, CHD7, GUCY2C, TTC21B, RPGRIP1L, SLC9A3, EDNRB, LCT, SPINK1, PKD1, BBS12, TCTN1, CELSR3, PKD2, TCTN3, LMF1, GLIS2, CYP27A1, APOA5, NRTN, SLC10A2, PRSS1, NEK8, UBR1, TMEM138, CFTR, EPCAM, NRG1, PEX2, SMPD1, MID1, SAR1B, B9D1, TTC7A, TTC37, PEX5, DGUOK, BBS10, NOTCH2, BBS7, AKR1D1, TMEM237, RMRP, RFX6, FAH, KIF7, SLC25A13, WDR35, OFD1, NCF2, SKIV2L, SOX10, NR1H4, ABCC2, FOXP3, MYCN, CEP164, IL10RB, TJP2, FANCC, PRKCSH, PEX6, ABCB4, FANCB, PEX12, ADAM17, PTF1A, VPS33B, CTRC, TMEM216, TMEM67, PEX10, BBS5, GANAB.

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## MITOCHONDRIAL PANEL (368 GENES)

AARS2, AASS, ABAT, ABCB7, ACACA, ACAD9, ACADM, ACADS, ACADVL, ACAT1, ACO2, ADAR, AFG3L2, AGK, AIFM1, AK2, ALAS2, ALDH3A2, AMPD1, AMT, APOPT1, APTX, ATP5A1, ATP5D, ATP5E, ATP7B, ATPAF2, AUH, BAG3, BCS1L, BOLA3, BTBD9, C12ORF65, C19ORF12, C19ORF70, C1QBP, CA5A, CARS2, CEP89, CHAT, CHCHD10, CLPB, CLPP, COA3, COA5, COA6, COA7, COASY, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX4I2, COX6A1, COX6B1, COX7B, COX8A, CPS1, CPT1A, CPT2, CYC1, CYCS, D2HGDH, DARS2, DES, DGUOK, DLAT, DLD, DNA2, DNAJC19, DNMT1, EARS2, ECHS1, ELAC2, ETFA, ETFB, ETFDH, ETHE1, FARS2, FASTKD2, FBXL4, FDX2, FH, FLAD1, FOXRED1, GAMT, GARS, GATM, GCDH, GDAP1, GFER, GFM1, GFM2, GLDC, GLRX5, GTPBP3, GYG2, HADH, HADHA, HADHB, HARS2, HCCS, HIBCH, HLCS, HMGCL, HMGCS2, HSD17B10, HSPD1, HTRA2, IARS2, IBA57, IDH2, IDH3B, IFIH1, ISCA1, ISCA2, ISCU, KARS, L2HGDH, LAMP2, LARS, LARS2, LIAS, LIPT1, LIPT2, LMBRD1, LONP1, LRPPRC, LYRM4, LYRM7, MARS2, MECR, MFF, MFN2, MGME1, MICU1, MIPEP, MPC1, MPV17, MRPL12, MRPL3, MRPL40, MRPL44, MRPS14, MRPS16, MRPS2, MRPS22, MRPS23, MRPS34, MRPS7, MSTO1, MTFMT, MTHFD1, MTO1, MTPAP, NADK2, NARS2, NAXE, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF7, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFS1, NFU1, NGLY1, NNT, NR2F1, NSUN3, NUBPL, NUP62, OGDH, OPA1, OPA3, OTC, OXCT1, PANK2, PARS2, PC, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1, PDSS2, PET100, PINK1, PITRM1, PMPCA, PMPCB, PNKD, PNPLA8, PNPT1, POLG, POLG2, POP1, PPA2, PPOX, PSAP, PUS1, QARS, QRSL1, RANBP2, RARS, RARS2, REEP1, RMND1, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, RRM2B, SACS, SAMHD1, SARS2, SCN1A, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHC, SDHD, SERAC1, SFXN4, SIRT1, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A21, SLC25A22, SLC25A26, SLC25A3, SLC25A32, SLC25A38, SLC25A4, SLC25A42, SLC25A46, SLC39A8, SLC52A2, SLC52A3, SLC6A8, SLC7A13, SPAST, SPG7, STAT2, STXBP1, SUCLA2, SUCLG1, SUCLG2, SUGCT, SURF1, TACO1, TANGO2, TARS2, TAZ, TFAM, TIMM50, TIMM8A, TIMMDC1, TK2, TMEM126A, TMEM126B, TMEM70, TOP1MT, TOP3A, TPK1, TRAP1, TREX1, TRIT1, TRMT10C, TRMT5, TRMU, TRNT1, TSFM, TTC19, TUFM, TWNK, TXN2, TYMP, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ, VARS2, WARS2, WDR45, WFS1, XPNPEP3, YARS2, YME1L1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MTND1, MT-ND2, MT-ND3, MT-ND4, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY.

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## COMPREHENSIVE NEPHROLOGY PANEL (401 GENES)

ABCC6, ACE, ACTB, ACTN4, ADA2, ADAMTS13, ADCY10, AGPAT2, AGT, AGTR1, AGXT, AHI1, ALG1, ALG8, ALG9, ALMS1, ALPL, AMN, ANKS6, ANLN, ANOS1, AP2S1, APOA1, APOC2, APOL1, APRT, AQP2, ARHGAP24, ARHGDI1, ARL13B, ARL6, ATP6VOA4, ATP6V1B1, ATP7B, AVP, AVPR2, B2M, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCS1L, BICC1, BMP4, BNC2, BRAF, BSND, C8ORF37, CA2, CACNA1D, CACNA1H, CASR, CBL, CC2D2A, CD151, CD2AP, CDC73, CDKN1C, CEP104, CEP120, CEP164, CEP19, CEP290, CEP41, CEP83, CEP89, CFH, CFHR5, CFI, CHD7, CHRM3, CHRNA3, CISD2, CLCN2, CLCN5, CLCNKB, CLDN16, CLDN19, CNNM2, COL4A1, COL4A3, COL4A4, COL4A5, COL4A6, COQ2, COQ6, COQ8B, COX10, COX14, CPLANE1, CPT2, CRB2, CREBBP, CRKL, CSPP1, CTNS, CUBN, CUL3, CYP11B1, CYP11B2, CYP17A1, CYP24A1, CYP27B1, CYP2R1, DCDC2, DCHS1, DGKE, DHCR7, DHTKD1, DICER1, DLC1, DMP1, DNAJB11, DNASE1L3, DSTYK, DZIP1L, EBP, EGF, EIF2AK3, ELP1, EMP2, ENPP1, EYA1, FAM20A, FAN1, FANCA, FAT1, FAT4, FBN3, FGF10, FGF20, FGF23, FGFR1, FGFR2, FN1, FOXC1, FOXC2, FOXI1, FOXP3, FRAS1, FREM1, FREM2, FXYD2, G6PC, GALNT3, GANAB, GATA3, GATM, GCM2, GLA, GLI3, GLIS2, GLIS3, GNA11, GNAS, GPC3, GPHN, GREB1L, GRHRP, GRIP1, GSN, HGD, HNF1A, HNF1B, HOGA1, HOXA13, HPRT1, HPS1, HPSE2, HSD11B2, IFT122, IFT140, IFT172, IFT27, IFT43, IFT74, INF2, INPP5E, INVS, IQCB1, ITGA3, ITGA6, ITGA8, ITGB4, ITSN2, JAG1, KANK1, KANK2, KANK4, KANSL1, KAT6B, KCNA1, KCNJ1, KCNJ10, KCNJ5, KCTD1, KIAA0586, KIF7, KLHL3, KRAS, LAGE3, LAMA5, LAMB2, LCAT, LDHA, LMNA, LMX1B, LPIN1, LRP2, LRP4, LRP5, LYZ, LZTFL1, LZTR1, MAFB, MAGED2, MAGI2, MAP2K1, MAP2K2, MAPKBP1, MEFV, MKKS, MKS1, MMACHC, MNX1, MOCOS, MOCS1, MOCS2, MRE11, MUT, MVK, MYCN, MYH9, MYO1E, NEK8, NF1, NLRP3, NOTCH2, NPHP1, NPHP3, NPHP4, NPHS1, NPHS2, NROB1, NR3C2, NRAS, NRIP1, NSD1, NSDHL, NUP107, NUP133, NUP160, NUP205, NUP85, NUP93, NXF5, OCRL, OFD1, OPLAH, OSGEP, PAX2, PBX1, PCBD1, PDE6D, PDSS1, PDSS2, PEX6, PHEX, PKD2, PKHD1, PLCE1, PLG, PMM2, PODXL, PREPL, PRKCSH, PRODH, PROKR2, PRPS1, PTPN11, PTPRO, RAF1, REN, RET, RIT1, RMND1, ROBO1, ROBO2, RPGRIP1L, RPL11, RPL26, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS7, RRM2B, SALL1, SALL4, SARS2, SCARB2, SCLT1, SCN4A, SCNN1A, SCNN1B, SCNN1G, SDCCAG8, SEC61A1, SEC63, SEMA3E, SGPL1, SHOC2, SI, SIX1, SIX2, SIX5, SLC12A1, SLC12A3, SLC22A12, SLC26A1, SLC2A2, SLC2A9, SLC34A1, SLC34A3, SLC36A2, SLC37A4, SLC3A1, SLC41A1, SLC4A1, SLC4A4, SLC5A1, SLC6A19, SLC7A7, SLC7A9, SLC9A3R1, SLIT2, SLX4, SMARCAL1, SMC1A, SOS1, SOS2, SOX17, SOX18, SRCAP, STX16, SYNPO, TBC1D8B, TBX18, TCTN1, TCTN2, TCTN3, TFAP2A, THBD, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TNS2, TP53RK, TP63, TPRKB, TRAPPC3, TRIM32, TRPC6, TRPM6, TSC1, TSC2, TTC21B, TTC8, TTR, UMOD, UQCC2, VDR, VHL, VIPAS39, VPS33B, WAS, WDPCP, WDR19, WDR73, WFS1, WNK1, WNK4, WNT4, WNT5A, WT1, XDH, XPNPEP3, XPO5, YRDC, ZNF423.

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## COMPREHENSIVE OPHTHALMOLOGY PANEL (438 GENES)

AASS, ABCA4, ABCB6, ABHD12, ACO2, ADAM9, ADAMTS10, ADAMTS17, ADAMTS18, ADAMTSL4, ADGRV1, ADIPOR1, AGBL5, AGK, AHI1, AIPL1, ALDH18A1, ALDH1A3, ALMS1, ANTXR1, AP3B1, APTX, ARHGEF18, ARL13B, ARL2BP, ARL6, ARMC9, ARMS2, ASPH, ATF6, ATOH7, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BEST1, BFSP1, BFSP2, BLOC1S3, BLOC1S6, BMP4, BMP7, C2, C3, C9, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CBS, CC2D2A, CDH23, CDH3, CDHR1, CEP104, CEP120, CEP164, CEP290, CEP41, CEP78, CERKL, CFB, CFH, CFI, CHD7, CHM, CHMP4B, CHN1, CHRDL1, CHST6, CIB2, CISD2, CLN3, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, CNTNAP2, COL11A1, COL11A2, COL17A1, COL18A1, COL2A1, COL4A1, COL5A1, COL8A2, COL9A1, COL9A2, COL9A3, COX7B, CRB1, CRX, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYGD, CRYGS, CSPP1, CST3, CTC1, CTDTP1, CTNNA1, CTNNB1, CWC27, CX3CR1, CYP1B1, CYP27A1, CYP4V2, DCN, DHDDS, DHX38, DRAM2, DTHD1, DTNBP1, EFEMP1, ELOVL4, EMC1, EPHA2, ERCC2, ERCC5, ERCC6, ERCC8, EYA1, EYS, FAM126A, FAM161A, FBLN5, FBN1, FBN2, FDXR, FLVCR1, FOXC1, FOXE3, FOXL2, FRAS1, FREM1, FREM2, FRMD7, FSCN2, FTL, FYCO1, FZD4, GALE, GALK1, GALT, GCNT2, GJA1, GJA3, GJA8, GNAT1, GNAT2, GNB3, GNPTG, GPR143, GPR179, GRIP1, GRM6, GSN, GUCA1A, GUCY2D, HCCS, HESX1, HGSNAT, HK1, HMCN1, HMX1, HPS1, HPS3, HPS4, HPS5, HPS6, HSF4, HTRA1, IDH3B, IFT140, IFT172, IFT81, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, JAG1, KCNV2, KERA, KIAA0586, KIAA0753, KIF11, KIF21A, KIF7, KIZ, KLHL7, KRT12, KRT3, LCA5, LCAT, LIM2, LMX1B, LOXHD1, LRAT, LRIT3, LRP2, LRP5, LTBP2, LYST, LZTFL1, MAF, MAK, MC1R, MERTK, MFN2, MFRP, MFSD8, MIP, MITF, MKKS, MKS1, MLPH, MMACHC, MT-TL1, MTPP, MVK, MYH9, MYO5A, MYO7A, MYOC, NAA10, NDP, NDUFS1, NEK2, NF2, NHS, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NYX, OAT, OCA2, OCRL, OFD1, OPA1, OPA3, OPTN, OTX2, OVOL2, P3H2, PANK2, PAX2, PAX6, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHOX2A, PHYH, PIKFYVE, PITPNM3, PITX2, PITX3, PLA2G5, PNPLA6, POC1B, POLG, PORCN, PQBP1, PRDM13, PRDM5, PRKCG, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, PRSS56, PXDN, RAB27A, RAB28, RAB3GAP1, RARB, RAX, RAX2, RBP3, RBP4, RD3, RDH11, RDH12, RDH5, RECQL4, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROBO3, ROM1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, RPGRIP1L, RRM2B, RS1, RTN4IP1, SAG, SALL4, SAMD11, SDCCAG8, SEMA4A, SETX, SHH, SIL1, SIX3, SIX6, SLC16A12, SLC24A1, SLC24A5, SLC25A4, SLC25A46, SLC33A1, SLC38A8, SLC45A2, SLC4A11, SLC52A2, SLC7A14, SMCHD1, SMOC1, SNRNP200, SNX10, SOX2, SPATA7, SPG7, SPP2, STRA6, SUOX, TACSTD2, TBK1, TCF4, TCTN1, TCTN2, TCTN3, TDRD7, TEAD1, TEK, TENM3, TFAP2A, TGFBI, TIMM8A, TIMP3, TK2, TMEM107, TMEM126A, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TMEM70, TOPORS, TRAF3IP1, TREX1, TRPM1, TSPAN12, TTC21B, TTC8, TTLL5, TTPA, TUB, TUBB3, TULP1, TYMP, TYR, TYRP1, UBIAD1, USH1C, USH1G, USH2A, VCAN, VIM, VPS13B, VSX2, WDPCP, WDR19, WDR36, WFS1, WRN, ZEB1, ZIC2, ZNF408, ZNF423, ZNF469, ZNF513.

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## **AUTOINFLAMMATORY AND AUTOIMMUNITY SYNDROMES PANEL (156 GENES)**

ACP5, ADA, ADA2, ADAM17, ADAR, AICDA, AIRE, ANKZF1, AP3B1, ARPC1B, ASAH1, BACH2, BLOC1S6, BTK, C17ORF62, CARD14, CARD8, CASP10, CASP8, CCBE1, CD27, CD3G, CD40, CD40LG, COPA, CR2, CTLA4, CYBA, CYBB, DCLRE1C, DDX58, DEF6, DKC1, DNASE1L3, DNASE2, DOCK8, DSG1, DUOX2, ELANE, FADD, FAS, FASLG, FCHO1, FOXP3, G6PC3, ICOS, IFIH1, IL10, IL10RA, IL10RB, IL1RN, IL21, IL21R, IL2RA, IL2RB, IL2RG, IL36RN, IRF2BP2, ITCH, ITGAM, ITGB2, ITK, JAK1, LIG4, LPIN2, LRBA, LYN, LYST, MAGT1, MEFV, MVK, NCF2, NCF4, NFAT5, NFKB1, NFKB2, NFKBIA, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, OAS1, ORAI1, OTULIN, PEPD, PIK3CD, PIK3R1, PLCG2, PNP, POLA1, POMP, PRF1, PRKCD, PSMA3, PSMB4, PSMB8, PSMG2, PSTPIP1, RAB27A, RAC2, RAG1, RAG2, RASGRP1, RBCK1, RFX5, RFXANK, RFXAP, RIPK1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF31, RTEL1, SAMHD1, SCO2, SH2D1A, SH3BP2, SI, SIAE, SKIV2L, SLC29A3, SLC37A4, SLC7A7, STAT1, STAT3, STAT4, STAT5B, STIM1, STX11, STXBP2, TBX1, TGFB1, TGFBR1, TGFBR2, TMEM173, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF6B, TNFSF12, TOP2B, TPP2, TREX1, TRNT1, TTC37, TTC7A, UNC13D, UNG, WAS, XIAP, ZAP70, ZNF341.

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## HEARING LOSS AND DEAFNESS PANEL (262 GENES)

ABHD12, ABHD5, ACOX1, ACTG1, ADCY1, ADGRV1, AIFM1, ALMS1, ANLN, ARSB, ARSG, ATP1A3, ATP2B2, ATP6V1B1, BCAP31, BCS1L, BSND, BTD, CABP2, CACNA1D, CCDC50, CD151, CD164, CDC14A, CDH23, CEACAM16, CEP250, CEP78, CHD7, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DBH, DCAF17, DCDC2, DFNA5, DFNB59, DIABLO, DIAPH1, DIAPH3, DMXL2, DNMT1, DSPP, EDN3, EDNRA, EDNRB, EFTUD2, ELMOD3, EPS8, EPS8L2, ESPN, ESRRB, EYA1, EYA4, FAM65B, FGF3, FGFR3, FOXC1, FOXI1, GALNS, GATA3, GDF6, GIPC3, GJA1, GJB1, GJB2, GJB3, GJB6, GLB1, GNS, GPSM2, GRHL2, GRXCR1, GRXCR2, GUSB, HARS, HARS2, HGF, HGSNAT, HOMER2, HSD17B4, HYAL1, IDS, IDUA, ILDR1, JAG1, KARS, KCNE1, KCNQ1, KCNQ4, KCNJ10, KITLG, LARS2, LHFPL5, LHX3, LOXHD1, LOXL3, LRP2, LRTOMT, MAN2B1, MARVELD2, MCM2, MEOX1, MET, MIR96, MITF, MPZ, MSRB3, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MT-RNR1, MT-RNR2, MT-TA, MT-TC, MT-TD, MT-TE, MT-TF, MT-TG, MT-TH, MT-TI, MT-TK, MT-TL1, MT-TL2, MT-TM, MT-TN, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MYH14, MYH7B, MYH9, MYO15A, MYO18B, MYO3A, MYO6, MYO7A, NAGLU, NARS2, NDRG1, NF2, NLRP3, NOG, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX3, PCDH15, PCGF2, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PMP22, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, RAI1, RDX, RMND1, ROR1, RPS6KA3, S1PR2, SCP2, SERAC1, SERPINB6, SGSH, SH3TC2, SIX1, SIX5, SLC12A2, SLC17A8, SLC22A4, SLC26A4, SLC26A5, SLC29A3, SLC44A4, SLC4A11, SLC52A2, SLC52A3, SLITRK6, SMPX, SNAI2, SOX10, SYNE4, TBC1D24, TCOF1, TECTA, TFAP2A, TIMM8A, TJP2, TMC1, TMEM126A, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TRRAP, TSPEAR, TUBB4B, TWNK, UBR1, USH1C, USH1G, USH2A, VCAN, WBP2, WFS1, WHRN, ZNF469.

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## CARDIOMETABOLIC DISORDERS PANEL (523 GENES)

A2ML1, ABCC9, ABCG5, ABCG8, ACADVL, ACTA1, ACTA2, ACTC1, ACTN2, ACVR2B, ACVRL1, ADAMTS2, AGL, AKAP9, ALMS1, ANK2, ANKRD1, ANO5, APOB, ATP2A1, ATP7A, B3GALNT2, B4GAT1, BAG3, BCOR, BIN1, BMPR1B, BMPR2, BRAF, CACNA1C, CACNA2D1, CACNB2, CALM1, CALM2, CALM3, CALR3, CAPN3, CASQ2, CAV1, CAV3, CBL, CBS, CCDC78, CFL2, CHD7, CHKB, CHRM2, CHST14, CNTN1, COL12A1, COL1A1, COL1A2, COL3A1, COL5A1, COL5A2, COL6A1, COL6A2, COL6A3, CPT2, CRELD1, CRTAP, CRYAB, CSRP3, CTF1, CTNNA3, DAG1, DEPDC5, DES, DMD, DNAJB6, DNAJC19, DNM2, DOLK, DPM1, DPM2, DPM3, DSC2, DSG2, DSP, DTNA, DYSF, EFEMP2, ELAC2, ELN, EMD, ENG, EYA4, FBN1, FBN2, FHL1, FHL2, FKBP14, FKRP, FKTN, FLNA, FLNC, FOXE3, FOXH1, GAA, GATA4, GATA6, GATAD1, GDF1, GDF2, GJA1, GJA5, GLA, GMPPB, GNE, GPC3, GPD1L, GYS1, HAMP, HAND1, HCN4, HFE, HJV, HNRNPDL, HRAS, ILK, ISPD, ITGA7, JAG1, JPH2, JUP, KBTBD13, KCNA1, KCNA5, KCND3, KCNE1, KCNE2, KCNE3, KCNE5, KCNH2, KCNJ2, KCNJ5, KCNJ8, KCNK3, KCNQ1, KCNQ2, KCNQ3, KCNT1, KLHL40, KLHL41, KRAS, LAMA2, LAMA4, LAMP2, LARGE1, LDB3, LDLR, LDLRAP1, LEFTY2, LIMS2, LMNA, LMOD3, LRRC10, MAP2K1, MAP2K2, MAT2A, MATR3, MED12, MED13L, MEGF10, MEIS2, MTM1, MTO1, MYBPC3, MYF6, MYH11, MYH6, MYH7, MYL2, MYL3, MYL4, MYLK, MYLK2, MYOM1, MYOT, MYOZ2, MYPN, NEB, NEBL, NEXN, NF1, NKX2-5, NKX2-6, NODAL, NOTCH1, NPPA, NR2F2, NRAS, NSD1, P3H1, PCDH19, PCSK9, PDLIM3, PKP2, PLEC, PLEKHM2, PLN, PLOD1, PNPLA2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PRDM16, PRKAG2, PRKG1, PRRT2, PTPN11, RAF1, RANGRF, RASA1, RBM20, RIT1, RRAS, RXYLT1, RYR1, RYR2, SCN10A, SCN1A, SCN1B, SCN2B, SCN3B, SCN4B, SCN5A, SCN8A, SCN9A, SDHA, SELENON, SEMA3E, SGCA, SGCB, SGCD, SGCG, SHOC2, SKI, SLC22A5, SLC2A1, SLC2A10, SLC39A13, SLC40A1, SLMAP, SMAD3, SMAD4, SMAD6, SMAD9, SNTA1, SOS1, SOS2, SPRED1, SQSTM1, STAC3, STIM1, SUN1, SUN2, SYNE1, SYNE2, TAZ, TBX1, TBX5, TCAP, TFR2, TGFB2, TGFB3, TGFB1, TGFB2, TIA1, TMEM43, TMEM70, TMPO, TNNC1, TNNI3, TNNT1, TNNT2, TNPO3, TOR1AIP1, TPM1, TPM2, TPM3, TRAPPC11, TRDN, TRIM32, TRPM4, TTN, TTR, TXNRD2, VCL, VCP, ZFPM2, ZIC3, ABCC8, APPL1, BLK, EIF2AK3, FOXP3, GCK, GLIS3, HNF1A, HNF1B, HNF4A, IER3IP1, INS, KCNJ11, KLF11, MNX1, NEUROD1, NEUROG3, NKX2-2, PAX4, PDX1, PPARG, PTF1A, RFX6, SLC19A2, WFS1, ZFP57, AIRE, CASR, CYP24A1, FAM111A, GATA3, GCM2, GNA11, GNAS, HADHA, HADHB, PDE4D, PTH, PTH1R, SLC34A1, STX16, TBCE, ADCY3, AFF4, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BDNF, C8ORF37, CEP164, CEP19, CEP290, CPE, CREBBP, CUL4B, DYRK1B, EP300, FBN3, GPR101, IFT172, IFT27, IFT74, KDM6A, KIDINS220, KIF7, KMT2D, KSR2, LEP, LEPR, LZTFL1, MAGEL2, MC3R, MC4R, MEGF8, MKKS, MKS1, MRAP2, NLGN2, NPY, NROB2, NTRK2, PCSK1, PHF6, POMC, PRMT7, RAB23, RAI1, RPS6KA3, SCLT1, SDCCAG8, SETD2, SH2B1, SIM1, TRAPPC3, TRAPPC9, TTC8, UCP3, VPS13B, WDPCP, ABCA1, ANGPTL3, APOA1, APOA5, APOC2, CETP, CREB3L3, CYP27A1, GPD1, GPIHBP1, LCAT, LIPA, LIPG, LMF1, LPL, LRP6, MTPP, SAR1B, APOA4, APOC3, CYP7A1, GALNT2, GCKR, LIPC, LIPI, MYLIP, PLTP, SCARB1, ZHX3, AAAS, ABCD1, ACAD9, ACADM, ACADS, ACADSB, ACAT1, ACSF3, ADK, AKT2, ALDOA, ALDOB, ALG3, ALG6, BCKDHA, BCKDHB, CA5A, CACNA1D, CDKN1C, COG7, CPT1A, CYP7B1, DBH, DBT, DDC, DGUOK, DLD, DMXL2, ENO3, ETFA, ETFB, ETFDH, FAH, FBP1, FLAD1, G6PC, GALE, GALK1, GALT, GBE1, GH1, GHR, GLUD1, GYG1, GYS2, HADH, HESX1, HK1, HMGCL, HMGCS2, HSD3B7, INSR, LDHA, LHX3, MLYCD, MPI, MPV17, NADK2, NNT, NROB1, NR3C1, OPLAH, OTX2, OXCT1, PC, PCK1, PCK2, PFKM, PGAM2, PGM1, PHGDH, PHKA1, PHKA2, PHKB, PHKG2, PMM2, PROP1, PYGL, PYGM, RBCK1, SERAC1, SLC16A1, SLC25A20, SLC25A32, SLC2A2, SLC37A4, SLC52A1, SLC52A2, SLC52A3, SOX2, SOX3, TBX19, TRMT10A, UCP2.

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## LEUKODYSTROPHY AND LEUKOENCEPHALOPATHY PANEL (729 GENES)

AARS, AARS2, ABAT, ABCA1, ABCD1, ABCD4, ABHD5, ACADS, ACBD5, ACER3, ACO2, ACOX1, ACP5, ACSF3, ACTA2, ACTB, ACY1, ADAR, ADGRG1, ADK, ADNP, ADSL, AGA, AHDC1, AHI1, AIFM1, AIMP1, AIMP2, ALDH18A1, ALDH3A2, ALDH5A1, ALDH6A1, ALG12, ALG13, ALG2, ALG6, ALG9, AMACR, AMPD2, AMT, ANK3, AP1S2, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APC2, APOPT1, ARCN1, ARFGF2, ARHGAP31, ARHGEF9, ARNT2, ARSA, ARX, ASL, ASNS, ASPA, ASS1, ASXL1, ASXL2, ATP13A2, ATP5A1, ATP6AP2, ATP6V1A, ATP7A, ATP7B, ATP8A2, ATPAF2, ATRN, ATRX, AUH, B3GALNT2, B4GALNT1, BCAP31, BCAT2, BCKDHA, BCKDHB, BCL11B, BCS1L, BICD2, BMP4, BOLA3, BPTF, BRAT1, BTD, C12ORF57, C12ORF65, C19ORF12, C2CD3, CACNA1A, CACNA1E, CARS2, CBS, CC2D2A, CCDC88A, CDC42, CDKL5, CEP290, CHMP1A, CLCN2, CLCN4, CLCN7, CLN2 (TPP1), CLN3, CLN5, CLN6, CLN8, CLP1, CLPP, CNNM2, CNOT1, CNTNAP1, CNTNAP2, COA7, COASY, COG7, COL18A1, COL3A1, COL4A1, COL4A2, COLGALT1, COQ2, COQ6, COQ7, COQ8A, COQ9, COX10, COX14, COX15, COX20, COX6B1, COX7B, COX8A, CPLANE1, CPLX1, CPS1, CRAT, CREBBP, CRIPT, CRLF1, CSF1R, CSPP1, CTBP1, CTC1, CTDTP1, CTNS, CTSB, CTSF, CUL4B, CYB5R3, CYFIP2, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS, DARS2, DBT, DCAF17, DCX, DDC, DDHD1, DDHD2, DDOST, DDX3X, DEAF1, DEGS1, DGUOK, DHCR24, DHFR, DHX37, DLD, DLL4, DMXL2, DNM1L, DNM2, DOCK6, DOCK7, DOLK, DONSON, DPAGT1, DPM1, DPM2, DPM3, DPYS, DYRK1A, EARS2, EDNRB, EHMT1, EIF2AK1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL1, ELOVL4, ENTPD1, EOGT, EPG5, EPRS, ERCC1, ERCC2, ERCC3, ERCC6, ERCC8, ETFA, ETFB, ETFDH, ETHE1, EXOSC2, EXOSC3, EXOSC8, EXOSC9, FA2H, FAM126A, FAR1, FARS2, FARSB, FASTKD2, FBXL4, FDX2, FGFR1, FH, FIG4, FKR, FKTN, FLVCR2, FOLR1, FOXC1, FOXG1, FOXRED1, FUCA1, FUK, GAA, GABBR2, GABRA1, GABRB1, GABRB2, GABRB3, GALT, GAN, GATAD2B, GCDH, GDAP1, GFAP, GFM1, GFM2, GJA1, GJB1, GJC2, GLA, GLB1, GLDC, GLRX5, GLUL, GLYCTK, GM2A, GMNN, GNAO1, GNS, GOT2, GPHN, GRIN1, GRM7, GTF2H5, GTPBP2, HACE1, HCN1, HEPACAM, HERC1, HEXA, HEXB, HIBCH, HIKESHI, HK1, HLCS, HMGCL, HNRNPU, HSD17B10, HSD17B4, HSPD1, IARS, IARS2, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, INPP5E, IREB2, ISCA1, ISCA2, ITPA, ITPR1, IVD, JAM3, KARS, KAT6B, KATNB1, KCNMA1, KCNT1, KCNJ10, KCNJ2, KCTD7, KDM1A, KIAA0556, KIAA0586, KIAA0753, KIDINS220, KIF1A, KIF1C, KLHL7, KMT2E, L2HGDH, LAGE3, LAMA1, LAMA2, LAMB1, LARGE1, LARS2, LETM1, LIAS, LIPT1, LIPT2, LONP1, LRPPRC, LYRM7, MAG, MAGEL2, MAN2B1, MANBA, MARS2, MAST1, MAT1A, MCCC1, MCCC2, MCOLN1, MDH2, MECP2, MED17, MED25, MEF2C, MFSD2A, MFSD8, MGP, MICU1, MKS1, MLC1, MLYCD, MMACHC, MMADHC, MOCS1, MOCS2, MOGS, MORC2, MPLKIP, MPV17, MPZ, MRPL12, MRPL44, MRPS16, MRPS22, MRPS34, MSTO1, MTFMT, MTHFR, MTHFS, MTO1, MTOR, MTR, MTRR, MUT, NACC1, NADK2, NAGA, NAGLU, NAGS, NANS, NARS2, NAXD, NAXE, NDRG1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA4, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB10, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEK1, NFE2L2, NFU1, NGLY1, NKX6-2, NOTCH1, NPC1, NPC2, NPHP1, NRXN1, NSUN3, NT5C2, NTRK2, NUBPL, NUP62, OAT, OCRL, OPA1, OPA3, OSGEP, OTC, PACS1, PAFAH1B1, PAH, PANK2, PARS2, PAX1, PC, PCCA, PCCB, PCDH12, PCGF2, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGK1, PHGDH, PHYH, PIGA, PIGB, PIGG, PIGL, PIGM, PIGN, PIGP, PIGQ, PIGT, PIGU, PIGV, PIK3C2A, PIK3R2, PLA2G6, PLAA, PLEKHG2, PLP1, PMM2, PMP22, PNKP, PNPT1, POLG, POLG2, POLR1A, POLR1C, POLR3A, POLR3B, POMGNT1, POMK, POMT1, POMT2, PPP1R15B, PPP2R1A, PPP3CA, PPT1, PRF1, PRKDC, PRODH, PROSC, PRPS1, PRUNE1, PSAP, PSAT1, PSPH, PTEN, PTPN23, PURA, PUS3, PYCR2, QARS, QRSL1, RAB11B, RAB3GAP1, RAB3GAP2, RARS, RARS2, RBPJ, REPS1, RERE, RHOBTB2, RIN2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RPGRIP1L, RPIA, RPS6KC1, RRM2B, RTTN, RXYLT1, SAMD9L, SAMHD1, SCN3A, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SEPSECS, SERAC1, SETBP1, SGSH, SH3TC2, SHOC2, SHPK, SLC12A2, SLC12A5, SLC12A6, SLC13A3, SLC13A5, SLC16A2, SLC17A5, SLC19A3, SLC1A2, SLC1A3, SLC1A4, SLC25A1, SLC25A12, SLC25A15, SLC25A19, SLC25A22, SLC25A4, SLC25A42, SLC25A46, SLC2A1, SLC30A10, SLC33A1, SLC35A2, SLC39A14, SLC39A8, SLC46A1, SLC6A19, SLC6A8, SLC6A9, SLC9A1, SLC9A6, SMC1A, SNAP29, SNIP1, SNORD118, SNRPB, SNX14, SOD1, SON, SOX10, SOX2, SPART, SPAST, SPATA5, SPG11, SPG7, SPTAN1, SQSTM1, SRD5A3, SSR4, ST3GAL5, STAG2, STAMBIP, STAT1, STAT2, STN1, STRADA, STX11, STXB1, STXP2, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYNE1, SYNJ1, TACO1, TAF2, TANGO2, TARS2, TBC1D24, TBCD, TBCE, TBCK, TBX1, TCF4, TCTN2, TIMM50, TIMMDC1, TM4SF20, TMEM106B, TMEM126B, TMEM165, TMEM216, TMEM67, TMEM70, TMTC3, TOE1, TP53RK, TPI1, TPK1, TRAPPC11, TRAPPC9, TREX1, TRMT10A, TRMT5, TSC1, TSC2, TSEN54, TTC19, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUFM, TWNK, TXN2, TYMP, UBA5, UBE2A, UBE3A, UFM1, UNC13D, UPB1, USP7, VARS2, VPS11, VPS13D, VPS33A, WARS2, WDR45, WDR73, WHSC1, WWOX, YARS, YME1L1, ZEB2, ZFYVE26, ZIC1, ZNF335, ALDH7A1, APP, CHMP2B, CP, CTSF, DNAJC5, DNMT1, FTL, GBE1, GRN, HTRA1, KIAA1161, KIF5A, LMNB1, MAPT, NOTCH3, PDGFRB, PDGFRB, PDYN, PHAX, PINK1, PRNP, PSEN1, RNF216, SCP2, SLC20A2, TREM2, TYROBP, VCP, VPS13A, XK, XPR1.

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## LIMB AND DIGITAL MALFORMATIONS PANEL (177 GENES)

ACVR1, ADAMTS10, ADAMTS17, AFF4, AHI1, ANKRD11, ARHGAP31, ARID1A, ARID1B, ARL13B, ARL6, B3GLCT, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BHLHA9, BMP2, BMP4, BMPR1B, BTRC, C2CD3, CACNA1C, CC2D2A, CCNQ, CDH3, CEP104, CEP120, CEP290, CEP41, CHSY1, CHUK, CKAP2L, CPLANE1, CREBBP, CSPP1, DDX59, DHCR7, DHODH, DLL4, DLX5, DLX6, DOCK6, DPF2, DVL1, DVL3, DYNC1I1, EOGT, EP300, ESCO2, EVC, EVC2, FAT1, FBLN1, FBN1, FBXW4, FGF10, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FLNA, FMN1, FRAS1, FREM2, FZD2, GDF5, GDF6, GJA1, GLI2, GLI3, GNAS, GSC, HDAC4, HDAC8, HOXA13, HOXD13, IFT57, IHH, INPP5E, KDM6A, KIAA0586, KIF7, KMT2A, KMT2D, LMBR1, LRP4, LTBP2, LTBP3, MAP3K20, MEGF8, MGP, MKKS, MKS1, MRE11, MYCN, NECTIN1, NECTIN4, NIPBL, NOG, NOTCH1, NPHP1, NPHP3, NSDHL, NXN, OFD1, PDE3A, PDE4D, PDE6D, PGM3, PHF6, PIGV, PITX1, POLR1A, PORCN, PRKAR1A, PRMT7, PTDSS1, PTHLH, RAB23, RAD21, RBM8A, RBPJ, RECQL4, ROR2, RPGRIP1L, SALL1, SALL4, SC5D, SDCCAG8, SF3B4, SMAD4, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SMOC1, SOST, SOX11, SOX9, TBX15, TBX3, TBX5, TCTN1, TCTN2, TCTN3, TGDS, THPO, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TP63, TRIM32, TRPS1, TRPV4, TTC21B, TTC8, VAC14, WDPCP, WNT10B, WNT3, WNT5A, WNT7A, ZNF423, ZSWIM6.

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## HEREDITARY CANCER PANEL (526 GENES)

ABL1, ABL2, ACD, ACVR1, ACVR1B, AFDN, AIP, AKT1, AKT2, AKT3, ALK, AMER1, ANKRD26, ANKS1A, AP2S1, APC, APEX1, APEX2, AR, ARAF, ARFRP1, ARHGEF12, ARID1A, ARI1B, ARID2, ASXL1, ATM, ATR, ATRX, AURKA, AURKB, AXIN1, AXIN2, AXL, BAP1, BARD1, BCAR1, BCL2, BCL2L1, BCL6, BCORL1, BCR, BLM, BMPR1A, BRAF, BRCA1, BRCA2, BRD4, BRI1, BTK, BUB1, BUB1B, BUB3, CAPN9, CARD11, CASP10, CASP8, CASR, CBF1, CBL, CBLB, CCND1, CCND2, CCND3, CD14, CD274, CD70, CD79A, CDC42, CDC73, CDH1, CDK12, CDK4, CDK6, CDK8, CDKN1A, CDKN1B, CDKN1C, CDKN2A, CDKN2B, CDKN2C, CEBPA, CEP57, CFTR, CHD2, CHD4, CHEK1, CHEK2, CLEC12B, CREBBP, CRLF2, CSF1R, CTCF, CTNNA1, CTNNA2, CTNNB1, CTRC, CUL3, CYLD, DAXX, DCLRE1C, DDB1, DDB2, DDR2, DDX10, DDX41, DICER1, DIS3L2, DKC1, DLST, DMC1, DNAH7, DNAJC21, DNMT3A, DOT1L, DROSHA, DSC2, DSG2, EDNRB, EFL1, EGFR, EGLN1, EIF3A, ELAC2, ELANE, ELP1, EME1, EME2, EMSY, ENG, EP300, EPCAM, EPHA3, EPHB1, ERBB2, ERBB3, ERBB4, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC6L2, ERCC8, ERG, ERFF1, ESR1, ESR2, ETV1, ETV4, ETV6, EXO1, EXT1, EXT2, EZH2, FAAP24, FABP2, FAM111B, FAN1, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FAT1, FBN1, FBXO24, FBXW7, FGF10, FGF14, FGF23, FGF3, FGF4, FGFR1, FGFR2, FGFR3, FGFR4, FH, FLCN, FLT1, FLT3, FLT4, G6PC3, GABRG1, GALNT14, GATA1, GATA2, GATA3, GATA4, GATA6, GF11, GLI1, GLI2, GLI3, GNA11, GNA13, GNAQ, GNAS, GPC3, GPR161, GPRIN1, GREM1, GRIN2A, GRM3, GSK3B, GTF2H1, GTF2H2, GTF2H3, GTF2H4, GTF2H5, HAVCR2, HAX1, HERPUD1, HGF, HMMR, HNF1A, HOXB13, HRAS, HSP90AA1, IDH1, IDH2, IGF1R, IGF2, IKBKE, IKZF1, IL1RN, IL7R, INHBA, INSR, IRF1, IRF2, IRF4, IRS2, JAK1, JAK2, JAK3, JUN, KAT6B, KDM6A, KDR, KEAP1, KEL, KIF1B, KIT, KITLG, KLF6, KLHL6, KLLN, KMT2A, KMT2C, KMT2D, KRAS, KRAS2, KRT76, LAMB4, LCP1, LIG4, LMO1, LRP5, LYN, LZTR1, MAGI2, MAP2K1, MAP2K2, MAP2K4, MAP3K1, MAP3K6, MAP3K9, MAX, MBD4, MC1R, MCL1, MCPH1, MDH2, MDM2, MDM4, MECOM, MED12, MEN1, MET, MGMT, MIR125A, MIR191, MITF, MLH1, MLH3, MLLT6, MMS19, MNAT1, MPG, MRE11, MSH2, MSH3, MSH4, MSH5, MSH6, MST1R, MTOR, MUS81, MUTYH, MX2, MYB, MYC, MYCL, MYCN, MYD88, NANP, NBAS, NBN, NEIL1, NEIL2, NEIL3, NEK10, NEK11, NEK2, NEK4, NF1, NF2, NFE2L2, NFKBIA, NHEJ1, NKX2-1, NLRP1, NME1, NOTCH1, NOTCH2, NOTCH3, NPM1, NQO2, NRAS, NSD1, NSUN2, NTHL1, NTRK1, NTRK2, NTRK3, NUMA1, NUP93, OCA2, OGG1, PAK3, PALB2, PALLD, PARP1, PARP2, PARP3, PAX5, PBRM1, PDE11A, PDGFRA, PDGFRB, PGR, PHB, PHOX2B, PIK3C2B, PIK3CA, PIK3CB, PIK3CG, PIK3R1, PIK3R2, PINK1, PKD1, PLA2G6, PLCE1, PLCG2, PLK2, PMS1, PMS2, PNKP, POLD1, POLE, POLH, POLK, POT1, PPM1D, PPP2R1A, PRDM1, PREX2, PRF1, PRKAR1A, PRKDC, PRKN, PROSER2, PRSS1, PRSS8, PTCH1, PTEN, PTPN11, PTPRD, PZP, RABL3, RAC1, RAD17, RAD50, RAD51, RAD51B, RAD51C, RAD51D, RAD52, RAD54B, RAD54L, RAF1, RANBP2, RARA, RASA2, RASEF, RB1, RB1CC1, RBBP8, RECQL, RECQL4, REST, RET, RFC1, RHBDF2, RICTOR, RINT1, RIPK1, RIT1, RNASL, RNF43, ROS1, RPA1, RPTOR, RRAS, RTEL1, RUNDC1, RUNX1, RUNX1T1, SAMD9, SAMD9L, SBDS, SDHA, SDHAF2, SDHB, SDHC, SDHD, SETD2, SF3B1, SHH, SHOC2, SLC22A18, SLC22A1L, SLC25A11, SLC34A2, SLC45A1, SLC45A2, SLIT2, SLX4, SMAD2, SMAD3, SMAD4, SMARCA4, SMARCB1, SMARCE1, SMO, SMUG1, SNCAIP, SOCS1, SON, SOS1, SOS2, SOX10, SOX2, SOX9, SPAG9, SPEN, SPINK1, SPOP, SPRED1, SPTA1, SRC, SRP72, STAG2, STAT3, STAT4, STK11, SUFU, SYK, TAF1, TBX3, TERC, TERF1, TERF2, TERF2IP, TERT, TET2, TGFB2, TGM6, TINF2, TMC6, TMEM127, TMPRSS2, TNFAIP3, TNFRSF14, TOP1, TOP2A, TP53, TSC1, TSC2, TSHR, U2AF1, UNG, UPF1, VEGFA, VHL, WRN, WRNIP1, WT1, XAB2, XPA, XPC, XPO1, XRCC1, XRCC2, XRCC3, XRCC4, XRCC5, XRCC6.

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## NEURODEVELOPMENTAL DISORDERS PANEL (241 GENES)

ACTB, ACTG1, ADNP, ADSL, AGA, AHDC1, ALDH5A1, ALDH7A1, AMER1, ANKRD11, AP1S2, ARG1, ARID1A, ARID1B, ARSA, ARX, ASNS, ASXL1, ATP1A3, ATP7A, ATRX, AUTS2, BCAP31, BRAF, BRAT1, BRD4, BRWD3, CACNA1A, CACNA1E, CAMK2B, CASK, CBL, CC2D2A, CDK13, CDKL5, CHD2, CHD7, CHD8, CLCN4, CLN2, CLN3, CLN5, CLN6, CLTC, CNTNAP2, COL4A1, CREBBP, CTNNB1, CUL3, DDC, DDX3X, DEAF1, DHCR7, DNMT1, DNMT3A, DOCK6, DPF2, DYNC1H1, DYRK1A, EEF1A2, EFTUD2, EHMT1, EP300, EZH2, FGD1, FOLR1, FOXP1, FOXP2, GABBR2, GABBR3, GABRG2, GALC, GAMT, GATAD2B, GATM, GLB1, GM2A, GNAO1, GNAS, GNS, GPC3, GRIA3, GRIN1, GRIN2A, GRIN2B, HDAC8, HEXA, HEXB, HGSNAT, HIVEP2, HNRNPB, HNRNPU, HRAS, HUWE1, IDS, IDUA, IGF1R, IL1RAPL1, IQSEC2, ITPR1, KANSL1, KAT6A, KAT6B, KCNA2, KCNB1, KCNH1, KCNQ2, KCNT1, KDM5C, KDM6A, KIF1A, KMT2A, KMT2B, KMT2D, KMT2E, KRAS, L1CAM, LZTR1, MAGEL2, MAN1B1, MAP2K1, MAP2K2, MBD5, MECP2, MED12, MED13L, MEF2C, MFSD8, MID1, MTOR, NAA10, NAA15, NAGLU, NALCN, NEXMIF, NF1, NFIA, NFIX, NGLY1, NHS, NIPBL, NONO, NPC1, NR2F1, NRAS, NRXN1, NSD1, NSUN2, OCRL, OPHN1, OTC, PACS1, PACS2, PAH, PCBD1, PCDH19, PDHA1, PGAP3, PHF21A, PHF6, PHIP, PLA2G6, PMM2, POLG, PPM1D, PPP1CB, PPP2R1A, PPP2R5D, PPP3CA, PPT1, PQBP1, PTEN, PTPN11, PTS, PURA, QDPR, RAD21, RAF1, RAI1, RBM10, RIT1, RPS6KA3, SATB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SETBP1, SETD5, SGSH, SHOC2, SIN3A, SLC13A5, SLC16A2, SLC2A1, SLC6A1, SLC6A8, SLC9A6, SMARCA2, SMARCA4, SMARCB1, SMARCE1, SMC1A, SMC3, SON, SOS1, SOS2, SOX11, SPAST, SPATA5, SPTAN1, STAG1, STXBP1, SURF1, SYNGAP1, TAF1, TBCK, TBL1XR1, TCF20, TCF4, TELO2, TRAPPC9, TRRAP, TSC1, TSC2, TUBA1A, UBE3A, UNC80, USP9X, VPS13B, WDR45, WWOX, ZBTB18, ZBTB20, ZC4H2, ZDHHC9, ZEB2, ZIC2, ZMIZ1, ZMYND11.

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## **SKELETAL AND CONNECTIVE TISSUE DISORDERS PANEL (416 GENES)**

ABCC6, ABL1, ACAN, ACP5, ACTA2, ACVR1, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL4, AEBP1, AFF4, AGA, AGPS, AIFM1, ALDH18A1, ALPL, AMER1, ANKH, ANO5, ARCN1, ARIH1, ARSB, ARSE, ASCC1, ASPM, ATP6VOA2, ATP6V1A, ATP6V1E1, ATP7A, ATR, ATRIP, B3GALT6, B3GAT3, B4GALT7, BGN, BMP1, BMP2, BMPER, BMPR1B, C1S, C2CD3, CA2, CANT1, CASR, CBS, CCDC8, CDC45, CDC6, CDK5RAP2, CDKN1C, CDT1, CENPJ, CEP120, CEP135, CEP152, CEP63, CEP97, CFAP410, CHST14, CHST3, CHUK, CLCN5, CLCN7, COG1, COG7, COL10A1, COL11A1, COL11A2, COL12A1, COL1A1, COL1A2, COL27A1, COL2A1, COL3A1, COL4A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COMP, CREB3L1, CRIPT, CRTAP, CSF1R, CSGALNACT1, CSPP1, CTNS, CTSA, CTSK, CUL7, CWC27, CYP27B1, CYP2R1, DCHS1, DDR2, DDRGK1, DHCR24, DIP2C, DLL1, DLL3, DLX3, DMP1, DMRT2, DNA2, DNMT3A, DONSON, DSE, DVL1, DVL3, DYM, DYNC2H1, DYNC2LI1, EBP, EFEMP2, EIF2AK3, ELN, ENPP1, ESCO2, EVC, EVC2, EXOC6B, EXOSC2, EXT1, EXT2, EXTL3, FAH, FAM111A, FAM20C, FAM46A, FAR1, FAT4, FBLN5, FBN1, FBN2, FGF23, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FKBP14, FLCN, FLNA, FLNB, FN1, FOXE3, FTO, FUCA1, FZD2, GALNS, GALNT3, GDF5, GDF6, GGCX, GHR, GHRHR, GHSR, GJA1, GLB1, GMNN, GNAS, GNE, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC6, GPX4, GSC, GUSB, GZF1, HCN4, HES7, HGSNAT, HPGD, HSPG2, HYAL1, IARS2, ICK, IDS, IDUA, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT57, IFT74, IFT80, IFT81, IGF1, IGF2, IHH, IMPAD1, INPPL1, INTU, JAG1, KAT6B, KIAA0586, KIAA0753, KIF22, KL, KMT2A, LARP7, LBR, LEMD3, LFNG, LIFR, LIG4, LMNA, LMX1B, LONP1, LOX, LOXL3, LRP4, LRP5, LRRK1, LTBP2, LTBP3, LTBP4, LZTS1, MAFB, MAN2B1, MANBA, MAP3K7, MAT2A, MATN3, MBTPS1, MBTPS2, MCM5, MCPH1, MED12, MEOX1, MESDC2, MESP2, MFAP5, MGP, MMP13, MMP14, MMP2, MMP9, MNX1, MSX2, MYH11, MYH3, MYLK, MYO18B, NAGLU, NANS, NBAS, NEK1, NEU1, NIN, NKX3-2, NOG, NOTCH1, NOTCH2, NPPC, NPR2, NPR3, NSDHL, NSMCE2, NTRK1, NXN, OBSL1, OCRL, ORC1, ORC4, ORC6, OSTM1, P3H1, P4HB, PAM16, PAPSS2, PCGF2, PCNT, PCYT1A, PDE4D, PEX5, PEX7, PGM3, PHEX, PIK3C2A, PISD, PKD2, PKDCC, PLK4, PLOD1, PLOD2, PLOD3, PLS3, POC1A, POLR1A, POP1, POR, PPIB, PPP3CA, PRDM5, PRKAR1A, PRKG1, PTSS1, PTH1R, PTHLH, PTPN11, PYCR1, RAB33B, RBBP8, RECQL4, RIN2, RIPPLY2, RMRP, RNU4ATAC, ROR2, RSPO2, RSPRY1, RTTN, RUNX2, SC5D, SEC24D, SERPINF1, SERPINH1, SETBP1, SFRP4, SGMS2, SGSH, SH3PXD2B, SKI, SLC17A5, SLC26A2, SLC29A3, SLC2A10, SLC2A2, SLC34A1, SLC34A3, SLC35D1, SLC39A13, SLCO2A1, SLC05A1, SMAD2, SMAD3, SMAD4, SMAD6, SMARCAL1, SNRPB, SNX10, SOX9, SP7, SPARC, SQSTM1, SRCAP, SUCO, SULF1, SUMF1, TAB2, TALDO1, TAPT1, TBCE, TBX15, TBX3, TBX5, TBX6, TBXAS1, TCIRG1, TCTEX1D2, TCTN3, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TMEM165, TMEM38B, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TRAF3IP1, TRAI, TRAPPC2, TREM2, TRIM37, TRIP11, TRIP4, TRMT10A, TRPS1, TRPV4, TTC21B, TUBGCP4, TUBGCP6, TYROBP, UBE3B, UPF3B, VAC14, VDR, VPS33A, WDR19, WDR34, WDR35, WDR4, WDR60, WISP3, WNT1, WNT3, WNT3A, WNT5A, XRCC4, XYLT1, XYLT2, ZMPSTE24, ZNF469, ZNF687.

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## IMMUNODEFICIENCY PANEL (429 GENES)

ACD, ACP5, ACTB, ADA, ADA2, ADAM17, ADAR, ADGRE2, AICDA, AIRE, AK2, ALG6, ALPK1, ANGPT1, ANKZF1, AP3B1, AP3D1, ARHGEF1, ARPC1B, ASAH1, ATM, ATP6AP1, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BLOC1S3, BLOC1S6, BTK, C17ORF62, C1QA, C1QB, C1QC, C1S, C2, C3, C5, C6, C7, C8A, C8B, C9, CARD11, CARD14, CARD8, CARD9, CARMIL2, CASP10, CASP8, CBL, CCBE1, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD55, CD59, CD79A, CD79B, CD81, CD8A, CDC42, CDCA7, CEBPE, CFB, CFD, CFH, CFI, CFP, CHD7, CIB1, CIITA, CLCN7, CLPB, COL7A1, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR2, CXCR4, CYBA, CYBB, CYP27A1, DBR1, DCLRE1C, DDX58, DEF6, DGAT1, DIAPH1, DKC1, DNAJC21, DNASE1L3, DNASE2, DNMT3B, DOCK2, DOCK8, DSG1, DTNBP1, DUOX2, EFL1, EIF2AK3, ELANE, EPG5, ERBIN, ERCC2, ERCC3, ERCC6L2, EXTL3, FADD, FANCA, FANCB, FANCE, FANCF, FANCI, FANCL, FAS, FASLG, FAT4, FCHO1, FERMT1, FERMT3, FNIP1, FOXI3, FOXN1, FOXP3, FPR1, G6PC, G6PC3, G6PD, GATA1, GATA2, GFI1, GINS1, GTF2E2, GTF2H5, GUCY2C, HAX1, HELLS, HMOX1, HPS1, HPS3, HPS4, HPS5, HPS6, HTRA2, HYOU1, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNGR1, IFNGR2, IGLL1, IKBKB, IKZF1, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL12RB2, IL17F, IL17RA, IL17RC, IL18BP, IL1RN, IL21, IL21R, IL23R, IL2RA, IL2RB, IL2RG, IL36RN, IL6R, IL6ST, IL7R, IRAK4, IRF2BP2, IRF4, IRF7, IRF8, IRF9, ISG15, ITCH, ITGAM, ITGB2, ITK, JAGN1, JAK1, JAK3, KAT6A, KDM6A, KMT2A, KMT2D, LAMTOR2, LAT, LCK, LCT, LIG1, LIG4, LIPA, LPIN2, LRBA, LRRC8A, LYN, LYST, MAD2L2, MAGT1, MALT1, MAP3K14, MCM4, MEFV, MKL1, MOGS, MPLKIP, MS4A1, MSN, MTHFD1, MVK, MYD88, MYO5B, MYSM1, NBAS, NBN, NCF2, NCF4, NCKAP1L, NCSTN, NEUROG3, NFAT5, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NOP10, NSMCE3, OAS1, ORAI1, OSTM1, OTULIN, PARN, PAX1, PEPD, PGM3, PIK3CD, PIK3R1, PLCG2, PLVAP, PMM2, PNLIP, PNP, POLA1, POLD1, POLD2, POLE, POLE2, POLR3A, POLR3F, POMP, PRF1, PRKCD, PRKDC, PSENEN, PSMA3, PSMB4, PSMB8, PSMG2, PSTPIP1, PTPRC, RAB27A, RAC2, RAG1, RAG2, RANBP2, RASGRP1, RBCK1, REL, RELA, RELB, RFWD3, RFX5, RFXANK, RFXAP, RHOH, RIPK1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF113A, RNF168, RNF31, RNU4ATAC, RORC, RPSA, RTEL1, SAMD9, SAMD9L, SAMHD1, SAR1B, SCO2, SEC61A1, SEMA3E, SERPING1, SGPL1, SH2D1A, SH3BP2, SH3KBP1, SI, SIAE, SKIV2L, SLC10A2, SLC26A3, SLC29A3, SLC35C1, SLC37A4, SLC39A7, SLC46A1, SLC51B, SLC5A1, SLC7A7, SLC9A3, SLX4, SMARCAL1, SMARCD2, SNX10, SP110, SPINK5, SPINT2, SPPL2A, SRP54, SRP72, STAT1, STAT2, STAT3, STAT4, STAT5B, STIM1, STK4, STN1, STX11, STX3, STXBP2, TAOK2, TAP1, TAP2, TAPBP, TAZ, TBX1, TCF3, TCIRG1, TCN2, TERC, TERT, TFRC, TGFB1, TGFB1R1, TGFB2, THBD, TICAM1, TIMM50, TINF2, TLR3, TLR7, TMC6, TMC8, TMEM173, TMPRSS15, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TNFRSF6B, TNFRSF9, TNFSF11, TNFSF12, TONSL, TOP2B, TP63, TPP2, TRAF3, TRAF3IP2, TREX1, TRNT1, TTC37, TTC7A, TYK2, UNC13D, UNC45A, UNC93B1, UNG, USB1, VAV1, VPS13B, VPS45, WAS, WDR1, WIPF1, WNT2B, WRAP53, XIAP, ZAP70, ZBTB24, ZCCHC8, ZNF341.

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## INFLAMMATORY BOWEL DISEASE (IBD) PANEL (125 GENES)

ADA, ADAM17, AICDA, AIRE, ANKZF1, ARPC1B, BACH2, BTK, C17ORF62, CARD8, CARMIL2, CASP8, CD19, CD3G, CD40, CD40LG, CD55, CD81, CHD7, CIITA, COL7A1, CR2, CTLA4, CYBA, CYBB, CYBC1, DCLRE1C, DKC1, DOCK8, DUOX2, FCHO1, FERMT1, FOXP3, FUT2, G6PC3, GUCY2C, HPS1, HPS4, HPS6, ICOS, IKBKB, IKZF1, IL10, IL10RA, IL10RB, IL21, IL21R, IL23R, IL2RA, IL2RB, IL2RG, IL7R, ITCH, ITGB2, JAK1, LCK, LIG4, LRBA, LYST, MALT1, MEFV, MVK, MYO5A, NCF2, NCF4, NFAT5, NFKB1, NFKB2, NLRC4, NLRP12, NOD2, NOP10, NPC1, PIK3CD, PIK3R1, PLCG2, POLA1, PRF1, PTEN, RAB27A, RAC1, RAC2, RAG1, RAG2, RET, RFX5, RFXANK, RFXAP, RIPK1, RTEL1, SH2D1A, SI, SKIV2L, SLC37A4, SLC9A3, SLCO2A1, STAT1, STAT3, STAT5B, STIM1, STX3, STXBP2, STXBP3, TAP1, TAP2, TERC, TERT, TGFB1, TGFB1R1, TGFB2, TNF2, TNFAIP3, TNFRSF13B, TRAF3, TRIM22, TTC37, TTC7A, UNC13D, UNG, WAS, WIPF1, XIAP, ZAP70, ZBTB24, ZNF341.

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## CARRIER SCREENING PANEL (302 GENES)

CFTR, SMN1, ABCB11, ABCC8, ABCD1, ACAD9, ACADM, ACADVL, ACAT1, ACOX1, ACSF3, ADA, ADAMTS2, ADGRG1, AGA, AGL, AGPS, AGXT, AIRE, ALDH3A2, ALDOB, ALG6, ALMS1, ALPL, AMT, AQP2, ARG1, ARSA, ARSB, ASL, ASNS, ASPA, ASS1, ATM, ATP6V1B1, ATP7A, ATP7B, ATRX, BBS1, BBS10, BBS12, BBS2, BCKDHA, BCKDHB, BCS1L, BLM, BSND, CAPN3, CBS, CDH23, CEP290, CERKL, CHM, CHRNE, CIITA, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGB3, COL27A1, COL4A3, COL4A4, COL4A5, COL7A1, CPS1, CPT1A, CPT2, CRB1, CTNS, CTSK, CYBA, CYBB, CYP11B1, CYP11B2, CYP17A1, CYP19A1, CYP21A2, CYP27A1, DBT, DCLRE1C, DHCR7, DHDDS, DLD, DMD, DNAH5, DNAI1, DNAI2, DYSF, EDA, EIF2B5, ELP1, EMD, ERCC6, ERCC8, ESCO2, ETFA, ETFDH, ETHE1, EVC, EVC2, EYS, F9, FAH, FAM161A, FANCA, FANCC, FANCG, FH, FKRP, FKTN, FMR1, G6PC, GAA, GALT, GALK1, GALT, GAMT, GBA, GBE1, GCDH, GFM1, GJB1, GJB2, GLA, GLB1, GLDC, GLE1, GNE, GNPTAB, GNPTG, GNS, GRHPR, HADHA, HAX1, HBA1, HBA2, HBB, HEXA, HEXB, HGSNAT, HJV, HLCS, HMGCL, HOGA1, HPS1, HPS3, HSD17B4, HSD3B2, HYAL1, HYLS1, IDS, IDUA, IL2RG, IVD, KCNJ11, LAMA2, LAMA3, LAMB3, LAMC2, LCA5, LDLR, LDLRAP1, LHX3, LIFR, LIPA, LOXHD1, LPL, LRPPRC, MAN2B1, MCOLN1, MED17, MESP2, MFSD8, MKS1, MLC1, MMAA, MMAB, MMACHC, MMADHC, MPI, MPL, MPV17, MTHFR, MTM1, MTRR, MTPP, MUT, MYO7A, NAGLU, NAGS, NBN, NDRG1, NDUFAF5, NDUFS6, NEB, NPC1, NPC2, NPHS1, NPHS2, NR2E3, NTRK1, OAT, OPA3, OTC, PAH, PC, PCCA, PCCB, PCDH15, PDHA1, PDHB, PEX1, PEX10, PEX12, PEX2, PEX6, PEX7, PFKM, PHGDH, PKHD1, PMM2, POMGNT1, PPT1, PROP1, PRPS1, PSAP, PTS, PUS1, PYGM, RAB23, RAG2, RAPSN, RARS2, RDH12, RMRP, RPE65, RPGRIP1L, RS1, RTEL1, SACS, SAMHD1, SEPSECS, SGCA, SGCB, SGCG, SGSH, SLC12A3, SLC12A6, SLC17A5, SLC22A5, SLC25A13, SLC25A15, SLC26A2, SLC26A4, SLC35A3, SLC37A4, SLC39A4, SLC4A11, SLC6A8, SLC7A7, SMARCAL1, SMPD1, STAR, SUMF1, TAT, TCIRG1, TECPR2, TFR2, TGM1, TH, TMEM216, TPP1, TRMU, TSFM, TTPA, TYMP, USH1C, USH2A, VPS13A, VPS13B, VPS45, VRK1, VSX2, WNT10A, XPA, XPC, ZFYVE26, BTB, F11, F2, F5, G6PD, GP1BA, GP9, HFE, HGD, MCCC1, MCCC2, MEFV, SERPINA1.

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## COMPREHENSIVE HEMATOLOGY PANEL (294 GENES)

ABCA3, ABCB7, ABCG5, ABCG8, ACTB, ACTN1, ADAMTS13, AK1, AK2, ALAS2, AMN, ANK1, ANKRD26, AP3B1, AP3D1, ARPC1B, ATM, ATR, ATRX, BLM, BLOC1S3, BLOC1S6, BRAF, BRCA1, BRCA2, BRIP1, C15ORF41, C6ORF25, CBL, CD59, CDAN1, CDC42, CDKN2A, CEBPA, CECR1, CLCN7, CLPB, CSF2RA, CSF3R, CTC1, CTLA4, CTSC, CUBN, CXCR4, CYB5R3, CYCS, DDX41, DHFR, DKC1, DNAJC21, DNASE2, DTNBP1, EFL1, EGLN1, ELANE, EPAS1, EPB41, EPB42, EPB42, EPCAM, EPOR, ERCC4, ERCC6L2, ETV6, F2, F5, F7, F8, F9, F10, F11, F12, F13A1, F13B, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FGA, FGB, FGG, FLI1, FLNA, FYB, G6PC3, G6PD, GATA1, GATA2, GBA, GCLC, GFI1, GFI1B, GGCX, GINS1, GLRX5, GP1BA, GP1BB, GP9, GPI, GPR143, GSS, HAVCR2, HAX1, HBA1, HBA2, HBB, HFE, HK1, HMOX1, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, IFNGR2, IKZF1, ITGA2, ITGA2B, ITGB3, ITK, JAGN1, JAK2, KCNN4, KIF23, KLF1, KRAS, LAMTOR2, LMAN1, LPIN2, LYST, MAGT1, MAP2K1, MAP2K2, MASTL, MCFD2, MECOM, MKL1, MLH1, MPL, MSH2, MSH6, MTR, MYH9, MYO5A, NAF1, NBEAL2, NBN, NF1, NHP2, NOP10, NRAS, NT5C3A, OBFC1, OCA2, P2RY12, PALB2, PARN, PAX5, PC, PDHA1, PDHX, PGK1, PGM3, PIEZO1, PKLR, PMS2, POT1, PRF1, PRKACG, PROC, PROS1, PTPN11, PUS1, RAB27A, RAD51C, RASGRP2, RBM8A, RECQL4, REN, RHAG, RIT1, RNF168, RPL5, RPL11, RPL15, RPL26, RPL27, RPL31, RPL35A, RPS7, RPS10, RPS17, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SEC23B, SERPINC1, SERPINF2, SFTPB, SFTPC, SH2D1A, SLC4A1, SLC11A2, SLC19A2, SLC25A38, SLC37A4, SLC45A2, SLC46A1, SLC4A1, SLFN14, SLX4, SMARCD2, SOS1, SPTA1, SPTB, SRC, SRP54, SRP72, STAT3, STX11, STXBP2, TBXA2R, TBXAS1, TCN2, TCIRG1, TERC, TERT, TF, THBD, THPO, TINF2, TMPRSS6, TP53, TPI1, TRNT1, TUBB1, TYR, TYRP1, UBE2T, UNC13D, USB1, VKORC1, VPS13B, VPS45, VWF, WAS, WDR1, WIPF1, WRAP53, XIAP, XRCC2, YARS2, ZCCHC8, FUT2, EPO, BPGM, NPM1, FLT3, DNMT3A, IDH1, IDH2, KIT, TET2, ASXL1, WT1, KMT2A, SRSF2, CALR, SETBP1, ZRSR2, SF3B1, EZH2, ETNK1, ABL1, VHL, U2AF1, PHF6, BCOR, CHECK2, SMC1A, SMC3.

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## NEUROLOGY COMPREHENSIVE PANEL (882 GENES)

ABAT, ABCA7, ABCB7, ABCD1, ABHD12, ABHD5, ACAD9, ACADL, ACADM, ACADVL, ACO2, ACSL4, ACTA1, ACTB, ACTG1, ADAR, ADCY5, ADGRG1, ADNP, ADSL, AFF2, AFG3L2, AGA, AGL, AGRN, AHCY, AHI1, AIFM1, AIMP1, AKT1, AKT3, ALAD, ALAS2, ALDH3A2, ALDH5A1, ALDH7A1, ALDOA, ALG13, ALS2, AMACR, AMPD1, AMPD2, AMT, ANG, ANO10, ANO3, ANO5, ANTXR1, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOE, APP, APTX, ARFGF2, ARG1, ARHGEF10, ARHGEF6, ARHGEF9, ARL13B, ARL6, ARSA, ARX, ASAH1, ASNS, ASPA, ASPM, ATAD3A, ATCAY, ATL1, ATL3, ATM, ATN1, ATP13A2, ATP1A2, ATP1A3, ATP2A1, ATP2B3, ATP6AP2, ATP6VOA2, ATP7A, ATP8A2, ATR, ATRX, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, B3GALNT2, B4GALNT1, BAG3, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCL11A, BCOR, BEAN1, BICD2, BRAT1, BRWD3, BSCL2, BTBD, CA8, CACNA1A, CACNA1C, CACNA1H, CACNA1S, CACNB4, CAMTA1, CAPN1, CAPN3, CASK, CASR, CAV3, CC2D1A, CC2D2A, CCDC88C, CCM2, CCND2, CCT5, CDK5RAP2, CDKL5, CDKN1C, CDON, CENPF, CENPJ, CEP152, CEP290, CEP41, CEP63, CFL2, CHAT, 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## COMPREHENSIVE EPILEPSY PANEL (591 GENES)

MT-TQ, NALCN, COX15, ALG3, GFM1, PCDH19, CPA6, MAP2K1, OFD1, PSAP, MT-RNR1, KANSL1, MICAL1, DNMT1L, FOXG1, ACY1, CLCN2, TMEM70, GRIA4, BRD2, ADRA2B, DNAJC5, TSEN54, SCN2A, CLN3, SETD1B, PLAA, IER3IP1, PRRT2, AMACR, COG8, RAB39B, FARS2, AGA, PNKD, SNAP25, ARFGEF2, ALDH4A1, FAM126A, MT-TL1, SLC25A1, GCSH, KCNH5, NTRK2, CACNA1B, DOCK7, PITRM1, RORA, TUBA1A, SSR4, MPDU1, NDST1, MT-ND5, PACS1, PCLO, MT-TH, SLC6A8, POLR3A, ST3GAL3, MGAT2, RBFOX3, CTSD, BRAT1, MTHFR, GNE, EPM2A, NSDHL, ZEB2, DENND5A, POLR3B, PPP2R1A, GUF1, NUBPL, PURA, SLC2A1, MAPK10, MT-ND1, GATM, KCNJ10, PDHA1, GAL, ARHGEF9, SPATA5, FOLR1, SLC25A42, ATAD1, DEAF1, KIF5A, MT-TM, NR2F1, GABRB1, FA2H, GAMT, PPP2CA, ATP1A1, CPT2, KCNH2, SLC35A1, SLC1A3, MT-ATP6, MEF2C, NTNG1, PEX19, PLA2G6, TRIT1, AUH, PIGW, SLC12A5, KCTD3, PLPBP, GLS, PRICKLE1, ACTL6B, RNASEH2C, HSPD1, PEX12, RNASET2, UNC80, PIGP, QDPR, KCTD7, PEX14, CCDC88C, STRADA, PLP1, KIF2A, TWNK, GNS, SGSH, GOSR2, MLC1, TBCK, GFAP, ALDH5A1, DPM2, SCN9A, ATP6V1A, LRPPRC, MED17, SLC25A22, MT-TE, MT-CO2, SIK1, MT-TD, CHRNA4, DEPDC5, ALDH7A1, NPC1, CTSF, TCF4, NEDD4L, HACE1, NECAP1, MTOR, EFHC1, GPHN, RHOTB2, GABRA1, MBOAT7, COX6B1, ATP7A, CASR, PTEN, TPK1, TBCE, ALG8, CHD2, CACNA1H, PEX10, EHMT1, PIGQ, GABBR2, ST3GAL5, PACS2, CLN5, BCKDK, RNASEH2B, DNMT1, NRXN1, SMC1A, NACC1, MT-TK, PRICKLE2, GLDC, SMARCA2, LMNB2, SLC1A4, SYN1, AMT, PLCB1, KCNH1, PIGA, CTC1, TSFM, KCNT2, CLN6, SLC4A10, RELN, ROGDI, ASAH1, SAMHD1, FUT8, SLC13A5, SCARB2, PIGV, TBX1, ARSA, CNKSR2, SERPINI1, MT-ND6, MT-TG, KCNT1, DDX3X, MT-TW, ARHGEF15, CDKL5, CASK, KDM5C, MARS2, SNX27, PRODH, KCNA1, CLTC, TNK2, CAMK2B, NPRL2, NAGLU, CHRNA7, NPC2, TRIM8, ADGRG1, DARS2, WARS2, MT-TR, NIPBL, TREX1, UBE3A, NKX6-2, SCN4A, MT-TS1, CYFIP2, PHGDH, CACNA1E, FUCA1, WWOX, NDUFS7, TPP1, UBE2A, TRAK1, PC, IRF2BPL, KPNA7, NEXMIF, ADGRV1, PEX3, ARG1, ITPA, OCLN, DCX, FGF12, TUBB2A, AKT3, MT-TV, CNPY3, IDS, DHPS, CSF1R, DYRK1A, PHF6, PHACTR1, WDR26, ATP2A2, RANBP2, NDUFAF3, PGK1, SHANK3, GRIA3, ALG13, BOLA3, ME2, TSC2, CSNK2B, STX1B, FBXO11, CUX2, MCPH1, CNTNAP2, GCH1, ATP1A2, MT-ND2, CLN8, LGI1, KCND2, ATP6AP2, HDAC8, EEF1A2, RUSC2, PPP3CA, ADNP, GPAA1, GRIK2, HEXA, SATB2, MT-TI, GLB1, COX10, GABRD, NDE1, PIGB, AP4B1, HNRNPU, ARX, IQSEC2, IBA57, NHLRC1, EML1, ALG12, SDHAF1, FAR1, DHFR, CACNB4, KCNAB2, MFSD8, YWHAG, EIF3F, MAP2K2, DOLK, PEX1, SLC19A3, ATIC, DIAPH1, PRIMA1, COQ9, EIF2B1, CARS2, NDUFA1, PYCR2, SCN10A, PUM1, TANGO2, GALC, PSAT1, EIF2B2, CACNA2D2, SDHA, GLUD1, ABCA2, LAMC3, EARS2, ALG1, CNTN2, P4HTM, SLC35C1, CYP27A1, LAMA2, HPD, ANKRD11, RORB, MT-TF, VAMP2, HCN1, COQ8A, NBEA, CACNA1G, KCNV2, ZIC2, GRIN2D, ALG6, CC2D2A, DPYD, SCN1B, PIGS, KIF1A, GRN, MOCS2, CHRNB2, HCN4, TSC1, KCNMA1, NEUROD2, SLC35A2, GRIN1, PNPO, NPRL3, CPLX1, PPT1, MT-TP, RAI1, ZNHIT3, FLNA, TK2, CUL4B, PQBP1, GABRB2, SCN3A, HEXB, KDM6A, MED12, NDUFS6, TBC1D24, PEX5, SCN8A, EIF2B5, CACNA1D, MRPL44, CAD, PIGC, KMT2D, RMND1, ADSL, SLC6A5, GCDH, COA7, CTSA, MT-ND4, CACNA1A, PEX6, DMXL2, ASPA, MT-ND3, ECM1, MDH2, NUS1, SLC46A1, ASXL3, MBD5, ASNS, PTPN23, SLC25A15, ARV1, ETHE1, KCNA2, DEGS1, NEU1, CCDC88A, KIF5C, SLC6A1, TBCD, PDSS2, RBFOX1, ABAT, ECHS1, HSD17B10, MT-TT, NSD1, SCO2, OPHN1, SCN1A, PARS2, SLC9A6, DPM1, PCNT, SMC3, PEX26, PAFAH1B1, COQ4, LIAS, SPTAN1, NF1, PMM2, SYNJ1, MOCS1, KCNB1, ADAR, GFM2, DHDDS, PTS, CLCN4, CRH, RAB11A, VPS13A, COG7, PEX7, COL4A2, FOXRED1, TBL1XR1, MT-TA, UBTF, CHRNA2, ALDH3A2, EMX2, SRGAP2, BTBD, MIPEP, HEPACAM, RARS2, SZT2, TSEN2, D2HGDH, GNB1, SLC17A5, SUOX, EIF2B4, KCNQ2, KCNC1, KCNK4, SYP, HGSNAT, ATP1A3, SLC1A2, STXBP2, SURF1, PNKP, UBA5, COQ2, DYNC1H1, KMT2E, COL18A1, AP3B2, KCNQ3, NFU1, ZDHHC9, SLC39A8, HIBCH, HTRA1, KRAS, SYNGAP1, GRIN2A, GRIN2B, FRRS1L, HTT, TUBB2B, SMS, WASF1, NDUFA2, MT-CYB, RNF13, SGCE, NDUFS8, HDAC4, RFT1, STXBP1, POLG, HCN2, PIGG, MOGS, SETBP1, TSEN34, PTCH1, FH, MECP2, COG5, GTPBP3, COL4A1, FASN, L2HGDH, TUBB4A, TUBA8, KCNQ5, DLD, NGLY1, CSTB, SERAC1, RAB11B, ATP13A2, GNAO1, PEX16, NDUFAF6, EIF2B3, RNASEH2A, AP4S1, GABRA2, PIGT, HECW2, NOTCH3, GLI2, SLC25A12, YY1, VPS13B, MT-TS2, SHH, DPAGT1, GABRB3, PIGN, SUMF1, GABRG2, NDUFV1, AP2M1, WDR45, ADAM22, ZSWIM6, PSPH, DPYS, MT-TL2, ALG9, PEX13, KCNJ11, PIGO, PEX2, RALA, SIX3.

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