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ENDOCRINOLOGY DISORDER PANEL (226 GENES)

The genetic variants in 226 genes analyzed in this panel are closely related to or, potentially, the cause of major endocrinological conditions.

Genes included in the Endocrinology Disorder Panel include:

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, FXYD2, 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, NR0B1, NR0B2, 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, RSPO1, 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)

The Gastroenterology Panel analyzes 146 genes associated with hereditary gastrointestinal diseases.

Genes included in the Gastroenterology Panel:

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 (129 GENES)

The Mitochondrial Panel analyzes 129 genes including mitochondrial DNA (mtDNA) genes to confirm a suspected diagnosis of the mitochondrial diseases.

Genes included in the Mitochondrial Panel:

ATP5F1A, ATP5F1D, ATP5F1E, ATPAF2, BCS1L, COA8, COX10, COX14, COX15, COX20, COX6B1, COX8A, CYC1, DGUOK, DLAT, DLD, ECHS1, FASTKD2, FBXL4, FDX2, FOXRED1, HADHA, HADHB, HMGCS2, LRPPRC, LYRM7, MGME1, MPC1, MPV17, MRM2, 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, MTFMT, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NUBPL, OPA1, PC, PDHA1, PDHB, PDHX, PET100, PNPLA8, POLG, POLG2, PUS1, RRM2B, SCN4A, SCO1, SDHA, SDHAF1, SDHD, SLC25A3, SLC25A4, SUCLA2, SUCLG1, SURF1, TACO1, TFAM, TIMMDC1, TK2, TMEM126B, TMEM70, TTC19, TWNK, TYMP, UQCC2, UQCC3, UQCRB, UQCRC2, UQCRQ, YARS2.

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AUTOIMMUNITY PANEL (111 GENES)

The Autoimmunity panel analyzes 111 genes associated with different autoimmune disorders.

Genes included in the Autoimmune Panel:

ABCC8, ACP5, ADA, ADA2, AGRN, AIRE, AKT2, ALG14, BANK1, BTK, C1GALT1C1, C1QA, C1QB, C1QC, C1R, C1S, C2, C4A, C8A, CASP10, CAV1, CD19, CD247, CD3G, CD81, CHAT, CHD7, COL13A1, COMT, CR2, CTLA4, DCLRE1C, DDX41, DNASE1, DNASE1L3, FADD, FAS, FASLG, FCGR2A, FCGR2B, FOXD3, FOXP3, GALC, GCK, GNAS, GP1BB, HLA-B, HLA-DRB1, ICOS, IL2RA, IL2RG, IL6, IL7R, INS, IRF5, ITCH, ITGAM, ITK, KCNJ11, KDM6A, KMT2D, KRAS, LCK, LIG4, LMNB2, LRBA, MASP2, MIF, MMP2, MS4A1, NBN, NFKB1, NFKB2, NHEJ1, NLRP1, NRAS, PAX4, PDX1, PEPD, PLAGL1, PLCG2, PNP, POLG, PRKCD, PTEN, PTPN22, RAG1, RAG2, RMRP, SEC23B, SERPING1, SLC25A1, SLC5A7, SNAP25, STAT1, STAT3, STAT4, STIM1, STX16, SYT2, TBX1, TNFAIP3, TNFRSF13B, TNFRSF13C, TREX1, TSHR, TTC7A, VAMP1, WAS, WIPF1, ZFP57.

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HEARING LOSS AND DEAFNESS PANEL (262 GENES)

The genetic variants in 262 genes analyzed in this panel are associated with syndromic and non-syndromic deafness.

Genes included in the Hearing Loss and Deafness Panel:

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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SKELETAL AND CONNECTIVE TISSUE DISORDERS PANEL (416 GENES)

The Skeletal and Connective Tissue Disorders Panel analyzes 416 genes associated with conditions affecting the skeletal system and connective tissues that can help establish a diagnosis and, in some cases, allow for appropriate management and surveillance for disease features based on the gene involved.

Genes included in the Skeletal and Connective Tissue Disorders Panel:

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, ATP6V0A2, 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, PTDSS1, 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, SLCO5A1, 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, TRAIP, 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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CARRIER SCREENING PANEL (302 GENES)

The Carrier Screening Panel is a 302-gene panel aimed at investigating the probability of transmission of a genetic disease from parents to children. Each of us can be a healthy carrier of a hereditary disease without being aware of it. Knowing your carrier status, in anticipation of a future pregnancy, is medical information of primary importance for the prevention of the development of autosomal recessive and X-linked diseases in children whose parents are healthy. If both partners are carriers of the same autosomal recessive disease, there is a 25% chance that they will pass on the pathogenic gene variants to their children in each pregnancy. In the case of diseases transmitted by the X chromosome, the risk of a healthy carrier mother giving birth to an affected male child is 50%, and the risk of giving birth to a healthy daughter, also a carrier, is 50%. This panel is recommended for couples who are planning a pregnancy and want to know if there is a risk of transmitting a genetic disease to the child. Testing is especially recommended if there are already family members with a hereditary disease, in case of pregnancy loss without other known causes, in case of blood relationship between partners, or in case they belong to a certain ethnic group.

Genes included in the Carrier Screening Panel include:

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, GALC, 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, MTTP, 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, BTD, F11, F2, F5, G6PD, GP1BA, GP9, HFE, HGD, MCCC1, MCCC2, MEFV, SERPINA1.

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INFLAMMATORY BOWEL DISEASE (IBD) PANEL (125 GENES)

The Inflammatory Bowel Disease Panel analyzes 125 genes known to be associated with IBD and immunodeficiency that can help establish a diagnosis and, in some cases, allow for appropriate management and surveillance for disease features based on the gene involved.

Genes included in the Inflammatory Bowel Disease Panel:

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, TGFBR1, TGFBR2, TINF2, TNFAIP3, TNFRSF13B, TRAF3, TRIM22, TTC37, TTC7A, UNC13D, UNG, WAS, WIPF1, XIAP, ZAP70, ZBTB24, ZNF341.

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PEDIATRIC PANEL (613 GENES)

The genetic variants analyzed in the Pediatric Panel are closely related to or, potentially, the cause of major pediatric conditions.

Genes included in the Pediatric Panel:

A2M, AAAS, ABCA12, ABCB11, ABCD1, ABL1, ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADVL, ACE, ACOX1, ADA, ADAMTS10, ADAMTS13, ADAMTS17, ADAMTS2, ADAMTSL2, ADAMTSL4, ADGRV1, AGA, AGL, AGPS, AGT, AGTR1, AGXT, AHI1, AIRE, ALDH3A2, ALDH5A1, ALDOB, ALG1, ALG6, ALMS1, ALPL, ALS2, AMACR, AMELY, AMPD1, AMT, ANTXR2, AP1S2, APOE, APP, APTX, AR, ARSA, ARSB, ARVCF, ARX, ASL, ASPA, ASS1, ATIC, ATM, ATP13A2, ATP1A3, ATP2A2, ATP6V0A2, ATP7A, ATP7B, ATP8B1, ATR, ATRX, AUH, AUTS2, AVPR1A, B4GALT1, BCKDHA, BCKDHB, BCS1L, BDNF, BGN, BLM, BMPR2, BRAF, BRCA2, BRIP1, BTD, BTK, CA2, CACNA1C, CASK, CBS, CD40LG, CD96, CDH23, CDKL5, CDKN1C, CEP290, CEP78, CFP, CFTR, CHD7, CHD8, CHM, CHRNA1, CHRNA7, CHRND, CHRNG, CIB2, CLDN1, CLDN19, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGB3, CNTNAP2, CNTNAP5, COL11A1, COL11A2, COL17A1, COL1A1, COL1A2, COL2A1, COL3A1, COL4A3, COL4A4, COL5A1, COL5A2, COL7A1, COMT, COQ2, CPS1, CPT1A, CPT2, CREBBP, CRLF1, CRTAP, CSF1R, CSTB, CTNS, CTSD, CTSF, CTSK, CXCR4, CYP11A1, CYP21A2, CYP27A1, CYP27B1, D2HGDH, DCLRE1C, DCTN1, DCX, DDB2, DDC, DGUOK, DHCR24, DHCR7, DKC1, DLD, DLL3, DMD, DMP1, DMPK, DNAJC19, DNAJC5, DNMT1, DNMT3B, DOCK4, DOLK, DPAGT1, DPM1, DPP10, DPP6, DPYD, DSP, EDA, EDN3, EDNRB, EFEMP2, EGR2, EHMT1, EIF2AK3, EIF4G1, ENPP1, EPM2A, ERBB3, ERCC2, ERCC3, ERCC4, ERCC5, ERCC6, ERCC8, ESCO2, ETFA, ETFB, ETFDH, ETHE1, F11, F5, FAH, FAM126A, FAM20C, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FBLN5, FBN1, FBN2, FBXO7, FGA, FGB, FGD1, FGD4, FGFR2, FGG, FH, FKRP, FKTN, FLI1, FLNA, FMR1, FOLR1, FOXG1, FOXN1, FOXP1, FOXP2, FOXP3, FRAS1, FREM2, FUCA1, G6PC3, G6PD, GAA, GABBR2, GABRB3, GABRG1, GALC, GALT, GATA1, GBA, GBE1, GCDH, GCH1, GCSH, GFM1, GJA1, GJA5, GJA8, GJB2, GJC2, GLA, GLB1, GLDC, GLE1, GLI3, GNA14, GNE, GNPTAB, GNS, GP1BB, GRHPR, GRIN2B, GRN, GRPR, GSS, GTF2H5, GUSB, H19, HADH, HADHA, HADHB, HAMP, HBA1, HBB, HESX1, HEXA, HEXB, HFE, HGD, HGSNAT, HIBCH, HIRA, HMGCL, HOXA1, HPRT1, HSD17B10, HSD17B4, HSPG2, HTRA2, HYLS1, IDUA, IGBP1, IGF1, IGF2, IGHMBP2, IKBKG, IL2RG, IMMP2L, INSR, INVS, IQCB1, ITGA6, ITGB4, IVD, JAK3, JMJD1C, KATNAL2, KCNJ1, KCNQ1, KCTD7, KDM5C, KIRREL3, KLHL3, KRT18, KRT8, KRT81, KRT83, KRT86, L1CAM, LAMA2, LAMA3, LAMB2, LAMB3, LAMC2, LAMC3, LBR, LHX3, LIFR, LMNA, LRP2, LRP5, LRPPRC, LRRK2, LYST, MAN2B1, MAPK10, MAPT, MAT2A, MBD5, MCCC2, MCOLN1, MECP2, MED12, MEF2C, MEFV, MET, MFSD8, MGAT2, MID1, MKS1, MLC1, MMAB, MMACHC, MOCS1, MOCS2, MOGS, MPI, MPL, MPO, MPV17, MPZ, MRPS16, MRPS22, MTHFR, MTM1, MTTP, MUTYH, MVK, MYO5A, MYO7A, NAGA, NAGS, NBN, NDP, NEB, NEGR1, NEU1, NEUROG3, NHLRC1, NHS, NIPBL, NLGN3, NLGN4X, NLGN4Y, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPHS1, NR0B1, NRXN1, NSD1, NTNG1, NTRK1, NUP62, OCRL, OFD1, OPA3, OPHN1, OSTM1, OTC, PAFAH1B1, PAH, PALB2, PARK7, PAX6, PC, PCDH15, PCDH19, PCDH9, PDE10A, PDHA1, PDHX, PDP1, PDSS1, PDSS2, PDZD7, PEX7, PHF6, PINK1, PIP5K1B, PKHD1, PKLR, PLA2G6, PLCE1, PLEKHG5, PLG, PLOD1, PLP1, PMM2, PMP22, PNKP, PNPO, POLG, POMGNT1, POMT1, POMT2, PON3, POU1F1, PPT1, PQBP1, PREPL, PRKAG2, PRKAR1A, PRKN, PRKRA, PRNP, PROC, PRODH, PROP1, PRPS1, PRX, PSAP, PSAT1, PSEN1, PSEN2, PTCHD1, PTEN, PTH1R, PTPN11, RAB23, RAB27A, RAB39B, RAB3GAP1, RAB3GAP2, RAD51C, RAG1, RAG2, RAI1, RAPSN, RBFOX1, RELN, REN, RHOBTB2, RMRP, RPGRIP1L, RPL10, RREB1, RS1, SACS, SATB2, SBDS, SCN1A, SCN2A, SCNN1A, SCNN1B, SCNN1G, SCO2, SEC24C, SERPINA1, SFTPA1, SFTPB, SFTPC, SGCA, SGSH, SH2D1A, SHANK2, SHANK3, SIL1, SKI, SLC12A1, SLC12A6, SLC16A2, SLC17A5, SLC25A15, SLC25A22, SLC26A2, SLC26A4, SLC2A10, SLC34A2, SLC35A1, SLC35C1, SLC35D1, SLC37A4, SLC3A1, SLC4A11, SLC6A3, SLC6A4, SLC6A8, SLC9A6, SLC9A9, SLX4, SMAD3, SMAD6, SMC1A, SMG6, SMN1, SMPD1, SNAP29, SNCA, SNCB, SNRPN, SOX10, SOX5, SP110, SPAST, SRY, ST3GAL5, ST7, STAR, STRA6, STXBP1, SUCLG1, SUOX, TAF1, TBCE, TBL1Y, TBX1, TCF4, TCIRG1, TFR2, TGFB2, TGFB3, TGFBR1, TGFBR2, TGM1, TH, TIMM8A, TK2, TMEM67, TNFRSF11B, TNNT1, TPP1, TREM2, TREX1, TRIM37, TSC1, TSC2, TSEN54, TSFM, TSHB, TSPY1, TSPYL1, TTN, TTPA, TYROBP, UBA1, UBE3A, UBR1, UCHL1, UFD1, UPF3B, UQCRB, UQCRQ, UROS, USH1C, USH1G, USH2A, VCAN, VDR, VLDLR, VPS13B, VPS33B, VPS35, WAS, WFS1, WHRN, WNT10A, WNT3, WNT7A, WT1, XIAP, XPA, XRCC2, ZDHHC9, ZEB2, ZIC3, ZMPSTE24, ZNF469.

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COMPREHENSIVE HEMATOLOGY PANEL (294 GENES)

The genetic variants in 294 genes analyzed in this panel are closely related to or, potentially, the cause of major hematologic conditions.

Genes included in the Comprehensive Hematology Panel:

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)

The Neurology Comprehensive Panel analyzes 882 genes associated with hereditary neurological diseases.

Genes included in the Neurology Comprehensive Panel:

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, ARFGEF2, ARG1, ARHGEF10, ARHGEF6, ARHGEF9, ARL13B, ARL6, ARSA, ARX, ASAH1, ASNS, ASPA, ASPM, ATAD3A, ATCAY, ATL1, ATL3, ATM, ATN1, ATP13A2, ATP1A2, ATP1A3, ATP2A1, ATP2B3, ATP6AP2, ATP6V0A2, 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, BTD, 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, CHCHD10, CHD2, CHD8, CHMP2B, CHN1, CHRNA1, CHRNA2, CHRNA4, CHRNB1, CHRNB2, CHRND, CHRNE, CHRNG, CISD2, CLCN1, CLCN2, CLCN4, CLN3, CLN5, CLN6, CLN8, CLPP, CNOT3, CNTN6, CNTNAP2, COASY, COL12A1, COL3A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COLQ, COQ2, COQ4, COQ5, COQ6, COQ7, COQ9, COX10, COX15, COX20, COX6A1, COX6B1, CP, CPOX, CPT1C, CPT2, CRYAB, CSF1R, CSTB, CTC1, CTDP1, CTNNB1, CTNND2, CTSD, CTSF, CUL4B, CWF19L1, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DARS2, DCAF17, DCAF8, DCTN1, DCX, DDC, DDHD1, DDHD2, DDX3X, DEPDC5, DES, DHCR24, DHCR7, DHFR, DHTKD1, DIS3L2, DKC1, DLG3, DMD, DNAJB2, DNAJB6, DNAJC19, DNAJC5, DNAJC6, DNM1, DNM1L, DNM2, DNMT1, DNMT3A, DOCK7, DOK7, DPAGT1, DPYD, DPYS, DST, DYNC1H1, DYRK1A, DYSF, EARS2, EBF3, ECHS1, ECM1, EED, EEF1A2, EEF2, EFHC1, EFTUD2, EGR2, EHMT1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELF2, ELK1, ELOVL4, ELOVL5, EMD, EMX2, EN2, ENO3, EPM2A, ERLIN2, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, EXOSC8, EZH2, FA2H, FAM126A, FANCB, FAR1, FARS2, FAT4, FBLN5, FBXL4, FBXO38, FBXO7, FDXR, FECH, FGD1, FGD4, FGF12, FGF14, FGF8, FGFR1, FH, FHL1, FIG4, FKRP, FKTN, FLAD1, FLNA, FLNC, FLVCR1, FLVCR2, FMR1, FOLR1, FOXG1, FOXH1, FOXP1, FOXRED1, FRMD7, FTSJ1, FUS, FXN, GAA, GABRA1, GABRB2, GABRB3, GABRG2, GALC, GAMT, GAN, GATM, GBA2, GBE1, GCDH, GCH1, GDAP1, GDI1, GFAP, GFM1, GFPT1, GJB1, GJC2, GK, GLB1, GLDC, GLI2, GLI3, GLRB, GMPPB, GNAL, GNAO1, GNB1, GNB4, GNE, GOSR2, GPC3, GPHN, GPR143, GPSM2, GRIA3, GRID2, GRIK2, GRIN1, GRIN2A, GRIN2B, GRM1, GRN, GSS, GTPBP3, GYG1, GYS1, HACE1, HADHA, HADHB, HARS2, HCCS, HCN1, HECW2, HEPACAM, HESX1, HEXA, HFE, HIBCH, HINT1, HK1, HMBS, HNRNPA1, HNRNPU, HPRT1, HSD17B10, HSPB1, HSPB3, HSPB8, HSPD1, HTRA1, HTT, HUWE1, IBA57, IDS, IFIH1, IGBP1, IGHMBP2, IL1RAPL1, INF2, INPP5E, IQSEC2, ISCU, ITGA7, ITM2B, ITPR1, KANSL1, KATNB1, KBTBD13, KCNA1, KCNA2, KCNB1, KCNC1, KCNC3, KCND3, KCNH1, KCNJ10, KCNJ2, KCNK18, KCNQ2, KCNQ3, KCNT1, KCTD7, KDM5C, KIF11, KIF1A, KIF1B, KIF1C, KIF21A, KIF5A, KIF7, KLF8, KLHL40, KLHL41, KMT2B, KMT5B, KPTN, KRIT1, L1CAM, L2HGDH, LAMA1, LAMA2, LAMB1, LAMB2, LAMC3, LAMP2, LARS2, LAS1L, LDB3, LDHA, LGI1, LIG4, LIMS2, LITAF, LMNA, LMNB1, LMOD3, LPIN1, LRPPRC, LRRK2, LRSAM1, LYRM7, MAGI2, MAGT1, MAOA, MAPT, MARS2, MBD5, MBTPS2, MCM3AP, MCPH1, MECP2, MED12, MED25, MEF2C, MEGF10, MFN2, MFSD2A, MFSD8, MICU1, MID1, MKKS, MKS1, MLC1, MME, MOCS1, MPDZ, MPZ, MRPL44, MTFMT, MTHFR, MTM1, MTMR2, MTOR, MTPAP, MTTP, MUSK, MYCN, MYH3, MYH7, MYOT, NACC1, NAGLU, NDE1, NDP, NDRG1, NDUFA1, NDUFAF5, NDUFAF6, NDUFS1, NDUFS2, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NEB, NECAP1, NEFH, NEFL, NEU1, NFIX, NFU1, NGF, NHEJ1, NHLRC1, NHS, NIPA1, NLGN3, NLGN4X, NODAL, NOL3, NOP56, NOTCH3, NPHP1, NR2F1, NRXN1, NSD1, NSDHL, NTRK1, NUBPL, NXF5, OCLN, OCRL, OFD1, OPA1, OPA3, OPHN1, OPTN, OTC, OTX2, PABPN1, PAFAH1B1, PAH, PAK3, PARK7, PAX6, PCDH19, PCNT, PDCD10, PDGFB, PDGFRB, PDHA1, PDK3, PDSS1, PDSS2, PDYN, PEX7, PFKM, PGAM2, PGK1, PGM1, PHF6, PHF8, PHGDH,

PHKA1, PHOX2A, PHYH, PI4KA, PIGA, PIGN, PIGO, PIGT, PIGV, PIK3CA, PIK3R2, PINK1, PLA2G6, PLCB1, PLEC, PLEKHG5, PLK4, PLP1, PMP22, PNKD, PNKP, PNPLA2, PNPLA6, PNPO, POGLUT1, POGZ, POLG, POLG2, POLR3A, POLR3B, POMGNT2, POMT1, PORCN, PPOX, PPP2R2B, PPT1, PQBP1, PRDM12, PRF1, PRICKLE1, PRIMA1, PRKCG, PRKRA, PRNP, PRODH, PRPS1, PRRT2, PRX, PSAP, PSEN1, PSEN2, PTCH1, PTCHD1, PTEN, PTS, PURA, PYCR2, PYGM, QDPR, RAB18, RAB39B, RAB3GAP1, RAB3GAP2, RAB7A, RAPSN, RARS2, RASA1, RBCK1, RBM10, REEP1, RELN, RFC1, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF135, RNF216, ROBO3, ROGDI, RPGRIP1L, RPL10, RPS6KA3, RRM2B, RTN2, RTN4IP1, RTTN, RUBCN, RYR1, SACS, SALL4, SAMHD1, SBF1, SBF2, SCARB2, SCN11A, SCN1A, SCN1B, SCN2A, SCN3A, SCN4A, SCN8A, SCN9A, SCO1, SCO2, SDHAF1, SELENON, SEPSECS, SERAC1, SERPINI1, SETD2, SETX, SGCA, SGCB, SGCD, SGCE, SGCG, SH3TC2, SHANK3, SHH, SHROOM4, SIGMAR1, SIK1, SIL1, SIX3, SLC12A5, SLC12A6, SLC13A5, SLC16A2, SLC19A3, SLC1A3, SLC20A2,SLC22A5, SLC25A1, SLC25A15, SLC25A20, SLC25A22, SLC25A26, SLC25A4, SLC25A46, SLC2A1, SLC33A1, SLC35A2, SLC38A8, SLC39A8, SLC46A1, SLC52A2, SLC52A3, SLC5A7, SLC6A1, SLC6A19, SLC6A3, SLC6A8, SLC9A6, SMAD3, SMC1A, SMCHD1, SMN1, SMN2, SMS, SNAP25, SNCA, SNX14, SOD1, SORL1, SOX10, SOX2, SOX3, SPAST, SPATA5, SPEG, SPG11, SPG21, SPG7, SPR, SPTAN1, SPTBN2, SPTLC1, SPTLC2, SQSTM1, SRPX2, ST3GAL3, ST3GAL5, STAMBP, STIL, STIM1, STUB1, STX1B, STXBP1, SUCLA2, SUMF1, SUOX, SURF1, SYN1, SYNE1, SYNGAP1, SYNJ1, SYP, SYT14, SZT2, TAF1, TANGO2, TARDBP, TBC1D24, TBCD, TBCE, TBCK, TBL1XR1, TBP, TCAP, TCF20, TCF4, TCTN1, TCTN2, TCTN3, TDP1, TFG, TGIF1, TGM6, TH, THAP1, TIMM8A, TK2, TMEM126A, TMEM126B, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM43, TMEM67, TNNT1, TNPO3, TOR1A, TOR1AIP1, TPM2, TPM3, TPP1, TRAPPC11, TREM2, TREX1, TRIM2, TRIP12, TRPV4, TSC1, TSC2, TSEN2, TSEN54, TSPAN7, TTBK2, TTC19, TTC8, TTN, TTPA, TTR, TUBA1A, TUBA4A, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP4, TUBGCP6, TYMP, UBA1, UBA5, UBE2A, UBE3A, UBQLN2, UNC80, UPF3B, UROD, UROS, USP9X, VAMP1, VAPB, VCP, VLDLR, VMA21, VPS13A, VPS35, VPS37A, VRK1, WDPCP, WDR26, WDR45, WDR62, WDR73, WDR81, WFS1, WNK1, WWOX, XRCC4, YWHAE, YY1, ZC4H2, ZCCHC12, ZDHHC9, ZEB2, ZFYVE26, ZIC2, ZNF41, ZNF423, ZNF592, ZNF674, ZNF711, ZNF81

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COMPREHENSIVE EPILEPSY PANEL (591 GENES)

The Comprehensive Epilepsy Panel analyzes 591 genes that are associated with both syndromic and nonsyndromic causes of epilepsy, a common neurological disease characterized by recurrent, unprovoked seizures.

Genes included in the Comprehensive Epilepsy Panel:

MT-TQ, NALCN, COX15, ALG3, GFM1, PCDH19, CPA6, MAP2K1, OFD1, PSAP, MT-RNR1, KANSL1, MICAL1, DNM1L, 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, RHOBTB2, GABRA1, MBOAT7, COX6B1, ATP7A, CASR, PTEN, TPK1, TBCE, ALG8, CHD2, CACNA1H, PEX10, EHMT1, PIGQ, GABBR2, ST3GAL5, PACS2, CLN5, BCKDK, RNASEH2B, DNM1, 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, BTD, 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.