CENTOGENE
GUIDING PRECISION MEDICINE

Gene List

Neurology Panels

Samples

CentoCard

EDTA Blood

Ready to use DNA

Buccal Swab

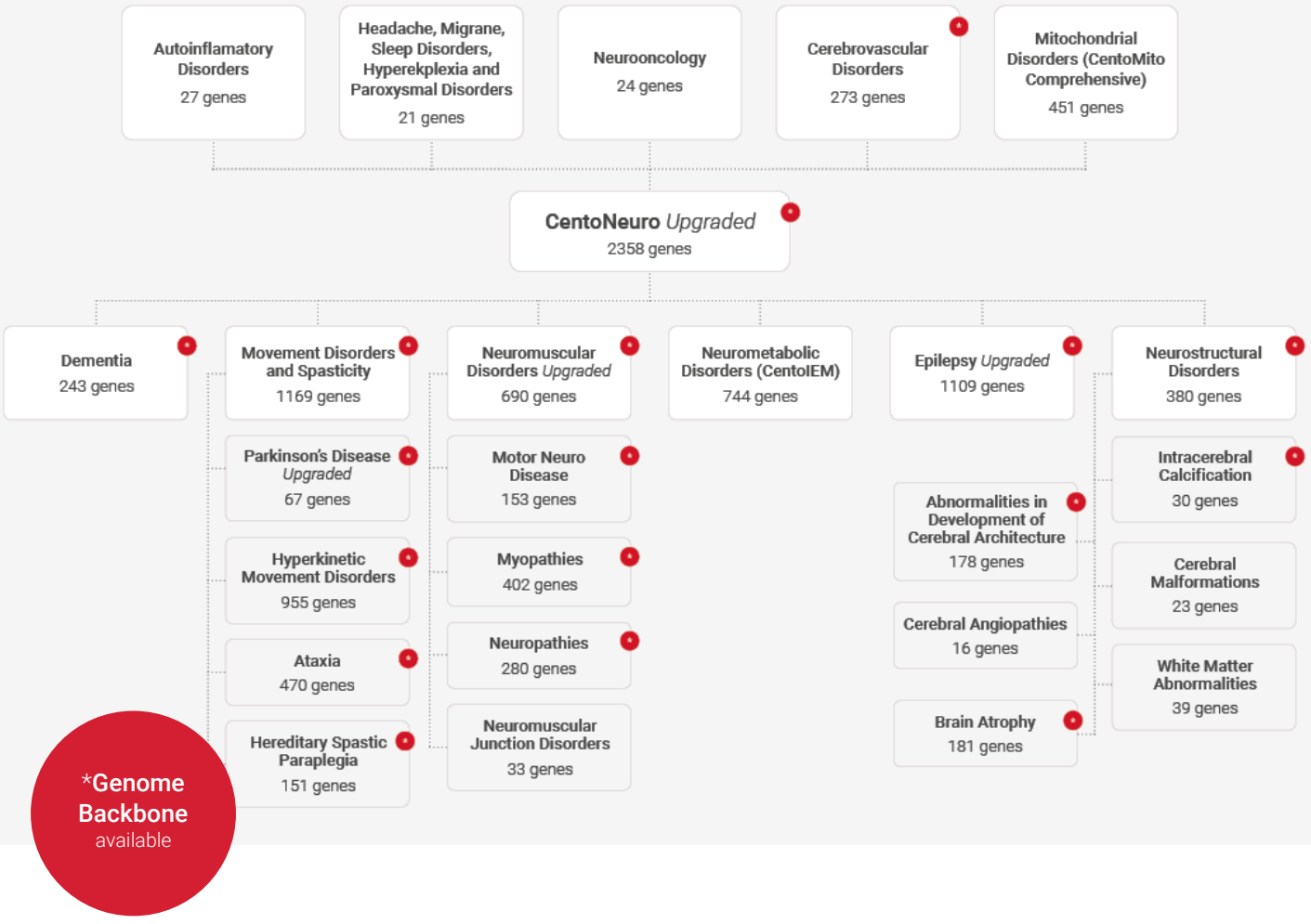
Saliva

Methods

NGS including CNV analysis

TAT

25 business days



CentoNeuro Upgraded Genome | 2358 genes

CentoNeuro Upgraded Genome is our largest neurology panel, designed to detect a wide range of neurological disorders. This panel is intended for patients with clinical features suggestive of epilepsies, dementia, movement disorders, and spasticity, including ataxia disorders and Parkinson's disease; neuromuscular disorders; cerebrovascular disorders; hyperekplexia; paroxysmal disorders; and neurostructural disorders, among others.

> This version of the panel is based on genome sequencing and includes the non-coding genes RNU4-2, RNU7-1, and SNORD118; repeat expansion screening of *AR, ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, C9orf72, CACNA1A, CNBP, CSTB, DMPK, FMR1, FXN, HTT, JPH3, NOP56, PABPN1, PHOX2B, PPP2R2B, PRNP,* and *TBP*; *GBA1* conversion analysis with its pseudogene; and *SMN1* CNV analysis.

Genes Included

AAAS, AARS1, AARS2, AASS, ABAT, ABCA1, ABCA2, ABCB11, ABCB4, ABCB6, ABCB7, ABCC2, ABCC6, ABCC8, ABCC9, ABCD1, ABCD3, ABCD4, ABCG5, ABCG8, ABHD12, ABHD16A, ABHD5, ACACA, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACBD6, ACE, ACER3, ACO2, ACOX1, ACOX2, ACP5, ACSF3, ACSL4, ACTA1, ACTA2, ACTB, ACTC1, ACTL6B, ACTN2, ACVRL1, ACY1, ADA, ADA2, ADAM10, ADAM22, ADAMTS10, ADAMTS13, ADAMTS15, ADAMTS2, ADAMTSL2, ADAR, ADARB1, ADAT3, ADCY5, ADCY6, ADGRG1, ADGRG6, ADGRV1, ADK, ADPRS, ADSL, ADSS1, AEBP1, AFF3, AFG2A, AFG2B, AFG3L2, AGA, AGK, AGL, AGO1, AGPAT2, AGPS, AGRN, AGTPBP1, AGXT, AHCY, AHI1, AIFM1, AIMP1, AIMP2, AIP, AK2, AKAP9, AKR1D1, AKT1, AKT2, AKT3, ALAD, ALAS2, ALDH18A1, ALDH2, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALK, ALKBH8, ALMS1, ALPL, ALS2, ALX1, ALX3, ALX4, AMACR, AMFR, AMH, AMN, AMPD1, AMPD2, AMT, ANG, ANGPTL3, ANK1, ANK2, ANK3, ANKLE2, ANKRD11, ANKS6, ANO10, ANO3, ANO4, ANO5, ANTXR1, ANTXR2, ANXA11, AP1G1, AP1S1, AP1S2, AP2M1, AP3B1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APC, APC2, APOA1, APOA2, APOA5, APOB, APOC2, APOE, APP, APPL1, APRT, APTX, AQP2, AR, ARF1, ARF3, ARFGEF1, ARFGEF2, ARG1, ARHGAP31, ARHGEF9, ARID1B, ARL13B, ARL3, ARL6, ARL6IP1, ARMC9, ARPC1B, ARSA, ARSB, ARSL, ARV1, ARX, ASAH1, ASCC1, ASCC3, ASH1L, ASL, ASNS, ASPA, ASPM, ASRGL1, ASS1, ASXL1, ASXL3, ATAD1, ATAD3A, ATCAY, ATG7, ATIC, ATL1, ATL3, ATM, ATN1, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2A1, ATP2A2, ATP2B1, ATP2B3, ATP5F1A, ATP5F1E, ATP5MC3, ATP5PO, ATP6AP1, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0C, ATP6V1A, ATP7A, ATP7B, ATP8A2, ATP8B1, ATPAF2, ATR, ATRX, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, AUH, AVP, AVPR2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, B9D1, B9D2, BAAT, BAG3, BAP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCAS3, BCKDHA, BCKDHB, BCKDK, BCORL1, BCS1L, BEAN1, BEST1, BET1, BICD2, BIN1, BLK, BLOC1S1, BLOC1S3, BLTP1, BMP6, BOLA3, BORCS8, BRAF, BRAT1, BRCA2, BRD4, BRF1, BSCL2, BTD, C12orf57, C19orf12, C1QBP, C1R, C2CD3, C2orf69, C5, C9orf72, CA2, CA5A, CA8, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1H, CACNA1I, CACNA1S, CACNA2D2, CAD, CAMK2D, CAMK2G, CAMSAP1, CAMTA1, CAPN1, CAPN3, CAPRIN1, CARD14, CARS2, CASK, CASP10, CASP8, CASQ1, CASQ2, CASR, CAT, CAV1, CAV3, CAVIN1, CBL, CBLIF, CBS, CBY1, CC2D2A, CCDC186, CCDC47, CCDC78, CCDC88A, CCDC88C, CCM2, CCND2, CCNF, CCT5, CD2AP, CD320, CD59, CDC42BPB, CDH2, CDK19, CDK5, CDKL5, CDKN2A, CDON, CEL, CELF2, CELSR1, CENPF, CEP104, CEP120, CEP152, CEP164, CEP290, CEP41, CEP55, CEP63, CEP83, CEP85L, CERS1, CERT1, CETP, CFAP410, CFAP43, CFL2, CHAMP1, CHAT, CHCHD10, CHCHD2, CHD1, CHD2, CHD4, CHD5, CHKA, CHKB, CHMP1A, CHMP2B, CHRNA1, CHRNA2, CHRNA3, CHRNA4, CHRNB1, CHRNB2, CHRND, CHRNE, CHRNG, CHST14, CHST3, CHST6, CHSY1, CIC, CILK1, CISD2, CIZ1, CLCF1, CLCN1, CLCN2, CLCN3, CLCN4, CLCN6, CLDN11, CLDN16, CLDN19, CLIC5, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, 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CentoNeuro Upgraded | 2355 genes

CentoNeuro Upgraded is our largest neurology panel, designed to detect a wide range of neurological disorders. This panel is intended for patients with clinical features suggestive of epilepsies, dementia, movement disorders, and spasticity, including ataxia disorders and Parkinson's disease; neuromuscular disorders; cerebrovascular disorders; hyperekplexia; paroxysmal disorders; and neurostructural disorders, among others. Please note that repeat expansion disorders, SMN1 CNV analysis, and GBA conversion analysis with its pseudo gene are not covered by this panel. If there is clinical suspicion of these conditions, the Genome version of this panel is recommended.

Genes Included

AAAS, AARS1, AARS2, AASS, ABAT, ABCA1, ABCA2, ABCB11, ABCB4, ABCB6, ABCB7, ABCC2, ABCC6, ABCC8, ABCC9, ABCD1, ABCD3, ABCD4, ABCG5, ABCG8, ABHD12, ABHD16A, ABHD5, ACACA, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACBD6, ACE, ACER3, ACO2, ACOX1, ACOX2, ACP5, ACSF3, ACSL4, ACTA1, ACTA2, ACTB, ACTC1, ACTL6B, ACTN2, ACVRL1, ACY1, ADA, ADA2, ADAM10, ADAM22, ADAMTS10, ADAMTS13, ADAMTS15, ADAMTS2, ADAMTSL2, ADAR, ADARB1, ADAT3, ADCY5, ADCY6, ADGRG1, ADGRG6, ADGRV1, ADK, ADPRS, ADSL, ADSS1, AEBP1, AFF3, AFG2A, AFG2B, AFG3L2, AGA, AGK, AGL, AGO1, AGPAT2, AGPS, AGRN, AGTPBP1, AGXT, AHCY, AHI1, AIFM1, AIMP1, AIMP2, AIP, AK2, AKAP9, AKR1D1, AKT1, AKT2, AKT3, ALAD, ALAS2, ALDH18A1, ALDH2, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALK, ALKBH8, ALMS1, ALPL, ALS2, ALX1, ALX3, ALX4, AMACR, AMFR, AMH, AMN, AMPD1, AMPD2, AMT, ANG, ANGPTL3, ANK1, ANK2, ANK3, ANKLE2, ANKRD11, ANKS6, ANO10, ANO3, ANO4, ANO5, ANTXR1, ANTXR2, ANXA11, AP1G1, AP1S1, AP1S2, AP2M1, AP3B1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APC, APC2, APOA1, APOA2, APOA5, APOB, APOC2, APOE, APP, APPL1, APRT, APTX, AQP2, AR, ARF1, ARF3, ARFGEF1, ARFGEF2, ARG1, ARHGAP31, ARHGEF9, ARID1B, ARL13B, ARL3, ARL6, ARL6IP1, ARMC9, ARPC1B, ARSA, ARSB, ARSL, ARV1, ARX, ASAH1, ASCC1, ASCC3, ASH1L, ASL, ASNS, ASPA, ASPM, ASRGL1, ASS1, ASXL1, ASXL3, ATAD1, ATAD3A, ATCAY, ATG7, ATIC, ATL1, ATL3, ATM, ATN1, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2A1, ATP2A2, ATP2B1, ATP2B3, ATP5F1A, ATP5F1E, ATP5MC3, ATP5PO, ATP6AP1, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0C, ATP6V1A, ATP7A, ATP7B, ATP8A2, ATP8B1, ATPAF2, ATR, ATRX, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, AUH, AVP, AVPR2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, B9D1, B9D2, BAAT, BAG3, BAP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCAS3, BCKDHA, BCKDHB, BCKDK, BCORL1, BCS1L, BEAN1, BEST1, BET1, BICD2, BIN1, BLK, BLOC1S1, BLOC1S3, BLTP1, BMP6, BOLA3, BORCS8, BRAF, BRAT1, BRCA2, BRD4, BRF1, BSCL2, BTD, C12orf57, C19orf12, C1QBP, C1R, C2CD3, C2orf69, C5, C9orf72, CA2, CA5A, CA8, CACNA1A, CACNA1B, CACNA1C, CACNA1D,

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INPP5E, INPP5K, INS, INSR, INTS11, INVS, IQCB1, IQSEC2, IRF2BPL, IRF6, ISCA1, ISCA2, ISCU, ISG15, ITGA7, ITGA8, ITIH4, ITK, ITM2B, ITPA, ITPR1, ITSN1, IVD, JAG1, JAG2, JAK2, JAM3, JPH2, JPH3, JUP, KANSL1, KARS1, KAT5, KAT6B, KAT8, KATNB1, KATNIP, KBTBD13, KCNA1, KCNA2, KCNA5, KCNB1, KCNC1, KCNC2, KCNC3, KCND2, KCND3, KCNH1, KCNH5, KCNJ1, KCNJ10, KCNJ11, KCNJ2, KCNK3, KCNK4, KCNMA1, KCNN2, KCNQ1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD17, KCTD3, KCTD7, KDM5C, KDM6A, KDM6B, KDR, KHK, KIAA0586, KIAA0753, KIDINS220, KIF14, KIF1A, KIF1B, KIF1C, KIF21A, KIF26A, KIF2A, KIF5A, KIF5C, KIF7, KIFBP, KLF11, KLHL20, KLHL40, KLHL41, KLHL7, KMT2B, KMT2D, KMT2E, KMT5B, KNG1, KPNA3, KPTN, KRAS, KRIT1, KRT5, KRT8, KRT86, KY, KYNU, L1CAM, L2HGDH, LAMA1, LAMA2, LAMA4, LAMA5, LAMB1, LAMB2, LAMC3, LAMP2, LARGE1, LARS1, LARS2, LAS1L, LAT, LBR, LCAT, LCT, LDB3, LDHA, LDLR, LDLRAP1, LETM1, LFNG, LGI1, LGI3, LGI4, LIAS, LIG3, LIPA, LIPC, LIPE, LIPI, LIPT1, LIPT2, LITAF, LMAN1, LMBRD1, LMBRD2, LMNA, LMNB1, 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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, MTTP, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MUSK, MVK, MYBPC1, MYBPC3, MYF5, MYH11, MYH14, MYH2, MYH3, MYH6, MYH7, MYH8, MYH9, MYL11, MYL2, MYL3, MYLK, MYMK, MYO18B, MYO5A, MYO9A, MYOD1, MYORG, MYOT, MYPN, MYRF, MYT1L, NAA60, NACC1, NADK2, NAGA, NAGLU, NAGS, NALCN, NANS, NAPB, NARS1, NARS2, NAXD, NAXE, NBAS, NBEA, NBEAL2, NCDN,

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TANC2, TANGO2, TAPT1, TARDBP, TARS2, TAT, TBC1D20, TBC1D23, TBC1D24, TBC1D2B, TBC1D32, TBCD, TBCE, TBCK, TBK1, TBL1XR1, TBP, TCAP, TCF4, TCIRG1, TCN2, TCOF1, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TECPR2, TEFM, TEK, TELO2, TENM4, TERT, TET3, TFE3, TFG, TFR2, TGFB1, TGFB2, TGFB3, TGFBR1, TGFBR2, TGIF1, TGM6, TH, THAP1, THAP11, THBD, THG1L, THSD1, TIA1, TIAM1, TIMM50, TIMM8A, TIMMDC1, TINF2, TK2, TMEM106B, TMEM107, TMEM126A, TMEM126B, TMEM138, TMEM151A, TMEM165, TMEM175, TMEM216, TMEM218, TMEM222, TMEM230, TMEM231, TMEM237, TMEM240, TMEM43, TMEM63B, TMEM63C, TMEM67, TMEM70, TMLHE, TMX2, TNFAIP3, TNFRSF1A, TNNC1, TNNI2, TNNI3, TNNT1, TNNT2, TNNT3, TNPO2, TNPO3, TNXB, TOE1, TOGARAM1, TOP3A, TOR1A, TOR1AIP1, TP53, TPI1, TPK1, TPM1, TPM2, TPM3, TPP1, TPP2, TRA2B, TRAF7, TRAIP, TRAK1, TRAPPC11, TRAPPC12, TRAPPC4, TRAPPC6B, TRAPPC9, TRDN, TREM2, TREX1, TRIM32, TRIM37, TRIM8, TRIP13, TRIP4, TRIT1, TRMT1, TRMT10A, TRMT10C, TRMT5, TRMU, TRNT1, TRPM3, TRPM6, TRPS1, TRPV4, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN34, TSEN54, TSFM, TSPOAP1, TTBK2, TTC19, TTC21B, TTC8, TTN, TTPA, TTR, TUBA1A, TUBA4A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUFM, TULP1, TUSC3, TWNK, TXNDC15, TYMP, TYROBP, U2AF2, UBA1, UBA5, UBAP1, UBAP2L, UBE2A, UBE3A, UBQLN2, UBR7, UBTF, UCHL1, UCP2, UFC1, UFM1, UFSP2, UGDH, UGP2, UGT1A1, UMOD, UMPS, UNC13D, UNC45B, UNC80, UNG, UPB1, UQCC2, UQCRB, UQCRC2, UQCRQ, UROC1, UROD, UROS, USH1C, USP18, USP7, VAC14, VAMP1, VAMP2, VAPB, VARS1, VARS2, VCAN, VCL, VCP, VHL, VIPAS39, VKORC1, VLDLR, VMA12, VMA21, VMA22, VPS11, VPS13A, VPS13B, VPS13C, VPS13D, VPS16, VPS33B, VPS35, VPS35L, VPS37A, VPS41, VPS4A, VPS50, VPS53, VRK1, VWA1, VWF, WARS1, WARS2, WASF1, WASHC5, WBP2, WDPCP, WDR11, WDR19, WDR35, WDR37, WDR45, WDR45B, WDR62, WDR73, WDR81, WFS1, WNK1, WNK3, WNK4, WNT7A, WRN, WWOX, XDH, XK, XPA, XPNPEP3, XPR1, XYLT1, XYLT2, YARS1, YARS2, YIF1B, YIPF5, YWHAG, YY1, YY1AP1, ZBTB18, ZC4H2, ZDHHC9, ZEB2, ZFHX3, ZFP57, ZFYVE26, ZFYVE27, ZIC2, ZMIZ1, ZMPSTE24, ZMYM2, ZNF142, ZNF335, ZNF423, ZNFX1, ZSWIM6

Movement Disorders and Spasticity Genome | 1169 genes

The movement disorders and spasticity panel Genome includes genes associated with disorders such as ataxia, hereditary spastic paraplegia, hyperkinetic movement disorders and Parkinson's disease.

> Recommended panel approach based on genome sequencing, including non-coding genes *RNU7-1* and *SNORD118*; repeat expansion screening of the genes *ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, C9orf72, CACNA1A, CSTB, DMPK, FMR1, FXN, HTT, JPH3, NOP56, PPP2R2B, PRNP, TBP* and *GBA1* conversion analysis with its pseudogene.

Genes Included

AAAS, AARS1, AARS2, AASS, ABAT, ABCA1, ABCA2, ABCB11, ABCB4, ABCB7, ABCD1, ABCD4, ABCG5, ABCG8, ABHD12, ABHD16A, ABHD5, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACBD6, ACER3, ACO2, ACOX1, ACSF3, ACTB, ACY1, ADA, ADAR, ADCY5, ADGRG1, ADPRS, ADSL, AFG2B, AFG3L2, AGA, AGK, AGL, AGPS, AGTPBP1, AGXT, AHI1, AIFM1, AIMP1, AKR1D1, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALK, ALMS1, ALPL, ALS2, AMACR, AMFR, AMN, AMPD2, AMT, ANKS6, ANO10, ANO3, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOA1, APOA5, APOB, APOC2, APOE, APRT, APTX, ARG1, ARL13B, ARL3, ARL6, ARL6IP1, ARMC9, ARSA, ARSB, ARSL, ARX, ASAH1, ASL, ASPA, ASS1, ATAD3A, ATCAY, ATG7, ATIC, ATL1, ATM, ATN1, ATP13A2, ATP1A2, ATP1A3, ATP2B3, ATP5MC3, ATP6AP1, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP7A, ATP7B, ATP8A2, ATP8B1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, AUH, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, B9D1, B9D2, BAAT, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCAS3, BCKDHA, BCKDHB, BCKDK, BCS1L, BEAN1, BICD2, BLOC1S1, BOLA3, BORCS8, BRF1, BSCL2, BTD, C19orf12, C1QBP, C2CD3, C9orf72, CA5A, CA8, CACNA1A, CACNA1G, CACNA2D2, CAD, CAMTA1, CAPN1, CAPRIN1, CASK, CAT, CBS, CBY1, CC2D2A, CCDC88C, CDK5, CENPF, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFAP410, CFAP43, CHCHD10, CHCHD2, CHKB, CHMP1A, CHMP2B, CHST14, CHST3, CHST6, CHSY1, CILK1, CISD2, CIZ1, CLCN2, CLDN11, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CNNM2, COA3, COA6, COA7, COA8, COASY, COG1, COG2, COG3, COG4, COG5, COG6, COG7, COG8, COL6A3, COQ2, COQ4, COQ6, COQ8A, COQ8B, COQ9, COX10, COX15, COX20, COX6A1, COX6B1, COX7B, COXFA4, CP, CPLANE1, CPOX, CPS1, CPT1A, CPT1C, CPT2, CRB2, CRPPA, CSF1R, CSGALNACT1, CSPP1, CSTB, CTBP1, CTH, CTNNB1, CTNS, CTSA, CTSC, CTSD, CTSK, CUBN, CWF19L1, CYC1, CYCS, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DAGLA, DARS1, DARS2, DBH, DBT, DCAF17, DCC, DCDC2, DCTN1, DCXR, DDC, DDHD1, DDHD2, DDOST, DDX3X, DDX59, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHODH, DHRSX, DHTKD1, DKC1, DLAT, DLD, DLG4, DMPK, DMXL2, DNA2, DNAH1, DNAJC12, DNAJC19, DNAJC3, DNAJC5, DNAJC6, DNM1L, DNMT1, DOCK3, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DPYS, DPYSL5, DSTYK, DYM, DYNC1H1, DYNC2H1, DYNC2I2, DYNC2LI1, DYNLT2B, EARS2, EBF3, EBP, ECHS1, EDEM3, EEF2, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4G1, ELAC2, ELOVL1, ELOVL4, ELOVL5, ENO3, ENTPD1, EOGT, EPG5, EPM2A, ERCC2, ERCC4, ERCC6, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOC3L2, EXOC8, EXOSC3, EXOSC5, EXOSC8, EXOSC9, EXT1, EXT2, FA2H, FAH, FAM149B1, FAR1, FARS2, FASTKD2, FBP1, FBXL4, FBXO7, FDXR, FECH, FGF14, FGFR2, FH, FITM2, FKRP, FKTN, FLAD1, FLVCR1, FMO3, FMR1, FOLR1, FOXG1, FOXP2, FOXRED1, FRMD4A, FRMD5, FTCD, FTH1, FTL, FUCA1, FUT8, FXN, G6PC1, G6PC3, GAA, GABRA1, GABRG2, GAD1, GALC, GALE, GALK1, GALNS, GALNT2, GALNT3, GALT, GAMT, GARS1, GATM, GBA1, GBA2, GBE1, GCDH, GCH1, GCLC, GDAP1, GDAP2, GEMIN5, GFAP, GFER, GFM1, GFPT1, GIGYF2, GJA1, GJC2, GK, GLA, GLB1, GLDC, GLI3, GLIS2, GLRA1, GLRB, GLRX5, GLS, GLUD1, GLUL, GLYCTK, GM2A, GMPPA, GMPPB, GNAL, GNAO1, GNB1, GNE, GNMT, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GOSR2, GPAA1, GPD1, GPHN, GPT2, GRHPR, GRID2, GRIN1, GRM1, GRN, GSN, GSS, GTPBP2, GTPBP3, GUSB, GYG1, GYS1, GYS2, HAAO, HACE1, HADH, HADHA, HADHB, HAMP, HARS1, HCCS, HCFC1, HECTD4, HECW2, HEXA, HEXB, HFE, HGD, HGSNAT, HIBCH, HIKESHI, HJV, HLCS, HMBS, HMGCL, HMGCS2, HNF1B, HNRNPH1, HOGA1, HPCA, HPD, HPDL, HPRT1, HPS1, HSD17B10, HSD17B4, HSD3B7, HSPD1, HTRA2, HTT, HYAL1, HYLS1, IARS2, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT54, IFT74, IFT80, INPP5E, INTS11, INVS, IQCB1, IRF2BPL, ISCU, ISG15, ITPA, ITPR1, ITSN1, IVD, JPH3, KARS1, KATNIP, KCNA1, KCNA2, KCNC1, KCNC3, KCND3, KCNJ10, KCNMA1, KCNN2, KCNQ2, KCNQ3, KCTD17, KCTD7, KDM5C, KIAA0586, KIAA0753, KIDINS220, KIF14, KIF1A, KIF1C, KIF5A, KIF7, KMT2B, KPNA3, KYNU, L1CAM, L2HGDH, LAMA1, LAMP2, LARGE1, LARS2, LBR, LCAT, LCT, LDHA, LDLR, LDLRAP1, LETM1, LFNG, LIAS, LIG3, LIPA, LIPC, LIPT1, LMBRD1, LNPK, LONP1, LPIN1, LPL, LRPPRC, LRRK2, LYST, LZTFL1, MAG, MAGT1, MAL, MAN1B1, MAN2B1, MANBA, MAOA, MAPK8IP3, MAPKBP1, MAPT, MARS1, MARS2, MAT1A, MCCC1, MCCC2, MCEE, MCOLN1, MDH2, MECR, MED27, MFF, MFN2, MFSD8, MGAT2, MGME1, MINPP1, MKKS, MKS1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MME, MMUT, MOCS1, MOCS2, MOGS, MORC2, MPDU1, MPI, MPV17, MRE11, MRPL3, MRPS22, MRPS34, MSMO1, MSTO1, MT-ATP6, MTCL1, MTFMT, MTHFR, MT-ND1, MT-ND6, MTO1, MTPAP, MTR, MTRFR, MTRR, MT-TK, MTTP, MVK, MYORG, NAA60, NAGA, NAGLU, NAGS, NARS2, NAXE, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB11, NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEK1, NEK8, NEU1, NFASC, NFU1, NGLY1, NHLRC1, NIPA1, NKX2-1, NKX6-2, NOP56, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPTX1, NR4A2, NRCAM, NSDHL, NSRP1, NT5C2, NT5C3A, NUBPL, NUP62, NUS1, OAT, OCLN, OCRL, OFD1, OGDHL, OPA1, OPA3, OPHN1, OPLAH, OTC, OXCT1, PACS2, PAH, PANK2, PARK7, PARS2, PAX2, PAX6, PC, PCBD1, PCCA, PCCB, PCDH12, PCDH19, PCK1, PCLO, PCSK9, PCYT2, PDE10A, PDE2A, PDE6D, PDE8B, PDGFB, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDX1, PDYN, PEPD, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGAP1, PGAP2, PGAP3, PGK1, PGM1, PGM3, PHGDH, PHKA1, PHKA2, PHKB, PHKG2, PHYH, PI4KA, PIBF1, PIGA, PIGL, PIGM, PIGN, PIGO, PIGS, PIGT, PIGV, PIGW, PINK1, PITRM1, PITX3, PKD1, PKD2, PKHD1, PLA2G6, PLP1, PMM2, PMPCA, PMPCB, PNKD, PNKP, PNP, PNPLA6, PNPO, PNPT1, POC1B, POLG, POLG2, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POR, POU4F1, PPA2, PPFIBP1, PPOX, PPP2R2B, PPP2R5D, PPT1, PRDM13, PRDX3, PRICKLE1, PRKAG2, PRKCG, PRKN, PRKRA, PRNP, PRODH, PRPS1, PRRT2, PSAP, PSAT1, PSEN1, PSPH, PTEN, PTF1A, PTRH2, PTRHD1, PTS, PUM1, PUS1, PYCR1, PYGL, PYGM, QDPR, RAB32, RAB39B, RAB3GAP2, RANBP2, RARS2, RBCK1, RBP4, REEP1, REEP2, RELN, RETREG1, RFC1, RFT1, RFXANK, RINT1, RMND1, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF170, RNF216, RNF220, RNU7-1,

ROBO3, RORA, RPGRIP1L, RPIA, RPL10, RRM2B, RTN2, RXYLT1, RYR1, SACS, SAMD12, SAMD9L, SAMHD1, SAR1B, SARS2, SBDS, SC5D, SCLT1, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SCO1, SCO2, SCP2, SCYL1, SDCCAG8, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SEPSECS, SERAC1, SETX, SGCE, SGSH, SHQ1, SI, SIL1, SKIC2, SKIC3, SLC12A3, SLC16A1, SLC16A2, SLC17A5, SLC18A2, SLC19A2, SLC19A3, SLC1A3, SLC1A4, SLC20A2, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC25A46, SLC2A1, SLC2A2, SLC30A10, SLC30A9, SLC33A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A14, SLC39A4, SLC39A8, SLC3A1, SLC40A1, SLC44A1, SLC46A1, SLC52A2, SLC52A3, SLC5A1, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SLC7A7, SLC7A9, SLC9A1, SLC9A6, SMPD1, SMPD4, SNAP25, SNCA, SNCB, SNORD118, SNX14, SPART, SPAST, SPG11, SPG21, SPG7, SPR, SPTAN1, SPTBN2, SPTLC1, SPTLC2, SPTSSA, SQSTM1, SRD5A3, SSR4, ST3GAL3, ST3GAL5, STN1, STS, STT3A, STUB1, STXBP1, SUCLA2, SUCLG1, SUFU, SUMF1, SUOX, SURF1, SVBP, SYNE1, SYNGAP1, SYNJ1, SYT1, TACO1, TAF1, TAF8, TAFAZZIN, TALDO1, TANGO2, TAPT1, TARS2, TAT, TBC1D23, TBC1D24, TBC1D32, TBK1, TBP, TCN2, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TECPR2, TENM4, TERT, TFG, TFR2, TGM6, TH, THAP1, THAP11, THG1L, TIMM50, TIMM8A, TINF2, TK2, TMEM106B, TMEM107, TMEM126B, TMEM138, TMEM151A, TMEM165, TMEM175, TMEM216, TMEM218, TMEM230, TMEM231, TMEM237, TMEM240, TMEM63C, TMEM67, TMEM70, TOE1, TOGARAM1, TOR1A, TPK1, TPP1, TRAPPC11, TREM2, TREX1, TRIM37, TRIP4, TRMU, TRNT1, TRPM6, TSEN15, TSEN2, TSEN34, TSEN54, TSFM, TSPOAP1, TTBK2, TTC19, TTC21B, TTC8, TTPA, TUBA1A, TUBA4A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUFM, TUSC3, TWNK, TXNDC15, TYMP, UBA5, UBAP1, UBE3A, UBTF, UCHL1, UGT1A1, UMOD, UMPS, UQCRQ, UROC1, UROD, UROS, VAC14, VAMP1, VAMP2, VARS2, VIPAS39, VKORC1, VLDLR, VMA12, VMA22, VPS11, VPS13A, VPS13B, VPS13D, VPS16, VPS33B, VPS35, VPS41, VPS4A, VPS53, VRK1, WASHC5, WDPCP, WDR19, WDR35, WDR45, WDR45B, WDR73, WDR81, WFS1, WWOX, XDH, XK, XPNPEP3, XPR1, XYLT1, XYLT2, YARS2, YIF1B, YY1, ZEB2, ZFHX3, ZFYVE26, ZNF423, ZSWIM6

Movement Disorders and Spasticity | 1167 genes

The movement disorders and spasticity panel includes genes associated with disorders such as ataxia, hereditary spastic paraplegia, hyperkinetic movement disorders and Parkinson's disease. Variants in the genes *ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, C9orf72, CACNA1A, CSTB, DMPK, FMR1, FXN, HTT, JPH3, NOP56, PPP2R2B, PRNP, TBP* are interrogated as sequence variants. If there is clinical suspicion of a repeat expansion disorder associated with one or more of the abovementioned genes, the Genome version of this panel is recommended.

Genes Included

AAAS, AARS1, AARS2, AASS, ABAT, ABCA1, ABCA2, ABCB11, ABCB4, ABCB7, ABCD1, ABCD4, ABCG5, ABCG8, ABHD12, ABHD16A, ABHD5, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACBD6, ACER3, ACO2, ACOX1, ACSF3, ACTB, ACY1, ADA, ADAR, ADCY5, ADGRG1, ADPRS, ADSL, AFG2B, AFG3L2, AGA, AGK, AGL, AGPS, AGTPBP1, AGXT, AHI1, AIFM1, AIMP1, AKR1D1, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALK, ALMS1, ALPL, ALS2, AMACR, AMFR, AMN, AMPD2, AMT, ANKS6, ANO10, ANO3, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOA1, APOA5, APOB, APOC2, APOE, APRT, APTX, ARG1, ARL13B, ARL3, ARL6, ARL6IP1, ARMC9, ARSA, ARSB, ARSL, ARX, ASAH1, ASL, ASPA, ASS1, ATAD3A, ATCAY, ATG7, ATIC, ATL1, ATM, ATN1, ATP13A2, ATP1A2, ATP1A3, ATP2B3, ATP5MC3, ATP6AP1, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP7A, ATP7B, ATP8A2, ATP8B1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, AUH, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, B9D1, B9D2, BAAT, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCAS3, BCKDHA, BCKDHB, BCKDK, BCS1L, BEAN1, BICD2, BLOC1S1, BOLA3, BORCS8, BRF1, BSCL2, BTD, C19orf12, C1QBP, C2CD3, C9orf72, CA5A, CA8, CACNA1A, CACNA1G, CACNA2D2, CAD, CAMTA1, CAPN1, CAPRIN1, CASK, CAT, CBS, CBY1, CC2D2A, CCDC88C, CDK5, CENPF, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFAP410, CFAP43, CHCHD10, CHCHD2, CHKB, CHMP1A, CHMP2B, CHST14, CHST3, CHST6, CHSY1, CILK1, CISD2, CIZ1, CLCN2, CLDN11, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CNNM2, COA3, COA6, COA7, COA8, COASY, COG1, COG2, COG3, COG4, COG5, COG6, COG7, COG8, COL6A3, COQ2, COQ4, COQ6, COQ8A, COQ8B, COQ9, COX10, COX15, COX20, COX6A1, COX6B1, COX7B, COXFA4, CP, CPLANE1, CPOX, CPS1, CPT1A, CPT1C, CPT2, CRB2, CRPPA, CSF1R, CSGALNACT1, CSPP1, CSTB, CTBP1, CTH, CTNNB1, CTNS, CTSA, CTSC, CTSD, CTSK, CUBN, CWF19L1, CYC1, CYCS, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DAGLA, DARS1, DARS2, DBH, DBT, DCAF17, DCC, DCDC2, DCTN1, DCXR, DDC, DDHD1, DDHD2, DDOST, DDX3X, DDX59, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHODH, DHRSX, DHTKD1, DKC1, DLAT, DLD, DLG4, DMPK, DMXL2, DNA2, DNAH1, DNAJC12, DNAJC19, DNAJC3, DNAJC5, DNAJC6, DNM1L, DNMT1, DOCK3, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DPYS, DPYSL5, DSTYK, DYM, DYNC1H1, DYNC2H1, DYNC2I2, DYNC2LI1, DYNLT2B, EARS2, EBF3, EBP, ECHS1, EDEM3, EEF2, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4G1, ELAC2, ELOVL1, ELOVL4, ELOVL5, ENO3, ENTPD1, EOGT, EPG5, EPM2A, ERCC2, ERCC4, ERCC6, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOC3L2, EXOC8, EXOSC3, EXOSC5, EXOSC8, EXOSC9, EXT1, EXT2, FA2H, FAH, FAM149B1, FAR1, FARS2, FASTKD2, FBP1, FBXL4, FBXO7, FDXR, FECH, FGF14, FGFR2, FH, FITM2, FKRP, FKTN, FLAD1, FLVCR1, FMO3, FMR1, FOLR1, FOXG1, FOXP2, FOXRED1, FRMD4A, FRMD5, FTCD, FTH1, FTL, FUCA1, FUT8, FXN, G6PC1, G6PC3, GAA, GABRA1, GABRG2, GAD1, GALC, GALE, GALK1, GALNS, GALNT2, GALNT3, GALT, GAMT, GARS1, GATM, GBA1, GBA2, GBE1, GCDH, GCH1, GCLC, GDAP1, GDAP2, GEMIN5, GFAP, GFER, GFM1, GFPT1, GIGYF2, GJA1, GJC2, GK, GLA, GLB1, GLDC, GLI3, GLIS2, GLRA1, GLRB, GLRX5, GLS, GLUD1, GLUL, GLYCTK, GM2A, GMPPA, GMPPB, GNAL, GNAO1, GNB1, GNE, GNMT, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GOSR2, GPAA1, GPD1, GPHN, GPT2, GRHPR, GRID2, GRIN1, GRM1, GRN, GSN, GSS, GTPBP2, GTPBP3, GUSB, GYG1, GYS1, GYS2, HAAO, HACE1, HADH, HADHA, HADHB, HAMP, HARS1, HCCS, HCFC1, HECTD4, HECW2, HEXA, HEXB, HFE, HGD, HGSNAT, HIBCH, HIKESHI, HJV, HLCS, HMBS, HMGCL, HMGCS2, HNF1B, HNRNPH1, HOGA1, HPCA, HPD, HPDL, HPRT1, HPS1, HSD17B10, HSD17B4, HSD3B7, HSPD1, HTRA2, HTT, HYAL1, HYLS1, IARS2, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, IFT122, IFT140,

IFT172, IFT27, IFT43, IFT52, IFT54, IFT74, IFT80, INPP5E, INTS11, INVS, IQCB1, IRF2BPL, ISCU, ISG15, ITPA, ITPR1, ITSN1, IVD, JPH3, KARS1, KATNIP, KCNA1, KCNA2, KCNC1, KCNC3, KCND3, KCNJ10, KCNMA1, KCNN2, KCNQ2, KCNQ3, KCTD17, KCTD7, KDM5C, KIAA0586, KIAA0753, KIDINS220, KIF14, KIF1A, KIF1C, KIF5A, KIF7, KMT2B, KPNA3, KYNU, L1CAM, L2HGDH, LAMA1, LAMP2, LARGE1, LARS2, LBR, LCAT, LCT, LDHA, LDLR, LDLRAP1, LETM1, LFNG, LIAS, LIG3, LIPA, LIPC, LIPT1, LMBRD1, LNPK, LONP1, LPIN1, LPL, LRPPRC, LRRK2, LYST, LZTFL1, MAG, MAGT1, MAL, MAN1B1, MAN2B1, MANBA, MAOA, MAPK8IP3, MAPKBP1, MAPT, MARS1, MARS2, MAT1A, MCCC1, MCCC2, MCEE, MCOLN1, MDH2, MECR, MED27, MFF, MFN2, MFSD8, MGAT2, MGME1, MINPP1, MKKS, MKS1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MME, MMUT, MOCS1, MOCS2, MOGS, MORC2, MPDU1, MPI, MPV17, MRE11, MRPL3, MRPS22, MRPS34, MSMO1, MSTO1, MT-ATP6, MTCL1, MTFMT, MTHFR, MT-ND1, MT-ND6, MTO1, MTPAP, MTR, MTRFR, MTRR, MT-TK, MTTP, MVK, MYORG, NAA60, NAGA, NAGLU, NAGS, NARS2, NAXE, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB11, NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEK1, NEK8, NEU1, NFASC, NFU1, NGLY1, NHLRC1, NIPA1, NKX2-1, NKX6-2, NOP56, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NPTX1, NR4A2, NRCAM, NSDHL, NSRP1, NT5C2, NT5C3A, NUBPL, NUP62, NUS1, OAT, OCLN, OCRL, OFD1, OGDHL, OPA1, OPA3, OPHN1, OPLAH, OTC, OXCT1, PACS2, PAH, PANK2, PARK7, PARS2, PAX2, PAX6, PC, PCBD1, PCCA, PCCB, PCDH12, PCDH19, PCK1, PCLO, PCSK9, PCYT2, PDE10A, PDE2A, PDE6D, PDE8B, PDGFB, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDX1, PDYN, PEPD, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGAP1, PGAP2, PGAP3, PGK1, PGM1, PGM3, PHGDH, PHKA1, PHKA2, PHKB, PHKG2, PHYH, PI4KA, PIBF1, PIGA, PIGL, PIGM, PIGN, PIGO, PIGS, PIGT, PIGV, PIGW, PINK1, PITRM1, PITX3, PKD1, PKD2, PKHD1, PLA2G6, PLP1, PMM2, PMPCA, PMPCB, PNKD, PNKP, PNP, PNPLA6, PNPO, PNPT1, POC1B, POLG, POLG2, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POR, POU4F1, PPA2, PPFIBP1, PPOX, PPP2R2B, PPP2R5D, PPT1, PRDM13, PRDX3, PRICKLE1, PRKAG2, PRKCG, PRKN, PRKRA, PRNP, PRODH, PRPS1, PRRT2, PSAP, PSAT1, PSEN1, PSPH, PTEN, PTF1A, PTRH2, PTRHD1, PTS, PUM1, PUS1, PYCR1, PYGL, PYGM, QDPR, RAB32, RAB39B, RAB3GAP2, RANBP2, RARS2, RBCK1, RBP4, REEP1, REEP2, RELN, RETREG1, RFC1, RFT1, RFXANK, RINT1, RMND1, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF170, RNF216, RNF220, ROBO3, RORA, RPGRIP1L, RPIA, RPL10, RRM2B, RTN2, RXYLT1, RYR1, SACS, SAMD12, SAMD9L, SAMHD1, SAR1B, SARS2, SBDS, SC5D, SCLT1, SCN1A, SCN1B, SCN2A, SCN8A, SCN9A, SCO1, SCO2, SCP2, SCYL1, SDCCAG8, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SEPSECS, SERAC1, SETX, SGCE, SGSH, SHQ1, SI, SIL1, SKIC2, SKIC3, SLC12A3, SLC16A1, SLC16A2, SLC17A5, SLC18A2, SLC19A2, SLC19A3, SLC1A3, SLC1A4, SLC20A2, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC25A46, SLC2A1, SLC2A2, SLC30A10, SLC30A9, SLC33A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A14, SLC39A4, SLC39A8, SLC3A1, SLC40A1, SLC44A1, SLC46A1, SLC52A2, SLC52A3, SLC5A1, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SLC7A7, SLC7A9, SLC9A1, SLC9A6, SMPD1, SMPD4, SNAP25, SNCA, SNCB, SNX14, SPART, SPAST, SPG11, SPG21, SPG7, SPR, SPTAN1, SPTBN2, SPTLC1, SPTLC2, SPTSSA, SQSTM1, SRD5A3, SSR4, ST3GAL3, ST3GAL5, STN1, STS, STT3A, STUB1, STXBP1, SUCLA2, SUCLG1, SUFU, SUMF1, SUOX, SURF1, SVBP, SYNE1, SYNGAP1, SYNJ1, SYT1, TACO1, TAF1, TAF8, TAFAZZIN, TALDO1, TANGO2, TAPT1, TARS2, TAT, TBC1D23, TBC1D24, TBC1D32, TBK1, TBP, TCN2, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TECPR2, TENM4, TERT, TFG, TFR2, TGM6, TH, THAP1, THAP11, THG1L, TIMM50, TIMM8A, TINF2, TK2, TMEM106B, TMEM107, TMEM126B, TMEM138, TMEM151A, TMEM165, TMEM175, TMEM216, TMEM218, TMEM230, TMEM231, TMEM237, TMEM240, TMEM63C, TMEM67, TMEM70, TOE1, TOGARAM1, TOR1A, TPK1, TPP1, TRAPPC11, TREM2, TREX1, TRIM37, TRIP4, TRMU, TRNT1, TRPM6, TSEN15, TSEN2, TSEN34, TSEN54, TSFM, TSPOAP1, TTBK2, TTC19, TTC21B, TTC8, TTPA, TUBA1A, TUBA4A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUFM, TUSC3, TWNK, TXNDC15, TYMP, UBA5, UBAP1, UBE3A, UBTF, UCHL1, UGT1A1, UMOD, UMPS, UQCRQ, UROC1, UROD, UROS, VAC14, VAMP1, VAMP2, VARS2, VIPAS39, VKORC1, VLDLR, VMA12, VMA22, VPS11, VPS13A, VPS13B, VPS13D, VPS16, VPS33B, VPS35, VPS41, VPS4A, VPS53, VRK1, WASHC5, WDPCP, WDR19, WDR35, WDR45, WDR45B, WDR73, WDR81, WFS1, WWOX, XDH, XK, XPNPEP3, XPR1, XYLT1, XYLT2, YARS2, YIF1B, YY1, ZEB2, ZFHX3, ZFYVE26, ZNF423, ZSWIM6

Parkinson's Disease Upgraded Genome | 67 genes

The Parkinson disease (PD) panel Upgraded Genome identifies all relevant pathophysiologically genetic variants for the development and treatment of PD. Characteristic features of PD include neuronal loss in specific areas of the substantia nigra and widespread intracellular protein synuclein accumulation. The disease is characterized by three core motor symptoms: tremor, muscle rigidity, and bradykinesia. This panel is based on genome sequencing, and it includes repeat expansion screening of the genes *ATN1, ATXN1, ATXN2, ATXN3, C9orf72, HTT, JPH3, PPP2R2B, TBP* and *GBA1* conversion analysis.

> Recommended panel approach based on genome sequencing, including repeat expansion screening of the genes *ATN1, ATXN1, ATXN2, ATXN3, C9orf72, HTT, JPH3, PPP2R2B, TBP* and *GBA1* conversion analysis with its pseudogene.

Genes Included

AN03, ARSA, ATN1, ATP13A2, ATP1A3, ATP6AP2, ATXN1, ATXN2, ATXN3, C19orf12, C9orf72, CHCHD2, COASY, CSF1R, DCTN1, DNAJC6, EIF4G1, FBXO7, FTL, GBA1, GCH1, GIGYF2, GNAL, GRN, HTRA2, HTT, ITSN1, JPH3, LRRK2, LYST, MAPT, NR4A2, OPA3, PANK2, PARK7, PDE8B, PDGFB, PINK1, PLA2G6, PPP2R2B, PPP2R5D, PRKN, PRKRA, PTRHD1, RAB32, RAB39B, SGCE, SLC30A10, SLC39A14, SLC6A3, SNCA, SNCB, SPG11, SPR, SYNJ1, TAF1, TBP, TH, THAP1, TMEM175, TMEM230, TOR1A, TUBB4A, UCHL1, VPS13A, VPS35, WDR45

Parkinson's Disease Upgraded | 67 genes

The Parkinson disease (PD) panel Upgraded identifies all relevant pathophysiologically genetic variants for the development and treatment of PD. Characteristic features of PD include neuronal loss in specific areas of the substantia nigra and widespread intracellular protein synuclein accumulation. The disease is characterized by three core motor symptoms: tremor, muscle rigidity, and bradykinesia. Variants in the genes ATN1, ATXN1, ATXN2, ATXN3, C9orf72, HTT, JPH3, PPP2R2B, TBP are interrogated as sequence variants. If there is clinical suspicion of a repeat expansion disorder associated with one or more of the abovementioned genes, the Genome version of this panel is recommended.

Genes Included

ANO3, ARSA, ATN1, ATP13A2, ATP1A3, ATP6AP2, ATXN1, ATXN2, ATXN3, C19orf12, C9orf72, CHCHD2, COASY, CSF1R, DCTN1, DNAJC6, EIF4G1, FBXO7, FTL, GBA1, GCH1, GIGYF2, GNAL, GRN, HTRA2, HTT, ITSN1, JPH3, LRRK2, LYST, MAPT, NR4A2, OPA3, PANK2, PARK7, PDE8B, PDGFB, PINK1, PLA2G6, PPP2R2B, PPP2R5D, PRKN, PRKRA, PTRHD1, RAB32, RAB39B, SGCE, SLC30A10, SLC39A14, SLC6A3, SNCA, SNCB, SPG11, SPR, SYNJ1, TAF1, TBP, TH, THAP1, TMEM175, TMEM230, TOR1A, TUBB4A, UCHL1, VPS13A, VPS35, WDR45

Hyperkinetic Movement Disorders Genome | 955 genes

The hyperkinetic movement disorders panel Genome includes genes associated with dystonia (e.g., *TOR1A, KMT2B*), Chorea (e.g., Huntington's disease, chorea-acanthocytosis), myoclonus. This panel includes repeat expansion screening of the *ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, C9orf72, CACNA1A, CSTB, DMPK, FXN, HTT, JPH3, NOP56, PPP2R2B, PRNP, TBP* genes.

> Recommended panel approach based on genome sequencing, including repeat expansion screening of the genes *ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, C9orf72, CACNA1A, CSTB, DMPK, FXN, HTT, JPH3, NOP56, PPP2R2B, PRNP, TBP. The panel includes the non-coding genes RNU7-1 and SNORD118.*

Genes Included

AAAS, AARS2, AASS, ABAT, ABCA1, ABCB11, ABCB4, ABCB7, ABCD1, ABCD4, ABCG5, ABCG8, ABHD12, ABHD5, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACBD6, ACER3, ACO2, ACOX1, ACSF3, ACTB, ACY1, ADA, ADAR, ADCY5, ADGRG1, ADSL, AFG2B, AFG3L2, AGA, AGK, AGL, AGPS, AGXT, AHI1, AIFM1, AKR1D1, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG3, ALG6, ALG8, ALG9, ALMS1, ALPL, AMACR, AMN, AMPD2, AMT, ANKS6, ANO10, ANO3, AP1S2, APOA1, APOA5, APOB, APOC2, APOE, APRT, APTX, ARG1, ARL13B, ARL6, ARSA, ARSB, ARSL, ARX, ASAH1, ASL, ASPA, ASS1, ATAD3A, ATCAY, ATIC, ATM, ATN1, ATP13A2, ATP1A2, ATP1A3, ATP5MC3, ATP6AP1, ATP6AP2, ATP6V0A2, ATP7A, ATP7B, ATP8A2, ATP8B1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, AUH, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALT1, B4GALT7, B4GAT1, B9D2, BAAT, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCKDHA, BCKDHB, BCKDK, BCS1L, BOLA3, BTD, C19orf12, C1QBP, C2CD3, C9orf72, CA5A, CA8, CACNA1A, CACNA1G, CAMTA1, CASK, CAT, CBS, CC2D2A, CENPF, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFAP410, CFAP43, CHCHD10, CHCHD2, CHKB, CHMP1A, CHMP2B, CHST14, CHST3, CHST6, CHSY1, CILK1, CISD2, CIZ1, CLCN2, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CLPB, CLPP, CNNM2, COA3, COA6, COA8, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL6A3, COQ2, COQ4, COQ6, COQ8A, COQ8B, COQ9, COX10, COX15, COX20, COX6A1, COX6B1, COX7B, COXFA4, CP, CPLANE1, CPOX, CPS1, CPT1A, CPT2, CRB2, CRPPA, CSF1R, CSPP1, CSTB, CTH, CTNS, CTSA, CTSC, CTSD, CTSK, CUBN, CWF19L1, CYC1, CYCS, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DBH, DBT, DCAF17, DCC, DCDC2, DCTN1, DCXR, DDC, DDHD2, DDX59, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHODH, DHTKD1, DKC1, DLAT, DLD, DMPK, DMXL2, DNA2, DNAH1, DNAJC12, DNAJC19, DNAJC5, DNAJC6, DNM1L, DNMT1, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DPYS, DYM, DYNC2H1, DYNC2I2, DYNC2LI1, DYNLT2B, EARS2, EBP, ECHS1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4G1, ELAC2, ELOVL4, ENO3, EPG5, EPM2A, ERCC6, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EXT1, EXT2, FA2H, FAH, FAR1, FARS2, FASTKD2, FBP1, FBXL4, FBXO7, FDXR, FECH, FGF14, FGFR2, FH, FITM2, FKRP, FKTN, FLAD1, FLVCR1, FMO3, FOLR1, FOXG1, FOXP2, FOXRED1, FTCD, FTL, FUCA1, FUT8, FXN, G6PC1, G6PC3, GAA, GABRA1, GABRG2, GALC, GALE, GALK1, GALNS, GALNT3, GALT, GAMT, GARS1, GATM, GBA1, GBA2, GBE1, GCDH, GCH1, GCLC, GDAP1, GFAP, GFER, GFM1, GFPT1, GIGYF2, GJC2, GK, GLA, GLB1, GLDC, GLI3, GLIS2, GLRA1, GLRB, GLRX5, GLUD1, GLUL, GLYCTK, GM2A, GMPPB, GNAL, GNAO1, GNB1, GNE, GNMT, GNPAT, GNPTAB, GNPTG, GNS, GOSR2, GPAA1, GPD1, GPHN, GRHPR, GRID2, GRIN1, GRM1, GRN, GSS, GTPBP2, GTPBP3, GUSB, GYG1, GYS1, GYS2, HAAO, HADH, HADHA, HADHB, HAMP, HCCS, HCFC1, HECW2, HEXA, HEXB, HFE, HGD, HGSNAT, HIBCH, HJV, HLCS, HMBS, HMGCL, HMGCS2, HNF1B, HNRNPH1, HOGA1, HPCA, HPD, HPRT1, HPS1, HSD17B10, HSD17B4, HSD3B7, HSPD1, HTRA2, HTT, HYAL1, HYLS1, IARS2, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT54, IFT80, INPP5E, INVS, IQCB1, IRF2BPL, ISCU, ISG15, ITPA, ITPR1, IVD, JPH3, KARS1, KCNA1, KCNC1, KCNC3, KCND3, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCTD17, KCTD7, KIAA0586, KIF1A, KIF1C, KIF7, KMT2B, KYNU, L2HGDH, LAMP2, LARGE1, LARS2, LBR, LCAT, LCT, LDHA, LDLR, LDLRAP1, LIAS, LIPA, LIPC, LIPT1, LMBRD1, LONP1, LPIN1, LPL, LRPPRC, LRRK2, LYST, LZTFL1, MAGT1, MAL, MAN1B1, MAN2B1, MANBA, MAOA, MAPKBP1, MAPT, MARS2, MAT1A, MCCC1, MCCC2, MCEE, MCOLN1, MDH2, MECR, MED27, MFF, MFN2, MFSD8, MGAT2, MGME1, MKKS, MKS1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPDU1, MPI, MPV17, MRE11, MRPL3, MRPS22, MRPS34, MSMO1, MT-ATP6, MT-ND1, MT-ND6, MT-TK, MTFMT, MTHFR, MTO1, MTPAP, MTR, MTRFR, MTRR, MTTP, MVK, MYORG, NAGA, NAGLU, NAGS, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB11, NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8,

NDUFV1, NDUFV2, NEK1, NEK8, NEU1, NFU1, NGLY1, NHLRC1, NKX2-1, NKX6-2, NOP56, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NR4A2, NSDHL, NT5C3A, NUBPL, NUP62, NUS1, OAT, OCLN, OCRL, OFD1, OPA1, OPA3, OPHN1, OPLAH, OTC, OXCT1, PAH, PANK2, PARK7, PARS2, PAX6, PC, PCBD1, PCCA, PCCB, PCDH12, PCDH19, PCK1, PCSK9, PDE10A, PDE2A, PDGFB, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDX1, PDYN, PEPD, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGAP2, PGAP3, PGK1, PGM1, PGM3, PHGDH, PHKA1, PHKA2, PHKB, PHKG2, PHYH, PIGA, PIGL, PIGM, PIGN, PIGO, PIGT, PIGV, PINK1, PITX3, PKD1, PKD2, PKHD1, PLA2G6, PLP1, PMM2, PMPCA, PNKD, PNKP, PNP, PNPLA6, PNPO, PNPT1, POLG, POLG2, POLR3A, POMGNT1, POMGNT2, POMT1, POMT2, POR, PPA2, PPOX, PPP2R2B, PPP2R5D, PPT1, PRKAG2, PRKCG, PRKN, PRKRA, PRNP, PRODH, PRPS1, PRRT2, PSAP, PSAT1, PSEN1, PSPH, PTEN, PTF1A, PTS, PUS1, PYCR1, PYGL, PYGM, QDPR, RAB39B, RANBP2, RARS2, RBCK1, RBP4, RELN, RFT1, RMND1, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF170, RNF216, RNU7-1, ROBO3, RPGRIP1L, RPIA, RPL10, RRM2B, RXYLT1, RYR1, SACS, SAMD12, SAMHD1, SAR1B, SARS2, SBDS, SC5D, SCN1A, SCN1B, SCN8A, SCN9A, SCO1, SCO2, SCP2, SDCCAG8, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SEPSECS, SERAC1, SETX, SGCE, SGSH, SHQ1, SI, SIL1, SKIC2, SKIC3, SLC12A3, SLC16A1, SLC16A2, SLC17A5, SLC18A2, SLC19A2, SLC19A3, SLC1A3, SLC20A2, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC25A46, SLC2A1, SLC2A2, SLC30A10, SLC30A9, SLC35A1, SLC35A2, SLC35C1, SLC35D1, SLC37A4, SLC39A14, SLC39A4, SLC39A8, SLC3A1, SLC40A1, SLC46A1, SLC52A2, SLC52A3, SLC5A1, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SLC7A7, SLC7A9, SLC9A6, SMPD1, SMPD4, SNCA, SNORD118, SNX14, SPG11, SPG7, SPR, SPTBN2, SPTLC1, SPTLC2, SQSTM1, SRD5A3, SSR4, ST3GAL3, ST3GAL5, STS, STT3A, STUB1, STXBP1, SUCLA2, SUCLG1, SUFU, SUMF1, SUOX, SURF1, SYNE1, SYNGAP1, SYNJ1, SYT1, TACO1, TAF1, TAFAZZIN, TALDO1, TANGO2, TARS2, TAT, TBC1D24, TBK1, TBP, TCN2, TCTN1, TCTN2, TCTN3, TENM4, TERT, TFR2, TGM6, TH, THAP1, TIMM50, TIMM8A, TINF2, TK2, TMEM107, TMEM126B, TMEM138, TMEM151A, TMEM165, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TMEM70, TOE1, TOR1A, TPK1, TPP1, TREM2, TREX1, TRIM37, TRMU, TRNT1, TRPM6, TSEN2, TSEN34, TSEN54, TSFM, TSPOAP1, TTBK2, TTC19, TTC21B, TTC8, TTPA, TUBA1A, TUBB2B, TUBB3, TUBB4A, TUFM, TUSC3, TWNK, TXNDC15, TYMP, UBE3A, UBTF, UCHL1, UGT1A1, UMOD, UMPS, UQCRQ, UROC1, UROD, UROS, VAC14, VAMP1, VAMP2, VARS2, VIPAS39, VKORC1, VLDLR, VMA22, VPS11, VPS13A, VPS13B, VPS13D, VPS16, VPS33B, VPS35, VPS41, VPS4A, VPS53, VRK1, WDPCP, WDR19, WDR35, WDR45, WDR73, WDR81, WFS1, WWOX, XDH, XK, XPNPEP3, XPR1, XYLT1, XYLT2, YARS2, YIF1B, YY1, ZNF423, ZSWIM6

Hyperkinetic Movement Disorders | 953 genes

The hyperkinetic movement disorders panel includes genes associated with Dystonia (e.g., *TOR1A, KMT2B*), Chorea (e.g., Huntington's disease, chorea-acanthocytosis), myoclonus. Please note that in this panel, variants in the genes *ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, C9orf72, CACNA1A, CSTB, DMPK, FXN, HTT, JPH3, NOP56, PPP2R2B, PRNP, TBP* are only interrogated as sequence variants. If there is medical suspicion of a repeat expansion disorder associated with any of the abovementioned genes, the Genome version of this panel is recommended.

Genes Included

AAAS, AARS2, AASS, ABAT, ABCA1, ABCB11, ABCB4, ABCB7, ABCD1, ABCD4, ABCG5, ABCG8, ABHD12, ABHD5, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACBD6, ACER3, ACO2, ACOX1, ACSF3, ACTB, ACY1, ADA, ADAR, ADCY5, ADGRG1, ADSL, AFG2B, AFG3L2, AGA, AGK, AGL, AGPS, AGXT, AHI1, AIFM1, AKR1D1, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG3, ALG6, ALG8, ALG9, ALMS1, ALPL, AMACR, AMN, AMPD2, AMT, ANKS6, ANO10, ANO3, AP1S2, APOA1, APOA5, APOB, APOC2, APOE, APRT, APTX, ARG1, ARL13B, ARL6, ARSA, ARSB, ARSL, ARX, ASAH1, ASL, ASPA, ASS1, ATAD3A, ATCAY, ATIC, ATM, ATN1, ATP13A2, ATP1A2, ATP1A3, ATP5MC3, ATP6AP1, ATP6AP2, ATP6V0A2, ATP7A, ATP7B, ATP8A2, ATP8B1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, AUH, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALT1, B4GALT7, B4GAT1, B9D2, BAAT, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCKDHA, BCKDHB, BCKDK, BCS1L, BOLA3, BTD, C19orf12, C1QBP, C2CD3, C9orf72, CA5A, CA8, CACNA1A, CACNA1G, CAMTA1, CASK, CAT, CBS, CC2D2A, CENPF, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CFAP410, CFAP43, CHCHD10, CHCHD2, CHKB, CHMP1A, CHMP2B, CHST14, CHST3, CHST6, CHSY1, CILK1, CISD2, CIZ1, CLCN2, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CLPB, CLPP, CNNM2, COA3, COA6, COA8, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL6A3, COQ2, COQ4, COQ6, COQ8A, COQ8B, COQ9, COX10, COX15, COX20, COX6A1, COX6B1, COX7B, COXFA4, CP, CPLANE1, CPOX, CPS1, CPT1A, CPT2, CRB2, CRPPA, CSF1R, CSPP1, CSTB, CTH, CTNS, CTSA, CTSC, CTSD, CTSK, CUBN, CWF19L1, CYC1, CYCS, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DBH, DBT, DCAF17, DCC, DCDC2, DCTN1, DCXR, DDC, DDHD2, DDX59, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHODH, DHTKD1, DKC1, DLAT, DLD, DMPK, DMXL2, DNA2, DNAH1, DNAJC12, DNAJC19, DNAJC5, DNAJC6, DNM1L, DNMT1, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DPYS, DYM, DYNC2H1, DYNC2I2, DYNC2LI1, DYNLT2B, EARS2, EBP, ECHS1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF4G1, ELAC2, ELOVL4, ENO3, EPG5, EPM2A, ERCC6, ETFA, ETFB, ETFDH, ETHE1, EVC, EVC2, EXOSC3, EXT1, EXT2, FA2H, FAH, FAR1, FARS2, FASTKD2, FBP1, FBXL4, FBXO7, FDXR, FECH, FGF14, FGFR2, FH, FITM2, FKRP, FKTN, FLAD1, FLVCR1, FMO3, FOLR1, FOXG1, FOXP2, FOXRED1, FTCD, FTL, FUCA1, FUT8, FXN, G6PC1, G6PC3, GAA, GABRA1, GABRG2, GALC, GALE, GALK1, GALNS, GALNT3, GALT, GAMT, GARS1, GATM, GBA1, GBA2, GBE1, GCDH, GCH1, GCLC, GDAP1, GFAP, GFER, GFM1, GFPT1, GIGYF2, GJC2, GK, GLA, GLB1, GLDC, GLI3, GLIS2, GLRA1, GLRB, GLRX5, GLUD1, GLUL, GLYCTK, GM2A, GMPPB, GNAL, GNAO1, GNB1, GNE, GNMT, GNPAT, GNPTAB, GNPTG, GNS, GOSR2, GPAA1, GPD1, GPHN, GRHPR, GRID2, GRIN1, GRM1, GRN, GSS, GTPBP2, GTPBP3, GUSB, GYG1, GYS1, GYS2, HAAO, HADH, HADHA, HADHB, HAMP, HCCS, HCFC1, HECW2, HEXA, HEXB, HFE, HGD, HGSNAT, HIBCH, HJV, HLCS, HMBS, HMGCL, HMGCS2, HNF1B, HNRNPH1, HOGA1, HPCA, HPD, HPRT1, HPS1, HSD17B10, HSD17B4, HSD3B7, HSPD1, HTRA2, HTT, HYAL1, HYLS1, IARS2, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT54, IFT80, INPP5E, INVS, IQCB1, IRF2BPL, ISCU, ISG15, ITPA, ITPR1, IVD,

JPH3, KARS1, KCNA1, KCNC1, KCNC3, KCND3, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCTD17, KCTD7, KIAA0586, KIF1A, KIF1C, KIF7, KMT2B, KYNU, L2HGDH, LAMP2, LARGE1, LARS2, LBR, LCAT, LCT, LDHA, LDLR, LDLRAP1, LIAS, LIPA, LIPC, LIPT1, LMBRD1, LONP1, LPIN1, LPL, LRPPRC, LRRK2, LYST, LZTFL1, MAGT1, MAL, MAN1B1, MAN2B1, MANBA, MAOA, MAPKBP1, MAPT, MARS2, MAT1A, MCCC1, MCCC2, MCEE, MCOLN1, MDH2, MECR, MED27, MFF, MFN2, MFSD8, MGAT2, MGME1, MKKS, MKS1, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPDU1, MPI, MPV17, MRE11, MRPL3, MRPS22, MRPS34, MSMO1, MT-ATP6, MT-ND1, MT-ND6, MT-TK, MTFMT, MTHFR, MTO1, MTPAP, MTR, MTRFR, MTRR, MTTP, MVK, MYORG, NAGA, NAGLU, NAGS, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB11, NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEK1, NEK8, NEU1, NFU1, NGLY1, NHLRC1, NKX2-1, NKX6-2, NOP56, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NR4A2, NSDHL, NT5C3A, NUBPL, NUP62, NUS1, OAT, OCLN, OCRL, OFD1, OPA1, OPA3, OPHN1, OPLAH, OTC, OXCT1, PAH, PANK2, PARK7, PARS2, PAX6, PC, PCBD1, PCCA, PCCB, PCDH12, PCDH19, PCK1, PCSK9, PDE10A, PDE2A, PDGFB, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDX1, PDYN, PEPD, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGAP2, PGAP3, PGK1, PGM1, PGM3, PHGDH, PHKA1, PHKA2, PHKB, PHKG2, PHYH, PIGA, PIGL, PIGM, PIGN, PIGO, PIGT, PIGV, PINK1, PITX3, PKD1, PKD2, PKHD1, PLA2G6, PLP1, PMM2, PMPCA, PNKD, PNKP, PNP, PNPLA6, PNPO, PNPT1, POLG, POLG2, POLR3A, POMGNT1, POMGNT2, POMT1, POMT2, POR, PPA2, PPOX, PPP2R2B, PPP2R5D, PPT1, PRKAG2, PRKCG, PRKN, PRKRA, PRNP, PRODH, PRPS1, PRRT2, PSAP, PSAT1, PSEN1, PSPH, PTEN, PTF1A, PTS, PUS1, PYCR1, PYGL, PYGM, QDPR, RAB39B, RANBP2, RARS2, RBCK1, RBP4, RELN, RFT1, RMND1, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF170, RNF216, ROBO3, RPGRIP1L, RPIA, RPL10, RRM2B, RXYLT1, RYR1, SACS, SAMD12, SAMHD1, SAR1B, SARS2, SBDS, SC5D, SCN1A, SCN1B, SCN8A, SCN9A, SCO1, SCO2, SCP2, SDCCAG8, SDHA, SDHAF1, SDHAF2, SDHB, SDHC, SDHD, SEC23B, SEPSECS, SERAC1, SETX, SGCE, SGSH, SHQ1, SI, SIL1, SKIC2, SKIC3, SLC12A3, SLC16A1, SLC16A2, SLC17A5, SLC18A2, SLC19A2, SLC19A3, SLC1A3, SLC20A2, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC25A46, SLC2A1, SLC2A2, SLC30A10, SLC30A9, SLC35A1, SLC35A2, SLC35C1, SLC35D1, SLC37A4, SLC39A14, SLC39A4, SLC39A8, SLC3A1, SLC40A1, SLC46A1, SLC52A2, SLC52A3, SLC5A1, SLC6A19, SLC6A3, SLC6A5, SLC6A8, SLC7A7, SLC7A9, SLC9A6, SMPD1, SMPD4, SNCA, SNX14, SPG11, SPG7, SPR, SPTBN2, SPTLC1, SPTLC2, SQSTM1, SRD5A3, SSR4, ST3GAL3, ST3GAL5, STS, STT3A, STUB1, STXBP1, SUCLA2, SUCLG1, SUFU, SUMF1, SUOX, SURF1, SYNE1, SYNGAP1, SYNJ1, SYT1, TACO1, TAF1, TAFAZZIN, TALDO1, TANGO2, TARS2, TAT, TBC1D24, TBK1, TBP, TCN2, TCTN1, TCTN2, TCTN3, TENM4, TERT, TFR2, TGM6, TH, THAP1, TIMM50, TIMM8A, TINF2, TK2, TMEM107, TMEM126B, TMEM138, TMEM151A, TMEM165, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TMEM70, TOE1, TOR1A, TPK1, TPP1, TREM2, TREX1, TRIM37, TRMU, TRNT1, TRPM6, TSEN2, TSEN34, TSEN54, TSFM, TSPOAP1, TTBK2, TTC19, TTC21B, TTC8, TTPA, TUBA1A, TUBB2B, TUBB3, TUBB4A, TUFM, TUSC3, TWNK, TXNDC15, TYMP, UBE3A, UBTF, UCHL1, UGT1A1, UMOD, UMPS, UQCRQ, UROC1, UROD, UROS, VAC14, VAMP1, VAMP2, VARS2, VIPAS39, VKORC1, VLDLR, VMA22, VPS11, VPS13A, VPS13B, VPS13D, VPS16, VPS33B, VPS35, VPS41, VPS4A, VPS53, VRK1, WDPCP, WDR19, WDR35, WDR45, WDR73, WDR81, WFS1, WWOX, XDH, XK, XPNPEP3, XPR1, XYLT1, XYLT2, YARS2, YIF1B, YY1, ZNF423, ZSWIM6

Ataxia panel Genome | 470 genes

The ataxia panel Genome includes genes associated with early and late onset type ataxias and also repeat expansion forms of ataxia. The panel includes repeat expansion screening of the *ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, CACNA1A, CSTB, FMR1, FXN, HTT, NOP56, PPP2R2B, PRNP, TBP* genes.

> Recommended panel approach based on genome sequencing, including repeat expansion screening of the genes *ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, CACNA1A, CSTB, FMR1, FXN, HTT, NOP56, PPP2R2B, PRNP, TBP.*

Genes Included

AAAS, AARS1, ABCA2, ABCB7, ABHD12, ACBD6, ACO2, ADCY5, ADGRG1, ADPRS, AFG3L2, AGTPBP1, AHI1, ALAS2, ALDH5A1, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, AMPD2, ANO10, AP1S2, APTX, ARL13B, ARL3, ARMC9, ARSA, ASL, ATAD3A, ATCAY, ATG7, ATM, ATN1, ATP1A2, ATP1A3, ATP2B3, ATP6AP1, ATP6V0A1, ATP6V0A2, ATP7B, ATP8A2, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, AUH, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, B9D1, B9D2, BBS1, BEAN1, BRF1, C2CD3, CA8, CACNA1A, CACNA1G, CACNA2D2, CAD, CAMTA1, CAPN1, CAPRIN1, CASK, CBY1, CC2D2A, CCDC88C, CDK5, CENPF, CEP104, CEP290, CEP41, CHMP1A, CHST14, CHST3, CHST6, CHSY1, CILK1, CLCN2, CLN5, CLN6, CLP1, CLPP, COA7, COASY, COG1, COG2, COG3, COG4, COG5, COG6, COG7, COG8, COQ4, COQ8A, COX20, CP, CPLANE1, CRB2, CRPPA, CSGALNACT1, CSPP1, CSTB, CTBP1, CWF19L1, CYP27A1, CYP2U1, DAG1, DAGLA, DARS2, DCC, DDHD2, DDOST, DDX59, DHCR7, DHDDS, DHRSX, DKC1, DLG4, DMXL2, DNAJC19, DNAJC3, DNAJC5, DNMT1, DOCK3, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYSL5, DYNC1H1, EBF3, EDEM3, EEF2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, EOGT, EPM2A, ERCC2, ERCC4, EVC, EVC2, EXOC3L2, EXOC8, EXOSC3, EXOSC5, EXOSC8, EXOSC9, EXT1, EXT2, FA2H, FAM149B1, FBXL4, FDXR, FGF14, FKRP, FKTN, FLVCR1, FMR1, FOLR1, FRMD4A, FRMD5, FTH1, FUT8, FXN, G6PC3, GALC, GALNT2, GALNT3, GBA2, GDAP2, GEMIN5, GFAP, GFPT1, GJC2, GLI3, GLRA1, GLRB, GLS, GMPPA, GMPPB, GNE, GORAB, GOSR2, GPAA1, GRID2, GRM1, GRN, GSN, HARS1, HEXA, HEXB, HMBS, HTT, HYLS1, IFT74, INPP5E, INTS11, IRF2BPL, ITPR1, KATNIP, KCNA1, KCNA2, KCNC3, KCND3, KCNJ10, KCNN2, KCNQ2, KCNQ3, KIAA0586, KIAA0753, KIF14, KIF1A, KIF1C, KIF7, LAMA1, LARGE1, LARS2, LETM1, LFNG, LIG3, LNPK, MAG, MAGT1, MAN1B1, MAPK8IP3, MARS2, MFN2, MFSD8, MGAT2, MINPP1, MKS1, MMACHC, MME, MOGS, MORC2, MPDU1, MPI, MRE11, MSTO1, MT-ATP6, MTCL1, MTFMT, MTPAP, MTTP, MVK, NAA60, NAGLU, NAXE, NFASC, NGLY1, NHLRC1, NKX2-1, NKX6-2, NOP56, NPC1, NPC2, NPHP1, NPHP3, NPTX1, NUS1, OFD1, OGDHL, OPA1, OPA3, OPHN1, PACS2, PAX2, PAX6,

PCLO, PDE6D, PDYN, PEX16, PEX2, PEX6, PGAP2, PGAP3, PGM1, PGM3, PHGDH, PI4KA, PIBF1, PIGA, PIGL, PIGM, PIGN, PIGO, PIGS, PIGT, PIGV, PIGW, PITRM1, PLA2G6, PMM2, PMPCA, PMPCB, PNKD, PNKP, PNPLA6, PNPT1, POC1B, POLG, POLG2, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POU4F1, PPP2R2B, PRDM13, PRDX3, PRICKLE1, PRKCG, PRNP, PRPS1, PRRT2, PTF1A, PTRH2, PUM1, RARS2, RELN, RFC1, RFT1, RFXANK, RNF170, RNF216, RNF220, ROBO3, RORA, RPGRIP1L, RXYLT1, SACS, SAMD9L, SAR1B, SCLT1, SCN1A, SCN2A, SCN8A, SCN9A, SCYL1, SEC23B, SEPSECS, SETX, SIL1, SLC17A5, SLC1A3, SLC25A46, SLC2A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A8, SLC44A1, SLC52A2, SLC6A5, SLC9A1, SLC9A6, SMPD4, SNAP25, SNX14, SPG7, SPR, SPTAN1, SPTBN2, SQSTM1, SRD5A3, SSR4, ST3GAL3, ST3GAL5, STT3A, STUB1, SUFU, SVBP, SYNE1, SYNGAP1, TANGO2, TAPT1, TBC1D23, TBC1D32, TBP, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TECPR2, TERT, TGM6, THAP11, THG1L, TINF2, TMEM106B, TMEM107, TMEM138, TMEM165, TMEM216, TMEM218, TMEM231, TMEM237, TMEM240, TMEM67, TOE1, TOGARAM1, TPP1, TRAPPC11, TRIP4, TSEN15, TSEN2, TSEN34, TSEN54, TTBK2, TTC19, TTPA, TUBA1A, TUBA4A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUSC3, TWNK, TXNDC15, UBA5, UBTF, UCHL1, VAMP1, VLDLR, VMA12, VMA22, VPS13B, VPS13D, VPS41, VPS53, VRK1, WDR73, WDR81, WFS1, WWOX, XYLT1, XYLT2, ZFHX3, ZFYVE26, ZNF423, ZSWIM6

Ataxia | 470 genes

The ataxia panel includes genes associated with early and late onset type ataxias. Repeat expansion analysis is not part of this panel. If there is clinical suspicion of repeat expansion form of ataxia, the Genome version of this panel is recommended.

Genes Included

AAAS, AARS1, ABCA2, ABCB7, ABHD12, ACBD6, ACO2, ADCY5, ADGRG1, ADPRS, AFG3L2, AGTPBP1, AHI1, ALAS2, ALDH5A1, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, AMPD2, ANO10, AP1S2, APTX, ARL13B, ARL3, ARMC9, ARSA, ASL, ATAD3A, ATCAY, ATG7, ATM, ATN1, ATP1A2, ATP1A3, ATP2B3, ATP6AP1, ATP6V0A1, ATP6V0A2, ATP7B, ATP8A2, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, AUH, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, B9D1, B9D2, BBS1, BEAN1, BRF1, C2CD3, CA8, CACNA1A, CACNA1G, CACNA2D2, CAD, CAMTA1, CAPN1, CAPRIN1, CASK, CBY1, CC2D2A, CCDC88C, CDK5, CENPF, CEP104, CEP290, CEP41, CHMP1A, CHST14, CHST3, CHST6, CHSY1, CILK1, CLCN2, CLN5, CLN6, CLP1, CLPP, COA7, COASY, COG1, COG2, COG3, COG4, COG5, COG6, COG7, COG8, COQ4, COQ8A, COX20, CP, CPLANE1, CRB2, CRPPA, CSGALNACT1, CSPP1, CSTB, CTBP1, CWF19L1, CYP27A1, CYP2U1, DAG1, DAGLA, DARS2, DCC, DDHD2, DDOST, DDX59, DHCR7, DHDDS, DHRSX, DKC1, DLG4, DMXL2, DNAJC19, DNAJC3, DNAJC5, DNMT1, DOCK3, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYSL5, DYNC1H1, EBF3, EDEM3, EEF2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, EOGT, EPM2A, ERCC2, ERCC4, EVC, EVC2, EXOC3L2, EXOC8, EXOSC3, EXOSC5, EXOSC8, EXOSC9, EXT1, EXT2, FA2H, FAM149B1, FBXL4, FDXR, FGF14, FKRP, FKTN, FLVCR1, FMR1, FOLR1, FRMD4A, FRMD5, FTH1, FUT8, FXN, G6PC3, GALC, GALNT2, GALNT3, GBA2, GDAP2, GEMIN5, GFAP, GFPT1, GJC2, GLI3, GLRA1, GLRB, GLS, GMPPA, GMPPB, GNE, GORAB, GOSR2, GPAA1, GRID2, GRM1, GRN, GSN, HARS1, HEXA, HEXB, HMBS, HTT, HYLS1, IFT74, INPP5E, INTS11, IRF2BPL, ITPR1, KATNIP, KCNA1, KCNA2, KCNC3, KCND3, KCNJ10, KCNN2, KCNQ2, KCNQ3, KIAA0586, KIAA0753, KIF14, KIF1A, KIF1C, KIF7, LAMA1, LARGE1, LARS2, LETM1, LFNG, LIG3, LNPK, MAG, MAGT1, MAN1B1, MAPK8IP3, MARS2, MFN2, MFSD8, MGAT2, MINPP1, MKS1, MMACHC, MME, MOGS, MORC2, MPDU1, MPI, MRE11, MSTO1, MT-ATP6, MTCL1, MTFMT, MTPAP, MTTP, MVK, NAA60, NAGLU, NAXE, NFASC, NGLY1, NHLRC1, NKX2-1, NKX6-2, NOP56, NPC1, NPC2, NPHP1, NPHP3, NPTX1, NUS1, OFD1, OGDHL, OPA1, OPA3, OPHN1, PACS2, PAX2, PAX6, PCLO, PDE6D, PDYN, PEX16, PEX2, PEX6, PGAP2, PGAP3, PGM1, PGM3, PHGDH, PI4KA, PIBF1, PIGA, PIGL, PIGM, PIGN, PIGO, PIGS, PIGT, PIGV, PIGW, PITRM1, PLA2G6, PMM2, PMPCA, PMPCB, PNKD, PNKP, PNPLA6, PNPT1, POC1B, POLG, POLG2, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POU4F1, PPP2R2B, PRDM13, PRDX3, PRICKLE1, PRKCG, PRNP, PRPS1, PRRT2, PTF1A, PTRH2, PUM1, RARS2, RELN, RFC1, RFT1, RFXANK, RNF170, RNF216, RNF220, ROBO3, RORA, RPGRIP1L, RXYLT1, SACS, SAMD9L, SAR1B, SCLT1, SCN1A, SCN2A, SCN8A, SCN9A, SCYL1, SEC23B, SEPSECS, SETX, SIL1, SLC17A5, SLC1A3, SLC25A46, SLC2A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A8, SLC44A1, SLC52A2, SLC6A5, SLC9A1, SLC9A6, SMPD4, SNAP25, SNX14, SPG7, SPR, SPTAN1, SPTBN2, SQSTM1, SRD5A3, SSR4, ST3GAL3, ST3GAL5, STT3A, STUB1, SUFU, SVBP, SYNE1, SYNGAP1, TANGO2, TAPT1, TBC1D23, TBC1D32, TBP, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TECPR2, TERT, TGM6, THAP11, THG1L, TINF2, TMEM106B, TMEM107, TMEM138, TMEM165, TMEM216, TMEM218, TMEM231, TMEM237, TMEM240, TMEM67, TOE1, TOGARAM1, TPP1, TRAPPC11, TRIP4, TSEN15, TSEN2, TSEN34, TSEN54, TTBK2, TTC19, TTPA, TUBA1A, TUBA4A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUSC3, TWNK, TXNDC15, UBA5, UBTF, UCHL1, VAMP1, VLDLR, VMA12, VMA22, VPS13B, VPS13D, VPS41, VPS53, VRK1, WDR73, WDR81, WFS1, WWOX, XYLT1, XYLT2, ZFHX3, ZFYVE26, ZNF423, ZSWIM6

Hereditary Spastic paraplegia Genome | 151 genes

The hereditary spastic paraplegia panel Genome includes genes associated with pure and complex types of *HSP* as well as repeat expansion forms (e.g., *SPG4, SPG20*, *SCAs* with spasticity). This panel includes repeat expansion screening of the *ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, CACNA1A, FXN, HTT, PPP2R2B, PRNP, TBP* genes.

> Recommended panel approach based on genome sequencing, including repeat expansion screening of the genes *ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, CACNA1A, FXN, HTT, PPP2R2B, PRNP, TBP. The panel includes the non-coding gene RNU7-1.*

Genes Included

ABCD1, ABHD16A, ACBD6, ACER3, ADAR, AFG2B, AFG3L2, AIMP1, ALDH18A1, ALDH3A2, ALK, ALS2, AMFR, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARG1, ARL6IP1, ATAD3A, ATL1, ATP13A2, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, B4GALNT1, BCAS3, BICD2, BLOC1S1, BORCS8, BSCL2, C19orf12, CACNA1A, CAPN1, CLDN11, COQ4, CPT1C, CTNNB1, CYP27A1, CYP2U1, CYP7B1, DARS1, DDHD1, DDHD2, DDX3X, DSTYK, ELOVL1, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FAR1, FARS2, FBXO7, FXN, GAD1, GALC, GBA2, GBE1, GCH1, GJA1, GJC2, GLRX5, GPT2, HACE1, HECTD4, HIKESHI, HMBS, HPDL, HSPD1, HTT, IBA57, IFIH1, KCNA2, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, KPNA3, L1CAM, LETM1, LYST, MAG, MAPK8IP3, MARS1, MARS2, MTPAP, MTRFR, NDUFA12, NIPA1, NKX6-2, NRCAM, NSRP1, NT5C2, OPA3, PCDH12, PCYT2, PGAP1, PI4KA, PLP1, PNPLA6, POLR3A, PPFIBP1, PPP2R2B, PRNP, PSEN1, RAB3GAP2, REEP1, REEP2, RETREG1, RINT1, RNASEH2B, RNF170, RTN2, SACS, SARS2, SERAC1, SLC16A2, SLC1A4, SLC25A15, SLC25A46, SLC2A1, SLC33A1, SPART, SPAST, SPG11, SPG21, SPG7, SPTAN1, SPTSSA, STN1, TAF8, TBP, TECPR2, TFG, TMEM63C, TUBB4A, UBAP1, UCHL1, VAMP1, WASHC5, WDR45B, ZEB2, ZFYVE26

Hereditary Spastic paraplegia | 150 genes

The hereditary spastic paraplegia panel includes genes associated with pure and complex types of *HSP*. Repeat expansion forms of *HSP* are not included in this panel. If there is clinical suspicion of repeat expansion form of *HSP*, the Genome version of this panel is recommended.

Genes Included

ABCD1, ABHD16A, ACBD6, ACER3, ADAR, AFG2B, AFG3L2, AIMP1, ALDH18A1, ALDH3A2, ALK, ALS2, AMFR, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARG1, ARL6IP1, ATAD3A, ATL1, ATP13A2, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, B4GALNT1, BCAS3, BICD2, BLOC1S1, BORCS8, BSCL2, C19orf12, CACNA1A, CAPN1, CLDN11, COQ4, CPT1C, CTNNB1, CYP27A1, CYP2U1, CYP7B1, DARS1, DDHD1, DDHD2, DDX3X, DSTYK, ELOVL1, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FAR1, FARS2, FBXO7, FXN, GAD1, GALC, GBA2, GBE1, GCH1, GJA1, GJC2, GLRX5, GPT2, HACE1, HECTD4, HIKESHI, HMBS, HPDL, HSPD1, HTT, IBA57, IFIH1, KCNA2, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, KPNA3, L1CAM, LETM1, LYST, MAG, MAPK8IP3, MARS1, MARS2, MTPAP, MTRFR, NDUFA12, NIPA1, NKX6-2, NRCAM, NSRP1, NT5C2, OPA3, PCDH12, PCYT2, PGAP1, PI4KA, PLP1, PNPLA6, POLR3A, PPFIBP1, PPP2R2B, PRNP, PSEN1, RAB3GAP2, REEP1, REEP2, RETREG1, RINT1, RNASEH2B, RNF170, RTN2, SACS, SARS2, SERAC1, SLC16A2, SLC1A4, SLC25A15, SLC25A46, SLC2A1, SLC33A1, SPART, SPAST, SPG11, SPG21, SPG7, SPTAN1, SPTSSA, STN1, TAF8, TBP, TECPR2, TFG, TMEM63C, TUBB4A, UBAP1, UCHL1, VAMP1, WASHC5, WDR45B, ZEB2, ZFYVE26

Autoinflammatory Disorders

Autoinflammatory Disorders | 27 genes

The autoinflammatory disorders panel includes genes associated with hereditary immunodeficiencies and inflammatory vasculopathies such as hereditary disorders of the peripheral nervous system with inflammatory components; hereditary systemic autoinflammatory diseases (SAIDs) with neurological involvement, and disorders mimicking acquired neuroinflammatory conditions.

Genes Included

ADA, ADA2, CARD14, GP6, IL17RA, IL1RN, IL36RN, LPIN2, MEFV, MVK, NLRC4, NLRP12, NLRP3, NOD2, OTULIN, PLCG2, PRF1, PSMB8, PSTPIP1, RBCK1, SEC23B, SH3BP2, SLC29A3, STING1, TNFAIP3, TNFRSF1A, UBA1

CentoIEM | 744 genes

Inborn Errors of Metabolism (IEM) largely impact human diseases. CentoIEM is a metabolic and liver disease gene panel that screens for an array of different disorders and contains genes responsible for diverse phenotypes, including intermediary metabolism, such as aminoacidopathies, organic acidurias, urea cycle disorders, sugar intolerance, mental disorders, and porphyrias, among others. Genes linked to cytoplasmic and mitochondrial energetic processes and metabolism affecting cellular organelles, such as lysosomal, peroxisomal, glycosylation, and cholesterol synthesis are also included.

Genes Included

AARS2, ABCA1, ABCB4, ABCC2, ABCC8, ABCD1, ABCD4, ABCG5, ABCG8, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACOX1, ACSF3, ACY1, ADA, ADAMTS10, ADAMTSL2, ADAR, ADGRG1, ADK, ADSL, AFG3L2, AGA, AGL, AGPAT2, AGPS, AGXT, AHCY, AIFM1, AIMP1, AIMP2, AKT2, AKT3, ALAD, ALAS2, ALDH18A1, ALDH3A2, ALDH4A1, ALDH5A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ALPL, ALS2, AMN, AMPD2, AMT, ANK1, ANTXR2, AP3B1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOA2, APOA5, APOB, APOC2, APOE, APP, APPL1, APTX, AQP2, ARG1, ARL6IP1, ARSA, ARSB, ASAH1, ASL, ASPA, ASS1, ATM, ATP13A2, ATP6V0A2, ATP7A, ATP7B, ATPAF2, ATRX, AUH, AVP, AVPR2, B3GALNT2, B4GALNT1, B4GALT1, BCAP31, BCKDHA, BCKDHB, BCS1L, BEST1, BICD2, BLK, BMP6, BOLA3, BRAT1, BSCL2, BTD, C19orf12, CA5A, CACNA1D, CAPN1, CASP10, CASP8, CAV1, CAVIN1, CBLIF, CBS, CCT5, CD320, CEL, CERS1, CETP, CISD2, CLCN2, CLN3, CLN5, CLN6, CLN8, CLPB, CLPP, COA7, COA8, COASY, COG1, COG4, COG5, COG6, COG7, COG8, COL11A2, COL2A1, COL4A1, COL4A2, COLGALT1, COQ2, COQ8A, COQ9, COX10, COX15, COX20, COX6B1, CP, CPOX, CPS1, CPT1A, CPT1C, CPT2, CSF1R, CTC1, CTH, CTLA4, CTNS, CTSA, CTSC, CTSD, CTSF, CTSK, CUBN, CYP11B1, CYP17A1, CYP19A1, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DBT, DCAF17, DDC, DDHD1, DDHD2, DDOST, DGUOK, DHCR7, DHDDS, DIABLO, DKC1, DLAT, DLD, DLL3, DNAJC5, DNM1L, DOLK, DPM1, DPM2, DPM3, DPYD, DSTYK, DYM, EARS2, ECHS1, EIF2AK3, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, ENO3, ENPP1, ENTPD1, EPB42, EPHX2, EPM2A, EPRS1, ERCC6, ERCC8, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, ETHE1, EXOSC3, F2, F5, FA2H, FADD, FAH, FARS2, FARSB, FAS, FASLG, FASTKD2, FBN1, FBP1, FBXL4, FDX2, FECH, FGF23, FH, FHL1, FLAD1, FOLR1, FOXA2, FOXP3, FOXRED1, FTL, FUCA1, G6PC1, G6PD, GAA, GABRB2, GALC, GALE, GALK1, GALNS, GALT, GAMT, GAN, GATA4, GATA6, GATM, GBA1, GBA2, GBE1, GCDH, GCK, GCSH, GFAP, GFER, GFM1, GFM2, GFPT1, GHR, GJA1, GJB1, GJC2, GK, GLA, GLB1, GLDC, GLIS3, GLRX5, GLUD1, GLUL, GM2A, GMPPA, GNE, GNMT, GNPAT, GNPTAB, GNPTG, GNS, GOSR2, GPC3, GRN, GTPBP2, GTPBP3, GUSB, GYG1, GYS1, GYS2, HACE1, HADH, HADHA, HADHB, HAMP, HCFC1, HEPACAM, HEXA, HEXB, HFE, HGD, HGSNAT, HIBCH, HIKESHI, HJV, HK1, HLCS, HMBS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HPD, HPRT1, HRAS, HSD17B10, HSD17B4, HSD3B2, HSPD1, HTRA1, HYAL1, HYCC1, IARS2, IBA57, IDS, IDUA, IER3IP1, IFIH1, IL2RA, INS, INSR, ISCA2, ITIH4, ITK, IVD, JAG1, JAM3, KCNC1, KCNJ10, KCNJ11, KCNT1, KCTD7, KDM6A, KHK, KIDINS220, KIF1A, KIF1C, KIF5A, KLF11, KMT2D, KRAS, L1CAM, L2HGDH, LAMA2, LAMB1, LAMP2, LARGE1, LAT, LCAT, LDB3, LDHA, LDLR, LDLRAP1, LIAS, LIPA, LIPC, LIPE, LIPI, LIPT1, LIPT2, LMBRD1, LMNA, LMNB1, LPIN1, LPL, LRBA, LRPPRC, LYRM7, LYST, MAG, MAGT1, MAN1B1, MAN2B1, MANBA, MARS1, MARS2, MCCC1, MCCC2, MCEE, MCOLN1, MECR, MFSD8, MGAT2, MGME1, MLC1, MLPH, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPDU1, MPI, MPV17, MRPL44, MRPS22, 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, MTFMT, MTHFR, MTPAP, MTR, MTRFR, MTRR, MYO5A, MYOT, NAGA, NAGLU, NAGS, NARS2, NAXD, NAXE, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF4, NDUFAF5, NDUFAF6, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEU1, NEUROD1, NEUROG3, NFE2L2, NFU1, NGLY1, NHLRC1, NIPA1, NKX2-2, NKX6-2, NOTCH3, NPC1, NPC2, NPR2, NRAS, NT5C2, NUBPL, OAT, OCLN, OCRL, OPA3, OSGEP, OTC, OXCT1, PAH, PANK2, PAX4, PC, PCCA, PCCB, PCK1, PCSK9, PCYT2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDX1, PEPD, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGAP1, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PHYH, PIK3R1, PKLR, PLA2G6, PLAA, PLCG2, PLIN1, PLP1, PLPBP, PMM2, PMPCB, PNPLA6, PNPO, PNPT1, POLG, POLR1C, POLR3A, POLR3B, POR, PPARG, PPOX, PPP1R17, PPT1, PRF1, PRICKLE1, PRKAG2, PRKCD, PRODH, PSAP, PSEN1, PTF1A, PTS, PYCR2, PYGL, PYGM, QDPR, RAB11B, RAB27A, RAB3GAP2, RAI1, RARS1, RARS2, RBCK1, REEP1, REEP2, RFT1, RFX6, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, RPIA, RRM2B, RTN2, SACS, SAMHD1, SCARB2, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SERAC1, SERPINI1, SGSH, SI, SLC13A3, SLC16A1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC1A4, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A20, SLC25A4, SLC25A46, SLC2A1, SLC2A2, SLC33A1, SLC35A1, SLC35A2, SLC35C1, SLC3A1, SLC40A1, SLC4A1, SLC6A19, SLC6A8, SLC6A9, SLC7A7, SLC7A9, SLCO1B1, SLCO1B3, SMPD1, SNTA1, SOX10, SPART, SPAST, SPG11, SPG21, SPG7, SPTA1, SPTB, SRD5A3, SSR4, STAT1, STAT3, STT3A, STT3B, STX11, STXBP2, SUCLA2, SUCLG1, SUGCT, SUMF1, SUOX, SURF1, SYNE1, TACO1, TAFAZZIN, TAT, TBC1D24, TCF4, TCN2, TECPR2, TFG, TFR2, TGFB1, TINF2, TK2, TMEM106B, TMEM165, TMEM70, TPK1, TPP1, TREM2, TREX1, TRMT10A, TRPV4, TSFM, TTC19, TUBB4A, TUFM, TUSC3, TWNK, TYMP, TYROBP, UBAP1, UCHL1, UCP2, UFM1, UGT1A1, UMPS, UNC13D, UNC80, UQCRQ, UROD, UROS, USH1C, VAMP1, VCP, VPS11, VPS37A, WARS2, WASHC5, WDR45, WDR45B, WFS1, ZFP57, ZFYVE26, ZFYVE27

Mitochondrial Disorders

CentoMito® Comprehensive | 451 genes

CentoMito comprehensive covers the entire mitochondrial genome with detection of heteroplasmy down to 15.0 % and tests for nuclear genes related to mitochondrial diseases. Mitochondrial diseases are genetic conditions that occur when mitochondria fail to produce enough energy for the cell. Genetic mutations related to mitochondria cause symptoms mainly in organs, that consume large amounts of energy. These organs include the eye, kidney, pancreas, blood, inner ear, colon, skeletal muscle, heart, and brain.

Genes Included

AARS2, AASS, ABAT, ABCB6, ABCB7, ABCD1, ABCD3, ACACA, ACAD8, ACAD9, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACO2, ACOX1, ACSF3, ACSL4, ADAR, AFG3L2, AGK, AGXT, AIFM1, AK2, ALAS2, ALDH18A1, ALDH2, ALDH3A2, ALDH4A1, ALDH5A1, ALDH6A1, ALDH7A1, AMACR, AMPD1, AMT, APTX, ATIC, ATP5F1A, ATP5F1E, ATP7B, ATPAF2, AUH, BAG3, BCKDHA, BCKDHB, BCKDK, BCS1L, BOLA3, BTD, C19orf12, C1QBP, CA5A, CARS2, CAT, CAVIN1, CEL, CHAT, CHCHD10, CISD2, CLPB, CLPP, COA5, COA6, COA7, COA8, COASY, COMT, COQ2, COQ4, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX14, COX15, COX20, COX4I2, COX6A1, COX6B1, COX7B, CPOX, CPS1, CPT1A, CPT1C, CPT2, CRBN, CYB5A, CYB5R3, CYC1, CYCS, CYP11A1, CYP11B1, CYP11B2, CYP24A1, CYP27A1, CYP27B1, D2HGDH, DARS2, DBT, DGUOK, DHCR24, DHODH, DHTKD1, DIABLO, DLAT, DLD, DMGDH, DNA2, DNAJC19, DNM1L, EARS2, ECHS1, ELAC2, EPHX2, ETFA, ETFB, ETFDH, ETHE1, FAH, FARS2, FASTKD2, FBXL4, FDX2, FECH, FH, FKBP10, FLAD1, FOXRED1, FXN, GAMT, GARS1, GATM, GCDH, GCSH, GDAP1, GFER, GFM1, GFM2, GK, GLDC, GLRX5, GLUD1, GLYCTK, GPI, GPT2, GPX1, GRHPR, GSR, GTPBP3, HADH, HADHA, HADHB, HAMP, HARS2, HAX1, HCCS, HIBCH, HINT1, HK1, HLCS, HMBS, HMGCL, HMGCS2, HOGA1, HSD17B10, HSD17B4, HSD3B2, HSPA9, HSPD1, HTRA2, IARS2, IBA57, IDH2, IDH3B, IFIH1, ISCA1, ISCA2, ISCU, IVD, KARS1, KRT5, KRT8, L2HGDH, LAMP2, LARS2, LIAS, LIPT1, LIPT2, LMBRD1, LONP1, LRPPRC, LYRM7, MAOA, MARS2, MCCC1, MCCC2, MCEE, MECR, MFF, MFN2, MGME1, MICU1, MIPEP, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MPC1, MPV17, MRPL3, MRPL44, MRPS16, MRPS2, MRPS22, MRPS34, MSRB3, MSTO1, 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, MTFMT, MTHFD1, MTO1, MTPAP, MTRFR, MTRR, NADK2, NAGS, NARS2, NAXE, NBAS, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA6, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB11, NDUFB3, NDUFB8, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NGLY1, NNT, NR2F1, NTHL1, NUBPL, NUP62, OAT, OGDH, OPA1, OPA3, OTC, OXCT1, P4HB, PAM16, PANK2, PARK7, PARS2, PC, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1, PDSS2, PDX1, PET100, PEX11B, PHYH, PINK1, PKLR, PMPCA, PMPCB, PNKD, PNPLA8, PNPO, PNPT1, POLG, POLG2, POP1, PPOX, PRODH, PSAP, PTRH2, PTS, PUS1, PYCR1, PYCR2, QDPR, RARS1, RARS2, RDH11, RMND1, RNASEH1, RNASEH2A, RNASEH2B, RNASEH2C, RPIA, RPL35A, RPS14, RRM2B, SACS, SARS2, SBDS, SCN1A, SCO1, SCO2, SDHA, SDHAF1, SDHAF2, SDHD, SECISBP2, SERAC1, SFXN4, SLC16A1, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A26, SLC25A3, SLC25A38, SLC25A4, SLC25A42, SLC25A46, SLC39A8, SLC52A2, SLC6A8, SLC9A6, SNAP29, SOD1, SOD2, SPAST, SPG7, SPR, SPTLC2, STAR, STAT2, STXBP1, SUCLA2, SUCLG1, SUGCT, SUOX, SURF1, TACO1, TAFAZZIN, TANGO2, TCIRG1, TFR2, TIMM50, TIMM8A, TIMMDC1, TK2, TMEM126A, TMEM126B, TMEM70, TMLHE, TOP3A, TPI1, TPK1, TREX1, TRIT1, TRMT10C, TRMT5, TRMU, TRNT1, TSFM, TTC19, TUBB3, TUFM, TWNK, TYMP, UNG, UQCC2, UQCRB, UQCRC2, UQCRQ, VARS2, WARS2, WDR45, WDR81, WFS1, XPNPEP3, YARS2

Neuromuscular Disorders *Upgraded*

Neuromuscular Disorders Upgraded Genome | 690 genes

This panel includes genes associated with motor neuron disease, myopathies, neuropathies and neuromuscular junction disorders. Repeat expansion screening for *AR, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, C9orf72, CACNA1A, CNBP, DMPK, FMR1, FXN, HTT, NOP56, PABPN1, PPP2R2B, PRNP, TBP* genes are included in the analysis. In addition, the panel includes screening for *GBA1* conversion analysis with its pseudogene and SMN1 CNV analysis.

> Recommended panel approach based on genome sequencing, including repeat expansion screening of the genes *AR, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, C9orf72, CACNA1A, CNBP, DMPK, FMR1, FXN, HTT, NOP56, PABPN1, PPP2R2B, PRNP, TBP; GBA1* conversion analysis with its pseudogene; and *SMN1* CNV analysis.

Genes Included

AARS1, ABCA1, ABCC9, ABCD1, ABHD12, ABHD16A, ABHD5, ACAD9, ACADM, ACADS, ACADVL, ACTA1, ACTC1, ACTN2, ADAMTS10, ADAMTS15, ADAR, ADCY6, ADGRG6, ADSL, ADSS1, AFG3L2, AGL, AGRN, AGTPBP1, AGXT, AIFM1, AIMP1, ALDH18A1, ALDH3A2, ALDOA, ALG14, ALG2, ALG3, ALS2, AMPD1, AMPD2, ANG, ANO5, ANTXR2, AP1S1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOA1, APTX, AR, ARG1, ARL6IP1, ARSA, ASAH1,

ASCC1, ASCC3, ASXL1, ATAD1, ATL1, ATL3, ATM, ATP13A2, ATP1A1, ATP1A2, ATP2A1, ATP7A, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, B3GALNT2, B4GALNT1, B4GAT1, BAG3, BCKDHB, BET1, BICD2, BIN1, BRAF, BSCL2, C19orf12, C9orf72, CACNA1A, CACNA1E, CACNA1S, CAPN1, CAPN3, CASQ1, CASQ2, CAV1, CAV3, CAVIN1, CCDC47, CCDC78, CD59, CEP55, CFL2, CHAT, CHCHD10, CHKB, CHMP1A, CHMP2B, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CLCF1, CLCN1, CNBP, CNTN1, CNTNAP1, COA7, COASY, COL12A1, COL13A1, COL25A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COL9A3, COLQ, COQ4, COQ8A, COX6A1, CPOX, CPT2, CRLF1, CRPPA, CRYAB, CSRP3, CTDP1, CUL4B, CYP27A1, CYP2U1, CYP7B1, DAG1, DARS1, DARS2, DCTN1, DDHD1, DDHD2, DEGS1, DES, DGUOK, DHCR24, DHH, DHTKD1, DHX16, DMD, DMPK, DNA2, DNAJB2, DNAJB4, DNAJB6, DNAJC3, DNM2, DNMT1, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DRP2, DSC2, DSG2, DSP, DST, DSTYK, DYNC1H1, DYSF, EBP, ECEL1, EGR2, ELP1, EMD, ENO3, ENTPD1, EPG5, ERBB3, ERBB4, ERCC1, ERCC5, ERCC6, ERCC8, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, EXOSC9, FA2H, FAH, FAM111B, FAM20C, FARS2, FBLN5, FBN2, FBXO38, FDX2, FGD4, FGF14, FGFR2, FGFR3, FHL1, FIG4, FILIP1, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLNA, FLNB, FLNC, FLVCR1, FMR1, FUS, FXN, FXR1, GAA, GAD1, GALC, GAN, GARS1, GBA1, GBA2, GBE1, GCH1, GDAP1, GFER, GFM2, GFPT1, GGPS1, GIPC1, GJB1, GJC2, GLA, GLDN, GLE1, GMPPB, GNB4, GNE, GOLGA2, GOSR2, GRN, GYG1, GYS1, HACD1, HACE1, HADH, HADHA, HADHB, HARS1, HFE, HIKESHI, HINT1, HK1, HMBS, HMGCR, HNRNPA1, HNRNPA2B1, HNRNPDL, HRAS, HSPB1, HSPB8, HSPD1, HSPG2, HTRA2, HTT, HYCC1, IARS2, IBA57, IGHMBP2, INF2, INPP5K, IRF6, ISCU, ITGA7, ITPR1, JAG1, JAG2, JPH2, JUP, KARS1, KAT6B, KBTBD13, KCNA1, KCNA2, KCNC3, KCNJ2, KCNK3, KDM5C, KIDINS220, KIF1A, KIF1B, KIF1C, KIF21A, KIF5A, KIF5C, KLHL40, KLHL41, KLHL7, KRAS, KY, L1CAM, LAMA2, LAMA4, LAMA5, LAMB2, LAMP2, LARGE1, LAS1L, LDB3, LDHA, LETM1, LGI3, LGI4, LITAF, LMNA, LMOD3, LMX1B, LPIN1, LRP12, LRP4, LRSAM1, LYST, MAG, MAGEL2, MAP1B, MAP2K1, MAP2K2, MAP3K20, MARS1, MARS2, MATR3, MCM3AP, MED12, MED25, MEGF10, MET, MFN2, MICU1, MLIP, MMACHC, MME, MORC2, MPV17, MPZ, MRE11, MSTO1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MTM1, MTMR2, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTPAP, MTRFR, 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, MTTP, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MUSK, MYBPC1, MYBPC3, MYF5, MYH14, MYH2, MYH3, MYH6, MYH7, MYH8, MYL11, MYL2, MYL3, MYMK, MYO18B, MYO9A, MYOD1, MYOT, MYPN, NAGA, NAGLU, NALCN, NDRG1, NEB, NEFH, NEFL, NEK1, NEK9, NEXN, NGF, NIPA1, NKX6-2, NOP56, NRAS, NT5C2, NTRK1, NUP88, OBSCN, OPA1, OPA3, OPTN, ORAI1, PABPN1, PAX7, PCDH12, PCYT2, PDHA1, PDK3, PDYN, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PFN1, PGAM2, PGAP1, PGK1, PGM1, PHKA1, PHKB, PHYH, PI4KA, PIEZO2, PIGS, PIP5K1C, PKP2, PLEC, PLEKHG5, PLN, PLOD1, PLOD2, PLP1, PMM2, PMP2, PMP22, PNKP, PNPLA2, PNPLA6, POGLUT1, POLG, POLG2, POLR3A, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC1, POPDC3, POR, PPA2, PPOX, PPP2R2B, PRDM12, PREPL, PRG4, PRKAG2, PRKCG, PRNP, PRPS1, PRX, PSEN1, PTEN, PTPN11, PTRH2, PUS1, PYGM, PYROXD1, RAB3GAP2, RAB7A, RAF1, RAPSN, RBCK1, RBM20, REEP1, REEP2, RETREG1, RFC4, RIPK4, RIT1, RNASEH2B, RRM2B, RTN2, RXYLT1, RYR1, RYR2, SACS, SARS2, SBF1, SBF2, SCARB2, SCARF2, SCN11A, SCN1A, SCN4A, SCN5A, SCN9A, SCP2, SCYL1, SCYL2, SELENOI, SELENON, SEPTIN9, SERAC1, SETX, SGCA, SGCB, SGCD, SGCG, SH3TC2, SIGMAR1, SIL1, SKI, SLC12A6, SLC16A2, SLC18A3, SLC1A3, SLC1A4, SLC22A12, SLC22A5, SLC25A1, SLC25A19, SLC25A4, SLC25A42, SLC25A46, SLC29A3, SLC2A1, SLC2A9, SLC33A1, SLC35A3, SLC52A2, SLC52A3, SLC5A7, SLC6A9, SMAD3, SMAD4, SMCHD1, SMN1, SMPD4, SMPX, SNAP25, SNRPN, SOD1, SORD, SOS1, SOX10, SPART, SPAST, SPEG, SPG11, SPG21, SPG7, SPTBN2, SPTBN4, SPTLC1, SPTLC2, SQSTM1, SRPK3, STAC3, STIM1, SUCLA2, SURF1, SVIL, SYNE1, SYT2, TAFAZZIN, TAMM41, TANGO2, TARDBP, TBCD, TBK1, TBP, TCAP, TDP1, TECPR2, TFG, TGFB2, TGFB3, TGFBR1, TGFBR2, TIA1, TK2, TMEM43, TNNC1, TNNI2, TNNI3, TNNT1, TNNT2, TNNT3, TNPO3, TOR1A, TOR1AIP1, TPM1, TPM2, TPM3, TRAPPC11, TRDN, TRIM32, TRIP4, TRPV4, TSEN2, TSEN34, TSEN54, TSFM, TTBK2, TTN, TTPA, TTR, TUBA4A, TUBB3, TUBB4A, TWNK, TYMP, UBA1, UBAP1, UBQLN2, UNC45B, VAMP1, VAPB, VCL, VCP, VIPAS39, VMA21, VPS13A, VPS33B, VRK1, VWA1, WARS1, WASHC5, WDR45B, WNK1, XK, XPA, YARS1, YARS2, ZC4H2, ZEB2, ZFYVE26, ZMPSTE24, ZNF335

Neuromuscular Disorders Upgraded | 690 genes

This panel includes genes associated with motor neuron disease, myopathies, neuropathies and neuromuscular junction disorders. Variants in the genes *AR, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, C9orf72, CACNA1A, CNBP, DMPK, FMR1, FXN, HTT, NOP56, PABPN1, PPP2R2B, PRNP, TBP* are only interrogated as sequence variants. If there is clinical suspicion of a repeat expansion disorder associated with one or more of the above mentioned genes, the Genome version of this panel is recommended. Please note that the most common variants associated with disease in *SMN1* are not covered by this NGS analysis.

Genes Included

AARS1, ABCA1, ABCC9, ABCD1, ABHD12, ABHD16A, ABHD5, ACAD9, ACADM, ACADS, ACADVL, ACTA1, ACTC1, ACTN2, ADAMTS10, ADAMTS15, ADAR, ADCY6, ADGRG6, ADSL, ADSS1, AFG3L2, AGL, AGRN, AGTPBP1, AGXT, AIFM1, AIMP1, ALDH18A1, ALDH3A2, ALDOA, ALG14, ALG2, ALG3, ALS2, AMPD1, AMPD2, ANG, ANO5, ANTXR2, AP1S1, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APOA1, APTX, AR, ARG1, ARL6IP1, ARSA, ASAH1, ASCC1, ASCC3, ASXL1, ATAD1, ATL1, ATL3, ATM, ATP13A2, ATP1A1, ATP1A2, ATP2A1, ATP7A, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, B3GALNT2, B4GALNT1, B4GAT1, BAG3, BCKDHB, BET1, BICD2, BIN1, BRAF, BSCL2, C19orf12, C9orf72, CACNA1A, CACNA1E, CACNA1S, CAPN1, CAPN3, CASQ1, CASQ2, CAV1, CAV3, CAVIN1, CCDC47, CCDC78, CD59, CEP55, CFL2, CHAT, CHCHD10, CHKB, CHMP1A, CHMP2B, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CLCF1, CLCN1, CNBP, CNTN1, CNTNAP1, COA7, COASY, COL12A1, COL13A1, COL25A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COL9A3, COLQ, COQ4, COQ8A, COX6A1, CPOX, CPT2, CRLF1, CRPPA, CRYAB, CSRP3, CTDP1, CUL4B, CYP27A1, CYP2U1, CYP7B1, DAG1, DARS1, DARS2, DCTN1, DDHD1, DDHD2, DEGS1, DES, DGUOK, DHCR24, DHH, DHTKD1, DHX16, DMD, DMPK,

DNA2, DNAJB2, DNAJB4, DNAJB6, DNAJC3, DNM2, DNMT1, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DRP2, DSC2, DSG2, DSP, DST, DSTYK, DYNC1H1, DYSF, EBP, ECEL1, EGR2, ELP1, EMD, ENO3, ENTPD1, EPG5, ERBB3, ERBB4, ERCC1, ERCC5, ERCC6, ERCC8, ERLIN1, ERLIN2, ETFA, ETFB, ETFDH, EXOSC3, EXOSC8, EXOSC9, FA2H, FAH, FAM111B, FAM20C, FARS2, FBLN5, FBN2, FBXO38, FDX2, FGD4, FGF14, FGFR2, FGFR3, FHL1, FIG4, FILIP1, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLNA, FLNB, FLNC, FLVCR1, FMR1, FUS, FXN, FXR1, GAA, GAD1, GALC, GAN, GARS1, GBA1, GBA2, GBE1, GCH1, GDAP1, GFER, GFM2, GFPT1, GGPS1, GIPC1, GJB1, GJC2, GLA, GLDN, GLE1, GMPPB, GNB4, GNE, GOLGA2, GOSR2, GRN, GYG1, GYS1, HACD1, HACE1, HADH, HADHA, HADHB, HARS1, HFE, HIKESHI, HINT1, HK1, HMBS, HMGCR, HNRNPA1, HNRNPA2B1, HNRNPDL, HRAS, HSPB1, HSPB8, HSPD1, HSPG2, HTRA2, HTT, HYCC1, IARS2, IBA57, IGHMBP2, INF2, INPP5K, IRF6, ISCU, ITGA7, ITPR1, JAG1, JAG2, JPH2, JUP, KARS1, KAT6B, KBTBD13, KCNA1, KCNA2, KCNC3, KCNJ2, KCNK3, KDM5C, KIDINS220, KIF1A, KIF1B, KIF1C, KIF21A, KIF5A, KIF5C, KLHL40, KLHL41, KLHL7, KRAS, KY, L1CAM, LAMA2, LAMA4, LAMA5, LAMB2, LAMP2, LARGE1, LAS1L, LDB3, LDHA, LETM1, LGI3, LGI4, LITAF, LMNA, LMOD3, LMX1B, LPIN1, LRP12, LRP4, LRSAM1, LYST, MAG, MAGEL2, MAP1B, MAP2K1, MAP2K2, MAP3K20, MARS1, MARS2, MATR3, MCM3AP, MED12, MED25, MEGF10, MET, MFN2, MICU1, MLIP, MMACHC, MME, MORC2, MPV17, MPZ, MRE11, MSTO1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MTM1, MTMR2, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTPAP, MTRFR, 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, MTTP, MT-TP, MT-TQ, MT-TR, MT-TS1, MT-TS2, MT-TT, MT-TV, MT-TW, MT-TY, MUSK, MYBPC1, MYBPC3, MYF5, MYH14, MYH2, MYH3, MYH6, MYH7, MYH8, MYL11, MYL2, MYL3, MYMK, MYO18B, MYO9A, MYOD1, MYOT, MYPN, NAGA, NAGLU, NALCN, NDRG1, NEB, NEFH, NEFL, NEK1, NEK9, NEXN, NGF, NIPA1, NKX6-2, NOP56, NRAS, NT5C2, NTRK1, NUP88, OBSCN, OPA1, OPA3, OPTN, ORAI1, PABPN1, PAX7, PCDH12, PCYT2, PDHA1, PDK3, PDYN, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PFN1, PGAM2, PGAP1, PGK1, PGM1, PHKA1, PHKB, PHYH, PI4KA, PIEZO2, PIGS, PIP5K1C, PKP2, PLEC, PLEKHG5, PLN, PLOD1, PLOD2, PLP1, PMM2, PMP2, PMP22, PNKP, PNPLA2, PNPLA6, POGLUT1, POLG, POLG2, POLR3A, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC1, POPDC3, POR, PPA2, PPOX, PPP2R2B, PRDM12, PREPL, PRG4, PRKAG2, PRKCG, PRNP, PRPS1, PRX, PSEN1, PTEN, PTPN11, PTRH2, PUS1, PYGM, PYROXD1, RAB3GAP2, RAB7A, RAF1, RAPSN, RBCK1, RBM20, REEP1, REEP2, RETREG1, RFC4, RIPK4, RIT1, RNASEH2B, RRM2B, RTN2, RXYLT1, RYR1, RYR2, SACS, SARS2, SBF1, SBF2, SCARB2, SCARF2, SCN11A, SCN1A, SCN4A, SCN5A, SCN9A, SCP2, SCYL1, SCYL2, SELENOI, SELENON, SEPTIN9, SERAC1, SETX, SGCA, SGCB, SGCD, SGCG, SH3TC2, SIGMAR1, SIL1, SKI, SLC12A6, SLC16A2, SLC18A3, SLC1A3, SLC1A4, SLC22A12, SLC22A5, SLC25A1, SLC25A19, SLC25A4, SLC25A42, SLC25A46, SLC29A3, SLC2A1, SLC2A9, SLC33A1, SLC35A3, SLC52A2, SLC52A3, SLC5A7, SLC6A9, SMAD3, SMAD4, SMCHD1, SMN1, SMPD4, SMPX, SNAP25, SNRPN, SOD1, SORD, SOS1, SOX10, SPART, SPAST, SPEG, SPG11, SPG21, SPG7, SPTBN2, SPTBN4, SPTLC1, SPTLC2, SQSTM1, SRPK3, STAC3, STIM1, SUCLA2, SURF1, SVIL, SYNE1, SYT2, TAFAZZIN, TAMM41, TANGO2, TARDBP, TBCD, TBK1, TBP, TCAP, TDP1, TECPR2, TFG, TGFB2, TGFB3, TGFBR1, TGFBR2, TIA1, TK2, TMEM43, TNNC1, TNNI2, TNNI3, TNNT1, TNNT2, TNNT3, TNPO3, TOR1A, TOR1AIP1, TPM1, TPM2, TPM3, TRAPPC11, TRDN, TRIM32, TRIP4, TRPV4, TSEN2, TSEN34, TSEN54, TSFM, TTBK2, TTN, TTPA, TTR, TUBA4A, TUBB3, TUBB4A, TWNK, TYMP, UBA1, UBAP1, UBQLN2, UNC45B, VAMP1, VAPB, VCL, VCP, VIPAS39, VMA21, VPS13A, VPS33B, VRK1, VWA1, WARS1, WASHC5, WDR45B, WNK1, XK, XPA, YARS1, YARS2, ZC4H2, ZEB2, ZFYVE26, ZMPSTE24, ZNF335

Motor Neuron Disease Genome | 153 genes

The Motor Neuron Disease Panel Genome includes genes associated with Upper motor neuron disease (MND) (e.g., hereditary spastic paraplegias, ALS), Lower MND (e.g., SMA, spinal and bulbar muscular atrophy) and mixed types of MND. This panel includes repeat expansion screening of the genes *AR, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, C9orf72, CACNA1A, DMPK, FXN, HTT, NOP56, PPP2R2B, TBP* and *SMN1* CNV screening.

> Recommended panel approach based on genome sequencing, including repeat expansion screening of the genes *AR, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, C9orf72, CACNA1A, DMPK, FXN, HTT, NOP56, PPP2R2B, TBP* and *SMN1* CNV analysis.

Genes Included

AARS1, ABCD1, ABHD16A, ADAR, AFG3L2, AIMP1, ALDH18A1, ALS2, AMPD2, ANG, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, AR, ARG1, ARL6IP1, ASAH1, ATL1, ATP13A2, ATP7A, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, B4GALNT1, BICD2, BSCL2, C19orf12, C9orf72, CACNA1A, CAPN1, CAV1, CHCHD10, CHMP2B, CYP27A1, CYP2U1, CYP7B1, DARS1, DCTN1, DDHD1, DDHD2, DMPK, DNAJB2, DSTYK, DYNC1H1, ENTPD1, ERBB4, ERLIN1, ERLIN2, EXOSC3, EXOSC8, FA2H, FARS2, FBXO38, FIG4, FUS, FXN, GAD1, GARS1, GBA2, GCH1, GJC2, GRN, HACE1, HFE, HIKESHI, HNRNPA1, HSPB1, HSPB8, HSPD1, HTT, IBA57, IGHMBP2, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, L1CAM, LYST, MAG, MARS1, MARS2, MTPAP, MTRFR, NEFH, NEK1, NIPA1, NKX6-2, NOP56, NT5C2, OPA3, OPTN, PCDH12, PCYT2, PFN1, PGAP1, PLEKHG5, PLP1, PNPLA6, POLR3A, PPP2R2B, PSEN1, RAB3GAP2, REEP1, REEP2, RNASEH2B, RTN2, SACS, SARS2, SERAC1, SETX, SIGMAR1, SLC16A2, SLC1A4, SLC25A46, SLC2A1, SLC33A1, SLC52A2, SLC52A3, SLC5A7, SMN1, SNRPN, SOD1, SPART, SPAST, SPG11, SPG21, SPG7, SYT2, TARDBP, TBK1, TBP, TECPR2, TFG, TRIP4, TRPV4, TUBA4A, TUBB4A, UBA1, UBAP1, UBQLN2, VAMP1, VAPB, VCP, VRK1, WASHC5, WDR45B, ZEB2, ZFYVE26

Motor Neuron Disease | 153 genes

The Motor Neuron Disease panel includes genes associated with Upper motor neuron disease (MND) (e.g., hereditary spastic paraplegias, ALS), Lower MND (e.g., SMA, spinal and bulbar muscular atrophy) and mixed types of MND. Variants in the genes *AR, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, C9orf72, CACNA1A, DMPK, FXN, HTT, NOP56, PPP2R2B, TBP*, are interrogated only as sequence variants. If there is clinical suspicion of a repeat expansion disorder associated with one or more of the above mentioned genes, the Genome version of this panel is recommended. Please note that the most common variants associated with disease in *SMN1* are not covered by this NGS analysis.

Genes Included

AARS1, ABCD1, ABHD16A, ADAR, AFG3L2, AIMP1, ALDH18A1, ALS2, AMPD2, ANG, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, AR, ARG1, ARL6IP1, ASAH1, ATL1, ATP13A2, ATP7A, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, B4GALNT1, BICD2, BSCL2, C19orf12, C9orf72, CACNA1A, CAPN1, CAV1, CHCHD10, CHMP2B, CYP27A1, CYP2U1, CYP7B1, DARS1, DCTN1, DDHD1, DDHD2, DMPK, DNAJB2, DSTYK, DYNC1H1, ENTPD1, ERBB4, ERLIN1, ERLIN2, EXOSC3, EXOSC8, FA2H, FARS2, FBXO38, FIG4, FUS, FXN, GAD1, GARS1, GBA2, GCH1, GJC2, GRN, HACE1, HFE, HIKESHI, HNRNPA1, HSPB1, HSPB8, HSPD1, HTT, IBA57, IGHMBP2, KDM5C, KIDINS220, KIF1A, KIF1C, KIF5A, L1CAM, LYST, MAG, MARS1, MARS2, MTPAP, MTRFR, NEFH, NEK1, NIPA1, NKX6-2, NOP56, NT5C2, OPA3, OPTN, PCDH12, PCYT2, PFN1, PGAP1, PLEKHG5, PLP1, PNPLA6, POLR3A, PPP2R2B, PSEN1, RAB3GAP2, REEP1, REEP2, RNASEH2B, RTN2, SACS, SARS2, SERAC1, SETX, SIGMAR1, SLC16A2, SLC1A4, SLC25A46, SLC2A1, SLC33A1, SLC52A2, SLC52A3, SLC5A7, SMN1, SNRPN, SOD1, SPART, SPAST, SPG11, SPG21, SPG7, SYT2, TARDBP, TBK1, TBP, TECPR2, TFG, TRIP4, TRPV4, TUBA4A, TUBB4A, UBA1, UBAP1, UBQLN2, VAMP1, VAPB, VCP, VRK1, WASHC5, WDR45B, ZEB2, ZFYVE26

Myopathies Genome | 402 genes

The Myopathies panel Genome includes genes associated with Muscular Dystrophies (e.g., Duchenne MD, Emery-Dreyfuss MD), Myotonia (e.g., Dystrophic myotonia type 1+2. non-dystrophic myotonia), metabolic, mitochondrial and rare myopathies (e.g. myofibrillary myopathy; centronuclear myopathies; nemaline myopathies). This panel includes screening for: *AR, CNBP, DMPK, PABPN1* repeat expansion; *GBA1* conversion analysis with its pseudogene; and *SMN1* CNV analysis.

> Recommended panel approach based on genome sequencing, including repeat expansion screening of the genes *AR, CNBP, DMPK, PABPN1; GBA1* conversion analysis with its pseudogene; and *SMN1* CNV analysis.

Genes Included

ABHD5, ACAD9, ACADM, ACADS, ACADVL, ACTA1, ACTC1, ACTN2, ADAMTS10, ADAMTS15, ADCY6, ADGRG6, ADSL, ADSS1, AGL, AGRN, AIMP1, ALDOA, ALG14, ALG2, ALG3, AMPD1, ANO5, ANTXR2, AR, ASCC1, ASCC3, ASXL1, ATAD1, ATP1A2, ATP2A1, B3GALNT2, B4GAT1, BAG3, BET1, BICD2, BIN1, CACNA1E, CACNA1S, CAPN3, CASQ1, CAV3, CAVIN1, CCDC47, CCDC78, CEP55, CFL2, CHAT, CHCHD10, CHKB, CHMP1A, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CLCF1, CLCN1, CNBP, CNTN1, CNTNAP1, COASY, COL12A1, COL13A1, COL25A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COL9A3, COLQ, COQ4, COQ8A, CPT2, CRLF1, CRPPA, CRYAB, CTDP1, CUL4B, DAG1, DES, DGUOK, DHCR24, DHX16, DMD, DMPK, DNA2, DNAJB4, DNAJB6, DNM2, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DYNC1H1, DYSF, EBP, ECEL1, EGR2, EMD, ENO3, EPG5, ERBB3, ERCC1, ERCC5, ERCC6, ERCC8, ETFA, ETFB, ETFDH, EXOSC3, EXOSC9, FAM111B, FAM20C, FBN2, FDX2, FGFR2, FGFR3, FHL1, FILIP1, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLNA, FLNB, FLNC, FXR1, GAA, GARS1, GBA1, GBE1, GDAP1, GFER, GFM2, GFPT1, GGPS1, GIPC1, GLDN, GLE1, GMPPB, GNE, GOLGA2, GOSR2, GRN, GYG1, GYS1, HACD1, HADH, HADHA, HADHB, HMGCR, HNRNPA1, HNRNPA2B1, HNRNPDL, HRAS, HSPB1, HSPB8, HSPG2, HTRA2, IBA57, IGHMBP2, INPP5K, IRF6, ISCU, ITGA7, JAG2, KAT6B, KBTBD13, KCNJ2, KCNK3, KIDINS220, KIF21A, KIF5C, KLHL40, KLHL41, KLHL7, KY, L1CAM, LAMA2, LAMP2, LARGE1, LDB3, LDHA, LETM1, LGI3, LGI4, LMNA, LMOD3, LMX1B, LPIN1, MAGEL2, MAP3K20, MATR3, MED12, MEGF10, MET, MFN2, MICU1, MLIP, MPZ, MSTO1, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MTM1, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTRFR, 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, MUSK, MYBPC1, MYBPC3, MYF5, MYH14, MYH2, MYH3, MYH7, MYH8, MYL11, MYL2, MYMK, MYO18B, MYO9A, MYOD1, MYOT, MYPN, NALCN, NEB, NEFL, NEK9, NUP88, OBSCN, ORAI1, PABPN1, PAX7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PI4KA, PIEZO2, PIGS, PIP5K1C, PLEC, PLOD1, PLOD2, PNPLA2, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC1, POPDC3, POR, PPA2, PRG4, PRKAG2, PUS1, PYGM, PYROXD1, RAPSN, RBCK1, RFC4, RIPK4, RRM2B, RXYLT1, RYR1, SCARF2, SCN1A, SCN4A, SCYL2, SELENON, SGCA, SGCB, SGCD, SGCG, SIL1, SKI, SLC18A3, SLC22A12, SLC22A5, SLC25A4, SLC25A42, SLC29A3, SLC2A9, SLC35A3, SLC52A3, SLC5A7, SLC6A9, SMAD3, SMAD4, SMCHD1, SMN1, SMPD4, SMPX, SPEG, SPTBN4, SQSTM1, SRPK3, STAC3, STIM1, SUCLA2, SVIL, SYNE1, SYT2, TAMM41, TANGO2, TBCD, TCAP, TGFB2, TGFB3, TGFBR1, TGFBR2, TIA1, TK2, TMEM43, TNNI2, TNNT1, TNNT3, TNPO3, TOR1A, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRDN, TRIM32, TRIP4, TRPV4, TSEN2, TSEN34, TSEN54, TSFM, TTN, TYMP, UBA1, UNC45B, VAMP1, VCP, VIPAS39, VMA21, VPS33B, VWA1, YARS2, ZC4H2, ZMPSTE24, ZNF335

Myopathies | 402 genes

The Myopathies panel includes genes associated with Muscular Dystrophies (e.g., Duchenne MD, Emery-Dreyfuss MD), Myotonia (e.g., Dystrophic myotonia type 1+2. non-dystrophic myotonia), metabolic, mitochondrial and rare myopathies (e.g. myofibrillary myopathy; centronuclear myopathies; nemaline myopathies). Variants in the genes *AR, CNBP, DMPK, PABPN1* are only interrogated as sequence variants. If there is clinical suspicion of a repeat expansion disorder associated with one or more of the above mentioned genes, the Genome version of this panel is recommended. Please note that the most common variants associated with disease in *SMN1* are not covered by this NGS analysis.

Genes Included

ABHD5, ACAD9, ACADM, ACADS, ACADVL, ACTA1, ACTC1, ACTN2, ADAMTS10, ADAMTS15, ADCY6, ADGRG6, ADSL, ADSS1, AGL, AGRN, AIMP1, ALDOA, ALG14, ALG2, ALG3, AMPD1, ANO5, ANTXR2, AR, ASCC1, ASCC3, ASXL1, ATAD1, ATP1A2, ATP2A1, B3GALNT2, B4GAT1, BAG3, BET1, BICD2, BIN1, CACNA1E, CACNA1S, CAPN3, CASQ1, CAV3, CAVIN1, CCDC47, CCDC78, CEP55, CFL2, CHAT, CHCHD10, CHKB, CHMP1A, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, CHST14, CLCF1, CLCN1, CNBP, CNTN1, CNTNAP1, COASY, COL12A1, COL13A1, COL25A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COL9A3, COLQ, COQ4, COQ8A, CPT2, CRLF1, CRPPA, CRYAB, CTDP1, CUL4B, DAG1, DES, DGUOK, DHCR24, DHX16, DMD, DMPK, DNA2, DNAJB4, DNAJB6, DNM2, DOK7, DOLK, DPAGT1, DPM1, DPM2, DPM3, DYNC1H1, DYSF, EBP, ECEL1, EGR2, EMD, ENO3, EPG5, ERBB3, ERCC1, ERCC5, ERCC6, ERCC8, ETFA, ETFB, ETFDH, EXOSC3, EXOSC9, FAM111B, FAM20C, FBN2, FDX2, FGFR2, FGFR3, FHL1, FILIP1, FKBP10, FKBP14, FKRP, FKTN, FLAD1, FLNA, FLNB, FLNC, FXR1, GAA, GARS1, GBA1, GBE1, GDAP1, GFER, GFM2, GFPT1, GGPS1, GIPC1, GLDN, GLE1, GMPPB, GNE, GOLGA2, GOSR2, GRN, GYG1, GYS1, HACD1, HADH, HADHA, HADHB, HMGCR, HNRNPA1, HNRNPA2B1, HNRNPDL, HRAS, HSPB1, HSPB8, HSPG2, HTRA2, IBA57, IGHMBP2, INPP5K, IRF6, ISCU, ITGA7, JAG2, KAT6B, KBTBD13, KCNJ2, KCNK3, KIDINS220, KIF21A, KIF5C, KLHL40, KLHL41, KLHL7, KY, L1CAM, LAMA2, LAMP2, LARGE1, LDB3, LDHA, LETM1, LGI3, LGI4, LMNA, LMOD3, LMX1B, LPIN1, MAGEL2, MAP3K20, MATR3, MED12, MEGF10, MET, MFN2, MICU1, MLIP, MPZ, MSTO1, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MTM1, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTRFR, 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, MUSK, MYBPC1, MYBPC3, MYF5, MYH14, MYH2, MYH3, MYH7, MYH8, MYL11, MYL2, MYMK, MYO18B, MYO9A, MYOD1, MYOT, MYPN, NALCN, NEB, NEFL, NEK9, NUP88, OBSCN, ORAI1, PABPN1, PAX7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PI4KA, PIEZO2, PIGS, PIP5K1C, PLEC, PLOD1, PLOD2, PNPLA2, POGLUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, POPDC1, POPDC3, POR, PPA2, PRG4, PRKAG2, PUS1, PYGM, PYROXD1, RAPSN, RBCK1, RFC4, RIPK4, RRM2B, RXYLT1, RYR1, SCARF2, SCN1A, SCN4A, SCYL2, SELENON, SGCA, SGCB, SGCD, SGCG, SIL1, SKI, SLC18A3, SLC22A12, SLC22A5, SLC25A4, SLC25A42, SLC29A3, SLC2A9, SLC35A3, SLC52A3, SLC5A7, SLC6A9, SMAD3, SMAD4, SMCHD1, SMN1, SMPD4, SMPX, SPEG, SPTBN4, SQSTM1, SRPK3, STAC3, STIM1, SUCLA2, SVIL, SYNE1, SYT2, TAMM41, TANGO2, TBCD, TCAP, TGFB2, TGFB3, TGFBR1, TGFBR2, TIA1, TK2, TMEM43, TNNI2, TNNT1, TNNT3, TNPO3, TOR1A, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRDN, TRIM32, TRIP4, TRPV4, TSEN2, TSEN34, TSEN54, TSFM, TTN, TYMP, UBA1, UNC45B, VAMP1, VCP, VIPAS39, VMA21, VPS33B, VWA1, YARS2, ZC4H2, ZMPSTE24, ZNF335

Neuromuscular junction disorders panel | 33 genes

The neuromuscular junction disorders panel includes genes associated with congenital myasthenic syndromes.

Genes Included

AGRN, ALG14, ALG2, CACNA1A, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COL13A1, COLQ, DOK7, DPAGT1, GFPT1, GMPPB, LAMA5, LAMB2, LRP4, MUSK, MYO9A, PLEC, PREPL, RAPSN, RYR1, SCN4A, SLC18A3, SLC25A1, SLC5A7, SNAP25, SYT2, TOR1AIP1, VAMP1

Neuropathies Genome | 280 genes

The Neuropathies panel Genome includes genes associated with all types of Charcot-Marie-Tooth disease and hereditary small fiber neuropathies. This panel includes repeat expansion screening of the genes *AR, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, DMPK, FMR1, FXN, NOP56, PPP2R2B, PRNP,* and *SMN1 CNV* screening.

> Recommended panel approach based on genome sequencing, including repeat expansion screening of the genes *AR, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, DMPK, FMR1, FXN, NOP56, PPP2R2B, PRNP* and *SMN1* CNV analysis.

Genes Included

AARS1, ABCA1, ABCC9, ABHD12, ACTC1, ACTN2, AGTPBP1, AGXT, AIFM1, ALDH3A2, ALS2, AP1S1, APOA1, APTX, AR, ARL6IP1, ARSA, ASAH1, ATL1, ATL3, ATM, ATP1A1, ATP7A, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, B4GALNT1, BAG3, BCKDHB, BICD2, BRAF, BSCL2, C19orf12, CASQ2, CAV3, CD59, CHCHD10, CNTNAP1, COA7, COQ8A, COX6A1, CPOX, CRYAB, CSRP3, CTDP1, CYP27A1, DARS2, DCTN1, DEGS1, DES, DGUOK, DHH, DHTKD1, DMD, DMPK, DNAJB2, DNAJC3, DNM2, DNMT1, DRP2, DSC2, DSG2, DSP, DST, DSTYK, DYNC1H1, EGR2, ELP1, EMD, ERBB3, ERCC6, ERCC8, ETFDH, EXOSC3, EXOSC8, FA2H, FAH, FBLN5, FBXO38, FGD4, FGF14, FIG4, FKTN, FLVCR1, FMR1, FXN, GAA, GALC, GAN, GARS1, GBA2, GDAP1, GJB1, GJC2, GLA, GLE1, GNB4, HADHA, HADHB, HARS1, HINT1, HK1, HMBS, HRAS, HSPB1, HSPB8, HYCC1, IARS2, IGHMBP2, INF2, ITPR1, JAG1, JPH2, JUP, KARS1, KCNA1, KCNA2, KCNC3, KIF1A, KIF1B, KIF5A, KRAS, L1CAM, LAMA4, LAMP2, LAS1L, LDB3, LITAF, LMNA, LRP12, LRSAM1, LYST, MAP1B, MAP2K1, MAP2K2, MARS1, MCM3AP, MED25, MFN2, MMACHC, MME, MORC2, MPV17, MPZ, MRE11, MT-ATP6, MT-RNR1, MT-TL1, MTMR2, MTRFR, MTTP, MYBPC3, MYH14, MYH6, MYH7, MYL2, MYL3, MYPN, NAGA, NAGLU, NDRG1, NEFH, NEFL, NEXN, NGF, NIPA1, NOP56, NRAS, NTRK1, OPA1, OPA3, PDHA1, PDK3, PDYN, PEX10, PEX7, PHYH, PKP2, PLEKHG5, PLN, PLP1, PMM2, PMP2, PMP22, PNKP, PNPLA6, POLG, POLR3A, PPOX, PPP2R2B, PRDM12, PRKAG2, PRKCG, PRNP, PRPS1, PRX, PTEN, PTPN11, PTRH2, RAB7A, RAF1, RBM20, REEP1, RETREG1, RIT1, RYR2, SACS, SBF1, SBF2, SCARB2, SCN11A, SCN5A, SCN9A, SCP2, SCYL1, SELENOI, SEPTIN9, SETX, SGCD, SH3TC2, SIGMAR1, SIL1, SLC12A6, SLC1A3, SLC25A19, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SMN1, SNRPN, SORD, SOS1, SOX10, SPART, SPAST, SPG11, SPG21, SPG7, SPTBN2, SPTBN4, SPTLC1, SPTLC2, SUCLA2, SURF1, SYT2, TAFAZZIN, TCAP, TDP1, TFG, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TRIP4, TRPV4, TTBK2, TTN, TTPA, TTR, TUBB3, TWNK, TYMP, UBA1, VAPB, VCL, VCP, VPS13A, VRK1, VWA1, WARS1, WASHC5, WNK1, XK, XPA, YARS1, ZFYVE26

Neuropathies | 280 genes

The Neuropathies panel includes genes associated with all types of Charcot-Marie-Tooth disease and hereditary small fiber neuropathies. Variants in the genes *AR, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, DMPK, FMR1, FXN, NOP56, PPP2R2B, PRNP* are only interrogated as sequence variants. If there is clinical suspicion of a repeat expansion disorder associated with one or more of the abovementioned genes, the Genome version of this panel is recommended. Please note that the most common variants associated with disease in SMN1 are not covered by this NGS analysis.

Genes Included

AARS1, ABCA1, ABCC9, ABHD12, ACTC1, ACTN2, AGTPBP1, AGXT, AIFM1, ALDH3A2, ALS2, AP1S1, APOA1, APTX, AR, ARL6IP1, ARSA, ASAH1, ATL1, ATL3, ATM, ATP1A1, ATP7A, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, B4GALNT1, BAG3, BCKDHB, BICD2, BRAF, BSCL2, C19orf12, CASQ2, CAV3, CD59, CHCHD10, CNTNAP1, COA7, COQ8A, COX6A1, CPOX, CRYAB, CSRP3, CTDP1, CYP27A1, DARS2, DCTN1, DEGS1, DES, DGUOK, DHH, DHTKD1, DMD, DMPK, DNAJB2, DNAJC3, DNM2, DNMT1, DRP2, DSC2, DSG2, DSP, DST, DSTYK, DYNC1H1, EGR2, ELP1, EMD, ERBB3, ERCC6, ERCC8, ETFDH, EXOSC3, EXOSC8, FA2H, FAH, FBLN5, FBXO38, FGD4, FGF14, FIG4, FKTN, FLVCR1, FMR1, FXN, GAA, GALC, GAN, GARS1, GBA2, GDAP1, GJB1, GJC2, GLA, GLE1, GNB4, HADHA, HADHB, HARS1, HINT1, HK1, HMBS, HRAS, HSPB1, HSPB8, HYCC1, IARS2, IGHMBP2, INF2, ITPR1, JAG1, JPH2, JUP, KARS1, KCNA1, KCNA2, KCNC3, KIF1A, KIF1B, KIF5A, KRAS, L1CAM, LAMA4, LAMP2, LAS1L, LDB3, LITAF, LMNA, LRP12, LRSAM1, LYST, MAP1B, MAP2K1, MAP2K2, MARS1, MCM3AP, MED25, MFN2, MMACHC, MME, MORC2, MPV17, MPZ, MRE11, MT-ATP6, MT-RNR1, MT-TL1, MTMR2, MTRFR, MTTP, MYBPC3, MYH14, MYH6, MYH7, MYL2, MYL3, MYPN, NAGA, NAGLU, NDRG1, NEFH, NEFL, NEXN, NGF, NIPA1, NOP56, NRAS, NTRK1, OPA1, OPA3, PDHA1, PDK3, PDYN, PEX10, PEX7, PHYH, PKP2, PLEKHG5, PLN, PLP1, PMM2, PMP2, PMP22, PNKP, PNPLA6, POLG, POLR3A, PPOX, PPP2R2B, PRDM12, PRKAG2, PRKCG, PRNP, PRPS1, PRX, PTEN, PTPN11, PTRH2, RAB7A, RAF1, RBM20, REEP1, RETREG1, RIT1, RYR2, SACS, SBF1, SBF2, SCARB2, SCN11A, SCN5A, SCN9A, SCP2, SCYL1, SELENOI, SEPTIN9, SETX, SGCD, SH3TC2, SIGMAR1, SIL1, SLC12A6, SLC1A3, SLC25A19, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SMN1, SNRPN, SORD, SOS1, SOX10, SPART, SPAST, SPG11, SPG21, SPG7, SPTBN2, SPTBN4, SPTLC1, SPTLC2, SUCLA2, SURF1, SYT2, TAFAZZIN, TCAP, TDP1, TFG, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TRIP4, TRPV4, TTBK2, TTN, TTPA, TTR, TUBB3, TWNK, TYMP, UBA1, VAPB, VCL, VCP, VPS13A, VRK1, VWA1, WARS1, WASHC5, WNK1, XK, XPA, YARS1, ZFYVE26

Cerebrovascular Disorders Genome | 273 genes

The panel includes genes associated with stroke (e.g., *CADASIL, CARASIL*), disorders associated with increased risk of stroke (e.g., Fabry disease, coagulopathies), vascular malformations and vasculopathies (e.g., Moyamoya disease, Ehlers-Danlos syndrome). This panel includes *CACNA1A* repeat expansion screening.

> Recommended cerebrovascular disorder panel approach based on genome sequencing, including *CACNA1A* repeat expansion screening. The panel includes the non-coding gene *SNORD118*.

Genes Included

ABCA1, ABCC6, ABCC9, ABCG5, ABCG8, ACAD9, ACE, ACP5, ACTA2, ACVRL1, ADA2, ADAMTS13, ADAMTS2, ADGRG1, AEBP1, AGXT, ANTXR1, APOA1, APOB, APP, ARX, ASS1, ATP7A, ATR, B3GALT6, B4GALT1, BRAF, C1R, CACNA1A, CBL, CBS, CCM2, CD59, CEP152, CEP63, CHD4, CISD2, CNOT3, COG6, COL1A1, COL3A1, COL4A1, COL4A2, COL5A1, COL5A2, COLGALT1, COQ2, COQ8A, CPAP, CPS1, CRB1, CSF1R, CST3, CTC1, CTSA, CYP11B1, CYP11B2, CYP26C1, DCX, DLD, DNA2, DOCK8, DPAGT1, DPM1, DPM3, DYRK1B, EFEMP2, ELN, ENG, ENPP1, EPAS1, EPHB4, EPOR, ERCC6, ERCC8, ESCO2, F10, F13A1, F13B, F2, F5, F7, F8, F9, FASTKD2, FBLN5, FBN1, FERMT3, FGA, FGB, FGG, FLNA, FLT4, FLVCR2, FOXC1, FOXE3, FOXF1, GAA, GANAB, GATA3, GCDH, GDF2, GFI1B, GGCX, GJA1, GJA5, GLA, GLMN, GNAQ, GP1BA, GPHN, GPR143, GUCY1A1, GYS1, HBB, HNF1B, HRAS, HSD11B2, HTRA1, IL1RN, IL6, ISCU, ITM2B, IVD, JAG1, JAK2, JAM3, KANSL1, KCNA5, KCNJ2, KCNQ1, KDR, KNG1, KRAS, KRIT1, LAMB1, LAMC3, LARGE1, LARS2, LDLR, LMAN1, LMBRD1, LMNA, LOX, LRPPRC, LTBP3, LZTR1, MAP2K1, MCCC1, MEF2C, MEFV, MFAP5, MFN2, MGAT2, MMACHC, MMUT, MOCS1, MOCS2, MPI, MTHFR, MYBPC3, MYH11, MYH7, MYLK, NBEAL2, NDE1, NF1, NLRP3, NOTCH1, NOTCH2, NOTCH3, NPPA, NRAS, OCLN, OPHN1, OTC, PAFAH1B1, PARS2, PCCA, PCCB, PCNT, PCSK9, PDCD10, PDE3A, PDE4D, PGM1, PIK3C2A, PIK3CA, PIK3R2, PKD1, PKD2, PLAU, PLG, PLIN1, PLOD1, PLOD3, PMM2, PNP, POMGNT1, POMT1, POMT2, PRKAR1A, PRKG1, PROS1, PTEN, PTPN11, RAF1, RASA1, RASGRP2, RBBP8, RBM20, RELN, RNF213, ROBO4, RRAS2, RTTN, RXYLT1, SAMHD1, SCN1B, SCN2B, SCN5A, SCNN1B, SCNN1G, SERPINC1, SERPINE1, SETD5, SHOC2, SLC19A2, SLC2A10, SMAD3, SMAD4, SMAD9, SMARCAL1, SNORD118, SOS1, SOS2, SPARC, SPRED2, STAMBP, STAT1, STAT2, STIM1, STING1, TEK, TGFB2, TGFB3, TGFBR1, TGFBR2, THBD, THSD1, TNFRSF1A, TNXB, TPP2, TRAIP, TREX1, TSC1, TSC2, TTR, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, USP18, VHL, VLDLR, VWF, WDR62, WFS1, WNK4, WRN, YY1AP1

Cerebrovascular Disorders | 272 genes

The panel includes genes associated with stroke (e.g., *CADASIL, CARASIL*), disorders associated with increased risk of stroke (e.g., Fabry disease, coagulopathies), vascular malformations and vasculopathies (e.g., Moyamoya disease, Ehlers-Danlos syndrome). Please note that in this panel, variants in *CACNA1A* are only interrogated as sequence variants. If there is clinical suspicion of a repeat expansion disorder associated with *CACNA1A*, the Genome version of this panel is recommended.

Genes Included

ABCA1, ABCC6, ABCC9, ABCG5, ABCG8, ACAD9, ACE, ACP5, ACTA2, ACVRL1, ADA2, ADAMTS13, ADAMTS2, ADGRG1, AEBP1, AGXT, ANTXR1, APOA1, APOB, APP, ARX, ASS1, ATP7A, ATR, B3GALT6, B4GALT1, BRAF, C1R, CACNA1A, CBL, CBS, CCM2, CD59, CEP152, CEP63, CHD4, CISD2, CNOT3, COG6, COL1A1, COL3A1, COL4A1, COL4A2, COL5A1, COL5A2, COLGALT1, COQ2, COQ8A, CPAP, CPS1, CRB1, CSF1R, CST3, CTC1, CTSA, CYP11B1, CYP11B2, CYP26C1, DCX, DLD, DNA2, DOCK8, DPAGT1, DPM1, DPM3, DYRK1B, EFEMP2, ELN, ENG, ENPP1, EPAS1, EPHB4, EPOR, ERCC6, ERCC8, ESCO2, F10, F13A1, F13B, F2, F5, F7, F8, F9, FASTKD2, FBLN5, FBN1, FERMT3, FGA, FGB, FGG, FLNA, FLT4, FLVCR2, FOXC1, FOXE3, FOXF1, GAA, GANAB, GATA3, GCDH, GDF2, GFI1B, GGCX, GJA1, GJA5, GLA, GLMN, GNAQ, GP1BA, GPHN, GPR143, GUCY1A1, GYS1, HBB, HNF1B, HRAS, HSD11B2, HTRA1, IL1RN, IL6, ISCU, ITM2B, IVD, JAG1, JAK2, JAM3, KANSL1, KCNA5, KCNJ2, KCNQ1, KDR, KNG1, KRAS, KRIT1, LAMB1, LAMC3, LARGE1, LARS2, LDLR, LMAN1, LMBRD1, LMNA, LOX, LRPPRC, LTBP3, LZTR1, MAP2K1, MCCC1, MEF2C, MEFV, MFAP5, MFN2, MGAT2, MMACHC, MMUT, MOCS1, MOCS2, MPI, MTHFR, MYBPC3, MYH11, MYH7, MYLK, NBEAL2, NDE1, NF1, NLRP3, NOTCH1, NOTCH2, NOTCH3, NPPA, NRAS, OCLN, OPHN1, OTC, PAFAH1B1, PARS2, PCCA, PCCB, PCNT, PCSK9, PDCD10, PDE3A, PDE4D, PGM1, PIK3C2A, PIK3CA, PIK3R2, PKD1, PKD2, PLAU, PLG, PLIN1, PLOD1, PLOD3, PMM2, PNP, POMGNT1, POMT1, POMT2, PRKAR1A, PRKG1, PROS1, PTEN, PTPN11, RAF1, RASA1, RASGRP2, RBBP8, RBM20, RELN, RNF213, ROBO4, RRAS2, RTTN, RXYLT1, SAMHD1, SCN1B, SCN2B, SCN5A, SCNN1B, SCNN1G, SERPINC1, SERPINE1, SETD5, SHOC2, SLC19A2, SLC2A10, SMAD3, SMAD4, SMAD9, SMARCAL1, SOS1, SOS2, SPARC, SPRED2, STAMBP, STAT1, STAT2, STIM1, STING1, TEK, TGFB2, TGFB3, TGFBR1, TGFBR2, THBD, THSD1, TNFRSF1A, TNXB, TPP2, TRAIP, TREX1, TSC1, TSC2, TTR, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, USP18, VHL, VLDLR, VWF, WDR62, WFS1, WNK4, WRN, YY1AP1

Dementia Genome | 243 genes

The dementia panel Genome includes genes associated familial forms of Alzheimer's disease, Frontotemporal lobe degeneration and vascular dementia, neuromuscular diseases and movement disorders associated with cognitive deficits (e.g., ALS-dementia complex, hereditary forms of Parkinson's disease, and hyperkinetic disorders).

> Recommended dementia panel approach based on genome sequencing, including repeat expansion screening of the genes *ATXN1*, *ATXN2*, *C9orf72*, *PRNP*.

Genes Included

ABCA1, ABCC6, ABCD4, ACE, ACOX2, ADAM10, AKAP9, ALG1, ALPL, ALS2, AMH, ANG, ANGPTL3, ANK3, ANXA11, APOB, APOE, APP, ARHGAP31, ARPC1B, ARSA, ASCC1, ASRGL1, ASXL3, ATL1, ATP7B, ATXN1, ATXN2, BCKDK, BICD2, BIN1, BLOC1S3, BRCA2, BRD4, BSCL2, C5, C9orf72, CASP10, CCNF, CD2AP, CELF2, CELSR1, CEP290, CHCHD10, CHMP2B, CHRNA3, CHRNA4, CIC, CLCN1, CLIC5, COG4, COQ4, CP, CPA1, CRBN, CSF1R, CTSC, CUBN, CYLD, CYP27A1, DAAM2, DAO, DCTN1, DEF6, DNM1, DSG2, DTYMK, DYSF, ENO3, ERBB4, ERLIN2, EWSR1, EXOSC5, FBP1, FIG4, FN1, FOXG1, FOXRED1, FTL, FUS, GABBR2, GANAB, GCDH, GLE1, GLIS3, GPAA1, GRN, HELLS, HEXA, HFE, HID1, HMOX1, HNRNPA1, HNRNPA2B1, HSPD1, IFT140, ITGA8, ITM2B, JAG2, JPH2, KANSL1, KIF5A, KMT2E, KMT5B, KRT86, LAMC3, LRRK2, LTBP4, MAGI2, MAPK8IP3, MAPT, MATR3, MCM8, MEF2C, MMP20, MMP21, MSH3, 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, MTHFD1, MTO1, MYH9, MYRF, MYT1L, NANS, NEFH, NEK1, NOTCH3, NPC1, OBSCN, OGDH, OPTN, PACS2, PANK2, PAX2, PCSK9, PEX5, PFN1, PGAP3, PHGDH, PHIP, PIGU, PIP5K1C, PLAU, PLCG2, POLR3B, PRKAR1B, PRNP, PSEN1, PSEN2, PSMC3, PSTPIP1, QRICH2, QRSL1, RARS1, RBFOX2, REEP1, REL, RUSC2, SCAPER, SCN4A, SDCCAG8, SERAC1, SETD1B, SETX, SIGMAR1, SLC24A1, SLC24A4, SLC52A3, SMAD3, SNCA, SNCB, SOD1, SORL1, SPAST, SPATA7, SPEN, SPG11, SPTB, SQSTM1, SS18L1, STAB1, STARD7, STT3A, TARDBP, TBK1, TCOF1, TET3, TFG, TIA1, TMEM106B, TP53, TREM2, TRMT1, TSPOAP1, TTN, TUBA4A, TULP1, TYROBP, UBE3A, UBQLN2, VAPB, VCP, VKORC1, VPS13C, WASHC5, WDR11, WDR73

Dementia | 243 genes

The dementia panel includes genes associated familial forms of Alzheimer's disease, frontotemporal lobe degeneration and vascular dementia, neuromuscular diseases and movement disorders associated with cognitive deficits (e.g., ALS-dementia complex, hereditary forms of Parkinson's disease, and hyperkinetic disorders). Please note that in this panel, variants in *ATXN1*, *ATXN2*, *C9orf72*, *PRNP* are only interrogated as sequence variants. If there is clinical suspicion of a repeat expansion disorder associated with *ATXN1*, *ATXN2*, *C9orf72*, *PRNP*, the Genome version of this panel is recommended.

Genes Included

ABCA1, ABCC6, ABCD4, ACE, ACOX2, ADAM10, AKAP9, ALG1, ALPL, ALS2, AMH, ANG, ANGPTL3, ANK3, ANXA11, APOB, APOE, APP, ARHGAP31, ARPC1B, ARSA, ASCC1, ASRGL1, ASXL3, ATL1, ATP7B, ATXN1, ATXN2, BCKDK, BICD2, BIN1, BLOC1S3, BRCA2, BRD4, BSCL2, C5, C9orf72, CASP10, CCNF, CD2AP, CELF2, CELSR1, CEP290, CHCHD10, CHMP2B, CHRNA3, CHRNA4, CIC, CLCN1, CLIC5, COG4, COQ4, CP, CPA1, CRBN, CSF1R, CTSC, CUBN, CYLD, CYP27A1, DAAM2, DAO, DCTN1, DEF6, DNM1, DSG2, DTYMK, DYSF, ENO3, ERBB4, ERLIN2, EWSR1, EXOSC5, FBP1, FIG4, FN1, FOXG1, FOXRED1, FTL, FUS, GABBR2, GANAB, GCDH, GLE1, GLIS3, GPAA1, GRN, HELLS, HEXA, HFE, HID1, HMOX1, HNRNPA1, HNRNPA2B1, HSPD1, IFT140, ITGA8, ITM2B, JAG2, JPH2, KANSL1, KIF5A, KMT2E, KMT5B, KRT86, LAMC3, LRRK2, LTBP4, MAGI2, MAPK8IP3, MAPT, MATR3, MCM8, MEF2C, MMP20, MMP21, MSH3, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MTHFD1, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTO1, 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, MYH9, MYRF, MYT1L, NANS, NEFH, NEK1, NOTCH3, NPC1, OBSCN, OGDH, OPTN, PACS2, PANK2, PAX2, PCSK9, PEX5, PFN1, PGAP3, PHGDH, PHIP, PIGU, PIP5K1C, PLAU, PLCG2, POLR3B, PRKAR1B, PRNP, PSEN1, PSEN2, PSMC3, PSTPIP1, QRICH2, QRSL1, RARS1, RBFOX2, REEP1, REL, RUSC2, SCAPER, SCN4A, SDCCAG8, SERAC1, SETD1B, SETX, SIGMAR1, SLC24A1, SLC24A4, SLC52A3, SMAD3, SNCA, SNCB, SOD1, SORL1, SPAST, SPATA7, SPEN, SPG11, SPTB, SQSTM1, SS18L1, STAB1, STARD7, STT3A, TARDBP, TBK1, TCOF1, TET3, TFG, TIA1, TMEM106B, TP53, TREM2, TRMT1, TSPOAP1, TTN, TUBA4A, TULP1, TYROBP, UBE3A, UBQLN2, VAPB, VCP, VKORC1, VPS13C, WASHC5, WDR11, WDR73

Epilepsy Upgraded Genome | 1109 genes

The epilepsy panel Upgraded Genome includes genes associated with monogenic epilepsies (e.g., Dravet syndrome, GEFS-Plus, autosomal dominant nocturnal frontal lobe epilepsy, progressive myoclonus epilepsies, benign familial neonatal epilepsy, and rare monogenic forms of complex inherited epilepsies including genetic generalized and genetic focal epilepsies) as well as epilepsy-associated genes (e.g., Developmental and epileptic encephalopathy). This panel includes repeat expansion screening of the genes *ATN1, CACNA1A, CSTB* and *GBA1* conversion analysis screening. The panel includes the non-coding gene *RNU4-2* and *SNORD118*.

> Recommended epilepsy panel approach based on genome sequencing, including repeat expansion screening of the genes *ATN1, CACNA1A, CSTB* and *GBA1* conversion analysis with its pseudogene. The panel includes the non-coding genes *RNU4-2* and *SNORD118*.

Genes Included

AARS1, AARS2, AASS, ABAT, ABCA2, ABCC8, ABCD1, ACAD9, ACADM, ACADS, ACADVL, ACOX1, ACTL6B, ACY1, ADA, ADAM22, ADAMTS10, ADAMTSL2, ADAR, ADARB1, ADAT3, ADGRG1, ADGRV1, ADPRS, ADSL, AFF3, AFG2A, AFG2B, AFG3L2, AGA, AGK, AGO1, AGPS, AIFM1, AIMP1, AIMP2, AKT1, AKT3, ALDH3A2, ALDH5A1, ALDH7A1, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALPL, AMPD2, AMT, ANK2, ANK3, ANKRD11, ANO4, ANTXR2, AP1G1, AP2M1, AP3B1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APC2, APP, APTX, ARF1, ARF3, ARFGEF1, ARFGEF2, ARG1, ARHGAP31, ARHGEF9, ARID1B, ARSA, ARSB, ARV1, ARX, ASAH1, ASH1L, ASL, ASNS, ASPA, ASS1, ASXL1, ASXL3, ATM, ATN1, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2A2, ATP2B1, ATP5F1A, ATP5PO, ATP6AP1, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0C, ATP6V1A, ATP7A, ATP7B, ATRX, AUH, B3GALNT2, B3GLCT, B4GALT1, BAP1, BCAP31, BCKDHA, BCKDHB, BCORL1, BCS1L, BEST1, BET1, BLOC1S1, BLTP1, BOLA3, BORCS8, BRAF, BRAT1, BSCL2, BTD, C12orf57, C19orf12, C2orf69, CA5A, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1H, CACNA1I, CACNA2D2, CAD, CAMK2D, CAMK2G, CAMSAP1, CAPRIN1, CARS2, CASK, CASR, CAV1, CBL, CBS, CC2D2A, CCDC115, CCDC186, CCDC88A, CCDC88C, CCND2, CDC42BPB, CDK19, CDKL5, CELF2, CEP85L, CERS1, CERT1, CHAMP1, CHD1, CHD2, CHD4, CHD5, CHKA, CHMP2B, CHRNA2, CHRNA4, CHRNB2, CIC, CLCN2, CLCN3, CLCN4, CLCN6, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLTC, CNKSR2, CNNM2, CNPY3, CNTN2, CNTNAP1, CNTNAP2, COA7, COA8, COASY, COG1, COG3, COG4, COG5, COG6, COG7, COG8, COL11A2, COL18A1, COL2A1, COL4A1, COL4A2, COLGALT1, COQ2, COQ4, COQ6, COQ8A, COQ9, COX10, COX15, COX20, COX6B1, CP, CPLX1, CPS1, CPSF3, CPT1A, CPT2, CREBBP, CRELD1, CRPPA, CSF1R, CSNK2A1, CSNK2B, CSTB, CTC1, CTNNA2, CTNS, CTSA, CTSC, CTSD, CTSF, CTSK, CTU2, CUL3, CUL4B, CUX1, CUX2, CYFIP2, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DBT, DCAF17, DCX, DDC, DDX3X, DEAF1, DEGS1, DENND5A, DENND5B, DEPDC5, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHPS, DHRSX, DHX16, DHX30, DIAPH1, DKC1, DLAT, DLD, DLL1, DLL3, DMXL2, DNAJC5, DNAJC6, DNM1, DNM1L, DOCK6, DOCK7, DOLK, DPAGT1, DPH5, DPM1, DPM2, DPM3, DPYD, DPYS, DTYMK, DYM, DYNC1H1, DYRK1A, EARS2, ECHS1, EEF1A2, EFTUD2, EHMT1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF3F, EIF4A2, EMC10, EML1, EMX2, ENTPD1, EPG5, EPM2A, EPRS1, ERCC6, ERCC8, ESAM, ETFA, ETFB, ETFDH, ETHE1, EXOC7, EXOSC3, EXT2, F2, F5, FA2H, FAH, FAM50A, FAR1, FARS2, FARSB, FASTKD2, FBXL4, FBXO11, FBXO28, FDX2, FGF12, FGF13, FGFR3, FH, FHL1, FIG4, FKRP, FKTN, FLNA, FLVCR2, FOLR1, FOXG1, FOXRED1, FRMD5, FRRS1L, FTL, FUCA1, FUT8, FXYD2, FZR1, GAA, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GALC, GALNS, GALNT2, GALT, GAMT, GAN, GATAD2B, GATM, GBA1, GBE1, GCDH, GCH1, GCSH, GFAP, GFER, GFM1, GFM2, GFPT1, GJA1, GJB1, GJC2, GLA, GLB1, GLDC, GLI3, GLRA1, GLRA2, GLRB, GLS, GLUD1, GLUL, GLYCTK, GM2A, GMPPA, GNAO1, GNAQ, GNB1, GNB2, GNB5, GNE, GNPAT, GNPTAB, GNPTG, GNS, GOSR2, GOT2, GPAA1, GPC3, GPHN, GRIA2, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRM7, GRN, GSS, GTPBP2, GTPBP3, GUSB, H3-3A, H3-3B, HACE1, HADHA, HADHB, HAX1, HCCS, HCFC1, HCN1, HCN2, HECTD4, HECW2, HEPACAM, HERC2, HEXA, HEXB, HGSNAT, HIBCH, HID1, HIKESHI, HLCS, HMGCL, HMGCS2, HNRNPH2, HNRNPR, HNRNPU, HOXA1, HPDL, HPRT1, HRAS, HSD17B10, HSD17B4, HSPD1, HTRA1, HTRA2, HYCC1, IARS2, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, IKBKG, IQSEC2, IRF2BPL, ISCA2, ITPA, IVD, JAG1, JAM3, KARS1, KAT5, KAT8, KATNB1, KCNA1, KCNA2, KCNB1, KCNC1, KCNC2, KCND2, KCNH1, KCNH5, KCNJ1, KCNJ10, KCNJ11, KCNK4, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KCTD7, KDM6B, KIF1A, KIF2A, KIF5A, KIF5C, KIFBP, KLHL20, KMT2E, KMT5B, KPTN, KRAS, L2HGDH, LAMA2, LAMB1, LAMP2, LARGE1, LARS1, LAT, LDB3, LETM1, LGI1, LIAS, LIPA, LIPT1, LIPT2, LMBRD2, LMNB1, LMNB2, LNPK, LRPPRC, LSS, LYRM7, LYST, MACF1, MADD, MAF, MAGI2, MAGT1, MAN1B1, MAN2B1, MANBA, MAP2K1, MAP2K2, MARS2, MAST1, MAST3, MBD5, MBOAT7, MCCC1, MCCC2, MCM3AP, MCOLN1, MDH2, MECP2, MECR, MED11, MED12, MED17, MED27, MEF2C, MFF, MFN2, MFSD8, MGAT2, MGME1, MINPP1, MLC1, MLPH, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPDU1, MPI, MPV17, MRPS22, MSX1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MTFMT, MTHFR, MTHFS, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTOR, MTR, MTRFR, 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, MYO5A, MYOT, NACC1, NAGA, NAGLU, NAGS, NAPB, NARS1, NARS2, NAXD, NAXE, NBAS, NBEA, NCDN, NDE1, NDP, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NECAP1, NEDD4L, NEU1, NEUROD2, NEXMIF, NF1, NFE2L2, NFU1, NGLY1, NHLRC1, NOTCH1, NOTCH3, NPC1, NPC2, NPR2, NPRL2, NPRL3, NR4A2, NRAS, NRROS, NRXN1, NSD1, NSD2, NSDHL, NSRP1, NTRK2, NUBPL, NUP214, NUS1, OAT, OCLN, OCRL, OGDHL, OPA1, OPA3, OPHN1, OSGEP, OTC, OTUD6B, OTX2, OXR1, P4HTM, PACS1, PACS2, PAFAH1B1, PAH, PAK1, PANK2, PARS2, PC, PCCA, PCCB, PCDH12, PCDH19, PCDHGC4, PCLO, PCYT2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDYN, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14,

PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGK1, PGM1, PGM2L1, PHACTR1, PHGDH, PHYH, PIDD1, PIGA, PIGB, PIGC, PIGG, PIGH, PIGK, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIK3CA, PIK3R2, PIP5K1C, PLA2G6, PLAA, PLCB1, PLCG2, PLEKHG2, PLP1, PLPBP, PLXNA1, PMM2, PMP22, PMPCB, PNKP, PNPO, PNPT1, POLG, POLG2, POLR1C, POLR3A, POLR3B, POMGNT1, POMK, POMT1, POMT2, PPFIBP1, PPIL1, PPP2CA, PPP2R1A, PPP3CA, PPT1, PRDM8, PRF1, PRICKLE1, PRICKLE2, PRMT7, PRODH, PRPF8, PRPS1, PRRT2, PSAP, PSAT1, PSEN1, PSMB8, PSPH, PTCD3, PTCH1, PTEN, PTF1A, PTPN23, PTS, PUM1, PURA, PYCR2, QARS1, QDPR, RAB11A, RAB11B, RAB18, RAB27A, RAB3GAP1, RAB3GAP2, RAC3, RAI1, RALA, RALGAPA1, RANBP2, RARS1, RARS2, RELN, RFT1, RHOBTB2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF13, RNF2, RNF216, RNU4-2, RNU4ATAC, ROGDI, RORA, RORB, RPIA, RRM2B, RTN4IP1, RTTN, RUSC2, RYR2, SAMD12, SAMHD1, SATB1, SATB2, SCAF4, SCAMP5, SCARB2, SCN1A, SCN1B, SCN2A, SCN2B, SCN3A, SCN8A, SCN9A, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SEC23B, SEC24D, SELENOI, SEMA6B, SEPSECS, SERAC1, SERPINI1, SETBP1, SETD1A, SETD1B, SETD5, SGCE, SGSH, SHH, SHQ1, SIK1, SIX3, SLC12A3, SLC12A5, SLC13A5, SLC16A2, SLC17A5, SLC19A3, SLC1A2, SLC1A4, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC25A4, SLC2A1, SLC31A1, SLC32A1, SLC33A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC38A3, SLC39A8, SLC45A1, SLC4A10, SLC5A6, SLC6A1, SLC6A19, SLC6A5, SLC6A8, SLC7A7, SLC9A6, SMARCA2, SMARCC2, SMC1A, SMPD1, SMS, SNAP25, SNF8, SNORD118, SNX27, SON, SOX10, SPART, SPG11, SPG7, SPR, SPTAN1, SPTBN1, SPTBN4, SRD5A3, SSR4, ST3GAL3, ST3GAL5, STAG1, STAMBP, STARD7, STAT1, STIL, STRADA, STT3A, STX1B, STXBP1, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYN1, SYNCRIP, SYNE1, SYNGAP1, SYNJ1, SZT2, TACO1, TAF8, TANC2, TANGO2, TBC1D20, TBC1D24, TBC1D2B, TBCD, TBCE, TBCK, TBL1XR1, TCF4, TDP2, TEFM, TELO2, TET3, TFE3, TGFB1, TGIF1, TIAM1, TIMM50, TIMM8A, TINF2, TK2, TMEM106B, TMEM126A, TMEM165, TMEM199, TMEM222, TMEM63B, TMEM70, TMX2, TNPO2, TPK1, TPP1, TRA2B, TRAF7, TRAK1, TRAPPC12, TRAPPC4, TRAPPC6B, TRAPPC9, TREM2, TREX1, TRIM8, TRIP13, TRIT1, TRPM3, TRPM6, TRPS1, TRPV4, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN34, TSEN54, TSFM, TTC19, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUFM, TUSC3, TWNK, TYMP, TYROBP, U2AF2, UBA5, UBAP2L, UBE2A, UBE3A, UBR7, UFC1, UFM1, UFSP2, UGDH, UGP2, UMPS, UNC80, UPB1, UQCRQ, USP18, USP7, VAMP2, VARS1, VARS2, VCP, VLDLR, VPS11, VPS50, WARS2, WASF1, WDR37, WDR45, WDR45B, WDR62, WDR73, WFS1, WNK3, WWOX, XK, YIF1B, YIPF5, YWHAG, ZBTB18, ZDHHC9, ZEB2, ZFYVE26, ZIC2, ZMIZ1, ZMYM2, ZNF142, ZNF335, ZNFX1

Epilepsy Upgraded | 1107 genes

The epilepsy panel Upgraded includes genes associated with monogenic epilepsies (e.g., Dravet syndrome, GEFS-Plus, autosomal dominant nocturnal frontal lobe epilepsy, progressive myoclonus epilepsies, benign familial neonatal epilepsy, and rare monogenic forms of complex inherited epilepsies including genetic generalized and genetic focal epilepsies) as well as epilepsy-associated genes (e.g., Developmental and epileptic encephalopathy). Please note that in this panel, variants in *ATN1, CACNA1A, CSTB* are only interrogated as sequence variants. If there is clinical suspicion of a repeat expansion disorder associated with *ATN1, CACNA1A, CSTB*, the Genome version of this panel is recommended.

Genes Included

AARS1, AARS2, AASS, ABAT, ABCA2, ABCC8, ABCD1, ACAD9, ACADM, ACADS, ACADVL, ACOX1, ACTL6B, ACY1, ADA, ADAM22, ADAMTS10, ADAMTSL2, ADAR, ADARB1, ADAT3, ADGRG1, ADGRV1, ADPRS, ADSL, AFF3, AFG2A, AFG2B, AFG3L2, AGA, AGK, AGO1, AGPS, AIFM1, AIMP1, AIMP2, AKT1, AKT3, ALDH3A2, ALDH5A1, ALDH7A1, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALPL, AMPD2, AMT, ANK2, ANK3, ANKRD11, ANO4, ANTXR2, AP1G1, AP2M1, AP3B1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APC2, APP, APTX, ARF1, ARF3, ARFGEF1, ARFGEF2, ARG1, ARHGAP31, ARHGEF9, ARID1B, ARSA, ARSB, ARV1, ARX, ASAH1, ASH1L, ASL, ASNS, ASPA, ASS1, ASXL1, ASXL3, ATM, ATN1, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP2A2, ATP2B1, ATP5F1A, ATP5PO, ATP6AP1, ATP6AP2, ATP6V0A1, ATP6V0A2, ATP6V0C, ATP6V1A, ATP7A, ATP7B, ATRX, AUH, B3GALNT2, B3GLCT, B4GALT1, BAP1, BCAP31, BCKDHA, BCKDHB, BCORL1, BCS1L, BEST1, BET1, BLOC1S1, BLTP1, BOLA3, BORCS8, BRAF, BRAT1, BSCL2, BTD, C12orf57, C19orf12, C2orf69, CA5A, CACNA1A, CACNA1B, CACNA1C, CACNA1D, CACNA1E, CACNA1G, CACNA1H, CACNA1I, CACNA2D2, CAD, CAMK2D, CAMK2G, CAMSAP1, CAPRIN1, CARS2, CASK, CASR, CAV1, CBL, CBS, CC2D2A, CCDC115, CCDC186, CCDC88A, CCDC88C, CCND2, CDC42BPB, CDK19, CDKL5, CELF2, CEP85L, CERS1, CERT1, CHAMP1, CHD1, CHD2, CHD4, CHD5, CHKA, CHMP2B, CHRNA2, CHRNA4, CHRNB2, CIC, CLCN2, CLCN3, CLCN4, CLCN6, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, CLTC, CNKSR2, CNNM2, CNPY3, CNTN2, CNTNAP1, CNTNAP2, COA7, COA8, COASY, COG1, COG3, COG4, COG5, COG6, COG7, COG8, COL11A2, COL18A1, COL2A1, COL4A1, COL4A2, COLGALT1, COQ2, COQ4, COQ6, COQ8A, COQ9, COX10, COX15, COX20, COX6B1, CP, CPLX1, CPS1, CPSF3, CPT1A, CPT2, CREBBP, CRELD1, CRPPA, CSF1R, CSNK2A1, CSNK2B, CSTB, CTC1, CTNNA2, CTNS, CTSA, CTSC, CTSD, CTSF, CTSK, CTU2, CUL3, CUL4B, CUX1, CUX2, CYFIP2, CYP27A1, CYP2U1, CYP7B1, D2HGDH, DAG1, DARS1, DARS2, DBT, DCAF17, DCX, DDC, DDX3X, DEAF1, DEGS1, DENND5A, DENND5B, DEPDC5, DGUOK, DHCR24, DHCR7, DHDDS, DHFR, DHPS, DHRSX, DHX16, DHX30, DIAPH1, DKC1, DLAT, DLD, DLL1, DLL3, DMXL2, DNAJC5, DNAJC6, DNM1, DNM1L, DOCK6, DOCK7, DOLK, DPAGT1, DPH5, DPM1, DPM2, DPM3, DPYD, DPYS, DTYMK, DYM, DYNC1H1, DYRK1A, EARS2, ECHS1, EEF1A2, EFTUD2, EHMT1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF3F, EIF4A2, EMC10, EML1, EMX2, ENTPD1, EPG5, EPM2A, EPRS1, ERCC6, ERCC8, ESAM, ETFA, ETFB, ETFDH, ETHE1, EXOC7, EXOSC3, EXT2, F2, F5, FA2H, FAH, FAM50A, FAR1, FARS2, FARSB, FASTKD2, FBXL4, FBXO11, FBXO28, FDX2, FGF12, FGF13, FGFR3, FH, FHL1, FIG4, FKRP, FKTN, FLNA, FLVCR2, FOLR1, FOXG1, FOXRED1, FRMD5, FRRS1L, FTL, FUCA1, FUT8, FXYD2, FZR1, GAA, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRD, GABRG2, GAD1, GALC, GALNS, GALNT2, GALT, GAMT, GAN, GATAD2B, GATM, GBA1, GBE1, GCDH, GCH1, GCSH, GFAP, GFER, GFM1, GFM2, GFPT1, GJA1, GJB1, GJC2, GLA, GLB1, GLDC, GLI3, GLRA1, GLRA2, GLRB, GLS, GLUD1, GLUL, GLYCTK, GM2A, GMPPA, GNAO1,

GNAQ, GNB1, GNB2, GNB5, GNE, GNPAT, GNPTAB, GNPTG, GNS, GOSR2, GOT2, GPAA1, GPC3, GPHN, GRIA2, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRM7, GRN, GSS, GTPBP2, GTPBP3, GUSB, H3-3A, H3-3B, HACE1, HADHA, HADHB, HAX1, HCCS, HCFC1, HCN1, HCN2, HECTD4, HECW2, HEPACAM, HERC2, HEXA, HEXB, HGSNAT, HIBCH, HID1, HIKESHI, HLCS, HMGCL, HMGCS2, HNRNPH2, HNRNPR, HNRNPU, HOXA1, HPDL, HPRT1, HRAS, HSD17B10, HSD17B4, HSPD1, HTRA1, HTRA2, HYCC1, IARS2, IBA57, IDH2, IDS, IDUA, IER3IP1, IFIH1, IKBKG, IQSEC2, IRF2BPL, ISCA2, ITPA, IVD, JAG1, JAM3, KARS1, KAT5, KAT8, KATNB1, KCNA1, KCNA2, KCNB1, KCNC1, KCNC2, KCND2, KCNH1, KCNH5, KCNJ1, KCNJ10, KCNJ11, KCNK4, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KCTD7, KDM6B, KIF1A, KIF2A, KIF5A, KIF5C, KIFBP, KLHL20, KMT2E, KMT5B, KPTN, KRAS, L2HGDH, LAMA2, LAMB1, LAMP2, LARGE1, LARS1, LAT, LDB3, LETM1, LGI1, LIAS, LIPA, LIPT1, LIPT2, LMBRD2, LMNB1, LMNB2, LNPK, LRPPRC, LSS, LYRM7, LYST, MACF1, MADD, MAF, MAGI2, MAGT1, MAN1B1, MAN2B1, MANBA, MAP2K1, MAP2K2, MARS2, MAST1, MAST3, MBD5, MBOAT7, MCCC1, MCCC2, MCM3AP, MCOLN1, MDH2, MECP2, MECR, MED11, MED12, MED17, MED27, MEF2C, MFF, MFN2, MFSD8, MGAT2, MGME1, MINPP1, MLC1, MLPH, MMAA, MMAB, MMACHC, MMADHC, MMUT, MOCS1, MOCS2, MOGS, MPDU1, MPI, MPV17, MRPS22, MSX1, MT-ATP6, MT-ATP8, MT-CO1, MT-CO2, MT-CO3, MT-CYB, MTFMT, MTHFR, MTHFS, MT-ND1, MT-ND2, MT-ND3, MT-ND4, MT-ND4L, MT-ND5, MT-ND6, MTOR, MTR, MTRFR, 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, MYO5A, MYOT, NACC1, NAGA, NAGLU, NAGS, NAPB, NARS1, NARS2, NAXD, NAXE, NBAS, NBEA, NCDN, NDE1, NDP, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NECAP1, NEDD4L, NEU1, NEUROD2, NEXMIF, NF1, NFE2L2, NFU1, NGLY1, NHLRC1, NOTCH1, NOTCH3, NPC1, NPC2, NPR2, NPRL2, NPRL3, NR4A2, NRAS, NRROS, NRXN1, NSD1, NSD2, NSDHL, NSRP1, NTRK2, NUBPL, NUP214, NUS1, OAT, OCLN, OCRL, OGDHL, OPA1, OPA3, OPHN1, OSGEP, OTC, OTUD6B, OTX2, OXR1, P4HTM, PACS1, PACS2, PAFAH1B1, PAH, PAK1, PANK2, PARS2, PC, PCCA, PCCB, PCDH12, PCDH19, PCDHGC4, PCLO, PCYT2, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PDYN, PET100, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGAP1, PGK1, PGM1, PGM2L1, PHACTR1, PHGDH, PHYH, PIDD1, PIGA, PIGB, PIGC, PIGG, PIGH, PIGK, PIGM, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIK3CA, PIK3R2, PIP5K1C, PLA2G6, PLAA, PLCB1, PLCG2, PLEKHG2, PLP1, PLPBP, PLXNA1, PMM2, PMP22, PMPCB, PNKP, PNPO, PNPT1, POLG, POLG2, POLR1C, POLR3A, POLR3B, POMGNT1, POMK, POMT1, POMT2, PPFIBP1, PPIL1, PPP2CA, PPP2R1A, PPP3CA, PPT1, PRDM8, PRF1, PRICKLE1, PRICKLE2, PRMT7, PRODH, PRPF8, PRPS1, PRRT2, PSAP, PSAT1, PSEN1, PSMB8, PSPH, PTCD3, PTCH1, PTEN, PTF1A, PTPN23, PTS, PUM1, PURA, PYCR2, QARS1, QDPR, RAB11A, RAB11B, RAB18, RAB27A, RAB3GAP1, RAB3GAP2, RAC3, RAI1, RALA, RALGAPA1, RANBP2, RARS1, RARS2, RELN, RFT1, RHOBTB2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF13, RNF2, RNF216, RNU4ATAC, ROGDI, RORA, RORB, RPIA, RRM2B, RTN4IP1, RTTN, RUSC2, RYR2, SAMD12, SAMHD1, SATB1, SATB2, SCAF4, SCAMP5, SCARB2, SCN1A, SCN1B, SCN2A, SCN2B, SCN3A, SCN8A, SCN9A, SCO1, SCO2, SDHA, SDHAF1, SDHB, SDHD, SEC23B, SEC24D, SELENOI, SEMA6B, SEPSECS, SERAC1, SERPINI1, SETBP1, SETD1A, SETD1B, SETD5, SGCE, SGSH, SHH, SHQ1, SIK1, SIX3, SLC12A3, SLC12A5, SLC13A5, SLC16A2, SLC17A5, SLC19A3, SLC1A2, SLC1A4, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A15, SLC25A19, SLC25A20, SLC25A22, SLC25A3, SLC25A4, SLC2A1, SLC31A1, SLC32A1, SLC33A1, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC38A3, SLC39A8, SLC45A1, SLC4A10, SLC5A6, SLC6A1, SLC6A19, SLC6A5, SLC6A8, SLC7A7, SLC9A6, SMARCA2, SMARCC2, SMC1A, SMPD1, SMS, SNAP25, SNF8, SNX27, SON, SOX10, SPART, SPG11, SPG7, SPR, SPTAN1, SPTBN1, SPTBN4, SRD5A3, SSR4, ST3GAL3, ST3GAL5, STAG1, STAMBP, STARD7, STAT1, STIL, STRADA, STT3A, STX1B, STXBP1, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYN1, SYNCRIP, SYNE1, SYNGAP1, SYNJ1, SZT2, TACO1, TAF8, TANC2, TANGO2, TBC1D20, TBC1D24, TBC1D2B, TBCD, TBCE, TBCK, TBL1XR1, TCF4, TDP2, TEFM, TELO2, TET3, TFE3, TGFB1, TGIF1, TIAM1, TIMM50, TIMM8A, TINF2, TK2, TMEM106B, TMEM126A, TMEM165, TMEM199, TMEM222, TMEM63B, TMEM70, TMX2, TNPO2, TPK1, TPP1, TRA2B, TRAF7, TRAK1, TRAPPC12, TRAPPC4, TRAPPC6B, TRAPPC9, TREM2, TREX1, TRIM8, TRIP13, TRIT1, TRPM3, TRPM6, TRPS1, TRPV4, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN34, TSEN54, TSFM, TTC19, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TUFM, TUSC3, TWNK, TYMP, TYROBP, U2AF2, UBA5, UBAP2L, UBE2A, UBE3A, UBR7, UFC1, UFM1, UFSP2, UGDH, UGP2, UMPS, UNC80, UPB1, UQCRQ, USP18, USP7, VAMP2, VARS1, VARS2, VCP, VLDLR, VPS11, VPS50, WARS2, WASF1, WDR37, WDR45, WDR45B, WDR62, WDR73, WFS1, WNK3, WWOX, XK, YIF1B, YIPF5, YWHAG, ZBTB18, ZDHHC9, ZEB2, ZFYVE26, ZIC2, ZMIZ1, ZMYM2, ZNF142, ZNF335, ZNFX1

Neurostructural disorders Genome | 380 genes

The neurostructural disorders panel Genome includes genes associated with abnormalities in the development of the cerebral architecture, brain atrophy, cerebral angiopathies, cerebral malformation, intracerebral calcification and white matter abnormalities. The panel includes repeat expansion screening for the genes: *ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, C9orf72, CACNA1A, CSTB, FMR1, FXN, HTT, NOP56, PPP2R2B, PRNP, TBP.*

> Recommended panel approach based on genome sequencing, including repeat expansion screening of the genes *ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, C9orf72, CACNA1A, CSTB, FMR1, FXN, HTT, NOP56, PPP2R2B, PRNP, TBP. The panel includes the non-coding gene SNORD118.*

Genes Included

AAAS, AARS1, ABCB7, ABCC6, ABCD1, ABHD12, ACE, ACOX1, ACP5, ACTA2, ACVRL1, ADA2, ADAR, ADGRG1, AFG3L2, AIMP1, ALAS2, ALX1, ALX3, ALX4, AMPD2, ANKLE2, ANO10, ANTXR1, AP1S2, APOE, APP, APTX, ARFGEF2, ARSA, ARX, ASPA, ASPM, ATAD3A, ATCAY, ATM, ATN1, ATP1A3, ATP2B3, ATP7A, ATP8A2, ATR, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, B3GALNT2, B4GAT1, BEAN1, BIN1, BRF1, C9orf72, CA2, CA8, CACNA1A, CACNA1G, CACNA2D2, CAMTA1, CASK, CBL, CCDC88C, CCM2, CDH2, CDK5, CDON, CEP152, CEP63, CHCHD10, CHD4, CHMP1A, CHMP2B, CLCN2, CLN6, CLP1, CNOT3, COASY, COG5, COL18A1, COL3A1, COL4A1, COL4A2, COL5A1, COQ8A, COX20, CP, CPAP, CRB1, CRPPA, CSF1R, CST3, CSTB, CTC1, CTSA, CTSF, CUL4B, CWF19L1, CYLD, CYP27A1, CYP2U1, DAG1, DAGLA, DARS2, DCC, DCX, DDHD2, DDX3X, DKC1, DMXL2, DNA2, DNAJC19, DNAJC5, DNMT1, DYNC1H1, EARS2, EEF2, EFEMP1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELN, ELOVL4, ELOVL5, EMX2, ENG, EPHB4, EPM2A, ERCC2, ERCC6, EXOSC3, EXOSC8, FARSA, FARSB, FBN1, FGF14, FGFR1, FKRP, FKTN, FLNA, FLT4, FLVCR1, FLVCR2, FMR1, FOLR1, FOXC1, FOXF1, FOXG1, FRMD4A, FUS, FXN, GALC, GBA2, GDF2, GFAP, GJC2, GLA, GLMN, GMPPB, GNAQ, GOSR2, GPAA1, GRID2, GRM1, GRN, GUCY1A1, HAAO, HBB, HEXA, HEXB, HMGA2, HTRA1, HTT, IFIH1, IL6, ITM2B, ITPR1, JAG1, JAM3, KANSL1, KCNA1, KCNC3, KCND3, KCNJ10, KDR, KIF1C, KIF26A, KIFBP, KRIT1, L1CAM, LAMB1, LAMC3, LARGE1, MACF1, MAN2C1, MAPT, MARS2, MAT1A, MEF2C, MMACHC, MRE11, MSX2, MT-ATP6, MTPAP, MTTP, MVK, MYH11, MYORG, NAGLU, NDE1, NF1, NFASC, NHLRC1, NKX6-2, NOP56, NOTCH3, NPC1, NPC2, NRROS, NUS1, OCLN, OFD1, OPA3, OPHN1, OPTN, OTX2, PAFAH1B1, PANX1, PAX2, PAX6, PCDH12, PCLO, PCNT, PDCD10, PDGFB, PDGFRB, PDYN, PEX16, PHGDH, PI4KA, PIK3CA, PIK3R2, PITX2, PKD1, PKD2, PLA2G6, PLCG2, PLP1, PMPCA, PNKP, PNPLA6, POLG, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PPP2R2B, PRDM13, PRICKLE1, PRKCG, PRNP, PRRT2, PSEN1, PSEN2, PTEN, PTF1A, QRICH1, RAB3GAP1, RARS2, RASA1, RBBP8, RELN, RIN2, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF170, RNF213, RNF216, ROBO1, ROBO3, RTTN, RXYLT1, SACS, SAMHD1, SAR1B, SATB2, SCN2A, SCN8A, SCP2, SEMA3A, SEPSECS, SETD5, SETX, SIL1, SLC1A3, SLC20A2, SLC2A1, SLC2A10, SLC9A6, SMAD3, SMAD4, SMAD9, SMARCAL1, SMPD4, SNORD118, SNX14, SORL1, SOX10, SPG7, SPTBN2, SQSTM1, SRD5A3, STAMBP, STUB1, SUMF1, SURF1, SYNE1, TARDBP, TBK1, TBP, TDP1, TEK, TERT, TGFB2, TGFBR1, TGFBR2, TGM6, THSD1, TINF2, TMEM106B, TMEM240, TOE1, TPP1, TRAIP, TREM2, TREX1, TSC1, TSEN15, TSEN2, TSEN34, TSEN54, TTBK2, TTC19, TTPA, TTR, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TWNK, UBQLN2, UCHL1, USP18, VAMP1, VCAN, VCP, VLDLR, VPS13D, VPS35L, VPS53, VRK1, WBP2, WDR73, WDR81, WFS1, WNT7A, WWOX, XPR1, YY1AP1, ZFYVE26

Neurostructural disorders | 379 genes

The neurostructural disorders panel includes genes associated with abnormalities in the development of the cerebral architecture, brain atrophy, cerebral angiopathies, cerebral malformation, intracerebral calcification and white matter abnormalities. Variants in the *ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, C9orf72, CACNA1A, CSTB, FMR1, FXN, HTT, NOP56, PPP2R2B, PRNP, TBP* genes are interrogated as sequence variants. If there is clinical suspicion of a repeat expansion disorder associated with one or more of the abovementioned genes, the Genome version of this panel is recommended.

Genes Included

AAAS, AARS1, ABCB7, ABCC6, ABCD1, ABHD12, ACE, ACOX1, ACP5, ACTA2, ACVRL1, ADA2, ADAR, ADGRG1, AFG3L2, AIMP1, ALAS2, ALX1, ALX3, ALX4, AMPD2, ANKLE2, ANO10, ANTXR1, AP1S2, APOE, APP, APTX, ARFGEF2, ARSA, ARX, ASPA, ASPM, ATAD3A, ATCAY, ATM, ATN1, ATP1A3, ATP2B3, ATP7A, ATP8A2, ATR, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, B3GALNT2, B4GAT1, BEAN1, BIN1, BRF1, C9orf72, CA2, CA8, CACNA1A, CACNA1G, CACNA2D2, CAMTA1, CASK, CBL, CCDC88C, CCM2, CDH2, CDK5, CDON, CEP152, CEP63, CHCHD10, CHD4, CHMP1A, CHMP2B, CLCN2, CLN6, CLP1, CNOT3, COASY, COG5, COL18A1, COL3A1, COL4A1, COL4A2, COL5A1, COQ8A, COX20, CP, CPAP, CRB1, CRPPA, CSF1R, CST3, CSTB, CTC1, CTSA, CTSF, CUL4B, CWF19L1, CYLD, CYP27A1, CYP2U1, DAG1, DAGLA, DARS2, DCC, DCX, DDHD2, DDX3X, DKC1, DMXL2, DNA2, DNAJC19, DNAJC5, DNMT1, DYNC1H1, EARS2, EEF2, EFEMP1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELN, ELOVL4, ELOVL5, EMX2, ENG, EPHB4, EPM2A, ERCC2, ERCC6, EXOSC3, EXOSC8, FARSA, FARSB, FBN1, FGF14, FGFR1, FKRP, FKTN, FLNA, FLT4, FLVCR1, FLVCR2, FMR1, FOLR1, FOXC1, FOXF1, FOXG1, FRMD4A, FUS, FXN, GALC, GBA2, GDF2, GFAP, GJC2, GLA, GLMN, GMPPB, GNAQ, GOSR2, GPAA1, GRID2, GRM1, GRN, GUCY1A1, HAAO, HBB, HEXA, HEXB, HMGA2, HTRA1, HTT, IFIH1, IL6, ITM2B, ITPR1, JAG1, JAM3, KANSL1, KCNA1, KCNC3, KCND3,

KCNJ10, KDR, KIF1C, KIF26A, KIFBP, KRIT1, L1CAM, LAMB1, LAMC3, LARGE1, MACF1, MAN2C1, MAPT, MARS2, MAT1A, MEF2C, MMACHC, MRE11, MSX2, MT-ATP6, MTPAP, MTTP, MVK, MYH11, MYORG, NAGLU, NDE1, NF1, NFASC, NHLRC1, NKX6-2, NOP56, NOTCH3, NPC1, NPC2, NRROS, NUS1, OCLN, OFD1, OPA3, OPHN1, OPTN, OTX2, PAFAH1B1, PANX1, PAX2, PAX6, PCDH12, PCLO, PCNT, PDCD10, PDGFB, PDGFRB, PDYN, PEX16, PHGDH, PI4KA, PIK3CA, PIK3R2, PITX2, PKD1, PKD2, PLA2G6, PLCG2, PLP1, PMPCA, PNKP, PNPLA6, POLG, POLR3A, POLR3B, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PPP2R2B, PRDM13, PRICKLE1, PRKCG, PRNP, PRRT2, PSEN1, PSEN2, PTEN, PTF1A, QRICH1, RAB3GAP1, RARS2, RASA1, RBBP8, RELN, RIN2, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF170, RNF213, RNF216, ROBO1, ROBO3, RTTN, RXYLT1, SACS, SAMHD1, SAR1B, SATB2, SCN2A, SCN8A, SCP2, SEMA3A, SEPSECS, SETD5, SETX, SIL1, SLC1A3, SLC20A2, SLC2A1, SLC2A10, SLC9A6, SMAD3, SMAD4, SMAD9, SMARCAL1, SMPD4, SNX14, SORL1, SOX10, SPG7, SPTBN2, SQSTM1, SRD5A3, STAMBP, STUB1, SUMF1, SURF1, SYNE1, TARDBP, TBK1, TBP, TDP1, TEK, TERT, TGFB2, TGFBR1, TGFBR2, TGM6, THSD1, TINF2, TMEM106B, TMEM240, TOE1, TPP1, TRAIP, TREM2, TREX1, TSC1, TSEN15, TSEN2, TSEN34, TSEN54, TTBK2, TTC19, TTPA, TTR, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TWNK, UBQLN2, UCHL1, USP18, VAMP1, VCAN, VCP, VLDLR, VPS13D, VPS35L, VPS53, VRK1, WBP2, WDR73, WDR81, WFS1, WNT7A, WWOX, XPR1, YY1AP1, ZFYVE26

Abnormalities in the Development of Cerebral Architecture Genome | 178 genes

The abnormalities in the development of cerebral architecture panel Genome includes genes associated with disorders affecting neuronal proliferation and migration, disorders affecting cortical organization and multiple aspects of cortical development, vascular malformations affecting brain structure, and regional hypoplasia. The panel includes repeat expansion screening of *FMR1*.

Recommended panel approach based on genome sequencing, including repeat expansion screening of *FMR1*.

Genes Included

ABCC6, ACE, ACTA2, ACVRL1, ADA2, ADGRG1, ALX1, ALX3, ALX4, AMPD2, ANKLE2, ANTXR1, APP, ARFGEF2, ARX, ASPM, ATAD3A, ATP7A, ATP8A2, ATR, B3GALNT2, B4GAT1, BRF1, CA2, CA8, CACNA1G, CASK, CBL, CCM2, CDH2, CDK5, CDON, CEP152, CEP63, CHD4, CHMP1A, CLP1, CNOT3, COASY, COL18A1, COL3A1, COL4A1, COL4A2, COL5A1, CPAP, CRB1, CRPPA, CTSA, CUL4B, CWF19L1, DAG1, DCC, DCX, DDX3X, DKC1, DNA2, ELN, EMX2, ENG, EPHB4, EXOSC3, EXOSC8, FBN1, FGFR1, FKRP, FKTN, FLNA, FLT4, FLVCR2, FMR1, FOXF1, FOXG1, FRMD4A, GDF2, GLA, GLMN, GMPPB, GNAQ, GUCY1A1, HBB, HMGA2, HTRA1, IL6, ITPR1, JAG1, KANSL1, KCNC3, KDR, KIFBP, KRIT1, L1CAM, LAMB1, LAMC3, LARGE1, MACF1, MEF2C, MSX2, MYH11, NDE1, NF1, NOTCH3, OCLN, OFD1, OPHN1, OTX2, PAFAH1B1, PAX6, PCLO, PCNT, PDCD10, PHGDH, PI4KA, PIK3CA, PIK3R2, PKD1, PKD2, PMPCA, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PRDM13, PTEN, PTF1A, RAB3GAP1, RARS2, RASA1, RBBP8, RELN, RIN2, RNF213, ROBO1, ROBO3, RTTN, RXYLT1, SAMHD1, SATB2, SEMA3A, SEPSECS, SETD5, SLC2A10, SMAD3, SMAD4, SMAD9, SMARCAL1, SMPD4, SNX14, SPTBN2, STAMBP, STUB1, TEK, TERT, TGFB2, TGFBR1, TGFBR2, THSD1, TINF2, TOE1, TRAIP, TSC1, TSEN15, TSEN2, TSEN34, TSEN54, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, VCAN, VLDLR, VPS35L, VPS53, VRK1, WDR81, YY1AP1

Abnormalities in the Development of Cerebral Architecture | 178 genes

The abnormalities in the development of cerebral architecture panel includes genes associated with disorders affecting neuronal proliferation and migration, disorders affecting cortical organization and multiple aspects of cortical development, vascular malformations affecting brain structure, and regional hypoplasia. *FMR1* repeat expansion analysis is not part of this panel; variants in this gene are only interrogated as sequence variants. If there is clinical suspicion associated with *FMR1* repeat expansion variants, the Genome version of this panel is recommended.

Genes Included

ABCC6, ACE, ACTA2, ACVRL1, ADA2, ADGRG1, ALX1, ALX3, ALX4, AMPD2, ANKLE2, ANTXR1, APP, ARFGEF2, ARX, ASPM, ATAD3A, ATP7A, ATP8A2, ATR, B3GALNT2, B4GAT1, BRF1, CA2, CA8, CACNA1G, CASK, CBL, CCM2, CDH2, CDK5, CDON, CEP152, CEP63, CHD4, CHMP1A, CLP1, CNOT3, COASY, COL18A1, COL3A1, COL4A1, COL4A2, COL5A1, CPAP, CRB1, CRPPA, CTSA, CUL4B, CWF19L1, DAG1, DCC, DCX, DDX3X, DKC1, DNA2, ELN, EMX2, ENG, EPHB4, EXOSC3, EXOSC8, FBN1, FGFR1, FKRP, FKTN, FLNA, FLT4, FLVCR2, FMR1, FOXF1, FOXG1, FRMD4A, GDF2, GLA, GLMN, GMPPB, GNAQ, GUCY1A1, HBB, HMGA2, HTRA1, IL6, ITPR1, JAG1, KANSL1, KCNC3, KDR, KIFBP, KRIT1, L1CAM, LAMB1, LAMC3, LARGE1, MACF1, MEF2C, MSX2, MYH11, NDE1, NF1, NOTCH3, OCLN, OFD1, OPHN1, OTX2, PAFAH1B1, PAX6, PCLO, PCNT, PDCD10, PHGDH, PI4KA, PIK3CA, PIK3R2, PKD1, PKD2, PMPCA, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PRDM13, PTEN, PTF1A, RAB3GAP1, RARS2, RASA1, RBBP8, RELN, RIN2, RNF213, ROBO1, ROBO3, RTTN, RXYLT1, SAMHD1, SATB2, SEMA3A, SEPSECS, SETD5, SLC2A10, SMAD3, SMAD4, SMAD9, SMARCAL1, SMPD4, SNX14, SPTBN2, STAMBP, STUB1, TEK, TERT, TGFB2, TGFBR1, TGFBR2, THSD1, TINF2, TOE1, TRAIP, TSC1, TSEN15, TSEN2, TSEN34, TSEN54, TUBA1A, TUBB, TUBB2A, TUBB2B, TUBB3, TUBG1, VCAN, VLDLR, VPS35L, VPS53, VRK1, WDR81, YY1AP1

Cerebral Angiopathies | 16 genes

The cerebral angiopathies panel includes genes associated with cerebral amyloid angiopathy, familial cerebral cavernous malformation (CCM) and hereditary hemorrhagic telangiectasia (HHT).

Genes Included

ACOX1, APOE, APP, COL4A1, COL4A2, CST3, EFEMP1, FOXC1, HAAO, HTRA1, ITM2B, NOTCH3, PITX2, TREX1, TTR, WBP2

Brain Atrophy Genome | 181 genes

The brain atrophy panel Genome includes associated with generalized brain atrophy and focal brain atrophy covering frontotemporal dementia, Alzheimer's disease and spinocerebellar ataxia. The panel includes repeat expansion screening for the genes: *ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, C9orf72, CACNA1A, CSTB, FMR1, FXN, HTT, NOP56, PPP2R2B, PRNP, TBP*.

> Recommended panel approach based on genome sequencing, including repeat expansion screening of the genes *ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, C9orf72, CACNA1A, CSTB, FMR1, FXN, HTT, NOP56, PPP2R2B, PRNP, TBP*.

Genes Included

AAAS, AARS1, ABCB7, ABHD12, AFG3L2, ALAS2, AMPD2, ANO10, AP1S2, APOE, APP, APTX, ARSA, ATCAY, ATM, ATN1, ATP1A3, ATP2B3, ATP8A2, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, BEAN1, BIN1, C9orf72, CA8, CACNA1A, CACNA1G, CACNA2D2, CAMTA1, CASK, CCDC88C, CHCHD10, CHMP1A, CHMP2B, CLCN2, CLN6, CLP1, COG5, COQ8A, COX20, CP, CSTB, CTSF, CWF19L1, CYLD, CYP27A1, CYP2U1, DAGLA, DARS2, DDHD2, DMXL2, DNAJC19, DNAJC5, DNMT1, DYNC1H1, EEF2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, EPM2A, EXOSC3, FGF14, FLVCR1, FMR1, FOLR1, FUS, FXN, GBA2, GFAP, GJC2, GOSR2, GPAA1, GRID2, GRM1, GRN, HEXA, HEXB, HTT, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1C, MAPT, MARS2, MMACHC, MRE11, MT-ATP6, MTPAP, MTTP, MVK, NAGLU, NFASC, NHLRC1, NKX6-2, NOP56, NPC1, NPC2, NUS1, OPA3, OPHN1, OPTN, PAX2, PAX6, PCLO, PDYN, PEX16, PLA2G6, PLCG2, PMPCA, PNKP, PNPLA6, POLG, POLR3A, PPP2R2B, PRICKLE1, PRKCG, PRNP, PRRT2, PSEN1, PSEn2, RARS2, RELN, RNF170, RNF216, SACS, SAR1B, SCN8A, SEPSECS, SETX, SIL1, SLC1A3, SLC2A1, SLC9A6, SNX14, SORL1, SPG7, SPTBN2, SQSTM1, SRD5A3, STUB1, SYNE1, TARDBP, TBK1, TBP, TDP1, TGM6, TMEM106B, TMEM240, TPP1, TREM2, TSEN2, TSEN34, TSEN54, TTBK2, TTC19, TTPA, TUBB4A, TWNK, UBQLN2, UCHL1, VAMP1, VCP, VLDLR, VPS13D, VPS53, VRK1, WDR73, WDR81, WFS1, WWOX, ZFYVE26

Brain Atrophy | 181 genes

The brain atrophy panel includes genes associated with generalized brain atrophy and focal brain atrophy covering frontotemporal dementia, Alzheimer's disease and spinocerebellar ataxia. Variants in the *ATN1, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, C9orf72, CACNA1A, CSTB, FMR1, FXN, HTT, NOP56, PPP2R2B, PRNP, TBP* genes are interrogated as sequence variants. If there is clinical suspicion of a repeat expansion disorder associated with one or more of the above mentioned genes, the Genome version of this panel is recommended.

Genes Included

AAAS, AARS1, ABCB7, ABHD12, AFG3L2, ALAS2, AMPD2, ANO10, AP1S2, APOE, APP, APTX, ARSA, ATCAY, ATM, ATN1, ATP1A3, ATP2B3, ATP8A2, ATXN1, ATXN10, ATXN2, ATXN3, ATXN7, ATXN8OS, BEAN1, BIN1, C9orf72, CA8, CACNA1A, CACNA1G, CACNA2D2, CAMTA1, CASK, CCDC88C, CHCHD10, CHMP1A, CHMP2B, CLCN2, CLN6, CLP1, COG5, COQ8A, COX20, CP, CSTB, CTSF, CWF19L1, CYLD, CYP27A1, CYP2U1, DAGLA, DARS2, DDHD2, DMXL2, DNAJC19, DNAJC5, DNMT1, DYNC1H1, EEF2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, EPM2A, EXOSC3, FGF14, FLVCR1, FMR1, FOLR1, FUS, FXN, GBA2, GFAP, GJC2, GOSR2, GPAA1, GRID2, GRM1, GRN, HEXA, HEXB, HTT, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1C, MAPT, MARS2, MMACHC, MRE11, MT-ATP6, MTPAP, MTTP, MVK, NAGLU, NFASC, NHLRC1, NKX6-2, NOP56, NPC1, NPC2, NUS1, OPA3, OPHN1, OPTN, PAX2, PAX6, PCLO, PDYN, PEX16, PLA2G6, PLCG2, PMPCA, PNKP, PNPLA6, POLG, POLR3A, PPP2R2B, PRICKLE1, PRKCG, PRNP, PRRT2, PSEN1, PSEn2, RARS2, RELN, RNF170, RNF216, SACS, SAR1B, SCN8A, SEPSECS, SETX, SIL1, SLC1A3, SLC2A1, SLC9A6, SNX14, SORL1, SPG7, SPTBN2, SQSTM1, SRD5A3, STUB1, SYNE1, TARDBP, TBK1, TBP, TDP1, TGM6, TMEM106B, TMEM240, TPP1, TREM2, TSEN2, TSEN34, TSEN54, TTBK2, TTC19, TTPA, TUBB4A, TWNK, UBQLN2, UCHL1, VAMP1, VCP, VLDLR, VPS13D, VPS53, VRK1, WDR73, WDR81, WFS1, WWOX, ZFYVE26

Cerebral Malformations | 23 genes

The cerebral malformations panel includes genes associated with lissencephaly and subcortical band heterotopia, polymicrogyria, and microcephaly.

Genes Included

ANKLE2, ARFGEF2, ARX, ASPM, CCM2, COL18A1, DCX, EMX2, FLNA, KIF26A, KIFBP, MAN2C1, PAFAH1B1, PANX1, PAX6, PDCD10, QRICH1, RAB3GAP1, RELN, SCN2A, TUBA1A, VLDLR, WNT7A

Intracerebral Calcification Genome | 30 genes

This panel includes genes associated with intracerebral calcification disorders. The non-coding gene *SNORD118* is also part of the genes of the panel.

Recommended panel approach based on genome sequencing, including the gene *SNORD118*.

Genes Included

ACP5, ADAR, AP1S2, COL4A1, CTC1, CYP2U1, ERCC6, FARSA, FARSB, IFIH1, JAM3, MAT1A, MYORG, NRROS, OCLN, PCDH12, PDGFB, PDGFRB, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SCN2A, SLC20A2, SNORD118, TINF2, TREM2, TREX1, USP18, XPR1

Intracerebral Calcification | 29 genes

This panel includes genes associated with intracerebral calcification disorders.

Genes Included

ACP5, ADAR, AP1S2, COL4A1, CTC1, CYP2U1, ERCC6, FARSA, FARSB, IFIH1, JAM3, MAT1A, MYORG, NRROS, OCLN, PCDH12, PDGFB, PDGFRB, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SCN2A, SLC20A2, TINF2, TREM2, TREX1, USP18, XPR1

White matter abnormalities | 39 genes

The white matter abnormalities panel includes genes associated with disorders such as hypomyelinating leukodystrophies, demyelinating leukodystrophies, and leukodystrophies with myelin vacuolization.

Genes Included

ABCD1, ADAR, AIMP1, ASPA, CLCN2, COL4A1, COL4A2, CSF1R, CYP27A1, DARS2, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ERCC2, FOXC1, GALC, GFAP, GJC2, GLA, MAPT, NOTCH3, PLP1, POLR3A, POLR3B, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, SACS, SCP2, SMARCAL1, SOX10, SUMF1, SURF1, TREX1, TUBB4A

Headache, migraine, sleep disorders, hyperekplexia, and paroxysmal disorders

Headache, Migraine, Sleep Disorders, Hyperekplexia, and Paroxysmal Disorders | 21 genes

This panel includes genes associated with familial hemiplegic episodic ataxia and CADASIL, sleep disorders, and hyperekplexia and paroxysmal disorders.

Genes Included

AIP, APC, ATM, BAP1, BRCA2, CDKN2A, CREBBP, ELP1, MLH1, MSH2, MSH6, NF1, NF2, PALB2, PHOX2B, PMS2, PTCH1, SMARCA4, SMARCB1, SUFU, TP53, TSC1, TSC2, VHL

Neurooncology | 24 genes

This panel includes genes associated with familial hemiplegic episodic ataxia and CADASIL, sleep disorders, and hyperekplexia and paroxysmal disorders.

Genes Included

AIP, APC, ATM, BAP1, BRCA2, CDKN2A, CREBBP, ELP1, MLH1, MSH2, MSH6, NF1, NF2, PALB2, PHOX2B, PMS2, PTCH1, SMARCA4, SMARCB1, SUFU, TP53, TSC1, TSC2, VHL

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