

## TIGER Panels Information Sheet

1. Ataxia (219 genes)
2. Charcot-Marie-Tooth (116 genes)
3. Complex Immune Dysregulation (641 genes)
4. Epilepsy (765 genes)
5. Growth and Related Disorders (520 genes)
6. Retinal Dystrophy (314 genes)
7. Movement Disorders (609 genes)
8. Spastic Paraplegia (75 genes)

### GENE CONTENT

#### Ataxia Panel (219 genes)

AARS2, ABCA2, ABCB7, ABCD1, ABHD12, ACO2, ADPRS, AFG3L2, AGTPBP1, AHI1, ALDH5A1, ANO10, APTX, ARL13B, ARL6, ATCAY, ATM, ATP13A2, ATP1A3, ATP2B3, ATP7B, ATP8A2, AUH, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEAN1, C12ORF4, CA8, CACNA1A, CACNA1G, CACNA2D2, CACNB4, CAMTA1, CAPN1, CASK, CC2D2A, CCDC88C, CEP290, CEP41, CHCHD10, CLCN2, CLN5, CLN6, CLN8, CLPB, CLPP, COA7, COASY, COQ2, COQ4, COQ8A, COX20, CP, CPLANE1, CSTB, CTBP1, CTDPI1, CTSB, CWF19L1, CYP27A1, CYP2U1, CYP7B1, DHPS, DNAJC19, DNAJC5, DNMT1, DOCK3, EBF3, EEF2, ELOVL4, ELOVL5, FA2H, FBXL4, FDXR, FGF14, FLVCR1, FMR1, FXN, GBA2, GFAP, GOSR2, GRID2, GRM1, GSS, HARS2, HEXB, HIBCH, INPP5E, IRF2BPL, ITM2B, ITPR1, KCNA1, KCNC3, KCND3, KCNJ10, KIF1C, KIF5A, KIF7, LAMA1, LARS2, LMNB1, LRPPRC, LRSAM1, MARS2, MECR, MGME1, MKKS, MKS1, MME, MRE11, MSTO1, MTFMT, MTPAP, MTPP, NDUFAF6, NDUFS2, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NKX2-1, NKX2-2, NOL3, NPC1, NPC2, NPHP1, NUBPL, OFD1, OPA1, OPHN1, PANK2, PAX6, PDYN, PEX10, PEX16, PEX2, PEX3, PEX6, PEX7, PHYH, PMM2, PNKD, PNKP, PNPLA6, POLG, PRKCG, PRRT2, PUM1, RNF216, RORA, RPGRIP1L, RUBCN, SACS, SAMD9L, SCYL1, SERAC1, SETX, SH3TC2, SIL1, SLC17A5, SLC1A3, SLC20A2, SLC25A46, SLC2A1, SLC52A2, SLC9A1, SLC9A6, SNX14, SPART, SPG11, SPG7, SPTBN2, SQSTM1, STUB1, STXB1, SUOX, SYNE1, SYT14, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TGM6, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TPP1, TRAPPC11, TRIM32, TTBK2, TTC19, TTC8, TTPA, TUBB4A, TWNK, UBA5, UBTX, UCHL1, VAMP1, VLDLR, VPS13D, VWA3B, WDPCP, WDR81, WFS1, WWOX, XRCC1, ZFYVE26, ZNF423

#### Charcot-Marie-Tooth Panel (116 genes)

AARS1, AGTPBP1, AIFM1, AMACR, ARHGEF10, ATAD3A, ATL1, ATL3, ATP1A1, ATP7A, BAG3, BICD2, BSCL2, CCT5, CHCHD10, COA7, COX10, COX6A1, CTDPI1, CYP27A1, DCAF8, DCTN1, DHTKD1, DNAJB2, DNM2, DNMT1, DST, DYNC1H1, EGR2, ELP1, FBLN5, FBXO38, FGD4, FIG4, FXN, GAN, GARS1, GDAP1, GJB1, GNB4, GNE, GSN, HADHB, HARS1, HINT1, HK1, HSPB1, HSPB8, IGHMBP2, INF2, KARS1, KIF1A, KIF1B, KIF5A, LDB3, LITAF, LMNA, LRSAM1, MARS1, MCM3AP, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, MTRFR, MYOT, NDRG1, NEFH, NEFL, NGF, NTRK1, PDK3, PLEKHG5, PMP2, PMP22, PNKP, POLG, POLG2, PRDM12, PRPS1, PRX, RAB7A, REEP1, RETREG1, SACS, SBF1, SBF2, SCN11A, SCN9A, SCYL1, SEPTIN9, SETX, SH3TC2, SLC12A6, SLC25A46, SLC52A2, SLC52A3, SMAD3, SPG11, SPTBN4, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TTR, TYMP, UBA1, VCP, WNK1, YARS1, ZFYVE26

#### Complex Immune Dysregulation Panel (641 genes)

ABCB7, ABCG5, ABCG8, ACD, ACP5, ACTB, ACTG1, ACTN1, ADA, ADA2, ADAM17, ADAMTS13, ADAMTS3, ADAR, ADIPOQ, ADIPOR1, ADIPOR2, AICDA, AIRE, AK2, ALAS2, ALPI, ANKRD11, ANKRD26, AP1S3, AP3B1, AP3D1, APOL1, ARHGEF1, ARPC1B, ATM, ATP6AP1, ATR, ATRX, B2M, BACH2, BCL10, BCL11B, BCO1, BLM, BLNK, BLOC1S3, BLOC1S6, BRAF, BRCA1, BRCA2, BRIP1, BTK, C1QA, C1QB, C1QBP, C1QC, C1R, C1S, C2, C3, C3AR1, C4BPA, C4BPB, C5, C5AR1, C5AR2, C6, C7, C8A, C8B, C8G, C9, CARD11, CARD14, CARD9, CARMIL2, CASP10, CASP8, CAVIN1, CBL, CCBE1, CCDC103, CCDC39, CCDC40, CCDC65, CCNK, CCNO, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CD93, CDAN1, CDC42, CDCA7, CDIN1, CDK9, CDKN2A, CEPPA, CEBPB, CENPF, CFAP298, CFAP300, CFB, CFD, CFH, CFHR5, CFI, CFP, CFTR, CHD7, CHEK2, CIB1, CIITA, CLCN7, CLEC7A, CLPB, CLU, COG6, COLEC11, COPA, CORO1A, CPT2, CR2, CREBBP, CRP, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTNBL1, CTSP1, CTSC, CXCR2, CXCR4, CYBA, CYBB, CYBC1, CYCS, CYP27A1, DBR1, DCLRE1B, DCLRE1C, DDX11, DDX41, DDX58, DEF6, DGAT1, DGKE, DHFR, DKC1, DNAAF1, DNAAF11, DNAAF2, DNAAF3, DNAAF4, DNAAF5, DNAAF6, DNAH1, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAJC21, DNAL1, DNASE1L3, DNASE2, DNMT3B, DOCK2, DOCK8, DRC1, DTNBP1, EFL1, EIF2AK3, ELANE, EP300, EPCAM, EPG5, EPO, ERBIN, ERCC2, ERCC3, ERCC4, ERCC6L2, ETV6, EXTL3, FAAP100, FAAP24, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANGC, FANCI, FANCL, FANCM, FAS, FASLG, FAT4, FCGR3A, FCHO1, FCN1, FCN2, FCN3, FERMT1, FERMT3, FLG, FLI1, FLNA, FOXN1, FOXP3, FPR1, FYB1, G6PC3, G6PD, GAS2L2, GAS8, GATA1, GATA2, GBA, GF11, GF11B, GINS1, GLRX5, GNE, GP1BA, GP1BB, GP9, GTF2H5, GUCY2C, HAVCR2, HAX1, HELLS, HMOX1, HNRNP, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, HSPA9, HYDIN, HYOU1, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNGR1, IFNGR2, IGLL1, IKBKB, IKZF1, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL12RB2, IL17F, IL17RA, IL17RC, IL18BP, IL1RN, IL21, IL21R, IL23R, IL2RA, IL2RB, IL2RG, IL36RN, IL6R, IL6ST, IL7R, INO80, IRAK1, IRAK4, IRF2BP2, IRF3, IRF4, IRF7, IRF8, IRF9, ISG15, ITCH, ITGA2, ITGA2B, ITGB2, ITGB3, ITK, JAGN1, JAK1, JAK2, JAK3, KDM1A, KDM6A, KIF23, KLF1, KMT2A, KMT2D, KRAS, LAMTOR2, LAT, LCK, LCT, LIG1, LIG4, LIPA, LPIN2, LRBA, LRRC8A, LYST, LZTR1, MAD2L2, MAGT1, MALT1, MAN2B1, MANBA, MAP2K1, MAP2K2, MAP3K14, MAP3K8, MASP1, MASP2, MASTL, MAT2A, MBL2, MCDIAS, MCM4, MECOM, MEFV, MLH1, MLPH, MOGS, MPL, MPO, MRAS, MRE11, MS4A1, MSH2, MSH6, MSN, MTHFD1, MVK, MYD88, MYH9, MYO5A, MYO5B, MYSM1, NAF1, NBAS, NBEAL2, NBN, NCF1, NCF2, NCF4, NCKAP1L, NCSTN, NEUROG3, NF1, NFAT5, NFE2L2, NFIL3, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLR4, NLRP1, NLRP12, NLRP3, NME8, NOD2, NOP10, NRAS, NSMCE2, NSMCE3, NUP214, OAS1, ODAD1, ODAD2, OFD1, ORAI1, OSTM1, OTUD6B, OTULIN, PALB2, PARN, PAX5, PEPD, PGM3, PIGA, PIK3CD, PIK3R1, PLCG2, PLEKHM1, PLG, PMM2, PMS2, PNP, POLA1, POLD1, POLD2, POLE, POLE2, POLR3A, POLR3C, POLR3F, POMP, POT1, PPP1CB, PRF1, PRG4, PRKACG, PRKCD, PRKDC, PSEN1, PSENE1, PSMB8, PSMG2, PSTPIP1, PTEN, PTPN11, PTPRC, PTX3, PUS1, RAB27A, RAC2, RAD50, RAD51, RAD51C, RAF1, RAG1, RAG2, RANBP2, RAP1A, RAP1B, RASA2, RASGRP1, RBCK1, RBM8A, RECQL4, REL, RELB, RELB, RFX5, RFXANK, RFXAP, RHOH, RIPK1, RIT1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RNU4ATAC, RORC, RPGR, RPL10, RPL11, RPL15, RPL18, RPL19, RPL26, RPL27, RPL35A, RPL36, RPL5, RPL9, RPS10, RPS14, RPS15, RPS15A, RPS19, RPS24, RPS26, RPS27, RPS27A, RPS28, RPS29, RPS7, RPSA, RRAS, RSPH1, RSPH3, RSPH4A, RSPH9, RTE1, RUNX1, SAMD9, SAMD9L, SAMHD1, SAR1B, SBDS, SBF2, SEC23B, SEC61A1, SEMA3E, SERPING1, SH2D1A, SH3BP2, SH3KBP1, SHOC2, SI, SKIV2L, SLC10A2, SLC19A2, SLC25A38, SLC26A3, SLC29A3, SLC35A1, SLC35C1, SLC37A4, SLC39A4, SLC39A7, SLC46A1, SLC5A1, SLC7A7, SLC9A3, SLC11A, SLC11B, SLC11A3, SLC11A4, SLC11A5, SLC11A6, SLC11A7, SLC11A8, SLC11A9, SLC11A10, SLC11A11, SLC11A12, SLC11A13, SLC11A14, SLC11A15, SLC11A16, SLC11A17, SLC11A18, SLC11A19, SLC11A20, SLC11A21, SLC11A22, SLC11A23, SLC11A24, SLC11A25, SLC11A26, SLC11A27, SLC11A28, 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SMARCAL1, SMARCD2, SNX10, SOS1, SOS2, SP110, SPAG1, SPINK5, SPINT2, SPPL2A, SPRED1, SRC, SRP54, SRP72, STAT1, STAT2, STAT3, STAT5B, STIM1, STING1, STK36, STK4, STN1, STX11, STX3, STXBP2, STXBP3, TAFAZZIN, TAP1, TAP2, TAPBP, TASP1, TBK1, TBX1, TCF3, TCIIRG1, TCN2, TERC, TERF2, TERF2IP, TERT, TFRC, TGFBI, TGFBR1, TGFBR2, THBD, THPO, THRA, THRB, TICAM1, TINF2, TIRAP, TLR3, TMC6, TMC8, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TNFRSF9, TNFRSF11, TNFRSF12, TOP2B, TP53, TPP2, TRAC, TRADD, TRAF3, TRAF3IP2, TREX1, TRIM22, TRNT1, TSR2, TTC37, TTC7A, TUBB1, TYK2, UBE2T, UNC119, UNC13D, UNC93B1, UNG, USB1, USP18, VPS13B, VPS45, VSG4, VTN, WAS, WDR1, WIPF1, WRAP53, XIAP, XRCC2, ZAP70, ZBTB24, ZCCHC8, ZMYND10, ZNF341

### Epilepsy Panel (765 genes)

AARS1, ABAT, ABCA2, ABCD1, ACOX1, ACTL6B, ACY1, ADAM22, ADAR, ADARB1, ADAT3, ADGRG1, ADNP, ADPRS, ADSL, AFF3, AFG3L2, AGA, AIFM1, AIMP1, AIMP2, AKT3, ALDH3A2, ALDH5A1, ALDH7A1, ALG1, ALG11, ALG13, ALG14, ALG3, ALG6, ALG8, ALG9, ALKBH8, ALPL, AMACR, AMPD2, AMT, ANKRD11, AP2M1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APC2, ARFGF2, ARG1, ARHGFE9, ARID1B, ARSA, ARV1, ARX, ASAH1, ASNS, ASPA, ASXL3, ATAD1, ATN1, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP5F1A, ATP6AP2, ATP6V0A2, ATP6V1A, ATP7A, ATRX, BCKDHA, BCKDHB, BCKDK, BCS1L, BOLA3, BRAF, BRAT1, BSCL2, BTBD, C12ORF57, CACNA1A, CACNA1B, CACNA1D, CACNA1E, CACNA1G, CACNA1H, CACNA2D2, CACNB4, CAD, CAMK2B, CARS2, CASK, CASR, CC2D1A, CC2D2A, CCDC88A, CCDC88C, CDC42BPB, CDK19, CDK9, CDKL5, CEP85L, CERS1, CERT1, CHD1, CHD5, CHRNA2, CHRNA4, CHRNB2, CIC, CILK1, CLCN2, CLCN4, CLN3, CLN5, CLN6, CLN8, CLPB, CLTC, CNKSR2, CNNM2, CNPY3, CNTNAP2, COA7, COA8, COG4, COG6, COG7, COG8, COL18A1, COL4A1, COL4A2, COQ2, COQ4, COQ6, COQ9, COX10, COX15, COX6B1, CPA6, CPLX1, CP2, CREBBP, CRPPA, CSF1R, CSNK2B, CSTB, CTC1, CTNNA2, CTSB, CTSF, CTU2, CUL3, CUL4B, CUX2, CYFIP2, CYP27A1, D2HGDH, DARS1, DARS2, DBT, DCX, DDC, DDX3X, DEAF1, DEGS1, DENND5A, DEPDC5, DHCR24, DHCR7, DHDDS, DHFR, DHPS, DHX16, DHX30, DIAPH1, DLL1, DMXL2, DNAJC5, DNAJC6, DNM1, DNM1L, DOCK7, DOLK, DPAGT1, DPM1, DPM2, DPYD, DPYS, DYNCH1H1, DYRK1A, EARS2, ECHS1, ECM1, EEF1A2, EFHC1, EFTUD2, EHMT1, EIF2AK2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF2S3, EIF3F, EMC10, EML1, EMX2, EPG5, EPM2A, EPRS1, ETFA, ETFB, ETFDH, ETHE1, EXOC7, EXOSC3, EXT2, FA2H, FAM126A, FAM50A, FAR1, FARS2, FASTKD2, FBXL4, FBXO11, fcsk, FDF1, FDX2, FGF12, FGF13, FGF3R3, FH, FKBP, FKTN, FLNA, FOLR1, FOXG1, FOXRED1, FRRS1L, FUCA1, FUT8, GABBR2, GABRA1, GABRA2, GABRA5, GABRB1, GABRB2, GABRB3, GABRG2, GAD1, GALT, GALNT2, GAMT, GBA, GCDH, GCH1, GCSH, GFAP, GFM1, GFM2, GJC2, GLB1, GLDC, GLI3, GLRA1, GLRB, GLS, GLUD1, GLUL, GLYCTK, GM2A, GNAO1, GNAQ, GNB1, GNB5, GNE, GOLGA2, GOSR2, GOT2, GPPA1, GPHN, GRIA2, GRIA3, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRN, GSS, GTPBP2, GTPBP3, GUF1, H3-3A, H3-3B, HACE1, HAX1, HCCS, HCFC1, HCN1, HCN2, HEATR5B, HECW2, HEPACAM, HERC2, HEXA, HEXB, HIBCH, HLCS, HMGCL, HNRNP2, HNRNP3, HNRNP4, HOXA1, HPDL, HPRT1, HRAS, HSD17B10, HSD17B4, HSPD1, HTRA1, HTRA2, HTT, IBA57, IER3IP1, IFIH1, IKBKG, IQSEC2, IRF2BPL, ITPA, KARS1, KAT5, KAT8, KATNB1, KCNA1, KCNA2, KCNB1, KCNC1, KCNH1, KCNJ10, KCNJ11, KCNK4, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KCTD7, KDM5C, KIAA1109, KIF1A, KIF2A, KIF5A, KIF5C, KIF6B, KMT2E, KPTN, KRAS, LZHGHD, LARGE1, LARS1, LGI1, LIAS, LIPT1, LIPT2, LMAN2L, LMBRD2, LMNB1, LMNB2, LNPB, LRPPRC, LSS, LYRM7, LYST, MACF1, MADD, MAF, MAGI2, MANBA, MAP2K1, MAP2K2, MARS2, MAST1, MBD5, MBOAT7, MDH2, MECP2, MED12, MED17, MED27, MEF2C, MFF, MFSB8, MINPP1, MIPEP, MLC1, MMACHC, MMADHC, MOCS1, MOCS2, MOGS, MPDU1, MRPL44, MTHFR, MTHFS, MTOR, MTR, NACC1, NAGA, NARS1, NARS2, NBEA, NCDN, NDE1, NDP, NDST1, NDUFA1, NDUFA10, NDUFA2, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFS1, NDUFS2, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NECAP1, NEDD4L, NEU1, NEUROD2, NEXMIF, NFU1, NGLY1, NHLRC1, NKX6-2, NOTCH3, NPRL2, NPRL3, NR2F1, NR4A2, NRROS, NRXN1, NSD1, NSDHL, NSF, NT5C2, NTRK2, NUBPL, NUS1, OCLN, OFD1, OPHN1, OPUD6B, OTX2, OXR1, P4HTM, PACS1, PACS2, PAFAH1B1, PAH, PAK1, PARS2, PCCA, PCCB, PCDH12, PCDH19, PCDHGCA, PCLO, PCYT2, PDHA1, PDHX, PDSS2, PET100, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PGK1, PGM2L1, PHACTR1, PHF6, PHGDH, PIGA, PIGB, PIGC, PIGG, PIGH, PIGK, PIGN, PIGO, PIGP, PIGQ, PIGS, PIGT, PIGU, PIGV, PIGW, PIK3R2, PITRM1, PLA2G6, PLAA, PLCB1, PLP1, PLPBP, PMM2, PMPCB, PNKP, PNPO, PNPT1, POLG, POLG2, POLR3A, POLR3B, POMGNT1, POMT1, POMT2, PP1L1, PPP2CA, PPP3CA, PPT1, PRICKLE1, PRICKLE2, PRIMA1, PRMT7, PRODH, PRRT2, PRUNE1, PSAP, PSAT1, PSPH, PTF1A, PTPN23, PTS, PUM1, PURA, PYCR2, QARS1, QDPR, RAB11A, RAB11B, RAB18, RAB39B, RAB3GAP1, RAB3GAP2, RALA, RALGAP1, RANBP2, RARS1, RARS2, RELN, RFT1, RHOTB2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF13, RNF2, RNF216, RNU4ATAC, ROGDI, RORA, RORB, RPIA, RRM2B, RTN4IP1, RTTN, RUSC2, RYR2, SAMHD1, SATB1, SCAF4, SCAMP5, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SCO1, SCO2, SDHA, SDHAF1, SEMA6B, SEPSECS, SERAC1, SERPINI1, SETBP1, SETD1A, SETD1B, SETD5, SGGH, SHANK3, SIK1, SIX3, SLC12A5, SLC13A5, SLC16A2, SLC19A3, SLC1A2, SLC1A4, SLC25A1, SLC25A12, SLC25A15, SLC25A22, SLC25A42, SLC2A1, SLC35A1, SLC35A2, SLC35A3, SLC39A8, SLC45A1, SLC46A1, SLC5A6, SLC6A1, SLC6A5, SLC6A8, SLC9A6, SMARCA2, SMARCC2, SMC1A, SMS, SNAP25, SNIP1, SNORD118, SNX27, SOX10, SPATA5, SPR, SPTAN1, SPTBN1, SPTBN4, SSR4, ST3GAL3, ST3GAL5, STAG1, STAMPB, STARD7, STRADA, STX1B, STXBP1, SUCLA2, SUCLG1, SUMF1, SUOX, SURF1, SYN1, SYNGAP1, SYNJ1, SZT2, TAF1, TANC2, TANGO2, TBC1D20, TBC1D24, TBC1D2B, TBCE, TBCE, TBCK, TBL1XR1, TCF4, TDP2, TEO2, TET3, TFE3, TIMM50, TK2, TMEM106B, TMEM222, TMEM70, TMX2, TNK2, TPK1, TPP1, TRAF7, TRAK1, TRAPPC12, TRAPPC4, TRAPPC6B, TREX1, TRIM8, TRIP13, TRIT1, TRPM3, TRPM6, TRRAP, TSC1, TSC2, TSEN15, TSEN2, TSEN34, TSEN54, TSFM, TTC19, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, TUBGCP2, TXNRD1, UBA5, UBE2A, UBE3A, UBR7, UBTF, UFC1, UFM1, UFPS2, UGDH, UGP2, UNC80, USP18, USP7, VAMP2, VARS1, VLDLR, VPS11, VPS13A, WARS2, WASF1, WDR26, WDR37, WDR45, WDR45B, WDR62, WDR73, WWOX, YIF1B, YIPF5, YWHAG, YY1, ZBTB18, ZDHHC9, ZEB2, ZFYVE26, ZMIZ1, ZNF142, ZNF335, ZNHIT3, ZSWIM6

### Growth and Related Disorders Panel (520 genes)

ACAN, ACP5, ACTB, ACTG1, ACVR1, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTSL2, AFF4, AGA, AGPS, AIFM1, AKT1, ALPL, ALX1, ALX3, ALX4, AMER1, AMMECR1, ANKH, ANKRD11, ANO5, ANTXR2, ARCN1, ARHGAP31, ARID1A, ARID1B, ARSB, ARSL, ASCC1, ASXL1, ATP6V0A2, ATR, B3GALT6, B3GAT3, B4GALT7, BCOR, BCS1L, BGN, BHLHA9, BLM, BMP1, BMP2, BMPER, BMPR1B, BPNT2, BRAF, BRCA2, BRIP1, C2CD3, CA2, CANT1, CASR, CBL, CC2D2A, CCDC47, CCDC8, CCN6, CCNQ, CDC42, CDC45, CDC6, CDH11, CDH3, CDK13, CDKN1C, CDT1, CENPE, CENPJ, CEP120, CEP152, CEP290, CEP63, CFAP410, CHD7, CHST14, CHST3, CHSY1, CILK1, CKAP2L, CLCN5, CLCN7, COG4, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL27A1, COL2A1, COL3A1, COL5A1, COL5A2, COL9A1, COL9A2, COL9A3, COMP, CREB3L1, CREBBP, CRIPT, CRLF1, CRTAP, CSF1R, CSPP1, CTSK, CUL7, CWC27, CYP27B1, CYP2R1, DDR2, DDRGK1, DDX58, DHCR24, DHCR7, DHODH, DLL3, DLL4, DLX3, DLX5, DMP1, DNAJC21, DNMT3A, DOCK6, DONSON, DSE, DVL1, DVL3, DYM, DYNCH2H1, DYNCH2I1, DYNCH2I2, DYNCH2LI1, DYNL2B, EBP, EDN1, EDNRA, EFL1, EFN1, EFTUD2, EIF2AK3, EIF4A3, ENAM, ENPP1, EOGT, ERCC4, ERF, ESCO2, EVC, EVC2, EXOSC2, EXT1, EXT2, EXTL3, EYA1, EZH2, FAM111A, FAM20A, FAM20C, FAM83H, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FBN1, FBN2, FERMT3, FGD1, FGF10, FGF23, FGF9, FGFR1, FGFR2, FGFR3, FIG4, FKBP10, FKBP14, FLNA, FLNB, FN1, FTO, FUCA1, FZD2, GALNS, GALNT3, GCM2, GDF3, GDF5, GDF6, GH1, GHR, GHRHR, GHSR, GJA1, GLB1, GLI1, GLI2, GLI3, GMNN, GNAI3, GNAS, GNPAT, GNPTAB, GNPTG, GNS, GORAB, GPC6, GSC, GUSB, GZF1, HAAO, HDAC4, HDAC8, HES7, HESX1, HNRNP4, HOXA11, HOXA13, HOXD13, HPGD, HRAS, HSPA9, HSPG2, IARS2, IDH2, IDS, IDUA, IFIH1, IFITM5, IFT122, IFT140, IFT172, IFT43, IFT52, IFT80, IFT81, IGF1, IGF1R, IGF2, IGFALS, IHH, IL1RN, INPPL1, INSR, INTU, IRF6, IRS1, KAT6B, KCNJ2, KDM1A, KDM6A, KIAA0586, KIAA0753, KIF22, KIF7, KL, KMT2A, KMT2D, KRAS, KYNU, LARP7, LBR, LEMD3, LFNG, LHX3, LHX4, LIFR, LMNA, LMX1B, LONP1, LPIN2, LRP4, LRP5, LTBP2, LTBP3, LZTR1, MAFB, MAN2B1, MANBA, MAP2K1, MAP2K2, MAP3K7, MATN3, MBTPS2, MECOM, MEGF8, MEOX1, MESP2, MET, MGP, MKS1, MMP13, MMP2, MMP9, MNX1, MSX2, MYCN, MYH3, MYO18B, NAGLU, NANS, NBAS, NEK1, NEU1, NF1, NFIX, NIPBL, NKX3-2, NOG, NOTCH1, NOTCH2, NPR2, NRAS, NSD1, NSDHL, OBSL1, OCLN, OFD1, ORC1, ORC4, ORC6, OSGEP, OSTM1, OTX2, P3H1, P4HB, PALB2, PAM16, PAPSS2, PAX3, PCGF2, PCNT, PCYT1A, PDE3A, PDE4D, PEX14, PEX19, PEX5, PEX7, PGM3, PHEX, PIGV, PIK3CA, PISD, PITX1, PITX2, PLCB4, PLEKHM1, PLK4, PLOD1, PLOD2, PLS3, POC1A, POLA1, POLR1A, POLR1C, POLR1D, POLR3A, POLR3B, POP1, POR, POU1F1, PPIB, PPP3CA, PRKAR1A, PRMT7, PROP1, PTDSS1, PTH1R, PTHLH, PTPN11, PUF60, PYCR1, RAB23,

RAB33B, RAD21, RAD51C, RAF1, RALA, RASA2, RBBP8, RBM8A, RBPJ, RECQL4, RIPPLY2, RIT1, RMRP, RNU4ATAC, ROR2, RPGRIP1L, RRAS, RSPRY1, RTTN, RUNX2, SALL1, SALL4, SBDS, SC5D, SEC24D, SERPINF1, SERPINH1, SETBP1, SETD2, SF3B4, SFRP4, SGMS2, SGSH, SH3BP2, SH3PXD2B, SHH, SHOC2, SHOX, SIX5, SKI, SLC10A7, SLC17A5, SLC26A2, SLC29A3, SLC34A3, SLC35D1, SLC39A13, SLC02A1, SLX4, SMAD2, SMAD3, SMAD4, SMARCA2, SMARCA4, SMARCAL1, SMARCB1, SMARCE1, SMC1A, SMC3, SNRPC, SNX10, SOS1, SOST, SOX11, SOX2, SOX3, SOX9, SP7, SPARC, SQSTM1, SRCAP, SRP54, STAG2, STAMBIP, STAT5B, SUMF1, TAB2, TALDO1, TAPT1, TBCE, TBX15, TBX19, TBX2, TBX3, TBX4, TBX5, TBX6, TBXAS1, TCF12, TCIRG1, TCOF1, TCTN3, TENT5A, TGDS, TGFB1, TGFB2, TGFB3, TGFB4, TGFB5, THPO, THRA, TMEM165, TMEM216, TMEM38B, TMEM67, TNFRSF11A, TNFRSF11B, TNFSF11, TONSL, TOP3A, TP63, TRAF3IP1, TRAPPC2, TREM2, TRIM37, TRIP11, TRMT10A, TRPS1, TRPV4, TRPV6, TTC21B, TUBGCP6, TWIST1, TYROBP, UFSP2, VDR, VIPAS39, WDR19, WDR35, WNT1, WNT10B, WNT5A, WNT7A, XRCC2, XRCC4, XYLT1, XYLT2, ZMPSTE24, ZSWIM6

### Retinal Dystrophy Panel (314 genes)

ABCA4, ABCG6, ABCD1, ABHD12, ACO2, ADAM9, ADAMTS18, ADGRV1, ADIPOR1, AGBL5, AHI1, AIPL1, ALMS1, AMACR, ARHGEF18, ARL13B, ARL2BP, ARL3, ARL6, ARMC9, ARR3, ARSG, ATF6, ATOH7, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEST1, C1QTNF5, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDH23, CDH3, CDHR1, CEP104, CEP120, CEP164, CEP19, CEP250, CEP290, CEP41, CEP78, CEP83, CERKL, CFAP410, CFAP418, CHM, CIB2, CISD2, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, COQ2, CPE, CPLANE1, CRB1, CRPPA, CRX, CSPP1, CTC1, CTNNA1, CTNNA1, CTSD, CWC27, CYP4V2, DHDDS, DHX38, DNAJC5, DRAM2, DTHD1, DYNC2H1, EFEMP1, ELOVL4, EMC1, ESPN, EXOSC2, EYS, FAM161A, FXR, FLVCR1, FRMD7, FZD4, GNAT1, GNAT2, GNB3, GNPTG, GPR143, GPR179, GRK1, GRM6, GUCA1A, GUCY2D, HARS1, HGSNAT, HK1, HMX1, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT81, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, JAG1, KATNIP, KCNJ13, KCNV2, KIAA0586, KIAA0753, KIAA1549, KIF11, KIF7, KIZ, KLHL7, LAMA1, LCA5, LRAT, LRIT3, LRP2, LRP5, LZTFL1, MAK, MERTK, MFN2, MFRP, MFSDB, MKKS, MKS1, MMACHC, MTPP, MVK, MYO7A, NAGLU, NDP, NEK2, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NYX, OAT, OCA2, OFD1, OPA1, OPA3, OPN1SW, OTX2, P3H2, PANK2, PAX2, PCARE, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDS1, PDS2, PDZD7, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, PISD, PITPNM3, PLA2G5, PLK4, PNPLA6, POC1B, POMGNT1, PPT1, PRCD, PRDM13, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, PRPS1, RAB28, RAX2, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, RTN4IP1, SAG, SAMD11, SCAPER, SCLT1, SDCCAG8, SEMA4A, SGSH, SLC24A1, SLC25A46, SLC45A2, SLC7A14, SNRNP200, SPATA7, SPP2, SRD5A3, TCTN1, TCTN2, TCTN3, TEAD1, TIMM8A, TIMP3, TMEM107, TMEM126A, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TOPORS, TPP1, TRAF3IP1, TREX1, TRIM32, TRPM1, TSPAN12, TTC21B, TTC8, TTL5, TTPA, TUB, TUBB4B, TUBGCP4, TUBGCP6, TULP1, TYR, TYRP1, USH1C, USH1G, USH2A, VCAN, VPS13B, WDPCP, WDR19, WFS1, WHRN, YME1L1, ZNF408, ZNF423, ZNF513

### Movement Disorders Panel (609 genes)

AARS2, AASS, ABCA2, ABCB7, ABCD1, ABHD12, ACO2, ACOX1, ACP5, ACTB, ADAR, ADCY5, ADPRS, ADSL, AFG3L2, AGTPBP1, AHI1, AIMP1, ALDH18A1, ALDH3A2, ALDH5A1, ALG1, ALG13, ALG6, ALG8, ALS2, AMACR, AMPD2, AMT, ANKLE2, ANO10, ANO3, AP1S2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, APTX, ARG1, ARL13B, ARL6, ARL6IP1, ARNT2, ARSA, ARX, ASNS, ASPA, ASXL1, ATAD3A, ATCA, ATLL1, ATM, ATP13A2, ATP1A3, ATP2B3, ATP6AP2, ATP7A, ATP7B, ATP8A2, ATRX, AUH, B3GALT6, B4GALNT1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCAP31, BCKDHA, BCKDHB, BCOR, BCS1L, BEAN1, BOLA3, BSCL2, BTBD, BUB1B, C12ORF4, C19ORF12, CA8, CACNA1A, CACNA1B, CACNA1D, CACNA1G, CACNA2D2, CACNB4, CAMTA1, CAPN1, CASK, CAV1, CC2D2A, CCDC88C, CCT5, CDS9, CEP120, CEP290, CEP41, CFH, CFHR1, CFHR3, CHCHD10, CHCHD2, CHMP1A, CIT, CLCN2, CLN5, CLN6, CLN8, CLP1, CLPB, CLPP, COA7, COASY, COG2, COG5, COG8, COL4A2, COQ2, COQ4, COQ5, COQ6, COQ8A, COX10, COX15, COX20, CP, CPLANE1, CPT1C, CSF1R, CSTB, CTBP1, CTC1, CTDP1, CTNNA1, CTSA, CTSD, CWF19L1, CYB5R3, CYP27A1, CYP2U1, CYP7B1, DARS1, DARS2, DBT, DCAF17, DCTN1, DDHD1, DDHD2, DDX3X, DHCR24, DHPS, DLAT, DLD, DNAJC12, DNAJC19, DNAJC5, DNAJC6, DNMT1, DOCK3, DPAGT1, DPH1, DPM1, DYNC1H1, EARS2, EBF3, EEF2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, ELOVL4, ELOVL5, ELP2, EML1, ENTPD1, ERLIN1, ERLIN2, ESCO2, EVC, EVC2, EXOSC3, EXOSC8, FA2H, FAR1, FARS2, FBXL4, FBXO7, FXR, FGF14, FGFR1, FITM2, FLNA, FLVCR1, FMR1, FOXG1, FOXRED1, FTL, FTO, FUCA1, FXN, GAA, GAD1, GALT, GAMT, GAN, GBA, GBA2, GBE1, GCDH, GCH1, GFAP, GFM1, GJA1, GJC2, GLA, GLB1, GLDC, GLRX5, GLS, GLYCTK, GM2A, GNAL, GNAO1, GNE, GOSR2, GPT2, GPX4, GRID2, GRM1, GRN, GSS, GTF2E2, GUCY1A1, H19, HACE1, HARS2, HEXA, HEXB, HIBCH, HIKESHI, HLCS, HPR1, HSD17B4, HSPB3, HSPD1, HTRA1, HTT, HYL1, IBA57, IDUA, IFIH1, IKBKG, INPP5E, IRF2BPL, ISCA2, ITM2B, ITPR1, JAM3, KATNB1, KCNA1, KCNA2, KCNC3, KCND3, KCNJ10, KCNJ6, KCNMA1, KCNQ2, KCNT1, KDM5C, KIDINS220, KIF1A, KIF1C, KIF2A, KIF5A, KIF5C, KIF7, KLC2, KMT2B, L1CAM, L2HGDH, LAGE3, LAMA1, LAMB1, LARS2, LIAS, LIPT1, LIPT2, LMNB1, LRP4, LRPPRC, LRRK2, LRSAM1, LYRM7, MAG, MAN2B1, MAPT, MARS2, MCEE, MCOLN1, MECP2, MECP3, MED17, MFF, MFSDB, MFSDB, MGME1, MIPEP, MKKS, MKS1, MLC1, MMACHC, MMADHC, MME, MOCS1, MOCS2, MPDU1, MRE11, MRPS22, MRPS34, MSTO1, MTFMT, MTHFR, MTO1, MTPAP, MTRFR, MTPP, MVK, NACC1, NADK2, NALCN, NARS2, NAXE, NDE1, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFB11, NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NEU1, NEXMIF, NIPA1, NKX2-1, NKX6-2, NOL3, NPC1, NPC2, NPHP1, NSUN2, NT5C2, NUBPL, NUP62, NUS1, OFD1, OPA1, OPA3, OPHN1, OSGEP, PAFAH1B1, PAH, PANK2, PARK7, PAX6, PC, PCLO, PDCD1, PDE10A, PDE8B, PDGFB, PDGFRA, PDGFRB, PDHA1, PDHB, PDHX, PDP1, PDS2, PDYN, PEX10, PEX12, PEX16, PEX2, PEX3, PEX6, PEX7, PGAP1, PGAP3, PHGDH, PHYH, PIEZO2, PIGA, PIGG, PIGN, PINK1, PLA2G6, PLCB1, PLG, PLP1, PMM2, PMPCA, PNKD, PNKP, PNP, PNPLA6, PNPLA8, POLG, POLG2, POLR1C, POLR3A, POLR3B, PPP3CA, PPT1, PRDM8, PRF1, PRKCG, PRKN, PRKRA, PRNP, PROC, PRPS1, PRRT2, PSAP, PSEN1, PUM1, PYCR2, RAB18, RAB27A, RAB39B, RAB3GAP1, RAB3GAP2, RARS1, RARS2, REEP1, REEP2, RFT1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF113A, RNF216, RORA, RPGRIP1L, RPIA, RTN2, RTN4IP1, RUBCN, SACS, SAMD9, SAMD9L, SAMHD1, SCN2A, SCN8A, SCP2, SCYL1, SDHA, SDHAF1, SDHD, SEPSECS, SERAC1, SETX, SGCE, SH3TC2, SIL1, SLC16A2, SLC17A5, SLC19A2, SLC19A3, SLC1A2, SLC1A3, SLC1A4, SLC20A2, SLC25A12, SLC25A15, SLC25A22, SLC25A46, SLC2A1, SLC30A10, SLC33A1, SLC39A14, SLC4A1, SLC52A2, SLC52A3, SLC6A19, SLC6A3, SLC6A8, SLC9A1, SLC9A6, SMG9, SMPD1, SNCA, SNX14, SOD1, SOX10, SOX2, SPART, SPAST, SPATA5, SPG11, SPG21, SPG7, SPR, SPTAN1, SPTBN2, SQSTM1, ST3GAL5, STAMBIP, STN1, STUB1, STXB1, SUCLA2, SUMF1, SUOX, SURF1, SYNE1, SYNGAP1, SYNJ1, SYT14, TAF1, TAF2, TANGO2, TARDBP, TBC1D20, TBCD, TBCE, TCIRG1, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TECPR2, TELO2, TFG, TGM6, TH, THAP1, TIMM8A, TIMMDC1, TMEM126B, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TNFSF11, TOR1A, TP53RK, TPI1, TPP1, TRAPPC11, TREM2, TREX1, TRIM32, TRMT5, TTBK2, TTC19, TTC8, TTPA, TUBB4A, TWNK, TYMP, UBA5, UBAP1, UBTF, UCHL1, VAC14, VAMP1, VLDLR, VPS13A, VPS13C, VPS13D, VPS35, VWA3B, WASHC4, WASHC5, WDPCP, WDR81, WFS1, WWOX, XPR1, XRCC1, ZFYVE26, ZNF423

### Spastic Paraplegia Panel (75 genes)

ABCD1, AFG3L2, ALDH18A1, ALS2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARG1, ARL6IP1, ATAD3A, ATLL1, B4GALNT1, BSCL2, BTBD, C19ORF12, CACNA1G, CAPN1, COASY, CTNNA1, CYP27A1, CYP2U1, CYP7B1, DARS1, DDHD1, DDHD2, ERLIN2, FA2H, FARS2, FXN, GALT, GBA2, GBE1, GCH1, GJC2, GPT2, HACE1, HSPD1, IBA57, IRF2BPL, KDM5C, KIDINS220, KIF1A, KIF5A, L1CAM, L2HGDH, MARS2, MTRFR, NIPA1, NKX6-2, NT5C2, PAH, PLP1, PNPLA6, RARS1, REEP1, RTN2, SACS, SETX, SLC16A2, SLC1A4, SLC25A15, SLC33A1, SPART, SPAST, SPG11, SPG7, SPR, TECPR2, TFG, TH, UBAP1, WASHC5, ZFYVE26