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TEST CODE	TEST NAME	NO. OF GENES	GENES INVOLVED	CATEGORY	DESCRIPTION
D4001	Childhood-Onset Epilepsy Panel	58	ADSL, CACNA1A, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLN3, CLN5, CLN6, CLN8, CNTNAP2, CSTB, CTSD, DYRK1A, EEF1A2, EPM2A, FOLR1, FOXG1, GABRA1, GABRB2, GABRB3, GABRG2, GAMT, GATM, GOSR2, GRIN1, GRIN2A, IQSEC2, KANSL1, KCNT1, KCTD7, LGI1, MAGI2, MBD5, MECP2, MEF2C, MFSD8, NHLRC1, NRXN1, PCDH19, PNKP, POLG, PPT1, PRICKLE1, SCN1A, SCN1B, SCN2A, SLC2A1, SLC6A8, SLC9A6, TBC1D24, TCF4, TPP1 (CLN2), UBE3A, WDR45, ZEB2	Neurology	Epilepsy
D4002	Comprehensive Epilepsy Panel	98	ADSL, ALDH7A1, ALG13, ARID1B, ARHGEF9, ARX, ATP1A2, ATP6AP2, ATRX, CACNA1A, CASK, CDKL5, CHD2, CHRNA2, CHRNA4, CHRNA7, CHRNB2, CLN3, CLN5, CLN6, CLN8, CNTNAP2, CREBBP, CSTB, CTSD, DNAJC5, DNM1, DYRK1A, EEF1A2, EHMT1, EPM2A, FOLR1, FOXG1, GABRA1, GABRB2, GABRB3, GABRG2, GAMT, GATM, GOSR2, GRIN1, GRIN2A, GRIN2B, HNRNPU, IQSEC2, KANSL1, KCNB1, KCNJ10, KCNQ2, KCNQ3, KCNT1, KCTD7, LGI1, MAGI2, MBD5, MECP2, MEF2C, MFSD8, NHLRC1, NR2F1, NRXN1, OPHN1, PCDH19, PHF6, PIGA, PIGO, PIGV, PLCB1, PNKP, PNPO, POLG, PPT1, PRICKLE1, PRRT2, QARS, SCARB2, SCN1A, SCN1B, SCN2A, SCN8A, SLC13A5, SLC25A22, SLC2A1, SLC6A8, SLC9A6, SMC1A, SPTAN1, STXBP1, SYNGAP1, TBC1D24, TCF4, TPP1 (CLN2), TSC1, TSC2, UBE3A, WDR45, WWOX, ZEB2	Neurology	Epilepsy
D4003	Infantile Epilepsy Panel	75	ADSL, ALDH7A1, ALG13, ARHGEF9, ARX, ATP6AP2, CACNA1A, CDKL5, CHD2, CHRNA7, CLN3, CLN5, CLN6, CLN8, CNTNAP2, CTSD, DNM1, DYRK1A, EEF1A2, FOLR1, FOXG1, GABRA1, GABRB2, GABRB3, GABRG2, GAMT, GATM, GRIN1, GRIN2A, GRIN2B, IQSEC2, KANSL1, KCNB1, KCNJ10, KCNQ2, KCNQ3, KCNT1, KCTD7, MAGI2, MBD5, MECP2, MEF2C, MFSD8, NR2F1, NRXN1, PCDH19, PIGA, PIGO, PIGV, PNKP, PNPO, POLG, PPT1, PRRT2, QARS, SCN1A, SCN1B, SCN2A, SCN8A, SLC13A5, SLC25A22, SLC2A1, SLC6A8, SLC9A6, SPTAN1, STXBP1, TBC1D24, TCF4, TPP1 (CLN2), TSC1, TSC2, UBE3A, WDR45, WWOX, ZEB2	Neurology	Epilepsy
D4004	Progressive Myoclonic Epilepsy Panel	17	CLN3, CLN5, CLN6, CLN8, CSTB, CTSD, DNAJC5, EPM2A, FOLR1, GOSR2, KCTD7, MFSD8, NHLRC1, PPT1, PRICKLE1, SCARB2, TPP1 (CLN2)	Neurology	Epilepsy
D4006	Tuberous Sclerosis Panel	2	TSC1, TSC2	Neurology	Epilepsy
D4007	Prenatal Tuberous Sclerosis Panel	2	TSC1, TSC2	Neurology	Epilepsy
D4008	Cerebellar Ataxia Panel	14	APTX, COQ8A, COQ2, COQ9, DNMT1, FXN, PDSS1, PDSS2, POLG, SACS, SETX, SYNE1, TTPA, VLDLR	Neurology	Movement Disorders
D4009	Episodic Ataxia Panel	4	CACNA1A, CACNB4, KCNA1, SLC1A3	Neurology	Movement Disorders
D4010	Myoclonic Dystonia Panel	3	DRD2, SGCE, TOR1A	Neurology	Movement Disorders
D4011	Spastic Paraplegia Panel Complete	58	ALDH18A1, ALS2, AMPD2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARL6IP1, ARSI, ATL1, ATP2B4, B4GALNT1, BICD2, BSCL2, C12ORF65, C19orf12, CCT5, CYP2U1, CYP7B1, DDHD1, DDHD2, ENTPD1, ERLIN1, ERLIN2, EXOSC3, FA2H, FLRT1, GBA2, GJC2, HSPD1, KIAA0196, KIF1A, KIF1C, KIF5A, L1CAM, MAG, MARS, NIPA1, NT5C2, PGAP1, PLP1, PNPLA6, RAB3GAP2, REEP1, REEP2, RTN2, SACS, SLC16A2, SLC33A1, SPAST, SPG11, SPG20, SPG21, SPG7, TECPR2, TFG, TTR, USP8, VAMP1, VPS37A, WDR48, ZFR, ZFYVE26, ZFYVE27	Neurology	Movement Disorders
D4012	Dystonia Panel	21	ANO3, ATP1A3, GCH1, GNAL, PARK2, PNKD, PRKRA, PRRT2, SGCE, SLC2A1, SLC6A3, SPR, TH, THAP1, TOR1A, TUBB4A, CIZ1, DRD2, HPCA, KCTD17, TOR1AIP1	Neurology	Movement Disorders

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D4013	Joubert and Meckel-Gruber Syndromes Panel	30	AHI1, ARL13B, B9D1, B9D2, C5orf42, CC2D2A, CEP104, CEP290, CEP41, CSPP1, INPP5E, KIAA0586, KIF7, MKS1, MRE11A, NPHP1, NPHP3, OFD1, PDE6D, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF4	Neurology	Neurological Disorders
D4014	Aicardi-Goutieres Syndrome Panel	7	ADAR, IFIH1, TREX1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1	Neurology	Neurological Disorders
D4015	Alzheimer Dementia and Dementia Panel	7	APOE, APP, PRNP, PSEN1, PSEN2, SORL1, TREM2	Neurology	Neurological Disorders
D4016	Brain iron Accumulation Syndromes Panel	11	ATP13A2, C19orf12, COASY, CP, DCAF17, FA2H, FTL, PANK2, PLA2G6, SCP2, WDR45	Neurology	Neurological Disorders
D4017	Pontocerebellar Hypoplasia Panel	9	CASK, TSEN2, TSEN34, TSEN54, OPHN1, RARS2, VRK1, EXOSC3, CHMP1A	Neurology	Neurological Disorders
D4018	Hereditary Parkinson's Disease & Parkinsonism Panel	17	ATP13A2, DCTN1, DNAJC6, FBXO7, GCH1, LRRK2, PARK2, PARK7, PINK1, PRKRA, SLC6A3, SNCA, SPR, TH, VPS35, CHCHD2, MAPT	Neurology	Neurological Disorders
D4019	Hereditary Dementia and Amyotrophic Lateral Sclerosis Panel	33	ALS2, ANG, APP, CHCHD10, DCTN1, ERBB4, FIG4, FUS, GRN, MAPT, NEFH, OPTN, PFN1, PRNP, PRPH, PSEN1, PSEN2, SNCA, SOD1, SPG11, TARDBP, TBK1, TFG, TUBA4A, UBQLN2, VAPB, VCP, CHMP2B, HNRNPA2B1, MATR3, SETX, SIGMAR1, SQSTM1	Neurology	Neurological Disorders
D4020	Comprehensive Neuropathies Panel	81	AARS, AIFM1, ATL1, ATL3, ATP7A, BICD2, BSCL2, CHCHD10, DCTN1, DNAJB2, DNM2, DNMT1, DST, DYNC1H1, EGR2, FAM134B, FBXO38, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GNB4, HARS, HINT1, HSPB1, HSPB8, IGHMBP2, IKBKAP, INF2, KIF1A, LITAF, LMNA, LRSAM1, MED25, MFN2, MORC2, MPZ, MTMR2, NDRG1, NEFL, NGF, NTRK1, PDK3, PLEKHG5, PMP22, PRPS1, PRX, RAB7A, REEP1, SBF2, SCN11A, SCN9A, SH3TC2, SIGMAR1, SLC52A2, SLC52A3, SLC5A7, SPG11, SPTLC1, SPTLC2, TFG, TRIM2, TRPV4, TTR, UBA1, VAPB, WNK1, YARS, CCT5, FLRT1, HSPB3, LAS1L, MARS, PRDM12, SCN10A, SETX, SLC25A46, SURF1, VRK1	Neurology	Neurological Disorders
D4021	Neuro-degeneration Panel	37	ABAT, ALDH5A1, ALDH7A1, AMT, ARHGEF9, DBH, DDC, GAD1, GCH1, GCSH, GLDC, GLRA1, GLRB, GPHN, MAOA, PCBD1, PHGDH, PNPO, PSAT1, PSPH, PTS, QDPR, SLC25A22, SLC6A3, SLC6A5, SPR, TH, ATP13A2, C19orf12, COASY, CP, DCAF17, FA2H, FTL, PANK2, PLA2G6, WDR45	Neurology	Neurological Disorders

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D4022	Hereditary Neuropathy Sequencing Panel	122	AARS, AIFM1, ALDH3A2, APTX, ATL1, ATL3, ATM, ATP7A, BICD2, BSCL2, CACNB4, CCT5, CHCHD10, COQ8A, CTDP1, DCTN1, DNAJB2, DNM2, DNMT1, DST, DYNC1H1, EGR2, FAM134B, FBXO38, FGD4, FGF14, FIG4, FLRT1, FXN, GAN, GARS, GDAP1, GJB1, GLA, GNB4, HARS, HINT1, HOXD10, HSPB1, HSPB3, HSPB8, IGHMBP2, IKBKAP, INF2, ITPR1, KCNA1, KCNC3, KIF1A, KIF1B, KIF5A, L1CAM, LAS1L, LITAF, LMNA, LRSAM1, MARS, MED25, MFN2, MORC2, MPZ, MRE11, MTMR2, MTTP, NDRG1, NEFL, NGF, NIPA1, NTRK1, PDK3, PEX7, PHYH, PLEKHG5, PLP1, PMP22, PNPLA6, POLG, PRDM12, PRKCG, PRPS1, PRX, RAB7A, REEP1, SACS, SBF2, SCN10A, SCN11A, SCN9A, SETX, SH3TC2, SIGMAR1, SIL1, SLC12A6, SLC1A3, SLC25A46, SLC52A2, SLC52A3, SLC5A7, SPAST, SPG11, SPG20, SPG21, SPG7, SPTBN2, SPTLC1, SPTLC2, SURF1, TDP1, TFG, TRIM2, TRPV4, TTBK2, TTPA, TTR, TWNK, UBA1, VAPB, VRK1, WASHC5, WNK1, YARS, ZFYVE26, ZFYVE27	Neurology	Neurological Disorders
D4023	Comprehensive Brain Malformations Panel	93	ACTB, ACTG1, ADGRG1, AHI1, AKT3, ARFGEF2, ARL13B, ARX, ASPM, ATP6V0A2, B3GALNT2, B3GNT1, B9D1, C5orf42, CASK, CC2D2A, CCND2, CEP290, CEP41, CHMP1A, CLP1, CSPP1, CUL4B, DCX, DYNC1H1, ERMARD, EXOSC3, FAT4, FKRP, FKTN, FLNA, GMPPB, GPSM2, IFT172, INPP5E, ISPD, KIAA1279, KIF2A, KIF5C, KIF7, LAMB1, LAMC3, LARGE, MKS1, NDE1, NPHP1, NPHP3, OCLN, OFD1 (CXORF5), OPHN1, PAFAH1B1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PQBP1, RAB18, RAB3GAP1, RAB3GAP2, RARS2, RELN, RPGRIP1L, RTTN, SEPSECS, SRD5A3, SRPX2, TBC1D20, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM5, TMEM67, TSEN2, TSEN34, TSEN54, TTC21B, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, VLDLR, VPS53, VRK1, WDR62	Neurology	Neurological Disorders
D4024	Cortical Brain Malformations Panel	56	ACTB, ACTG1, ADGRG1, AKT3, ARFGEF2, ARX, ASPM, ATP6V0A2, B3GALNT2, B3GNT1, CCND2, CUL4B, DCX, DYNC1H1, ERMARD, FAT4, FKRP, FKTN, FLNA, GMPPB, GPSM2, ISPD, KIAA1279, KIF2A, KIF5C, LAMB1, LAMC3, LARGE, NDE1, OCLN, PAFAH1B1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PQBP1, RAB18, RAB3GAP1, RAB3GAP2, RELN, RTTN, SRD5A3, SRPX2, TBC1D20, TMEM5, TUBA1A, TUBA8, TUBB, TUBB2A, TUBB2B, TUBB3, TUBB4A, TUBG1, VLDLR, WDR62	Neurology	Neurological Disorders
D4025	Lissencephaly Panel	24	ACTB, ACTG1, ARX, ATP6V0A2, B3GALNT2, B3GNT1, DCX, FKRP, FKTN, GMPPB, ISPD, LAMB1, LARGE, NDE1, PAFAH1B1, POMGNT1, POMGNT2, POMT1, POMT2, RELN, TMEM5, TUBA1A, VLDLR, WDR62	Neurology	Neurological Disorders
D4026	Ceroid Lipofuscinosis Panel	13	ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, PPT1, TPP1	Neurology	Neurological Disorders

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D4046	Focused Autism and Intellectual Disability Panel	264	ADNP, ADSL, AHDC1, AKAP9, ALDH5A1, ALDH7A1, ALG6, AMT, ANK2, ANKRD11, ANXA1, AP3B2, AP4B1, ARFGEF2, ARID1B, ARID2, ARX, ASAH1, ASH1L, ASPM, ASXL1, ASXL3, ATP10A, ATP1A3, AUTS2, BCKDK, BCL11A, BRAF, BRAT1, C5orf42, CACNA1C, CACNA1D, CACNA1E, CACNA1H, CASK, CBL, CC2D1A, CDC42BPB, CDKL5, CEP135, CHAMP1, CHD2, CHD7, CHD8, CHKB, CHMP1A, CIB2, CIC, CREBBP, CSMD1, CTCF, CTNNA3, CTNNB1, CUL3, DDX3X, DEAF1, DHCR7, DLG2, DMD, DNMT3A, DPP6, DPYD, DYNC1H1, DYRK1A, EBF3, EFR3A, EFTUD2, EHMT1, ELOVL4, ELP4, EP300, ERCC6, FBXL4, FOXG1, FOXP1, FOXP2, GATAD2B, GATM, GPC4, GPHN, GRIA1, GRIK2, GRIN1, GRIN2B, HCN1, HDAC8, HECW2, HEPACAM, HERC2, HIVEP2, HNRNPU, IL1RAPL1, IQSEC2, ITPR1, ITSN1, KANSL1, KAT6A, KAT6B, KATNAL2, KCNB1, KCNJ10, KCNQ2, KCNQ3, KDM5B, KDM5C, KDM6A, KDM6B, KIAA2022, KIF11, KMT2A, KMT2C, KMT2D, LAMA2, LAMB1, LARP7, LRP2, MAGEL2, MBD5, MECP2, MED13, MED13L, MEF2C, MET, MTOR, MYH10, MYO5A, MYT1L, NALCN, NAV2, NBEA, NCKAP1, NEDD9, NFIA, NFIX, NGLY1, NIPBL, NLGN1, NRXN1, NSD1, NSUN2, OCRL, OXTR, PACS1, PAH, PARK2, PAX5, PAX6, PCCA, PCCB, PCDH19, PDHA1, PER2, PGAP1, PHF2, PHF21A, PHIP, PIK3R2, PLA2G6, POGZ, PON1, PRICKLE2, PRKD1, PRODH, PTCHD1, PTEN, PURA, RAI1, RIMS1, ROBO2, SATB2, SBF1, SCN1A, SCN2A, SCN9A, SEMA5A, SETBP1, SETD1A, SETD2, SETD5, SGSH, SHANK1, SHANK2, SHANK3, SLC12A5, SLC2A1, SLC35A2, SLC6A1, SLC6A3, SLC6A8, SLC7A3, SLC9A6, SMAD4, SMARCA2, SMARCA4, SMC1A, SON, SOX11, SOX5, SPAST, SPEN, SPP2, SRCAP, STAG1, STXBP1, SURF1, SYNGAP1, TAF1, TAF6, TBL1XR1, TBR1, TCF20, TCF4, TCF7L2, TCOF1, TMLHE, TPP1, TRAPPC9, TRIO, TRIP12, TRPC6, TRRAP, TSC1, TSC2, UBE2A, UBE3A, UPF3B, USP9X, WAC, WDFY3, WDR45, WWOX, YTHDC1, YY1, ZBTB18, ZBTB20, ZEB2, ZMYND11, ZWILCH, CYP27A1, FOLR1, HPRT1, MID1, NHS, NR1I3, PHF6, PNKP, SMARCB1, AFF2, AP1S2, ATRX, CNTNAP2, FGD1, L1CAM, MED12, NLGN3, NLGN4X, OPHN1, PAFAH1B1, PQBP1, PTPN11, RAB39B, RELN, VPS13B, FMR1	Neurology	Neurological Disorders
D4027	Autism and Intellectual Disability Panel	2439	264 Primary genes associated with Intellectual Disability (including *FMR1*). 2,175 Secondary genes that have been associated, or thought to be involved with, and Autism or Autism-like features (not all are proven monogenic causes). Please contact us for the full gene list.	Neurology	Neurological Disorders
D4028	Wilson Disease Test	1	ATP7B	Neurology	Neurological Disorders
D4029	Childhood Ataxia with Central Nervous System Hypomyelin-ation	5	EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5	Neurology	Neurological Disorders
D4030	Comprehensive Myopathy Panel	52	ACTA1, ANO5, ATP2A1, BAG3, BIN1, CACNA1S, CAV3, CCDC78, CFL2, CNTN1, COL6A1, COL6A2, COL6A3, CPT2, CRYAB, DES, DNAJB6, DNM2, DYSF, FHL1, FKBP14, FLNC, GNE, KBTBD13, KCNJ2, KLHL40, KLHL41, LDB3, LMNA, LMOD3, MATR3, MEGF10, MTM1, MYH7, MYL2, MYOT, MYPN, NEB, RYR1, SCN4A, SELENON, SQSTM1, STAC3, STIM1, TIA1, TNNT1, TPM2, TPM3, TTN, VCP, COL12A1, MYF6	Neurology	Neuromuscular Disorders
D4031	Congenital Myopathy Panel	28	ACTA1, BIN1, CCDC78, CFL2, CNTN1, COL6A1, COL6A2, COL6A3, DNM2, FKBP14, KBTBD13, KLHL40, KLHL41, LMOD3, MEGF10, MTM1, MYH7, MYPN, NEB, RYR1, SELENON, STAC3, TNNT1, TPM2, TPM3, TTN, COL12A1, MYF6	Neurology	Neuromuscular Disorders

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D4032	Comprehensive Muscular Dystrophy Panel	56	ANO5, B3GALNT2, B4GAT1, CAPN3, CAV3, CHKB, COL6A1, COL6A2, COL6A3, DAG1, DES, DMD, DNAJB6, DPM1, DPM2, DPM3, DYSF, EMD, FHL1, FKRP, FKTN, GAA, GMPPB, ISPD, ITGA7, LAMA2, LARGE1, LMNA, MYOT, PLEC, PNPLA2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, SGCA, SGCB, SGCD, SGCG, TCAP, TMEM5, TNPO3, TRAPPC11, TRIM32, TTN, COL12A1, HNRNPDL, LIMS2, SUN1, SUN2, SYNE1, SYNE2, TMEM43, TOR1AIP1, SMCHD1	Neurology	Neuromuscular Disorders
D4033	Congenital Muscular Dystrophy Panel	27	B3GALNT2, B4GAT1, CHKB, COL6A1, COL6A2, COL6A3, DAG1, DMD, DPM1, DPM2, DPM3, FKRP, FKTN, GMPPB, ISPD, ITGA7, LAMA2, LARGE1, LMNA, POMGNT1, POMGNT2, POMK, POMT1, POMT2, TCAP, TMEM5, COL12A1	Neurology	Neuromuscular Disorders
D4034	Congenital Myasthenic Syndrome Panel	21	AGRN, ALG2, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, COLQ, DOK7, DPAGT1, GFPT1, MUSK, RAPSN, ALG14, GMPPB, LAMB2, LRP4, PLEC, PREPL, SCN4A, SNAP25	Neurology	Neuromuscular Disorders
D4035	Comprehensive Neuromuscular Disorders Panel	131	ACTA1, AGRN, ALG14, ALG2, ANO5, ATP2A1, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, CACNA1S, CAPN3, CAV3, CCDC78, CFL2, CHAT, CHKB, CHRNA1, CHRNB1, CHRND, CHRNE, CLCN1, CNTN1, COL12A1, COL6A1, COL6A2, COL6A3, COLQ, CPT2, CRYAB, DAG1, DES, DMD, DNAJB6, DNM2, DOK7, DPAGT1, DPM1, DPM2, DPM3, DYNC1H1, DYSF, EMD, FHL1, FKBP14, FKRP, FKTN, FLNC, GAA, GFPT1, GMPPB, GNE, HNRNPA2B1, HNRNPDL, IGHMBP2, ISPD, ITGA7, KBTBD13, KCNJ2, KLHL40, KLHL41, LAMA2, LAMB2, LAMP2, LARGE1, LDB3, LIMS2, LMNA, LMOD3, LRP4, MATR3, MEGF10, MTM1, MUSK, MYF6, MYH2, MYH7, MYL2, MYOT, MYPN, NEB, PHKA1, PLEC, PLEKHG5, PMP22, PNPLA2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PREPL, PYGM, RAPSN, RYR1, SCN4A, SELENON, SGCA, SGCB, SGCD, SGCG, SIL1, SMCHD1, SNAP25, SQSTM1, STAC3, STIM1, SUN1, SUN2, SYNE1, SYNE2, TAZ, TCAP, TIA1, TMEM43, TMEM5, TNNI2, TNNT1, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TRPV4, TTN, UBA1, VCP, VMA21, VRK1	Neurology	Neurological Disorders
D4036	Arthrogryposis Panel	11	TPM2, MYBPC1, MYH3, TNNT3, TNNI2, MYH8, FBN2, PIEZO2, ECEL1, DOK7, RAPSN	Neurology	Neurological Disorders
D4037	Charcot Marie Tooth Disease Panel	62	AARS, AIFM1, ARHGEF10, BSCL2, COX6A1, DHTKD1, DNAJB2, DNM2, DNMT1, DYNC1H1, EGR2, FAM134B, FBLN5, FGD4, FIG4, GAN, GARS, GDAP1, GJB1, GNB4, HARS, HINT1, HK1, HOXD10, HSPB1, HSPB8, IGHMBP2, IKBKAP, INF2, KARS, KIF1B, KIF5A, LITAF, LMNA, LRSAM1, MARS, MED25, MFN2, MORC2, MPZ, MTMR2, NDRG1, NEFL, PDK3, PLEKHG5, PMP22, PRPS1, PRX, RAB7A, REEP1, SBF1, SBF2, SH3TC2, SLC12A6, SLC25A46, SPG11, SURF1, TFG, TRIM2, TRPV4, VCP, YARS	Neurology	Neurological Disorders
D4045	Comprehensive DMD Test (Sequencing and Deletion/ Duplication)	1	DMD	Neurology	Neuromuscular Disorders
D4043	Adrenoleu-kodystrophy Panel	15	ABCD1, ACOX1, HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26	Neurology	X-Linked Disorders
D5000	ABCD1 XALD Gene Sequencing	1	ABCD1	Neurology	X-Linked Disorders

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D4100	Arrhythmogenic Cardiomyopathy Panel	24	ACTN2, DES, DSC2, DSG2, DSP, EMD, FLNC, JUP, LMNA, PKP2, PLN, PRKAG2, RBM20, RYR2, SCN5A, TMEM43, TNNI3, TNNT2, TTN, ANKRD1, CTNNA3, LDB3, PDLIM3, TGFB3	Cardiology	Cardiomyopathy
D4101	Comprehensive Cardiomyopathy Panel	122	ABCC9, ACTC1, ACTN2, AGL, ALPK3, BAG3, CACNA1C, CAV3, CRYAB, CSRP3, DES, DMD, DOLK, DSC2, DSG2, DSP, EMD, EYA4, FHL1, FKRP, FKTN, FLNC, GAA, GLA, HCN4, JUP, LAMP2, LMNA, MYBPC3, MYH7, MYL2, MYL3, PKP2, PLN, PRKAG2, RAF1, RBM20, RYR2, SCN5A, SGCD, SLC22A5, TAZ, TCAP, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL, ANKRD1, CALR3, CHRM2, CTF1, CTNNA3, DTNA, FHL2, GATA4, GATA6, GATAD1, ILK, JPH2, LAMA4, LDB3, LRRC10, MYH6, MIB1, MTND1, MTND5, MTND6, MTTD, MTTG, MTTH, MTTI, MTTK, MTTL1, MTTL2, MTTM, MTTQ, MTTS1, MTTS2, MURC, MYLK2, MYOM1, MYOZ2, MYPN, NEBL, NEXN, NKX2-5, NPPA, PDLIM3, PLEKHM2, PRDM16, TGFB3, TMPO, TXNRD2, A2ML1, BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NF1, NRAS, PTPN11, RASA1, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1, ACADVL, ALMS1, CPT2, DNAJC19, ELAC2, MTO1, SDHA, TMEM70	Cardiology	Cardiomyopathy
D4102	Dilated Cardiomyopathy Panel	69	ABCC9, ACTC1, ACTN2, BAG3, CAV3, CRYAB, CSRP3, DES, DMD, DOLK, DSC2, DSG2, DSP, EMD, EYA4, FKRP, FKTN, FLNC, JUP, LAMP2, LMNA, MYBPC3, MYH7, PKP2, PLN, RAF1, RBM20, RYR2, SCN5A, SGCD, SLC22A5, TAZ, TCAP, TMEM43, TNNC1, TNNI3, TNNT2, TPM1, TTN, TTR, VCL, ANKRD1, CHRM2, CTF1, FHL2, GATA4, GATA6, GATAD1, ILK, LAMA4, LDB3, LRRC10, MYH6, MYPN, NEBL, NEXN, NKX2-5, NPPA, PDLIM3, PLEKHM2, PRDM16, TMPO, TXNRD2, ACADVL, ALMS1, CPT2, DNAJC19, SDHA, TMEM70	Cardiology	Cardiomyopathy
D4103	Hypertrophic Cardiomyopathy Panel	60	ACTC1, ACTN2, AGL, BAG3, CACNA1C, CAV3, CSRP3, DES, FHL1, FLNC, GAA, GLA, LAMP2, MYBPC3, MYH7, MYL2, MYL3, PLN, PRKAG2, TCAP, TNNC1, TNNI3, TNNT2, TPM1, TTR, VCL, ANKRD1, CALR3, GATA4, JPH2, LDB3, MYH6, MYLK2, MYOM1, MYOZ2, MYPN, NEXN, PDLIM3, A2ML1, BRAF, CBL, HRAS, KRAS, MAP2K1, MAP2K2, NF1, NRAS, PTPN11, RAF1, RASA1, RIT1, RRAS, SHOC2, SOS1, SOS2, SPRED1, ACADVL, CPT2, ELAC2, MTO1	Cardiology	Cardiomyopathy
D4104	Sudden Cardiac Arrest (SCA) Panel	11	ANK2, CASQ2, CAV3, KCND3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, RYR2, SCN5A	Cardiology	Cardiomyopathy
D4105	Arrhythmia Comprehensive Panel	73	ABCC9, ACTN2, ANK2, CACNA1C, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, DES, DSC2, DSG2, DSP, EMD, FLNC, GPD1L, HCN4, JUP, KCNA5, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, LMNA, MYL4, NKX2-5, PKP2, PLN, PRKAG2, RBM20, RYR2, SCN5A, TMEM43, TNNI3, TNNT2, TRDN, TTN, AKAP9, ANKRD1, CACNA2D1, CTNNA3, GJA5, KCND3, KCNE3, KCNE5, KCNJ5, KCNJ8, KCNK3, LDB3, NPPA, PDLIM3, RANGRF, SCN10A, SCN1B, SCN2B, SCN3B, SCN4B, SLMAP, SNTA1, TGFB3, TRPM4, DEPDC5, KCNQ2, KCNQ3, KCNT1, PCDH19, PRRT2, SCN1A, SCN8A, SCN9A, SLC2A1	Cardiology	Arrhythmia
D4106	Long QT Syndrome Panel	17	ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNQ1, SCN5A, TRDN, AKAP9, KCNJ5, SCN4B, SNTA1	Cardiology	Arrhythmia
D4107	Short QT Syndrome Panel	6	CACNA1C, CACNB2, KCNH2, KCNJ2, KCNQ1, CACNA2D1	Cardiology	Arrhythmia
D4108	Brugada Syndrome Panel	9	CACNA1C, CACNB2, GPD1L, HCN4, KCNE3, SCN1B, SCN3B, SCN5A, SLMAP	Cardiology	Arrhythmia

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D4109	Cardiomyopathy and Skeletal Muscle Disease Panel	158	ABCC9, ACTA1, ACTC1, ACTN2, AGL, ANO5, ATP2A1, B3GALNT2, B4GAT1, BAG3, BIN1, CACNA1C, CAPN3, CAV3, CCDC78, CFL2, CHKB, CNTN1, COL6A1, COL6A2, COL6A3, CPT2, CRYAB, CSRP3, DAG1, DES, DMD, DNAJB6, DNM2, DOLK, DPM1, DPM2, DPM3, DSC2, DSG2, DSP, DYSF, EMD, EYA4, FHL1, FKBP14, FKRP, FKTN, FLNC, GAA, GLA, GMPPB, GNE, HCN4, ISPD, ITGA7, JUP, KBTBD13, KLHL40, KLHL41, LAMA2, LAMP2, LARGE1, LMNA, LMOD3, MATR3, MEGF10, MTM1, MYBPC3, MYH7, MYL2, MYL3, MYOT, MYPN, NEB, PKP2, PLEC, PLN, PNPLA2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PRKAG2, RAF1, RBM20, RYR1, RYR2, SCN5A, SELENON, SGCA, SGCB, SGCD, SGCG, SLC22A5, SQSTM1, STAC3, STIM1, TAZ, TCAP, TIA1, TMEM43, TMEM5, TNNC1, TNNI3, TNNT1, TNNT2, TNPO3, TPM1, TPM2, TPM3, TRAPPC11, TRIM32, TTN, TTR, VCL, VCP, ANKRD1, CALR3, CHRM2, COL12A1, CTF1, CTNNA3, DTNA, FHL2, GATA4, GATA6, GATAD1, HNRNPDL, ILK, JPH2, LAMA4, LDB3, LIMS2, LRRC10, MYF6, MYH6, MYLK2, MYOM1, MYOZ2, NEBL, NEXN, NKX2-5, NPPA, PDLIM3, PLEKHM2, PRDM16, SUN1, SUN2, SYNE1, SYNE2, TGFB3, TMPO, TOR1AIP1, TXNRD2, ACADVL, ALMS1, DNAJC19, ELAC2, MTO1, SDHA, TMEM70	Cardiology	Cardiomyop-athy
D4110	Marfan/TAAD Panel	23	ACTA2, CBS, COL3A1, COL5A1, COL5A2, FBN1, FBN2, FLNA, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PRKG1, SKI, SLC2A10, SMAD3, SMAD4, TGFB2, TGFB3, TGFBR1, TGFBR2	Cardiology	Connective Tissue
D4111	Marfan Syndrome	1	FBN1	Cardiology	Connective Tissue

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TEST CODE	TEST NAME	NO. OF GENES	GENES INVOLVED	CATEGORY	DESCRIPTION
D4200	Colorectal Cancer Panel	25	APC, AXIN2, BMPR1A, CDH1, CHEK2, EPCAM, GREM1, MLH1, MSH2, MSH6, MUTYH, PMS2, POLD1, POLE, PTEN, SMAD4, STK11, TP53, ATM, BLM, BUB1B, ENG, FLCN, GALNT12, MLH3	Hereditary Cancer	Colon
D4201	Pancreatic Cancer Panel	29	APC, ATM, BMPR1A, BRCA1, BRCA2, CDKN2A, EPCAM, MEN1, MLH1, MSH2, MSH6, NF1, PALB2, PMS2, SMAD4, STK11, TP53, TSC1, TSC2, VHL, CDK4, FANCC, PALLD, CASR, CFTR, CTRC, PRSS1, SPINK1, XRCC2	Hereditary Cancer	Pancreatic
D4202	Melanoma Cancer Panel	9	BAP1, BRCA2, CDK4, CDKN2A, MITF, POT1, PTEN1, RB1, TP53	Hereditary Cancer	Skin
D4203	Multiple Endocrine Neoplasias/ Paraganglioma/ Peochromocy-toma Panel	14	MAX, NF1, RET, SDHA, SDHAF2, SDHB, SDHC, SDHD, TMEM127, VHL, EGLN1, FH, KIF1B, MEN1	Hereditary Cancer	Other
D4204	Renal Cancer Panel	30	BAP1, CDC73, CDKN1C, DICER1, DIS3L2, EPCAM, FH, FLCN, GPC3, MET, MLH1, MSH2, MSH6, PMS2, PTEN, SDHB, SDHC, SMARCA4, SMARCB1, TP53, TSC1, TSC2, VHL, WT1, BUB1B, CEP57, MITF, PALB2, SDHA, SDHD	Hereditary Cancer	Renal
D4205	Comprehensive Cancer Panel	82	ALK, APC, ATM, AXIN2, BAP1, BARD1, BLM, BMPR1A, BRCA1, BRCA2, BRIP1, CASR, CDC73, CDH1, CDK4, CDKN1B, CDKN1C, CDKN2A, CEBPA, CHEK2, DICER1, DIS3L2, EGFR, EPCAM, FANCC, FH, FLCN, GATA2, GPC3, GREM1, HOXB13, HRAS, KIT, MAX, MEN1, MET, MITF, MLH1, MSH2, MSH6, MUTYH, NBN, NF1, NF2, PALB2, PDGFRA, PHOX2B, PMS2, POLD1, POLE, POT1, PRKAR1A, PTCH1, PTEN, RAD50, RAD51C, RAD51D, RB1, RECQL4, RET, RUNX1, SDHA, SDHAF2, SDHB, SDHC, SDHD, SMAD4, SMARCA4, SMARCB1, SMARCE1, STK11, SUFU, TERC, TERT, TMEM127, TP53, TSC1, TSC2, VHL, WRN, WT1, XRCC2	Hereditary Cancer	Multiple
D4206	Breast/Ovarian Cancer Panel	20	ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, EPCAM, FANCC, MLH1, MSH2, MSH6, NBN, PALB2, PMS2, PTEN, RAD51C, RAD51D, TP53, XRCC2	Hereditary Cancer	Breast/Ovarian
D4207	Pediatric Tumor Panel	27	ALK, APC, CDC73 (HRPT2), DICER1, EPCAM, MEN1, MLH1, MSH2, MSH6, NF1, NF2, PHOX2B, PMS2, PRKAR1A, PTCH1, PTEN, RB1, RET, SMARCA4, SMARCB1, STK11, SUFU, TP53, TSC1, TSC2, VHL, WT1	Hereditary Cancer	Pediatric
D4208	Prostate Cancer Panel	12	ATM, BRCA1, BRCA2, CHEK2, EPCAM, HOXB13, MLH1, MSH2, MSH6, NBN, PMS2, TP53	Hereditary Cancer	Prostate
D4209	Hereditary Breast and Ovarian Cancer Syndrome Panel	2	BRCA1, BRCA2	Hereditary Cancer	Breast/Ovarian
D4210	Breast and Gyn Cancers Panel	35	ATM, BARD1, BRCA1, BRCA2, BRIP1, CDH1, CHEK2, DICER1, EPCAM, MLH1, MSH2, MSH6, NBN, NF1, PALB2, PMS2, PTEN, RAD50, RAD51C, RAD51D, SMARCA4, STK11, TP53, AKT1, CDC73, FAM175A, FANCC, MRE11, MUTYH, PIK3CA, POLD1, RINT1, SDHB, SDHD, XRCC2	Hereditary Cancer	Breast/Gyn
D4211	Tuberous Sclerosis Panel	2	TSC1, TSC2	Hereditary Cancer	Other

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D4300	Microphthalmia/ Anophthalmia/ Coloboma Spectrum Panel	48	ABCB6, ALDH1A3, BCOR, BMP4, CHD7, ERCC1, ERCC2, ERCC5, ERCC6, FOXE3, FOXL2, FRAS1, FREM1, FREM2, GDF3, GDF6, GJA1, GRIP1, HCCS, HMGB3, HMX1, MAB21L2, MFRP, NAA10, NDP, OCRL, OTX2, PAX2, PAX6, PRSS56, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, RBP4, SALL2, SHH, SIX3, SIX6, SMOC1, SOX2, STRA6, TBC1D20, TENM3, TFAP2A, VAX1, VSX2	Ophthalmology	
D4301	Progressive External Ophthalmoplegia (PEO)/Optic Atrophy Nuclear Gene Panel	55	ACO2, ALG13, ALG3, APTX, AUH, C10ORF2, C12ORF65, CISD2, CLPB, COX7B, DARS, DDHD2, DGUOK, DNA2, DNAJC19, DNM1L, DPM1, EARS2, FH, GYG2, ISCA2, MCEE, MFF, MFN2, MGME1, MOGS, MTFMT, MTO1, MTPAP, NARS2, NDUFAF3 (C3ORF60), NR2F1, OPA1, OPA3, PDHX, PDSS1, POLG, POLG2, PRPS1, RRM2B, SLC19A2, SLC19A3, SLC25A4, SPG7, SRD5A3, STT3B, SUCLA2, TACO1, TIMM8A, TK2, TMEM126A, TSFM, TYMP, VARS2, WFS1	Ophthalmology	
D4302	Achromatopsia, Cone, and Cone-rod Dystrophy Panel	36	ABCA4, ADAM9, AIPL1, BEST1, C8orf37, CABP4, CACNA1F, CACNA2D4, CDHR1, CEP290, CERKL, CNGA3, CNGB3, CNNM4, CRX, GNAT2, GUCA1A, GUCA1B, GUCY2D, KCNV2, PAX6, PDE6C, PDE6H, PITPNM3, PROM1, PRPH2, RAX2, RBP4, RDH5, RGS9, RGS9BP, RIMS1, RPGR, RPGRIP1, SEMA4A, UNC119	Ophthalmology	
D4303	Retinitis Pigmentosa Panel	66	ABCA4, AIPL1, BBS1, BEST1, C1QTNF5, C2orf71, C8orf37, CA4, CERKL, CHM, CLN3, CLRN1, CNGA1, CNGB1, CRB1, CRX, CYP4V2, DHDDS, EYS, FAM161A, FLVCR1, FSCN2, GUCA1B, GUCY2D, IDH3B, IMPDH1, IMPG2, KLHL7, LRAT, MAK, MERTK, NR2E3, NRL, OFD1, PDE6A, PDE6B, PDE6G, PRCD, PROM1, PRPF3, PRPF31, PRPF6, PRPF8, PRPH2, RBP3, RBP4, RDH12, RGR, RHO, RLBP1, ROM1, RP1, RP2, RP9, RPE65, RPGR, RPGRIP1, SAG, SEMA4A, SNRNP200, SPATA7, TOPORS, TTC8, TULP1, USH2A, ZNF513	Ophthalmology	
D4304	Retina/ Photoreceptor Dystrophy Panel	121	ABCA4, ADAM9, AIPL1, BBS1, BEST1, C1QTNF5, C2orf71, C8orf37, CA4, CABP4, CACNA1F, CACNA2D4, CDH3, CDHR1, CEP290, CERKL, CHM, CLN3, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, CRB1, CRX, CYP4V2, DHDDS, EFEMP1, ELOVL4, EYS, FAM161A, FLVCR1, FSCN2, FZD4, GNAT1, GNAT2, GPR179, GRM6, GUCA1A, GUCA1B, GUCY2D, IDH3B, IMPDH1, IMPG2, IQCB1, KCNJ13, KCNV2, KLHL7, LCA5, LRAT, LRIT3, LRP5, MAK, MERTK, MFN2, NDP, NR2E3, NRL, NYX, OAT, OFD1, OPA1, OPA3, OTX2, PAX6, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PITPNM3, PLA2G5, PRCD, PROM1, PRPF3, PRPF31, PRPF6, PRPF8, PRPH2, RAX2, RBP3, RBP4, RD3, RDH12, RDH5, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP2, RP9, RPE65, RPGR, RPGRIP1, SAG, SEMA4A, SLC24A1, SNRNP200, SPATA7, TIMM8A, TIMP3, TMEM126A, TOPORS, TRPM1, TSPAN12, TTC8, TULP1, UNC119, USH2A, VCAN, ZNF513	Ophthalmology	
D4305	Macular Dystrophy/ Degeneration/ Stargardt Disease Panel	15	ABCA4, BEST1, CDH3, CNGB3, EFEMP1, ELOVL4, FSCN2, GUCA1B, PROM1, PRPH2, RBP4, RDH12, RPGR, RPGRIP1, TIMP3	Ophthalmology	

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D4306	Eye Disorders Comprehensive Panel	211	ABCA4, ABHD12, ADAM9, ADGRV1, AHI1, AIPL1, ALMS1, ARL13B, ARL6, ATP13A2, B3GLCT, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BCOR, BEST1, BMP4, C10orf11, C1QTNF5, C2orf71, C5orf42, C8orf37, CA4, CABP4, CACNA1F, CACNA2D4, CC2D2A, CDH23, CDH3, CDHR1, CEP290, CEP41, CERKL, CHM, CIB2, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNNM4, COL11A1, COL11A2, COL2A1, COL4A1, COL9A1, COL9A2, CRB1, CRX, CTSD, CYP1B1, CYP27A1, CYP4V2, DHDDS, EFEMP1, ELOVL4, EYS, FAM161A, FLVCR1, FOXC1, FOXE3, FRAS1, FREM1, FREM2, FSCN2, FZD4, GNAT1, GNAT2, GPR143, GPR179, GRIP1, GRM6, GRN, GUCA1A, GUCA1B, GUCY2D, HARS, HCCS, IDH3B, IMPDH1, IMPG2, INVS, IQCB1, KCNJ13, KCNV2, KCTD7, KIF7, KLHL7, LCA5, LRAT, LRIT3, LRP5, LZTFL1, MAK, MERTK, MFN2, MFRP, MFSD8, MKKS, MKS1, MTTP, MYO7A, MYOC, NDP, NPHP1, NPHP3, NPHP4, NR2E3, NRL, NYX, OAT, OCA2, OCRL, OFD1, OPA1, OPA3, OTX2, PAX6, PCDH15, PDE6A, PDE6B, PDE6C, PDE6G, PDE6H, PEX7, PHYH, PITPNM3, PITX2, PITX3, PLA2G5, PPT1, PRCD, PROM1, PRPF3, PRPF31, PRPF6, PRPF8, PRPH2, RAX2, RBP3, RBP4, RD3, RDH12, RDH5, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RP1, RP2, RP9, RPE65, RPGR, RPGRIP1, RPGRIP1L, RS1, SAG, SDCCAG8, SEMA4A, SLC24A1, SLC24A5, SLC45A2, SMOC1, SNRNP200, SOX2, SPATA7, STRA6, TCTN1, TCTN2, TCTN3, TIMM8A, TIMP3, TMEM126A, TMEM216, TMEM237, TMEM67, TOPORS, TPP1, TRIM32, TRPM1, TSPAN12, TTC21B, TTC8, TULP1, TYR, TYRP1, UNC119, USH1C, USH1G, USH2A, VAX1, VCAN, VSX2, WDPCP, WFS1, WHRN, WT1, ZNF423, ZNF513	Ophthalmology	
D4307	Flecked-retina Disorders Panel	6	ABCA4, PLA2G5, PRPH2, RDH5, RHO, RLBP1	Ophthalmology	
D4308	Usher Syndrome Panel	12	ABHD12, ADGRV1, CDH23, CIB2, CLRN1, HARS, MYO7A, PCDH15, USH1C, USH1G, USH2A, WHRN	Ophthalmology	
D4309	Albinism Panel	28	AP3B1, BLOC1S3, BLOC1S6, C10ORF11, DTNBP1, EDN3, EDNRB, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, KIT, LYST, MC1R, MITF, MLPH, MYO5A, OCA2, PAX3, RAB27A, SLC24A5, SLC45A2, SNAI2, SOX10, TYR, TYRP1	Ophthalmology	
D4310	Alport Syndrome Panel	3	COL4A3, COL4A4, COL4A5	Ophthalmology	
D4311	Bardet Biedl Panel	21	ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CCDC28B, CEP290, IFT27, LZTFL1, MKKS, MKS1, SDCCAG8, TMEM67, TRIM32, TTC8, WDPCP	Ophthalmology	
D4702	Pulmonary Fibrosis and Hermansky-Pudlak Syndrome Panel	16	ABCA3, AP3B1, BLOC1S3, BLOC1S6, CSF2RA, DTNBP1, ELMOD2, HPS1, HPS3, HPS4, HPS5, HPS6, SFTPB, SFTPC, SFTPD, TERT	Ophthalmology	

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D4400	Deafness Non-Syndromic Sensorineural Autosomal Dominant Panel	30	ACTG1, CCDC50, COCH, COL11A2, CRYM, DFNA5, DIABLO, DIAPH1, DIAPH3, EYA4, GJB2, GJB3, GJB6, GRHL2, KCNQ4, MIR96, MYH14, MYH9, MYO6, MYO7A, POU3F4, POU4F3, PRPS1, SIX1, SLC17A8, SMPX, TECTA, TJP2, TMC1, WFS1	Hearing Disorders	
D4401	Deafness Non-Syndromic Sensorineural Autosomal Recessive Panel	46	CDH23, CLDN14, COL11A2, DFNB31, DFNB59, ESPN, ESRRB, FOXI1, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRXCR1, HGF, ILDR1, KCNJ10, LHFPL5, LOXHD1, LRTOMT, MARVELD2, MSRB3, MYO15A, MYO3A, MYO6, MYO7A, OTOA, OTOF, PCDH15, POU3F4, PRPS1, PTPRQ, RDX, SERPINB6, SLC12A1, SLC26A4, SLC26A5, SMPX, STRC, TECTA, TMC1, TMIE, TMPRSS3, TPRN, TRIOBP, USH1C	Hearing Disorders	
D4402	Waardenburg Syndrome Panel	7	EDN3, EDNRB, MITF, PAX3, SNAI2, SOX10, TYR	Hearing Disorders	
D4403	Usher Syndrome Panel	12	ABHD12, ADGRV1, CDH23, CIB2, CLRN1, HARS, MYO7A, PCDH15, USH1C, USH1G, USH2A, WHRN	Hearing Disorders	
D4310	Alport Syndrome Panel	3	COL4A3, COL4A4, COL4A5	Hearing Disorders	
D4405	Treachers Collins Syndrome Sequencing Panel	3	TCOF1, POLR1C, POLR1D	Hearing Disorders	

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D4500	Ceroid Lipofuscinosis Panel	13	ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, DNAJC5, GRN, KCTD7, MFSD8, PPT1, TPP1	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D4501	Comprehensive Lysosomal Storage Disorders Panel	54	AGA, ARSA, ARSB, ASAH1, ATP13A2, TPP1 (CLN2), CLN3, CLN5, CLN6, CLN8, CTNS, CTSA, CTSD, CTSF, CTSK, DNAJC5, FUCA1, GAA, GALC, GALNS, GBA, GLA, GLB1, GM2A, GNPTAB, GNPTG, GNS, GRN, GUSB, HEXA, HEXB, HGSNAT, HYAL1, IDS, IDUA, KCTD7, LAMP2, LIPA, MAN2B1, MANBA, MCOLN1, MFSD8, NAGA, NAGLU, NEU1, NPC1, NPC2, PPT1, PSAP, SGSH, SLC17A5, SMPD1, SUMF1, CHIT1	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D4502	Comprehensive Mucopoly-saccharidoses (MPS) Panel	23	ARSB, GALNS, GLB1, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, NAGLU, SGSH, AGA, CTSA, CTSK, FUCA1, GNPTAB, GNPTG, MAN2B1, MANBA, MCOLN1, NAGA, NEU1, SLC17A5	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D4503	Farber Lipogranuloma-tosis Test	1	ASAH1	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D5033	GLA Gene Sequencing	1	GLA	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D4505	GM2 Gangliosidosis Panel	3	GM2A, HEXA, HEXB	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D4506	Krabbe Disease Panel	2	GALC, PSAP	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D4507	Lysosomal Acid Lipase Deficiency	1	LIPA	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D4508	Metachromatic Leukodystrophy Panel	7	ARSA, PSAP, SUMF1, ASPA, GALC, HEXA, HEXB	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D4509	Mucolipidosis Panel	4	GNPTAB, GNPTG, MCOLN1, NEU1	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D5058	SUMF1 Gene Sequencing	1	SUMF1	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D4511	Comprehensive Neuronal Ceroid Lipofuscinoses Panel	13	TPP1 (CLN2), CLN3, CLN5, CLN6, CLN8, CTSD, KCTD7, MFSD8, PPT1, ATP13A2, CTSF, DNAJC5, GRN	Metabolic/ Mitochondrial	Lysosomal Storage Disorder

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TEST CODE	TEST NAME	NO. OF GENES	GENES INVOLVED	CATEGORY	DESCRIPTION
D5057	SMPD1 Gene Sequencing	1	SMPD1	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D4513	Niemann-Pick Type C Panel	3	NPC1, NPC2, LIPA	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D4514	Oligo-saccharidoses Panel	23	AGA, CTSA, CTSK, FUCA1, MAN2B1, MANBA, NAGA, SLC17A5, ARSB, GALNS, GLB1, GNPTAB, GNPTG, GNS, GUSB, HGSNAT, HYAL1, IDS, IDUA, MCOLN1, NAGLU, NEU1, SGSH	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D5025	GAA Gene Sequencing	1	GAA	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D4516	Prosaposin Deficiency Test	1	PSAP	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D4517	Sandhoff Disease Test	1	HEXB	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D4518	Tay-Sachs Disease Test	1	HEXA	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D4043	X-Linked Adrenoleuko• dystrophy Panel	15	ABCD1, ACOX1, HSD17B4, PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D5009	ARSB Gene Sequencing	1	ARSB	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D5031	GALC Gene Sequencing	1	GALC	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D5028	GALNS Gene Sequencing	1	GALNS	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D5032	GBA Gene Sequencing	1	GBA	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D5034	GLB1 Gene Sequencing	1	GLB1	Metabolic/ Mitochondrial	Lysosomal Storage Disorder

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TEST CODE	TEST NAME	NO. OF GENES	GENES INVOLVED	CATEGORY	DESCRIPTION
D5035	GUSB Gene Sequencing	1	GUSB	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D5042	IDS Gene Sequencing	1	IDS	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D5041	IDUA Gene Sequencing	1	IDUA	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D4528	NAGLU MP-SIIIB Gene Sequencing	1	NAGLU	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D4044	ABCD1 Gene Sequencing	1	ABCD1	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D4530	Cystinosis Test	1	CTNS	Metabolic/ Mitochondrial	Lysosomal Storage Disorder
D4531	Non Ketotic Hyperglycinemia Panel	3	ATM, GCSH, GLDC	Metabolic/ Mitochondrial	Metabolic Disorders
D4532	Urea Cycle Disorders Panel	15	ALDH18A1, ARG1, ASL, ASS1, CPS1, NAGS, OAT, OTC, SLC25A13, SLC25A15, CA5A, GLUD1, GLUL, SLC7A7, UMPS	Metabolic/ Mitochondrial	Metabolic Disorders
D4533	Low C0 Test	1	SLC22A5	Metabolic/ Mitochondrial	Metabolic Disorders
D4534	Elevated C0/ (C16+C18) Test	1	CPT1A	Metabolic/ Mitochondrial	Metabolic Disorders
D4535	Elevated C3 Panel	9	BTD, HLCS, MMAA, MMAB, MMACHC, MMADHC, MUT, PCCA, PCCB	Metabolic/ Mitochondrial	Metabolic Disorders
D4536	Elevated C3-DC Test	1	MLYCD	Metabolic/ Mitochondrial	Metabolic Disorders
D4537	Elevated C4 Panel	3	ACAD8, ACADS, ETHE1	Metabolic/ Mitochondrial	Metabolic Disorders
D4538	Elevated C4-OH Panel	2	HADH, HIBCH	Metabolic/ Mitochondrial	Metabolic Disorders
D4539	Elevated C4 & C5 Panel	7	ETFA, ETFB, ETFDH, ETHE1, SLC52A1, SLC52A2, SLC52A3	Metabolic/ Mitochondrial	Metabolic Disorders
D4540	Elevated C5 Panel	2	ACADSB, IVD	Metabolic/ Mitochondrial	Metabolic Disorders

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TEST CODE	TEST NAME	NO. OF GENES	GENES INVOLVED	CATEGORY	DESCRIPTION
D4541	Elevated C5-OH Panel	12	ACAT1, AUH, BTD, DNAJC19, HLCS, HMGCL, HSD17B10, MCCC1, MCCC2, OPA3, SERAC1, TAZ	Metabolic/ Mitochondrial	Metabolic Disorders
D4542	Elevated C16-OH, C16:1-OH, C18-OH & C18:1-OH Panel	2	HADHA, HADHB	Metabolic/ Mitochondrial	Metabolic Disorders
D4543	Elevated C16, C16:1, C18, & C18:1 Panel	2	CPT2, SLC25A20	Metabolic/ Mitochondrial	Metabolic Disorders
D4544	Elevated Arginine Test	1	ARG1	Metabolic/ Mitochondrial	Metabolic Disorders
D4545	Elevated Citrulline Panel	5	ASL, ASS1, PC, SLC25A13, DLD	Metabolic/ Mitochondrial	Metabolic Disorders
D4546	Elevated Glycine Panel	62	AMT, GLDC, GCSH, LIAS, NFU1, SLC6A9, ACAD8, ACADSB, ACAT1, ACSF3, ASPA, AUH, BCKDHA, BCKDHB, BTD, D2HGDH, DBT, DHTKD1, DLD, DNAJC19, ETFA, ETFB, ETFDH, ETHE1, FBP1, FH, FTCD, GCDH, GSS, HIBCH, HLCS, HMGCL, HSD17B10, IDH2, IVD, L2HGDH, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MUT, NFU1, OGDH, OPA3, OPLAH, OXCT1, PCCA, PCCB, POLG, PPM1K, SERAC1, SLC13A5, SLC25A1, SLC25A19, SUCLA2, SUCLG1, TAZ, TMEM70	Metabolic/ Mitochondrial	Metabolic Disorders
D4547	Elevated Leucine Panel	5	BCKDHA, BCKDHB, DBT, DLD, PPM1K	Metabolic/ Mitochondrial	Metabolic Disorders
D4548	Elevated Methionine Panel	6	AHCY, CBS, GNMT, MAT1A, FAH, SLC25A13	Metabolic/ Mitochondrial	Metabolic Disorders
D4549	Elevated Phenylalanine Panel	6	GCH1, PAH, PCBD1, PTS, QDPR, SPR	Metabolic/ Mitochondrial	Metabolic Disorders
D4550	Elevated Proline Panel	2	ALDH4A1, PRODH	Metabolic/ Mitochondrial	Metabolic Disorders
D4551	Elevated Succinylacetone Test	1	FAH	Metabolic/ Mitochondrial	Metabolic Disorders
D4552	Elevated Tyrosine Panel	3	FAH, HPD, TAT	Metabolic/ Mitochondrial	Metabolic Disorders
D4553	Alkaptonuria Test	1	HGD	Metabolic/ Mitochondrial	Metabolic Disorders

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TEST CODE	TEST NAME	NO. OF GENES	GENES INVOLVED	CATEGORY	DESCRIPTION
D4554	Combined Methylmalonic Acidemia and Homocystinuria Panel	11	ABCD4, AMN, CD320, CUBN, GIF, HCFC1, LMBRD1, MMACHC, MMADHC, TCN1, TCN2	Metabolic/ Mitochondrial	Metabolic Disorders
D4555	Cystinuria Panel	3	PREPL, SLC3A1, SLC7A9	Metabolic/ Mitochondrial	Metabolic Disorders
D4556	Disorders of Serine Biosynthesis Panel	3	PHGDH, PSAT1, PSPH	Metabolic/ Mitochondrial	Metabolic Disorders
D4557	Glycine Encephalopathy Panel	6	AMT, GCSH, GLDC, LIAS, NFU1, SLC6A9	Metabolic/ Mitochondrial	Metabolic Disorders
D4558	Homocystinuria Panel	19	CBS, MTHFR, MTR, MTRR, ABCD4, AMN, CD320, CUBN, GIF, HCFC1, LMBRD1, MMACHC, MMADHC, TCN1, TCN2, AHCY, CBS, GNMT, MAT1A	Metabolic/ Mitochondrial	Metabolic Disorders
D4559	Hyperphenylal-aninemia Panel	6	GCH1, PAH, PCBD1, PTS, QDPR, SPR	Metabolic/ Mitochondrial	Metabolic Disorders
D4560	Hyperprolinemia Panel	2	ALDH4A1, PRODH	Metabolic/ Mitochondrial	Metabolic Disorders
D4561	Maple Syrup Urine Disease Panel	5	BCKDHA, BCKDHB, DBT, PPM1K, DLD	Metabolic/ Mitochondrial	Metabolic Disorders
D4562	Tyrosinemia Panel	3	FAH, HPD, TAT	Metabolic/ Mitochondrial	Metabolic Disorders
D4563	Galactosemia Panel	3	GALE, GALK1, GALT	Metabolic/ Mitochondrial	Metabolic Disorders
D5024	G6PD Gene Sequencing	1	G6PD	Metabolic/ Mitochondrial	Metabolic Disorders
D4565	Glucose Transporter Type 1 Deficiency Syndrome Test	1	SLC2A1	Metabolic/ Mitochondrial	Metabolic Disorders
D4566	Comprehensive Glycogen Storage Disease Panel	23	AGL, ALDOA, ENO3, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, PFKM, PGAM2, PHKA1, PHKA2, PHKB, PHKG2, PYGL, PYGM, RBCK1, SLC2A2, SLC37A4	Metabolic/ Mitochondrial	Metabolic Disorders
D4567	Liver Glycogen Storage Disease Panel	11	AGL, FBP1, G6PC, GBE1, GYS2, PHKA2, PHKB, PHKG2, PYGL, SLC2A2, SLC37A4	Metabolic/ Mitochondrial	Metabolic Disorders

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TEST CODE	TEST NAME	NO. OF GENES	GENES INVOLVED	CATEGORY	DESCRIPTION
D4568	Muscle Glycogen Storage Disease Panel	15	ALDOA, ENO3, GAA, GBE1, GYG1, GYS1, LAMP2, LDHA, PFKM, PGAM2, PHKA1, PHKB, PYGM, RBCK1, PGM1	Metabolic/ Mitochondrial	Metabolic Disorders
D4569	Hereditary Fructose Intolerance Test	1	ALDOB	Metabolic/ Mitochondrial	Metabolic Disorders
D4570	Rare Carbohydrate Disorders Panel	2	FBP1, SLC5A1	Metabolic/ Mitochondrial	Metabolic Disorders
D4571	Congenital Disorders of Glycosylation Panel	103	ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ATP6V0A2, B3GLCT, CHST14, COG1, COG2, COG4, COG5, COG6, COG7, COG8, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, G6PC3, GFPT1, GMPPA, GMPPB, MAGT1, MAN1B1, MGAT2, MOGS, MPDU1, MPI, NGLY1, PGM1, PGM3, PMM2, RFT1, SEC23B, SLC35A1, SLC35A2, SLC35C1, SRD5A3, SSR4, TMEM165, TRIP11, TUSC3, ALG14, B4GALT1, DDOST, NUS1, PIGM, RPN2, SEC23A, SLC35A3, ST3GAL3, STT3A, STT3B, B3GALNT2, B4GAT1, FKRP, FKTN, GNE, ISPD, LARGE1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, TMEM5, B3GALT6, B3GAT3, B4GALNT1, B4GALT7, C1GALT1C1, CHST3, CHST6, CHSY1, DSE, EOGT, EXT1, EXT2, GALNT3, LFNG, PAPSS2, PIGA, PIGL, PIGM, PIGN, PIGO, PIGQ, PIGT, PIGV, PIGW, POFUT1, POGLUT1, SLC26A2, SLC35D1, ST3GAL5, XYLT1	Metabolic/ Mitochondrial	Metabolic Disorders
D4572	Fatty Acid Oxidation Defects Panel	22	ACADM, ACADS, ACADSB, ACADVL, CPT1A, CPT2, ETFA, ETFB, ETFDH, HADH, HADHA, HADHB, HMGCL, HMGCS2, MLYCD, NADK2, SLC22A5, SLC25A20, DECR1, SLC52A1, SLC52A2, SLC52A3	Metabolic/ Mitochondrial	Metabolic Disorders
D4573	Ketogenesis Disorders Panel	2	HMGCL, HMGCS2	Metabolic/ Mitochondrial	Metabolic Disorders
D4574	Congenital Hypothyroidism	8	PAX8, TSHB, TSHR, NKX2, DUOX2, SLC5A5, TG, TPO	Metabolic/ Mitochondrial	Metabolic Disorders
D4575	Ketolysis Disorders Panel	2	ACAT1, OXCT1	Metabolic/ Mitochondrial	Metabolic Disorders
D5002	ACADM Gene Sequencing	1	ACADM	Metabolic/ Mitochondrial	Metabolic Disorders
D4577	Multiple Acyl-CoA Dehydrogenase Deficiency Panel	6	ETFA, ETFB, ETFDH, SLC52A1, SLC52A2, SLC52A3	Metabolic/ Mitochondrial	Metabolic Disorders
D5005	ACADVL Gene Sequencing	1	ACADVL	Metabolic/ Mitochondrial	Metabolic Disorders
D4579	Copper Metabolism Disorders Panel	5	AP1S1, ATP7A, ATP7B, CP, SLC33A1	Metabolic/ Mitochondrial	Metabolic Disorders

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TEST CODE	TEST NAME	NO. OF GENES	GENES INVOLVED	CATEGORY	DESCRIPTION
D4580	Organic Acidemias Panel	56	ACAD8, ACADSB, ACAT1, ACSF3, ASPA, AUH, BCKDHA, BCKDHB, BTD, D2HGDH, DBT, DNAJC19, ETFA, ETFB, ETFDH, ETHE1, FBP1, FTCD, GCDH, GSS, HIBCH, HLCS, HMGCL, HSD17B10, IDH2, IVD, L2HGDH, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MUT, OPA3, OPLAH, OXCT1, PCCA, PCCB, POLG, PPM1K, SERAC1, SLC25A1, SUCLA2, SUCLG1, TAZ, TMEM70, DHTKD1, DLD, FH, NFU1, OGDH, SLC13A5, SLC25A19	Metabolic/ Mitochondrial	Metabolic Disorders
D4581	2-Hydroxyglutaric Aciduria Panel	4	D2HGDH, IDH2, L2HGDH, SLC25A1	Metabolic/ Mitochondrial	Metabolic Disorders
D3007	MCCC1 and MCCC2 Gene Sequencing	2	MCCC1, MCCC2	Metabolic/ Mitochondrial	Metabolic Disorders
D4583	3-Methylglu-taconic Aciduria Panel	7	AUH, CLPB, DNAJC19, OPA3, SERAC1, TAZ, TMEM70	Metabolic/ Mitochondrial	Metabolic Disorders
D4584	Congenital Adrenal Hyperplasia Panel (by Sanger)	7	CYP21A2, CYP17A1, CYP11A1, CYP11B1, HSD3B2, STAR, ACTHR	Metabolic/ Mitochondrial	Metabolic Disorders
D4585	Barth Syndrome Test	1	TAZ	Metabolic/ Mitochondrial	Metabolic Disorders
D5014	BTD Gene Sequencing	1	BTD	Metabolic/ Mitochondrial	Metabolic Disorders
D4587	Canavan Disease Test	1	ASPA	Metabolic/ Mitochondrial	Metabolic Disorders
D5030	GCDH Gene Sequencing	1	GCDH	Metabolic/ Mitochondrial	Metabolic Disorders
D4589	Methylmalonic Acidemia Panel	18	MMAA, MMAB, MMADHC, MCEE, MUT, SUCLA2, SUCLG1, ACSF3, ABCD4, AMN, CD320, CUBN, GIF, HCFC1, LMBRD1, MMACHC, TCN1, TCN2	Metabolic/ Mitochondrial	Metabolic Disorders
D4590	Multiple Carboxylase Deficiency Panel	2	BTD, HLCS	Metabolic/ Mitochondrial	Metabolic Disorders
D4591	Propionic Acidemia Panel	9	PCCA, PCCB, MMAA, MMAB, MMADHC, MMACHC, MUT, BTD, HLCS	Metabolic/ Mitochondrial	Metabolic Disorders
D4592	Purine Metabolism Disorders Panel	10	ADA, ADSL, AMPD1, HPRT1, GPHN, MOCOS, MOCS1, PNP, XDH, SUOX	Metabolic/ Mitochondrial	Metabolic Disorders

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TEST CODE	TEST NAME	NO. OF GENES	GENES INVOLVED	CATEGORY	DESCRIPTION
D4593	2-Ketoglutarate Dehydrogenase Deficiency Panel	4	DLD, OGDH, SLC25A19, DHTKD1	Metabolic/ Mitochondrial	Metabolic Disorders
D4594	Citrate Transporter Deficiency Test	1	SLC13A5	Metabolic/ Mitochondrial	Metabolic Disorders
D4595	Dihydrolipoamide Dehydrogenase Deficiency Test	1	DLD	Metabolic/ Mitochondrial	Metabolic Disorders
D4596	Fumarase Deficiency Test	1	FH	Metabolic/ Mitochondrial	Metabolic Disorders
D4597	Pyruvate Carboxylase Deficiency Test	1	PC	Metabolic/ Mitochondrial	Metabolic Disorders
D4598	Pyruvate Dehydrogenase Deficiency Panel	8	DLAT, DLD, LIAS, MPC1, PDHA1, PDHB, PDHX, PDP1	Metabolic/ Mitochondrial	Metabolic Disorders
D4599	Treatable Neurometabolic Disorders Panel	133	ABCD1, ACAT1, AGA, ALDH5A1, ALDH7A1, AMN, AMT, ARG1, ARSA, ASL, ASS1, ATP7A, ATP7B, AUH, BCKDHA, BCKDHB, BTD, CBS, TPP1 (CLN2), CP, CPS1, CUBN, CYP27A1, DBT, DHCR7, DLAT, DLD, ETFA, ETFB, ETFDH, ETHE1, GAMT, GATM, GCDH, GCH1, GCSH, GIF, GLA, GLDC, GLUD1, GUSB, HLCS, HMGCL, HMGCS2, HSD17B10, IDS, IDUA, IVD, LIPA, LMBRD1, MAN2B1, MCCC1, MCCC2, MMAA, MMAB, MMACHC, MMADHC, MOCS1, MTHFR, MTR, MTRR, MUT, NAGS, NPC1, NPC2, OTC, OXCT1, PAH, PANK2, PCBD1, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, PHGDH, PNPO, PPM1K, PSAT1, PSPH, PTS, QDPR, SGSH, SLC19A3, SLC25A13, SLC25A15, SLC2A1, SLC6A8, SPR, TAT, TH, ABAT, ADSL, AP1S1, ATP13A2, BCKDK, C19orf12, CLN3, CLN5, CLN6, CLN8, COASY, CTSD, D2HGDH, DBH, DCAF17, DDC, FA2H, FTL, GAD1, GNS, GPHN, HEXA, HEXB, HGSNAT, HPRT1, IDH2, KCTD7, L2HGDH, MAOA, MFSD8, MOCOS, NAGLU, PLA2G6, POLG, PPT1, SLC13A5, SLC33A1, SLC6A3, SUOX, WDR45, XDH	Metabolic/ Mitochondrial	Metabolic Disorders
D5050	PAH Gene Sequencing	1	PAH	Metabolic/ Mitochondrial	Metabolic Disorders
D4700	Hereditary Hemochroma-tosis Panel	5	HAMP, HFE, HFE2, SLC40A1, TFR2	Metabolic/ Mitochondrial	Metabolic Disorders
D4604	ATP7A-Related Disorders	1	ATP7A	Metabolic/ Mitochondrial	Metabolic Disorders
D4605	Wilson Disease Test	1	ATP7B	Metabolic/ Mitochondrial	Metabolic Disorders

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TEST CODE	TEST NAME	NO. OF GENES	GENES INVOLVED	CATEGORY	DESCRIPTION
D4606	Comprehensive Mitochondrial Nuclear Gene Panel	254	AARS, AARS2, ABCB11, ABCB4, ABCB7, ABCD4, ACAD9, ACADM, ACADVL, ACO2, ACSF3, ADCK3 (CABC1:COQ8), ADCK4, AFG3L2, AGK, AGL, AIFM1, ALAS2, ALDOA, ALDOB, ALG1, ALG11, ALG13, ALG2, ALG3, ALG6, ALG9, AMACR, APOPT1, APTX, ARG1, ASL, ASS1, ATP5A1, ATP5E, ATP7B, ATP8B1, ATPAF2 (ATP12), AUH, B4GALT1, BCKDHA, BCKDHB, BCS1L, BOLA3, C10ORF2, C12ORF65, C19orf12, CA5A, CARS2, CHKB, CISD2, CLPB, COA5 (C2ORF64), COA6, COASY, COG4, COG5, COG6, COG7, COG8, COQ2, COQ4, COQ6, COQ9, COX10, COX14 (C12ORF62), COX15, COX20 (FAM36A), COX4I2, COX6A1, COX6B1, COX7B, CPS1, CPT1A, CPT2, CYC1, DARS, DARS2, DBT, DDHD1, DDHD2, DDOST, DGUOK, DLAT, DLD, DMGDH, DNA2, DNAJC19, DNM1L, DNM2, DOLK, DPAGT1, DPM1, DPM3, EARS2, ECHS1, ELAC2, ENO3, ETFA, ETFB, ETFDH, ETHE1, FAH, FARS2, FASTKD2, FBP1, FBXL4, FDX1L, FH, FLAD1, FOXRED1, G6PC, GAA, GAMT, GARS, GATM, GBE1, GCDH, GFER, GFM1 (EFG1), GFM2, GLRX5, GMPPA, GSS, GTPBP3, GYG1, GYG2, GYS1, GYS2, HADHA, HADHB, HARS2, HCFC1, HIBCH, HLCS, HMGCL, HMGCS2, HSD17B10, HSPD1, IARS2, IBA57, ISCA2, ISCU, IVD, LAMP2, LARS, LARS2, LDHA, LIAS, LIPT1, LMBRD1, LRPPRC, LYRM4, LYRM7, MARS, MARS2, MCCC1, MCCC2, MCEE, MFF, MFN2, MGAT2, MGME1, MICU1, MLYCD, MMAA, MMAB, MMACHC, MMADHC (C2ORF25), MOGS, MPC1 (BRP44L), MPDU1, MPI, MPV17, MRPL12, MRPL3, MRPL44, MRPS16, MRPS22, MRPS7, MTFMT, MTO1, MTPAP, MTR, MTRR, MUT, NADK2, NAGS, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA4, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3 (C3ORF60), NDUFAF4 (C6ORF66), NDUFAF5, NDUFAF6, NDUFAF7 (C2ORF56), NDUFB3, NDUFB9, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFS1, NFU1, NGLY1, NR2F1, NUBPL, OPA1, OPA3, OTC, PARS2, PC, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PET100, PFKM, PGAM2, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PMM2, PNPT1, POLG, POLG2, PRKAG2, PRPS1, PTRH2, PUS1, PYGM, QARS, RANBP2, RARS, RARS2, REEP1 (C2ORF23)	Metabolic/ Mitochondrial	Mitochondrial Disorders
D4607	Mitochondrial Encephalopathy/ Leigh Syndrome Nuclear Gene Panel	146	AARS2, ACAD9, ACO2, ADCK3 (CABC1:COQ8), AFG3L2, AIFM1, APOPT1, APTX, ATP5A1, ATP5E, ATPAF2 (ATP12), AUH, BCS1L, BOLA3, C10ORF2, C12ORF65, CA5A, COG8, COQ2, COQ4, COQ6, COQ9, COX10, COX14 (C12ORF62), COX15, COX20 (FAM36A), COX6B1, CPT1A, CPT2, CYC1, DARS, DARS2, DGUOK, DLAT, DLD, DNM1L, EARS2, ECHS1, ETFDH, ETHE1, FARS2, FASTKD2, FBP1, FBXL4, FH, FOXRED1, GCDH, GFER, GFM1 (EFG1), GFM2, GTPBP3, GYG2, HIBCH, HLCS, HSPD1, IARS2, IBA57, ISCA2, LARS2, LIAS, LIPT1, LRPPRC, LYRM7, MARS2, MFF, MFN2, MPC1 (BRP44L), MPV17, MRPL44, MRPS22, MTFMT, MTPAP, NADK2, NARS2, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA2, NDUFA4, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3 (C3ORF60), NDUFAF4 (C6ORF66), NDUFAF5, NDUFAF6, NDUFAF7 (C2ORF56), NDUFB3, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NFU1, NUBPL, PC, PCCA, PCCB, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PET100, PNPT1, POLG, RANBP2, RARS2, RMND1, RRM2B, SCO1, SCO2, SDHA, SDHAF1, SERAC1, SLC19A3, SLC22A5, SLC25A1, SLC25A15, SLC25A19, SLC25A22, SLC35A2, STXBP1, SUCLA2, SUCLG1, SURF1, TACO1, TARS2, TK2, TMEM70, TPK1, TRMU, TSFM, TTC19, TUFM, TYMP, UQCC2, UQCC3, UQCRQ, VARS2	Metabolic/ Mitochondrial	Mitochondrial Disorders
D4608	Zellweger Syndrome Panel	12	PEX1, PEX2, PEX3, PEX5, PEX6, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX26	Metabolic/ Mitochondrial	Peroxisomal Disorders
D4609	Adult Refsum Disease Panel	2	PEX7, PHYH	Metabolic/ Mitochondrial	Peroxisomal Disorders
D4610	Rhizomelic Chondrodys-plasia Punctata Spectrum Panel	3	AGPS, GNPAT, PEX7	Metabolic/ Mitochondrial	Peroxisomal Disorders

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D4611	Zellweger Spectrum Disorder Panel	15	ACOX1, AMACR, HSD17B4, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6	Metabolic/ Mitochondrial	Peroxisomal Disorders
D4612	Polycystic Kidney Panel	5	BICC1, PKD1, PKD2, NOTCH2, PKHD1	Metabolic/ Mitochondrial	Renal Disorders
D4613	Nephrotic Syndrome Panel	37	ACTN4, ANLN, APOL1, ARHGAP24, ARHGDIA, CD2AP, CFH, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CUBN, DGKE, EMP2, INF2, ITGA3, ITGB4, KANK4, LAMB2, LMX1B, MEFV, MYH9, MYO1E, NEIL1, NPHS1, NPHS2, PDSS2, PLCE1, PTPRO, SCARB2, SMARCAL1, TRPC6, TTC21B, WT1	Metabolic/ Mitochondrial	Renal Disorders

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TEST CODE	TEST NAME	NO. OF GENES	GENES INVOLVED	CATEGORY	DESCRIPTION
D4700	Hereditary Hemochroma-tosis Panel	5	HAMP, HFE, HFE2, SLC40A1, TFR2	Other	Blood Disorders
D4701	Hereditary Thrombophilia Panel	7	F2, F5, PROC, PROS1, SERPINC1, F9, MPL	Other	Blood Disorders
D4702	Pulmonary Fibrosis and Hermansky-Pudlak Syndrome Panel	16	ABCA3, AP3B1, BLOC1S3, BLOC1S6, CSF2RA, DTNBP1, ELMOD2, HPS1, HPS3, HPS4, HPS5, HPS6, SFTPB, SFTPC, SFTPD, TERT	Other	Blood Disorders
D4703	Ciliopathies Panel	102	AHI1, ANKS6, ARL13B, ARL6, ARMC4, B9D1, B9D2, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C21orf59, C5orf42, CC2D2A, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CEP104, CEP120, CEP164, CEP290, CEP41, CEP83, CSPP1, DCDC2, DNAAF1 , DNAAF2, DNAAF3, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAI1, DNAI2, DNAL1, DRC1, DYNC2H1, DYX1C1, EVC, EVC2, GAS8, GLIS2, IFT122, IFT140, IFT172, IFT80, INPP5E, INVS, IQCB1, KIAA0586, KIF7, LRRC6, MCIDAS, MKKS, MKS1, MRE11A, NEK1, NEK8, NME8, NPHP1, NPHP3, NPHP4, OFD1, PDE6D, PKD2, PKHD1, RPGR, RPGRIP1L, RSPH1, RSPH3, RSPH4A, RSPH9, SDCCAG8, SPAG1, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TRIM32, TTC21B, TTC8, WDPCP, WDR19, WDR34, WDR35, WDR60, XPNPEP3, ZMYND10, ZNF423	Other	Ciliopathies
D4704	Bardet Biedl Panel	21	ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CCDC28B, CEP290, IFT27, LZTFL1, MKKS, MKS1, SDCCAG8, TMEM67, TRIM32, TTC8, WDPCP	Other	Ciliopathies
D4705	Heterotaxy Panel	11	ACVR2B, CFAP53, CFC1, CRELD1, FOXH1, GDF1, LEFTY2, MMP21, NKX2-5, NODAL, ZIC3	Other	Ciliopathies
D4706	Joubert and Meckel-Gruber Syndromes Panel	30	AHI1, ARL13B, B9D1, B9D2, C5orf42, CC2D2A, CEP104, CEP290, CEP41, CSPP1, INPP5E, KIAA0586, KIF7, MKS1, MRE11A, NPHP1, NPHP3, OFD1, PDE6D, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF4	Other	Ciliopathies
D4707	Cornelia de Lange Syndrome Panel	8	AFF4, HDAC8, KMT2A, NIPBL, RAD21, SMC1A, SMC3, TAF6	Other	Congenital Abnormalities Syndrome
D4708	Noonan and RASopathies Panel	19	A2ML1, ACTB, ACTG1, BRAF, CBL, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, RIT1, SHOC2, SOS1, SOS2, SPRED1	Other	Congenital Abnormalities Syndrome
D4709	Prenatal Noonan Spectrum Disorders Panel	11	BRAF, HRAS, KRAS, MAP2K1, MAP2K2, NRAS, PTPN11, RAF1, RIT1, SHOC2, SOS1	Other	Congenital Abnormalities Syndrome

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D4710	Overgrowth and Macrocephaly Syndromes Panel	25	AKT2, AKT3, CDKN1C, CUL4B, DIS3L2, DNMT3A, EZH2, GLI3, GPC3, KPTN, MED12, NF1, NFIX, NPR2, NSD1, PHF6, PIK3R2, PTEN, SETD2, SPRED1, DICER1, EED, PDGFRB, RNF125, UPF3B	Other	Congenital Abnormalities Syndrome
D4711	Treachers Collins Syndrome Sequencing Panel	3	TCOF1, POLR1C, POLR1D	Other	Congenital Abnormalities Syndrome
D4712	Hyper-IgE Syndromes Panel	4	DOCK8, PGM3, SPINK5, STAT3	Other	Immuno Deficiency
D4713	B-Negative SCID Panel	9	ADA, AK2, DCLRE1C (ARTEMIS), LIG4, NHEJ1, PRKDC, RAC2, RAG1, RAG2	Other	Immuno Deficiency
D4714	B-Positive SCID Panel	17	ATM, CD3D, CD3E, CD3Z, CORO1A, DOCK8, FOXN1, IL2RG, IL7R, JAK3, ORAI1, PNP, PTPRC, RMRP, STIM1, TBX1, ZAP70	Other	Immuno Deficiency
D3006	SCID Gene Sequencing-Panel	26	ADA, AK2, ATM, CD3D, CD3E, CD3Z, CORO1A, DCLRE1C (ARTEMIS), DOCK8, FOXN1, IL2RG, IL7R, JAK3, LIG4, NHEJ1, ORAI1, PNP, PRKDC, PTPRC, RAC2, RAG1, RAG2, RMRP, STIM1, TBX1, ZAP70	Other	Immuno Deficiency
D4716	Common Variable Immune Deficiency (CVID) Panel	4	ICOS, NFKB2, TNFRSF13B, TNFRSF13C	Other	Immuno Deficiency
D4717	Mendelian Susceptibility to Mycobacterial Disease Panel	16	ACP5, ADAR, CYBB, GATA2, IFNGR1, IFNGR2, IL12B, IL12RB1, IRAK4, IRF8, ISG15, MYD88, SAMHD1, STAT1, STAT2, TYK2	Other	Immuno Deficiency
D4718	Premature Ovarian Failure: Sequencing Panel and FMR1 CGG Repeat Analysis	21	BMP15, CYP17A1, CYP19A1, DIAPH2, EIF2B2, EIF2B3, EIF2B5, FIGLA, FMR1, FOXL2, FSHR, GALT, GDF9, HFM1, LHCGR, LMNA, NOBOX, NR5A1, POF1B, POR, PSMC3IP	Other	Infertility
D4719	Female Infertility Panel	7	BMP15, CYP21A2, FSHR, LHB, LHCGR, TUBB8, ZP1	Other	Infertility
D4720	Male Infertility Panel	5	AR, CATSPER1, CFTR, FSHR, LHCGR	Other	Infertility
D4310	Alport Syndrome Panel	3	COL4A3, COL4A4, COL4A5	Other	Renal Disorders

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D4722	Nephrotic Syndrome Panel	37	ACTN4, ANLN, APOL1, ARHGAP24, ARHGDIA, CD2AP, CFH, COL4A3, COL4A4, COL4A5, COQ2, COQ6, COQ8B, CRB2, CUBN, DGKE, EMP2, INF2, ITGA3, ITGB4, KANK4, LAMB2, LMX1B, MEFV, MYH9, MYO1E, NEIL1, NPHS1, NPHS2, PDSS2, PLCE1, PTPRO, SCARB2, SMARCAL1, TRPC6, TTC21B, WT1.	Other	Renal Disorders
D4723	Cystinosis Test	1	CTNS	Other	Renal Disorders
D4724	Skeletal Dysplasia With Increased Bone Density: Sequencing Panel	22	ANKH, CA2, CLCN7, COL1A1, CTSK, DLX3, FERMT3, GALNT3, HPGD, LEMD3, LRP4, LRP5, OSTM1, RASGRP2, SOST, TBXAS1, TCIRG1, TGFB1, TNFRSF11A, TNFRSF11B, TNFSF11, TYROBP	Other	Skeletal Dysplasia
D4725	Limb Malformation: Sequencing Panel	46	ARHGAP31, BMP2, BMPR1B, CC2D2A, CDH3, CEP290, CHSY1, ESCO2, FBLN1, FBXW4, FGF10, FGFR2, FGFR3, FMN1, GDF5, GLI3, GNAS, GREM1, HDAC4, HOXD13, IHH, KIF7, LMBR1, LRP4, MGP, MKS1, MYCN, NIPBL, NOG, PIGV, PITX1, PTHLH, RECQL4, ROR2, RPGRIP1L, SALL1, SALL4, SHH, SOX9, TBX15, TBX3, TBX5, THPO, TP63, WNT3, WNT7A	Other	Skeletal Dysplasia
D4726	Cornelia de Lange Syndrome Panel	8	AFF4, HDAC8, KMT2A, NIPBL, RAD21, SMC1A, SMC3, TAF6	Other	Skeletal Dysplasia
D4610	Rhizomelic Chondrodys-plasia Punctata Spectrum Panel	3	AGPS, GNPAT, PEX7	Other	Skeletal Dysplasia
D4729	Periodic Fever Syndromes Panel	12	ADA2, ELANE, LPIN2, MEFV, MVK, NLRC4, NLRP12, NLRP3, PSMB8, PSTPIP1, TNFRSF1A, TRNT1	Other	Others
D4730	Familial Mediterranean Fever Panel	12	MEFV, ADA2, ELANE, LPIN2, MVK, NLRC4, NLRP12, NLRP3, PSMB8, PSTPIP1, TNFRSF1A, TRNT1	Other	Others
D4731	Obesity Panel	35	ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP290, CUL4B, DYRK1B, GNAS, IFT27, LEP, LEPR, LZTFL1, MAGEL2, MC4R, MKKS, MKS1, NR0B2, NTRK2, PCSK1, PHF6, POMC, SDCCAG8, SIM1, TRIM32, TTC8, UCP3, VPS13B, WDPCP	Other	Others
D4732	Pulmonary Disease: Comprehensive Sequencing Panel	52	ABCA3, ACVRL1, AP3B1, ASCL1, BDNF, BLOC1S3, BLOC1S6, BMPR2, CCDC39, CCDC40, CFTR, CSF2RA, DNAAF1, DNAAF2, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DOCK8, DTNBP1, EDN3, EFEMP2, ELMOD2, ELN, ENG, FBLN5, FLCN, GDNF, HPS1, HPS3, HPS4, HPS5, HPS6, LTBP4, NME8, PHOX2B, RET, RSPH4A, RSPH9, SCNN1A, SCNN1B, SCNN1G, SERPINA1, SFTPB, SFTPC, SFTPD, SMAD9, STAT3, TERT, TSC1, TSC2	Other	Others
D4733	MODY Panel	16	ABCC8, BLK, CEL, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, NKX2-2, PAX4, PDX1, RFX6, ZFP57	Other	Others

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D4734	Hereditary Hemorrhagic Telangiectasia Panel	7	ACVRL1, ADAM17, ENG, GDF2, PTPN14, RASA1, SMAD4	Other	Others
D4735	Cerebral Cavernous Malformations Panel	3	CCM2, KRIT1, PDCD10	Other	Others
D5016	CFTR Gene Sequencing	1	CFTR	Other	Others