

NGS PANELS

Next Generation Sequencing Tests

Farabi Medical Laboratory, established in Erbil, Iraq in 2012, is a premier private laboratory specializing in molecular diagnostics. We serve patients at all stages of life, from prenatal diagnosis to elderly care, and are committed to staying at the forefront of advanced laboratory science.

This catalog presents our comprehensive Next-Generation Sequencing (NGS) test panels, organized by category. Each panel includes the genes tested, enabling clinicians to select the right panel for their diagnostic needs.

Dermatology Test Panels

Adams-Oliver Syndrome Panel

ARHGAP31, DLL4, DOCK6, EOGT, KCTD1, NOTCH1, RBPJ, UBR1

Albinism Panel

AP3B1, BLOC1S3, BLOC1S6, C10ORF11, DTNBP1, EDN3, EDNRB, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, MC1R, MITF, MLPH, MYO5A, OCA2, PAX3, RAB27A, SLC24A5, SLC38A8, SLC45A2, TYR, TYRP1

Cutis Laxa Panel

ALDH18A1, ATP6V0A2, ATP7A, EFEMP2, ELN, FBLN5, GORAB, LTBP4, PYCR1, SLC2A10

Dyskeratosis Congenita Panel

ACD, AK2, CTC1, DKC1, NAF1, NHP2, NOP10, PARN, POT1, RTEL1, TERC, TERT, TINF2, USB1, WRAP53

Ectodermal Dysplasia Panel

BCS1L, CDH3, DSP, EDA, EDAR, EDARADD, ERCC2, EVC, EVC2, GJB2, GJB6, HOXC13, HR, IFT122, JUP, LRP6, MPLKIP, PAX9, PORCN, PRKD1, PVRL4, RMRP, TP63, WDR35, WNT10A

Ehlers-Danlos Syndrome Panel

ABCC6, ADAMTS2, ADAMTS12, AEBP1, ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, CHST3, COL11A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, GORAB, PLOD1, PYCR1, SLC39A13, SMAD3, TGFB2, TGFB1, TGFB2, ZNF469

Epidermolysis Bullosa Panel

ATP2C1, CDSN, COL17A1, COL7A1, CSTA, DSG1, DSG2, DSG4, DSP, DST, EXPH5, FERMT1, GRIP1, ITGA3, ITGA6, ITGB4, KLHL24, KRT1, KRT14, KRT5, LAMA3, LAMB3, LAMC2, PKP1, PLEC, TGM5

Hereditary Acrodermatitis Enteropathica Panel

SLC30A2, SLC39A4

Hereditary Melanoma and Skin Cancer Panel

BAP1, BRCA1, BRCA2, CDK4, CDKN2A, DDB2, ERCC2, ERCC3, ERCC4, ERCC5, MITF, POT1, PTCH1, PTEN, SUFU, TP53, WRN, XPA, XPC

Hermansky-Pudlak Syndrome Panel

ABCA3, AP3B1, AP3D1, BLOC1S3, BLOC1S6, DKC1, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, OCA2, SFTPB, SFTPC, SLC45A2, TERC, TERT, TINF2, TYR, TYRP1

Ichthyosis Panel

ABCA12, ABHD5, ALDH3A2, ALOX12B, ALOXE3, CASP14, CDSN, CERS3, CSTA, CYP4F22, EBP, ERCC2, FLG, GJA1, GJB2, GJB3, GJB4, KDSR, KRT1, KRT10, KRT2, KRT9, LIPN, LOR, MBTPS2, MPLKIP, NIPAL4, OSMR, PEXT7, PHYH, PNPLA1, SDR9C7, SLC27A4, SPINK5, ST14, STS, SUMF1, TGM1, TGM5

Neurofibromatosis Panel

KIT, KITLG, LZTR1, NF1, NF2, PTPN11, RAF1, SMARCB1, SPRED1

Pachyonychia Congenita Panel

AAGAB, KRT16, KRT17, KRT6A, KRT6B, KRT6C, TRPV3

Palmoplantar Keratoderma Panel

AAGAB, AQP5, CTSC, DSG1, DSP, ENPP1, GJB2, GJB4, GJB6, JUP, KRT1, KRT14, KRT16, KRT17, KRT6A, KRT6B, KRT6C, KRT9, LOR, MBTPS2, PKP1, SERPINB7, SLURP1, SMARCA1, TRPV3, WNT10A

Progeria and Progeroid Syndromes Panel

AGPAT2, ALDH18A1, B4GALT7, BLM, BSCL2, COL3A1, ERCC2, ERCC4, ERCC5, ERCC6, ERCC8, GORAB, LMNA, PYCR1, RECQL4, WRN, ZMPSTE24

Tuberous Sclerosis Panel

TSC1, TSC2

Waardenburg Syndrome Panel

EDN3, EDNRB, KIT, MITF, PAX3, SNAI2, SOX10

Xeroderma Pigmentosum Panel

DDB2, ERCC1, ERCC2, ERCC3, ERCC4, ERCC5, POLH, XPA, XPC

Immunology Test Panels

Severe Combined Immunodeficiency Panel ADA, AK2, ATM, BCL11B, BLM, CARD11, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD8A, CIITA, CORO1A, DCLRE1C, DNMT3B, DOCK8, EPG5, FOXN1, IFNGR1, IKBKB, IL12RB1, IL2RA, IL2RG, IL7R, IRF8, ITGB2, ITK, JAK3, LAT, LCK, LIG4, LRBA, MAGT1, MALT1, MAP3K14, MSN, NHEJ1, NSMCE3, ORAI1, PARN, PGM3, PIK3CD, PMS2, PNP, POLE, POLE2, PRKDC, PTPRC, RAC2, RAG1, RAG2, RFX5, RFXANK, RFXAP, RHOH, RMRP, RTEL1, SH2D1A, SMARCA1, SP110, SPINK5, STAT1, STAT2, STAT3, STAT5B, STIM1, STK4, TAP1, TAP2, TAPBP, TBX1, TFRC, TNFRSF4, TYK2, UNC119, WAS, ZAP70

Bone Marrow Failure Syndrome Panel ACD, ACTB, AK2, ALAS2, ANKRD26, AP3B1, ATM, ATR, BLM, BLOC1S3, BLOC1S6, BRAF, BRCA1, BRCA2, BRIP1, C15ORF41, C6ORF25, CBL, CDAN1, CDKN2A, CEBPA, CLPB, CSF2RA, CSF3R, CTC1, CTSC, CXCR4, DDX41, DKC1, DNAJC21, DNASE2, DTNBP1, EFL1, ELANE, EPCAM, ERCC4, ERCC6L2, ETV6, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, G6PC3, GATA1, GATA2, GFI1, GINS1, GLRX5, GP1BA, HAX1, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, IFNGR2, IKZF1, ITGA2B, ITK, JAGN1, KCNN4, KRAS, LAMTOR2, LYST, MAGT1, MAP2K1, MAP2K2, MECOM, MKL1, MLH1, MPL, MSH2, MSH6, MYH9, MYO5A, NAF1, NBN, NF1, NHP2, NOP10, NRAS, OBFC1, PALB2, PARN, PAX5, PGM3, PMS2, POT1, PRF1, PTPN11, RAB27A, RAC2, RAD51C, RBM8A, RECQL4, RIT1, RPL11, RPL15, RPL26, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SEC23B, SH2D1A, SLC19A2, SLC25A38, SLC37A4, SLX4, SMARCD2, SOS1, SRP54, SRP72, STX11, STXBP2, TBXAS1, TERC, TERT, THPO, TINF2, TP53, TUBB1, UBE2T, UNC13D, USB1, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, XIAP, XRCC2, ZCCHC8

Hemophagocytic Lymphohistiocytosis Panel FADD, FAS, FASLG, ITK, LYST, MAGT1, MYO5A, PRF1, RAB27A, RECQL4, SH2D1A, STX11, STXBP2, UNC13D, XIAP

Dyskeratosis Congenita Panel ACD, AK2, CTC1, DKC1, NAF1, NHP2, NOP10, PARN, POT1, RTEL1, TERC, TERT, TINF2, USB1, WRAP53

Complement System Disorder Panel ADIPOQ, ADIPOR1, ADIPOR2, ARMC4, C11ORF70, C1QA, C1QB, C1QBP, C1QC, C1S, C2, C3, C3AR1, C4BPA, C4BPB, C5, C5AR1, C5AR2, C6, C7, C8A, C8B, C8G, C9, CCDC103, CCDC114, CCDC39, CCDC40, CCDC65, CCNO, CD46, CD55, CD59, CD93, CFB, CFD, CFH, CFI, CFP, CLU, COLEC11, CR2, CRP, DGKE, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNALI1, DRC1, DYX1C1, FCN1, FCN2, FCN3, GAS2L2, HYDIN, LRRC6, MASP1, MASP2, MAT2A, MCIDAS, NME8, OFD1, PIGA, PTX3, RSPH1, RSPH4A, RSPH9, SERPING1, SPAG1, STK36, THBD, VSIG4, VTN, ZMYND10

Chronic Granulomatous Disease Panel C17ORF62, CYBA, CYBB, G6PD, NCF1, NCF2, NCF4, NOD2

Congenital Neutropenia Panel ACTB, CLPB, CSF2RA, CSF3R, CTSC, EFL1, ELANE, G6PC3, GATA2, GFI1, GINS1, HAX1, IFNGR2, JAGN1, LAMTOR2, LYST, MKL1, PGM3, RAC2, SBDS, SLC37A4, SMARCD2, SRP54, SRP72, VPS13B, VPS45, WAS, WDR1

Genetic Counseling

We are the first lab to offer medical genetics services in North Iraq and the only center that provides scientific genetic counseling by an experienced medical practitioner in collaboration with well-known genetic counseling clinics in the region. Genetic counseling is a service to help clients or their families who are at risk of developing a heritable disease and to translate scientific genetic knowledge into practical understandable information. In the counseling process, clients are informed about the nature of the risk, the probability of increasing the severity of the disease or transmitting it to future generations, and the options available to deal with the risk.

Immunology Test Panels

Primary Immunodeficiency Panel

ACD, ACP5, ACTB, ADA, ADAM17, ADAR, AICDA, AIRE, AK2, ALPI, AP3B1, AP3D1, ARHGEF1, ARPC1B, ATM, ATP6AP1, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BTK, C17ORF62, C1QA, C1QB, C1QC, C1S, C2, C3, C5, C6, C7, C8A, C8B, C9, CARD11, CARD14, CARD9, CASP10, CASP8, CCB1, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD4, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDC42, CDCA7, CDK9, CEBPE, CECR1, CFB, CFD, CFH, CFI, CFP, CFTR, CHD7, CIITA, CLCN7, CLPB, COG6, COLEC11, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR4, CYBA, CYBB, CYP27A1, DBR1, DCLRE1C, DDX58, DGAT1, DGKE, DIAPH1, DKC1, DNAJC21, DNASE1L3, DNASE2, DNMT3B, DOCK2, DOCK8, DSG1, EFL1, ELANE, EPG5, ERCC6L2, EXTL3, FADD, FANCA, FAS, FASLG, FAT4, FCGR3A, FCHO1, FERMT3, FOXN1, FOXP3, G6PC, G6PC3, G6PD, GATA2, GFI1, GINS1, GUCY2C, HAVCR2, HAX1, HELLS, HMOX1, HYOU1, ICOS, IFIH1, IFNAR2, IFNGR1, IFNGR2, IGLL1, IKBKB, IKZF1, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17RA, IL17RC, IL1RN, IL21, IL21R, IL23R, IL2RA, IL2RB, IL2RG, IL36RN, IL6R, IL6ST, IL7, IL7R, IRAK4, IRF2BP2, IRF4, IRF7, IRF8, ISG15, ITGB2, ITK, JAGN1, JAK1, JAK3, KRAS, LAMTOR2, LAT, LCK, LIG1, LIG4, LPIN2, LRBA, LYST, MAGT1, MALT1, MAP3K14, MASP1, MEFV, MKL1, MOGS, MRE11A, MSN, MTHFD1, MVK, MYD88, MYO5A, NBN, NCF1, NCF2, NCF4, NCSTN, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLR4, NLRP1, NLRP12, NLRP3, NOD2, NOP10, NRAS, NSMCE3, OBFC1, OFD1, ORAI1, OTULIN, PARN, PEPD, PGM3, PIGA, PIK3CD, PIK3R1, PLCG2, PMS2, PNP, POLA1, POLD1, POLE, POLE2, POMP, PRF1, PRG4, PRKCD, PRKDC, PSENEN, PSMB4, PSMB8, PSTPIP1, PTPRC, RAB27A, RAC2, RAG1, RAG2, RANBP2, RASGRP1, RBCK1, RECQL4, REL, RELB, RFX5, RFXANK, RFXAP, RHOH, RIPK1, RLTTPR, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RNU4ATAC, RORC, RPSA, RTEL1, SAMD9, SAMD9L, SAMHD1, SBDS, SEC61A1, SERPING1, SH2D1A, SLC29A3, SLC35C1, SLC37A4, SLC39A7, SLC46A1, SLC7A7, SMARCA1, SMARCD2, SP110, SPINK5, SPPL2A, SRP54, SRP72, STAT1, STAT2, STAT3, STAT5B, STIM1, STK4, STX11, STXBP2, TAP1, TAP2, TAPBP, TAZ, TBX1, TCF3, TCN2, TERC, TERT, TFRC, TGFB1, THBD, TNF2, TLR3, TMC6, TMC8, TMEM173, TNFAIP3, TNFRSF13B, TNFRSF1A, TNFRSF4, TNFRSF9, TRAF3IP2, TREX1, TRNT1, TTC37, TTC7A, TYK2, UBA1, UNC119, UNC13D, UNC93B1, UNG, USB1, USP18, VPS13B, VPS45, WAS, WDRI, WIPF1, WRAP53, XIAP, ZAP70, ZBTB24, ZNF341

Primary Immunodeficiency (PID) and Primary Ciliary Dyskinesia (PCD) Panel

ACD, ACP5, ACTB, ADA, ADAM17, ADAR, AICDA, AIRE, AK2, ALPI, AMN, AP3B1, AP3D1, ARHGEF1, ARMCA, ARPC1B, ATM, ATP6AP1, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BTK, C17ORF70, C1QA, C1QB, C1QC, C1S, C2, C21ORF59, C3, C5, C6, C7, C8A, C8B, C9, CARD11, CARD14, CARD9, CASP10, CASP8, CCB1, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD4, CD40, CD40LG, CD46, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDC42, CDCA7, CDK9, CEBPE, CECR1, CENPF, CFAP57, CFB, CFD, CFH, CFI, CFP, CFTR, CHD7, CIITA, CLCN7, CLPB, COG6, COLEC11, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR4, CYBA, CYBB, CYP27A1, DBR1, DCLRE1C, DDX58, DGAT1, DGKE, DIAPH1, DKC1, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNAJC21, DNAL1, DNASE1L3, DNASE2, DNMT3B, DOCK2, DOCK8, DRC1, DSG1, DYX1C1, EFL1, ELANE, EPG5, ERCC6L2, EXTL3, FADD, FANCA, FAS, FASLG, FAT4, FCGR3A, FCHO1, FERMT3, FOXN1, FOXP3, G6PC, G6PC3, G6PD, GAS2L2, GAS8, GATA2, GFI1, GINS1, GUCY2C, HAVCR2, HAX1, HELLS, HMOX1, HYDIN, HYOU1, ICOS, IFIH1, IFNAR2, IFNGR1, IFNGR2, IGLL1, IKBKB, IKZF1, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17RA, IL17RC, IL1RN, IL21, IL21R, IL23R, IL2RA, IL2RB, IL2RG, IL36RN, IL6R, IL6ST, IL7, IL7R, INVS, IRAK4, IRF2BP2, IRF4, IRF7, IRF8, ISG15, ITGB2, ITK, JAGN1, JAK1, JAK3, KRAS, LAMTOR2, LAT, LCK, LIG1, LIG4, LPIN2, LRBA, LRRC56, LRRC6, LYST, MAGT1, MALT1, MAP3K14, MASP1, MCIDAS, MCM4, MEFV, MKL1, MOGS, MRE11A, MSN, MTHFD1, MVK, MYD88, MYO5A, NBN, NCF1, NCF2, NCF4, NCSTN, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLR4, NLRP1, NLRP12, NLRP3, NME8, NOD2, NOP10, NRAS, NSMCE3, OBFC1, OFD1, ORAI1, OTULIN, PARN, PEPD, PGM3, PIGA, PIH1D3, PIK3CD, PIK3R1, PLCG2, PMS2, PNP, POLA1, POLD1, POLE, POLE2, POMP, PRF1, PRG4, PRKCD, PRKDC, PSENEN, PSMB4, PSMB8, PSTPIP1, PTPRC, RAB27A, RAC2, RAG1, RAG2, RANBP2, RASGRP1, RBCK1, RECQL4, REL, RELB, RFX5, RFXANK, RFXAP, RHOH, RIPK1, RLTTPR, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RNU4ATAC, RORC, RPGR, RPSA, RSPH1, RSPH3, RSPH4A, RSPH9, RTEL1, SAMD9, SAMD9L, SAMHD1, SBDS, SEC61A1, SERPING1, SH2D1A, SLC29A3, SLC35C1, SLC37A4, SLC39A7, SLC46A1, SLC7A7, SMARCA1, SMARCD2, SP110, SPAG1, SPINK5, SPPL2A, SRP54, SRP72, STAT1, STAT2, STAT3, STAT5B, STIM1, STK36, STK4, STX11, STXBP2, TAP1, TAP2, TAPBP, TAZ, TBX1, TCF3, TCN2, TERC, TERT, TFRC, TGFB1, THBD, TNF2, TLR3, TMC6, TMC8, TMEM173, TNFAIP3, TNFRSF13B, TNFRSF1A, TNFRSF4, TNFRSF9, TRAF3IP2, TREX1, TRNT1, TTC25, TTC37, TTC7A, TYK2, UBA1, UNC119, UNC13D, UNC93B1, UNG, USB1, USP18, VPS13B, VPS45, WAS, WDRI, WIPF1, WRAP53, XIAP, ZAP70, ZBTB24, ZMYND10, ZNF341

Autoinflammatory Syndrome Panel

ACP5, ADAM17, ADAR, CARD14, CECR1, COPA, DDX58, DNASE1L3, DNASE2, ELANE, IFIH1, IL10, IL10RA, IL10RB, IL1RN, IL36RN, ISG15, JAK1, LPIN2, MEFV, MVK, NLR4, NLRP1, NLRP12, NLRP3, NOD2, OTULIN, PLCG2, POMP, PRG4, PSENEN, PSMB4, PSMB8, PSTPIP1, RIPK1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, SLC29A3, TMEM173, TNFAIP3, TNFRSF1A, TREX1, TRNT1, TTC7A, UBA1

Malformations Panel

Limb Malformations Panel ARHGAP31, ARID1A, ARID1B, ATR, BHLHA9, BRCA2, BRIP1, DHODH, DLL4, DLX5, DOCK6, EOGT, ERCC4, ESCO2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FGF10, GDF5, HDAC8, KYNU, NIPBL, NOTCH1, NSDHL, PALB2, PITX1, PUF60, RAD21, RAD51C, RBM8A, RBPJ, RECQL4, SALL1, SALL4, SF3B4, SLX4, SMC1A, SMC3, TBX3, TBX5, TP63, WNT7A, XRCC2

Exostosis and Related Disorders Panel EXT1, EXT2, PTPN11

Facial Dysostosis and Related Disorders Panel ALPL, ALX3, ALX4, CREBBP, DHODH, DLL3, EFN1, EFTUD2, EHMT1, EP300, EVC, EVC2, HDAC8, HSPG2, LIFR, MYCN, NIPBL, POLR1A, POLR1C, POLR1D, SF3B4, SMC1A, SMC3, SRCAP, TCOF1, TWIST1, UBE2A

Gastrointestinal Atresia Panel CDK9, CHD7, CLMP, DHCR7, EFTUD2, FANCB, FANCC, GLI3, MID1, MNX1, MYCN, PTF1A, RFX6, SOX2, TTC7A

Heterotaxy and Situs Inversus Panel ACVR2B, ANKS6, ARMC4, C21ORF59, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CFAP53, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNAL1, DDX1, COG1, FOXH1, GDF1, INVS, LEFTY2, LRRC6, MMP21, NODAL, PIH1D3, PITRM1, PKD1L1, SPAG1, TTC25, ZIC3, ZMYND10

Comprehensive Skeletal Dysplasias and Disorders Panel

ACAN, ACP5, ACVR1, ADAMTS10, ADAMTS17, ADAMTSL2, AGA, AGPS, AIFM1, AKT1, ALPL, ALX1, ALX3, ALX4, AMER1, ANKH, ANKRD11, ANO5, ANTXR2, ARCN1, ARHGAP31, ARID1B, ARSB, ARSE, ATP6V0A2, ATR, B3GALT6, B3GALT3, B4GALT7, BGN, BHLHA9, BMP1, BMP2, BMPER, BMPR1B, C21ORF2, C2CD3, CA2, CANT1, CASR, CC2D2A, CDC45, CDC6, CDH3, CDKN1C, CDT1, CENPE, CEP120, CEP152, CEP290, CHST14, CHST3, CHSY1, CKAP2L, CLCN5, CLCN7, COG1, COG4, COL10A1, COL11A1, COL11A2, COL1A1, COL1A2, COL27A1, COL2A1, COL9A1, COL9A2, COL9A3, COMP, CREB3L1, CREBBP, CRIP1, CRLF1, CRTAP, CSF1R, CSPP1, CTSA, CTSK, CUL7, CYP27B1, CYP2R1, DDR2, DDX58, DHCR24, DHODH, DLL3, DLL4, DLX3, DLX5, DMP1, DNAJC21, DNMT3A, DOCK6, DONSON, DSE, DVL1, DVL3, DYM, DYNC2H1, DYNC2L1, EBP, EDN1, EDNRA, EFL1, EFN1, EFTUD2, EIF2AK3, EIF4A3, ENAM, ENPP1, EOGT, EP300, ERF, ESCO2, EVC, EVC2, EXT1, EXT2, EXT3, EZH2, FAM111A, FAM20A, FAM20C, FAM46A, FAM83H, FANCB, FANCC, FBN1, FBN2, FERMT3, FGF10, FGF23, FGF9, FGF11, FGF12, FGF13, FGF14, FGF15, FGF16, FGF17, FGF18, FGF19, FGF20, FGF21, FGF22, FGF23, FGF24, FGF25, FGF26, FGF27, FGF28, FGF29, FGF30, FGF31, FGF32, FGF33, FGF34, FGF35, FGF36, FGF37, FGF38, FGF39, FGF40, FGF41, FGF42, FGF43, FGF44, FGF45, FGF46, FGF47, FGF48, FGF49, FGF50, FGF51, FGF52, FGF53, FGF54, FGF55, FGF56, FGF57, FGF58, FGF59, FGF60, FGF61, FGF62, FGF63, FGF64, FGF65, FGF66, FGF67, FGF68, FGF69, FGF70, FGF71, FGF72, FGF73, FGF74, FGF75, FGF76, FGF77, FGF78, FGF79, FGF80, FGF81, FGF82, FGF83, FGF84, FGF85, FGF86, FGF87, FGF88, FGF89, FGF90, FGF91, FGF92, FGF93, FGF94, FGF95, FGF96, FGF97, FGF98, FGF99, FGF100, FGF101, FGF102, FGF103, FGF104, FGF105, FGF106, FGF107, FGF108, FGF109, FGF110, FGF111, FGF112, FGF113, FGF114, FGF115, FGF116, FGF117, FGF118, FGF119, FGF120, FGF121, FGF122, FGF123, FGF124, FGF125, FGF126, FGF127, FGF128, FGF129, FGF130, FGF131, FGF132, FGF133, FGF134, FGF135, FGF136, FGF137, FGF138, FGF139, FGF140, FGF141, FGF142, FGF143, FGF144, FGF145, FGF146, FGF147, FGF148, FGF149, FGF150, FGF151, FGF152, FGF153, 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Malformations Panel

Holoprosencephaly Panel

CDON, FGF8, FGFR1, FOXH1, GLI2, GLI3, NODAL, PTCH1, SHH, SIX3, TGIF1, ZIC2

Kabuki Syndrome Panel

CHD7, EYA1, FLNB, HNRNP, IRF6, KDM1A, KDM6A, KMT2D, SIX5

Comprehensive Short Stature Syndrome Panel

ACTB, ACTG1, AMMECR1, ARCN1, ATR, B3GAT3, BCS1L, BLM, BMP2, BRAF, CBL, CCDC8, CDC45, CDC6, CDT1, CENPJ, CEP152, CEP63, COL27A1, CREBBP, CUL7, DHCR7, DONSON, EP300, FGD1, FGFR3, FN1, GH1, GHR, GHRHR, GHRSR, GLI2, GNAS, HDAC8, HESX1, HRAS, IDUA, IGF1, IGF1R, IGFALS, INSR, IRS1, KRAS, LARP7, LFNG, LHX3, LHX4, LZTR1, MAP2K1, MAP2K2, NIPBL, NOTCH2, NRAS, OBSL1, ORC1, ORC4, ORC6, OSGEP, OTX2, PCNT, PISD, PITX2, POC1A, POP1, POU1F1, PPP3CA, PRMT7, PROP1, PTPN11, PUF60, RAD21, RAF1, RALA, RASA2, RBBP8, RIT1, RNU4ATAC, RRAS, RTTN, SGMS2, SHOC2, SHOX, SMARCA2, SMARCE1, SMC1A, SMC3, SOS1, SOX11, SOX2, SOX3, SRCAP, STAT5B, TALDO1, TBX19, TBX2, TBX3, TOP3A, TRIM37, TRMT10A, XRCC4

Arthrogryposes Panel

ACTA1, ADGRG6, AGRN, BIN1, CACNA1E, CASK, CFL2, CHAT, CHRNA1, CHRNBI, CHRNA2, CHRN, CHST14, CHUK, CNTNAP1, COL6A2, COLQ, DHCR24, DOK7, DPAGT1, ECEL1, EGR2, ERBB3, ERCC5, ERCC6, EXOSC3, FBN2, FHL1, FKBP10, FKTN, FLVCR2, GBA, GBE1, GFPT1, GLDN, GLE1, KAT6B, KIAA1109, KLHL40, LGI4, LMNA, MPZ, MTM1, MUSK, MYBPC1, MYH2, MYH3, MYH8, NALCN, NEB, PIEZO2, PLOD2, PMM2, PPP3CA, RAPSN, RARS2, RIPK4, SCO2, SELENON, SMN1, SMN2, TGFBI3, TK2, TNNI2, TNNT1, TNNT3, TPM2, TPM3, TRPV4, TSEN2, TSEN54, UBA1, VIPAS39, VPS33B, VRK1, ZBTB42, ZC4H2

Chondrodysplasia Punctata Panel

AGPS, ARSE, EBP, GNPAT, LBR, MGP, NSDHL, PEX14, PEX19, PEX7

Craniosynostosis Panel

ALPL, ALX3, ALX4, BMP4, CDC45, EDNRB, EFNBI, ERF, ESCO2, FGFR1, FGFR2, FGFR3, FLNB, FREM1, GDF5, GLI3, IFT122, IFT140, IL11RA, MASP1, MEGF8, MSX2, NOG, PAX3, POR, RAB23, RECQL4, SKI, SOX10, SPECC1L, TCF12, TGFBR1, TGFBR2, TWIST1, TWIST2, WDR19, WDR35, ZIC1

Lymphatic Malformations and Related Disorders Panel

ADAMTS3, CCBE1, FAT4, FLT4, FOXC2, GATA2, GJC2, KIF11, PIEZO1, PIK3CA, RASA1, SOX18

Lissencephaly Panel

ACTB, ACTG1, ARX, ATP6V0A2, CIT, CRADD, DCX, FKTN, ISPD, KATNB1, KIAA1109, LAMB1, LARGE, MACF1, PAFAH1B1, POMGNT2, POMT1, RELN, TMTC3, TUBA1A, TUBB2B, TUBG1, VLDLR, YWHAE

Macrocephaly / Overgrowth Syndrome Panel

AKT1, AKT3, ASPA, ASXL2, BRWD3, CCND2, CDKN1C, CHD8, CUL4B, DHCR24, DIS3L2, DNMT3A, EED, EIF2B5, EZH2, GFAP, GLI3, GPC3, GPM2, GRIA3, HEPACAM, HUWE1, KDM1A, KIAA0196, KIF7, KPTN, L1CAM, MED12, MLC1, MPDZ, NFIB, NFIX, NSD1, OFD1, PIGA, PIK3CA, PIK3R2, PTCH1, PTEN, RAB39B, RNF135, SETD2, SYN1, TMEM94, TSC1, TSC2, UPF3B, ZBTB20

Microcephaly and Pontocerebellar Hypoplasia Panel

AKT3, AMPD2, ASPM, ASXL1, ASXL3, ATR, CASK, CCDC47, CDK5RAP2, CENPF, CENPJ, CEP152, CEP63, CSNK2A1, DONSON, DYNC1H1, DYRK1A, EFTUD2, EXOSC3, GFM1, GPT2, KANSL1, KATNB1, KIF11, LGI4, MBD5, MCPH1, MED17, MFSD2A, MIPER, MRE11A, MYCN, MYO18B, NCAPD3, NDE1, NHEJ1, OPHN1, PAFAH1B1, PCDH12, PCLO, PCNT, PHGDH, PLK4, PNKP, POMT1, PQBP1, QARS, RARS2, RTTN, SEPSECS, SLC1A4, SMARCA2, SMARCE1, SOX11, STAG2, STAMBP, STIL, TBC1D20, TBC1D23, THOC6, TMTC3, TOE1, TOP3A, TRMT10A, TSEN2, TSEN54, TUBB, TUBB2B, TUBGCP4, TUBGCP6, UBE3B, VARS, VRK1, WDR62, WDR73, XRCC4, ZNF148, ZNF335

Malformations Panel

Metaphyseal Dysplasia Panel

ANKH, CDKN1C, COL10A1, FGFR3, FLNA, MMP13, MMP9, PISD, PTH1R, RMRP, RUNX2, SBDS

Micromelic Dysplasia Panel

ADAMTS10, ADAMTS12, BMP1B, DVL1, EXT1, FBN1, FGFR3, GDF5, GNAS, IFT122, IFT140, IHH, INPPL1, LIFR, LTBP2, NPR2, PRKARIA, ROR2, SHOX, SLC35D1, SMAD4, SOX9, TRIP11, TRPS1, WDR19, WDR35, WNT5A

Meier-Gorlin Syndrome Panel

CDC45, CDC6, CDT1, ORC1, ORC4, ORC6

Neurofibromatosis Panel

KIT, KITLG, LZTR1, NF1, NF2, PTPN11, RAF1, SMARCB1, SPRED1

Neuronal Migration Disorder Panel

ACTB, ACTG1, ADGRG1, AKT3, ARFGEF2, ARX, ATP6V0A2, B3GALNT2, COL4A1, COL4A2, DCX, DYNC1H1, EMX2, FAT4, FH, FKTN, FLNA, FLVCR2, GMPPB, GPSM2, ISPD, KATNB1, KIF1BP, KIF7, L1CAM, LAMA2, LAMB1, LAMC3, LARGE, MACF1, MED12, MEF2C, MPDZ, NDE1, NSDHL, OCLN, PAFAH1B1, PHGDH, PIK3CA, PIK3R2, POMGNT2, POMT1, RAB18, RAB3GAP1, RAB3GAP2, RELN, RTTN, SEPSECS, SRPX2, TMEM5, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBG1, VLDLR, WDR62, YWHAE

Osteogenesis Imperfecta Panel

ALPL, ARCN1, B3GAT3, B4GALT7, BMP1, CLCN5, COL1A1, COL1A2, CREB3L1, CRTAP, FAM46A, FGF23, FKBP10, IFITM5, LRP5, MBTPS2, P3H1, P4HB, PHEX, PLOD2, PLS3, PPIB, SEC24D, SERPINF1, SERPINH1, SGMS2, SLC29A3, SLC34A3, SP7, SPARC, TAPT1, TMEM38B, WNT1

Osteopetrosis and Dense Bone Dysplasia Panel

AMER1, ANKH, CA2, CLCN7, COL1A1, CTSK, DLX3, FAM20C, FERMT3, GJA1, LEMD3, LRP4, LRP5, OSTM1, PLEKHM1, PTDSS1, PTH1R, SLC29A3, SLC20A1, SNX10, SOST, TCIRG1, TGFB1, TNFRSF11A, TNFRSF11B, TNFSF11, TYROBP

Polymicrogyria Panel

ADGRG1, AKT3, FH, GPSM2, KIF1BP, LAMC3, NDE1, NSDHL, OCLN, PI4KA, RAB18, SNAP29, SRPX2, TBC1D20, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, WDR62

Seckel Syndrome Panel

ATR, CENPJ, CEP152, CEP63, PCNT, RBBP8

Septo-Optic Dysplasia Panel

HESX1, OTX2, PAX6, SOX2

Cerebral Cavernous Malformation Panel

CCM2, KRIT1, PDCD10, RASA1

Short Rib Dysplasia / Asphyxiating Thoracic Dysplasia Panel

CSPP1, DYNC2H1, EVC, EVC2, GLI2, IFT122, IFT140, IFT172, IFT52, IFT80, NEK1, TCTEX1D2, TCTN3, TTC21B, WDR19, WDR34, WDR35, WDR60

Malformations Panel

Spondylometaphyseal / Spondyloepi-(meta)-physeal Dysplasia Panel

ACAN, ACP5, B3GALT6, BGN, CANT1, CHST3, COL11A1, COL11A2, COL2A1, DDR2, DYM, EIF2AK3, FN1, HSPG2, INPPL1, KIF22, LONP1, MATN3, MMP13, NANS, NKX3-2, PAM16, PAPSS2, PCYT1A, PISD, POP1, RAB33B, RMRP, RSPRY1, SLC39A13, SMARCAL1, TONSL, TRAPPC2, TRPV4, UFSP2, WISP3

Vascular Malformations Panel

ACVRL1, CCM2, ELMO2, ENG, EPHB4, GDF2, GLMN, KRIT1, PDCD10, PIK3CA, PTEN, RASA1, SMAD4, SOX18, STAMBP, TEK

Cleft Lip/Palate and Associated Syndromes Panel

ARHGAP29, COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, CTNND1, FOXE1, GRHL3, IRF6, KDM6A, KMT2D, MSX1, SATB2, SPECC1L, TBX2, TBX22, TGDS, TP63, TXNL4A, ZSWIM6

Hirschsprung Disease Panel

BDNF, CELSR3, EDN3, EDNRB, KIF1BP, L1CAM, MITE, NRG1, NRTN, PAX3, PHOX2B, RET, RMRP, SOX10, ZEB2

Ophthalmology Test Panels

Achromatopsia Panel

ATF6, CNGA3, CNGB3, GNAT2, PDE6C, PDE6H, RGS9, RGS9BP

Albinism Panel

AP3B1, BLOC1S3, BLOC1S6, C10ORF11, DTNBP1, EDN3, EDNRB, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, MC1R, MITE, MLPH, MYO5A, OCA2, PAX3, RAB27A, SLC24A5, SLC38A8, SLC45A2, TYR, TYRP1

Bardet-Biedl Syndrome Panel

ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP19, CEP290, CPE, IFT172, IFT27, LZTFL1, MKKS, MKS1, PNPLA6, SCAPER, SDCCAG8, TMEM67, TRIM32, TTC8, WDPCP

Flecked Retina Disorders Panel

ABCA4, CHM, CYP4V2, ELOVL4, PLA2G5, PROM1, PRPH2, RDH5, RHO, RLBP1, RS1, VPS13B

Ectopia Lentis Panel

AASS, ADAMTS10, ADAMTS17, ADAMTSL4, ASPH, BCOR, CBS, COL18A1, FBN1, LTBP2, P3H2, PORCN, SUOX, VSX2

Glaucoma Panel

CNTNAP2, COL4A1, CYP1B1, FOXC1, FOXE3, LMX1B, LTBP2, MAE, MYOC, OPA1, OPA3, OPTN, PAX6, PITX2, PXDN, TBK1, TEK, TMEM126A, WDR36

Corneal Dystrophy Panel

CHRD1, CHST6, COL17A1, COL5A1, COL8A2, CYP4V2, DCN, FOXE3, GJA8, GRHL2, GSN, KERA, KRT12, KRT3, LCAT, LOXHD1, MAE, NLRP3, OVOL2, PIKFYVE, PITX2, PRDM5, SLC4A11, TACSTD2, TCF4, TGFBI, UBIAD1, ZEB1, ZNF469

Leber Congenital Amaurosis Panel

AIPL1, ALMS1, BBS4, CABP4, CEP290, CNGA3, CRB1, CRX, CWC27, DTHD1, GUCY2D, IDH3A, IMPDH1, IQCB1, KCNJ13, LCA5, LRAT, MERTK, MYO7A, NMNAT1, RD3, RDH12, RDH5, RPE65, RPGRIP1, SPATA7, TUBB4B, TULP1

Ophthalmology Test Panels

Joubert Syndrome Panel

AHI1, ARL13B, ARMC9, B9D1, B9D2, C21ORF2, C5ORF42, CC2D2A, CEP104, CEP120, CEP164, CEP290, CEP41, CSPP1, INPP5E, KIAA0556, KIAA0586, KIAA0753, KIF7, MKS1, NPHP1, NPHP3, OFD1, PDE6D, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423

Cataract Panel

ABCB6, ADAMTS18, ADAMTS14, AGK, ALDH18A1, BCOR, BFSP1, BFSP2, CHMP4B, COL11A1, COL18A1, COL2A1, COL4A1, CRYAA, CRYAB, CRYBA1, CRYBA4, CRYBB1, CRYBB2, CRYBB3, CRYGC, CRYGD, CRYGS, CTDPI, CYP27A1, DNMBP, EPHA2, ERCC2, ERCC5, ERCC6, ERCC8, EYA1, FAM126A, FOXE3, FTL, FYCO1, FZD4, GALE, GALK1, GALT, GCNT2, GJA1, GJA3, GJA8, HSF4, LEMD2, LIM2, MAE, MIR, MYH9, NDP, NF2, NHS, OCRL, OPA3, P3H2, PAX6, PITX3, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RECQL4, SIL1, SIPA1L3, SLC16A12, SLC33A1, TBC1D20, TDRD7, TFAP2A, TMEM70, VIM, VSX2, WFS1, WRN, XYLT2

Macular Dystrophy Panel

ABCA4, BEST1, C1QTNF5, CDH3, CERKL, CNGB3, CRB1, CRX, CTNNA1, DRAM2, EFEMP1, ELOVL4, IMPG1, IMPG2, KCNV2, MFSD8, NMNAT1, PRDM13, PROM1, PRPH2, RAX2, RDH12, RDH5, RLBP1, RP1L1, RPGR, RS1, TIMP3

Microphthalmia, Anophthalmia and Anterior Segment Dysgenesis Panel

ABCB6, ADAMTS18, ALDH1A3, BCOR, BMP4, BMP7, CDK9, CHD7, COL4A1, COX7B, CPAMD8, CYP1B1, ERCC2, ERCC5, ERCC6, FOXC1, FOXE3, FOXL2, FRAS1, FREM1, FREM2, GJA1, GRIP1, HCCS, HESX1, HMX1, MAB21L2, MFRP, MITE, NAA10, NDP, OCRL, OTX2, PAX2, PAX6, PITX2, PQBP1, PRSS56, PXDN, RAB18, RAB3GAP1, RAB3GAP2, RARB, RAX, RBP4, SHH, SIPA1L3, SIX3, SIX6, SLC38A8, SMCHD1, SMOC1, SOX2, STRA6, TBC1D20, TENM3, TFAP2A, VPS13B, VSX2, YAP1, ZIC2

Congenital Stationary Night Blindness Panel

CABP4, CACNA1F, CACNA2D4, CYP4V2, FRMD7, GNAT1, GNB3, GPR179, GRK1, GRM6, LRIT3, NYX, PDE6B, RDH5, RHO, RLBP1, RPE65, SAG, SLC24A1, TRPM1

Retinal Dystrophy Panel

ABCA4, ABCC6, 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, C21ORF2, C2ORF71, C5ORF42, C8ORF37, CA4, CABP4, CACNA1F, CACNA2D4, CAPN5, CC2D2A, CDH23, CDH3, CDHR1, CEP104, CEP120, CEP164, CEP19, CEP250, CEP290, CEP41, CEP78, CEP83, CERKL, CHM, CIB2, CISD2, CLN3, CLN5, CLN6, CLN8, CLRN1, CNGA1, CNGA3, CNGB1, CNGB3, CNM4, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, COQ2, CPE, CRB1, CRX, CSPP1, CTC1, CTNNA1, CTNNB1, CTSD, CWC27, CYP4V2, DFN3B1, DHDDS, DHX38, DNAJC5, DRAM2, DTHD1, DYNC2H1, EFEMP1, ELOVL4, EMC1, ESPN, EXOSC2, EYS, FAM161A, FDXR, FLVCR1, FRMD7, FZD4, GNAT1, GNAT2, GNB3, GNPTG, GPR143, GPR179, GRK1, GRM6, GUCA1A, GUCY2D, HARS, HGSNAT, HK1, HMX1, IDH3A, IDH3B, IFT140, IFT172, IFT27, IFT81, IMPDH1, IMPG1, IMPG2, INPP5E, INVS, IQCB1, ISPD, JAG1, KCNJ13, KCNV2, KIAA0556, KIAA0586, KIAA0753, KIAA1549, KIF11, KIF7, KIZ, KLHL7, LAMA1, LCA5, LRAT, LRIT3, LRP2, LRP5, LZTFL1, MAK, MERTK, MFN2, MFRP, MFSD8, MKKS, MKS1, MMACHC, MTTP, MVK, MYO7A, NAGLU, NDP, NEK2, NMNAT1, NPHP1, NPHP3, NPHP4, NR2E3, NR2F1, NRL, NYX, OAT, OCA2, OFD1, OPA1, OPA3, OPN1SW, OTX2, P3H2, PANK2, PAX2, PCDH15, PCYT1A, PDE6A, PDE6B, PDE6C, PDE6D, PDE6G, PDE6H, PDSS1, PDSS2, 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, PRPF2, PRPS1, RAB28, RAX2, RBP3, RBP4, RCBTB1, RD3, RDH11, RDH12, RDH5, REEP6, RGR, RGS9, RGS9BP, RHO, RIMS1, RLBP1, ROM1, RPI, 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, TPPI, TRAF3IP1, TREX1, TRIM32, TRPM1, TSPAN12, TTC21B, TTC8, TLLS, TTPA, TUB, TUBB4B, TUBGCP4, TUBGCP6, TULP1, TYR, TYRP1, USH1C, USH1G, USH2A, VCAN, VPS13B, WPCR, WDR19, WFS1, YME1L1, ZNF408, ZNF423, ZNF513

Ophthalmology Test Panels

Cone Rod Dystrophy Panel ABCA4, ADAM9, ADAMTS18, AIPL1, ARHGEF18, BEST1, C21ORF2, C8ORF37, CABP4, CACNA1F, CACNA2D4, CDHR1, CEP250, CEP78, CERKL, CLN3, CNGA3, CNGB3, CNNM4, CRB1, CRX, CYP4V2, ELOVL4, GNAT2, GUCA1A, GUCY2D, KCNV2, MERTK, PDE6C, PDE6H, PITPNM3, POC1B, PROM1, PRPH2, RAB28, RAX2, RDH5, RGS9, RGS9BP, RIMS1, RPGR, RPGRIP1, SEMA4A, TTLL5

Senior-Loken Syndrome Panel CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SCLT1, SDCCAG8, TRAF3IP1

Septo-Optic Dysplasia Panel HESX1, OTX2, PAX6, SOX2

Stickler Syndrome Panel COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, LRP2, VCAN

Usher Syndrome Panel ABHD12, ADGRV1, ARSG, CDH23, CEP250, CEP78, CIB2, CLRN1, DFN3B1, ESPN, HARS, MYO7A, PCDH15, PDZD7, PEX1, PEX6, PRPS1, TUBB4B, USH1C, USH1G, USH2A

Vitreoretinopathy Panel ATOH7, BEST1, CAPN5, COL11A1, COL11A2, COL18A1, COL2A1, COL9A1, COL9A2, COL9A3, CTC1, CTNNB1, FZD4, KCNJ13, KIF11, LRP5, NDP, NR2E3, P3H2, RCBTB1, RS1, TSPAN12, VCAN, ZNF408

Retinitis Pigmentosa Panel ABCA4, ABHD12, ADIPOR1, AGBL5, AHI1, AIPL1, ARHGEF18, ARL2BP, ARL3, ARL6, BBS1, BBS2, BEST1, C1QTNF5, C21ORF2, C2ORF71, C8ORF37, CA4, CACNA1F, CDHR1, CEP290, CERKL, CHM, CLN3, CLRN1, CNGA1, CNGB1, CNGB3, CRB1, CRX, CTNNA1, CWC27, CYP4V2, DHDDS, DHX38, DYNC2H1, EYS, FAM161A, FLVCR1, GNPTG, GUCY2D, HGSNAT, HK1, IDH3A, IDH3B, IFT140, IMPDH1, IMPG2, INPP5E, KIAA1549, KIZ, KLHL7, LCA5, LRAT, MAK, MERTK, MFRP, MVK, NEK2, NMNAT1, NR2E3, NRL, OAT, OFD1, PDE6A, PDE6B, PDE6G, PEX1, PEX2, PEX7, PHYH, PITPNM3, PLA2G5, POMGNT1, PRCD, PROM1, PRPF3, PRPF31, PRPF4, PRPF6, PRPF8, PRPH2, RBP3, RBP4, RCBTB1, RD3, RDH12, RDH5, REEP6, RGR, RHO, RIMS1, RLBP1, ROM1, RP1, RP1L1, RP2, RPE65, RPGR, RPGRIP1, RS1, SAG, SAMD11, SCAPER, SCLT1, SEMA4A, SLC7A14, SNRNP200, SPATA7, SPP2, TIMP3, TOPORS, TTC8, TTPA, TUB, TULP1, USH1C, USH2A, VPS13B, WDR19, ZNF408, ZNF513

Neuro-Ophthalmology Panel ACO2, AFG3L2, ANTXR1, APTX, ATAD3A, AUH, C10ORF2, C12ORF65, C19ORF12, CHN1, CISD2, DNAJC19, DNMI1L, FDXR, FRMD7, GPR143, HESX1, ISCA2, KIF21A, MEER, MFN2, MGME1, MTPAP, NARS2, NDUFAF3, NDUFS1, NR2F1, OPA1, OPA3, OTX2, PAX6, PDSS1, PHOX2A, POLG, PRPS1, ROBO3, RRM2B, RTN4IP1, SALL4, SETX, SLC19A2, SLC19A3, SLC25A4, SLC25A46, SLC38A8, SLC52A2, SNX10, SOX2, SPG7, SUCLA2, TIMM8A, TK2, TMEM126A, TSFM, TUBB3, TYMP, UCHL1, WFS1, YME1L1, ZNHIT3

Optic Atrophy Panel ACO2, AFG3L2, ATAD3A, AUH, C12ORF65, C19ORF12, CISD2, DNAJC19, DNMI1L, FDXR, ISCA2, MEER, MFN2, MGME1, MTPAP, NARS2, NDUFAF3, NDUFS1, NR2F1, OPA1, OPA3, PDSS1, POLG, PRPS1, RTN4IP1, SLC19A2, SLC19A3, SLC25A46, SLC52A2, SNX10, SPG7, SUCLA2, TIMM8A, TMEM126A, TSFM, UCHL1, WFS1, YME1L1, ZNHIT3

Endocrinology Test Panels

Abnormal Genitalia/ Disorders of Sex Development Panel	AMH, AMHR2, ANOS1, AR, ARX, ATRX, BCOR, CDK9, CDKN1C, CEP41, CHD4, CHD7, CREBBP, CYB5A, CYP11A1, CYP11B1, CYP17A1, CYP19A1, CYP21A2, DHCR7, DHH, DYNC2H1, ERCC3, FEZF1, FGF17, FGF8, FGFR1, FIG4, FRAS1, FSHB, GATA4, GNRH1, GNRHR, HS6ST1, HSD17B3, HSD3B2, IL17RD, IRF6, KISS1, KISS1R, LEP, LEPR, LHB, LHCGR, MAMLD1, MAP3K1, MKRN3, MKS1, NR0B1, NR5A1, NSMF, POLR3B, POR, PROK2, PROKR2, PROP1, RNF216, RSPO1, SAMD9, SGPL1, SOX10, SOX2, SOX9, SRD5A2, SRY, STAR, TAC3, TACR3, TOE1, TSPYL1, WDR11, WT1, ZFPM2		
Glucocorticoid Deficiency Panel	AAAS, MC2R, MRAP, NNT, NR3C1, POMC, STAR	Hyperlipidemia Panel	ABCA1, ABCG5, ABCG8, ALMS1, APOA1, APOA5, APOB, APOC2, APOC3, APOE, CREB3L3, CYP27A1, GPD1, GPIHBP1, LDLR, LDLRAP1, LIPA, LMF1, LPL, PCSK9
Hyperparathyroidism Panel	AIRE, AP2S1, CASR, CDC73, CDKN1A, CDKN1B, CDKN2B, CDKN2C, GCM2, GNA11, MEN1, PTH, RET, TRPV6	Hypoglycemia, Hyperinsulinism and Ketone Metabolism Panel	ABCC8, ACAT1, ACSF3, AGL, ALDOA, ALDOB, ENO3, EPM2A, FBP1, G6PC, GAA, GBE1, GCK, GLUD1, GYG1, GYS1, GYS2, HADH, HMGCL, HMGCS2, HNF1A, HNF4A, INSR, KCNJ11, LAMP2, LDHA, MPV17, NHLRC1, OXCT1, PC, PCK1, PDX1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PTF1A, PYGL, PYGM, RBCK1, SLC16A1, SLC2A2, SLC37A4, UCP2
Hypomagnesemia Panel	BSND, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, CNNM4, EGF, FAM111A, FXYD2, HNF1B, KCNA1, KCNJ10, MAGT1, NIPA2, PCBD1, SARS2, SLC12A3, TRPM6	Hypothyroidism and Resistance to Thyroid Hormone Panel	DUOX2, DUOX2A, FOXE1, GNAS, HESX1, IGSF1, NKX2-1, NKX2-5, PAX8, POU1F1, PROP1, SECISBP2, SLC16A2, SLC26A4, SLC5A5, TBL1X, TG, THRA, THRB, TPO, TSHB, TSHR
Kallmann Syndrome Panel	ANOS1, CDK9, CHD4, CHD7, DHCR7, FEZF1, FGF17, FGF8, FGFR1, FSHB, GNRH1, GNRHR, HS6ST1, IL17RD, KISS1, KISS1R, LEP, LEPR, LHB, NR0B1, NSMF, POLR3B, PROK2, PROKR2, PROP1, RNF216, SOX10, SOX2, TAC3, TACR3, WDR11	MODY Panel	ABCC8, APPL1, BLK, CEL, GATA6, GCK, HNF1A, HNF1B, HNF4A, INS, KCNJ11, KLF11, NEUROD1, PAX4, PDX1, RFX6, WFS1
Monogenic Obesity Panel	ADCY3, ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP19, CEP290, CPE, CUL4B, DYRK1B, GNAS, KSR2, LEP, LEPR, MAGEL2, MC3R, MC4R, MKKS, MKS1, NR0B2, NTRK2, PCSK1, PHF6, PHIP, POMC, PPARG, SDCCAG8, SH2B1, SIM1, TRIM32, TTC8, TUB, UCP3, VPS13B, WPCP	Premature Ovarian Failure Panel	BMP15, CYP17A1, CYP19A1, FOXL2, FSHR, GALT, GNAS, LHCGR, LMNA, NOBOX, NR5A1, POLG, POR, STAG3, STAR, WT1
Comprehensive Monogenic Diabetes Panel	ABCC8, APPL1, BLK, CEL, EIF2AK3, FOXP3, GATA6, GCK, GLIS3, GLUD1, HADH, HNF1A, HNF1B, HNF4A, INS, INSR, KCNJ11, KLF11, NEUROD1, NEUROG3, PAX4, PDX1, PPARG, PTF1A, RFX6, SLC16A1, SLC2A2, UCP2, WFS1, ZFP57		

Gastroenterology Panels

Gastrointestinal Atresia Panel	CDK9, CHD7, CLMP, DHCR7, EFTUD2, FANCB, FANCC, GLI3, MID1, MNX1, MYCN, PTF1A, RFX6, SOX2, TTC7A	Hirschsprung Disease Panel	BDNF, CELSR3, EDN3, EDNRB, KIF1BP, L1CAM, MITE, NRG1, NRTN, PAX3, PHOX2B, RET, RMRP, SOX10, ZEB2
Cholestasis Panel	ABCB11, ABCB4, ABCC2, AKR1D1, ATP8B1, BAAT, CFTR, CLDN1, CREB3L3, CYP7B1, DCDC2, DGUOK, EPCAM, FAH, HSD3B7, JAG1, LCT, LIPA, LMF1, MKS1, MPV17, MYO5B, NEUROG3, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NR1H4, PEX1, PEX10, PEX12, PEX2, PEX26, PEX5, PEX6, SCYL1, SERPINA1, SLC25A13, SLC26A3, SLCO1B1, SLCO1B3, SMPD1, SPINT2, TJP2, TMEM216, TRMU, TTC37, UGT1A1, VIPAS39, VPS33B	Congenital Diarrhea Panel	ADAM17, ALPI, CYP27A1, DGAT1, EPCAM, FOXP3, GUCY2C, IL10, IL10RA, IL10RB, IL21, LCT, LIPA, MVK, MYO5B, NCF2, NEUROG3, SAR1B, SI, SKIV2L, SLC10A2, SLC26A3, SLC5A1, SLC9A3, SPINT2, STX3, TTC37, TTC7A, XIAP
Polycystic Liver Disease Panel	GANAB, LRP5, PKD1, PKD2, PRKCSH, SEC63	Pancreatitis Panel	APOA5, APOC2, CFTR, CPA1, CTSC, GPIHBP1, PRSS1, SPINK1, UBR1
Congenital Hepatic Fibrosis Panel	AHI1, ANKS6, ARL13B, ARL6, B9D1, B9D2, BAAT, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C5ORF42, CC2D2A, CEP164, CEP290, CEP41, DCDC2, GLIS2, INPP5E, INVS, IQCB1, KIF7, LIPA, MKKS, MKS1, NEK8, NPHP1, NPHP3, NPHP4, NR1H4, OFD1, PKD2, PKHD1, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TRIM32, TTC21B, TTC8, WDR19, WDR35, ZNF423		

Hematology Test Panels

Anemia Panel	ABCB7, ADAMTS13, AK1, ALAS2, AMN, ANK1, ATM, ATR, ATRX, BLM, BRCA2, BRIP1, C15ORF41, CDAN1, CLCN7, CUBN, CYB5R3, DHFR, DNAJC21, DNASE2, EFL1, EPB42, ERCC4, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCE, FANCG, FANCI, FANCL, FANCM, G6PD, GATA1, GCLC, GPI, GSS, HBA1, HBA2, HBB, HFE, KIF23, KLF1, LPIN2, MTR, NBN, NT5C3A, PALB2, PC, PDHA1, PDHX, PIEZO1, PKLR, PUS1, RAD51C, REN, RHAG, RPL11, RPL15, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, SBDS, SEC23B, SLC11A2, SLC19A2, SLC25A38, SLC4A1, SLX4, SPTA1, SPTB, SRP54, TCN2, TE, THBD, TMPRSS6, TPST1, XRCC2, YARS2		
Diamond-Blackfan Anemia Panel	GATA1, RPL11, RPL15, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7	Coagulation Factor Deficiency Panel	F10, F11, F12, F13A1, F2, F5, F7, F8, F9, FGA, FGB, FGG, GGCX, LMAN1, VKORC1, VWF
Dyskeratosis Congenita Panel	ACD, AK2, CTC1, DKC1, NAF1, NHP2, NOP10, PARN, POT1, RTEL1, TERC, TERT, TINF2, USB1, WRAP53	Red Blood Cell Membrane Disorder Panel	ANK1, EPB41, EPB42, RHAG, SLC4A1, SPTA1, SPTB
Fanconi Anemia Panel	ATM, ATR, BLM, BRCA2, BRIP1, CXCR4, ERCC4, ERCC6L2, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCE, FANCG, FANCI, FANCL, FANCM, NBN, PALB2, RAD51C, SLX4, UBE2T, XRCC2	Hemophagocytic Lymphohistiocytosis Panel	FADD, FAS, FASLG, ITK, LYST, MAGT1, MYO5A, PRF1, RAB27A, RECQL4, SH2D1A, STX11, STXBP2, UNC13D, XIAP

Hematology Test Panels

Hereditary Leukemia Panel

ANKRD26, ATM, BLM, BRAF, BRCA1, BRCA2, CBL, CDKN2A, CEBPA, DDX41, DKC1, EFL1, EPCAM, ETV6, FANCA, GATA2, HAVCR2, HRAS, IKZF1, KRAS, MAP2K1, MAP2K2, MLH1, MSH2, MSH6, NBN, NF1, NRAS, PAX5, PMS2, PTPN11, RIT1, RUNX1, SAMD9, SAMD9L, SBDS, SOS1, SRP72, TERC, TERT, TINF2, TP53

Hermansky-Pudlak Syndrome Panel

ABCA3, AP3B1, AP3D1, BLOC1S3, BLOC1S6, DKC1, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, OCA2, SFTPB, SFTPC, SLC45A2, TERC, TERT, TINF2, TYR, TYRP1

Bleeding Disorder/ Coagulopathy Panel

ABCG5, ABCG8, ACTN1, ADAMTS13, ANKRD26, AP3B1, ARPC1B, BLOC1S3, BLOC1S6, CYCS, DTNBP1, EFL1, ETV6, F10, F11, F12, F13A1, F2, F5, F7, F8, F9, FGA, FGB, FGG, FLI1, FLNA, FYB, GATA1, GBA, GFI1B, GGCX, GP1BA, GP1BB, GP9, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2, ITGB3, LMAN1, MASTL, MCFD2, MECOM, MPL, MYH9, NBEAL2, P2RY12, PRKACG, PROC, PROS1, RASGRP2, RBM8A, RUNX1, SERPINC1, SERPINF2, SLFN14, SRC, SRP54, TBXA2R, THBD, THPO, TUBB1, VKORC1, VWF, WAS, WIPF1

Congenital Neutropenia Panel

ACTB, CLPB, CSF2RA, CSF3R, CTSC, EFL1, ELANE, G6PC3, GATA2, GFI1, GINS1, HAX1, IFNGR2, JAGN1, LAMTOR2, LYST, MKL1, PGM3, RAC2, SBDS, SLC37A4, SMARCD2, SRP54, SRP72, VPS13B, VPS45, WAS, WDR1

Bone Marrow Failure Syndrome Panel

ACD, ACTB, AK2, ALAS2, ANKRD26, AP3B1, ATM, ATR, BLM, BLOC1S3, BLOC1S6, BRAF, BRCA1, BRCA2, BRIP1, C15ORF41, C6ORF25, CBL, CDAN1, CDKN2A, CEBPA, CLPB, CSF2RA, CSF3R, CTC1, CTSC, CXCR4, DDX41, DKC1, DNAJC21, DNASE2, DTNBP1, EFL1, ELANE, EPCAM, ERCC4, ERCC6L2, ETV6, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, G6PC3, GATA1, GATA2, GFI1, GINS1, GLRX5, GP1BA, HAX1, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, IFNGR2, IKZF1, ITGA2B, ITK, JAGN1, KCNN4, KRAS, LAMTOR2, LYST, MAGT1, MAP2K1, MAP2K2, MECOM, MKL1, MLH1, MPL, MSH2, MSH6, MYH9, MYO5A, NAF1, NBN, NF1, NHP2, NOP10, NRAS, OBFC1, PALB2, PARN, PAX5, PGM3, PMS2, POT1, PRF1, PTPN11, RAB27A, RAC2, RAD51C, RBM8A, RECQL4, RIT1, RPL11, RPL15, RPL26, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SEC23B, SH2D1A, SLC19A2, SLC25A38, SLC37A4, SLX4, SMARCD2, SOS1, SRP54, SRP72, STX11, STXBP2, TBXA1, TERC, TERT, THPO, TINF2, TP53, TUBB1, UBE2T, UNC13D, USB1, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, XIAP, XRCC2, ZCCHC8

Platelet Function Disorder Panel

AP3B1, ARPC1B, BLOC1S3, BLOC1S6, DTNBP1, GBA, GP1BA, GP1BB, GP9, HPS1, HPS3, HPS4, HPS5, HPS6, ITGA2B, ITGB3, NBEAL2, P2RY12, RASGRP2, RUNX1, TBXA2R, THPO, WIPF1

Thrombocytopenia Panel

ABCG5, ABCG8, ACTN1, ADAMTS13, ANKRD26, ARPC1B, CYCS, EFL1, ETV6, FLI1, FLNA, FYB, GATA1, GBA, GFI1B, GP1BA, GP1BB, GP9, HOXA11, ITGA2, ITGA2B, ITGB3, MASTL, MECOM, MPL, MYH9, NBEAL2, PRKACG, RBM8A, RUNX1, SLFN14, SRC, SRP54, THBD, TUBB1, WAS, WIPF1

Comprehensive Hematology Panel

ABCA3, ABCB7, ABCG5, ABCG8, ACD, ACTB, ACTN1, ADAMTS13, AK1, AK2, ALAS2, AMN, ANK1, ANKRD26, AP3B1, AP3D1, ARPC1B, ATM, ATR, ATRX, BLM, BLOC1S3, BLOC1S6, BRAF, BRCA1, BRCA2, BRIP1, C15ORF41, C6ORF25, CBL, CD59, CDAN1, CDC42, CDKN2A, CEBPA, CECR1, CLCN7, CLPB, CSF2RA, CSF3R, CTC1, CTLA4, CTSC, CUBN, CXCR4, CYB5R3, CYCS, DDX41, DHFR, DKC1, DNAJC21, DNASE2, DTNBP1, EFL1, EGLN1, ELANE, EPAS1, EPB41, EPB42, EPCAM, EPOR, ERCC4, ERCC6L2, ETV6, F10, F11, F12, F13A1, F13B, F2, F5, F7, F8, F9, FADD, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FAS, FASLG, FGA, FGB, FGG, FLI1, FLNA, FYB, G6PC3, G6PD, GATA1, GATA2, GBA, GCLC, GFI1, GFI1B, GGCX, GINS1, GLRX5, GP1BA, GP1BB, GP9, GPI, GPR143, GSS, HAVCR2, HAX1, HBA1, HBA2, HBB, HFE, HK1, HMOX1, HOXA11, HPS1, HPS3, HPS4, HPS5, HPS6, HRAS, IFNGR2, IKZF1, ITGA2, ITGA2B, ITGB3, ITK, JAGN1, JAK2, KCNN4, KIF23, KLF1, KRAS, LAMTOR2, LMAN1, LPIN2, LYST, MAGT1, MAP2K1, MAP2K2, MASTL, MCFD2, MECOM, MKL1, MLH1, MPL, MSH2, MSH6, MTHFD1, MTR, MYH9, MYO5A, NAF1, NBEAL2, NBN, NF1, NHP2, NOP10, NRAS, NT5C3A, OBFC1, OCA2, P2RY12, PALB2, PARN, PAX5, PC, PDHA1, PDHX, PGK1, PGM3, PIEZO1, PKLR, PMS2, POT1, PRF1, PRKACG, PROC, PROS1, PTPN11, PUS1, RAB27A, RAC2, RAD51C, RASGRP2, RBM8A, RECQL4, REN, RHAG, RIT1, RNF168, RPL11, RPL15, RPL26, RPL27, RPL31, RPL35A, RPL5, RPS10, RPS19, RPS24, RPS26, RPS28, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SEC23B, SERPINC1, SERPINF2, SFTPB, SFTPC, SH2D1A, SLC11A2, SLC19A2, SLC25A38, SLC37A4, SLC45A2, SLC46A1, SLC4A1, SLFN14, SLX4, SMARCD2, SOS1, SPTA1, SPTB, SRC, SRP54, SRP72, STAT3, STX11, STXBP2, TBXA2R, TBXA1, TCN2, TERC, TERT, TF, THBD, THPO, TINF2, TMPRSS6, TP53, TPI1, TRNT1, TUBB1, TYR, TYRP1, UBE2T, UNC13D, USB1, VKORC1, VPS13B, VPS45, VWF, WAS, WDR1, WIPF1, WRAP53, XIAP, XRCC2, YARS2, ZCCHC8

Cardiology Test Panels

Hypertrophic Cardiomyopathy (HCM) Panel

ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AGL, ALPK3, APOA1, BAG3, BRAF, CACNA1C, CBL, COX15, CPT2, CSRP3, DES, ELAC2, EPG5, FBXL4, FHL1, FHOD3, FLNC, FXN, GAA, GLA, GSK3B, HRAS, JPH2, KLHL24, KRAS, LAMP2, MAP2K1, MAP2K2, MIPER, MTO1, MYBPC3, MYH7, MYL2, MYL3, NDUFAF2, PLN, PRKAG2, PTPN11, RAF1, RIT1, SLC25A4, SOS1, TNNC1, TNNI3, TNNT2, TPM1, TTR, VCL

Long QT Syndrome (LQTS) Panel

AKAP9, ANK2, CACNA1C, CALM1, CALM2, CALM3, CAV3, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, NOS1AP, SCN5A, SLC12A3, TECRL, TRDN

Cardiomyopathy Panel

AARS2, ABCC6, ABCC9, ACAD9, ACADVL, ACTA1, ACTC1, ACTN2, AGK, AGL, ALMS1, ALPK3, ANO5, APOA1, BAG3, BRAF, CACNA1C, CALR3, CAPN3, CASQ2, CASZ1, CBL, CDH2, CHKB, CHRM2, COX15, CPT2, CRYAB, CSRP3, CTNNA3, DBH, DES, DMD, DNAJC19, DOLK, DPM3, DSC2, DSG2, DSP, DTNA, DYSE, EEF1A2, ELAC2, EMD, EPG5, ETFA, ETFB, ETFDH, FBXL4, FBXO32, FHL1, FHOD3, FKR3, FKTN, FLNC, FOXD4, FOXRED1, FXN, GAA, GATA4, GATA6, GATAD1, GATC, GBE1, GFM1, GLA, GLB1, GMPPB, GSK3B, GTPBP3, GUSB, HADHA, HAND1, HCN4, HFE, HRAS, IDUA, ILK, ISPD, JPH2, JUP, KLHL24, KRAS, LAMA2, LAMP2, LARGE, LDB3, LEMD2, LMNA, LMOD2, LRRC10, LZTR1, MAP2K1, MAP2K2, MAP3K8, MIPER, MLYCD, MTO1, MYBPC3, MYBPHL, MYH6, MYH7, MYL2, MYL3, MYL4, MYOT, MYPN, MYR, NDUFAF2, NDUFB11, NEXN, NF1, NKX2-5, NONO, NRAP, NRAS, PCCA, PCCB, PKP2, PLEC, PLEKHM2, PLN, PNPLA2, PPA2, PPCS, PPP1CB, PRDM16, PRKAG2, PTPN11, QRSL1, RAF1, RASA2, RBCK1, RBM20, RIT1, RMND1, RRAS, RYR2, SCN5A, SCNN1B, SCNN1G, SCO1, SCO2, SDHA, SELENON, SGCA, SGCB, SGCD, SGCG, SHOC2, SLC22A5, SLC25A20, SLC25A4, SMCHD1, SOS1, SOS2, SPFG, SPRED1, TAB2, TAZ, TBX20, TBX5, TCAP, TGF3, TMEM43, TMEM70, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TRIM32, TSFM, TTN, TTR, VCL, VCP, VPS13A, XK

Congenital Structural Heart Disease Panel

ABL1, ACTA2, ACTB, ACTC1, ACTG1, ACVR1, ACVR2B, ADAMTS10, ADAMTS17, AFF4, AMMECR1, ARHGAP31, ARID1A, ARID1B, B3GAT3, BCOR, BMPR2, BRAF, C12ORF57, C2CD3, CASZ1, CBL, CDK13, CDK9, CDKN1C, CFAP53, CHD4, CHD7, CHRM2, CREBBP, CRELD1, CTC1, DHCR7, DLL4, DOCK6, EFTUD2, EHMT1, EIF2AK4, ELN, ENG, EOGT, EP300, EVC, EVC2, FKTN, FLNA, FLT4, FOXC1, FOXF1, FOXH1, FOXP1, GATA4, GATA5, GATA6, GDF1, GJA1, GJA5, GPC3, HAND1, HAND2, HDAC8, HNRNP, HOXA1, HRAS, JAG1, KDM6A, KMT2D, KRAS, KYN, LEFTY2, LZTR1, MED12, MED13L, MEGF8, MEIS2, MMP21, MYCN, MYO18B, MYR, NAA15, NEXN, NF1, NIPBL, NKX2-5, NKX2-6, NODAL, NONO, NOTCH1, NOTCH2, NR2F2, NSD1, PITX2, PKD1L1, PLD1, PPP1CB, PRDM6, PRKD1, PTPN11, PUF60, RAB23, RAF1, RBM10, RECQL4, RERE, RIT1, SALL4, SMARCB1, SMC1A, SMC3, SOS1, SOS2, STAG2, STRA6, TAB2, TBX1, TBX20, TBX5, TFAP2B, TGDS, TLL1, TMEM94, TWIST1, ZEB2, ZFPM2, ZIC3

Liddle Syndrome Panel

SCNN1B, SCNN1G

Noonan Syndrome Panel

ACTB, ACTG1, BMP2, BRAF, CBL, CCNK, CDC42, EPHB4, FGD1, HRAS, KAT6B, KRAS, LZTR1, MAP2K1, MAP2K2, MAP3K8, MRAS, NF1, NF2, NRAS, NSUN2, PPP1CB, PTPN11, RAF1, RASA1, RASA2, RIT1, RRAS, SASH1, SHOC2, SMARCB1, SOS1, SOS2, SPRED1, STAMBP, SYNGAP1

Marfan Syndrome Panel

ABL1, ADAMTS10, ADAMTS17, ADAMTSL4, AEBP1, B3GAT3, BGN, CBS, COL11A1, COL11A2, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, DLG4, EFEMP2, FBN1, FBN2, FLCN, LOX, MAT2A, MED12, PLOD1, SKI, SLC2A10, SMAD3, SMAD6, TGF3, TGFBR1, TGFBR2, UPF3B, VCAN, ZDHHC9

Pulmonary Artery Hypertension (PAH) Panel

ABCC8, ACVRL1, AQP1, ATP13A3, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, FOXF1, GDF2, KCNA5, KCNK3, KLF2, NFU1, NOTCH3, RASA1, SARS2, SMAD4, SMAD9, SOX17, STRA6, TBX4

Left Ventricular Non-Compaction Cardiomyopathy (LVNC) Panel

ABCC9, BAG3, CTNNA3, DES, DMD, DSC2, DSG2, DSP, DTNA, EMD, FBXO32, FLNC, HCN4, JPH2, JUP, LAMP2, LMNA, MIPER, MYBPC3, MYH6, MYH7, PKP2, PLEKHM2, PLN, RAF1, RBM20, RYR2, SCN5A, TCAP, TNNT2, TPM1, TTN, VCL

Cardiology Test Panels

Aorta Panel

ABCC6, ABL1, ACTA2, ADAMTS10, ADAMTS17, ADAMTS2, ADAMTS14, ALDH18A1, ATP7A, B3GAT3, BGN, CBS, COL1A1, COL1A2, COL2A1, COL3A1, COL4A5, COL5A1, COL5A2, COLGALT1, EFEMP2, ELN, ENPP1, FBLN5, FBN1, FBN2, FKBP14, FLNA, FOXE3, GATA5, HCN4, LOX, MAT2A, MED12, MFAP5, MYH11, MYLK, NOTCH1, PLOD1, PLOD3, PRKG1, SKI, SLC2A10, SLC39A13, SMAD2, SMAD3, SMAD4, SMAD6, TGFB2, TGFB3, TGFBRI, TGFBRI2, ZDHHC9

Arrhythmia Panel

ABCC9, AKAP9, ANK2, BAG3, CACNA1C, CACNB2, CALM1, CALM2, CALM3, CASQ2, CAV3, CDH2, CTNNA3, DBH, DES, DSC2, DSG2, DSP, FLNC, GATA6, HADHA, HCN4, JUP, KCNA5, KCNE1, KCNE2, KCNH2, KCNJ2, KCNJ5, KCNQ1, LDB3, LEMD2, LMNA, MYH6, MYH7, MYL4, NKX2-5, NOS1AP, NUP155, PKP2, PLN, PPA2, PRKAG2, RBM20, RYR2, SALL4, SCN10A, SCN1B, SCN3B, SCN5A, SLC12A3, TANGO2, TBX5, TECRL, TGFB3, TMEM43, TNNI3, TNNI3K, TNNT2, TRDN, TRPM4, TTN

Arrhythmogenic Right Ventricular Cardiomyopathy (ARVC) Panel

BAG3, CDH2, CTNNA3, DES, DSC2, DSG2, DSP, FLNC, JUP, LDB3, LEMD2, LMNA, MYH7, NKX2-5, PKP2, PLN, RYR2, SCN5A, TGFB3, TMEM43, TTN

Brugada Syndrome Panel

CACNA1C, CACNB2, HCN4, KCNH2, SCN3B, SCN5A, TRPM4

Dilated Cardiomyopathy (DCM) Panel

ABCC6, ABCC9, ACADVL, ACTA1, ACTC1, ACTN2, ALMS1, ALPK3, APOA1, BAG3, CASZ1, CHKB, CHRM2, CPT2, DES, DMD, DNAJC19, DOLK, DPM3, DSC2, DSG2, DSP, DYSE, EEF1A2, EMD, EPG5, ETFA, ETFB, ETFDH, FBXO32, FHOD3, FKRP, FKTN, FLNC, FOXD4, GATA4, GATA6, GATC, GBE1, GLB1, GSK3B, HAND1, HCN4, ILK, JPH2, JUP, KLHL24, LAMP2, LDB3, LEMD2, LMNA, LMOD2, LRRIC10, MLYCD, MYBPC3, MYBPHL, MYH6, MYH7, MYL4, NEXN, NKX2-5, NRAP, PCCA, PCCB, PKP2, PLEKHM2, PLN, PPCS, PRDM16, QRS1, RAF1, RBCK1, RBM20, RMND1, SCN5A, SLC22A5, SPEG, TAB2, TAZ, TBX20, TBX5, TCAP, TMEM43, TNNC1, TNNI3, TNNI3K, TNNT2, TOR1AIP1, TPM1, TTN, TTR, VCL, VPS13A

Hereditary Hemorrhagic Telangiectasia (HHT) Panel

ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4

Ehlers-Danlos Syndrome Panel

ABCC6, ADAMTS2, ADAMTS12, AEBP1, ALDH18A1, ATP6V0A2, ATP6V1A, ATP6V1E1, ATP7A, B3GALT6, B4GALT7, BGN, C1R, C1S, CBS, CHST14, CHST3, COL11A1, COL1A1, COL1A2, COL2A1, COL3A1, COL5A1, COL5A2, DSE, EFEMP2, ELN, FBLN5, FBN1, FBN2, FKBP14, FLNA, GORAB, PLOD1, PYCR1, SLC39A13, SMAD3, TGFB2, TGFBRI, TGFBRI2, ZNF469

Heterotaxy and Situs Inversus Panel

ACVR2B, ANKS6, ARMC4, C21ORF59, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CFAP53, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH11, DNAH5, DNAI1, DNAI2, DNALI, DDX1C1, FOXH1, GDF1, INVS, LEFTY2, LRRC6, MMP21, NODAL, PIH1D3, PITRM1, PKD1L1, SPAG1, TTC25, ZIC3, ZMYND10

Hyperlipidemia Panel

ABCA1, ABCG5, ABCG8, ALMS1, APOA1, APOA5, APOB, APOC2, APOC3, APOE, CREB3L3, CYP27A1, GPD1, GPIHBP1, LDLR, LDLRAP1, LIPA, LMF1, LPL, PCSK9

Ear Nose Throat Test Panels

Alport Syndrome Panel	CD151, COL4A3, COL4A4, COL4A5, COL4A6, MYH9	Branchio-Oto-Renal (BOR) Syndrome Panel	EYA1, SIX1, SIX5, TFAP2A
Comprehensive Hearing Loss and Deafness Panel	ABHD12, ABHD5, ACOX1, ACTB, ACTG1, ADCY1, ADGRV1, AIFM1, ALMS1, AMMECR1, ANKH, ANLN, ARSG, ATP2B2, ATP6V0A4, ATP6V1B1, ATP6V1B2, BCS1L, BDP1, BSND, BTBD, C10ORF2, CABP2, CACNA1D, CATSPER2, CCDC50, CD151, CD164, CDC14A, CDC42, CDH23, CDK9, CDKN1C, CEACAM16, CEP250, CEP78, CHD7, CHSY1, CIB2, CISD2, CLDN14, CLIC5, CLPP, CLRN1, COCH, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, CRYM, DCAF17, DCDC2, DFNA5, DFNB31, DFNB59, DIABLO, DIAPH1, DIAPH3, DLX5, DMXL2, DNMT1, DSPP, EDN3, EDNRA, EDNRB, EFTUD2, EIF3E, ELMOD3, EPS8, EPS8L2, ESPN, ESRRB, EYA1, EYA4, FAM136A, FAM65B, FDXR, FGF3, FGFR2, FGFR3, FITM2, FOXC1, FOXI1, GATA3, GDF6, GIPC3, GJA1, GJB2, GJB3, GJB6, GPSM2, GREB1L, GRHL2, GRXCR1, GRXCR2, HARS, HARS2, HGF, HOMER2, HOXA2, HOXB1, HSD17B4, ILDR1, KARS, KCNE1, KCNJ10, KCNQ1, KCNQ4, KIT, KITLG, KMT2D, LARS2, LHFPL5, LHX3, LMX1A, LOXHD1, LRP2, LRTOMT, MAN2B1, MANBA, MARVELD2, MASP1, MCM2, MET, MGP, MIR96, MITE, MPZL2, MSRB3, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, NDP, NDRG1, NEFL, NF2, NLRP3, NOG, NR2F1, OPA1, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PAX1, PAX3, PCDH15, PCGF2, PDE1C, PDZD7, PEX1, PEX11B, PEX12, PEX13, PEX14, PEX2, PEX26, PEX5, PEX6, PEX7, PHYH, PISD, PMP22, PNPT1, POLR1C, POLR1D, POU3F4, POU4F3, PRPS1, PTPRQ, RAI1, RDX, REST, RMND1, ROR1, RPS6KA3, S1PR2, SALL1, SALL4, SEMA3E, SERAC1, SERPINB6, SH3TC2, SIX1, SIX2, SIX5, SLC17A8, SLC19A2, SLC22A4, SLC26A4, SLC26A5, SLC29A3, SLC33A1, SLC44A4, SLC4A11, SLC52A2, SLC52A3, SLITRK6, SMAD4, SMPX, SNAI2, SOX10, SPATA5, STAG2, STRC, SUCLA2, SUCLG1, SYNE4, SYT2, TBC1D24, TBL1X, TBX1, TCOF1, TECTA, TFAP2A, TIMM8A, TJP2, TMC1, TMEM126A, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TRMU, TRRAP, TSHZ1, TSPEAR, TUBB4B, TYR, UBR1, USH1C, USH1G, USH2A, VCAN, WBP2, WFS1, XYLT2		
Hereditary Hemorrhagic Telangiectasia (HHT) Panel	ACVRL1, ENG, EPHB4, GDF2, RASA1, SMAD4	Pendred Syndrome Panel	FOXI1, KCNJ10, SLC26A4
Non-Syndromic Hearing Loss Panel	ACTG1, ADCY1, ATP2B2, BDP1, BSND, CABP2, CCDC50, CD164, CDC14A, CDH23, CEACAM16, CIB2, CLDN14, CLIC5, COCH, COL11A2, COL4A6, CRYM, DCDC2, DFNA5, DFNB31, DFNB59, DIABLO, DIAPH1, DIAPH3, DMXL2, DSPP, ELMOD3, EPS8, EPS8L2, ESPN, ESRRB, EYA4, FAM65B, GIPC3, GJB2, GJB3, GJB6, GPSM2, GRHL2, GRXCR1, GRXCR2, HGF, HOMER2, ILDR1, KARS, KCNQ4, LHFPL5, LMX1A, LOXHD1, LRTOMT, MARVELD2, MET, MIR96, MPZL2, MSRB3, MYH14, MYH9, MYO15A, MYO3A, MYO6, MYO7A, NARS2, OSBPL2, OTOA, OTOF, OTOG, OTOGL, P2RX2, PCDH15, PDE1C, PNPT1, POU3F4, POU4F3, PRPS1, RDX, S1PR2, SERPINB6, SIX1, SLC17A8, SLC22A4, SLC26A4, SLC26A5, SLITRK6, SMPX, STRC, SYNE4, TBC1D24, TECTA, TJP2, TMC1, TMEM132E, TMIE, TMPRSS3, TNC, TPRN, TRIOBP, TSPEAR, USH1C, WBP2, WFS1		
Syndromic Hearing Loss Panel	ABHD12, ACTG1, ADGRV1, ALMS1, ANKH, ARSG, ATP6V1B1, ATP6V1B2, BCS1L, BSND, BTBD, C10ORF2, CACNA1D, CD151, CDH23, CDK9, CDKN1C, CEP250, CEP78, CHD7, CHSY1, CIB2, CLPP, CLRN1, COL11A1, COL11A2, COL2A1, COL4A3, COL4A4, COL4A5, COL4A6, COL9A1, COL9A2, COL9A3, DCAF17, DFNB31, DLX5, DNMT1, EDN3, EDNRB, EIF3E, ESPN, EYA1, FDXR, FGF3, FITM2, FOXI1, GATA3, GJA1, HARS, HARS2, HOXB1, KCNE1, KCNJ10, KCNQ1, KIT, LARS2, LRP2, MAN2B1, MANBA, MGP, MITE, MYH9, MYO7A, NDP, NLRP3, PAX3, PCDH15, PDZD7, PEX1, PEX26, PEX6, PISD, POLR1C, POLR1D, PRPS1, SALL1, SEMA3E, SIX1, SIX5, SLC19A2, SLC26A4, SLC52A2, SLC52A3, SLITRK6, SMAD4, SNAI2, SOX10, TBL1X, TCOF1, TFAP2A, TIMM8A, TSHZ1, TUBB4B, TYR, USH1C, USH1G, USH2A, VCAN, WFS1, XYLT2		
Stickler Syndrome Panel	COL11A1, COL11A2, COL2A1, COL9A1, COL9A2, COL9A3, LRP2, VCAN	Waardenburg Syndrome Panel	EDN3, EDNRB, KIT, MITE, PAX3, SNAI2, SOX10
Usher Syndrome Panel	ABHD12, ADGRV1, ARSG, CDH23, CEP250, CEP78, CIB2, CLRN1, DFNB31, ESPN, HARS, MYO7A, PCDH15, PDZD7, PEX1, PEX6, PRPS1, TUBB4B, USH1C, USH1G, USH2A		

Metabolic Test Panels

Aicardi-Goutieres Syndrome Panel

ADAR, IFIH1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1

Glycogen Storage Disorder Panel

AGL, ALDOA, ENO3, EPM2A, FBP1, G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, NHLRC1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PYGL, PYGM, RBCK1, SLC2A2, SLC37A4

Comprehensive Metabolism Panel

ABCC8, ABCD1, ABCD3, ABCD4, ACAD8, ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACOX1, ACSF3, ACY1, ADA, ADAMTSL2, ADAR, ADCK3, ADK, ADSL, AGA, AGK, AGL, AGPAT2, AGPS, AGXT, AHCY, AKT2, ALAD, ALAS2, ALDH5A1, ALDH7A1, ALDOA, ALDOB, ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, AMACR, AMN, AMPD1, AMT, ANO10, ANO5, ANTXR2, APRT, APTX, ARG1, ARSA, ARSB, ASAH1, ASL, ASPA, ASS1, ATIC, ATP13A2, ATP2A1, ATP6V0A2, ATP7B, AUH, B3GALNT2, B3GLCT, B4GALT1, B4GAT1, BCKDHA, BCKDHB, BCS1L, BOLA3, BSCL2, BSND, BTD, C10ORF2, C12ORF65, CA5A, CACNA1S, CAPN3, CASQ1, CASR, CAV1, CAV3, CBS, CD320, CHKB, CIDEA, CLCN1, CLCNKB, CLDN16, CLDN19, CLN3, CLN5, CLN6, CLN8, CLPB, CNM2, CNM4, COG1, COG4, COG5, COG6, COG7, COG8, COL11A2, COL2A1, COQ2, COQ4, COQ5, COQ6, COQ7, COQ9, CPOX, CPS1, CPT1A, CPT2, CTDP1, CTH, CTNS, CTSA, CTSC, CTSD, CTSK, CUBN, D2HGDH, DAG1, DBT, DDOST, DGUOK, DHCR7, DHDDS, DHODH, DLD, DMD, DNAJB6, DNAJC12, DNMI1L, DOLK, DPAGT1, DPM1, DPM2, DPM3, DPYD, DPYS, DYM, DYSF, EBP, ECHS1, EGF, EMD, ENO3, EPM2A, ETFA, ETFB, ETFDH, FAH, FAM111A, FBP1, FBXL4, FDX1L, FECH, FH, FHL1, FKBP, FKTN, FLAD1, FLNA, FLNB, FMO3, FOLR1, FOXRED1, FUCA1, FUT8, FXSD2, G6PC, GAA, GALT, GALK1, GALNS, GALT, GAMT, GATM, GBA, GBE1, GCDH, GCH1, GCK, GCSH, GFM1, GIF, GLA, GLB1, GLDC, GLRX5, GLUD1, GLUL, GM2A, GMPPA, GMPBP, GNE, GNMT, GNPAT, GNPTAB, GNPTG, GNS, GPC3, GPHN, GUSB, GYG1, GYS1, GYS2, HADH, HADHA, HADHB, HAMP, HCF1, HEXA, HEXB, HFE, HFE2, HGD, HGSNAT, HIBCH, HLCS, HMBS, HMGCL, HMGCS2, HNF1A, HNF1B, HNF4A, HPD, HPRT1, HRAS, HSD17B10, HSD17B4, HYAL1, IDH2, IDS, IDUA, IFIH1, INSR, ISCU, IVD, KCNA1, KCNJ10, KCNJ11, KCNJ2, L2HGDH, LAMA2, LAMP2, LARGE, LCT, LDB3, LDHA, LIAS, LIPA, LIPE, LIPT1, LMBRD1, LMNA, LPIN1, MAGT1, MAN1B1, MAN2B1, MANBA, MCCC1, MCCC2, MCEE, MCOLN1, MFN2, MFSD8, MGAT2, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MOCOS, MOCS1, MOCS2, MOGS, MPDU1, MPI, MPV17, MTHFR, MTR, MTRR, MUT, MYH3, MYOT, NAGA, NAGLU, NAGS, NBAS, NDUFAF2, NDUFS1, NEU1, NFU1, NGLY1, NHLRC1, NIPA2, NPC1, NPC2, NT5C3A, OAT, OPA1, OPA3, OTC, OXCT1, PAH, PC, PCBD1, PCCA, PCCB, PCK1, PDHA1, PDHB, PDHX, PDSS1, PDSS2, PDX1, PEPD, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PHYH, PLIN1, PMM2, PNP, PNPLA2, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PPARG, PPOX, PPT1, PRKAG2, PRKAG3, PRODH, PRPS1, PSAP, PTF1A, PTRF, PTS, PYGL, PYGM, QDPR, RAI1, RBCK1, REN, RFT1, RNASEH2A, RNASEH2B, RNASEH2C, RRM2B, RYR1, SAMHD1, SARS2, SCN4A, SEC23B, SERAC1, SERPINA1, SGCA, SGCB, SGCD, SGCG, SGSH, SI, SIL1, SLC12A3, SLC16A1, SLC17A5, SLC22A5, SLC25A1, SLC25A13, SLC25A15, SLC25A20, SLC25A26, SLC25A3, SLC25A4, SLC2A1, SLC2A2, SLC30A10, SLC35A1, SLC35A2, SLC35C1, SLC37A4, SLC39A4, SLC3A1, SLC40A1, SLC46A1, SLC5A1, SLC6A19, SLC6A8, SLC6A9, SLC7A7, SLC7A9, SMPD1, SPG7, SRD5A3, SSR4, STAC3, STT3A, STT3B, SUCLA2, SUCLG1, SUGCT, SUMF1, SUOX, TALDO1, TANGO2, TAT, TAZ, TBC1D4, TCAP, TCF4, TCN2, TFR2, TIMM8A, TK2, TMEM126A, TMEM165, TMEM70, TNPO3, TPMT, TPP1, TREX1, TRIM32, TRIM37, TRPM6, TUSC3, TYMP, UCP2, UMOD, UMPS, UPB1, UROD, UROS, WFS1, XDH, ZMPSTE24

Metabolic Test Panels

Hyperammonemia and Urea Cycle Disorder Panel

ACADM, ACADS, ACADVL, ARG1, ASL, ASS1, BCKDHA, BCKDHB, CA5A, CPS1, CPT1A, CPT2, DBT, DLD, ETFA, ETFB, ETFDH, GLUD1, GLUL, HADHA, HADHB, HCFC1, HLCS, HMGCL, HMGS2, IVD, MCCC1, MCCC2, MMAA, MMAB, MMACHC, MMADHC, MUT, NAGS, NBAS, OAT, OTC, PC, PCCA, PCCB, SLC22A5, SLC25A13, SLC25A15, SLC25A20, SLC7A7, SUCLA2, SUCLG1, TMEM70, UMPS

Hypoglycemia, Hyperinsulinism and Ketone Metabolism Panel

ABCC8, ACAT1, ACSF3, AGL, ALDOA, ALDOB, ENO3, EPM2A, FBP1, G6PC, GAA, GBE1, GCK, GLUD1, GYG1, GYS1, GYS2, HADH, HMGCL, HMGS2, HNF1A, HNF4A, INSR, KCNJ11, LAMP2, LDHA, MPV17, NHLRC1, OXCT1, PC, PCK1, PDX1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PRKAG3, PTF1A, PYGL, PYGM, RBCK1, SLC16A1, SLC2A2, SLC37A4, UCP2

Hyperphenylalaninemia Panel

DNAJC12, GCH1, PAH, PCBD1, PTS, QDPR

Hypomagnesemia Panel

BSND, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, CNNM4, EGF, FAM111A, FXD2, HNF1B, KCNA1, KCNJ10, MAGT1, NIPA2, PCBD1, SARS2, SLC12A3, TRPM6

Nephrolithiasis Panel

ADCY10, AGXT, ALPL, APRT, ATP6V0A4, ATP6V1B1, ATP7B, BSND, CA2, CASR, CLCN5, CLCNKA, CLCNKB, CLDN16, CLDN19, CLPB, CYP24A1, FAM20A, FOXI1, GNA11, GPHN, GRHR, HNF4A, HOGA1, HPRT1, KCNJ1, MAGED2, MOCOS, MOCS1, MOCS2, OCRL, PRPS1, SLC12A1, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC7A9, SLC9A3R1, UMOD, VDR, XDH

Nonketotic Hyperglycinemia / Glycine Encephalopathy Panel

AMT, BOLA3, GCSH, GLDC, GLRX5, LIAS, LIPT1, NFU1, SLC6A9

Organic Acidemia/ Aciduria & Cobalamin Deficiency Panel

ABCD4, ACADSB, ACAT1, ACSF3, ADK, AHCY, AMN, BCKDHA, BCKDHB, BCS1L, CBS, CD320, CLPB, CTH, CUBN, D2HGDH, DBT, DLD, ETFA, ETFB, ETFDH, FLAD1, GCDH, GIF, GNMT, HCFC1, HIBCH, HMGCL, IDH2, IVD, L2HGDH, LMBRD1, MCCC1, MCCC2, MCEE, MLYCD, MMAA, MMAB, MMACHC, MMADHC, MTHFR, MTR, MTRR, MUT, PCCA, PCCB, PEPD, SERAC1, SLC25A1, SUCLA2, SUCLG1, SUGCT, TCN2, UMPS

Periodic Paralysis Panel

CACNA1S, CLCN1, KCNJ2, SCN4A

Peroxisomal Disorders Panel

ABCD1, ABCD3, ACOX1, AGPS, AGXT, AMACR, DYM, EBP, GNPAT, HSD17B4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, SUGCT, TRIM37

Porphyrria Panel

ALAD, ALAS2, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS

Purine and Pyrimidine Metabolism Disorders Panel

ADA, ADSL, AMPD1, APRT, ATIC, DHODH, DPYD, DPYS, GPHN, HPRT1, MOCOS, MOCS1, NT5C3A, PNP, PRPS1, REN, TPMT, UMOD, UMPS, UPB1, XDH

Cystinuria Panel

SLC3A1, SLC7A9

Tyrosinemia Panel

FAH, HPD, TAT

Fatty Acid Oxidation Syndrome Panel

ACAD8, ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADVL, ALDH5A1, CPT1A, CPT2, ECHS1, ETFA, ETFB, ETFDH, GLUD1, HADH, HADHA, HADHB, HMGCL, HMGS2, HSD17B10, LPIN1, PPARG, SLC22A5, SLC25A20, TAZ

Metabolic Test Panels

Homocystinuria Core Panel	CBS, MTHFR, MTR, MTRR	Hereditary Hemochromatosis Panel	HAMP, HFE, HFE2, SLC40A1, TFR2
Coenzyme q10 Deficiency Panel	ADCK3, ANO10, APTX, COQ2, COQ4, COQ5, COQ6, COQ7, COQ9, ETFA, ETFB, ETFDH, PDSS1, PDSS2, SLC25A26	Congenital Mono- and Disaccharide Disorders Panel	ALDOB, GALE, GALK1, GALT, LCT, SI, SLC2A1, SLC2A2, SLC5A1
Congenital Disorders of Glycosylation Panel	ALG1, ALG11, ALG12, ALG13, ALG2, ALG3, ALG6, ALG8, ALG9, ATP6V0A2, B3GLCT, B4GALT1, COG1, COG4, COG5, COG6, COG7, COG8, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, FUT8, GMPPA, GNE, MAGT1, MAN1B1, MGAT2, MOGS, MPDU1, MPI, NGLY1, PGM1, PMM2, RFT1, SEC23B, SLC35A1, SLC35A2, SLC35C1, SRD5A3, SSR4, STT3A, STT3B, TMEM165, TUSC3		
Congenital and Familial Lipodystrophy Panel	AGPAT2, AKT2, BSCL2, CAV1, CIDEC, LIPE, LMNA, PLIN1, PPARG, PTRF, TBC1D4, ZMPSTE24	Creatine Metabolism Deficiency Panel	GAMT, GATM, SLC6A8
Metabolic Liver Failure Panel	ALDOB, ATP7B, FAH, GALE, GALK1, GALT, LIPA, MPI, NPC1, NPC2, PHKA2, PHKB, PHKG2, PYGL, SERPINA1, SMPD1	Mitochondrial DNA Depletion Syndrome Panel	AGK, APTX, AUH, C10ORF2, C12ORF65, DGUOK, FBXL4, MFN2, MPV17, NDUFS1, OPA1, OPA3, POLG, POLG2, RRM2B, SLC25A3, SLC25A4, SPG7, SUCLA2, SUCLG1, TIMM8A, TK2, TMEM126A, TYMP, WFS1
Metabolic Myopathy and Rhabdomyolysis Panel	ACAD9, ACADL, ACADM, ACADVL, ADCK3, AGL, AHCY, ALDOA, AMPD1, ANO5, ATP2A1, B3GALNT2, B4GAT1, C10ORF2, CAPN3, CASQ1, CAV3, CHKB, COQ2, CPT2, CTDP1, DAG1, DGUOK, DMD, DNAJB6, DPM1, DPM2, DYSE, EMD, ENO3, ETFA, ETFB, ETFDH, FDX1L, FHL1, FKRP, FKTN, FLAD1, GAA, GBE1, GMPPB, GYG1, GYS1, HADHA, HADHB, ISCU, LAMA2, LAMP2, LARGE, LDHA, LPIN1, MYH3, OPA1, OPA3, PDSS2, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PNPLA2, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PYGM, RBCK1, RRM2B, RYR1, SCN4A, SGCA, SGCB, SGCD, SGCG, SIL1, SLC22A5, SLC25A20, STAC3, SUCLA2, SUCLG1, TANGO2, TCAP, TK2, TNPO3, TRIM32, TYMP		
Monogenic Obesity Panel	ADCY3, ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP19, CEP290, CPE, CUL4B, DYRK1B, GNAS, KSR2, LEP, LEPR, MAGEL2, MC3R, MC4R, MKKS, MKS1, NR0B2, NTRK2, PCSK1, PHF6, PHIP, POMC, PPARG, SDCCAG8, SH2B1, SIM1, TRIM32, TTC8, TUB, UCP3, VPS13B, WDPCP		
Lysosomal Disorders and Mucopolysaccharidosis Panel	ABCC8, ACY1, ADAMTSL2, ADSL, AGA, ALDH5A1, ALDH7A1, AMT, ANTXR2, ARG1, ARSA, ARSB, ASAH1, ASPA, ATP13A2, BTBD, CLN3, CLN5, CLN6, CLN8, COL11A2, COL2A1, CTNS, CTSB, CTSC, CTSD, CTSK, DHCR7, DPYD, DYM, ETFB, ETFB, ETFDH, FH, FOLR1, FUCA1, GAA, GALT, GALNS, GAMT, GBA, GCDH, GLA, GLB1, GLDC, GM2A, GNE, GNPTAB, GNPTG, GNS, GPC3, GUSB, HEXA, HEXB, HGSNAT, HRAS, HYAL1, IDS, IDUA, L2HGDH, LAMA2, LAMP2, LDB3, LIPA, MAN1B1, MAN2B1, MANBA, MCOLN1, MFSD8, MOCOS1, MOCOS2, MYOT, NAGA, NAGLU, NEU1, NPC1, NPC2, PEX1, PEX10, PEX12, PEX13, PEX16, PEX26, PEX3, PEX5, PEX6, PGK1, PHYH, PPT1, PRODH, PSAP, QDPR, RAI1, SGSH, SLC17A5, SLC25A15, SLC46A1, SMPD1, SUMF1, SUOX, TCF4, TPP1		

Nephrology Test Panels

Alport Syndrome Panel	CD151, COL4A3, COL4A4, COL4A5, COL4A6, MYH9	Bardet-Biedl Syndrome Panel	ALMS1, ARL6, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C8ORF37, CEP19, CEP290, CPE, IFT172, IFT27, LZTFL1, MKKS, MKS1, PNPLA6, SCAPER, SDCCAG8, TMEM67, TRIM32, TTC8, WDPCP
Bartter Syndrome Panel	AP2S1, BSND, CASR, CLCNKA, CLCNKB, GNA11, KCNJ1, MAGED2, SLC12A1, SLC12A3	Branchio-Oto-Renal (BOR) Syndrome Panel	EYA1, SIX1, SIX5, TFAP2A
Diabetes Insipidus Panel	AQP2, AVP, AVPR2	Hemolytic Uremic Syndrome Panel	ADAMTS13, C3, CD46, CFB, CFH, CFHR5, CFI, DGKE, THBD
Hypophosphatemic Rickets Panel	ALPL, CLCN5, CYP27B1, CYP2R1, DMP1, ENPP1, FAH, FGF23, KL, PHEX, SLC34A1, SLC34A3, VDR	Hypomagnesemia Panel	BSND, CASR, CLCNKB, CLDN16, CLDN19, CNNM2, CNNM4, EGF, FAM111A, FXYP2, HNF1B, KCNA1, KCNJ10, MAGT1, NIPA2, PCBD1, SARS2, SLC12A3, TRPM6
Joubert Syndrome Panel	AHI1, ARL13B, ARMC9, B9D1, B9D2, C21ORF2, C5ORF42, CC2D2A, CEP104, CEP120, CEP164, CEP290, CEP41, CSPP1, INPP5E, KIAA0556, KIAA0586, KIAA0753, KIF7, MKS1, NPHP1, NPHP3, OFD1, PDE6D, RPGRIP1L, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TTC21B, ZNF423	Cystic Kidney Disease Panel	ANKS6, CEP164, CEP290, CEP83, COL4A1, CRB2, DCDC2, DNAJB11, DZIP1L, EYA1, GANAB, GLIS2, HNF1B, IFT172, INVS, IQCB1, JAG1, LRP5, MAPKBP1, NEK8, NOTCH2, NPHP1, NPHP3, NPHP4, OFD1, PAX2, PKD1, PKD2, PKHD1, PRKCSH, RPGRIP1L, SDCCAG8, SEC61A1, SEC63, SIX5, TMEM67, TSC1, TSC2, TTC21B, UMOD, VHL, WDR19, ZNF423
Liddle Syndrome Panel	SCNN1B, SCNN1G	Meckel Syndrome Panel	B9D1, B9D2, CC2D2A, CEP290, KIF14, MKS1, NPHP3, RPGRIP1L, TCTN2, TMEM107, TMEM216, TMEM231, TMEM67
Monogenic Obesity Panel	ADCY3, ALMS1, ARL6, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, CEP19, CEP290, CPE, CUL4B, DYRK1B, GNAS, KSR2, LEP, LEPR, MAGEL2, MC3R, MC4R, MKKS, MKS1, NR0B2, NTRK2, PCSK1, PHF6, PHIP, POMC, PPARG, SDCCAG8, SH2B1, SIM1, TRIM32, TTC8, TUB, UCP3, VPS13B, WDPCP	Nephrolithiasis Panel	ADCY10, AGXT, ALPL, APRT, ATP6V0A4, ATP6V1B1, ATP7B, BSND, CA2, CASR, CLCN5, CLCNKA, CLCNKB, CLDN16, CLDN19, CLPB, CYP24A1, FAM20A, FOXI1, GNA11, GPHN, GRHPR, HNF4A, HOGA1, HPRT1, KCNJ1, MAGED2, MOCOS, MOC51, MOC52, OCRL, PRPS1, SLC12A1, SLC22A12, SLC26A1, SLC2A9, SLC34A1, SLC34A3, SLC3A1, SLC4A1, SLC7A9, SLC9A3R1, UMOD, VDR, XDH
Nephronophthisis Panel	ANKS6, CEP164, CEP290, CEP83, DCDC2, GLIS2, IFT172, INVS, IQCB1, MAPKBP1, NEK8, NPHP1, NPHP3, NPHP4, RPGRIP1L, SDCCAG8, TMEM67, TTC21B, WDR19, ZNF423	Nephrotic Syndrome Panel	ACTN4, ADCK4, ANLN, APOL1, ARHGAP24, ARHGDIA, CD2AP, COL4A3, COL4A4, COL4A5, COQ2, COQ6, CRB2, CUBN, DGKE, DLC1, EMP2, FAN1, FAT1, FN1, INF2, ITGA3, KANK1, KANK2, KANK4, LAGE3, LAMB2, LMX1B, IYZ, MAFB, MAGI2, MYH9, MYO1E, NPHS1, NPHS2, NUP107, NUP133, NUP205, NUP85, NUP93, OSGEP, PAX2, PDSS2, PLCE1, PTPRO, SCARB2, SGPL1, SMARCAL1, TBC1D8B, TP53RK, TPRKB, TRIM8, TRPC6, TTC21B, TTR, WDR4, WDR73, WT1, XPO5

Nephrology Test Panels

Polycystic Kidney Disease Panel

DNAJB11, DZIP1L, GANAB, HNF1B, JAG1, LRP5, NOTCH2, PKD1, PKD2, PKHD1, PRKCSH, SEC61A1, SEC63

Primary Hyperoxaluria Panel

AGXT, GRHPR, HOGA1

Primary Ciliary Dyskinesia Panel

ARMC4, C11ORF70, C21ORF59, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CENPF, CFAP57, CFTR, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAH1, DNAI2, DNAJB13, DNAL1, DRC1, DYX1C1, GAS2L2, GAS8, HYDIN, INVS, LRRC56, LRRC6, MCIDAS, NME8, OFD1, PIH1D3, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, STK36, TTC25, ZMYND10

Pseudohypoaldosteronism Panel

CUL3, HSD11B2, KCNJ5, KLHL3, NR3C2, SCNN1A, SCNN1B, SCNN1G, WNK1, WNK4

Renal Malformation Panel

ACE, ACTG2, AGT, BMP4, CDC5L, CHD1L, DSTYK, EYA1, FAM58A, FANCB, FOXC2, FREM1, FREM2, GATA3, GREB1L, HNF1B, KYNU, PAX2, PBX1, PUF60, REN, RET, ROBO2, SALL1, SIX1, SIX5, WT1

Renal Tubular Acidosis Panel

ATP6V0A4, ATP6V1B1, CA2, SLC4A1, SLC4A4

Ciliopathy Panel

ACVR2B, ADAMTS9, AHI1, ALMS1, ANKS6, ARL13B, ARL3, ARL6, ARMC9, ATXN10, B9D1, B9D2, BBIP1, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, C21ORF2, C2CD3, C5ORF42, C8ORF37, CC2D2A, CENPF, CEP104, CEP120, CEP164, CEP19, CEP290, CEP41, CEP55, CEP83, CPE, CRB2, CSPP1, DCD2, DDX59, DHCR7, DYNC2H1, DYNC2L1, EVC, EVC2, FAM58A, FAN1, GLI2, GLI3, GLIS2, HYL1, ICK, IFT122, IFT140, IFT172, IFT27, IFT43, IFT52, IFT57, IFT74, IFT80, IFT81, INPP5E, INTU, INVS, IQCB1, KIAA0556, KIAA0586, KIAA0753, KIF14, KIF7, LEFTY2, LZTFL1, MAPKBP1, MKKS, MKS1, NEK1, NEK8, NODAL, NPHP1, NPHP3, NPHP4, OFD1, PDE6D, PIBF1, PKD1, PKD2, PKHD1, PMM2, PNPLA6, POC1B, RPGRIP1L, SCAPER, SCLT1, SDCCAG8, SUFU, TCTEX1D2, TCTN1, TCTN2, TCTN3, TMEM107, TMEM138, TMEM216, TMEM231, TMEM237, TMEM67, TRAF3IP1, TRIM32, TTC21B, TTC8, USP9X, WDR34, WDR35, WDR60, XPNPEP3, ZIC3, ZNF423

Senior-Loken Syndrome Panel

CEP290, INVS, IQCB1, NPHP1, NPHP3, NPHP4, SCLT1, SDCCAG8, TRAF3IP1

Pulmonology Test Panels

Pulmonary Artery Hypertension (PAH) Panel

ABCC8, ACVRL1, AQP1, ATP13A3, BMPR1B, BMPR2, CAV1, EIF2AK4, ENG, FOXF1, GDF2, KCNA5, KCNK3, KLF2, NFU1, NOTCH3, RASA1, SARS2, SMAD4, SMAD9, SOX17, STRA6, TBX4

Interstitial Lung Disease Panel

ABCA3, CSF2RA, CSF2RB, DKC1, ELMOD2, FAM111B, FOXF1, HPS1, HPS4, ITGA3, NAF1, NF1, NKX2-1, PARN, RTEL1, SFTPA1, SFTPA2, SFTPB, SFTPC, SLC34A2, SLC7A7, SMPD1, STAT3, STRA6, TERC, TERT, TINF2, TMEM173, TSC1, TSC2

Bronchiectasis Panel

ARHGEF1, C11ORF70, CCDC39, CCDC40, CFTR, DNAAF1, DNAAF2, DNAH11, DNAH5, DNAH9, DNAI1, DNAI2, DNAL1, GAS2L2, MCIDAS, NME8, POLD1, RSPH4A, RSPH9, SCNN1A, SCNN1B, STK36

Central Hypoventilation and Apnea Panel

CHAT, CHRNA1, CHRN1, CHRNA2, CHRNA3, COLQ, EDN3, GLRA1, MECP2, PHOX2B, RAPSIN, RET, SCN4A, SLC6A5, ZEB2

Pulmonology Test Panels

Hermansky-Pudlak Syndrome Panel

ABCA3, AP3B1, AP3D1, BLOC1S3, BLOC1S6, DKC1, DTNBP1, GPR143, HPS1, HPS3, HPS4, HPS5, HPS6, LYST, OCA2, SFTPB, SFTPC, SLC45A2, TERC, TERT, TNF2, TYR, TYRP1

Cystic Lung Disease Panel

EFEMP2, ELN, FBLN5, FLCN, LTBP4, SERPINA1, TSC1, TSC2

Neonatal Respiratory Distress – Surfactant Dysfunction Panel

ABCA3, FOXF1, NKX2-1, SFTPB, SFTPC

Primary Ciliary Dyskinesia Panel

ARMC4, C11ORF70, C21ORF59, CCDC103, CCDC114, CCDC151, CCDC39, CCDC40, CCDC65, CCNO, CENPF, CFAP57, CFTR, DNAAF1, DNAAF2, DNAAF3, DNAAF5, DNAH1, DNAH11, DNAH5, DNAH8, DNAH9, DNAI1, DNAI2, DNAJB13, DNALI1, DRC1, DYX1C1, GAS2L2, GAS8, HYDIN, INVS, LRRC56, LRRC6, MCIDAS, NME8, OFD1, PIH1D3, RPGR, RSPH1, RSPH3, RSPH4A, RSPH9, SPAG1, STK36, TTC25, ZMYND10

Neurology Test Panels

Amyotrophic Lateral Sclerosis Panel

ALS2, ANG, ATL1, BSCL2, CHCHD10, CHMP2B, DCTN1, FIG4, FUS, GBE1, GRN, HEXA, HNRNP1, HSPD1, KIAA0196, KIF5A, MATR3, OPTN, PFN1, PRF1, REEP1, SETX, SLC52A2, SLC52A3, SOD1, SPAST, SPG11, SPG20, SQSTM1, TARDBP, TIA1, TUBA4A, UBQLN2, VAPB, VCP

Dementia Panel

ABCA7, APOE, APP, CHMP2B, CSF1R, FUS, GRN, MAPT, PRNP, PSEN1, PSEN2, RNF216, SIGMAR1, SNCA, SORL1, TARDBP, TREM2, TUBA4A, UBE3A, UBQLN2, VCP

Charcot-Marie-Tooth Panel

AARS, AGTPBP1, AIFM1, AMACR, ARHGEF10, ATAD3A, ATL1, ATL3, ATP1A1, ATP7A, BAG3, BICD2, BSCL2, C12ORF65, CCT5, CHCHD10, COA7, COX10, COX6A1, CTDP1, CYP27A1, DCAF8, DCTN1, DHTKD1, DNAJB2, DNMT2, DNMT1, DST, DYNC1H1, EGR2, FAM134B, FBLN5, FBXO38, FGD4, FIG4, FXN, GAN, GARS, GDAP1, GJB1, GNB4, GNE, GSN, HADHB, HARS, HINT1, HK1, HSPB1, HSPB8, IGHMBP2, IKBKAP, INF2, KARS, KIF1A, KIF1B, KIF5A, LDB3, LITAF, LMNA, LRSAM1, MARS, MCM3AP, MED25, MFN2, MME, MORC2, MPV17, MPZ, MTMR2, MYOT, NDRG1, NEFH, NEFL, NGF, NTRK1, PDK3, PLEKHG5, PMP2, PMP22, PNKP, POLG, POLG2, PRDM12, PRPS1, PRX, RAB7A, REEP1, SACS, SBF1, SBF2, SCN11A, SCN9A, SCYL1, SEPT9, SETX, SH3TC2, SLC12A6, SLC25A46, SLC52A2, SLC52A3, SMAD3, SPG11, SPTBN4, SPTLC1, SPTLC2, SURF1, TFG, TRIM2, TRPV4, TTR, TYMP, UBA1, VCP, WNK1, YARS, ZFYVE26

Ataxia Panel

AARS2, ABCA2, ABCB7, ABCD1, ABHD12, ACO2, ADCK3, ADPRHL2, AFG3L2, AGTPBP1, AH1I, ALDH5A1, ANO10, APTX, ARL13B, ARL6, ATCAY, ATM, ATP13A2, ATP1A3, ATP2B3, ATP7B, ATP8A2, AUH, BBS1, BBS10, BBS12, BBS2, BBS4, BBS5, BBS7, BBS9, BEAN1, C10ORF2, C12ORF4, C5ORF42, CA8, CACNA1A, CACNA1G, CACNA2D2, CACNB4, CAMTA1, CAPN1, CASK, CC2D2A, CCDC88C, CEP290, CEP41, CHCHD10, CLCN2, CLN5, CLN6, CLN8, CLPB, CLPP, COA7, COASY, COQ2, COQ4, COX20, CP, CSTB, CTBP1, CTDP1, CTSA, 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, MECP, MGME1, MKKS, MKS1, MME, MRE11A, MSTO1, MTFMT, MTPAP, MTPP, NDUFAF6, NDUFS2, NDUFS4, NDUFS7, NDUFS8, NDUFV1, NKX2-1, NKX6-2, 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, SLC25A15, SLC25A46, SLC2A1, SLC52A2, SLC9A1, SLC9A6, SNX14, SPG11, SPG20, SPG7, SPTBN2, SQSTM1, STUB1, STXBPI, SUOX, SYNE1, SYT14, TCTN1, TCTN2, TCTN3, TDP1, TDP2, TGM6, TMEM138, TMEM216, TMEM231, TMEM237, TMEM240, TMEM67, TPP1, TRAPPC11, TRIM32, TTBK2, TTC19, TTC8, TTPA, TUBB4A, UBA5, UBTF, UCHL1, VAMP1, VLDLR, VPS13D, VWA3B, WDCPC, WDR81, WFS1, WWOX, XRCC1, ZFYVE26, ZNF423

Dystonia Panel

ADCY5, ANO3, ATP1A3, BCAP31, CACNA1B, CACNA1G, DCAF17, DNAJC12, FA2H, FITM2, GCH1, GNAL, KCNMA1, KMT2B, MECP, MIPER, PDE10A, PDGFB, PDGFRB, PNKD, PRKRA, PRRT2, SGCE, SLC2A1, SLC39A14, SPR, TH, THAP1, TOR1A, UBTF, VAC14

Emery-Dreifuss Muscular Dystrophy Panel

DMD, EMD, FHL1, LMNA, TMEM43, TTN

Neurology Test Panels

Macrocephaly / Overgrowth Syndrome Panel

AKT1, AKT3, ASPA, ASXL2, BRWD3, CCND2, CDKN1C, CHD8, CUL4B, DHCR24, DIS3L2, DNMT3A, EED, EIF2B5, EZH2, GFAP, GLI3, GPC3, GPSM2, GRIA3, HEPACAM, HUWE1, KDM1A, KIAA0196, KIF7, KPTN, L1CAM, MED12, MLC1, MPDZ, NFIB, NFIX, NSD1, OFD1, PIGA, PIK3CA, PIK3R2, PTCH1, PTEN, RAB39B, RNF135, SETD2, SYN1, TMEM94, TSC1, TSC2, UPF3B, ZBTB20

Metabolic Epilepsy Panel

ABAT, ADSL, AGA, ALDH5A1, ALDH7A1, AMT, ARG1, BTD, D2HGDH, DHFR, DNM1L, DPYD, DPYS, ETFA, ETFB, ETFDH, ETHE1, FH, GAMT, GCDH, GCH1, GLDC, GNE, GPHN, HIBCH, L2HGDH, MIPER, MOCS1, MTHFR, NDUFAF3, NDUFS6, PGK1, PNPO, POLG, PRODH, PTS, QDPR, SERAC1, SLC25A1, SLC25A15, SLC25A42, SLC2A1, SLC39A8, SLC46A1, SUOX, TPK1, WARS2

Spastic Paraplegia Panel

ABCD1, AFG3L2, ALDH18A1, ALS2, AP4B1, AP4E1, AP4M1, AP4S1, AP5Z1, ARG1, ARL6IP1, ATAD3A, ATIL1, B4GALNT1, BSCL2, BTD, C12ORF65, C19ORF12, CACNA1G, CAPN1, COASY, CTNNB1, CYP27A1, CYP2U1, CYP7B1, DARS, DDHD1, DDHD2, ERLIN2, FA2H, FARS2, FXN, GALT, GBA2, GBE1, GCH1, GJC2, GPT2, HACE1, HSPD1, IBA57, IRF2BPL, KDM5C, KIAA0196, KIDINS220, KIF1A, KIF5A, L1CAM, L2HGDH, MARS2, NIPA1, NKX6-2, NT5C2, PAH, PLP1, PNPLA6, RARS, REEP1, RTN2, SACS, SETX, SLC16A2, SLC1A4, SLC25A15, SLC33A1, SPAST, SPG11, SPG20, SPG7, SPR, TECPR2, TFG, TH, UBAP1, ZFYVE26

Epilepsy Panel

AARS, ABAT, ABCA2, ABCD1, ACTL6B, ACY1, ADAM22, ADAR, ADNP, ADPRHL2, ADSL, AFG3L2, AGA, AIFM1, AIMP1, ALDH3A2, ALDH5A1, ALDH7A1, ALG13, ALG6, ALKBH8, AMACR, AMT, ANKRD11, AP2M1, AP3B2, AP4B1, AP4E1, AP4M1, AP4S1, APOPT1, ARG1, ARHGEF9, ARID1B, ARSA, ARV1, ARX, ASAH1, ASNS, ASPA, ASXL3, ATAD1, ATP13A2, ATP1A1, ATP1A2, ATP1A3, ATP6V1A, ATRX, BCKDK, BRAT1, BTD, C12ORF57, CACNA1A, CACNA1B, CACNA1D, CACNA1E, CACNA1G, CACNA1H, CACNA2D2, CACNB4, CAD, CAMK2B, CARS2, CASK, CASR, CC2D1A, CDK9, CDKL5, CERS1, CHD2, CHRNA2, CHRNA4, CHRN2, CLCN2, CLCN4, CLN3, CLN5, CLN6, CLN8, CLTC, CNKSR2, CNPY3, CNTNAP2, COA7, COL4A1, COL4A2, COL4A3BP, COQ2, COQ4, COX15, COX6B1, CPLX1, CPT2, CSF1R, CSNK2B, CSTB, CTC1, CTSD, CTSE, CUL4B, CUX2, CYFIP2, CYP27A1, D2HGDH, DARS, DARS2, DCX, DDC, DDX3X, DEAF1, DEGS1, DENND5A, DEPD5, DHDDS, DHFR, DHPS, DIAPH1, DMXL2, DNAJC5, DNM1, DNM1L, DOCK7, DOLK, DPAGT1, DPM1, DPM2, DPYD, DPYS, DYNC1H1, DYRK1A, EARS2, ECHS1, ECM1, EEF1A2, EFHC1, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EIF3F, EML1, EPM2A, EPRS, ETFA, ETFB, ETFDH, ETHE1, FA2H, FAM126A, FAR1, FARS2, FDF1, FDX1L, FGF12, FH, FKTN, FLNA, FOLR1, FOXG1, FOXRED1, FRRS1L, FUT8, GABBR2, GABRA1, GABRB1, GABRB2, GABRB3, GABRG2, GALT, GAMT, GCDH, GCH1, GCSH, GFAP, GFM1, GFM2, GJC2, GLB1, GLDC, GLRB, GLS, GLUD1, GNAO1, GNB1, GNE, GOLGA2, GOSR2, GPAA1, GPHN, GRIA3, GRIA4, GRIK2, GRIN1, GRIN2A, GRIN2B, GRIN2D, GRN, GTPBP3, GUF1, HACE1, HCN1, HCN2, HECW2, HEPACAM, HIBCH, HNRNP, HSD17B10, HSPD1, HTRA1, HTT, IBA57, ICK, IER3IP1, IFIH1, IQSEC2, IRF2BPL, ITPA, KCNA1, KCNA2, KCNB1, KCNC1, KCNH1, KCNJ10, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KCTD7, KDM5C, KIAA1715, KIAA2022, KIF1A, KIF5A, KIF5C, KMT2E, L2HGDH, LGI1, LIAS, LMNB1, LMNB2, LRPPRC, LYRM7, MACF1, MAGI2, MARS2, MBD5, MBOAT7, MDH2, MECP2, MED12, MED17, MEF2C, MFS2, MIPER, MLC1, MOCS1, MOCS2, MRPL44, MTFMT, MTHFR, MTOR, NACC1, NBEA, NDS1, NDUFAF3, NDUFAF5, NDUFAF6, NDUFS2, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NECAP1, NEU1, NEUROD2, NFU1, NHLRC1, NKX6-2, NOTCH3, NPRL2, NPRL3, NR2F1, NRXN1, NSDHL, NT5C2, NTRK2, NUBPL, NUS1, OCLN, OFD1, OPHN1, P4HTM, PACS1, PACS2, PAFAH1B1, PARS2, PCDH19, PEX1, PEX10, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PGK1, PHACTR1, PHF6, PIGA, PIGB, PIGC, PIGG, PIGN, PIGO, PIGR, PIGQ, PIGS, PIGT, PIGV, PIGW, PITRM1, PLAA, PLCB1, PLP1, PNKP, PNPO, POLG, POLR3A, POLR3B, PPP2CA, PPP3CA, PPT1, PRICKLE1, PRICKLE2, PRIMA1, PRODH, PROSC, PRRT2, PRUNE, PSAP, PSAT1, PTPN23, PTS, PUM1, PURA, PYCR2, QARS, QDPR, RAB11A, RAB11B, RAB39B, RALA, RARS, RELN, RHOTB2, RMND1, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF13, RNF216, ROGDI, RORA, RORB, RUSC2, SAMHD1, SCARB2, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCN9A, SCO1, SDHAF1, SERAC1, SERPIN1, SETBP1, SETD1B, SGSH, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC1A2, SLC1A4, SLC25A1, SLC25A15, SLC25A22, SLC25A42, SLC2A1, SLC35A1, SLC35A2, SLC39A8, SLC46A1, SLC6A1, SLC6A5, SLC6A8, SLC9A6, SMARCA2, SMC1A, SMS, SNAP25, SNORD118, SOX10, SPATA5, SPTAN1, SPTBN4, SSR4, ST3GAL3, ST3GAL5, STRADA, STX1B, STXBP1, SUMF1, SUOX, SYN1, SYNGAP1, SYNJ1, SZT2, TAF1, TANGO2, TBC1D20, TBC1D24, TBCD, TBCE, TBCK, TBL1XR1, TCF4, TK2, TPK1, TPP1, TRAK1, TREX1, TRIM8, TRIT1, TSC1, TSC2, TSFM, TTC19, TUBA1A, TUBB2A, TUBB2B, TUBB4A, UBA5, UBE2A, UBE3A, UBTF, UNC80, VAMP2, VARS, VPS13A, WARS2, WASF1, WDR26, WDR45, WWOX, YWHAG, YY1, ZDHHC9, ZEB2, ZFYVE26, ZNHIT3, ZSWIM6

Epileptic Encephalopathy Panel

ABAT, ACTL6B, ADAM22, ADAR, ADPRHL2, ADSL, ALDH7A1, ALG13, AMT, AP2M1, AP3B2, APOPT1, ARHGEF9, ARX, ASNS, ATP6V1A, BRAT1, CACNA1A, CACNA1B, CACNA1E, CASK, CDKL5, CHD2, CLCN4, CLTC, CNKSR2, CNPY3, CNTNAP2, COX6B1, CPT2, CYFIP2, D2HGDH, DCX, DENND5A, DNM1, DNM1L, DOCK7, ECHS1, EEF1A2, ETHE1, FAR1, FARS2, FGF12, FLNA, FOXG1, FRRS1L, GABBR2, GABRA1, GABRB2, GABRB3, GABRG2, GAMT, GLDC, GNAO1, GPHN, GRIN1, GRIN2A, GRIN2B, GTPBP3, HCN1, HECW2, HEPACAM, HIBCH, HNRNP, HTT, KCNA2, KCNB1, KCNMA1, KCNQ2, KCNQ3, KCNQ5, KCNT1, KCNT2, KCTD3, KIF1A, LRPPRC, LYRM7, MBD5, MDH2, MECP2, MED17, MEF2C, MOCS1, MRPL44, MTFMT, MTHFR, NACC1, NDUFAF6, NDUFS2, NDUFS4, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NECAP1, NEUROD2, NRXN1, NUBPL, PARS2, PCDH19, PHACTR1, PIGA, PIGB, PIGR, PIGQ, PIGS, PLAA, PLCB1, PNKP, PNPO, POLG, PPP3CA, PROSC, PTPN23, PURA, RMND1, RNASEH2A, RNASEH2B, ROGDI, SAMHD1, SCN1A, SCN1B, SCN2A, SCN3A, SCN8A, SCO1, SDHAF1, SERAC1, SIK1, SLC12A5, SLC13A5, SLC19A3, SLC25A1, SLC25A22, SLC2A1, SLC35A2, SLC6A8, SLC9A6, SNAP25, SPTAN1, ST3GAL3, ST3GAL5, STXBP1, SYN1, SYNGAP1, SYNJ1, SZT2, TBC1D24, TBCD, TBCE, TBCK, TCF4, TRAK1, TREX1, TRIM8, TSC1, TSC2, TTC19, UBA5, UBE3A, UNC80, VARS, WARS2, WDR45, WWOX, ZEB2

Neurology Test Panels

Creatine Metabolism Deficiency Panel	GAMT, GATM, SLC6A8	LGMD and Congenital Muscular Dystrophy Panel	ANO5, B3GALNT2, BICD2, CAPN3, CAV3, CHKB, COL4A1, COL4A2, CRYAB, DES, DMD, DNAJB6, DYSE, FKRP, FKTN, GAA, GMPPB, GOLGA2, INPP5K, ISPD, ITGA7, LAMA2, LARGE, LIMS2, LMNA, MAP3K20, MEGF10, MSTO1, MYH7, MYO18B, MYOT, PNPLA2, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PYROXD1, RYR1, SELENON, SGCA, SGCB, SGCD, SGCG, SMCHD1, SPEG, SPTBN4, SYNE1, TCAP, TNPO3, TOR1AIP1, TRAPPC11, TRIM32, TTN, VMA21
Lissencephaly Panel	ACTB, ACTG1, ARX, ATP6V0A2, CIT, CRADD, DCX, FKTN, ISPD, KATNB1, KIAA1109, LAMB1, LARGE, MACF1, PAFAH1B1, POMGNT2, POMT1, RELN, TMTC3, TUBA1A, TUBB2B, TUBG1, VLDLR, YWHAЕ	Leukodystrophy and Leukoencephalopathy Panel	ABCD1, ADAR, AIFM1, AIMP1, ALDH3A2, AP4B1, AP4E1, AP4M1, AP4S1, APOPT1, ARSA, ASPA, CLCN2, COA7, COL4A1, COX15, COX6B1, CSF1R, CTC1, CYP27A1, D2HGDH, DARS, DARS2, DEGS1, EARS2, EIF2B1, EIF2B2, EIF2B3, EIF2B4, EIF2B5, EPRS, FA2H, FAM126A, FDX1L, FOLR1, FOXRED1, GALT, GFAP, GFM1, GJC2, HEPACAM, HIBCH, HSPD1, HTRA1, IBA57, L2HGDH, LMNB1, LYRM7, MARS2, MLC1, MRPL44, MTFMT, NDUFAF5, NFU1, NKX6-2, NOTCH3, NT5C2, NUBPL, PLP1, POLR3A, POLR3B, PSAP, PYCR2, RARS, RNASEH2A, RNASEH2B, RNASEH2C, RNASET2, RNF216, SAMHD1, SCO1, SDHAF1, SERAC1, SLC1A4, SNORD118, SOX10, SUMF1, TREX1, TTC19, TUBB4A, ZFYVE26
Metabolic Myopathy and Rhabdomyolysis Panel	ACAD9, ACADL, ACADM, ACADVL, ADCK3, AGL, AHCY, ALDOA, AMPD1, ANO5, ATP2A1, B3GALNT2, B4GAT1, C10ORF2, CAPN3, CASQ1, CAV3, CHKB, COQ2, CPT2, CTDP1, DAG1, DGUOK, DMD, DNAJB6, DPM1, DPM2, DYSE, EMD, ENO3, ETFA, ETFB, ETFDH, FDX1L, FHL1, FKRP, FKTN, FLAD1, GAA, GBE1, GMPPB, GYG1, GYS1, HADHA, HADHB, ISCU, LAMA2, LAMP2, LARGE, LDHA, LPIN1, MYH3, OPA1, OPA3, PDSS2, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKB, PNPLA2, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PYGM, RBCK1, RRM2B, RYR1, SCN4A, SGCA, SGCB, SGCD, SGCG, SIL1, SLC22A5, SLC25A20, STAC3, SUCLA2, SUCLG1, TANGO2, TCAP, TK2, TNPO3, TRIM32, TYMP		
Microcephaly and Pontocerebellar Hypoplasia Panel	AKT3, AMPD2, ASPM, ASXL1, ASXL3, ATR, CASK, CCDC47, CDK5RAP2, CENPE, CENPJ, CEP152, CEP63, CSNK2A1, DONSON, DYNC1H1, DYRK1A, EFTUD2, EXOSC3, GFM1, GPT2, KANSL1, KATNB1, KIF11, LIG4, MBD5, MCPH1, MED17, MFSD2A, MIEP, MRE11A, MYCN, MYO18B, NCAPD3, NDE1, NHEJ1, OPHN1, PAFAH1B1, PCDH12, PCLO, PCNT, PHGDH, PLK4, PNKP, POMT1, PQBP1, QARS, RARS2, RTTN, SEPECS, SLC1A4, SMARCA2, SMARCE1, SOX11, STAG2, STAMBP, STIL, TBC1D20, TBC1D23, THOC6, TMTC3, TOE1, TOP3A, TRMT10A, TSEN2, TSEN54, TUBB, TUBB2B, TUBGCP4, TUBGCP6, UBE3B, VARS, VRK1, WDR62, WDR73, XRCC4, ZNF148, ZNF335		
Holoprosencephaly Panel	CDON, FGF8, FGFR1, FOXH1, GLI2, GLI3, NODAL, PTCH1, SHH, SIX3, TGIF1, ZIC2	Coenzyme q10 deficiency Panel	ADCK3, ANO10, APTX, COQ2, COQ4, COQ5, COQ6, COQ7, COQ9, ETFA, ETFB, ETFDH, PDSS1, PDSS2, SLC25A26
Collagen Type VI-Related Disorders Panel	COL12A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3	Congenital Myasthenic Syndromes Panel	AGRN, CHAT, CHRNA1, CHRNB1, CHRND, CHRNE, CHRNG, COLQ, DOK7, DPAGT1, FLAD1, GFPT1, LAMB2, MUSK, MYO9A, PLEC, PREPL, RAPSN, SCN4A, STIM1, SYT2
Septo-Optic Dysplasia Panel	HESX1, OTX2, PAX6, SOX2	Cerebral Cavernous Malformation Panel	CCM2, KRIT1, PDCD10, RASA1

Neurology Test Panels

X-linked Intellectual Disability Panel	ABCD1, ACSL4, AFF2, AP1S2, ARHGEF6, ARHGEF9, ARX, ATP6AP2, ATP7A, ATRX, BCOR, BRWD3, CASK, CDKL5, CLCN4, CNKSR2, CUL4B, DCX, DDX3X, DKC1, DLG3, ELK1, FANCB, FGD1, FLNA, FMR1, FRMPD4, FTSJ1, GDI1, GK, GPC3, GRIA3, HCCS, HDAC8, HPRT1, HSD17B10, HUWE1, IDS, IGBP1, IL1RAPL1, IQSEC2, KDM5C, KIAA2022, KLF8, L1CAM, LAMP2, MAGT1, MAOA, MBTPS2, MECP2, MED12, MID1, MSL3, MTM1, NDP, NDUFA1, NHS, NLGN3, NLGN4X, NONO, NSDHL, NXF5, OCRL, OFD1, OPHN1, OTC, PAK3, PCDH19, PDHA1, PGK1, PHF6, PHF8, PLP1, PORCN, PQBP1, PRPS1, PTCHD1, RAB39B, RBM10, RLIM, RPL10, RPS6KA3, SHROOM4, SLC16A2, SLC6A8, SLC9A6, SMC1A, SMS, SOX3, SRPX2, SYN1, SYP, TAF1, THOC2, TIMM8A, TSPAN7, UBE2A, UPF3B, USP9X, ZC4H2, ZCCHC12, ZDHHC9, ZNF41, ZNF674, ZNF711, ZNF81		
Nemaline Myopathy Panel	ACTA1, BIN1, CFL2, KBTBD13, KLHL40, KLHL41, LMOD3, MTM1, MYO18B, NEB, TNNT1, TPM2, TPM3	Otizm Spektrum Bozukları Paneli <i>Autism Spectrum Disorders Panel</i>	ADNP, BCL11A, C12ORF4, CACNA1C, CC2D1A, CNOT3, CNTN6, COL4A3BP, CSNK2A1, CTNND2, DHCR7, EHMT1, EN2, FBXO11, FOXP1, GAMT, KMT2E, KMT5B, MBOAT7, MECP2, NBA, NFIB, NLGN3, NLGN4X, NSD1, POGZ, PTCHD1, PTEN, RPL10, SHANK3, TBRI, TCF20, TRIP12, TSC1, TSC2, VAMP2, WASF1, ZSWIM6
Migraine Panel	ATP1A2, ATP1A3, CACNA1A, KCNK18, NOTCH3, POLG, PRRT2, SCN1A, SLC1A3, SLC2A1		
Comprehensive Muscular Dystrophy / Myopathy Panel	ACAD9, ACADVL, ACTA1, ADCK3, ANO5, ATP2A1, B3GALNT2, B4GAT1, BAG3, BICD2, BIN1, CAPN3, CASQ1, CAV3, CFL2, CHKB, CNTN1, COL12A1, COL4A1, COL4A2, COL6A1, COL6A2, COL6A3, COQ2, CRYAB, DAG1, DES, DGUOK, DMD, DNAJB6, DPM3, DYSE, EMD, FDX1L, FHL1, FKRP, FKTN, FLAD1, FLNC, GAA, GBE1, GMPPB, GOLGA2, GYG1, HNRNPDL, INPP5K, ISCU, ISPD, ITGA7, KBTBD13, KLHL40, KLHL41, LAMA2, LAMP2, LARGE, LDB3, LIMS2, LMNA, LMOD3, MAP3K20, MEGF10, MICU1, MME, MSTO1, MTM1, MYH7, MYO18B, MYOT, NEB, PABPN1, PGK1, PHKA1, PHKB, PLEC, PNPLA2, POGUT1, POLG, POLG2, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PYGM, PYROXD1, RBCK1, RRM2B, RYR1, SCN4A, SELENON, SEPT9, SGCA, SGCB, SGCD, SGCG, SIL1, SLC22A5, SLC25A20, SMCHD1, SMN1, SMN2, SPEG, SPTBN4, STAC3, SUCLA2, SUCLG1, SYNE1, TANGO2, TCAP, TIA1, TMEM126B, TMEM43, TNNT1, TNPO3, TOR1AIP1, TPM2, TPM3, TRAPPC11, TRIM32, TSFM, TTN, TYMP, VMA21, VPS13A		
NCL and Progressive Myoclonic Epilepsy Panel	AFG3L2, ASAH1, ATP13A2, BRAT1, CERS1, CLN3, CLN5, CLN6, CLN8, CSTB, CTSD, CTSE, DNAJC5, EPM2A, FARS2, FOLR1, GABRB2, GOSR2, GRN, KCNC1, KCTD7, MFSD8, NEU1, NHLRC1, POLG, PPT1, PRICKLE1, SCARB2, SERPINI1, TBC1D24, TPP1	Neuronal Migration Disorder Panel	ACTB, ACTG1, ADGRG1, AKT3, ARFGEF2, ARX, ATP6V0A2, B3GALNT2, COL4A1, COL4A2, DCX, DYNC1H1, EMX2, FAT4, FH, FKTN, FLNA, FLVCR2, GMPPB, GPM2, ISPD, KATNB1, KIF1BP, KIF7, L1CAM, LAMA2, LAMB1, LAMC3, LARGE, MACF1, MED12, MEF2C, MPDZ, NDE1, NSDHL, OCLN, PAFAH1B1, PHGDH, PIK3CA, PIK3R2, POMGNT2, POMT1, RAB18, RAB3GAP1, RAB3GAP2, RELN, RTTN, SEPSEC, SRPX2, TMEM5, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, TUBG1, VLDLR, WDR62, YWHAE
Neuro-Ophthalmology Panel	ACO2, AFG3L2, ANTXR1, APTX, ATAD3A, AUH, C10ORF2, C12ORF65, C19ORF12, CHN1, CISD2, DNAJC19, DNM1L, FDXR, FRMD7, GPR143, HESX1, ISCA2, KIF21A, MECP, MFN2, MGME1, MTPAP, NARS2, NDUFAF3, NDUFS1, NR2F1, OPA1, OPA3, OTX2, PAX6, PDSS1, PHOX2A, POLG, PRPS1, ROBO3, RRM2B, RTN4IP1, SALL4, SETX, SLC19A2, SLC19A3, SLC25A4, SLC25A46, SLC38A8, SLC52A2, SNX10, SOX2, SPG7, SUCLA2, TIMM8A, TK2, TMEM126A, TSFM, TUBB3, TYMP, UCHL1, WFS1, YME1L1, ZNHIT3		
Parkinson Disease Panel	ATP1A2, ATP1A3, ATP7B, C19ORF12, CHCHD10, CHCHD2, CP, CSF1R, DCTN1, DNAJC12, DNAJC5, DNAJC6, FBXO7, FTL, GBA, GCH1, GRN, LRRK2, MAPT, NUS1, PANK2, PARK2, PARK7, PDE10A, PDE8B, PDGFB, PDGFRB, PINK1, PLA2G6, POLG, PRKRA, PSEN1, RAB39B, SLC20A2, SLC30A10, SLC39A14, SLC6A3, SNCA, SPR, SYNJ1, TH, VPS13A, VPS13C, VPS35, XPR1		
Porphyria Panel	ALAD, ALAS2, CPOX, FECH, HFE, HMBS, PPOX, UROD, UROS	Tuberous Sclerosis Panel	TSC1, TSC2
Periodic Paralysis Panel	CACNA1S, CLCN1, KCNJ2, SCN4A	Polymicrogyria Panel	ADGRG1, AKT3, FH, GPM2, KIF1BP, LAMC3, NDE1, NSDHL, OCLN, PI4KA, RAB18, SNAP29, SRPX2, TBC1D20, TUBA1A, TUBA8, TUBB2A, TUBB2B, TUBB3, WDR62