AMINO ACID DISORDERS		
PANEL	**SUBPANEL**	**GENES**
Homocystinuria	**Hypermethioninemia**	*ADK, AHCY, CBS, GNMT, MAT1A, SLC25A13*
	Hypomethioninemia	*MTHFR, MTR, MTRR*
Phenylketonuria	**PAH Deficiency**	*PAH* (sequencing + reflex MLPA as needed)
	Biopterin Deficiencies	*DNAJC12, GCH1, PCBD1, PTS, QDPR, SPR* (for PKU panel, PAH will be done with reflex to these genes)
Tyrosinemia	**Elevated Succinylacetone**	*FAH, GSTZ1*
	Elevated Tyrosine	*HPD, TAT*
Urea Cycle Diseases	**High citrulline**	*ASS1, SLC25A13*
	High ASA	*ASL*
	Low citrulline	*CPS1, NAGS, OTC*
	Other	*ARG1, CA5A, GLUL, GLUD1, OAT, SLC7A7, SLC25A2, SLC25A15*
Maple Syrup Urine Disease		*BCKDHA, BCKDHB, DBT, DLD*

ORGANIC ACID DISORDERS		
PANEL	**SUBPANEL**	**GENES**
Multiple carboxylase Deficiency	**Biotinidase Deficiency**	*BTD*
	Other	*CA5A, HLCS*
Propionic / Methylmalonic acidemias	**PA**	*PCCA, PCCB*
	MMA	*ACSF3, ALDH6A1, MCEE, MLYCD, MMAA, MMAB, MMUT, SUCLA2, SUCLG1*
	MMA + Homocysteinemia	*ABCD4, AMN, CBLIF, CD320, CUBN, HCFC1, LMBRD1, MMACHC, MMADHC, TCN1, TCN2*
Isovaleric acidemia		*IVD, ACADSB, FLAD1*
Glutaric aciduria Type 1		*GCDH*
Isobutyryl-CoA dehydrogenase deficiency		*ACAD8*
Succinic semialdehyde dehydrogenase deficiency		*ALDH5A1*
b-ketothiolase deficiency		*ACAT1*
Guanidinoacetate Methyltransferase Deficiency		*GAMT*

FATTY ACID OXIDATION DISORDERS	
PANEL	**GENES**
Carnitine Uptake Deficiency	*SLC22A5*
MCAD Deficiency	*ACADM (c.985A>G common mutation and del/dup +/- reflex sequencing)*
LCHAD/MTP Deficiency	*HADHA, HADHB (HADHA c.1528G>C + reflex sequencing)*
VLCAD Deficiency	*ACADVL*
MADD/Glutaric Aciduria Type2	*ETFA, ETFB , ETFDH, FLAD1, SLC52A2, SLC52A3, SLC52A1*
CPT2 Deficiency	*CPT2*
CACT Deficiency	*SLC25A20*
CPT1 Deficiency	*CPT1A (p.Pro479Leu common mutation + reflex sequencing)*
Other FAOD	*ACAA2, ACAD9, ACADL, ACADS, ECHS1, HADH*

CONGENITAL ADRENAL HYPERPLASIA	
21-Hydroxylase Deficiency	*CYP21A2* (includes MLPA and long-range PCR analyses for CNVs and common rearrangements)
Other	*ARMC5, CYP11B1, CYP11B2, CYP17A1, HSD3B2, POR, PRKAR1A, STAR*

GALACTOSEMIA	
GALT Deficiency	*GALT*
Other	*GALK1, GALE, GALM, GLUT2 (SLC2A2)*

INBORN ERRORS OF IMMUNITY	
ADA Deficiency	*ADA*
Chronic Granulomatous Disease	*CYBA, CYBB, CYBC1, NCF1*, NCF2, NCF4, G6PD (*limited coverage due to high homology with duplicated regions in genome)*

Severe Combined/Primary Immune Deficiency Panel (251 genes)

(Augmented exome slice with Sanger backfill of underlined genes + reflex MLPA of *DOCK8* as needed)

ACD, ACP5, ADA, ADA2, ADAM17, ADAR, AICDA, AIRE, AK2, AP3B1, ARHGEF1, ARPC1B, ATM, ATP6AP1, B2M, BACH2, BCL10, BCL11B, BLM, BLNK, BTK, CYBC1, C1QA, C1QB, C1QC, C1S, C2, C3, CARD11, CARD14, CARD9, CARMIL2, CASP10, CASP8, CD19, CD247, CD27, CD3D, CD3E, CD3G, CD40, CD40LG, CD70, CD79A, CD79B, CD81, CD8A, CDCA7, CFD, CFI, CFP, CHD7, CIITA, COPA, CR2, CTLA4, CTPS1, CTSC, CXCR4, CYBA, CYBB, DBR1, DCLRE1C, DKC1, DNASE2, DNMT3B, DOCK2, DOCK8, EBF1, EPG5, ERCC6L2, EXTL3, FADD, FAS, FASLG, FCHO1, FERMT3, FOXN1, FOXP3, G6PD, GATA2, GFI1, GINS1, HELLS, ICOS, IFIH1, IFNAR2, IFNGR1, IFNGR2, IGHM, IGLL1, IKBKB, IKZF1, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL17RA, IL17RC, IL1RN, IL21, IL21R, IL23R, IL2RA, IL2RB, IL2RG, IL36RN, IL6ST, IL7R, IRAK4, IRF2BP2, IRF8, ISG15, ITGB2, ITK, JAK1, JAK3, KRAS, LAMTOR2, LAT, LCK, LIG1, LIG4, LRBA, LRRC8A, LYST, MAGT1, MALT1, MAP3K14, MEFV, MRTFA, MOGS, MSN, MTHFD1, MVK, MYD88, MYO5A, NBN, NCF2, NCF4, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NOP10, NRAS, NSMCE3, ORAI1, OTULIN, PARN, PEPD, PGM3, PIK3CD, PIK3R1, PLCG2, PMS2, PNP, POLD1, POLE, POLE2, PRF1, PRKCD, PRKDC, PSMB8, PSTPIP1, PTPRC, RAB27A, RAC2, RAG1, RAG2, RASGRP1, RBCK1, RELA, RELB, RFX5, RFXANK, RFXAP, RHOH, RIPK1, RMRP, RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RORC, RTEL1, SAMHD1, SBDS, SEMA3E, SERPING1, SH2D1A, SLC29A3, SLC35C1, SLC39A7, SLC7A7, SMARCAL1, SP110, SPINK5, SPPL2A, STAT1, STAT2, STAT3, STIM1, STING1, STK4, STX11, STXBP2, TAP1, TAP2, TAPBP, TCF3, TCN2, TERC, TERT, TFRC, TGFB1, TINF2, TMC6,

TMC8, TNFAIP3, TNFRSF13B, TNFRSF1A, TNFRSF4, TNFRSF9, TRAC, TRAF3IP2, TREX1, TRNT1, TTC37, TTC7A, TYK2, UNC13D, UNC93B1, UNG, USP18, WAS, WDR1, WIPF1, WRAP53, XIAP, ZAP70, ZBTB24, ZNF341

MITOCHONDRIAL DISEASES (AUGMENTED EXOME SLICES WITH SANGER BACKFILL OF UNDERLINED GENES LISTED)

Full Mitochondrial Disease Nuclear Gene Panel (425 genes)

AARS2, ABAT, ABCB7, ACACB, ACAD8, ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACLY, ACO2, ACSL5, ACSM3, ADAR, ADSL, AFG3L2, AGK, AGXT2, AIFM1, AK2, AKAP10, AKR7A2, ALDH18A1, ALDH1B1, ALDH5A1, ALDH6A1, ALDH7A1, ALG3, AMPD1, AMT, ANTXR1, AS3MT, ATIC, ATP1A3, ATP10D, ATP5F1A, ATP5F1B, ATP5F1C, ATP5F1D, ATP5F1E, ATP5MC1, ATP5MC2, ATP5MC3, ATP5ME, ATP5MF, ATP5MG, ATP5MGL, ATP5PO, ATP5PB, ATP5PD, ATP5PF, ATPAF1, ATPAF2, AUH, BCKDHA, BCKDHB, BCS1L, BOLA3, BTD, C1QBP, C19orf12, CA5A, CARS2, CCDC88A, CEP89, CHCHD10, CHDH, CHKB, CISD2, CLN3, CLPB, CLPP, CLYBL, COA1, COA3, COA4, COA5, COA6, COA7, COA8, COASY, COMT, COQ2, COQ4, COQ5, COQ6, COQ7, COQ8A, COQ8B, COQ9, COX10, COX11, COX14, COX15, COX16, COX17, COX18, COX19, COX20, COX4I1, COX4I2, COX5A, COX5B, COX6A1, COX6A2, COX6B1, COX6B2, COX6C, COX7A1, COX7A2, COX7B, COX7C, COX8A, CPT1A, CPT1B, CPT2, CYC1, CYCS, CYP11A1, CYP11B1, CYP11B2, D2HGDH, DARS2, DBT, DDAH1, DGUOK, DLAT, DLD, DNA2, DNAJC19, DNAJC30, DNM1L, DNMT1, EARS2, ECHS1, ELAC2, ERAL1, ETFA, ETFB, ETFDH, ETHE1, FA2H, FARS2, FASTKD2, FBXL4, FDX2, FDXR, FH, FLAD1, FOXRED1, FXN, GARS1, GATB, GATC, GATM, GCDH, GCSH, GDAP1, GFER, GFM1, GFM2, GLDC, GLRX5, GLS, GTPBP3, GYG2, HADHA, HADHB, HARS2, HCCS, HIBCH, HLCS, HMGCL, HMGCS2, HSD17B10, HSPA9, HSPD1, IARS2, IBA57, IDH2, IDH3A, IDH3B, ISCA1, ISCA2, ISCU, IVD, KARS1, KIF5A, KIF21A, KLC2, KYNU, L2HGDH, LARS1, LARS2, LIAS, LIPT1, LIPT2, LMBRD1, LONP1, LPIN1, LRPPRC, LYRM4, LYRM7, MARS2, MCEE, MDH2, MECR, MFF, MFN2, MGME1, MICOS13, MICU1, MIPEP, MLYCD, MMAA, MMAB, MMACHC, MPV17, MRM2, MRPL12, MRPL3, MRPL44, MRPS14, MRPS16, MRPS22, MRPS23, MRPS34, MRPS7, MTFMT, MTO1, MTPAP, MTRFR, MTRR, MMUT, NADK2, NARS2, NAXE, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA3, NDUFA5, NDUFA7, NDUFA8, NDUFA9, NDUFAB1, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF7, NDUFAF8 (C17ORF89), NDUFB1, NDUFB6, NDUFB10, NDUFB11, NDUFB2, NDUFB3, NDUFB4, NDUFB5, NDUFB7, NDUFB8, NDUFB9, NDUFC1, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NDUFV3, NFS1, NFU1, NR2F1, NSUN3, NUBPL, OPA1, OPA3, OXA1L, OXCT1, PANK2, PARS2, PC, PCCA, PCCB, PCK2, PDHA1, PDHB, PDHX, PDK3, PDP1, PDSS1, PDSS2, PET100, PET117, PHOX2A, PITRM1, PLA2G6, PLP1, PMPCA, PMPCB, PNPLA8, PNPT1, POLG, POLG2, PPA2, PRPS1, PTCD3, PTS, PUS1, QARS1, QDPR, QRSL1, RARS1, RARS2, RMND1, RNASEH1, ROBO3, RRM2B, RTN4IP1, SACS, SARS2, SCO1, SCO2, SCP2, SDHA, SDHAF1, SDHAF2, SDHAF3, SDHAF4, SDHB, SDHC, SDHD, SERAC1, SFXN4, SLC19A2, SLC19A3, SLC22A5, SLC25A1, SLC25A12, SLC25A13, SLC25A19, SLC25A20, SLC25A21, SLC25A26, SLC25A3, SLC25A32, SLC25A38, SLC25A4, SLC25A42, SLC25A46, SLC52A2, SLC52A3, SNX10, SPATA5, SPG7, SPR, STAR, SUCLA2, SUCLG1, SUOX, SURF1, TACO1, TAFAZZIN, TARS2, TCIRG1, TCN2, TIMM22, TIMM44, TIMM50, TIMM8A, TIMMDC1, TK2, TMEM126A, TMEM126B, TMEM65, TMEM70, TOMM20, TOP3A, TPK1, TRIT1, TRMT10C, TRMT5, TRMU, TRNT1, TSFM, TTC19, TUBB3, TUBB4A, TUFM, TUSC3, TWNK, TXN2, TYMP, UCHL1, UNG, UQCC1, UQCC2, UQCC3, UQCR10, UQCR11 , UQCRB, UQCRC1, UQCRC2, UQCRFS1, UQCRH, UQCRQ, VARS2, WARS2, WDR73, WFS1, YARS2, YME1L1

Mitochondrial Encephalopathy / Leigh Disease Panel (117 genes)

AARS2, ACAD9, ACO2, AFG3L2, AIFM1, ATP5F1E, ATPAF2, BCS1L, BOLA3, COQ2, COQ8A, COQ9, COX7A1, COX10, COX14, COX15, COX20, COX4I1, COX4I2, COX6B1, DARS2, DGUOK, DLAT, DLD, DNM1L, EARS2, ETFDH, ETHE1, FARS2, FASTKD2, FH, FOXRED1, GFER, GFM1, GFM2, HLCS, HSPD1, LARS2, LIAS, LMBRD1, LRPPRC, MARS2, MFN2, MPV17, MRPS16, MTFMT, MTPAP, NDUFA1, NDUFA10, NDUFA11, NDUFA12, NDUFA13, NDUFA2, NDUFA7, NDUFA8, NDUFA9, NDUFAF1, NDUFAF2, NDUFAF3, NDUFAF4, NDUFAF5, NDUFAF6, NDUFAF7, NDUFB6, NDUFS1, NDUFS2, NDUFS3, NDUFS4, NDUFS5, NDUFS6, NDUFS7, NDUFS8, NDUFV1, NDUFV2, NDUFV3, NFU1, NUBPL, PC, PDHA1, PDHB, PDHX, PDP1, PDSS1, PDSS2, PNPT1, POLG, RARS2, RMND1, RRM2B, SCO1, SCO2, SDHA, SDHAF1, SDHAF2, SDHB, SDHD, SERAC1, SLC19A3, SUCLA2, SUCLG1, SURF1, TACO1, TIMM44, TK2, TMEM70, TOMM20, TPK1, TRMU, TSFM, TTC19, TUFM, TUSC3, TWNK, TYMP, UQCRB, UQCRQ, YARS2

mtDNA Depletion And Deletion Panel (19 genes)

AGK, DGUOK, DNA2, FBXL4, GFER, MFN2, MGME1, MPV17, OPA1, OPA3, POLG, POLG2, RRM2B, SLC25A4, SUCLA2, SUCLG1, TK2, TYMP, TWNK

Progressive External Ophthalmoplegia (PEO) And Optic Atrophy (77 genes)

ACO2, AFG3L2, ALG3, ANTXR1, ATP1A3, AUH, C19orf12, C1QBP, CCDC88A, CISD2, CLN3, DGUOK, DNA2, DNAJC19, DNAJC30, DNM1L, DNMT1, FA2H, FDX2, FDXR, FH, GYG2, IBA57, ISCA2, KIF21A, KLC2, MECR, MFF, MFN2, MGME1, MICOS13, MTFMT, MTO1, MTPAP, MTRFR, NARS2, NDUFAF3, NDUFS1, NR2F1, OPA1, OPA3, PANK2, PDHX, PDSS1, PHOX2A, PLA2G6, PLP1, POLG, POLG2, PRPS1, RNASEH1, ROBO3, RRM2B, RTN4IP1, SLC19A2, SLC19A3, SLC25A4, SLC25A46, SLC52A2, SLC52A3, SNX10, SPG7, SUCLA2, TACO1, TCIRG1, TIMM8A, TK2, TMEM126A, TSFM, TUBB3, TUBB4A, TWNK, TYMP, UCHL1, WDR73, WFS1, YME1L1

Pyruvate Dehydrogenase Complex Deficiency (16 genes)

BOLA3, DLAT, DLD, LIAS, LIPT1, LIPT2, NFU1, PC, PDHA1, PDHB, PDHX, PDK3, PDP1, SLC19A2, SLC19A3, TPK1