Gene and Variant List for Panels Offered at NSO

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*
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		*ACADSB, FLAD1, IVD*
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*
SCAD Deficiency	*ACADS*
MCAD Deficiency	*ACADM* (sequencing + reflex MLPA as needed)
LCHAD/MTP Deficiency	*HADHA, HADHB*
VLCAD Deficiency	*ACADVL*
MADD/Glutaric Aciduria Type2	*ETFA, ETFB, ETFDH, FLAD1, SLC52A1, SLC52A2, SLC52A3*
CPT2 Deficiency	*CPT2*
CACT Deficiency	*SLC25A20*
CPT1 Deficiency	*CPT1A*
ECHS1 Deficiency	*ECHS1*
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*
PEROXISOMAL STORAGE DISORDERS	
XL-Adrenoleukodystrophy	*ABCD1*
Zellweger Spectrum	*ACBD5, ACOX1, HSD17B4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7*
OTHER GENETIC DISORDERS	
Cystic Fibrosis	*CFTR* common mutation panel only: see table below for complete list of variants
Spinal Muscular Atrophy	*SMN1* and *SMN2* copy number only
INBORN ERRORS OF IMMUNITY	
ADA Deficiency	*ADA*
Aicardi-Goutières syndrome	*ADAR, IFIH1, RNASEH2A, RNASEH2B, RNASEH2C, SAMHD1, TREX1*
Chronic Granulomatous Disease	*CYBA, CYBB, CYBC1, NCF1*, NCF2, NCF4, G6PD (*limited coverage due to high homology with duplicated regions in genome)*

Last update June 2026

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, 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, CYBC1, 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 (437 genes)

AARS2, ABAT, ABCB7, ACACB, ACAD8, ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACLY, ACO2, ACSL5, ACSM3, ADAR, ADSL, AFG3L2, AGK, AGL, 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, GAA GARS1, GATB, GATC, GATM, GBE1, GCDH, GCSH, GDAP1, GFER, GFM1, GFM2, GLDC, GLRX5, GLS, GTPBP3, GYG2, GYS1, 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, LDHA, 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, PFKM, PGAM2, PGM1, PHKA1, PHKB, PHOX2A, PITRM1, PLA2G6, PLP1, PMPCA, PMPCB, PNPLA8, PNPT1, POLG, POLG2, PPA2, PRPS1, PTCD3, PTS, PUS1, PYGM, 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, SLC16A1, 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 Panel (16 genes)
BOLA3, DLAT, DLD, LIAS, LIPT1, LIPT2, NFU1, PC, PDHA1, PDHB, PDHX, PDK3, PDP1, SLC19A2, SLC19A3, TPK1
Hydroxyglutaric Aciduria Panel (4 genes)
L2HGDH, D2HGDH, IDH2, SLC25A1

***CFTR* Common Mutation Panel Variants NM_000492.4**		
DNA level nomenclature	**Protein level nomenclature**	**Historical nomenclature**
c.54-5940_273+10250del	p.?	CFTRdele2,3
c.254G>A	p.(Gly85Glu)	G85E
c.262_263del	p.(Leu88IlefsTer22)	394delTT
c.350G>A	p.(Arg117His)	R117H
c.366T>A	p.(Tyr122Ter)	Y122X
c.489+1G>T	p.?	621+1G>T
c.579+1G>T	p.?	711+1G>T
c.948del	p.(Phe316LeufsTer12)	1078delT
c.1000C>T	p.(Arg334Trp)	R334W
c.1040G>C	p.(Arg347Pro)	R347P
c.1040G>A	p.(Arg347His)	R347H
c.1210-12T[5/7/9]	p.?	5/7/9T
c.1364C>A	p.(Ala455Glu)	A455E
c.1466C>A	p.(Ser489Ter)	S489X
c.1519_1521del	p.(Ile507del)	I507del
c.1521_1523del	p.(Phe508del)	F508del/df508
c.1558G>T	p.(Val520Phe)	V520F
c.1585-1G>A	p.?	1717-1G>A
c.1624G>T	p.(Gly542Ter)	G542X
c.1646G>A	p.(Ser549Asn)	S549N
c.1647T>G	p.(Ser549Arg)	S549R
c.1652G>A	p.(Gly551Asp)	G551D
c.1657C>T	p.(Arg553Ter)	R553X
c.1675G>A	p.(Ala559Thr)	A559T
c.1679G>C	p.(Arg560Thr)	R560T
c.1679G>A	p.(Arg560Lys)	R560K
c.1766+1G>A	p.?	1898+1G>A
c.1766+1G>C	p.?	1898+1G>C
c.1766+1G>T	p.?	1898+1G>T
c.1766+5G>T	p.?	1898+5G>T
c.2051_2052delinsG	p.(Lys684SerfsTer38)	2183AA>G
c.2052del	p.(Lys684AsnfsTer38)	2184delA
c.2175dup	p.(Glu726ArgfsTer4)	2307insA
c.2657+2_2657+3insA	p.?	2789+2insA/2657+2insA
c.2657+5G>A	p.?	2789+5G>A
c.2988+1G>A	p.?	3120+1G>A
c.3276C>A/G	p.(Tyr1092Ter)	Y1092X
c.3302T>A	p.(Met1101Lys)	M1101K
c.3454G>C	p.(Asp1152His)	D1152H
c.3484C>T	p.(Arg1162Ter)	R1162X
c.3528del	p.(Lys1177SerfsTer15)	3659delC
c.3718-2477C>T	p.?	3849+10kbC>T
c.3744del	p.(Lys1250ArgfsTer9)	3876delA
c.3764C>A	p.(Ser1255Ter)	S1255X
c.3773dup	p.(Leu1258PhefsTer7)	3905insT
c.3846G>A	p.(Trp1282Ter)	W1282X
c.3909C>G	p.(Asn1303Lys)	N1303K