

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	ARMCS, 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 <u>common mutation panel only</u> : see table below for complete list of variants	
Spinal Muscular Atrophy	SMN1 and SMN2 <u>copy number only</u>	
SEVERE COMBINED IMMUNE DEFICIENCIES		
Adenosine Deaminase Deficiency	ADA	
PNP Deficiency	PNP	

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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, ATP5ME, ATP5MG, ATP5MGL, ATP5PO, ATP5PB, ATP5PD, ATP5PF, ATPAF1, ATPAF2, AUH, BCKDHA, BCKDHB, BCS1L, BOLA3, BTD, C1QB, C1QBP, C19orf12, CASA, 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, NDUFAB2, NDUFAB3, NDUFAB4, NDUFAB5, NDUFAB7, NDUFAB8, NDUFAB9, 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, PKCK, 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, TWINK, TXN2, TYMP, UCHL1, UNG, UQCC1, UQCC2, UQCC3, UQCR10, UQCR11, UQCRB, UQCRC1, UQCRC2, UQCRFS1, UQCRH, UQCRO, 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, NDUFAB1, NDUFAB2, NDUFAB3, NDUFAB4, NDUFAB5, NDUFAB7, NDUFAB8, NDUFAB9, NDUFC1, 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, SDHAF3, SDHAF4, SDHB, SDHC, SDHD, SERAC1, SLC19A3, SUCLA2, SUCLG1, SURF1, TACO1, TIMM44, TK2, TMEM70, TOMM20, TPK1, TRMU, TSFM, TTC19, TUFM, TUSC3, TWINK, TYMP, UQCRB, UQCRO, 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, TWINK

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, NDUFAB3, 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, TWINK, 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