MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Inborn Errors of Metabolism Custom Gene Panel

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of variants in coding regions and intron/exon boundaries of the genes analyzed. NGS and/or a polymerase chain reaction (PCR)-based quantitative method is performed to test for the presence of deletions and duplications in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript[a]
AASS	NM_005763.4
ABAT	NM_020686.6
ABCA1	NM_005502.4
ABCB11	NM_003742.4
ABCB4	NM_000443.4
ABCC2	NM_000392.5
ABCC8	NM_000352.5
ABCD1	NM_000033.4
ABCD3	NM_002858.4
ABCD4	NM_005050.4
ABCG5	NM_022436.3
ABCG8	NM_022437.3
ABHD12	NM_001042472.3
ABHD5	NM_016006.6
ACAA2	NM_006111.3
ACACA	NM_198839.2
ACAD8	NM_014384.2
ACAD9	NM_014049.5
ACADL	NM_001608.4
ACADM	NM_000016.5
ACADS	NM_000017.4
ACADSB	NM_001609.4
ACADVL	NM_000018.4
ACAT1	NM_000019.4
ACAT2	NM_005891.3
ACOT9	NM_001037171.2
ACOX1	NM_004035.7
ACOX3	NM_003501.3
ACSF3	NM_174917.5
ACY1	NM_000666.3
ADA	NM_000022.4
ADA2	NM_001282225.2
ADAMTS6	NM_197941.4
ADCY5	NM_183357.2
ADK	NM_001123.3
ADSL	NM_000026.4
AGA	NM_000027.4

Gene	Reference Transcript[a]
AGK	NM_018238.4
AGL	NM_000642.3
AGPAT2	NM_006412.4
AGPS	NM_003659.4
AGXT2	NM_031900.4
AHCY	NM_000687.4
AICDA	NM_020661.4
AK1	NM_000476.2
AK2	NM_001625.4
AKR1D1	NM_005989.4
AKT2	NM_001626.6
ALAD	NM_000031.6
ALAS2	NM_000032.5
ALDH18A1	NM_002860.4
ALDH4A1	NM_003748.4
ALDH5A1	NM_001080.3
ALDH6A1	NM_005589.4
ALDH7A1	NM_001182.5
ALDOA	NM_000034.3
ALDOB	NM_000035.4
ALDOC	NM_005165.3
ALG1	NM_019109.5
ALG11	NM_001004127.3
ALG12	NM_024105.4
ALG13	NM_001099922.3
ALG14	NM_144988.4
ALG2	NM_033087.4
ALG3	NM_005787.6
ALG5	NM_013338.5
ALG6	NM_013339.4
ALG8	NM_024079.5
ALG9	NM_024740.2 [GRCh38(hg38)]
AMACR	NM_014324.6
AMN	NM_030943.3
AMPD1	NM_000036.2
AMPD2	NM_001257360.1
AMT	NM_000481.4

Targeted Genes and Methodology Details for Inborn Errors of Metabolism Custom Gene Panel (continued)

Gene	Reference Transcript[a]
AOX1	NM_001159.4
APOA1	NM_000039.2
APOA5	NM_052968.5
APOB	NM_000384.3
APOC2	NM_000483.5
APOE	NM_000041.4
APRT	NM_000485.3
ARCN1	NM_001655.5
ARG1	NM_000045.4
ARG2	NM_001172.4
ARSA	NM_000487.6
ARSB	NM_000046.5
ARSL	NM_000047.3
ARV1	NM_022786.3
ASAH1[c]	NM_177924.5
ASL	NM_000048.4
ASNS	NM_133436.3
ASPA	NM_000049.4
ASS1	NM_000050.4
ATIC	NM_004044.7
ATP13A2	NM_022089.4
ATP5F1E	NM_006886.4
ATP6AP1	NM_001183.6
ATP6V0A2	NM_012463.4
ATP7A	NM_000052.7
ATP7B	NM_000053.4
ATP8B1	NM_005603.6
ATPAF2	NM_145691.4
AUH	NM_001698.2
B3GALNT2	NM_152490.5
B3GALT6[b,c]	NM_080605.4
B3GAT3	NM_012200.4
B3GLCT[c]	NM_194318.4
B4GALNT1	NM_001478.5
B4GALT1	NM_001497.3
B4GALT7	NM_007255.3
B4GAT1	NM_006876.3
BAAT	NM_001701.4
BCKDHA	NM_000709.4
BCKDHB	NM_183050.4
BCKDK	NM_005881.4
BCS1L	NM_004328.5
BDH1	NM_004051.5

Gene	Reference Transcript[a]
BOLA3	NM_212552.3
BRAF	NM_004333.6
BSCL2	NM_032667.6
BTD	NM_000060.4
C15orf41	NM_001130010.3
C1GALT1C1	NM_152692.4
CA5A	NM_001739.2
CAD	NM_004341.5
CANT1	NM_138793.4
CAT	NM_001752.4
CAV1	NM_001753.5
CAVIN1	NM_012232.6
CBL	NM_005188.4
CBLIF	NM_005142.3
CBS	NM_000071.2
CC2D2A	NM_001080522.2
CCBE1	NM_133459.4
CCDC115	NM_032357.4
CD320	NM_016579.4
CDA	NM_001785.3
CDAN1	NM_138477.4
CFTR[c]	NM_000492.4
CHIT1	NM_003465.3
CHKA	NM_001277.3
CHKB	NM_005198.4
CHRNA1	NM_001039523.3
CHRND	NM_000751.3
CHRNG	NM_005199.5
CHST14	NM_130468.3
CHST3	NM_004273.5
CHST6	NM_021615.5
CHST8	NM_001127896.2
CHSY1[b,c]	NM_014918.5
CIDEC	NM_001199623.1
CISD2	NM_001008388.5
CLCNKA	NM_004070.4
CLCNKB[c]	NM_000085.5
CLDN1	NM_021101.5
CLN3	NM_001042432.1
CLN5	NM_006493.4
CLN6	NM_017882.3
CLN8	NM_018941.4
CLPB	NM_030813.6

Targeted Genes and Methodology Details for Inborn Errors of Metabolism Custom Gene Panel (continued)

Gene	Reference Transcript[a]
CLPX	NM_006660.5
COG1	NM_018714.3
COG2	NM_007357.3
COG4	NM_015386.3
COG5	NM_006348.3
COG6[c]	NM_020751.3
COG7	NM_153603.4
COG8	NM_032382.4
COL2A1	NM_001844.5
CP[c]	NM_000096.4
CPOX	NM_000097.7
CPS1	NM_001875.5
CPT1A	NM_001876.4
CPT2	NM_000098.3
CRPPA	NM_001101426.4
CTH	NM_001902.6
CTNS	NM_001031681.2
CTSA	NM_000308.3
CTSD	NM_001909.5
CTSF	NM_003793.4
CTSK	NM_000396.4
CUBN	NM_001081.4
CYP27A1	NM_000784.4
CYP2U1	NM_183075.3
CYP7A1	NM_000780.4
CYP7B1	NM_004820.5
D2HGDH	NM_152783.5
DBH	NM_000787.4
DBT	NM_001918.4
DCDC2	NM_016356.5
DDC	NM_000790.4
DDHD1	NM_001160147.2
DDOST	NM_005216.4
DECR1	NM_001359.2
DGAT1	NM_012079.6
DGKE	NM_003647.3
DGUOK	NM_080916.3
DHCR24	NM_014762.4
DHCR7	NM_001360.2
DHDDS	NM_024887.3
DHFR[c]	NM_000791.4
DHODH	NM_001361.5
DHTKD1	NM_018706.7

Gene	Reference Transcript[a]
DLAT	NM_001931.5
DLD	NM_000108.5
DMGDH	NM_013391.3
DNAJC12	NM_021800.3
DNAJC19	NM_145261.4
DNAJC5	NM_025219.3
DNM1L	NM_012062.5
DOLK	NM_014908.4
DPAGT1	NM_001382.4
DPM1	NM_003859.2
DPM2	NM_003863.3
DPM3	NM_153741.2
DPYD	NM_000110.4
DPYS	NM_001385.3
DSE	NM_013352.4
DUOX2[b,c]	NM_014080.4
EBP	NM_006579.3
ECHS1	NM_004092.4
ECI1	NM_001919.4
EHHADH	NM_001966.4
ENO3	NM_001976.5
EOGT	NM_001278689.2
EPM2A	NM_005670.4
ETFA[c]	NM_000126.4
ETFB	NM_001985.3
ETFDH	NM_004453.4
ETHE1	NM_014297.5
EXT1	NM_000127.2
EXT2	NM_207122.1
FAH	NM_000137.3
FAR1	NM_032228.6
FAT4	NM_024582.4
FBP1	NM_000507.4
FBXL4	NM_012160.4
FCSK	NM_145059.3
FECH	NM_000140.4
FGA	NM_021871.4
FGB	NM_005141.4
FGFR3	NM_000142.4
FGG	NM_000509.5
FHL1	NM_001449.5
FKRP	NM_024301.5
FKTN	NM_001079802.1

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Gene	Reference Transcript[a]
FLAD1	NM_025207.5
FMO3	NM_001002294.3
FOLR1	NM_016725.3
FOXC2	NM_005251.3
FOXP3	NM_014009.4
FOXRED1	NM_017547.4
FTCD	NM_006657.3
FTL	NM_000146.4
FUCA1	NM_000147.4
FUT8	NM_178155.3
FXN	NM_000144.5
G6PC	NM_000151.4
G6PC3	NM_138387.3
G6PD	NM_001042351.3
GAA	NM_000152.5
GALC[c]	NM_000153.4
GALE	NM_000403.4
GALK1	NM_000154.2
GALM	NM_138801.3
GALNS	NM_000512.5
GALNT2	NM_004481.5
GALNT3	NM_004482.4
GALT	NM_000155.4
GAMT	NM_000156.6
GATA1	NM_002049.4
GATM	NM_001482.3
GBA	NM_000157.4
GBE1	NM_000158.4
GCDH	NM_000159.4
GCH1[b,c]	NM_000161.3
GCK	NM_000162.5
GCLC	NM_001498.4
GCSH	NM_004483.5
GET4	NM_015949.3
GFAP	NM_002055.5
GFER	NM_005262.3
GFM1	NM_024996.5
GFPT1[c]	NM_002056.4
GGT5	NM_004121.3
GK[c]	NM_000167.5
GLA[c]	NM_000169.2
GLB1	NM_000404.4
GLDC	NM_000170.2

Gene	Reference Transcript[a]
GLIS3	NM_152629.3
GLRA1	NM_000171.4
GLRB[c]	NM_000824.5
GLRX5	NM_016417.3
GLUD1	NM_005271.5
GLUL	NM_002065.6
GM2A	NM_000405.5
GMPPA	NM_205847.3
GMPPB	NM_013334.3
GNE	NM_001128227.3
GNMT	NM_018960.6
GNPAT[c]	NM_014236.4
GNPTAB[c]	NM_024312.5
GNPTG	NM_032520.5
GNS[c]	NM_002076.4
GOLIM4[c]	NM_014498.5
GORASP2	NM_001201428.2
GPD1[c]	NM_005276.4
GPHN	NM_020806.4
GPIHBP1	NM_178172.6
GRN	NM_002087.3
GSS	NM_000178.4
GUSB	NM_000181.4
GYG1	NM_004130.3
GYS1	NM_002103.5
GYS2	NM_021957.4
HADH	NM_005327.5
HADHA[c]	NM_000182.5
HADHB	NM_000183.3
HAL[c]	NM_002108.4
HCFC1	NM_005334.3
HEXA	NM_000520.6
HEXB[c]	NM_000521.4
HFE	NM_000410.3
HGD	NM_000187.4
HGSNAT	NM_152419.3
HK1	NM_000188.2
HLCS	NM_000411.8
HMBS	NM_000190.4
HMGCL	NM_000191.3
HMGCS2	NM_005518.4
HNF1A	NM_000545.6
HNF1B	NM_000458.4

Targeted Genes and Methodology Details for Inborn Errors of Metabolism Custom Gene Panel (continued)

Gene	Reference Transcript[a]
HNF4A	NM_175914.4
HPD	NM_002150.3
HPRT1[b,c]	NM_000194.3
HRAS	NM_005343.4
HSD17B10	NM_004493.3
HSD17B4	NM_001199291.3
HSD3B7	NM_025193.4
HTRA2	NM_013247.4
HYAL1	NM_153281.1
IBA57	NM_001010867.4
IDH1	NM_005896.3
IDH2	NM_002168.3
IDS	NM_000202.8
IDUA	NM_000203.5
IHH	NM_002181.4
IMPDH1	NM_000883.4
IMPDH2	NM_000884.3
INSR	NM_000208.4
INVS	NM_014425.5
ISCA2	NM_194279.4
ITPA	NM_033453.4
IVD	NM_002225.5
IYD	NM_203395.3
JAG1	NM_000214.3
KAT6B	NM_012330.4
KCNH1	NM_172362.3
KCNJ11	NM_000525.3
KCTD7	NM_153033.4
KHK	NM_000221.3
KIAA0586[b,c]	NM_001244189.2
KIF23	NM_138555.4
KLF1	NM_006563.5
KMT2D	NM_003482.3
KRAS	NM_004985.5
L2HGDH[c]	NM_024884.3
LAMP2	NM_002294.3
LARGE1	NM_004737.6
LBR	NM_002296.4
LCAT[b,c]	NM_000229.2
LDHA	NM_005566.4
LDLRAP1	NM_015627.3
LFNG[b,c]	NM_001040167.2
LIAS	NM_006859.4

Gene	Reference Transcript[a]
LIPA	NM_000235.4
LIPC	NM_000236.3
LIPE	NM_005357.4
LIPG	NM_006033.4
LIPT1	NM_145199.3
LMBRD1[c]	NM_018368.4
LMF1	NM_022773.4
LPIN1	NM_145693.4
LPIN2	NM_014646.2
LPL[b,c]	NM_000237.3
LZTR1	NM_006767.4
MADD	NM_003682.4
MAGT1	NM_032121.5
MAN1B1	NM_016219.5
MAN2B1	NM_000528.4
MAN2B2	NM_015274.3
MANBA	NM_005908.4
MAOA[c]	NM_000240.3
MAOB	NM_000898.5
MAP2K1	NM_002755.3
MAP2K2	NM_030662.3
MAT1A	NM_000429.3
MAT2A	NM_005911.6
MBTPS1	NM_003791.4
MCCC1	NM_020166.5
MCCC2	NM_022132.5
MCEE	NM_032601.4
MCM6	NM_005915.6
MCOLN1	NM_020533.3
MFSD8	NM_152778.3
MGAT1	NM_001114618.1
MGAT2	NM_002408.4
MGLL	NM_007283.6
MID1	NM_000381.4
MKS1	NM_017777.4
MLYCD	NM_012213.3
MMAA	NM_172250.3
MMAB	NM_052845.4
MMACHC	NM_015506.3
MMADHC	NM_015702.3
MMUT	NM_000255.4
MOCOS	NM_017947.4
MOCS1	NM_005943.5

Targeted Genes and Methodology Details for Inborn Errors of Metabolism Custom Gene Panel (continued)

Gene	Reference Transcript[a]
MOCS2	NM_176806.4
MOCS3	NM_014484.5
MOGS	NM_006302.3
MPC1[b,c]	NM_016098.4
MPDU1	NM_004870.4
MPI	NM_002435.3
MPV17	NM_002437.5
MRPL3	NM_007208.4
MRPS22	NM_020191.4
MSMO1	NM_006745.5
MTHFD1	NM_005956.4
MTHFD2L	NM_001144978.2
MTHFR	NM_005957.5
MTHFS	NM_006441.3
MTR	NM_000254.2
MTRR	NM_002454.3
MTTP	NM_000253.3
MVK	NM_000431.4
NADK2	NM_001085411.3
NAGA	NM_000262.3
NAGLU	NM_000263.4
NAGS	NM_153006.3
NDP	NM_000266.4
NDUFB11	NM_019056.6
NDUFS4	NM_002495.4
NEU1	NM_000434.4
NFU1	NM_001002755.3
NGLY1	NM_018297.4
NHLRC1	NM_198586.3
NNT	NM_012343.4
NOTCH2[b,c]	NM_024408.4
NPC1[c]	NM_000271.5
NPC2	NM_006432.4
NPHP1[c]	NM_000272.4
NPHP3[c]	NM_153240.5
NPHP4	NM_015102.5
NR1H4[c]	NM_005123.4
NRAS	NM_002524.5
NSDHL	NM_015922.3
NT5C3A[c]	NM_016489.13
NUS1[c]	NM_138459.5
OAT	NM_000274.4
OGDH	NM_002541.4

Gene	Reference Transcript[a]
OPA1	NM_015560.2
OPA3	NM_025136.4
OPLAH	NM_017570.5
OTC	NM_000531.6
OXCT1[b,c]	NM_000436.4
PAH	NM_000277.3
PANK2	NM_153638.3
PAPSS2	NM_001015880.2
PAX8	NM_003466.4
PC	NM_000920.4
PCBD1	NM_000281.4
PCCA[c]	NM_000282.4
PCCB	NM_000532.5
PCK1	NM_002591.4
PCK2	NM_004563.4
PCSK9	NM_174936.4
PCYT1A	NM_005017.4
PDHA1	NM_000284.4
PDHA2	NM_005390.5
PDHB	NM_000925.4
PDHX	NM_003477.3
PDP1	NM_018444.4
PDX1	NM_000209.4
PDXK	NM_003681.5
PEPD	NM_000285.4
PEX1	NM_000466.3
PEX10	NM_153818.1
PEX11B	NM_003846.3
PEX12	NM_000286.3
PEX13	NM_002618.4
PEX14	NM_004565.3
PEX16	NM_057174.2
PEX19	NM_002857.3
PEX2	NM_000318.3
PEX26	NM_017929.6
PEX3	NM_003630.3
PEX5	NM_001131023.1
PEX6	NM_000287.4
PEX7	NM_000288.4
PFKM	NM_000289.6
PGAM2	NM_000290.4
PGAP2	NM_001256240.2
PGAP3	NM_033419.5

Targeted Genes and Methodology Details for Inborn Errors of Metabolism Custom Gene Panel (continued)

Gene	Reference Transcript[a]
PGK1[c]	NM_000291.4
PGM1	NM_002633.3
PGM2	NM_018290.4
PGM3	NM_001199917.2
PHGDH	NM_006623.4
PHKA1[c]	NM_002637.4
PHKA2[c]	NM_000292.3
PHKB[c]	NM_000293.3
PHKG2	NM_000294.3
PHYH	NM_006214.4
PIEZO1	NM_001142864.4
PIGA	NM_002641.3
PIGL	NM_004278.4
PIGM	NM_145167.3
PIGN[c]	NM_176787.5
PIGO	NM_032634.4
PIGT	NM_015937.6
PIGV	NM_017837.3
PIGW	NM_178517.4
PIGY	NM_001042616.2
PIPOX	NM_016518.3
PKHD1	NM_138694.4
PKLR	NM_000298.6
PLA2G6	NM_003560.4
PLIN1	NM_002666.5
PLPBP	NM_007198.4
PMM1	NM_002676.3
PMM2	NM_000303.3
PNP	NM_000270.3
PNPLA2	NM_020376.4
PNPLA6	NM_006702.5
PNPLA8	NM_015723.5
PNPO	NM_018129.4
POFUT1	NM_015352.2
POGLUT1[c]	NM_152305.3
POLG	NM_002693.2
POMGNT1	NM_017739.3
POMGNT2	NM_032806.6
POMK	NM_032237.5
POMT1	NM_007171.3
POMT2	NM_013382.5
POR	NM_000941.3
PPARG	NM_015869.4

Gene	Reference Transcript[a]
PPM1K	NM_152542.5
PPOX	NM_000309.5
PPT1	NM_000310.3
PRDX1	NM_002574.3
PREPL	NM_006036.4
PRKAG2[c]	NM_016203.4
PRKCSH	NM_002743.3
PRODH	NM_016335.5
PRPS1	NM_002764.4
PSAP	NM_002778.4
PSAT1	NM_058179.4
PSPH[c]	NM_004577.4
PTDSS1	NM_014754.3
PTH1R	NM_000316.3
PTPN11	NM_002834.4
PTS	NM_000317.3
PYCR1	NM_006907.4
PYCR2	NM_013328.4
PYCR3	NM_023078.6
PYGL	NM_002863.5
PYGM	NM_005609.4
PYY	NM_004160.5
QDPR	NM_000320.3
RAF1	NM_002880.3
RASA1	NM_002890.3
RBCK1	NM_031229.4
RFT1	NM_052859.4
RIT1	NM_006912.6
RNF216	NM_207111.4
RPIA	NM_144563.3
RPL11	NM_000975.5
RPL35A	NM_000996.4
RPL5	NM_000969.5
RPS10	NM_001014.5
RPS19	NM_001022.4
RPS24	NM_033022.4
RPS26	NM_001029.5
RXYLT1	NM_014254.3
SAR1B	NM_001033503.3
SARDH	NM_007101.4
SC5D	NM_006918.5
SCARB1	NM_005505.5
SCGB1D2	NM_006551.4

Gene	Reference Transcript[a]
SCP2[c]	NM_002979.5
SEC23A	NM_006364.4
SEC23B	NM_006363.6
SEC63	NM_007214.5
SERAC1[c]	NM_032861.4
SERPINA1	NM_000295.5
SGSH	NM_000199.5
SHH	NM_000193.4
SHMT1	NM_004169.5
SHOC2	NM_007373.3
SHPK	NM_013276.4
SLC10A1	NM_003049.4
SLC10A2	NM_000452.3
SLC10A7	NM_001300842.3
SLC16A1	NM_003051.3
SLC16A2	NM_006517.5
SLC17A5	NM_012434.5
SLC18A2	NM_003054.6
SLC19A1	NM_194255.4
SLC19A2	NM_006996.3
SLC19A3	NM_025243.4
SLC1A1	NM_004170.6
SLC22A5	NM_003060.4
SLC25A1	NM_005984.5
SLC25A13	NM_014251.3
SLC25A15	NM_014252.4
SLC25A19	NM_021734.4
SLC25A20	NM_000387.6
SLC25A29	NM_001039355.3
SLC25A32	NM_030780.5
SLC25A38	NM_017875.4
SLC26A2	NM_000112.4
SLC27A5	NM_012254.3
SLC2A1	NM_006516.3
SLC2A10	NM_030777.4
SLC2A2	NM_000340.2
SLC2A3	NM_006931.3
SLC2A4	NM_001042.3
SLC34A1	NM_003052.5
SLC35A1[c]	NM_006416.5
SLC35A2	NM_001042498.3
SLC35A3[c]	NM_012243.3
SLC35C1	NM_018389.5

Gene	Reference Transcript[a]
SLC35D1[c]	NM_015139.3
SLC36A2	NM_181776.3
SLC36A4	NM_001286139.2
SLC37A4	ENST00000545985.1
SLC39A8	NM_022154.5
SLC3A1	NM_000341.4
SLC46A1	NM_080669.6
SLC52A1	NM_001104577.1
SLC52A2	NM_024531.5
SLC52A3	NM_033409.4
SLC5A1	NM_000343.4
SLC5A2	NM_003041.4
SLC5A5	NM_000453.3
SLC6A19	NM_001003841.3
SLC6A20	NM_020208.4
SLC6A5	NM_004211.5
SLC6A8	NM_005629.4
SLC6A9	NM_201649.4
SLC7A7	NM_001126106.2
SLC7A9	NM_014270.5
SMPD1	NM_000543.5
SOS1	NM_005633.3
SOS2[c]	NM_006939.4
SOX18[b,c]	NM_018419.3
SPR	NM_003124.5
SRD5A3	NM_024592.5
SRR	NM_021947.3
SSR3	NM_007107.4
SSR4	NM_006280.3
ST3GAL3	NM_006279.5
ST3GAL5	NM_003896.4
STS	NM_000351.6
STT3A	NM_001278503.2
STT3B	NM_178862.3
STXBP1	NM_003165.4
SUCLA2	NM_003850.2
SUCLG1	NM_003849.4
SUGCT[b,c]	NM_024728.2
SUMF1	NM_182760.4
SUOX	NM_000456.3
SYP	NM_003179.2
TALDO1	NM_006755.2
TANGO2	NM_152906.7

Targeted Genes and Methodology Details for Inborn Errors of Metabolism Custom Gene Panel (continued)

Gene	Reference Transcript[a]
TAT	NM_000353.3
TAZ	NM_000116.5
TBC1D24	NM_001199107.2
TCN1	NM_001062.4
TCN2	NM_000355.4
TF	NM_001063.4
TG	NM_003235.5
TH	NM_199292.3
THAP11	NM_020457.3
THRA	NM_199334.4
TIMM50	NM_001001563.5
TJP2	NM_004817.4
TKT[b,c]	NM_001258028.1
TMEM165	NM_018475.5
TMEM199	NM_152464.3
TMEM216	NM_001173990.3
TMEM70	NM_017866.6
TPH1	NM_004179.3
TPK1	NM_022445.4
TPO	NM_000547.5
TPP1	NM_000391.4
TRAPPC11	NM_021942.6
TRAPPC9	NM_031466.7
TRIM37	NM_015294.6
TRIP11	NM_004239.4
TRMU	NM_018006.5

Gene	Reference Transcript[a]
TSHB	NM_000549.5
TSHR	NM_000369.3
TSTA3	NM_003313.4
TTC19	NM_017775.4
TUFM	NM_003321.5
TUSC3[c]	NM_006765.4
UCP2	NM_003355.2
UGT1A1	NM_000463.3
UMPS	NM_000373.4
UPB1	NM_016327.3
UQCRB	NM_006294.4
UQCRC2	NM_003366.4
UQCRQ	NM_014402.5
UROD	NM_000374.5
UROS	NM_000375.3
VARS2	NM_001167734.1
VIPAS39	NM_022067.4
VMA21	NM_001017980.3
VPS33A	NM_022916.6
VPS33B	NM_018668.4
WDR35	NM_001006657.2
WFS1	NM_006005.3
XDH	NM_000379.4
XYLT1	NM_022166.4
ZNF143	NM_003442.6a

[a] Reference transcript numbers may be updated due to transcript re-versioning. Always refer to the final patient report for gene transcript information referenced at the time of testing.

[b] There are regions of this gene that cannot be effectively amplified for sequencing as a result of technical limitations of the assay, including regions of homology, high GC content, and repetitive sequences.

[c] There are regions of this gene that cannot be effectively analyzed for the presence of copy number variants.

To verify if a specific region/exon/variant is covered by this assay, contact a laboratory genetic counselor at 800-533-1710.

Available Inborn Errors of Metabolism Panels

Test ID	Test Name	Genes
2OHGP	2-Hydroxyglutaric Aciduria Gene Panel	*D2HGDH, IDH2, L2HGDH, SLC25A1*
3MGAP	3-Methylglutaconic Aciduria Panel	*AGK, ATP5F1E, ATPAF2, AUH, CLPB, CPS1, DNAJC19, GFER, HMGCL, HTRA2, OPA3, POLG, SERAC1, SUCLA2, TAZ, TIMM50, TMEM70*
APGP	Acute Porphyria Gene Panel	*ALAD, CPOX, HMBS, PPOX*
CDGGP	Congenital Disorders of Glycosylation Gene Panel, Varies	*ALDOB, ALDOC, ALG1, ALG11, ALG12, ALG13, ALG14, ALG2, ALG3, ALG5, ALG6, ALG8, ALG9, ARCN1, ARV1, ATP6AP1, ATP6V0A2, B3GALNT2, B3GALT6, B3GAT3, B3GLCT, B4GALNT1, B4GALT1, B4GALT7, B4GAT1, C1GALT1C1, CCDC115, CHST14, CHST3, CHST6, CHST8, CHSY1, COG1, COG2, COG4, COG5, COG6, COG7, COG8, CRPPA, DDOST, DHDDS, DOLK, DPAGT1, DPM1, DPM2, DPM3, DSE, EOGT, EXT1, EXT2, FCSK, FKRP, FKTN, FUT8, G6PC3, GALE, GALK1, GALNT2, GALNT3, GALT, GET4, GFM1, GFPT1, GMPPA, GMPPB, GNE, GNPTAB, GOLIM4, GORASP2, LARGE1, LFNG, MAGT1, MAN1B1, MAN2B2, MBTPS1, MGAT1, MGAT2, MOGS, MPDU1, MPI, MPV17, NGLY1, NUS1, PAPSS2, PGAP2, PGAP3, PGM1, PGM2, PGM3, PIGA, PIGL, PIGM, PIGN, PIGO, PIGT, PIGV, PIGW, PMM1, PMM2, POFUT1, POGLUT1, POMGNT1, POMGNT2, POMK, POMT1, POMT2, PRKCSH, RFT1, RXYLT1, SEC23A, SEC23B, SEC63, SLC10A7, SLC26A2, SLC35A1, SLC35A2, SLC35A3, SLC35C1, SLC35D1, SLC37A4, SLC39A8, SRD5A3, SSR3, SSR4, ST3GAL3, ST3GAL5, STT3A, STT3B, STXBP1, SYP, TF, TMEM165, TMEM199, TRAPPC11, TRAPPC9, TRIP11, TSTA3, TUSC3, VMA21, XYLT1*
CHLGP	Cholestasis Gene Panel	*ABCB11, ABCB4, ABCC2, ABCG5, ABCG8, ABHD5, ACOX1, AGL, AGPAT2, AKR1D1, ALDOA, ALDOB, AMACR, ARSB, ASAH1, ATP8B1, BAAT, BSCL2, CAVIN1, CC2D2A, CFTR, CIDEC, CLDN1, CYP27A1, CYP7A1, CYP7B1, DCDC2, DGUOK, DHCR7, EHHADH, FAH, FBP1, FUCA1, G6PC, GAA, GALNS, GBA, GBE1, GLB1, GNE, GNPTAB, GNS, GUSB, HADHA, HGSNAT, HNF1B, HSD17B4, HSD3B7, IDS, IDUA, INVS, JAG1, KCNH1, LIPA, MAN2B1, MKS1, MPV17, MVK, NAGLU, NEU1, NOTCH2, NPC1, NPC2, NPHP1, NPHP3, NPHP4, NR1H4, PEPD, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHKA2, PHKB, PHKG2, PKHD1, PNPLA2, POLG, PRKAG2, PSAP, PYGL, SCP2, SERPINA1, SGSH, SLC10A1, SLC10A2, SLC17A5, SLC25A13, SLC27A5, SLC37A4, SLC7A7, SMPD1, SUMF1, TALDO1, TJP2, TMEM216, TRIM37, TRMU, UGT1A1, VIPAS39, VPS33A, VPS33B*
CLADP	Congenital Lactic Acidosis Panel	*ACAD9, AGK, DLD, ECHS1, FBXL4, FLAD1, FOXRED1, GFER, HADHA, HADHB, HLCS, MRPL3, MRPS22, NDUFB11, NDUFS4, OGDH, PC, PDHA1, PDHX, PDP1, SLC19A2, SLC19A3, SLC25A19, SUCLG1, TMEM70, TPK1, UQCRC2, VARS2*
CYSGP	Cystinuria Gene Panel	*SLC3A1, SLC7A9, PREPL*
DHCRZ	Smith Lemli Optiz, DHCR7 Gene, Full Gene Analysis	*DHCR7*
GA2P	Glutaric Aciduria Type II Gene Panel	*ETFA, ETFB, ETFDH, FLAD1, SLC52A1, SLC52A2, SLC52A3, TANGO2*
GALZ	Galactosemia, *GALT* Gene, Full Gene Analysis	*GALT*
GSDGP	Glycogen Storage Disease Gene Panel	*AGL, ALDOA, ENO3, EPM2A, FBP1 , G6PC, GAA, GBE1, GYG1, GYS1, GYS2, LAMP2, LDHA, NHLRC1, PFKM, PGAM2, PGK1, PGM1, PHKA1, PHKA2, PHKB, PHKG2, PRKAG2, PYGL, PYGM, RBCK1, SLC2A2, SLC37A4*
HEXBZ	Sandhoff Disease, HEXB Gene, Full Gene Analysis	*HEXB*
HFAOP	Fatty Acid Oxidation Gene Panel	*ACAA2, ACACA, ACAD8, ACAD9, ACADL, ACADM, ACADS, ACADSB, ACADVL, ACAT1, ACAT2, ACOT9, ALDH5A1, CPT1A, CPT2, DECR1, ECHS1, ECI1, ETFA, ETFB, ETFDH, ETHE1, FLAD1, GLUD1, HADH, HADHA, HADHB, HMGCL, HMGCS2, HSD17B10, LPIN1, MLYCD, NADK2, OPA1, PPARG, SLC22A5, SLC25A20, SLC25A29, SLC25A32, SLC52A1, SLC52A2, SLC52A3, TANGO2, TAZ*

Available Inborn Errors of Metabolism Panels

Test ID	Test Name	Genes
KETGP	Ketone Disorders Gene Panel	*ACAA2, ACAT1, ACAT2, AKT2, BDH1, HMGCL, HMGCS2, OXCT1, SLC16A1*
LSDGP	Lysosomal Storage Disease Gene Panel	*AGA, ARSA, ARSB, ASAH1, ATP13A2, CHIT1, CLN3, CLN5, CLN6, CLN8, CTNS, CTSA, CTSD, CTSF, CTSK, DNAJC5, FUCA1, GAA, GALC, GALNS, GBA, GFAP, GLA, GLB1, GM2A, GNPTAB, GNPTG, GNS, GRN, GUSB, HEXA, HEXB, HGSNAT, HYAL1, IDS, IDUA, KCTD7, LAMP2, LIPA, MAN2B1, MANBA, MCOLN1, MFSD8, NAGA, NAGLU, NEU1, NPC1, NPC2, PANK2, PPT1, PSAP, SGSH, SLC17A5, SMPD1, SUMF1, TPP1*
MMAGP	Methylmalonic Aciduria Gene Panel	*ABCD4, ACSF3, ALDH6A1, AMN, CD320, CUBN, CBLIF, HCFC1, LMBRD1, MCEE, MMAA, MMAB, MMACHC, MMADHC, MTHFR, MTR, MTRR, MMUT, PRDX1, SUCLA2, SUCLG1, TCN1, TCN2, THAP11, ZNF143*
MPAGP	Methylmalonic Aciduria-Propionic Aciduria Combined Gene Panel	*ABCD4, ACSF3, ALDH6A1, AMN, CD320, CUBN, DMGDH, CBLIF, HCFC1, LMBRD1, MCEE, MMAA, MMAB, MMACHC, MMADHC, MTHFR, MTR, MTRR, MMUT, PCCA, PCCB, PRDX1, SUCLA2, SUCLG1, TCN1, TCN2, THAP11, ZNF143*
MSUDP	Maple Syrup Urine Disease Gene Panel	*BCKDHA, BCKDHB, BCKDK, DBT, DLD, PPM1K*
NCLGP	Neuronal Ceroid Lipofuscinosis (Batten Disease) Gene Panel	*ATP13A2, CLN3, CLN5, CLN6, CLN8, CTSD, CTSF, CTSK, DNAJC5, GRN, KCTD7, MFSD8, PANK2, PPT1, SGSH, TPP1*
PCGP	Porphyria Comprehensive Gene Panel	*ALAD, ALAS2, CLPX, CPOX, FECH, GATA1, HFE, HMBS, PPOX, UROD, UROS*
PDGP	Peroxisomal Disorder Gene Panel	*ABCD1, ABCD3, ACOX1, ACOX3, AGPS, AMACR, CAT, DNM1L, GNPAT, HSD17B4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, SCP2, SUGCT, TRIM37*
PHEGP	Phenylalanine Disorders Gene Panel	*DDC, DNAJC12, GCH1, PAH, PCBD1, PTS, QDPR, SLC18A2, SPR, TH*
TYRGP	Tyrosine Disorders Gene Panel	*FAH, HGD, HPD, TAT*
UCDP	Urea Cycle Disorders Gene Panel	*ALDH18A1, ARG1, ARG2, ASL, ASS1, CA5A, CPS1, GLUD1, GLUL, NAGS, OAT, OTC, SLC25A13, SLC25A15, SLC7A7, UMPS*