



The following applies to CGPH / Custom Gene Panel, Hereditary, Next-Generation Sequencing. Testing is performed to evaluate for the presence of variants in coding regions and extending to +/- 30 base pairs of adjacent intronic sequence on either side of the coding exons of the genes analyzed. In addition, the analysis will cover select non-coding variants. Next-generation sequencing and/or a polymerase chain reaction-based quantitative method is performed to test for the presence of copy number variants (CNV) in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

This list is current from January 2026 to the present. This document is intended to highlight additional evaluations for variants of high clinical interest as well as technical limitations. However, this document does not comprehensively reflect all genomic regions covered by this test. For questions regarding transcripts, genes or regions covered, contact the laboratory at 800-533-1710.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
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	chr17:g.7123159TCCCAGCATGCCCC>TT (c.-144_-132del13insT)	-
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	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
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	-	-
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	chr9:g.104198194C>T (c.-214G>A); chr9:g.104197990C>G (c.-11+1G>C); chr9:g.104183575A>T (c.*516T>A)	-
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	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
AMACR	NM_014324.6	-	-
AMN	NM_030943.3	-	-
AMPD1	NM_000036.2	-	-
AMPD2	NM_001257360.1	-	-
AMT	NM_000481.4	-	-
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	chr6:g.131901748T>C (c.306-611T>C)	-
ARG2	NM_001172.4	-	-
ARSA	NM_000487.6	-	-
ARSB	NM_000046.5	-	-
ARSL	NM_000047.3	-	-
ARV1	NM_022786.3	-	-
ASAH1	NM_177924.5	-	CNVs may not be detected in exon 4
ASL	NM_000048.4	-	-
ASNS	NM_133436.3	-	-
ASPA	NM_000049.4	-	-
ASS1	NM_000050.4	chr9:g.133332669G>A (c.175-1119G>A)	-
ATIC	NM_004044.7	-	-
ATP13A2	NM_022089.4	-	-
ATP5F1E	NM_006886.4	-	-
ATP6AP1	NM_001183.6	-	-
ATP6VOA2	NM_012463.4	-	-
ATP7A	NM_000052.7	-	-
ATP7B	NM_000053.4	chr13:g.52586149T>C (c.-676A>G); chr13:g.52585933C>T (c.-460G>A); chr13:g.52585920G>A (c.-447C>T); chr13:g.52585915C>T (c.-442G>A); chr13:g.52585897_52585911del (c.-436_-422del15); chr13:g.52585861G>A (c.-388C>T); chr13:g.52585683T>A (c.-210A>T); chr13:g.52585606T>G (c.-133A>C); chr13:g.52585601T>G (c.-128A>C); chr13:g.52585603_52585607del (c.-128_-124delAGCCG); chr13:g.52585596G>T (c.-123C>A); chr13:g.52534477A>T (c.1947-19T>A); chr13:g.52518439A>T (c.3061-12T>A)	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>ATP8B1</i>	NM_005603.6	-	-
<i>ATPAF2</i>	NM_145691.4	-	-
<i>AUH</i>	NM_001698.2	-	-
<i>B3GALNT2</i>	NM_152490.5	-	-
<i>B3GALT6</i>	NM_080605.4	-	-
<i>B3GAT3</i>	NM_012200.4	-	-
<i>B3GLCT</i>	NM_194318.4	-	CNVs may not be detected in exon 3
<i>B4GALNT1</i>	NM_001478.5	-	-
<i>B4GALT1</i>	NM_001497.3	-	-
<i>B4GALT7</i>	NM_007255.3	-	-
<i>B4GAT1</i>	NM_006876.3	-	-
<i>BAAT</i>	NM_001701.4	-	-
<i>BCKDHA</i>	NM_000709.4	-	-
<i>BCKDHB</i>	NM_183050.4	-	-
<i>BCKDK</i>	NM_005881.4	-	-
<i>BCS1L</i>	NM_004328.5	-	-
<i>BDH1</i>	NM_004051.5	-	-
<i>BOLA3</i>	NM_212552.3	-	-
<i>BRAF</i>	NM_004333.6	-	-
<i>BSCL2</i>	NM_032667.6	-	-
<i>BTD</i>	NM_000060.4	chr3:g.15687154G>A(c.*159G>A)	-
<i>C15orf41</i>	NM_001130010.3	-	-
<i>C1GALT1C1</i>	NM_152692.4	-	-
<i>CA5A</i>	NM_001739.2	-	-
<i>CAD</i>	NM_004341.5	-	-
<i>CANT1</i>	NM_138793.4	-	-
<i>CAT</i>	NM_001752.4	-	-
<i>CAV1</i>	NM_001753.5	-	-
<i>CAVIN1</i>	NM_012232.6	-	-
<i>CBL</i>	NM_005188.4	-	-
<i>CBLIF</i>	NM_005142.3	-	-
<i>CBS</i>	NM_000071.2	-	-
<i>CC2D2A</i>	NM_001080522.2	-	-
<i>CCBE1</i>	NM_133459.4	-	-
<i>CCDC115</i>	NM_032357.4	-	-
<i>CD320</i>	NM_016579.4	-	-
<i>CDA</i>	NM_001785.3	-	-
<i>CDAN1</i>	NM_138477.4	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>CFTR</i>	NM_000492.4	<p>Poly T tract; TG repeat region for 5T alleles only; deletion/duplication analysis; chr7:g.117179040AGAAT>A (c.870-1113_870-1110del); chr7:g.117199500G>A (c.1393-18G>A); chr7:g.117227784T>A (c.1585-9T>A); chr7:g.117227785G>A (c.1585-8G>A); chr7:g.117218381A>G (c.1585-9412A>G); chr7:g.117229521A>G (c.1680-886A>G); chr7:g.117229524A>G (c.1680-883A>G); chr7:g.117229530G>T (c.1680-877G>T); chr7:g.117246713T>G (c.2909-15T>G); chr7:g.117250260A>T (c.2989-313A>T); chr7:g.117251609A>G (c.3140-26A>G); chr7:g.117251619T>A (c.3140-16T>A); chr7:g.117266272C>G (c.3469-1304C>G); chr7:g.117267864A>G (c.3717+40A>G); chr7:g.117280015C>T (c.3718-2477C>T); chr7:g.117288374A>G (c.3874-4522A>G)</p>	CNVs may not be detected in exon 13
<i>CHIT1</i>	NM_003465.3	-	-
<i>CHKA</i>	NM_001277.3	-	-
<i>CHKB</i>	NM_005198.4	-	-
<i>CHRNA1</i>	NM_001039523.3	-	-
<i>CHRND</i>	NM_000751.3	-	-
<i>CHRNG</i>	NM_005199.5	-	-
<i>CHST14</i>	NM_130468.3	-	-
<i>CHST3</i>	NM_004273.5	-	-
<i>CHST6</i>	NM_021615.5	-	-
<i>CHST8</i>	NM_001127896.2	-	-
<i>CHSY1</i>	NM_014918.5	-	-
<i>CIDEC</i>	NM_001199623.1	-	-
<i>CISD2</i>	NM_001008388.5	-	-
<i>CLCNKA</i>	NM_004070.4	-	-
<i>CLCNKB</i>	NM_000085.5	-	CNVs may not be detected in exon 20
<i>CLDN1</i>	NM_021101.5	-	-
<i>CLN3</i>	NM_001042432.1	-	-
<i>CLN5</i>	NM_006493.4	-	-
<i>CLN6</i>	NM_017882.3	-	-
<i>CLN8</i>	NM_018941.4	-	-
<i>CLPB</i>	NM_030813.6	-	-
<i>CLPX</i>	NM_006660.5	-	-
<i>COG1</i>	NM_018714.3	-	-
<i>COG2</i>	NM_007357.3	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
COG4	NM_015386.3	-	-
COG5	NM_006348.3	-	-
COG6	NM_020751.3	-	CNVs may not be detected in exon 4
COG7	NM_153603.4	-	-
COG8	NM_032382.4	-	-
COL2A1	NM_001844.5	-	-
CP	NM_000096.4	-	CNVs may not be detected in exon 19
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	NM_000791.4	-	CNVs may not be detected in exon 6
DHODH	NM_001361.5	-	-
DHTKD1	NM_018706.7	-	-
DLAT	NM_001931.5	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>DLD</i>	NM_000108.5	-	-
<i>DMGDH</i>	NM_013391.3	-	-
<i>DNAJC12</i>	NM_021800.3	-	-
<i>DNAJC19</i>	NM_145261.4	-	-
<i>DNAJC5</i>	NM_025219.3	-	-
<i>DNM1L</i>	NM_012062.5	-	-
<i>DOLK</i>	NM_014908.4	-	-
<i>DPAGT1</i>	NM_001382.4	-	-
<i>DPM1</i>	NM_003859.2	-	-
<i>DPM2</i>	NM_003863.3	-	-
<i>DPM3</i>	NM_153741.2	-	-
<i>DPYD</i>	NM_000110.4	chr1:g.98045449G>C (c.1129-5923C>G)	Variants will not include pharmacogenomics interpretation. Consider DPYDZ / Dihydropyrimidine Dehydrogenase, DPYD Full Gene Sequencing, Varies for pharmacogenomics interpretation.
<i>DPYS</i>	NM_001385.3	-	-
<i>DSE</i>	NM_013352.4	-	-
<i>DUOX2</i>	NM_014080.4	-	Sequence variants and CNVs may not be detected in exon 7
<i>EBP</i>	NM_006579.3	-	-
<i>ECHS1</i>	NM_004092.4	-	-
<i>ECI1</i>	NM_001919.4	-	-
<i>EHHADH</i>	NM_001966.4	-	-
<i>ENO3</i>	NM_001976.5	-	-
<i>EOGT</i>	NM_001278689.2	-	-
<i>EPM2A</i>	NM_005670.4	-	-
<i>ETFA</i>	NM_000126.4	-	CNVs may not be detected in exon 12
<i>ETFB</i>	NM_001985.3	-	-
<i>ETFDH</i>	NM_004453.4	-	-
<i>ETHE1</i>	NM_014297.5	-	-
<i>EXT1</i>	NM_000127.2	-	-
<i>EXT2</i>	NM_207122.1	chr11:g.44265893G>A (c.*56G>A); chr11:g.44265938G>A (c.*101G>A)	-
<i>FAH</i>	NM_000137.3	-	-
<i>FAR1</i>	NM_032228.6	-	-
<i>FAT4</i>	NM_024582.4	-	-
<i>FBP1</i>	NM_000507.4	-	-
<i>FBXL4</i>	NM_012160.4	-	-
<i>FCSK</i>	NM_145059.3	-	-
<i>FECH</i>	NM_000140.4	chr18:g.55238820A>G (c.315-48T>C)	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>FGA</i>	NM_021871.4	-	-
<i>FGB</i>	NM_005141.4	-	-
<i>FGFR3</i>	NM_000142.4	-	-
<i>FGG</i>	NM_000509.5	-	-
<i>FHL1</i>	NM_001449.5	-	-
<i>FKRP</i>	NM_024301.5	-	-
<i>FKTN</i>	NM_001079802.1	-	-
<i>FLAD1</i>	NM_025207.5	-	-
<i>FMO3</i>	NM_001002294.3	-	-
<i>FOLR1</i>	NM_016725.3	-	-
<i>FOXC2</i>	NM_005251.3	-	-
<i>FOXP3</i>	NM_014009.4	-	-
<i>FOXRED1</i>	NM_017547.4	-	-
<i>FTCD</i>	NM_006657.3	-	-
<i>FTL</i>	NM_000146.4	-	-
<i>FUCA1</i>	NM_000147.4	-	-
<i>FUT8</i>	NM_178155.3	-	-
<i>FXN</i>	NM_000144.5	-	-
<i>G6PC</i>	NM_000151.4	-	-
<i>G6PC3</i>	NM_138387.3	-	-
<i>G6PD</i>	NM_001042351.3	-	Variants will not include pharmacogenomics interpretation. Consider G6PDZ / Glucose-6-Phosphate Dehydrogenase (G6PD) Full Gene Sequencing, Varies for pharmacogenomics interpretation.
<i>GAA</i>	NM_000152.5	chr17:g.78078341T>A (c.-32-13T>A); chr17:g.78090422A>G(c.2190-345A>G)	-
<i>GALC</i>	NM_000153.4	-	CNVs may not be detected in exon 6
<i>GALE</i>	NM_000403.4	-	-
<i>GALK1</i>	NM_000154.2	-	-
<i>GALM</i>	NM_138801.3	-	-
<i>GALNS</i>	NM_000512.5	-	-
<i>GALNT2</i>	NM_004481.5	-	-
<i>GALNT3</i>	NM_004482.4	-	-
<i>GALT</i>	NM_000155.4	chr9:g.34646583_34646586del (c.-119_-116delGTCA); chr9:g.34646606T>G (c.-96T>G); chr9:g.34647597G>A (c.328+33G>A); chr9:g.34648519T>A (c.687+66T>A); chr9:g.34649617C>T (c.1059+56C>T)	-
<i>GAMT</i>	NM_000156.6	-	-
<i>GATA1</i>	NM_002049.4	-	-
<i>GATM</i>	NM_001482.3	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>GBA</i>	NM_000157.4	-	-
<i>GBE1</i>	NM_000158.4	-	-
<i>GCDH</i>	NM_000159.4	-	-
<i>GCH1</i>	NM_000161.3	-	CNVs may not be detected in exon 4
<i>GCK</i>	NM_000162.5	-	-
<i>GCLC</i>	NM_001498.4	-	-
<i>GCSH</i>	NM_004483.5	-	-
<i>GET4</i>	NM_015949.3	-	-
<i>GFAP</i>	NM_002055.5	-	-
<i>GFER</i>	NM_005262.3	-	-
<i>GFM1</i>	NM_024996.5	-	-
<i>GFPT1</i>	NM_002056.4	-	CNVs may not be detected in exon 5
<i>GGT5</i>	NM_004121.3	-	-
<i>GK</i>	NM_000167.5	-	CNVs may not be detected in exon 19
<i>GLA</i>	NM_000169.2	chrX:g.100654735C>T (c.640-801G>A); chrX:g.100654764A>G (c.640-830T>C); chrX:g.100654793G>A (c.640-859C>T)	-
<i>GLB1</i>	NM_000404.4	-	-
<i>GLDC</i>	NM_000170.2	-	-
<i>GLIS3</i>	NM_152629.3	-	-
<i>GLRA1</i>	NM_000171.4	-	-
<i>GLRB</i>	NM_000824.5	-	CNVs may not be detected in exon 4
<i>GLRX5</i>	NM_016417.3	-	-
<i>GLUD1</i>	NM_005271.5	-	-
<i>GLUL</i>	NM_002065.6	-	-
<i>GM2A</i>	NM_000405.5	-	-
<i>GMPPA</i>	NM_205847.3	-	-
<i>GMPPB</i>	NM_013334.3	-	-
<i>GNE</i>	NM_001128227.3	-	-
<i>GNMT</i>	NM_018960.6	-	-
<i>GNPAT</i>	NM_014236.4	-	CNVs may not be detected in exons 15 and 16
<i>GNPTAB</i>	NM_024312.5	-	CNVs may not be detected in exon 4
<i>GNPTG</i>	NM_032520.5	-	-
<i>GNS</i>	NM_002076.4	-	CNVs may not be detected in exons 2 and 7
<i>GOLIM4</i>	NM_014498.5	-	CNVs may not be detected in exons 2 and 3
<i>GORASP2</i>	NM_001201428.2	-	-
<i>GPD1</i>	NM_005276.4	-	CNVs may not be detected in exon 8
<i>GPHN</i>	NM_020806.4	-	-
<i>GPIHBP1</i>	NM_178172.6	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
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	NM_000182.5	-	CNVs may not be detected in exon 14
HADHB	NM_000183.3	-	-
HAL	NM_002108.4	-	CNVs may not be detected in exon 11
HCFC1	NM_005334.3	-	-
HEXA	NM_000520.6	-	-
HEXB	NM_000521.4	-	CNVs may not be detected in exon 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	-	-
HNF4A	NM_175914.4	-	-
HPD	NM_002150.3	-	-
HPRT1	NM_000194.3	-	CNVs may not be detected in exon 5
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	chrX:g.148586802-148586978del (c.-311_-135del177); chrX:g.148578704C>T (c.709-657G>A); chrX:g.148568762 (c.1007-133A>G); IDS/IDSP1 inversion	-
IDUA	NM_000203.5	chr4:g.980786T>C (c.-87T>C)	-
IHH	NM_002181.4	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>IMPDH1</i>	NM_000883.4	-	-
<i>IMPDH2</i>	NM_000884.3	-	-
<i>INSR</i>	NM_000208.4	-	-
<i>INVS</i>	NM_014425.5	-	-
<i>ISCA2</i>	NM_194279.4	-	-
<i>ITPA</i>	NM_033453.4	-	-
<i>IVD</i>	NM_002225.5	-	-
<i>IYD</i>	NM_203395.3	-	-
<i>JAG1</i>	NM_000214.3	-	-
<i>KAT6B</i>	NM_012330.4	-	-
<i>KCNH1</i>	NM_172362.3	-	-
<i>KCNJ11</i>	NM_000525.3	-	-
<i>KCTD7</i>	NM_153033.4	-	-
<i>KHK</i>	NM_000221.3	-	-
<i>KIAA0586</i>	NM_001244189.2	-	Sequence variants and CNVs may not be detected in exons 6 and 33
<i>KIF23</i>	NM_138555.4	-	-
<i>KLF1</i>	NM_006563.5	-	-
<i>KMT2D</i>	NM_003482.3	-	-
<i>KRAS</i>	NM_004985.5	-	-
<i>L2HGDH</i>	NM_024884.3	-	CNVs may not be detected in exon 6
<i>LAMP2</i>	NM_002294.3	-	-
<i>LARGE1</i>	NM_004737.6	-	-
<i>LBR</i>	NM_002296.4	-	-
<i>LCAT</i>	NM_000229.2	-	Sequence variant and CNVs may not be detected in exon 6
<i>LDHA</i>	NM_005566.4	-	-
<i>LDLRAP1</i>	NM_015627.3	-	-
<i>LFNG</i>	NM_001040167.2	-	-
<i>LIAS</i>	NM_006859.4	-	-
<i>LIPA</i>	NM_000235.4	-	-
<i>LIPC</i>	NM_000236.3	-	-
<i>LIPE</i>	NM_005357.4	-	-
<i>LIPG</i>	NM_006033.4	-	-
<i>LIPT1</i>	NM_145199.3	-	-
<i>LMBRD1</i>	NM_018368.4	-	CNVs may not be detected in exon 12
<i>LMF1</i>	NM_022773.4	-	-
<i>LPIN1</i>	NM_145693.4	-	-
<i>LPIN2</i>	NM_014646.2	-	-
<i>LPL</i>	NM_000237.3	-	Sequence variants and CNVs may not be detected in exon 10
<i>LZTR1</i>	NM_006767.4	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>MADD</i>	NM_003682.4	-	-
<i>MAGT1</i>	NM_032121.5	-	-
<i>MAN1B1</i>	NM_016219.5	-	-
<i>MAN2B1</i>	NM_000528.4	-	-
<i>MAN2B2</i>	NM_015274.3	-	-
<i>MANBA</i>	NM_005908.4	-	-
<i>MAOA</i>	NM_000240.3	-	-
<i>MAOB</i>	NM_000898.5	-	-
<i>MAP2K1</i>	NM_002755.3	-	-
<i>MAP2K2</i>	NM_030662.3	-	-
<i>MAT1A</i>	NM_000429.3	-	-
<i>MAT2A</i>	NM_005911.6	-	-
<i>MBTPS1</i>	NM_003791.4	-	-
<i>MCCC1</i>	NM_020166.5	-	-
<i>MCCC2</i>	NM_022132.5	-	-
<i>MCEE</i>	NM_032601.4	-	-
<i>MCM6</i>	NM_005915.6	-	-
<i>MCOLN1</i>	NM_020533.3	-	-
<i>MFSD8</i>	NM_152778.3	-	-
<i>MGAT1</i>	NM_001114618.1	-	-
<i>MGAT2</i>	NM_002408.4	-	-
<i>MGLL</i>	NM_007283.6	-	-
<i>MID1</i>	NM_000381.4	-	-
<i>MKS1</i>	NM_017777.4	-	-
<i>MLYCD</i>	NM_012213.3	-	-
<i>MMAA</i>	NM_172250.3	-	-
<i>MMAB</i>	NM_052845.4	-	-
<i>MMACHC</i>	NM_015506.3	-	-
<i>MMADHC</i>	NM_015702.3	-	-
<i>MMUT</i>	NM_000255.4	-	-
<i>MOCOS</i>	NM_017947.4	-	-
<i>MOCS1</i>	NM_005943.5	-	-
<i>MOCS2</i>	NM_176806.4	-	-
<i>MOCS3</i>	NM_014484.5	-	-
<i>MOGS</i>	NM_006302.3	-	-
<i>MPC1</i>	NM_016098.4	-	Sequence variants and CNVs may not be detected in exon 2
<i>MPDU1</i>	NM_004870.4	-	-
<i>MPI</i>	NM_002435.3	-	-
<i>MPV17</i>	NM_002437.5	-	-
<i>MRPL3</i>	NM_007208.4	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
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	NM_024408.4	-	Sequence variants and CNVs in exons 1–4 will not be detected
NPC1	NM_000271.5	chr18:g.21132700C>T (c.1554-1009G>A)	-
NPC2	NM_006432.4	-	-
NPHP1	NM_000272.4	-	CNVs may not be detected in exon 3
NPHP3	NM_153240.5	-	CNVs may not be detected in exons 8 and 22
NPHP4	NM_015102.5	-	-
NR1H4	NM_005123.4	-	CNVs may not be detected in exon 3
NRAS	NM_002524.5	-	-
NSDHL	NM_015922.3	-	-
NT5C3A	NM_016489.13	-	CNVs may not be detected in exons 5 and 6
NUS1	NM_138459.5	-	CNVs may not be detected in exon 5
OAT	NM_000274.4	-	-
OGDH	NM_002541.4	-	-
OPA1	NM_015560.2	-	-
OPA3	NM_025136.4	-	-
OPLAH	NM_017570.5	-	-
OTC	NM_000531.6	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>OXCT1</i>	NM_000436.4	-	Sequence variants and CNVs in exon 17 may not be detected
<i>PAH</i>	NM_000277.3	-	-
<i>PANK2</i>	NM_153638.3	-	-
<i>PAPSS2</i>	NM_001015880.2	-	-
<i>PAX8</i>	NM_003466.4	-	-
<i>PC</i>	NM_000920.4	-	-
<i>PCBD1</i>	NM_000281.4	-	-
<i>PCCA</i>	NM_000282.4	-	CNVs may not be detected in exon 10
<i>PCCB</i>	NM_000532.5	-	-
<i>PCK1</i>	NM_002591.4	-	-
<i>PCK2</i>	NM_004563.4	-	-
<i>PCSK9</i>	NM_174936.4	-	-
<i>PCYT1A</i>	NM_005017.4	-	-
<i>PDHA1</i>	NM_000284.4	-	-
<i>PDHA2</i>	NM_005390.5	-	-
<i>PDHB</i>	NM_000925.4	-	-
<i>PDHX</i>	NM_003477.3	-	-
<i>PDP1</i>	NM_018444.4	-	-
<i>PDX1</i>	NM_000209.4	-	-
<i>PDXK</i>	NM_003681.5	-	-
<i>PEPD</i>	NM_000285.4	-	-
<i>PEX1</i>	NM_000466.3	-	-
<i>PEX10</i>	NM_153818.1	-	-
<i>PEX11B</i>	NM_003846.3	-	-
<i>PEX12</i>	NM_000286.3	-	-
<i>PEX13</i>	NM_002618.4	-	-
<i>PEX14</i>	NM_004565.3	-	-
<i>PEX16</i>	NM_057174.2	-	-
<i>PEX19</i>	NM_002857.3	-	-
<i>PEX2</i>	NM_000318.3	-	-
<i>PEX26</i>	NM_017929.6	-	-
<i>PEX3</i>	NM_003630.3	-	-
<i>PEX5</i>	NM_001131023.1	-	-
<i>PEX6</i>	NM_000287.4	-	-
<i>PEX7</i>	NM_000288.4	-	-
<i>PFKM</i>	NM_000289.6	-	-
<i>PGAM2</i>	NM_000290.4	-	-
<i>PGAP2</i>	NM_001256240.2	-	-
<i>PGAP3</i>	NM_033419.5	-	-
<i>PGK1</i>	NM_000291.4	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>PGM1</i>	NM_002633.3	chr1:g.64124734G>A (c.1600-523G>A)	-
<i>PGM2</i>	NM_018290.4	-	-
<i>PGM3</i>	NM_001199917.2	-	-
<i>PHGDH</i>	NM_006623.4	-	-
<i>PHKA1</i>	NM_002637.4	-	-
<i>PHKA2</i>	NM_000292.3	-	-
<i>PHKB</i>	NM_000293.3	-	CNVs may not be detected in exons 6 and 11
<i>PHKG2</i>	NM_000294.3	-	-
<i>PHYH</i>	NM_006214.4	-	-
<i>PIEZO1</i>	NM_001142864.4	-	-
<i>PIGA</i>	NM_002641.3	-	-
<i>PIGL</i>	NM_004278.4	-	-
<i>PIGM</i>	NM_145167.3	chr1:g.160001799G>C (c.-270C>G)	-
<i>PIGN</i>	NM_176787.5	-	CNVs may not be detected in exon 14
<i>PIGO</i>	NM_032634.4	-	-
<i>PIGT</i>	NM_015937.6	-	-
<i>PIGV</i>	NM_017837.3	-	-
<i>PIGW</i>	NM_178517.4	-	-
<i>PIGY</i>	NM_001042616.2	-	-
<i>PIPOX</i>	NM_016518.3	-	-
<i>PKHD1</i>	NM_138694.4	-	-
<i>PKLR</i>	NM_000298.6	-	-
<i>PLA2G6</i>	NM_003560.4	-	-
<i>PLIN1</i>	NM_002666.5	-	-
<i>PLPBP</i>	NM_007198.4	-	-
<i>PMM1</i>	NM_002676.3	-	-
<i>PMM2</i>	NM_000303.3	chr16:g.8926102C>T (c.640-15479C>T)	-
<i>PNP</i>	NM_000270.3	-	-
<i>PNPLA2</i>	NM_020376.4	-	-
<i>PNPLA6</i>	NM_006702.5	-	-
<i>PNPLA8</i>	NM_015723.5	-	-
<i>PNPO</i>	NM_018129.4	-	-
<i>POFUT1</i>	NM_015352.2	-	-
<i>POGLUT1</i>	NM_152305.3	-	CNVs may not be detected in exon 10
<i>POLG</i>	NM_002693.2	-	-
<i>POMGNT1</i>	NM_017739.3	-	-
<i>POMGNT2</i>	NM_032806.6	-	-
<i>POMK</i>	NM_032237.5	-	-
<i>POMT1</i>	NM_007171.3	-	-
<i>POMT2</i>	NM_013382.5	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>POR</i>	NM_000941.3	-	-
<i>PPARG</i>	NM_015869.4	-	-
<i>PPM1K</i>	NM_152542.5	-	-
<i>PPOX</i>	NM_000309.5	-	-
<i>PPT1</i>	NM_000310.3	-	-
<i>PRDX1</i>	NM_002574.3	-	-
<i>PREPL</i>	NM_006036.4	-	-
<i>PRKAG2</i>	NM_016203.4	-	CNVs may not be detected in exon 13
<i>PRKCSH</i>	NM_002743.3	-	-
<i>PRODH</i>	NM_016335.5	-	-
<i>PRPS1</i>	NM_002764.4	-	-
<i>PSAP</i>	NM_002778.4	chr10:g.73583679G>T (c.777+1915C>A)	-
<i>PSAT1</i>	NM_058179.4	-	-
<i>PSPH</i>	NM_004577.4	-	-
<i>PTDSS1</i>	NM_014754.3	-	-
<i>PTH1R</i>	NM_000316.3	-	-
<i>PTPN11</i>	NM_002834.4	-	-
<i>PTS</i>	NM_000317.3	-	-
<i>PYCR1</i>	NM_006907.4	-	-
<i>PYCR2</i>	NM_013328.4	-	-
<i>PYCR3</i>	NM_023078.6	-	-
<i>PYGL</i>	NM_002863.5	-	-
<i>PYGM</i>	NM_005609.4	-	-
<i>PYY</i>	NM_004160.5	-	-
<i>QDPR</i>	NM_000320.3	-	-
<i>RAF1</i>	NM_002880.3	-	-
<i>RASA1</i>	NM_002890.3	-	-
<i>RBCK1</i>	NM_031229.4	-	-
<i>RFT1</i>	NM_052859.4	-	-
<i>RIT1</i>	NM_006912.6	-	-
<i>RNF216</i>	NM_207111.4	-	-
<i>RPIA</i>	NM_144563.3	-	-
<i>RPL11</i>	NM_000975.5	-	-
<i>RPL35A</i>	NM_000996.4	-	-
<i>RPL5</i>	NM_000969.5	-	-
<i>RPS10</i>	NM_001014.5	-	-
<i>RPS19</i>	NM_001022.4	-	-
<i>RPS24</i>	NM_033022.4	-	-
<i>RPS26</i>	NM_001029.5	-	-
<i>RXYLT1</i>	NM_014254.3	-	-
<i>SAR1B</i>	NM_001033503.3	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>SARDH</i>	NM_007101.4	-	-
<i>SC5D</i>	NM_006918.5	-	-
<i>SCARB1</i>	NM_005505.5	-	-
<i>SCGB1D2</i>	NM_006551.4	-	-
<i>SCP2</i>	NM_002979.5	-	CNVs may not be detected in exon 7
<i>SEC23A</i>	NM_006364.4	-	-
<i>SEC23B</i>	NM_006363.6	chr20:g.18491776A>G (c.221+76A>G); chr20:g.18492791C>T (c.222-78C>T); chr20:g.18523885_18523888del (c.1665+69_1665+72delCTTA); chr20:g.18535856dup (c.2214+39dupA); chr20:g.18535896T>G (c.2214+79T>G); chr20:g.18535916G>A (c.2214+99G>A)	-
<i>SEC63</i>	NM_007214.5	-	-
<i>SERAC1</i>	NM_032861.4	-	CNVs may not be detected in exon 3
<i>SERPINA1</i>	NM_000295.5	-	-
<i>SGSH</i>	NM_000199.5	-	-
<i>SHH</i>	NM_000193.4	-	-
<i>SHMT1</i>	NM_004169.5	-	-
<i>SHOC2</i>	NM_007373.3	-	-
<i>SHPK</i>	NM_013276.4	-	-
<i>SLC10A1</i>	NM_003049.4	-	-
<i>SLC10A2</i>	NM_000452.3	-	-
<i>SLC10A7</i>	NM_001300842.3	-	-
<i>SLC16A1</i>	NM_003051.3	-	-
<i>SLC16A2</i>	NM_006517.5	-	-
<i>SLC17A5</i>	NM_012434.5	-	-
<i>SLC18A2</i>	NM_003054.6	-	-
<i>SLC19A1</i>	NM_194255.4	-	-
<i>SLC19A2</i>	NM_006996.3	-	-
<i>SLC19A3</i>	NM_025243.4	-	-
<i>SLC1A1</i>	NM_004170.6	-	-
<i>SLC22A5</i>	NM_003060.4	-	-
<i>SLC25A1</i>	NM_005984.5	-	-
<i>SLC25A13</i>	NM_014251.3	-	-
<i>SLC25A15</i>	NM_014252.4	-	-
<i>SLC25A19</i>	NM_021734.4	-	-
<i>SLC25A20</i>	NM_000387.6	-	-
<i>SLC25A29</i>	NM_001039355.3	-	-
<i>SLC25A32</i>	NM_030780.5	-	-
<i>SLC25A38</i>	NM_017875.4	-	-
<i>SLC26A2</i>	NM_000112.4	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
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	NM_006416.5	-	CNVs may not be detected in exon 1
SLC35A2	NM_001042498.3	-	-
SLC35A3	NM_012243.3	-	CNVs may not be detected in exon 6
SLC35C1	NM_018389.5	-	-
SLC35D1	NM_015139.3	-	CNVs may not be detected in exon 11
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	NM_006939.4	-	-
SOX18	NM_018419.3	-	-
SPR	NM_003124.5	-	-
SRD5A3	NM_024592.5	-	-
SRR	NM_021947.3	-	-
SSR3	NM_007107.4	-	-
SSR4	NM_006280.3	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>ST3GAL3</i>	NM_006279.5	-	-
<i>ST3GAL5</i>	NM_003896.4	-	-
<i>STS</i>	NM_000351.6	-	-
<i>STT3A</i>	NM_001278503.2	-	-
<i>STT3B</i>	NM_178862.3	-	-
<i>STXBP1</i>	NM_003165.4	-	-
<i>SUCLA2</i>	NM_003850.2	-	-
<i>SUCLG1</i>	NM_003849.4	-	-
<i>SUGCT</i>	NM_024728.2	-	Sequence variants and CNVs may not be detected in exon 13
<i>SUMF1</i>	NM_182760.4	-	-
<i>SUOX</i>	NM_000456.3	-	-
<i>SYP</i>	NM_003179.2	-	-
<i>TALDO1</i>	NM_006755.2	-	-
<i>TANGO2</i>	NM_152906.7	-	-
<i>TAT</i>	NM_000353.3	-	-
<i>TAZ</i>	NM_000116.5	-	-
<i>TBC1D24</i>	NM_001199107.2	-	-
<i>TCN1</i>	NM_001062.4	-	-
<i>TCN2</i>	NM_000355.4	-	-
<i>TF</i>	NM_001063.4	-	-
<i>TG</i>	NM_003235.5	-	-
<i>TH</i>	NM_199292.3	-	-
<i>THAP11</i>	NM_020457.3	-	-
<i>THRA</i>	NM_199334.4	-	-
<i>TIMM50</i>	NM_001001563.5	-	-
<i>TJP2</i>	NM_004817.4	-	-
<i>TKT</i>	NM_001258028.1	-	Sequence variants and CNVs may not be detected in exon 5
<i>TMEM165</i>	NM_018475.5	chr4:g.56284334G>A (c.792+182G>A)	-
<i>TMEM199</i>	NM_152464.3	-	-
<i>TMEM216</i>	NM_001173990.3	-	-
<i>TMEM70</i>	NM_017866.6	-	-
<i>TPH1</i>	NM_004179.3	-	-
<i>TPK1</i>	NM_022445.4	-	-
<i>TPO</i>	NM_000547.5	-	-
<i>TPP1</i>	NM_000391.4	-	-
<i>TRAPPC11</i>	NM_021942.6	-	-
<i>TRAPPC9</i>	NM_031466.7	-	-
<i>TRIM37</i>	NM_015294.6	-	-
<i>TRIP11</i>	NM_004239.4	-	-

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

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>TRMU</i>	NM_018006.5	-	-
<i>TSHB</i>	NM_000549.5	-	-
<i>TSHR</i>	NM_000369.3	-	-
<i>TSTA3</i>	NM_003313.4	-	-
<i>TTC19</i>	NM_017775.4	-	-
<i>TUFM</i>	NM_003321.5	-	-
<i>TUSC3</i>	NM_006765.4	-	CNVs may not be detected in exon 10
<i>UCP2</i>	NM_003355.2	-	-
<i>UGT1A1</i>	NM_000463.3	Promotor TA repeat region c.-111 to c.-19	Variants will not include pharmacogenomics interpretation. Consider UGTFZ / UDP-Glucuronosyltransferase 1A1 (UGT1A1), Full Gene Sequencing, Varies for pharmacogenomics interpretation.
<i>UMPS</i>	NM_000373.4	-	-
<i>UPB1</i>	NM_016327.3	-	-
<i>UQCRB</i>	NM_006294.4	-	-
<i>UQCRC2</i>	NM_003366.4	-	-
<i>UQCRQ</i>	NM_014402.5	-	-
<i>UROD</i>	NM_000374.5	-	-
<i>UROS</i>	NM_000375.3	-	-
<i>VARS2</i>	NM_001167734.1	-	-
<i>VIPAS39</i>	NM_022067.4	-	-
<i>VMA21</i>	NM_001017980.3	-	-
<i>VPS33A</i>	NM_022916.6	-	-
<i>VPS33B</i>	NM_018668.4	-	-
<i>WDR35</i>	NM_001006657.2	-	-
<i>WFS1</i>	NM_006005.3	-	-
<i>XDH</i>	NM_000379.4	-	-
<i>XYLT1</i>	NM_022166.4	-	-
<i>ZNF143</i>	NM_003442.6	-	-

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

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
ABCD1	ABCD1 Gene Analysis	ABCD1
ACADM	ACADM Gene Analysis	ACADM
ACADV	ACADVL Gene Analysis	ACADVL
APGP	Acute Porphyria Gene Panel	ALAD, CPOX, HMBS, PPOX
BTD	BTD Gene Analysis	BTD
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, FKR, 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
DPYDZ	DPYD Full Gene Sequencing	DPYD
DHCRZ	Smith Lemli Optiz, DHCR7 Gene, Full Gene Analysis	DHCR7

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

Available Inborn Errors of Metabolism Panels (continued)

Test ID	Test Name	Genes
GA2P	Glutaric Aciduria Type II Gene Panel	ETFA, ETFB, ETFDH, FLAD1, SLC52A1, SLC52A2, SLC52A3, TANGO2
GAAN	GAA Gene Analysis	GAA
GALC	GALC Gene Analysis	GALC
GALZ	Galactosemia, GALT Gene, Full Gene Analysis	GALT
GBA	GBA1 Gene Analysis	GBA1
GLA	GLA Gene Analysis	GLA
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
HEXAN	HEXA Gene Analysis	HEXA
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, EC1, 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
IDS	IDS Gene Analysis	IDS
IDUA	IDUA Gene Analysis	IDUA
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

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

Available Inborn Errors of Metabolism Panels (continued)

Test ID	Test Name	Genes
<i>PCGP</i>	Porphyria Comprehensive Gene Panel	<i>ALAD, ALAS2, CLPX, CPOX, FECH, GATA1, HFE, HMBS, PPOX, UROD, UROS</i>
<i>PDGP</i>	Peroxisomal Disorder Gene Panel	<i>ABCD1, ABCD3, ACOX1, ACOX3, AGPS, AMACR, CAT, DNMT1L, GNPAT, HSD17B4, PEX1, PEX10, PEX11B, PEX12, PEX13, PEX14, PEX16, PEX19, PEX2, PEX26, PEX3, PEX5, PEX6, PEX7, PHYH, SCP2, SUGCT, TRIM37</i>
<i>PHEGP</i>	Phenylalanine Disorders Gene Panel	<i>DDC, DNAJC12, GCH1, PAH, PCBD1, PTS, QDPR, SLC18A2, SPR, TH</i>
<i>TYRGP</i>	Tyrosine Disorders Gene Panel	<i>FAH, HGD, HPD, TAT</i>
<i>UCDP</i>	Urea Cycle Disorders Gene Panel	<i>ALDH18A1, ARG1, ARG2, ASL, ASS1, CA5A, CPS1, GLUD1, GLUL, NAGS, OAT, OTC, SLC25A13, SLC25A15, SLC7A7, UMPS</i>
<i>WNDZ</i>	ATP7B Full Gene Analysis	<i>ATP7B</i>