

## Targeted Genes and Methodology Details for Inborn Errors of Immunity/Bone Marrow Failure/ Telomeropathy/Pulmonary Fibrosis/Very Early Onset IBD/Pancreatitis Custom Gene Panel

The following applies to Inborn Errors of Immunity/Bone Marrow Failure/Telomeropathy/Pulmonary Fibrosis/Very Early Onset IBD/Pancreatitis Custom Gene Panel. Testing is performed to evaluate for the presence of variants in coding regions and extending to +/- 10 base pairs of adjacent intronic sequence on either side of the coding exons of the genes analyzed. In addition, the analysis will cover select noncoding 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 July 2025 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, or 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
<i>ABCA3</i>	NM_001089.3	chr16:g.2358644C>T (c.1112-20G>A); chr16:g.2333457G>A (c.3863-98C>T)	-
<i>ABCB7</i>	NM_004299.6	-	-
<i>ACD</i>	NM_001082486.2	-	-
<i>ACP5</i>	NM_001111035.3	-	-
<i>ACTB</i>	NM_001101.5	-	-
<i>ADA</i>	NM_000022.4	chr20:g.43249076C>T (c.976-34G>A); chr20:g.43248503A>T (c.1079-15T>A)	-
<i>ADA2</i>	NM_001282225.2	-	-
<i>ADAM17</i>	NM_003183.6	-	-
<i>ADAR</i>	NM_001111.5	-	-
<i>AICDA</i>	NM_020661.4	-	-
<i>AIRE</i>	NM_000383.4	-	-
<i>AK2</i>	NM_001625.4	-	-
<i>ALAS2</i>	NM_000032.5	chrX:g.55054636 to chrX:g.55054634 (c.-15-2188 to c.-15-2186); chrX:g.55054628_55054663del (c.-15-2214_-15-2179del)	-
<i>ALPI</i>	NM_001631.5	-	-
<i>ANGPT1</i>	NM_001146.5	-	-
<i>ANKRD26</i>	NM_014915.2	chr10:g.27389395 to chr10:g.27389368 (c.-140 to c.-113)	CNV analysis in exon 19 is not performed
<i>ANKZF1</i>	NM_018089.3	-	-
<i>AP1S3</i>	NM_001039569.2	-	-
<i>AP3B1</i>	NM_003664.4	-	-
<i>AP3D1</i>	NM_001261826.3	-	-
<i>APOL1</i>	NM_003661.4	-	-
<i>ARHGEF1</i>	NM_199002.2	-	-
<i>ARPC1B</i>	NM_005720.4	-	-
<i>ASAH1</i>	NM_177924.5	-	CNV analysis in exon 4 is not performed
<i>ATM</i>	NM_000051.3	chr11:g.108138753A>G (c.2639-384A>G); chr11:g.108139118_108139130del (c.2639-19_2639-7del); chr11:g.108141212_108141215del (c.2839-579_2839-576del); chr11:g.108164028A>G (c.4612-12A>G); chr11:g.108179837A>G (c.5763-1050A>G); chr11:g.108217993_108218019del (c.8585-13_8598del)	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>ATP6AP1</i>	NM_001183.6	-	-
<i>ATR</i>	NM_001184.4	-	-
<i>B2M</i>	NM_004048.3	-	-
<i>BACH2</i>	NM_021813.4	-	-
<i>BCL10</i>	NM_003921.5	-	-
<i>BCL11B</i>	NM_138576.4	-	-
<i>BLM</i>	NM_000057.4	-	-
<i>BLNK</i>	NM_013314.3	-	-
<i>BLOC1S6</i>	NM_012388.3	-	-
<i>BRCA1</i>	NM_007294.4	-	-
<i>BRCA2</i>	NM_000059.3	-	-
<i>BRIP1</i>	NM_032043.3	-	-
<i>BTK</i>	NM_000061.2	chrX:g.100641212T>C (c.-193A>G); chrX:g.100629827T>C (c.142-205A>G); chrX:g.100629416A>C (c.240+108T>G); chrX:g.100629415G>T (c.240+109C>A); chrX:g.100613695G>T (c.895-11C>A); chrX:g.100613290_100613300del (c.1102+2_1102+12del); chrX:g.100612471_100612472insTTTTTCTACC (c.1177+28_1177+29insCACCTTTTTCT); chrX:g.100609705T>G (c.1567-23A>C); chrX:g.100609705T>C (c.1567-23A>G); chrX:g.100609693_100609696del (c.1567-12_1567-9del)	-
<i>C15orf41 (CDIN1)</i>	NM_001130010.3	-	-
<i>C1QA</i>	NM_015991.4	-	-
<i>C1QB</i>	NM_000491.5	-	-
<i>C1QC</i>	NM_172369.5	-	-
<i>C1R</i>	NM_001733.7	Whole gene deletion and duplication performed	CNV analysis in individual exons 1-7 is not performed
<i>C1S</i>	NM_201442.4	-	-
<i>C2</i>	NM_000063.6	chr6:g.31902068_31902095del (c.841_849+19del); chr6:g.31911326_31911347del (c.1567+22_1567+43del)	-
<i>C3</i>	NM_000064.4	-	-
<i>C5</i>	NM_001735.3	-	-
<i>C6</i>	NM_000065.4	-	-
<i>C7</i>	NM_000587.4	chr5:g.40931143T>A (c.63-23T>A)	CNV analysis in exon 14 is not performed
<i>C8A</i>	NM_000562.3	chr1:g.57351588G>A (c.856-12G>A)	-
<i>C8B</i>	NM_000066.4	-	-
<i>C8G</i>	NM_000606.3	-	-
<i>C9</i>	NM_001737.5	-	-
<i>CARD11</i>	NM_032415.6	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>CARD14</i>	NM_024110.4	-	-
<i>CARD9</i>	NM_052813.5	-	-
<i>CARMIL2</i>	NM_001013838.3	-	-
<i>CASP10</i>	NM_032977.4	-	-
<i>CASP8</i>	NM_001228.4	-	-
<i>CASR</i>	NM_000388.4	-	-
<i>CBL</i>	NM_005188.4	chr11:g.119148864_119148876del (c.1096-12_1096del); chr11:g.119148865_119148889del (c.1096-11_1109del); chr11:g.119148768_119149410del (c.1096-108_1418del); chr11:g.119148997_119149018del (c.1217_1227+11del)	-
<i>CCBE1</i>	NM_133459.4	-	-
<i>CD19</i>	NM_001770.6	-	-
<i>CD247</i>	NM_198053.2	-	-
<i>CD27</i>	NM_001242.4	-	-
<i>CD28</i>	NM_006139.4	-	-
<i>CD3D</i>	NM_000732.4	-	-
<i>CD3E</i>	NM_000733.4	-	CNV analysis in exon 4 is not performed
<i>CD3G</i>	NM_000073.2	-	-
<i>CD40</i>	NM_001250.6	-	-
<i>CD40LG</i>	NM_000074.3	-	-
<i>CD46</i>	NM_002389.4	-	CNV analysis in exon 2 is not performed
<i>CD48</i>	NM_001778.4	-	-
<i>CD55</i>	NM_000574.5	-	-
<i>CD59</i>	NM_203330.2	-	-
<i>CD70</i>	NM_001252.5	-	-
<i>CD79A</i>	NM_001783.4	-	-
<i>CD79B</i>	NM_000626.4	-	-
<i>CD81</i>	NM_004356.3	-	CNV analysis in exon 1 is not performed
<i>CD8A</i>	NM_001768.6	-	-
<i>CDAN1</i>	NM_138477.4	-	-
<i>CDC42</i>	NM_001791.4	-	-
<i>CDCA7</i>	NM_031942.5	-	-
<i>CEBPA</i>	NM_004364.4	-	-
<i>CEBPE</i>	NM_001805.3	-	-
<i>CEL</i>	NM_001807.5	-	Sequence and CNV analysis in exons 1, 8, 9, and 11 are not performed
<i>CFB</i>	NM_001710.6	-	-
<i>CFD</i>	NM_001928.4	-	-
<i>CFH</i>	NM_000186.4	-	CNV analysis in exon 20 is not performed
<i>CFI</i>	NM_000204.4	-	CNV analysis in exon 7 is not performed
<i>CFP</i>	NM_002621.2	-	-

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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.117179041_117179044del (c.870-1113_870-1110del);                      chr7:g.117199500G&gt;A (c.1393-18G&gt;A);                      chr7:g.117227784T&gt;A (c.1585-9T&gt;A);                      chr7:g.117227785G&gt;A (c.1585-8G&gt;A);                      chr7:g.117218381A&gt;G (c.1585-9412A&gt;G);                      chr7:g.117229521A&gt;G (c.1680-886A&gt;G);                      chr7:g.117229524A&gt;G (c.1680-883A&gt;G);                      chr7:g.117229530G&gt;T (c.1680-877G&gt;T);                      chr7:g.117246713T&gt;G (c.2909-15T&gt;G);                      chr7:g.117250260A&gt;T (c.2989-313A&gt;T);                      chr7:g.117251609A&gt;G (c.3140-26A&gt;G);                      chr7:g.117251619T&gt;A (c.3140-16T&gt;A);                      chr7:g.117266272C&gt;G (c.3469-1304C&gt;G);                      chr7:g.117267864A&gt;G (c.3717+40A&gt;G);                      chr7:g.117280015C&gt;T (c.3718-2477C&gt;T);                      chr7:g.117288374A&gt;G (c.3874-4522A&gt;G)</p>	CNV analysis in exon 13 is not performed
<i>CHD7</i>	NM_017780.4	chr8:g.61763035 to chr8:g.61763039 (c.5405-17 to c.5405-13)	-
<i>CHEK2</i>	NM_007194.4	-	-
<i>CIB1</i>	NM_006384.4	-	-
<i>CIITA</i>	NM_000246.3	-	-
<i>CLCN7</i>	NM_001287.6	-	CNV analysis in exon 17 is not performed
<i>CLDN2</i>	NM_020384.4	-	-
<i>CLPB</i>	NM_030813.6	-	-
<i>COL7A1</i>	NM_000094.3	<p>chr3:g.48629915C&gt;T (c.977-15G&gt;A);                      chr3:g.48616971T&gt;C (c.5236-23A&gt;G);                      chr3:g.48613354T&gt;C (c.5821-19A&gt;G);                      chr3:g.48610175_48610195del (c.6832-23_6832-3del);                      chr3:g.48605981_48605985delinsTGAA                      (c.7759-18_7759-14delinsTTCA);                      chr3:g.48605248_48605263del (c.7929+11_7929+26del);                      chr3:g.48604008A&gt;T (c.8305-12T&gt;A);                      chr3:g.48602928_48602947del (c.8441-15_8445del);                      chr3:g.48602928_48602948del (c.8441-14_8447del)</p>	-
<i>COPA</i>	NM_004371.4	-	-
<i>CORO1A</i>	NM_007074.3	-	CNV analysis in exon 11 is not performed
<i>CPA1</i>	NM_001868.4	-	-
<i>CR2</i>	NM_001006658.2	-	-
<i>CSF2RA</i>	NM_006140.5	-	-
<i>CSF2RB</i>	NM_000395.3	-	-
<i>CSF3R</i>	NM_000760.4	-	-
<i>CTC1</i>	NM_025099.6	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>CTLA4</i>	NM_005214.5	-	-
<i>CTPS1</i>	NM_001905.4	-	-
<i>CTRC</i>	NM_007272.3	-	-
<i>CTSC</i>	NM_001814.6	chr11:g.88070895G>T (c.-55C>A)	-
<i>CXCR2</i>	NM_001557.4	-	-
<i>CXCR4</i>	NM_003467.3	-	-
<i>CXXC5</i>	NM_016463.9	-	-
<i>CYBA</i>	NM_000101.4	chr16:g.88712620G>C (c.288-15C>G)	-
<i>CYBB</i>	NM_000397.4	chrX:g.37639262 to chrX:g.37639267 (c.-69 to c.-64); chrX:g.37641327_37641330delinsGAA (c.46-14_46-11delinsGAA); chrX:g.37642715_37642731del (c.142-28_142-12del); chrX:g.37642731delinsACCTCTCTAG (c.142-12delinsACCTCTCTAG)	-
<i>CYBC1</i>	NM_001033046.4	-	-
<i>CYCS</i>	NM_018947.6	-	-
<i>DBR1</i>	NM_016216.4	-	-
<i>DCLRE1B</i>	NM_022836.4	-	-
<i>DCLRE1C</i>	NM_001033855.3	-	-
<i>DDX41</i>	NM_016222.4	-	-
<i>DDX58 (RIGI)</i>	NM_014314.4	-	-
<i>DEF6</i>	NM_022047.4	-	-
<i>DGAT1</i>	NM_012079.6	-	-
<i>DGKE</i>	NM_003647.3	chr17:g.54925466A>G ( c.888+40A>G)	-
<i>DHFR</i>	NM_000791.4	-	CNV analysis in exon 6 is not performed
<i>DIAPH1</i>	NM_005219.5	-	-
<i>DKC1</i>	NM_001363.5	chrX:g.153991099C>G (c.-142C>G)	-
<i>DNAJC21</i>	NM_001012339.3	-	-
<i>DNASE1</i>	NM_005223.3	-	-
<i>DNASE1L3</i>	NM_004944.4	-	-
<i>DNASE2</i>	NM_001375.3	-	-
<i>DNMT3B</i>	NM_006892.4	chr20:g.31395557G>A (c.2421-11G>A)	-
<i>DOCK2</i>	NM_004946.3	-	-
<i>DOCK8</i>	NM_203447.3	chr9:g.317025 to chr9:g.317028 (c.742-18 to c.742-15); chr9:g.372260_372348del (c.2083_2109+62del)	-
<i>DUOX2</i>	NM_014080.4	-	CNV analysis in exon 7 is not performed
<i>EFL1</i>	NM_024580.6	-	-
<i>ELANE</i>	NM_001972.4	-	-
<i>EPCAM</i>	NM_002354.3	chr2:g.47596641T>C (c.-4T>C); chr2:g.47606078A>G (c.556-14A>G)	-
<i>EPG5</i>	NM_020964.3	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>EPO</i>	NM_000799.4	-	-
<i>ERBIN</i>	NM_001253697.2	-	CNV analysis in exon 7 is not performed
<i>ERCC4</i>	NM_005236.3	-	-
<i>ERCC6L2</i>	NM_020207.5	-	-
<i>ETV6</i>	NM_001987.5	-	-
<i>EXTL3</i>	NM_001440.4	-	-
<i>F12</i>	NM_000505.3	-	-
<i>FADD</i>	NM_003824.3	-	-
<i>FANCA</i>	NM_000135.4	chr16:g.89864654G>T (c.893+920C>A); chr16:g.89849346T>C (c.1567-20A>G); chr16:g.89836805T>C (c.2223-138A>G); chr16:g.89836111T>C (c.2504+134A>G); chr16:g.89831215G>C (c.2778+83C>G); chr16:g.89816056A>C (c.3239+82T>G); chr16:g.89805128_89805135del (c.4261-19_4261-12del)	-
<i>FANCB</i>	NM_001018113.3	-	-
<i>FANCC</i>	NM_000136.3	-	-
<i>FANCD2</i>	NM_033084.5	chr3:g.10083186C>G (c.696-121C>G); chr3:g.10102127T>G (c.1766+40T>G); chr3:g.10106024T>G (c.1948-16T>G)	CNV analysis in exons 13, 14, and 17 is not performed
<i>FANCE</i>	NM_021922.3	-	-
<i>FANCF</i>	NM_022725.4	-	-
<i>FANCG</i>	NM_004629.1	-	-
<i>FANCI</i>	NM_001113378.1	-	-
<i>FANCL</i>	NM_018062.3	-	-
<i>FANCM</i>	NM_020937.4	-	CNV analysis in exon 8 is not performed
<i>FAS</i>	NM_000043.6	-	-
<i>FASLG</i>	NM_000639.3	chr1:g.172628081T>C (c.-261T>C)	-
<i>FAT4</i>	NM_024582.4	-	-
<i>FCGR3A</i>	NM_000569.7	-	CNV analysis is not performed
<i>FCHO1</i>	NM_015122.3	-	-
<i>FERMT1</i>	NM_017671.4	chr20:g.6103422T>C (c.-20A>G); chr20:g.6074846C>T (c.1139+740G>A)	CNV analysis in exon 9 is not performed
<i>FERMT3</i>	NM_031471.6	-	-
<i>FLI1</i>	NM_002017.5	-	-
<i>FLNA</i>	NM_001456.3	chrX:g.153592001A>T (c.2280+389T>A); chrX:g.153581590_153581601del (c.5999-27_5999-16del)	-
<i>FOXI3</i>	NM_001135649.3	-	-
<i>FOXN1</i>	NM_003593.2	-	-
<i>FOXP3</i>	NM_014009.4	chrX:c.-105_-23+1303del1386 (also known as g.del-6247_-4859); chrX:g.49114969C>A (c.-7G>T); chrX:g.49106919 to chrX:g.49106917 (c.*876 to c.*878)	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>FYB1</i>	NM_001465.6	-	Sequence and CNV analysis in exon 5 is not performed, CNV analysis in exon 18 is not performed
<i>G6PC</i> ( <i>G6PC1</i> )	NM_000151.4	-	-
<i>G6PC3</i>	NM_138387.3	-	-
<i>G6PD</i>	NM_001042351.3	-	-
<i>GATA1</i>	NM_002049.4	chrX:g.48652176C>T (c.871-24C>T)	-
<i>GATA2</i>	NM_032638.5	chr3:g.128202114 to chr3:g.128202177 (c.1017+526 to c.1017+589 corresponding to a highly conserved intronic region); chr3: g.128200787_128200806dup (c.1018-17_1020dup)	-
<i>GFI1</i>	NM_005263.5	-	-
<i>GFI1B</i>	NM_004188.7	-	-
<i>GIN1</i>	NM_021067.5	chr20:g.25388397A>G (c.-60A>G); chr20:g.25388409C>G (c.-48C>G);	-
<i>GLA</i>	NM_000169.2	chrX:g.100653945A>T (c.640-11T>A); chrX:g.100654735C>T (c.640-801G>A); chrX:g.100653097C>T (c.1000-10G>A)	-
<i>GLRX5</i>	NM_016417.3	-	-
<i>GP1BA</i>	NM_000173.7	-	-
<i>GP1BB</i>	NM_000407.5	chr22:g.19710933C>G (c.-160C>G)	-
<i>GP9</i>	NM_000174.4	-	-
<i>GUCY2C</i>	NM_004963.4	-	-
<i>HAVCR2</i>	NM_032782.5	-	-
<i>HAX1</i>	NM_006118.4	-	-
<i>HELLS</i>	NM_018063.5	-	CNV analysis in exon 4 is not performed, sequence and CNV analysis in exon 7 is not performed
<i>HMOX1</i>	NM_002133.3	-	-
<i>HOXA11</i>	NM_005523.6	-	-
<i>HPS1</i>	NM_000195.5	chr10:g.100193862C>T (c.399-14G>A)	-
<i>HPS3</i>	NM_032383.5	-	-
<i>HPS4</i>	NM_022081.5	-	-
<i>HPS6</i>	NM_024747.6	-	-
<i>HS3ST6</i>	NM_001009606.4	-	Sequence and CNV analysis in exon 1 is not performed
<i>HYOU1</i>	NM_006389.5	-	-
<i>ICOS</i>	NM_012092.4	-	-
<i>ICOSLG</i>	NM_015259.6	-	-
<i>IFIH1</i>	NM_022168.4	-	-
<i>IFNAR1</i>	NM_000629.3	-	-
<i>IFNAR2</i>	NM_207585.2	-	-

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<i>IFNGR1</i>	NM_000416.2	-	CNV analysis in exon 7 is not performed
<i>IFNGR2</i>	NM_005534.3	-	-
<i>IGHM</i>	ENST00000390559.2	Whole gene deletion and duplication performed	CNV analysis in individual exons 1–4 is not performed
<i>IGLL1</i>	NM_020070.4	-	-
<i>IKBKB</i>	NM_001556.3	-	-
<i>IKBKG</i>	NM_001099857.3	chrX:g.153775960G>C (c.-16G>C); chrX:g.153788599A>T (c.519-23A>T); chrX:g.153787731C>T (c.518+866C>T); 11.7 kb deletion spanning <i>IKBKG</i> exons 4–10	CNV analysis in exons 3–10 is not performed with the exception of the common 11.7 kb deletion spanning exons 4–10
<i>IKZF1</i>	NM_001291845.2; NM_006060.6	-	CNV analysis in exons 4–8 is not performed
<i>IKZF3</i>	NM_012481.5	-	-
<i>IL10</i>	NM_000572.3	-	-
<i>IL10RA</i>	NM_001558.3	-	-
<i>IL10RB</i>	NM_000628.5	-	-
<i>IL12B</i>	NM_002187.3	-	-
<i>IL12RB1</i>	NM_005535.3	-	CNV analysis in exons 6 and 17 is not performed
<i>IL12RB2</i>	NM_001559.2	-	CNV analysis in exons 2 and 15 is not performed
<i>IL17F</i>	NM_052872.4	-	-
<i>IL17RA</i>	NM_014339.7	-	-
<i>IL17RC</i>	NM_153461.4	-	-
<i>IL18BP</i>	NM_173042.2	-	-
<i>IL1RN</i>	NM_173841.2; NM_173842.3	-	-
<i>IL21</i>	NM_021803.4	-	-
<i>IL21R</i>	NM_021798.4	-	-
<i>IL23R</i>	NM_144701.3	-	-
<i>IL2RA</i>	NM_000417.3	-	-
<i>IL2RB</i>	NM_000878.5	-	-
<i>IL2RG</i>	NM_000206.2	chrX:g.70331494G>A (c.-105C>T); chrX:g.70330553T>C (c.270-15A>G); chrX:g.70329246_70329257del (c.595-15_595-4del)	-
<i>IL36RN</i>	NM_012275.3	-	-
<i>IL37</i>	NM_014439.3	-	-
<i>IL6R</i>	NM_000565.4	-	-
<i>IL6ST</i>	NM_002184.4	-	-
<i>IL7R</i>	NM_002185.5	chr5:g.35867853G>A (c.379+288G>A)	-
<i>IRAK4</i>	NM_016123.3	chr12:g.44178047A>G (c.1188+520A>G)	-
<i>IRF2BP2</i>	NM_182972.2	-	-

# Targeted Genes and Methodology Details for Inborn Errors of Immunity/Bone Marrow Failure/ Telomeropathy/Pulmonary Fibrosis/Very Early Onset IBD/Pancreatitis Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>IRF3</i>	NM_001571.6	-	-
<i>IRF4</i>	NM_002460.4	-	-
<i>IRF7</i>	NM_004031.4	-	-
<i>IRF8</i>	NM_002163.4	-	-
<i>IRF9</i>	NM_006084.5	-	-
<i>ISG15</i>	NM_005101.4	-	-
<i>ITCH</i>	NM_031483.7	-	-
<i>ITGA2B</i>	NM_000419.5	chr17:g.42463181G>T (c.408+11C>A); chr17:g.42458507T>C (c.1211-78A>G); chr17:g.42457859_42457871del (c.1440-13_1440-1del); chr17:g.42455177A>T (c.2095-19T>A); chr17:g.42449567A>G (c.*165T>C)	-
<i>ITGB2</i>	NM_000211.5	chr21:g.46321660A>C (c.500-12T>G); chr21:g.46320404G>T (c.742-14C>A)	-
<i>ITGB3</i>	NM_000212.2	-	-
<i>ITK</i>	NM_005546.3	-	-
<i>ITPKB</i>	NM_002221.3	-	-
<i>JAGN1</i>	NM_032492.4	-	-
<i>JAK1</i>	NM_002227.4	-	-
<i>JAK3</i>	NM_000215.3	chr19:g.17946035C>T (c.1915-11G>A); chr19:g.17943239C>T (c.2680+89G>A)	-
<i>KDM1A</i>	NM_001009999.3	-	-
<i>KDM6A</i>	NM_021140.3	-	-
<i>KIF23</i>	NM_138555.4	-	-
<i>KLF1</i>	NM_006563.5	chr19:g.12998108G>A (c.-154C>T)	-
<i>KMT2A</i>	NM_001197104.1	-	-
<i>KMT2D</i>	NM_003482.3	-	-
<i>KNG1</i>	NM_001102416.3	-	-
<i>KRAS</i>	NM_004985.5	-	-
<i>LACC1</i>	NM_001128303.2	-	-
<i>LAMTOR2</i>	NM_014017.4	chr1:g.156028185C>A (c.*23C>A)	-
<i>LAT</i>	NM_001014987.2	-	-
<i>LCK</i>	NM_001042771.2	-	-
<i>LCP2</i>	NM_005565.5	-	CNV analysis in exon 8 is not performed
<i>LCT</i>	NM_002299.4	-	-
<i>LIG1</i>	NM_000234.3	-	-
<i>LIG4</i>	NM_002312.3	-	-
<i>LPIN2</i>	NM_014646.2	-	-
<i>LRBA</i>	NM_006726.4	-	CNV analysis in exons 20 and 39 is not performed
<i>LSM11</i>	NM_173491.4	-	-

# Targeted Genes and Methodology Details for Inborn Errors of Immunity/Bone Marrow Failure/ Telomeropathy/Pulmonary Fibrosis/Very Early Onset IBD/Pancreatitis Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
LYN	NM_002350.4	-	-
LYST	NM_000081.4	-	-
MAD2L2	NM_001127325.1	-	-
MAGT1	NM_032121.5	-	-
MALT1	NM_006785.4	-	CNV analysis in exon 8 is not performed
MAN2B2	NM_015274.3	-	-
MAP3K14	NM_003954.5	-	-
MASP1	NM_139125.3	-	-
MASP2	NM_006610.4	-	-
MBL2	NM_000242.2	-	-
MCM10	NM_182751.3	-	-
MCM4	NM_005914.4	-	-
MDM4	NM_002393.5	-	-
MECOM	NM_001105078.4	-	-
MEFV	NM_000243.2	-	-
MLH1	NM_000249.3	-	-
MOGS	NM_006302.3	-	-
MPIG6B	NM_025260.3	-	-
MPL	NM_005373.3	-	-
MRTFA	NM_020831.6	-	CNV analysis in exon 3 is not performed
MS4A1	NM_152866.2	-	-
MSH2	NM_000251.3	-	-
MSH6	NM_000179.2	-	-
MSN	NM_002444.3	-	-
MTHFD1	NM_005956.4	-	-
MVK	NM_000431.4	-	-
MYD88	NM_002468.5	-	-
MYH9	NM_002473.5	-	-
MYO5B	NM_001080467.3	-	-
MYOF	NM_013451.4	-	-
MYSM1	NM_001085487.3	-	-
NAF1	NM_138386.3	-	-
NBAS	NM_015909.4	-	-
NBEAL2	NM_015175.3	-	CNV analysis in exon 1 is not performed
NBN	NM_002485.4	-	CNV analysis in exon 16 is not performed
NCF1	NM_000265.6	chr7:g.74191615_74191616del (c.75_76del)	CNV analysis is not performed
NCF2	NM_000433.3	-	-
NCF4	NM_000631.5; NM_013416.3	-	-
NCSTN	NM_015331.3	-	-

# Targeted Genes and Methodology Details for Inborn Errors of Immunity/Bone Marrow Failure/ Telomeropathy/Pulmonary Fibrosis/Very Early Onset IBD/Pancreatitis Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>NEUROG3</i>	NM_020999.4	-	-
<i>NF1</i>	NM_000267.3	-	-
<i>NFAT5</i>	NM_138714.4	-	-
<i>NFE2L2</i>	NM_006164.5	-	-
<i>NFKB1</i>	NM_003998.4	-	-
<i>NFKB2</i>	NM_001077494.3	-	-
<i>NFKBIA</i>	NM_020529.2	-	-
<i>NHEJ1</i>	NM_024782.2	-	-
<i>NHP2</i>	NM_017838.3	-	-
<i>NLRC4</i>	NM_021209.4	-	CNV analysis in exon 2 is not performed
<i>NLRP1</i>	NM_033004.4	-	-
<i>NLRP12</i>	NM_144687.3	-	-
<i>NLRP3</i>	NM_004895.4	-	-
<i>NOD2</i>	NM_022162.3	Variants reported to be associated with risk for Yao syndrome, including the non-coding variant c.2798+158C>T	-
<i>NOPI0</i>	NM_018648.3	-	-
<i>NPAT</i>	NM_002519.3	-	CNV analysis in exon 3 is not performed
<i>NRAS</i>	NM_002524.5	-	-
<i>NSMCE3</i>	NM_138704.4	-	-
<i>OAS1</i>	NM_016816.4	-	-
<i>ORAI1</i>	NM_032790.3	-	-
<i>OSTM1</i>	NM_014028.4	-	-
<i>OTULIN</i>	NM_138348.6	-	-
<i>PALB2</i>	NM_024675.4	chr16:g.23649285A>T (c.109-12T>A); chr16:g.23625426_23625431del (c.3114-16_3114-11del)	-
<i>PARN</i>	NM_002582.4	-	CNV analysis in exons 6 and 19 is not performed
<i>PAX1</i>	NM_006192.5	-	-
<i>PAX5</i>	NM_016734.3	-	-
<i>PCSK1</i>	NM_000439.5	-	-
<i>PEPD</i>	NM_000285.4	-	-
<i>PGM3</i>	NM_001199917.2	-	-
<i>PIK3CD</i>	NM_005026.5	-	-
<i>PIK3R1</i>	NM_181523.3	-	-
<i>PLCG2</i>	NM_002661.5	-	-
<i>PLG</i>	NM_000301.3	-	-
<i>PLVAP</i>	NM_031310.3	-	-
<i>PMM2</i>	NM_000303.3	chr16:g.8926102C>T (c.640-15479C>T)	-
<i>PNLIP</i>	NM_000936.4	-	-
<i>PNP</i>	NM_000270.3	chr14:g.20942914G>A (c.286-18G>A)	-

# Targeted Genes and Methodology Details for Inborn Errors of Immunity/Bone Marrow Failure/ Telomeropathy/Pulmonary Fibrosis/Very Early Onset IBD/Pancreatitis Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>POLA1</i>	NM_016937.4	chrX:g.24744696A>G (c.1375-354A>G)	CNV analysis in exons 12, 17, 27, and 28 is not performed
<i>POLD1</i>	NM_002691.4	-	-
<i>POLE</i>	NM_006231.4	-	-
<i>POLE2</i>	NM_002692.4	-	CNV analysis in exons 8 and 19 is not performed
<i>POLR3A</i>	NM_007055.4	chr10:g.79769277C>T (c.1909+18G>A); chr10:g.79769273C>T (c.1909+22G>A); chr10:g.79743781A>G (c.3337-11T>C)	-
<i>POLR3C</i>	NM_006468.8	-	-
<i>POMP</i>	NM_015932.6	chr13:g.29233225TC>T (c.-95del)	-
<i>POT1</i>	NM_015450.3	-	CNV analysis in exon 5 is not performed
<i>PRF1</i>	NM_001083116.3	-	-
<i>PRKACG</i>	NM_002732.3	-	-
<i>PRKCD</i>	NM_006254.4	-	-
<i>PRKDC</i>	NM_006904.7	-	-
<i>PRSS1</i>	NM_002769.5	-	-
<i>PSEENEN</i>	NM_172341.4	-	-
<i>PSMA3</i>	NM_002788.4	-	-
<i>PSMB10</i>	NM_002801.4	-	-
<i>PSMB4</i>	NM_002796.3	-	-
<i>PSMB8</i>	NM_148919.4	-	-
<i>PSMB9</i>	NM_002800.5	-	-
<i>PSMG2</i>	NM_020232.5	-	-
<i>PSTPIP1</i>	NM_003978.5	-	-
<i>PTPN11</i>	NM_002834.4	-	-
<i>PTPRC</i>	NM_002838.5	-	-
<i>PTPRJ</i>	NM_002843.4	-	-
<i>PUS1</i>	NM_025215.6	-	-
<i>RAB27A</i>	NM_004580.5	-	-
<i>RAC2</i>	NM_002872.5	-	-
<i>RAD50</i>	NM_005732.4	-	-
<i>RAD51</i>	NM_002875.5	-	-
<i>RAD51C</i>	NM_058216.3	-	-
<i>RAG1</i>	NM_000448.2	-	-
<i>RAG2</i>	NM_000536.4	-	-
<i>RASGRP1</i>	NM_005739.4	-	-
<i>RBCK1</i>	NM_031229.4	-	-

# Targeted Genes and Methodology Details for Inborn Errors of Immunity/Bone Marrow Failure/ Telomeropathy/Pulmonary Fibrosis/Very Early Onset IBD/Pancreatitis Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>RBM8A</i>	NM_005105.4	chr1:g.145507648G>T (c.-19G>T); chr1:g.145507646G>A (c.-21G>A); chr1:g.145507765G>C (c.67+32G>C); chr1:g.145508462C>A (c.206-13C>A); chr1:g.145509217C>G (c.*6C>G)	-
<i>RECQL4</i>	NM_004260.3	chr8:g.145739527_145739550del (c.1878+32_1879-27del)	-
<i>REL</i>	NM_002908.4	-	CNV analysis in exon 9 is not performed
<i>RELA</i>	NM_021975.4	-	-
<i>RELB</i>	NM_006509.4	-	-
<i>RFWD3</i>	NM_018124.4	-	-
<i>RFX5</i>	NM_000449.3	-	-
<i>RFXANK</i>	NM_003721.4	chr19:g.19308305_19308330del (c.338-25_338del); chr19:g.19308411_19308468del (c.419_438+38del)	-
<i>RFXAP</i>	NM_000538.3	-	-
<i>RHOH</i>	NM_004310.5	-	-
<i>RIPK1</i>	NM_003804.6	chr6: g.3083547_3083567del (c.688_688+20del)	-
<i>RMRP (NME1)</i>	NR_003051.3	5' UTR variants from n.-25 through the transcription initiation site	CNV analysis is not performed
<i>RNASEH2A</i>	NM_006397.2	-	-
<i>RNASEH2B</i>	NM_024570.4	chr13:g.51501530G>A (c.65-13G>A)	CNV analysis in exon 8 is not performed
<i>RNASEH2C</i>	NM_032193.4	-	-
<i>RNF168</i>	NM_152617.4	-	-
<i>RNF31</i>	NM_017999.5	-	-
<i>RNU4ATAC</i>	NR_023343.1	-	CNV analysis is not performed
<i>RNU7-1</i>	NR_023317.1	-	CNV analysis is not performed
<i>RORC</i>	NM_005060.4	-	-
<i>RPA1</i>	NM_002945.5	-	-
<i>RPL11</i>	NM_000975.5	-	-
<i>RPL15</i>	NM_002948.4	-	CNV analysis in exon 4 is not performed
<i>RPL18</i>	NM_000979.4	-	-
<i>RPL26</i>	NM_000987.5	-	-
<i>RPL27</i>	NM_000988.5	-	-
<i>RPL35</i>	NM_007209.4	-	-
<i>RPL35A</i>	NM_000996.4	-	-
<i>RPL5</i>	NM_000969.5	-	-
<i>RPS10</i>	NM_001014.5	-	-
<i>RPS15A</i>	NM_001019.5	-	-
<i>RPS17</i>	NM_001021.6	Whole gene deletion and duplication performed	CNV analysis in individual exons 1–5 is not performed
<i>RPS19</i>	NM_001022.4	-	-
<i>RPS24</i>	NM_033022.4	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>RPS26</i>	NM_001029.5	-	-
<i>RPS27</i>	NM_001030.6	-	-
<i>RPS28</i>	NM_001031.5	-	-
<i>RPS29</i>	NM_001032.4	-	-
<i>RPS7</i>	NM_001011.4	-	-
<i>RPSA</i>	NM_002295.6	-	-
<i>RTEL1</i>	NM_001283009.2; NM_032957.5	chr20:g.62316953_62317030del (c.1266+3_1266+80del)	-
<i>RUNX1</i>	NM_001754.4	-	-
<i>SAMD9</i>	NM_017654.4	-	-
<i>SAMD9L</i>	NM_152703.5	-	-
<i>SAMHD1</i>	NM_015474.3	-	-
<i>SAR1B</i>	NM_001033503.3	-	-
<i>SASH3</i>	NM_018990.4	-	-
<i>SBDS</i>	NM_016038.4	-	-
<i>SEC23B</i>	NM_006363.6	-	-
<i>SEC61A1</i>	NM_013336.4	-	-
<i>SEMA3E</i>	NM_012431.3	-	-
<i>SERPING1</i>	NM_000062.3	chr11:g.57365567G>T (c.-22-155G>T); chr11:g.57365720A>C (c.-22-2A>C); chr11:g.57365720A>G (c.-22-2A>G); chr11:g.57365721G>A (c.-22-1G>A); chr11:g.57373471A>G (c.686-12A>G)	-
<i>SFTPA1</i>	NM_005411.5	-	CNV analysis in exons 3, 5, and 6 is not performed
<i>SFTPA2</i>	NM_001098668.4	-	CNV analysis in exons 3–6 is not performed
<i>SFTPB</i>	NM_198843.3	-	-
<i>SFTPC</i>	NM_003018.4	-	-
<i>SH2D1A</i>	NM_002351.4	-	-
<i>SH3BP2</i>	NM_003023.4	-	-
<i>SH3KBP1</i>	NM_031892.3	-	-
<i>SI</i>	NM_001041.4	-	CNV analysis in exons 7, 23, 39, and 46 is not performed
<i>SKIV2L</i> ( <i>SKIC2</i> )	NM_006929.5	-	-
<i>SLC10A2</i>	NM_000452.3	-	-
<i>SLC19A2</i>	NM_006996.3	-	-
<i>SLC25A38</i>	NM_017875.4	-	-
<i>SLC26A3</i>	NM_000111.3	-	-
<i>SLC29A3</i>	NM_018344.6	-	CNV analysis in exon 1 is not performed
<i>SLC2A2</i>	NM_000340.2	-	-
<i>SLC35C1</i>	NM_018389.5	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>SLC37A4</i>	NM_001164277.1	-	-
<i>SLC39A4</i>	NM_130849.4	chr8:g.145641963C>T (c.192+19G>A)	-
<i>SLC39A7</i>	NM_006979.3	-	-
<i>SLC46A1</i>	NM_080669.6	-	-
<i>SLC51B</i>	NM_178859.4	-	-
<i>SLC5A1</i>	NM_000343.4	-	-
<i>SLC7A7</i>	NM_001126106.2	-	-
<i>SLC9A3</i>	NM_004174.4	-	CNV analysis in exon 16 is not performed
<i>SLFN14</i>	NM_001129820.2	-	-
<i>SLX4</i>	NM_032444.4	-	-
<i>SMARCAL1</i>	NM_014140.4	-	-
<i>SMARCD2</i>	NM_001098426.2	-	-
<i>SNX10</i>	NM_001199835.1	-	-
<i>SP110</i>	NM_004509.4	-	-
<i>SPINK1</i>	NM_003122.4	chr5:g.147211193G>A (c.-53C>T)	-
<i>SPINK5</i>	NM_006846.3	chr5:g.147465956T>A (c.283-12T>A); chr5:g.147484503G>A (c.1431-12G>A); chr5:g.147491511G>A (c.1820+53G>A)	-
<i>SPINT2</i>	NM_021102.4	-	-
<i>SPPL2A</i>	NM_032802.4	-	CNV analysis in exon 10 is not performed
<i>SRC</i>	NM_005417.4	-	-
<i>SRP54</i>	NM_003136.4	-	-
<i>SRP72</i>	NM_006947.4	-	CNV analysis in exon 17 is not performed
<i>STAT1</i>	NM_007315.3	-	-
<i>STAT2</i>	NM_005419.4	-	-
<i>STAT3</i>	NM_139276.2	chr17:g.40478306G>A (c.1282-89C>T)	-
<i>STAT5B</i>	NM_012448.4	-	CNV analysis in exons 6–8 is not performed
<i>STIM1</i>	NM_003156.3	-	-
<i>STING1</i>	NM_198282.4	-	-
<i>STK4</i>	NM_006282.5	-	-
<i>STN1</i>	NM_024928.5	-	-
<i>STX11</i>	NM_003764.4	-	-
<i>STX3</i>	NM_004177.5	-	-
<i>STXBP2</i>	NM_006949.4	chr19:g.7705756_7705763del (c.326-30_326-23del); chr19:g.7705763_7705770del (c.326-23_326-16del)	-
<i>SYK</i>	NM_003177.7	-	-
<i>TAP1</i>	NM_000593.5	-	-
<i>TAP2</i>	NM_000544.3	-	-
<i>TAPBP</i>	NM_003190.4	-	-
<i>TAZ</i> ( <i>TAFAZZIN</i> )	NM_000116.5	chrX:g.153641699G>A (c.284+110G>A)	-

# Targeted Genes and Methodology Details for Inborn Errors of Immunity/Bone Marrow Failure/ Telomeropathy/Pulmonary Fibrosis/Very Early Onset IBD/Pancreatitis Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>TBK1</i>	NM_013254.4	-	Sequence and CNV analysis in exon 16 may not be reported
<i>TBX1</i>	NM_080647.1	-	-
<i>TCF3</i>	NM_001136139.4; NM_003200.5	-	-
<i>TCIRG1</i>	NM_006019.4	chr11:g.67816893 to chr11:g.67816910 (c.1887+132 to c.1887+149)	-
<i>TCN2</i>	NM_000355.4	chr22:g.31011112A>T (c.581-176A>T)	-
<i>TERC</i>	NR_001566.1	chr3:g.169482906G>A (n.-58C>G); chr3:169482870G>A (n.-22C>T)	CNV analysis is not performed
<i>TERF1</i>	NM_017489.3	-	-
<i>TERF2IP</i>	NM_018975.4	-	-
<i>TERT</i>	NM_198253.3	-	-
<i>TET2</i>	NM_001127208.2	-	-
<i>TFRC</i>	NM_003234.4	-	-
<i>TGFB1</i>	NM_000660.7	-	-
<i>TGFBR1</i>	NM_004612.4	-	-
<i>TGFBR2</i>	NM_003242.6	-	-
<i>THBD</i>	NM_000361.3	-	-
<i>THPO</i>	NM_000460.4	chr3:g.184094078C>A (c.-31G>T); chr3:g.184094097del (c.-47del)	-
<i>TICAM1</i>	NM_182919.3	-	-
<i>TINF2</i>	NM_001099274.3	-	-
<i>TIRAP</i>	NM_148910.2	-	-
<i>TLR3</i>	NM_003265.2	-	-
<i>TLR7</i>	NM_016562.4	-	-
<i>TLR8</i>	NM_138636.5	chrX:g.12924697C>G (c.-129C>G)	-
<i>TMC6</i>	NM_007267.7	-	-
<i>TMC8</i>	NM_152468.5	-	-
<i>TMPRSS15</i>	NM_002772.3	-	CNV analysis in exon 8 is not performed
<i>TNFAIP3</i>	NM_006290.4	-	-
<i>TNFRSF11A</i>	NM_003839.4	-	-
<i>TNFRSF13B</i>	NM_012452.3	-	-
<i>TNFRSF13C</i>	NM_052945.3	-	-
<i>TNFRSF1A</i>	NM_001065.4	chr12:g.6443045C>T (c.194-14G>A)	-
<i>TNFRSF4</i>	NM_003327.4	-	-
<i>TNFRSF9</i>	NM_001561.6	-	-
<i>TNFSF11</i>	NM_003701.4	-	-
<i>TNFSF12</i>	NM_003809.3	-	-
<i>TNFSF13</i>	NM_003808.3	-	-
<i>TOLLIP</i>	NM_019009.4	-	-

# Targeted Genes and Methodology Details for Inborn Errors of Immunity/Bone Marrow Failure/ Telomeropathy/Pulmonary Fibrosis/Very Early Onset IBD/Pancreatitis Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>TOP2B</i>	NM_001068.3	-	-
<i>TP53</i>	NM_000546.5	chr17:g.7579601G>C (c.97-11C>G)	-
<i>TPP2</i>	NM_003291.4	-	CNV analysis in exon 28 is not performed
<i>TRAC</i>	ENST00000611116.1	-	CNV analysis is not performed
<i>TRAF3</i>	NM_003300.4	-	-
<i>TRAF3IP2</i>	NM_147686.4	-	-
<i>TREX1</i>	NM_033629.6	-	-
<i>TRIM22</i>	NM_006074.5	-	-
<i>TRNT1</i>	NM_182916.3	-	-
<i>TRPV6</i>	NM_018646.6	-	-
<i>TSR2</i>	NM_058163.3	-	-
<i>TTC37</i> ( <i>SKIC3</i> )	NM_014639.3	-	-
<i>TTC7A</i>	NM_020458.4	-	-
<i>TUBB1</i>	NM_030773.4	-	-
<i>TYK2</i>	NM_003331.5	-	-
<i>UBE2T</i>	NM_014176.4	-	-
<i>UBR1</i>	NM_174916.3	-	-
<i>UNC13D</i>	NM_199242.2	chr17:g.73839908 to chr17:g.73839907 (c.118-308 to c.118-307); chr17:g.73827442C>T (c.2448-13G>A); chr17:g.73826245C>T (c.2831-13G>A); common 253kb inversion	
<i>UNC45A</i>	NM_018671.5	-	-
<i>UNC93B1</i>	NM_030930.4	-	-
<i>UNG</i>	NM_080911.3	-	-
<i>USB1</i>	NM_024598.4	-	-
<i>USP18</i>	NM_017414.4	-	CNV analysis in exons 7–11 is not performed
<i>VHL</i>	NM_000551.3	chr3:g.10183478_10183488dup (c.-54_-44dup); chr3:g.10184445 to chr3:g.10184687 (c.340+574 to 340+816); chr3:g.10191460T>A (c.464-11T>A); chr3:g.10191656C>G (c.*7C>G)	-
<i>VPS13B</i>	NM_017890.4	chr8:g.100479619T>G (c.3446-23T>G)	-
<i>VPS45</i>	NM_007259.5	-	-
<i>VWF</i>	NM_000552.4	-	Sequence and CNV analysis is only reported for exon 28 (can detect majority of Von Willibrand disease-associated variants)
<i>WAS</i>	NM_000377.3	chrX:g.48547690_48547698delinsATCTGCAGACC (c.1339-19_1339-11delinsATCTGCAGACC)	-
<i>WDR1</i>	NM_017491.5	-	-
<i>WIPF1</i>	NM_001077269.1	-	-

## Targeted Genes and Methodology Details for Inborn Errors of Immunity/Bone Marrow Failure/ Telomeropathy/Pulmonary Fibrosis/Very Early Onset IBD/Pancreatitis Custom Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>WNT2B</i>	NM_024494.3	-	-
<i>WRAP53</i>	NM_018081.2	-	-
<i>XIAP</i>	NM_001167.3	-	-
<i>XRCC2</i>	NM_005431.2	-	-
<i>YARS2</i>	NM_001040436.3	-	-
<i>ZAP70</i>	NM_001079.3	chr2:g.98349927G>A (c.838-80G>A); chr2:g.98354447G>A (c.1624-11G>A)	-
<i>ZBTB24</i>	NM_014797.2	-	-
<i>ZCCHC8</i>	NM_017612.5	-	-
<i>ZNF341</i>	NM_032819.4	-	-
<i>ZNFX1</i>	NM_021035.3	-	-

## Available Inborn Errors of Immunity/Bone Marrow Failure/Telomeropathy/Pulmonary Fibrosis/ Very Early Onset IBD/Pancreatitis Custom Gene Panels

Test ID	Test Name	Genes
SCCNP	Severe Congenital and Cyclic Neutropenia Gene Panel	<i>AK2, AP3B1, AP3D1, CD40LG, CEBPE, CLPB, CSF3R, CXCR2, CXCR4, DNAJC21, EFL1, ELANE, G6PC3, GATA2, GFI1, GINS1, HAX1, JAGN1, LYST, RAC2, SBDS, SLC37A4, SMARCD2, SRP54, TAZ (TAFAZZIN), USB1, VPS13B, VPS45, WAS, WIPF1</i>
TELDP	Telomere Biology Disorders Gene Panel	<i>ACD, CTC1, DKC1, LIG4, NAF1, NHP2, NOP10, PARN, POT1, RPA1, RTEL1, STN1, TERC, TERT, TINF2, USB1, WRAP53, ZCCHC8</i>
IMMAU	Inborn Errors of Immunity with Immune Dysregulation and Autoimmunity Gene Panel	<i>AIRE, BACH2, CARD11, CASP10, CASP8, CD3G, CTLA4, DEF6, FADD, FASLG, FERMT1, FOXP3, IL10, IL10RA, IL10RB, IL2RA, IL2RB, ITCH, JAK1, LRBA, ORAI1, PEPD, PRKCD, RIPK1, STAT3, STAT5B, STIM1, TET2, TGFB1, TPP2</i>
VIRID	Viral Susceptibility, Defects in Intrinsic and Innate Immunity, Gene Panel	<i>CARMIL2, CD27, CD70, CIB1, CTPS1, CXCR4, DBR1, IFIH1, IFNAR1, IFNAR2, IRF3, IRF7, IRF9, MAGT1, POLR3A, POLR3C, PRKCD, RASGRP1, SH2D1A, STAT1, STAT2, TLR3, TLR7, TLR8, TMC6, TMC8, TNFRSF9, TRAF3, UNC93B1, XIAP</i>
HPANP	Hereditary Pancreatitis Gene Panel	<i>CFTR, CTRC, PRSS1, SPINK1</i>
HIESG	Hyper IgE Syndrome Gene Panel	<i>AIRE, CARD11, CARD9, CARMIL2, DOCK8, ERBIN, IL6R, IL6ST, IL17RA, IL17RC, IL17F, PGM3, SPINK5, STAT1, STAT3, TGFB1, TGFB2, TRAF3IP2, TYK2, WAS, ZNF341</i>
ALPSG	Autoimmune Lymphoproliferative Syndrome (ALPS) Gene Panel	<i>ADA2, CARD11, CASP10, CASP8, CTLA4, DEF6, FADD, FAS, FASLG, IL2RA, IL2RB, ITK, LRBA, MAGT1, PIK3CD, PIK3R1, PRKCD, RASGRP1, SH2D1A, STAT3, STK4, TET2, TNFAIP3, TNFRSF9, TPP2, XIAP</i>

# Targeted Genes and Methodology Details for Inborn Errors of Immunity/Bone Marrow Failure/ Telomeropathy/Pulmonary Fibrosis/Very Early Onset IBD/Pancreatitis Custom Gene Panel (continued)

## Available Inborn Errors of Immunity/Bone Marrow Failure/Telomeropathy/Pulmonary Fibrosis/ Very Early Onset IBD/Pancreatitis Custom Gene Panels (continued)

Test ID	Test Name	Genes
BTKSG	Bruton Tyrosine Kinase (BTK), Full Gene Sequencing	<i>BTK</i>
GATAS	GATA-Binding Protein 2 (GATA2), Full Gene Sequencing	<i>GATA2</i>
CFTRN	Cystic Fibrosis Transmembrane Conductance Regulator (CFTR), Full Gene Analysis	<i>CFTR</i>
SCIDP	Severe Combined Immunodeficiency (SCID) Gene Panel	<i>ADA, AK2, ATM, BCL11B, CARD11, CD247, CD3D, CD3E, CD3G, CD8A, CHD7, CIITA, CORO1A, DCLRE1C, DOCK2, DOCK8, EXTL3, FOXN1, IKZF1, IL2RA, IL2RG, IL7R, JAK3, LAT, LCP2, LIG4, MTHFD1, NBN, NHEJ1, ORAI1, PAX1, PNP, POLE2, PRKDC, PTPRC, RAC2, RAG1, RAG2, RFX5, RFXANK, RFXAP, RMRP (NME1), SEMA3E, SMARCAL1, STIM1, TBX1, TTC7A, WAS, WIPF1, ZAP70</i>
EBLPD	Epstein Barr Virus (EBV) Susceptibility and Lymphoproliferative Disorders Gene Panel	<i>ATM, CARD11, CARMIL2, CD27, CD70, CORO1A, CTLA4, CTPS1, DEF6, GATA2, ITK, LRBA, MAGT1, PIK3CD, PRF1, PRKCD, RASGRP1, SH2D1A, STK4, STX11, STXBP2, TET2, TNFRSF9, UNC13D, XIAP</i>
HLHGP	Primary Hemophagocytic Lymphohistiocytosis (HLH) Gene Panel	<i>ADA, AP3B1, AP3D1, BLOC1S6, CD27, CD70, CDC42, CORO1A, CTPS1, IFNAR2, ITK, LYST, MAGT1, MVK, NLRC4, PRF1, RAB27A, SH2D1A, SLC7A7, STX11, STXBP2, UNC13D, XIAP</i>
BCELL	B-Cell/Antibody Deficiency Gene Panel	<i>ADA, ADA2, AICDA, ATP6AP1, BLNK, BTK, CARD11, CD19, CD27, CD40, CD40LG, CD70, CD79A, CD79B, CD81, CDCA7, CTLA4, CR2, CXCR4, DCLRE1C, DNMT3B, GATA2, ICOS, IGHM, IGLL1, IKBKG, IKZF1, IKZF3, IL21, IL21R, IRF2BP2, KDM6A, KMT2A, KMT2D, LIG1, LRBA, MOGS, MS4A1, NFKB1, NFKB2, PIK3CD, PIK3R1, PLCG2, PRKCD, RAC2, RAG1, RAG2, RNF168, SEC61A1, SH2D1A, SH3KBP1, SLC39A7, TCF3, TNFRSF13B, TNFRSF13C, TNFSF12, TNFSF13, TOP2B, TRNT1, UNG, XIAP</i>
AUTOG	Autoinflammatory Disorders Gene Panel	<i>ACP5, ADA, ADA2, ADAM17, ADAR, AIRE, ALPI, AP3B1, AP3D1, ARPC1B, ASAH1, C1QA, C1QB, C1QC, C1R, C1S, C2, CARD11, CARD14, CASP10, CASP8, CD3G, CD40LG, CD48, CD55, CDC42, COPA, CTLA4, DDX58 (RIGI), DNASE1, DNASE1L3, DNASE2, DOCK8, FADD, FOXP3, GATA2, HAVCR2, ICOS, IFIH1, IKBKG, IL10, IL10RA, IL10RB, IL1RN, IL2RA, IL2RB, IL2RG, IL36RN, ISG15, ITCH, ITGB2, ITK, JAK1, LACC1, LIG4, LPIN2, LRBA, LSM11, LYN, LYST, MEFV, MVK, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, OAS1, OTULIN, PIK3CD, PIK3R1, PLCG2, POLA1, POMP, PRF1, PRKCD, PSMA3, PSMB10, PSMB4, PSMB8, PSMB9, PSMG2, PSTPIP1, RAB27A, RBCK1, RIPK1, RNASEH2A, RNASEH2B, RNASEH2C, RNF31, RNU7-1, SAMD9L, SAMHD1, SH2D1A, SH3BP2, SKIV2L (SKIC2), SLC29A3, SLC37A4, STAT1, STAT2, STAT3, STIM1, STING1, STX11, STXBP2, TLR7, TNFAIP3, TNFRSF1A, TPP2, TREX1, TRNT1, UNC13D, USP18, WAS, WDR1, XIAP, ZAP70</i>
EOIBD	Early Onset Monogenic Inflammatory Bowel Disease (IBD) Gene Panel	<i>ADA, ADAM17, AICDA, AIRE, ALPI, ANKZF1, ARPC1B, ASAH1, BACH2, BTK, CARMIL2, CASP8, CD3G, CD40LG, CD55, COL7A1, CTLA4, CYBA, CYBB, CYBC1, DCLRE1C, DEF6, DGAT1, DKC1, DOCK8, DUOX2, EPCAM, FCHO1, FERMT1, FOXP3, G6PC (G6PC1), G6PC3, GUCY2C, HPS1, HPS3, HPS4, HPS6, ICOS, IFIH1, IKBKG, IL10, IL10RA, IL10RB, IL21, IL21R, IL2RA, IL2RB, IL2RG, IL7R, ITCH, ITGB2, JAK1, LCT, LIG4, LRBA, MALT1, MEFV, MVK, MYO5B, NCF1, NCF2, NCF4, NEUROG3, NFKBIA, NLRC4, PAX1, PCSK1, PIK3CD, PIK3R1, PLCG2, PLVAP, POLA1, RAG1, RAG2, RIPK1, RTEL1, SH2D1A, SI, SKIV2L (SKIC2), SLC10A2, SLC26A3, SLC37A4, SLC39A4, SLC51B, SLC5A1, SLC9A3, SPINT2, STAT1, STAT3, STAT5B, STIM1, STX3, STXBP2, TGFB1, TGFB1, TGFB2, TLR3, TNFAIP3, TRIM22, TRNT1, TTC37 (SKIC3), TTC7A, UNC45A, WAS, WIPF1, XIAP, ZAP70, ZBTB24</i>

# Targeted Genes and Methodology Details for Inborn Errors of Immunity/Bone Marrow Failure/ Telomeropathy/Pulmonary Fibrosis/Very Early Onset IBD/Pancreatitis Custom Gene Panel (continued)

## Available Inborn Errors of Immunity/Bone Marrow Failure/Telomeropathy/Pulmonary Fibrosis/ Very Early Onset IBD/Pancreatitis Custom Gene Panels (continued)

Test ID	Test Name	Genes
IEICP	Inborn Errors of Immunity Comprehensive Gene Panel	ACD, ACP5, ADA, ADA2, ADAM17, ADAR, AICDA, AIRE, AK2, ALPI, ANGPT1, ANKZF1, AP3B1, AP3D1, ARPC1B, ASAH1, ATM, ATP6AP1, BACH2, BCL10, BCL11B, BLM, BLNK, BLOC1S6, BTK, C1QA, C1QB, C1QC, C1R, C1S, C2, C3, C5, C6, C7, C8A, C8B, C9, CARD11, CARD14, CARD9, CARMIL2, CASP10, CASP8, CCBE1, CD19, CD247, CD27, CD28, CD3D, CD3E, CD3G, CD40, CD40LG, CD46, CD48, CD55, CD59, CD70, CD79A, CD79B, CD81, CD8A, CDC42, CDCA7, CEBPE, CFB, CFD, CFH, CFI, CFP, CFTR, CHD7, CIB1, CIITA, CLCN7, CLPB, COPA, CORO1A, CR2, CSF2RA, CSF2RB, CSF3R, CTC1, CTLA4, CTPS1, CTSC, CXCR2, CXCR4, CYBA, CYBB, CYBC1, DBR1, DCLRE1C, DDX58 (RIGI), DEF6, DGAT1, DKC1, DNAJC21, DNASE1, DNASE1L3, DNASE2, DNMT3B, DOCK2, DOCK8, DUOX2, EFL1, ELANE, EPG5, ERBIN, ERCC6L2, EXTL3, F12, FADD, FAS, FASLG, FAT4, FCHO1, FERMT1, FERMT3, FOXP1, FOXP3, G6PC (G6PC1), G6PC3, G6PD, GATA2, GFI1, GINS1, GLA, HAVCR2, HAX1, HELLS, HMOX1, HPS1, HPS3, HPS4, HPS6, ICOS, ICOSLG, IFIH1, IFNAR1, IFNAR2, IFNGR1, IFNGR2, IGHM, IGLL1, IKBKB, IKBKG, IKZF1, IKZF3, IL10, IL10RA, IL10RB, IL12B, IL12RB1, IL12RB2, IL17F, IL17RA, IL17RC, IL1RN, IL21, IL21R, IL23R, IL2RA, IL2RB, IL2RG, IL36RN, IL6R, IL6ST, IL7R, IRAK4, IRF2BP2, IRF3, IRF7, IRF8, IRF9, ISG15, ITCH, ITGB2, ITK, ITPKB, JAGN1, JAK1, JAK3, KDM6A, KMT2A, KMT2D, KNG1, LACC1, LAT, LCK, LCP2, LIG1, LIG4, LPIN2, LRBA, LSM11, LYN, LYST, MAGT1, MALT1, MAP3K14, MASP2, MCM10, MEFV, MOGS, MRTFA, MS4A1, MSN, MTHFD1, MVK, MYD88, MYSM1, NBAS, NBN, NCF1, NCF2, NCF4, NCSTN, NFE2L2, NFKB1, NFKB2, NFKBIA, NHEJ1, NHP2, NLRC4, NLRP1, NLRP12, NLRP3, NOD2, NSMCE3, OAS1, ORAI1, OSTM1, OTULIN, PARN, PAX1, PEPD, PGM3, PIK3CD, PIK3R1, PLCG2, PLG, PMM2, PNP, POLA1, POLD1, POLE, POLE2, POLR3A, POLR3C, POMP, PRF1, PRKCD, PRKDC, PSENEN, PSMA3, PSMB10, PSMB4, PSMB8, PSMB9, PSMG2, PSTPIP1, PTPRC, RAB27A, RAC2, RAG1, RAG2, RASGRP1, RBCK1, RBM8A, REL, RELA, RELB, RFX5, RFXANK, RFXAP, RHOH, RIPK1, RMRP (NME1), RNASEH2A, RNASEH2B, RNASEH2C, RNF168, RNF31, RNU4ATAC, RNU7-1, RORC, RPSA, RTEL1, SAMD9, SAMD9L, SAMHD1, SBDS, SEC61A1, SEMA3E, SERPING1, SH2D1A, SH3BP2, SH3KBP1, SKIV2L (SKIC2), SLC29A3, SLC35C1, SLC37A4, SLC39A7, SLC46A1, SLC7A7, SMARCAL1, SMARCD2, SNX10, SP110, SPINK5, SPPL2A, SRP54, STAT1, STAT2, STAT3, STAT5B, STIM1, STING1, STK4, STX11, STXBP2, SYK, TAP1, TAP2, TAPBP, TAZ (TAFAZZIN), TBX1, TCF3, TCIRG1, TCN2, TERC, TERT, TET2, TFRC, TGFB1, TGFBR1, TGFBR2, THBD, TINF2, TIRAP, TLR3, TLR7, TLR8, TMC6, TMC8, TNFAIP3, TNFRSF11A, TNFRSF13B, TNFRSF13C, TNFRSF1A, TNFRSF4, TNFRSF9, TNFSF11, TNFSF12, TNFSF13, TOP2B, TPP2, TRAC, TRAF3, TRAF3IP2, TREX1, TRNT1, TTC37 (SKIC3), TTC7A, TYK2, UNC13D, UNC93B1, UNG, USB1, USP18, VPS13B, VPS45, WAS, WDR1, WIPF1, WRAP53, XIAP, ZAP70, ZBTB24, ZNF341, ZNFX1
COMID	Combined Humoral and Cell-Mediated Immunodeficiency Gene Panel	ADA, AK2, ARPC1B, ATM, BCL10, BCL11B, BLM, CARD11, CD247, CD28, CD3D, CD3E, CD3G, CD40, CD40LG, CD70, CD8A, CDCA7, CHD7, CIITA, CORO1A, CTLA4, DCLRE1C, DNMT3B, DOCK2, DOCK8, EPG5, ERCC6L2, EXTL3, FCHO1, FOXP1, GINS1, HELLS, ICOS, ICOSLG, IKBKB, IKBKG, IKZF1, IL21, IL21R, IL2RA, IL2RB, IL2RG, IL7R, ITK, ITPKB, JAK3, KDM6A, KMT2A, KMT2D, LAT, LCK, LCP2, LIG1, LIG4, LRBA, MAGT1, MALT1, MAP3K14, MCM10, MSN, MTHFD1, MYSM1, NBN, NFKBIA, NHEJ1, NSMCE3, ORAI1, PAX1, PGM3, PNP, POLD1, POLE, POLE2, PRKDC, PSMB9, PTPRC, RAC2, RAG1, RAG2, RASGRP1, RBCK1, REL, RELA, RELB, RFX5, RFXANK, RFXAP, RHOH, RMRP (NME1), RNF168, RNF31, RNU4ATAC, SEC61A1, SEMA3E, SH2D1A, SKIV2L (SKIC2), SMARCAL1, SP110, STAT3, STAT5B, STIM1, STK4, TAP1, TAP2, TAPBP, TAZ (TAFAZZIN), TBX1, TFRC, TNFRSF4, TRAC, TTC37 (SKIC3), TTC7A, WAS, WIPF1, ZAP70, ZBTB24
AECDP	Angioedema and Complement Disorders Gene Panel	ANGPT1, C1QA, C1QB, C1QC, C1R, C1S, C2, C3, C5, C6, C7, C8A, C8B, C9, CD46, CD55, CD59, CFB, CFD, CFH, CFI, CFP, F12, KNG1, MASP2, PLG, SERPING1, THBD

# Targeted Genes and Methodology Details for Inborn Errors of Immunity/Bone Marrow Failure/ Telomeropathy/Pulmonary Fibrosis/Very Early Onset IBD/Pancreatitis Custom Gene Panel (continued)

## Available Inborn Errors of Immunity/Bone Marrow Failure/Telomeropathy/Pulmonary Fibrosis/ Very Early Onset IBD/Pancreatitis Custom Gene Panels (continued)

Test ID	Test Name	Genes
BMFGP	Inherited Bone Marrow Failure Gene Panel	<i>ABCB7, ACD, ADA, ADA2, AK2, ALAS2, ANKRD26, AP3B1, AP3D1, ARPC1B, BLOC1S6, BRCA1, BRCA2, BRIP1, C15orf41 (CDIN1), CD27, CDAN1, CEBPA, CLCN7, CLPB, CSF3R, CTC1, CXCR2, CXCR4, CYCS, DDX41, DHFR, DIAPH1, DKC1, DNAJC21, DNMT3B, EFL1, ELANE, EPO, ERCC4, ERCC6L2, ETV6, FANCA, FANCB, FANCC, FANCD2, FANCE, FANCF, FANCG, FANCI, FANCL, FANCM, FLI1, FYB1, G6PC3, GATA1, GATA2, GF11, GF11B, GINS1, GLA, GLRX5, GP1BA, GP1BB, GP9, HAX1, HOXA11, IKZF1, ITGA2B, ITGB3, JAGN1, KDM1A, KIF23, KLF1, LIG4, LPIN2, LYST, MAD2L2, MDM4, MECOM, MPIG6B, MPL, MTHFD1, MYH9, MYSM1, NAF1, NBEAL2, NBN, NHP2, NOP10, OSTM1, PALB2, PARN, PAX5, PGM3, PIK3CD, POT1, PRKACG, PUS1, RAB27A, RAC2, RAD50, RAD51, RAD51C, RBM8A, RECQL4, RFWD3, RMRP (NME1), RPA1, RPL11, RPL15, RPL18, RPL26, RPL27, RPL35, RPL35A, RPL5, RPS10, RPS15A, RPS17, RPS19, RPS24, RPS26, RPS27, RPS28, RPS29, RPS7, RTEL1, RUNX1, SAMD9, SAMD9L, SBDS, SEC23B, SLC19A2, SLC25A38, SLC37A4, SLC46A1, SLFN14, SLX4, SMARCAL1, SMARCD2, SNX10, SRC, SRP54, SRP72, STAT3, STIM1, STN1, STXBP2, SYK, TAZ (TAFAZZIN), TCIRG1, TCN2, TERC, TERT, THPO, TINF2, TNFRSF11A, TNFRSF13B, TNFSF11, TP53, TRNT1, TSR2, TUBB1, UBE2T, USB1, VPS13B, VPS45, VWF, WAS, WDR1, WIPF1, WRAP53, XIAP, XRCC2, YARS2, ZCCHC8</i>
PHCGD	Phagocytic Disorders and Chronic Granulomatous Disease Gene Panel	<i>CEBPE, CFTR, CSF2RA, CSF2RB, CTSC, CYBA, CYBB, CYBC1, FERMT3, G6PD, ITGB2, MRTFA, NCF1, NCF2, NCF4, RAC2, SLC35C1, SLC7A7, SMARCD2, WDR1</i>
IPFGP	Idiopathic Pulmonary Fibrosis Gene Panel	<i>ABCA3, ACD, AP3B1, CTC1, DKC1, HPS1, HPS4, NAF1, NHP2, NOP10, PARN, POT1, RNF168, RPA1, RTEL1, SFTPA1, SFTPA2, SFTPB, SFTPC, STN1, STING1, TERC, TERT, TINF2, USB1, WRAP53, ZCCHC8</i>
PANGP	Expanded Pancreatitis Gene Panel	<i>CASR, CEL, CFTR, CLDN2, CPA1, CTRC, PRSS1, SPINK1, TRPV6</i>
Custom Gene Ordering Only	NA	<i>ACTB, AP1S3, APOL1, ARHGEF1, ATR, B2M, C8G, CBL, CHEK2, CXXC5, DCLRE1B, DGKE, FCGR3A, FLNA, FOXI3, HS3ST6, HYOU1, IL18BP, IL37, IRF4, LAMTOR2, MAN2B2, MBL2, MCM4, MLH1, MSH2, MSH6, MYOF, NF1, NFAT5, NPAT, NRAS, PTPN11, PTPRJ, SASH3, SLC2A2, TBK1, TERF1, TERF2IP, TICAM1, TOLLIP, VHL</i>