

Targeted Genes and Methodology Details for Early Onset Monogenic Inflammatory Bowel Disease (IBD) Gene Panel

The following applies to EOIBD / Early Onset Monogenic Inflammatory Bowel Disease (IBD) 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 numbers 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 2023 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>ADA</i>	NM_000022.4	chr20:g.43249076C>T (c.976-34G>A); chr20:g.43248503A>T (c.1079-15T>A)	-
<i>ADAM17</i>	NM_003183.6	-	-
<i>AICDA</i>	NM_020661.4	-	-
<i>AIRE</i>	NM_000383.4	-	-
<i>ALPI</i>	NM_001631.5	-	-
<i>ANKZF1</i>	NM_018089.3	-	-
<i>ARPC1B</i>	NM_005720.4	-	-
<i>ASAH1</i>	NM_177924.5	-	CNV analysis in exon 4 is not performed
<i>BACH2</i>	NM_021813.4	-	-
<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+12del11); chrX:g.100612471_100612472insTTTTTCTACC (c.1177+28_1177+29insAGAAAAAGGT); chrX:g.100609705T>G (c.1567-23A>C); chrX:g.100609705T>C (c.1567-23A>G); chrX:g.100609693_100609696del (c.1567-12_1567-9delTTTG)	-
<i>CARMIL2</i>	NM_001013838.3	-	-
<i>CASP8</i>	NM_001228.4	-	-
<i>CD3G</i>	NM_000073.2	-	-
<i>CD40LG</i>	NM_000074.3	-	-
<i>CD55</i>	NM_000574.5	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>COL7A1</i>	NM_000094.3	chr3:g.48629915C>T (c.977-15G>A); chr3:g.48616971T>C (c.5236-23A>G); chr3:g.48613354T>C (c.5821-19A>G); chr3:g.48610175_48610195del (c.6832-23_6832-3del); chr3:g.48605981_48605985delinsTGAA (c.7759-18_7759-14delCATCTinsTTCA); chr3:g.48605248_48605263del (c.7929+11_7929+26del); chr3:g.48604008A>T (c.8305-12T>A); chr3:g.48602928_48602947del (c.8441-15_8445del); chr3:g.48602928_48602948del (c.8441-14_8447del)	-
<i>CTLA4</i>	NM_005214.5	-	-
<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>DCLRE1C</i>	NM_001033855.3	-	-
<i>DEF6</i>	NM_022047.4	-	-
<i>DGAT1</i>	NM_012079.6	-	-
<i>DKC1</i>	NM_001363.5	chrX:g.153991099C>G (c.-142C>G)	-
<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>EPCAM</i>	NM_002354.3	chr2:g.47596641T>C (c.-4T>C); chr2:g.47606078A>G (c.556-14A>G)	-
<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>FOXP3</i>	NM_014009.4	chrX:g.49119820_49121205del (c.-105_-23+1303del, 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)	-
<i>G6PC(G6PC1)</i>	NM_000151.4	-	-
<i>G6PC3</i>	NM_138387.3	-	-
<i>GUCY2C</i>	NM_004963.4	-	-
<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>ICOS</i>	NM_012092.4	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>IFIH1</i>	NM_022168.4	-	-
<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)	CNV analysis in exons 3–10 is not performed
<i>IL10</i>	NM_000572.3	-	-
<i>IL10RA</i>	NM_001558.3	-	-
<i>IL10RB</i>	NM_000628.5	-	-
<i>IL21</i>	NM_021803.4	-	-
<i>IL21R</i>	NM_021798.4	-	-
<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>IL7R</i>	NM_002185.5	chr5:g.35867853G>A (c.379+288G>A)	-
<i>ITCH</i>	NM_031483.7	-	-
<i>ITGB2</i>	NM_000211.5	chr21:g.46321660A>C (c.500-12T>G); chr21:g.46320404G>T (c.742-14C>A)	-
<i>JAK1</i>	NM_002227.4	-	-
<i>LCT</i>	NM_002299.4	-	-
<i>LIG4</i>	NM_002312.3	-	-
<i>LRBA</i>	NM_006726.4	-	CNV analysis in exons 20 and 39 is not performed
<i>MALT1</i>	NM_006785.4	-	CNV analysis in exon 8 is not performed
<i>MEFV</i>	NM_000243.2	-	-
<i>MVK</i>	NM_000431.4	-	-
<i>MYO5B</i>	NM_001080467.3	-	-
<i>NCF2</i>	NM_000433.3	-	-
<i>NCF4</i>	NM_000631.5; NM_013416.3	-	-
<i>NEUROG3</i>	NM_020999.4	-	-
<i>NFKBIA</i>	NM_020529.2	-	-
<i>NLRC4</i>	NM_021209.4	-	CNV analysis in exon 2 is not performed
<i>PAX1</i>	NM_006192.5	-	-
<i>PCSK1</i>	NM_000439.5	-	-
<i>PIK3CD</i>	NM_005026.5	-	-
<i>PIK3R1</i>	NM_181523.3	-	-
<i>PLCG2</i>	NM_002661.5	-	-
<i>PLVAP</i>	NM_031310.3	-	-
<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

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
<i>RAG1</i>	NM_000448.2	-	-
<i>RAG2</i>	NM_000536.4	-	-
<i>RIPK1</i>	NM_003804.6	chr6:g.3083547_3083567del (c.688_688+20del)	-
<i>RTEL1</i>	NM_032957.5	-	-
<i>SH2D1A</i>	NM_002351.4	-	-
<i>SI</i>	NM_001041.4	-	CNV analysis in exons 7, 23, 39, and 46 is not performed
<i>SKIV2L(SKIC2)</i>	NM_006929.5	-	-
<i>SLC10A2</i>	NM_000452.3	-	-
<i>SLC26A3</i>	NM_000111.3	-	-
<i>SLC37A4</i>	NM_001164277.1	-	-
<i>SLC39A4</i>	NM_130849.4	chr8:g.145641963C>T (c.192+19G>A)	-
<i>SLC51B</i>	NM_178859.4	-	-
<i>SLC5A1</i>	NM_000343.4	-	-
<i>SLC9A3</i>	NM_004174.4	-	CNV analysis in exon 16 is not performed
<i>SPINT2</i>	NM_021102.4	-	-
<i>STAT1</i>	NM_007315.3	-	-
<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>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>TGFB1</i>	NM_000660.7	-	-
<i>TGFBR1</i>	NM_004612.4	-	-
<i>TGFBR2</i>	NM_003242.6	-	-
<i>TLR3</i>	NM_003265.2	-	-
<i>TNFAIP3</i>	NM_006290.4	-	-
<i>TRIM22</i>	NM_006074.5	-	-
<i>TRNT1</i>	NM_182916.3	-	-
<i>TTC37(SKIC3)</i>	NM_014639.3	-	-
<i>TTC7A</i>	NM_020458.4	-	-
<i>UNC45A</i>	NM_018671.5	-	-
<i>WAS</i>	NM_000377.3	chrX:g.48547690_48547698delinsATCTGCAGACC (c.1339-19_1339-11delinsATCTGCAGACC)	-
<i>WIPF1</i>	NM_001077269.1	-	-
<i>XIAP</i>	NM_001167.3	-	-
<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	-	-