MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Hyper-IgE Syndrome Gene Panel

The following applies to HIESG / Hyper-IgE Syndrome Gene Panel, Varies. 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 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 March 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, 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
AIRE	NM_000383.4	-	-
CARD11	NM_032415.6	-	-
CARD9	NM_052813.5	-	-
CARMIL2	NM_001013838.3	-	-
DOCK8	NM_203447.3	c.742-18 to c.742-15; c.2083_2109+62del89	-
ERBIN	NM_001253697.2	-	CNV analysis in exon 7 is not performed
IL6R	NM_000565.4	-	-
IL6ST	NM_002184.4	-	-
IL17RA	NM_014339.7	-	-
IL17RC	NM_153461.4	-	-
IL17F	NM_052872.4	-	-
PGM3	NM_001199917.2	-	-
SPINK5	NM_006846.3	c.283-12T>A; c.1431-12G>A; c.1820+53G>A	-
STAT1	NM_007315.3	-	-
STAT3	NM_139276.2	c.1282-89C>T	-
TGFBR1	NM_004612.4	-	-
TGFBR2	NM_003242.6	-	-
TRAF3IP2	NM_147686.4	-	-
TYK2	NM_003331.5	-	-
WAS	NM_000377.3	c.1339-19_1339-11delinsATCTGCAGACC	-
ZNF341	NM_032819.4	-	-