MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Autoinflammatory Disorders Gene Panel

The following applies to AUTOG / Autoinflammatory Disorders 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 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
ACP5	NM_001111035.3	-	-
ADA	NM_000022.4	chr20:g.43249076C>T (c.976-34G>A); chr20:g.43248503A>T (c.1079-15T>A)	-
ADA2	NM_001282225.2	-	-
ADAM17	NM_003183.6	-	-
ADAR	NM_001111.5	-	-
AIRE	NM_000383.4	-	-
ALPI	NM_001631.5	-	-
AP3B1	NM_003664.4	-	-
AP3D1	NM_001261826.3	-	-
ARPC1B	NM_005720.4	-	-
ASAH1	NM_177924.5	-	CNV analysis in exon 4 is not performed
C1QA	NM_015991.4	-	-
C1QB	NM_000491.5	-	-
C1QC	NM_172369.5	-	-
C1R	NM_001733.7	-	CNV analysis in exons 1–7 is not performed
C1S	NM_201442.4	-	-
C2	NM_000063.6	chr6:g.31902068_31902095del (c.841_849+19del); chr6:g.31911326_31911347del (c.1567+22_1567+43del)	-
CARD11	NM_032415.6	-	-
CARD14	NM_024110.4	-	-
CASP10	NM_032977.4	-	-
CASP8	NM_001228.4	-	-
CD3G	NM_000073.2	-	-
CD40LG	NM_000074.3	-	-
CD48	NM_001778.4	-	-
CD55	NM_000574.5	-	-
CDC42	NM_001791.4	-	-

Targeted Genes and Methodology Details for Autoinflammatory Disorders Gene Panel (continued)

Gene	Reference Transcript	Additional Evaluations	Technical Limitations
COPA	NM_004371.4	-	-
CTLA4	NM_005214.5	-	-
DDX58	NM_014314.4	-	-
DNASE1	NM_005223.3	-	-
DNASE1L3	NM_004944.4	-	-
DNASE2	NM_001375.3	-	-
DOCK8	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)	-
FADD	NM_003824.3	-	-
FOXP3	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)	-
GATA2	NM_032638.5	chr3:g.128202114 through chr3:g.128202177 (c.1017+526 through c.1017+589, corresponding to a highly conserved intronic region); chr3:g.128200787_128200806dup (c.1018-17_1020dup)	-
HAVCR2	NM_032782.5	-	-
ICOS	NM_012092.4	-	-
IFIH1	NM_022168.4	-	-
IKBKG	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
IL10	NM_000572.3	-	-
IL10RA	NM_001558.3	-	-
IL10RB	NM_000628.5	-	-
IL1RN	NM_173841.2; NM_173842.3	-	-
IL2RA	NM_000417.3	-	-
IL2RB	NM_000878.5	-	-
IL2RG	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)	-
IL36RN	NM_012275.3	-	-
ISG15	NM_005101.4	-	-
ITCH	NM_031483.7	-	-
ITGB2	NM_000211.5	chr21:g.46321660A>C (c.500-12T>G); chr21:g.46320404G>T (c.742-14C>A)	-
ITK	NM_005546.3	-	-
JAK1	NM_002227.4	-	-
LACC1	NM_001128303.2	-	-
LIG4	NM_002312.3	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
LPIN2	NM_014646.2	-	-
LRBA	NM_006726.4	-	CNV analysis in exons 20 and 39 is not performed
LSM11	NM_173491.4	-	-
LYN	NM_002350.4	-	-
LYST	NM_000081.4	-	-
MEFV	NM_000243.2	-	-
MVK	NM_000431.4	-	-
NLRC4	NM_021209.4	-	CNV analysis in exon 2 is not performed
NLRP1	NM_033004.4	-	-
NLRP12	NM_144687.3	-	-
NLRP3	NM_004895.4	-	-
NOD2	NM_022162.3	Variants reported to be associated with risk for Yao syndrome, including the noncoding variant chr16:g.50756774C>T (c.2798+158C>T)	-
OAS1	NM_016816.4	-	-
OTULIN	NM_138348.6	-	-
PIK3CD	NM_005026.5	-	-
PIK3R1	NM_181523.3	-	-
PLCG2	NM_002661.5	-	-
POLA1	NM_016937.4	chrX:24744696A>G (c.1375-354A>G)	CNV analysis in exons 12, 17, 27, and 28 is not performed
POMP	NM_015932.6	chr13:g.29233227del (c.-95del)	-
PRF1	NM_001083116.3	-	-
PRKCD	NM_006254.4	-	-
PSMA3	NM_002788.4	-	-
PSMB10	NM_002801.4	-	-
PSMB4	NM_002796.3	-	-
PSMB8	NM_148919.4	-	-
PSMB9	NM_002800.5	-	-
PSMG2	NM_020232.5	-	-
PSTPIP1	NM_003978.5	-	-
RAB27A	NM_004580.5	-	-
RBCK1	NM_031229.4	-	-
RIPK1	NM_003804.6	chr6:g.3083547_3083567del (c.688_688+20del)	-
RNASEH2A	NM_006397.2	-	-
RNASEH2B	NM_024570.4	chr13:g.51501530G>A (c.65-13G>A)	CNV analysis in exon 8 is not performed
RNASEH2C	NM_032193.4	-	-

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Gene	Reference Transcript	Additional Evaluations	Technical Limitations
RNF31	NM_017999.5	-	-
RNU7-1	NR_023317.1	-	CNV analysis is not performed
SAMD9L	NM_152703.5	-	-
SAMHD1	NM_015474.3	-	-
SH2D1A	NM_002351.4	-	-
SH3BP2	NM_003023.4	-	-
SKIV2L(SKIC2)	NM_006929.5	-	-
SLC29A3	NM_018344.6	-	Sequence and CNV analysis in exon 1 is not performed
SLC37A4	NM_001164277.1	-	-
STAT1	NM_007315.3	-	-
STAT2	NM_005419.4	-	-
STAT3	NM_139276.2	chr17:g.40478306G>A (c.1282-89C>T)	-
STIM1	NM_003156.3	-	-
STING1	NM_198282.4	-	-
STX11	NM_003764.4	-	-
STXBP2	NM_006949.4	chr19:g.7705756_7705763del (c.326-30_326-23del); chr19:g.7705763_7705770del (c.326-23_326-16del)	-
TLR7	NM_016562.4	-	-
TNFAIP3	NM_006290.4	-	-
TNFRSF1A	NM_001065.4	chr12:g.6443045C>T (c.194-14G>A)	-
TPP2	NM_003291.4	-	CNV analysis in exon 28 is not performed
TREX1	NM_033629.6	-	-
TRNT1	NM_182916.3	-	-
UNC13D	NM_199242.2	chr17:g.73839908 to chr17:g.73839907C>T (c.118-308 to c.118-307); chr17:g.73827442C>T (c.2448-13G>A); chr17:g.73826245C>T (c.2831-13G>A)	-