MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Noonan Syndrome and Related Conditions Gene Panel

The following applies to NSRGG / Noonan Syndrome and Related Conditions 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 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 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 November 2022 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
ACTB	NM_001101.5	-	-
ACTG1	NM_001614.5	-	-
BRAF	NM_004333.6	-	-
CBL	NM_005188.4	-	-
HRAS	NM_005343.4	-	-
KRAS	NM_004985.5	-	-
LZTR1	NM_006767.4	-	-
MAP2K1	NM_002755.3	-	-
MAP2K2	NM_030662.3	-	-
MRAS	NM_012219.4	-	-
NRAS	NM_002524.5	-	-
PPP1CB	NM_206876.1	-	-
PTPN11	NM_002834.4	-	-
RAF1	NM_002880.3	-	-
RIT1	NM_006912.6	-	-
RRAS2	NM_012250.6	-	-
SHOC2	NM_007373.3	-	-
SOS1	NM_005633.3	-	-
SOS2	NM_006939.4	-	CNV may not be detected in exon 18.
SPRED1	NM_152594.3	-	-