MAYO CLINIC LABORATORIES | Targeted Genes and Methodology Details for Severe Congenital and Cyclic Neutropenia Gene Panel

The following applies to SCCNP / Severe Congenital and Cyclic Neutropenia 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
AK2	NM_001625.4	-	-
AP3B1	NM_003664.4	-	-
AP3D1	NM_001261826.3	-	-
CD40LG	NM_000074.3	-	-
CEBPE	NM_001805.3	-	-
CLPB	NM_030813.6	-	-
CSF3R	NM_000760.4	-	-
CXCR2	NM_001557.4	-	-
CXCR4	NM_003467.3	-	-
DNAJC21	NM_001012339.3	-	-
EFL1	NM_024580.6	-	CNV analysis in exon 15 is not performed
ELANE	NM_001972.4	-	-
G6PC3	NM_138387.3	-	-
GATA2	NM_032638.5	c.1017+526_1017+589 (corresponding to a highly conserved intronic region); c.1018-17_1020	-
GFI1	NM_005263.5	-	-
GINS1	NM_021067.5	c.-60A>G; c.-48C>G	-
HAX1	NM_006118.4	-	-
JAGN1	NM_032492.4	-	-
LYST	NM_000081.4	-	-
RAC2	NM_002872.5	-	-
SBDS	NM_016038.4	-	CNV analysis in exon 2 is not performed
SLC37A4	NM_001164277.1	-	-
SMARCD2	NM_001098426.2	-	-
SRP54	NM_003136.4	-	-
TAZ(TAFAZZIN)	NM_000116.5	c.284+110G>A	-
USB1	NM_024598.4	-	-
VPS13B	NM_017890.4	c.3446-23T>G	-
VPS45	NM_007259.5	-	-
WAS	NM_000377.3	c.1339-19_1339-11delinsATCTGCAGACC	-
WIPF1	NM_001077269.1	-	-