



The following applies to GNSPD / Platelet Storage Pool Deficiency Gene Panel, Next-Generation Sequencing. 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 June 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
<i>ABCC4</i>	NM_005845.4	-	-
<i>AP3B1</i>	NM_003664.4	-	-
<i>AP3D1</i>	NM_001261826.3	-	-
<i>BLOC1S3</i>	NM_212550.5	-	-
<i>BLOC1S5</i>	NM_201280.3	-	-
<i>BLOC1S6</i>	NM_012388.3	-	-
<i>DTNBP1</i>	NM_032122.4	-	-
<i>FLI1</i>	NM_002017.5	-	-
<i>GFI1B</i>	NM_004188.7	c.-20 to c.-1	-
<i>HPS1</i>	NM_000195.5	c.988-50 to c.988-1	-
<i>HPS3</i>	NM_032383.5	-	-
<i>HPS4</i>	NM_022081.5	-	-
<i>HPS5</i>	NM_181507.2	-	-
<i>HPS6</i>	NM_024747.6	-	-
<i>LYST</i>	NM_000081.4	c.9107-30 to c.9107-1	Sequence variants and CNV in exon 30 may not be detected or reported
<i>NBEA</i>	NM_015678.4	-	Sequence variants and CNV in exons 2, 6-13, and 16 may not be detected or reported
<i>NBEAL2</i>	NM_015175.3	-	Analyses for sequence variants and CNV in exon 1 will not be performed
<i>ORAI1</i>	NM_032790.3	-	-
<i>PLAU</i>	NM_002658.5	NM_001145031.2 Exon 1	-
<i>STIM1</i>	NM_003156.3	NM_001382567.1 Exon 11, NM_001277961.1 Exon 11, NM_001277962.1 Exon 12	
<i>STXBP2</i>	NM_006949.4	c.326-40 to c.326-1	-
<i>VIPAS39</i>	NM_022067.4	-	-
<i>VPS33B</i>	NM_018668.4	-	-
<i>WAS</i>	NM_000377.3	c.1339-30 to c.1339-1	-