MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Telomere Biology Disorders Gene Panel

The following applies to TELDP / Telomere Biology Disorders 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 to providers 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
ACD	NM_001082486.2	-	-
CTC1	NM_025099.6	-	-
DKC1	NM_001363.5	c.-142C>G	-
LIG4	NM_002312.3	-	-
NAF1	NM_138386.3	-	-
NHP2	NM_017838.3	-	-
NOP10	NM_018648.3	-	-
PARN	NM_002582.4	-	CNV analysis in exons 6; 19 is not performed.
POT1	NM_015450.3	-	CNV analysis in exon 5 is not performed.
RPA1	NM_002945.5	-	-
RTEL1	NM_032957.5	-	-
STN1	NM_024928.5	-	-
TERC	NR_001566.1	n.-58C>G; n.-22C>T	CNV analysis is not performed.
TERT	NM_198253.3	-	-
TINF2	NM_001099274.3	-	-
USB1	NM_024598.4	-	-
WRAP53	NM_018081.2	-	-
ZCCHC8	NM_017612.5	-	-