MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Hereditary Wilms Tumor Panel

Next-generation sequencing (NGS) and/or Sanger sequencing is performed to test for the presence of variants in coding regions and intron/exon boundaries of the genes analyzed. NGS and/or a polymerase chain reaction (PCR)-based quantitative method is performed to test for the presence of deletions and duplications in the genes analyzed. Confirmation of select reportable variants may be performed by alternate methodologies based on internal laboratory criteria.

Genomic Build: GRCh37 (hg19) unless otherwise specified

Gene	Reference Transcript[a]
BLM	NM_000057.4
BUB1B	NM_001211.5
CDC73	NM_024529.4
DIS3L2[b,c]	NM_152383.4
GPC3	NM_004484.4
REST	NM_005612.5
TP53	NM_000546.5
TRIP13	NM_004237.4
WT1	NM_024426.6

[a] Reference transcript numbers may be updated due to transcript re-versioning. Always refer to the final patient report for gene transcript information referenced at the time of testing.

[b] There are regions of this gene that cannot be effectively evaluated by sequencing as a result of technical limitations of the assay, including regions of homology, high GC content, and repetitive sequences.

[c] There are regions of this gene that cannot be effectively analyzed for the presence of copy number variants.

To verify if a specific region/exon/variant is covered by this assay, contact a laboratory genetic counselor at 800-533-1710.