MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for Histone H3.3 Mutations

Next-generation sequencing (NGS) is performed to test for the presence of single nucleotide variations, deletions, and insertions in coding regions and intron/exon boundaries of the genes listed. When appropriate, alterations detected are confirmed by an independent reference method, such as Sanger sequencing. Default reportable range offset is +/-2 base pairs around each targeted exon region.

Genomic Build: GRCh37 (hg19) unless otherwise specified

As a result of technical limitations of the assay (including regions of homology, high GC content, and repetitive sequences), there are regions of some genes that cannot be effectively evaluated. Refer to gene regions table below for complete gene coverage information. To verify if a specific region/exon/variant is covered by this assay, contact the laboratory at 800-533-1710.

Gene	Exon	Chromosome	Genomic Start	Genomic Stop	Reference Transcript
H3-3A	Ex2	chr1	226252051	226252182	NM_002107
H3-3A	Ex3	chr1	226253355	226253512	NM_002107
H3-3B	Ex2	chr17	73775126	73775257	NM_005324
H3-3B	Ex3	chr17	73774889	73775046	NM_005324
H3-3B	Ex4	chr17	73774674	73774806	NM_005324