MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for FH Mutation Analysis

The following applies to TFH / FH Mutation Analysis, Next-Generation Sequencing. Next-generation sequencing 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.

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.

Genomic Build: GRCh37 (hg19) unless otherwise specified.

Gene	Exon	Chromosome	Genomic Start	Genomic End	Transcript
FH	Ex1	chr1	241682889	241683024	NM_000143
FH	Ex2	chr1	241680480	241680618	NM_000143
FH	Ex3	chr1	241676901	241677015	NM_000143
FH	Ex4	chr1	241675265	241675445	NM_000143
FH	Ex5	chr1	241671901	241672087	NM_000143
FH	Ex6	chr1	241669301	241669470	NM_000143
FH	Ex7	chr1	241667340	241667547	NM_000143
FH	Ex8	chr1	241665741	241665872	NM_000143
FH	Ex9	chr1	241663735	241663892	NM_000143
FH	Ex10	chr1	241661126	241661272	NM_000143