MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for ESR1 Mutation Analysis

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 Stop	Reference Transcript
ESR1	Ex2	chr6	152129046	152129501	NM_001122740
ESR1	Ex3	chr6	152163730	152163924	NM_001122740
ESR1	Ex4	chr6	152201788	152201908	NM_001122740
ESR1	Ex5	chr6	152265306	152265645	NM_001122740
ESR1	Ex6	chr6	152332789	152332931	NM_001122740
ESR1	Ex7	chr6	152382124	152382261	NM_001122740
ESR1	Ex8	chr6	152415518	152415705	NM_001122740
ESR1	Ex9	chr6	152419865	152420103	NM_001122740