MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for *BRAF/KIT* 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 guanine-cytosine [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
BRAF	Ex1	chr7	140624364	140624505	NM_004333
BRAF	Ex2	chr7	140549909	140550014	NM_004333
BRAF	Ex3	chr7	140534407	140534674	NM_004333
BRAF	Ex4	chr7	140508690	140508797	NM_004333
BRAF	Ex5	chr7	140507758	140507864	NM_004333
BRAF	Ex6	chr7	140501210	140501362	NM_004333
BRAF	Ex7	chr7	140500160	140500283	NM_004333
BRAF	Ex8	chr7	140494106	140494269	NM_004333
BRAF	Ex9	chr7	140487346	140487386	NM_004333
BRAF	Ex10	chr7	140482819	140482959	NM_004333
BRAF	Ex11	chr7	140481374	140481495	NM_004333
BRAF	Ex12	chr7	140477789	140477877	NM_004333
BRAF	Ex13	chr7	140476710	140476890	NM_004333
BRAF	Ex14	chr7	140453985	140454035	NM_004333
BRAF	Ex15	chr7	140453073	140453195	NM_004333
BRAF	Ex16	chr7	140449085	140449220	NM_004333
BRAF	Ex17	chr7	140439610	140439748	NM_004333
BRAF	Ex18	chr7	140434395	140434572	NM_004333
KIT	Ex1	chr4	55524180	55524250	NM_000222
KIT	Ex2	chr4	55561676	55561949	NM_000222
KIT	Ex3	chr4	55564448	55564733	NM_000222
KIT	Ex4	chr4	55565794	55565934	NM_000222
KIT	Ex5	chr4	55569888	55570060	NM_000222
KIT	Ex6	chr4	55573262	55573455	NM_000222
KIT	Ex7	chr4	55575588	55575707	NM_000222
KIT	Ex8	chr4	55589748	55589866	NM_000222
KIT	Ex9	chr4	55592021	55592218	NM_000222
KIT	Ex10	chr4	55593382	55593492	NM_000222
KIT	Ex11	chr4	55593580	55593710	NM_000222
KIT	Ex12	chr4	55593987	55594095	NM_000222

Targeted Genes and Methodology Details for *BRAF/KIT* Mutation Analysis (continued)

Gene	Exon	Chromosome	Genomic Start	Genomic Stop	Reference Transcript
KIT	Ex13	chr4	55594175	55594289	NM_000222
KIT	Ex14	chr4	55595499	55595653	NM_000222
KIT	Ex15	chr4	55597492	55597587	NM_000222
KIT	Ex16	chr4	55598035	55598166	NM_000222
KIT	Ex17	chr4	55599234	55599360	NM_000222
KIT	Ex18	chr4	55602662	55602777	NM_000222
KIT	Ex19	chr4	55602885	55602988	NM_000222
KIT	Ex20	chr4	55603339	55603448	NM_000222
KIT	Ex21	chr4	55604593	55604725	NM_000222