MAYO CLINIC LABORATORIES

Targeted Genes and Methodology Details for BRAF/KIT Mutation Analysis

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
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