Seraseq® ctDNA v2 Mutation Mix and Reference Material (AF2%, AF1%, AF0.5%, AF0.25%, AF0.125%, WT)

For Research Use Only. Not for use in diagnostic procedures.

These products contain fragmented DNA from GM24385 cells and plasmids of spiked in variants in 0.1x TE buffer or synthetic plasma.

This file lists the genomic coordinates for the spike in variants included in the material numbers 0710-0139~0710-0144, 0710-0203~0710-0205, 0710-0207~0710-0208.

Gene	Nucleotide Change	Protein Change	Transcript	GRCh37 Location	GRCh38 Location	COSMIC ID	Type of Nucleotide Alteration
AKT1	c.49G>A	p.E17K	NM_001382430.1	14:105246551	14:104780214	COSM33765	SNV
APC	c.4348C>T	p.R1450*	NM_000038.6	5:112175639	5:112839942	COSM13127	SNV
APC	c.4666dup	p.T1556fs	NM_000038.6	5:112175957	5:112840260	COSM18561	Insertion
ATM	c.1058_1059del	p.C353fs	NM_000051.4	11:108117847_108117848	11:108247120_108247121	COSM21924	Deletion
BRAF	c.1799T>A	p.V600E	NM_004333.6	7:140453136	7:140753336	COSM476	SNV
CTNNB1	c.121A>G	p.T41A	NM_001904.4	3:41266124	3:41224633	COSM5664	SNV
EGFR	c.2236_2250del	p.E746_A750del	NM_005228.5	7:55242466_55242480	7:55174773_55174787	COSM6225	Deletion
EGFR	c.2310_2311insGGT	p.D770_N771insG	NM_005228.5	7:55249012_55249013	7:55181319_55181320	COSM12378	Insertion
EGFR	c.2369C>T	p.T790M	NM_005228.5	7:55249071	7:55181378	COSM6240	SNV
EGFR	c.2573T>G	p.L858R	NM_005228.5	7:55259515	7:55191822	COSM6224	SNV
ERBB2	c.2313_2324dup	p.Y772_A775dup	NM_004448.4	17:37880984_37880995	17:39724731_39724742	COSM20959	Insertion
FGFR3	c.746C>G	p.S249C	NM_000142.5	4:1803568	4:1801841	COSM715	SNV
FLT3	c.2503G>T	p.D835Y	NM_004119.3	13:28592642	13:28018505	COSM783	SNV
FOXL2	c.402C>G	p.C134W	NM_023067.4	3:138665163	3:138946321	COSM33661	SNV
GNA11	c.626A>T	p.Q209L	NM_002067.5	19:3118942	19:3118944	COSM52969	SNV
GNAQ	c.626A>C	p.Q209P	NM_002072.5	9:80409488	9:77794572	COSM28758	SNV
GNAS	c.601C>T	p.R201C	NM_000516.7	20:57484420	20:58909365	COSM27887	SNV
IDH1	c.394C>T	p.R132C	NM_005896.4	2:209113113	2:208248389	COSM28747	SNV
JAK2	c.1849G>T	p.V617F	NM_004972.4	9:5073770	9:5073770	COSM12600	SNV
KIT	c.2447A>T	p.D816V	NM_000222.3	4:55599321	4:54733155	COSM1314	SNV
KRAS	c.35G>A	p.G12D	NM_004985.5	12:25398284	12:25245350	COSM521	SNV
MPL	c.1544G>T	p.W515L	NM_005373.3	1:43815009	1:43349338	COSM18918	SNV
NPM1	c.860_863dup	p.W288fs	NM_002520.7	5:170837544_170837547	5:171410540_171410543	COSM17559	Insertion
NRAS	c.182A>G	p.Q61R	NM_002524.5	1:115256529	1:114713908	COSM584	SNV
PDGFRA	c.1694_1695insA	p.S566fs	NM_006206.6	4:55141048_55141049	4:54274881_54274882	COSM28053	Insertion
PDGFRA	c.2525A>T	p.D842V	NM_006206.6	4:55152093	4:54285926	COSM736	SNV
PIK3CA	c.1633G>A	p.E545K	NM_006218.4	3:178936091	3:179218303	COSM763	SNV
PIK3CA	c.3140A>G	p.H1047R	NM_006218.4	3:178952085	3:179234297	COSM775	SNV
PIK3CA	c.3204_3205insA	p.*1069Mext*3	NM_006218.4	3:178952149_178952150	3:179234361_179234362	NA	Insertion
PTEN	c.741dup	p.P248fs	NM_000314.8	10:89717716	10:87957959	COSM4986	Insertion
PTEN	c.800del	p.K267fs	NM_000314.8	10:89717775	10:87958018	COSM5809	Deletion

RET	c.2753T>C	p.M918T	NM_020975.6	10:43617416	10:43121968	COSM965	SNV
SMAD4	c.1394dup	p.A466fs	NM_005359.6	18:48603093	18:51076723	COSM14105	Insertion
TP53	c.267del	p.S90fs	NM_000546.6	17:7579420	17:7676102	COSM18610	Deletion
TP53	c.524G>A	p.R175H	NM_000546.6	17:7578406	17:7675088	COSM10648	SNV
TP53	c.723del	p.C242fs	NM_000546.6	17:7577558	17:7674240	COSM6530	Deletion
TP53	c.743G>A	p.R248Q	NM_000546.6	17:7577538	17:7674220	COSM10662	SNV
TP53	c.818G>A	p.R273H	NM_000546.6	17:7577120	17:7673802	COSM10660	SNV

Translocation	5' Transcript	5' Breakpoint GRCh37	5' Breakpoint GRCh38	3' Transcript	3' Breakpoint GRCh37	3' Breakpoint GRCh38
NCOA4::RET	NM_001145263.2	10:51584251	10:46011571	NM_020975.6	10:43610593	10:43115146
TPR::ALK	NM_003292.3	1:186325172	1:186356039	NM_004304.5	2:29446944	2:29224079

NOTE: Genomic coordinates use the 1-based coordinate system.