# 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 |

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|---|---|---|---|---|---|---|---|
| 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.