

# Seraseq™ ctDNA v2 Reference Materials

Gene ID	HGVS	Amino Acid Change	Mutation Type
AKT1	c.49G>A	p.E17K	SNV
APC	c.4666_4667insA	p.T1556fs*3	Insertion in homopolymer (7N)
APC	c.4348C>T	p.R1450*	SNV
ATM	c.1058_1059delGT	p.C353fs*5	Deletion
BRAF	c.1799T>A	p.V600E	SNV
CTNNB1	c.121A>G	p.T41A	SNV
EGFR	c.2236_2250del15	p.E746_A750delELREA	Deletion
EGFR	c.2310_2311insGGT	p.D770_N771insG	Insertion
EGFR	c.2573T>G	p.L858R	SNV
EGFR	c.2369C>T	p.T790M	SNV
ERBB2	c.2324_2325ins12	p.A775_G776insYVMA	Insertion
FGFR3	c.746C>G	p.S249C	SNV
FLT3	c.2503G>T	p.D835Y	SNV
FOXL2	c.402C>G	p.C134W	SNV
GNA11	c.626A>T	p.Q209L	SNV
GNAQ	c.626A>C	p.Q209P	SNV
GNAS	c.601C>T	p.R201C	SNV
IDH1	c.394C>T	p.R132C	SNV
JAK2	c.1849G>T	p.V617F	SNV
KIT	c.2447A>T	p.D816V	SNV
KRAS	c.35G>A	p.G12D	SNV
MPL	c.1544G>T	p.W515L	SNV
NCOA4-RET	NCOA4{NC_000010.10}:r.1_1014+1312_RET {NC_000010.10}:r.2327-1437_5659	N/A	Gene Fusion (DNA)
NPM1	c.863_864insTCTG	p.W288fs*12	Insertion
NRAS	c.182A>G	p.Q61R	SNV
PDGFRA	c.1694_1695insA	p.S566fs*6	Insertion
PDGFRA	c.2525A>T	p.D842V	SNV
PIK3CA	c.3204_3205insA	p.N1068fs*4	Insertion
PIK3CA	c.1633G>A	p.E545K	SNV
PIK3CA	c.3140A>G	p.H1047R	SNV
PTEN	c.800delA	p.K267fs*9	Deletion in homopolymer (6N>5N)
PTEN	c.741_742insA	p.P248fs*5	Insertion
RET	c.2753T>C	p.M918T	SNV
SMAD4	c.1394_1395insT	p.A466fs*28	Insertion
TP53	c.723delC	p.C242fs*5	Deletion
TP53	c.263delC	p.S90fs*33	Deletion in homopolymer (5N>4N)
TP53	c.524G>A	p.R175H	SNV
TP53	c.818G>A	p.R273H	SNV
TP53	c.743G>A	p.R248Q	SNV
TPR-ALK	TPR{NC_000001.10}:r.1_2185+246_ALK {NC_000002.11}:r.4125-550_6265	N/A	Gene Fusion (DNA)

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