Q-Seq Reference Standards

Tru-Q 1 (5%) Mutation List

Mutation	Genome Change	Cosmic IDs	Validated	Allelic Frequency (%)
Engineered Mutations				
BRAF V600E	g.chr7:140453136A>T	COSM476	✔	8
BRAF V600K	g.chr7:140453136_140453137AC>TT	COSM473	✔	4
EGFR G719S*	g.chr7:55241707G>A	COSM6252	✔	16.7
EGFR T790M	g.chr7:55249071C>T	COSM6240	✔	4.2
FLT3 ΔI836	g.chr10:28592637_28592639delGAT	COSM797	✔	5
IDH1 R132C	g.chr2:209113113G>A	COSM28747	✔	5
JAK2 V617F	g.chr9:5073770G>T	COSM12600	✔	5
KRAS G12A	g.chr12:25398284C>G	COSM522	✔	5
KRAS G12R	g.chr12:25398285C>G	COSM518	✔	5
KRAS G13D*	g.chr12:25398281C>T	COSM532	✔	25
MEK1 P124L	g.chr15:66729163C>T	COSM1315861	✔	5
NOTCH L1601P	g.chr9:139399366A>G	COSM12771	✔	4.8
NRAS Q61K	g.chr1:115256530G>T	COSM580	✔	5
PIK3CA H1047R*	g.chr3:178952085A>G	COSM775	✔	30
Endogenous Onco-relevant Mutations				
ABL2 P986fs	g.chr1:179077445_179077445delG	N/A	✔	5
ALK P1543S	g.chr2:29416326G>A	N/A	✔	20
APC R2714C	g.chr5:112179431C>T	N/A	✔	20
ARID1A P1562fs	g.chr1:27101402_27101402delC	N/A	✔	20
BRCA2 A1689fs	g.chr13:32913559_32913559delA	N/A	✔	20
CCND2 3'UTR	g.chr12:4409271_4409271delA	N/A	✔	20
CDH1 3'UTR	g.chr16:68867505_68867505delA	N/A	✘	5
CDX2 V306fs	g.chr13:28537278_28537278delC	N/A	✘	45
CTNNB1 S33Y	g.chr3:41266101C>A	N/A	✔	20
CTNNB1 S45del	g.chr3:41266134_41266136delCTT	N/A	✔	25
EP300 K291fs	g.chr22:41522009_41522009delA	N/A	✔	5
FANCA E345fs	g.chr16:89858927_89858928delCT	N/A	✘	5
FBXW7 G667fs	g.chr4:153244156_153244156delC	N/A	✔	20
FGFR1 P150L	g.chr8:38285611G>A	N/A	✔	5
FLT3 S985fs	g.chr13:28578215_28578216delGA	N/A	✔	25
FLT3 V197A	g.chr13:28626706A>G	N/A	✔	25
IDH1 S261L	g.chr2:209106786G>A	N/A	✔	25
MET V237fs	g.chr7:116339848_116339848delT	N/A	✔	20
MLH1 L323M	g.chr3:37061883C>A	N/A	✔	5
NF1 L626fs	g.chr17:29552144_29552144delT	N/A	✔	5
NF2 P275fs	g.chr22:30060991_30060991delC	N/A	✘	5
NOTCH1 P668S	g.chr9:139409754G>A	N/A	✔	19
NTRK1 5'UTR	g.chr1:156785545_156785545delC	N/A	✔	5
PDGFRA G426D	g.chr4:55138600G>A	N/A	✔	20

To view the list of Secondary Endogenous Mutations, please contact:

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