

# SeraSeq™ VariantFlex™ Catalog

FLEXIBLE, READY-TO-CONFIGURE DNA MUTATIONS AND RNA GENE FUSIONS

**TABLE 1**

List of available mutations with gene ID, COSMIC identifier, and mutation type.

Gene ID	COSMIC Identifier	Mutation Type	HGVS Nomenclature	Amino Acid
EGFR	COSM6225	Deletion	c.2236_2250del15	p.E746_A750delELREA
SMAD4	COSM14105	Insertion	c.1394_1395insT	p.A466fs*28
ERBB2	COSM682/20959	Insertion	c.2324_2325ins12	p.A775_G776insYVMA
APC	COSM18561	Insertion in HP 7N	c.4666_4667insA	p.T1556fs*3
NCOA4-RET	NA	Structural Variant	NA	NA
TPR-ALK	NA	Structural Variant	NA	NA
MPL	COSM18918	Substitution	c.1544G>T	p.W515L
PIK3CA	COSM763	Substitution	c.1633G>A	p.E545K
PDGFRA	COSM736	Substitution	c.2525A>T	p.D842V
KIT	COSM1314	Substitution	c.2447A>T	p.D816V
APC	COSM13127	Substitution	c.4348C>T	p.R1450*
GNAQ	COSM28758	Substitution	c.626A>C	p.Q209P
AKT1	COSM33765	Substitution	c.49G>A	p.E17K
GNA11	COSM52969	Substitution	c.626A>T	p.Q209L
TP53	COSM18610	Deletion 5N >4N	c.263delC	p.S90fs*33
PIK3CA	COSM12464	Insertion	c.3204_3205insA	p.N1068fs*4
PDGFRA	COSM28053	Insertion	c.1694_1695insA	p.S566fs*6
EGFR	COSM12378	Insertion	c.2310_2311insGGT	p.D770_N771insG
ATM	COSM21924	Deletion	c.1058_1059delGT	p.C353fs*5
IDH1	COSM28747	Substitution	c.394C>T	p.R132C
PIK3CA	COSM775	Substitution	c.3140A>G	p.H1047R
FGFR3	COSM715	Substitution	c.746C>G	p.S249C
EGFR	COSM6240	Substitution	c.2369C>T	p.T790M
RET	COSM965	Substitution	c.2753T>C	p.M918T
FLT3	COSM783	Substitution	c.2503G>T	p.D835Y
BRAF	COSM476	Substitution	c.1799T>A	p.V600E
FOXL2	COSM33661	Substitution	c.402C>G	p.C134W
NPM1	COSM17559	Insertion	c.863_864insTCTG	p.W288fs*12
TP53	COSM6530	Deletion	c.723delC	p.C242fs*5
PTEN	COSM5809	Deletion 6N > 5N	c.800delA	p.K267fs*9
PTEN	COSM4986	Insertion	c.741_742insA	p.P248fs*5
NRAS/CSDE1	COSM584	Substitution	c.182A>G	p.Q61R
CTNNB1	COSM5664	Substitution	c.121A>G	p.T41A
EGFR	COSM6224	Substitution	c.2573T>G	p.L858R

Gene ID	COSMIC Identifier	Mutation Type	HGVS Nomenclature	Amino Acid
JAK2	COSM12600	Substitution	c.1849G>T	p.V617F
KRAS	COSM521	Substitution	c.35G>A	p.G12D
TP53	COSM10660	Substitution	c.818G>A	p.R273H
TP53	COSM10662	Substitution	c.743G>A	p.R248Q
TP53	COSM10648	Substitution	c.524G>A	p.R175H
GNAS	COSM27887	Substitution	c.601C>T	p.R201C

**TABLE 2**

List of available RNA gene fusions with gene name, primary tissue, and exon partners.

RNA Fusion	Primary Cancer Tissue	5' Partner- Exon #	3' Partner- Exon #
EML4-ALK	Lung	EML4 Exon 13	ALK Exon 20
NPM1-ALK	Lymphoid	NPM1 exon 5	ALK Exon 20
KIF5B-RET	Lung	KIF5B Exon 24	Ret Exon 11
NCOA4-RET	Thyroid	NCOA4 Exon 8	RET exon 12
CD74-ROS1	Lung	CD74 Exon 6	Ros 1 Exon 34
SLC34A2-ROS1	Lung, Stomach	SLC34A Exon 4	Ros 1 Exon 34
TPM3-NTRK1	Lung, Large Intestine	TPM3 Exon 8	NTRK1 Exon 10
TFG-NTRK1	Thyroid (rare)	TFG Exon 5	NTRK1 Exon 10
FGFR3-BAIAP2L1	Urinary tract (rare)	FGFR3 Exon 18	BAIAP2L1 Exon 2
FGFR3-TACC3	Urinary tract, CNS	FGFR3 exon 18	TACC3 Exon 11
PAX8-PPARG	Thyroid	Pax8 Exon 8	PPARG Exon 1
ETV6-NTRK3	Kidney, Breast, Soft Tissue	ETV6 Exon 5	NTRK3 Exon 13



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