

EGFR exon 19 deletions subtypes in patients with NSCLC

Original publication: “Molecular characteristics and clinical outcomes of EGFR exon 19 indel subtypes to EGFR TKIs in NSCLC patients”, Su et al; [Oncotarget. 2017; 8:111246-111257](https://doi.org/10.1186/1745-6215-8-111)

Table 2 from that publication with additional columns “dbSNP” and “Observations” describing the deletion in terms of frame-context and relation to presence in dbSNP and COSMIC

Highlighted rows refer to “somatic” haplotypes at https://replicongenetics.com/helpv2/#EGFR_help

	Amino acid change	Base pair change	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	Cases	%	Cosmic ID	Mis sed*	dbSNP	Observations
			Protein: I K E L R E A T S P K A N K E I L D E A						
			atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc						
1	p.E746_A750 del(1)	c.2235_2249del15	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	198	45.0	COSM6223		rs121913421	Agrees with COSMIC Deletion: “c.2235_2249del15” 15nuc deletion in-frame.
2	p.E746_T751 >KV	c.2235_2252>gggt	atc aag gaa tta aga gaa gca acGGTa tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	1	0.2	NA	1		Delins: “c.2235_2252>” 18 nuc deletion plus a 3 nuc insert (net 15 del: frame preservation after missense)
3	p.E746_A750 >HS	c.2236_2248>ctaa	atc aag gaa tta aga gaa gCAT Tca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	1	0.2	NA	1		Delins: “c.2236_2248” 13 nuc deletion plus 4 nuc insert (net: 9 nuc insert: frame preservation after missense)
4	p.E746_A750 del(3)	c.2236_2249del14	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	3	0.7	NA	3		Deletion: “c.2236_2249del14” 14 nuc deletion, so frame disruption, possibly early termination or missense after? 1 nuc less deletion than entry 5
5	p.E746_A750 del(2)	c.2236_2250del15	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	86	19.6	COSM6225		rs727504233	Agrees with COSMIC Deletion: annotation “c.2236_2250del15” 15 nuc deletion in-frame.
6	p.E746_T750 del(4)	c.2236_2251>a	atc aaggaa tta aga gaa gca aAca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	1	0.2	NA	1		Delins: annotation “c.2236_2251>a” 16 nuc deletion, then 1 nuc insert. (net 15 del: frame preservation, but no missense annotated) 1 bp insert compared to entry 5
7	p.E746_T751 >FPT	c.2236_2251>tttccaa	atc aag gaa tta aga gaa gca aTTTCCAAca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	1	0.2	NA	1		Delins: annotation “c.2236_2251>tttccaa” 16 nuc deletion , plus 7 nuc insert (net 9 del: frame preservation after missense)
8	p.E746_T751 >L	c.2236_2252>ct	atc aag gaa tta aga gaa gca acCTa tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	1	0.2	COSM51502	1	rs727504402	Delins: “c.2236_2252>” 17 nuc deletion, plus 2 nuc insert (net 15 del: frame preservation after missense)
9	p.E746_S752 >IP	c.2236_2255>atacc	atc aag gaa tta aga gaa gca aca tcA TAC Ct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	1	0.2	NA	1		Delins: “c.2236_2255>atacc” 20 nuc deletion plus 5 nuc insert (net 15 del: frame preservation after missense)
10	p.E746_P753 >MS	c.2236_2257>atgt	atc aag gaa tta aga gaa gca aca tct cATGTcg aaa gcc aac aag gaa atc ctc gat gaa gcc	1	0.2	NA	1		Delins: “c.2236_2257>atgt” a 22 nuc deletion plus 4 nuc insert (net 18 del: frame preservation after missense)

	Amino acid change	Base pair change	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc Protein: I K E L R E A T S P K A N K E I L D E A	Cases	%	Cosmic ID	Mis sed*	dbSNP	Observations
11	p.E746_P753 >MS	c.2236_2257>atgt c	atc aag gaa tta aga gaa gca aca tct cATGTCcg aaa gcc aac aag gaa atc ctc gat gaa gcc	1	0.2	NA	1		Delins: “c.2236_2257>atgtc” a 22 nuc deletion plus 5 nuc insert (net 17 del: frame disruption after missense?) 1 more nuc insert than entry 10 Agrees with COSMIC deletion: “c.2237_2251del15” a 15 nuc deletion (frame preservation after missense?) Delins: “c.2237_2253>caccaact” 17 nuc deletion plus 8 nuc insert (net 9 deletion: frame preservation after missense) Delins: “c.2237_2253>ttgct” 17 nuc deletion plus 5 nuc insert (net 12 deletion: frame preservation after missense)
12	p.E746_T751 >A	c.2237_2251del15	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	3	0.7	COSM12678		rs121913425	
13	p.E746_T751 >APT	c.2237_2253>caccaact	atc aag gaa tta aga gaa gca aca CACCAACTtct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	1	0.2	NA	1		
14	p.E746_T751 >VA	c.2237_2253>ttgct	atc aag gaa tta aga gaa gca acaTT GCT tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	3	0.7	12416	3		
15	p.E746_S752 >V	c.2237_2255>t	atc aag gaa tta aga gaa gca aca tcTt ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	7	1.6	COSM12384		rs727504258	Agrees with COSMIC Delins: “c.2237_2255>t” 19 nuc deletion, plus 1 nuc insert (net 18 deletion: frame preservation after missense) Agrees with COSMIC delins: “c.2237_2257>tct” 21 nuc deletion, plus 3 nuc insert (net 18 deletion: frame preservation after missense) similar to entry 15
16	p.E746_P753 >VS	c.2237_2257>tct	atc aag gaa tta aga gaa gca aca tct cTCTcg aaa gcc aac aag gaa atc ctc gat gaa gcc	3	0.7	COSM18427	3	rs727504282	delins: “c.2237_2257>tct” 21 nuc deletion, plus 3 nuc insert (net 18 deletion: frame preservation after missense) similar to entry 15
17	p.E746_S752 >V(2)	c.2237_2257>ttc	atc aag gaa tta aga gaa gca aca tct cTTCcg aaa gcc aac aag gaa atc ctc gat gaa gcc	14	3.2	NA	14		delins: “c.2237_2257>ttc” 21 nuc deletion, plus 3 nuc insert (net 18 deletion: frame preservation after missense) similar to entry 15
18	p.E746_K754 >GG	c.2237_2261>tcgg	atc aag gaa tta aga gaa gca aca tct ccg aaTCGGa gcc aac aag gaa atc ctc gat gaa gcc	1	0.2	NA	1		delins: “c.2237_2261>tcgg” 25 nuc deletion plus 4 nuc insert (net 21 deletion: frame preservation + missense)
19	p.E746_E749 del	c.2238_2247del10	atc aag gaatta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	2	0.5	NA	2		Deletion: “c.2238_2247del10” 10 nuc deletion: frame disruption – outcome not obvious
20	p.E746_E749 >P	c.2238_2249>gcc	atc aag gaa tta aga gaa gc GCCa aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	3	0.9	NA	3		delins: “c.2238_2249>gcc” 12 nuc deletion plus 3 nuc insertion (net 9 deletion: frame preservation after missense) Agrees with COSMIC
21	p.L747_E749 del	c.2239_2247del9	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	2	0.5	COSM6218		No dbsnp	Deletion: “c.2239_2247del9” 9 nuc deletion. Frame preservation 133195 does not exist in COSMIC. closest position-match is COSM4170220 but that is c.2239_2250delinsCCG not
22	p.L747_A750 >P	c.2239_2250>cca	atc aag gaa tta aga gaa gcaCCA aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	15	3.4	COSM133195	16		c.2239_2250delinsCCA delins: “c.2239_2250>cca” 12 nuc deletion and 3 nuc

	Amino acid change	Base pair change	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc Protein: I K E L R E A T S P K A N K E I L D E A	Cases	%	Cosmic ID	Mis sed*	dbSNP	Observations
									insert (net: 9 nuc deletion: frame preservation after missense) Disagrees with COSMIC so mis-annotation & this one appears not to be in cosmic.
23	p.L747_T751 >P	c.2239_2253>cca	atc aag gaa tta aga gaa gca aca CCA tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	5	1.1	COSM51527	4	No dbsnp	delins: “c.2239_2253>cca” 15 nuc deletion, plus 3nuc insert (net: 12 nuc deletion: frame preservation after missense) Agrees with COSMIC delins: “c.2239_2253>aat” 15 nuc deletion and 3 nuc insert (net: 12 nuc deletion: frame preservation after missense)
24	p.L747_T751 >N	c.2239_2253>aat	atc aag gaa tta aga gaa gca aca AAT tct ccg aaa gcc aac aag gaa atc ctc gat	1	0.2	COSM51503		No dbsnp	delins: “c.2239_2254>ccga” 16 nuc deletion and 4 nuc insert (net 12 nuc deletion - frame preservation after missense) delins: “c.2239_2256>aattcg” 18 nuc deletion, 6 nuc insert(net: 12 nuc deletion, frame preservation after missense)
25	p.L747_S752 >PT	c.2239_2254>ccg a	atc aag gaa tta aga gaa gca aca tCCGAct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	1	0.2	NA	1		delins: “c.2239_2256>caaata” 18 nuc deletion, 6 nuc insert(net: 12 nuc deletion, frame preservation after missense)
26	p.L747_P753 >NS	c.2239_2256>aatt cg	atc aag gaa tta aga gaa gca aca tct ccg AATTCGaaa gcc aac aag gaa atc ctc gat gaa gcc	1	0.2	NA	1		delins: “c.2239_2256>caaata” 18 nuc deletion, 6 nuc insert(net: 12 nuc deletion, frame preservation after missense)
27	p.L747_S752 >PI	c.2239_2256>caaa ta	atc aag gaa tta aga gaa gca aca tctCAAATAccg aaa	1	0.2	NA	1		Agrees with COSMIC deletion: “c.2239_2256del18” 18 nuc deletion only & frame preservation. delins: “c.2239_2259>tc” 21 nuc deletion plus 2 nuc insert (net: 19 nuc deletion: frame disruption after missense– outcome not obvious)
28	p.L747_S752 del	c.2239_2256del18	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	6	1.4	COSM6255		rs121913440	deletion: “c.2239_2262del24” 24 nuc deletion-only
29	p.L747_P753 >S	c.2239_2259>tc	atc aag gaa tta aga gaa gca aca tct ccg TCaaagcc aac aag	1	0.2	NA	1		Agrees with COSMIC deletion: “c.2240_2251del12” 12 nuc deletion frame preservation after missense.
30	p.L747_K754 del	c.2239_2262del24	atc aag gaa tta aga gaa gca aca tct ccg aaagcc aac aag gaa atc ctc gat gaa gcc	1	0.2	COSM24970	1	No dbsnp	Agrees with COSMIC deletion: “c.2240_2254del15” 15 nuc deletion only. Frame preservation.
31	p.L747_T751 >S	c.2240_2251del12	atc aag gaa tta aga gaa gca aca tct ccg aaa	1	0.2	COSM6210		rs121913441	Agrees with COSMIC deletion: “c.2240_2257del18” 18 nuc deletion. Frame preservation after missense.
32	p.L747_T751 del	c.2240_2254del15	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	19	4.3	COSM12369		No dbsnp	
33	p.L747_P753 >S	c.2240_2257del18	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	37	8.4	COSM12370		rs121913438	
34	p.A750_I759 >PT	c.2248_2276>ccaa c	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa	1	0.2	5023004	1		delins: “c.2248_2276>ccaac” 29 nuc deletion and 5 nuc

	Amino acid change	Base pair change	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc Protein: I K E L R E A T S P K A N K E I L D E A atCCAACc ctc gat gaa gcc	Cases	%	Cosmic ID	Mis sed*	dbSNP	Observations
35	p.T751_I759>T	c.2253_2275del23	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	1	0.2	NA	1		insert (net 24 nuc deletion: frame preservation after missense) deletion: "c.2253_2275del23" 23 nuc deletion. Frame disruption after missense – outcome not obvious
36	p.S752_I759del(2)	c.2253_2276del24	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	2	0.5	COSM13556	2	rs727504232	1bp less than entry 36 Agrees with COSMIC deletion: "c.2253_2276del24" 24 nuc deletion in-frame.
37	p.T751_I759>N	c.2252_2276>a	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atAc ctc gat gaa gcc	1	0.2	COSM96856	1		Agrees with COSMIC delins: "c.2252_2276>a" 25 nuc deletion, 1 nuc insert (net: 24 nuc deletion frame preservation after missense) Delins: "c.2252_2281>atctct" 30 nuc deletion, 6 nuc insert (net: 24 nuc deletion frame preservation after missense)
38	p.T751_D761>NLY	c.2252_2281>atctct	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gATCTCTat gaa gcc	1	0.2	NA	1		
39	p.S752_I759del	c.2254_2277del24	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	3	0.7	6255	3		Must be an error as COSM6255 already in table (entry 28) with different annotation
40	untyped			8	1.8				
				440			72		

	Amino acid change	Base pair change	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc Protein: I K E L R E A T S P K A N K E I L D E A atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	Cases	%	Cosmic ID	Mis sed*	Dbsnp	Observations
1 som1	p.E746_A750 del(1)	c.2235_2249del15	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	198	45.0	COSM6223		rs121913421	Agrees with COSMIC Deletion: “c.2235_2249del15” 15nuc deletion in-frame.
14 som2	p.E746_T751 >VA	c.2237_2253>ttgct	atc aag gaa tta aga gaa gca acaTT GCT tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	3	0.7	12416	3		Delins: “c.2237_2253>ttgct” 17 nuc deletion plus 5 nuc insert (net 12 deletion: frame preservation after missense)
15 som3	p.E746_S752 >V	c.2237_2255>t	atc aag gaa tta aga gaa gca aca tcTt ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	7	1.6	COSM12384		rs727504258	Agrees with COSMIC Delins: “c.2237_2255>t” 19 nuc deletion, plus 1 nuc insert (net 18 deletion: frame preservation after missense)
16 som4	p.E746_P753 >VS	c.2237_2257>tct	atc aag gaa tta aga gaa gca aca tct cTCTcg aaa gcc aac aag gaa atc ctc gat gaa gcc	3	0.7	COSM18427	3	rs727504282	Agrees with COSMIC delins: “c.2237_2257>tct” 21 nuc deletion, plus 3 nuc insert (net 18 deletion: frame preservation after missense) similar to entry 15
32 som6	p.L747_T751 del	c.2240_2254del15	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	19	4.3	COSM12369		No dbsnp	Agrees with COSMIC deletion: “c.2240_2254del15” 15 nuc deletion only. Frame preservation.
39 som5	p.S752_I759d el	c.2254_2277del24	atc aag gaa tta aga gaa gca aca tct ccg aaa gcc aac aag gaa atc ctc gat gaa gcc	3	0.7	6255	3		Must be an error as COSM6255 already in table (entry 28) with different annotation