

DNA Alteration	Patient ID	Localization	Other info	Mapping of alterations (Hg19)	Mutation
Frameshift deletion					
ALT1	1149	Ex 10	10-bp deletion	g.7573999-7574008het_delTGTTCCGAGA	yes
ALT2	3036	Ex 10	7-bp deletion	g.7574000-7574006delTTCCGAG	yes
ALT3	3048	Ex 10	1-bp deletion	g.7574013-7574013het_delC	yes
Frameshift insertion					
ALT4	3477	Ex 11	1-bp insertion	g.7572933-7572933dupA	yes
Non-frameshift deletion					
ALT5	1144	Ex 6	3-bp deletion	g.7578275-7578277delCTC	yes
ALT6	1133	Ex 10	6-bp insertion	g.7574026-7574026dupGGCGTG	yes
ALT7	2557	Ex 5	27-bp insertion	g.7578447-7578447dupCATCTACAAGCAGTCACAGCACATGAC	yes
Non-synonymous SNV					
ALT8	1133, 1134, 1135, 1136, 1137, 1138, 1140, 1142, 1143, 1145, 1146, 1706, 2220, 2557, 2967, 3033, 3039, 3041, 3043, 3044, 3045, 3047, 3048, 3050,	Ex 4	Single substitution	g.7579472-7579472C>G	no

3053, 3057, 3058, 3061,
3062, 3143, 4718

ALT9	3143	Ex 5	Single substitution	g.7578394-7578394A>C	yes
ALT10	3044	Ex 5	Single substitution	g.7578406-7578406G>GA	yes
ALT11	3056	Ex 5	Single substitution	g.7578413-7578413G>A	yes
ALT12	3047	Ex 5	Single substitution	g.7578457-7578457G>GC	yes
ALT13	3052	Ex 5	Single substitution	g.7578526-7578526G>GT	yes
ALT14	1134	Ex 8	Single substitution	g.7577095-7577095C>CA	yes
ALT15	1134	Ex 8	Single substitution	g.7577118-7577118G>GT	yes
ALT16	1142, 1706	Ex 8	Single substitution	g.7577120-7577120G>GA	yes
ALT17	2967	Ex 8	Single substitution	g.7577129-7577129T>T/G	yes

Splicing mutation

ALT18	1135	intronic	10-bp deletion	g.7578296-7578305het_delTCACTGATTG	yes
ALT19	3041	intronic	11-bp deletion	g.7578298-7578308delCCTCACTGATT	yes

Stop-gain SNV

ALT20	1136	Ex 10	Single substitution	g.7574003-7574003C>T	yes
ALT21	4718	Ex 6	Single substitution	g.7578212-7578212C>CT	yes

Synonymous SNV

ALT22	1133, 1134, 1136, 1146, 1149, 2557, 3044, 3055, 3058, 3062, 3143, 4718	Ex 3	Single substitution	g.7579579-7579579G>GA	no
--------------	--	------	---------------------	-----------------------	----

Silent mutations

ALT23	1133, 1134, 1135, 1136, 1137, 1138, 1140, 1142, 1143, 1144, 1145, 1146, 1149, 1706, 2220, 2557, 2967, 3033, 3036, 3043, 3044, 3045, 3047, 3048, 3050, 3052, 3053, 3055, 3056, 3057, 3058, 3060, 3061, 3062, 3143	Ex 11, UTR3	Single substitution	g.7572890-7572890A>T	no
ALT24	1133, 1134, 1136, 1137, 1138, 1140, 1142, 1143, 1145, 1706, 2220, 2557, 2967, 3033, 3039, 3041, 3043, 3044, 3045, 3047, 3048, 3052, 3053, 3057, 3058, 3061, 3062, 3143, 4718	Ex 2, UTR5	Single substitution	g.7579801-7579801C>G	no
ALT25	1135, 3050	intronic	Single substitution	g.7576841-7576841T>TC	no
ALT26	1135	intronic	Single substitution	g.7577188-7577188T>TC	no
ALT27	1146	intronic	Single substitution	g.7579596-7579596C>CT	no
ALT28	1146	intronic	Single substitution	g.7579596-7579596C>CT	no
ALT29	1140, 1146, 3477	intronic	Single substitution	g.7579619-7579619C>CA	no
ALT30	1133, 1134, 1136, 1137, 1138, 1140, 1142, 1143, 1145, 1146, 1149, 1706, 2220, 2557, 3033, 3036, 3039, 3041, 3043, 3044, 3045, 3047, 3048, 3050, 3052, 3053, 3055, 3056, 3057, 3058, 3060, 3061, 3062, 3143, 3477, 4718	intronic	16-bp deletion	g.7579644-7579659del16	no
