

**Additional File 4: Table Splicing Mutation Descriptions**

Exon/Intron	Nucleotide	Effect	Protein	GOF/LOF	DNE	Splicing info	COSMIC ID	Functional source	Count
Intron 2	c.96+1G>A	splicing	p.?	LOF	No DNE	consensus SD	COSM44435	Implied	1
Exon 3	c.75_96del	frameshift	p.L26fs	LOF	No DNE	TRUE	COSM45386	Implied	1
Exon 4	c.374?del	frameshift	p.T125fs	LOF	No DNE	TRUE	NA	Implied	1
Exon 4	c.374_375insA	frameshift	p.T125fs	LOF	No DNE	TRUE	NO-ID	Implied	1
Exon 4	c.374C>A	missense	p.T125K	LOF	Unknown	TRUE	COSM44073	(94)	2
Exon 4	c.375G>A	splicing	p.?	LOF	No DNE	TRUE	NA	Implied	1
Exon 4	c.375G>C	splicing	p.?	LOF	No DNE	TRUE	COSM46152	Implied	2
Exon 4	Unknown	frameshift	p.T125fs	LOF	No DNE	TRUE	NA	Implied	1
Exon 4	Unknown	splicing	p.?	LOF	No DNE	TRUE	NA	(80)	1
Intron 4	c.?(i4-intron_G>A)	splicing	p.?	LOF	No DNE	TRUE	NA	Implied	1
Intron 4	c.370_375+1delins1	splicing	p.?	LOF	No DNE	TRUE	NO-ID	Implied	1
Intron 4	c.375+1_375+2ins1	splicing	p.?	LOF	No DNE	consensus SD	NO-ID	Implied	1
Intron 4	c.375+1del	splicing	p.?	LOF	No DNE	consensus SD	COSM318394	Implied	1
Intron 4	c.375+1G>A	splicing	p.?	LOF	No DNE	consensus SD	COSM45304	Implied	1
Intron 4	c.375+1G>T	splicing	p.?	LOF	No DNE	consensus SD	COSM69405	Implied	2
Intron 4	c.375+2T>G	splicing	p.?	LOF	No DNE	consensus SD	COSM46043	Implied	1
Intron 4	c.375del	splicing	p.?	LOF	No DNE	consensus SD	NO-ID	Implied	1
Intron 4	c.375G>T	splicing	p.?	LOF	No DNE	TRUE	COSM45940	Implied	1
Intron 4	c.376-1G>A	splicing	p.?	LOF	No DNE	consensus SA	COSM6900	Implied	7
Intron 4	c.376-1G>C	splicing	p.?	LOF	No DNE	consensus SA	COSM45809	Implied	2
Intron 4	c.376-2A>C	splicing	p.?	LOF	No DNE	consensus SA	COSM46049	Implied	1
Intron 4	c.376-2A>G	splicing	p.?	LOF	No DNE	consensus SA	COSM45672	Implied	3
Intron 4	c.376-2delA	splicing	p.?	LOF	No DNE	consensus SA	COSM45658	Implied	1
Exon 5	c.?(556del18)	in-frame deletion	p.D186del6	Unknown	Unknown	TRUE	NA	NA	1

Exon 5	c.375_376ins18	in-frame insertion	p.Y126ins6	Unknown	Unknown	TRUE	NO-ID	NA	1
Exon 5	c.376_389del	frameshift	p.Y126fs	LOF	No DNE	TRUE	COSM45206	Implied	1
Exon 5	c.376_396del	in-frame deletion	p.Y126del7	Unknown	Unknown	TRUE	COSM44405	NA	2
Exon 5	c.376T>A	missense	p.Y126N	Unknown	Unknown	TRUE	COSM44380	NA	3
Exon 5	c.376T>G	missense	p.Y126D	LOF	No DNE	TRUE	COSM43900	(94)	1
Exon 5	c.377A>G	missense	p.Y126C	LOF	DNE	TRUE	COSM11517	(95)	2
Exon 5	c.378C>G	nonsense	p.Y126X	LOF	No DNE	TRUE	COSM10862	Implied	1
Exon 5	c.392A>T	missense	p.N131I	Unknown	Unknown	cryptic SA	COSM44794	NA	1
Exon 5	c.393_394ins2	frameshift	p.K132fs	LOF	No DNE	cryptic SA	NO-ID	Implied	1
Exon 5	c.393_395del	in-frame deletion	p.N131del	Unknown	Unknown	cryptic SA	COSM44212	NA	1
Exon 5	c.394A>G	missense	p.K132E	LOF	DNE	cryptic SA	COSM10813	(98)	8
Exon 5	c.395A>C	missense	p.K132T	Unknown	Unknown	cryptic SA	COSM43912	NA	1
Exon 5	c.395A>G	missense	p.K132R	LOF	DNE	cryptic SA	COSM11582	(94)	2
Exon 5	c.396G>C	missense	p.K132N	LOF	DNE	cryptic SA	COSM43963	(94)	10
Exon 5	c.396G>T	missense	p.K132N	LOF	DNE	cryptic SA	COSM10991	(94)	1
Exon 5	c.556G>C	missense	p.D186H	Unknown	Unknown	TRUE	COSM44837	NA	2
Exon 5	c.556G>T	missense	p.D186Y	Unknown	Unknown	TRUE	NO-ID	NA	1
Exon 5	c.559del	frameshift	p.G187fs	LOF	No DNE	TRUE	NO-ID	Implied	1
Exon 5	c.559G>A	missense	p.G187S	Unknown	Unknown	TRUE	COSM44026	NA	1
Exon 5	c.559G>T	missense	p.G187C	Unknown	Unknown	TRUE	COSM45275	NA	2
Exon 5	c.560G>A	missense	p.G187D	Unknown	Unknown	TRUE	COSM44023	NA	1
Exon 5	Unknown	frameshift	p.N131fs	LOF	No DNE	cryptic SA	NA	Implied	1
Exon 5	Unknown	in-frame deletion	p.N131del	Unknown	Unknown	cryptic SA	NA	NA	2
Exon 5	Unknown	frameshift	p.K132fs	LOF	No DNE	cryptic SA	NA	Implied	1
Exon 5	Unknown	missense	p.K132N	LOF	DNE	cryptic SA	COSM10827	(94)	1
Exon 5	Unknown	missense	p.Y126C	LOF	DNE	TRUE	NA	(95)	2

Exon 5	Unknown	missense	p.Y126D	LOF	No DNE	TRUE	NA	(94)	2
Exon 5	Unknown	missense	p.Y126C	LOF	DNE	TRUE	NA	(95)	2
Exon 5	Unknown	nonsense	p.Y126X	LOF	No DNE	TRUE	NA	Implied	1
Exon 5	Unknown	splicing	p.?	LOF	No DNE	TRUE	NA	(80)	1
Intron 5	c.559+1G>A	splicing	p.?	LOF	No DNE	consensus SD	COSM6901	Implied	4
Intron 5	c.559+1G>C	splicing	p.?	LOF	No DNE	consensus SD	COSM33648	Implied	1
Intron 5	c.559+1G>T	splicing	p.?	LOF	No DNE	consensus SD	COSM44268	Implied	1
Intron 5	c.560-1G>A	splicing	p.?	LOF	No DNE	consensus SA	COSM43753	Implied	1
Intron 5	c.560-1G>C	splicing	p.?	LOF	No DNE	consensus SA	COSM43872	Implied	2
Intron 5	c.560-2A>C	splicing	p.?	LOF	No DNE	consensus SA	COSM45031	Implied	2
Intron 5	c.560-2A>T	splicing	p.?	LOF	No DNE	consensus SA	COSM45026	Implied	1
Intron 5	c.560-3T>G	splicing	p.?	LOF	No DNE	consensus SA	COSM46059	Implied	1
Exon 6	c.670G>T	nonsense	p.E224X	LOF	No DNE	TRUE	COSM44922	Implied	1
Exon 6	c.672G>C	missense	p.E224D	Unknown	Unknown	TRUE	COSM44945	NA	1
Exon 6	c.672G>T	missense	p.E224D	Unknown	Unknown	TRUE	COSM11451	NA	1
Exon 6	Unknown	splicing	p.?	LOF	No DNE	TRUE	NA	(80)	1
Intron 6	c.672+1G>C	splicing	p.?	LOF	No DNE	consensus SD	COSM45367	Implied	1
Intron 6	c.672+1G>T	splicing	p.?	LOF	No DNE	consensus SD	COSM13586	Implied	3
Intron 6	c.672+2T>A	splicing	p.?	LOF	No DNE	consensus SD	COSM165066	Implied	1
Intron 6	c.672+2T>G	splicing	p.?	LOF	No DNE	consensus SD	COSM6907	Implied	2
Intron 6	c.673-1G>A	splicing	p.?	LOF	No DNE	consensus SA	COSM43751	Implied	2
Intron 6	c.673-1G>C	splicing	p.?	LOF	No DNE	TRUE	COSM45675	Implied	2
Intron 6	c.673-2A>C	splicing	p.?	LOF	No DNE	consensus SA	COSM29760	Implied	1
Intron 6	c.673-2A>G	splicing	p.?	LOF	No DNE	consensus SA	COSM6908	Implied	8
Intron 6	Unknown	splicing	p.?	LOF	No DNE	consensus SA	NA	Implied	1
Intron 6	Unknown	splicing	p.?	LOF	No DNE	consensus SA	NA	(84)	1
Exon 7	c.768_769ins1	frameshift	p.T256fs	LOF	No DNE	ALTERNATIVE	COSM45238	Implied	1
Exon 7	c.770T>A	missense	p.L257Q	LOF	DNE	ALTERNATIVE	COSM43530	(94)	5
Exon 7	c.770T>C	missense	p.L257P	LOF	DNE	ALTERNATIVE	COSM43842	(94)	3
Exon 7	c.772del	frameshift	p.E258fs	LOF	No DNE	ALTERNATIVE	COSM45001	Implied	1

Exon 7	c.772G>A	missense	p.E258K	GOF	DNE	ALTERNATIVE	COSM10988	(99)	1
Exon 7	c.772G>C	missense	p.E258Q	Unknown	Unknown	ALTERNATIVE	COSM10751	NA	2
Exon 7	c.772G>T	nonsense	p.E258X	LOF	No DNE	ALTERNATIVE	COSM43568	Implied	4
Exon 7	c.773A>G	missense	p.E258G	LOF	Unknown	ALTERNATIVE	COSM44168	(94)	2
Exon 7	c.781_782insA	frameshift	p.S261fs	LOF	No DNE	TRUE	NO-ID	Implied	1
Exon 7	c.783_806del	in-frame deletion	p.S261del8	Unknown	Unknown	TRUE	COSM45574	NA	1
Exon 7	Unknown	nonsense	p.E258X	LOF	No DNE	ALTERNATIVE	COSM43568	Implied	1
Exon 7	Unknown	splicing	p.?	LOF	No DNE	TRUE	NA	(80)	1
Intron 7	c.?(i7-intron_G>A)	splicing	p.?	LOF	No DNE	TRUE	NA	Implied	1
Intron 7	c.782+1G>A	splicing	p.?	LOF	No DNE	consensus SD	COSM98254	Implied	3
Intron 7	c.782+1G>T	splicing	p.?	LOF	No DNE	TRUE	COSM44640	Implied	2
Intron 7	c.783-1G>T	splicing	p.?	LOF	No DNE	consensus SA	COSM6913	Implied	1
Intron 7	Unknown	splicing	p.?	LOF	No DNE	TRUE	NA	(81)	1
Exon 8	c.784_786del	in-frame deletion	p.G262del3	Unknown	Unknown	TRUE	COSM45437	NA	1
Exon 8	c.784_797del	frameshift	p.G262fs	LOF	No DNE	TRUE	NO-ID	Implied	1
Exon 8	c.785G>T	missense	p.G262V	Unknown	Unknown	TRUE	COSM11198	NA	2
Exon 8	c.805_841del	frameshift	p.S269fs	LOF	No DNE	cryptic SA	NO-ID	Implied	1
Exon 8	c.805_851del	frameshift	p.S269fs	LOF	No DNE	cryptic SA	NO-ID	Implied	1
Exon 8	c.916C>T	nonsense	p.R306X	LOF	No DNE	TRUE	COSM10663	Implied	26
Exon 8	c.916del	frameshift	p.R306fs	LOF	No DNE	TRUE	NO-ID	Implied	1
Exon 8	Unknown	frameshift	p.G262fs	LOF	No DNE	TRUE	NA	Implied	1
Exon 8	Unknown	in-frame deletion	p.G262_N263del	Unknown	Unknown	TRUE	NA	NA	1
Exon 8	Unknown	nonsense	p.R306X	LOF	No DNE	TRUE	NA	Implied	1
Intron 8	c.919+1G>A	splicing	p.?	LOF	No DNE	consensus SD	COSM97868	Implied	1
Intron 8	c.919+1G>T	splicing	p.?	LOF	No DNE	consensus SA	COSM13584	Implied	3
Intron 8	c.920-1_920GCdelinsTT	splicing	p.?	LOF	No DNE	consensus SA	NO-ID	Implied	1
Intron 8	c.920-1G>C	splicing	p.?	LOF	No DNE	consensus SA	COSM5947890	Implied	1

Intron 8	c.920-1G>T	splicing	p.?	LOF	No DNE	consensus SA	COSM95740	Implied	3
Intron 8	c.920-2A>G	splicing	p.?	LOF	No DNE	TRUE	COSM33650	Implied	4
Exon 9	c.923_?del	in-frame deletion	p.?	LOF	No DNE	TRUE	NA	Implied	1
Exon 9	c.991C>T	nonsense	p.Q331X	LOF	No DNE	TRUE	COSM11354	Implied	11
Exon 9	Unknown	frameshift	p.P322fs	LOF	No DNE	ALTERNATIVE	NA	Implied	1
Exon 9	Unknown	frameshift	p.Q331fs	LOF	No DNE	TRUE	NA	Implied	1
Exon 9	Unknown	nonsense	p.Q331X	LOF	No DNE	TRUE	NA	Implied	1
Intron 9	c.993+1G>A	splicing	p.?	LOF	No DNE	consensus SD	COSM44295	Implied	4
Intron 9	c.993+1G>C	splicing	p.?	LOF	No DNE	consensus SD	COSM119021	Implied	1
Intron 9	c.993+1G>T	splicing	p.?	LOF	No DNE	consensus SD	COSM6918	Implied	3
Intron 9	c.993+2T>A	splicing	p.?	LOF	No DNE	consensus SD	COSM707067	Implied	1
Intron 9	c.994-1G>A	splicing	p.?	LOF	No DNE	consensus SD	COSM69404	Implied	1
Exon 10	c.1023_1024delinsTT	nonsense	p.R342X	LOF	No DNE	FALSE	COSM44906	Implied	1
Exon 10	c.993_994ins14	frameshift	p.Q331fs	LOF	No DNE	TRUE	NO-ID	Implied	1
Exon 10	Unknown	splicing	p.?	LOF	No DNE	TRUE	NA	(80)	2
Intron 10	c.1101-1G>A	splicing	p.?	LOF	No DNE	consensus SA	COSM6967285	Implied	1
Unknown	Unknown	splicing	p.?	LOF	No DNE	TRUE	NA	(81)	4
Unknown	Unknown	splicing	p.?	LOF	No DNE	TRUE	NA	(84)	7
Unknown	Unknown	splicing	p.?	LOF	No DNE	TRUE	NA	(86)	4
Unknown	Unknown	splicing	p.?	LOF	No DNE	TRUE	NA	(87)	2
Unknown	Unknown	splicing	p.?	LOF	No DNE	consensus SD	NA	(90)	1
Unknown	Unknown	splicing	p.?	LOF	No DNE	TRUE	NA	(90)	1

Ref ID, study supplemental reference; DNE, dominant negative activity; GOF, gain of function mutation; LOF, loss of function mutation; DNE+, dominant negative activity present; DNE-, no dominant negative activity;