

Additional File 3: Table. Master list of TP53 coding mutations in study and classifications

Exon	Codon	Nucleotide change description(s)	Effect	Protein	Function	DNE	COSMIC ID(s)	Ref ID	Total count
2	20	Unknown	frameshift	p.S20fs	LOF	No DNE	NA	Implied	1
3	26	c.75_96del	frameshift	p.L26fs	LOF	No DNE	COSM45386	Implied	1
3	27	Unknown	missense	p.P27L	Unknown	Unknown	NA	NA	1
3	27	Unknown	missense	p.P27S	Unknown	Unknown	NA	NA	1
3	29	c.85ins1	frameshift	p.N29fs	LOF	No DNE	NO-ID	Implied	1
4	33	Unknown	nonsense	p.E33X	LOF	No DNE	NA	Implied	1
4	34	Unknown	frameshift	p.P34fs	LOF	No DNE	NA	Implied	1
4	35	Unknown	frameshift	p.L35fs	LOF	No DNE	NA	Implied	1
4	37	c.109del	frameshift	p.S37fs	LOF	No DNE	NO-ID	Implied	1
4	38	c.112C>T	nonsense	p.Q38X	LOF	No DNE	COSM46286	Implied	1
4	40	c.117_118ins1	frameshift	p.M40fs	LOF	No DNE	NO-ID	Implied	2
4	44	c.130del, Unknown	frameshift	p.M44fs	LOF	No DNE	COSM45151	Implied	2
4	46	c.136_153del	in-frame insertion	p.S46ins6	Unknown	Unknown	NO-ID	NA	1
4	47	c.139_142del, c.139del	frameshift	p.P47fs	LOF	No DNE	NO-ID, NO-ID	Implied	2
4	49	Unknown	frameshift	p.D49fs	LOF	No DNE	NA	NA	1
4	50	c.147_148ins1, Unknown	frameshift	p.I50fs	LOF	No DNE	NO-ID	Implied	2
4	50	Unknown	frameshift	p.I50X	LOF	No DNE	NA	Implied	1
4	51	Unknown	in-frame deletion	p.E51_Q52del	Unknown	Unknown	NA	NA	1
4	51	Unknown	frameshift	p.E51fs	LOF	No DNE	NA	Implied	1
4	51	Unknown	nonsense	p.E51X	LOF	No DNE	NA	Implied	3
4	52	Unknown	frameshift	p.Q52fs	LOF	No DNE	NA	Implied	1
4	53	c.156del	frameshift	p.W53fs	LOF	No DNE	COSM6806434	Implied	1
4	53	c.158G>A, c.159G>A, Unknown	nonsense	p.W53X	LOF	No DNE	COSM44760, COSM44488	Implied	3

4	54	c.159_160ins1, c.160del, Unknown	frameshift	p.F54fs	LOF	No DNE	COSM46027, NO-ID	Implied	5
4	56	c.166G>T	nonsense	p.E56X	LOF	No DNE	COSM12168	Implied	2
4	62	c.184_203del	frameshift	p.E62fs	LOF	No DNE	NO-ID	Implied	1
4	62	c.184G>T, Unknown	nonsense	p.E62X	LOF	No DNE	COSM10827	Implied	3
4	67	Unknown	frameshift	p.P67fs	LOF	No DNE	NA	Implied	1
4	68	c.201_202ins1, c.202del	frameshift	p.E68fs	LOF	No DNE	COSM45214, COSM45213	Implied	3
4	69	Unknown	frameshift	p.A69fs	LOF	No DNE	NA	Implied	1
4	73	c.216_217ins1, Unknown	frameshift	p.V73fs	LOF	No DNE	COSM44466	Implied	2
4	74	Unknown	frameshift	p.A74fs	LOF	No DNE	NA	Implied	1
4	75	Unknown	frameshift	p.P75fs	LOF	No DNE	NA	Implied	1
4	77	c.229C>G	missense	p.P77A	Unknown	Unknown	COSM44287	NA	1
4	77	Unknown	frameshift	p.P77fs	LOF	No DNE	NA	Implied	1
4	78	c.232_244del	frameshift	p.A78fs	LOF	No DNE	NO-ID	Implied	1
4	80	c.238del	frameshift	p.P80fs	LOF	No DNE	NO-ID	Implied	1
4	81	c.241del, Unknown	frameshift	p.T81fs	LOF	No DNE	NO-ID	Implied	2
4	83	c.247del, c.249_250ins4, Unknown	frameshift	p.A83fs	LOF	No DNE	COSM6911698, NO-ID	Implied	4
4	84	Unknown	in-frame deletion	p.A84_S90del	Unknown	Unknown	NA	NA	1
4	86	Unknown	frameshift	p.A86fs	LOF	No DNE	NA	Implied	1
4	89	c.265ins11, c.267del	frameshift	p.P89fs	LOF	No DNE	NO-ID, NO-ID	Implied	2
4	90	Unknown	frameshift	p.S90fs	LOF	No DNE	NA	Implied	3
4	91	c.273G>A, Unknown	nonsense	p.W91X	LOF	No DNE	COSM44492	Implied	7
4	92	Unknown	frameshift	p.P92fs	LOF	No DNE	NA	NA	1
4	93	Unknown	frameshift	p.L93fs	LOF	No DNE	NA	Implied	2
4	94	c.281C>A	nonsense	p.S94X	LOF	No DNE	COSM45500	Implied	2
4	96	c.285_286ins1	frameshift	p.S96fs	LOF	No DNE	COSM45212	Implied	1

4	99	c.295del	frameshift	p.S99fs	LOF	No DNE	COSM44688	Implied	1
4	101	c.301del	frameshift	p.K101fs	LOF	No DNE	NO-ID	Implied	1
4	102	c.304del, Unknown	frameshift	p.T102fs	LOF	No DNE	COSM45766	Implied	4
4	103	c.307_325del	frameshift	p.Y103fs	LOF	No DNE	NO-ID	Implied	1
4	104	c.310_321delins2	frameshift	p.Q104fs	LOF	No DNE	NO-ID	Implied	1
4	104	c.310C>T	nonsense	p.Q104X	LOF	No DNE	COSM10886	Implied	3
4	105	c.315_320del	frameshift	p.G105fs	LOF	No DNE	NO-ID	Implied	1
4	106	c.316_321delins4, c.317_326del, Unknown	frameshift	p.S106fs	LOF	No DNE	NO-ID, NO-ID	Implied	4
4	107	c.319_320del, c.319_329del	frameshift	p.Y107fs	LOF	No DNE	NO-ID, NO-ID	Implied	2
4	107	c.319T>G	missense	p.Y107D	Unknown	Unknown	COSM46103	NA	3
4	107	c.320A>G	missense	p.Y107C	Unknown	Unknown	COSM44994	NA	1
4	107	c.321C>G, c.321C>A, Unknown	nonsense	p.Y107X	LOF	No DNE	COSM11448, COSM45040	Implied	8
4	107	Unknown	in-frame deletion	p.Y107_F109del	Unknown	Unknown	NA	NA	1
4	108	c.322_323ins1, c.322_326del, Unknown	frameshift	p.G108fs	LOF	No DNE	NO-ID, NO-ID	Implied	9
4	109	c.325T>G	missense	p.F109V	Unknown	Unknown	COSM48817	NA	1
4	109	c.326T>C	missense	P.F109S	Unknown	Unknown	COSM45169	NA	2
4	109	Unknown	in-frame deletion	p.F109_R110del	Unknown	Unknown	NA	NA	1
4	110	c.327_328ins6	in-frame insertion	p.R110ins2	Unknown	Unknown	COSM45158	NA	1
4	110	c.328C>T	missense	p.R110C	Unknown	Unknown	COSM43682	NA	1
4	110	c.328del, Unknown	frameshift	p.R110fs	LOF	No DNE	COSM44669	Implied	12
4	110	c.329G>A	missense	p.R110H	Unknown	Unknown	COSM46115	NA	4
4	110	c.329G>C	missense	p.R110P	LOF	Unknown	COSM11250	(94)	7
4	110	c.329G>T	missense	p.R110L	GOF	No DNE	COSM10716	(94)	1

4	110	Unknown	in-frame deletion	p.R110del	Unknown	Unknown	NA	NA	1
4	111	c.332T>A	missense	p.L111Q	LOF	Unknown	COSM44630	(94)	2
4	111	c.332T>C	missense	p.L111P	LOF	Unknown	COSM44045	(94)	8
4	111	Unknown	frameshift	p.L111fs	LOF	No DNE	NA	Implied	3
4	113	c.337T>G	missense	p.F113V	LOF	Unknown	COSM11498	(94)	2
4	113	c.338T>G	missense	p.F113C	LOF	Unknown	COSM10717	(94)	2
4	113	Unknown	in-frame deletion	p.F113_H115del	Unknown	Unknown	NA	NA	1
4	113	Unknown	frameshift	p.F113fs	LOF	No DNE	NA	Implied	1
4	114	c.341_354del	frameshift	p.L114fs	LOF	No DNE	COSM45221	Implied	1
4	115	Unknown	frameshift	p.H115fs	LOF	No DNE	NA	Implied	2
4	116	c.?(346del12)	in-frame deletion	p.S116del4	Unknown	Unknown	NA	NA	1
4	117	c.349del	frameshift	p.G117fs	LOF	No DNE	NO-ID	Implied	1
4	118	c.354_355ins1, Unknown	frameshift	p.T118fs	LOF	No DNE	NO-ID	Implied	2
4	120	c.358A>G	missense	p.K120E	LOF	Unknown	COSM44827	(94)	2
4	121	c.362_363del, c.363_369del	frameshift	p.S121fs	LOF	No DNE	COSM45223, NO-ID	Implied	2
4	122	c.364_378delins1, Unknown	frameshift	p.V122fs	LOF	No DNE	NO-ID	Implied	3
4	124	c.369_370ins1, Unknown	frameshift	p.C124fs	LOF	No DNE	NO-ID	Implied	3
4	124	c.372C>A, Unknown	nonsense	p.C124X	LOF	No DNE	COSM326716	Implied	3
4	125	c.374?del, c.374_375ins1, Unknown	frameshift	p.T125fs	LOF	No DNE	NO-ID, NO-ID	Implied	3
4	125	c.374C>A	missense	p.T125K	LOF	Unknown	COSM44073	(94)	2
5	126	c.375_376ins18	in-frame insertion	p.Y126ins6	Unknown	Unknown	NO-ID	NA	1
5	126	c.376_389del	frameshift	p.Y126fs	LOF	No DNE	COSM45206	Implied	1
5	126	c.376_396del	in-frame deletion	p.Y126del7	Unknown	Unknown	COSM44405	NA	2

5	126	c.376T>A	missense	p.Y126N	Unknown	Unknown	COSM44380	NA	3
5	126	c.376T>G, Unknown	missense	p.Y126D	LOF	No DNE	COSM43900	(94)	3
5	126	c.377A>G, Unknown	missense	p.Y126C	LOF	DNE	COSM11517	(95)	4
5	126	c.378C>G, Unknown	nonsense	p.Y126X	LOF	No DNE	COSM10862	Implied	2
5	127	c.379T>A	missense	p.S127T	Unknown	Unknown	COSM53285	NA	1
5	127	c.379T>C	missense	p.S127P	LOF	DNE	COSM44687	(94)	4
5	127	c.380C>T	missense	p.S127F	LOF	Unknown	COSM44226	(94)	6
5	128	c.382C>T	missense	p.P128S	Unknown	Unknown	COSM44397	NA	1
5	128	c.383del	frameshift	p.P128fs	LOF	No DNE	COSM44396	Implied	2
5	130	c.388C>G	missense	p.L130V	LOF	Unknown	COSM11462	(96)	2
5	130	c.388C>T	missense	p.L130F	LOF	Unknown	COSM11449	(97)	1
5	130	c.389T>G	missense	p.L130R	Unknown	Unknown	COSM44063	NA	1
5	131	c.392A>T	missense	p.N131I	Unknown	Unknown	COSM44794	NA	1
5	131	c.392del, Unknown	frameshift	p.N131fs	LOF	No DNE	COSM45882	Implied	2
5	131	c.393_395del, Unknown	in-frame deletion	p.N131del	Unknown	Unknown	COSM44212	NA	3
5	132	c.393_394ins2, Unknown	frameshift	p.K132fs	LOF	No DNE	NO-ID	Implied	2
5	132	c.394A>G	missense	p.K132E	LOF	DNE	COSM10813	(94,98)	8
5	132	c.395A>C	missense	p.K132T	Unknown	Unknown	COSM43912	NA	1
5	132	c.395A>G	missense	p.K132R	LOF	DNE	COSM11582	(94)	2
5	132	c.396G>C, c.396G>T, Unknown	missense	p.K132N	LOF	DNE	COSM43963, COSM10991	(94)	12
5	133	c.?(397del19), c.397del, c.398_399delins1, Unknown	frameshift	p.M133fs	LOF	No DNE	NO-ID, COSM45586, NO-ID	Implied	4
5	133	c.397A>G	missense	p.M133V	Unknown	Unknown	COSM43950	NA	2
5	133	c.398T>A	missense	p.M133K	Unknown	Unknown	COSM11781	(94)	3
5	133	c.398T>G	missense	p.M133R	LOF	No DNE	COSM43730	(94)	1

5	133	Unknown	in-frame deletion	p.M133_139del	Unknown	Unknown	COSM43775	NA	1
5	134	c.400T>C, c.402T>A	missense	p.F134L	LOF	Unknown	COSM44654, COSM45167	(94)	5
5	134	c.400T>G	missense	p.F134V	Unknown	Unknown	COSM43941	(94)	1
5	134	c.401T>G	missense	p.F134C	Unknown	Unknown	COSM43949	(94)	5
5	134	Unknown	missense	p.F134I	LOF	Unknown	NA	(94,99)	1
5	135	c.402_403ins2, c.403dup, c.404del, Unknown	frameshift	p.C135fs	LOF	No DNE	COSM45148, COSM6960879, COSM5369713	Implied	5
5	135	c.403T>C	missense	p.C135R	LOF	Unknown	COSM10684	(94)	3
5	135	c.403T>G	missense	p.C135G	LOF	Unknown	COSM44829	(94)	1
5	135	c.404G>A	missense	p.C135Y	LOF	DNE	COSM10801	(94)	10
5	135	c.404G>T	missense	p.C135F	LOF	DNE	COSM10647	(94)	5
5	135	c.405C>A	nonsense	p.C135X	LOF	No DNE	COSM44319	Implied	1
5	135	c.405C>G, Unknown	missense	p.C135W	LOF	No DNE	COSM44219	(94)	6
5	136	c.406_421del, c.406_428del, c.407del, c.409_438del	frameshift	p.Q136fs	LOF	No DNE	NO-ID, COSM6906745, NO-ID, COSM46236	Implied	4
5	136	c.406C>G	missense	p.Q136E	Unknown	Unknown	COSM43767	(94)	2
5	136	c.406C>T, Unknown	nonsense	p.Q136X	LOF	No DNE	COSM11166	Implied	7
5	136	c.407A>C	missense	p.Q136P	Unknown	Unknown	COSM45089	NA	1
5	136	Unknown	missense	p.Q136H	Unknown	Unknown	NA	NA	1
5	138	c.412_428del, Unknown	frameshift	p.A138fs	LOF	No DNE	NO-ID	Implied	4
5	138	c.412G>A	missense	p.A138T	Unknown	Unknown	COSM44821	NA	2
5	138	c.412G>C	missense	p.A138P	LOF	DNE	COSM11188	(94,100)	3
5	138	c.413C>T	missense	p.A138V	LOF	No DNE	COSM43818	(94,101)	6
5	139	c.?(415del9)	in-frame deletion	p.K139_136del	Unknown	Unknown	NA	NA	1
5	139	c.414_425del, Unknown	in-frame deletion	p.K139_P142del	Unknown	Unknown	NO-ID	NA	2

5	139	c.415A>C	missense	p.K139Q	Unknown	Unknown	COSM45846	NA	1
5	139	c.415A>T	nonsense	p.K139X	LOF	No DNE	COSM44678	Implied	2
5	139	c.417G>C, c.417G>T	missense	p.K139N	Unknown	Unknown	COSM44101, COSM44220	NA	2
5	139	Unknown	frameshift	p.K139fs	LOF	No DNE	NA	Implied	1
5	140	c.418_427del, c.418del	frameshift	p.T140fs	LOF	No DNE	NO-ID, COSM44596	Implied	2
5	141	c.420_421ins2, c.420del, c.421_428del, Unknown	frameshift	p.C141fs	LOF	No DNE	COSM45165, COSM69019, COSM45463	Implied	4
5	141	c.421T>C	missense	p.C141R	LOF	Unknown	COSM43901	(94)	3
5	141	c.422G>A	missense	p.C141Y	LOF	DNE	COSM43708	(102)	16
5	141	c.422G>T	missense	p.C141F	Unknown	Unknown	COSM44911	NA	1
5	141	c.423_430del	nonsense	p.C141X	LOF	No DNE	NO-ID	Implied	1
5	142	c.423_425del	in-frame deletion	p.P142del	Unknown	Unknown	COSM44824	NA	1
5	142	c.424del	frameshift	p.P142fs	LOF	No DNE	NO-ID	Implied	1
5	143	c.427del, Unknown	frameshift	p.V143fs	LOF	No DNE	COSM45108	Implied	2
5	143	c.427G>A	missense	p.V143M	Unknown	Unknown	COSM43878	NA	2
5	143	c.428T>C	missense	p.V143A	GOF	DNE	COSM11306	(103,104)	2
5	144	c.430_432del	in-frame deletion	p.Q144del	Unknown	Unknown	NO-ID	NA	2
5	144	c.430C>T, Unknown	nonsense	p.Q144X	LOF	No DNE	COSM11245	Implied	8
5	144	c.431A>C	missense	p.Q144P	LOF	Unknown	COSM44205	(94)	1
5	144	c.432G>T	missense	p.Q144H	Unknown	Unknown	COSM45961	NA	1
5	145	c.433_439del	frameshift	p.L145fs	LOF	No DNE	COSM45790	Implied	1
5	145	c.434T>A	missense	p.L145Q	Unknown	Unknown	COSM44137	NA	3
5	145	c.434T>C	missense	p.L145P	LOF	Unknown	COSM43899	(94)	2
5	145	c.434T>G	missense	p.L145R	LOF	Unknown	COSM45351	(94)	2
5	146	c.437_438delins1	frameshift	p.W146fs	LOF	No DNE	COSM45791	Implied	1

5	146	c.437G>A, c.438G>A	nonsense	p.W146X	LOF	No DNE	COSM43609, COSM10727	Implied	7
5	147	c.439del	frameshift	p.V147fs	LOF	No DNE	COSM44698	Implied	2
5	147	c.440T>A	missense	p.V147D	Unknown	Unknown	COSM11273	NA	1
5	148	c.444_445ins1, c.445_449del, c.445_457del	frameshift	p.D148fs	LOF	No DNE	COSM44210, NO-ID, COSM45210	Implied	4
5	148	c.445_477del	in-frame deletion	p.D148del11	Unknown	Unknown	NO-ID	NA	1
5	149	Unknown	frameshift	p.S149fs	LOF	No DNE	NA	Implied	3
5	150	c.448_466del	frameshift	p.T150fs	LOF	No DNE	NO-ID	Implied	1
5	151	c.451C>A	missense	p.P151T	LOF	No DNE	COSM43911	(94)	1
5	151	c.451C>G	missense	p.P151A	Unknown	Unknown	COSM44944	NA	5
5	151	c.451C>T, Unknown	missense	p.P151S	GOF	DNE	COSM10905	(105)	15
5	151	c.451del, c.453_454ins1	frameshift	p.P151fs	LOF	No DNE	NO-ID, COSM43894	Implied	2
5	151	c.452C>A	missense	p.P151H	GOF	DNE	COSM11476	(94,106)	3
5	151	c.452C>G	missense	p.P151R	LOF	DNE	COSM44003	(94)	1
5	152	c.454_466del, c.454del, c.455del, Unknown	frameshift	p.P152fs	LOF	No DNE	COSM44983, COSM45890, COSM43792	Implied	5
5	152	c.454C>T	missense	p.P152S	Unknown	Unknown	COSM43582	NA	1
5	152	c.455C>T	missense	p.P152L	GOF	No DNE	COSM10790	(94,106)	1
5	153	Unknown	frameshift	p.P153fs	LOF	No DNE	NA	Implied	2
5	154	c.460G>A	missense	p.G154S	Unknown	Unknown	COSM43692	NA	1
5	154	c.461G>T	missense	p.G154V	LOF	Unknown	COSM6815	(94)	1
5	154	Unknown	frameshift	p.G154fs	LOF	No DNE	NA	Implied	1
5	155	c.463_470del, Unknown	frameshift	p.T155fs	LOF	No DNE	COSM44009	Implied	2
5	155	c.463A>C	missense	p.T155P	Unknown	Unknown	COSM10912	NA	2
5	155	c.464C>A, Unknown	missense	p.T155N	LOF	DNE	COSM11218	(94)	2

5	155	c.464C>T	missense	p.T155I	LOF	DNE	COSM44033	(97)	1
5	155	Unknown	in-frame deletion	p.T155_163del	LOF	No DNE	NA	Implied	1
5	156	c.466_472del	frameshift	p.R156fs	LOF	No DNE	COSM44479	NA	1
5	156	c.466C>G	missense	p.R156G	Unknown	Unknown	COSM45154	NA	2
5	156	c.467G>A	missense	p.R156H	LOF	DNE	COSM43739	(94,107)	3
5	156	c.467G>C	missense	p.R156P	GOF	DNE	COSM10760	(94)	2
5	156	Unknown	in-frame deletion	p.R156del	Unknown	Unknown	NA	NA	1
5	157	c.468_469ins1, c.468_469ins4, c.468del, c.469_473del, Unknown	frameshift	p.V157fs	LOF	No DNE	COSM45054, COSM45217, COSM43710, COSM45111	Implied	5
5	157	c.469G>C	missense	p.V157L	Unknown	Unknown	COSM45120	NA	1
5	157	c.469G>T	missense	p.V157F	GOF	DNE	COSM10670	(94,108)	12
5	157	c.470T>A	missense	p.V157D	LOF	Unknown	COSM44329	(94)	2
5	157	c.470T>G	missense	p.V157G	Unknown	Unknown	COSM43903	NA	1
5	157	Unknown	in-frame deletion	p.V157_R158del	Unknown	Unknown	NA	NA	1
5	158	c.472C>G	missense	p.R158G	LOF	No DNE	COSM11087	(94)	3
5	158	c.472C>T	missense	p.R158C	Unknown	Unknown	COSM43848	(94,109)	1
5	158	c.473G>A	missense	p.R158H	GOF	No DNE	COSM10690	(94)	2
5	158	c.473G>C	missense	p.R158P	LOF	DNE	COSM43615	(94)	1
5	158	c.473G>T	missense	p.R158L	LOF	DNE	COSM10714	(94)	1
5	158	c.474_475ins6	in-frame insertion	p.R158ins2	Unknown	Unknown	COSM45160	NA	1
5	158	Unknown	frameshift	p.R158fs	LOF	No DNE	NA	Implied	3
5	159	c.475G>C	missense	p.A159P	LOF	Unknown	COSM43836	(94)	2
5	159	c.476C>A	missense	p.A159D	Unknown	Unknown	COSM11496	(94)	2
5	159	c.476C>T, Unknown	missense	p.A159V	LOF	Unknown	COSM11148	(94)	13
5	159	Unknown	frameshift	p.A159fs	LOF	No DNE	NA	Implied	1
5	161	c.481_484del, Unknown	frameshift	p.A161fs	LOF	No DNE	COSM46234	Implied	2

5	161	c.481G>A	missense	p.A161T	LOF	Unknown	COSM10739	(94,110)	3
5	161	c.482C>A	missense	p.A161D	LOF	No DNE	COSM11323	(107)	3
5	161	c.483_485del	in-frame deletion	p.A161del	Unknown	Unknown	NO-ID	NA	1
5	162	c.485del, Unknown	frameshift	p.I162fs	LOF	No DNE	COSM5195325	Implied	3
5	162	c.485T>A, Unknown	missense	p.I162N	LOF	No DNE	COSM11966	(94)	2
5	162	c.485T>C	missense	p.I162T	Unknown	Unknown	NO-ID	NA	1
5	162	c.487_489del	in-frame deletion	p.I162_Y163delinsN	Unknown	Unknown	COSM4770726	NA	1
5	163	c.487del	frameshift	p.Y163fs	LOF	No DNE	COSM45194	Implied	1
5	163	c.487T>A	missense	p.Y163N	GOF	DNE	COSM44623	(94)	3
5	163	c.487T>C, Unknown	missense	p.Y163H	LOF	No DNE	COSM43846	(94)	2
5	163	c.487T>G	missense	p.Y163D	Unknown	Unknown	COSM44216	(94)	1
5	163	c.488A>G, Unknown	missense	p.Y163C	GOF	DNE	COSM10808	(94,107,111)	24
5	163	c.489C>G	nonsense	p.Y163X	LOF	No DNE	COSM43820	Implied	1
5	164	c.490_499del	frameshift	p.K164fs	LOF	No DNE	NO-ID	Implied	1
5	164	c.490A>C	missense	p.K164Q	Unknown	Unknown	COSM44521	NA	2
5	164	c.490A>G	missense	p.K164E	LOF	Unknown	COSM10762	(94)	1
5	165	c.493C>T, Unknown	nonsense	p.Q165X	LOF	No DNE	COSM43632	Implied	10
5	165	c.494_495ins1	frameshift	p.Q165fs	LOF	No DNE	COSM45697	Implied	1
5	166	c.?(496del3point)	in-frame deletion	p.S166del	Unknown	Unknown	NA	NA	1
5	166	c.497C>G	nonsense	p.S166X	LOF	No DNE	COSM44467	Implied	1
5	166	c.497C>T	missense	p.S166L	Unknown	Unknown	COSM44289	NA	1
5	166	Unknown	frameshift	p.S166fs	LOF	No DNE	NA	Implied	1
5	167	c.499C>G	missense	p.Q167E	Unknown	Unknown	COSM46066	NA	1
5	167	c.499C>T, Unknown	nonsense	p.Q167X	LOF	No DNE	COSM11333	Implied	9
5	167	c.500A>G	missense	p.Q167R	LOF	No DNE	COSM43709	(94)	1
5	167	c.500A>T	missense	p.Q167L	Unknown	Unknown	COSM45342	NA	2
5	167	c.500del, c.501del	frameshift	p.Q167fs	LOF	No DNE	COSM45226, COSM437555	Implied	2

5	168	c.502_508del, c.504del	frameshift	p.H168fs	LOF	No DNE	NO-ID, COSM46299	Implied	2
5	168	c.502C>T	missense	p.H168Y	LOF	DNE	COSM43861	(94)	1
5	168	c.503A>C, Unknown	missense	p.H168P	LOF	No DNE	COSM44808	(111)	5
5	168	c.503A>T	missense	p.H168L	Unknown	Unknown	COSM44801	NA	2
5	168	Unknown	missense	p.H168R	LOF	DNE	NA	(94)	1
5	169	c.504_505ins2	frameshift	p.M169fs	LOF	No DNE	COSM45220	Implied	1
5	169	c.505A>G	missense	p.M169V	Unknown	Unknown	COSM44431	NA	2
5	169	c.507G>A	missense	p.M169I	Unknown	Unknown	COSM44126	NA	2
5	171	c.511_514del, c.511del, c.513_514ins1, Unknown	frameshift	p.E171fs	LOF	No DNE	COSM437546, COSM46095, COSM45207	Implied	4
5	171	c.511G>T	missense	p.E171X	LOF	No DNE	COSM10996	Implied	1
5	171	c.512A>G	missense	p.E171G	Unknown	Unknown	COSM44732	NA	1
5	171	c.512A>T	missense	p.E171V	Unknown	Unknown	COSM44796	NA	1
5	172	c.514del	frameshift	p.V172fs	LOF	No DNE	COSM45906	Implied	2
5	172	c.514G>T	missense	p.V172F	LOF	No DNE	COSM44240	(94,112)	2
5	173	c.517G>A, Unknown	missense	p.V173M	LOF	DNE	COSM11084	(111)	9
5	173	c.517G>C, c.517G>T	missense	p.V173L	GOF	DNE	COSM44057, COSM43559	(111)	15
5	173	c.518T>C	missense	p.V173A	LOF	Unknown	COSM44327	(94)	1
5	173	c.518T>G	missense	p.V173G	GOF	No DNE	COSM44383	(94)	1
5	174	c.520_536del, c.520del, c.522del, Unknown	frameshift	p.R174fs	LOF	No DNE	COSM46393, COSM45316, COSM44725	Implied	5
5	174	Unknown	in-frame deletion	p.R174_E180>R	Unknown	Unknown	NA	NA	1
5	175	c.523_534delins3	in-frame deletion	p.R175del3	Unknown	Unknown	COSM45845	NA	1
5	175	c.523C>A, Unknown	missense	p.R175S	LOF	Unknown	COSM43931	(113)	5
5	175	c.523C>G, Unknown	missense	p.R175G	LOF	No DNE	COSM10870	(113)	3

5	175	c.523del, c.525_526ins1	frameshift	p.R175fs	LOF	No DNE	COSM45936, COSM45319	Implied	3
5	175	c.524G>A, Unknown	missense	p.R175H	GOF	DNE	COSM10648	(94,114)	138
5	175	c.524G>C, Unknown	missense	p.R175P	LOF	No DNE	COSM45416	(94)	2
5	175	c.524G>T	missense	p.R175L	LOF	No DNE	COSM10718	(94,115)	2
5	176	c.?(526del6)	in-frame deletion	p.C176_P177del	Unknown	Unknown	NA	NA	1
5	176	c.525_526delins2, c.526T>A, c.527G>C, Unknown	missense	p.C176S	GOF	DNE	COSM46255, COSM44146, COSM44645	(107)	7
5	176	c.526_537del	in-frame deletion	p.C176del4AA	Unknown	Unknown	NO-ID	NA	1
5	176	c.526del	frameshift	p.C176fs	LOF	No DNE	COSM44759	Implied	3
5	176	c.526T>C	missense	p.C176R	LOF	DNE	COSM44948	(94,116)	2
5	176	c.527G>A, Unknown	missense	p.C176Y	GOF	DNE	COSM10687	(94,117)	6
5	176	c.527G>T	missense	p.C176F	LOF	DNE	COSM10645	(94)	8
5	176	c.528C>A	nonsense	p.C176X	LOF	No DNE	COSM43734	Implied	1
5	176	c.528C>G	missense	p.C176W	LOF	Unknown	COSM11114	(94)	2
5	177	c.529_537del	in-frame deletion	p.P177del3	Unknown	Unknown	COSM46181	NA	1
5	177	c.529_540del	frameshift	p.P177del4	Unknown	Unknown	NO-ID	NA	1
5	177	c.530C>A, Unknown	missense	p.P177H	LOF	DNE	COSM45326	(94,118)	2
5	177	c.530C>G	missense	p.P177R	LOF	Unknown	COSM10651	NA	1
5	177	Unknown	in-frame deletion	p.P177_C182del	Unknown	Unknown	NA	NA	2
5	178	c.531_532del, c.533del, Unknown	frameshift	p.H178fs	LOF	No DNE	COSM6502246, COSM6960219	Implied	5
5	178	c.533A>C	missense	p.H178P	LOF	DNE	COSM44215	(107)	3
5	178	c.534C>A, c.534C>G, Unknown	missense	p.H178Q	Unknown	Unknown	COSM11998, COSM46163	NA	3
5	179	c.535C>A	missense	p.H179N	LOF	DNE	COSM44151	(94,119)	2
5	179	c.535C>G	missense	p.H179D	Unknown	Unknown	COSM44776	NA	3
5	179	c.535C>T, Unknown	missense	p.H179Y	GOF	DNE	COSM10768	(94,120)	9
5	179	c.536A>C	missense	p.H179P	LOF	Unknown	COSM44218	(94,119)	1

5	179	c.536A>G, Unknown	missense	p.H179R	GOF	DNE	COSM10889	(105,107,114)	23
5	179	c.536A>T	missense	p.H179L	LOF	DNE	COSM43635	(94)	4
5	179	c.537T>A, c.537T>G	missense	p.H179Q	GOF	DNE	COSM44214, COSM11249	(105)	5
5	180	c.538G>A	missense	p.E180K	LOF	DNE	COSM43772	(102)	2
5	180	c.538G>C	missense	p.E180Q	Unknown	Unknown	COSM45705	NA	1
5	180	c.538G>T, Unknown	nonsense	p.E180X	LOF	No DNE	COSM43597	Implied	2
5	180	c.539A>G	missense	p.E180G	Unknown	Unknown	COSM87457	NA	1
5	181	c.541C>G	missense	p.R181G	Unknown	DNE	COSM45475	(94)	1
5	181	c.541C>T	missense	p.R181C	GOF	DNE	COSM11090	(94)	7
5	181	c.542G>A, Unknown	missense	p.R181H	LOF	DNE	COSM10738	(94)	2
5	181	Unknown	frameshift	p.R181fs	LOF	No DNE	NA	Implied	1
5	182	c.544del	frameshift	p.C182fs	LOF	No DNE	COSM46042	Implied	1
5	182	c.544T>C	missense	p.C182R	Unknown	Unknown	COSM44563	NA	1
5	182	c.546C>A	nonsense	p.C182X	LOF	No DNE	COSM45562	Implied	1
5	183	c.548C>G	nonsense	p.S183X	LOF	No DNE	COSM10706	Implied	11
5	183	c.548C>T	missense	p.S183L	LOF	Unknown	COSM44300	(96)	1
5	185	c.553del, c.554del	frameshift	p.S185fs	LOF	No DNE	COSM45314, COSM45698	Implied	3
5	185	c.555C>G	missense	p.S185R	Unknown	Unknown	COSM45701	NA	2
5	186	c.?(556del18)	in-frame deletion	p.D186del6AA	Unknown	Unknown	NA	NA	1
5	186	c.556G>C	missense	p.D186H	Unknown	Unknown	COSM44837	NA	2
5	186	c.556G>T	missense	p.D186Y	Unknown	Unknown	NO-ID	NA	1
5	187	c.559del	frameshift	p.G187fs	LOF	No DNE	NO-ID	Implied	1
5	187	c.559G>A	missense	p.G187S	Unknown	Unknown	COSM44026	NA	1
5	187	c.559G>T	missense	p.G187C	Unknown	Unknown	COSM45275	NA	2
5	187	c.560G>A	missense	p.G187D	Unknown	Unknown	COSM44023	NA	1
6	189	c.566_574del	in-frame deletion	p.A189del3	Unknown	Unknown	COSM45231	NA	1
6	189	c.566del	frameshift	p.A189fs	LOF	No DNE	NO-ID	Implied	1

6	190	c.568C>A	missense	p.P190T	Unknown	Unknown	COSM44438	NA	1
6	190	c.569C>T	missense	p.P190L	LOF	Unknown	COSM43657	(94)	7
6	190	c.571_584del, Unknown	frameshift	p.P190fs	LOF	No DNE	NO-ID	Implied	3
6	191	c.570_571ins1, Unknown	frameshift	p.P191fs	LOF	No DNE	NO-ID	Implied	3
6	192	c.574C>T	nonsense	p.Q192X	LOF	No DNE	COSM10733	Implied	32
6	192	c.575A>G	missense	p.Q192R	Unknown	Unknown	COSM44849	NA	1
6	193	c.577C>T	missense	p.H193Y	LOF	Unknown	COSM10672	(94)	4
6	193	c.578A>C, Unknown	missense	p.H193P	LOF	Unknown	COSM43833	(94)	3
6	193	c.578A>G, Unknown	missense	p.H193R	LOF	DNE	COSM10742	(102)	25
6	193	c.578A>T	missense	p.H193L	LOF	No DNE	COSM11066	(107)	11
6	193	c.579del, Unknown	frameshift	p.H193fs	LOF	No DNE	COSM45230	Implied	2
6	194	c.580_581ins1, c.581del, Unknown	frameshift	p.L194fs	LOF	No DNE	COSM45702, COSM45205	Implied	3
6	194	c.580C>T	missense	p.L194F	GOF	DNE	COSM10995	(94,121)	4
6	194	c.581T>C	missense	p.L194P	LOF	Unknown	COSM43827	(122)	4
6	194	c.581T>G, Unknown	missense	p.L194R	GOF	No DNE	COSM44571	(94,123)	23
6	194	Unknown	in-frame insertion	p.L194_195insS IL	Unknown	Unknown	NA	NA	1
6	195	c.583A>T	missense	p.I195F	LOF	No DNE	COSM44633	(94)	2
6	195	c.584T>A	missense	p.I195N	LOF	Unknown	COSM44877	(94)	1
6	195	c.584T>C, Unknown	missense	p.I195T	LOF	No DNE	COSM11089	(94,124)	27
6	195	Unknown	frameshift	p.I195fs	LOF	No DNE	NA	(94)	3
6	196	c.586_607del	frameshift	p.R196fs	LOF	No DNE	NO-ID	Implied	1
6	196	c.586C>T, Unknown	nonsense	p.R196X	LOF	No DNE	COSM10705	Implied	34
6	196	c.587G>C	missense	p.R196P	LOF	DNE	COSM43814	(94)	5
6	197	c.589_594del	in-frame deletion	p.V197del2	Unknown	Unknown	NO-ID	NA	1
6	197	c.589G>A	missense	p.V197M	LOF	No DNE	COSM43779	(105)	2
6	197	c.589G>C	missense	p.V197L	LOF	No DNE	COSM45265	(94,125)	1

6	197	c.590del	frameshift	p.V197fs	LOF	No DNE	NO-ID	Implied	1
6	197	c.590T>G	missense	p.V197E	LOF	DNE	COSM43905	(94)	1
6	197	c.590T>G	missense	p.V197G	Unknown	Unknown	COSM43905	NA	2
6	198	c.592G>A	missense	p.E198K	Unknown	Unknown	COSM43690	NA	1
6	198	c.592G>T	nonsense	p.E198X	LOF	No DNE	COSM44241	Implied	7
6	199	c.595G>T	nonsense	p.G199X	LOF	No DNE	COSM44537	Implied	2
6	199	c.596G>A	missense	p.G199E	Unknown	Unknown	COSM43989	NA	1
6	199	c.596G>T, Unknown	missense	p.G199V	Unknown	Unknown	COSM44140	NA	5
6	200	c.600T>G	missense	p.N200K	Unknown	Unknown	NO-ID	NA	1
6	200	Unknown	frameshift	p.N200fs	LOF	No DNE	NA	Implied	2
6	201	c.601del, c.602_603ins1	frameshift	p.L201fs	LOF	No DNE	NO-ID	Implied	3
6	202	c.605G>T	missense	p.R202L	Unknown	Unknown	COSM44925	NA	1
6	203	c.608_609ins1	frameshift	p.V203fs	LOF	No DNE	COSM44982	Implied	1
6	204	c.610del, c.611_612ins1	frameshift	p.E204fs	LOF	No DNE	COSM44011, COSM44981	Implied	4
6	204	c.610G>A	missense	p.E204K	Unknown	Unknown	COSM43990	NA	1
6	204	c.610G>T, Unknown	nonsense	p.E204X	LOF	No DNE	COSM10804	Implied	8
6	205	c.613T>A	missense	p.Y205N	LOF	Unknown	COSM45685	(94)	2
6	205	c.613T>C	missense	p.Y205H	Unknown	Unknown	COSM43642	NA	1
6	205	c.613T>G	missense	p.Y205D	LOF	Unknown	COSM43844	(95)	2
6	205	c.614A>C, c.614A>G	missense	p.Y205S	Unknown	Unknown	COSM44169, COSM43947	NA	4
6	205	c.614A>G	missense	p.Y205C	LOF	No DNE	COSM43947	(95)	10
6	205	c.614A>T	missense	p.Y205F	Unknown	Unknown	COSM11351	NA	1
6	205	c.615T>G	nonsense	p.Y205X	LOF	No DNE	COSM44924	Implied	2
6	205	Unknown	in-frame deletion	p.Y205_T211del	Unknown	Unknown	NA	NA	1
6	206	c.618G>C	missense	p.L206F	Unknown	Unknown	COSM44797	NA	1
6	208	c.623A>T	missense	p.D208V	LOF	Unknown	COSM44249	(94)	3
6	208	c.624C>A	missense	p.D208E	Unknown	Unknown	COSM45707	NA	1

6	208	Unknown	missense	p.D208G	Unknown	Unknown	NA	NA	1
6	209	c.625_626del, c.626_627del, c.626del, Unknown	frameshift	p.R209fs	LOF	No DNE	NO-ID, COSM6482, COSM45438	Implied	14
6	209	c.625A>T	nonsense	p.R209X	LOF	No DNE	COSM11290	Implied	1
6	210	c.628del, Unknown	frameshift	p.N210fs	LOF	No DNE	NO-ID	Implied	2
6	210	c.629A>C	missense	p.N210T	Unknown	Unknown	COSM45699	NA	1
6	211	c.631_632del, c.631del, Unknown	frameshift	p.T211fs	LOF	No DNE	COSM45222, COSM44371	Implied	4
6	211	c.631A>G	missense	p.T211A	Unknown	Unknown	COSM44238	NA	2
6	211	c.632C>T	missense	p.T211I	LOF	Unknown	COSM43939	(95)	1
6	212	c.634_635del	frameshift	p.F212fs	LOF	No DNE	NO-ID	Implied	2
6	212	c.634T>A	missense	p.F212I	Unknown	Unknown	COSM45703	NA	1
6	212	c.634T>C	missense	p.F212L	Unknown	Unknown	COSM45477	NA	1
6	212	c.635T>C	missense	p.F212S	Unknown	Unknown	COSM46214	NA	1
6	213	c.637C>T, Unknown	nonsense	p.R213X	LOF	No DNE	COSM10654	Implied	64
6	213	c.638G>A	missense	p.R213Q	LOF	DNE	COSM10735	(107,126)	2
6	213	c.638G>T	missense	p.R213L	LOF	Unknown	COSM43650	(94)	2
6	213	Unknown	frameshift	p.R213fs	LOF	No DNE	NA	Implied	1
6	214	c.640_641del, c.642_643del, Unknown	frameshift	p.H214fs	LOF	No DNE	NO-ID, COSM46269	Implied	3
6	214	c.640C>T	missense	p.H214Y	Unknown	Unknown	COSM44112	NA	2
6	214	c.641A>G, Unknown	missense	p.H214R	GOF	No DNE	COSM43687	(94,127)	9
6	214	Unknown	in-frame deletion	p.H214_E221del	Unknown	Unknown	NA	NA	1
6	215	c.643_645dup	in-frame insertion	p.S215dup	Unknown	Unknown	NO-ID	NA	1
6	215	c.643_658del, c.644del	frameshift	p.S215fs	LOF	No DNE	NO-ID, COSM44887	Implied	3
6	215	c.643A>G, Unknown	missense	p.S215G	LOF	Unknown	COSM43951	(94,128)	2

6	215	c.644G>A	missense	p.S215N	LOF	Unknown	COSM44093	(94)	1
6	215	c.644G>T	missense	p.S215I	GOF	Unknown	COSM11450	(94)	4
6	215	c.645T>G	missense	p.S215R	Unknown	Unknown	COSM44979	NA	3
6	216	c.645_646ins2, Unknown	frameshift	p.V216fs	LOF	No DNE	COSM45216	Implied	2
6	216	c.646_648del	in-frame deletion	p.V216del	Unknown	Unknown	NO-ID	NA	1
6	216	c.646_651del	in-frame deletion	p.V216_V217del	Unknown	Unknown	NO-ID	NA	1
6	216	c.646G>A	missense	p.V216M	LOF	DNE	COSM10667	(129)	15
6	216	c.646G>T	missense	p.V216L	Unknown	Unknown	COSM11210	NA	1
6	216	c.647T>A, Unknown	missense	p.V216E	LOF	Unknown	COSM44274	(94,99)	2
6	217	c.649_662del, c.651_652ins5, Unknown	frameshift	p.V217fs	LOF	No DNE	NO-ID, NO-ID	Implied	3
6	217	c.650T>C	missense	p.V217A	Unknown	Unknown	COSM45110	NA	2
6	218	c.652_654del	in-frame deletion	p.V218del	Unknown	Unknown	COSM6496	NA	3
6	218	c.652del, Unknown	frameshift	p.V218fs	LOF	No DNE	COSM1324795	Implied	2
6	218	c.652G>A	missense	p.V218M	Unknown	Unknown	COSM44683	NA	1
6	218	c.652G>T	missense	p.V218L	Unknown	Unknown	COSM44159	NA	1
6	218	c.653T>A	missense	p.V218E	Unknown	Unknown	COSM44317	NA	2
6	220	c.658_659ins1	frameshift	p.Y220fs	LOF	No DNE	COSM44978	Implied	1
6	220	c.658T>A, Unknown	missense	p.Y220N	Unknown	Unknown	COSM44672	NA	6
6	220	c.658T>C, Unknown	missense	p.Y220H	GOF	No DNE	COSM44637	(107)	5
6	220	c.659A>C, Unknown	missense	p.Y220S	LOF	DNE	COSM43850	(94)	7
6	220	c.659A>G, Unknown	missense	p.Y220C	GOF	DNE	COSM10758	(94,105)	52
6	220	c.660T>A, Unknown	nonsense	p.Y220X	LOF	No DNE	COSM43605	Implied	2
6	221	c.661del	frameshift	p.E221fs	LOF	No DNE	COSM44276	Implied	1
6	221	c.661G>T	nonsense	p.E221X	LOF	No DNE	COSM44817	Implied	1
6	222	c.664C>A	missense	p.P222T	Unknown	Unknown	COSM43607	NA	1
6	222	c.665_666ins4	frameshift	p.P222fs	LOF	No DNE	COSM8599684	Implied	1
6	222	c.665C>T	missense	p.P222L	Unknown	Unknown	COSM44606	NA	1

6	223	c.668C>T	missense	p.P223L	LOF	No DNE	COSM12193	(94)	1
6	223	Unknown	frameshift	p.P223fs	LOF	No DNE	NA	Implied	1
6	224	c.670G>T	nonsense	p.E224X	LOF	No DNE	COSM44922	Implied	1
6	224	c.672G>C, c.672G>T	missense	p.E224D	Unknown	Unknown	COSM44945, COSM11451	NA	2
7	226	c.676del	frameshift	p.G226fs	LOF	No DNE	NO-ID	Implied	1
7	226	c.676G>C	missense	p.G226R	Unknown	Unknown	NO-ID	NA	1
7	226	c.677G>T	missense	p.G226V	LOF	No DNE	COSM44399	(94)	1
7	227	c.679_680del, c.681_682ins1	frameshift	p.S227fs	LOF	No DNE	NO-ID, COSM69213	Implied	2
7	228	c.682G>A	missense	p.D228N	Unknown	Unknown	COSM44398	NA	2
7	228	c.683A>G	missense	p.D228G	Unknown	Unknown	COSM43960	NA	1
7	229	c.685T>C	missense	p.C229R	LOF	DNE	COSM45612	(94,129)	1
7	229	c.686_687del, Unknown	frameshift	p.C229fs	LOF	No DNE	COSM44360	Implied	5
7	229	c.686G>A	missense	p.C229Y	Unknown	Unknown	COSM44313	NA	1
7	229	c.687T>A	nonsense	p.C229X	LOF	No DNE	COSM45394	Implied	2
7	230	c.688del	frameshift	p.T230fs	LOF	No DNE	COSM45157	Implied	1
7	230	c.689C>A	missense	p.T230N	Unknown	Unknown	COSM43806	NA	1
7	231	c.691A>G	missense	p.T231A	Unknown	Unknown	COSM43980	NA	1
7	232	c.694_699del	in-frame deletion	p.I232_H233del	Unknown	Unknown	NO-ID	NA	1
7	232	c.695T>A	missense	p.I232N	Unknown	Unknown	COSM10715	NA	1
7	232	c.695T>C	missense	p.I232T	LOF	DNE	COSM44601	(94)	2
7	232	c.695T>G	missense	p.I232S	Unknown	Unknown	COSM45045	NA	2
7	233	c.697_704del	frameshift	p.H233fs	LOF	No DNE	NO-ID	Implied	1
7	233	c.698A>T	missense	p.H233L	Unknown	Unknown	COSM45959	NA	1
7	234	c.700_702ins3	in-frame deletion	p.Y234ins	Unknown	Unknown	NO-ID	NA	1
7	234	c.700T>A	missense	p.Y234N	Unknown	Unknown	COSM43956	NA	1
7	234	c.700T>C	missense	p.Y234H	LOF	No DNE	COSM11152	(94)	9

7	234	c.701A>G	missense	p.Y234C	LOF	Unknown	COSM10725	(94)	19
7	234	c.702C>A	nonsense	p.Y234X	LOF	No DNE	COSM45114	Implied	1
7	235	c.703_705del	in-frame deletion	p.N235del	Unknown	Unknown	COSM44784	NA	1
7	235	c.704_709del	in-frame deletion	p.N235_Y236del	Unknown	Unknown	COSM44333	NA	1
7	235	c.704A>G	missense	p.N235S	Unknown	Unknown	COSM43616	NA	1
7	235	c.705_706ins1	frameshift	p.N235fs	LOF	No DNE	COSM44394	Implied	1
7	236	c.706T>A	missense	p.Y236N	Unknown	Unknown	COSM43826	NA	1
7	236	c.706T>C	missense	p.Y236H	LOF	Unknown	COSM44326	(94)	4
7	236	c.706T>G	missense	p.Y236D	LOF	DNE	COSM43602	(94)	1
7	236	c.707A>C	missense	p.Y236S	LOF	No DNE	COSM44693	(94)	1
7	236	c.707A>G	missense	p.Y236C	LOF	DNE	COSM10731	(94)	8
7	236	c.708C>A, Unknown	nonsense	p.Y236X	LOF	No DNE	COSM43564	Implied	2
7	237	c.709A>G	missense	p.M237V	LOF	Unknown	COSM44525	(94)	1
7	237	c.709del	frameshift	p.M237fs	LOF	No DNE	COSM45162	Implied	2
7	237	c.710T>A	missense	p.M237K	LOF	Unknown	COSM43952	(94)	1
7	237	c.711G>A, c.711G>T, Unknown	missense	p.M237I	GOF	DNE	COSM10834, COSM11063	(105)	23
7	238	c.712_714delins2	frameshift	p.C238fs	LOF	No DNE	NO-ID	Implied	1
7	238	c.712T>A, c.713G>C	missense	p.C238S	LOF	DNE	COSM43700, COSM44653	(130)	3
7	238	c.712T>C	missense	p.C238R	LOF	Unknown	COSM44321	(94)	3
7	238	c.712T>G	missense	p.C238G	LOF	DNE	COSM46336	(94)	2
7	238	c.713G>A, Unknown	missense	p.C238Y	LOF	DNE	COSM11059	(94)	15
7	238	c.713G>T, Unknown	missense	p.C238F	LOF	DNE	COSM43778	(94)	7
7	238	c.714T>G	missense	p.C238W	LOF	Unknown	COSM44676	(94)	1
7	239	c.713_714ins1	frameshift	p.N239X	LOF	No DNE	NO-ID	Implied	2

7	239	c.714_715ins1, c.715_716del, c.715_716ins1, c.715_725del, c.717_718ins1, c.717_718ins7, Unknown	frameshift	p.N239fs	LOF	No DNE	COSM44155, NO-ID, NO-ID, NO-ID, COSM5988164, COSM45232	Implied	7
7	239	c.715A>G	missense	p.N239D	LOF	Unknown	COSM10777	(94)	10
7	239	c.716A>C	missense	p.N239T	LOF	DNE	COSM43801	(94,131)	2
7	239	c.716A>G	missense	p.N239S	GOF	DNE	COSM44094	(94,132)	3
7	239	c.716A>T	missense	p.N239I	Unknown	Unknown	COSM307333	NA	1
7	239	Unknown	in-frame deletion	p.N239_S240del	Unknown	Unknown	NA	NA	1
7	239	Unknown	in-frame insertion	p.N239_S240ins T	Unknown	Unknown	NA	NA	1
7	240	c.718A>C	missense	p.S240R	Unknown	Unknown	COSM45804	NA	1
7	240	c.718A>G	missense	p.S240G	LOF	DNE	COSM43973	(94)	1
7	240	c.719G>T	missense	p.S240I	Unknown	Unknown	COSM43660	NA	2
7	241	c.721del, c.722del	frameshift	p.S241fs	LOF	No DNE	COSM43645, NO-ID	Implied	6
7	241	c.721T>C	missense	p.S241P	LOF	Unknown	COSM44578	(94)	2
7	241	c.721T>G	missense	p.S241A	LOF	DNE	COSM44224	(94)	1
7	241	c.722_724del	in-frame deletion	p.241_242del	Unknown	Unknown	COSM45168	NA	2
7	241	c.722C>A	missense	p.S241Y	Unknown	Unknown	COSM10935	NA	2
7	241	c.722C>G	missense	p.S241C	LOF	Unknown	COSM10709	(96)	4
7	241	c.722C>T, Unknown	missense	p.S241F	GOF	DNE	COSM10812	(94,102)	14

7	242	c.723_724ins8, c.723del, c.724del	frameshift	p.C242fs	LOF	No DNE	NO-ID, COSM6530, COSM45826	Implied	12
7	242	c.724T>A, c.725G>C, Unknown	missense	p.C242S	LOF	Unknown	COSM44935, COSM11133	(94,133)	4
7	242	c.724T>C	missense	p.C242R	LOF	Unknown	COSM11738	(94)	1
7	242	c.724T>G	missense	p.C242G	Unknown	Unknown	COSM44135	NA	2
7	242	c.725G>A	missense	p.C242Y	GOF	DNE	COSM10646	(94)	9
7	242	c.725G>T	missense	p.C242F	LOF	DNE	COSM10810	(130)	4
7	242	c.726C>A	nonsense	p.C242X	LOF	No DNE	COSM44378	Implied	1
7	242	c.726C>G	missense	p.C242W	Unknown	Unknown	COSM11356	NA	1
7	243	c.727_740delins1	Frameshift	p.M243fs	LOF	No DNE	NO-ID	Implied	1
7	243	c.728T>C, c.728_729delins2	missense	p.M243T	LOF	Unknown	COSM44536, NO-ID	(96)	3
7	243	c.729_730delins2	missense	p.M243_G244d elinsIS	Unknown	Unknown	COSM6951415	NA	1
7	243	c.729G>A, c.729G>T, Unknown	missense	p.M243I	Unknown	Unknown	COSM44129, NO-ID	NA	5
7	244	c.727_732dup	In-frame insertion	p.G244_G245ins MG	Unknown	Unknown	COSM6022948	NA	1
7	244	c.730_732del	in-frame deletion	p.G244del1	Unknown	Unknown	NO-ID	NA	1
7	244	c.730_737del, c.732_736del	Frameshift	p.G244fs	LOF	No DNE	NO-ID, COSM45937	Implied	2
7	244	c.730G>A	missense	p.G244S	LOF	DNE	COSM10941	(94)	8
7	244	c.730G>T	missense	p.G244C	LOF	Unknown	COSM11524	(134)	4
7	244	c.731G>A	missense	p.G244D	LOF	DNE	COSM10883	(94)	7
7	244	c.731G>T	missense	p.G244V	LOF	DNE	COSM43652	(96)	2
7	244	c.732_733ins3	in-frame insertion	p.G244ins1	Unknown	Unknown	NO-ID	NA	1
7	245	c.733_745del	Frameshift	p.G245fs	LOF	No DNE	NO-ID	Implied	1
7	245	c.733G>A	missense	p.G245S	GOF	DNE	COSM6932	(102,107)	36

7	245	c.733G>T	missense	p.G245C	GOF	DNE	COSM11081	(102)	6
7	245	c.734G>A	missense	p.G245D	GOF	DNE	COSM43606	(102,107)	14
7	245	c.734G>C	missense	p.G245A	LOF	Unknown	COSM43965	(94)	1
7	245	c.734G>T	missense	p.G245V	GOF	DNE	COSM11196	(105)	9
7	246	c.736A>G	missense	p.M246V	GOF	DNE	COSM43555	(94)	6
7	246	c.736A>T	missense	p.M246L	GOF	DNE	COSM45992	(94)	1
7	246	c.736del	frameshift	p.M246X	LOF	No DNE	COSM44903	Implied	1
7	246	c.737T>A	missense	p.M246K	LOF	Unknown	COSM44103	(94,135)	1
7	246	c.737T>C	missense	p.M246T	LOF	Unknown	COSM11355	(94)	1
7	246	c.737T>G	missense	p.M246R	LOF	DNE	COSM11376	(107)	3
7	246	c.738G>A, Unknown	missense	p.M246I	LOF	DNE	COSM44310	(94)	4
7	247	c.739_740delinsTT	missense	p.N247F	LOF	Unknown	COSM44980	(94)	1
7	247	c.740_744delins5	missense	p.N247_R248del ins2	Unknown	Unknown	COSM44977	NA	1
7	247	c.740A>T	missense	p.N247I	LOF	DNE	COSM43995	(94)	3
7	248	c.742C>G	missense	p.R248G	LOF	DNE	COSM11564	(94)	6
7	248	c.742C>T, Unknown	missense	p.R248W	GOF	DNE	COSM10656	(102)	73
7	248	c.742del	frameshift	p.R248fs	LOF	No DNE	COSM45116	Implied	1
7	248	c.743G>A, Unknown	missense	p.R248Q	GOF	DNE	COSM10662	(94,107)	104
7	248	c.743G>C	missense	p.R248P	LOF	Unknown	COSM11491	(102)	1
7	248	c.743G>T	missense	p.R248L	LOF	DNE	COSM6549	(94)	3
7	249	c.745_750del	in-frame deletion	p.R249_P250del	Unknown	Unknown	NO-ID	NA	1
7	249	c.745A>G	missense	p.R249G	LOF	Unknown	COSM10668	(94)	5
7	249	c.745A>T	missense	p.R249W	LOF	DNE	COSM43629	(94)	3
7	249	c.746G>C	missense	p.R249T	GOF	Unknown	COSM43665	(94)	3
7	249	c.746G>T	missense	p.R249M	LOF	DNE	COSM43871	(94)	3
7	249	c.747G>T	missense	p.R249S	GOF	DNE	COSM10817	(107,123)	4
7	250	c.748C>A	missense	p.P250T	Unknown	Unknown	COSM45939	NA	1
7	250	c.748C>G	missense	p.P250A	Unknown	Unknown	COSM44096	NA	3
7	250	c.749C>T, Unknown	missense	p.P250L	LOF	DNE	COSM10771	(94)	5

7	251	c.750_751ins4, c.751del, c.753del	frameshift	p.I251fs	LOF	No DNE	COSM45818, COSM44124, NO-ID	Implied	4
7	251	c.751A>C	missense	p.I251L	Unknown	Unknown	COSM10931	NA	1
7	251	c.752T>G	missense	p.I251S	LOF	Unknown	COSM43829	(94)	2
7	251	Unknown	missense	p.I251N	Unknown	Unknown	NA	NA	1
7	252	c.754_762del	in-frame deletion	p.L252del3	Unknown	Unknown	COSM44457	NA	1
7	252	c.754del	frameshift	p.L252fs	LOF	No DNE	COSM44541	Implied	1
7	252	Unknown	missense	p.L252P	GOF	Unknown	NA	(94)	1
7	253	c.757A>C	missense	p.T253P	Unknown	Unknown	COSM45980	NA	1
7	253	c.757A>G	missense	p.T253A	LOF	Unknown	COSM45322	(94)	1
7	253	Unknown	frameshift	p.T253fs	LOF	No DNE	NA	Implied	1
7	254	c.761T>A	missense	p.I254N	LOF	Unknown	COSM44535	(94)	2
7	254	c.761T>C	missense	p.I254T	LOF	DNE	COSM44058	(94,129)	1
7	254	c.761T>G	missense	p.I254S	Unknown	Unknown	COSM45035	NA	1
7	254	Unknown	frameshift	p.I254fs	LOF	No DNE	NA	Implied	1
7	255	c.763_765del, c.765_767del, Unknown	in-frame deletion	p.I255del	LOF	Unknown	NO-ID, COSM69135	(136)	6
7	255	c.763A>T	missense	p.I255F	LOF	Unknown	COSM43651	(94,137)	4
7	255	c.764T>A	missense	p.I255N	Unknown	Unknown	COSM11244	NA	1
7	255	c.764T>C	missense	p.I255T	LOF	Unknown	COSM11181	NA	1
7	255	c.764T>G	missense	p.I255S	Unknown	Unknown	COSM10788	NA	4
7	255	c.765_766ins1, Unknown	frameshift	p.I255fs	LOF	No DNE	COSM45204	Implied	3
7	255	c.765_766ins12	in-frame insertion	p.I255ins4	Unknown	Unknown	NO-ID	NA	1
7	256	c.766_774del	in-frame deletion	p.T256del3	Unknown	Unknown	NO-ID	NA	1
7	256	c.766A>G	missense	p.T256A	Unknown	No DNE	COSM44544	(94)	1
7	256	c.766A>T	missense	p.T256S	Unknown	Unknown	COSM44662	NA	1

7	256	c.766del, c.768_769ins1, Unknown	frameshift	p.T256fs	LOF	No DNE	COSM45526, COSM45238	Implied	3
7	257	c.770T>A	missense	p.L257Q	LOF	DNE	COSM43530	(94)	5
7	257	c.770T>C	missense	p.L257P	LOF	DNE	COSM43842	(94)	3
7	258	c.772del	frameshift	p.E258fs	LOF	No DNE	COSM45001	Implied	1
7	258	c.772G>A	missense	p.E258K	GOF	DNE	COSM10988	(94,99)	1
7	258	c.772G>C	missense	p.E258Q	Unknown	Unknown	COSM10751	NA	2
7	258	c.772G>T, Unknown	nonsense	p.E258X	LOF	No DNE	COSM43568	Implied	5
7	258	c.773A>G	missense	p.E258G	LOF	Unknown	COSM44168	(94)	2
7	259	c.775del	frameshift	p.D259fs	LOF	No DNE	COSM44253	Implied	1
7	259	c.775G>A	missense	p.D259N	LOF	No DNE	COSM44117	(94)	2
7	259	c.775G>T	missense	p.D259Y	LOF	DNE	COSM11552	(94)	2
7	260	c.778_779del, Unknown	frameshift	p.S260fs	LOF	No DNE	COSM45768	Implied	3
7	261	c.781_782ins1	frameshift	p.S261fs	LOF	No DNE	NO-ID	Implied	1
7	261	c.783_806del	in-frame deletion	p.S261del8	Unknown	Unknown	COSM45574	NA	1
8	262	c.784_786del	in-frame deletion	p.G262del3	Unknown	Unknown	COSM45437	NA	1
8	262	c.784_797del, Unknown	frameshift	p.G262fs	LOF	No DNE	NO-ID	Implied	2
8	262	c.785G>T	missense	p.G262V	Unknown	Unknown	COSM11198	NA	2
8	262	Unknown	in-frame deletion	p.G262_N263de l	Unknown	Unknown	NA	NA	1
8	263	c.787A>G	missense	p.N263D	Unknown	Unknown	COSM45752	NA	1
8	264	c.790_792del	in-frame deletion	p.L264del	Unknown	Unknown	NO-ID	NA	2
8	264	c.790C>G	missense	p.L264V	Unknown	Unknown	COSM45704	NA	1
8	264	c.790del	frameshift	p.L264fs	LOF	No DNE	NO-ID	Implied	1
8	265	c.793_?del	large delins	p.(large delins)	LOF	No DNE	NA	Implied	1
8	265	c.793C>A	missense	p.L265M	LOF	Unknown	COSM45393	(94,138)	2

8	265	c.794T>C	missense	p.L265P	LOF	DNE	COSM11011	(94)	4
8	265	c.794T>G	missense	p.L265R	Unknown	Unknown	COSM44092	NA	1
8	265	Unknown	frameshift	p.L265fs	LOF	No DNE	NA	Implied	1
8	266	c.796del, Unknown	frameshift	p.G266fs	LOF	No DNE	NO-ID	Implied	2
8	266	c.796G>A, c.796G>C, Unknown	missense	p.G266R	GOF	No DNE	COSM10794, COSM11205	(94)	5
8	266	c.796G>T	nonsense	p.G266X	LOF	No DNE	COSM44891	implied	1
8	266	c.797_?del	large delins	p.(large delins)	LOF	No DNE	NA	Implied	1
8	266	c.797G>A, Unknown	missense	p.G266E	GOF	No DNE	COSM10867	(107)	12
8	266	c.797G>T	missense	p.G266V	LOF	No DNE	COSM10958	(107)	8
8	267	c.799_807del	in-frame deletion	p.R267del3	Unknown	Unknown	NO-ID	NA	1
8	267	c.799C>G	missense	p.R267G	Unknown	Unknown	COSM45822	NA	1
8	267	c.799C>T	missense	p.R267W	LOF	DNE	COSM11183	(94,139)	1
8	267	c.800G>A	missense	p.R267Q	LOF	No DNE	COSM43923	(94)	1
8	267	c.800G>C	missense	p.R267P	GOF	Unknown	COSM11392	(140)	1
8	268	c.803A>G	missense	p.N268S	Unknown	Unknown	COSM43981	NA	1
8	268	Unknown	frameshift	p.N268fs	LOF	No DNE	NA	Implied	1
8	269	c.805_841del, c.805_851del	frameshift	p.S269fs	LOF	No DNE	NO-ID, NO-ID	Implied	2
8	270	c.808T>A	missense	p.F270I	Unknown	Unknown	COSM43809	NA	1
8	270	c.808T>C, c.810T>G	missense	p.F270L	LOF	Unknown	COSM44262, COSM45297	(94)	4
8	270	c.809T>C, Unknown	missense	p.F270S	GOF	No DNE	COSM11305	(107)	5
8	270	c.809T>G	missense	p.F270C	LOF	DNE	COSM43621	(94,141)	2
8	271	c.810_811ins1, Unknown	frameshift	p.E271fs	LOF	No DNE	COSM45666	Implied	2
8	271	c.811G>A	missense	p.E271K	LOF	Unknown	COSM10719	(94)	2
8	271	c.811G>C	missense	p.E271Q	Unknown	Unknown	COSM43706	NA	2

8	271	c.811G>T	nonsense	p.E271X	LOF	No DNE	COSM43750	Implied	1
8	271	c.812A>G	missense	p.E271G	Unknown	Unknown	COSM43879	NA	1
8	272	c.814G>A	missense	p.V272M	LOF	DNE	COSM10891	(94)	9
8	272	c.814G>C, c.814G>T	missense	p.V272L	LOF	No DNE	COSM45898, COSM10859	(96,102)	3
8	272	c.815T>A	missense	p.V272E	Unknown	Unknown	COSM44580	NA	1
8	272	c.815T>G	missense	p.V272G	LOF	No DNE	COSM44870	(95)	3
8	273	c.817C>G, c.817C>T	missense	p.R273G	LOF	DNE	COSM43843, COSM10659	(94,142)	3
8	273	c.817C>T, Unknown	missense	p.R273C	GOF	DNE	COSM10659	(94,142)	50
8	273	c.817ins3	in-frame insertion	p.R273ins1	Unknown	Unknown	NO-ID	NA	1
8	273	c.818_819ins1, c.819_820ins1	frameshift	p.R273fs	LOF	No DNE	NO-ID, COSM5030952	Implied	6
8	273	c.818G>A, Unknown	missense	p.R273H	GOF	DNE	COSM10660	(94,107)	87
8	273	c.818G>C	missense	p.R273P	GOF	DNE	COSM43896	(105)	5
8	273	c.818G>T, Unknown	missense	p.R273L	GOF	DNE	COSM10779	(94)	8
8	274	c.820_821del	frameshift	p.V274fs	LOF	No DNE	NO-ID	Implied	1
8	274	c.820G>C	missense	p.V274L	LOF	No DNE	COSM44443	(94)	1
8	274	c.820G>T	missense	p.V274F	GOF	DNE	COSM10769	(94)	3
8	274	c.821T>A	missense	p.V274D	LOF	Unknown	COSM44448	(94)	2
8	274	c.821T>C	missense	p.V274A	LOF	Unknown	COSM44393	(94)	4
8	274	c.821T>G	missense	p.V274G	LOF	Unknown	COSM43945	(95)	2
8	275	c.823del, c.824del, c.831_840del	frameshift	p.C275fs	LOF	No DNE	COSM45251, COSM44178, NO-ID	Implied	3
8	275	c.823T>C	missense	p.C275R	Unknown	Unknown	COSM43902	NA	2
8	275	c.823T>G	missense	p.C275G	Unknown	Unknown	COSM11501	NA	1
8	275	c.824G>A, Unknown	missense	p.C275Y	GOF	DNE	COSM10893	(94)	9
8	275	c.824G>T, Unknown	missense	p.C275F	GOF	No DNE	COSM10701	(107)	5
8	275	c.825T>G	missense	p.C275W	LOF	Unknown	COSM43823	(94)	2

8	276	c.826_831del	in-frame deletion	p.A276_C277del	Unknown	Unknown	COSM45714	NA	1
8	276	c.826G>A	missense	p.A276T	Unknown	Unknown	COSM44114	NA	1
8	276	c.826G>C	missense	p.A276P	LOF	DNE	COSM43663	(94)	3
8	276	c.827C>A	missense	p.A276D	LOF	Unknown	COSM45268	(94,143)	1
8	276	c.827C>G, Unknown	missense	p.A276G	LOF	Unknown	COSM45695	(94)	2
8	277	c.829T>G	missense	p.C277G	Unknown	Unknown	COSM45074	NA	1
8	277	c.830G>A	missense	p.C277Y	LOF	DNE	COSM43737	(94,144)	2
8	277	c.830G>T	missense	p.C277F	LOF	Unknown	COSM10749	(94,145)	3
8	277	c.831_832ins21	in-frame insertion	p.C277ins7	Unknown	Unknown	NO-ID	NA	1
8	277	c.831T>A	nonsense	p.C277X	LOF	No DNE	COSM44972	Implied	1
8	277	c.831T>G	missense	p.C277W	LOF	Unknown	COSM45299	(94,146)	1
8	278	c.832_833del	frameshift	p.P278fs	LOF	No DNE	COSM6444763	Implied	1
8	278	c.832C>A, c.832C>G	missense	p.P278T	LOF	No DNE	COSM43697, COSM10814	(94)	4
8	278	c.832C>G	missense	p.P278A	LOF	Unknown	COSM10814	(94,147)	3
8	278	c.832C>T, c.833C>T, Unknown	missense	p.P278S	LOF	DNE	COSM10939, COSM10863	(140)	15
8	278	c.833C>A	missense	p.P278H	LOF	DNE	COSM43755	(94)	3
8	278	c.833C>G, Unknown	missense	p.P278R	LOF	No DNE	COSM10887	(94)	9
8	278	c.833C>T, Unknown	missense	p.P278L	LOF	DNE	COSM10863	(102)	7
8	278	c.834_835ins15	in-frame insertion	p.P278ins5	Unknown	Unknown	NO-ID	NA	1
8	279	c.835_840del	in-frame deletion	p.G279_R280del	Unknown	Unknown	NO-ID	NA	1
8	279	c.835del	frameshift	p.G279fs	LOF	No DNE	NO-ID	Implied	1
8	279	c.836G>A, Unknown	missense	p.G279E	GOF	DNE	COSM43714	(94,148)	3
8	280	c.838A>G	missense	p.R280G	LOF	DNE	COSM11123	(94)	2
8	280	c.838A>T	nonsense	p.R280X	LOF	No DNE	COSM44390	Implied	1
8	280	c.838del	frameshift	p.R280fs	LOF	No DNE	COSM46183	Implied	1
8	280	c.839G>A, Unknown	missense	p.R280K	LOF	DNE	COSM10728	(94)	8

8	280	c.839G>C, Unknown	missense	p.R280T	GOF	DNE	COSM10724	(94)	16
8	280	c.839G>T	missense	p.R280I	LOF	DNE	COSM11287	(94)	1
8	280	c.840A>C, Unknown	missense	p.R280S	LOF	DNE	COSM44233	(94)	4
8	281	c.841G>A	missense	p.D281N	GOF	DNE	COSM43596	(94)	3
8	281	c.841G>C	missense	p.D281H	LOF	Unknown	COSM10943	(94)	3
8	281	c.841G>T	missense	p.D281Y	GOF	DNE	COSM11516	(94)	1
8	281	c.842A>C	missense	p.D281A	LOF	Unknown	COSM11665	NA	1
8	281	c.842A>G, Unknown	missense	p.D281G	GOF	DNE	COSM11232	(94)	3
8	281	c.842A>T	missense	p.D281V	LOF	DNE	COSM45729	(94)	1
8	281	c.843C>A, c.843C>G	missense	p.D281E	GOF	DNE	COSM43906, COSM43837	(94)	2
8	282	c.844C>G	missense	p.R282G	GOF	DNE	COSM10992	(94)	11
8	282	c.844C>T, Unknown	missense	p.R282W	GOF	DNE	COSM10704	(94,105,149)	29
8	282	c.844del, Unknown	frameshift	p.R282fs	LOF	No DNE	COSM43813	Implied	3
8	282	c.845G>A	missense	p.R282Q	GOF	Unknown	COSM44338	(94,120)	1
8	282	c.845G>C	missense	p.R282P	LOF	DNE	COSM44306	(94,129)	2
8	282	c.845G>T	missense	p.R282L	Unknown	Unknown	COSM44470	NA	1
8	283	c.848G>A	missense	p.R283H	GOF	DNE	COSM11483	(94,144)	2
8	283	c.848G>C	missense	p.R283P	LOF	Unknown	COSM10743	(96)	2
8	284	c.850_851ins1, Unknown	frameshift	p.T284fs	LOF	No DNE	NO-ID	Implied	2
8	285	c.853_854del, Unknown	frameshift	p.E285fs	LOF	No DNE	NO-ID	Implied	2
8	285	c.853G>A, Unknown	missense	p.E285K	LOF	DNE	COSM10722	(94)	33
8	285	c.853G>T, Unknown	nonsense	p.E285X	LOF	No DNE	COSM44388	Implied	4
8	286	c.856del, c.857_863del	frameshift	p.E286fs	LOF	No DNE	COSM45277, NO-ID	Implied	2
8	286	c.856G>A	missense	p.E286K	LOF	DNE	COSM10726	(95)	12

8	286	c.856G>C	missense	p.E286Q	LOF	Unknown	COSM44250	(94)	1
8	286	c.857A>C	missense	p.E286A	LOF	DNE	COSM45892	(94)	1
8	286	c.857A>G	missense	p.E286G	LOF	DNE	COSM43565	(94)	3
8	286	c.857A>T	missense	p.E286V	LOF	Unknown	COSM43936	(94)	1
8	287	c.859G>T	nonsense	p.E287X	LOF	No DNE	COSM44133	Implied	1
8	287	c.887_915del, Unknown	frameshift	p.E287fs	LOF	No DNE	NO-ID	Implied	3
8	288	c.862_866del, c.862del, Unknown	frameshift	p.N288fs	LOF	No DNE	NO-ID, NO-ID	Implied	3
8	289	Unknown	frameshift	p.L289fs	LOF	No DNE	NA	Implied	1
8	290	Unknown	frameshift	p.R290fs	LOF	No DNE	NA	Implied	1
8	291	c.871A>T	nonsense	p.K291X	LOF	No DNE	COSM44475	Implied	2
8	291	c.872A>G	missense	p.K291R	Unknown	Unknown	COSM43747	NA	1
8	291	c.873G>C	missense	p.K291N	LOF	Unknown	COSM44446	(94)	1
8	292	c.874_875del, c.877_880del	frameshift	p.K292fs	LOF	No DNE	NO-ID	Implied	3
8	292	c.875A>G	missense	p.K292R	Unknown	Unknown	COSM43624	NA	1
8	293	c.877_891del	in-frame deletion	p.G293del5	Unknown	Unknown	NO-ID	NA	1
8	294	c.880G>A	missense	p.E294K	Unknown	Unknown	COSM44127	NA	1
8	294	c.880G>T	nonsense	p.E294X	LOF	No DNE	COSM10856	Implied	8
8	295	c.882_883ins1, c.882_885delins3, c.883del	frameshift	p.P295fs	LOF	No DNE	COSM45700, NO-ID, NO-ID	Implied	4
8	295	c.884C>T	missense	p.P295L	Unknown	Unknown	COSM45233	NA	3
8	296	c.887A>T	missense	p.H296L	Unknown	Unknown	COSM44522	NA	1
8	297	c.889C>G	missense	p.H297D	Unknown	Unknown	NO-ID	NA	1
8	297	c.889ins5	frameshift	p.H296fs	LOF	No DNE	NO-ID	Implied	1
8	297	c.890A>C	missense	p.H297P	Unknown	Unknown	COSM45478	NA	1
8	298	c.892_896del	frameshift	p.E298fs	LOF	No DNE	NO-ID	Implied	1
8	298	c.892G>C	missense	p.E298Q	Unknown	Unknown	COSM45938	(94)	2

8	298	c.892G>T, Unknown	nonsense	p.E298X	LOF	No DNE	COSM10710	Implied	7
8	299	c.896T>C	missense	p.L299P	Unknown	Unknown	COSM46159	NA	1
8	299	Unknown	frameshift	p.L299fs	LOF	No DNE	NA	Implied	2
8	300	c.898_901del, Unknown	frameshift	p.P300fs	LOF	No DNE	NO-ID	Implied	2
8	301	c.901del, Unknown	frameshift	p.P301fs	LOF	No DNE	NO-ID	Implied	3
8	301	c.902C>A	missense	p.P301Q	Unknown	Unknown	COSM43873	NA	1
8	302	Unknown	frameshift	p.G302fs	LOF	No DNE	NA	Implied	2
8	303	Unknown	frameshift	p.S303fs	LOF	No DNE	NA	Implied	2
8	304	Unknown	frameshift	p.T304fs	LOF	No DNE	NA	NA	2
8	305	c.912_913ins1, Unknown	frameshift	p.K305fs	LOF	No DNE	COSM45458	Implied	2
8	306	c.916C>T, Unknown	nonsense	p.R306X	LOF	No DNE	COSM10663	Implied	27
8	306	c.916del	frameshift	p.R306fs	LOF	No DNE	NO-ID	Implied	1
9	308	c.923_?del	in-frame deletion	p.(large delins)	LOF	No DNE	NA	Implied	1
9	308	Unknown	frameshift	p.L308fs	LOF	No DNE	NA	Implied	1
9	310	c.929del	frameshift	p.N310fs	LOF	No DNE	COSM6925440	Implied	1
9	311	c.931A>C	missense	p.N311H	Unknown	Unknown	COSM45618	NA	1
9	313	c.937_941del	frameshift	p.S313fs	LOF	No DNE	NO-ID	Implied	1
9	314	Unknown	nonsense	p.S314X	LOF	No DNE	NA	Implied	1
9	317	c.?(949_ins5&C>T), c.949del	frameshift	p.Q317fs	LOF	No DNE	NO-ID, COSM43573	Implied	2
9	317	c.949C>T	nonsense	p.Q317X	LOF	No DNE	COSM10786	Implied	7
9	318	c.951_952ins1, Unknown	frameshift	p.P318fs	LOF	No DNE	NO-ID	Implied	3
9	319	c.955del	frameshift	p.K319fs	LOF	No DNE	NO-ID	Implied	1
9	320	c.958A>T, Unknown	nonsense	p.K320X	LOF	No DNE	COSM44335	Implied	2
9	321	c.963del	frameshift	p.K321fs	LOF	No DNE	COSM44876	Implied	1
9	322	Unknown	frameshift	p.P322fs	LOF	No DNE	NA	Implied	1
9	325	c.973G>T	nonsense	p.G325X	LOF	No DNE	COSM307331	Implied	2

9	325	Unknown	missense	p.G325E	LOF	Unknown	NA	(94)	1
9	328	c.982_987del	in-frame deletion	p.F328del2AA	Unknown	Unknown	NO-ID	NA	1
9	328	Unknown	frameshift	p.F328fs	LOF	No DNE	NA	Implied	1
9	330	c.989T>G, Unknown	missense	p.L330R	LOF	No DNE	COSM45643	(94,150)	2
9	331	c.991C>T, Unknown	nonsense	p.Q331X	LOF	No DNE	COSM11354	Implied	12
10	331	c.993_994ins14, Unknown	frameshift	p.Q331fs	LOF	No DNE	NO-ID	Implied	2
10	333	c.997del, Unknown	frameshift	p.R333fs	LOF	No DNE	NO-ID	Implied	4
10	334	c.1000C>G	missense	p.G334R	LOF	No DNE	NO-ID	(93,94)	2
10	334	c.1000G>T	missense	p.G334W	Unknown	Unknown	COSM48816	(94)	1
10	334	Unknown	missense	p.G334V	LOF	DNE	NA	(94)	1
10	336	c.1006G>T	nonsense	p.E336X	LOF	No DNE	COSM11291	Implied	1
10	337	c.1009C>T, Unknown	missense	p.R337C	LOF	No DNE	COSM11071	(94)	8
10	337	c.1010G>A	missense	p.R337H	LOF	No DNE	COSM43882	(94)	1
10	337	c.1010G>T	missense	p.R337L	LOF	No DNE	COSM11411	(94)	3
10	338	c.1013_1014ins18	in-frame insertion	p.F338ins6	Unknown	Unknown	NO-ID	NA	1
10	338	Unknown	frameshift	p.F338fs	LOF	No DNE	NA	Implied	1
10	339	c.1015_1022del, c.1015del, Unknown	frameshift	p.E339fs	LOF	No DNE	COSM5442033, COSM5230441	Implied	3
10	339	c.1015G>T, Unknown	nonsense	p.E339X	LOF	No DNE	COSM11286	Implied	3
10	340	c.1017_1018ins2, Unknown	frameshift	p.M340fs	LOF	No DNE	NO-ID	Implied	2
10	341	c.1022T>C	missense	p.F341S	LOF	No DNE	COSM1386561	(94)	1
10	341	c.1022T>G	missense	p.F341C	LOF	No DNE	COSM45744	(94,151)	1
10	342	c.1023_1024delins2, c.1024C>T, Unknown	nonsense	p.R342X	LOF	No DNE	COSM44906, COSM11073	Implied	34

10	342	c.1024del, c.1025del, Unknown	frameshift	p.R342fs	LOF	No DNE	COSM18597, COSM46154	Implied	5
10	342	c.1025G>C	missense	p.R342P	LOF	No DNE	COSM45276	(94)	6
10	344	c.1032_1042del	frameshift	p.L344fs	LOF	No DNE	NO-ID	Implied	1
10	344	Unknown	missense	p.L344R	LOF	No DNE	NA	NA	1
10	345	Unknown	in-frame deletion	p.N345_L348del insM	Unknown	Unknown	NA	NA	1
10	347	c.1039G>A	missense	p.A347T	LOF	No DNE	COSM45717	(94,152)	1
10	348	Unknown	missense	p.L348F	LOF	No DNE	COSM46348	(94)	3
10	348	Unknown	missense	p.L348S	LOF	No DNE	NA	(94)	1
10	349	c.1045G>T	nonsense	p.E349X	LOF	No DNE	COSM10770	Implied	2
10	349	Unknown	in-frame deletion	p.E349_G356del	Unknown	Unknown	NA	NA	1
10	349	Unknown	frameshift	p.E349fs	LOF	No DNE	NA	Implied	2
10	351	c.1051A>T	nonsense	p.K351X	LOF	No DNE	COSM1522202	Implied	1
11	374	Unknown	frameshift	p.G374fs	LOF	No DNE	NA	Implied	1
11	375	c.1123C>T	nonsense	p.Q375X	LOF	No DNE	COSM307348	Implied	1
11	382	c.1146del	frameshift	p.K382fs	LOF	No DNE	COSM13747	Implied	1
11	390	Unknown	frameshift	p.P390fs	LOF	No DNE	NA	Implied	1
11	393	c.1177del4, Unknown	frameshift	p.D393fs	LOF	No DNE	NO-ID	Implied	2

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;