

**Supplemental table 1****Mutations included in the non-N215S group.**

Mutations	Number of patients	Mutations	Number of patients
<i>Missense</i>		<i>Nonsense</i>	
A13P	2	Q107X	2
A143T	9	R220X	1
A257P	1	R227X	20
A309P	2	R301X	2
A31V	3	S78X	1
C52G	2	W209X	1
D93Y	1	W227X	1
E338K	5	W277X	2
G261D	1	Y184X	2
G261V	1	<i>Small deletions and other rearrangements</i>	
G361A	3	c.1223del6	1
G361R	3	c.359_364 del TAGCTA del	2
H46Y	1	c.402 del T	1
I303N	1	c.466delG	2
I317T	9	c.520delT	8
I91T	1	c.700_702 del GAT del	3
L166P	5	c.717-718 het del AA	1
L372P	2	c.717del2	1
M42V	2	c.748_801+8del62	2
N33D	3	c.802-3_802-2 delCA	3
P205T	13	exon 1 deletion	3
P293H	2	exon 7 deletion	1
P409T	3	c.589het-dupA	1
Q221P	1		
Q279H	1		
Q280H	2		
R112C	1		
R112H	2		
R301P	1		
R301Q	9		
R342Q	3		
R49L	2		
S126G	1		
T410I	2		
T410L	1		
V269A	2		
V316E	2		