

Table 2: Non-Synonymous Coding Variants

Amino Acid	Codon	Position	Genotype (number of patients)
Val 7 Leu	<u>G</u> TC --> <u>C</u> TC	within signal peptide	G/G (10) C/G (11) C/C (5)
Met 1016 Val	<u>A</u> TG --> <u>G</u> TG	Y position of repeat	A/A (26) A/G (1) G/G (0)
Gln 1316 Glu	<u>C</u> AA --> <u>G</u> AA	Y position of repeat	C/C (26) C/G (1) G/G (0)
Gln 1334 His	<u>C</u> AA --> <u>C</u> AC	Y position of repeat	A/A (11) A/C (13) G/G (3)