

Nucleotide change	Amino-acid change	Novel/Previously described	Allelic frequency in control population (%)
c.-84-55G>T		Novel	0
c.-84-54G>A		Novel	0
c.-20C>G		Novel	0
c.18C>T	D6D	Novel	4.95
c.112_131del	G38Qfs21X	Novel	0
c.153-155del	G52Afs56X	Novel	0
c.249C>T	Y83Y	Novel	0.50
c.684C>T	P228P	Novel	0
c.822C>T	G274G	Novel	0
c.927T>C	H309H	Previously described rs139884	56.98
c.1257T>C	S419S	Novel	0
c.*131G>A		Novel	0