

S3: ON-LINE ONLY. Description, frequency and distribution of missense

B2M mutations and predicted *in silico* effect of protein structure and function

(*occurred in conjunction with a pathogenic mutation).

Exon	Genomic change	Amino acid change	Frequency	Effect on protein structure/function
1	c.14T>C	p.(Val5Ala)	1	Unlikely
1	c.35T>C	p.(Leu12Pro)	1*	Uncertain
1	c.37C>T	p.(Leu13Phe)	1	Unlikely
1	c.38T>C	p.(Leu13Pro)	1*	Uncertain
1	c.38T>G	p.(Leu13Arg)	1*	Uncertain
2a	c.133T>C	p.(Cys45Arg)	1*	Likely
2a	c.152A>G	p.(His51Arg)	1	Likely
2a	c.235G>A	p.(Asp79Asn)	1*	Likely
2a	c.274C>T	p.(Pro92Ser)	1	Likely
2a	c.293A>G	p.(Tyr98Cys)	1*	Likely
2a	c.299G>C	p.(Cys100Ser)	1	Likely