Table 4.5: Summary of the variations found during sequencing, giving the specifications of the variants including: variation type, the cases it was found in, their reference and the significance of the variation.

Exon number	Variation type	Exon/Intron	Variation	Cases present	References	Significance
2	Substitution	Intron (upstream)	G>A 87921(refseq) Chr11: 2527911	S20, S29, S45	rs28730661	Benign
2	Substitution	Intron (downstream)	G>A 88033(refseq) Chr11: 2528023	C2, C5, C6, C9, C10, C11, C13	rs39750811	Benign
3	Substitution	Intron (upstream)	G>A 130628(refseq) Chr11: 2570618	S1, S5, S6, S7, S10, S16, S17, S25, S26, S32, S33, S36, S38, S43, S46, S47, S60, S62, S63	rs28730752 (Duzkale *et al.*, 2013)	Benign
8	Substitution, silent	Exon	G>A c.720 p.240 =Gln	C13	rs199473663 (Crotti *et al.*, 2007)	Likely benign

Table 4.5(continued): Summary of the variations found during sequencing, giving the specifications of the variants including: variation type, the cases it was found in, their reference and the significance of the variation.

Exon number	**Variation type**	**Exon/Intron**	**Variation**	**Cases present**	**References**	**Significance**
10	Substitution, missense	Exon	G>C c.1007 Ser336Thr	S7, S9, S11, S13, S29, S33, S34	rs184636161	Predicted as benign
13	Substitution, silent	Exon	G>A c.1257 p.419 =Ser	C19 S24	rs1057128 (Frost *et al.*, 2010)	Benign
13	Substitution	Intron (Downstream)	A>G 336100(refseq) Chr11: 2776090	C4, C6, C19, S1, S4, S5, S7, S10, S11, S24, S25, S27, S28, S31, S34, S37, S38, S41, S42, S48, S49, S50, S54, S56, S58, S65	rs163150 (Alders *et al.*, 2009)	Benign