Supplementary Table 1: benign variations identified in the untranslated regions (UTR).						
Patient III-1 (family A)	variant	dbSNP	1000 Genomes MAF	Highest Population MAF	Zigosity	**Clinical Significance (Clin Var)**
5'UTR	c.-872_-871insTC	rs112192237	0,47	0,49	Hetero	With Uncertain significance,other allele
	c.-591delT	rs11356115	0,43	0,5	Hetero	With Benign allele
3'UTR	c.*1285A>G	rs1061651	0,28	0,48	Homo	With Benign allele
	c.*1510A>G	rs1061657	0,26	0,49	Homo	With Benign allele
Patient IV-2 (family B)	variant	dbSNP	1000 Genomes MAF	Highest Population MAF	Zigosity	**Clinical Significance (Clin Var)**
5'UTR	c.-872_-871insTC	rs112192237	0,47	0,49	Hetero	With Uncertain significance,other allele
	c.-591delT	rs11356115	0,43	0,5	Hetero	With Benign allele
	c.-417T>G	rs36202980	0,027	0,09	Hetero	With Benign allele
	c.-184C>T	rs2242442	0,25	0,48	Hetero	With Benign allele
3'UTR	c.*423G>C	rs3741698	0,41	0,49	Hetero	With Benign allele
	c.*739A>G	rs8853	0,5	0,5	Hetero	With Benign allele
	c.*1285A>G	rs1061651	0,28	0,48	Hetero	With Benign allele
	c.*1510A>G	rs1061657	0,26	0,49	Hetero	With Benign allele