

Table 2. Results of sequencing analysis showing the novel mutation and the known polymorphisms (1,2) in the CaSR of proband and family genetic profile

Patient	DNA (Gen Bank Accession number NM:000388.3)	DNA change	Protein change	Result
Z1	Intron3	c.492+19G>A homozygous	-	Polymorphism1
Z2	Intron3	c.492+19G>A homozygous	-	Polymorphism1
Z3	Intron3	c.492+19G>A heterozygous	-	Polymorphism1
Z1	Exon 4	c. 772_773delGTinsA heterozygous	pVal258ArgfsX47	Mutation
Z2	Exon 4	c. 772_773delGTinsA heterozygous	pVal258ArgfsX47	Mutation
Z3	Exon 4	c. 772_773delGTinsA heterozygous	pVal258ArgfsX47	Mutation
Z1	Ex7-3'UTR	c.*60A>T homozygous	-	Polymorphism2
Z2	Ex7-3'UTR	c.*60A>T homozygous	-	Polymorphism2
Z3	Ex7-3'UTR	c.*60A>T heterozygous	-	Polymorphism2

Z1: ZS, proband; Z2: ZP, sister; Z3: ZB, brother; Polymorphism1: rs9869985; Polymorphism2: rs 4677948.