Appendix 2. Molecular analysis of *KCNV2* polymorphisms

Pt	Exon	Nucleotide substitution	Amino acid change	Hom/het	SIFT	Polyphen 2		HSF Matrix				Allelic frequency observed by EVS	Reference
					Prediction	Prediction	Hum var score (0-1)	Site affected	Wildtype CV	Mutant CV	CV % variation		
1, 2, 3, 4	1	c.183 C>G	p.Gly61Gly (cording-synonymous)	Hom	Torelant	NA	NA	Acceptor	65.85	65.22	No change (-0.95 %)	7647/13006	db SNP (rs10967705)
1, 2, 3, 4	1	c.795 C>G	p.Ala265Ala (cording-synonymous)	Hom	Torelant	NA	NA	Donor	69.7	69.94	No change (-4.22%)	5636/13006	db SNP (rs12237048)

Pt = patient; Hom = homozygous; Het = heterozygous; SIFT = sorting Intolerant from Tolerance; HSF = human splicing finder program; CV = consensus values; EVS = exome variant server.