

Table 1. Results of in-silico analysis of rare missense *ADGRV1* mutations with allele frequency < 0.0005 reported by Myers KA *et al.* and this study

Base change	Codon change	rs ID	gnomAD	Polyphen-2	Mutation Taster	Grantham (<100)	GERP (>4.4)	Protein Domain	Inheritance
c.2021A>G	p.Tyr674Cys	rs761220696	4/17562 (EA) 1/110184 (EU) 5/242476 (All)	Benign (0.001)	Polymorphism	Radical 194	-5.21	Calx-β5	Father
*c.2039A>G	p.Asp680Gly	rs547076322	7/17866 (EA) 7/247616 (All)	Damaging (1)	Disease Causing	Conservative 94	5.87	Calx-β5	Mother
c.2261T>C	p.Val754Ala	rs374609813	0/19516 (EA) 22/30544 (SA) 13/128162 (EU) 39/280148 (All)	Damaging (0.682)	Disease Causing	Conservative 64	5.09	Extracellular, links Calx-β5 and Calx-β6	Unknown
c.5722G>A	p.Asp1908Asn	rs757418364	0/19490 (EA) 9/128042 (EU) 9/279986 (All)	Damaging (0.976)	Disease Causing	Conservative 23	5.56	Calx-β13	Mother
c.8266G>A	p.Gly2756Arg	rs546198768	0/18314 (EA) 11/117330 (EU) 15/257862 (All)	Damaging (1)	Disease Causing	Radical 125	5.15	Calx-β19	Mother
c.9466A>G	p.Ile3156Val	rs372484022	0/19532 (EA) 57/30568 (SA) 7/127064 (EU) 69/279132 (All)	Benign (0.274)	Disease Causing	Conservative 29	1.86	Calx-β22	Mother
c.13228G>A	p.Glu4410Lys	rs371970388	0/19532 (EA) 17/126258 (EU) 18/273998 (All)	Damaging (0.676)	Polymorphism	Conservative 56	1.39	Calx-β30	Unknown
c.13495C>T	p.Arg4499Cys	rs567519802	0/17780 (EA) 6/29982 (SA) 7/245774 (All)	Damaging (0.998)	Disease Causing	Radical 180	5.97	Extracellular, links Calx-β30 and Calx-β31	Unknown
c.13568G>C	p.Ser4523Thr	rs376673439	0/17816 (EA) 11/111494 (EU) 12/246276 (All)	Damaging (0.996)	Disease Causing	Conservative 58	5.78	Calx-β31	Father

*Identified in the present Study

Reference sequence number: NM_032119.3 NP_115495.3

Abbreviations: rsID, reference ID in dbSNP; gnomAD, The Genome Aggregation Database; EA, East Asian; EU, non-Finnish Europe; SA, South Asian; All, Total population; Polyphen-2, Polymorphism Phenotyping v2;) AF (Allele Frequency); Grantham, Grantham scores for conservative < 100; GERP, Genomic Evolutionary Rate Profiling for conservative > 4.4