

Supplementary Table S1. Information on the variants that remained after the filtering steps

Chrom	Position	Ref	Alt	State	Gene Name	Accession number	Exon	Nucleotide change	Amino Acid change	SNP ID	<i>in silico</i> analysis			Frequency		Reference	
											Polyphen2_HVAR_score	SIFT_score	PROVEAN_score	HGVD	ExAC		ExAC East Asian
1	46194329	T	C	Ref/Alt	<i>POMGNT1</i>	NM_001290129	8	c.758A>G	p.Y253C	rs200789546	0.999 (damaging)	0 (damaging)	-7.15 (deleterious)	0	0	0	
5	90642725	A	C	Ref/Alt	<i>ADGRV1</i>	NM_032119	12	c.2330A>C	p.E777A	rs78627723	0.939 (damaging)	0.172 (tolerated)	-3.04 (deleterious)	0.00657895	0.0001	0.0017	
5	149884549	G	A	Alt/Alt	<i>PDE6A</i>	NM_000440	16	c.1957C>T	p.R653X	rs753942596	not evaluated	not evaluated	not evaluated	0	0.00003302	0	Perez-Carro R et al., 2016, Sci Rep
10	71741838	C	T	Ref/Alt	<i>CDH23</i>	NM_022124	36	c.4762C>T	p.R1588W	rs137937502	0.996 (damaging)	not evaluated	not evaluated	0.00513539	0.0002	0.0019	Miyagawa M et al. 2013, Plos One
11	8094153	G	A	Alt/Alt	<i>TUB</i>	NM_177972	4	c.361G>A	p.G121S	rs768447598	0.063 (benign)	0.464 (tolerated)	-0.42 (neutral)	0.00520833	0.00002506	0.0001	
17	8014012	A	G	Ref/Alt	<i>GUCY2D</i>	NM_000180	12	c.2396A>G	p.D799G	not reported	0.247 (benign)	0.02 (damaging)	-5.64 (deleterious)	0	0	0	
X	85894235	C	T	Ref/Alt	<i>CHM</i>	NM_000390	12	c.1463G>A	p.R488Q	not reported	0.478 (possibly damaging)	0.211 (tolerated)	-1.56 (neutral)	0	0	0	

Chro = chromosome, Ref = reference allele, Alt = alternative allele, The position of variants is determined using the Human Genome reference (GRCh37/hg19), HGVD = human genetic variation database (<http://www.hgvd.genome.med.kyoto-u.ac.jp/index.html>), ExAC = Exome Aggregation Consortium database (<http://exac.broadinstitute.org>), Polyphen-2 (<http://genetics.bwh.harvard.edu/pph2/>), SIFT (<http://sift.jcvi.org>), PROVEAN (<http://provean.jcvi.org>)