

Supplementary Table 2

Ten most frequent alleles in the cohort			
Gene	Variant on cDNA level	Variant and consequence on amino acid level	Number of alleles
<i>ABCA4</i>	c.5603A>T	p.(Asn1868Ile)	50
<i>ABCA4</i>	c.5882G>A	p.(Gly1961Glu)	34
<i>ABCA4</i>	c.[1622T>C;3113C>T]	p.[(Leu541Pro);(Ala1038Val)]	25
<i>CNGB3</i>	c.1148del	p.(Thr383Ilefs*13)	16
<i>ABCA4</i>	c.2588G>C	p.(Gly863Ala)	13
<i>USH2A</i>	c.11864G>A	p.(Trp3955*)	13
<i>CDHR1</i>	c.783G>A	p.(Pro261=);p.(Asp214_Pro261del)	11
<i>ABCA4</i>	c.5461-10T>C	p.(Thr1821Valfs*13);p.(Thr1821Aspfs*6)	10
<i>USH2A</i>	c.2299del	p.(Glu767Serfs*21)	9
<i>USH2A</i>	c.4714C>T	p.(Leu1572Phe)	9

Refseq transcripts: *ABCA4* (NM_000350.2), *USH2A* (NM_206933.2), *CNGB3* (NM_019098.4), *CDHR1* (NM_033100.3).