

Table S4 Exome data-filtering approach used to select for candidate variants in the myopia pedigree

Analytical strategy	Data					
Patient	Proband (II-3)					
Database	1000 Genomes (CHS)		dbSNP (Global)		ExAc	
Total variants	38281		38281		38281	
Variant type	SNV	INDEL	SNV	INDEL	SNV	INDEL
	35751	2530	35751	2530	35751	2530
Rare variants (MAF<1% in 1000 Genomes (CHS)/dbSNP/ExAc or not available in databases)	3139	Omitted*	3443	Omitted*	7987	Omitted*
SNV(MS/SS/NS)&INDEL	868	577	952	577	710	577
Variants within the candidate regions	47	5	47	5	32	5
Variants segregating with disease	<b>7</b>	<b>0</b>	<b>7</b>	<b>0</b>	<b>7</b>	<b>0</b>
Myopia-related and heterozygously inherited gene	<b>2</b>	<b>0</b>	<b>2</b>	<b>0</b>	<b>2</b>	<b>0</b>
Candidate variants	2 (located in <i>NDUFAF7</i> and <i>RHO</i> , respectively)					

CHS is the sub-database of 1000 Genomes, which represents population samples that are Southern Han Chinese.

SNV, single nucleotide variants including MS (missense variants), SS (splice site variants), and NS (nonsense variants). INDEL including MNV (multiple nucleotide variation with alleles of common length > 1) and DIV (deletion/insertion variation). \*, the INDELS-filtering procedure based on MAF was omitted, because the frequency information of INDEL is usually not complete in 1000 Genomes, dbSNP, and ExAc.