

Appendix 1

A list of primers used in this study, including those utilized in the amplification of the L cone gene and each exon by Long Range PCR (LRP), sequencing of each L cone exon 5 variant, construction and cloning of L cone opsin constructs, site-directed mutagenesis, and quantitative PCR (qPCR).

Primer	Primer sequence (5' – 3' orientation)	Location and function
FG	GAGGCGAGGCTACGGAGT	Forward primer targeted to the upstream promoter region of the first L opsin gene in the L/M cone opsin array. Used in Long Range PCR (LRP) to amplify L opsin from genomic DNA (gDNA).
E6	GCAGTGAAAGCCTCTGTGACT	Reverse primer targeted to exon 6 of L opsin gene. Used in LRP to amplify L opsin from genomic DNA.
2F	GCACTGGTATCGACAGGCG	Exon 2 specific primer used in a second PCR following LRP of the L opsin gene.
2R	CAGTATATGGATGTGAGGC	Exon 2 specific primer used in a second PCR following LRP of the L opsin gene.
3F	CTCAGTCCGTGGAGCCCTGAATTC	Exon 3 specific primer used in a second PCR following LRP of the L opsin gene.
3R	ACATTGATAGACATTGCACGCTCA	Exon 3 specific primer used in a second PCR following LRP of the L opsin gene.
4F	GGTGA CTCCACAGAATTGAT	Exon 4 specific primer used in a second PCR following LRP of the L opsin gene.
4R	CTGATTCTCATCGCTGGATCT	Exon 4 specific primer used in a second PCR following LRP of the L opsin gene.
5F	CTATGCCTGGGTACCTGCCTC	Exon 5 specific primer used in a second PCR following LRP of the L opsin gene.
5R	CTTATCAGAGACATGATTCCAGTGG	Exon 5 specific primer used in a second PCR following LRP of the L opsin gene.
PE-L-cone-F	GCGCGAATTCCACCATGGCCCAGCAGTGGAGCCTC	Cloning primers to permit the insertion of the L cone opsin sequence (i.e. exons 1-6, including introns and 5) into the pMT4 mammalian expression vector.
E4-I4R	GAATCGGGGGGCTTACCGCTCGGATGGCCAG	Cloning primers to permit the insertion of the L cone opsin sequence (i.e. exons 1-6, including introns and 5) into the pMT4 mammalian expression vector.
E4-I4F	CTGGCCATCCGAGCGGTAAGCCCCCGATTCT	Cloning primers to permit the insertion of the L cone opsin sequence (i.e. exons 1-

		6, including introns and 5) into the pMT4 mammalian expression vector.
I4-E5R	CTGCTGCTTTGCCACCTAAGGGAGACAGGG	Cloning primers to permit the insertion of the L cone opsin sequence (i.e. exons 1-6, including introns and 5) into the pMT4 mammalian expression vector.
I4-E5F	CCCTGTCTCCCTTAGGTGGCAAAGCAGCAG	Cloning primers to permit the insertion of the L cone opsin sequence (i.e. exons 1-6, including introns and 5) into the pMT4 mammalian expression vector.
E5-I5R	ATGGTGTTGCTTACCTGCCGGTTCATAAAG	Cloning primers to permit the insertion of the L cone opsin sequence (i.e. exons 1-6, including introns and 5) into the pMT4 mammalian expression vector.
E5-I5F	TTTATGAACCGGCAGGTAAGCAACACCATC	Cloning primers to permit the insertion of the L cone opsin sequence (i.e. exons 1-6, including introns and 5) into the pMT4 mammalian expression vector.
I5-6R	ATGCAGTTTCGAACTGGAAGGACAGAAAAG	Cloning primers to permit the insertion of the L cone opsin sequence (i.e. exons 1-6, including introns and 5) into the pMT4 mammalian expression vector.
I5-E6F	TTTCTGTCCTTCCAGTTTCGAACTGCATC	Cloning primers to permit the insertion of the L cone opsin sequence (i.e. exons 1-6, including introns and 5) into the pMT4 mammalian expression vector.
PE-L-cone-R	CGGCGTCGACGCTGCAGGCGATACCGAGGACA	Cloning primers to permit the insertion of the L cone opsin sequence (i.e. exons 1-6, including introns and 5) into the pMT4 mammalian expression vector.
E4F	TCATCATGCTCTGCTACCTCCAAG	To identify novel variants in myopic patients from gDNA.
E5R	GTCACCTCCTTCTCTGCCTTCTG	To identify novel variants in myopic patients from gDNA.
E5F	ACGCCTTCCACCCTTTGATG	To identify novel variants in myopic patients from gDNA.
E6R	TCAGAGCCATCGTCAACCTTCTTC	To identify novel variants in myopic patients from gDNA.
Var970_E5_R	CTGCCGGTTCATAATGACATAGATAACGGG	Primers specific to L cone exon 5 for the introduction of variants by site directed mutagenesis. Paired with either PE-L-cone-F or PE-L-cone-R.
Var971_E5_R	CTGCCGGTTCATATAGACATAGATAACGGG	Primers specific to L cone exon 5 for the introduction of variants by site directed

		mutagenesis. Paired with either PE-L-cone-F or PE-L-cone-R.
Var973_E5_R	CTGCCGGTTCACAAAGACATAGATAACGGG	Primers specific to L cone exon 5 for the introduction of variants by site directed mutagenesis. Paired with either PE-L-cone-F or PE-L-cone-R.
Var970_E5_F	CCCGTTATCTATGTCATTATGAACCGGCAG	Primers specific to L cone exon 5 for the introduction of variants by site directed mutagenesis. Paired with either PE-L-cone-F or PE-L-cone-R.
Var971_E5_F	CCCGTTATCTATGTCTATATGAACCGGCAG	Primers specific to L cone exon 5 for the introduction of variants by site directed mutagenesis. Paired with either PE-L-cone-F or PE-L-cone-R.
Var973_E5_F	CCCGTTATCTATGTCTTTGTGAACCGGCAG	Primers specific to L cone exon 5 for the introduction of variants by site directed mutagenesis. Paired with either PE-L-cone-F or PE-L-cone-R.
Ex4F	GCAGTCTTACATGATTGTCCTCATGG	Quantitative PCR (qPCR) primers.
In4R	CCTCAAATGAGTCCACTGAGACTCC	qPCR primers.
In4F	ATGCCACCTCCAATTCTAGAACTCC	qPCR primers.
Ex5R	GAAAGAGTCTGAATCCACCCAGAAG	qPCR primers.
Ex5F	CCTACACCTTCTTCGCATGCTTTG	qPCR primers.
In5R	CCACCTGGAATCATGTCTCTGATAAG	qPCR primers.
In5F	AGTCACCATCAACTGCTAAGTCAG	qPCR primers.
Ex6R	GGAAGAAGGTTGACGATGGCTCT	qPCR primers.
EFlaF	AGGTGATTATCCTGAACCATCC	Internal control qPCR primers.
EFlaR	AAAGGTGGATAGTCTGAGAAGC	Internal control qPCR primers.
RPL13aF	CATAGGAAGCTGGGAGCAAG	Internal control qPCR primers.
RPL13aR	GCCCTCCAATCAGTCTTCTG	Internal control qPCR primers.
In1F	GACGAGGTCAACAGATAGAGATCATCC	qPCR primers.
In1R	TTTGAGACAAGAGTCTCACTCTGTGG	qPCR primers.
In2F	CACATCCATATACTGAAGGGTTCTGGAG	qPCR primers.
In2R	GTTGTCTAGCTCTGTATCTGTCTCCTC	qPCR primers.
In3F	GTTTGCATTCCAGGTCTCTTTCTTGC	qPCR primers.
In3R	GGAACGTTCTCGAGGAAAGAACATTC	qPCR primers.
5'-UTR-F	CCACATTCACACTCTGTGACTTAGAG	qPCR primers.

5'-UTR-R	CTTCTGACCTTCCTACCTCGTTTGAAG	qPCR primers.
3'-UTR-F	ACTATCTGTGTGTGCTCTTCAGTTCC	qPCR primers.
3'-UTR-R	CCCACTAATCTGCTTTCTGTCTCTATGG	qPCR primers.
Ex4F	GCAGTCTTACATGATTGTCCTCATGG	qPCR primers.
Ex5R	GAAAGAGTCTGAATCCACCCAGAAG	qPCR primers.
Ex5F	CCTACACCTTCTTCGCATGCTTTG	qPCR primers.
Ex6R	GGAAGAAGGTTGACGATGGCTCT	qPCR primers.
