Supplementary Figure S1- A schematic diagram of mutations in the cone opsin gene array. (A) mutations identified in the current study. (B) a summary of all LIAVA/S related haplotypes reported in the literature to date.

A

Genotype

MOL0110
MOL0432
C203R
MOL0057
LIAVA
MOL0250
LIAVS
MOL0267
LIAVS
MOL0283
V120M
LIAVA
MOL0152

Cone ERG
Normal
Extinguished
Reduced / Normal
Reduced

Diagnosis
Trichromat
BCM
Maculopathy
CRD/CD

B

Genotype

PEDIGREE H
LIAVS
LIAVA
A376
LIAVA
NC
LIAVA
110
ND
LIAVA
RS072630
LIAVA
ND
ND
MF

Phenotype
BCM
Protanope
Deuteranope
Deuteranope
Protanope
MF

Reference
Nathans et al., 1989
Ueyama et al., 2004
Carroll et al., 2004
Neitz et al., 2004
Neitz et al., 2004
Crognale et al., 2004

Note: the order of the second and third genex could not be determined