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| Table S2. LIAVA haplotype and arrays that cause red-green color vision defects | | | | | | | | |
| Subject ID | Age (yr) | Color vision | Genotype | SER  OD | SER  OS | BCVA  OD | BCVA  OS | Photopic  ERG |
| \*MOL0152 III:2 | 5 | ND | LLIAVA - MMVVVA | -5.5 | -6.5 | 20/100 | 20/200 | reduced |
| \*MOL0152 III:3 | 5 | ND | LLIAVA - MMVVVA | -8 | -8.75 | 20/67 | 20/50 | reduced |
| BCM160-23130 | 6 | impaired | LLIAVA - MMVVVA | -3.75 | -5 | 20/67 | 20/40 | reduced |
| \*MOL0152 III:1 | 7 | ND | LLIAVA - MMVVVA | -9.25 | -9.5 | 20/40 | 20/50 | reduced |
| ZD314-18057 | 10 | impaired | LLIAVA - MMVVVA | -6.5 | -6 | 20/100 | 20/100 | reduced |
| JC\_0609 | 11 | Protanope | LLIAVA - MMVVVA | -14 | -13.75 | 20/20 | 20/20 | ND |
| BCM51-12359 | 12 | impaired | LLIAVA - MMVVVA | -14.5 | -14.5 | 20/100 | 20/100 | reduced |
| MM\_0142 | 13 | Protanope | LLIAVA – MMVVVA | -2 | -2 | ND | ND | ND |
| \*MM\_0145 | 15 | Protanope | MLIAVA -  MMVVVA | 0 | 0 | 20/20 | 20/30 | ND |
| \*MM\_0144 | 19 | Protanope | MLIAVA - MMVVVA | -8 | -8.25 | 20/70 | 20/40 | ND |
| JC\_0196 | 28 | Protanope | LLIAVA-MMVVVA-M | -6 | -5 | 20/40 | 20/40 | ND |
| JC\_0195 | 29 | Protanope | LLIAVA-MMVVVA-M | -11.5 | -12 | 20/60 | 20/60 | ND |
| JC\_0084 | 38 | Deuteranope | L - MLIAVA | -2.5 | -2.5 | 20/20 | 20/15 | ND |
| \*MOL0512 III:2, III:3, III:1 are relatives see reference [1] for pedigree; MM\_0145 and MM\_0144 are brothers, JC\_0195 and JC\_0196 are brothers, genotype of MM\_0145 not done, presumed to be same as brother. Subject IDs starting with MOL are from reference [1], those starting with BCM or ZD are from reference [2], those starting with MM or JC are from references [3,4]. ND = not done. | | | | | | | | |

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| Table S1. LIAVA haplotype and arrays that cause blue cone monochromacy | | | | | |
| Subject ID | Age (yr) | Genotype | SER  OD | SER  OS | BCVA |
| \*BCM73-20770 | 0.25 | MLIAVA | -8 | -9 | 20/100 |
| \*BCM73-20537 | 3 | MLIAVA | -4 | -4 | 20/100 |
| S16 | 10 | LLIAVA- LLIAVA-MMVVVA | <-6 D | <-6 D | 20/60 |
| \*MOL0057 III:3 | 12 | MLIAVA | -6 | -6 | 20/160 |
| BCM72-17075 | 12 | LLIAVA-MLIAVA-MMVVVA | 0.5 | 0.5 | 20/200 |
| \*BCM73-17481 | 14 | MLIAVA | -14 | -14 | 20/400 |
| BCM93-19164 | 14 | MLIAVA | -5 | -3 | 20/67 |
| S17 | 18 | LLIAVA- LLIAVA-MMVVVA | <-6 D | <-6 D | 20/40 |
| \*MOL0057 III:I | 20 | MLIAVA | -18 | -18 | 20/320 |
| MM\_0155 | 30 | LLIAVA - MLIAVA | ND | ND | ND |
| \*BCM73-16953 | 51 | MLIAVA | -12 | -12 | 20/133 |
| S21 | 73 | LLIAVA | <-6 D | <-6 D | 20/200 |
| \*BCM73-20770, BCM73-20537, BCM73-16953 are relatives, see ref 6 for pedigree; MOL0057 III:3 and III:1 are relatives, see reference 15 for pedigree. Subjects with IDs starting with BCM are from reference [2], those starting with S are from reference [5], those starting with MOL are from reference [1]. MM\_0155 is from reference [3]. BCM = blue cone monochromacy, XLCD = X-linked cone dysfunction. | | | | | |

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| Table S3. MIAVA haplotype and arrays that cause red-green color vision defects | | | | | |
| Subject ID | Age (yr) | Genotype | SER  OD | SER  OS | BCVA |
| BCM101-19818 | 3 | LLIAVA-MMIAVA | -6 | -6 | 20/200 |
| \*ZD379-19195 | 7 | LLIAVA-MMIAVA | 1 | 1 | 20/125 |
| \*S22 | 9 | LLIAVA- LMIAVA-MMIAVA | <-6 D | <-6 D | 20/125 |
| \*ZD379-19194 | 12 | LLIAVA-MMIAVA | ND | ND | 20/125 |
| \*S23 | 68 | LLIAVA- LMIAVA-MMIAVA | >-3D | >-3D | 20/200 |
|  |  |  |  |  |  |
| \*S24 | 34 | LMIAVA- LMIAVA-MMVVVA | <-6 D | <-6 D | 20/80 |
| \*S25 | 51 | LMIAVA- LMIAVA-MMVVVA | <-6 D | <-6 D | 20/63 |
| Subjects ZD379-19195, -19194 are relatives see pedigree in reference 6. Subjects S22 and S23 are relatives, and S24 and S25 are relatives, see pedigrees in reference [5]. Subjects IDs starting with BCM or ZD are from reference, those starting with S are from reference [2]. | | | | | |

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| Table S4. LVAVA haplotype and red-green color vision deficiency | | | | | | | | | | |
| Subject ID | Age  Yrs | Color Vision | Diag. | Genotype | AL  OD | AL  OS | SER  OD | SER  OS | BCVA | Photopic  ERG |
| BCM194-25474 | 5 | Test not valid | BCM | LLVAVA | ND | ND | -16 | -16.25 | 20/40 | reduced |
| S18 | 10 | impaired | XLCD | LLVAVA | ND | ND | <-6D | <-6D | 20/50 | reduced |
| \*JC\_11445 | 26 | Deuteranope | XLCD | LLVAVA – LMVVVA – M – M | 25.79 | 25.65 | ND | ND | ND | ND |
| \*JC\_0758 | 33 | deuteranomalous | XLCD | LLVAVA – LMVVVA – M – M | 29.18 | 28.53 | -12.75 | -10.00 | 20/50 | ND |
| JC\_0347 | 33 | deuteranope | XLICD | LLVAVA | 24.77 | 24.25 | -3.3 | -2 | 20/200 | ND |
| JC\_0683 | 36 | deuteranomalous | XLCD | LLVAVA – LMVVVA – M – M | 27.03 | 26.70 | -8.5 | -8.5 | 20/15 | ND |
| BCM66-16407 | 41 | impaired | BCM | LLVAVA | ND | ND | -13 | -14 | 20/67 | absent |
| S26 | 80 | Impaired  progressive | XLCD | LLVAVA-LLVAVA-MMVVVA | ND | ND | >-3 D | >-3 D | 20/125 | reduced |
|  |  |  |  |  |  |  |  |  |  |  |
| \*BCM112-22852 | 6 | impaired | BCM/CRD | MLVAVA | ND | ND | -9 | -9.25 | 20/67 | reduced |
| \*JC\_0451 | 12 | Protanope | XLCD | MLVAVA – M – M | 28.1 | 27.6 | -9.5 | -9 | 20/20 | ND |
| \*JC\_0448 | 14 | Protanope | XLCD | MLVAVA – M – M | 27.75 | 27.51 | -11.5 | -10.75 | 20/25 | ND |
| \*BCM112-23518 | 14 | impaired | BCM/CRD | MLVAVA | ND | ND | -24 | -23 | 20/125 | reduced |
| \*JC\_0447 | 17 | Protanope | XLCD | MLVAVA – M – M | 26.54 | 26.52 | -9.75 | -9 | 20/20 | ND |
| \*JC\_10340 | 32 | Protanope | XLCD | MLVAVA – MMVVVA – M | 28.96 | 28.08 | -17.5 | -13 | 20/20 | ND |
| S19 | 40 | impaired | XLCD | MLVAVA | ND | ND | <-6D | <-6D | 20/80 | reduced |
| JC\_0564 | 45 | protanope | XLICD | MLVAVA | 27.08 | 26.58 | -5.8 | -4.9 | 20/100 | reduced |
| S20 | 49 | impaired | XLCD | MLVAVA | ND | ND | <-6D | <-6D | 20/63 | reduced |
| \*JC\_11445 and JC\_11445 are brothers; JC\_0683 and JC\_0758 are cousins: JC\_0451, JC\_0448, and JC\_0447 are brothers and JC\_10340 is their cousin; BCM112-22852 and BCM112-23518 are from the same family. Subject IDs starting with BCM are from reference [2], subject IDs starting with JC\_ are from references [3,4], subject IDs starting with S are from reference [5]. CRD=cone-rod dystrophy. | | | | | | | | | | |

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| Table S5. LVAVA haplotypes and arrays for normal color vision | | | | | | | | | |
| Subject ID | Age  Yrs | Color Vision | Diag. | Genotype | SER  OD | SER  OS | BCVA  OD | BCVA  OS |
| \*BCM133-23364 | 1 | ND | BCM | LLVAVA – MLVAIA | -5 | -5 | ND | ND |
| \*BCM133-20960 | 10 | impaired | BCM | LLVAVA – MLVAIA | -5 | -6 | 20/33 | 20/40 |
| \*BCM133-20961 | 32 | ND | BCM | LLVAVA – MLVAIA | -20.75 | -19 | 20/33 | 20/55 |
|  |  |  |  |  |  |  |  |  |
| \*HM346IV:2 | 15 | Normal | high myopia | LLVAVA – M | -11 | -12.25 | 20/20 | 20/30 |
| \*HM346III:7 | 27 | ND | high myopia | LLVAVA – M | -12.5 | -12.5 | 20/67 | 20/80 |
| \*HM346II:4 | 47 | Normal | high myopia | LLVAVA – M | -13.75 | -14 | 20/200 | 20/125 |
| \*HM346II:2 | 54 | ND | high myopia | LLVAVA – M | -18.25 | -16.25 | 20/200 | 20/100 |
|  |  |  |  |  |  |  |  |  |
| \*HM295 IV4 | 6 | Normal | high myopia | LLVAVA - M | -10.5 | -10.5 | ND | ND |
| \*HM295 IV2 | 12 | Normal | high myopia | LLVAVA - M | -8.25 | -9 | 20/30 | 20/25 |
| \*HM295 IV3 | 12 | Normal | high myopia | LLVAVA - M | -9.25 | -9.25 | 20/40 | 20/40 |
| \*HM295 IV7 | 14 | Normal | high myopia | LLVAVA - M | -9.25 | -10.25 | 20/30 | 20/30 |
| \*HM295 IV1 | 19 | Normal | high myopia | LLVAVA - M | -6 | -6.25 | 20/25 | 20/30 |
| \*HM295 IV5 | 31 | Normal | high myopia | LLVAVA - M | -15 | -15 | 20/30 | 20/40 |
| \*HM295 II:5 | 76 | Normal | high myopia | LLVAVA - M | ND | ND | ND | ND |
|  |  |  |  |  |  |  |  |  |
| BCM126-20616 | 41 | impaired | BCM | LLVAVA-MLVAVA | ND | ND | 20/80 | 20/100 |
| \*Subject IDs beginning with BCM are from the same reference and BCM133- are from the same family, and are from reference [2], subject IDs beginning with HM are from reference [6]. | | | | | | | | | |

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| Table S6. MVAVA haplotype and arrays for normal color vision | | | | | | | | | |
| Subject ID | Age  Yrs | Color  Vision | genotype | AL  OD  OS | OD SER | OS SER | BCVA OD | BCVA OS | Photopic ERG |
| Orosz IV:5 | 40 | ND | LLVAVA – MMVAVA | 25.16  25.51 | -4.25 | -5.25 | 20/20 | 20/20 | ND |
| Orosz IV:5 | 57 | Deuteranomaly | LLVAVA – MMVAVA | ND | NA | NA | 20/25 | 20/40 | ND |
| Orosz IV:5 | 62 | impaired | LLVAVA – MMVAVA | ND | NA | NA | 20/25 | 20/40 | absent |
|  |  |  |  |  |  |  |  |  |  |
| Orosz IV:7 | 51 | impaired | LLVAVA – MMVAVA | 27.82  27.57 | -5.75 | -6.5 | 20/50 | 20/33 | absent |
|  |  |  |  |  |  |  |  |  |  |
| Orosz V:1 | 32 | ND | LLVAVA – MMVAVA | 31.44  30.72 | ND | ND | 20/20 | 20/20 | ND |
| Orosz V:1 | 38 | Impaired | LLVAVA – MMVAVA | ND | -20.0 | -18.0 | 20/20 | 20/20 | absent |
| Orosz V:1 | 46 | impaired | LLVAVA – MMVAVA | 33.11  32.28 | -21.0 | -19.75 | 20/20 | 20/25 | ND |
|  |  |  |  |  |  |  |  |  |  |
| Orosz VI:3 | 42 | impaired | LLVAVA – MMVAVA | 31.35  32.49 | -22.5 | -24.5 | 20/20 | 20/20 | ND |
|  |  |  |  |  |  |  |  |  |  |
| Orosz VI:6 | 11 | normal | LLVAVA – MMVAVA | ND | -6.25 | -6.5 | 20/40 | 20/20 | reduced |
| All subjects are from the same Hungarian family in reference [7]. | | | | | | | | | |

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| Table 8. LIAVS haplotype | | | | | | |
| Subject ID | Age (Yr) | Genotype | SER OD | SER OS | BCVA OD | BCVA OS |
| \*MOL0250 III:2 | 50 | LLIAVS | -2.25 | -1.75 | 20/67 | 20/67 |
| ***\*+MOL0250 IV:3*** | 24 | LLIAVS | -4.00 | -5.50 | 20/67 | 20/67 |
| ***\*+MOL0250 IV:3*** | 27 | LLIAVS | ND | ND | 20/67 | 20/67 |
| ***\*+MOL0250 IV:3*** | 32 | LLIAVS | -2.75 | -5.00 | 20/80 | 20/100 |
| MOL0267 III:1 | 34 | LLIAVS | ND | ND | 20/67 | 20/67 |
| BCM72-16874 | 71 | LLIAVS-MLIAVA-MMVVVA | 1.75 | 0.75 | 20/200 | 20/200 |
| \*members of the same family, see reference [1] for pedigree. +same individual examined over an 8 year period. BCM72-16874 is from reference [2]. | | | | | | |

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