

**S1 Table.** Clinical characteristics of the BCM patients

| Patient/<br>Family <sup>a</sup> | Age at<br>visits (y) | Mutation class: mutation detail                     | Visual acuity <sup>b</sup>  | Refraction <sup>c</sup> |
|---------------------------------|----------------------|---|-----------------------------|-------------------------|
| P1/F1                           | 5,8                  | Deletion: <i>OPN1LW</i> , <i>OPN1MW</i> (partial)   | 20/100                      | -12.50                  |
| P2/F1                           | 8,10                 | Deletion: <i>OPN1LW</i> , <i>OPN1MW</i> (partial)   | 20/80                       | -3.75                   |
| P3/F1                           | 11,13                | Deletion: <i>OPN1LW</i> , <i>OPN1MW</i> (partial)   | 20/63                       | -12.75                  |
| P4/F1                           | 48,50                | Deletion: <i>OPN1LW</i> , <i>OPN1MW</i> (partial)   | 20/100                      | -10.25                  |
| P5/F2                           | 7                    | Deletion: LCR, <i>OPN1LW</i> (partial)              | 20/100                      | -3.25                   |
| P6/F2                           | 12                   | Deletion: LCR, <i>OPN1LW</i> (partial)              | 20/100                      | -5.00                   |
| P7/F3                           | 5,7                  | Deletion: LCR, <i>OPN1LW</i> (partial)              | 20/400 <sup>d</sup> -20/200 | -0.50                   |
| P8/F3                           | 10,19                | Deletion: LCR, <i>OPN1LW</i> (partial)              | 20/100                      | -5.00                   |
| P9/F3                           | 16,25                | Deletion: LCR, <i>OPN1LW</i> (partial)              | 20/100                      | -6.00                   |
| P10/F3                          | 19,28                | Deletion: LCR, <i>OPN1LW</i> (partial)              | 20/125                      | -6.50                   |
| P11-P14 <sup>e</sup>            |                      |   |                             |                         |
| P15/F6                          | 14                   | Deletion: LCR, <i>OPN1LW</i> (partial)              | 20/100                      | -4.00                   |
| P16/F7                          | 28                   | Deletion: LCR, <i>OPN1LW</i> (partial)              | 20/125                      | -8.50                   |
| P17/F8                          | 33                   | Deletion: LCR, <i>OPN1LW</i> (partial)              | 20/100                      | -3.75                   |
| P18/F9                          | 35                   | Deletion: LCR, <i>OPN1LW</i> (partial)              | 20/63                       | -5.50                   |
| P19/F10                         | 43                   | Deletion: LCR, <i>OPN1LW</i> (partial)              | 20/80                       | -6.00                   |
| P20/F11                         | 55                   | Deletion: LCR, <i>OPN1LW</i> (partial)              | 20/80                       | -6.50                   |
| P21/F12                         | 18                   | Deletion: <i>OPN1LW</i> , <i>OPN1MW</i>             | 20/160-20/100               | -8.50                   |
| P22/F12                         | 72                   | Deletion: <i>OPN1LW</i> , <i>OPN1MW</i>             | 20/125-20/100               | -3.00                   |
| P23/F13                         | 30                   | Deletion: LCR                                       | 20/100                      | -10.00                  |
| P24/F14                         | 27                   | Deletion: LCR                                       | 20/100-20/125               | -8.50                   |
| P25/F15                         | 18                   | Deletion: LCR, <i>OPN1LW</i> (partial)              | 20/200-20/125               | -11.25                  |
| P26/F15                         | 24                   | Deletion: LCR, <i>OPN1LW</i> (partial)              | 20/70-20/80                 | -9.25                   |
| P27/F16                         | 39                   | Missense: p.C203R in a single red-green hybrid gene | 20/80                       | -1.00                   |
| P28/F17                         | 13                   | Missense: p.C203R                                   | 20/80                       | -5.50                   |
| P29/F17                         | 35                   | Missense: p.C203R                                   | 20/100                      | -5.25                   |

*OPN1LW*, long-wave-sensitive opsin-1 gene; *OPN1MW*, medium-wave-sensitive opsin-1 gene; LCR, locus control region.

<sup>a</sup>Patients P1-P20 have been described previously [4].

<sup>b</sup>Best corrected visual acuity at most recent visit; similar in the two eyes; otherwise, specified individually, as RE-LE.

<sup>c</sup>Refraction at most recent visit; spherical equivalent; average of both eyes.

<sup>d</sup>Eye with macular coloboma.

<sup>e</sup>Patients P11-P14 are not included because detailed visual function measurements were not performed.