**Appendix A:** Ophthalmic features present in each of the affected family members.

|  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Identity (as per Figure 1) | Laterality | Phenotype | | | | | | |
| **Nystagmus** | **Coloboma** | **Cataract** | **Myopia** | **Pupillary Membrane** | **Microcornea** | **Strabismus** |
| **II.2** | RE | 3 | 1 | 1 | Plano | 0 | 0 | 0 |
| LE | 2 | 1 | +1.00 | 0 | 0 | 0 |
| **II.3** | RE | 2 | 3 | 3 | 25mm | 0 | 0 | 1 |
| LE | 3 | 1 | -6.00 | 1 | 9 | 1 |
| **II.5** | RE | 2 | 3 | 4 | 26mm | 0 | 10 | 0 |
| LE | 4 | 4 | 24mm | 0 | 9 | 0 |
| **II.7** | RE | 0 | N/V | 3 | 22mm | 0 | 0 | 2 |
| LE | 0 | 2 | +4.50 | 0 | 0 | 0 |
| **II.9** | RE | 2 | 4 | 4 | 24mm | 0 | 0 | 0 |
| LE | 4 | 2 | -1.50 | 0 | 0 | 0 |
| **II.11** | RE | 2 | 4 | 4 | -2.50 | 0 | 0 | 0 |
| LE | 4 | 1 | -1.50 | 0 | 0 | 0 |
| **III.1\*** | RE | 1 | 1 | 0 | 24mm | 0 | 0 | 0 |
| LE | 1 | 1 | 24mm | 0 | 0 | 0 |
| **III.5** | RE | 2 | 0 | 0 | 25mm | 0 | 0 | 1 |
| LE | 0 | 0 | 24mm | 0 | 0 | 1 |
| **III.6** | RE | 1 | 1 | 2 | -1.00 | 0 | 0 | 0 |
| LE | 1 | 2 | -2.25 | 1 | 0 | 0 |
| **III.8** | RE | 2 | 2 | 0 | -10.00 | 0 | 0 | 1 |
| LE | 2 | 0 | -9.50 | 0 | 0 | 1 |
| **III.17** | RE | 3 | 2 | 4 | 25mm | 0 | 0 | 0 |
| LE | 3 | 4 | 24mm | 0 | 0 | 0 |
| **III.19\*** | RE | 3 | 4 | 1 | -7.50 | 0 | 0 | 0 |
| LE | 4 | 0 | -11.00 | 0 | 0 | 0 |
| **III.21** | RE | 0 | 2 | 0 | 22mm | 0 | 0 | 0 |
| LE | 2 | 0 | 21.5mm | 0 | 0 | 0 |
| **III.22\*** | RE | 2 | 1 | 4 | 24mm | 0 | 0 | 0 |
| LE | 1 | 1 | -5.00 | 1 | 0 | 0 |
| **III.23** | RE | 2 | 1 | 0 | 21mm | 0 | 0 | 0 |
| LE | 1 | 0 | 21mm | 0 | 0 | 2 |
| **III.25** | RE | 2 | 2 | 0 | -5.50 | 0 | 0 | 0 |
| LE | 2 | 0 | -6.00 | 0 | 0 | 0 |
| **III.27\*** | RE | 2 | 2 | 1 | -11.00 | 0 | 0 | 0 |
| LE | 3 | 1 | -10.00 | 0 | 0 | 0 |
| **IV.1\*** | RE | 1 | 3 | 1 | 22mm | 1 | 9 | 1 |
| LE | 3 | 1 | 22mm | 1 | 9 | 0 |
| **IV.5\*** | RE | 2 | 5 | 3 | 22mm | 0 | 0 | 0 |
| LE | 5 | 3 | 21mm | 0 | 0 | 0 |
| **Key:**  **\*** = Whole genome sequencing   * **RE** = Right Eye, **LE** = Left Eye * **Nystagmus**: 0 = Not present, 1 = subtle, gaze-evoked, 2 = prominent, fast phase in direction of gaze, 3 = Gross, rotatory * **Coloboma**: 0 = Not present, 1 = Large optic disc and cup, 2 = Small disc coloboma, 3 = Large disc coloboma, 4 = Disc and Retinochoroidal colobomas, 5 = Iris and Disc colobomas * **Cataract**: 0 = Not present, 1 = Anterior Polar, 2 = Visually significant, 3 = Dense Cataract, 4 = Pseudophakic or Aphakic * **Myopia**: Spherical equivalent provided where possible, axial length (mm) provided in remaining eyes * **Pupillary Membrane Remnants**: 0 = Not Present, 1 = Present * **Microcornea**: 0 = Vertical diameter > 11.5mm, Vertical diameter (in mm) (where vertical diameter < 11.5mm) * **Tropia**: 0 = Not present, 1 = Esotropia, 2 = Exotropia * **N/V**: No/inadequate view of fundus | | | | | | | | |