Appendix 3. Clinical phenotypes in the six probands with *COL2A1* mutation.

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Family ID | Inheri-  tance patterns | Gen-der | Age at  (year) | | BCVA  (OD/OS) | Myopia refraction errors | | Axis length  (OD/OS mm) | Cata-ract | Vitreous  anoma-  lies | Cause for RD | Age at onset of RD  (year) | Systemic phenotype | | |
| pre-  sent | diag-nosis | OD | OS | Hear-ing loss | Skeletal abnor-malities | Midfacial dysplasia |
| QT1527 | SC | F | 29 | 28 | 0.32/0.12 | -13.75D | -17.50D | 28.56/28.12 | - | Mem. | PRD | 28.0 | - | - | + |
| QT851 | AD | M | 6 | 1.5 | NLP/0.05 | NA | +6.50D\* | 16.70/26.54 | + | NA | GRT | 1.5 | - | - | + |
| QT1589 | AD | F | 6 | 6 | 0.5/0.5 | -3.75D | -2.00D | 23.89/23.78 | - | Mem. | - | - | - | ED | + |
| QT1016 | AD | M | 31 | 27 | 0.08/NLP | -25.25D | NA | 29.84/16.40 | + | Mem. | GRT | 7.0 | - | - | + |
| QT1791 | AD | M | 4 | 3.5 | 0.15/0.15 | -22.00D | -21.50D | 29.96/29.97 | - | Mem. | - | - | - | - | + |
| HM470 | SC | M | 15 | 6 | 0.1/0.1 | -12.25D | -12.50D | NA | - | Mem. | - | - | - | - | + |

Note: SC=sporadic case; AD=autosomal dominant inheritance; M=male; F=female; BCVA=best corrected visual acuity; OD=the right eye; OS=the left eye; NLP=no light perception; NA=not available; Mem.=membranous congenital vitreous anomaly; RD=retinal detachment; PRD=perivascular retinal degeneration; GRT=giant retinal tear; ED=epiphysial dysplasia of both wrists; +=the phenotype exists; -=the phenotype does not exist; \*=aphakic eye and after silicone oil injection.