Appendix 4. Clinical phenotypes of the 10 patients without mutations in *COL2A1* or *COL11A1.*

|  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| 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 | diagno-sis | OD | OS | Hear-ing loss | Skeletal abnor-malities | Midfacial dysplasia |
| QT606 | SC | M | 34 | 27 | 0.1/0.2 | -30.00D | -25.50D | NA | - | B-scan | - | - | - | - | + |
| QT618 | SC | M | 27 | 20 | 0.3/0.3 | -12.00D | -14.75D | 29.57/30.59 | Cor. | B-scan | - | - | - | - | + |
| QT846 | SC | M | 28 | 21 | 1.0/1.0 | -17.00D | -15.00D | 32.55/31.99 | - | Hypo. | - | - | - | - | + |
| QT1172 | AD | M | 6 | 3 | 0.1/0.1 | -17.25D | -14.50D | 29.83/29.35 | Cor. | B-scan | - | - | - | - | + |
| QT1211 | AD | M | 25 | 22 | LP/FC/5cm | NA | NA | NA | All | B-scan | GRT | 15 | Mixed | - | + |
| QT1547 | SC | M | 24 | 24 | 0.2/0.3 | <-20.00D | <-20.00D | 37.23/36.63 | - | Mem. | PRD | 13 | - | - | + |
| QT1553 | SC | F | 7 | 5 | 0.15/0.2 | -14.50D | -15.25D | 30.51/30.71 | - | Mem. | - | - | - | PC | MP |
| HM932 | AD | M | 7 | 6 | 1.0/0.15 | 0D | -12.25D | 22.69/27.16 | - | Hypo. | - | - | - | - | + |
| HM964 | AD | F | 9 | 6 | 0.4/0.5 | -17.00D | -15.37D | 33.91/33.86 | Pos. | B-scan | - | - | - | PO | MP |
| QT1574 | AD | M | 53 | 52 | 0.3/1.0 | <-9.00D | <-9.00D | 35.61/29.92 | Cor. | Hypo. | - | - | - | - | + |

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; LP=light perception; FC=finger count; NA=not available; Cor.=cortical opacity; All=all opacity; Pos.=posterior subcapsular opacities; B-scan=the opacity observed by B-scan; Hypo.=hypoplastic congenital vitreous anomaly; Mem.=membranous congenital vitreous anomaly; RD=retinal detachment; GRT=giant retinal tear; PRD=perivascular retinal degeneration; PC=pigeon chest; PO= precocious osteoarthrosis; +=the phenotype exists; -=the phenotype does not exist; MP= the facial phenotype of bulb eyes, a flat nose and mandibular protrusion.