**Supplementary Table S1: The steps used to filter for autosomal recessive variants in this study.**

| Filtering criteria | Proband/ II-2 | Elder brother/ II-1 |
|---|---|---|
| Homozygous or heterozygous variants in affected individuals[a] | 24162 | 24251 |
| 193 autosomal recessive genes listed in the RetNet database[b] | 388 | 370 |
| Allele frequency of ≤ 0.005 in the public SNP databases[c] | 7 | 4 |
| Two or more variants in one gene | 2 (*CDHR1*) | 2 (*CDHR1*) |

[a]The variants localized in the coding exonic regions and in the intronic regions within 5 bp of the exon-flanking boundaries (± 5 bp) were included.

[b]The reported 193 genes responsible for autosomal recessive inherited retinal dystrophy (accessed 8th October 2017 (https://sph.uth.edu/retnet/)).

[c]The following databases were used: 1000 Genomes database (http://www.1000genomes.org/), Exome Aggregation Consortium database (http://exac.broadinstitute.org/), Human Genetic Variation Database (HGVD) (http://www.genome.med.kyoto-u.ac.jp/SnpDB/), and Tohoku Medical Megabank Organization database (ToMMo) (https://ijgvd.megabank.tohoku.ac.jp/).