Appendix 1. Filtering steps used to select for autosomal recessive variants in the study.

|  |  |  |
| --- | --- | --- |
|  | Patient | |
| Filtering criteria | JU#1303 | JU#1085 |
| Homozygous or heterozygous variants in affected individualsa | 24068 | 23819 |
| 188 autosomal recessive genes listed in the RetNet databaseb | 365 | 362 |
| Non-homozygous variants in parents | 163 | 155 |
| Allele frequency of ≤ 0.005 in the public SNP databasesc | 9 | 7 |
| Two or more variants in one gene | 4（*RPE65* and *NBAS*) | 4（*RPE65* and *ALMS1*) |

a Variants localized in coding exonic regions and in intronic regions within 5 bp of the exon-flanking boundaries (±5 bp), were included.

b 188 genes have been reported to be responsible for autosomal

recessive inherited retinal disease. Accessed 23th August 2017 (https://sph.uth.edu/retnet/).

c 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/).