Appendix 2. Filtering steps used to select for de novo dominant variants in the study.

|  |  |  |
| --- | --- | --- |
|  | Patient | |
| Filtering criteria | JU#1303 | JU#1085 |
| Heterozygous variants in affected individualsa | 14304 | 13764c |
| 75 autosomal dominant genes listed in the RetNet databaseb | 85 | 53d |
| No variants in parents | 0 | 0 |

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

b 76 genes have been reported to be responsible for autosomal dominant inherited retinal disease. Accessed 23th August 2017 (https://sph.uth.edu/retnet/).

c JU1085 patient is male, therefore including hemizygous variants.

d Additionally, we screened variants in the known 13 X-linked inherited retinal dystrophy genes.