|  |  |  |  |  |  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- | --- |
| Supplementary Table 6. Variant types of genes in families with different diseases. | | | | | | | |  |  |  |  |  |
| Gene | ACHM | | | CORD | | | OCT | | | LCA | | |
|  | Trun+Trun | Trun+Mis | Mis+Mis | Trun+Trun | Trun+Mis | Mis+Mis | Trun+Trun | Trun+Mis | Mis+Mis | Trun+Trun | Trun+Mis | Mis+Mis |
| CNGA3 | 17 | 34 | 225 | 6 | 18 | 31 | 0 | 1 | 2 | 0 | 0 | 2 |
| CNGB3 | 748 | 40 | 12 | 13 | 1 | 3 | 1 | 0 | 0 | 2 | 0 | 0 |
| GNAT2 | 9 | 1 | 0 | 2 | 0 | 2 | 0 | 0 | 0 | 0 | 0 | 0 |
| PDE6C | 15 | 5 | 10 | 5 | 1 | 3 | 0 | 0 | 0 | 0 | 0 | 1 |
| ATF6 | 9 | 0 | 3 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| PDE6H | 2 | 0 | 0 | 1 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
|  |  |  |  |  |  |  |  |  |  |  |  |  |
|  |  |  |  |  |  |  |  |  |  |  |  |  |
| Gene | Unclassified IRD | | | RP | | | CSNB | | | MD | | |
|  | Trun+Trun | Trun+Mis | Mis+Mis | Trun+Trun | Trun+Mis | Mis+Mis | Trun+Trun | Trun+Mis | Mis+Mis | Trun+Trun | Trun+Mis | Mis+Mis |
| CNGA3 | 2 | 3 | 16 | 1 | 0 | 3 | 0 | 1 | 0 | 0 | 0 | 0 |
| CNGB3 | 9 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 1 | 1 | 2 |
| GNAT2 | 1 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| PDE6C | 1 | 1 | 1 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |
| ATF6 | 1 | 0 | 1 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 1 |
| PDE6H | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 |