**Table S4. Summary of compound heterozygous variants per proband, before and after filtration.**

|  |  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- | --- |
| **Patient** | **Compound heterozygous variants** | **Compound heterozygous variants \*** | **Compound heterozygous variants \*\*** | **Validated a compound heterozygous variants** | **Validated a compound heterozygous variants that are true compound heterozygotes** | **Validated a true compound heterozygous variants that segregate with disease** | **Corresponding number of genes** |
| AU070811 | 72 | 44 (61%) | 35 (80%) | 23 (66%) | 4 | 4 | 2 |
| AU035204 | 68 | 45 (66%) | 37 (82%) | 26 (70%) | 4 | 4 | 2 |
| AU081204 | 68 | 48 (71%) | 46 (96%) | 32 (70%) | 0 | 0 | 0 |
| AU075308 | 54 | 37 (69%) | 32 (86%) | 20 (62%) | 8 | 6 | 3 |
| AU1328302 | 72 | 49 (68%) | 43 (88%) | 31 (72%) | 2 | 0 | 0 |
| AU1261301 | 82 | 45 (55%) | 39 (87%) | 27 (69%) | 6 | 0 | 0 |
| AU1353302 | 80 | 58 (72%) | 55 (95%) | 39 (71%) | 4 | 2 | 1 |
| AU1252302 | 56 | 28 (50%) | 23 (82%) | 19 (83%) | 2 | 0 | 0 |
| AU037103 | 40 | 26 (65%) | 25 (96%) | 14 (56%) | 2 | 2 | 1 |
| AU1019301 | 58 | 29 (50%) | 25 (86%) | 20 (80%) | 6 | 4 | 2 |
| AU1388301 | 70 | 50 (71%) | 43 (86%) | 37 (86%) | 2 | 0 | 0 |
| AU1196301 | 74 | 56 (76%) | 52 (93%) | 41 (79%) | 4 | 0 | 0 |
| AU022203 | 90 | 67 (74%) | 63 (94%) | 32 (51%) | 2 | 2 | 1 |
| AU000504 | 54 | 36 (67%) | 33 (92%) | 21 (64%) | 8 | 0 | 0 |
| AU039903 | 66 | 49 (74%) | 42 (86%) | 20 (48%) | 6 | 6 | 3 |
| AU062504 | 68 | 41 (60%) | 33 (80%) | 23 (70%) | 1 | 0 | 0 |

For each proband, variants that are candidates for being compound heterozygotes were validated. Parental genotypes were used for segregation analysis to determine which variants are true compound heterozygotes.

\* Successful Sequenom design

\*\* Successful Sequenom run

a Variants validated by Sequenom analysis