**Suppl Table 1.** ExpansionHunter results in the family. 24 genes were analyzed for pathogenic repeat expansions.

|  |  |  |  |  |  |  |
| --- | --- | --- | --- | --- | --- | --- |
| **CHR** | **Position** | **ref** | **Gene** | **Repeat unit** | **Repeats risk cut-off** | **N patients with repeats > risk cut-off** |
| 3 | 63898360 | G | ATXN7 | GCA | 37 | 0 |
| 3 | 128891419 | A | CNBP | CAGG | 75 | 0 |
| 4 | 3076603 | C | HTT | CAG | 36 | 0 |
| 4 | 41747989 | A | PHOX2B | GCN | 24 | 0 |
| 5 | 146258290 | A | PPP2R2B | GCT | 55 | 0 |
| 6 | 16327864 | G | ATXN1 | TGC | 39 | 0 |
| 6 | 170870994 | G | TBP | GCA | 47 | 0 |
| 9 | 27573526 | C | C9ORF72 | GGCCCC | 30 | 0 |
| 9 | 71652202 | A | FXN | GAA | 66 | 0 |
| 12 | 7045879 | C | ATN1 | CAG | 49 | 0 |
| 12 | 50898784 | T | DIP2B | GGC | 75 | 0 |
| 12 | 112036753 | G | ATXN2 | GCT | 32 | 0 |
| 13 | 70713515 | A | ATXN8OS | CTG | 74 | 0 |
| 14 | 92537353 | C | ATXN3 | GCT | 61 | 0 |
| 16 | 87637893 | C | JPH3 | CTG | 66 | 0 |
| 18 | 53253386 | G | TCF4 | CAG | 40 | 0 |
| 19 | 13318672 | C | CACNA1A | CTG | 21 | 0 |
| 19 | 46273462 | C | DMPK | CAG | 50 | 0 |
| 20 | 2633379 | G | NOP56 | GGCCTG | 650 | 0 |
| 21 | 45196324 | G | CSTB | CGCGGGGCGGGG | 40 | 0 |
| 22 | 46191234 | G | ATXN10 | ATTCT | 500 | 0 |
| X | 66765158 | T | AR | GCA | 38 | 0 |
| X | 146993568 | G | FMR1 | CGG | 200 | 0 |
| X | 147582151 | A | AFF2 | GCC | 200 | 0 |