Supplementary Figure 1: Consensus sequences of the GGC repeat expansions of \textit{NOTCH2NLC} in three index patients.
Supplementary Table 1. Top 30 candidate genes in three index patients by LRS and STR-Scoring method.

| Num. | PATTERN | Gene             | F1-III-5 | S1     | F1-IV-5 | Healthy Individuals ERC Range | Healthy Individuals Cubic Mean | Score |
|------|---------|------------------|----------|--------|---------|-------------------------------|-------------------------------|-------|
| 1    | GGC     | NOTCH2NLC        | 203/0.00 | 306/0.00 | 68/0.00 | 11-36                          | 23.58                         | 4.77  |
| 2    | TTCC    | TRPM2            | 902/0.00 | 153/0.67 | 1101/0.00 | 3-598                         | 336.68                        | 0.88  |
| 3    | TCTC    | PRDM16           | 260/0.09 | 311/0.00 | 290/0.01 | 76-299                        | 140.62                        | 0.67  |
| 4    | TCCT    | LYRM4-AS1        | 145/0.03 | 153/0.03 | 122/0.06 | 49-174                        | 75.47                         | 0.55  |
| 5    | CCCT    | PCBP3            | 104/0.02 | 111/0.01 | 106/0.02 | 63-117                        | 72.93                         | 0.46  |
| 6    | GGGA    | CELSR1           | 647/0.01 | 724/0.01 | 581/0.06 | 45-774                        | 399.98                        | 0.41  |
| 7    | GAA     | EYS              | 1895/0.02 | 1248/0.24 | 1868/0.03 | 21-2184                     | 1111.32                        | 0.32  |
| 8    | TCTC    | JAKMIP3          | 936/0.00 | 988/0.00 | 147/0.92 | 24-859                        | 571.26                        | 0.30  |
| 9    | CCCT    | BOD1             | 85/0.06  | 114/0.02 | 105/0.03 | 45-122                        | 61.28                         | 0.28  |
| 10   | TATA    | STK32B           | 135/0.03 | 137/0.02 | 139/0.00 | 56-137                        | 107.55                        | 0.27  |
| 11   | ACTAAC  | TMEM132D         | 159/0.02 | 160/0.02 | 58/0.75  | 17-167                         | 82.48                         | 0.27  |
| 12   | TCCC    | AP2A2            | 162/0.00 | 136/0.06 | 49/0.08  | 6-146                         | 57.58                         | 0.23  |
| 13   | TCCC    | CNOT3;PRPF31     | 954/0.00 | 146/0.34 | 130/0.42 | 3-920                         | 320.31                        | 0.22  |
| 14   | CCTCT   | LINC02492        | 158/0.04 | 169/0.04 | 160/0.04 | 60-181                         | 134.13                        | 0.20  |
| 15   | TATA    | NBDY             | 109/0.01 | 101/0.04 | 102/0.04 | 0-110                          | 87.28                         | 0.19  |
| 16   | AGTGG   | TMEM132B         | 310/0.10 | 319/0.03 | 299/0.15 | 36-415                         | 192.46                        | 0.18  |
| 17   | CCTCT   | ROHOBTB1         | 150/0.02 | 144/0.14 | 149/0.02 | 18-167                         | 118.00                        | 0.16  |
| 18   | CCT     | CAMTA1           | 632/0.00 | 586/0.00 | 602/0.00 | 327-580                        | 524.31                        | 0.16  |
| 19   | ACT     | CTNND2           | 157/0.00 | 152/0.15 | 155/0.02 | 40-155                         | 125.36                        | 0.15  |
| 20   | TGTGGAGGGG | TRPM2       | 128/0.03 | 127/0.03 | 36/0.71  | 14-177                         | 83.89                         | 0.15  |
|   | Sequence | Gene            |峭点 | 峭点 | 峭点 | 峭点 |
|---|----------|----------------|------|------|------|------|
| 21| GAG      | DNAH17         | 430/0.03 | 415/0.05 | 478/0.00 | 82-453 |
| 22| ATAT     | RGS20          | 309/0.00 | 305/0.00 | 302/0.00 | 16-289 |
| 23| TTATA    | FAM230E        | 562/0.00 | 244/0.51 | 365/0.15 | 12-554 |
| 24| CTCC     | RXRA           | 116/0.00 | 67/0.83  | 105/0.02 | 50-150 |
| 25| TTCC     | ZBTB34         | 360/0.00 | 64/0.87  | 63/0.89  | 0-161  |
| 26| TTC      | DMD            | 197/0.01 | 26/0.11  | 22/0.62  | 0-233  |
| 27| ACAC     | OSGIN1         | 216/0.04 | 139/0.80 | 217/0.04 | 0-249  |
| 28| TCCT     | SYN3           | 228/0.01 | 227/0.02 | 168/0.71 | 150-231|
| 29| CCCTC    | PKD1P1;PKD1P4-NPIPA8 | 10/0.04 | 178/0.04 | 6/0.35  | 0-228  |
| 30| ATATATT  | FAM230J        | 138/0.88 | 209/0.03 | 216/0.01 | 3-272  |