

**Table S4 – List of 80 Synonymous and Missense variants that have been experimentally tested in mini-gene constructs and do not cause changes in splicing.** This group consists of variants that occur both naturally (i.e. SNPs) [152] or have been created artificially by mutagenesis [73,103,153]. References are in the main text of the paper.

| <b>SNP No.</b> | <b>Chr</b> | <b>Position</b> | <b>Gene</b>       | <b>Potential Coding Effect</b> | <b>Mutation (Protein)</b> | <b>Mutation (DNA)</b> | <b>Reference</b> |
|----------------|------------|-----------------|-------------------|--------------------------------|---------------------------|-----------------------|------------------|
| 1              | 21         | 21668058        | <b>NCAM2</b>      | Missense                       | L->P                      | T->C                  | [152]            |
| 2              | 5          | 13882799        | <b>DNAH5</b>      | Synonymous                     | A->A                      | C->T                  | [152]            |
| 3              | 12         | 103233689       | <b>TXNRD1</b>     | Synonymous                     | L->L                      | C->T                  | [152]            |
| 4              | 1          | 181451239       | <b>LAMC2</b>      | Synonymous                     | S->S                      | C->T                  | [152]            |
| 5              | 1          | 27552384        | <b>SYTL1</b>      | Missense                       | Q->R                      | A->G                  | [152]            |
| 6              | 2          | 98516378        | <b>INPP4A</b>     | Synonymous                     | T->T                      | G->A                  | [152]            |
| 7              | 2          | 24378462        | <b>ITSN2</b>      | Missense                       | V->I                      | G->A                  | [152]            |
| 8              | 12         | 27758994        | <b>MRPS35</b>     | Missense                       | G->R                      | G->A                  | [152]            |
| 9              | 4          | 9531265         | <b>SLC2A9</b>     | Missense                       | V->I                      | G->A                  | [152]            |
| 10             | 4          | 96325345        | <b>UNC5C</b>      | Missense                       | M->T                      | T->C                  | [152]            |
| 11             | 15         | 84607033        | <b>AGBL1</b>      | Synonymous                     | D->D                      | C->T                  | [152]            |
| 12             | 1          | 17174367        | <b>MFAP2</b>      | Synonymous                     | H->H                      | T->C                  | [152]            |
| 13             | 2          | 218976025       | <b>CTDSP1</b>     | Synonymous                     | H->H                      | C->T                  | [152]            |
| 14             | 16         | 83480360        | <b>CRISPLD2</b>   | Synonymous                     | A->A                      | C->T                  | [152]            |
| 15             | 18         | 75574115        | <b>CTDP1</b>      | Missense                       | T->M                      | C->T                  | [152]            |
| 16             | 5          | 38406294        | <b>EGFLAM</b>     | Missense                       | W->R                      | T->C                  | [152]            |
| 17             | 12         | 108102248       | <b>ACACB</b>      | Synonymous                     | D->D                      | T->C                  | [152]            |
| 18             | 22         | 29187448        | <b>SEC14L3</b>    | Synonymous                     | D->D                      | T->C                  | [152]            |
| 19             | 12         | 116154297       | <b>NOS1</b>       | Synonymous                     | D->D                      | C->T                  | [152]            |
| 20             | 11         | 68462250        | <b>IGHMBP2</b>    | Missense                       | T->K                      | C->A                  | [152]            |
| 21             | 2          | 160516321       | <b>PLA2R1</b>     | Missense                       | G->S                      | G->A                  | [152]            |
| 22             | 7          | 158520087       | <b>VIPR2</b>      | Synonymous                     | N->N                      | C->T                  | [152]            |
| 23             | 7          | 21565758        | <b>DNAH11</b>     | Synonymous                     | N->N                      | C->T                  | [152]            |
| 24             | 17         | 68922486        | <b>SDK2</b>       | Synonymous                     | T->T                      | G->A                  | [152]            |
| 25             | 10         | 102255805       | <b>SEC31B</b>     | Synonymous                     | H->H                      | T->C                  | [152]            |
| 26             | 17         | 70121684        | <b>CD300E</b>     | Missense                       | G->R                      | G->A                  | [152]            |
| 27             | 15         | 39953792        | <b>SPTBN5</b>     | Missense                       | R->H                      | G->A                  | [152]            |
| 28             | 4          | 154733172       | <b>KIAA0922</b>   | Synonymous                     | H->H                      | C->T                  | [152]            |
| 29             | 3          | 133701313       | <b>DNAJC13</b>    | Missense                       | A->S                      | G->T                  | [152]            |
| 30             | 2          | 200962201       | <b>LOC26010</b>   | Synonymous (5' UTR)            | NA                        | A->G                  | [152]            |
| 31             | 5          | 41036100        | <b>AC114967.2</b> | Synonymous                     | Y->Y                      | C->T                  | [152]            |
| 32             | 12         | 47351994        | <b>C12orf41</b>   | Synonymous                     | T->T                      | A->G                  | [152]            |
| 33             | 10         | 113925369       | <b>GPAM</b>       | Missense                       | E->G                      | A->G                  | [152]            |
| 34             | 3          | 113403806       | <b>SLC9A10</b>    | Missense                       | S->I                      | G->T                  | [152]            |
| 35             | 3          | 155501581       | <b>DHX36</b>      | Synonymous                     | S->S                      | G->C                  | [152]            |
| 36             | 4          | 87925530        | <b>PTPN13</b>     | Missense                       | Y->D                      | T->G                  | [152]            |
| 37             | 15         | 38443137        | <b>DISP2</b>      | Missense                       | P->A                      | C->G                  | [152]            |
| 38             | 9          | 83456967        | <b>TLE1</b>       | Synonymous                     | E->E                      | A->G                  | [152]            |
| 39             | X          | 14797068        | <b>FANCB</b>      | Synonymous (5' UTR)            | NA                        | C->G                  | [152]            |
| 40             | 6          | 41106119        | <b>UNC5CL</b>     | Synonymous                     | C->C                      | C->T                  | [152]            |
| 41             | 11         | 60422917        | <b>PRPF19</b>     | Synonymous                     | S->S                      | C->T                  | [152]            |
| 42             | 11         | 85646271        | <b>EED</b>        | Synonymous                     | L->L                      | C->T                  | [152]            |
| 43             | 3          | 37042476        | <b>MLH1</b>       | Missense                       | K->N                      | G->T                  | [153]            |
| 44             | 3          | 37042191        | <b>MLH1</b>       | Synonymous                     | L->L                      | G->T                  | [153]            |
| 45             | 3          | 37042232        | <b>MLH1</b>       | Synonymous                     | A->A                      | C->T                  | [153]            |
| 46             | 3          | 37042239        | <b>MLH1</b>       | Missense                       | Q->H                      | G->C                  | [153]            |
| 47             | 3          | 37042240        | <b>MLH1</b>       | Missense                       | M->L                      | A->T                  | [153]            |
| 48             | 3          | 37042297        | <b>MLH1</b>       | Missense                       | K->Q                      | A->C                  | [153]            |
| 49             | 3          | 37042335        | <b>MLH1</b>       | Missense                       | E->D                      | G->C                  | [153]            |
| 50             | 3          | 37042363        | <b>MLH1</b>       | Missense                       | A->S                      | G->C                  | [153]            |
| 51             | 3          | 37042376        | <b>MLH1</b>       | Missense                       | D->V                      | A->T                  | [153]            |

|    |    |           |             |            |      |      |       |
|----|----|-----------|-------------|------------|------|------|-------|
| 52 | 3  | 37042406  | <b>MLH1</b> | Missense   | A->V | C->T | [153] |
| 53 | 3  | 37042432  | <b>MLH1</b> | Synonymous | L->L | T->C | [153] |
| 54 | 3  | 37042454  | <b>MLH1</b> | Missense   | G->G | G->C | [153] |
| 55 | 7  | 116975945 | <b>CFTR</b> | Missense   | L->F | A->C | [103] |
| 56 | 7  | 116975973 | <b>CFTR</b> | Missense   | N->D | A->G | [103] |
| 57 | 7  | 116976002 | <b>CFTR</b> | Missense   | S->R | C->A | [103] |
| 58 | 7  | 116976048 | <b>CFTR</b> | Missense   | D->N | G->A | [103] |
| 59 | 7  | 116976081 | <b>CFTR</b> | Missense   | L->M | T->A | [103] |
| 60 | 7  | 116976081 | <b>CFTR</b> | Synonymous | L->L | T->C | [103] |
| 61 | 7  | 116976083 | <b>CFTR</b> | Missense   | L->F | G->C | [103] |
| 62 | 7  | 116976087 | <b>CFTR</b> | Missense   | V->F | G->T | [103] |
| 63 | 17 | 26689881  | <b>NF1</b>  | Missense   | Y->N | T->A | [73]  |
| 64 | 17 | 26689881  | <b>NF1</b>  | Missense   | Y->H | T->C | [73]  |
| 65 | 17 | 26689881  | <b>NF1</b>  | Missense   | Y->D | T->G | [73]  |
| 66 | 17 | 26689882  | <b>NF1</b>  | Missense   | Y->S | A->C | [73]  |
| 67 | 17 | 26689882  | <b>NF1</b>  | Missense   | Y->C | A->G | [73]  |
| 68 | 17 | 26689882  | <b>NF1</b>  | Missense   | Y->F | A->T | [73]  |
| 69 | 17 | 26689884  | <b>NF1</b>  | Missense   | N->H | A->C | [73]  |
| 70 | 17 | 26689884  | <b>NF1</b>  | Missense   | N->D | A->G | [73]  |
| 71 | 17 | 26689884  | <b>NF1</b>  | Missense   | N->Y | A->T | [73]  |
| 72 | 17 | 26689885  | <b>NF1</b>  | Missense   | N->T | A->C | [73]  |
| 73 | 17 | 26689885  | <b>NF1</b>  | Missense   | N->S | A->G | [73]  |
| 74 | 17 | 26689885  | <b>NF1</b>  | Missense   | N->I | A->T | [73]  |
| 75 | 17 | 26689887  | <b>NF1</b>  | Missense   | S->R | A->C | [73]  |
| 76 | 17 | 26689887  | <b>NF1</b>  | Missense   | S->G | A->G | [73]  |
| 77 | 17 | 26689887  | <b>NF1</b>  | Missense   | S->C | A->T | [73]  |
| 78 | 17 | 26689888  | <b>NF1</b>  | Missense   | S->N | G->A | [73]  |
| 79 | 17 | 26689888  | <b>NF1</b>  | Missense   | S->I | G->T | [73]  |
| 80 | 17 | 26689888  | <b>NF1</b>  | Missense   | S->T | G->C | [73]  |