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Supplemental Data

βIV Spectrinopathies Cause Profound Intellectual Disability, Congenital Hypotonia, and Motor Axonal Neuropathy

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Supplemental Fig. 1. Alignment of human pathogenic *SPTBN4* variants with and their equivalent locations in mouse *Sptbn4*.

Gln605*

```
Human   599   LRFSQLQGYQPCDPQVICNRVNHVHGCLAELQEQAARRRAELEASRSLWALLQELEEAES  658
              LRFSQLQGYQPCDPQVICNRVNHVHGCL+ELQEQAARRRAELEASRSLWALLQELEEAES
Mouse   600   LRFSQLQGYQPCDPQVICNRVNHVHGCLSELQEQAARRRAELEASRSLWALLQELEEAES  659
```

Trp903*

```
Human   899   EKEQWLLSMRVPDSLDDVEVVQHRFESLDQEMNSLMGRVLDVNHTVQELVEGGHPSSDEV  958
              EKEQWLL+MRVPDSLDDVEVVQHRFESLDQEMNSLMGRVLDVN TVQELVEGGHPSSDEV
Mouse   895   EKEQWLLAMRVPDSLDDVEVVQHRFESLDQEMNSLMGRVLDVNQTVQELVEGGHPSSDEV  954
```

Glu1274*

```
Human   1259  VQAAEGLLRQGNIYGEQAQEAVTRLLEKNQENQLRAQQWMQKLHDQLELQHFLRDCHELD  1318
              VQAAE LLRQGN YGEQAQEAV RLLEK+QENQLRAQQWMQKL DQL LQHFLRDCHELD
Mouse   1255  VQAAESLLRQGNAYGEQAQEAVARLLEKSQENQLRAQQWMQKLLDQLVLQHFLRDCHELD  1314
```

Gln1277Argfs*4

```
Query   1259  VQAAEGLLRQGNIYGEQAQEAVTRLLEKNQENQLRAQQWMQKLHDQLELQHFLRDCHELD  1318
              VQAAE LLRQGN YGEQAQEAV RLLEK+QENQLRAQQWMQKL DQL LQHFLRDCHELD
Sbjct   1255  VQAAESLLRQGNAYGEQAQEAVARLLEKSQENQLRAQQWMQKLLDQLVLQHFLRDCHELD  1314
```

Arg504Gln

```
Human   479   AAYEERVQGVAELAQALAAEGYYDIRRVAAQRDSVLRQWALLTGLVGARRTRLEQNLALQ  538
              AAYEERVQGVAELAQALAAEGYYD RRVAAQRDSVLRQWALLTGLVGARRTRLEQNLALQ
Mouse   480   AAYEERVQGVAELAQALAAEGYYDARRVAAQRDSVLRQWALLTGLVGARRTRLEQNLALQ  539
```

Arg2435Cys

```
Human   2398  SRSAPAQGGSAPAPPPPPTHTVQHEGFLLRKRELDANRKSSNRSWVSLYCVLSKGELGFY  2457
              SRSAPAQGGSAPAPPPPPTHTVQHEGFLLRKRELDANRKSSNRSWVSLYCVLSKGELGFY
Mouse   2395  SRSAPAQGGSAPAPPPPPTHTVQHEGFLLRKRELDANRKSSNRSWVSLYCVLSKGELGFY  2454
```

Ala2485Leufs*31

```
Human   2458  KDSKGPASGSTHGGEPLLSLHKATSEVASDYKKKKHVFKLQTQDGSEFLLQAKDEEEMNG  2517
               KDSKGPASG THGGEPLLSLHKATSEVASDYKKKKHVFKLQTQDGSEFLLQAKDEEEMNG
Mouse   2455  KDSKGPASGGTHGGEPLLSLHKATSEVASDYKKKKHVFKLQTQDGSEFLLQAKDEEEMNG  2514
```