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## Supplemental Data

# Loss-of-Function Mutations in *FRRS1L*

# Lead to an Epileptic-Dyskinetic Encephalopathy

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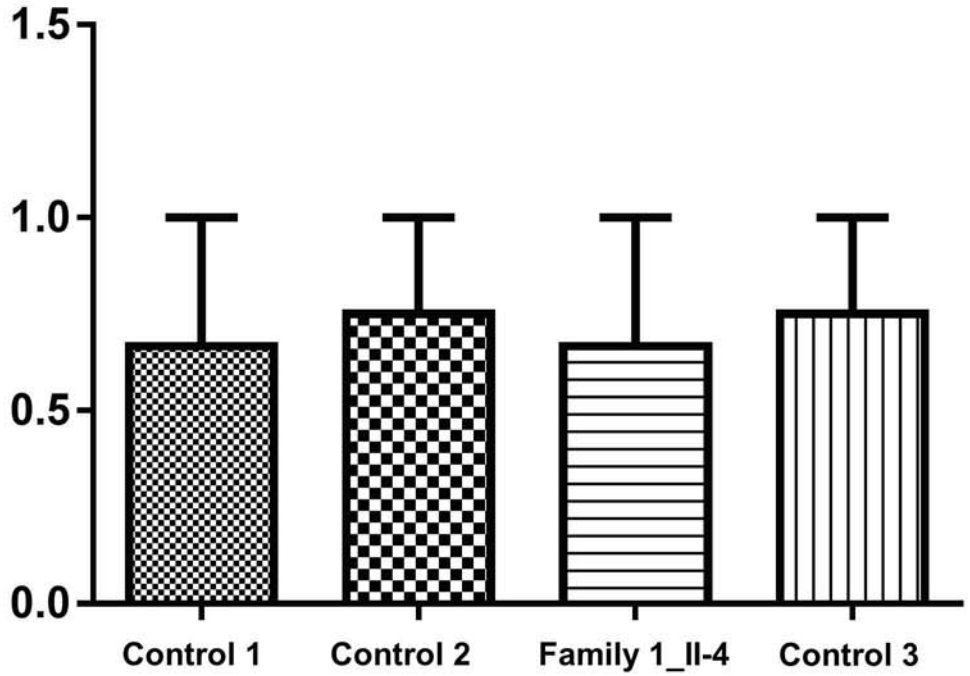


**Figure S1. RT-PCR shows similar amounts of *FRRS1L* transcript in c.961C>T affected cells when compared to controls (mean ± SEM from n = 3 experiments)**

**Table S1. Regions of homozygosity shared by index family members**

| Chromosome | Start | End | Size (Mb) |
|---|---|---|---|
| **1** | 147,779,938 | 151,347,700 | 3.5 |
| **3** | 62,613 | 4,878,792 | 4.8 |
| **9** | 104,622,396 | 119,149,440 | 14.5 |
| **16** | 31,901,547 | 35,220,544 | 3.3 |
| **X** | 61,932,503 | 65,386,406 | 3.5 |

**Table S2. Rare heterozygous *FRRS1L* sequence variants identified in a cohort of >500 individuals with neurodegenerative disease**

| *FRRS1L* variant | ESP6500 | ExAc | Phenotype |
|---|---|---|---|
| **c.469T>A; (p.C157S)** | - | 8.13E-06 | Ataxia, optic nerve atrophy |
| **c.469T>A; (p.C157S)** | - | 8.13E-06 | Ataxia, intellectual disability |
| **c.721delG; (p.A241fs)** | - | 8.13E-06 | Multiple system atrophy |
| **c.686T>C; (p.V229A)** | 0.000231 | 7.40E-04 | Ataxia |
| **c.885A>G; (p.I295M)** | 0.000077 | 1.63E-05 | Multiple system atrophy |