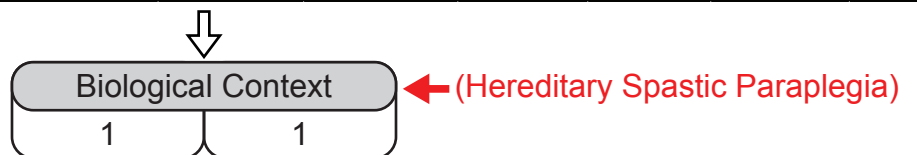


| Chromosome | Position | Gene Region | Gene Symbol | Transcript ID | Transcript Variant | Protein Variant | Translation Impact | SIFT Function Prediction | PolyPhen-2 Function Prediction | Regulatory Site |
|------------|-----------|-------------|-----------------|--------------------------------|--------------------|-----------------|--------------------|--------------------------|--------------------------------|------------------|
| 10 | 104852895 | Splice Site | <i>NT5C2</i> | NM_012229.4; NM_001134373.2 | c.1159+1G>T | | | | | Splice Site Loss |
| 20 | 25456907 | Exonic | <i>NINL</i> | NM_025176.4 | c.3020delC | p.P1007fs*43 | frameshift | | | |
| 20 | 36855621 | Exonic | <i>KIAA1755</i> | NM_001029864.1 | c.1987C>T | p.R663W | missense | Damaging | Possibly Damaging | |



| Chromosome | Position | Gene Region | Gene Symbol | Transcript ID | Transcript Variant | Protein Variant | Translation Impact | SIFT Function Prediction | PolyPhen-2 Function Prediction | Regulatory Site |
|------------|-----------|-------------|--------------|--------------------------------|--------------------|-----------------|--------------------|--------------------------|--------------------------------|------------------|
| 10 | 104852895 | Splice Site | <i>NT5C2</i> | NM_012229.4; NM_001134373.2 | c.1159+1G>T | | | | | Splice Site Loss |

| Gene | Allele Frequency in 108 Qatari Genomes | | | |
|-----------------|--|--------------|--------------------------|---------|
| | Homozygous for Variant | Heterozygous | Homozygous for Reference | No Call |
| <i>NT5C2</i> | 0 | 0 | 108 | 0 |
| <i>NINL</i> | 0 | 0 | 108 | 0 |
| <i>KIAA1755</i> | 0 | 4 | 104 | 0 |

Figure S1: IVA analysis of WGS data. Filters applied (see methods) revealed 3 variants in 3 genes (NT5C2, NINL and KIAA1755) at the genetic analysis filter that showed in IVA to be compatible with Mendelian recessive inheritance (upper panel). Detailed description of those 3 variants is shown in the upper table. Frequency of those 3 genetic variants in the Qatari population is shown in the bottom table with KIAA1755 variant detected as heterozygous at a frequency of 3.7% in Qatari genomes. When biological context filter is applied and hereditary spastic paraplegia is specified as the disease, the two variants, NINL and KIAA1755 ones, are filtered out leading to one single variant in *NT5C2* (middle table).