SUPPLEMENTARY FILE

Table S1. Exome sequencing summary data

	Case 1	**Case 5**	**Case 9**
Total reads	105 898 538	140 882 234	120 519 058
Total aligned reads	101 729 359	139 641 721	119 375 959
Mean read length	93.05	97.99	96.92
Mean coverage	107.01X	149.8X	131.45X
Coverage in CCDS region			
5X	97.70%	97.90%	96.90%
10X	97.00%	97.40%	95.20%
20X	94.80%	96.20%	91.80%
30X	91.20%	94.40%	88.50%

Table S2. Filtering strategy employed to identify the causative variants in *SPG7*

	Case 1	**Case 5**	**Case 9**
Total variants	195 088	285 572	278 372
Non-synonymous/splicing/indel variants	11 876	11 756	11 955
After excluding variants reported in 1000genomes and EVS datasets (frequency >3%)	1 878	1 761	1 926
After excluding variants observed in our in-house database (frequency >1.5%)	484	357	430
Genes with homozygous or compound heterozygous variants	43	18	24
Genes with variants not homozygous or compound heterozygous in our in-house database	22	12	14
Gene associated with an ataxia phenotype	1	2*	1

**SYNE1* and *SPG7*

Table S3. Genotypes of SNPs in patients with the p.(Ala510Val) mutation in *SPG7*

SNP ID	**rs11076708**	**rs564705**	**rs12447947**	**A510V**	**rs258327**	**rs461115**	**rs7200990**	**rs3809643**	**rs7196459**
Position	**88 808 141**	**88 946 955**	**89 199 651**	**89 613 145**	**89 629 218**	**89 703 519**	**89 836 507**	**90 124 273**	**90 141 477**
Case 1	C	A	G	C	G	T	T	T	G
	C	G	A	T	G	T	C	T	G
Case 5	C	G	A	C	G	T	C	G	G
	C	G	A	T	G	T	C	T	G
Case 9	T	G	A	C	G	T	T	T	G
	T	G	A	T	G	T	C	T	G

*Reference sequence hg19