S1 Table Mutation detected in the reported patients

Patient's code	MUTATION	Type	GENE	Reference
#1	**c.1413+1G>A**	**Splicing**	**SPG4**	[S1]
#2	**c.1620_1623delGACT/InsC**	**Deletion**	**SPG4**	
#3	**c.1620_1623delGACT/InsC**	**Deletion**	**SPG4**	
#4	**exon 17 deletion (MLPA)**	**Deletion**	**SPG4**	
#5	**exon 17 deletion (MLPA)**	**Deletion**	**SPG4**	
#6	**c.1709A>G p.K570R**	**Missense**	**SPG4**	
#7	**c.1763C>A p.S588Y**	**Missense**	**SPG4**	
#8	**c.1634G>C p.S545X**	**Truncating**	**SPG4**	
#9	**c.1291C>T p.R431X**	**Truncating**	**SPG4**	[S2]
#10	**c.1222A>G, p.M408V**	**Missense**	**SPG3a**	[S10]
#11	**c.1222A>G, p.M408V**	**Missense**	**SPG3a**	[S10]
#12	**c.1222A>G, p.M408V**	**Missense**	**SPG3a**	[S10]
#13	**c.1222A>G, p.M408V**	**Missense**	**SPG3a**	[S10]
#14	**c.889A>G p.Y297A exon 4 / c.260G>T p.G87V**	**Missense**	**SPG5**	[S12]
#15	**c.889A>G p.Y297A exon 4 / c.260G>T p.G87V**	**Missense**	**SPG5**	[S12]
#16	**c.605G>A; p.S202N**	**Missense**	**SPG10**	[S16]
#17	**exon 2-5 Deletion (MLPA)**	**Deletion**	**SPG31**	[S23]
#18	**c.733_734delAT/p.M245VfsX / c.7081insT p.F2361FfsX**	**truncating**	**SPG11**	[S18]
#19	**c.1450G>C p.G484R**	**Missense**	**SPG4**	
#20	**exon 16 deletion (MLPA)**	**Deletion**	**SPG4**	
#21	**c.1456C>T p.R486C**	**Missense**	**SPG5**	[S12]
#22	**c.1456C>T p.R486C**	**Missense**	**SPG5**	[S12]
#23	c.1620_1623delGACT/InsC	Deletion	SPG4	
#24	exon 17 deletion (MLPA)	Deletion	SPG4	
#25	exon 17 deletion (MLPA)	Deletion	SPG4	
#26	c.1291C>T p.R431X	Truncating	SPG4	[S2]

#27	c.1222A>G, p.M408V	Missense	SPG3a	[S10]
#28	c.583C>G p.L195V	Missense	SPG4	[S3]
#29	c.1507C>T p.R503W	Missense	SPG4	[S4]
#30	c.1092G>T, p.R364S	Missense	SPG4	
#31	c.439G>T p.E147X	Truncating	SPG4	[S1]
#32	c.439G>T p.E147X	Truncating	SPG4	
#33	c.583C>G p.L195V	Missense	SPG4	[S3]
#34	c.1246-1G>T IVS9	Missense	SPG4	
#35	c.1821G>C p.W607C	Missense	SPG4	[S3]
#36	c.1322 -2 A>C	Splicing	SPG4	
#37	c.1685G>A p.R562Q	Missense	SPG4	[S5]
#38	c.439G>T p.E147X	Truncating	SPG4	[S1]
#39	c.326C>G p.P109R	Missense	SPG4	[S6]
#40	c.1537G>A p.G471D	Missense	SPG4	[S7]
#41	c.854A>T p.H285L	Missense	SPG5	[S13]
#42	c.854A>T p.H285L	Missense	SPG5	[S13]
#43	c.679C>T , p.R227X //c.1231G>A p.D412N	Truncating/Missense	SPG7	[S14]
#44	c.609C>G, p.S203C	Missense	SPG10	[S17]
#45	c.609C>G, p.S203C	Missense	SPG10	[S17]
#46	c.5014G>T E1672X / c.6100C>T R2034X	Truncating	SPG11	[S18]
#47	c.733-734delAT/p.M245VfsX / c.6157G>A p.V2053M	Truncating/Missense	SPG11	[S18; S19]
#48	c.2833A>G p.R945GfsX950	Truncating	SPG11	[S18]
#49	c.3935C>A p.S1312X	Truncating	SPG15	[S22]
#50	c.1550G>T p.L517W	Missense	SPG4	

#51	c.404A>G p.E135G	Missense	SPG4	
#52	c.1496G>A p.R499H	Missense	SPG4	[S8]
#53	c.334G>T p.E112X	Truncating	SPG4	[S9]
#54	c.715C>T, p.R239C	Missense	SPG3a	[S11]
#55	c.650G>A, p.R217Q;	Missense	SPG3a	[S11]
#56	c.1328G>C p.G443A	Missense	SPG5	[S13]
#57	c.1529C>T, p.A510V	Missense	SPG7	[S14]
#58	c.1529C>T, p.A510V	Missense	SPG7	[S14]
#59	c.233T>A p.Leu78X	Truncating	SPG7	[S15]
#60	c.408_428del21 p.E136_I142del	Deletion	SPG11	
#61	c.349G>T p.E117X / c.2833A>G p.R945G	Truncating/Missense	SPG11	[S18; S19]
#62	c.442+1G>C IVS2 / c.1735+2_7del AAGT IVS8	Splicing	SPG11	[S20]
#63	c. 5986_5987insT p.C1996LfsX1999 / c.6418C>T Q2140X	Truncating	SPG11	[S21]
#64	c.1203delA p.K401KfsX	Truncating	SPG11	[S18]
#65	c.442+1G>C IVS2 / c.4161 -1G>A	Splicing	SPG11	[S20]
#66	c.6832_6833delAG p.S2278LfsX2338	Truncating	SPG11	[S21]
#67	c.1523T>A p.I508N	Missense	SPG15	[S22]
#68	c.1523T>A p.I508N	Missense	SPG15	[S22]
#69	c.509A>G p.Y170C	Missense	SPG35	[S24]
#70	c.509A>G p.Y170C	Missense	SPG35	[S24]

Note: In bold patients that underwent the DTI and MRS protocol (n=22). References are found in the final part of the present document.
Abbreviations: MLPA = Multiplex Ligation-dependent Probe Amplification, SPG = Spastic Paraplegia Gene.