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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]

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c.1222A>G, p.M408V	Missense	SPG3a	[S10]
c.583C>G p.L195V	Missense	SPG4	[S3]
c.1507C>T p.R503W	Missense	SPG4	[S4]
c.1092G>T, p.R364S	Missense	SPG4	
c.439G>T p.E147X	Truncating	SPG4	[S1]
c.439G>T p.E147X	Truncating	SPG4	
c.583C>G p.L195V	Missense	SPG4	[S3]
c.1246-1G>T IVS9	Missense	SPG4	
c.1821G>C p.W607C	Missense	SPG4	[S3]
c.1322 -2 A>C	Splicing	SPG4	
c.1685G>A p.R562Q	Missense	SPG4	[S5]
c.439G>T p.E147X	Truncating	SPG4	[S1]
c.326C>G p.P109R	Missense	SPG4	[S6]
c.1537G>A p.G471D	Missense	SPG4	[S7]
c.854A>T p.H285L	Missense	SPG5	[S13]
c.854A>T p.H285L	Missense	SPG5	[S13]
c.679C>T, p.R227X //c.1231G>A p.D412N	Truncating/Missense	SPG7	[S14]
c.609C>G, p.S203C	Missense	SPG10	[S17]
c.609C>G, p.S203C	Missense	SPG10	[S17]
c.5014G>T E1672X / c.6100C>T R2034X	Truncating	SPG11	[S18]
c.733- 734delAT/p.M245VfsX / c.6157G>A p.V2053M	Truncating/Missense	SPG11	[S18; S19]
c.2833A>G p.R945GfsX950	Truncating	SPG11	[S18]
c.3935C>A p.S1312X	Truncating	SPG15	[S22]
c.1550G>T p.L517W	Missense	SPG4	
	c.583C>G p.L195V c.1507C>T p.R503W c.1092G>T, p.R364S c.439G>T p.E147X c.439G>T p.E147X c.583C>G p.L195V c.1246-1G>T IVS9 c.1821G>C p.W607C c.1322 -2 A>C c.1685G>A p.R562Q c.439G>T p.E147X c.326C>G p.P109R c.1537G>A p.G471D c.854A>T p.H285L c.854A>T p.H285L c.679C>T, p.R227X //c.1231G>A p.D412N c.609C>G, p.S203C c.609C>G, p.S203C c.5014G>T E1672X / c.6100C>T R2034X c.733- 734delAT/p.M245VfsX / c.6157G>A p.V2053M c.2833A>G p.R945GfsX950 c.3935C>A p.S1312X	c.583C>G p.L195V Missense c.1507C>T p.R503W Missense c.1092G>T, p.R364S Missense c.439G>T p.E147X Truncating c.439G>T p.E147X Truncating c.583C>G p.L195V Missense c.1246-1G>T IVS9 Missense c.1821G>C p.W607C Missense c.1322 - 2 A>C Splicing c.1685G>A p.R562Q Missense c.1326C>G p.P109R Missense c.1537G>A p.G471D Missense c.854A>T p.H285L Missense c.854A>T p.H285L Missense c.679C>T, p.R227X Truncating/Missense c.609C>G, p.S203C Missense c.5014G>T E1672X / Truncating c.6100C>T R2034X c.733- 734delAT/p.M245VfsX / c.6157G>A p.V2053M c.2833A>G p.R945GfsX950 Truncating c.3935C>A p.S1312X Truncating Truncating Truncating Truncating Truncating Truncating Truncating	c.583C>G p.L195V Missense SPG4 c.1507C>T p.R503W Missense SPG4 c.1092G>T, p.R364S Missense SPG4 c.439G>T p.E147X Truncating SPG4 c.439G>T p.E147X Truncating SPG4 c.583C>G p.L195V Missense SPG4 c.1246-1G>T IVS9 Missense SPG4 c.122G>C p.W607C Missense SPG4 c.1322 -2 A>C Splicing SPG4 c.1685G>A p.R562Q Missense SPG4 c.439G>T p.E147X Truncating SPG4 c.326C>G p.P109R Missense SPG4 c.326C>G p.P109R Missense SPG4 c.854A>T p.H285L Missense SPG5 c.854A>T p.H285L Missense SPG5 c.679C>T, p.R227X Truncating/Missense SPG10 c.609C>G, p.S203C Missense SPG10 c.5014G>T E1672X / c.6100C>T R2034X Truncating SPG11 c.5014G>T E1672A / c.6157G>A p.V2053M Truncating SPG11 c.6157G>A p.V2053M

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#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.