Column1	Column2	Column3	Column4	Column5	Column6	Column7
Study/ Population	Gender	Age at Onset	Mutation		Protein Change	
			Allele One	Allele Two	Protein One	Protein Two
SPG7 and Impaired Emotional Connection. Zhang et al. (American)	M	34	c.1529 C>T	c.2271delG	p. Ala510Val	p.Met757fs
A series of Greek Children with pure HSP: Clinical features and genetic findings. A. Polymeris et al. (Greek)	M	6.5	c.397C>T		p.Arg133Trp	
Genetic and Phenotypic characerization of complex HSP. E Kara et al. (Kenyan)	M	39	c.233T>A	c.233T>A	p.Leu78*	p.Leu78*
Genetic and Phenotypic characerization of complex HSP. E Kara et al. (UK)	M	14	c.1672A>T	c.1672A>T	p.Lys558	p.Lys558
Genetic and Phenotypic characerization of complex HSP. E Kara et al. (UK)	M	38	c.1523G>A	c.1529C>T	p.Arg508His	p.Ala510Val
Genetic and Phenotypic characerization of complex HSP. E Kara et al. (UK)	F	46	c.1450_1458del		p.Arg485_Glu487del	
Genetic and Phenotypic characerization of complex HSP. E Kara et al. (UK)	F	16	c.1529C>T	c.2102A>C	p.Ala510Val	p.His701Pro
Genetic background of the HSP phenotypes in Hungary- An analysis of 58 probands (Hungarian)	?	38	c.233T>A		p.Leu78*	
Genetic background of the HSP phenotypes in Hungary- An analysis of 58 probands (Hungarian)	M	46	c.1529C>T	c.233T>A	p.Ala510Val	p.Leu78*
Genetic background of the HSP phenotypes in Hungary- An analysis of 58 probands (Hungarian)	?	31	c.233T>A	c.233T>A	p.Leu78*	p.Leu78*
Genetic background of the HSP phenotypes in Hungary- An analysis of 58 probands (Hungarian)	?	49	c.233T>A	c.233T>A	p.Leu78*	p.Leu78*
Genetic background of the HSP phenotypes in Hungary- An analysis of 58 probands (Hungarian)	M	24	c.1529C>T	c.1031G>A	p.Ala510Val	p.Gly344Asp
A founder mutation p.H701P identified as a major cause of SPG7 in Norway. (Norwegian)	?	7	c.2102A>C	c.2102A>C	p.His701Pro	p.His701Pro
A founder mutation p.H701P identified as a major cause of SPG7 in Norway. (Norwegian)	?	8	c.2102A>C	c.2102A>C	p.His701Pro	p.His701Pro
A founder mutation p.H701P identified as a major cause of SPG7 in Norway. (Norwegian)	?	15	c.2102A>C	c.1454_1462del	p.His701Pro	p.Arg485_Glu487del
A founder mutation p.H701P identified as a major cause of SPG7 in Norway. (Norwegian)	?	27	c.2102A>C	c.1454_1462del	p.His701Pro	p.Arg485_Glu487del
A founder mutation p.H701P identified as a major cause of SPG7 in Norway. (Norwegian)	?	43	c.1053dup	c.1053dup	p.Gly352Argfs*44	p.Gly352Argfs*45
A founder mutation p.H701P identified as a major cause of SPG7 in Norway. (Norwegian)	?	20	c.2102A>C	c.1529C>T	p.His701Pro	p.Ala510Val
A founder mutation p.H701P identified as a major cause of SPG7 in Norway. (Norwegian)	?	24	c.2102A>C	c.1529C>T	p.His701Pro	p.Ala510Val
A founder mutation p.H701P identified as a major cause of SPG7 in Norway. (Norwegian)	?	36	c.2102A>C	c.1529C>T	p.His701Pro	p.Ala510Val
A founder mutation p.H701P identified as a major cause of SPG7 in Norway. (Norwegian)	?	22	c.2102A>C	c.1529C>T	p.His701Pro	p.Ala510Val
A founder mutation p.H701P identified as a major cause of SPG7 in Norway. (Norwegian)	?	20	c.1672A>T	c.1529C>T	p.Lys558*	p.Ala510Val
A founder mutation p.H701P identified as a major cause of SPG7 in Norway. (Norwegian)	?	36	c.1672A>T	c.1529C>T	p.Lys558*	p.Ala510Val
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	20	c.1529C>T	c.2249C>T	p.Ala510Val	p.Pro750Leu
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	55	c.1529C>T	c.2249C>T	p.Ala510Val	p.Pro750Leu
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	40	c.1529C>T	c.2249C>T	p.Ala510Val	p.Pro750Leu
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	32	c.1529C>T	c.2249C>T	p.Ala510Val	p.Pro750Leu
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	15	c.1529C>T	c.1715C>T	p.Ala510Val	p.Ala572Val
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	43	c.1529C>T	c.1715C>T	p.Ala510Val	p.Ala572Val
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	50	c.1529C>T	c.1715C>T	p.Ala510Val	p.Ala572Val
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	45	c.1529C>T	c.1715C>T	p.Ala510Val	p.Ala572Val
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	25	c.1529C>T	c.988-1G>A Novel	p.Ala510Val	p.Arg333C
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	28	c.1529C>T	c.1715C>T	p.Ala510Val	p.Ala572Val
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	32	c.1045G>A	c.2249C>T	p.Gly349Ser	p.Pro750Leu
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	40	c.1045G>A	c.2249C>T	p.Gly349Ser	p.Pro750Leu
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	48	c.1045G>A	c.2249C>T	p.Gly349Ser	p.Pro750Leu
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French	?	30	c.1529C>T	c.233T>A	p.Ala510Val	p.Leu78Thr

Canadian)						
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	20	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	25	c.988-1G>A Novel	c.1529C>T	p.Arg333C	p.Ala510Val
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	37	c.988-1G>A Novel	c.1529C>T	p.Arg333C	p.Ala510Val
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	25	c.233T>A	c.988-1G>A Novel	p.Leu78Thr	p.Arg333C
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	40	c.988-1G>A Novel	c.988-1G>A Novel	p.Arg333C	p.Arg333C
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	35	c.988-1G>A Novel	c.988-1G>A Novel	p.Arg333C	p.Arg333C
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	39	c.988-1G>A Novel	c.988-1G>A Novel	p.Arg333C	p.Arg333C
SPG7 Mutations explain a significant proportion of French Canadian spastic ataxia cases (French Canadian)	?	30	c.473_474del Novel	c.988-1G>A Novel	p.Leu158GlyInfsThr30	p.Arg333C
Canadian)						
Abnormal paraplegin expression in swollen neurites, tu and alpha-synuclein pathology in a case of hereditary paraplegia SPG7 with an Ala510Val Mutation (Caucasian, ?German)	M	59	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
SPG7 Mutations are a common cause of undiagnosed Ataxia (UK)	?	35	c.1529C>T	c.1715C>T	p.Ala510Val	p.Ala572Val
SPG7 Mutations are a common cause of undiagnosed Ataxia (UK)	?	35	c.1529C>T	c.1715C>T	p.Ala510Val	p.Ala572Val
SPG7 Mutations are a common cause of undiagnosed Ataxia (UK)	?	25	c.1529C>T	c.1192C>T	p.Ala510Val	p.Arg398*
SPG7 Mutations are a common cause of undiagnosed Ataxia (UK)	?	28	c.1529C>T	c.1192C>T	p.Ala510Val	p.Arg398*
SPG7 Mutations are a common cause of undiagnosed Ataxia (UK)	?	33	c.1529C>T	c.233T>A	p.Ala510Val	p.Leu78*
SPG7 Mutations are a common cause of undiagnosed Ataxia (UK)	?	32	c.1529C>T	c.1715C>T	p.Ala510Val	p.Ala572Val
SPG7 Mutations are a common cause of undiagnosed Ataxia (UK)	?	45	c.1529C>T	c.1715C>T	p.Ala510Val	p.Ala572Val
SPG7 Mutations are a common cause of undiagnosed Ataxia (UK)	?	"teens"	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
SPG7 Mutations are a common cause of undiagnosed Ataxia (UK)	?	29	c.1529C>T	c.228T>C	p.Ala510Val	p.Ile743Thr
SPG7 Mutations are a common cause of undiagnosed Ataxia (UK)	?	10	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
SPG7 Mutations are a common cause of undiagnosed Ataxia (UK)	?	49	c.1529C>T	c.1053dup	p.Ala510Val	p.Gly352Argfs*44
SPG7 Mutations are a common cause of undiagnosed Ataxia (UK)	?	54	c.1529C>T	c.1225_1229del	p.Ala510Val	p.Gly352Argfs*49
SPG7 Mutations are a common cause of undiagnosed Ataxia (UK)	?	46	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
SPG7 Mutations are a common cause of undiagnosed Ataxia (UK)	?	46	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
SPG7 Mutations are a common cause of undiagnosed Ataxia (UK)	?	18	c.1529C>T	c.1715C>T	p.Ala510Val	p.Ala572Val
SPG7 Mutations are a common cause of undiagnosed Ataxia (UK)	?	"Childhood"	c.1529C>T	c.1053dup	p.Ala510Val	p.Gly352Argfs*44
SPG7 Mutations are a common cause of undiagnosed Ataxia (UK)	?	40	c.1529C>T	c.1053dup	p.Ala510Val	p.Gly352Argfs*44
Predominant cerebellar phenotype in spastic paraplegia 7 SPG7 (Japanese)	M	32	c.1192C>T	c.1192C>T	p.R398X	p.Arg398*
Predominant cerebellar phenotype in spastic paraplegia 7 SPG7 (Japanese)	M	50	c.1192C>T	c.1192C>T	p.Arg398*	p.Arg398*
Early-onset optic neuropathy as initial presentation in SPG7 (Italian)	M	Optic:Childhood Gait:30	c.538>A	c.1045G>A	p.Val180Met	p.Gly349Ser
Hereditary spastic parapresis in adults. A clinical and genetic perspective from Tuscany (Italian)	M	27	Macrodeletion of exons 13-17	c.1529C>T		p.Ala510Val
Hereditary spastic parapresis in adults. A clinical and genetic perspective from Tuscany (Italian)	F	35	c.1529C>T	c.2213_2214insAfr	p.Ala510Val	
Hereditary spastic parapresis in adults. A clinical and genetic perspective from Tuscany (Italian)	M	46	c.233T>A*	c.233T>A*	p.Leu78*	p.Leu78*
Hereditary spastic parapresis in adults. A clinical and genetic perspective from Tuscany (Italian)	M	53	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
Hereditary spastic parapresis in adults. A clinical and genetic perspective from Tuscany (Italian)	M	34	c.1617delCfr	c.1617delCfr	p.Ser539*	p.Ser539*
Spastic Paraplegia Type 7 is associated with multiple mitochondrial DNA deletions	M	7	c.2102A>C	c.2102A>C	p.His701Pro	p.His701Pro
Spastic Paraplegia Type 7 is associated with multiple mitochondrial DNA deletions	F	8	c.2102A>C	c.2102A>C	p.His701Pro	p.His701Pro
Spastic Paraplegia Type 7 is associated with multiple mitochondrial DNA deletions	F	15	c.2102A>C	c.1454_1462del	p.His701Pro	p.Arg485_1462
Spastic Paraplegia Type 7 is associated with multiple mitochondrial DNA deletions	M	27	c.2102A>C	c.1454_1462del	p.His701Pro	p.Arg485_1462

Identification of a novel homozygous SPG7 mutation in a japanese patient with spastic ataxia: Making efficient diagnosis using exome sequencing for autosomal recessive cerebellar ataxia and spastic paraplegia (Japanese)	M	34	c.1342dup	c.1342dup	p.His448Profs*12	p.His448Profs*12
Targeted next generation sequencing in SPAST-negative HSP. (Australian)	M	42	c.1045G>A	c.1454_1462del	p.Gly349Ser	p.Arg485_Glu487del
Targeted next generation sequencing in SPAST-negative HSP. (Australian)	M	60	c.1454_1462del	c.1529C>T	p.Arg485_Glu487del	p.Ala510Val
Targeted next generation sequencing in SPAST-negative HSP. (Australian)	M	48	c.1454_1462del	c.1529C>T	p.Arg485_Glu487del	p.Ala510Val
Targeted next generation sequencing in SPAST-negative HSP. (Australian)	M	46	c.1529C>T	c.1572C>G	p.Ala510Val	p.Ser576Trp
Autosomal recessive HSP- Clinical and genetic characteristics of a well-defined cohort (UK)	M	22	c.415C>T	c.1053dupC	p.Arg139*	p.Gly352Argfs*44
Autosomal recessive HSP- Clinical and genetic characteristics of a well-defined cohort (Caucasian)	M	10	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
Autosomal recessive HSP- Clinical and genetic characteristics of a well-defined cohort (Caucasian)	M	43	c.1529C>T	c.861dup	p.Ala510Val	p.Asn288*
Autosomal recessive HSP- Clinical and genetic characteristics of a well-defined cohort (French-Canadian)	M	10	c.1045G>A	c.1996G>C	p.Gly349Ser	p.Gly666Arg
Autosomal recessive HSP- Clinical and genetic characteristics of a well-defined cohort (Caucasian)	M	30	c.759-1G>T	c.1450_1458del	p.Met1_Asn253delfs*35	p.Arg485_Glu487del
Autosomal recessive HSP- Clinical and genetic characteristics of a well-defined cohort (French-Canadian)	M	40	c.1529C>T	c.4delG	p.Ala510Val	p.Ala2fs*64
Autosomal recessive HSP- Clinical and genetic characteristics of a well-defined cohort (East Indian)	F	48	del exon 6 Novel	del exon 6 Novel		
Autosomal recessive HSP- Clinical and genetic characteristics of a well-defined cohort (East Indian)	M	35	del exon 6 Novel	del exon 6 Novel		
Autosomal recessive HSP- Clinical and genetic characteristics of a well-defined cohort (Lebanese/ French-Canadian)	M	38	c.1996G>C	del exons 12-14 Novel	p.Gly666Arg	
Autosomal recessive HSP- Clinical and genetic characteristics of a well-defined cohort (Caucasian)	F	30	c.1529C>T		p.Ala510Val	
Autosomal recessive HSP- Clinical and genetic characteristics of a well-defined cohort (Caucasian)	M	42	c.1529C>T		p.Ala510Val	
Autosomal recessive HSP- Clinical and genetic characteristics of a well-defined cohort (Caucasian)	M	50	c.1529C>T		p.Ala510Val	
The p.Ala510val mutation in the SPG7 gene is the most common mutation causing adult onset neurogenic disease in patients of ritish ancestry. (UK ancestry)	?	54	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
The p.Ala510val mutation in the SPG7 gene is the most common mutation causing adult onset neurogenic disease in patients of ritish ancestry. (UK ancestry)	?	46	c.1529C>T	c.1053dup	p.Ala510Val	p.Gly352Argfs*44
The p.Ala510val mutation in the SPG7 gene is the most common mutation causing adult onset neurogenic disease in patients of ritish ancestry. (UK ancestry)	?	33	c.1529C>T	c.1450_1458del	p.Ala510Val	p.Arg485_Glu487del
The p.Ala510val mutation in the SPG7 gene is the most common mutation causing adult onset neurogenic disease in patients of ritish ancestry. (UK ancestry)	?	39			p.Arg599Leu Novel	
The p.Ala510val mutation in the SPG7 gene is the most common mutation causing adult onset neurogenic disease in patients of ritish ancestry. (Australian)	?	39	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
The p.Ala510val mutation in the SPG7 gene is the most common mutation causing adult onset neurogenic disease in patients of ritish ancestry. (UK ancestry)	?	42	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
The p.Ala510val mutation in the SPG7 gene is the most common mutation causing adult onset neurogenic disease in patients of ritish ancestry. (UK ancestry)	?	55	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
The p.Ala510val mutation in the SPG7 gene is the most common mutation causing adult onset neurogenic disease in patients of ritish ancestry. (UK ancestry)	?	61	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
The p.Ala510val mutation in the SPG7 gene is the most common mutation causing adult onset neurogenic disease in patients of ritish ancestry. (UK ancestry)	?	60	c.1045G>A	c.1529C>T	p.Gly349Ser	p.Ala510Val
The p.Ala510val mutation in the SPG7 gene is the most common mutation causing adult onset neurogenic disease in patients of ritish ancestry. (UK ancestry)	?	38	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
SPG7 in patients with spasticity and/or optic atrophy (French)	F	36	c.1519C>T	c.1529C>T	p.Gln507*	p.Ala510Val
SPG7 in patients with spasticity and/or optic atrophy (French)	M	39	c.233T>A	c.1450_1458del	p.Leu78*	p.Arg485_Glu487del
SPG7 in patients with spasticity and/or optic atrophy (French)	M	40	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
SPG7 in patients with spasticity and/or optic atrophy (French)	F	46	c.1 A>T	c.1529C>T	p.Met1?	p.Ala510Val
SPG7 in patients with spasticity and/or optic	F	18	c.1450_1458del	c.1450_1458del	p.Arg485_Glu487del	p.Arg485_Glu487del

atrophy (French)						
SPG7 in patients with spasticity and/or optic atrophy (French)	M	25	c.1450_1458del	c.1529C>T	p.Arg485_Glu487del	p.Ala510Val
SPG7 in patients with spasticity and/or optic atrophy (French)	M	37	c.1 A>G	c.850_851delins	p.Met1?	p.Phe284Profs*45
SPG7 in patients with spasticity and/or optic atrophy (French)	F	40	c.1450_1458del	c.1529C>T	p.Arg485_Glu487del	p.Ala510Val
SPG7 in patients with spasticity and/or optic atrophy (French)	F	42	c.233T>A	c.1450_1458del	p.Leu78*	p.Arg485_Glu487del
SPG7 in patients with spasticity and/or optic atrophy (French)	F	44	c.1-?_286+?	c.1529C>T	p.0?	p.Ala510Val
SPG7 in patients with spasticity and/or optic atrophy (French)	F	44	c.233T>A	c.233T>A	p.Leu78*	p.Leu78*
SPG7 in patients with spasticity and/or optic atrophy (Maroccon)	M	32	c.618+3G>C	c.2240T>C	p.Arg2017Trp	p.Ile747Thr
SPG7 in patients with spasticity and/or optic atrophy (French)	F	34	c.1450_1458del	c.1529C>T	p.Arg485_Glu487del	p.Ala510Val
SPG7 in patients with spasticity and/or optic atrophy (French)	F	37	c.1049_1077del	c.1529C>T	p.Pro350Glyinf*36	p.Ala510Val
SPG7 in patients with spasticity and/or optic atrophy (French)	M	40	c.2216dupA	c.2216dupA	p.Asp739Lysfs*3	p.Asp739Lysfs*3
SPG7 in patients with spasticity and/or optic atrophy (French)	M	?	c.1053dup	c.1529C>T	p.Gly352Argfs*44	p.Ala510Val
SPG7 in patients with spasticity and/or optic atrophy (French)	F	52	c.376+1G>T	c.1045G>A	p.?	p.Gly349Ser
SPG7 in patients with spasticity and/or optic atrophy (French)	F	45	c.1529C>T	c.1715C>T	p.Ala510Val	p.Ala572Val
SPG7 in patients with spasticity and/or optic atrophy (French)	M	20	c.1529C>T	c.2249C>T	p.Ala510Val	p.Pro750Leu
SPG7 in patients with spasticity and/or optic atrophy (UK)	F	30	c.1053dup	c.1529C>T	p.Gly352Argfs*44	p.Ala510Val
SPG7 in patients with spasticity and/or optic atrophy (French)	F	30	c.1529C>T	c.2249C>T	p.Ala510Val	p.Pro750Leu
SPG7 in patients with spasticity and/or optic atrophy (French)	M	31	c.1369C>T	c.1529C>T	p.Arg457*	p.Ala510Val
SPG7 in patients with spasticity and/or optic atrophy (French)	F	43	c.861dup	c.1529C>T	p.Asn288*	p.Ala510Val
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	10	c.1409G>A	c.1409G>A	p.Arg470Gln	p.Arg470Gln
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	27	c.1409G>A	c.1409G>A	p.Arg470Gln	p.Arg470Gln
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	43	c.1454_1462del	c.1454_1462del	p.Arg485_Glu487del	p.Arg485_Glu487del
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	47	c.1454_1462del	c.1454_1462del	p.Arg485_Glu487del	p.Arg485_Glu487del
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	?	c.1454_1462del	c.1454_1462del	p.Arg485_Glu487del	p.Arg485_Glu487del
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	37	c.1454_1462del	c.1454_1462del	p.Arg485_Glu487del	p.Arg485_Glu487del
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	36	c.1454_1462del	c.1672A>T	p.Arg485_Glu487del	p.Lys558*
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	28	c.1454_1462del	c.1672A>T	p.Arg485_Glu487del	p.Lys558*
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	34	c.1454_1462del	c.2115_2131del	p.Arg485_Glu487del	p.Leu706fs
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	38	c.1454_1462del	c.2115_2131del	p.Arg485_Glu487del	p.Leu706fs
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	53	c.1529C>T	c.1454_1462del	p.Ala510Val	p.Arg485_Glu487del
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	36	c.1529C>T	c.1454_1462del	p.Ala510Val	p.Arg485_Glu487del
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	47	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	44	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	52	c.1529C>T	c.1937-2del	p.Ala510Val	?
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	45	c.1529C>T	c.1937-2del	p.Ala510Val	?
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	21	c.1529C>T	c.861dup	p.Ala510Val	p.Asn288*
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	24	c.1529C>T	c.861dup	p.Ala510Val	p.Asn288*
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	46	c.861dup	c.2228T>C	p.Asn288*	p.Ile743Thr
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	48	c.861dup	c.2228T>C	p.Asn288*	p.Ile743Thr
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	33	c.861dup	c.861dup	p.Asn288*	p.Asn288*
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	33	c.861dup	c.861dup	p.Asn288*	p.Asn288*
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	?	c.1529C>T	c.1045G>A	p.Ala510Val	p.Gly349Ser
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	?	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	?	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	?	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	?	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	42	c.1045G>A	c.2069C>T	p.Gly349Ser	p.Pro690Leu
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	40	c.1045G>A	c.2090A>C	p.Gly349Ser	p.Gln697Pro
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	42	c.1045G>A	c.2115_2131del	p.Gly349Ser	p.Leu706fs

Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	60	c.1147G>T	c.1822C>T	p.Gly383*	p.Gln608*
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	34	c.1454_1462del	c.2228T>C	p.Arg485_Glu487del	p.Ile743Thr
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	47	c.1454_1462del	c.233T>A	p.Arg485_Glu487del	p.Leu78*
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	38	c.1529C>T	c.1454_1462del	p.Ala510Val	p.Arg485_Glu487del
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	22	c.1529C>T	c.1454_1462del	p.Ala510Val	p.Arg485_Glu487del
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	43	c.1529C>T	c.1454_1462del	p.Ala510Val	p.Arg485_Glu487del
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	33	c.1529C>T	c.1454_1462del	p.Ala510Val	p.Arg485_Glu487del
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	57	c.1529C>T	c.1454_1462del	p.Ala510Val	p.Arg485_Glu487del
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	47	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	53	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	60	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	58	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	44	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	37	c.1529C>T	c.2014G>A	p.Ala510Val	p.Gly672Arg
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	50	c.1529C>T	c.2115_2131del	p.Ala510Val	p.Leu706fs
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	27	c.1529C>T	c.2115_2131del	p.Ala510Val	p.Leu706fs
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	20	c.1529C>T	c.2115_2131del	p.Ala510Val	p.Leu706fs
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	55	c.1529C>T	c.759-1G>T	p.Ala510Val	p.Met1_Asn253delfs*35
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	51	c.1529C>T	c.759-1G>T	p.Ala510Val	p.Met1_Asn253delfs*35
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	63	c.1672A>T	c.1672A>T	p.Lys558*	p.Lys558*
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	16	c.1894G>A	c.1984delinsPhe	p.Gly632Arg	p.Val662fs
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	20	c.2115_2131del	c.2115_2131del	p.Leu706fs	p.Leu706fs
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	16	c.2115_2131del	c.2115_2131del	p.Leu706fs	p.Leu706fs
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	10	c.3G>A	c.3G>A	p.Ala2Thr	p.Ala2Thr
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	51	c.618+3G>C	c.2219A>G	p.Arg2017Trp	p.Tyr740C
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	?	c.1045G>A	c.1450_1458del	p.Gly349Ser	p.Arg485_Glu487del
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	?	c.1529C>T	c.1045G>A	p.Ala510Val	p.Gly349Ser
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	?	c.1529C>T	c.1045G>A	p.Ala510Val	p.Gly349Ser
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	F	?	c.1529C>T	c.1053dup	p.Ala510Val	p.Gly352Argfs*44
Genotype-phenotype correlations in SPG7: a study in a large Dutch cohort (Dutch)	M	?	c.1529C>T	c.1454_1462del	p.Ala510Val	p.Arg485_Glu487del
SPG7 mutational screening in spastic paraplegia patients supports a dominant effect for some mutations and a pathogenic role for p.A510V (Spanish)	?	39	c.1061G>C	c.1715C>T	p.Gly354Ala	p.Ala572Val
SPG7 mutational screening in spastic paraplegia patients supports a dominant effect for some mutations and a pathogenic role for p.A510V (Spanish)	?	44	c.1987A>T		p.Lys663*	
SPG7 mutational screening in spastic paraplegia patients supports a dominant effect for some mutations and a pathogenic role for p.A510V (Spanish)	?	32	c.661A>T		p.Lys221*	
SPG7 mutational screening in spastic paraplegia patients supports a dominant effect for some mutations and a pathogenic role for p.A510V (Spanish)	?	52	c.1198C>T		p.Arg400Trp	
SPG7 mutational screening in spastic paraplegia patients supports a dominant effect for some mutations and a pathogenic role for p.A510V (Spanish)	?	29	c.1044_1045insA		p.Leu348_Gly349insLeu	
SPG7 mutational screening in spastic paraplegia patients supports a dominant effect for some mutations and a pathogenic role for p.A510V (Spanish)	?	39	c.1645G>A		p.Val549Met	
SPG7 mutational screening in spastic paraplegia patients supports a dominant effect for some mutations and a pathogenic role for p.A510V (Spanish)	?	20	c.233T>A		p.Leu78*	
SPG7 mutational screening in spastic paraplegia patients supports a dominant effect for some mutations and a pathogenic role for p.A510V (Spanish)	?	40	c.773_774del*		p.Val258Glyfs*30	
SPG7 mutational screening in spastic paraplegia patients supports a dominant effect for some mutations and a pathogenic role for p.A510V (Spanish)	?	18	c.759-1G>T		p.Met1_Asn253delfs*35	
SPG7 mutational screening in spastic paraplegia patients supports a dominant effect for some						

mutations and a pathogenic role for p.A510V (Spanish)	?	25	c.376G>C		p.Glu126Argfs*5	
SPG7 mutational screening in spastic paraplegia patients supports a dominant effect for some mutations and a pathogenic role for p.A510V (Spanish)	?	26	c.376G>C	c.1529C>T	p.Glu126Argfs*5	p.Ala510Val
SPG7 mutational screening in spastic paraplegia patients supports a dominant effect for some mutations and a pathogenic role for p.A510V (Spanish)	?	36	c.1982T>C	c.1529C>T	p.Met661Thr	p.Ala510Val
SPG7 mutational screening in spastic paraplegia patients supports a dominant effect for some mutations and a pathogenic role for p.A510V (Spanish)	?	48	c.184-?_1324+?del	c.1529C>T	p.Ser62_Asp441delfs*24	p.Ala510Val
SPG7 mutational screening in spastic paraplegia patients supports a dominant effect for some mutations and a pathogenic role for p.A510V (Spanish)	?	50	c.1777A>T	c.1529C>T	p.Lys593*	p.Ala510Val
SPG7 mutational screening in spastic paraplegia patients supports a dominant effect for some mutations and a pathogenic role for p.A510V (Spanish)	?	47	c.1529C>T		p.Ala510Val	
SPG7 mutational screening in spastic paraplegia patients supports a dominant effect for some mutations and a pathogenic role for p.A510V (Spanish)	?	1	c.1529C>T		p.Ala510Val	
SPG7 mutational screening in spastic paraplegia patients supports a dominant effect for some mutations and a pathogenic role for p.A510V (Spanish)	?	44	c.1529C>T		p.Ala510Val	
SPG7 mutational screening in spastic paraplegia patients supports a dominant effect for some mutations and a pathogenic role for p.A510V (Spanish)	?	50	c.1529C>T		p.Ala510Val	
Amplicon-based high throughput pooled sequencing identifies mutations in CYP781 and SPG7 in sporadic spastic paraplegia in patients (German)	M	?	c.739C>T	c.1045G>A	p.Arg247*	p.Gly349Ser
Amplicon-based high throughput pooled sequencing identifies mutations in CYP781 and SPG7 in sporadic spastic paraplegia in patients (German)	M	46	c.1057_1085del29	c.1552+1G>T	p.Fs353-384*385	Splicing
Amplicon-based high throughput pooled sequencing identifies mutations in CYP781 and SPG7 in sporadic spastic paraplegia in patients (German)	M	58	c.1192C>T	c.2275G>A	p.Arg398*	p.Ala759Thr
Amplicon-based high throughput pooled sequencing identifies mutations in CYP781 and SPG7 in sporadic spastic paraplegia in patients (German)	M	36	c.86G>A	c.86G>A	p.Trp29*	p.Trp29*
Amplicon-based high throughput pooled sequencing identifies mutations in CYP781 and SPG7 in sporadic spastic paraplegia in patients (German)	M	30	c.415C>T	c.1057_1085del29	p.Arg139*	p.Fs353-384*385
Amplicon-based high throughput pooled sequencing identifies mutations in CYP781 and SPG7 in sporadic spastic paraplegia in patients (German)	M	14	c.1038_1039ins33	c.1730G>A	p.Leu346_Leu347ins11	p. Gly577Asp
Amplicon-based high throughput pooled sequencing identifies mutations in CYP781 and SPG7 in sporadic spastic paraplegia in patients (German)	F	52	c.1045G>A	c.2216dupA	p.Gly349Ser	p.Asn739fs
Amplicon-based high throughput pooled sequencing identifies mutations in CYP781 and SPG7 in sporadic spastic paraplegia in patients (German)	?	?	c.1450_1458del	c.1529C>T	p.Arg485_Glu487del	p.Ala510Val
Amplicon-based high throughput pooled sequencing identifies mutations in CYP781 and SPG7 in sporadic spastic paraplegia in patients (German)	?	?	c.1031G>A	c.1529C>T	p.Gly344Asp	p.Ala510Val
Amplicon-based high throughput pooled sequencing identifies mutations in CYP781 and SPG7 in sporadic spastic paraplegia in patients (German)	M	28	c.1192C>T	c.1529C>T	p.Arg398*	p.Ala510Val
Amplicon-based high throughput pooled sequencing identifies mutations in CYP781 and SPG7 in sporadic spastic paraplegia in patients (German)	M	48	c.1193G>A	c.1529C>T	p.Arg398Gln	p.Ala510Val
Amplicon-based high throughput pooled sequencing identifies mutations in CYP781 and SPG7 in sporadic spastic paraplegia in patients (German)	F	?	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
Amplicon-based high throughput pooled sequencing identifies mutations in CYP781 and SPG7 in sporadic spastic paraplegia in patients (German)	M	25	c.21_23dupA		p.Leu8delinsLeu	
Amplicon-based high throughput pooled sequencing identifies mutations in CYP781 and SPG7 in sporadic spastic paraplegia in patients (German)	M	45	c.1529C>T	c.1529C>T	p.Ala510Val	p.Ala510Val
A novel Splice Site Mutation in the SPG7 Gene causing widespread fiber damage in Homozygous and Heterozygous Subjects (German)	F	40	c1552+1G>T	c1552+1G>T	frameshift by two bases after Amino Acid 483 leading to a stop of transcription after amino acid 556	frameshift by two bases after Amin leading to a stop of transcription a acid 557
HSP caused by the novel mutation 1047insC in the SPG7 gene (Italian)	M	34	c.1047insC	c.1047insC	p.Pro350Thr	p.Pro350Thr
HSP caused by the novel mutation 1047insC in						

the SPG7 gene (Italian)	F	45	c.1047insC	c.1047insC	p.Pro350Thr	p.Pro350Thr
HSP caused by the novel mutation 1047insC in the SPG7 gene (Italian)	M	44	c.1047insC	c.1047insC	p.Pro350Thr	p.Pro350Thr
A clinical, genetic and biochemical characterization of SPG7 Mutations in a large Cohort of Patients with HSP (Italian)	?	41	c.233T>A	c.233T>A	p.Leu78*	p.Leu78*
A clinical, genetic and biochemical characterization of SPG7 Mutations in a large Cohort of Patients with HSP (Italian)	?	60	c.233T>A	c.233T>A	p.Leu78*	p.Leu78*
A clinical, genetic and biochemical characterization of SPG7 Mutations in a large Cohort of Patients with HSP (Italian)	?	40	c.233T>A	c.233T>A	p.Leu78*	p.Leu78*
A clinical, genetic and biochemical characterization of SPG7 Mutations in a large Cohort of Patients with HSP (Italian)	?	40	c.1616delC	c.1447_1778 del331	p.Val540fs	p.Glu484_Lys593 del119
A clinical, genetic and biochemical characterization of SPG7 Mutations in a large Cohort of Patients with HSP (Italian)	?	50	c.698T>C		p.Leu233Pro	
A clinical, genetic and biochemical characterization of SPG7 Mutations in a large Cohort of Patients with HSP (Italian)	?	38	c.1636G>A		p.Glu546Lys	
A clinical, genetic and biochemical characterization of SPG7 Mutations in a large Cohort of Patients with HSP (Italian)	?	25	c.2191G>A		p.Ala731Thr	
A clinical, genetic and biochemical characterization of SPG7 Mutations in a large Cohort of Patients with HSP (Italian)	?	35	c.2216dupA		p.Asn739fs	
A clinical, genetic and biochemical study of SPG7 mutations in hereditary spastic paraplegia. (British)	M	11	c.28G>A	c.1729G>A	p.Ala10Ser	p.Gly577Ser
A clinical, genetic and biochemical study of SPG7 mutations in hereditary spastic paraplegia. (British)	F	14	c.1057-1085del29	c.1715C>T	p.Fs353-384*385	p.Ala572Val
A clinical, genetic and biochemical study of SPG7 mutations in hereditary spastic paraplegia. (British)	M	19	c.1450_1458del	c.2026T>C	p.Arg485_Glu487del	p.Phe676Leu
Paraplegin mutations in sproadic adult-onset upper motor neuron syndromes (Dutch)	F	42	c.1454-1462del	c.1529C>T	p.Arg485_Glu487del	p.Ala510Val
Paraplegin mutations in sproadic adult-onset upper motor neuron syndromes (Dutch)	F	39	c.1454-1462del	c.1529C>T	p.Arg485_Glu487del	p.Ala510Val
Paraplegin mutations in sproadic adult-onset upper motor neuron syndromes (Dutch)	M	36	c.1454-1462del	c.1529C>T	p.Arg485_Glu487del	p.Ala510Val
Paraplegin mutations in sproadic adult-onset upper motor neuron syndromes (Dutch)	M	39	c.1454-1462del	c.1454-1462del	p.Arg485_Glu487del	p.Arg485_Glu487del
Paraplegin mutations in sproadic adult-onset upper motor neuron syndromes (Dutch)	F	37	c.1529C>T	c.2014G>A	p.Ala510Val	p.Gly672Arg
Paraplegin mutations in sproadic adult-onset upper motor neuron syndromes (Dutch)	M	34	c.1454-1462del	c.2228T>C	p.Arg485_Glu487del	p.Ile743Thr
Paraplegin mutations in sproadic adult-onset upper motor neuron syndromes (Dutch)	F	35	c.1045G>A	c.2069C>T	p.Gly349Ser	p.Pro690Leu
A novel form of autosomal recessive HSP caused by a new SPG7 mutation (Turkish)	M	25	c.2075G>C	c.2075G>C	p.Ser692Thr	p.Ser692Thr
A novel form of autosomal recessive HSP caused by a new SPG7 mutation (Turkish)	F	16	c.2075G>C	c.2075G>C	p.Ser692Thr	p.Ser692Thr
A novel form of autosomal recessive HSP caused by a new SPG7 mutation (Turkish)	M	10	c.2075G>C	c.2075G>C	p.Ser692Thr	p.Ser692Thr

Study/population: No changes
Gender: Where unknown - ?
Age at onset: Where unknown - ?
Allele one: all AA changed into three letter coding
Allele two: all AA changed into three letter coding
Protein one: all AA changed into three letter coding
Spasticity : "+"- present; "++"- moderate; "+++"- severe; "-"- absent; ?- not indicated
Cerebellar signs: "+"- present; "++"- moderate; "+++"- severe; "-"- absent; ?- not indicated
Myopathic signs: "+"- present; "++"- moderate; "+++"- severe; "-"- absent; ?- not indicated
Optic atrophy: "+"- present; "++"- moderate; "+++"- severe; "-"- absent; ?- not indicated
CPEO: "+"- present; "++"- moderate; "+++"- severe; "-"- absent; ?- not indicated
Other: no blank spaces left
Clinical diagnosis: blank paces filled; sporadic HSP > sporadic spastic paraparesis