CORRIGENDUM

Molecular epidemiology and clinical spectrum of hereditary spastic paraplegia in the Japanese population based on comprehensive mutational analyses

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Incorrect description in Table 2 in the above-mentioned article was noticed after its publication.

1. In the title for Table 2, 'Mutation and clinical summary of Japanese HSP patients' should be 'Mutations and clinical summaries of Japanese HSP patients'.

2. The nucleotide description for p.R562Q of *SPAST* should be c.1685G>A instead of c.1688G>A.

3. The position of deletion of *SPAST* (intron 16 ~ 3' region) should be Chr2: 32231942_32290423 which causes 58 482-bp deletion instead of Chr2: 32290425-32231940.

4. The position of the *REEPI* deletion (intron 3 ~ intron 5) should be Chr2:86326359_86338423 which causes 12 065-bp deletion instead of Chr2: 86326358-86338428.

5. The nucleotide description for p.K1614* of *SPG11* should be c.4840A>T instead of c.4840T>A.

6. In the description of *SPAST* deletions involving intron16 to 3'-UTR and exon 4 to intron 7, 'Chr2' should be added to clarify the position of the deletions.

7. The incorrect description of the foot note 'All the patients presented the pure form.' should be deleted.

These correction does not alter the results and their interpretation as discussed in the paper.

We apologize for this mistake.

Table 2 Mutations and clinical summaries of Japanese HSP patients

	Mutation	Amino-acid change	Detecting method	Family history	Age at onset of index patient	Clinical phenotype	References
SPG4							
Exon 1	c.139 A>T	p.K47*	R	AD	40s	Pure	Novel
Exon 1	c.155 A>G	p.Y52C	R	Sporadic with consanguineous parents	49 y.o.	Pure	Novel
Exon 1	c.283_323 del	p.A95Afs	D	AD	40 y.o.	Pure	Novel
Exon 1	c.343_352 dup	p.V118Afs	D	AD	35 y.o.	Pure	Novel
Exon 2	c.422–425 delAGAA	p.Q141fs	D	AD	36 y.o.	Pure	Novel
Exon 2	c.422–425 delAGAA	p.Q141fs	D	AD	51 y.o.	Pure	Novel
Exon 2	c.422–425 delAGAA	p.Q141fs	D	Sporadic	35 y.o.	Pure	Novel
Exon 3	c.532 C>T	p.Q178*	R	AD	33 y.o.	Pure	Novel
Exon 5	c.734 C>G	p.S245*	R	AD	Teens	Pure	Known 35
Exon 5	c.838 C>T	p.Q280*	R	AD	~ 6 y.o.	Pure	Novel
Exon 6	c.871 delG	p.G291Vfs	D	AD	20 y.o.	Pure	Novel
Intron 6	c.1005-2 A>G	IVS6-2A>G	R	AD	2 y.o.	Pure	Known 4
Exon 7	c.1014 delT	p.A338Afs	D	AD	40s	Pure	Novel
Exon 8	c.1105 A>C	p.T369P	R	AD	38 y.o.	Pure	Novel
Exon 8	c.1141 C>T	p.F381L	R	Sporadic	< 6 y.o.	Pure	Known 4
Exon 8	c.1141 C>T	p.F381L	R	AD	Late 50s	Pure	Known 4
Intron 8	c.1173+1G>A	IVS8+1G>A	R	AD	46 y.o.	Pure	Known 3
Exon 11	c.1378 C>T	p.R460C	R	AD	27 y.o.	Pure	Known 36
Exon 12	c.1426_1427 delGG	p.G476Rfs	D	AD	39 y.o.	Pure	Novel
Intron 12	c.1493+2T>C	IVS12+2T>C	R	Sporadic	40 y.o.	Pure	Known 35
Exon 13	c.1504 A>T	p.K502*	R	AD	30 y.o.	Pure	Novel
Exon 13	c.1507 C>T	p.R503W	R	AD	~ 10 y.o.	Pure	Known 37
Exon 15	c.1646 insT	p.L549Lfs	D	AD	34 y.o.	Pure	Novel
Exon 15	c.1646 insT	p.L549Lfs	D	AD	47 y.o.	Pure	Novel
Exon 15	c.1646 T>C	p.L549P	R	AD	< 15 y.o.	Pure	Novel
Exon 15	c.1685 G>A	p.R562Q	R	AD	~ 10 y.o.	Pure	Known 38
Exon 17	c.1741 C>T	p.R581*	R	AD	14 y.o.	Pure	Known 39
Promoter ~ intron 1	del Chr2: 32136286–32145830 (9545 bp)	Del ex1	aCGH	AD	40 y.o.	Pure	Novel
Intron 1 ~ 3' region	(> 170 kb)	Del ex2-17	aCGH	Affected sibling	24 y.o.	Pure	Novel
Intron 16 ~ 3' region	del Chr2: 32231942–32290423 (58 482 bp)	Del ex17	aCGH	AD	58 y.o.	Pure	Novel
Intron 16 ~ 3'-UTR	del Chr2: 32229622–32234715 (5094 bp)	Del ex17	aCGH	AD	52 y.o.	Pure	Novel
Exon 4 ~ intron 7	dup Chr2: 32177411–32199467 (22 057 bp)+insAGT	Tandem duplication (part of ex4–ex7)	aCGH	AD	< 6 y.o.	Pure	Novel
SPG3A							
Exon 12	c.1243 C>T	p.R415W	R	AD	12 y.o.	Pure	Known 40
Exon 12	c.1483 C>T	p.R495W	R	Sporadic	~ 12 y.o.	Pure	Known 41
SPG8							
Exon 13	c.1749 A>C	p.R583S	R	AD	50 y.o.	Pure	Novel
Intron 10 ~ exon 15	del Chr8: 126138189–126142822 (4634 bp)	Del exon 11–15	aCGH	AD	64 y.o.	Pure	Novel
SPG17							
Exon 2	c.107 G>A (c.299 G>A)	p.C36Y (p.C100Y)	R	AD	~ 10 y.o.	Complicated	Novel
SPG31							
Exon 2	c.87 insA	p.K30Kfs	D	AD	8 y.o.	Pure	Novel
Intron 3 ~ intron 5	del Chr2: 86326359–86338423 (12 065 bp)	Del exon 4–5	aCGH	AD	~ 12 y.o.	Pure	Novel
SPG11							
Intron 18	c.3291+1G>T	IVS18+1G->T (homozygous)	D	Consanguinity affected siblings	20 y.o.	Complicated	Known 42
Intron 18	c.3291+1G>T	IVS18+1G->T (homozygous)	D	Consanguinity affected sibling	25 y.o.	Complicated	Known 42
Intron 8 and intron 38	c.1735+2 delT, c.6999+5 delG	IVS8+2 delT, IVS38+5 delG	D, D	Sporadic	22 y.o.	Complicated	Novel
Exon 20 and exon 28	c.3491 G>A, c.4840 A>T	p.W1164*, p.K1614*	D, D	Sporadic	18 y.o.	Complicated	Novel
Intron 7 ~ intron 8 and exon25	del Chr15: 42709367–42715955 (6589 bp) c.4426 insAT	Del exon 8, p.C1476Yfs	aCGH	Affected sibling	2 y.o.	Complicated	Novel
SPG21							
Exon 4	c.322G>C	p.A108P (homozygous)	R	Familial	60 y.o.	Complicated	Novel

Abbreviations: aCGH, array-based comparative genomic hybridization analysis; AD, autosomal dominant; D, direct nucleotide sequence analysis; Del, deletion; dup, duplication; R, resequencing microarray analysis; UTR, untranslated region; y.o., years old.

+1 of nucleotides is the first A of the start codon (ATG). The NCBI36/hg18 assembly is used as the reference genome.