PLOS ONE

CORRECTION

Correction: Targeted high throughput sequencing in hereditary ataxia and spastic paraplegia

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The last four lines are missing from the legend for Table 3. Please see the complete legend and table here.

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Table 3. List of variants of uncertain significance.

Chr	Gene	Disorder	OMIM phenotype#	Individual identity	Genomic position (Hg19/ GRCH 37)	Transcript	cDNA position	Amino acid position	LOVD variant ID	Zygosity	PhyloP score, in-silico pathogenicity predictions, CADD	Allele frequency in ExAC	Number of affected individuals carrying the variant in the respective family	Main phenotype—additional features
1	*KCND3* [a]	SCA19 (AD)	607346	HCT-088	g.112322852T>C	NM_004980.4	c.1456A>G	p.(Thr486Ala)	162993	het	4.40/m,p/19.2	0.001461	1	comp AT–pyramidal and extrapyramidal signs
3	*ITPR1*	SCA15/ 29 (AD)	606658/ 117360	HCT-029	g.4735396G>A	NM_001168272.1	c.4207G>A	p.(Val1403Met)	162994	het	4.08/s,m,p/17.1	0.00004663	1	pure HSP—none
3	*ITPR1* [b]	SCA15/ 29 (AD)	606658/ 117360	HCT-077	g.4810224G>A	NM_001168272.1	c.5710G>A	p.(Glu1904Lys)	162995	het	3.68/s,m/13.3	0.000008432	1#	comp AT—early onset, spastic AT
11	*BSCL2*	SPG17 (AD)	270685	HCT-044	g.62462158C>A	NM_001122955.3	c.512G>T	p.(Arg171Leu)	162996	het	2.14/s,m,p/19.3	0.000008322	1	pure HSP—amyotrophy, neuropathy
11	*SPTBN2*	SCA5 (AD)	600224	HCT-086	g.66453485T>G	NM_00694.2	c.7030A>C	p.(Ser2344Arg)	162997	het	1.66/p/15.1	0.00001679	1#	comp AT—neuropathy
11	*SPTBN2* [c]	SCA5 (AD)	600224	HCT-071	g.66453406C>T	NM_00694.2	c.7109G>A	p.(Arg2370His)	162998	het	5.86/s,m,p/33	0.0001252	1#	pure AT—none
12	*KIF5A*	SPG10 (AD)	604187	HCT-082	g. 57970109C>T	NM_004984.2	c.2146C>T	p.(Arg716Trp)	162999	het	3.60/s,m,p/24.6	0.00005826	1#	comp AT—episodic
15	*TTBK2*	SCA11 (AD)	604432	HCT-115	g.43132604C>G	NM_173500.3	c.245G>C	p.(Gly82Ala)	163000	het	5.21/s,m,p/16	0.0002898	1	comp AT—spastic AT
16	*BEAN1*	SCA31 (AD)	117210	HCT-087	g.66503607T>A	NM_001178020.2	c.128T>A	p.(Ile43Lys)	163001	het	3.35/s,m,p/25.3	-	2	comp AT—lower limb paresis, neuropathy
19	*RTN2*	SPG12 (AD)	604805	HCT-057	g.45996535C>A	NM_005619.3	c.916G>T	p.(Val306Phe)	163002	het	2.71/p/15.6	-	3	pure AT—none

Abbreviations: Chr, chromosome; AD, autosomal dominant; AR, autosomal recessive; OMIM, online Mendelian inheritance in man; cDNA, complementary deoxyribonucleic acid; Zygosity, heterozygous (het), compound heterozygous (c.het), homozygous (hom); LOVD, Leiden open variation database; CADD, combined annotation dependent depletion score, also called as a PHRED score PhyloP, evolutionary conservation score at specific nucleotide position; s, damaging prediction by SIFT (http://sift.jcvi.org); m, damaging prediction by MutationTaster (http://www.mutationtaster.org); p, damaging prediction by PolyPhen-2 (http://genetics.bwh.harvard.edu/pph2/); ExAC, exome aggregation consortium (http://exac.broadinstitute.org); comp, complex; AT, ataxia; HSP, hereditary spastic paraplegia. OMIM gene identifiers: *KCND3* (605411), *ITPR1* (147265), *BSCL2* (606158), *SPTBN2* (604985), *KIF5A* (602821), *TTBK2* (611695).

a Another variant, c.929G>A, p.(Arg310Gln) in *CYP7B1* was found heterozygously in this individual.

b Another variant, c.2261C>T, p.(Pro754Leu) was found in *SPG7* heterozygously in this individual.

c Another variant, c.2228T>C, p.(Ile743Thr) was found in *SPG7* heterozygously in this individual.

#, No additional samples of affected and/or unaffected individuals were available for segregation analysis.

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Reference

1. Iqbal Z, Rydning SL, Wedding IM, Koht J, Pihlstrøm L, Rengmark AH, et al. (2017) Targeted high throughput sequencing in hereditary ataxia and spastic paraplegia. PLoS ONE 12(3): e0174667. https://doi.org/10.1371/journal.pone.0174667. https://doi.org/10.1371/journal.pone.0174667 PMID: 28362824