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CORRIGENDUM

Corrigendum: Genetic diagnosis of Duchenne/Becker muscular dystrophy using next-generation sequencing: validation analysis of *DMD* mutations

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Correction to: *Journal of Human Genetics* (2016) **61**, 483-489; doi:10.1038/jhg.2016.7; published online 25 February 2016

Incorrect description in the above article was noticed after its publication.

1. In Table 1, we showed 'Patient no. 30: Ex20 c.2612A>C, p.Lys871Thr, Missense'.

2. Although the patient had this variant, he was later diagnosed as having facioscapulohumeral muscular dystrophy (FSHD).
In the section Materials and methods: Patients, we want to add these sentences written in italic.

Patients
We chose 67 cases with different mutations to test the clinical utility of our method: 17 cases with common deletions, 20 cases with common duplications (two of them with discontinuous double duplications) and 30 cases with point mutations, including nonsense mutations, deletions, insertions, splice regions and missense mutations (Table 1). *Patient 30 had a variant with unknown significance in DMD. He was later found to have a pathogenic 10 kb D4Z4 allele at 4q35 by Southern blot analysis and diagnosed as facioscapulohumeral muscular dystrophy (FSHD) after publication. Although his muscle pathology was reported from other hospital described faint and patchy staining pattern, the variant c.2612A>C is likely to be a benign polymorphism.* All deletion/duplication patterns are frequently diagnosed by MLPA in our laboratory, as shown in Supplementary Tables S1 and S2 and Supplementary Figures S1 and S2. Among 30 small mutations, 10 mutations were located within five or more consecutive bases (P16, 18, 19, 22, 25, 26, 27, 28, 29 and 30) and 5 mutations were deletion/insertion of more than nine bases (P12, 17, 21, 23 and 24). These mutations were predicted to be difficult to detect by Ion Torrent sequencer. All clinical information and materials used in this study were obtained for diagnostic purposes with written informed consent. This study was approved by the ethics committee of the National Center of Neurology and Psychiatry.

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

The authors would like to apologize for this mistake.

Table 1 67 cases with different mutations diagnosed by MLPA or Sanger sequencing

	Exon ID	*Nucleotide change*	*Protein change*	*Mutation*
1	EX25	c.3408_3409delinsGT	p.Gln1137*	Small insertions
2	EX3	c.160_162delCTC	p.Leu54del	Small deletions
3	EX44	c.6337delA	p.Ile2113*	Small deletions
4	EX21	c.2674delA	p.Ile892Phefs*4	Small deletions
5	EX53	c.7693C>T	p.Gln2565*	nonsense
6	EX6	c.434G>C	p.Arg145Pro	missense
7	EX69	c.10011C>G	p.Cys3337Trp	missense
8	EX66	c.9568C>T	p.Arg3190	nonsense
9	E27	c.3765dupT	p.Gly1256Trpfs*15	Small insertions
10	E28	c.3909dupT	p.Glu1304*	Small insertions
11	int2	c.94-1G>T		Splice donor variant
12	E59	c.8821_8822insAGGCCACTTCAAG	p.Asp2944Glyfs*6	Small insertions
13	E68	c.9816dupT	p.Lys3273*	Small insertions
14	int3	c.187-10_187-6delTTGTT		Splice region variant
15	E20	c.2591delC	p.Ser800Argfs*9	Small deletions
16	E22	c.2808dupT	p.Asp937*	Small insertions
17	EX14	c.1632-15_1639dup52	p.Thr547Ilefs*16	Small insertions
18	E21	c.2673_2674delAAinsG	p.Ile892Phefs*4	Small insertions
19	E74	c.10453_10454delCT	p.Leu3485Glufs*5	Small deletions
20	int54	c.8218-2A>G		Splice donor variant
21	E20	c.2430_2443delCCGGTGGATCGAAT	p.Arg811Leufs*6	Small deletions
22	E48	c.6986delA	p.Lys2329Serfs*9	Small deletions
23	E26	c.3454_3479del26	p.Leu1152Lysfs*17	Small deletions
24	E48	c.6923_6933del11	p.Ala2308Glufs*6	Small deletions
25	E74	c.10454delT	p.Leu3485Argfs*11	Small deletions
26	E24	c.3257dupA	p.Gln1087Alafs*11	Small insertions
27	E74	c.10453dupC	p.Leu3485Prpfs*6	Small insertions
28	E38	c.5413dupG	p.Val1805Glyfs*10	Small insertions
29	E15	c.1732A>T	p.Lys578*	nonsense
30[a]	E20	c.2612A>C	p.Lys871Thr	Missense variant
31	EX45			del
32	EX45-47			del
33	EX45-48			del
34	EX45-50			del
35	EX45-52			del
36	EX45-55			del
37	EX48-50			del
38	EX48-52			del
39	EX49-50			del
40	EX51			del
41	EX45-53			del
42	EX45-54			del
43	EX48			del
44	EX50-52			del
45	EX2-17			del
46	EX5-47			del
47	EX8-28			del
48	EX2			dup
49	EX2-7			dup
50	EX3-7			dup
51	EX3-13			dup
52	EX8,9			dup
53	EX8-11			dup
54	EX8-17			dup
55	EX17-19			dup
56	EX49-50			dup
57	EX50-55			dup
58	EX3-9			dup
59	EX3-30			dup
60	EX8-41			dup

Table 1 (Continued)

	Exon ID	*Nucleotide change*	*Protein change*	*Mutation*
61	EX18-48			dup
62	EX28-55			dup
63	EX56-67			dup
64	EX8-29			dup
65	EX34-44			dup
66	EX2-6, EX10-18			dup
67	EX45-53, EX56-60			dup

Abbreviations: del, deletion; dup, duplication; MLPA, multiplex ligation-dependent probe amplification.
[a]P30 has this variant; however, he was diagnosed as having FSHD after its publication.