

Additional file 1: Clinical and genetic information of patients with small mutation.

Patient	Age	Diagnosis	Ambulatory	Age at loss of ambulation	Mutation type	Exon	DNA change	Protein(predicted)	Report	Carrier status of mother
1	11.8	DMD	Yes		Nonsense	44	c.6370G>T	p.Glu2124*	unreported	Unknown
2	22.6	DMD	No	11.0	Nonsense	41	c.5889C>T	p.Arg1967*	reported	Yes
3	8.4	DMD	Yes		Nonsense	8	c.829C>T	p.Gln277*	reported	Yes
4	11.6	DMD	Yes		Small del	40	c.5602_5605delAGAA	p.Arg1868Glufs*5	reported	Unknown
5	12.9	DMD	No	9.0	Nonsense	55	c.8038C>T	p.2680Arg*	reported	Yes
6	8.7	DMD	Yes		Splicing	int32	c.4518+2T>C		reported	Yes
7	13.0	DMD	No	11.0	Small del	10	c.1076delA	p.Glu359Glyfs*17	unreported	Unknown
8	14.2	DMD	No	12.0	Nonsense	65	c.9445C>T	p.Gln3149*	reported	Unknown
9	10.4	DMD	Yes		Nonsense	59	c.8713C>T	p.Arg2905*	reported	Unknown
10	9.1	DMD	Yes		Nonsense	10	c.998C>A	p.Ser333*	reported	Yes
11	9.1	DMD	Yes		Nonsense	12	c.1465C>T	p.Gln489*	reported	Yes
12	7.6	DMD	Yes		Small del	56	c.8227delG	p.Gly2743Valfs*21	unreported	Yes
13	9.9	DMD	Yes		Nonsense	8	c.826C>T	p.Gln276*	reported	Unknown
14	13.3	DMD	No	9.0	Small del	6	c.522_523delTA	p.His174Glnfs*3	unreported	No
15	12.7	DMD	No	9.0	Nonsense	22	c.2832T>G	p.Tyr944*	unreported	Unknown
16	11.3	DMD	Yes		Nonsense	23	c.3087G>A	p.Trp1029*	reported	Yes
17	8.7	DMD	Yes		Nonsense	6	c.433c>T	p.Arg145*	reported	Unknown
18	8.1	DMD	Yes		Splicing	56	c.8390G>A		unreported	Unknown
19	6.0	DMD	Yes		Nonsense	58	c.8608C>T	p.Arg2870*	reported	Yes
20	9.5	DMD	Yes		Splicing	int17	c.2169-2A>G		reported	Unknown
21	9.0	DMD	Yes		Small del	12	c.1412delC	p.Thr471Lysfs*16	unreported	Yes
22	8.2	DMD	Yes		Nonsense	27	c.3622C>T	p.Gln1208*	reported	Unknown
23	8.9	DMD	Yes		Small del	70	c.10101_10103delAG	p.Glu3367del	reported	No
24	10.5	DMD	Yes		Nonsense	30	c.4117C>T	p.Gln1373*	reported	Unknown
25	12.5	IMD	Yes		Nonsense	5	c.336G>A	p.Trp112*	reported	Yes
26	14.0	DMD	No	7.0	Nonsense	59	c.8812G>T	p.Glu2938*	unreported	Unknown
27	10.6	DMD	Yes		Nonsense	56	c.8475G>A	p.Trp2915*	unreported	Unknown
28	12.0	DMD	No	9.0	Small del	45	c.6439-104253_6615-13966del	p.Glu2147Alafs*17	reported	Unknown
29	9.0	DMD	Yes		Nonsense	70	c.10108C>T	p.Arg3370*	reported	Unknown
30	10.1	DMD	Yes		Nonsense	14	c.1684C>T	p.Gln562*	reported	Yes
31	9.2	DMD	Yes		Nonsense	8	c.748G>T	p.Glu250*	reported	No
32	14.4	DMD	No	11.0	Small del	13	c.1593delA	p.Glu531Aspfs*52	unreported	No
33	11.6	DMD	No	7.0	Nonsense	64	c.9337C>T	p.Arg3113*	reported	Unknown
34	9.0	DMD	Yes		Small del	53	c.7755delG	p.Trp2585Cysfs*12	unreported	Yes
35	10.1	DMD	Yes		Nonsense	6	c.433c>T	p.Arg145*	reported	Unknown
36	5.6	DMD	Yes		Nonsense	44	c.6423C>A	p.Tyr2141*	reported	No
37	10.1	DMD	Yes		Splicing	int2	c.44-1G>T		unreported	Yes
38	14.3	IMD	Yes		Small ins	20	c.2462-2463insA	p.Glu821Glufs*	unreported	Unknown
39	12.9	IMD	Yes		Nonsense	7	c.583C>T	p.Arg195*	reported	Unknown
40	13.0	IMD	Yes		Nonsense	70	c.10141C>T	p.Arg3381*	reported	Unknown
41	11.9	DMD	No	9.0	Nonsense	58	c.8608C>T	p.Arg2870*	reported	No
42	7.2	DMD	Yes		Small del	51	c.7341delA	p.Gln2447Hisfs*13	unreported	Yes
43	12.2	DMD	No	9.0	Nonsense	55	c.8038C>T	p.Arg2680*	reported	Unknown
44	7.2	DMD	Yes		Splicing	int70	c.10228+1G>A		reported	Unknown
45	14.9	DMD	No	12.0	Nonsense	57	c.8420G>A	p.Trp2807*	reported	Unknown
46	7.7	DMD	Yes		Nonsense	25	c.3328G>T	p.Glu1110*	reported	Yes
47	6.7	DMD	Yes		Small del	23	c.2962delT	p.Ser988Leufs*16	unreported	Yes
48	9.7	DMD	Yes		Nonsense	66	c.9568C>T	p.Arg3190*	reported	Yes
49	9.5	DMD	Yes		Small del	16	c.1872delA	p.Lys624Asnfs*8	unreported	Unknown
50	9.4	DMD	Yes		Small del	13	c.1533-1536del	p.His512Trpfs*4	reported	Unknown
51	8.6	DMD	No	8.0	Nonsense	35	c.4870C>T	p.Gln1624*	reported	No
52	12.6	IMD	Yes		Nonsense	30	c.4142T>G	p.Leu1381*	unreported	No
53	9.8	DMD	Yes		Splicing	int10	c.1150-2A>G		unreported	Yes
54	10.5	DMD	Yes		Small ins	42	c.6077-6078insA	p.Asp2026Glufs*4	unreported	Unknown
55	11.1	DMD	Yes		Small ins	27	c.3772dupT	p.Cys1258Leufs*1	unreported	Unknown
56	6.6	DMD	Yes		Splicing	int11	c.1114-2A>T		unreported	Unknown
57	4.8	DMD	Yes		Nonsense	23	c.3151C>T	p.Arg1051*	reported	Unknown
58	7.1	DMD	Yes		Small del	33	c.4583delA	p.Gln1528Argfs*18	unreported	Unknown
59	4.3	DMD	Yes		Small del	26	c.3466-3467del	p.Val1156Lysfs*7	unreported	Yes
60	11.7	DMD	Yes		Small ins	33	c.4561-4562insA	p.Met1521Asnfs*2	unreported	Unknown
61	6.4	DMD	Yes		Nonsense	33	c.4567G>T	p.Glu1523*	unreported	Unknown
62	15.9	DMD	No	12.0	Nonsense	33	c.4660G>T	p.Glu1554*	unreported	Unknown
63	20.8	IMD	No	14.0	Nonsense	16	c.1843C>T	p.Gln615*	reported	Unknown
64	8.2	DMD	Yes		Small ins	3	c.130dupC	p.Leu44Profs*45	unreported	Unknown
65	7.3	DMD	Yes		Nonsense	32	c.4483C>T	p.Gln1495*	reported	Unknown
66	11.8	DMD	No	8.0	Splicing	int56	c.8390+1G>A		reported	Unknown
67	6.2	DMD	Yes		Nonsense	63	c.9249G>A	p.Trp3083*	unreported	No

68	10.4	DMD	Yes		Nonsense	32	c.4414C>T	p.Gln1472*	reported	Yes
69	8.9	DMD	Yes		Nonsense	26	c.3580C>T	p.Gln1194*	reported	Unknown
70	6.8	DMD	Yes		Nonsense	20	c.2527G>T	p.Glu843*	reported	Unknown
71	15.0	IMD	Yes		Nonsense	17	c.2006T>C	p.Val669*	unreported	Yes
72	10.8	DMD	No	7.0	Nonsense	41	c.5899C>T	p.Arg1967*	reported	Yes
73	14.0	IMD	Yes		Nonsense	21	c.2758c>T	p.Gln920*	reported	Unknown
74	13.3	DMD	No	9.0	Nonsense	16	c.1938T>A	P.Cys646*	unreported	Yes
75	8.5	DMD	Yes		Nonsense	12	c.1339C>T	p.Gly447*	unreported	Yes
76	9.6	DMD	Yes		Small del	17	c.2034delG	p.Thr679Glnfs*3	unreported	Yes
77	5.5	DMD	Yes		Nonsense	33	c.4618G>T	p.Glu1540*	reported	Unknown
78	9.9	DMD	Yes		Splicing	int46	c.6762+2T>A		unreported	Yes
79	9.1	DMD	Yes		Nonsense	7	c.583C>T	p.Arg195*	reported	No
80	8.5	DMD	Yes		Small del	24	c.3205delG	p.Val1069Phefs*3	unreported	Yes
81	9.9	DMD	Yes		Splicing	int27	c.3417+1G>T		unreported	Yes
82	4.3	DMD	Yes		Nonsense	7	c.583C>T	p.Arg195*	reported	Unknown
83	6.3	DMD	Yes		Nonsense	19	c.2302C>T	p.Arg768*	reported	Unknown
84	5.2	DMD	Yes		Nonsense	22	c.2869C>T	p.Gln957*	reported	No
85	3.0	DMD	Yes		Small ins	12	c1427-1428insGA	p.Glu476Lysfs*11	unreported	Unknown
86	5.7	DMD	Yes		Nonsense	11	c.1255C>T	p.Gln419*	reported	Yes
87	4.9	DMD	Yes		Nonsense	6	c.433C>T	p.Arg145*	reported	Unknown
88	4.3	DMD	Yes		Nonsense	50	c.7288G>T	p.Gly2430*	unreported	Yes
89	3.3	DMD	Yes		Nonsense	3	c.119A>T	p.Lys1707*	unreported	Yes
90	4.1	DMD	Yes		Missense	55	c.7828C>A	p.Pro2610Thr	unreported	Yes
91	12.1	DMD	No	10.0	Small del	16	c.1979delA	p.Lys660Argfs*17	unreported	No
92	2.3	DMD	Yes		Nonsense	60	c.9029C>G	p.Ser3010*	reported	Yes
93	5.8	DMD	Yes		Nonsense	19	c.2368C>T	p.Gln790*	reported	Unknown
94	7.0	DMD	Yes		Nonsense	58	c.8608c>T	p.Arg2870*	reported	Yes
95	2.7	DMD	Yes		Splicing	int5	c.334-1G>A		unreported	Yes
96	3.2	DMD	Yes		Small del	42	c.6006_6007del	p.Glu2003Asnfs*1	unreported	Unknown
97	1.8	DMD	Yes		Nonsense	19	c.2302C>T	p.Arg768*	reported	Yes
98	9.9	DMD	Yes		Splicing	int46	c.6742+3T>A		unreported	Yes
99	2.3	DMD	Yes		Nonsense	24	c.3259C>T	p.Gln1087*	reported	Yes
100	11.0	DMD	Yes		Nonsense	25	c.3426C>A	p.Cys1142*	unreported	Unknown
101	5.3	DMD	Yes		Missense	6	c.496G>T	p.Gly166cys	unreported	No
102	2.9	DMD	Yes		Splicing	int52	c7661-1G>A		reported	Yes
103	6.7	DMD	Yes		Nonsense	20	c.2605C>T	p.Gln869*	reported	Yes
104	7.8	DMD	Yes		Nonsense	44	c.6292C>T	p.Arg2098*	reported	Yes
105	9.6	DMD	Yes		Nonsense	22	c.2832T>A	p.Tyr944*	unreported	No
106	12.9	IMD	Yes		Nonsense	27	c.3769A>T	p.Lys1257*	unreported	Unknown
107	8.0	DMD	Yes		Nonsense	10	c.984C>T	P.Arg1662*	unreported	Yes
108	3.2	DMD	Yes		Nonsense	26	c.3580C>T	p.Gln1194*	reported	Yes
109	10.1	DMD	Yes		Nonsense	18	c.2260C>T	p.Arg754*	unreported	Yes
110	11.7	DMD	Yes		Nonsense	10	c.1062G>A	p.Trp354*	reported	Unknown
111	6.9	DMD	Yes		Nonsense	30	c.4108C>T	p.Gln1370*	reported	Unknown
112	10.6	DMD	Yes		Nonsense	9	c.853G>T	p.Gly285*	reported	Unknown
113	10.0	DMD	No	9.0	Nonsense	43	c.6256G>T	p.Glu2086*	unreported	Yes
114	5.3	DMD	Yes		Nonsense	57	c.8443C>T	p.Gln2815*	reported	No
115	6.8	DMD	Yes		Nonsense	55	c.8194A>T	p.Lys2732*	reported	No
116	9.7	DMD	Yes		Splicing	int5	c.358-2A>G		reported	Yes
117	17.1	BMD	Yes		Nonsense	53	c.7672C>T	p.Gln2558*	reported	No
118	4.8	DMD	Yes		Nonsense	7	c.541C>T	p.Gln181*	unreported	No
119	6.9	DMD	Yes		Nonsense	17	c.1997C>A	p.ser666*	unreported	Unknown
120	3.2	DMD	Yes		Nonsense	25	c.3061C>T	p.Gln1021*	unreported	Yes
121	12.5	IMD	Yes		Nonsense	5	c.336G>A	p.Trp112*	reported	Yes
122	11.0	DMD	Yes		Nonsense	7	c.583C>T	p.Arg195*	reported	Unknown
123	13.9	DMD	No	12.0	Nonsense	2	c.77A>G	p.Asn26*	reported	Yes
124	13.5	IMD	Yes		Nonsense	25	c.3028G>T	p.Glu1010*	unreported	Yes
125	5.6	DMD	Yes		Nonsense	32	c.4405C>T	p.Gln1469*	reported	Yes
126	1.8	DMD	Yes		Small del	42	c.5996_5999del	p.Thr1999Ilefs*2	unreported	Yes
127	11.2	DMD	Yes		Nonsense	39	c.5530C>T	p.Arg1844*	reported	Yes
128	16.3	IMD	No	13.0	Splicing	int21	c.2804-1G>T		reported	No
129	7.4	DMD	Yes		Nonsense	6	c.433C>T	p.Arg145*	reported	Yes
130	7.0	DMD	Yes		Nonsense	11	c.1305G>A	p.Ser443*	unreported	Unknown
131	10.8	DMD	Yes		Small del	13	c.1593delA	p.Glu531Aspfs*52	unreported	Yes
132	7.7	DMD	Yes		Small ins	13	c.1540dupG	p.val514Glyfs*5	reported	Unknown
133	7.1	DMD	Yes		Nonsense	44	c.6310G>T	p.Glu2104*	reported	Unknown
134	5.5	DMD	Yes		Nonsense	59	c.8713C>T	p.Arg2905*	reported	Unknown
135	4.7	DMD	Yes		Small del	16	c.1978_1979del	p.Lys660Glufs*59	reported	Unknown
136	9.6	DMD	Yes		Nonsense	18	c.2213C>G	p.Ser738*	unreported	Unknown
137	7.4	DMD	Yes		Nonsense	44	c.6436A>T	p.Lys2146*	unreported	Yes
138	7.8	DMD	Yes		Small del	40	c.5697del	Lys1899Asnfs*2	reported	Yes

139	2.7	DMD	Yes		Missense	27	c.3774C>G	p.Cys1258Trp	unreported	Yes
140	5.2	DMD	Yes		Nonsense	14	c.1702C>T	p.Gln568*	reported	Yes
141	5.5	DMD	Yes		Nonsense	44	c.6292C>T	p.Arg2093*	unreported	Yes
142	2.6	DMD	Yes		Nonsense	66	c.9568C>T	p.Arg3190*	reported	Yes
143	2.8	DMD	Yes		Nonsense	7	c.559C>T	p.Gln187*	unreported	Yes
144	8.1	DMD	Yes		Splicing	int56	c.8300-IG>C		unreported	No
145	9.7	DMD	Yes		Nonsense	15	c.1777C>T	p.Gln593*	reported	No
146	9.1	DMD	Yes		Small del	8	c.676-678delAAG	p.Lys226del	reported	No
147	12.2	IMD	Yes		Small ins	3	c.115-117 insA	p.Asn39Lysfs *5	unreported	Unknown
148	9.3	DMD	Yes		Nonsense	44	c.6373c>T	p.Gln2125*	reported	Yes
149	14.8	DMD	No	8.0	Small del	47	c.6807-6810delACAA	p.Leu2270Metfs*9	unreported	Unknown
150	11.0	DMD	Yes		Nonsense	55	c.8215C>T	p.Gln2739*	unreported	Unknown
151	9.7	DMD	Yes		Nonsense	6	c.436C>T	p.Gln146*	reported	Yes
152	10.0	DMD	Yes		Small del	52	c.7559delT	p.Leu2520Trpfs*18	unreported	Yes
153	12.6	IMD	Yes		Small del	20	c.2471_2474del	p.Asn824Serfs*21	unreported	Yes
154	9.9	DMD	Yes		Nonsense	34	c.4732A>T	P.Lys1578*	unreported	Unknown
155	11.9	DMD	No	8.0	Nonsense	58	c.8608C>T	p.Arg2870*	reported	Unknown
156	8.9	DMD	Yes		Missense	37	c.5234G>A	p.Arg1745His	reported	Unknown
157	8.5	DMD	Yes		Small ins	16	c.1898dupA	Asn633Lysfs*2	unreported	Yes
158	11.5	DMD	Yes		Small del	70	c.10101_10103del	p.Glu3367del	reported	Yes
159	7.3	DMD	Yes		Missense	1	c.1A>T	p.Met1Leu	unreported	Unknown
160	10.3	DMD	Yes		Small ins	70	c.1648dupC	p.Arg550Profs*17	unreported	Yes
161	9.4	DMD	Yes		Small del	13	c.1533_1536del	His512Trpfs*4	reported	Unknown
162	10.2	DMD	Yes		Small ins	69	c.10027_10028insT	p.Ser3343Phefs*9	unreported	Unknown
163	10.7	DMD	Yes		Splicing	int56	c.8391-1G>A		reported	Yes
164	12.8	IMD	Yes		Missense	22	c.2949G>T	p.Gln983His	reported	Unknown
165	6.2	DMD	Yes		Nonsense	41	c.5851C>T	p.Gln1951*	reported	Unknown
166	10.3	DMD	Yes		Small ins	74	c.10453-10454insC	p.Leu3485Profs*6	unreported	Unknown
167	13.1	IMD	Yes		Nonsense	14	c.1702C>T	p.Gln568*	reported	Yes
168	7.1	DMD	Yes		Nonsense	10	c.998C>A	p.Ser333*	reported	Yes
169	10.6	DMD	Yes		Nonsense	14	c.1638G>A	p.Trp546*	reported	Yes
170	24.7	BMD	Yes		Splicing	int18	c.2254-2A>T		unreported	Yes
171	6.0	DMD	Yes		Small del	8	c.676-678delAAG	p.Lys226del	reported	Unknown
172	12.9	IMD	Yes		Splicing	int20	c.2435-2A>T		unreported	Unknown
173	11.0	DMD	Yes		Small ins	20	c.2462-2463insA	p.Glu821Glufs*4	unreported	Unknown
174	10.9	DMD	Yes		Nonsense	23	c.3034delC	p.Leu1012*	unreported	Unknown
175	7	DMD	Yes		Nonsense	11	c.1305G>A	p.Ser443*	unreported	No
176	12.0	IMD	Yes		Nonsense	43	c.6223C>T	p.Gln2075*	reported	No
177#	6.6	DMD	Yes		Splicing	int13	c.1114-1 G>A*		unreported	Yes
178#	6.6	DMD	Yes		Splicing	int13	c.1114-1 G>A*		unreported	Yes
179	10.9	DMD	Yes		Small ins	22	c.2864dupT	Gln956Profs*13	unreported	Yes
180	13.9	IMD	Yes		Nonsense	20	c.2518G>T	p.Gln840*	reported	Unknown
181	8.3	DMD	Yes		Splicing	int6	c.531-2A>G		unreported	Unknown
182	37.5	BMD	Yes		Nonsense	1	c.11G>A	p.Trp4*	reported	Unknown
183	4.6	DMD	Yes		Nonsense	1	c.12G>A	p.Trp4*	unreported	Yes
184	6.3	DMD	Yes		Nonsense	19	c.2302C>T	p.Arg768*	reported	Yes
185	5.7	DMD	Yes		Nonsense	11	c.1255C>T	p.Gln419*	unreported	Yes
186	8.9	DMD	Yes		Nonsense	60	c.8944C>T	p.Arg2982*	reported	Yes
187	4.9	DMD	Yes		Nonsense	6	c.433C>T	p.Arg145*	reported	Yes
188	6.0	DMD	Yes		Nonsense	58	c.8608C>T	p.Arg2870*	reported	Yes
189	11.2	DMD	Yes		Nonsense	39	c.5530C>T	p.Arg1844*	reported	Unknown
190	7.3	DMD	Yes		Nonsense	35	c.4996C>T	p.Arg1666*	reported	Unknown
191	6.4	DMD	Yes		Nonsense	35	c.4561G>T	p.Glu1523*	unreported	Yes
192	5.5	DMD	Yes		Nonsense	33	c.4618G>T	p.Glu1540*	reported	Yes
193	10.1	DMD	Yes		Nonsense	18	c.2260C>T	p.Arg754*	unreported	Yes
194	11.0	DMD	Yes		Nonsense	32	c.4375C>T	p.Arg1459*	reported	Unknown
195	8.0	DMD	Yes		Nonsense	10	c.984C>T	p.Arg1662*	unreported	Yes
196	4.3	DMD	Yes		Nonsense	7	c.583C>T	p.Arg195*	reported	Yes
197	9.2	DMD	No	9.0	Small del	33	c.4587delG	p.Lys1529Asnfs*13	reported	Yes

The patients with # are brothers.