

Comprehensive genetic diagnosis of patients with Duchenne/ Becker muscular dystrophy (DMD/BMD) and pathogenicity analysis of splice site variants in the *DMD* gene^{*#}

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Table S1 Oligonucleotide primers for detection of mutations in *DMD* gene

Exon	Size(bp)	Forward primer sequence	Reverse primer sequence
1	479	5'-TTTAAAGCCTACTGGAGC-3'	5'-TACGGAAGTATCAACTAATC-3'
2	518	5'-TGGCTGGTCTCAAACACTAC-3'	5'-TTCTGCTGCTTACTCCTT-3'
3	615	5'-CCCAGGTAGATTGTGGTC-3'	5'-TCATACGAGGTTGCTTTA-3'
4	541	5'-TTTCTGTCATTGGTGGTC-3'	5'-CACAATCAGGCATACACG-3'
5	552	5'-GGGTGCTTGTGTTGTTA-3'	5'-CCTATGATTGGGACTT-3'
6	532	5'-TCTCATTACTAATGGCCCTAA-3'	5'-CACTTTCACGCTCCGCTA-3'
7	686	5'-GACTATGGGCATTGGTTG-3'	5'-ACAGAAGTAGTGGCAGGA-3'
8	277	5'-TTCATTATCCTTTAGAGTC-3'	5'-ATAGTAGCTGTCTTTACAC-3'
9	712	5'-TGCCGGATTGAAGAGTAC-3'	5'-CCATTAGCAGCCTTTGTG-3'
10	681	5'-CTCCCTATTGTCTGTATCTG-3'	5'-GAAGCCAAACGAGTGTAT-3'
11	408	5'-GGGAATATAAATCTAAATGGCCACAA-3'	5'-GTGTGCCTTGGGAACAACTGAGA-3'
12	498	5'-GGGGTGACTGATAGTGGCCTTAC-3'	5'-GGGGACTTATCAAGCCATTGC-3'
13	477	5'-GCAGAAAATTGGCTTGAATGGTT-3'	5'-CCCATCCGAGTTAGTTACTATTG-3'
14	341	5'-GCGTACATAGGAGACTGA-3'	5'-TCTGAAAGCCATGCACTA-3'
15	679	5'-CTCAAATGGCAACGTCTT-3'	5'-CCAGTCCCAGTCTCAGGT-3'
16	552	5'-CCAGGCTTATTCTGTGAT-3'	5'-ATGTTCCCTCGTGGTTCTA-3'
17	438	5'-TTTTCTTTGCCACTCCA-3'	5'-TTGCGATAGTGATTTCTGTGA-3'
18	452	5'-GTCAGGCAGGAGTCTCAG-3'	5'-GGCATCCCTAGTCAGTCA-3'
19	686	5'-GAAAGGGCAAGTAGAAGT-3'	5'-GTAAAGGTTATCCCGAAG-3'
20	475	5'-TCAGTCTGTGGTTTCAGG-3'	5'-TCCGGGTATCTACGTCTT-3'
21	469	5'-ATGGCTGGTGATAGAGGC-3'	5'-AGGTTGTAGGGAGAATGG-3'
22	414	5'-TACAGGCTGATTTGTGAA-3'	5'-GATAAGCGTGCTTTATTG-3'
23	363	5'-ATTATTCATCAATTAGGGTAAATG-3'	5'-GATGCTGAAGGTCAAATGCTTATG-3'
24	682	5'-AGGCTATAAAGACACCAT-3'	5'-GTAGCATCTAACACCAC-3'
25	635	5'-AAATCCACCTCACAATAAC-3'	5'-AAAGTAACGGTGAAGGGA-3'
26	453	5'-CTTATGGGAAATTGAGTG-3'	5'-AGAACCAGGAAAGAGCAG-3'
27	515	5'-ACAAAT5'-CCATACCTCCAT-3'	5'-TAACCACTATGCCTCACA-3'
28	479	5'-GACTGCAATGTTGGTGCTTA-3'	5'-TATTTGGTACTTGACCTCTTTTA-3'
29	509	5'-AGTTCTCAGTCCGCAACA-3'	5'-CTGGCATTGGATTGTCTC-3'

30	699	5'-GTGATGACGATGACCCTA-3'	5'-CTTCCTACCTACCTCCAA-3'
31	638	5'-GTAGAGGTGGTTGAGGAG-3'	5'-ATCGAAACTTCATGGAGAC-3'
32	636	5'-GCTGCCCAAAGAGTCCTG-3'	5'-TGCGTATTTGCCACCAGA-3'
33	328	5'-ATGGAATAGCAATTAAGGGGATCT-3'	5'-TTATTGCCCGTTGCTTTAC-3'
34	318	5'-AAAAGTTCCAAATAAGTGGTTATA-3'	5'-TTCCTTACAAAATCATATTATGTG-3'
35	482	5'-ATAGTTACCCAACAATGAAG-3'	5'-TGAATTAAGAGCCAGCAT-3'
36	602	5'-CCCAGCAGAAGAGTG-3'	5'-AAGGGAAAGATAAAGGAAG-3'
37	652	5'-TTGCTCACTCGCTCTGTT-3'	5'-ATCGGTAGTGCTTTATGTTG-3'
38	429	5'-TATCTTGCATGTATGTTTC-3'	5'-TCAGTTGGAGACTTATCT-3'
39	628	5'-AGGCTATGAGCACAGTATCT-3'	5'-TCGGTTATTCTGGCAGTTAG-3'
40	584	5'-AAGATGAGGGACGCAAAT-3'	5'-TGGGCTAACATGAGTAAGAAGT-3'
41	509	5'-GTTATTGAGCGAGGATGA-3'	5'-TACAGAAGCCCAAAGTGA-3'
42	596	5'-TTTTAGCCAGTTTGTGA-3'	5'-GTGGTTCCTTTCTGTCCC-3'
43	403	5'-AGAATTGCAACACCAATTTGCTACC-3'	5'-TGAGAGTGATACTTCTTTTTCCCTGTC-3'
44	509	5'-AATGCCAATAGTCCAAAATA-3'	5'-GTCCAGATGTGCTGAAGATA-3'
45	693	5'-TCGTATCCACGATCACTA-3'	5'-GCCTTTCACCCCTGCTTAT-3'
46	318	5'-GTTTGTGTCCAGTTTGCATTAAC-3'	5'-GGAGAAAACCAATGATTGAATTA-3'
47	579	5'-ACCCAAGAGGCTGATGAA-3'	5'-TGAAGCACCCAGGAAACA-3'
48	408	5'-TAAACATTTGGCTTATGCCTTGA-3'	5'-TGGTGCCTGTGCCTATTGTGGTTAT-3'
49	459	5'-TATGTACCAGGCAGAAAT-3'	5'-CACCAGGACAGAAATGAG-3'
50	530	5'-TATTTGTAGGGTGGTTGG-3'	5'-TTCCTTGTGGGTGTATCT-3'
51	398	5'-GGCTCTTTAGCTTGTGTTTC-3'	5'-GTGGGAAATGGTCTAGGAG-3'
52	676	5'-TCTCCATTTGAGCCTTTA-3'	5'-AGCCCAGATGACAACTAA-3'
53	390	5'-TTCCTCCAGACTAGCATTACTAC-3'	5'-CATTTACAGCTTTAACGTGATT-3'
54	268	5'-GACACTCCAAC TAGAGATTTTC-3'	5'-CCCATTATTACAGCCAAC-3'
55	482	5'-TATGAAACCTCCTCTGTG-3'	5'-GGAAATGCCTGACTTACT-3'
56	510	5'-CATTCATCGCTTGTTCCT-3'	5'-TAGCTGCCTCCTCATTT-3'
57	315	5'-ACATGGTACGCTGCTGTT-3'	5'-ATTGACCCCTGGGTGAGA-3'
58	529	5'-ACAAGTTCTGAGCACCCA-3'	5'-CAGGCTCTTAAATGGAAAA-3'
59	679	5'-GAGCCTGCCTGAAGTAA-3'	5'-TTGTGAAAGACGGACTGA-3'
60	361	5'-CCCTAAAGAGAATAAGCCCAGGTA-3'	5'-TCCTATCCTCACAAATATTACCATGAA-3'
61	614	5'-TGTACCCTCCCTAATGTTT-3'	5'-AATTGGCCTTCCTCTTCTA-3'
62	550	5'-TTGACCTCCTTGCCCTTC-3'	5'-TGCTGCTTTCTGCCTTTT-3'
63	541	5'-CACATTGGATGGGTGAAG-3'	5'-TAGGAAGGTGCCACTGCT-3'
64	579	5'-GAACTTAGCCTTCCTGGTT-3'	5'-AATTATGGATGCCGATGC-3'
65	548	5'-ATATCTTACCCAATGCC-3'	5'-TAGGTCCACAGCTTCCAG-3'
66	674	5'-CTCTAGGAAAGGGTCAGT-3'	5'-GAAATTCTGTAGGGTGGT-3'
67	382	5'-TGTGGAAATACTGGCTAC-3'	5'-TACTGCCTACTGAAGAGC-3'
68	458	5'-AGCCTTAGGAGTGGGTAG-3'	5'-GCAACTGGCACAGGAGAT-3'
69	485	5'-AGTCTGTGCTCCTGG-3'	5'-TGGCGTCAAACCTACC-3'
70	295	5'-GGCAGAAGACTGGAGTGGTCATTA-3'	5'-TGGGAGTGAAGGAGGGTGTTC-3'
71	236	5'-GCTATTGCTTTCCATGGTTCATAC-3'	5'-TAAACAGAACAAAAGAGAACCAAG-3'
72	539	5'-TCGGAATTATAGGATGTG-3'	5'-GTTAGAGGGCAAGTTGAT-3'
73	639	5'-GGGTAGTGTAAGGTAGGGAGT-3'	5'-CAAATAAGAGTAAGGCAAAC-3'

74	644	5'-AATACACTCCTGAGTCCCTA-3'	5'-CCAAGCCAATAACCCTAC-3'
75	652	5'-AAATCCTTTGCTGCTTAC-3'	5'-GATGCTTGTTGCACCTAT-3'
76	423	5'-CTAAAGTAGGAAGGGTCA-3'	5'-AGATAGAGCTGCCAGAAT-3'
77	508	5'-GGCAGAAGAATCGTTGA-3'	5'-ACACCAGTTGGGTAGGGA-3'
78	527	5'-AGTCCAGCCTCTATGTCG-3'	5'-GAGCCTGAATCTCACTAACC-3'
79	628	5'-TGACGATGGTTGGGTGCT-3'	5'-ATGCGGGAATCAGGAGTT-3'

Table S2 Deletions in *DMD* gene detected by MLPA

Patient NO.	Gender	Family history	MLPA result	Inheritance
P1	Male	No	EX45_54	<i>De novo</i>
P2	Male	No	EX45_49	<i>De novo</i>
P3	Male	Yes	EX45_53	Maternal
P4	Male	No	EX5_6*	Maternal
P5	Male	No	EX45_47	<i>De novo</i>
P6	Male	No	EX8_17	<i>De novo</i>
P7	Male	No	EX49_50	<i>De novo</i>
P8	Male	No	EX44_47	Maternal
P9	Male	Yes	EX48_49	Maternal
P10	Male	No	EX46_50	Maternal
P11	Male	No	EX51	Maternal
P12	Male	Yes	EX48_50	Maternal
P13	Male	No	EX8_16	Maternal
P14	Male	No	EX51	<i>De novo</i>
P15	Male	No	EX48_50	<i>De novo</i>
P16	Male	No	EX45_48	Maternal
P17	Male	No	EX48_52	<i>De novo</i>
P18	Male	Yes	EX45_53	Maternal
P19	Male	No	EX48_52	Maternal
P20	Male	No	EX51_53	Maternal
P21	Male	No	EX3	Maternal
P22	Male	No	EX48_52	<i>De novo</i>
P23	Male	No	EX46_48	Maternal
P24	Male	Yes	EX21	Maternal
P25	Male	No	EX45_52	<i>De novo</i>
P26	Male	No	EX50_52	<i>De novo</i>
P27	Male	No	EX50_52	<i>De novo</i>
P28	Male	No	EX19	<i>De novo</i>
P29	Male	Yes	EX52_54	Maternal
P30	Male	No	EX48_50	Maternal
P31	Male	No	EX42_43	Maternal
P32	Male	No	EX50	Maternal
P33	Male	No	EX44_47	Maternal
P34	Male	Yes	EX48_50	Maternal
P35	Male	No	EX46_51	<i>De novo</i>
P36	Male	No	EX20_22	<i>De novo</i>
P37	Male	No	EX3_49	<i>De novo</i>
P38	Male	No	EX48_52	Maternal
P39	Male	No	EX45_52	Maternal
P40	Male	No	EX14_19	<i>De novo</i>
P41	Male	No	EX45_54	<i>De novo</i>
P42	Male	No	EX45	Maternal

P43	Male	No	EX12_33 [*]	Maternal
P44	Male	No	EX46_52	Maternal
P45	Male	Yes	EX8_18	Maternal
P46	Male	No	EX50_52	<i>De novo</i>
P47	Male	No	EX45_47	Maternal
P48	Male	No	EX48_50	<i>De novo</i>
P49	Male	No	EX5_7	Maternal
P50	Male	No	EX49_50	Maternal
P51	Male	No	EX45_52	<i>De novo</i>
P52	Male	No	EX46_47	<i>De novo</i>
P53	Male	Yes	EX48_52	Maternal
P54	Male	Yes	EX46_47	<i>De novo</i>
P55	Male	No	EX51	<i>De novo</i>
P56	Male	No	EX3_17	<i>De novo</i>
P57	Male	No	EX45_52	<i>De novo</i>
P58	Male	No	EX45_50	Maternal
P59	Male	No	EX51	Maternal
P60	Male	No	EX45_47	<i>De novo</i>
P61	Male	No	EX49_53	Maternal
P62	Male	Yes	EX46_51	Maternal
P63	Male	No	EX45_48	Maternal
P64	Male	No	EX38_43	<i>De novo</i>
P65	Male	No	EX50	Maternal
P66	Male	No	EX49_50	Maternal
P67	Male	No	EX46_49	Maternal
P68	Male	No	EX46_49	Maternal
P69	Male	No	EX51	<i>De novo</i>

EX: exon; *: novel mutation.

Table S3 Duplications in *DMD* gene detected by MLPA

Patient NO.	Grander	Family history	MLPA result	Inheritance
P70	Male	No	EX3_16	Maternal
P71	Male	Yes	EX2	Maternal
P72	Male	No	EX8_9	Maternal
P73	Male	No	EX2_34	Maternal
P74	Male	No	EX53_55	<i>De novo</i>
P75	Male	Yes	EX56_63	Maternal
P76	Male	No	EX10_44	Maternal
P77	Male	No	EX56_70*	Maternal
P78	Male	No	EX10_11	<i>De novo</i>

EX: exon; *: novel mutation.

Table S4 Point mutations in *DMD* gene detected by NGS

Patient NO.	Grander	Family history	Exon/ Intron	Variants	Change of protein	Report	Mutation Type	Inheritance
P79	Male	No	EX26	c.3487C>T	p.Gln1163Ter	Novel	Nonsense	<i>De novo</i>
P80	Male	No	EX16	c.1886C>G	p.Ser629Ter	Known	Nonsense	Maternal
P81	Male	Yes	EX6	c.497G>T	p.Gly166Val	Known	Missense	Maternal
P82	Male	No	EX55	c.8086_8087 insC	p.Leu2696ProfsX14	Novel	Frameshift	Maternal
P83	Male	No	EX23	c.2962del T	p.Ser988LeufsX16	Known	Frameshift	Maternal
P84	Male	No	EX35	c.4857del A	p.Lys1619LysfsX10	Known	Frameshift	Maternal
P85	Male	No	EX67	c.9689delA	p.Asp3230AlafsX53	Novel	Frameshift	Maternal
P86	Male	Yes	EX17	c.2125C>T	p.Gln709Ter	Known	Nonsense	Maternal
P87	Male	Yes	EX72	c.10279C>T	p.Gln3427Ter	Known	Nonsense	Maternal
P88	Male	No	EX67	c.9669C>T	p.Arg3223Ter	Novel	Nonsense	Maternal
P89	Male	No	EX21	c.2664 C>T	p.Gln888Ter	Novel	Nonsense	<i>De novo</i>
P90	Male	No	EX14	c.1638G>A	p.Trp546Ter	Known	Nonsense	Maternal
P91	Male	No	EX13	c.1489C>T	p.Gln497Ter	Known	Nonsense	<i>De novo</i>
P92	Male	No	EX54	c.7899G>A	p.Trp2633Ter	Novel	Nonsense	Maternal
P93	Male	No	EX43	c.6283C>T	p.Arg2095Ter	Known	Nonsense	Maternal
P94	Male	No	EX30	c.4131delA	p.Lys1377AsnfsX5	Novel	Frameshift	Maternal
P95	Male	No	EX62	c.9204_9207delCAAAA	p.Asn3068LysfsX20	Known	Frameshift	Maternal
P96	Male	No	EX53	c.7822G>T	p.Glu2608Ter	Known	Nonsense	Maternal
P97	Male	Yes	EX35	c.4996C>T	p.Arg1666Ter	Known	Nonsense	Maternal
P98	Male	Yes	EX70	c.10141C>T	p.Arg3381Ter	Known	Nonsense	Maternal
P99	Male	No	IN10	c.1149+1G>A	Unknow	Novel	Splice	Maternal
P100	Male	No	IN10	c.1150-2A>G	Unknow	Known	Splice	Maternal

EX: exon; IN: intron.