Supp. Table S1. *NEB* variants identified in 143 nemaline myopathy families

Family ID	Exon/Intron; variant in cDNA; in protein sequence	Additional details
A: Families with the severe form of NM (n = 24)		
F25 cons	int18; c.1675-2A>G; p.?	HOZ
F367 cons	int32; c.3255+2dup; p.?	HOZ
F407 cons	ex173; c.24475_24479dup; p.Asn8160fs	HOZ , ALT3
F88 cons	ex180; c.25200del; p.Phe8400fs B-Fam13, D-Fam4	HOZ
F34 cons	del ex55; c.7431+1916_7536+372del, p.Arg2478_Asp2512del F-Pat7, Q-Pat2	HOZ, Ashk. 2.5 kb del
F105 cons	del ex55; c.7431+1916_7536+372del, p.Arg2478_Asp2512del C, F-Pat2	HOZ, Ashk. 2.5 kb del
F326	del ex55; c.7431+1916_7536+372del, p.Arg2478_Asp2512del F-Pat14 ex132; c.20213dup; p.Asp6738fs	Ashk. 2.5 kb del
F370	del ex55; c.7431+1916_7536+372del, p.Arg2478_Asp2512del L-Pat2 int108; c.17118+1G>C; p.? L-Pat2	Ashk. 2.5 kb del
F374	ex120; c.18808C>T; p.Arg6270* L-Pat3 del ex55; c.7431+1916_7536+372del, p.Arg2478_Asp2512del L-Pat3	
F181	del 5'end-ex24; Chr2.hg19:g.(152,545,083_152,545,531)_(152,599,010_152,608,650)del P-Fam3 ex129; c.19913G>C; p.Arg6638Pro D-Fam55	53-64 kb del
Bos227-1	ex57; c.7647A>G; p.Tyr2549* ex64; c.9046C>T; p.Arg3016*	ALT1
F47	int32; c.3255+1G>T; p.? D-Fam53 ex8; c.549C>A; p.Tyr183* D-Fam53	
F383	ex107; c.16921G>T; p.Glu5641* K int32; c.3255+1G>A; p.? K	
BOS-32	int32; c.3255+1G>A; p.? ex136; c.C20659T; p.Arg6887*	
F51	int4; c.78+1G>A; p.?	

Family ID	Exon/Intron; variant in cDNA; in protein sequence	Additional details
	int36; c.3987+1_+2inv; p.?	
F109	ex81; c.12048_12049del; p.Lys4016fs [H] int13; c.1152+1G>T; p.? [H]	
F110	ex28; c.2827C>T; p.Gln943* int145; c.21628-2A>T; p.?	
F121	int81; c.12330+5G>A; p.? [D-Fam5] ex139; c.20975_20976del; p.Lys6992fs [D-Fam5]	
F134	ex180; c.25205_25206del; p.Tyr8402* [B-Fam12] ex33; c.3473_3488del; p.Asn1187fs [D-Fam1]	
F146	int147; c.21840+5_+13del; p.? [B-Fam1, D-Fam2] ex148; c.21898C>T; p.Arg7300* [B-Fam1, D-Fam2]	
F207	int132; c.20262+5G>A; p.? int11; c.928-7_-5del; p.?	
F375	ex9; c.694del; p.Gln232fs ex58; c.7964A>G; p.Tyr2655Cys	
G17539	int58; c.8160+1G>A; p.Asn2653_His2720del [M-Pat2] ex46; c.5783_5784del; p.Tyr1928fs [M-Pat2]	
G21781	ex45; c.5574C>G; p.Tyr1858* [M-Pat1, Q-P1] int122; c.19101+5G>A; p.? [M-Pat1, Q-P1]	
B: Families with the intermediate form of NM (n = 15), * sib has typical form of NM		
F178 cons *	del ex55; c.7431+1916_7536+372del, p.Arg2478_Asp2512del [C, F-Fam3]	HOZ, Ashk. 2.5 kb del
G22593 cons	ex174;c.24541_24544dup; p.Val8182fs [Q-P9]	HOZ, ALT3
G36659 cons	ex177; c.24735_24736del; p.Ala8246* [Q-P7]	HOZ, ALT3
F263	int4; c.78+1G>A; p.? [F-Fam11] del ex55; c.7431+1916_7536+372del, p.Arg2478_Asp2512del [F-Fam11]	Ashk. 2.5 kb del
F148	int23; c.2212-1G>C; p.? [D-Fam10] del ex55; c.7431+1916_7536+372del, p.Arg2478_Asp2512del [F-Fam10]	Ashk. 2.5 kb del
P8	ex139; c.20928G>T; p.Gly6976 [Q-P8]	

Family ID	Exon/Intron; variant in cDNA; in protein sequence	Additional details
	ex172; c.24274del; p.Arg8125fs Q-P8	ALT3
SYD-F1*	ex173; c.24407_24410dup; p.Leu8137fs	ALT3
F176	ex163; c.23521C>T; p.Gln7841* D-Fam12 ex122; c.19097G>T; p.Ser6366Ile D-Fam12	FIN1
TRE-F1	ex122; c.19097G>T; p.Ser6366Ile ex42; c.5061G>A p.Trp1687*	FIN1
F1 *	ex151; c.22249A>C; p.Thr7417Pro B-Fam2 ex122; c.19048_19057del; p.Thr6350fs D-Fam13	FIN2 FIN3
F8	ex122; c.19048_19057del; p.Thr6350fs D-Fam30 int32; c.3255+1G>A; p.? D-Fam30	FIN3
F77 *	ex58; c.7964A>G; p.Tyr2655Cys ex78; c.11628G>A; p.Trp3876*	
F17	int36; c.3987+1G>A; p.? ex138; c.20845dup; p.Thr6949fs	
F332	int36; c.3987+1G>A; p.? ex7; c.412C>T; p.Arg138*	
F60	ex135; c.20554G>T; p.Glu6852* ex180; c.25241T>G; p.Leu8414*	
C: Families with the typical form of NM (n = 51) , * sib has intermediate form of NM		
F117 cons	int32; c.3255+1G>T; p.? D-Fam14	HOZ
F300 cons	int60; c.8373+5G>A; p.?	HOZ
F94 cons	ex154; c.22584G>C; p.Gln7528His A-Fam4, D-Fam21	HOZ
F119 cons	ex181; c.25279G>T; p.Glu8427* A-Fam5, D-Fam23	HOZ
F178 cons*	del ex55; c.7431+1916_7536+372del, p.Arg2478_Asp2512del C, F-Fam3	HOZ, Ashk. 2.5 kb del
SYD-F1*	ex173; c.24407_24410dup; p.Leu8137fs	HOZ ALT3
F328 cons	ex173; c.2444dup; p.Pro8147fs	HOZ, ALT3
AG-F1	ex172; c.24355_24358dup; p.Thr8120fs O	ALT3

Family ID	Exon/Intron; variant in cDNA; in protein sequence	Additional details
	ex175; c.24632_24633del; p.Pro8211fs [O]	ALT3
F4	int65; c.9414+1G>T, p.? ex61; c.8381A>T; p.Tyr2794Phe	ALT1
F318	ex173; c.24480_24483del; p.Gln8161fs del ex73-int73; c.10798_10872+839del; p.?	ALT3 1 kb del
F340	ex49; c.6385C>T; p.Gln2129* ex172; c.24370_ 24373del; p.Val8126fs	 ALT3
SYD-F2	ex172; c.24372_24375del; p.Arg8125fs ex142; c.21210T>A, p.Tyr7070*	ALT3
SYD-F3	ex172; c.24314_24317dup, p.Tyr8107fs ex14; c.1172_1173del, p.Tyr391*	ALT3
F349	ex175; c.24684G>A; p.? ex119; c.18676C>T; p.Gln6226*	ALT3
G19106	ex175; c.24684G>A; p.? [Q-P11] ex119; c.18676C>T; p.Gln6226* [Q-P11]	ALT3
F238	int17; c.1569+1G>A; p.? [Q-P14] ex176; c.24711del; p.Ala8238fs [Q-P14]	 ALT3
F305	int32; c.3255+1G>T; p.? int83/91/99; c.12639+3_+6delAAGT; p.?	 TRI
F160	del ex55; c.7431+1916_7536+372del, p.Arg2478_Asp2512del [F-Fam12] int79; c.11910+5G>C; p.? [D-Fam17]	Ashk. 2.5 kb del
F229	del ex55; c.7431+1916_7536+372del, p.Arg2478_Asp2512del [F-Fam13] int151; c.22266+1_+9del; p.? [D-Fam52]	Ashk. 2.5 kb del
F211	ex155; c.22489C>T; p.Arg7497* del ex43-45; c.5238+335_5764-407del; p.Arg1747_Asp1921del	 2.2 kb del
F45	int36; c.3987+1_+2inv; p.? [D-Fam24] del ex53-54; c.6916-163_7431+211del; p.Tyr2306_Asp2477del [L-Fam4]	 1 kb del
F9	ex163; c.23500_23503dup; p.Leu7835fs [A-Fam2]	

Family ID	Exon/Intron; variant in cDNA; in protein sequence	Additional details
	ex122; c.19097G>T; p.Ser6366Ile D-Fam31	FIN1
F11	ex122; c.19097G>T; p.Ser6366Ile D-Fam36 ex75; c.11164C>T; p.Arg3722*	FIN1
F3	ex136; c.20665C>T; p.Gln6889* ex122; c.19097G>T; p.Ser6366Ile D-Fam35	 FIN1
F298	ex122; c.19097G>T; p.Ser6366Ile D-Fam37 ex50; c.6678del; p.Lys2226fs	FIN1
F1*	ex151;c.22249A>C; p.Thr7417Pro B-Fam2 ex122; c.19048_19057del; p.Thr6350fs D-Fam13	FIN2 FIN3
F12	ex61; c.8392_8395dup; p.Arg2799fs D-Fam32 ex151;c.22249A>C; p.Thr7417Pro B-Fam3	 FIN2
F5	ex75; c.11083T>C; p.Tyr3695His D-Fam15 ex158; c.22933G>T; p.Glu7645* D-Fam15	
F6	int35; c.3879+1G>A; p.? ex156; c.22746del; p.Met7582fs A-Fam1, D-Fam28	
F10	ex182; c.25441C>T; p.Arg8481* ex61; c.8458_8459del; p.Ser2820fs D-Fam39	
F14	ex161; c.23245C>T; p.Arg7749* D-Fam50 ex7; c.410A>G; p.Tyr137Cys D-Fam50	
F56	ex38; c.4337G>T; p.Gly1446Val ex158; c.22936C>T; p.Arg7646*	
F77 *	ex58; c.7964A>G; p.Tyr2655Cys ex78; c.11628G>A; p.Trp3876*	
F80	int22; c.2106+3A>C; p.? I int3; c.36+2dup; p.? I	
F83	ex72; c.10528G>C; p.Ala3510Pro D-Fam25 ex18; c.1623del; p.Asp542fs	
F106	int5; c.294+2T>C; p.? D-Fam19	

Family ID	Exon/Intron; variant in cDNA; in protein sequence	Additional details
	ex25; c.2320C>G; p.Tyr773* D-Fam19	
F107	int11; c.928-1G>A; p.? D-Fam20 ex18; c.1657_1665delins8; p.Ala553fs D-Fam20	
F112	int35; c.3879+5G>T; p.? ex70; c.10261C>T; p.Gln3421*	
F118	ex152; c.22275C>G; p.Tyr7425* B-Fam4 int43; c.5343+5G>A; p.? D-Fam16	
F129	ex13; c.1054_1055del; p.Tyr352* ex38; c.4337G>T; p.Gly1446Val	
F136	int28; c.2835+5G>C; p.? D-Fam54 ex164; c.23600_23601del; p.Thr7867fs B-Fam5	
F200	ex56; c.7641T>A; p.Ser2547Arg int152; c.22378-1G>A; p.?	
F274	int56; c.7645-2A>G; p.? ex18; c.1623del; p.Pro541fs	
F279	int43; c.5343+5G>A; p.? Q-P10 ex153; c.22378del; p.Val7460fs Q-P10	
F319	ex140; c.21076C>T; p.Arg7026* ex129; c.19944G>A; p.Ser6648	
G33120	ex6; c.300dup; p.Tyr101fs Q-P6 int49; c.6496-1G>A; p.? Q-P6	
PG-F1	ex58; c.8031_8041del; p.Lys2677fs ex182; c.25424del; p.Thr8475fs	
SYD-F4	int29; c.2943+1G>A; p.? int80; c.12018+1G>A; p.?	
SYD-F5	int45; c.5763+5G>A; p.? int25; c.2415+1G>A; p.?	
SYD-F6	ex81; c.12160delT, p.Trp4054fs	

Family ID	Exon/Intron; variant in cDNA; in protein sequence	Additional details
	int35; c.3880-2A>G; p.?	
Bos153-1	int13; c.1152+1G>A; p.? int29; c.2943+1G>A; p.?	
EST-F1	*	
D: Families with the mild form of NM (n = 7)		
F333	ex130; c.19992_19999dup; p.Asp6667fs del ex43-45; c.5238+335_5764-407del; p.Arg1747_Asp1921del	2.2 kb del
F155	ex54; c.7291G>A; p.Glu2431Lys ex55; c.7523_7526del; p.Ile2508fs D-Fam41	
F202	int158; c.23011-2A>G; p.? D-Fam44 ex3; c.8A>G; p.Asp3Gly	
F231	ex69; c.10043_10046del; p.Val3348fs Q-P13 ex49; c.6388G>C; p.Ala2130Pro Q-P13	
F237	int155; c.22696-3C>G; p.? Q-P12 ex148; c.21901_21915delinsT; p.Pro7301fs Q-P12	
F344	ex61; c.8381A>T; p.Tyr2794Phe int164; c.23662-1G>A; p.?	
PG-F2	int18; c.1674+1G>T; p.? ex159; c.22909G>T; p.Glu7672*	
E: NM families with unusual features, i.e. "other forms" of NM (n = 18).		
Mutations in alternatively spliced exons 63-66 (ALT1), which are always expressed as a block.		
F270 (typical)	ex63; c.8897A>G; p.Tyr2966Cys ex73; c.10701del; p.Lys3567fs	Axial involvement predominant, limb muscles relatively spared in all three families
F284 (typical)	ex151; c.22249A>C; p.Thr7417Pro D-Fam37 ex65; c.9357C>A; p.Cys3119*	
F386 (severe)	int32; c.3255+1G>T; p.? ex66; c.9465del; p.Pro3155fs	
Mutation in the triplicate region of eight exons (TRI: three homologous sets: ex82-89, ex90-97, ex98-105)		

Family ID	Exon/Intron; variant in cDNA; in protein sequence	Additional details
F36 (severe)	ex86/94/102; c.13066del; p.Tyr4356fs [Q-P3] ex110; c.17535G>A; p.Glu5845 [Q-P3]	Cleft palate, oesophageal atresia, clubfeet
F139 (typical)	int29; c.2943+1G>A; p.? int89/97/105; c.13788+1G>A; p.?	Non-selective muscle weakness
Mutations in ex143 – 144 (ALT2). Expressed in a mutually exclusive way - never in the same isoform.		
F187 (typical)	ex58; c.8031_8041del; p.Lys2677fs [D-Fam51] ex143; c.21406C>T; p.Gln7136*	Ophthalmoplegia, FSH-like pattern of muscle weakness
F390 (typical)	ex144: c.21506C>A; p.Ser7169*	Unusual pattern of selective weakness, dystrophic biopsy. Difficulties in waking from anaesthesia, HOZ
Mutations in ex167-173 (ALT3). Expressed independently of each other		
F189 (mild)	ex170; c.24218C>A; p.Ser8073* [D-Fam42] ex7; c.471delT; p.Ile157fs [D-Fam42]	Selective axial weakness
F79 (mild)	ex171; c.24285_24288dup; p.His8097fs [B-Fam9, D-Fam43]	Rigid neck in F79. Unusual selective weakness pattern in both families, both HOZ mutations
F81 (typical)	ex171;c.24282_24283del; p.Arg8094fs [B-Fam10, D-Fam22]	
F127 (severe)	ex171; c.24294_24297dup; p.Glu8098fs [B-Fam7] ex119; c.18676C>T; p.Gln6226* [D-Fam49]	Fasciculations
F206 (intermediate)	ex172; c.24372_24375dup; p.Val8126fs [D-Fam9] ex48; c.6078del; p.Lys2026Asnfs*62	Tremor/ myoclonus. Fasciculations
F389 (typical)	ex173;c.24407_24410dup; p.Leu8137fs	Unusual pattern of selective weakness
F382 (typical)	ex173;c.24444_24447del; p.Leu8148fs ex167; c.23910_23911del; p.Arg7970fs	Fasciculations, type 2 predominance
F324 (severe)	int32; c.3255+1G>A; p.? ex175; c.24632_24633del; p.Pro8211fs	Unusual pattern of selective weakness
F31 (severe)	ex163; c.23526_23527del; p.Arg7842fs [A-Fam3, D-Fam3, Q-P4&P5-N] ex177; c.24792_24793del; p.Glu8265fs [A-Fam3, D-Fam3, Q-P4&P5-N]	Arthrogryposis, micropenis, hypospadias
Mutations in non-alternatively spliced exons		
F316 (typical)	ex68; c.9727_9728del; p.Leu3243fs	Fever subsequent to anesthesia on three

Family ID	Exon/Intron; variant in cDNA; in protein sequence	Additional details
	ex122; c.19097G>T; p.Ser6366Ile	occasions, unusually severe feeding problems
F309 (mild)	ex78; c.11610C>A; p.Tyr3870* dup 5'-ex51; Chr2.hg19:g.(152,511,101_152,511,615)_(152,614,714_?)dup	Normal muscle strength of upper limbs 80 kb dup in *NEB* (>103 kb altogether)
F: NM families with unspecified form of NM (n = 32)		
PG-F3	int4; c.78+1G>A, p.?	HOZ
PG-F4	del ex55; c.7431+1916_7536+372del, p.Arg2478_Asp2512del	HOZ, Ashk. 2.5 kb del
F114	int112; c.17737-2A>T; p.? ex144; c.21423del; p.Lys7141fs	 ALT2
PG-F5	int66; c.9619-2A>G; p.? ex173; c.24473_24476dup; p.His8259fs	ALT1 ALT3
F385	int83/91/99; c.12639+1G>A; p? ex173; c.24493_.24494del; p.Ser8165fs	TRI ALT3
PG-F6	int85/93/101; c.13059+5G>A; p.? ex170; c24218C>A, p.Ser8073*	TRI ALT3
PG-F7	ex158; c.22936C>T; p.Arg7646* ex170; c.24131_24134dup; p.Tyr8045fs	 ALT3
PG-F8	int152; c.22377+2dup; p.? ex172; c.24324_24328dup; p.Leu8106fs	 ALT3
PG-F9	ex175; c.24632_24633del; p.Pro8211fs ex172; c.24313_24212dup; p.Leu8102fs	ALT3 ALT3
PG-F10	int32; c.3255+1G>T, p.? ex173; c.24407_24410dup; p.Leu8137fs	 ALT3
F111	ex174; c.24559C>T; p.Arg8187* ex161; c.23340del; p.Ala7780fs	ALT3
PG-F11	ex115; c.18211del; p.Leu6070fs ex174; c.24500_24503dup; p.Val8167fs	 ALT3
PG-F12	ex61; c.8425C>T; p.Arg2809*	

Family ID	Exon/Intron; variant in cDNA; in protein sequence	Additional details
	ex172; c.24317T>A; p.Leu8106*	ALT3
PG-F13	int80; c.12018+1G>A; p.? ex173; c.24458_24461dup; p.Met8254fs	ALT3
PG-F14	int47; c.6075+2T>G; p.? ex175; c.24654_24655del; p.Arg8218fs	ALT3
PG-F15	int32; c.3255+1G>A; p.? ex175; c.24654_24655del; p.Arg8218fs	ALT3
PG-F16	int32; c.3255+1G>C; p.? ex177; c.24735_24736del; p.Ala8246*	ALT3
PG-F17	int32; c.3255+1G>A; p.? Int82/90/98; c.12534+1G>C; p.?	TRIPL
PG-F18	int18; c.1674+2T>C; p.? ex85/93/101; c.12996G>A; p.Trp4332*	TRI
PG-F19	int32; c.3252_3255+3del; p.Ser1084Argfs*10 del ex77; 11289+541_11601+1451del	2.5 kb del
PG-F20	ex163; c.23378del; p.Met7828Serfs*25 del ex77; 11289+541_11601+1451del	2.5 kb del
F267	ex138; c.20845dup; p.Thr6949Asnfs*16 int36; c.3987+1G>A; p.?	
F372	int8; c.612+1G>A; p.? ex75; c.11164C>T; p.Arg3722*	
F249	ex110; c.17457G>A; p.Trp5819* int13; c.1152+5G>A; p.?	
CMH000102 & 103	ex132; c.20253del; p.Ala6751Alafs*8 ex35; c.3874A>G; p.Ser1292Gly	
PG-F21	int13; c.1152+1G>A; p.? Int107; c.17013+1G>T; p.?	
PG-F22	int32; c.3255+2_+3insTT; p.?	

Family ID	Exon/Intron; variant in cDNA; in protein sequence	Additional details
	ex33; c.3390T>G; p.Tyr1130*	
PG-F23	int32; c.3255+1G>A; p.? ex138: c.20803C>T; p.Gln6935*	
PG-F24	int32; c.3255+1G>T; p.? ex58; c.8031_8041del; p.Lys2677Asnfs*7	
PG-F25	int32; c.3255+1G>A; p.? ex118; c.18579+1G>A; p.?	
PG-F26	int43; c.5343+5G>A; p.? ex112; c.17654G>A; p.Trp5885*	
EST-F1	ex53; c.6937C>T; p.Arg2313* ex180; c.25176G>A; p.Trp8392	

A-F: Families with the **A: severe** (n = 24), **B: intermediate** (n = 15), **C: typical** (n = 51), **D: mild** (n = 7), **E: other** (n = 18) and **F: unknown** (n = 32) forms of NM (n = 143; sibs in four families have typical and intermediate forms of NM). If only one variant is indicated, the mutation is homozygous. Mutations/family IDs in previous publications are marked as superscripts following the mutation. Note that three families include patients with both intermediate and typical forms of NM. Reference sequence: c.NM_001271208.1 (contains all of the 183 exons including the triplicated region).

In Supp. Table S1 and Table 2: Cons = consanguineous; HOZ = homozygous variant, del ex55 = Ashk. = Ashkenazi Jewish Founder mutation; Finnish founder mutations: FIN1 = a missense mutation Ser > Ile in exon 122, FIN2 = a missense mutation Thr > Pro in exon 151, FIN3 = a frameshift mutation in exon 122; ALT1 = mutation in the alternatively spliced exons 63-66 (always expressed together), ALT2 = mutation in the alternatively spliced exons 143 or 144, ALT3 = mutation in the alternatively spliced exons 167-177 (expressed independently), TRI = mutation in the triplicate region.

Published mutations in Supp. Table S1 and Table 2: A = Pelin et al., 1999, B = Pelin et al., 2002, C = Anderson et al., 2004, D = Lehtokari et al., 2006, E = Wallgren-Pettersson et. al., 2007, F = Lehtokari and Greenleaf et al., 2009, G = Romero and Lehtokari et al., 2009, H = Lawlor et al., 2011, I = Ochala et al., 2011, J = Lehtokari et al., 2011, K = Kapoor et al., 2013, L = Yonath et al., 2012 , M = Böhm et al., 2013, N = Scoto et al., 2013, O = Gajda et al., 2013, P = Kiiski et al., 2013, Q = Malfatti et al., 2014, R = Malfatti et al, in preparation.