

Table S1 Clinical features of 84 *LMNA*-related muscular dystrophy

Patient	Phenotype	Sex	Age of onset	Age at last review/Maximum motor milestone	Contractures	Spinal deformities	Highest CK level (U/L)	ECG	UCG	EMG	Muscle biopsy
P1	EDMD	F	1y	Deceased 20y/walking	Elbow, knee	Scoliosis	451	Atrial rest, atrioventricular block	Enlargement of the right heart and decreased systolic and diastolic function of the whole heart	Myogenic	Dystrophic (9y)
P2	L-CMD	F	Birth	16y/walking	Elbow, knee, ankle	Lordosis, rigid spine	623	Sinus arrhythmia	n/a	n/a	n/a
P3	L-CMD	M	Birth	Deceased 7y/sitting	Elbow, knee, ankle, hip		1248	n/a	n/a	Neurogenic	n/a
P4	L-CMD	F	Birth	10y7m/sitting	Ankle, knee, hip	Lordosis	882	Normal	Normal	Myogenic	Dystrophic, Inflammatory change (1.5y)
P5	EDMD	F	1y3m	10y9m/running	Elbow	Rigid spine	427	Normal	Normal	Myogenic	Dystrophic (3y)
P5's Father	EDMD	M	43y	43y/running	Elbow, ankle	Rigid spine,	293	sinus bradycardi	n/a	n/a	n/a

P6	EDMD	F	2y	16y/walking	Elbow, ankle	scoliosis Scoliosis, rigid spine	244.9	Normal	Normal	Normal	n/a
P7	L-CMD	M	Birth	9y1m/sitting	Ankle, hip		511	Normal	Normal	Myogenic	Dystrophic (1y7m)
P8	L-CMD	M	Birth	8y4m/walking		Scoliosis	1653	Normal	Normal	Myogenic	Dystrophic, Inflammator y change (10m)
P9	L-CMD	M	13m	12y2m/walking	Ankle, knee, hip		749	Sinus arrhythmia	Normal	Normal	Dystrophic (5y)
P10	L-CMD	F	Birth	7y5m/sitting			3766	Sinus arrhythmia	left ventricular false tendon	Normal	Dystrophic, Inflammator y change (7m)
P11	L-CMD	F	Birth	11y11m/walking	Ankle, knee, hip	Lordosis	371	Normal	Normal	Normal	Dystrophic (4y)
P12	EDMD	F	4y	11y11m/running	Elbow	Rigid spine	554	Sinus rhythm, frequent atrial extrasystol e	Normal	Myogenic	Myopathic (5y)
P13	L-CMD	M	1y3m	9y8m/walking	Ankle	Lordosis	1046	Normal	Normal	Myogenic	Dystrophic (2y11m)

P14	L-CMD	F	Birth	8y4m/sitting	Knee, hip, ankle	Lordosis	1062	Sinus tachycardia	Normal	Normal	Dystrophic (1y)
P15	L-CMD	F	Birth	7y6m/sitting	Ankle		1817	Sinus tachycardia	Normal	n/a	Dystrophic, Inflammatory change (1y2m)
P16	EDMD	M	1y2m	11y10m/walking	Elbow, ankle		568	Normal	Normal	Normal	Dystrophic, Inflammatory change (6y)
P17	EDMD	F	1y	13y4m/walking	Elbow, ankle	Lordosis	992	Normal	Normal	Myogenic	n/a
P18	L-CMD	F	Birth	7y9m/sitting	Ankle		195	n/a	n/a	Myogenic	Dystrophic (1y8m)
P19	L-CMD	M	Birth	6y9m/walking	Ankle	Lordosis	484	Normal	Normal	n/a	Dystrophic (1y3m)
P20	L-CMD	M	Birth	6y3m/sitting	Ankle	Lordosis	1568.6	n/a	n/a	n/a	n/a
P21	EDMD	M	1y6m	11y/running			1089	Sinus tachycardia	Normal	Myogenic	n/a
P22	L-CMD	F	6m	7y4m/sitting	Ankle	Lordosis	770	Normal	Normal	n/a	n/a
P23	L-CMD	M	Birth	5y11m/sitting		Lordosis	871	Normal	Normal	Neurogenic	Myopathic, Inflammatory change (1y4m)

P24	EDMD	M	2y	18y/running	Elbow, ankle	Rigid spine	691	Sinus arrhythmia	n/a	Myogenic	n/a
P25	EDMD	F	2y	8y7m/running	Ankle		1005	Sinus arrhythmia	Normal	Myogenic	Dystrophic (4y)
P26	EDMD	F	1y6m	9y8m/walking	Elbow, ankle	Lordosis	927	Sinus arrhythmia	Normal	Myogenic	n/a
P27	L-CMD	M	Birth	5y3m/walking			2648	Sinus tachycardia	Normal	Normal	n/a
P28	L-CMD	M	Birth	10y7m/walking			493	Normal	Normal	Myogenic	n/a
P29	L-CMD	M	Birth	4y10m/sitting		Lordosis	1738	Normal	Normal	n/a	n/a
P30	L-CMD	M	Birth	6y2m/walking	Ankle	Lordosis	1428	Normal	Normal	Myogenic	n/a
P31	L-CMD	M	Birth	9y6m/walking	Elbow, ankle, hip	Lordosis	727	Sinus arrhythmia	Normal	Normal	n/a
P32	EDMD	F	2y	5y8m/walking	Ankle		950	Normal	Normal	Myogenic	n/a
P33	EDMD	M	1y2m	8y5m/running	Ankle	Lordosis	774	Normal	Normal	Myogenic	n/a
P34	EDMD	M	2y8m	7y3m/running	Ankle		1200	Normal	Normal	n/a	Dystrophic (2y)
P35	L-CMD	F	Birth	5y5m/walking		Lordosis	723	Normal	Normal	n/a	n/a
P36	L-CMD	M	Birth	3y4m/sitting		Lordosis	793	Sinus tachycardia	Normal	n/a	Myopathic (7m)

P37	L-CMD	M	Birth	7y4m/walking		Lordosis	2241	Sinus tachycardia	Normal	Myogenic	Dystrophic, Inflammatory change (1y9m)
P38	L-CMD	M	Birth	3y6m/sitting	Ankle		1530.8	Normal	Normal	Myogenic	n/a
P39	L-CMD	M	Birth	3y7m/sitting			929	n/a	n/a	Neurogenic	n/a
P40	L-CMD	F	Birth	6y3m/walking			370	Normal	Normal	Myogenic	n/a
P41	L-CMD	F	Birth	2y5m/sitting			n/a	Normal	Normal	Myogenic	n/a
P42	EDMD	M	1y	7y3m/running		Lordosis	528	Sinus arrhythmia	Normal	Myogenic	n/a
P43	EDMD	M	1y6m	7y5m/walking	Elbow, ankle		462	Normal	Normal	Normal	Dystrophic, Inflammatory change (3.5y)
P44	EDMD	M	2y	3y5m/running			1356	n/a	n/a	Normal	n/a
P45	EDMD	M	1y3m	4y5m/running			967	Sinus tachycardia	Normal	Myogenic	n/a
P46	L-CMD	F	Birth	2y9m/walking		Lordosis	1674	Normal	Normal	Myogenic	n/a
P47	EDMD	M	1y3m	3y1m/walking		Lordosis	2595	Sinus arrhythmia, premature beat	Patent foramen ovale	Myogenic	Dystrophic (2.5y)

P48	LGMD1B	M	1y3m	6y8m/running			278	Normal	Normal	n/a	n/a
P48's Mother	LGMD1B	F	1y3m	35y/running			162	Normal	Normal	n/a	n/a
P49	EDMD	M	1y6m	2y10m/running	Ankle		3278	Normal	Normal	n/a	n/a
P50	EDMD	M	2y	4y4m/walking			1027	Sinus tachycardia	left ventricular false tendon	Myogenic	n/a
P51	L-CMD	F	4m	3y4m/sitting aided	Ankle	Lordosis	1455	Normal	Normal	Normal	n/a
P52	EDMD	F	10y	24y/walking	Ankle		223	Normal	Normal	Myogenic	Dystrophic (24y)
P53	EDMD	M	1y	19y/walking	Elbow, ankle		929	Normal	Normal	Myogenic	Dystrophic (19y)
P54	EDMD	F	3y	29y/walking	Elbow, ankle	Rigid spine	n/a	Atrioventricular block	Normal	Myogenic	Dystrophic (29y)
P55	EDMD	M	2y	4y/walking	Ankle		1361	Normal	Normal	Myogenic	Dystrophic (4y)
P56	EDMD	F	4y	14y/walking	Ankle	Scoliosis	820	Sinus tachycardia	Normal	Myogenic	Dystrophic (14y)
P57	LGMD1B	F	1y3m	3y/walking			2623	Normal	Normal	Myogenic	Dystrophic (3y)
P58	LGMD1B	M	1y	2y6m/walking			413	Normal	Normal	Myogenic	Dystrophic (2.5y)
P59	LGMD1B	F	11y	16y/running			710	Sinus	Normal	Myogenic	Dystrophic

								tachycardi		(16y)
								a		
P60	LGMD1B	F	1y	3y/walking			350	Normal	Normal	Myogenic Dystrophic (3y)
P61	L-CMD	M	11m	2y6m/sitting	Ankle		1046	Normal	Normal	Myogenic Dystrophic (2.5y)
P62	L-CMD	M	2m	1y6m/sitting aided	Ankle, wrist		445	Normal	Normal	Myogenic Dystrophic, Inflammatory change (1.5y)
P63	L-CMD	F	1y	4y6m/sitting aided			736	Normal	Normal	Myogenic Dystrophic (4.5y)
P64	L-CMD	F	8m	8y/sitting aided	Knee, ankle		372	Normal	Normal	Myogenic Dystrophic, Inflammatory change (8y)
P65	L-CMD	F	6m	3y/sitting aided			898	Normal	Normal	Myogenic Dystrophic, Inflammatory change (3y)
P66	LGMD1B	M	1y2m	3y/walking			904	Normal	Normal	Myogenic Dystrophic (3y)
P66's Father	LGMD1B	M	5y	38y/walking	Ankle		800	Normal	Normal	Myogenic Dystrophic (38y)
P67	L-CMD	M	1y	2y3m/walking	Ankle	Lordosis	435	Normal	Normal	n/a

P68	L-CMD	F	1y	1y6m/walking		Lordosis	1506	Normal	Normal	n/a	n/a
P69	L-CMD	F	1y	3y3m/walking		Lordosis	2208	Normal	Normal	Myogenic	n/a
P70	L-CMD	M	2y	3y/walking		Lordosis	733	Normal	Normal	Myogenic	n/a
P71	LGMD1B	F	2y	4y5m/walking			2116	Normal	Normal	n/a	n/a
P72	EDMD	F	2y	12y/walking	Elbow, ankle	Scoliosis	500	Normal	Normal	Myogenic	n/a
P73	LGMD1B	F	2y	4y1m/walking			1789	Normal	Normal	n/a	n/a
P74	LGMD1B	F	2y	3y11m/walking	Ankle		799	Normal	Normal	n/a	n/a
P75	EDMD	F	3y	14y3m/walking	Elbow, ankle	Rigid spine	615	Atrioventri- cular block	Normal	n/a	n/a
P76	EDMD	F	3y	6y3m/walking	Ankle		1705	Normal	Normal	Myogenic	n/a
P77	EDMD	F	1y6m	6y5m/walking	Ankle		349	Normal	Normal	Myogenic	Dystrophic (6.4y)
P78	L-CMD	F	3m	6y11m/sitting aided	Wrist, hip, knee	Rigid spine	1156	Normal	Normal	n/a	Dystrophic (6.9y)
P79	EDMD	M	2y	8y2m/walking	Elbow, ankle		1259	Normal	Normal	Normal	Myopathic (8.2)
P80	EDMD	F	3y	10y3m/walking	Elbow, ankle		1186	Normal	Normal	Myogenic	Myopathic (10.3)
P81	L-CMD	F	3m	3y6m/sitting aided			1779	Normal	Normal	Normal	n/a

Abbreviations: L-CMD, *LMNA*-related congenital muscular dystrophy; EDMD, Emery-Dreifuss muscular dystrophy; LGMD1B, Limb-girdle muscular dystrophy type 1B; F, female; M, male; y, year; m, month; CK, creatine kinase; ECG, electrocardiogram; UCG, ultrasound cardiography; EMG, electromyography; n/a, not available. The upper limit of the normal CK level in our research team is 195 U/L.

Table S2 Univariate and multivariate Cox regression analysis of ambulance loss and cardiac involvement

	Variables	Univariate analysis			Multivariate analysis		
		HR	95% CI	<i>p</i> value	HR	95% CI	<i>p</i> value
Ambulance loss	Gender	0.308	0.079-1.198	0.089	0.381	0.065-2.225	0.284
	Age onset	2.244	0.355-14.162	0.039	2.571	0.346-19.096	0.356
	Mutation*	0.691	0.161-2.962	0.619	0.287	0.054-1.519	0.142
	Orthopedic complications	0.610	0.119-3.121	0.553	0.444	0.066-2.970	0.402
Cardiac involvement	Gender	1.398	0.519-3.769	0.508	2.315	0.758-7.071	0.141
	Age onset	0.634	0.191-2.103	0.457	0.432	0.109-1.702	0.432
	Mutation*	0.961	0.267-3.453	0.951	0.958	0.289-3.173	0.944
	Orthopedic complications	0.244	0.068-0.879	0.031	0.188	0.016-2.231	0.186

Abbreviations: CI, confidence interval; HR, hazard ratio. *Mutations were divided into missense group and non-missense group. Bold values are statistically significant ($p < 0.05$).

Table S3 Genetic analysis of *LMNA*-related muscular dystrophy

Patient	Phenotype	<i>LMNA</i> exon/intron	Protein domain affected	Nucleotide change	Predicted amino acid change	Inheritance	Novel/reported
P1	EDMD	Exon1	Head	c.94_96delAAG	p.K32del	<i>De novo</i>	Reported
P2	L-CMD	Exon2	Coil 1B	c.422T>C	p.L141P	<i>De novo</i>	Reported
P3	L-CMD	Exon1	Head	c.91G>A	p.E31K	<i>De novo</i>	Reported
P4	L-CMD	Exon4	Coil 2A	c.745C>T	p.R249W	<i>De novo</i>	Reported
P5	EDMD	Exon9	Ig-like domain	c.1580G>C	p.R527P	Paternal	Reported
P5's Father	EDMD	Exon9	Ig-like domain	c.1580G>C	p.R527P	Paternal	Reported
P6	EDMD	Exon1	Coil 1A	c.116A>G	p.N39S	<i>De novo</i>	Reported
P7	L-CMD	Exon6	Linker of coil 2B and NLS	c.1151A>G	p.E384G	<i>De novo</i>	Reported
P8	L-CMD	Intron8	Ig-like domain	c.1489-14_1489-7del	p.I497_E536del	<i>De novo</i>	Reported
P9	L-CMD	Exon1	Head	c.91_93delGAG	p.E31del	<i>De novo</i>	Reported
P10	L-CMD	Exon6	Coil 2B	c.1117A>G	p.I373V	<i>De novo</i>	Reported
P11	L-CMD	Exon1	Head	c.94_96delAAG	p.K32del	<i>De novo</i>	Reported
P12	EDMD	Exon4	Coil 2A	c.746G>A	p.R249Q	<i>De novo</i>	Reported
P13	L-CMD	Exon9	Ig-like domain	c.1558T>C	p.W520R	<i>De novo</i>	Reported
P14	L-CMD	Exon1	Coil 1A	c.117T>G	p.N39K	<i>De novo</i>	Reported
P15	L-CMD	Exon1	Coil 1A	c.143G>C	p.R48P	<i>De novo</i>	Reported
P16	EDMD	Exon6	Coil 2B	c.1124C>G	p.A375G	<i>De novo</i>	Novel
P17	EDMD	Exon5	Coil 2B	c.832G>C	p.A278P	<i>De novo</i>	Reported
P18	L-CMD	Exon6	Coil 2B	c.1118T>A	p.I373N	<i>De novo</i>	Reported
P19	L-CMD	Exon6	Coil 2B	c.1147G>A	p.E383K	<i>De novo</i>	Reported
P20	L-CMD	Exon4	Coil 2A	c.745C>T	p.R249W	<i>De novo</i>	Reported
P21	EDMD	Exon9	Ig-like domain	c.1540T>A	p.W514R	<i>De novo</i>	Reported

P22	L-CMD	Exon4	Coil 2A	c.745C>T	p.R249W	<i>De novo</i>	Reported
P23	L-CMD	Exon4	Coil 2A	c.745C>T	p.R249W	<i>De novo</i>	Reported
P24	EDMD	Exon4	Linker of coil 2A and 2B	c.778_780delAAG	p.K260del	<i>De novo</i>	Reported
P25	EDMD	Exon7	Ig-like domain	c.1357C>T	p.R453W	<i>De novo</i>	Reported
P26	EDMD	Exon1	Coil 1A	c.116A>G	p.N39S	<i>De novo</i>	Reported
P27	L-CMD	Exon7	Ig-like domain	c.1372T>C	p.S458P	<i>De novo</i>	Novel
P28	L-CMD	Exon7	Linker of coil 2B and NLS	c.1163G>C	p.R388P	<i>De novo</i>	Novel
P29	L-CMD	Exon1	Head	c.95_97delAGG	p.E33del	<i>De novo</i>	Novel
P30	L-CMD	Exon6	Coil 2B	c.1072G>A	p.E358K	<i>De novo</i>	Reported
P31	L-CMD	Exon6	Coil 2B	c.1072G>A	p.E358K	<i>De novo</i>	Reported
P32	EDMD	Exon6	Coil 2B	c.1126T>C	p.Y376H	<i>De novo</i>	Novel
P33	EDMD	Exon4	Linker of coil 2A and 2B	c.788T>C	p.L263P	<i>De novo</i>	Reported
P34	EDMD	Exon5	Coil 2B	c.907T>C	p.S303P	<i>De novo</i>	Reported
P35	L-CMD	Exon1	Coil 1A	c.116A>G	p.N39S	<i>De novo</i>	Reported
P36	L-CMD	Exon6	Linker of coil 2B and NLS	c.1150G>A	p.E384K	<i>De novo</i>	Novel
P37	L-CMD	Exon7	Ig-like domain	c.1325T>G	p.V442G	<i>De novo</i>	Novel
P38	L-CMD	Exon4	Coil 2A	c.745C>T	p.R249W	<i>De novo</i>	Reported
P39	L-CMD	Exon4	Coil 2A	c.745C>T	p.R249W	<i>De novo</i>	Reported
P40	L-CMD	Exon4	Coil 2A	c.745C>T	p.R249W	<i>De novo</i>	Reported
P41	L-CMD	Exon1	Head	c.92_94delAGA	p.K32del	<i>De novo</i>	Reported
P42	EDMD	Exon1	Coil 1B	c.317T>C	p.L106P	<i>De novo</i>	Novel
P43	EDMD	Exon4	Coil 2A	c.746G>A	p.R249Q	<i>De novo</i>	Reported
P44	EDMD	Exon5	Coil 2B	c.905T>C	p.L302P	<i>De novo</i>	Reported
P45	EDMD	Intron4	Coil 2B	c.810+2delT	p.(L271_Q312del)	<i>De novo</i>	Novel
P46	L-CMD	Exon1	Coil 1A	c.122G>C	p.R41P	<i>De novo</i>	Novel

P47	EDMD	Exon1	Head	c.94A>G	p.K32E	<i>De novo</i>	Reported
P48	LGMD1B	Exon6	Coil 2B	c.1004delG	p.(R336Gfs*144)	Maternal	Novel
P48's	LGMD1B	Exon6	Coil 2B	c.1004delG	p.(R336Gfs*144)	Maternal	Novel
Mother							
P49	EDMD	Exon6	Coil 2B	c.1110C>A	p.D370E	<i>De novo</i>	Novel
P50	EDMD	Exon6	Coil 2B	c.1072G>A	p.E358K	<i>De novo</i>	Reported
P51	L-CMD	Exon4	Coil 2A	c.745C>T	p.R249W	<i>De novo</i>	Reported
P52	EDMD	Exon6	Coil 2B	c.1090G>A	p.D364N	<i>De novo</i>	Novel
P53	EDMD	Exon6	Linker of coil 2B and NLS	c.1152G>C	p.E384D	<i>De novo</i>	Novel
P54	EDMD	Exon8	Ig-like domain	c.1478A>C	p.Q493P	<i>De novo</i>	Novel
P55	EDMD	Exon7	Ig-like domain	c.1357C>T	p.R453W	<i>De novo</i>	Reported
P56	EDMD	Exon7	Ig-like domain	c.1357C>T	p.R453W	<i>De novo</i>	Reported
P57	LGMD1B	Exon4	Linker of coil 1B and 2A	c.695G>A	p.G232E	<i>De novo</i>	Reported
P58	LGMD1B	Exon7	Linker of coil 2B and NLS	c.1163G>C	p.R388P	<i>De novo</i>	Novel
P59	LGMD1B	Exon6	Coil 2B	c.1106T>C	p.L369P	<i>De novo</i>	Novel
P60	LGMD1B	Exon7	Linker of coil 2B and NLS	c.1159C>G	p.L387V	<i>De novo</i>	Reported
P61	L-CMD	Exon9	Ig-like domain	c.1558T>C	p.W520R	<i>De novo</i>	Novel
P62	L-CMD	Exon4	Coil 2A	c.745C>T	p.R249W	<i>De novo</i>	Reported
P63	L-CMD	Exon1	Head	c.92_94delAGA	p.K32del	<i>De novo</i>	Reported
P64	L-CMD	Exon4	Coil 2A	c.745C>T	p.R249W	<i>De novo</i>	Reported
P65	L-CMD	Exon6	Linker of coil 2B and NLS	c.1151A>G	p.E384G	<i>De novo</i>	Reported
P66	LGMD1B	Exon9	Ig-like domain	c.1580G>C	p.R527P	<i>De novo</i>	Reported
P66's	LGMD1B	Exon9	Ig-like domain	c.1580G>C	p.R527P	<i>De novo</i>	Reported
Father							
P67	L-CMD	Exon4	Coil 2A	c.746G>A	p.R249Q	<i>De novo</i>	Reported

P68	L-CMD	Exon1	Head	c.96G>T	p.K32N	<i>De novo</i>	Novel
P69	L-CMD	Exon9	Ig-like domain	c.1580G>C	p.R527P	<i>De novo</i>	Reported
P70	L-CMD	Exon4	Coil 2A	c.745C>T	p.R249W	<i>De novo</i>	Reported
P71	LGMD1B	Exon7	Coil 2B	c.1336G>T	p.D446Y	<i>De novo</i>	Novel
P72	EDMD	Exon8	Ig-like domain	c.1406T>C	p.I469T	<i>De novo</i>	Novel
P73	LGMD1B	Exon7	Ig-like domain	c.1357C>T	p.R453W	<i>De novo</i>	Reported
P74	LGMD1B	Exon1	Head	c.95A>C	p.K32T	<i>De novo</i>	Novel
P75	EDMD	Exon5	Coil 2B	c.890_901delTCCGCAT CGACA	p.I297_D300del	<i>De novo</i>	Novel
P76	EDMD	Exon7	Ig-like domain	c.1366A>G	p.N456D	<i>De novo</i>	Reported
P77	EDMD	Exon1	Head	c.92_94delAGA	p.K32del	<i>De novo</i>	Reported
P78	L-CMD	Exon4	Coil 2A	c.745C>T	p.R249W	<i>De novo</i>	Reported
P79	EDMD	Exon7	Ig-like domain	c.1357C>T	p.R453W	<i>De novo</i>	Reported
P80	EDMD	Exon7	Ig-like domain	c.1357C>T	p.R453W	<i>De novo</i>	Reported
P81	L-CMD	Exon1	Head	c.94_96delAAG	p.K32del	<i>De novo</i>	Reported

Abbreviations: L-CMD, *LMNA*-related congenital muscular dystrophy; EDMD, Emery-Dreifuss muscular dystrophy; LGMD1B, Limb-girdle muscular dystrophy type 1B; NLS, nuclear location signal; Ig, immunoglobulin.