

Supplementary table 2. Phenotypic information for the patients harbouring *TRIM32* pathogenic variants or variants of unknown significance.

	Patient 5	Patient 14	Patient 15	Patient 27	Patient 28	Patient 35	Patient 36	Patient 37	Patient 39	
Sex	Male	Female	Female	Male	Female	Female	Female	Male	Female	
Ethnicity	Turkish	Belgian	Belgian	Bosnian Serb	Serbian	Italian	Polish	British	British	
Variant(s)	p.Arg155Asnfs Ter29	p.Glu192Glyfs Ter7	p.Glu192Glyfs Ter7	p.Asp487Asn	p.Asp487Asn	p.Arg596Gly	p.Arg613Ter	p.Ala231Glnfs Ter21	p.Arg394His	
	p.Arg155Asnfs Ter29	p.Ala388Val	p.Ala388Val	p.Asp487Asn	p.Asp487Asn	p.Arg596Gly	63.5 kb deletion	p.Met370Cysfs Ter10	p.Arg394His	
Region	Coiled-coil	Coiled-coil, NHL repeats	Coiled-coil, NHL repeats	NHL repeats	NHL repeats	NHL repeats	NHL repeats	Intervening, NHL repeats	NHL repeats	
Onset, y	10	30	30	28	25	14	34	Early 30s	45	
Last exam, y	40	56	48	38	54	42	45	42	53	
Presenting symptoms	Proximal weakness, waddling gait, difficulty climbing stairs	Exercise intolerance, difficulty climbing stairs, myalgia	Exercise intolerance, difficulty climbing stairs	Fatigue, gait difficulties, lower limb cramps, myalgia	Lower limb weakness, back pain	Scoliosis, joint laxity	Proximal lower and upper limb weakness, difficulty climbing stairs	Waddling gait, difficulty climbing stairs	Proximal weakness, myalgia	
Serum CK (U/l)	↑ (1 450)	↑ (802)	↑ (443)	↑ (1 189)	↑ (276)	↑ (≤ 2 000)	↑ (317)	↑ (1 844)	↑ (500)	
Weakness	Proximal UL	Yes	Yes	No	Yes	Yes	Mild	Yes	No	Yes
	Proximal LL	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes	Yes
	Distal UL	No	No	No	Yes	No	Mild	No	No	No
	Distal LL	Yes	Yes	Yes	Yes	Yes	No	No	Yes	No
	Other	Mild facial	Abdominal, paraspinal	Abdominal, paraspinal	Abdominal, mild facial, scapular	Abdominal, mild facial, scapular	Paraspinal	Abdominal	No	Neck flexors, scapular
Ambulatory aid, y	Wheelchair, 30	Wheelchair, 48	Wheelchair, 46	None	Wheelchair, 53	Cane, mid-30s	None	None	Unilateral, 50	
FVC (% of predicted)	ND	105	103	Normal	109	82	109	104	100	
Echocardiography	Normal	Normal	Mildly aberrant contraction pattern	Normal	Normal	Minor atrial septal aneurysm	Slight regurgitation of mitral and tricuspid valve	Normal	Normal	
Biopsy, y	Myopathic, 39	Dystrophic, 40	Dystrophic, 36	Dystrophic, 30	Myopathic, 30	Myopathic, 14	Myopathic, 42	Dystrophic, 39	Myopathic, 45	
Vacuoles	No	No	Scattered fibers	Scattered fibers	No	No	No	No	Scattered fibers	

Supplementary table 2. Phenotypic information for the patients harbouring *TRIM32* pathogenic variants or variants of unknown significance (continued).

	Patient 16	Patient 38	Patient 40	
Sex	Female	Male	Male	
Ethnicity	Persian	Pakistani	Persian	
Variant(s)	p.Ile291Ser	p.Leu163Pro	p.Leu163Pro	
	p.Ile291Ser	p.Leu163Pro	p.Leu163Pro	
Region	Intervening	Coiled-coil	Coiled-coil	
Onset, y	19	3	32	
Last exam, y	48	21	40	
Presenting symptoms	Proximal muscle weakness, exercise intolerance	Difficulty rising from sitting and walking	Proximal lower limb weakness, myalgia, mild exercise intolerance	
Serum CK (U/l)	Normal (120)	↑ (6 500)	↑ (398)	
Weakness	Proximal UL	Yes	Yes	No
	Proximal LL	Yes	Yes	Yes
	Distal UL	Yes	No	No
	Distal LL	Yes	No	No
	Other	No	Facial, scapular	Scapular
Ambulatory aid, y	Bilateral, 42	One crutch, 20	None	
FVC (% of predicted)	92	Normal	97	
Echocardiography	Normal	Normal	Normal	
Biopsy, y	Myopathic, 40	Myopathic, 15	Dystrophic, 39	
Vacuoles	Scattered fibers	No	Scattered fibers	

Upper panel: patients with pathogenic *TRIM32* variants. **Lower panel:** patients with a variant of unknown significance in *TRIM32*. VUS, variant of unknown significance; y, age in years; CK, creatine kinase; ↑, increased; UL, upper limbs; LL, lower limbs; FVC, forced vital capacity; ND, not done