

Supplementary table 1. All *TRIM32* variants detected in the MYO-SEQ cohort of patients with unexplained proximal muscle weakness.

Patient	Location		Genotype	Variant	Predicted deleteriousness			ClinVar	ExAC v3 allele frequency	ACMG
	hg19 co-ordinates (chr9)	Protein change			PolyPhen-2	MutationTaster2	FATHMM			
1	119460027	-	Het.	Syn.	No data	No data	No data	No data	0.00029	No data
2	119460338	p.Arg106His	Het.	Mis.	Probably damaging	Disease-causing	Tolerated	No data	0.00000	No data
3	119460351	-	Het.	Syn.	No data	No data	No data	No data	0.00003	No data
4	119460425	p.Thr135Ile	Het.	Mis.	Benign	Disease-causing	Tolerated	No data	0.00021	No data
6	119460518	p.Arg166Gln	Het.	Mis.	Benign	Disease-causing	Tolerated	No data	0.00004	No data
7	119460542	p.Ser174Phe	Het.	Mis.	Benign	Disease-causing	Tolerated	No data	0.00017	No data
8	119460542	p.Ser174Phe	Het.	Mis.	Benign	Disease-causing	Tolerated	No data	0.00017	No data
9	119460542	p.Ser174Phe	Het.	Mis.	Benign	Disease-causing	Tolerated	No data	0.00017	No data
11	119460579	p.Gln186His	Het.	Mis.	Possibly damaging	Disease-causing	Tolerated	Uncertain	0.00205	No data
12	119460579	p.Gln186His	Het.	Mis.	Possibly damaging	Disease-causing	Tolerated	Uncertain	0.00205	No data
13	119460579	p.Gln186His	Het.	Mis.	Possibly damaging	Disease-causing	Tolerated	Uncertain	0.00205	No data
17	119460922	p.Val301Ile	Het.	Mis.	Benign	Disease-causing	Damaging	No data	0.00000	No data
19	119461113	p.Lys364Asn	Het.	Mis.	Possibly damaging	Disease-causing	Damaging	No data	0.00000	No data
20	119461212	-	Het.	Syn.	No data	No data	No data	No data	0.00002	No data
21	119461217	p.Thr399Ile	Het.	Mis.	Benign	Disease-causing	Damaging	No data	0.00000	No data
22	119461243	p.Arg408Cys	Het.	Mis.	Possibly damaging	Disease-causing	Damaging	Uncertain	0.00143	No data
23	119461243	p.Arg408Cys	Het.	Mis.	Possibly damaging	Disease-causing	Damaging	Uncertain	0.00143	No data
24	119461244	p.Arg408His	Het.	Mis.	Benign	Disease-causing	Damaging	No data	0.00008	No data
25	119461407	-	Het.	Syn.	No data	No data	No data	No data	0.00002	No data
26	119461478	p.Thr486Ile	Het.	Mis.	Benign	Disease-causing	Tolerated	No data	0.00000	No data
29	119461480	p.Asp487Asn	Het.	Mis.	Probably damaging	Disease-causing	Damaging	Pathogenic	0.00000	No data
30	119461480	p.Asp487Asn	Het.	Mis.	Probably damaging	Disease-causing	Damaging	Pathogenic	0.00000	No data
31	119461480	p.Asp487Asn	Het.	Mis.	Probably damaging	Disease-causing	Damaging	Pathogenic	0.00000	No data
32	119461480	p.Asp487Asn	Het.	Mis.	Probably damaging	Disease-causing	Damaging	Pathogenic	0.00000	No data
33	119461792	p.Val591Met	Het.	Mis.	Probably damaging	Disease-causing	Damaging	No data	0.00002	No data
34	119461792	p.Val591Met	Het.	Mis.	Probably damaging	Disease-causing	Damaging	No data	0.00002	No data

Supplementary 1. All *TRIM32* variants detected in the MYO-SEQ cohort of patients with unexplained proximal muscle weakness (continued).

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	hg19 co-ordinates (chr9)	Protein change			PolyPhen-2	MutationTaster2	FATHMM			
5	119460478	p.Arg155AsnfsTer29	Hom.	FS	No data	Disease-causing	No data	No data	0.00000	Pathogenic
5*	119460733	p.Arg238Cys	Hom.	Mis.	Probably damaging	Disease-causing	Damaging	No data	0.00002	VUS
10*	119460579	p.Gln186His	Hom.	Mis.	Possibly damaging	Disease-causing	Tolerated	Uncertain	0.00205	VUS
14	119460593	p.Glu192GlyfsTer7	Het.	FS	No data	Disease-causing	No data	No data	0.00000	Pathogenic
14	119461184	p.Ala388Val	Het.	Mis.	Possibly damaging	Disease-causing	Tolerated	No data	0.00000	Pathogenic
15	119460593	p.Glu192GlyfsTer7	Het.	FS	No data	Disease-causing	No data	No data	0.00000	Pathogenic
15	119461184	p.Ala388Val	Het.	Mis.	Possibly damaging	Disease-causing	Tolerated	No data	0.00000	Pathogenic
16†	119460893	p.Ile291Ser	Hom.	Mis.	Benign	Disease-causing	Damaging	No data	0.00000	VUS
18*	119460944	p.Ala308Val	Het.	Mis.	Benign	Disease-causing	Damaging	No data	0.00001	VUS
18	119461222	p.Lys401Ter	Het.	Stop	No data	Disease-causing	No data	No data	0.00001	Pathogenic
27	119461480	p.Asp487Asn	Hom.	Mis.	Probably damaging	Disease-causing	Damaging	Pathogenic	0.00000	Pathogenic
28	119461480	p.Asp487Asn	Hom.	Mis.	Probably damaging	Disease-causing	Damaging	Pathogenic	0.00000	Pathogenic
35	119461807	p.Arg596Gly	Hom.	Mis.	Possibly damaging	Disease-causing	Damaging	No data	0.00001	Likely pathogenic
36	119461858	p.Arg613Ter	Het.	Stop	No data	Disease-causing	No data	No data	0.00006	Pathogenic
36	119453455-119516944	63.5 kb deletion	Het.	CNV	No data	No data	No data	No data	No data	Pathogenic

Upper panel: MYO-SEQ patients with single heterozygous rare *TRIM32* variants. **Lower panel:** MYO-SEQ patients with non-synonymous homozygous or compound heterozygous rare variants in *TRIM32*. Het., heterozygous; Hom., homozygous; Syn., synonymous; Mis., missense; FS, frameshift; Stop, stop gained; CNV, copy number variation; VUS, variant of unknown significance.

* Phenotype explained by other variant(s).

† Variant of unknown significance, no candidate variant(s) in other known myopathy genes identified.