Table e-1. Genetic variants identified among congenital myopathy patients diagnosed in adulthood

Gene	State	Variant	Families	ACMG classification	Selected references
ACTA1	Het.	c.149G>A; p.Gly50Asp	1	VUS	(1)
ACTA1	Hom.	c.460G>T; p.Val154Leu	2	Likely pathogenic	(2) (same AA change)
ACTA1	Het.	c.587C>T; p.Thr196Ile	3	Likely pathogenic	
ACTA1	Het.	c.757G>C; p.Gly253Arg	4	VUS	(3)
ACTA1	Het.	c.811A>G; p.Met271Val	5	VUS	(4)
CACNA1S	Het.	c.1583G>T; p.Arg528Leu	6	VUS	(5)
DNM2	Het.	c.1106G>A; p.Arg369Gln	7	Pathogenic	(6-8)
MYH2	Comp. het.	c.1546T>G; p.Phe516Val	8	VUS	
MYH2	Comp. het.	c.3331C>T; p.Gln1111*	8	Likely pathogenic	
MYH7	Het.	c.1358G>A; p.Arg453His	9	Pathogenic	(9)
MYH7	Het.	c.2594A>G; p.Lys865Arg	10	Likely pathogenic	(10, 11)
MYH7	Comp. het.	c.5459G>A; p.Arg1820Gln	11	VUS	(12)
RYR1	Het.	c.1841G>T; p.Arg614Leu	12	Pathogenic	(13, 14)
RYR1	Het.	c.5132A>G; p.Tyr1711Cys	13	Likely pathogenic	(15)
RYR1	Het.	c.6617C>T; p.Thr2206Met	14, 15, 16	Pathogenic	(13, 16)
RYR1	Het.	c.7300G>A; p.Gly2434Arg	17	Pathogenic	(13, 16)
RYR1	Het.	c.4711A>G, p.Ile1571Val; c.10097G>A, p.Arg3366His; c.11798A>G, p.Tyr3933Cys	18	Likely pathogenic	(17)
RYR1	Comp. het.	c.10648C>T; p.Arg3550Trp	19	Likely benign	(18)
RYR1	Het.	c.10690_10703del; p.Glu3564Serfs*20	20	Likely pathogenic	
RYR1	Het.	c.11126C>T; p.Ala3709Val	21	VUS	
RYR1	Het./comp. het.	c.12315_12328del; p.Glu4106Alafs*8	19	Pathogenic	(18)
RYR1	Het.	c.14818G>A; p.Ala4940Thr	22, 23, 24	Pathogenic	(16, 19-22)
SELENON	Comp. het.	c.713dupA; p.Asn238Lysfs*63	25, 26	Pathogenic	(23)
SELENON	Comp. het.	c.943G>A; p.Gly315Ser	26, 27, 28	Pathogenic	(23-25)
SELENON	Comp. het.	c.1028T>G; p.Met343Arg	25	VUS	(26)
SELENON	Comp. het.	c.1332_1334delCAA; p.Asn444del	27	Likely pathogenic	
SELENON	Comp. het.	c.1405C>T; p.Arg469Trp	28	Likely pathogenic	(27)
TIA1	Comp. het.	c.1150G>A; p.Glu384Lys	11	Pathogenic	(12, 28)
TPM2	Het.	c.349G>A; p.Glu117Lys	29	Likely pathogenic	(29, 30)

Note: for variants classified as VUS, pathogenicity was supported by patients' phenotypes, histological findings and/or variant segregation, as detailed in the body of the manuscript and in previous publications (see references).

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