

Patient	Age	Sex	Age onset	Hyper CK-emia at age	CK level (U/l) at study onset	First symptoms	Pheno-type	Nucleotide change	Protein predicted	Western Blotting	Immu-no-histochemi-stry
1*	26	f	19	19	4948	Climbing stairs impaired	LGM D	Exon 9: c.863dupA (p.Asp288GlufsX40), Exon 30: c.3191_3196dup (p.Ala1064_Glu1065dup) [§]	present	no specific band	negative
2*	30	f	23	18	6395	Climbing stairs impaired	LGM D	Exon 9: c.863dupA (p.Asp288GlufsX40), Exon 30: c.3191_3196dup (p.Ala1064_Glu1065dup) [§]	present	not done	not done
3	38	f	24	13	5865	Calf atrophy, tiptoe walking impaired	MM	Exon 4: c.265C>T (p.Arg89X), Exon 44: c.4872_4876delinsCCCC (p.Glu1624AspfsX10)	absent	no specific band	negative
4	33	m	27	27	4128	Fatigue, running impaired	LGM D	Exon 26: 2779delG (p.Ala927LeufsX21) homozygous	absent	no specific band	negative
5	34	m	33	14	4685	Climbing stairs impaired	LGM D	Exon 24: c.2367C>A (p.Ser789Arg), Exon 53: c.5979dupA (p.Glu1994ArgfsX3)	present	no specific band	negative
6 [†]	26	m	17	20	6345	Fatigue, climbing stairs	LGM D	Exon 21: c.1976dupT (p.Val660GlyfsX12) homozygous	absent	not done	negative
7	44	f	35	35	1932	Calf atrophy, tiptoe walking impaired	MM	Exon 29: c.3118C>T (p.Arg1040Trp), Exon 53: c.5979dupA (p.Glu1994ArgfsX3)	present	severely reduced	negative
8 [‡]	27	f	18	14	2903	Getting up from squatting and tiptoe walking impaired	LGM D	Exon 43: c.4756C>T (p.Arg1586X), Exon 45: c.5022delT (p.Phe1674LeufsX48)	absent	no specific band	negative
9 [‡]	20	m	18	6	7237	Getting up from squatting and tiptoe walking impaired	LGM D	Exon 43: c.4756C>T (p.Arg1586X), Exon 45: c.5022delT (p.Phe1674LeufsX48)	absent	not done	not done
10 [‡]	30	f	16	21	2487	Climbing stairs impaired	LGM D	Exon 43: c.4756C>T (p.Arg1586X), Exon 45: c.5022delT (p.Phe1674LeufsX48)	absent	not done	not done
11	35	m	30	18	12620	Calf atrophy, tiptoe walking impaired	MM	Exon 5: c.401C>T (p.Pro134Leu), Exon 45: c.5022delT (p.Phe1674LeufsX48)	present	mildly reduced	reduced
12	24	f	13	13	9099	Calf hypertrophy and pronounced tiptoe walking, walking on heels impaired	DMA T	Exon 4: c.3113dupC (p.Leu105ProfsX43), Intron 49: c.5525+3 A>G (splice)	unknown	no specific band	negative
13	27	m	18	21	7785	Climbing stairs impaired	DMA T	Exon 4: c.3113dupC (p.Leu105ProfsX43), Intron 49: c.5525+3 A>G (splice)	unknown	not done	not done
14*	36	f	22	22	4996	Tiptoe walking and climbing stairs impaired	MM	Exon 6: c.610C>T (p.Arg204X), Exon 22: c.2113G>A (p.Val705Met), Intron 49: c.5525+1G>A (splice)	present	not done	not done
15*	32	m	20	21	6480	Tiptoe walking impaired	MM	Exon 6: c.610C>T (p.Arg204X), Exon 22: c.2113G>A (p.Val705Met), Intron 49: c.5525+1G>A (splice)	present	no specific band	negative
16	25	f	22	15	7536	Tiptoe walking and climbing stairs impaired	LGM D	Exon 34: c.3832C>T (p.Glu1278X), deletion of exon 31 heterozygous	unknown	no specific band	negative
17	60	f	25	20	1989	Climbing stairs impaired	LGM D	Exon 6: c.610C>T (p.Arg204X), Exon 12: c.1120G>C (p.Val374Leu)	present	no specific band	severely reduced
18	26	m	18	22	7995	Calf atrophy, climbing stairs impaired	LGM D	deletion of exons 37-40 homozygous	absent	no specific band	negative
19 [†]	19	f	17	17	7539	Myalgia, tiptoe walking and climbing stairs impaired	LGM D	Exon 21: c.1976dupT (p.Val660GlyfsX12) homozygous	absent	not done	not done
20 [†]	19	f	17	17	5442	Tiptoe walking and climbing stairs impaired	LGM D	Exon 21: c.1976dupT (p.Val660GlyfsX12) homozygous	absent	not done	not done
21	39	m	17	17	3324	Calf atrophy, tiptoe walking and climbing stairs impaired	LGM D	Exon 7: c.763delC (p.His255ThrfsX33), Exon 29: c.3059dupC (p.Glu1021GlyfsX11)	absent	no specific band	negative
22	36	m	20	20	2825	Climbing stairs impaired	LGM D	Exon 4: c.331C>T (p.Gln111X) homozygous	absent	no specific band	negative
23	42	m	32	32	3861	Calf atrophy, climbing stairs impaired	LGM D	Intron 28: c.3031+2T>C (splice), Exon 34: c.3832C>T (p.Gln1278X)	present	not done	negative
24 [‡]	31	m	22	22	3941	Proximal leg and calf atrophy, climbing stairs and tiptoe walking impaired	LGM D	Exon 30: c.3321delA (p.Ala1108LeufsX12) homozygous	absent	no specific band	unknown
25 [‡]	27	m	22	22	5114	Proximal leg atrophy, climbing stairs impaired	LGM D	Exon 30: c.3321delA (p.Ala1108LeufsX12) homozygous	absent	not done	not done

f = female, m = male, red = protein present, black = protein absent, blue = unknown.

*, †, ‡, § = sibs from five different families, § = polymorphism