

Table 1

Nerve	Index		Sister		Mother	
	CMAP/mV	mNCV/ms ⁻¹	CMAP/mV	mNCV/ms ⁻¹	CMAP/mV	mNCV/ms ⁻¹
N. ulnaris [4,0 mV / 50 ms ⁻¹]	23	68	19	61	15	57
N. tibialis [5,0 mV / 40 ms ⁻¹]	11	51	24	51	6,4	40
N. peroneus [4,0 mV / 41 ms ⁻¹]	9,9	47	n.d.	n.d.	n.d.	n.d.
Nerve	Index		Sister		Mother	
	SNAP/ μ V	sNCV/ms ⁻¹	SNAP/ μ V	sNCV/ms ⁻¹	SNAP/ μ V	sNCV/ms ⁻¹
N. radialis [16 μ V / 55 ms ⁻¹]	8,8	67	15	55	6,1	58
N. suralis [3,8 μ V / 39 ms ⁻¹]	1,6	50	2,7	43	\emptyset	n.a.
Muscle	Index		Sister		Mother	
	ASA	MUAP/mV	ASA	MUAP/mV	ASA	MUAP/mV
M. deltoideus [1,5 mV]	\emptyset	2,0	\emptyset	2,7	\emptyset	6,4
M. IOD1 [2,3 mV]	\emptyset	7,5	n.d.	n.d.	\emptyset	12
M. vastus medialis [2,0 mV]	\emptyset	16	\emptyset	28	\emptyset	28
M. tibialis anterior [1,6 mV]	\emptyset	4,3	n.d.	n.d.	n.d.	n.d.

Table 2

Mutation						
DNA	Protein	Expression	RING domain	Transmission	Reference	
c.1913-1G>A	p.Glu638Ala/fsX7	∅	n.a.	recessive	Guernsey et al. [2010]	
c.2122-2123insGC	p.Leu708Arg/fsX28	n.d.	disrupted	dominant	Weterman et al. [2012]	
c.2047-1G>A	p.Ala683ProfsX3	+	disrupted	dominant	Nicolaou et al. [2013]	
c.2046+1G>T	p.Glu682_Ala683ins21	n.d.	disrupted	dominant	herein	