Supplementary Materials

TABLES

Table 1. T1 Scoring Scale

Grade	Mercuri et al. 2002	Komblum et al. 2006	Fisher et al. 2008	Garibaldi et al.
0		Normal apparence	Normal appearance	Normal apparence
1	Normal apparence	Discrete moth-eaten appearance with sporadic T1 hyperintense area	Mild: traces of increased signal intensity on the T1-weighted MR equences	Minimal: minimal/initial replacement (one isolated area)
2	Mild involvement: Early moth-eaten appearance with scattered small areas of increased signal or with numerous discrete areas of increased signal with beginning confluence, comprising less than 30% of the volume of the individual muscle	a. Moderate moth-eaten appearance with numerous scattered T1 hyperintense areas b. Late moth-eaten appearance with numerous confluent T1 hyper-intense areas	Moderate: increased T1-weighted signal intensity with beginning confluence in less than 50% of the muscle	Mild: <50% volume replacement
3	Moderate involvement: Late moth-eaten appearance with numerous discrete areas of increased signal with beginning confluence, comprising 30-60% of the volume of the individual muscle	Complete fatty degeneration, replacement of muscle by connective tissue and fat	Severe: increased T1-weighted signal intensity with beginning confluence in more than 50% of the muscle	Moderate: ≥50% replacement
4	Severe involvement: Washed-out appearance, fuzzy appearance due to confluent areas of increased signal or an end-stage appearance, with muscle replaced by increased density connective tissue and fat, and only a rim of fascia and neurovascular structures distinguishable		End-stage appearance, entire muscle replaced by increased density of connective tissue and fat	Severe: complete replacement (≥99%);
5				End-stage atrophy: complete muscle atrophy fibro-fatty replacement not-evaluable

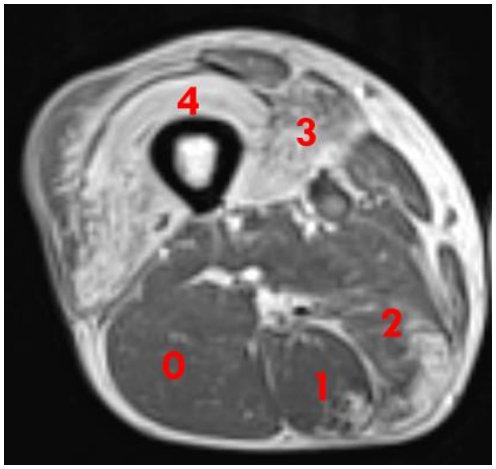
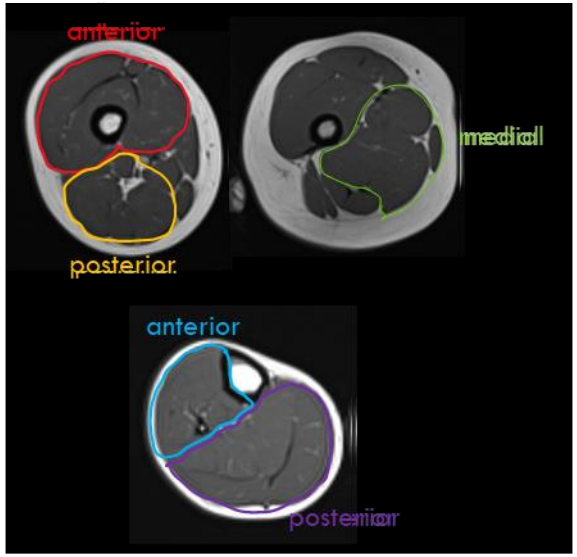


Table 2. Atrophy Scoring system and scale

Lower limbs' compartments

Thigh

Leg

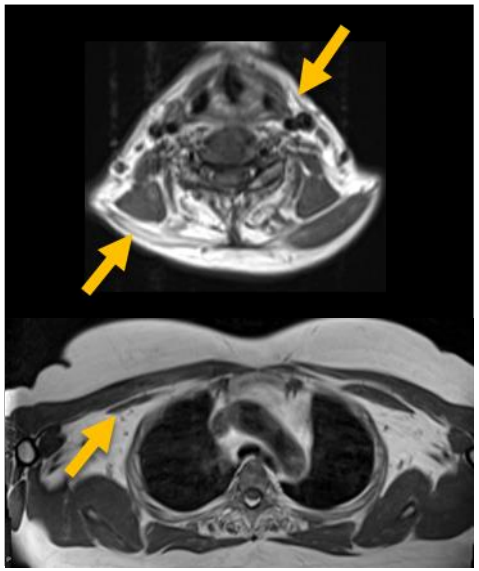


Upper body muscles

Sternocleidomastoideus

Trapezius

Pectoralis



Grade	Brogna et al.2020	Garibaldi et al.
0	Normal	Normal muscle bulk
1	Mild peripheral muscle volume loss	Minimal: peripheral muscle volume loss
2	< 50% of muscle volume loss	Mild: < 50% of muscle volume loss
3	≥ 50% of muscle volume loss	Moderate: ≥ 50% of muscle volume loss
4		Severe: complete muscle atrophy

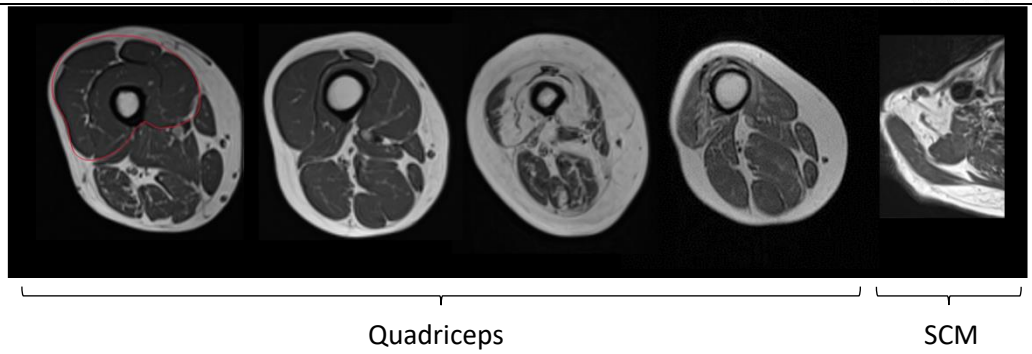


Table 3. Average and Median T1 score per muscle

	Average T1 score	Median T1 score
Gastrocnemius Medialis	2,205223881	3
Sternocleidomastoideus	1,73148148	0
Soleus	1,604477612	1
Flexor Hallucis Longus	1,425373134	1
Multifidus	1,369402985	1
Genioglossus	1,14285714	1
Paraspinous (Thoracic)	1,13913043	1
Paraspinous (Cervical)	1,06086957	1
Tibialis Anterior	1,041044776	0
Gastrocnemius Lateralis	0,98880597	0
Extensor Digitorum/Allucis Longus	0,925373134	0
Vastus Intermedius	0,891791045	1
Vastus Medialis	0,861940299	0
Gluteus Minimus	0,850746269	1
Peroneus Longus/Brevis	0,77238806	0
Rectus Abdominis	0,76119403	0
Vastus Lateralis	0,757462687	0
Iliocostalis Lomborum	0,697761194	0
Tensor Fasciae Latae	0,679104478	0
Semimembranosus	0,611940299	0
Latissimus Dorsi	0,53913043	0
Sartorius	0,537313433	0
Biceps Femoris Caput Brevis	0,496268657	0
Obliquus Ext/Int/Trasv	0,470149254	0
Pectoralis Minor	0,39473684	0
Trapezius	0,33043478	0
Gracilis	0,320895522	0
Tibialis Posterior/Flexor Digitorum Longus	0,317164179	0
Semitendinosus	0,276119403	0
Biceps Femoris Caput Longum	0,25	0
Adductor Longus	0,182835821	0
Rectus Femoris	0,171641791	0
Average upper body T1 score	0,75	
Average lower body T1 score	1,47	

**distal-proximal gradient: 100% (83/83). Not evaluable in the remaining patients because of total muscle sparing; UL: up*

Table 4. characteristics of patients

Patient	Age at MRI	Age at Disease onset (ys)	Clinical form (0= cong, 1=infa, 2=juv, 3=classic, 4=late)	Disease duration	Sex (M=0, F=1)	CTG range	CTG average	Expansion_Class (1= E1; 2= E2; 3= E3)	Expansion_Class Revised (1= E1a; 2=E1b; 3= E2a; 4= E2b; 5=E2c; 6=E3a; 7=E3b)	MIRS at 1st MRI	T1 LL	T1 UL	T1 TOT	STIR	atrophy SCM	Marble
p1	49	10	1	39	0	350-780	565	2	4	3	36	6	42	16	no	1
p2	43	0	0	43	0	570-1080	825	2	5	4	116	67	183	10	yes	0
p3	44	21	3	23	0	250	250	2	3	3	25	12	37	9	1	0
p4	50	40	3	10	1	680-1250	965	3	5	4	90			1	n.a.	0
p5	53	40	3	13	1	785-965	875	2	5	4	118	40	158	0	yes	0
p6	53	16	2	37	0	96	96	1	1	4	114	44	158	8	yes	0
p7	15	8	1	7	0	350	350	2	3	1	2	0	2	0	no	0
p8	37	22	3	15	1	130	130	2	2	3	24	3	27	6	no	0
p9	25	20	2	5	1	50-1000	475	2	3	2	2	0	2	0	no	0
p10	56	56	4	0	0	50-85	70	1	1	1	13	0	13	4	no	1
p11	55	51	4	4	0	70	70	1	1	2	0	1	1	0	no	1
p12	27	16	2	11	1	390-500	445	2	3	3	18	1	19	4	no	0
p13	29	19	2	10	1	200-450	325	2	3	3	0	0	0	0	no	0
p14	54	18	2	36	0	1000	1000	3	6	4	108	48	156	10	yes	0
p15	20	18	2	2	0	150-1000	575	2	4	3	0	1	1	5	no	0
p16	57	47	4	10	0	90-240	210	2	3	4	112	34	146	8	2	0
p17	54	53	4	1	0	150-1000	575	2	4	3	3	9	12	2	no	1
p18	26	0	0	26	1	1000-1500	1250	3	6	3	73	50	123	7	yes	0
p19	18	0	0	18	1	1000	1000	3	6	3	13	4	17	4	no	0
p20	58	35	3	23	1	250	250	2	3	5	140			4	n.a.	0
p21	35	10	1	25	0	372-850	611	2	4	3	23	0	23	11	no	0
p22	31	16	2	15	0	400-570	485	2	3	3	75	28	103	19	no	0
p23	33	0	0	33	0	940-1250	1095	3	6	3	6	8	14	7	1	1
p24	27	25	3	2	0	150-1000	575	2	4	3	16	1	17	4	no	0
p25	57	57	4	0	0	50-150	100	1	2	1	0	3	3	2	no	1
p26	54	30	3	24	0	500	500	2	4	4	117	35	152	9	yes	0
p27	63	53	4	10	0	82	82	1	1	3	11	5	16	5	no	0
p28	55	25	3	30	0	120	120	1	2	3	66	13	79	22	no	0
p29	44	30	3	14	0	360	360	2	3	4	88			20	n.a.	0

p30	41	35	3	6	0	150-1000	425	2	3	2	4	2	6	6	no	1
p31	50	35	3	15	1	85	85	1	1	3	3	0	3	2	no	0
p32	47	40	3	7	0	255-580	417	2	4	4	99	11	110	17	no	1
p33	53	48	4	5	0	120	120	1	2	2	0	0	0	4	no	0
p34	65	55	4	10	0	120	120	1	2	2	1	9	10	0	yes	1
p35	53	35	3	18	1	300	300	2	3	3	24	4	28	2	no	0
p36	24	12	2	12	0	350	350	2	3	2	2	0	2	4	no	0
p37	50	46	4	4	0	670	670	2	4	3	89			7	n.a.	0
p38	48	15	2	33	0	110	110	1	2	4	113	45	158	10	yes	0
p39	30	30	3	0	0	150	150	1	2	2	2	0	2	0	no	0
p40	72	72	4	0	0	64	64	1	1	1	12	2	14	0	no	1
p41	58	37	3	21	1	300	300	2	3	4	44	21	65	7	yes	0
p42	76	30	3	46	0	500-700	600	2	4	3	37	8	45	16	no	1
p43	47	25	3	22	0	350	350	2	3	3	24	5	29	10	no	1
p44	46	30	3	16	1	180	180	2	2	3	36	8	44	15	no	1
p45	20	10	1	10	0	1000	1000	3	6	2	0	1	1	0	no	0
p46	54	25	3	29	0	450-530	480	2	3	3	53	16	69	19	no	0
p47	44	0	0	44	0	1250-1800	1525	3	7	3	96	34	130	6	yes	0
p48	57	30	3	27	0	350-800	575	2	4	3	96	24	120	2	yes	0
p49	53	53	4	0	1	45	45	1	1	2	15	3	18	3	no	0
p50	71	71	4	0	0	70	70	1	1	1	8	3	11	5	no	1
p51	53	19	2	34	1	500-1000	750	2	5	4	81	28	109	11	yes	0
p52	36	28	3	8	0	490-630	560	2	4	3	30	22	52	17	yes	1
p53	42	42	4	0	1	73	73	1	1	1	2	0	2	0	no	0
p54	35	28	3	7	0	150-1000	425	2	3	3	20	1	21	12	no	0
p55	40	29	3	11	1	220-365	292	2	3	3	25	5	30	8	no	0
p56	75	75	4	0	0	55-70	62	1	1	2	18	8	26	0	no	1
p57	81	81	4	0	1	50	50	1	1	2	37	8	45	0	no	1
p58	47	20	2	27	1	400-600	500	2	4	3	75	20	95	3	3	1
p59	59	15	2	44	1	150-250	200	2	3	3	25	2	27	4	no	0
p60	48	25	3	23	0	80-200	140	1	2	3	22	16	38	18	yes	0
p61	36	17	2	19	0	130-210	170	1	2	3	5	0	5	5	no	1
p62	38	12	2	26	0	500	500	2	4	3	49			25	n.a.	0
p63	45	15	2	30	0	125	125	1	2	4	121	36	157	21	yes	0

p64	47	44	4	3	0	>350	350	2	3	3	49	27	76	20	yes	0
p65	43	35	3	8	1	>350	350	2	3	3	93	38	131	10	yes	0
p66	74	68	4	6	0	85	85	1	1	2	36	16	52	7	no	0
p67	22	16	2	6	1	85-150	117	1	2	1	2	0	2	3	no	0
p68	31	5	1	26	0	400	400	2	3	3	19	7	26	18	no	0
p69	33	27	3	6	1	150-1000	425	2	3	3	42	7	49	13	no	0
p70	23	10	1	13	0	489	489	2	3	2	4	5	9	6	no	1
p71	53	12	2	41	1	550	550	2	4	4	119	36	155	6	yes	0
p72	46	30	3	16	1	>170	170	2	2	4	23	5	28	7	no	0
p73	57	35	3	22	1	>170	170	2	2	4	30	2	32	6	no	0
p74	42	10	1	32	0	300	300	2	3	3	27	7	34	11	no	0
p75	44	18	2	26	0	>170	170	2	2	3	128	60	188	20	yes	0
p76	34	29	3	5	1	667	667	2	4	2	5	1	6	0	no	0
p77	36	16	2	20	0	50-150	100	1	2	2	9			2	n.a.	0
p78	48	39	3	9	0	91	91	1	1	2	1	12	13	1	yes	0
p79	39	25	3	14	1	165-330	250	2	3	2	4	11	15	2	yes	0
p80	54	28	3	26	1	250	250	2	3	2	97	22	119	0	yes	0
p81	40	27	3	13	0	400-630	515	2	4	3	31			11	n.a.	0
p82	48	5	1	43	1	350	350	2	3	3	49	22	71	13	yes	0
p83	43	30	3	13	0	120-350	235	2	3	3	14			2	n.a.	0
p84	29	20	2	9	0	200	200	2	3	2	9	0	9	3	n.a.	0
p85	24	16	2	8	0	700	700	2	4	2	2	0	2	0	no	0
p86	35	10	1	25	1	415-935	675	2	4	4	97	13	110	24	no	0
p87	72	60	4	12	0	>350	350	2	3	2	60	22	82	4	yes	0
p88	41	11	2	30	1	250-1100	675	2	4	3	79	32	111	10	yes	0
p89	60	30	3	30	1	288-389	338	2	3	4	42			12	n.a.	0
p90	43	33	3	10	1	1000-1400	1200	3	6	4	53			14	n.a.	0
p91	41	17	2	24	1	250	250	2	3	3	44			3	n.a.	0
p92	59	40	3	19	0	55-3395	1725	3	7	2	20	1	21	9	no	0
p93	46	38	3	8	0	250	250	2	3	4	82	27	109	13	yes	0
p94	51	16	2	35	0	>350	350	2	3	4	51	15	66	2	yes	0
p95	28	27	3	1	0	>250	250	2	3	2	9	5	14	8	n.a.	0
p96	39	19	2	20	0	300-450	375	2	3	3	2	0	2	4	n.a.	0
p97	41	28	3	13	0	100-275	187	2	2	3	18			1	n.a.	1
p98	42	15	2	27	0	400	400	2	3	3	20	2	22	1	n.a.	0
p99	56	13	2	43	1	335-650	490	2	3	4	111	12	123	23	n.a.	0
p100	35	15	2	20	1	210	210	2	3	2	33	4	37	7	no	0

p101	53	40	3	13	1	74	74	1	1	2	4	0	4	2	no	0
p102	54	50	4	4	0	130-350	240	2	3	3	15	4	19	2	no	1
p103	42	30	3	12	1	20-200	110	2	2	4	67			18	n.a.	0
p104	44	20	2	24	0	75-330	202	2	3	2	1			2	n.a.	0
p105	22	15	2	7	0	220-600	211	2	3	2	2	1	3	0	no	0
p106	22	8	1	14	1	500-1000	750	2	5	2	1			9	n.a.	0
p107	55	46	4	9	1	170	170	2	2	2	0	0	0	0	no	0
p108	34	22	3	12	0	>350	350	2	3	3	11	1	12	0	no	0
p109	61	60	4	1	0	>400	400	2	3	2	58	12	70	11	n.a.	0
p110	34	20	2	14	1	>400	400	2	3	1	20			8	n.a.	0
p111	51	47	4	4	1	380	380	2	3	2	28	13	41	3	no	0
p112	68	40	3	28	0	200	299	2	3	3	90	21	111	7	yes	0
p113	39	39	3	0	0	80-145	112	1	2	2	43	10	53	5	no	0
p114	22	8	1	14	0	200-400	300	2	3	2	0	0	0	2	no	0
p115	49	15	2	34	0	333-900	616	2	4	4	65	17	82	20	yes	0
p116	60	30	3	30	0	>250	250	2	3	4	73	13	86	2	n.a.	1
p117	29	15	2	14	1	>350	350	2	3	3	39	0	39	3	no	1
p118	51	15	2	36	0	300	300	2	3	4	68	28	96	10	yes	0
p119	47	45	4	2	0	>400	400	2	3	3	55	20	75	1	yes	0
p120	48	48	4	0	0	91	91	1	1	1	6			3	n.a.	0
p121	49	35	3	14	0	90-250	170	2	2	2	30			4	n.a.	0
p122	49	44	4	5	0	110-300	220	2	3	3	26	4	30	4	no	0
p123	71	60	4	11	1	130-350	240	2	3	3	14	8	22	3	no	0
p124	38	15	2	23	1	1100	110	1	2	3	65	9	74	17	no	0
p125	50	30	3	20	1	300-1100	700	2	4	4	67	21	88	21	yes	0
p126	68	33	3	35	1	200-300	250	2	3	4	56			16	n.a.	0
p127	12	5	1	7	0	>400	400	2	3	3	2	0	2	0	no	0
p128	57	45	4	12	0	115-250	180	2	2	4	33	17	50	12	yes	1
p129	46	8	1	38	1	730	730	2	4	4	67	25	92	2	yes	1
p130	49	40	3	9	1	>350	350	2	3	4	9	4	13	1	no	0
p131	20	18	2	2	0	>200	200	2	3	2	0	11	11	0	yes	0
p132	68	65	4	3	1	125-140	262	2	3	4	32	2	34	4	no	1
p133	51	40	3	11	1	>300	300	2	3	4	94	25	119	12	yes	0
p134	73	55	4	18	1	75-350	212	2	3	1	12	5	17	0	no	0