

Supplementary Table 1: SMA genetic screening results for copy numbers of *SMN*.

Gene	Sample	Phenotype	MLPA				ddPCR			
			Test 1		Test 2		Test 1		Test 2	
			Ratio	Copy Number	Ratio	Copy Number	Ratio	Copy Number	Ratio	Copy Number
<i>SMN1-E7</i>										
	P-1*	SMA Type I	0	0	0	0	0.01	0	0	0
	P-2	SMA Type II	0	0	0	0	0	0	0	0
	P-3	SMA Type III	0	0	0	0	0.01	0	0.01	0
	P-4	SMA Type III	0	0	0	0	0	0	0	0
	P-5	SMA Type III	0	0	0	0	0	0	0	0
	P-6	SMA Type III	0	0	0	0	0	0	0	0
	P-7	<i>SMN1</i> (1+0) carrier [†]	0.52	1	0.63	1	0.97	1	0.95	1
	P-8	<i>SMN1</i> (1+0) carrier	0.49	1	0.6	1	0.96	1	0.99	1
	P-9	<i>SMN1</i> (1+0) carrier	0.44	1	0.56	1	0.8	1	0.83	1
	P-10	Normal Control	1.15	2	1.37	3	2.01	2	1.98	2
	F1-II-1 [‡]	SMA-asymptomatic	0.05	0	0	0	0	0	0	0
	F1-II-1-f [§]	SMA-asymptomatic	0	0	0	0	/	0	/	0
	F1-II-2	SMA Type III	0.05	0	0	0	0.01	0	0.01	0
	F1-II-2-f	SMA Type III	0	0			/	0	/	0
	F2-II-1	<i>SMN1</i> (1+0) carrier	0.43	1	0.54	1	1.1	1	1.09	1
	F2-II-2	<i>SMN1</i> (c.844C>T+1) carrier	1.05	2	1.29	2-3	1.04	1	1.06	1
	F2-II-3	<i>SMN1</i> (c.844C>T+1) carrier	0.72	1-2	0.87	2	0.87	1	0.91	1
	F2-II-4	<i>SMN1</i> (1+0) carrier	0.48	1	0.6	1	0.84	1	0.86	1
	F2-III-2	<i>SMN1</i> (c.844C>T+1) carrier	1.18	2	1.41	3	1.03	1	1.06	1
	F2-III-4	SMA Type I <i>SMN1</i> (c.844C>T+0) [¶]	0.55	1	0.68	1-2	0	0	0	0
	F3-I-1	<i>SMN1</i> (2+0) carrier ^{**}	1	2	1.24	2-3	1.91	2	1.89	2
	F3-I-2	<i>SMN1</i> (c.815A>G+1) carrier ^{††}	1.04	2	1.29	2-3	1.97	2	1.95	2
	F3-II-1	SMA Type II <i>SMN1</i> (c.815A>G+0) ^{**}	0.51	1	0.62	1	0.97	1	0.97	1
	i-1 ^{§§}	SMA Type I	0.08	0	0	0	0	0	0	0
	i-2	SMA Type I	0	0	0	0	/	0	0	0
	i-3	SMA Type II	0	0	0	0	0.01	0	0.01	0
	SD-019	Reference Selection DNA	1.04	2	0.97	2	1.96	2	2.1	2
<i>SMN2-E7</i>										
	P-1	SMA Type I	1.58	3	2.01	4	2.97	3	3.02	3
	P-2	SMA Type II	1.47	3	2.03	4	2.89	3	3.05	3
	P-3	SMA Type III	1.67	2-3	2.18	>4	3.07	3	3.03	3
	P-4	SMA Type III	4.02	>4	2.83	>4	3.98	4	3.9	4
	P-5	SMA Type III	1.48	3	1.99	4	3.02	3	2.92	3

P-6	SMA Type III	2.22	> 4	2.97	> 4	3.82	4	4.04	4
P-7	<i>SMNI</i> (1+0) carrier	1.44	3	1.82	4	1.99	2	1.99	2
P-8	<i>SMNI</i> (1+0) carrier	1.04	2	1.38	3	1.97	2	1.97	2
P-9	<i>SMNI</i> (1+0) carrier	0.94	2	1.25	2-3	1.71	2	1.7	2
P-10	Normal Control	0.48	1	0.51	1	0.99	1	1	1
F1-II-1	SMA-asymptomatic	1.2	2	2.1	4	3.93	4	3.88	4
F1-II-1-f	SMA-asymptomatic	1.74	3-4	1.85	4	3.85	4	3.99	4
F1-II-2	SMA Type III	1.76	4	1.91	4	3.82	4	3.88	4
F1-II-2-f	SMA Type III	4.46	> 4			3.88	4	3.96	4
F2-II-1	<i>SMNI</i> (1+0) carrier	1.07	2	1.4	3	2.04	2	2.05	2
F2-II-2	(c.844C>T+1) carrier <i>SMNI</i>	1.11	2	1.42	3	2.08	2	2.03	2
F2-II-3	(c.844C>T+1) carrier <i>SMNI</i>	0.99	2	1.27	2-3	1.85	2	1.92	2
F2-II-4	<i>SMNI</i> (1+0) carrier	1.06	2	1.38	3	1.8	2	1.91	2
F2-III-2	(c.844C>T+1) carrier <i>SMNI</i>	1.14	2	1.42	3	2.09	2	2.09	2
F2-III-4	SMA Type I <i>SMNI</i> (c.844C>T+0)	0.69	1-2	0.83	2	2.22	2	2.22	2
F3-I-1	<i>SMNI</i> (2+0) carrier <i>SMNI</i>	1.72	3-4	2.17	> 4	3.02	3	2.91	3
F3-I-2	(c.815G>A+1) carrier <i>SMNI</i>	1.09	2	1.42	3	1.96	2	1.98	2
F3-II-1	SMA Type II <i>SMNI</i> (c.815G>A+0)	1.69	3-4	2.17	> 4	2.89	3	2.93	3
i-1	SMA Type I	1	2	1.34	3	2.06	2	2.1	2
i-2	SMA Type I	2.88	> 4	3.12	> 4	3.01	3	3.04	3
i-3	SMA Type II	2.53	> 4	2.53	> 4	3.09	3	3.07	3
SD-019	Reference Selection DNA	1.07	2	0.99	2	2.04	2	2.01	2

*P: PBMC (Peripheral Blood Mononuclear Cells); † *SMNI* (1+0) carrier: heterogeneous deletion of *SMNI*; ‡ F: Family; § f: fibroblast; || *SMNI* (c.844C>T+1) carrier: have two copies of *SMNI* with one variant c. 844C>T; ¶ *SMNI* (c.844C>T+0): have one copy of *SMNI* with the variant c. 844C>T; ** *SMNI* (2+0) carrier: an allele that contains an *SMNI* duplication in cis, while the other allele contains no *SMNI*; †† *SMNI* (c.815A>G+1) carrier: have two copies of *SMNI* with one variant c.815A>G; †† *SMNI* (c.815A>G+0): have one copy of *SMNI* with the variant c.815A>G; §§ i: iPSCs (induced Pluripotent Stem Cells).