

Supplementary Information

S1 figure: Comprehensive clinical details of sarcoglycanopathy cases in our cohort.

Patient ID	Age at onset (Years)	Age at Evaluation (Years)	Age of Loss of Ambulation /assistance needed (Years)	creatine kinase level (IU/L)	Gender	Consanguinity	Family History	Calf Muscle Hypertrophy	Tendon Contracture	Alpha expression	Beta expression	Gamma expression	Delta expression	Mobility status at evaluation	Current Mobility status	Unique clinical findings		
																Scapular winging	Muscle pain	Macroglossia
SG-1	2	8	-	11109	M													
SG-2	4	5	-	16093	F													
SG-3	7	11	14	NA	M													
SG-4	4	15	-	NA	M													
SG-5	9	10	11	10492	M													
SG-6	3	11	-	1073	F													
SG-7	5	6	14	18869	M													
SG-8	2.5	9	10	17210	F													
SG-9	3	8	11	7474	M													
SG-10	4	9	13	5846	M													
SG-11	14	19	20	7730	M													
SG-12	2	9	10	2088	F													
SG-13	2	8	-	5770	M													

	Independently ambulant		Need physical assistance		Wheel chair Bound		Bedbound		Loss of followup		Present		Absent
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S1 table: The genotypes of 5 markers selected surrounding the c.544T>G (p.Thr182Pro) (rs751427686) mutation for the cases and unaffected related controls.

Pedigree #	ID	rs10009426	rs6824707	rs2271046	rs751427686	rs35414474	rs17611952
Ped1	SG-1	G/G	A/A	T/T	G/G	G/G	T/T
Ped2	SG-2	G/G	A/A	T/T	G/G	G/G	T/T
Ped3	SG-3	G/G	A/A	T/T	G/G	G/G	T/T
Ped3	SG-3-S	G/G	A/A	T/T	G/G	G/G	T/T
Ped3	SG-3-P1	G/G	A/A	T/T	G/T	C/G	A/T
Ped4	SG-4-P1	G/G	A/A	T/T	G/T	C/G	A/T
Ped4	SG-4-P2	A/G	G/A	A/T	G/T	C/G	A/T
Ped4	SG-4	G/G	A/A	T/T	G/G	G/G	T/T
Ped5	SG-5	G/G	A/A	T/T	G/G	G/G	T/T
Ped6	SG-6	G/G	A/A	T/T	G/G	G/G	T/T
Ped6	SG-6-P1	G/G	A/A	T/T	G/T	G/G	T/T
Ped6	SG-6-P2	G/G	A/A	T/T	G/T	C/G	A/T
Ped7	SG-7	G/G	A/A	T/T	G/G	G/G	T/T
Ped7	SG-7-P1	A/G	G/A	A/T	G/T	G/G	T/T
Ped8	SG-8	G/G	A/A	T/T	G/G	G/G	T/T
Ped8	SG-8-P1	A/G	G/A	A/T	G/T	C/G	A/T
Ped8	SG-8-P2	G/G	A/A	T/T	G/T	C/G	T/T
Ped9	SG-9	G/G	A/A	T/T	G/G	G/G	T/T
Ped9	SG-9-P1	G/G	A/A	T/T	G/T	C/G	A/T
Ped9	SG-9-P2	A/G	G/A	A/T	G/T	C/G	A/T
Ped10	SG-10	G/G	A/A	T/T	G/G	G/G	T/T
Ped10	SG-10-P1	G/G	A/A	T/T	G/T	C/G	A/T
Ped10	SG-10-P2	G/G	A/A	T/T	G/T	G/G	T/T
Ped11	SG-11	A/G	G/A	A/T	G/G	G/G	T/T
Ped12	SG-12	G/G	A/A	T/T	G/G	C/G	T/T
Ped12	SG-12-P1	A/G	G/A	A/T	G/T	C/G	A/T
Ped13	SG-13	G/G	A/A	T/T	G/G	G/G	T/T

S2 table: The genotypes of 5 markers selected surrounding the c.544T>G (p.Thr182Pro) (rs751427686) mutation for the south Indian population (N=87) and north Indian population (N=63).

South Indian population:

	rs10009426	rs6824707	rs2271046	rs35414474	rs17611952
1	A/G	G/A	A/T	G/G	T/T
2	A/A	G/G	A/A	G/G	T/T
3	G/G	A/A	T/T	C/C	A/A
4	A/A	G/G	A/A	G/C	T/A
5	G/G	A/A	T/T	C/C	A/A
6	A/A	G/G	A/A	G/C	T/A
7	A/A	G/G	A/A	C/C	A/A
8	A/G	G/A	A/T	C/C	A/A
9	A/G	G/A	A/T	C/C	A/A
10	G/G	A/A	T/T	G/C	T/A
11	G/G	A/A	T/T	G/C	T/A
12	A/A	G/G	A/A	G/C	T/A
13	A/G	G/A	A/T	G/C	T/A
14	A/G	G/A	A/T	G/C	T/A
15	A/A	G/G	A/A	G/C	T/A
16	A/G	G/A	A/T	C/C	A/A
17	A/G	G/A	A/T	C/C	A/A
18	A/A	G/G	A/A	C/C	A/A
19	A/A	G/G	A/A	C/C	A/A
20	G/G	A/A	T/T	G/C	T/A
21	A/G	G/A	A/T	G/C	T/A
22	A/G	G/A	A/T	G/C	T/A
23	G/G	A/A	T/T	C/C	A/A
24	A/A	G/G	A/A	C/C	A/A
25	A/G	G/A	A/T	G/C	T/A
26	G/G	A/A	T/T	C/C	A/A
27	A/G	G/A	A/T	C/C	A/A
28	G/G	A/A	T/T	G/C	T/A
29	G/G	A/A	T/T	C/C	A/A
30	A/G	G/A	A/T	C/C	A/A
31	A/G	G/A	A/T	C/C	A/A
32	A/G	G/A	A/T	C/C	A/A
33	G/G	A/A	T/T	C/C	A/A
34	G/G	A/A	T/T	G/G	T/T
35	G/G	A/A	T/T	C/C	A/A
36	A/G	G/A	A/T	G/C	T/A
37	G/G	A/A	T/T	C/C	A/A
38	G/G	A/A	T/T	G/C	T/A
39	G/G	A/A	T/T	G/C	T/A
40	G/G	A/A	T/T	G/G	T/T
41	A/G	G/A	A/T	C/C	A/A
42	A/G	G/A	A/T	G/G	T/T
43	A/G	G/A	A/T	C/C	A/A
44	A/G	G/A	A/T	G/C	T/A
45	A/A	G/G	A/A	G/C	T/A
46	A/G	G/A	A/T	C/C	A/A
47	A/G	G/A	A/T	C/C	A/A
48	A/A	G/G	A/A	C/C	A/A
49	A/A	G/G	A/A	C/C	A/A
50	G/G	A/A	T/T	C/C	A/A
51	G/G	A/A	T/T	C/C	A/A
52	A/G	G/A	A/T	C/C	A/A
53	A/G	G/A	A/T	C/C	A/A

54	G/G	A/A	T/T	C/C	A/A
55	A/G	G/A	A/T	C/C	A/A
56	G/G	A/A	T/T	G/C	T/A
57	G/G	A/A	T/T	G/G	T/T
58	A/G	G/A	A/T	C/C	A/A
59	G/G	A/A	T/T	C/C	A/A
60	A/G	G/A	A/T	G/C	T/A
61	A/A	G/G	A/A	G/C	T/A
62	A/G	G/A	A/T	C/C	A/A
63	A/G	G/A	A/T	G/C	T/A
64	A/A	G/G	A/A	C/C	A/A
65	A/G	G/A	A/T	G/C	T/A
66	A/A	G/G	A/A	C/C	A/A
67	A/G	G/A	A/T	C/C	A/A
68	A/A	G/G	A/A	C/C	A/A
69	A/G	G/A	A/T	G/C	T/A
70	A/G	G/A	A/T	C/C	A/A
71	A/G	G/A	A/T	G/C	T/A
72	A/A	G/G	A/A	C/C	A/A
73	A/A	G/G	A/A	G/G	T/T
74	A/G	G/A	A/T	G/C	T/A
75	A/A	G/G	A/A	G/C	T/A
76	A/G	G/A	A/T	C/C	A/A
77	G/G	A/A	T/T	C/C	A/A
78	A/G	G/A	A/T	C/C	A/A
79	G/G	A/A	T/T	G/C	T/A
80	A/G	G/A	A/T	C/C	A/A
81	A/G	G/A	A/T	C/C	A/A
82	A/G	G/A	A/T	G/C	T/A
83	A/G	G/A	A/T	G/C	T/A
84	A/G	G/A	A/T	G/C	T/A
85	G/G	A/A	T/T	C/C	A/A
86	A/G	G/A	A/T	C/C	A/A
87	A/G	G/A	A/T	G/C	T/A

North Indian population:

	rs10009426	rs6824707	rs2271046	rs35414474	rs17611952
1	A/G	G/A	A/T	C/C	A/A
2	A/G	G/A	A/T	G/G	T/T
3	A/G	G/A	A/T	C/C	A/A
4	A/G	G/A	A/T	C/C	A/A
5	A/G	G/A	A/T	C/C	A/A
6	A/G	G/A	A/T	C/C	A/A
7	A/A	G/G	A/A	C/C	A/A
8	A/A	G/G	A/A	C/C	A/A
9	A/G	G/A	A/T	G/G	T/T
10	G/G	A/A	T/T	C/C	A/A
11	G/G	A/A	T/T	G/C	T/A
12	A/A	G/G	A/A	C/C	A/A
13	A/G	G/A	A/T	G/C	T/A
14	G/G	A/A	T/T	G/C	T/A
15	G/G	A/A	T/T	C/C	A/A
16	A/G	G/A	A/T	C/C	A/A
17	A/G	G/A	A/T	C/C	A/A
18	A/G	G/A	A/T	G/C	T/A
19	A/A	G/G	A/A	G/C	T/A
20	G/G	A/A	T/T	C/C	A/A
21	A/G	G/A	A/T	G/C	T/A
22	G/G	A/A	T/T	G/C	T/A
23	G/G	A/A	T/T	C/C	A/A

24	A/A	G/G	A/A	G/C	T/A
25	A/G	G/A	A/T	C/C	A/A
26	A/A	G/G	A/A	G/C	T/A
27	G/G	A/A	T/T	C/C	A/A
28	G/G	A/A	T/T	C/C	A/A
29	A/G	G/A	A/T	G/G	T/T
30	G/G	A/A	T/T	C/C	A/A
31	A/A	G/G	A/A	C/C	A/A
32	A/G	G/A	A/T	G/C	T/A
33	A/G	G/A	A/T	C/C	A/A
34	A/A	G/G	A/A	G/G	T/T
35	A/G	G/A	A/T	G/C	T/A
36	G/G	A/A	T/T	C/C	A/A
37	G/G	A/A	T/T	C/C	A/A
38	A/G	G/A	A/T	G/C	T/A
39	A/A	G/G	A/A	G/C	T/A
40	A/G	G/A	A/T	C/C	A/A
41	A/A	G/G	A/A	C/C	A/A
42	A/G	G/A	A/T	C/C	A/A
43	A/G	G/A	A/T	G/C	T/A
44	G/G	A/A	T/T	C/C	A/A
45	A/G	G/A	A/T	G/C	T/A
46	A/G	G/A	A/T	C/C	A/A
47	A/G	G/A	A/T	C/C	A/A
48	G/G	A/A	T/T	C/C	A/A
49	A/A	G/G	A/A	G/C	T/A
50	A/G	G/A	A/T	G/C	T/A
51	A/A	G/G	A/A	C/C	A/A
52	G/G	A/A	T/T	C/C	A/A
53	A/A	G/G	A/A	C/C	A/A
54	G/G	A/A	T/T	C/C	A/A
55	A/A	G/G	A/A	C/C	A/A
56	A/G	G/A	A/T	G/C	T/A
57	A/G	G/A	A/T	G/C	T/A
58	A/G	G/A	A/T	C/C	A/A
59	G/G	A/A	T/T	G/C	T/A
60	A/G	G/A	A/T	G/G	T/T
61	A/A	G/G	A/A	C/C	A/A
62	G/G	A/A	T/T	C/C	A/A
63	A/A	G/G	A/A	C/C	A/A

S3 table: The frequency of the haplotypes reconstructed from the SNPs rs10009426, rs6824707, rs2271046, rs35414474 and rs17611952 in the background of c. 544T>G mutation in cases, related controls and unrelated south and north Indian population.

Haplotypes	Frequency in cases (N=14)	Frequency in unrelated south Indian control population (N=87)	CHISQ	P-value
H1'	0.9	0.1519	34.03	5.43E-09
H2'	0	0.3825	5.993	0.01436
H3'	0	0.3531	5.301	0.02131
H4'	0.1	0.1124	0.0147	0.9035

Haplotypes	Frequency in cases (N=14)	Frequency in unrelated north Indian control population (N=63)	CHISQ	P-value
H1'	0.9	0.1109	41.09	1.46E-10
H2'	0	0.4049	6.481	0.0109
H3'	0	0.357	5.333	0.02092
H4'	0.1	0.1272	0.06245	0.8027

Haplotypes	Frequency in related controls (N=13)	Frequency in unrelated south Indian control population (N=87)	CHISQ	P-value
H1'	0.4766	0.1504	15.47	8.38E-05
H2'	0.2542	0.3841	1.643	0.1998
H3'	0.09196	0.3515	7.029	0.008022
H4'	0.1003	0.114	0.04241	0.8368
H5'	0.07692	0	13.52	0.000236

Haplotypes	Frequency in related controls (N=13)	Frequency in unrelated north Indian control population (N=63)	CHISQ	P-value
H1'	0.4644	0.1088	19.35	1.09E-05
H2'	0.2663	0.4071	1.807	0.1789
H3'	0.07981	0.3548	7.65	0.005676
H4''	0.1125	0.1293	0.0552	0.8143
H5'	0.07692	0	9.822	0.001725

S4 table: Proportion of loci shares to be zero alleles (Z0), one allele (Z1), or two alleles (Z=2) and PI_HAT values between individual pairs of the same family pedigree.

Individual ID1	Individual ID2	Z0	Z1	Z2	PI_HAT
SG-3-S	SG-3-P1	0.0365	0.8178	0.1457	0.5546
SG-3-S	SG-3	0.0462	0.8522	0.1016	0.5277
SG-3-P1	SG-3	0.2554	0.4159	0.3287	0.5366
SG-4-P1	SG-4-P2	0.8951	0.049	0.056	0.0804
SG-4-P1	SG-4	0.0401	0.8628	0.097	0.5284
SG-4-P2	SG-4	0.0401	0.8805	0.0794	0.5196
SG-6-P1	SG-6-P2	0.7309	0.1414	0.1277	0.1984
SG-6-P1	SG-6	0.1046	0.6902	0.2052	0.5503
SG-6-P2	SG-6	0.0936	0.697	0.2094	0.5579
SG-7-P1	SG-7	0.062	0.7099	0.228	0.583
SG-8-P1	SG-8-P2	0.8748	0	0.1252	0.1252
SG-8-P1	SG-8	0.0632	0.8589	0.0779	0.5073
SG-8-P2	SG-8	0.0535	0.8483	0.0982	0.5223
SG-9-P1	SG-9-P2	1	0	0	0
SG-9-P1	SG-9	0.0426	0.8897	0.0677	0.5126
SG-9-P2	SG-9	0.0401	0.8929	0.067	0.5134
SG-10-P1	SG-10-P2	0.9036	0.042	0.0544	0.0754
SG-10-P1	SG-10	0.0693	0.8734	0.0573	0.494
SG-10-P2	SG-10	0.0645	0.7995	0.1361	0.5358
SG-12-P1	SG-12	0.0511	0.8063	0.1427	0.5458

S5 table: Proportion of loci shares to be zero alleles (Z0), one allele (Z1), or two alleles (Z=2) and PI_HAT values between seemingly unrelated 33 individual pairs from different pedigrees sharing a novel relationship with Z1 and Z2 IBD share.

Family ID1	Individual ID1	Family ID2	Individual ID2	Z0	Z1	Z2	PI_HAT
Ped2	SG-2	Ped10	SG-10-P2	0.9011	0	0.0989	0.0989
Ped3	SG-3-S	Ped4	SG-4-P1	0.9298	0	0.0702	0.0702
Ped3	SG-3-S	Ped4	SG-4-P2	0.9604	0	0.0396	0.0396
Ped3	SG-3-S	Ped4	SG-4	0.9494	0	0.0506	0.0506
Ped3	SG-3-S	Ped8	SG-8-P1	0.9486	0.0092	0.0422	0.0468
Ped3	SG-3-S	Ped9	SG-9	0.945	0	0.055	0.055
Ped3	SG-3-S	Ped10	SG-10-P1	0.9304	0.0335	0.0361	0.0529
Ped3	SG-3-S	Ped10	SG-10-P2	0.9243	0.0108	0.065	0.0703
Ped3	SG-3-S	Ped12	SG-12-P1	0.8708	0.1085	0.0207	0.075
Ped4	SG-4-P1	Ped10	SG-10-P2	0.9421	0	0.0579	0.0579
Ped4	SG-4-P1	Ped12	SG-12-P1	0.9231	0.043	0.0339	0.0554
Ped4	SG-4-P2	Ped6	SG-6-P2	0.932	0	0.068	0.068
Ped4	SG-4-P2	Ped10	SG-10-P1	0.9425	0.0218	0.0357	0.0466
Ped4	SG-4-P2	Ped10	SG-10	0.953	0	0.047	0.047
Ped4	SG-4-P2	Ped12	SG-12-P1	0.8987	0.0665	0.0348	0.068
Ped4	SG-4	Ped6	SG-6-P2	0.9166	0	0.0834	0.0834
Ped4	SG-4	Ped10	SG-10-P1	0.9492	0	0.0508	0.0508
Ped4	SG-4	Ped10	SG-10-P2	0.9373	0	0.0627	0.0627
Ped4	SG-4	Ped12	SG-12-P1	0.889	0.0691	0.0419	0.0764
Ped6	SG-6-P2	Ped10	SG-10-P1	0.9259	0	0.0741	0.0741
Ped6	SG-6-P2	Ped10	SG-10-P2	0.9133	0.0348	0.0519	0.0693
Ped6	SG-6-P2	Ped10	SG-10	0.924	0	0.076	0.076
Ped6	SG-6-P2	Ped12	SG-12-P1	0.9245	0	0.0755	0.0755
Ped8	SG-8-P1	Ped10	SG-10-P2	0.9548	0	0.0452	0.0452
Ped8	SG-8-P1	Ped12	SG-12-P1	0.9583	0.0011	0.0406	0.0411
Ped8	SG-8-P2	Ped10	SG-10-P2	0.9106	0	0.0894	0.0894
Ped8	SG-8-P2	Ped12	SG-12-P1	0.9274	0	0.0726	0.0726
Ped8	SG-8	Ped10	SG-10-P2	0.9229	0	0.0771	0.0771
Ped8	SG-8	Ped12	SG-12-P1	0.9583	0.0072	0.0345	0.0381
Ped9	SG-9-P2	Ped12	SG-12-P1	0.9612	0	0.0388	0.0388
Ped10	SG-10-P1	Ped12	SG-12-P1	0.8951	0.0611	0.0438	0.0743
Ped10	SG-10-P2	Ped12	SG-12-P1	0.8902	0.0473	0.0624	0.0861
Ped10	SG-10	Ped12	SG-12-P1	0.9121	0.0523	0.0356	0.0617