CentreID and PtID (sex)	Molecular genetics (mutation)	Age at onset (years)– symptom(s) at onset	Age at imaging	Severity at imaging	Distribution of weakness
IT1_1 (M)	SGCA [c.229C>T/p.R77C] + [c.739G>A/p.V247M]	13, difficulties in running	34	Not able to run	Proximal
IT1_2 (M)	SGCG [c.342dupT/p.A115CfsX41] + [c.525delT/p.F175LfsX20]	4, difficulties in running	10	Not able to run	Proximal
IT1_3 (F)	SGCG [c.525delT/p.F175LfsX20] + [c.525delT/p.F175LfsX20]	4, difficulties in running	11	Not able to run	Proximal
IT1_4 (F)	SGCG [c.525delT/p.F175LfsX20] + [c.525delT/p.F175LfsX20]	15, difficulties in climbing stairs	50	Non-ambulant since age 42	Generalized
IT1_5 (F)	SGCA [c.229C>T/p.R77C] + [c.850C>T/p.R284C]	3rd decade, difficulties in climbing stairs	50	Not able to run	Proximal
IT1_6 (M)	SGCA [c.308T>C/p.I103T] + [c.480dupC/p.L164TfsX27]	hyperCKemia at age 2, difficulties in climbing stairs at age 5	8	Runs with difficulties	Mild proximal
IT1_7 (F)	SGCG [c.525delT/p.F175LfsX20] + [c.525delT/p.F175LfsX20]	hyperCKemia at age 2, toe walking at age 6	9	Not able to run	Generalized
IT1_8 (F)	SGCA [c.308T>C/p.I103T] + [c.850C>T/p.R284C]	12, myalgias and exercise intolerance	29	Not able to run	Proximal
IT1_9 (M)*	SGCA [c.229C>T/p.R77C] + [c.739G>A/p.V247M]	6, asymptomatic hyperCKemia	9	Asymptomatic	None
IT1_10 (F)*	SGCA [c.229C>T/p.R77C] + [c.739G>A/p.V247M]	10, asymptomatic hyperCKemia	10	Asymptomatic	None
IT1_11 (F)	SGCA [c.850C>T/p.R284C] + [c.850C>T/p.R284C]	45, hyperCKemia	54	Not able to run	Proximal
IT2_1 (F)	SGCA [c.739G>A/p.V247M] + [c.739G>A/p.V247M]	11, hyperCKemia, post-exercise muscle pain and myoglobinuria	40	Not able to run	Proximal
IT2_2 (M)	SGCA [c.850C>T/p.R284C] + [c.850C>T/p.R284C]	10, hyperCKemia, post-exercise muscle pain	36	Not able to run	Proximal
IT3_1 (F)	SGCA [c.229C>T/p.R77C] + ?	4, difficulties in running	4	Runs with difficulties	Proximal
IT3_2 (M)	SGCA [c.89del/p.P30HfsX17] + [c.89del/p.P30HfsX17]	4,difficulties in running	7	Not able to run	Proximal
IT3_3 (M)	SGCA [c.409G>A/p.E137K] + [c.739G>A/p.V247M]	8, difficulties in climbing stairs	24	Not able to run	Proximal
IT3_4 (F)#	SGCB [c.265G>A/p.V89M] + [c.265G>A/p.V89M]	9, difficulties in running	20	Not able to run	Proximal
IT3_5 (F)#	SGCB [c.265G>A/p.V89M] + [c.265G>A/p.V89M]	9, difficulties in running and climbing stairs	15	Not able to run	Proximal
IT3_6 (M)	SGCA [c. 229C>T/p.R77C] + [c.1005C>A/p.C335X]	9, difficulties in running and climbing stairs	21	Not able to run	Proximal
IT3_7 (M)	SGCG [c.801_802del/p.P268RfsX48] + [c.385+2T>C/p.?]	7, difficulties in running and climbing stairs	29	Non-ambulant since age 22	Generalized
IT3_8 (M)	SGCD [c.353_357del/p.T119SfsX17] + [c.353_357del/p.T119SfsX17]	4, difficulties in running and climbing stairs	12	Non-ambulant since age 11	Generalized
IT3_9 (M)	SGCA [c.92T>C/p.L31P] + [c.92T>C/p.L31P]	3, toe walking	11	Not able to run	Proximal
IT4_1 (M)^	SGCB [c.377_384dup/p.G129QfsX2] + [c.377_384dup/p.G129QfsX2]	3, walking difficulties	11	Non-ambulant since age 10	Proximal
IT4_2 (F)	SGCA [c.739G>A/p.V247M] + [c.739G>A/p.V247M]	9 , myalgia, high CK	16	Asymptomatic	None

IT4_3 (M)	SGCG [c.525delT/p.F175LfsX20] + [c.525delT/p.F175LfsX20]	7, high CK, walking difficulties	18	Ambulant with support	Proximal
IT4_4 (M)^	SGCB [c.377_384dup/p.G129QfsX2] + [c.377_384dup/p.G129QfsX2]	2, walking difficulties	8	Not able to run	Proximal
IT4_5 (F)	SGCA [c.271G>A/p.G91S] + [c.292 C>T/p.R98C]	3, proximal weakness	10	Not able to run	Proximal
IT5_1 (M)	SGCB [c.377_384dup/T126RfsX5] + [c.377_384dup/T126RfsX5]	4, difficulties in running, early fatigue	9	Runs with difficulties	Proximal
IT5_2 (F)	SGCG [c.87dup/p.G30WfsX30] + [c.87dup/p.G30WfsX30]	6, proximal lower limb weakness	32	Non ambulant since age 14	Generalized
IT5_3 (F)	SGCA [c.739G>A/p.Val247Met] + [c.739G>A/p.Val247Met]	38, exertional pain	49	Able to run	Mild proximal
IT5_4 (F)	SGCG [c.371G>A/p.G124D] + [c.371G>A/p.G124D]	6, myalgias	16	Able to run	Mild proximal
IT5_5 (F)	SGCB [c.552T>G/p.Y184X] + [c.552T>G/p.Y184X]	5, running difficulaties, proximal lower limb weakness	14	Non ambulant since age 11	Generalized
IT5_6 (F)[§]	SGCA [c.850C>T/p.R284C] + [c.850C>T/p.R284C]	20, walking difficulties	59	Ambulant with support	Proximal
IT5_7 (M)[§]	SGCA [c.850C>T/p.R284C] + [c.850C>T/p.R284C]	Asymptomatic	57	Asymptomatic	none
IT5_8 (M)	SGCA [c.541C>T/p.R181C]+ [c.86dupA/p.H29QfsX15]	9,asymptomatic hyperCKemia	26	asymptomatic	none
ES1_1 (M)**	SGCA [c.307A>T/p.I103F] + [c.700G>A/p.D234N]	6, difficulties in running	36	Ambulant with support	Proximal
ES1_2 (F)**	SGCA [c.307A>T/p.I103F] + [c.700G>A/p.D234N]	8, pain during exercise	34	Ambulant with support	Proximal
ES1_3 (F)	SGCA [c.724G>T/p.V242F] + [c.850C>T/p.R284C]	22, muscle pain and weakness	27	Not able to run	proximal
ES1_4 (F)[##]	SGCA [c.585-31_585-24del/p.?] + [c.585-31_585-24del/p.?]	Second decade, proximal weakness	49	Not able to run	Proximal
ES1_5 (F)[##]	SGCA [c.585-31_585-24del/p.?] + [c.585-31_585-24del/p.?]	40, proximal and axial weakness	47	Not able to run	Proximal
ES1_6 (F)	SGCA [c.488dupG/p.L164TfsX27] + [c.661C>T/p.R221C]	Second decade, proximal lower limb weakness	51	Not able to run	Proximal
ES2_1 (M)	SGCB [c.949_956del/p.T317KfsX49] + ?	11, exercise intolerance and myoglobinuria	30	Not able to run	Proximal
ES2_2 (F)	SGCG [c.579-?_702+?del (ex07del)] + [c.579-?_702+?del (ex07del)]	8, myalgia, proximal weakness	11	Not able to run	Proximal
ES2_3 (F)	SGCG [c.525delT/p.F175LfsX20] + [c.525delT/p.F175LfsX20]	4, hyperckemia	4	Able to run	Mild proximal
EN1_1(M)	SGCG [c.787G>A/p.E263K] + [c.787G>A/p.E263K]	8, waddling gait	11	Not able to run	Proximal
EN1_2 (M)	SGCG [c.787G>A/p.E263K] + [c.787G>A/p.E263K]	6, waddling gait, frequent falls	17	Non-ambulant since age 13	Proximal
EN2_1 (M)	SGCB [c.82_86del/p.E28KfsX5] + [c.82_86del/p.E28KfsX5]	2 , walking difficulties	5	Runs with difficulties	Proximal
EN2_2 (M)	SGCG [c.158T>C/p.L53P] +?	9, intermittent toe walking	14	Runs with difficulties	Proximal

EN2_3 (F)	SGCA [c.229C>T/p.R77C] + [c.229C>T/p.R77C]	First decade, walking difficulties	19	Non-ambulant since age 14	Proximal
DE1_1(M)	SGCA [c.100C>T/p.R34C] + [c.229C>T/p.R77C]	2, running difficulties	11	Not able to run	Proximal
DE1_2 (F)	SGCA [c.229C>T/p.R77C] + [c.229C>T/p.R77C]	4, difficulties in running	21	Ambulant with support	Proximal
NE1_1 (M)^^	SGCG [c.505+2T>C/p.?]+ [c.505+2T>C/p.?]	6, walking difficulties	22	Non-ambulant since age 12	Generalized
NE1_2 (M)^^	SGCG [c.505+2T>C/p.?] + [c.505+2T>C/p.?]	7, walking difficulties	18	Non-ambulant since age 13	Generalized
FR1_1 (F)	SGCB [c.290G>A/p.C97Y] + [c.654_655del/p.K219SfsX2]	6, walking difficulties	16	Non-ambulant since age 8	Generalized
FR1_2 (M)	SGCA [c.229C>T/p.R77C] + [c.403C>T/p.Q135X]	4, difficulties climbing stairs	10	Runs with difficulties	Proximal
CZ1_1 (F)	SGCG [c.87dup/p.G30WfsX30] + [c.87dup/p.G30WfsX30]	7, walking difficulties	8	Not able to run	Proximal
CZ1_2 (F)	SGCA [c.739G>A/p.V247M] + [c.402C>G/p.Y134X]	9, hyperCKemia	9	Asymptomatic	None
FI1_1 (F)	SGCA [c.229C>T/p.R77C] + [c.229C>T/p.R77C]	7, proximal weakness, walking difficulties	21	Ambulant with support	Proximal
FI1_2 (M)	SGCA [c.229C>T/p.R77C] + [c.229C>T/p.R77C]	6, difficulties in running and climbing stairs	21	Non-ambulant since age 19	Proximal
FI1_3 (F)	SGCA [c.229C>T/p.R77C]+ [c.229C>T/p.R77C]	4, clumsiness, Gower's sign	35	Non-ambulant since age 21	Generalized
FI1_4 (F)	SGCA [c.229C>T/p.R77C] + [c.229C>T/p.R77C]	5, running difficulties	33	Non-ambulant since age 25	Generalized
FI1_5 (F)	SGCA [c.229C>T/p.R77C] + [c.229C>T/p.R77C]	5, walking difficulties	13	Not able to run	Proximal
US1_1 (F)[§§]	SGCB [c.355A>T/p.I119F] + [c.355A>T/p.I119F]	Toe walking at age 3, walking difficulties at age 5	9	Not able to run	Proximal
US1_2 (M)[§§]	SGCB [c.355A>T/p.I119F] + [c.355A>T/p.I119F]	3, hyperCKemia, difficulties in running	6	Not able to run	Proximal
US1_3 (M)	SGCG [g.23,860,250_23,874,950del (del of 14.7kb including exon 6)] + [g. 23,511,150_24,910,750del (del of 1.4Mb including the entire SGCG gene + 8 adjacent genes)]	2, difficulties in jumping	6	Runs with difficulties	Proximal
RU1_1 (M)	SGCA [c.229C>T/p.R77C] + [c.271G>A/p.G91S]	2, walking difficulties and frequent fallings	6	Runs with difficulties	Proximal
DK1_1 (M)	SGCG [c.205G>C/p.G69R] + [c.385G>A/p.G129S]	10, walking difficulties and muscle pain	41	Ambulant with support	Proximal
DK1_2 (M)	SGCB [c.341C>T/p.S114F] + [c.543C>G/p.S181R]	10, walking and running difficulties	36	Not able to run	Proximal
DK1_3 (M)	SGCA [c.229C>T/p.R77C] + [c.739G>A/p.V247M]	12, walking difficulties	59	Not able to run	Proximal

Supplementary Table 1: summary of clinical and genetic features of all the patients
Legend: *, [#], ^, [§], **, [##], ^^, [§§]: siblings
SGCA : NM_000023.2; *SGCG* : NM_000231.2; *SGCB* : NM_000232.4; *SGCD* : NM_000337.5