

Supplementary Table 2

## Clinical features

Patient	Age/Gender	Disease duration (years)	UE Muscle strength (I/D)	LE Muscle strength (P/I/D)	Reflexes (UE/Knee/Ankle)	UE Sensory involvement (T/Pa/V/Po)	LE Sensory involvement (T/Pa/V/Po)	Atrophy	Deformities	Mobility
A 4.2	44/F	39	5/5	5/5/5	2/2/2	2/2/2/2	2/2/2/2	None	Cavus foot	Ambulatory
A 5.1	15/M	12	5/5	5/4/5	2/2/2	2/2/2/2	2/2/2/2	None	Cavus foot + claw	Ambulatory
A 5.2	18/F	16	5/5	5/4/5	2/2/2	2/2/2/2	2/2/2/2	None	Cavus foot	Ambulatory
A 5.5	22/F	08	5/5	5/5/5	2/2/2	2/2/2/2	2/2/2/2	Distal LE	Cavus foot	Ambulatory
B 5.88	37/M	23	5/5	5/4/3	2/3/2	2/2/2/2	2/2/0/0	Distal LE	Cavus foot + claw	Ambulatory
B 5.93	40/F	38	5/5	5/4/3	3/3/2	2/2/2/2	2/2/0/1	Distal LE	Cavus foot + claw	Ambulatory
B 6.28	19/F	17	5/5	5/4/4	2/2/2	2/2/2/2	2/2/1/1	Distal LE	Cavus foot	Ambulatory
B 6.29	22/M	20	5/5	5/4/3	3/3/0	2/2/2/2	2/2/2/2	Distal LE	Cavus foot	Ambulatory
BC 4.2	59/F	33	5/4	5/4/3	2/1/0	2/2/2/2	2/2/2/2	Distal UE + LE	Cavus foot	Cane
BC 5.1	26/M	24	5/5	4/4/4	3/3/2	2/2/2/2	2/2/2/2	Distal UE + LE	Cavus foot + claw	Ambulatory
BC 5.5	30/F	15	5/5	4/4/4	3/2/0	2/2/2/2	2/2/0/0	None	Cavus foot	Ambulatory
BC 5.8	32/M	31	5/4	4/3/3	3/0/0	2/2/1/1	2/2/0/1	Distal UE + LE	Cavus foot + claw	Wheelchair
C 3.6	80/M	60	3/3	3/1/0	2/0/0	2/2/1/1	2/2/0/0	Distal UE + LE	None	Wheelchair
C 5.68	38/F	28	5/5	5/4/4	2/2/0	2/2/2/2	2/2/1/2	None	Cavus foot	Ambulatory
C 5.79	36/M	26	5/5	5/4/3	2/1/0	2/2/2/2	2/2/1/2	Distal LE	Cavus foot	Ambulatory
C 5.87	44/M	34	5/5	5/4/3	3/3/0	2/2/2/2	2/2/1/1	Distal LE	Cavus foot	Ambulatory
D 4.8	61/F	43	4/4	3/2/2	3/0/0	2/2/1/1	2/2/0/0	Distal UE + LE	Cavus foot + varus	Wheelchair
D 4.28	69/F	51	5/5	4/3/1	2/0/0	2/2/2/2	2/2/0/1	Distal UE + LE	Cavus foot + varus	Walker
D 5.97	32/M	21	5/5	5/4/3	2/2/0	2/2/2/2	2/2/1/2	Distal LE	Cavus foot	Ambulatory
D 5.100	35/F	20	5/5	5/4/4	2/2/0	2/2/2/2	2/2/0/2	None	Cavus foot + claw	Ambulatory
D 5.106	40/F	05	5/5	5/4/4	2/0/0	2/2/2/2	2/2/0/2	None	Cavus foot + claw	Ambulatory
D 5.109	45/M	38	5/5	3/2/1	0/0/0	2/2/2/2	2/2/0/0	Distal LE	Cavus Foot + varus	Wheelchair
D 5.111	48/M	34	5/5	4/4/3	2/3/0	2/2/2/2	2/2/0/2	Distal LE	Cavus foot + claw	Ambulatory
D 5.131	39/F	13	5/5	4/5/5	2/0/0	2/2/2/2	2/2/1/0	Distal LE	Cavus foot	Ambulatory
D 5.136	43/F	18	5/5	4/4/4	2/1/0	2/2/2/2	2/2/1/1	None	Cavus foot	Ambulatory
D 6.61	09/F	07	5/5	5/5/5	2/3/2	2/2/2/2	2/2/2/2	None	None	Ambulatory
D 6.146	16/F	02	5/5	5/5/4	2/2/2	2/2/2/2	2/2/2/2	Distal LE	Cavus foot	Ambulatory
D 6.147	20/F	19	5/5	5/4/3	2/3/2	2/2/2/2	2/2/2/0	Distal LE	None	Ambulatory
D 6.177	36/M	34	5/5	5/5/5	2/2/2	2/2/2/2	2/2/0/1	None	Cavus foot	Ambulatory
D 6.178	38/F	36	5/5	4/3/2	2/4/4	2/2/2/2	2/2/0/1	Distal UE + LE	Cavus foot	Walker
E 4.39	75/M	20	5/5	5/4/4	1/2/1	2/2/2/2	2/2/2/2	Distal LE	None	Cane
E 5.93	47/F	05	5/5	5/5/4	2/2/1	2/2/2/2	2/2/1/1	None	Cavus foot	Ambulatory
E 6.70	9/F	05	5/5	5/5/4	2/2/1	2/2/2/2	2/2/1/1	Distal LE	Cavus foot	Ambulatory
EF1 4.3	52 F	50	5/5	5/3/2	0/0/0	2/2/1/1	2/2/1/1	Distal UE + LE	Cavus foot + varus	Cane
EF1 4.5	57/F	47	5/5	4/3/3	3/3/0	2/2/2/2	2/2/2/2	Distal LE	Cavus foot + varus	Cane
EF1 4.12	70/M	30	5/5	5/4/3	2/1/0	2/2/2/2	2/2/0/0	Distal LE	None	Cane
EF1 4.14	72/M	44	4/4	3/2/1	0/0/0	2/2/1/1	2/2/1/1	Distal UE + LE	Cavus foot	Wheelchair
EF1 5.46	46/F	39	5/5	5/4/4	2/2/2	2/2/2/2	2/2/0/0	Distal LE	Cavus foot	Ambulatory
EF1 5.53	51/M	40	5/5	5/5/5	2/2/0	2/2/2/2	2/2/2/2	None	None	Ambulatory
F 5.243	70/F	02	5/5	5/5/5	2/2/2	2/2/2/2	2/2/2/1	None	Cavus foot	Ambulatory
F 6.357	30/M	22	5/5	5/4/4	3/4/4	2/2/2/2	2/2/2/1	Distal LE	None	Ambulatory
F 6.363	35/M	18	5/4	5/4/4	2/2/1	2/2/2/2	2/2/1/1	None	Cavus foot	Ambulatory
F 6.370	49/F	10	5/5	5/5/5	2/3/1	2/2/2/2	2/2/2/2	None	Cavus foot	Ambulatory
F 6.372	52/M	37	5/4	5/4/3	3/4/4	2/2/2/2	2/2/1/1	Distal UE + LE	Cavus foot	Cane

UE, upper extremity; LE, lower extremity; P, proximal (knee extensor or flexor); I, intermediate (hand extensor or flexor (UE); foot extensor or flexor (LE)); D, distal (intrinsic hand muscles (UE), intrinsic foot muscles (LE)). Motor scale: 5, normal; 4, mild weakness; 3, ability to lift against gravity; 2, not able to lift against gravity, but movement visible; 1, no movement but tendon contraction visible; 0, complete paralysis. Reflexes: 4, clonus; 3 increased; 2, normal; 1, reduced; 0, absent; Sensory involvement/sense of vibration or position: T, touch; Pa, pain; V, vibration; Po, position; 2, normal; 1, mildly reduced; distally to wrist level (UE) or malleoli level (LE), 0, severely reduced; distally to elbow level (UE) or knee level (LE). Deformities: claw, claw toes.