

Supplemental Table 2

Pedigree Designation	II-5	III-2	IV-1	IV-2
Fibroblast Designation	N/A	Patient 1	Patient 2	Patient 3
Age at last assessment	N/A	58	23	24
Sex	F	F	M	F
Onset age (years)		9	<1 (hearing loss), 10 (foot weakness)	4 (hearing loss), 10 (foot weakness)
Age deceased	86	N/A	N/A	N/A
Initial symptom	N/A	Tripping	Hearing loss	Hearing loss
Other medical history	N/A	Diabetes type II Intrinsic hand muscles, anterior/posterior calf and intrinsic foot muscles; pes cavus present	Mild language delay	Attention-deficit disorder
Atrophy	N/A	Hyporeflexia arms, areflexia in legs	Atrophy calves and feet, mild.	Intrinsic hand muscles, calf muscles and intrinsic foot muscles
Reflexes			Absent Achilles reflex	Reduced in arms and Achilles tendon
Muscle power (MRC scale)				
Shoulder abduction	N/A	5	5	5
Elbow flexion	N/A	5	5	5
Elbow extension	N/A	4-	5	5
Wrist extension	N/A	4-	5	4+
Wrist flexion	N/A	4-	5	4+
Finger extension	N/A	3	5	4
Finger flexion	N/A	3	5	4
Finger abduction	N/A	3	5-	4
Hip flexion	N/A	5	5	5
Knee extension	N/A	5	5	5
Knee flexion	N/A	5	5	5
Ankle dorsiflexion	N/A	1	4	3
Ankle plantarflexion	N/A	3	5-	4

Sensory symptoms	N/A	Numbness, dysesthesiae below knees	-	-
Sensory exam	N/A	Reduced cold sensation below knees; normal vibration sense	Normal	Normal
Sensorineural hearing loss	+	+	+++	++
Dysarthria	N/A	-	+	-
Pulmonary function tests	N/A	Normal	Normal	Normal
Echocardiogram	N/A	N/A	N/A	Normal
				Fatty infiltration of gastrocnemius, soleus and extensor digitorum muscles. Asymmetric/mild involvement of left anterior compartment muscles in lower legs.
Muscle MRI	N/A	Lipomatous atrophy of calf musculature	N/A	
Neurophysiology	N/A	Consistent with axonal polyneuropathy	N/A	N/A