

eTable 1 Clinical and laboratory data for affected individuals reported with R159H variant.

Family	Age/ (Age deceased) (years)	Sex	Myopathy	FTD	PDB	ALS	Age of Onset, Myopathy (years)	Age of Onset, FTD (years)	EMG&NCS	Muscle Biopsy
Greek Family ²⁵	(62)	M	+	+	-	-	45	61	Low amplitude of peroneal and tibial nerves	Fibroadipose tissue replacement
	(63)	F	-	+	-	-	N/A	63	N/A	N/A
	(53)	M	-	-	-	+	N/A	N/A	N/A	N/A
	(59)	M	-	+	-	-	N/A	55	N/A	N/A
Netherland Family ²⁴	(61)	F	+	-	-	+	59	N/A	N/A	N/A
	N/A	M	N/A	+	N/A	N/A	N/A	N/A	N/A	N/A
	N/A	M	N/A	+	N/A	N/A	N/A	N/A	N/A	N/A
	N/A	M	N/A	+	N/A	N/A	N/A	N/A	N/A	N/A
Spanish Family ²⁶	(68)	M	+	+	-	-	N/A	64	N/A	N/A
	(73)	F	-	+	-	-	N/A	65	N/A	N/A
Austrian Family ^{11,27}	76	F	+	-	+	-	76	N/A	N/A	N/A
	71	F	+	-	+	-	63	N/A	brief and small amplitude muscle action potentials	N/A
	66	F	+	+	+	-	62	64	N/A	N/A
	60	M	+	-	+	-	49	N/A		Atrophic cells. Rimmed vacuoles
Belgian Family#1 ¹⁸	36	M	+	-	+	-	29	N/A	Neurogenic and myogenic features	Neurogenic necrotic and regenerating fibers with rimmed vacuoles and endomysial inflammatory infiltrates.
	66	M	+	-	-	-	58	N/A	Myopathic features	Rimmed vacuoles.
Belgian Family#2 ²⁷	(67)	F	-	+	-	-	N/A	N/A	N/A	N/A
	(68)	M	-	+	-	-	N/A	N/A	N/A	N/A
	(71)	F	-	+	-	-	N/A	63	N/A	N/A
	(68)	M	-	+	-	-	N/A	62	N/A	N/A
	(68)	M	-	+	-	-	N/A	60	N/A	N/A
	(72)	F	-	+	-	-	N/A	64	N/A	N/A
Belgian Family#3 ²⁷	57	F	-	+	-	-	N/A	45	N/A	N/A
	57	F	-	+	-	-	N/A	44	N/A	N/A
	57	M	-	+	+	-	N/A	49	N/A	N/A
	54	M	-	-	+	-	N/A	N/A	N/A	N/A
Mean/total	60.1/ (66.5)	N=26 M/F 15/11	39%	70%	33%	9%	55.1	58.4		

Abbreviations: PDB Paget's disease of bone, FTD frontotemporal dementia, EMG electromyography, NCS nerve conduction study, ALS amyotrophic lateral sclerosis. N/A=Not available, N=Number, M=Male, F=Female