Figure S1. Validation of GGC repeat expansion and intranuclear inclusions in NIID-affected individuals. (A-B) Representative electropherogram of the RP-PCR assay (left) and the GC-PCR assay (right) revealed abnormal expanded repeats in NIID-affected case subjects (A), and normal repeats in control subjects (B). (C-D) Representative skin biopsy sample showed positive p62-staining (C) and ubiquitin-staining (D) intranuclear inclusions (arrows) in most NIID-affected case subjects. Scale bars, 10μm.(E) Representative electron microscopy imaging displayed intranuclear inclusions (arrow). Scale bars, 1μm.

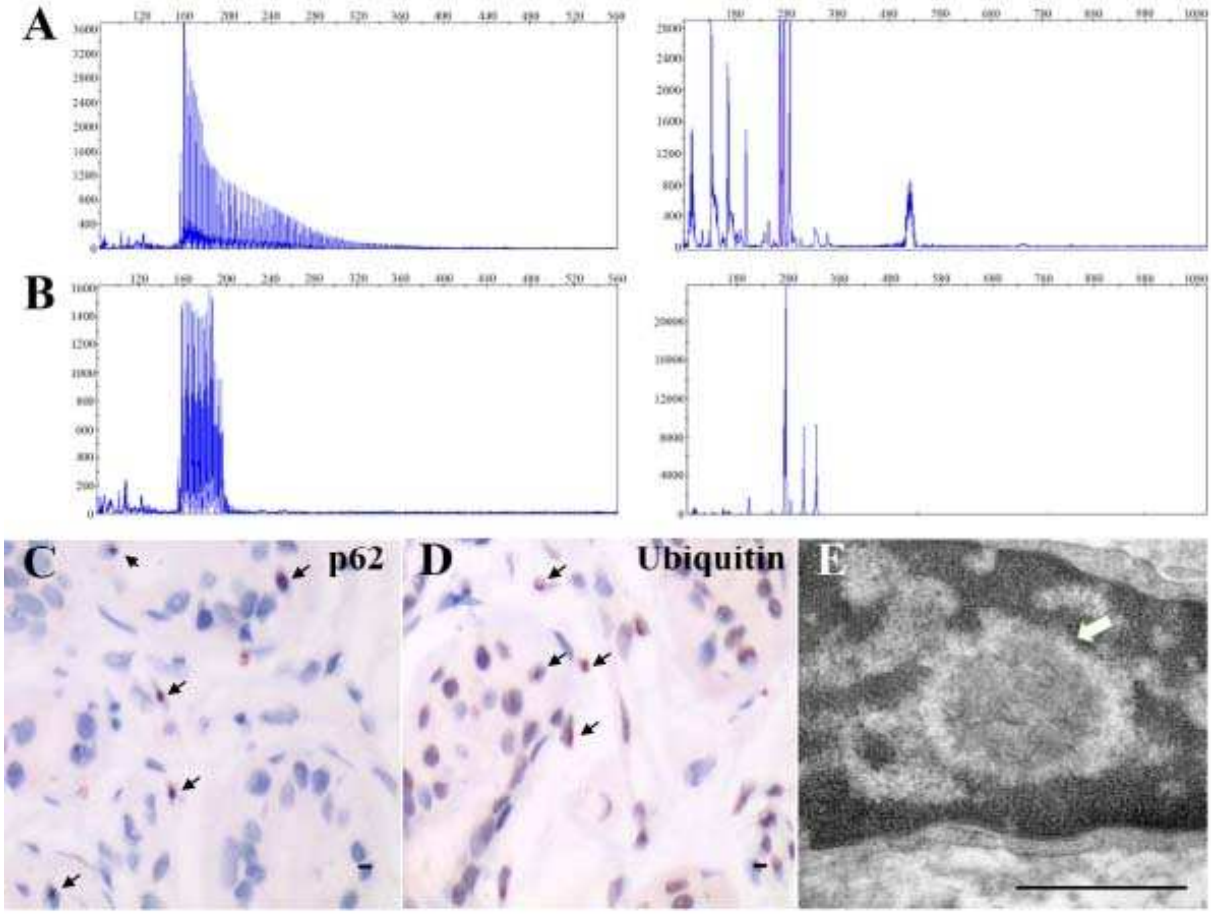

Table S1. Initial manifestations in different clinical phenotypes of NIID.

Initial symptoms	Total	Clinical Phenotype			
		Dementia-dominant type (n=94)	Movement disorder -dominant type (n=63)	Muscle weakness-dominant type (n=29)	Paroxysmal symptom-dominant type (n=61)
Muscle weakness	10.1%	0.0%	0.0%	86.2%	0.0%
Cognitive impairment	23.1%	60.6%	0.0%	0.0%	0.0%
Movement disorders	22.3%	0.0%	87.3%	0.0%	0.0%
Paroxysmal symptoms	32.8%	25.5%	4.8%	6.9%	85.2%
Autonomic dysfunctions	11.7%	13.8%	7.9%	6.9%	14.8%

Table S2. Neurophysiological examination of the muscle weakness-dominant phenotype.

Case	Sex	Age at evaluation	Ulnar motor		Median motor		Tibial motor		Peroneal motor		Ulnar sensory		Median sensory		Sural sensory		Needle EMG/ report
			Amp, mV (>7)	CV, m/s (>50)	Amp, mV (>7)	CV, m/s (>50)	Amp, mV (>7)	CV, m/s (>40)	Amp, mV (>3)	CV, m/s (>40)	Amp, μV (>7)	CV, m/s (>45)	Amp, μV (>7)	CV, m/s (>45)	Amp, μV (M>8; F>6)	CV, m/s (>40)	
M-1	M	69	13.0	37.8	11.9	40.7	8.0	31.2	4.5	34.8	NR	NR	5.4	37.2	7.9	34.7	Neurogenic damage/ Mixed demyelinating and axonal sensorimotor neuropathy
M-2	F	65	4.4	46.2	3.0	44.4	1.8	36.5	1.1	38.6	ND	ND	12.6	37.0	1.4	39.8	ND/ Sensorimotor neuropathy with axonal damage predominant
M-3	M	70	7.3	36.1	7.2	40.5	NR	NR	NR	NR	11.6	38.0	10.1	48.9	NR	NR	Neurogenic damage/ Mixed demyelinating and axonal sensorimotor neuropathy
M-4	M	51	12.1	48.2	11.4	52.9	9.5	35.1	NR	NR	8.3	49.6	10.7	51.1	12.2	42.6	Neurogenic damage/ Mixed demyelinating and axonal motor neuropathy
M-5	M	61	1.3	42.5	0.6	26.4	4.0	29.4	NR	NR	NR	NR	5.2	41.3	6.2	34.6	Neurogenic damage/ Mixed demyelinating and axonal sensorimotor neuropathy
M-7	M	54	9.5	45.4	4.9	51.6	9.5	33.7	0.9	37.2	NR	NR	3.7	49.1	NR	NR	Neurogenic damage/ Mixed demyelinating and axonal sensorimotor neuropathy
M-11	M	22	5.3	36.7	4.0	36.3	2.4	29.0	1.4	28.3	10.0	41.4	9.4	34.1	4.3	32.3	Neurogenic damage/ Sensorimotor neuropathy with axonal damage predominant
M-12	M	23	12.8	46.2	17.3	47.5	15.6	38.7	8.7	39.0	16.0	55.6	44.3	61.9	27.1	57.0	Neurogenic damage/ Mixed demyelinating and axonal motor neuropathy
M-13	M	32	7.3	47.2	14.2	47.7	2.4	39.4	1.5	34.2	4.6	47.4	6.2	54.5	3.5	40.7	Neurogenic damage/ Sensorimotor neuropathy with axonal damage predominant
M-15	M	34	15.6	45.6	18.3	46.1	15.3	34.7	14.1	36.2	6.8	41.4	12.5	48.3	9.3	37.5	Neurogenic damage/ Mixed demyelinating and axonal sensorimotor neuropathy
M-16	F	48	5.3	50.0	6.0	50.0	NR	NR	NR	NR	6.4	42.3	5.7	43.3	NR	NR	Neurogenic damage/ Mixed demyelinating and axonal sensorimotor neuropathy
M-18	M	43	6.4	46.8	3.4	44.6	1.8	39.6	6.8	39.5	9.6	53.6	11.6	55.7	NR	NR	Neurogenic damage/ Mixed demyelinating and axonal sensorimotor neuropathy
M-19	M	41	3.4	41.8	10.4	41.1	NR	NR	1.3	36.3	9.3	59.7	0.2	39.2	2.1	40.1	Neurogenic damage/ Mixed demyelinating and axonal sensorimotor neuropathy
M-20	M	47	9.1	36.1	7.1	41.1	1.0	28.2	NR	NR	2.8	35.5	3.5	43.8	7.9	32.3	Neurogenic damage/ Mixed demyelinating and axonal sensorimotor neuropathy

M-21	M	49	8.7	37.8	8.7	38.1	NR	NR	0.2	21.0	7.3	36.2	15.1	37.4	NR	NR	Neurogenic damage/ Mixed demyelinating and axonal sensorimotor neuropathy
M-22	M	48	9.7	50.0	7.1	39.7	NR	NR	1.4	34.7	3.3	39.4	7.1	34.9	1.9	35.4	Neurogenic damage/ Mixed demyelinating and axonal sensorimotor neuropathy
M-25	M	72	13.1	60.2	12.2	60.8	21.1	43.2	5.5	41.0	7.5	50.0	11.0	51.2	8.7	53.3	Neurogenic damage/ Pure axonal motor neuropathy
M-26	M	53	4.0	35.9	0.3	23.1	1.0	27.2	NR	NR	3.7	29.7	7.3	31.9	NR	NR	Neurogenic damage/ Mixed demyelinating and axonal sensorimotor neuropathy

Amp=amplitude; CV=conduction velocity; F=female; M=male; ND=not done; NR=not response.

Table S3. Spearman's rho coefficients for correlations between GGC repeat sizes and clinical symptoms. $^{*}P < 0.05$ were considered as being statistically significant.

	GGC repeat sizes
Cognitive impairment	-0.072 (p=0.318)
Abnormal behavior	-0.037 (p=0.612)
Muscle weakness	0.142* (p<0.05)
Bulbar paralysis	0.156* (p<0.05)
Limb weakness	0.132 (p=0.067)
Sensory disturbance	0.044 (p=0.543)
Movement disorders	-0.047 (p=0.518)
Tremor	0.006 (p=0.934)
Rigidity	-0.172* (p<0.05)
Bradykinesia	-0.115 (p=0.109)
Ataxia	0.003 (p=0.966)
Paroxysmal symptom	-0.160* (p<0.05)
Disturbance of consciousness	-0.191* (p<0.05)
Encephalitic episodes	-0.133 (p=0.064)
Stroke-like episodes	-0.132 (p=0.067)
Generalized convulsions	-0.081 (p=0.259)
Chronic headache	-0.022 (p=0.757)
Autonomic dysfunction	-0.041 (p=0.567)
Bladder dysfunction	-0.081 (p=0.263)
Miosis	-0.082 (p=0.258)
Orthostatic hypotension	0.008 (p=0.917)
Emesis	-0.006 (p=0.929)
Visual loss	-0.027 (p=0.709)

Table S4. Comparison analysis of GGC repeat sizes between patients with different initial symptoms. *P < 0.05 were considered as being statistically significant.

	Muscle weakness	Cognitive impairment	Movement disorders	Paroxysmal symptoms	Autonomic dysfunctions	*p*
GGC repeat sizes, median (IQR)	143 (106.00-235.00)	117 (103.00-135.00)	124 (100.50-142.00)	120 (104.00-138.75)	122 (102.00-189.00)	0.226

Table S5. Spearman's rho coefficients for correlations between GGC repeat sizes and age at onset, scores of MMSE, MoCA, FAB, and NPI. *P < 0.05 were considered as being statistically significant.

	Age at onset	MMSE	MoCA	FAB	NPI
GGC repeat sizes	-0.196* (p<0.05)	0.004 (p=0.960)	0.013 (p=0.900)	0.010 (p=0.930)	0.059 (p=0.546)