

Supplementary Table 1. Clinical features of the patients

	Proband	Proband's sister
Sex	Male	Female
Onset age	Early teens	Late teens
Initial symptoms	Visual disturbance and delayed puberty	Gait disturbance
Key clinical features	Cerebellar ataxia, hypogonadotropic hypogonadism, and chorioretinal dystrophy	Cerebellar ataxia, hypogonadotropic hypogonadism, and chorioretinal dystrophy
Other neurological symptoms	Dysarthria, swallowing difficulty, gaze-evoked nystagmus, and peripheral neuropathy	Dysarthria and alopecia
Endocrinological assessments		
Luteinizing hormone (1.5–9.3 mIU/mL)	0.51	0.303
Follicle-stimulating hormone (males: 1.5–12.4 mIU/mL, follicular phase: 3.5–12.5 mIU/mL, luteal phase: 1.7–7.7 mIU/mL, postmenopause: 25.8–134.8 mIU/mL, menopause: 25.8–134.8 mIU/mL)	0.86	0.394
Estrogen total (pg/mL)	486.0	-
Estradiol (pg/mL)	-	18.66
Testosterone (2.8–8.0 ng/mL)	0.23	-
Prolactin (males: 4.1–18.4 ng/mL, females: 4.79–23.3 ng/mL)	8.87	7.73
Thyroid-stimulating hormone (0.27–4.2 mIU/mL)	6.0	4.45
Free thyroxine (0.93–1.71 ng/dL)	0.98	1.08
Growth hormone (ng/mL)	-	3.19
Cortisol (2–28 mg/dL)	-	7.91
ACTH (7–10 a.m.: 7.2–63.3 pg/mL)	-	36.3
Somatomedin-C (155.3–667.1 ng/mL)	-	119.8
NCS	Left peroneal motor and sensory neuropathy	Not done
EMG (measured right-side muscles: triceps, extensor digitorum communis, vastus lateralis, and tibialis anterior)	Insertion activity: normal Spontaneous activity: no fibrillations, no positive sharp waves, no fasciculation, no myotonic response, and no BHP Motor-unit potentials: no reduced amplitudes, normal duration, no polyphasic waves, no giant waves, and full interference pattern	Not done

Normal ranges are within parentheses.

ACTH, adrenocorticotropic hormone; BHP, bizarre high-frequency potential; EMG, electromyography; NCS, nerve conduction study.