

Supplementary Table 1: Mitochondrial enzyme analysis for II.1

	Measure (nmol/min/mg protein)	Reference range (nmol/min/mg protein)	% activity	% citrate synthetase control	% complex II ratio
Complex I	42	19-72	100	79	135
Complex II	34	26-63	76	58	
Complex II+III	23	30-76	50	39	67
Complex III	16.3	12.8-50.9	56	41	72
Complex IV	2.71	3.3-9.1	41	32	55
Citrate synthetase	167	85-179	129		

Supplementary Table 2: Variant impact algorithm scores

	NM_020247.4:c.830T>C		NG_012825.2:c.1506+1G>A	
	<i>Prediction</i>	<i>Score</i>	<i>Prediction</i>	<i>Score</i>
Provean	Damaging	-5.49		n/a
phyloP 100 vertebrates		7.9019		9.798
phyloP 20 mammals		0.998		0.953
MutationAssessor	High	3.5299		n/a
MutationTaster	Disease-causing		Disease-causing	
PolyPhen-2 (HumVar)	Probably damaging	0.971		n/a
SIFT	Deleterious	0		n/a
ClinVar clinical significance	likely pathogenic			n/a
CADD	(phred-scaled)	28		n/a
MetaSVM	Damaging	0.747		n/a
SNPs&GO PANTHER	Disease	0.978		n/a
dbNSFP ADA score				0.9999

Supplementary Table 3: Ataxia assessment for II.1 and II.2

Case		II.1		II.2	
Date		0	10 months	0	10 months
Posture and Gait	Walking capacity	2 /8	2 /8	3 /8	3/8
	Gait speed	1/4	1/4	1/4	1/4
	Standing, eyes open	1/6	1/6	3/6	3/6
	Stance, eyes open	0/4	0/4	1/4	0/4
	Body sway, eyes open	1/4	1/4	1/4	1/4
	Body sway, eyes closed	1/4	1/4	2/4	1/4
	Quality of sitting	1/4	0/4	1/4	0/4
	TOTAL	7/34	6/34	12/34	9/34
Kinetic functions	Knee tibia	3+3/4	2+2/4	4+3/4	3+3/4
	Action tremor (heel-knee)	3+3/4	1+1/4	4+4/4	3+3/4
	Finger nose - dysmetria	3+3/4	2+2/4	3+3/4	3+3/4
	Finger nose – intention tremor	2+2/4	2+2/4	2+2/4	2+2/4
	Finger-finger (action tremor)	2+2/4	1+1/4	2+2/4	2+2/4
	Pronation-supination alternating	2+2/4	2+2/4	2+2/4	2+2/4
	Drawing Archimedes spiral	1/4	1/4	2/4	2/4
	TOTAL	31/52	21/52	35/52	32/52
Speech disorders	Dysarthria - fluency	2/4	2/4	2/4	2/4
	Dysarthria - clarity	0/4	0/4	0/4	0/4
	TOTAL	2/8	2/8	2/8	2/8
Oculomotor	Gaze evoked nystagmus	0/3	0/3	0/3	0/3
	Ocular pursuit	0/2	0/2	0/2	0/2
	Saccadic dysmetria	0/1	0/1	0/1	0/1
	TOTAL	0/6	0/6	0/6	0/6
TOTAL		40/100	29/100	49/100	43/100