

Supplemental Table 2. Neurological examination of CS probands

Age at Exam (years)	Mutation cDNA position	Mutation Protein Position	Vocalizations	Extraocular Movements	Tone, limb or side	Strength, or pattern of weakness	Tremor	Deep tendon reflex (DTR)	Babinski left or right	Gait: base, posture
6-10	c.1380_1389delinsGTCTTGGGAGACT	p.N460Kfs*12	Present	Esotropia, L>R	Normal	Full	None	Normal and symmetric	Absent	Wide base
11-15	c.680+5G>A	-	Present	Esotropia, s.p. surgical correction	Normal	Full	None	Could not assess	Could not assess	Wide base
11-15	c.899+1del	-	Present	Esotropia, L	Could not assess	Full	None	Normal and symmetric	B/L	Unable to walk
11-15	c.680+1G>A	-	~ 5 words	Normal	Normal	Full	None	Normal and symmetric	Absent	Normal
16-20	c.526-1>A	-	Present	Strabismus, s.p. surgical correction	Low-normal	Could not assess	None	High amplitude patellar reflexes with crossed-adductor response	Right	Wide base
16-20	c.630dup	p.I211Yfs*53	Present	Strabismus, s.p. incomplete surgical correction	Low-normal	Full	Upper extremity action tremor	High amplitude throughout	Left	Wide base
21-25	c.540_547dup	p.F183fs*1	Present	Normal	Increased tone, R>L	Full	Left arm intermittent postural tremor	Normal and symmetric	Absent	Wide base
21-25	c.1237-556_*92046del; 120.7kb deletion	-	Present	Normal	Increased tone, L>R	Mild Left hemiparesis	None	Normal and symmetric	Left	Posturing of upper extremities
26-30	c.630dup	p.I211Yfs*53	Present	Normal	Increased tone, Left leg	Full	None	High amplitude throughout; symmetric	B/L	Wide base with toe walking
26-30	c.1148G>A	p.G383D	Present	Normal	Normal	Full	None	Normal and symmetric	Absent	Wide base
26-30	c.1569G>A	p.W523*	Present	Normal	Normal	Full	Left leg tremor	Normal and symmetric	Unable to assess	Wide base
26-30	c.630dup	p.I211Yfs*53	Present	Strabismus, s.p. surgical correction	Increased tone	Full	Upper extremity action tremor	High amplitude throughout	B/L	Wide base, diminished arm swing

L=Left. R=Right. L>R=Left greater than right. R>L=Right greater than left. s.p.=status post.