

**Supplemental Table 1: Published cases of dystonia with a CACNA1A mutation**

Reference	Age of onset	Initial symptoms	Additional Symptoms	Family history	Genetic mutation	MRI findings	Treatments tried
10	42 years	Writer's cramp	A few years following initial symptoms, developed limb ataxia and later axial ataxia	Father: Parkinson's disease D Paternal Family with tremors	CAG 22 repeat expansion in CACNA1A gene, with 12 repeats detected in another allele	Marked cerebellar atrophy with sparing of the cortex and brainstem	Trihexyphenidyl with satisfactory improvement of dystonia
10	15 years	Lower limb dystonia, then cervical dystonia	Mild axial and limb ataxia detected on initial exam	Negative	CACNA1A, C2324 G < A: novel mutation, heterozygous, autosomal dominant, computer modelling suggests pathogenicity  PNKP, C1029 + 2 T < C, heterozygous, autosomal recessive, known pathological mutation  ATP7B, C2544 C < T, autosomal recessive, variant of unknown significance	Unremarkable	Levodopa: No improvement  Botox injection: Some improvement  Cyclobenzaprine: Some improvement  Acetazolamide: No improvement
11	20s	Poor balance	Hand dystonia with writer's cramp	Unknown	c.1748G>A missense mutation in CACNA1A gene	Cerebellar atrophy	

12	15 years	Exercise-induced diplopia	Later developed ataxia, then, at age 59, cervical and arm dystonia	Paternal grandmother: Episodic dizziness and headache  Son: Dizziness and ataxia	CACNA1A, a C-to-T substitution at exon 29 (c.4963C→T) resulted in the creation of a stop codon in place of glutamine (Q1561X) and the subsequent truncation of the protein	Unknown	Acetazolamide improved episodic ataxia  Clonazepam and carbamazepine improved dystonia but worsened ataxia and dysarthria  For neck dystonia, botulinum toxin used
12	5 years	Episodic ataxia	At age 47 developed blepharospasm	Unremarkable	CACNA1A, deletion of a C in exon 20 (c.3772delC) resulted in a frameshift and a predictive truncation of the putative protein at the start of exon 21 at c.3839	Mild midline atrophy of cerebellar vermis	Acetazolamide, diazepam, phenytoin, carbamazepine, propranolol, verapamil, meclizine, nortriptyline, clonazepam (all with no improvement)  Botulinum toxin for blepharospasm with some initial improvement