Supplementary Table 3: Clinical features in Episodic Ataxia Type 1					
Family (mutation)	**Subject (Sex;age)**	**Ataxic phenotype**	**Seizures**	**Neuro-myotonia**	**Other**
Family A (R167M)	I:1 (F; 55)	Onset age 12; gait ataxia and dysarthria lasting 1-2 min; precipitated by change in position and sudden movement; Frequency peaked in adolescence 8 per day and receded in adulthood.	No	Yes	
	II:3 * (M; 25)	Onset age 15 during sport; gait and limb ataxia with dysarthria lasting 4-5 min; predictably occur during sport.; triggered by startle and change in position; frequency diminished in adulthood with altered activity. Episodes necessitated job change.	No	Yes	Hearing impairment attributed to chronic otitis media in infancy resulting in delayed speech.
Family B (C185T)	I:1 (F; 55)	Onset age 10; gait and limb ataxia usually brief but last up to 10 min; brought on by dancing, sport, sudden movement, change in position intercurrent illness; frequency now 3-4 per month.	No	No	
	II:3 (M; 37)	Onset age 10; phenotype dominated by flexion posturing of distal limbs, carpopedal spasm and muscle stiffness which can last hours often triggered by fever or exertion.	No	Yes	Hearing impairment attributed to childhood measles
	II:4 (F; 29) *	Onset age 12; gait and limb ataxia with dysarthria; triggered by exertion, change in position, fever, sudden movement; duration less than 10 min; 1-2 episodes per year; abating with age; also experiences muscle stiffness and carpopedal spasm (less profound than sibling)	No	Yes	Two of four children have intermittent muscle stiffness and difficulty walking during febrile illnesses.
Family C (I407M)	II:2 (F; 92)	Onset age 10; 1-2 min duration; predominantly gait ataxia and dysarthria; sometimes blurred vision. No episodes since age 34.	No	Yes	Hearing impairment
	III:1 (F; 62)	Onset age 14; gait ataxia and dysarthria; 1 min duration; peak frequency in adolescence, nil since age 34; predictably triggered by sport; sometimes triggered by febrile illness.	No	Yes	
	III:2 (F; 58)	Onset age 4; gait and limb ataxia and dysarthria; predictably triggered by sport; also triggered by sudden movement, change in position, emotion. Daily episodes in youth, now 5 per year.	No	Yes	Mild hearing impairment

	III:4* (M; 64)	Onset age 13, prodrome of flash in head; gait and limb ataxia with dysarthria and blurred vision; predictably triggered by sport; duration 1 min;	No	Yes	
	IV:1 (F; 37)	Onset age 13; prodrome followed by gait ataxia and dysarthria; frequency peaked age 19, now occur every few months; triggered by intercurrent illness.	No	Yes	
	IV:2 (M; 33)	Onset age 12; gait ataxia only lasting 30 – 60 s (no dysarthria); predictably triggered by sport; events peaked in adolescence every 2-3 weeks; now seldom occur.	No	Yes	
	IV:6 (M; 29)	Onset age 8; experiences prodrome; gait and limb ataxia, dysarthria, blurred vision; duration 30 s; occur several times per week; triggered by sport, hunger, emotion, sudden movement, startle, change in position, fever, .	No	Yes	
Family D (A242P)	II:1 (F; 64)	Onset age 8; gait and limb ataxia, dysarthria, blurred vision; duration 2 min; 1-2 episodes per year;	No	Yes	Right laterocollis
	III:1 (F; 35)	Onset age 6, gait and limb ataxia, dysarthria, muscle stiffness and hemifacial spasm; triggers include dancing, diving, stress, exercise, sport; frequency 1-2 per month diminishing with time.	No	Yes	Acetazolamide no benefit
	III:2* (F; 30)	Onset age 6; gait and limb ataxia, dysarthria and muscle spasm with thumb adduction; episodes occur every few months and last 30 s; triggered by sport, exertion, change of temperature, diving into water and sometimes fever and emotion	Yes; 2 nocturnal seizures from age 22; controlled with lamotrigine.	Yes	

*Indicates proband