Supplementary Table 3. Published ARCA1/SYNE1 case series of non-Asian populations displaying a broad and diverse spectrum of clinical manifestations

ARCA1 series	Gros-Louis et al.[10]	Dupré et al.[11]	Synofzik et al.[12]	Mademan et al.[13]	Gama et al.[14]	Indelicato et al.[15]
No. of patients	53	64	26	7	6	5†
Ethnicity or region	FC	FC	Turkish, Italian, French, German, Belgian, Moroccan, Algerian	Turkish, Italian, German, Belgian	Brazil	Austria
Mean or median age at onset ± SD (range) (y/o)	30.4 (mean) ± N/A (17–46)	34.79 (mean) ± 7.62 (dysarthria)/31.60 (mean) ± 7.81 (ataxia)	22 (median) ± N/A (6–40)	14 (median) ± N/A (6–42)*	43.3 (mean) ± 11 (26–53)	26 (median) ± N/A (13–29)
Cerebellar signs						
Gaze-evoked nystagmus	7/53 (13.2%)	6/64 (9.4%)	N/A	N/A	2/6 (33.3%)	N/A
Abnormal saccades	16/51 (31.4%)	20/64 (31.2%)	4/26 (15.4%)	N/A	1/6 (16.7%)	N/A
Abnormal pursuit	23/51 (45.1%)	28/64 (43.8%)	N/A	N/A	0/6 (0)	1/1 (100%)
Cerebellar dysarthria	53/53 (100%)	64/64 (100%)	N/A	N/A	6/6 (100%)	1/1 (100%)
Limb ataxia	52/53 (98.1%)	58/64 (90.6%)	N/A	N/A	5/6 (83.3%)	1/1 (100%)
Ataxic gait	52/53 (98.1%)	63/64 (98.4%)	N/A	N/A	5/6 (83.3%)	3/4 (75%)
Relatively pure ataxia	53/53 (100%)	64/64 (100%)	5/26 (19.2%)	0/8 (0%)	3/6 (50%)	3/5 (60%)
Cerebellar atrophy on magnetic resonance imaging or computed tomography	34/34 (100%)	50/50 (100%)	26/26 (100%)	7/8 (87.5%)	6/6 (100%)	5/5 (100%)
Other manifestations						
Brisk reflexes	14/53 (26.4%)	21/64 (32.8%)	N/A	N/A	4/6 (66.7%)	2/2 (100%)
Babinski's sign	N/A	4/64 (6.2%)	N/A	N/A	2/6 (33.3%)	1/1 (100%)
Motor neuron dysfunction	N/A	N/A	15/26 (57.7%)	7/8 (87.5%)	1/6 (16.7%)	2/5 (40%)
Dystonia	N/A	N/A	1/26 (3.8%)	1/8 (12.5%)	2/6 (33.3%)	N/A
Reduced vibratory sensation	N/A	N/A	3/26 (11.5%)	N/A	N/A	N/A
Intellectual disability	N/A	N/A	3/26 (11.5%)	N/A	N/A	N/A
Cognitive impairment	N/A	N/A	N/A	2/8 (25%)	1/6 (16.7%)	N/A
Dysphagia	N/A	N/A	3/26 (11.5%)	N/A	N/A	N/A
Skeletal abnormalities	N/A	N/A	9/26 (34.6%)	2/8 (25%)	N/A	N/A
Respiratory dysfunction	N/A	N/A	3/26 (11.5%)	1/8 (12.5%)	N/A	1/5 (20%)
Myocardial involvement	N/A	N/A	N/A	N/A	N/A	1/5 (20%)
CK elevation	N/A	N/A	4/26 (15.4%)	2/8 (25%)	N/A	1/1 (100%)
Peripheral neuropathy	N/A	N/A	2/19 (10.5%)	5/7 (71.4%)	N/A	1/4 (25%)
Abnormal EMG changes	N/A	N/A	4/14 (28.6%)	4/6 (66.7%)	1/6 (16.7%)	2/4(50%)
Abnormal MEP	N/A	N/A	6/9 (66.7%)	2/5 (40%)	N/A	2/5 (40%)

*the disease started in 4/8 patients (50%) with nonataxic features, namely, facial muscle fasciculations, speech disturbances, spasticity, and cognitive deficits; †genetic testing of one patient revealed a carrier status for spinal muscular atrophy (heterozygous deletion of exon 7). No., number; FC, French–Canadian; SD, standard deviation; y/o, year old; N/A, not applicable or not available; CK, creatine kinase; EMG, electromyography; MEP, motor-evoked potential.