Supplementary Table 1. Summary of the demographic, genetic, and clinical aspects of *SYNE1* mutations reported in the East Asian population until 2022

No.	Case ID	Ethnicity	AAO (y/o)	Initial symptom	Mutation	Phenotype	Upper motor neuron disease	Lower motor neuron disease	Peripheral neuropathy	MRI finding	NCV study	EMG changes	EEG changes	Other manifestations	Reference
1	Proband	Taiwanese	30	Frequent falls	Hetero p.N641Mfs*20 + p.Q1295X	Pure CA	N/A	N/A	N/A	Cerebellar atrophy	Normal	Normal	Normal	N/A	Our study
2	Patient 1	Japanese	6	Running difficulty	Homo p.R7486fs7488X + Homo p.G185R	CA plus	Spastic-ataxic gait	Tongue atrophy, multiple muscle atrophy	N/A	Cerebellar and brainstem atrophy	Decreased CMAP	Chronic denervation	N/A	Respiratory failure	Izumi et al. (2013)[1]
3	Patient 2	Japanese	36	Ataxic gait	Homo p.R3597X	Pure CA	N/A	N/A	N/A	Cerebellar atrophy	N/A	N/A	N/A	N/A	Izumi et al. (2013)[1]
4	Patient 3	Japanese	27	Ataxic gait	Homo p.Y4534fs4539X	Pure CA	N/A	N/A	N/A	Cerebellar atrophy	N/A	N/A	N/A	N/A	Izumi et al. (2013)[1]
5	Patient 1 (IV-5)	Japanese	22	Slurred speech	Homo p.Q2282Sfs*3	Pure CA	N/A	N/A	N/A	Cerebellar atrophy	N/A	N/A	N/A	N/A	Yoshinaga et al. (2017)[2]
6	Patient 2 (IV-1)	Japanese	30	Gait difficulty	Homo p.Q2282Sfs*3	Pure CA	N/A	N/A	N/A	Cerebellar atrophy	N/A	N/A	N/A	N/A	Yoshinaga et al. (2017)[2]
7	Proband	Japanese	11	Frequent falls	Homo p.R6380*	CA plus	Progressive bulbar palsy, Brisk tendon reflexes, Babinski's sign (+)	Tongue atrophy with fasciculation	N/A	Cerebellar and brainstem atrophy	Decreased CMAP	Chronic denervation	N/A	Respiratory failure Chilaiditi syndrome	Kume et al. (2019)[3]
8	Proband's younger sister	Japanese	19	Walking difficulty	Homo p.R6380*	CA plus	Progressive bulbar palsy, Brisk tendon reflexes, Babinski's sign (+)	Tongue atrophy with fasciculation	N/A	Cerebellar and brainstem atrophy	N/A	Chronic denervation	N/A	Respiratory failure Chilaiditi syndrome	Kume et al. (2019)[3]
9	Proband	Japanese	12	Decreased ability to exercise	Hetero p.Arg7155* + p.Lys7612Serfs*28	CA plus	Lower limb spasticity, pathologic reflexes	Distal muscle weakness, atrophy of hand muscles	N/A	Mild cerebellar atrophy	N/A	Chronic denervation	N/A	*Pes cavus*, hand tremor, mild cognitive decline, restrictive ventilation impairment; JALS	Naruse et al. (2021)[4]
10	Case I	Korean	39	Slurred speech	Hetero p.Q1741X + c.26153+1G > A	Pure CA	N/A	N/A	N/A	Cerebellar atrophy	N/A	N/A	N/A	N/A	Kim et al. (2019)[5]
11	Case II	Korean	39	Body swayed while skating	Hetero p.I2995V + p.E5112D	Pure CA	N/A	N/A	N/A	Cerebellar atrophy	N/A	N/A	N/A	N/A	Kim et al. (2019)[5]
12	III-1 in Family 1	Chinese	10	MND	Homo p.Arg7190Ter	CA plus	N/A	Multiple muscle atrophy	N/A	Cerebellar atrophy	Decreased CMAP	Chronic neurogenic	Fronto-temporal slow waves and paroxysmal sharp-slow wave	Intellectual disability bilateral *pes cavus*, mild ankle arthrogryposis	Peng et al. (2018)[6]
13	III-2 in Family 1	Chinese	9	Ataxia	Homo p.Arg7190Ter	CA plus	N/A	Subclinical MND	N/A	Normal	Decreased CMAP	Chronic denervation	Normal	Low normal IQ	Peng et al. (2018)[6]
14	II-2 in Family 2	Chinese	15	Cognitive impairment	Hetero p.Trp6228Ter + p.Arg5982Ter	CA plus	Spastic-ataxic gait, increased muscle tension, hyperreflexia	N/A	N/A	Cerebellar atrophy	Normal	Chronic denervation	N/A	Cognitive decline	Peng et al. (2018)[6]
15	1-IV2	Chinese	23	Gait ataxia	Homo p.L4224Pfs*16	Pure CA	N/A	N/A	N/A	Cerebellar atrophy	N/A	N/A	N/A	N/A	Duan et al. (2021)[7]
16	2-V1	Chinese	18	Spastic gait	Hetero c.20826+1G>T + p.R8652*	CA plus	Hypertonia, pathologic reflexes	N/A	N/A	Cerebellar atrophy	N/A	Chronic neurogenic	Normal	N/A	Duan et al. (2021)[7]
17	3-IV1	Chinese	12	Distal muscle atrophy and weakness	Homo p.L7747Afs*28	CA plus	Hypertonia, pathologic reflexes	Distal muscle atrophy and weakness	N/A	N/A	N/A	N/A	N/A	*Pes cavus*, scoliosis, JALS	Duan et al. (2021)[7]
18	3-IV2	Chinese	10	Distal muscle atrophy and weakness	Homo p.L7747Afs*28	CA plus	Hypertonia, pathologic reflexes	Distal muscle atrophy and weakness, and fibrillation tongue	N/A	Cerebellar and spinal atrophy	N/A	Active and chronic denervation	Normal	*Pes cavus*, JALS	Duan et al. (2021)[7]
19	4-II1	Chinese	24	Ataxic gait, dysarthria	Hetero p.Q1498Rfs*3 + p.V7875Afs*47	CA plus	N/A	N/A	N/A	Cerebellar atrophy	N/A	Normal	Normal	Mild cognitive decline	Duan et al. (2021)[7]
20	5-II1	Chinese	18	Ataxic gait	Hetero p.Q7319* + p.T7926K	CA plus	N/A	N/A	N/A	N/A	N/A	N/A	N/A	Mild cognitive decline	Duan et al. (2021)[7]
21	5-II4	Chinese	15	Intellectual disability	Hetero p.Q7319* + p.T7926K	CA plus	Hypertonia, pathologic reflexes	N/A	N/A	Cerebellar and slight fronto-temporal atrophy	N/A	Chronic neurogenic	Fronto-temporal slow waves	Intellectual disability	Duan et al. (2021)[7]
22	6-II2	Chinese	27	Gait ataxia	Hetero p.D4368Tfs*2 + p.R6156L	Pure CA	N/A	N/A	N/A	Cerebellar atrophy	N/A	N/A	N/A	N/A	Duan et al. (2021)[7]
23	Case 7	Chinese	9	Gait disturbance	Hetero p.K1094X + p.E5273fs	CA plus	N/A	N/A	N/A	Cerebellar atrophy	N/A	N/A	N/A	External ophthalmoplegia, psychiatric symptoms, myoclonic jerks	Cheng et al. (2021)[8]
24	Case 8	Chinese	19	Gait disturbance	Homo p.E3053fs	Pure CA	N/A	N/A	N/A	N/A	N/A	N/A	N/A	Dysphagia	Cheng et al. (2021)[8]
25	Case 9	Chinese	21	Gait disturbance	Hetero p.L6946fs + c.23765+1G>A	CA plus	N/A	N/A	Sensory motor polyneuropathy	Cerebellar atrophy	Sensorimotor neuropathy	N/A	N/A	Tremor, dizziness, pes cavus, intellectual disability, ankylosing spondylitis	Cheng et al. (2021)[8]
26	Case 10	Chinese	25	Gait disturbance	Hetero c.909+1G>A + p.R3479X	CA plus	N/A	N/A	N/A	Cerebellar atrophy	N/A	N/A	N/A	N/A	Cheng et al. (2021)[8]
27	Case 11	Chinese	53	Gait disturbance	Hetero p.R85X + p.H5844fs	CA plus	N/A	N/A	Sensory motor polyneuropathy	Cerebellar atrophy	Sensorimotor neuropathy	N/A	N/A	Dysphagia	Cheng et al. (2021)[8]
28	Proband	Chinese	48	Unsteady gait	Homo p.S2526Sfs*8	CA plus	N/A	N/A	N/A	Cerebellar atrophy	N/A	N/A	N/A	Dysphagia, cognitive decline	Qian et al. (2022)[9]

No., number order; Case ID, individual code name in the respective literature; AAO, age at onset; y/o, years old; MRI, magnetic resonance imaging; NCV, nerve conduction velocity; EMG, electromyography; EEG, electroencephalography; Hetero, heterozygous; pure CA, pure cerebellar ataxia; CMAP, compound muscle action potential; N/A, not applicable or not available; Homo, homozygous; CA plus, cerebellar ataxia with complicated non-cerebellar damage; MND, motor neuron disease; JALS, juvenile amyotrophic lateral sclerosis; IQ, intelligence quotient.