Supplementary Table 1. Demographic, clinical features, investigation findings and follow-up details of the cohort

Variables	Patient-1	Patient-2	Patient-3	Patient-4	Patient-5	Patient-6	Patient-7
			History				
Age/AAO/Gender	23y/7y/F	27y/5y/M	28y/3y/M	15y/5y/F	27y/5y/F	42y/7y/M	23y/4y/M
FH/Consanguinity	+/+	+/+	-/-	-/+	-/-	-/+	-/+
Presenting symptoms	Swaying while walking, slurring of speech, head tremors and tilt to left side	Walking and running difficulty, urinary disturbance, diminished hearing	Delayed milestones, Imbalance while walking, Abnormal posturing of hands	Imbalance while walking, speech disturbance, posturing of hand, difficulty in writing	Imbalance while walking and slippage of footwear	Imbalance while walking	Imbalance while walking, falls, stiffness of lower limbs
GDD/ID	-/-	+/+	+/+	-/-	-/-	-/-	-/-
			Examination findings				
Cognition	Normal	Abnormal	Normal	Normal	Normal	Normal	Normal
Eye movements	Normal	EOM full, gaze evoked nystagmus, dysmetric saccades, broken pursuits	EOM full, bilateral horizontal gaze evoked nystagmus, intermittent upbeat nystagmus, dysmetric saccades	EOM full, gaze evoked nystagmus, saccades and pursuits normal.	EOM full, gaze evoked nystagmus, broken pursuits	EOM full, gaze evoked nystagmus, dysmetric saccades	EOM full, gaze evoked nystagmus, dysmetric saccades, broken pursuits
Fundus	Hypermyelinated fibres radiating from optic disc	Normal	Hypermyelinated fibres radiating from optic disc	Hypermyelinated fibres radiating from optic disc	Normal	Normal	Hypermyelinated fibres radiating from optic disc
Speech	Ataxic	Ataxic	Ataxic	Ataxic	Ataxic	Ataxic	Ataxic
Tone	Spasticity in B/L LL	Spasticity in B/L LL	Moderate spasticity in both UL, Severe spasticity in B/L LL	B/L LL limb spasticity with distal hypotonia	Distal hypotonia in B/L UL and spasticity in B/L LL	B/L LL spasticity	B/L LL spasticity
Weakness	Normal	Normal in B/L UL, Grade 4 in B/L LL	Normal	Finger and toe weakness	B/L toe weakness	Normal	Normal
Deep tendon reflexes	3+	Knee jerk +3, Ankle Jerk absent	3+	3+	3+ except ankle jerk absent	3+ except ankle jerk absent	3+ except ankle jerk absent
Plantar	Extensor	Extensor	Extensor	Extensor	Extensor	Extensor	Extensor
Sensory system	Normal	JPS impaired at B/L great toes, vibration impaired in B/L LL, Romberg positive	Normal	Vibration and JPS impaired in B/L LL	Normal	Normal	Normal
Ataxia	+	+	+	+	+	+	+
Dystonia	Cervical and left-hand dystonia	-	Bilateral hand dystonia	Bilateral finger dystonia	Bilateral hand dystonia	-	Bilateral hand dystonia
Gait	Ataxic	Spastic	Spastic ataxic	Spastic ataxic	Spastic ataxic	Spastic ataxic	Spastic
Additional findings	-	Pes-cavus	B/L Achilles tendon contracture	Bald tongue, flat foot, wasting of thenar and hypothenar eminence	B/L Achilles tendon contracture, pes-cavus, hammer toes	Pes-cavus	-
			Electrophysiology				
VEP	NA	Prolonged	Prolonged	Prolonged	Prolonged	NA	Prolonged
BEP	NA	Prolonged	Prolonged	Prolonged	Prolonged	NA	Absent

SSEP	NA	Prolonged	Prolonged	Absent	Prolonged	NA	Prolonged
NCS	NA	Sensorimotor demyelinating neuropathy	Sensorimotor demyelinating neuropathy	Sensorimotor demyelinating neuropathy	Sensorimotor demyelinating neuropathy	Sensorimotor demyelinating neuropathy	Sensorimotor demyelinating neuropathy
OCT	NA	NA	Increased thickness of RNFL	Increased thickness of RNFL	NA	NA	Increased thickness of RNFL
Magnetic resonance imaging							
Perithalamic T2 hyperintensity	+	+	+	+	+	+	+
Superior vermian atrophy	+	+	+	+	+	+	+
Tigroid appearance in Pons	+	+	+	+	+	+	+
Bulky pons	+	+	+	+	+	+	+
Parietal atrophy	-	+	-	-	+	-	-
Callosal atrophy	-	+	+	-	+	-	+
Arachnoid cyst	-	+	+	-	+	-	-
Atrophy of Cord	+	+	+	Not done	+	+	+
Genetics[#]							
Zygosity	Homozygous	Homozygous	Comp-Het	Homozygous	Homozygous	Homozygous	Homozygous
Variant	c.6000_6004del p.Arg2002Cysfs*25	c.8240T>A p.Ile2747Asn	c.10686_10689del p.Phe3562Leufs*8 c.3810del p.Phe1270Leufs*4	c.13531del p.Glu4511Asnfs*9	c.11356G>T p.Glu3786Ter	c.13469A>C p.Tyr4490Ser	c.1908del p.Lys63Serfs*15
Consequence	Frameshift truncation	Missense	Frameshift truncation	Frameshift truncation	Stop Gain	Missense	Frameshift truncation
Novel (ACMG criteria)[&]	No	Yes (PM2PP1,3M,4M)	No/Yes (PVS1PM2PP4)	Yes (PVS1PM2PP4)	Yes (PVS1PM2PP4)	Yes (PM2PP3M4M)	Yes (PVS1PM2PP4)
Treatment and follow-up							
Treatment	Baclofen – 20mg/d Clonazepam – 1mg/d	Baclofen – 15mg/d Amantadine-200mg/d	Baclofen – 30mg/day	Baclofen – 20mg/d	Baclofen – 30mg/d	Baclofen – 20mg/d	Baclofen – 10mg/d Amantadine-200mg/d
Follow-up	Mild improvement at one-year	Mild improvement at six-months	Status-quo at six-months	Mild improvement at nine-months	Lost to follow-up	Lost to follow-up	Mild improvement at six-months

+: present; -: Absent, #: Reference transcript ID of *SACS* gene NM_014363.6; &: ACMG criteria for pathogenicity provided only if novel.

ACMG: American College of Medical Genetics, BAER: Brainstem auditory evoked potential, B/L: Bilateral, Comp-Het: Compound heterozygous, d: Day, EOM: Extra ocular movements, F: Female, FH: Family history, GDD: Global developmental delay, ID: Intellectual disability, JPS: Joint position sense, LL: Lower Limb, M: Male, mg: Milligram, NA: Not available, NCS: Nerve conduction study, OCT: Ocular computerized tomography, RNFL: Retinal nerve fibre layer, SSEP: Somatosensory evoked potential, UL: Upper limb. VEP: Visual evoked potential