Supplementary Table 2. Clinical, radiological, electrophysiological and genetic details of patients with ARSACS reported from India

Author, year, State	Subject	Age	AAO	Gender	Con/FH	Presenting symptoms	Examination	NCS	MRI	Genetic	Consequence	OCT	Region
Current study, 2024, Karnataka	1	23	7	F	Yes/Yes	Swaying while walking, slurring of speech, head tremors and tilt to left side	Hypermyelinated fibres radiating from optic disc, ataxic dysarthria, bilateral LL spasticity, hyperreflexia, extensor plantar response, cerebellar ataxia, cervical and left-hand dystonia	NA	Linear T2 pontine hypointensity, superior vermian atrophy, bulky pons, lateral thalamic hyperintensity, cord atrophy	Hom; c.6000_6004del	Frameshift deletion	NA	Tamil Nadu
	2	27	5	M	Yes/Yes	Walking and running difficulty, urinary disturbance, diminished hearing	Cognitive impairment, nystagmus, dysmetric saccades, broken pursuit, ataxic speech, spasticity bilateral LL, bilateral lower limb weakness, brisk knee jerk, absent ankle jerk, extensor plantar response, impaired bilateral LL JPS and vibration, ataxia, pes cavus	SMDN	Linear T2 pontine hypointensity, superior vermian atrophy, bulky pons, lateral thalamic hyperintensity, bilateral parietal atrophy, callosal atrophy, posterior fossa arachnoid cyst, cord atrophy	Hom; c.8240T>A	Missense	NA	Andhra Pradesh
	3	28	3	M	No/No	Delayed milestones, Imbalance while walking, Abnormal posturing of hands,	Hypermyelinated fibres radiating from optic disc, nystagmus, intermittent upbeat nystagmus, dysmetric saccades, ataxic speech, spasticity bilateral UL and LL, hyperreflexia, extensor plantar response, ataxia, bilateral hand dystonia, bilateral Achillis tendon contracture	SMDN	Linear T2 pontine hypointensity, superior vermian atrophy, bulky pons, lateral thalamic hyperintensity, callosal atrophy, posterior fossa arachnoid cyst, cord atrophy	CH; c.10686_10689del/c.3810del	Frameshift deletion	Bilateral thickening of RNFL	Karnataka
	4	15	5	F	Yes/No	Imbalance while walking, speech disturbance, posturing of hand, difficulty in writing	Hypermyelinated fibres radiating from optic disc, nystagmus, ataxic speech, spasticity bilateral LL, distal limb weakness, hyperreflexia, extensor plantar response, impaired vibration and JPS in bilateral LL, ataxia, bilateral finger dystonia	SMDN	Linear T2 pontine hypointensity, superior vermian atrophy, bulky pons, lateral thalamic hyperintensity	Hom; c.13531del	Frameshift deletion	Bilateral thickening of RNFL	Karnataka
	5	27	5	F	No/No	Imbalance while walking and slippage of footwear	Nystagmus, broken pursuit, ataxic speech, distal UL hypotonia, spasticity bilateral LL, bilateral toe weakness, hyperreflexia except absent ankle jerk, extensor plantar response, ataxia, bilateral hand dystonia, pes cavus, Achilles tendon contracture	SMDN	Linear T2 pontine hypointensity, superior vermian atrophy, bulky pons, lateral thalamic hyperintensity, bilateral parietal atrophy, callosal atrophy, posterior fossa arachnoid cyst, cord atrophy	Hom; c.11356G>T	Stop gain	NA	West Bengal
	6	42	7	M	Yes/No	Imbalance while walking	Nystagmus, dysmetric saccades, ataxic speech, spasticity bilateral LL, hyperreflexia except absent ankle jerk, extensor plantar response, ataxia, pes cavus	SMDN	Linear T2 pontine hypointensity, superior vermian atrophy, bulky pons, lateral thalamic hyperintensity, cord atrophy	Hom; c.13469A>C	Missense	NA	Tamil Nadu
	7	23	4	M	Yes/No	Imbalance while walking, falls, stiffness of lower limbs	Hypermyelinated fibres radiating from optic disc, nystagmus, dysmetric saccades, broken pursuit, ataxic speech, spasticity bilateral LL, hyperreflexia except absent ankle jerk, extensor plantar response, ataxia, bilateral hand dystonia	SMDN	Linear T2 pontine hypointensity, superior vermian atrophy, bulky pons, lateral thalamic hyperintensity, callosal atrophy, cord atrophy	Hom; c.1908del	Frameshift deletion	Bilateral thickening of RNFL	Karnataka
Divya KP et al, 2023, Kerala	1	23	1	5 males, 2 females	Yes/No	NA	Scanning dysarthria, spasticity, cerebellar signs, normal fundus, nystagmus, peripheral neuropathy, pes cavus, hyporeflexia, extensor plantar response	SMDN	Linear T2 pontine hypointensity, cerebellar and superior vermian atrophy, FLAIR T2 hyperintensity of lateral pons merging with the MCP, bilateral parietal atrophy, thinning of corpus callosum, lateral thalamic hyperintensity, marginal basal ganglia mineralization, DTI thinning of CST	Hom; c.13132C>T	Stop gain	NA	6 Kerala, 1 Tamil Nadu
	2	26	19		Yes/Yes	NA	ID, scanning dysarthria, spasticity, cerebellar signs, normal fundus, nystagmus, peripheral neuropathy, pes cavus, absent JPS and vibration sensation, areflexia, extensor plantar response	SMDN	Linear T2 pontine hypointensity, cerebellar and superior vermian atrophy, FLAIR T2 hyperintensity of lateral pons merging with the MCP, posterior fossa arachnoid cyst, bilateral parietal atrophy, thinning of corpus callosum, lateral thalamic hyperintensity, marginal basal ganglia mineralization, DTI thinning of CST	Hom; c.12851_12854del	Frameshift deletion	NA	
	3	23	18		Yes/No	NA	ID, scanning dysarthria, spasticity, cerebellar signs, normal fundus, nystagmus, peripheral neuropathy, pes cavus, areflexia, extensor plantar response	SMDN	Linear T2 pontine hypointensity, cerebellar and superior vermian atrophy, FLAIR T2 hyperintensity of lateral pons merging with the MCP, posterior fossa arachnoid cyst, bilateral parietal atrophy, thinning of corpus callosum, lateral thalamic hyperintensity, marginal basal ganglia mineralization, DTI thinning of CST			NA	
	4	55	37		No/Yes	NA	Scanning dysarthria, spasticity, cerebellar signs, normal fundus, peripheral neuropathy, pes cavus, sensory dysfunction, areflexia, extensor plantar response	SMDN	Linear T2 pontine hypointensity, cerebellar and superior vermian atrophy, FLAIR T2 hyperintensity of lateral pons merging with the MCP, bilateral parietal atrophy, thinning of corpus callosum, lateral thalamic hyperintensity, marginal basal ganglia mineralization, DTI thinning of CST	Hom; c.8793del	Frameshift deletion	NA	
	5	34	12		No/No	NA	Scanning dysarthria, spasticity, cerebellar signs, peripapillary nerve fibre thickening, nystagmus, peripheral neuropathy, pes cavus, areflexia, extensor plantar response	SMDN	Linear T2 pontine hypointensity, cerebellar and superior vermian atrophy, FLAIR T2 hyperintensity of lateral pons merging with the MCP, thickening of MCP, posterior fossa arachnoid cyst, bilateral parietal atrophy, thinning of corpus callosum, marginal basal ganglia mineralization, DTI thinning of CST	Het; c.8793del	Frameshift deletion	Thick RNFL	

	6	20	14		Yes/No	NA	Scanning dysarthria, spasticity, cerebellar signs, bilateral RNFL thickening, nystagmus, peripheral neuropathy, pes cavus, areflexia, extensor plantar response	SMDN	Linear T2 pontine hypointensity, cerebellar and superior vermian atrophy, FLAIR T2 hyperintensity of lateral pons merging with the MCP, thickening of MCP, posterior fossa arachnoid cyst, bilateral parietal atrophy, thinning of corpus callosum, lateral thalamic hyperintensity, marginal basal ganglia mineralization, DTI thinning of CST	Hom; c.2439_2440del	Frameshift deletion	Thick RNFL	
	7	28	19		No/No	NA	Scanning dysarthria, spasticity, cerebellar signs, peripapillary nerve fibre thickening, nystagmus, peripheral neuropathy, pes cavus, JPS and vibration sensation impaired, hyporeflexia, extensor plantar response	SMDN	Linear T2 pontine hypointensity, cerebellar and superior vermian atrophy, FLAIR T2 hyperintensity of lateral pons merging with the MCP, thickening of MCP, bilateral parietal atrophy, thinning of corpus callosum, lateral thalamic hyperintensity, marginal basal ganglia mineralization, DTI thinning of CST	Hom; c.8793del	Frameshift deletion	Thick RNFL	
Karuvath RH et al, 2021, Karnataka	NA	6	Early childhood	M	NA/No	Ataxia and delayed motor development	dysarthria, dysmetria, dysdiadokokinesia, brisk DTR in lower limbs	SMAN	Linear T2/FLAIR pontine hypointensity, enlarged pons, discrete atrophy of the superior vermis	NA		NA	NA
Sheetal S et al, 2021, Kerala	NA	31	Early childhood	M	No/Yes	Walking difficulty, slurring of speech	Bilateral pes-cavus, scanning speech, normal fundus, slow saccades, GEN, spasticity, mild weakness, brisk DTR in UL and absent in LL, extensor plantar response, impaired LL vibration, impaired tandem walking	SMDN	Superior vermian atrophy, striped pons, thinning of the splenium of the corpus callosum	Hom; c.8793del	Frameshift deletion	NA	Kerala
Agarwal A et al, 2020, Uttar Pradesh	NA	28	3	M	No/No	Progressive gait ataxia, recurrent falls, dysarthria, lower limb stiffness, thinning of feet	Bilateral pes cavus, hammer toes, lower limb spasticity, brisk DTR, extensor plantar, sensory and fundus normal, nystagmus, appendicular and gait ataxia	SMDN	Superior cerebellar vermian atrophy, pontine stripes,	CH; c.4232T>G/c.8132C>T	Stop gain/missense	NA	NA
Sheetal S et al, 2020, Kerala	NA	32	Early childhood	F	No/Yes	Motor developmental delay, unclear speech, running difficulty, sway either side while walking, drop objects, hand tremors, illegible handwriting,	Thinly built, MMSE-28/28, spastic-ataxic speech, 6/12 acuity, fundus normal, slow saccades, broken pursuit, GEN, UL and LL spasticity, 4/5 power, DTR brisk, extensor plantar, appendicular ataxia, spastic-ataxic gait, loss of proprioception till med mall	SMAN	stripe pons, enlarged pons, thickened rostral CC, super vermian atrophy, bi-thalamic stripes	Hom; c.8793delA	Frameshift deletion	NA	Kerala
Shakya S et al, 2019, Delhi	AT1027	35	21	M	No/No	NA	Cerebellar ataxia, Impaired pursuit eye movement, Babinski sign	NA	NA	CH; c.12851_12854del/c.10686_10689del	Frameshift deletion	NA	NA
	AT3450	10	1	F	Yes/No	NA	Ataxia, Nystagmus, Areflexia, SM Neuropathy	NA	Diffuse cerebellar atrophy	Hom; c.12613A>G	Missense	NA	NA
	AT2488	37	22	F	No/Yes	NA	Ataxia, Nystagmus, Slow saccades, Hyperreflexia, Babinski sign, Hammer toe	NA	NA	CH; c.1071dupT/c.7276C>T	Frameshift duplication/Stop-gain	NA	NA
Kuchay RAH et al, 2019, Jammu & Kashmir	NA	28	14 months	M	Yes/No	Slowly progressive spastic-ataxic disorder and mild intellectual disability	Spastic-ataxic gait, lower limb spasticity, extensor plantar sign, broken-up smooth pursuit, gaze evoked nystagmus, loss of ankle jerk, distal lower limb weakness and atrophy, dysarthria	NA	Hypointense pontine strips, hyperintense lateral pons, atrophy of superior vermis and thinning of corpus callosum T2 hyperintense thalamic rim	Hom; c.8605delT	Frameshift deletion	Bilateral thickening of RNFL	Kashmir
Biswas A et al, 2018, Tamil Nadu	NA	9	NA	M	NA/NA	Slowly progressive spastic ataxic syndrome	slowly progressive spastic ataxic syndrome	SMAN	Linear T2/FLAIR pontine hypointensity, enlarged pons, discrete atrophy of the superior vermis, corpus callosum thinning, hyperintense thalamic rim	NA		NA	NA
Agarwal PA et al, 2017, Maharashtra	NA	12	4	M	No/No	Difficulty walking, delay in reaching motor milestones, unclear speech	Myelinated retinal nerve fibres radiating from optic disc, mildly abnormal extraocular movements with saccadic pursuit, normal saccades, no oculo-motor apraxia, spastic-ataxic dysarthria, upper limb ataxia, impaired tandem gait, pes cavus, anormal DTR in UL and absent in LL, normal tone	SMAN	Superior vermian atrophy, linear striped pontine hypointensities, mild cervical atrophy	Hom; c.11690_11693dup	Frameshift duplication	Bilateral thickening of RNFL	NA
Menon MS et al, 2016, Kerala	NA	20	2	M	No/No	Motor and language delay, recurrent falls, swaying ether side while walking, tremulousness in UL om reaching out towards objects, drop objects, slippage of slippers with awareness, handwriting abnormality, hand weakness, decreased interaction, violent outbursts	Thin built, high arched palate, long slender fingers, hypertelorism, right-handed, MMSE 30, 6/12 acuity, normal fundus, horizontal GEN, rebound nystagmus, saccadic hypermetria, small muscle wasting hand foot, 5/5, except hand and foot, DTR brisk except absent AJ, plantar flexor, cerebellar UL and LL, wide based gait, difficulty turning, impaired tandem, head nodding occasionally, sensory normal.	SMDN	Pons stria, superior cerebellar and vermis atrophy	Hom; c.8844delT	Frameshift deletion	NA	Kerala
Faruq et al, 2014, Delhi	AT1573	20	9	F	Yes/Yes	Gait ataxia, hand incoordination, speech impairment	Nystagmus, broken pursuit, brisk DTR, extensor plantar response, spasticity, mild lower limb weakness, pes-cavus, impaired LL proprioception, ataxia	SMN	NA	Hom; c.2117dup	Frameshift duplication	NA	NA
	AT1925	22	15	M	Yes/Yes	Gait ataxia, hand incoordination, speech impairment	Nystagmus, extensor plantar response, spasticity, pes-cavus, ataxia	NA	Superior vermian atrophy			NA	NA
	AT1926	16	11	M	Yes/Yes	Gait ataxia, hand incoordination, speech impairment	Nystagmus, Pes-cavus, ataxia	NA	Superior vermian atrophy			NA	NA

Age and age at onset in years. AAO: Age at onset; CH: Compound heterozygous; Con: Consanguinity; DTR: Deep tendon reflex; F: Female; FH: Family history; FLAIR: Fluid attenuated inversion recovery; GEN: Gaze evoked nystagmus; Het: Heterozygous; Hom: Homozygous; LL: Lower limb; M: Males; MCP: Middle cerebellar peduncle; MMSE: Mini Mental state examination; MRI: Magnetic resonance imaging; NA: Not available; NCS: Nerve conduction study; OCT: Optical coherence tomography; RNFL: Retinal nerve fibre layer; SMAN: Sensory motor axonal neuropathy; SMDN: Sensory motor demyelinating polyneuropathy;

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