Table 2. Study characteristics

Study, year *Country*	**N**	**Inclusion criteria**	**Population**	**Type of testing & genes investigated**	**Genetic cause** n (%))	**Additional information**
Barghigiani, 2022 *Italy*	62	LOA, sporadic at time of first visit. Acquired etiologies and GAA or CAG expansions were preliminary excluded and other targeted tests were performed in selected patients.	Age at onset 56.8 ± 8.8 years [38 to 80], disease duration 8.32 ± 5.93 clinical diagnoses at first visit (n) probable MSA-C (39) possible MSA-C (5), SAOA (18) Sex (F/M (%F)) 28/34 (45)	Testing for CANVAS only (RFC1- AAGGG) Standard PCR, RP-PCR, LR-PCR, and sanger sequencing. Southern blotting.	9 (14.5%)	6 patients in SAOA group 3 patients with MSA-C
Bogdan, 2022 *France*	205	Sporadic ILOCA age of onset>40 y, progressive evolution for last 3 months, absence of family history of cerebellar ataxia	Age at inclusion 62.6±9.4 Age at onset 57.9±9.7 Disease duration 4.7±5.2 years. Sex (ratio M/F) 1.30	The following genes were investigated FMR1 and FXN SCA 1, 2, 3, 6, 7, 17 and DRPLA MELAS, MERRF, and PFBC. If this first-tier analysis was negative, sequencing was performed of a panel including 392 genes related to CA and movement disorders and screening for RFC1 It is unclear from the paper what the genetic work-up was. All patients had FMR1 and FXN testing; the other genes are tested only in a subpopulation. FGF14 not tested.	20 (9.8%)	11 genetic causes in 20 patients, including SPG7 (n = 4), RFC1-associated CANVAS (n = 3), SLC20A2 (n = 3), very-late-onset Friedre ich's ataxia (n = 2), FXTAS (n = 2), SCA3 (n = 1), SCA17 (n = 1), DRPLA (n = 1), MYORG (n = 1), MELAS (n = 1), and a mitochondriopathy (n = 1)
Cortese, 2019 *UK*	150	sporadic CANVAS or late-onset ataxia >35 years	Not provided	Testing for CANVAS (RFC1- AAGGG) WES (7 individuals), RP-PCR (whole group), long range PCR. Southern blot, qRT-PCR, RNA-seq	33 (22%)	32/51 (63%) for LOCA and sensory neuronopathy 11/12 (92%) for full CANVAS syndrome No population defined
Genis, 2025 *Germany*	50	Specific phenotype: patients with a vestibulo-cerebellar ataxia of very late onset (LOVCA)	Age at onset 69.5 [60 to 77] Sex (F/M (%F))	Molecular studies of PolyQ expansions were performed in 35 subjects. In five patients, a molecular study for RFC1 AAGGG repeat expansions was undertaken	15 (37.5%)	

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		Episodes of gait ataxia Mainly downbeat nystagmus Excluded acquired causes of ataxia Excluded patients with CANVAS clinical criteria	21/29(42) Familial/sporadic 10/40	In 28 patients, we searched for variants in the known episodic ataxia genes (*KCNA1*, CACNA1A, and *CACNB4, SLC1A3*), We also screened 36 patients for FGF 14GAA expansions After a post-mortem study gave a diagnosis of a prob able adult polyglucosan body disease (APBD), a search for variants in the GBE1 gene was undertaken. Exome sequencing data were fil tered by 28 genes previously associated with glycogenosis		
Iruzubieta, 2023 *Spain*	107	Age of onset >30 years Progressive ataxie No diagnosis of MSA-C No acquired cause consecutive series of 107 patients family history not mentioned	Age at onset 55 [30 to 82] Sex (F/M (%F)) 47/60 (44) Previously known molecular diagnosis 43 out of 107	Targeted next-generation sequencing gene panels 40% of patients had a previously known molecular diagnosis (43/107). The remaining 64 patients with genetically undefined LOCA were screened for FGF14 GAA expansions. In patients without a molecular diagnosis, 83% (53/64) were screened for intronic FXN GAA repeat expansions, 92% (59/64) for SCA 1, 2, 3, 6, 7, 12, 17, and DRPLA, 100% (64/64) for RFC1expansions, and 61% (39/64) for FMR1 premutation. Targeted next-generation sequencing gene panels were performed in 34% of these patients (22/64) and did not identify pathogenic variants.	61/107 (57%)	
	64	Subgroup of patients without previously known molecular diagnosis	Not provided	FGF-14	18/64 (28%)	
Kartanou 2024 Greek	160	160 patients with LOCA (age at onset >30 years), remaining undiagnosed following screening for SCA1,2,3,6,7, Friedreich ataxia (FRDA), FXTAS and CANVAS were included in this study	Age at onset 60.5 +/- 12.3 range, 34–80	Combination of long rangePCR and bidirectional repeat-primedPCRs.	19 (12%)	Only FGF14 tested
Kontogeorgiou, 2021	67	age of onset ≥35 years	Age 60.1 ± 10.1 [36 to 80]	Screening for CANVAS only - sizing PCR and visualization on agarose gel	2 (3%)	

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Greece		SCA1-3,6,7 and Friedreich ataxia-negative	Age at onset 52.9 ± 10.3 [35 to 72] Sex (F/M (%F)) 40/27 (56%) Familial/sporadic ataxia 7/63	- RP-PCR		
Koutsis, 2024 *Greece*	206	≥30 years of age good DNA quality suspected hereditary ataxia 1995-2023 206 consecutive Greek LOCA patients	Age 60.1 ± 11.2 [35 to 87] Age at onset 52.5±11.4 [30 to 80] Sex (F/M (%F)) 107/99 (51.9) Disease duration 9 [3 to 34] Familial/sporadic ataxia 57/149	PCR, repeat primed-PCR (RP-PCR), agarose gel electrophoresis and fragment analysis were performed to detect the pathogenic tandem repeat expansions. For the diagnosis of FRDA, SCA1,2,3,6,7 and FXTAS, well-established laboratory guidelines were followed. For the diagnosis of CANVAS, cases with no band on sizing PCR, positive RP-PCR for the pathological AAGGG expansion, and negative RP-PCR for the non-pathological AAAGG and AAAAG expan sions were considered positive for biallelic pathological AAGGG ex pansions For the diagnosis of SCA27B, cases with FGF14 expansions longer than a threshold of 250 GAA repeat units were considered positive Analysis did not include NGS to pick up conventional mutations	Sporadic: 34 (22.8%) Total cohort: 62 (30.1%)	In sporadic LOCA FRDA n=4, SCA2 n=2, FXTAS n=2, SCA27B n=13, RFC1 n=12
Montaut, 2021 *France*	100	163 patients with sporadic ataxia of unknown cause after exhaustive investigations were recruited in 3 tertiary centers. 100 ILOA, 21 IEOA, and 42 patients with possible or probable MSA-C.	Age NR Age at onset in ILOA 56 [40 to 78] Sex (F/M (%F)) 43/57(43%)	RFC1 only Repeat Primed-PCR (RP-PCR) and southern blot analysis	15/100 (15%) in the ILOA group	

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Pellerin, 2023 *Canada*	66	Progressive ataxia, onset ≥30 years no clinical features suggestive of MSA-c; exclusion of acquired disease; negative results on testing with an ataxia gene panel and on screening for repeat expansions that cause common spinocerebellar ataxias, fragile X–associated tremor–ataxia syndrome, Friedreich's ataxia, and CANVAS (cerebella rataxia, neuropathy, and vestibular areflexia syndrome).	Age at onset of episodes* 54±14 Age at onset permanent ataxia* 59±12 Sex (F/M (%F))* 36/32 (53%) Familial/sporadic ataxia* 58/10	FGF-14 GAA>250 PCR Lr-PCR Rp-PCR Targeted long-read nanopore sequencing was performed on specimens from 104 patients	40 (61%)	
Pellerin, 2023 *Germany*	228		Age at onset of episodes* 62±7 Age at onset permanent ataxia* 60±11 Sex (F/M (%F))* 24/24 (50%) Familial/sporadic ataxia* 22/22		42 (18%)	
Pellerin, 2023 *Australia*	20		Age at onset of episodes* 48 Age at onset permanent ataxia* 57±11 Sex (F/M (%F))* 0/3 (0%)		3 (15%)	

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			Familial/sporadic ataxia* 1/2			
Pellerin, 2023 *India*	31		Age at onset of episodes* 67 Age at onset permanent ataxia* 62±9 Sex (F/M (%F))* 0/3 (0%) Familial/sporadic ataxia* 1/2		3 (10%)	
Satolli, 2024 *Italy*	239	LOCA onset ≥30 years of age Exclusion of acquired disease. sporadic adult-onset cerebellar ataxia (SAOA)	Age (median [IQR] 62 [18] Age at onset NR Sex (F/M (%F)) NR	FGF14 only tested SCA27B ≥250 repeats were considered pathogenic LR-PCR and RP-PCR	Total: Late onset ataxia 21 (9.8%)	
Traschutz, 2021 *Turkey*	105 index patients (cohort B)	cross-European cohort A (70 families) with ≥2 features of CANVAS or ataxia with chronic cough (ACC) and (2) Turkish cohort B (105 families) with unselected late-onset ataxia Exclusion of SCA1,2,3 in 44% of patients and of Friedreich in 33%, negative WES in 10% of patients	Age [range] [38 to 71] Age at onset NR Sex (F/M (%F)) NR Familial/sporadic ataxia NR	RFC1 only PCR, Southern blot, whole-exome/genome sequencing based approaches)	Prevalence of RFC1 disease was 67% in cohort A, 14% in unselected cohort B, 68% in clinical CANVAS, and 100% in ACC	No distinction sporadic versus familial.

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Wan, 2020	104	unexplained sporadic ataxia with onset after 35 years of age, excluding known genetic causes, including (CAG)n/ (GAA)n expansions (spinocerebellar ataxia [SCA]1, 2, 3, 6, 7, 12, 17, dentatorubral–pallidoluysian atrophy [DRPLA], Friedreich's ataxia [FRDA]), other oligonucleotide expansions, and conventional mutations	Age Age at onset 50.3±6.6 Sex (F/M (%F)) 48/56 (46%) Disease duration 1.9±2.0	CANVAS – RFC1 (biallelic (AAGGG) expansion) PCR Southern Blot WES	1 (1.0%)	biallelic (AAGGG)exp in 1 SAOA patient and 3 MSA patients

Values are presented as mean±SD [range] or median [range]; * values for patients with FGF-14 related ataxia.
Abbreviations: LOA – late onset ataxia; LOCA – late onset cerebellar ataxia