

Table 3 prevalence of (probable) pathogenic findings

Study	N	Total yield (%)	CAN VAS	SCA 27B	SPG7	vLO FA	FXT AS	SCA 1 (ATX N1)	SCA 2	SCA3 (ATX N3)	SCA 6	SCA 7	SCA 17	DRP LA	PFB C	WF M	Gordon Holmes	ScAR 10	MT-TW	SPG -10	Friedreich	Mit. Disease	Episodic ataxia
Barghigiani, 2022	62	14.5	9 (14.5 %)																				
Bogdan, 2022	205	9.8	3 (1,5 %)		4 (2,0 %)	2 (1,0 %)	2 (1,0 %)			1 (0,5 %)			1 (0,5 %)	1 (0,5 %)	4 (2,0 %)							2 (1,0 %)	
Cortese, 2019	150	22	33 (22%)																				
Genis, 2025	40	37.5		15 (37.5%)																			
Iruzubieta, 2023	107	57	16 (16.8 %)	18 (17%)	9 (14.8 %)			1 (0.9%)		2 (1.9%)	3 (2.8 %)					2 (1.9 %)	1 (0.9 %)	1 (0.9 %)	1 (0.9 %)	1 (0.9 %)	2 (1.9%)		2(1.9 %)
	64	28		18 (28%)																			
Kartanou, 2024	160	11.9	19 (11.9 %)																				
Konto-georgiou, 2021	67	3.0	2 (3%)																				
Koutsis, 2024	149	22.8	12 (8.0 %)	13 (8.7%)			1 (0.7 %)		1 (0.7 %)			1 (0.7 %)									4 (2.7%)		
Montaut, 2021	100	15.0	15																				
Ouyang, 2024	399	3.5		14 (3.5%)																			
Pellerin, 2023	66	60.6		40 (61%)																			
	228	18.4		42 (18%)																			

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	20	15.0		3 (15%)																			
	31	9.7		3 (10%)																			
Satolli, 2024	396																						
Traschutz, 2021	105*	14.3	14%																				
Wan, 2020	104	1.0	1 (1.0 %)																				

*Number of families