

Supplementary Table 2: Summary of the clinical presentations of the 43 patients in whom a *CSF1R* mutation was not found.

Case	Sex	Age at onset (y)	Clinical Features					MRI Features	Additional
			Cognitive Decline	Behavioural Change	Seizures	Spasticity	Ataxia	WML	
1	m	28	-	-	-	+	-	+	
2	f	25	-	-	+	-	-	+	
3	f	43	+	+	-	+	-	+	
4	f	36	-	-	-	+	-	+	Vertigo, migraine
4	f	53	+	+	-	-	-	+	
5	m	33	+	+	-	-	-	+	AD Family History
6	m	51	+	-	-	-	-	+	AD Family History
7	f	42	+	-	-	-	-	+	
8	f	55	+	+	-	-	-	+	
9	m	57	+	+	-	-	-	+	AD Family History
10	m	18	+	+	-	+	-	+	
11	m	52	+	+	-	+	-	+	Myoclonus
12	m	35	-	-	-	-	-	+	Migraine
13	f	76	-	-	-	+	-	+	Lower limb sensory disturbance
14	m	57	+	+	-	-	+	+	
15	f	21	-	-	-	+	+	+	
16	m	18	-	+	-	-	-	+	Upper limb tremor
17	m	46	+	-	-	-	-	+	
18	m	64	+	-	-	-	+	+	Myoclonus
19	m	30	-	-	-	+	+	+	

20	m	64	-	-	-	+	+	+	
21	f	53	+	-	-	+	-	+	Vertigo
22	m	18	+	+	-	-	-	+	
23	m	50	+	+	-	-	-	+	
24	f	47	-	-	-	-	+	+	
25	f	42	+	+	-	-	-	+	AD Family History
26	m	60	+	-	-	-	+	+	
27	f	57	+	+	-	-	+	+	
28	m	37	+	+	-	-	-	+	
29	f	18	-	-	+	+	-	+	
30	m	44	-	-	-	-	-	+	Visual Change, dysarthria
31	f	44	+	-	-	-	+	+	Dysarthria
32	m	45	-	-	-	+	+	+	
33	m	18	+	+	-	+	-	+	Myoclonus
34	m	22	+	+	-	-	-	+	
35	f	22	+	-	-	-	-	+	
36	f	44	+	-	+	+	-	+	
37	f	56	+	-	-	-	-	+	
38	m	23	+	-	-	-	-	+	
39	m	34	+	-	-	-	-	+	
40	f	68	+	-	-	-	-	+	
41	f	35	+	-	-	+	-	+	
42	m	42	-	-	-	-	-	+	
43	f	54	-	-	+	+	-	+	

WML: Symmetric T2 hyperintense white matter lesions, m: male, f: female, +: present, -:absent, AD: Autosomal Dominant