Linkage or genetic variant in *TMEM240*	**Ethnicity/nr of cases**	**Age of ataxia onset (AO)**	**Ataxia/Rate of progression**	**Cerebellar atrophy (CA)/Other abnormalities**	**ID**	**Diagnosed as cerebral palsy?**	**Other neurological features than ataxia and ID**	**Author**
Linkage to chromosome 7p21.3-p15.1	1 large French family/15 patients*	6-60 years	Y/Slow	NA	2 patients with ID 64 and 70, in 8 MMS 24-29	No	Akinesia and hyporeflexia	Vuillame et al, Annals of Neurology, 2002
Linkage to chromosome 7p21.3-p15.1	Same family reported by Vuillame et al/ 16 patients	1-30 years	Y/Slow	Yes	Mild cognitive impairment in some	No	Parkinsonian signs in the index case Neuropathological findings in one 50 y.o. woman who died of gastric cancer: severe loss of Purkinje cells and Bergmann's gliosis	Delplanque et al, The Cerebellum, 2008
c.509C>T (P170L) in families A,B and C c.489C>T (Y163*) c.346C>T (R116C) c.239C>T (T80M) c.511C>T (R171W) c.445G>A (E149K)	8 French families including the family (A) reported by Vuillame et al. Family B: 2 patients Families C-H: 1 case each 15 patients (7 of them were from the previously described family)	1-61, most during childhood	Y/Slow	CA in all tested patients (11/15)	Yes, mild to severe	No, but patients had delayed motor development	Behavior disorders (aggressive behavior, apathy, impulsivity)	Delplanque et al, Brain, 2014
c.509C>T (P170L)	I Chinese man (both healthy parents lack the variant in TMEM240)	33 years (clumsiness and tremor)	Y/	Mild CA	Mild mental retardation	No	Hyperreflexia Frontal behavior disorder	Zeng et al, Scientific Reports, 2016
c.509C>T (P170L)	Japanese family/3 cases	5, 30 and 31 years	Y/slow	Yes in 2 studied patients	Yes, MR diagnosed in the index case	No	Psychomotor retardation and cognitive impairment A patient died of a heart attack; neuropathological findings similar to what Delplanque et al reported before	Yahikozawa et al, The Cerebellum, 2018
c.509C>T (P170L) in the German and Dutch families	3 families: German (2 patients), Dutch (1 patient) and Colombian (2 patients)	2-40 years (most in childhood)	Y/Slow	All patients did a brain MRI, only 2 had CA	Yes	No	Myoclonus, hypokinesia, however DAT scan was normal in one case	Traschütz et al, PRD 2019

c.196G > A (p.G66R) in the Colombian family							One of the patients had a transient improvement	
c.196G > A (p.G66R)	1 family in the US/4 patients (father and 3 children), Caucasian ethnicity	Birth-5 years	Y/Slow	Brain MRI was normal in all the children	Neurodevelopmental disorder	No, but all 3 were diagnosed as having neurodevelopmental delay	Myoclonus, hypotonia, chorea, anxiety Oculomotor apraxia in one	Burdekin et al, Journal of Child Neurology 2020
c.509C>T (P170L) in families A and B c.239C>T (T80M) in family C c.196G > A (p.G66R) in family D	5 patients: 2 of them in one Libyan family (A); 3 Italian families (B, C and D, 1 case each)	0-62 years	Y/slow	CA in 3 out of 4 patients 1 pat with delayed myelination	Yes Cognitive delay or CCAS	No	1 with RBD, 1 with strabismus, 1 with hearing loss 1 pat with dystonic posturing None of the patients had hypokinetic features	Riso et al, European Journal of Neurology 2021
c.509C>T (P170L)	1 Brazilian pat (Italian ancestry?)	51 years	Y/Slow	Mild CA	"Cognition was normal"	No	Tremor and dystonia	Camargo et al, European Journal of Neurology 2021

Table e-1: Summary of reported SCA21 patients. Cerebellar atrophy is common but not a universal trait. CCAS: cerebellar cognitive affected syndrome; ID: intellectual disability; NA: No assessed; RBD: REM sleep behavior disorder. *Average AO was 17.4 ±7.4 y.