

Additional Table 1. Phenotypic features of SCA28 patients

Code	Sex	Yrs at onset	Mutation	Yrs at Exam	Severity	Gait ataxia	Limb ataxia	Dysarthria	Horiz. gaze-evoked nystagmus	Ophthalmoplegia	Lower Limbs Reflexes	Ptosis	Bilateral extensor plantar response	Other signs
P1F	F	28	Gly671Arg	50	Mod.	Mod.	Mod.	Mod.	No	Upward, horizontal	+	No	No	-
P2F	F	34	Gly671Arg	39	Mod.	Mod.	Mod.	Mod.	Yes	Horizontal	+	Yes	No	-
P3F	F	29	Gly671Arg	29	None	Mild	Mild	None	Yes	No	+	No	No	-
P4F	F	53	Met666Thr	53	Mod.	Mod.	Mod.	Severe	Yes	Vertical	N	Yes	No	Severe depression
P5F	F	50	Met666Val	77	Mod.	Severe	Mild	Mod.	No	Vertical, horizontal	+	Yes	Yes	-
P1M	M	20	Met666Thr	25	Mod.	Mod.	Mild	Mod.	Yes	No	N	No	No	Head tremor, cognitive difficulties
P2M	M	6	Met666Arg	10	Mild	Mild	Mild	None	No	No	N	No	No	behaviour problems
P3M	M	14	Met666Val	38	None	Mild	Mild	None	Yes	No	+	No	Unilateral	-
P4M	M	10	Thr654Ile	43	Mod.	Mod.	Mod.	Severe	Yes	No	N	No	No	Head tremor, IQ78
P5M	M	25	Met666Val	58	Mild	Mod.	Mod.	Mod.	Yes	Upward	+	Yes	-	-

Notes: Mod., moderate; N, normal; +, augmented;