Appendix

Supplementary Tables and Figures

Association of serum neurofilament light (sNfL) and disease severity in patients with spinocerebellar ataxia type 3

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Table e-1 Subject characteristic in MRI subgroup and Non-MRI subgroup of ataxic stage *ATXN3* mutation carriers

	MRI subgroup	Non-MRI subgroup
Sample size	50	148
Female	25 (50%)	69 (47%)
Expanded CAG repeat of *ATXN3*	72 (70-74)	72 (70-74)
Age (years)	42.06 (8.94)	43.65 (10.38)
Age of onset (years)	35.15 (8.29)	34.57 (8.95)
Disease duration (years)	6.5 (4-8)	8 (5-12.75)
SARA score	11.26 (3.54)	13.50 (10.5-22.38)
INAS score	4 (3-6)	6 (4-7)
Cross-sectional Annual SARA progression	1.96 (1.34-2.34)	1.79 (1.39-2.58)
Serum NfL (pg/ml)	34.76 (27.56-43.50)	37.34 (30.66-48.00)
Serum NfL (ln pg/ml)	3.56 (0.30)	3.63 (0.32)

A total of 198 ataxic stage *ATXN3* mutation carriers were divided into two groups: MRI subgroup (50 ataxic stage *ATXN3* mutation carriers, who received MRI examination) and Non-MRI subgroup (the other 148 ataxic stage *ATXN3* mutation carriers, who did not receive MRI examination). Quantitative data were described as mean (standard deviation) if normally distributed, or median (interquartile range) if non-normally distributed. SARA: scale for the assessment and rating of ataxia. INAS: inventory of non-ataxia symptoms. Cross-sectional annual SARA progression was estimated by the quotient of each subject's SARA score and their disease duration. Natural log-transformation of serum NfL produced plausibly normal distributions, described as ln pg/mL, and were used for all analyses. The MRI subgroup and Non-MRI subgroup did not differ significantly in sex ($\chi^2(1) = 0.190$, $p = 0.663$), expanded CAG repeat of *ATXN3* (U=3843.5, z=-0.125, p=0.901, two-sided Mann–Whitney U tests), age (t(196)=1.00, p= 0.318), age of onset (t(196)=-0.413, p= 0.680), cross-sectional annual SARA progression (U=3845.5, z=-0.118, p=0.906), serum NfL (t(196)=1.621, p=0.107). Compared with Non-MRI group, the MRI subgroup was lower in disease duration (U=2831.0, z=-2.952, p=0.003), SARA score (U=2648.5, z=-3.454, p=0.001), and INAS score (U=2634.0, z=-3.520, p=0.0004).

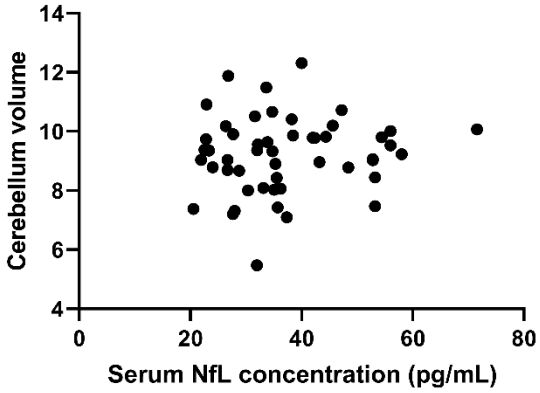

Figure e-1 Association between serum NfL (sNfL) and cerebellum volume in the MRI subgroup. sNfL concentration was not correlated with cerebellum volumes, expressed as percentages of total intracranial volume, tested by using Pearson correlation test (P = 0.489).