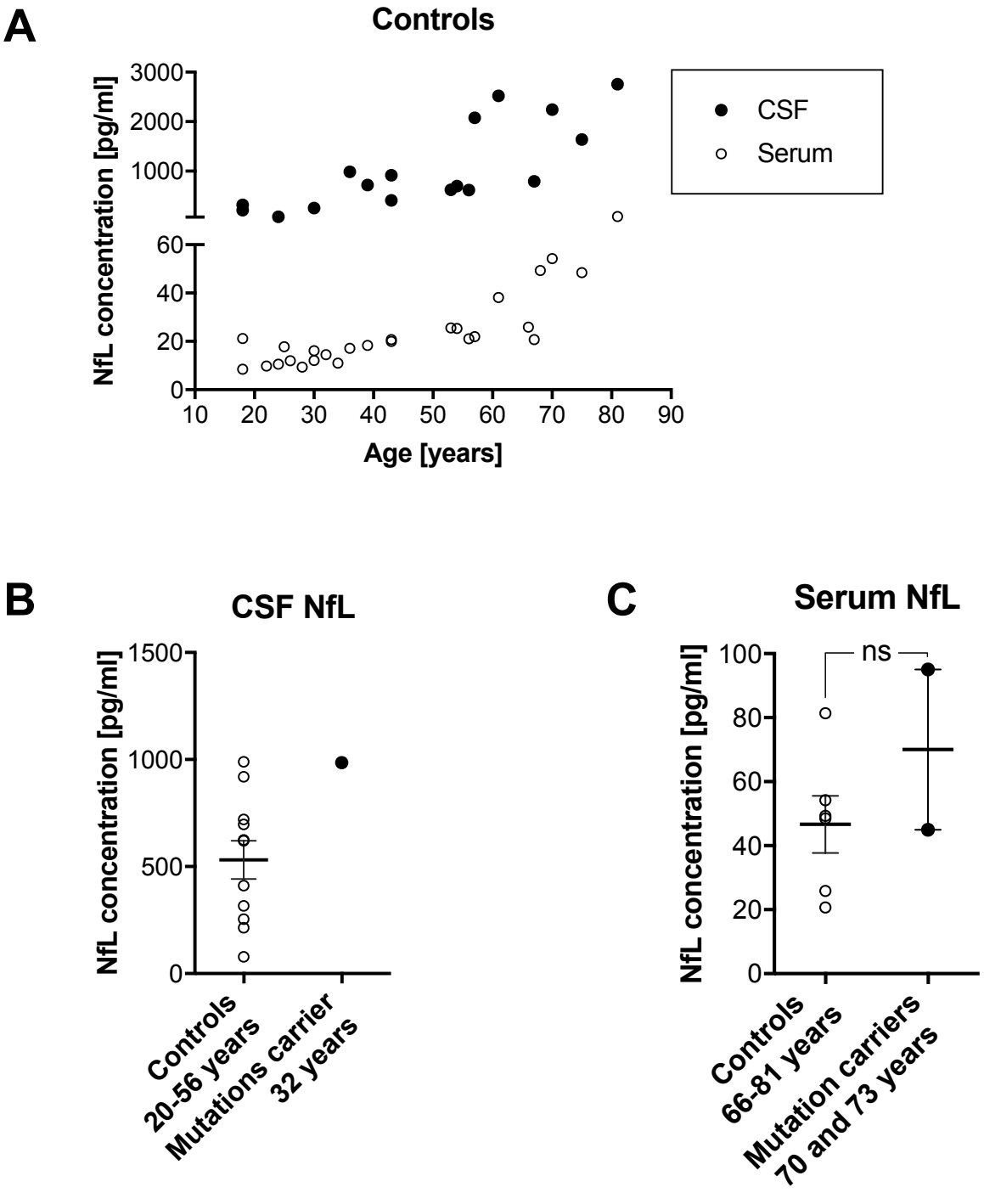

Figure S1. NfL in serum and/or CSF of healthy controls and presymptomatic *CSF1R* mutation carriers. **A**, NfL concentrations in serum and CSF of healthy controls are age-dependent (serum: r=0.849, 95% CI=0.681-0.932, r^2=0.720, p<0.0001, n=26; CSF: r=0.775, 95% CI=0.470-0.915, r^2=0.601, p=0.0003, n=17) and correlate closely with each other (r=0.668, 95% CI=0.262-0.873, r^2=0.440, p=0.0042, n=17). **B**, CSF NfL is elevated in a young presymptomatic *CSF1R* mutation carrier compared to age-matched controls (mutation carrier: 985.4 pg/ml, n=1, controls: 531,2 ± 89.6 pg/ml, 95% CI=331.6-730.8, n=11). **C**, There is no significant difference between serum NfL of elder *CSF1R* mutation carriers and age-matched controls (mutation carriers: 70.1 ± 25.1 pg/ml, 95% CI 35.4-25.1, n=2; controls: 46.7 ± 8.9 pg/ml, 95% CI 23.8-69.6, n=6; p=0.6429), most likely reflecting the lack of neurodegeneration in these mutation carriers, who escaped disease manifestation by an unknown mechanism for at least 70 years.

Data in B, C represent mean ± SEM. CI, confidence interval; CSF, cerebrospinal fluid; NfL, Neurofilament light chain; SEM, standard error of the mean.