**Supplemental figure 3**

MRI of the brain in patient 4 at age 10 years showed; supra- and infratentorial atrophy with widening of sulci, thinning of corpus callosum and narrowing of folia in vermis cerebelli and cerebellar hemispheres on sagittal T1-weighted (a) and axial T2 TIRM dark fluid (b) images; and increased signaling in globi pallidi and thalami (c), midbrain (d) and brain stem (e), cerebellum (d) and U-fibers of supratentorial white matter (f) in axial T2-weighted images.