**SUPPLEMENTARY MATERIALS**

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**Figure 1. Brain Magnetic Resonance Imaging**

Brain MRI was performed on Patient IV-4 with a 3-Tesla MRI scanner using a 12-channel head

coil. T1 weighted spin echo and T2 weighted fluid-attenuated inversion recovery imaging (FLAIR) sequences were used. Cortical and cerebellar atrophy are noted in sagittal (A-C) and axial (D-E) sections. T2 weighted images reveal enlarged lateral (H-J) and third (L) ventricles and bilaterally reduced hippocampal volume (K-L). White matter loss is evident, especially in the frontal region (G) and corpus callosum (H). Abnormal white matter also includes hypointensity in the globus pallidus (F).