Supplementary Online Content


eFigure 1. Neuropathologic Profile of Patient 1

eFigure 2. Neuropathologic Profile of Patient 2

This supplementary material has been provided by the authors to give readers additional information about their work.
eFigure 1. Neuropathologic Profile of Patient 1
Sections form the hippocampus (A; CA2), frontal cortex (B; Layers III and IV), amygdala (C), thalamus (D; anterior thalamus), midbrain (E; substantia nigra), and cerebellum (F) were processed with hematoxylin and eosin (H&E) as well as immunohistochemistry, including antibodies against phospho-tau, α-synuclein, C9RANT, p62/sequestosome-1, and TDP-43. Shown with H&E, all regions have healthy neuronal populations and an absence of gliosis. Many regions throughout the brain display p62 and C9RANT pathology including the hippocampus, frontal cortex, amygdala, thalamus and cerebellum (A-D, F). C9RANT and p62 pathology is quite infrequent in the midbrain and the rest of the brainstem (E). Tau pathology, namely isolated neurofibrillary tangles and neuropil threads, as well as TDP-43 pathology is seen sparsely in the amygdala (C) and other areas not shown here (entorhinal and occipitotemporal cortices). No α-synuclein inclusions were seen in any of these regions. Measure bar is equal to 30 µm for the large H&E image and 10 µm for the smaller immunohistochemistry images.
eFigure 2. Neuropathologic Profile of Patient 2
Sections form the hippocampus (A; CA2), frontal cortex (B; Layers III and IV), amygdala (C), thalamus (D; anterior thalamus), midbrain (E; substantia nigra), and cerebellum (F) were processed with hematoxylin and eosin (H&E) as well as immunohistochemistry, including antibodies against phospho-tau, α-synuclein, C9RANT, p62/sequestosome-1, and TDP-43. Shown with H&E, all regions have healthy neuronal populations and an absence of gliosis. Many regions throughout the brain display p62 and C9RANT pathology including the hippocampus, frontal cortex, amygdala, thalamus and cerebellum (A-D, F). C9RANT and p62 pathology is quite infrequent in the midbrain and the rest of the brainstem (E). Immunohistochemistry with α-synuclein reveals Lewy neurites in the midbrain (E) as well as Lewy bodies and neurites in the amygdala (C). Tau immunohistochemistry revealed sparse neurofibrillary tangles in the amygdala as well as neuropil threads in the amygdala and hippocampus (A). Rare TDP-43-immunoreactive neuronal cytoplasmic inclusions were found in the frontal cortex (B) and in the amygdala (C). Diffuse TDP-43 expression is also seen in some neurons in the substantia nigra (E). Measure bar is equal to 30 µm for the large H&E image and 10 µm for the smaller immunohistochemistry images.