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NEUROPATHOLOGICAL ANALYSIS OF FOUR FRAGILE X SYNDROME (FXS) AUTOPSY BRAINS

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Post-mortem brain tissue is difficult to obtain from individuals with the Fragile X Syndrome (FXS), as these individuals live a normal life span, and often reside with their families or in assisted living facilities; autopsies are not usually performed. Publications that detail the pathology of this disorder are few in number. There is an extensive literature on the MRI and fMRI abnormalities in individuals with FXS, clearly identifying regions of functional pathology. Recently we have performed neuropathological examinations on newly harvested autopsy brain tissue from four patients with FXS who had been followed in our clinics ranging in age from 21 to 74 years. We have analyzed the cerebral cortex, basal ganglia, hippocampal formation, brainstem, and cerebellum with histological and immunocytochemical techniques. We have identified focal neuronal migration abnormalities in the hippocampus, along with prominent vascular hyalinosis in white matter. Ubiquitin staining has shown fine granular depositions in the neuropil and white matter. Clinical, molecular and neuropathological correlations will be discussed.

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