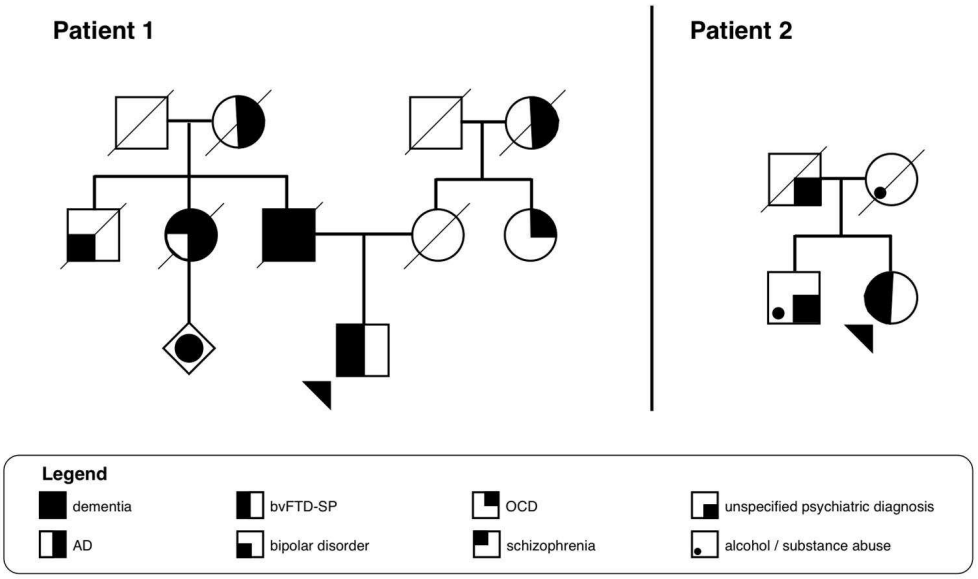

Family pedigrees for Patients 1 and 2. Proband is indicated by black triangle. AD – Alzheimer's disease, bvFTD-SP – slowly progressive behavioral variant frontotemporal dementia, OCD – obsessive-compulsive disorder.
129x77mm (300 x 300 DPI)

Description of neuropathological findings in father of Patient 1.

After Patient 1 was found to harbor a C9ORF72 mutation we were able to obtained limited brain autopsy specimens from the patient's father. These studies revealed an unclassifiable subtype of FTLD-TDP with TDP-43-immunoreactive dots, threads, and neuronal cytoplasmic inclusions (NCIs) that were sparse in frontal and entorhinal cortex and moderate in hippocampal CA1/subiculum, accompanied by hippocampal sclerosis. In the hippocampus, frequent ubiquitin-positive, TDP-43-negative small, round NCIs were identified in dentate gyrus, along with scattered round or stellate NCIs in all subfields of Ammon's horn. In addition, mild argyrophilic grain disease affecting medial temporal lobe structures (especially CA2) was present. Scattered diffuse amyloid plaques were seen in frontal cortex, and no alpha-synuclein pathology was identified.