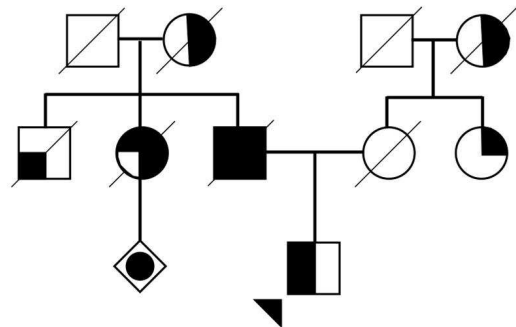
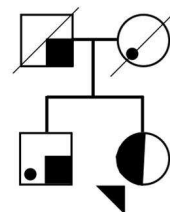


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Patient 1



Patient 2



**Legend**

dementia	bvFTD-SP	OCD	unspecified psychiatric diagnosis
AD	bipolar disorder	schizophrenia	alcohol / substance abuse

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)

For Review Only

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2  
3 Description of neuropathological findings in father of Patient 1.  
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5

6  
7 After Patient 1 was found to harbor a C9ORF72 mutation we were able to obtain limited  
8  
9 brain autopsy specimens from the patient's father. These studies revealed an unclassifiable  
10  
11 subtype of FTLD-TDP with TDP-43-immunoreactive dots, threads, and neuronal cytoplasmic  
12  
13 inclusions (NCIs) that were sparse in frontal and entorhinal cortex and moderate in  
14  
15 hippocampal CA1/subiculum, accompanied by hippocampal sclerosis. In the hippocampus,  
16  
17 frequent ubiquitin-positive, TDP-43-negative small, round NCIs were identified in dentate gyrus,  
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19 along with scattered round or stellate NCIs in all subfields of Ammon's horn. In addition, mild  
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21 argyrophilic grain disease affecting medial temporal lobe structures (especially CA2) was  
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23 present. Scattered diffuse amyloid plaques were seen in frontal cortex, and no alpha-synuclein  
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25 pathology was identified.  
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