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A 5-Year Clinical Course of Phenocopy Syndrome of Behavioral Variant Frontotemporal Dementia: Case Report and Literature Review

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Background and Objective

- Frontotemporal dementia (FTD) is a neurodegenerative disease which results in progressive changes in executive functioning, behavior, or language.¹
- FTD encompasses three major clinical phenotypes including the behavioral variant, semantic variant of primary progressive aphasia, and nonfluent/agrammatic primary progressive aphasia.¹
- The behavioral variant of FTD (bvFTD) is characterized by a gradual decline in personality, social conduct, and cognitive. It is diagnosed by meeting three of six criteria: behavioral disinhibition, apathy, loss of empathy or sympathy, compulsive/ritualistic behaviors, hyperorality and/or dietary changes, and executive deficits with relative preservation of episodic memory and visuospatial skills.²
- In 2006, Davies et al. discovered a new subset of FTD called phenocopy syndrome of bvFTD (phFTD).³ These patients presented with similar symptoms to those of bvFTD, however this condition tended to have slow or minimal progression in cognitive deficits.^{3,4}
- It is called phenocopy because it appeared to “copy” the phenotype seen in bvFTD
- This case focuses on a now 70-year-old male with phFTD who has been living with a diagnosis of FTD for five years, with symptoms for about fifteen years.

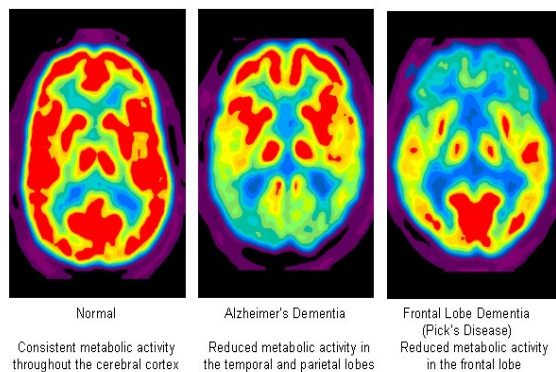


Figure 1: Example of the changes commonly seen on PET scan in Alzheimer's and Frontotemporal dementia.¹⁴ The patient featured in our study exhibited minimal changes on PET scan.

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Case Presentation

- A 66-year-old male presented to the outpatient office in 2018 with his wife reporting about ten years of symptoms that included emotional outbursts, socially inappropriate behavior, apathy, insatiable appetite, and mild short term memory loss.
- Medical history was significant for major depression, type two diabetes mellitus, hypercholesterolemia, and hypertension.
- His initial physical exam was unremarkable besides the following: his affect was pleasant but defensive and disagreeing with his wife throughout which is his baseline presentation. Patient's thought rate was slowed, his process was goal directed, and had decreased associations. Patient's fund of knowledge was seemingly within normal limits, but he demonstrated poor judgment and insight into the full extent of his cognitive impairments.
- MRI in 2018 showed hippocampi volume in the 1st percentile, inferior lateral ventricle volume in the 99th percentile and lateral ventricle volume in the 89th percentile.
- Positron Emission Tomography (PET) scan showed no gross diffuse or focal metabolic abnormalities.
- Neuropsychiatric testing in 2019 showed decline in basic attention and encoding information. He was generally able to retain what he learned. He had borderline impairments in verbal abstract reasoning and semantic fluency. . Based on these findings, he was diagnosed with bvFTD.
- Patient Has a history of emotional outburst
- Initially was on quetiapine but did not help him and was transitioned to Nuedexta (dextromethorphan-quinine) 20mg-10mg every twelve hours.
- Patient has lived away from his wife in their second home on his own and continues to do so as of the date of this report.
- He still drives locally and in familiar places since he has passed multiple driving evaluations.
- His results on the Trail-Making Part B cognitive test have improved over the last three visits with his time on each being: 100 seconds, 90 seconds, and 55 seconds.
- Activities of daily living are currently three out of six with deficits in bathing, continence, and toileting.
- His wife reports difficulties in word finding, but minimal signs have been seen during office visits.
- Last Mini Mental Status Exam (MMSE) was a 30/30
- Despite being unable to fulfill all his ADLs and IADLs, he has shown only moderate signs of deterioration.

Discussion

- The patient presented in this report meets the criteria of phFTD regarding the slow onset of progression.
- Typically, PET scans of FTD patients indicate hypometabolism in the frontal, anterior temporal, and anterior cingulate cortices.⁸ This patient showed no signs of any metabolic changes, resembling other studies indicating a lack of neuroimaging findings in phFTD.
- A study conducted by Steketee et al. also found cortical brain atrophy in patients with phFTD.⁹
- Despite the finding of hippocampal atrophy on MRI, which would typically indicate Alzheimer's dementia, it was not considered given his clinical findings, neuropsychiatric testing, and negative PET scan.
- The reasoning for his hippocampus changes are unclear currently.
- Vascular dementia was also ruled out given his lack of blood vessel abnormalities and absence of stepwise decline in his executive functioning.
- Hornberger et al. and Bussè et al. found a correlation with depression and phFTD and suggested that it may assist in distinguishing between bvFTD and phFTD.^{11,12} Our patient adds another example to the literature of an individual with suspected phFTD and a previous diagnosis of major depression.
- Limited studies in the published literature discuss management of patients with phFTD.
- Based on experiences from this case, treating symptoms of phFTD appears to be the correct approach at this time. Further studies investigating different interventions for patients with phFTD would provide clarity and additional assistance in managing and treating this disorder.

Conclusion

- This case presents a clinical summary of an individual with phenocopy variant of frontotemporal dementia over the course of five years since his diagnosis.
- The clinical presentation depicted in this case demonstrated similar unique characteristics to others presented within the literature such as changes seen on MRI and a comorbid depression diagnosis. .

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