Diagnosis of Frontotemporal Dementia is Often in Non-Specialist Settings and Complicated by Non-FTD Dementia and Psychiatric Diagnoses: Patient and Provider Findings from Analysis of a Large US Real-World Dataset

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OBJECTIVE

To present data from a retrospective, observational real-world data (RWD) cohort study that provides further insights into the epidemiology, patient localization, use of genetic testing, and brain imaging in patients with frontotemporal dementia (FTD), in order to inform an understanding of diagnostic and referral pathways.

BACKGROUND

- FTD is an important and progressive type of early onset dementia, typically manifesting with behavioral/language symptoms, for which there are no approved therapeutics.[1,2]
- Ongoing trials of novel therapies target the genetic subset (5–10%) with progranulin mutations (FTD-*GRN*).[3–5]
- RWD offers an opportunity to gain insights into FTD epidemiology, diagnostic and referral pathways, and utilization of brain imaging and genetic testing, thereby identifying potential opportunities to improve diagnosis and referral.
- Here we present the results of a retrospective, observational RWD cohort study characterizing FTD patients, providers and their use of diagnostic/imaging assessments.

METHODS

- FTD patients were identified in the OM1 RWD covered cohort of ~350 million US lives from January 1, 2013–October 4, 2024. This dataset covered ~1/3 of practicing neurologists and ~1/4 of practicing psychiatrists, respectively. All patients had either medical claims (most common), pharmacy claims, electronic health records (EHR; ~100 million lives), laboratory and/or government data.
- FTD diagnosis was defined by ≥2 FTD diagnosis codes (ICD-10: G31.0 or G31.09) at least 30 days apart.
- The analysis focused on a 6-year period around the first available FTD diagnosis (±3-year observation period).
- Demographic information was captured on or prior to the first FTD diagnosis.
- Claims and electronic medical records data were assessed for concurrent diagnoses, medication use, specialist consultation and use of imaging during the ±3-year observation period. Additional analysis sought to identify the specialty of the healthcare provider associated with the diagnosis of FTD and post-FTD Alzheimer's disease (AD).

RESULTS

- FTD diagnoses were identified in 41,353 patient records across the US, with a mean age of 73 years, 51% female, 90% Caucasian race, and 93% non-Hispanic/non-Latino ethnicity. The age distribution of FTD is outlined in **Figure 1**.
- Only 27% of patients had any neurologist or mental health (MH) professional consultation in the ±3-year observation period.
  - Consultation rates remained consistently low before and after FTD diagnosis (**Figure 2**).
- Concurrent non-FTD dementias were reported in 47% of patients during the pre-diagnosis period, rising to 99% of patients in the post-diagnosis period.
  - Psychiatric comorbidities were also very common (82%) during the ±3-year observation period (**Figure 3**).
- The most common medications reflected psychiatric comorbidities (**Figure 4**). Anxiolytics (47%) and antidepressants (37%) were the most common, with medication patterns remaining consistent across the 6-year period.
- Only 40% of patients had claims for brain imaging, including MRI or CT over the 6-year period.
  - Overall, CT was more common than MRI; more patients received MRI pre-diagnosis than post-diagnosis (**Figure 5**).
- FTD and other dementia diagnoses were infrequently made by specialists, with neurology and psychiatry associated with only 17% and 3% of FTD diagnoses, respectively.
  - Together, primary care physicians, physician-extenders and trainees were more likely to diagnose FTD and post-FTD AD than specialists (**Figure 6**).

Figure 1: Age distribution of FTD

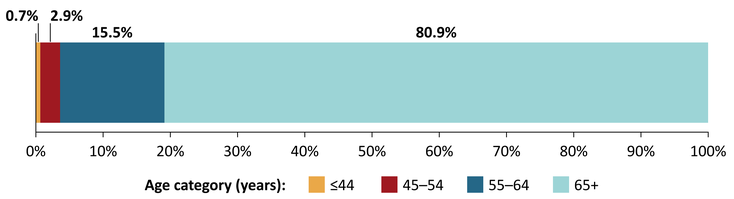
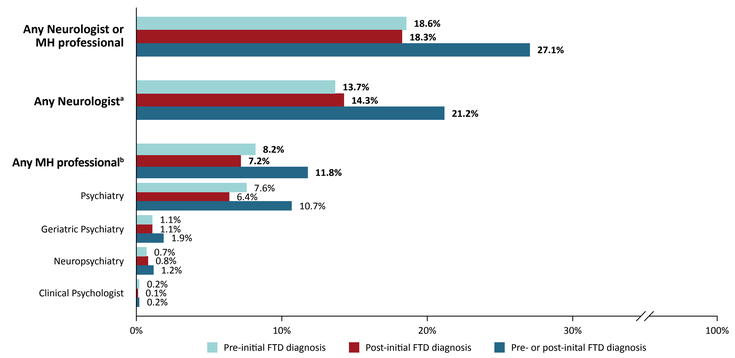


Figure 2: Proportion of FTD patients who had neurologist or MH professional consultations



[a] Includes: neurologist, neurosurgeon. **[b]** Includes: psychiatrists, psychologists, geriatric psychiatrists, neuropsychiatrists, neuropsychologists, other or unknown MH provider. Specialists with >0.1% representation are shown.

Figure 3: Proportion of FTD patients with concurrent diagnoses

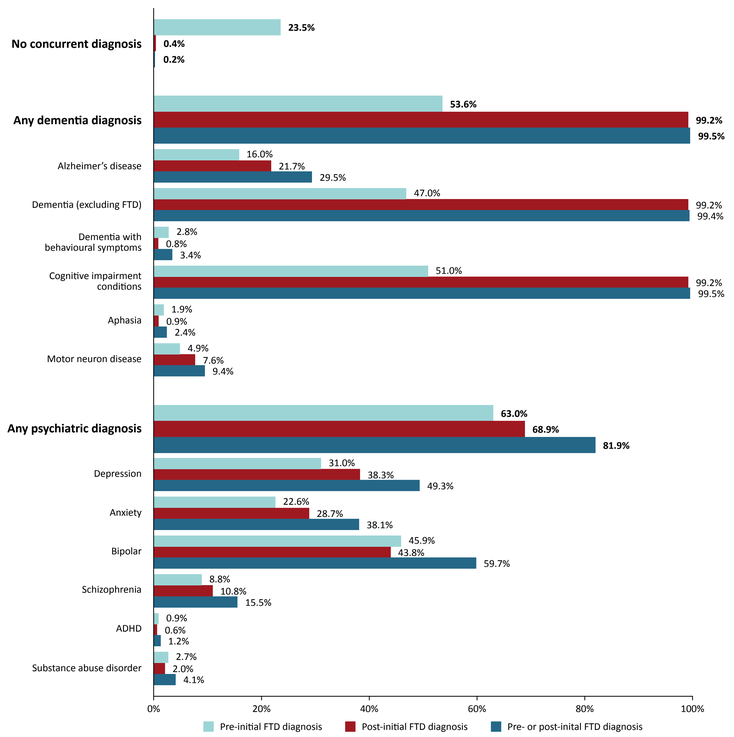


Figure 4: Proportion of FTD patients with concurrent medication use

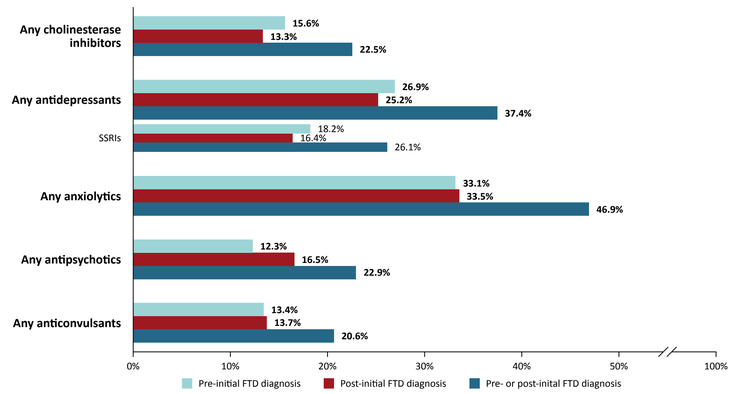


Figure 5: Proportion of FTD patients who received diagnostic or imaging assessments

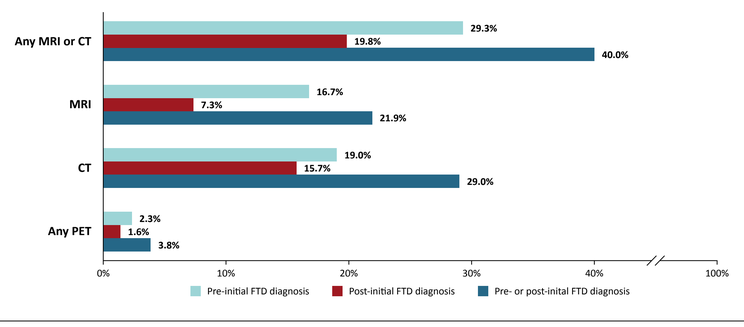
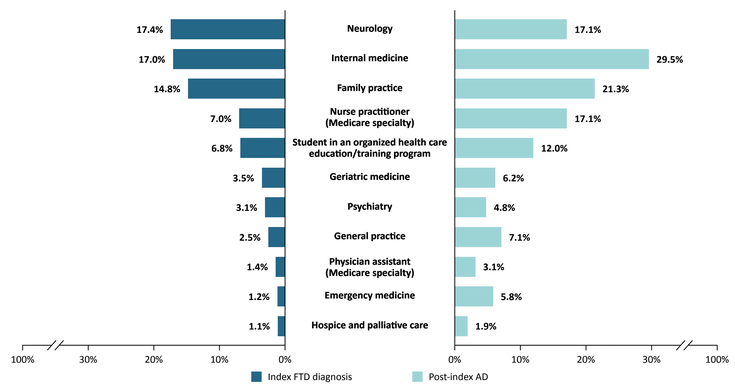


Figure 6: Specialities of the FTD and AD diagnosis provider



Specialties with >1% representation are shown. A provider may have more than one specialty and, therefore, be counted under multiple specialties. Patients may have multiple post-index Alzheimer's diagnoses across multiple provider specialties.

CONCLUSIONS

- **The lack of specialist consultation, limited brain imaging, and the high prevalence of concurrent diagnoses of AD and non-FTD dementias (particularly after the initial FTD diagnosis) suggest challenges in accurate FTD diagnosis. Further, the high prevalence of psychiatric disorders in the absence of MH consultation was unexpected.**
- **Patients with FTD (and AD) diagnoses are predominantly associated with primary care clinicians, suggesting a need for policy and educational efforts to enhance referral pathways alongside disease awareness initiatives.**
- **The primary limitation of this work is the uncertainty in diagnostic accuracy, as the study relies solely on ICD-10 codes associated with claims from multiple providers without verification of actual FTD diagnoses or who made the diagnosis. The claims data, therefore, may not always align in time with EHR data.**

REFERENCES: [1]Young JJ, *et al*. Ther Adv Psychopharmacol 2018;8:33–48; [2]Riedl L, *et al*. Neuropsychiatr Dis Treat 2014;10:297–310; [3]Sevigny J, *et al*. Nature Medicine 2024;30:1406–15; [4] Chan JYC, *et al.* Presented at ISFTD 2024; Thursday-P049; [5]Greaves CV and Rohrer JD. J Neurol 2019;266:2075–86.

ABBREVIATIONS: **AD:** Alzheimer's disease; **ADHD:** attention-deficit hyperactivity disorder; **EHR:** electronic health record; **FTD:** frontotemporal dementia; **CT:** computerised tomography; **ICD:** 10th revision of the International Classification of Diseases; **MH:** mental health; **MRI:** magnetic resonance imaging; **PET:** positron emission tomography; **RWD:** real-world data; **SD:** standard deviation; **SSRI:** selective serotonin reuptake inhibitor; **US:** United States.

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