

Not So Rapidly Progressive Dementia: A Case of Variably Protease-Sensitive Prionopathy (VPSPr)

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KEY POINTS

1. VPSPr with valine homozygosity at codon 129 presents with early neuropsychiatric symptoms and cognitive decline, mimicking frontotemporal degeneration and may evade standard diagnostic techniques.
2. VPSPr should be considered in slower-progressing cases of rapidly progressive dementia.

INTRODUCTION

METHODS & RESULTS

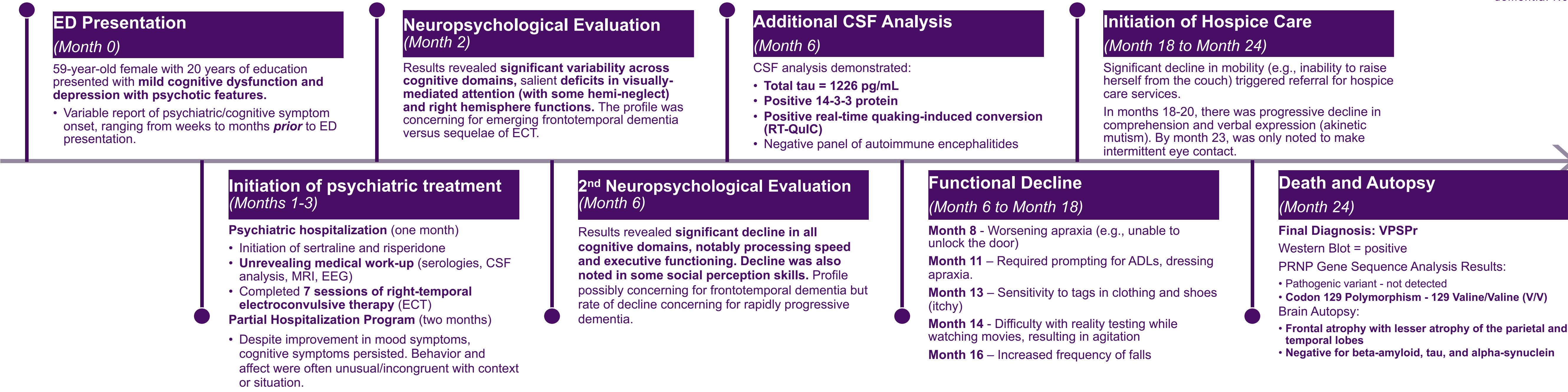
DISCUSSION

REFERENCES

Sporadic prion diseases (SPDs) are phenotypically heterogeneous, making them difficult to diagnose in the setting of slower disease progression. Initial neuropsychiatric changes and marked cognitive decline, in the absence of classical signs (e.g., ataxia, myoclonus), further complicates differential diagnosis. We present a neuropathologically-confirmed case of variably protease-sensitive prionopathy (VPSPr) - a rare, molecular subtype of sporadic prion disease, with valine homozygosity at codon 129, which has been associated with early neuropsychiatric and cognitive symptoms.

Figure 1

Timeline Illustrating Symptom Presentation and Course of Care



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