Neuropsychiatry and behavioral neurology/Dementia

Atypical case of VV1 Creutzfeldt-Jakob disease subtype

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Abstract

Background: Creutzfeldt-Jakob disease (CJD) is rare form of rapidly progressive dementia due to the presence of abhorrent prion protein and affects 1-1.5 cases per million per year. An estimated 85% of these cases are sporadic, and the remaining 5-15% develop CJD from inherited mutations of the prion gene. Sporadic CJD (sCJD) is further subdivided into six subtypes based on genetic polymorphisms, with the VV1 subtype occurring at a rate of 1 case per one-hundredth million population per year. These subtypes have been shown to correlate with age of onset, clinical course, disease features and duration. Clinical characteristics of the VV1 subtype has been reported to show, early age of onset (39 years), disease duration of 15 months, elevated 14-3-3 and total tau in the CSF, absent PSWCs on electroencephalography (EEG), and magnetic resonance imaging (MRI) hyperintensities in the cerebral cortex with usual negative signal in the basal ganglia or thalamus.

Method: We present an atypical case of the rarest VV1 sCJD subtype. Contrary to current data on VV1, our patient presented with an unusual age at onset (63 years), longer disease duration (20 months), and positive signal in the basal ganglia on MRI. The real-time quaking-induced conversion (RT-QuIC) was negative.

Result: Presenting clinical symptoms included paranoid thoughts and agitation, progressing with rapid memory decline, prosopagnosia, and later development of myoclonus and mutism. Other findings showed positive antithyroid peroxidase antibodies (anti-TPO), absent PSWCs on EEG, positive 14-3-3 and elevated tau on CSF. High dose steroid therapy treatment was administered based on positive anti-TPO findings, which failed to illicit any improvement and the patient continued to decline.

Conclusion: To our knowledge, only two confirmed cases with the VV1 subtype, including our patient, have been reported to have a negative result on RT-QuIC. This may indicate a unique characteristic to VV1 and aid in the diagnostic work up in suspected sCJD cases with this finding. However, given the rarity of our patient’s subtype, and the relatively novel RT-QuIC, current data is based on a small number of cases and larger cohorts of confirmed VV1 cases with RT-QuIC testing need to be reported.