Abstract

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A "strange" psicosis

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Background

Prion diseases, a group of disorders caused by abnormally shaped proteins called prions, occur in sporadic (Jakob-Creutzfeldt disease), genetic (genetic Jakob Creutzfeldt disease, Gerstmann-Sträussler-Scheinker syndrome, and fatal familial insomnia), and acquired (kuru, variant Jakob-Creutzfeldt disease, and iatrogenic Jakob-Creutzfeldt disease) forms. It can be difficult to diagnose because of their varied presentations[1].

Case history

A 61-year-old previously healthy female experienced behavioral disturbances for a month, for which she was admitted to our ED with a diagnosis of psicosis. She presented irritability, dystonia and right arm hyperreflexia, without fever or meningeal signs. Routine laboratory tests resulted unremarkable except for increased CRP. Brain MRI highlighted a slight T2 hyper intensity and restricted diffusion of striatum and thalamus (Fig. 1). A cerebrospinal fluid (CSF) sample showed positivity of the real-time quaking-induced conversion RT-QuIC analysis (RT-QuIC). A diagnosis of probable Sporadic JCD was made and the patient was referred to neurological ward for the continuation of the diagnostic-therapeutic process.. She died a month later.

Discussion

Human prion diseases are a group of rare neurodegenerative conditions, that occur in most of the developed world at a rate of 1 to 1.5 cases per million per year. Prion diseases are unique in medicine in that they can occur by three mechanisms: spontaneous (sporadic), genetic (familial), and acquired (infectious/ transmitted). The model of prion disease is that the pathologic disease-causing misfolded form of the prion protein, PrPSc (in which “Sc” stands for scrapie, the prion disease of sheep and goats) acts as a template, such that when it comes into contact with a prion protein, PrPC (in which “C” stands for the normal, cellular form of the protein), it transforms PrPC into PrPSc, resulting in two prions with following exponential transformation and accumulation of prions. Sporadic JCD is the most common form with fatal prognosis. The spectrum of possible symptoms is highly heterogeneous and included rapidly progressive dementia, cerebellar ataxia, and myoclonus, as well as behavioral and psychiatric symptoms. Ancillary tests, such as CSF, EEG, and brain MRI, each with varying utility, can help with sporadic JCD diagnosis. Brain MRI, particularly diffusion-weighted imaging and apparent diffusion coefficient sequences, showing restricted diffusion in the cortical or deep nuclei gray matter has high diagnostic utility, with high sensitivity and specificity. The RT-QuIC assays have made a considerable impact on its clinical diagnosis. This technique exploits the ability of the misfolded pathological form of prion protein (PrP^{Sc}) found in CSF to induce conversion of normal PrP to the misfolded form, which subsequently aggregates with current sensitivity of 92% and specificity until to 100%. To date, effective therapeutics are not available and there is a role for judicious use of pharmacological agents for neuropsychiatric symptoms in some patients[2]. Earlier diagnosis of human prion diseases will allow for a better quality of life for patients should current experimental therapies become a consolidated reality.

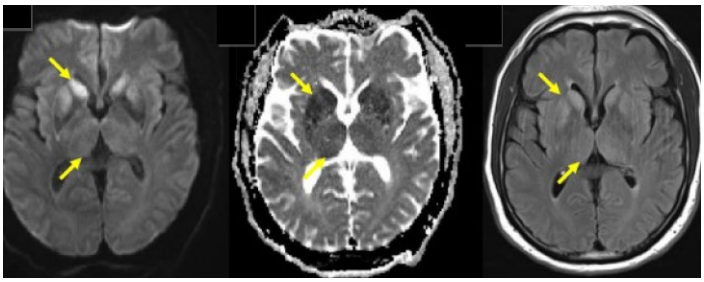

Figure 1. Brain MRI showed restricted diffusion in caudate nucleus, putamen (yellow arrows), and thalamus (less impressive, predominantly in the pulvinar, yellow arrows) in both hemispheres; associated hyper intensities on DWI (D) and FLAIR images (F).

References

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