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RESEARCH ARTICLE

A CASE OF VARIANT CREUTZFELDT–JAKOB DISEASE: THE CRUCIAL ROLE OF MRI

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ABSTRACT

Variant Creutzfeldt–Jakob disease is a rare pathology that represents the infectious form of prion disease. We report the case of a 38-old man presenting the variant Creutzfeldt–Jakob disease reveled by an atypical clinical presentation and evoked thanks to MRI. Brain MRI has a crucial role in the detection of prion disease and to guide biological tests for diagnosis confirmation.

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INTRODUCTION

Prion diseases are rare lethal neurodegenerative conditions that occur when the normal cellular prion protein is converted into an abnormal form which is neurotoxic. Prion disease may be due to genetic, infectious, or sporadic origins. We report the case of a 38-old man presenting a rapidly progressive disturbance of consciousness with brain MRI lesions compatible with prion disease whose infectious origin has been proven by laboratory data. We underline through this article the role of MRI for the detection of this rare pathology.

Case report:

A 38-old immunocompromised man, who was admitted in the intensive care unit for a rapidly progressive disturbance of consciousness evolved into a coma. MRI was performed promptly and showed a bilateral and symmetric hyperintensity on T2 weighted images (Figure 1) and on the Diffusion sequence (Figure 2), lesions are focused in caudate nucleus and lenticular nucleus, without enhancement after contrast and without other cerebral abnormality.

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Prion disease was evoked, and an etiological investigation of the disease has been launched. Electrophoresis data demonstrated an infectious origin with the typical misfolded form of the prion protein (PrP^{Sc}).

DISCUSSION

The human prion diseases represent several rare disorders in which the pathophysiologic event is the conformational changes in the prion-related protein gene (PRNP). These changes result in a misfolded form of the protein within the brain (Degnan, 2014). The worldwide incidence of this disease range between 1 and 2 new cases per million individuals (Holman, 2010). The most common form of human prion diseases is the sporadic Creutzfeldt-Jakob disease and a minority of cases (15%) are due to mutations of the prion protein gene. Prion diseases can be also transmissible as iatrogenic form and zoonotic form (caused by ingestion of mad-cow-infected beef), these forms correspond to variant Jakob-Creutzfeldt disease, iatrogenic Jakob-Creutzfeldt disease, and kuru (Ironside, 2017); without forgetting the genetic forms of the disease (Gerstmann-Sträussler-Scheinker syndrome, genetic Jakob-Creutzfeldt disease, and fatal familial insomnia) (Geschwind, 2015). It is important to note that the sporadic form of the disease is the most common, followed by genetic and acquired types (Geschwind, 2007)

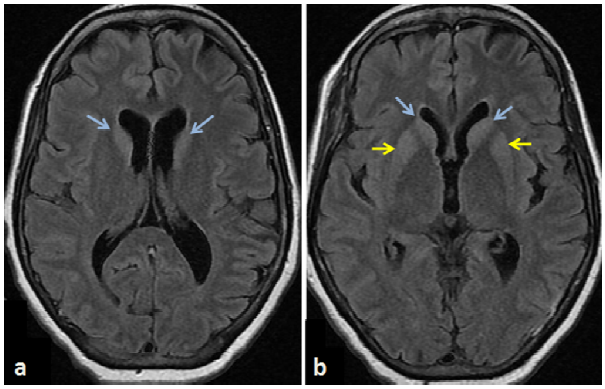

Figure 1. Brain MRI in T2 weighted images in axial section; showing signal abnormality as a bilateral and symmetric hyperintensity interesting caudate nucleus (blue arrows) and lenticular nucleus (yellow arrows).

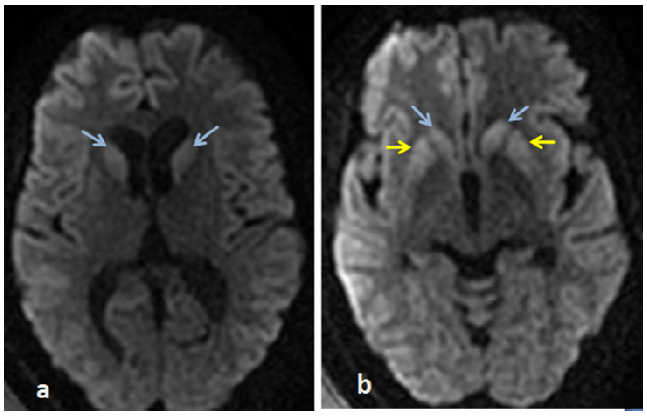

Figure 2. Diffusion sequence images showing a restriction in the lesions in the caudate nucleus (blue arrows) and lenticular nucleus (yellow arrows).

Prion diseases are heterogeneous in their clinicopathologic presentation and make usually a diagnosis challenging. Patients can present dysautonomia, pyramidal and extrapyramidal signs, rapidly progressive dementia, and disturbance of consciousness up to coma. Creutzfeldt-Jakob disease is characterized by cerebellar dysfunction and myoclonus that progresses to corticobasal degeneration with supranuclear palsy (Degnan, 2014). The imaging findings of Jakob-Creutzfeldt disease are hyperintensities in T2 FLAIR and Diffusion sequences within the basal ganglia and thalamus, with reciprocal changes on apparent diffusion coefficient ADC. Recent studies show high sensitivity and specificity for the Diffusion and apparent diffusion coefficient ranging between 92 and 96% (Geschwind, 2015). Diffusion is considered as the most sensitive technique for the detection of Jakob-Creutzfeldt disease (Tschampa, 2007).

The variant Jakob-Creutzfeldt disease is characterized by the "hockey stick sign " or the "pulvinar sign" (75% of cases), that appear as confluent hyperintensity within the dorsomedial thalamus and the posterior thalamus (pulvinar) (Degnan, 2014).

Brain abnormalities detected in our patient by MRI are not specifics of Jakob-Creutzfeldt disease or variant Jakob-Creutzfeldt disease but evoke a prion disease origin which guided the etiological investigations. High cortical signal intensity is also described in the literature, that involve usually the cingulum, insula, superior frontal cortex, and the cortical areas near the midline (Tschampa, 2007). These MRI changes are more sensitive and specific than other methods including electroencephalography that shows periodic sharp waves; the detection of CSF protein 14-3-3 or neuron-specific enolase (Geschwind, 2008; Vitali *et al*., 2001). Despite rare cases reported in the literature of atypical imaging finding in prion disease mimicking other pathological entities (Tschampa *et al*., 2007; Bittar, 2020), imaging and especially MRI is essential in making the diagnosis of prion disease and represent the most sensitive method.

Conclusion

MRI plays a crucial role in the detection of prion disease and to guide biological investigations in order to determine the type of the disease and its origin.

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