

CONTRIBUTION OF MAGNETIC RESONANCE TO THE DIAGNOSIS OF CREUTZFELDT-JAKOB DISEASE

CONTRIBUIÇÃO DA RESSONÂNCIA MAGNÉTICA NO DIAGNÓSTICO DA DOENÇA DE CREUTZFELDT-JAKOB

Leonardo Cordenonzi Pedroso de Albuquerque¹, Juliana Avila Duarte², Artur Francisco Schumacher Schuh¹

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1 Serviço de Neurologia, Hospital de Clínicas de Porto Alegre. Porto Alegre, RS, Brazil.

2 Serviço de Radiologia, Hospital de Clínicas de Porto Alegre. Porto Alegre, RS, Brazil.

Corresponding author:

Juliana Avila Duarte

E-mail: juavila2003@yahoo.com.br

Serviço de Radiologia

Hospital de Clínicas de Porto Alegre

Rua Ramiro Barcelos, 2350.

90035-903, Porto Alegre, RS, Brazil.

A 71 year-old woman presented to our hospital with acute onset of progressive cognitive impairment and psychotic symptoms. Five weeks earlier, she noticed memory impairment and diplopia. After 2 weeks, she was admitted to a psychiatric hospital due to delusions, agitation and hallucinations. After 3 weeks, she was referred to our hospital with progressive deterioration, especially impairment of language and gait. At admission, examination showed aphasia, myoclonic jerks, apraxic gait, and Babinski's sign. No other neurological signs were noticed, and general medical examination was unremarkable. She had no previous psychiatric or significant medical conditions.

Laboratory tests showed mild elevation in anti-thyroid peroxidase titer. Other tests were normal, including complete blood count and tests for liver and renal function, electrolytes, TSH, vitamin B12, inflammatory markers, and HIV and syphilis antibody. Electroencephalogram (EEG) showed a semi-periodic pattern of widespread sharp-wave discharges with periods of arrhythmic widespread theta waves. FLAIR brain magnetic resonance imaging (MRI) (figure 1) showed hyperintensity and decreased diffusivity in the basal ganglia, specially in the head of caudate and putamen, as well as in the cortex of frontal lobes bilaterally, left parietal lobe and bilateral occipital lobes on diffusion weighted images (DWI) (figure 2) and apparent diffusion map. There was no gadolinium enhancement. All of these findings led to diagnosis of probable Creutzfeldt-Jakob disease (CJD)¹.

CJD is a fatal prion-related neurodegenerative disease. This condition is categorized into sporadic CJD (sCJD), familial CJD (fCJD) and variant CJD (vCJD)². sCJD is the most common variant, accounting for 85-90% of the cases, with an incidence of 1-1.5 per million annually¹. fCJD is related to mutations in the prion protein gene (PRNP) on chromosome 20, and accounts for 10-15% of the cases. vCJD is related to bovine spongiform encephalopathy and accounts for 1% of the cases^{1,2}.

CJD primarily presented with cognitive impairment, behavioral symptoms, and sensory and motor dysfunction. sCJD often presents with dementia, generally associated with ataxia and myoclonus, while vCJD has prominent behavioral symptoms, and may be misdiagnosed as a psychiatric illness. These diseases are rapidly progressive, with median survival of 5 months for sCJD, while vCJD has a slightly protracted course^{2,3}.

The definite diagnosis requires pathological confirmation by biopsy. The cerebrospinal fluid (CSF) can be examined for biomarkers, such the 14-3-3 protein, but its use is controversial for high sensitivity and low specificity¹. EEG shows a pattern with periodic sharp and slow wave complexes, with sensitivity of 65% and specificity of 80%¹. FLAIR and DWI MRI show hyperintensities in the cortex, putamen and caudate nucleus, providing a sensitivity and specificity of more than 90%².

In a recent study comparing MRI and CSF biomarkers, MRI had a 97% accuracy in predicting the CJD diagnosis, versus 79.6% of total tau protein (T-tau), 70.4% of 14-3-3 protein, and 71.4% of

neuron specific enolase⁴, and is currently the best non-invasive test for CJD diagnosis².

Unfortunately, the disease is untreatable, invariably progressive, and fatal in 100% of the cases.

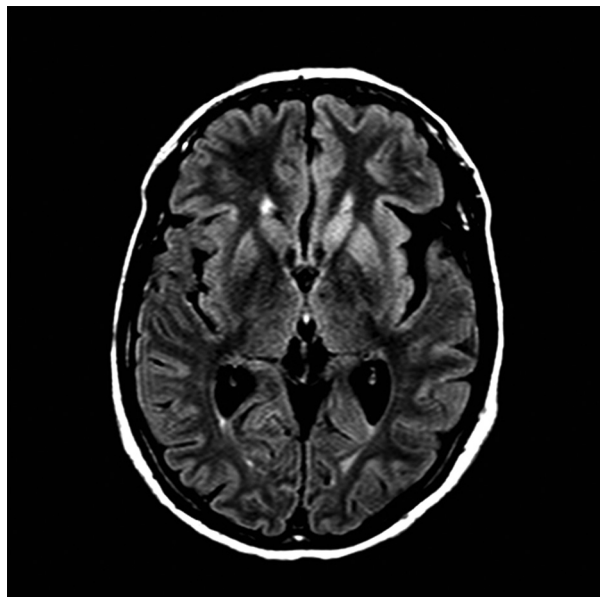


Figure 1: Axial flair weighted image – Frontal, temporal and occipital lobes showing discrete cortical and basal ganglia hyperintensities.

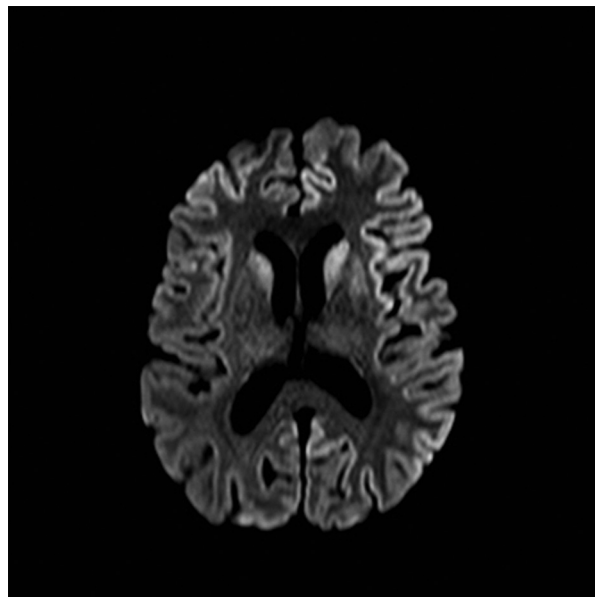


Figure 2: Diffusion weighted image and apparent diffusion map (not shown here) revealing restricted diffusion on the frontal, temporal and occipital lobes, strongly suggestive of Creutzfeldt Jakob disease.

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