DIAGNOSTIC CHALLENGES IN CREUTZFELDT-JAKOB DISEASE: A CASE STUDY

DESAFIOS DIAGNÓSTICOS NA DOENÇA DE CREUTZFELDT-JAKOB: UM ESTUDO DE CASO

DESAFÍOS DIAGNÓSTICOS EN LA ENFERMEDAD DE CREUTZFELDT-JAKOB: UN ESTUDIO DE CASO

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ABSTRACT

Creutzfeldt-Jakob Disease (CJD) is a severe and rapidly progressive neurological disease, with a median survival of 6 to 12 months, caused by the abnormal accumulation of prion protein in the brain. It is characterized by rapid dementia, myoclonus, tremors, ataxia, and motor impairments. Due to its nonspecific clinical presentation, diagnosis is challenging and often delayed, but the association of clinical findings with complementary examinations, such as magnetic resonance imaging, cerebrospinal fluid analysis, and EEG, contributes to its early identification. This report describes the case of a 73-year-old male patient, previously healthy, residing in a rural area of Minas Gerais, who presented with subacute onset of mental confusion, associated with rigidity, incoherent speech, stereotyped movements, and progressive loss of autonomy. Initially treated with antipsychotics and antidepressants without response, he was admitted to a tertiary hospital, where laboratory tests ruled out infections and metabolic disorders. Magnetic resonance imaging (MRI) revealed cortical hyperintensities in the right frontal, parietal, and occipital lobes, as well as involvement of the caudate nucleus, and cerebrospinal fluid analysis revealed positivity for the 14-3-3 protein, consistent with sporadic CJD. During the course of the disease, the patient presented with

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severe neurological deterioration, requiring prolonged care, and bilateral laminar subdural hemorrhage as an associated complication. Therefore, this case highlights the importance of comprehensive assessment in elderly patients with mental confusion, especially after intensive pharmacological treatments. The interaction between pre-existing neurological conditions, medication use, and metabolic factors should be carefully considered.

Keywords: Creutzfeldt-Jakob Disease. Neurology. Clinical Management. Prion. Neurodegeneration. Differential Diagnosis. Electroencephalogram. RT-QuIC. Cognitive Decline.

RESUMO

A Doença de Creutzfeldt-Jakob (DCJ) é uma enfermidade neurológica grave e rapidamente progressiva, com sobrevida média de 6 a 12 meses, causada pelo acúmulo anormal de proteína priônica no cérebro. Caracteriza-se por demência rápida, mioclonias, tremores, ataxia e alterações motoras. Devido à apresentação clínica inespecífica, o diagnóstico é desafiador e muitas vezes tardio, mas a associação de achados clínicos com exames complementares, como ressonância magnética, líquor e EEG, contribui para sua identificação precoce. Este relato descreve o caso de um paciente masculino, 73 anos, previamente hígido, residente em área rural de Minas Gerais, que apresentou quadro de confusão mental de início subagudo, associado a rigidez, fala desconexa, movimentos estereotipados e progressiva perda de autonomia. Inicialmente tratado com antipsicóticos e antidepressivos sem resposta, foi admitido em hospital terciário, onde exames laboratoriais descartaram infecções e alterações metabólicas. A ressonância magnética evidenciou hiperintensidades corticais nos lobos frontais, parietal e occipital direitos, além de acometimento do núcleo caudado, e a análise do líquor revelou positividade para a proteína 14-3-3, compatível com DCJ esporádica. Durante a evolução, o paciente apresentou deterioração neurológica grave, necessitando de cuidados prolongados, e hemorragia subdural laminar bilateral como complicação associada. Portanto, este caso ressalta a importância da avaliação abrangente em pacientes idosos com confusão mental, especialmente após tratamentos farmacológicos intensivos. A interação entre condições neurológicas prévias, uso de medicamentos e fatores metabólicos deve ser cuidadosamente considerada.

Palavras-chave: Doença de Creutzfeldt-Jakob. Neurologia. Manejo Clínico. Prion. Neurodegeneração. Diagnostico Diferencial. Eletroencefalograma. RT-QuIC. Declínio Cognitivo.

RESUMEN

La enfermedad de Creutzfeldt-Jakob (ECJ) es una enfermedad neurológica grave y de rápida progresión, con una mediana de supervivencia de 6 a 12 meses, causada por la acumulación anormal de proteína priónica en el cerebro. Se caracteriza por demencia rápida, mioclonías, temblores, ataxia y deterioro motor. Debido a su presentación clínica inespecífica, el diagnóstico es difícil y a menudo tardío, pero la asociación de los hallazgos clínicos con exámenes complementarios, como la resonancia magnética, el análisis del líquido cefalorraquídeo y el electroencefalograma (EEG), contribuye a su identificación temprana. Este informe describe el caso de un paciente masculino de 73 años, previamente sano, residente en una zona rural de Minas Gerais, quien presentó un cuadro de confusión mental de inicio subagudo, asociado a rigidez, habla incoherente, movimientos estereotipados y pérdida progresiva de autonomía. Inicialmente tratado con antipsicóticos y antidepresivos sin respuesta, ingresó en un hospital terciario, donde las pruebas de laboratorio descartaron

infecciones y trastornos metabólicos. La resonancia magnética (RM) reveló hiperintensidades corticales en los lóbulos frontal, parietal y occipital derechos, así como afectación del núcleo caudado. El análisis del líquido cefalorraquídeo reveló positividad para la proteína 14-3-3, compatible con ECJ esporádica. Durante la evolución de la enfermedad, el paciente presentó un deterioro neurológico grave que requirió cuidados prolongados, y una hemorragia subdural laminar bilateral como complicación asociada. Por lo tanto, este caso resalta la importancia de una evaluación integral en pacientes de edad avanzada con confusión mental, especialmente después de tratamientos farmacológicos intensivos. Se debe considerar cuidadosamente la interacción entre las afecciones neurológicas preexistentes, el uso de medicamentos y los factores metabólicos.

Palabras clave: Enfermedad de Creutzfeldt-Jakob. Neurología. Manejo Clínico. Prión. Neurodegeneración. Diagnóstico Diferencial. Electroencefalograma. RT-QuIC. Deterioro Cognitivo.

1 INTRODUCTION

Creutzfeldt-Jakob disease (CJD) is a rare, progressive and fatal condition of the central nervous system, belonging to the group of transmissible spongiform encephalopathies. It is mainly characterized by a rapidly evolving dementia, accompanied by multifocal motor and neurological changes (Brasil, 2020). Caused by prions, which are infectious particles formed only by abnormal and extremely resistant proteins, CJD presents relevant diagnostic challenges (Brasil, 2020; Zerret al., 2024).

According to the World Health Organization (WHO), definitive confirmation of the diagnosis still depends on neuropathological tests performed after death, although clinical, epidemiological, and laboratory criteria allow classifications as possible or probable cases (Brasil, 2020). The rarity and complexity of the disease reinforce the importance of recognizing atypical clinical presentations, especially in the face of uncharacteristic initial symptoms (Salehi et al., 2022).

CJD can manifest in three main clinical forms: sporadic, hereditary, and iatrogenic. The sporadic form is the most common, accounting for about 85% of cases, appearing without an identifiable cause, usually between the ages of 55 and 75, and characterized by rapidly progressive dementia, ataxia, and myoclonus. The hereditary form, which accounts for 10% to 15% of cases, is related to mutations in the PRNP gene and follows an autosomal dominant pattern of inheritance, and may present earlier onset and symptoms similar to those of the sporadic form. The iatrogenic form, on the other hand, is extremely rare and results from accidental transmission through medical procedures, such as the use of contaminated neurosurgical instruments, transplants of human tissues, or the administration of growth hormone derived from cadavers (Zerr et al., 2024).

Between 2005 and 2021, 1,576 suspected cases of CJD were reported in Brazil, with a higher concentration in the South, Southeast, and Northeast regions of the country. The states with the highest number of notifications were São Paulo, responsible for approximately one third of the records (32.5%), followed by Paraná (12.0%) and Minas Gerais (10%). The most affected age group was 55 to 74 years, representing about 60% of suspected cases, which reinforces the predominance of the disease in older individuals. Regarding gender, the distribution was relatively balanced, with a slight predominance of women (53.6%) compared to men (46.3%), highlighting the importance of epidemiological surveillance and attention to clinical signs, especially in older populations (Brasil, 2020).

CJD has a very varied clinical spectrum, which contributes to the complexity of the diagnosis. Among the most frequent symptoms are rapidly progressing dementia, cerebellar ataxia, and myoclonus, but other manifestations may also be present, making it difficult to

identify the disease early (Hermann et al., 2021). In view of this heterogeneity, the use of high-performance biomarkers becomes essential to increase diagnostic accuracy and assist in differentiating CJD from other neurological conditions with a similar presentation (Sato, 2021).

Currently, there are still no effective therapies for CJD, and diagnostic confirmation remains a major clinical challenge. The diagnosis is based on a combination of typical neuropsychiatric manifestations, imaging tests such as magnetic resonance imaging, electroencephalogram (EEG), and the presence of specific proteins in the cerebrospinal fluid, such as 14-3-3 (Hermann et al., 2021).

Biomarkers such as elevated levels of total tau protein in cerebrospinal fluid have been widely used to support the diagnosis of Creutzfeldt-Jakob Disease However, the reliability of some of these markers, especially the 14-3-3 protein and the tau protein itself, is still the subject of controversy. Although useful as indicators of rapid neuronal injury, both markers have important limitations in terms of specificity. The 14-3-3 protein, for example, may be increased in other acute neurological conditions, such as viral encephalitis, meningitis, strokes, prolonged epilepsies, and other rapidly evolving neurodegenerative diseases, which can lead to false-positive results. Similarly, tau protein elevation is also observed in other neurodegenerative pathologies, such as late-stage Alzheimer's disease (Zerr, 2022).

In addition, the sensitivity of these tests may vary according to the clinical stage of CJD, being less effective in the early stages of the disease, which compromises their usefulness as an early diagnostic tool. The interpretation of the results also depends on the standardization of the laboratory methods used, which can generate variations between diagnostic centers. For these reasons, although these biomarkers continue to be used as support criteria in the classification of probable or possible cases, their application should be done with caution and always associated with other clinical, radiological, and electroencephalographic parameters (Hermann et al., 2021; Noor et al., 2024).

Although progressive cognitive loss is the most common form of CJD onset, atypical presentations in the early stages can make diagnosis difficult and lead to misinterpretations (Nooret al., 2024). In cases with isolated symptoms, such as visual changes or cerebellar signs, it is important to consider variants of the disease (Usluet al., 2020).

When the clinical suspicion remains, tests such as magnetic resonance imaging and EEG should be repeated throughout the follow-up, since the findings may become more evident with the progression of the condition. As there is no cure for CJD currently, treatment is centered on supportive care, with a focus on symptom relief and patient comfort (Rasheedet al., 2024).

In view of this scenario, it is noteworthy that the scarcity of in-depth clinical studies on uncommon manifestations of CJD reinforces the need to report cases that deviate from the classic pattern of the disease, contributing to expand the knowledge of health professionals in the face of conditions that are difficult to diagnose.

Atypical presentations can delay the proper identification of CJD, leading to mistaken initial treatments and prolonged suffering of the patient and their families. This Case Report is justified by the relevance of sharing a real clinical experience that highlights the challenges involved in the suspicion and confirmation of a rare condition, with the aim of enriching medical practice, sensitizing multidisciplinary teams and promoting more assertive conducts in the face of rapidly evolving neurological diseases.

In this perspective, the present study aims to report and analyze an atypical manifestation of Creutzfeldt-Jakob Disease, through a clinical case study, in order to discuss the diagnostic and therapeutic challenges involved, as well as to reflect on the importance of early recognition of non-classical presentations of this rare neurodegenerative disease.

2 METHODOLOGY

This is an observational, descriptive, case report study with theoretical foundation, which presents the clinical manifestation, diagnosis, treatment and evolution of a patient with Creutzfeldt-Jakob Disease (CJD).

The bibliographic search was carried out in the PubMed, SciELO and Science Direct databases, using descriptors related to the disease, with the objective of subsidizing the discussion and contextualizing the case in the current scientific scenario.

The study was carried out in a public hospital located in the eastern region of the state of Minas Gerais. As it is a rare condition, the focus of the research was on the detailed analysis of a single patient, describing in detail the clinical, laboratory and radiological aspects observed during follow-up.

The information was obtained by reviewing the patient's medical record, available in the Medical and Statistical Archive of the institution. To describe the data, a structured instrument was used, covering variables such as initial symptoms, complementary tests performed, differential diagnoses, therapeutic conducts instituted and clinical evolution.

During the document analysis, medical records, imaging reports (such as magnetic resonance imaging), cerebrospinal fluid and electroencephalogram exams, as well as relevant clinical notes were examined, ensuring the integrity and consistency of the information collected.

The description of complementary information occurred through a detailed review of the clinical record, allowing the comparison of the observed findings with the data described in the scientific literature, with the aim of contributing to the improvement of knowledge about CJD and its diagnostic and evolutionary particularities.

The patient's legal guardians were duly informed about the objectives and procedures of the research, and expressed agreement by signing the Informed Consent Form (ICF).

3 CASE REPORT

A 73-year-old male patient, previously healthy, living in a rural area of the eastern region of Minas Gerais, driver, married, accompanied by his grandson, who reported a history of Bell's palsy for four years and cataract surgery for about 60 days. He had previously used carvedilol (Ablok), suspended due to hypotension, and haloperidol drops (2mg/ml - 10 drops/day).

The companion reported that approximately 45 days after the beginning of the investigation, the patient presented acute mental confusion, with subacute and progressive onset. He was initially evaluated by a psychiatrist, with a diagnostic hypothesis of mood disorder, and treatment with venlafaxine was instituted, later replaced by olanzapine and mirtazapine, without satisfactory clinical response. The patient evolved with progressive cognitive worsening, with disconnected speech, muscle rigidity, more evident in the left limdium, but without the "cogwheel" stiffness pattern.

During hospitalization, the patient presented neurological and clinical worsening, rapidly and progressively, becoming dependent for all activities of daily living. He developed stereotyped hand movements, suggestive of parkinsonism, as well as paresthesias in the distal upper limbs.

Magnetic resonance imaging (MRI) of the brain, performed on 10/03/2024, revealed mild cortical hyperintensity in FLAIR, with restricted diffusion in the right frontal, parietal, and occipital lobes, in addition to involvement of the caudate nucleus. Diagnostic hypotheses included ischemic lesions, metabolic causes, and, less likely, prion disease.

About twelve days later (10/15/2024), the patient was admitted to a tertiary hospital in the state of Minas Gerais, presenting with a progressive confusional state. At the initial neurological examination, he was autopsychically oriented, but disoriented in time and space, with comprehension and execution preserved for simple commands, a pattern that was lost during hospitalization.

Initial laboratory tests showed mild renal dysfunction (creatinine: 1.61 mg/dL), blood count without significant alterations, and negative serology for HIV, hepatitis B and C, and VDRL.

A new MRI, performed on 10/17/2024, demonstrated cortical hypersignal on FLAIR and diffusion restriction affecting the superior frontal gyri, posterior cingulate, and right striatum, without contrast-enhanced enhancement. Filling failures suggestive of possible cerebral venous thrombosis were also observed, to be clarified.

On 10/21/2024, a lumbar puncture was performed, which showed cerebrospinal fluid with normal cellularity (3 cells/mm³, predominant mononuclear), slightly elevated proteins (58 mg/dL), normal glucose, and non-reactive VDRL. The molecular panel for infectious agents was negative.

With the clinical and neurological worsening, the patient was transferred to the Intensive Care Unit (ICU), with fluctuations in the level of consciousness, episodes of agitation, seizures, and supraventricular tachyarrhythmia. Levetiracetam 500 mg/day was instituted for subclinical seizure control, in addition to intensive clinical support.

A new brain MRI scan, on 10/30/2024, confirmed the persistence of cortical changes and identified bilateral laminar subdural hematomas. \e the lumbar puncture repeat, on 10/30/2024, showed cerebrospinal fluid with 2 cells/mm³, slightly elevated proteins (68.6 mg/dL) and positive testing for neuronal protein 14-3-3, strongly suggestive of prion disease.

On 10/31/2024, the reference laboratory (FUNED) confirmed the positivity of the 14-3-3 protein in the cerebrospinal fluid, a finding that, together with the clinical pattern and the results of the magnetic resonance imaging, contributed to the establishment of the diagnosis, on 11/01/2024, of Prion Disease, probably sporadic Creutzfeldt-Jakob Disease (CJD).

During hospitalization, the patient received pulse therapy with methylprednisolone, empiric acyclovir, and broad-spectrum antibiotic therapy at times of infectious suspicion, without significant neurological response.

In view of the condition presented, the patient evolved with severe and progressive neurological deterioration, with loss of verbal and visual contact, absence of response to commands, decortication posture and total dependence. On 11/07/2024, he was transferred to the long-term care ward, under the monitoring of the palliative care team, according to the family's desire not to carry out invasive measures.

After hospital discharge, the patient was followed up at home, with the neurology team, together with the palliative care team, with the main focus on adjusting the doses of anticonvulsants, due to the persistence of stereotyped movements and myoclonus. Non-invasive measures were instituted in order to avoid therapeutic obstinacy, prioritizing comfort

and human dignity. During this period, the neurological condition remained stable, with no evidence of new relevant clinical manifestations, compatible with the progressive and inexorable course of sporadic Creutzfeldt-Jakob disease.

At home, the patient presented an episode of community-acquired pneumonia, treated with levofloxacin, with a good initial response, however, approximately one month later, he evolved with a new episode of aspiration pneumonia, and treatment with broad-spectrum therapeutic antibiotic, piperacillin/tazobactam, was instituted at home, without the need for hospitalization at that time.

However, during treatment, the patient presented with an episode of bronchoaspiration, accompanied by progressive clinical deterioration, characterized by acute respiratory failure and hemodynamic instability. The patient developed septic symptoms with a probable pulmonary focus, culminating in death at home, despite adequate support measures and antibiotic therapy.

This outcome reflects the rapidly progressive and fatal character of sporadic Creutzfeldt-Jakob disease, whose clinical course tends to be marked by continuous neurological decline, secondary infectious complications, and therapeutic limitation due to the absence of course-modifying treatment to date.

The case reinforces the importance of early diagnosis, multidisciplinary follow-up and an approach focused on the comfort and dignity of the patient, especially in the advanced stages of the disease, in which palliative care plays an essential role in maintaining quality of life and supporting the family.

4 DISCUSSION

Creutzfeldt-Jakob Disease (CJD) is a rare spongiform encephalopathy, being the most common form among human prion diseases, with an incidence of approximately one case per million inhabitants per year (Sitammagari; Masood, 2021). It is a neurodegenerative disease caused by abnormal prions, predominantly accumulated in brain and eye tissue, especially in the retina and optic nerve. Clinically, it is characterized by rapidly progressing cognitive decline, associated with myoclonus and pyramidal signs, in addition to typical electroencephalographic findings that aid in the diagnosis. However, the clinical course may be nonspecific and similar to that of other neurological diseases, which makes early recognition difficult (Uttley et al., 2020).

The present case clearly reflects many of the classic aspects of CJD, where the patient initially presented with subacute dementia, rigidity, incoherent speech, parkinsonian signs, and rapid progression to total dependence. These findings are in line with traditional

descriptions of the disease, in which accelerated cognitive decline, associated with focal neurological manifestations such as myoclonus and rigidity, make up the most characteristic clinical picture (Appleby et al., 2021; Haddad et al., 2022; Jurcau et al., 2024).

Despite this, CJD can manifest in a heterogeneous way, with variations that make early recognition difficult. In some cases, the initial manifestations mimic psychiatric disorders, focal dementias (with alterations in language or visual perception), or parkinsonian syndromes, which often leads to inappropriate initial therapeutic approaches. In the case described, the introduction of antipsychotics and antidepressants before the neurological hypothesis reflects a pattern observed in other reports in the literature, in which the atypical presentation contributes to the delay in diagnosis (Oliveira et al., 2021; Appleby et al., 2021).

The definitive diagnosis of sporadic CJD is confirmed only by postmortem neuropathological examination, by means of brain biopsy or autopsy. However, according to the criteria of the World Health Organization (WHO), the diagnosis can be classified as possible, probable, or definitive, based on the combination of clinical picture, epidemiological history, and results of complementary tests (Ministry of Health, 2025). Among these, magnetic resonance imaging (MRI) and electroencephalogram (EEG) may present findings characteristic of the disease, being fundamental to support clinical suspicion (Jurcau et al., 2024).

In this context, one of the highlights of the present case was the use of MRI with FLAIR and diffusion (DWI) sequences, showing cortical hyperintensities in the right frontal, parietal, and occipital lobes, as well as involvement of the caudate nucleus, and then evolution of the findings to other structures (superior frontal gyri, posterior cingulate, striatum).

Similar to the present case, Farnetano et al. (2019) reported a 74-year-old female patient from Ubá (MG) with rapidly progressing dementia, cerebellar ataxia, and myoclonus, who was later diagnosed with sporadic CJD, whose DWI-weighted MRI of the head showed cortical hyperintensity in the frontal lobes and in the caudate nuclei and bilateral thalamus. Similarly, Appleby et al. (2021) described a 21-year-old man in the United States with cognitive decline, behavioral changes, and progressive motor disorders, whose MRI demonstrated asymmetric cortical hyperintensity and involvement of the caudate nuclei, findings compatible with prion disease.

Reinforcing these findings, Park et al. (2021) found in their meta-analyses that DWI has a sensitivity of around 91% (95% CI: 84-95%) and specificity around 97% (95% CI: 94-99%) when used to diagnose sporadic CJD in patients with rapidly progressive dementia.

In the present case, the diffusion alterations were initially detected and later persisted and evolved, which reinforces the usefulness of repeating neuroimaging studies when the

clinical picture progresses. This also highlights that even in cases without cogwheel stiffness or other specific early signs, MRI findings can strongly guide the diagnosis.

In addition to the characteristic neuroimaging findings, cerebrospinal fluid (CSF) analysis for the detection of the 14-3-3 protein is one of the main diagnostic criteria for the sporadic form of CJD, helping to confirm the clinical suspicion and differential diagnosis in relation to other rapidly progressive encephalopathies (Hermann et al., 2021).

The positivity of the 14-3-3 protein in the cerebrospinal fluid observed in this patient reinforces the findings described in the literature on CJD. According to Sahyouni et al. (2024), the detection of the 14-3-3 protein in CSF allowed the confirmation of the diagnosis of sporadic CJD after the interpretation of the tests. In a study conducted by Senesi et al. (2023) in an Australian cohort of patients with sporadic CJD, the analysis of the 14-3-3 protein by western blot (WB) showed a sensitivity of 87.5% and specificity of 66.7%, while the 14-3-3 ELISA assay demonstrated a sensitivity of 81.3% and specificity of 84.4%. The authors also highlighted that the CSF real-time tremor-induced conversion assay (RT-QuIC) had the best diagnostic performance, with a sensitivity of 92.7% and specificity of 100%, and is considered the most reliable test for confirming the disease.

However, it is important to note that the positivity of the 14-3-3 protein is not exclusive to CJD, and may occur in other conditions associated with acute neuronal injury, such as stroke, encephalitis, and metabolic disorders (Hermann et al., 2021). Therefore, the interpretation of this finding should always be done in conjunction with clinical and neuroimaging data. In the present case, serology for infectious agents was negative, and the exclusion of other causes consistently reinforced the diagnostic suspicion of sporadic CJD.

In view of this scenario, it is important to emphasize that to date, there are no specific therapies that substantially alter the course of CJD (Hall; Masood, 2025). Treatments are generally supportive, with palliative care when there is already clear deterioration, such as the condition of the patient reported in this case (Uttley et al., 2020). The literature confirms that empirical interventions rarely bring significant clinical benefit, given that the disease is essentially degenerative and rapidly progressing (Miranda et al., 2022; Sahyouni et al., 2024).

It is noteworthy that the prognosis of CJD is largely unfavorable, with rapid progression to total dependence and death within a few months. The present case followed this typical course, characterized by progressive neurological deterioration, loss of responsiveness, and need for long-term care, culminating in the adoption of palliative measures and the decision to avoid invasive interventions, in accordance with ethical principles of end-of-life care. The literature reinforces that CJD has an invariably fatal outcome, usually less than a year after

the onset of symptoms, although early recognition of the disease can help in clinical management and attenuation of the patient's suffering (Uttley et al., 2020).

Despite the detailed characteristics presented in this report, it is important to recognize its limitations, such as the absence of post-mortem confirmation, possible variations in laboratory methods for detecting the 14-3-3 protein, the absence of EEG, and the potential selection bias inherent in studies involving a single patient. Even so, this case can add value to the literature by describing the evolution of CJD in a previously healthy elderly patient, with the onset of a psychiatric condition and compatible neuroimaging and cerebrospinal fluid findings, reinforcing the relevance of early clinical suspicion and repeated tests in view of the progression of the condition.

5 CONCLUSION

The case presented here highlights the diagnostic complexity of sporadic Creutzfeldt-Jakob disease, a rare and fatal neurodegenerative disease characterized by rapid cognitive deterioration and multifocal neurological signs. The patient's clinical trajectory reinforces that initial nonspecific manifestations, such as behavioral changes and mental confusion, can simulate psychiatric disorders, delaying the definitive diagnosis.

The association between typical magnetic resonance imaging findings, especially in FLAIR and diffusion sequences, and the positivity of the 14-3-3 protein in the cerebrospinal fluid, demonstrated great diagnostic value, corroborating what is described in the international literature. It is noteworthy that, even in the absence of specific treatment, early recognition of the disease is essential to avoid futile therapeutic conducts, allow family planning, and ensure referral to appropriate palliative care.

Finally, this report contributes to the expansion of clinical knowledge about CJD in the Brazilian context, emphasizing the importance of diagnostic suspicion in cases of rapidly progressive dementia and the need for training health professionals to recognize its early signs.

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