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CASE REPORT

Probable Creutzfeldt-Jakob Disease in a Resource-Limited Setting: a case report and diagnostic challenges

Enfermedad de Creutzfeldt-Jakob probable en un contexto de recursos limitados: reporte de caso y desafíos diagnósticos

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ABSTRACT

Creutzfeldt-Jakob disease is a rare, transmissible, and invariably fatal spongiform encephalopathy caused by the accumulation of abnormal prion proteins in the central nervous system. We report the case of a middle-aged female patient from eastern Bolivia who presented with a six-month history of diplopia, progressive ataxia, generalized myoclonus, and rapidly progressive cognitive decline. A comprehensive diagnostic workup excluded infectious, metabolic, and autoimmune etiologies. Electroencephalography revealed periodic triphasic complexes, while brain magnetic resonance imaging demonstrated bilateral hyperintensities in the basal ganglia and cortical involvement, findings highly suggestive of Creutzfeldt-Jakob disease. According to the 2018 diagnostic criteria of the National Creutzfeldt-Jakob Disease Research & Surveillance Unit, the case was classified as probable prion disease. Neuropathological confirmation was not feasible due to logistical constraints. This case highlights the importance of considering prion diseases in the differential diagnosis of rapidly progressive dementias, particularly in low-resource settings where clinical assessment and neuroimaging play a central diagnostic role.

Keywords: Creutzfeldt-Jakob Disease; Prions; Dementia, Rapidly Progressive; Myoclonus; Magnetic Resonance Imaging.

RESUMEN

La enfermedad de Creutzfeldt-Jakob es una encefalopatía espongiforme transmisible, rara e invariablemente fatal, causada por la acumulación de proteínas priónicas anómalas en el sistema nervioso central. Se presenta el caso de una paciente femenina de edad media, procedente del oriente boliviano, con un cuadro de seis meses de evolución caracterizado por diplopía, ataxia progresiva, mioclonías generalizadas y deterioro cognitivo rápidamente progresivo. El abordaje diagnóstico incluyó un exhaustivo descarte de etiologías infecciosas, metabólicas y autoinmunes. El electroencefalograma mostró complejos periódicos trifásicos, mientras que la resonancia magnética cerebral evidenció hiperintensidades bilaterales en ganglios basales y compromiso cortical, hallazgos altamente sugestivos de enfermedad de Creutzfeldt-Jakob. De acuerdo con los criterios diagnósticos del National Creutzfeldt-Jakob Disease Research & Surveillance Unit, el caso fue clasificado como una prionopatía probable. La confirmación neuropatológica no fue posible por limitaciones logísticas. Este reporte resalta la importancia de considerar esta entidad dentro del diagnóstico diferencial de las demencias rápidamente progresivas, especialmente en contextos con recursos diagnósticos limitados, donde la correcta interpretación clínica, electroencefalográfica y de neuroimagen resulta fundamental.

Palabras clave: Enfermedad de Creutzfeldt-Jakob; Priones; Demencia Rápidamente Progresiva; Mioclonía; Resonancia Magnética.

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INTRODUCTION

Creutzfeldt–Jakob disease (CJD) is a transmissible, neurodegenerative, and invariably fatal spongiform encephalopathy caused by the accumulation of abnormal prion proteins in the central nervous system. It was first described by Hans Gerhard Creutzfeldt (1920) and Alfons Jakob (1921) as a form of subacute dementia associated with spongiform changes in the cortex and basal ganglia.[1,2,3] Studies by Gajdusek and Zigas in the 1950s, on the basis of the description of *kuru* in Papua New Guinea, subsequently established the transmissible nature of these diseases.[4] In 1982, Stanley Prusiner formulated the prion hypothesis as an infectious particle composed exclusively of protein, a concept that revolutionized the understanding of these pathologies and earned him the Nobel Prize.[5]

From an epidemiological point of view, CJD has an estimated global incidence of 1 to 2 cases per million inhabitants per year, with the sporadic form being the most common (approximately 85–90 % of cases), followed by genetic, iatrogenic, and acquired variants.[2,6,7] Although it is a rare disease, it is among the leading causes of rapidly progressive dementia (RPD) in adults, with a median survival of less than one year from symptom onset.[8,9,10] In Europe and North America, there are well-established surveillance systems, such as the National Creutzfeldt–Jakob Disease Research & Surveillance Unit (NCJDRSU) in the United Kingdom, which have made it possible to standardize clinical, electroencephalographic, and neuroimaging diagnostic criteria.[11,12]

In Latin America, CJD continues to be underdiagnosed because of its low frequency, the limited availability of advanced confirmatory tests (such as RT-QuIC), and clinical overlap with other causes of PDD, especially infectious, metabolic, and autoimmune causes.[10,11] In Bolivia, the national scientific literature records only one previously published case, reflecting both the rarity of the entity and the diagnostic difficulties in resource-limited settings. This scarce local documentation leads to low initial clinical suspicion and delays appropriate diagnostic guidance in patients with accelerated neurological deterioration.

From a theoretical perspective, early recognition of CJD is essential for differentiating it from other potentially treatable causes of PDD, avoiding unnecessary interventions, and optimizing palliative care. In clinical practice, the combination of characteristic neurological manifestations (rapidly progressive dementia, myoclonus, ataxia, and visual disturbances), together with typical findings on electroencephalogram and magnetic resonance imaging, allows a high-probability diagnosis to be established even in the absence of neuropathological confirmation.[9,12] In regions such as Bolivia, where definitive studies are not always available, the correct application of clinical and paraclinical diagnostic criteria is particularly important.

The objective of this report is to describe the clinical, diagnostic, and paraclinical approach to a probable case of Creutzfeldt–Jakob disease, emphasizing clinical, electroencephalographic, and neuroimaging findings, as well as the usefulness of international diagnostic criteria in the context of limited resources, to contribute to the timely recognition of this entity at the national and regional levels.

CASE REPORT

A middle-aged female patient from the department of Beni (Bolivia), with no known neurological history, presented with a clinical picture of approximately six months of evolution, initially characterized by intermittent diplopia and progressive gait disturbance, followed by marked ataxia and sudden onset myoclonus, initially focal and later generalized, without a defined rhythmic pattern. The condition progressed rapidly, associated with cognitive impairment, behavioral changes with irritability, emotional lability, and episodes of fluctuating confusion, until a state of almost complete functional dependence was reached.

Upon admission, the patient was in a supine position, with little interaction with her surroundings. Neurological examination revealed spontaneous myoclonus in both upper extremities, repetitive flexion–extension movements in the right lower extremity, and asymmetric bilateral intentional tremors. She responded inconsistently to verbal stimuli, uttering monosyllables and alternating between episodes of crying and unmotivated laughter. She had horizontal nystagmus, with no focal motor deficit or meningeal signs. Osteotendinous reflexes were preserved, and no adenopathies were evident. From a systemic point of view, the patient was hemodynamically stable, afebrile, and hydrated.

Her personal history included a diagnosis of hyperthyroidism four years earlier, without regular treatment. She reported no history of cardiovascular risk, regular medication use, or known allergies. A relevant epidemiological finding was the regular consumption of wild animal meat from the region, as reported by family members. The patient had initially been evaluated in the city of Trinidad, where basic laboratory tests and a cranial CT scan were performed without conclusive findings, which is why she was subsequently transferred to the city of Cochabamba for specialized evaluation.

During her hospitalization, serial laboratory tests were performed, which revealed no relevant hematological, metabolic, or infectious abnormalities. A cerebrospinal fluid study revealed a clear appearance, preserved glucorraquia, and mild hyperproteinorraquia without pleocytosis. Bacterial cultures, tests for mycobacteria and fungi, viral panels, and serological tests for syphilis were negative. The immunological profile, including antinuclear antibodies, anti-DNA, complement, antiphospholipid antibodies, anti-Ro, and anti-La, was found to be within normal ranges, ruling out an autoimmune or paraneoplastic etiology.

The thyroid profile showed a slight increase in T4, which is why metabolic encephalopathy associated with thyroid dysfunction was initially considered, and thiamine supplementation was started without a clinical response. Given the unfavorable evolution and the ruling out of infectious, metabolic, and deficiency causes, the study was expanded to include autoimmune and atypical encephalopathies, including prionopathies. In a therapeutic trial, pulses of methylprednisolone were administered, and no neurological improvement was observed.

To rule out underlying neoplastic pathology, tomographic studies of the neck, chest, abdomen, and pelvis were performed, which revealed no pathological findings. An electroencephalogram subsequently revealed a pattern characterized by slow sharp wave-like triphasic periodic complexes, with a periodicity of less than two seconds, a finding highly suggestive of Creutzfeldt–Jakob disease

(figure 1).

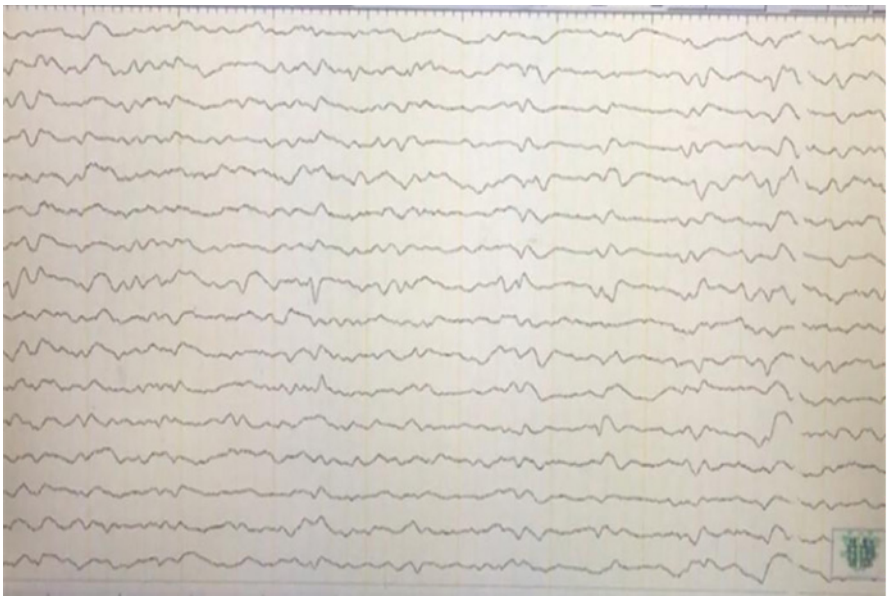


Figure 1. Electroencephalogram with characteristic triphasic periodic complexes

Brain MRI revealed areas of bilateral hyperintensity in the caudate nuclei, globus pallidus, and thalamus, as well as frontal cortical hyperintensity in the left hemisphere, which is consistent with the patterns described in prion encephalopathies (figure 2).

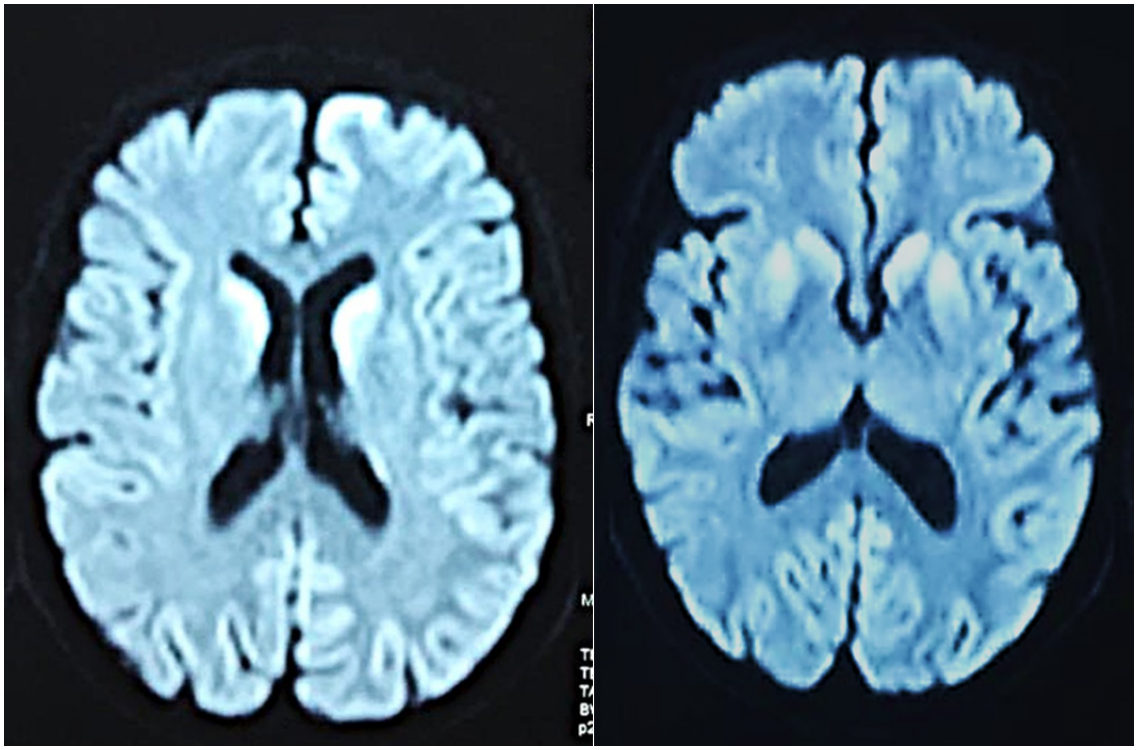


Figure 2. Brain MRI with hyperintensities in the basal ganglia and cortical involvement

Given the combination of a compatible clinical picture, characteristic electroencephalographic findings, and typical neuroimaging abnormalities, the diagnostic criteria of the UK National Creutzfeldt–Jakob Disease Research & Surveillance Unit (NCJDRSU), 2018, were applied, and the case was classified as probable Creutzfeldt–Jakob disease. A definitive diagnosis through neuropathological study could not be made because the patient was transferred to her place of origin, which prevented the collection of brain samples.

DISCUSSION

The case presented is clinically consistent with the sporadic form of Creutzfeldt–Jakob disease (CJD), the most common variant worldwide, and faithfully reproduces the classic triad described in the international literature: rapidly progressive dementia,

myoclonus, and ataxia, which is present in approximately 70–80 % of patients in advanced stages of the disease.[7,9] The subacute course, with irreversible cognitive decline of less than one year, is consistent with the clinical patterns reported in large European and North American series.[10,11,12]

From a diagnostic point of view, this case illustrates the value of a stepwise and systematic approach to rapidly progressive dementia (RPD), initially prioritizing the exclusion of potentially treatable etiologies, such as central nervous system infections, metabolic disorders, and autoimmune encephalopathies, before prionopathy is considered.[9,10,11] The absence of significant abnormalities in the cerebrospinal fluid, except for mild hyperproteinorraquia, is consistent with what is described in sporadic CJD, where inflammatory findings are usually absent.[12]

Electroencephalograms play a key role in the diagnostic orientation, revealing triphasic periodic complexes, which are highly specific in advanced stages of the disease, although with variable sensitivity depending on the clinical stage.[9,12] However, recent studies have reported that the diagnostic performance of EEG has progressively declined in the face of advances in neuroimaging and more sensitive molecular techniques.[10]

In this context, brain magnetic resonance imaging emerged as the main diagnostic pillar. The bilateral hyperintensities in the basal ganglia and cortical involvement observed in this case correspond to patterns widely described in sporadic CJD and have a sensitivity of more than 90 % in established clinical stages.[10,11,12] Several studies have shown that MRI, particularly when diffusion sequences are used, outperforms EEG in the early detection of disease and is a major criterion in current diagnostic systems.[9,12]

Although techniques such as RT-QuIC in cerebrospinal fluid have shown a specificity close to 100 % and a sensitivity greater than 90 %, their availability remains limited in many low- and middle-income countries, including Bolivia.[12] In this scenario, the correct application of standardized clinical and paraclinical criteria, such as those of the NCJDRSU 2018, is essential for establishing a high-probability diagnosis, as was the case in the present case.

A particular aspect of epidemiological interest is the history of consumption of wild animal meat, as reported by family members. Although this information does not allow us to establish a causal relationship or classify the case as an acquired variant, it is historically reminiscent of the transmission of kuru described by Gajdusek and Zigas.[4] This history should be interpreted with caution and only as a contextual element, without inferring unproven transmission mechanisms, especially in the absence of neuropathological or molecular evidence.

From a therapeutic point of view, this case reaffirms the absence of curative treatments for CJD. The lack of response to pulse corticosteroids reinforces the differential diagnosis against autoimmune encephalopathies, while management is inevitably oriented toward palliative and supportive measures, including control of agitation, myoclonus, and behavioral disorders, as recommended by international guidelines.[13,14]

The main limitation of this report is the inability to confirm the diagnosis by neuropathological study, which is considered the gold standard, because of the patient's transfer to her place of origin. Furthermore, advanced molecular tests such as RT-QuIC were not available, reflecting a common structural limitation in settings with limited diagnostic resources. However, these limitations do not invalidate the diagnosis of probable CJD, given the strict fulfillment of widely validated clinical, electroencephalographic, and neuroimaging criteria.

This case highlights the importance of considering Creutzfeldt–Jakob disease in the differential diagnosis of all rapidly progressive dementia, even in regions where it is rarely reported. The systematic application of international diagnostic criteria, supported by magnetic resonance imaging and electroencephalogram, allows for high-probability diagnoses to be established in the absence of advanced confirmatory tests. Strengthening clinical training in the PRD and promoting neurological surveillance networks at the national and regional levels are recommended, with the aim of improving the timely recognition of this entity, optimizing palliative management, and contributing to better epidemiological documentation of prion diseases in Latin America.

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None.

CONFLICT OF INTEREST

The authors declare that there is no conflict of interest.

AUTHOR CONTRIBUTION

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Writing – revision and editing: Arnaldo Zambrana Campana, Gaby Elizabeth Bordon Ortiz, Rommer Alex Ortega-Martinez.

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