

## Remembering Creutzfeldt Jakob Disease: Case Report

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### Abstract

Creutzfeldt-Jakob disease (CJD) is a rare and fatal neurodegenerative disorder and represents the most common prion human disease. It occurs at a rate of one case per million per year, with an onset age between 50 and 70 years and a median time to death of 5 months. The pathogenesis of sporadic CJD is represented by the conformational transition of a host-encoded cellular prion protein (PrP<sup>c</sup>) into an infectious and pathogenic isoform, which tends to propagate progressively into multiple regions of the brain. CJD remains a particularly challenging diagnosis for physicians, mainly because of its rarity and clinical heterogeneity. Common neurological features are cerebellar ataxia, rapidly progressive dementia, and/or myoclonus. Psychiatric manifestations, in particular psychosis, behavioral changes, depression, and sleep disturbances, are frequent in the early stages of the disease. Some tests can be helpful in providing support for diagnosis, will not changing the prognosis but will improving the care of patients.

**Keywords:** Creutzfeldt Jakob Disease (CJD); Prion Protein (PrP<sup>c</sup>)

### Introduction

Creutzfeldt Jakob disease (CJD) is a rare and fatal subacute spongiform encephalopathy. It has a nual incidence of 1:1 million patients, evolving to death between six months and one year. It affects patients, preferably, between the fifth and sixth decades and there is no discrimination regarding gender. Approximately 85% of cases belong to the sporadic form of the disease; the other 15% consist of the genetic form (Gerstmann-Sträussler-Scheinker-Scheinker syndrome). Clinically, it is characterized by a rapidly progressive dementia accompanied by myoclonus and pyramidal signs, however, the manifestations may be absent, making diagnosis difficult. The reason why we have recorded this case is due to its relatively low incidence.

### Case Report

Female patient, 68 years old, hospitalized by psychiatry due to differential. Family members report that the patient was previously healthy and independent, however, for 3 months she started with tremors, behavior alteration, psychomotor agitation and sleep disorders. In two months, he evolved with myoclonic s crisis and signs of pyramidal release, submitted to cranial MRI that showed diffuse cerebral atrophy and EEG presenting periodic complexes with bilateral diffuse projection. Protein 14-3-3 was performed in the CSF with positive result. The patient died after 5 months of evolution.

### Discussion

Transmissible spongiform encephalopathies (TSE) are rare neurological disorders that maintain similarity to neurodegenerative diseases. TSE is transmitted by a prion protein that accumulates in the CNS leading to rapidly progressive dementia, cerebellar syndrome, myofasciculation, mutism, visual disturbances, and pyramidal/extrapyramidal signs. Regarding the diagnosis, the EEG shows periodic complexes of the tipon, bi or triphasic stupendous that appear more frequently in the first three months of the disease. The detection protein 14-3-3 in the CSF has a specificity of 99%, however, this protein is not pathognomonic. Some cranial MRI present a hypersignal on T2-and-long diffusion, located in the deep cortical regions and nuclei of the base. Biopsy of the pharyngeal tonsil is described as an adjunct procedure in the diagnosis of the disease in more advanced phases, due to prion replication in the lymph for reticular system. Brain biopsy and immunohistochemistry are considered the gold standard for diagnosis. There is no cure for CJD [1-4].

### Conclusion

Creutzfeldt-Jakob disease is an uncommon and fatal disease; however, early diagnosis can enable the implementation of an important palliative care program.

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