

CASE REPORT

Sporadic Creutzfeldt-Jakob disease

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Abstract

Creutzfeldt-Jakob disease (CJD) is a rare disease that belongs to the category of transmissible spongiform encephalopathies. The condition is invariably fatal and progresses with severe dementia with psychiatric signs and symptoms, as well as cortical, subcortical and cerebellar signs. We present a case of a woman aged 54 years, who developed CJD without risk factors for the disease. The usual manners of transmission could not be confirmed for this patient, who died ten months after the onset of symptoms. CJD is also a serious health care concern, since patients who are still asymptomatic may be donating blood for transfusions or for blood products.

Key words: creutzfeldt-jakob disease, encephalopathy, prion.

INTRODUCTION

Transmissible spongiform encephalopathies are fatal neurodegenerative diseases that affect mammals including humans. Creutzfeldt-Jakob disease (CJD) belongs to a group of these encephalopathies and is caused by abnormal folding of a host-based cellular prion protein PrP.¹ CJD is the commonest form of human prion disease, leading to severe neurodegeneration and invariably death. The neuropathology of CJD consists of spongiform change, astro- and microgliosis, and neuronal loss occurring diffusely in the brain.² CJD is rare, poorly understood and leads to serious public health concerns,³ since it is highly infectious. For example, use of human tissue in surgery, such as dura mater grafts, has been shown to be strongly associated with development of CJD in the patient receiving the infected tissue.⁴ Other forms of iatrogenic contamination include contaminated growth hormone derived from human cadavers with undiagnosed CJD infections, and a small number of additional cases may have been caused by neurosurgical instruments.⁵ When a patient with suspected CJD has a clear history of possible contamination with infected tissue, the diagnosis may be easier. However, for sporadic cases of CJD, in which there is no clear

evidence for the origin of the infection, the diagnosis may be very difficult. In fact, patients with CJD are commonly diagnosed as having viral encephalitis, paraneoplastic disorders, depression, vertigo, Alzheimer disease, stroke, unspecified dementia, central nervous system vasculitis, peripheral neuropathy and Hashimoto encephalopathy.⁶ Here, we present a case of CJD in a woman who had never travelled abroad or undergone neurosurgery or any other procedure with an imminent risk of CJD, and who had been a vegetarian for many years.

CASE REPORT

Ethical considerations: a relative of the patient acting as her legal guardian authorized the use of this case for scientific purposes provided confidentiality was guaranteed.

The patient was a 53-year-old Caucasian female who was a primary school teacher. Ten months before admission, she started to develop moments of apathy and aggression. She progressed with global cognitive deficit, impaired coordination and balance. The clinical symptoms developed over a three-week period. After admission, her condition continued to progress, with development of neurological signs and symptoms. Myoclonus was also observed. Verbal and written expression aphasia, inability to perform simple calculations, cog-wheeling rigidity and cerebellar signs progressed continuously over a period of four weeks. The medical condition

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worsened with severe apathy, dementia and pyramidal signs in all limbs.

Her cerebral spinal fluid was normal, except for the positive 14-3-3 protein immunoblot. Her brain magnetic resonance imaging (MRI) showed severe cortical and subcortical atrophy of the brain and cerebellum. Her electroencephalogram showed delta waves as the basic rhythm with bursts of peak-waves lasting two to four seconds in all areas of the brain.

Her diagnosis of CJD was confirmed by the clinical signs and symptoms, the laboratory and imaging examinations, and the polymorphic codon mutation V129v that is typical of classic CJD.

Over the course of ten months, the patient's condition became progressively worse, culminating in death due to pneumonia and sepsis. Her brain presented typical spongiform disease, as shown in Figure 1.

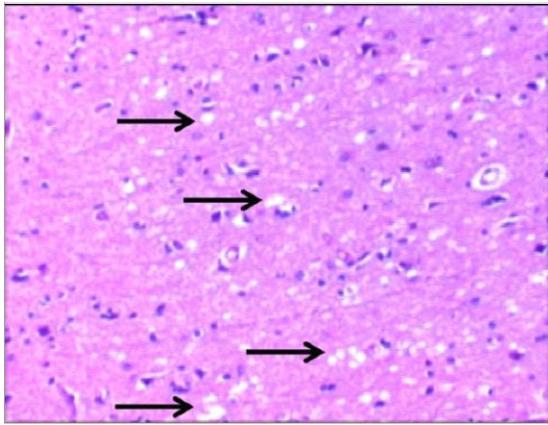


Figure 1 Anatomic- pathological aspect of the brain in a patient with Creutzfeldt-Jakob disease. Arrows point to some areas of neuronal loss and spongiform appearance.

DISCUSSION

CJD is a rare and fatal condition, for which there is no treatment. The polymorphic codon mutation V129v found in our patient is associated with classical CJD with a severe phenotype⁷, but can be found in 10% of the normal population as well. Differential diagnosis with other forms of degenerative neurological diseases is imperative, since CJD can be infectious. There is good evidence for transmission of prion disease among human beings through blood, blood components, several

tissues, growth hormone and even fertility treatments.⁸ Due to the difficulties of testing for prion diseases in asymptomatic individuals and the lack of absolute biomarkers for the disease, measures like prion filtration may be necessary, at least when dealing with blood.⁹ However, despite all possible care that may be implemented in the coming years, sporadic cases such as the one described above continue to appear and must be promptly recognized. Reports of original cases like this one are important to maintain awareness of the disease in the population.

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