

CREUTZFELDT-JAKOB DISEASE

Creutzfeldt-Jakob disease (CJD) is a rare type of dementia that is produced by an infectious particle called a prion. The term "prion" was coined in 1982 and stands for proteinaceous infectious particle lacking nucleic acid. This neurological disease occurs in approximately one per million persons. The typical clinical features of Creutzfeldt-Jakob disease include a rapid progression, i.e., severe dementia in less than 18 months, myoclonic jerks, and an abnormal electroencephalogram, i.e., brain wave test.

The presenting symptoms of Creutzfeldt-Jakob (CJD) are typically subtle. Approximately 1/3 of patients demonstrates vague symptoms such as fatigue, poor sleep, and diminished appetite. Another 1/3 develops non-focal neurological symptoms such as confusion, memory loss, or abnormal behavior. The symptoms progress and the usual duration of survival is approximately five months. The EEG pattern shows a characteristic periodic bi-phasic or tri-phasic spiking complex superimposed on a slow background rhythm. EEG abnormalities are present in up to 90% of patients if sufficient numbers of tracing are performed. Myoclonis, i.e., jerking motions of the extremities are present in most patients.

A variant of Creutzfeldt-Jakob disease (CJD) is present in Great Britain that some researchers believe may result from eating beef from cattle that have bovine spongiform encephalopathy (BSE). Patients with the variant Creutzfeldt-Jakob (v-CJD) are typically younger persons who frequently present with psychiatric symptoms that may mimic schizophrenia. These individuals tend to survive longer than Creutzfeldt-Jakob (CJD) patients, i.e., 14 months for v-CJD versus five months for CJD. Both types of patients develop terminal symptoms that include mutism and an akinetic state that requires intensive nursing care.

Scientists poorly understand the prion that produces Creutzfeldt-Jakob (CJD). The specific molecular biological causes of prion-mediated diseases are unknown. Brain damage results from aberrant conversion of a normal cellular protein into an abnormal protein, i.e., PrP^{sc}, within the brain. This abnormal protein has a different shape, i.e., folding, than its normal brain counterpart, and this abnormal folding propagates itself through brain tissue. Prions cause a variety of brain disorders in humans and other species. Mad Cow disease, i.e., the prion-mediated disorder termed “bovine spongiform encephalopathy (BSE)” closely resembles Creutzfeldt-Jakob disease. Scrapie is a similar disorder in sheep. These non-human prions are transmitted by consumption of brain tissue used in animal feeds and share many biological similarities. The treatment strategies for prion disease focus on prevention. Public health experts focus on avoidance of high-risk materials such as brain or spinal cord from cows. Future therapies will include compounds that block the abnormal protein foldings or the message to fold.

The mode of spread for prions in humans is unclear, except by infected tissue from the human nervous system. Spinal fluid, brain tissue, pituitary tissue, and infected human corneas are able to transmit the prion. There is no documented transmission by human blood, saliva, urine, or other bodily excretions, although universal precautions should always be used for a patient with Creutzfeldt-Jakob disease. Standard disinfection methodologies are not effective against prions. Surgical personnel who perform brain biopsies on patients with suspected prion disease should consult with infection control specialist to assure adequate sterilization of neurosurgical instruments. Standard fixation techniques such as embalming, and formalin fixation do not inactivate this infectious particle. Funeral home staff are often hesitant to embalm persons with CJD; however, there is no documented transmission to mortuary staff. Neuropathologists and autopsy staff must take special precautions to prevent the spread of this disease by autopsy contamination. Several reports document spread by non-brain tissue including

dural flaps and pericardial tissue; however, these materials may have include nervous tissue.

There is no known treatment for Creutzfeldt-Jakob's disease. Standard antibiotic and antiviral medications do not affect the prion. A spinal fluid test is available that identifies the prion protein (PrPsc); however, this test is not 100% sensitive. Physical examinations, neurological assessments, or brain imaging studies cannot confirm Creutzfeldt-Jakob's disease. Brain biopsy or autopsy is required to confirm the diagnosis.

Creutzfeldt-Jakob (CJD) is a rare cause of dementia that attracts considerable media attention. This prion-mediated dementia may be related to mad cow disease. The typical CJD patient has a short survival and the mode of transmission is unknown. The risk to the general population is quite low.

REFERENCES

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