

Creutzfeldt-Jakob Disease

Definition

Creutzfeldt-Jakob Disease (CJD) is a rare, rapidly progressive, fatal, degenerative disorder of the central nervous system. Death often occurs within six to 12 months from the onset of symptoms; however, in some cases, the course is longer and can be mistaken for Alzheimer's disease. This illness is both mysterious and complex in its origins and presentation. Due to its rarity and rapid course, clinicians, affected individuals and families are stunned by its fatal outcome.

CJD belongs to a group of transmissible spongiform encephalopathies (brain degeneration without inflammation, resembling a sponge) affecting both humans and animals alike. Related diseases in humans include Gerstmann-Sträussler-Scheinker disease (GSS), fatal familial insomnia (FFI) and kuru. In animals, the spongiform encephalopathies appear as scrapie in sheep, chronic wasting disease in elk and deer, transmissible mink encephalopathy, and bovine spongiform encephalopathy (BSE) in cows, also termed "mad cow disease." BSE first appeared in Britain's herds over 10 years ago and scientists think cattle contracted the disease by eating feed contaminated with scrapie-infected sheep tissue added as a protein supplement.

Cause

Current theory and research suggests that a "proteinaceous infectious particle" or prion plays a fundamental role in the development of Creutzfeldt-Jakob and other prion diseases. Prions are thought to transform normal, benign protein molecules into infectious, deadly ones by altering the shape of the healthy molecules, setting off a chain reaction that results in brain cell death. The cause of this is still unknown and a source of great speculation for those who study the prion diseases.

Modes of Transmission

Sporadic (sCJD): This is the most common type and occurs at random, with no apparent predisposing factors. Approximately one in 1 million people are affected with the disease each year, with no regard to gender. It occurs most frequently between the ages of 55 to 65, although both younger and older persons have been affected.

Acquired or iatrogenic (iCJD): Presently, the only proven manner for contracting CJD from an infected person is through exposure to infected brain or spinal cord tissue or blood. Transmission through this means is rare but has occurred in some cases involving corneal transplants, implantation of electrodes in the brain, and contaminated surgical equipment.

Familial (fCJD): The inherited form of CJD is rare, comprising approximately 10 percent of cases. The disease is 60 times more common than usual among inhabitants of one region of Slovakia, and 40 times higher among Libyans who immigrated to Israel many years ago. These familial cases exhibit a mutation in the gene coding for the prion protein (PRNP).

Variant (vCJD): In the mid-1990s in England, a significant number of cases of the new variant CJD (vCJD) were identified and diagnosed in a group of young individuals. These cases raise the possibility that BSE or "mad cow disease" can be transmitted to humans if they ingest infected beef. Variant CJD (vCJD) has never been reported in the USA.

Clinical Features

CJD has the same cognitive losses (short-term memory loss, difficulty with calculation) that accompany other irreversible dementing illnesses, but individuals may also present with other symptoms such as:

- Myoclonus (involuntary jerking movements)
- Visual disturbances
- Motor dysfunction (balance difficulty while walking)
- Dramatic psychiatric symptoms (paranoia, thought disorders, agitation)
- Heightened startle reflex
- Advanced symptoms may include: dementia, akinetic mutism (inability to speak or move), seizure activity, abnormal posturing, coma, and eventually death caused by secondary infection or pneumonia.

Diagnosis

The probable diagnosis is made by clinical presentation of symptoms, the rapidly progressive course, along with a comprehensive neurological examination. There are tests that support the diagnosis but there can only be a definitive diagnosis in life with a brain biopsy and after death at autopsy. Brain biopsy is not recommended in most situations.

Treatment

There is no cure for CJD. The treatment focus is on providing medical, environmental and emotional supports to the affected person, as well as education and emotional support to caregivers. It is a devastating disease that affects the family profoundly. Therefore, it is imperative that clinicians be available for education and supportive care from the onset of the disease well into the bereavement period.

Family and Health Provider Concerns

There has been no evidence that CJD is transmitted by human contact. Health care providers must always practice Universal Precautions when caring for any individual. Such is the case with a person who has CJD or other prion diseases. However, health care workers must be aware that this disease is resistant to traditional modes of sterilization and disinfectant. Therefore, equipment that comes in contact with hazardous wastes such as blood, spinal fluid, cerebral nervous tissue must be handled and disposed of cautiously and within the guidelines set forth by the Centers for Disease Control.

Important resources:

For information and support www.cjdfoundation.org

For tracking of CJD and other prion diseases in the United States: The National Prion Disease Pathology Surveillance Center (supported by the Center of Disease Control and Prevention).

www.cjdsurveillance.com

Please call Clarissa Rentz MSN, APRN at (513) 721-4284 for more information about diagnosis, CJD Surveillance, and support services for individuals and families dealing with this and other dementing illnesses. (References/bibliography available upon request.)