CREUTZFELDT-JAKOB DISEASE FOUNDATION, INC.

Supporting Families Affected by Prion Disease

CJD FACT SHEET

What is CJD?

Creutzfeldt-Jakob Disease (CJD) is a rare, fatal brain disorder within a group of illnesses called prion diseases. The incidence of CJD cases worldwide is one to two cases, per million individuals, per year. In the United States this statistic translates to nearly 500-600 new cases per year. There is one CJD death per every 6,000 deaths in the U.S. each year. Eighty-five percent of CJD cases are sporadic, meaning there is no known cause.

There are three causes of prion disease:

- ***Sporadic*** (sCJD)
- ***Genetic*** (also known as familial)
- ***Acquired*** (variant CJD; vCJD and iatrogenic CJD; iCJD)

In the early stages of the disease, CJD patients may exhibit failing memory, behavior changes, impaired coordination, and/or visual disturbances. As the illness progresses, mental deterioration becomes more pronounced, and involuntary movements, blindness, weakness of extremities, and, ultimately, coma may occur. CJD usually occurs later in life and typically leads to death within a few months to one year following the onset of symptoms.

Types/Causes of CJD

Sporadic CJD

- **Cause unknown** — the normal prion protein spontaneously misfolds into a disease-causing form and induces other normal prion proteins around it to misfold, in a similar pattern to other protein-related neurodegenerative diseases such as Alzheimer's or Parkinson's disease. The misfolded form of the prion protein also causes damage to brain cells.

Genetic Prion Disease

- Caused by a genetic mutation of the prion protein gene
- There are nearly 60 prion disease mutations, which cause varying symptoms, including the following diseases:
  - **Genetic** (e.g., familial) CJD (gCJD)
  - **Fatal Familial Insomnia** (FFI)
  - **Gerstmann-Sträussler-Scheinker Disease** (GSS)

Iatrogenic CJD has been transmitted by:

- Contaminated neurosurgical instruments
- Contaminated dura mater transplants obtained from cadavers
- Contaminated corneal transplants
- Contaminated human growth hormone obtained from cadavers

Variant CJD has been transmitted by:

- Beef contaminated by bovine spongiform encephalopathy (BSE) (or mad cow disease)
- Contaminated blood or blood plasma transfusion from individuals with vCJD

The sporadic form of CJD is the most prevalent form of CJD affecting nearly 500-600 new people in the United States each year. The genetic form of prion disease accounts for 10-15% of cases. Acquired CJD, which includes iatrogenic and variant CJD, accounts for less than 1% of all cases. As of 2024, there are no known cases of vCJD that have been acquired within the United States. More information can be found on **www.cjdfoundation.org**.

Diagnosis of prion disease is challenging and is often made from clinical observation and eliminating the possibility of other diseases. Key clinical diagnostics for prion disease include electroencephalogram (EEG), brain MRI, and cerebral spinal fluid (CSF) tests including Real-Time Quaking Induced Conversion (RT-QuIC) as well as tests for 14-3-3 and total tau proteins. The diagnosis of CJD, as well as its cause, can only be definitively confirmed through a brain biopsy or autopsy. (If CJD is

suspected, brain biopsy is not recommended.)

Autopsy determines the type of prion disease and whether there is a genetic mutation. One of the most important parts of diagnosing prion disease, however, is ruling out other treatable conditions. For more information, call **800-659-1991.**

Public Health Concerns

CJD is NOT "Mad Cow Disease." Bovine Spongiform Encephalopathy (BSE), the technical term for Mad Cow Disease, occurs only in cows. The first documented case of BSE found in the United States occurred in Washington State in December 2003 in a cow imported from Canada. The first endemic case found in cattle was announced in 2005 in Texas.

Eating beef contaminated with BSE can cause the variant form of CJD (vCJD) in humans. vCJD usually affects young people. No cases of vCJD have been thought to have been acquired within the United States.

A deeper understanding of all forms of CJD would provide vital direction to scientists in studying prion disease, improve public health, and support the ultimate goal of finding a cure. In particular, brain autopsy for suspected CJD patients is critical to learning where the disease originates, how it progresses, and how to treat it.

Funerals

There are no special interment, entombment, inurnment, or cremation requirements for patients with CJD. Interment of bodies in closed caskets does not present a significant risk of environmental contamination and cremated remains can be considered sterile, as the infectious agent does not survive incineration-range temperatures. Information for funeral and crematory practitioners is available on the CDC website and from the CJD Foundation and National Prion Disease Pathology Surveillance Center.

Chronic Wasting Disease (CWD)

Chronic Wasting Disease (CWD), a prion disease in deer and elk (i.e., cervids), appears to be environmentally contagious among members of its own species. First recognized in 1967 in Colorado, CWD has since spread from 2 states in 2000 to 32 states in 2024. Affected prions from the body fluids of deer and elk have been shown to be able to be taken up into the roots and leaves of plants as well as soil. Therefore, fields contaminated by a cervid herd can later spread CWD to other herds.

CWD has not been proven to cross the species barrier. However, studies in progress seem to demonstrate the potential for CWD from the brain and meat of affected cervids to be transmitted to swine and macaques (monkeys) through feeding. Health and wildlife authorities continue to monitor and study this possible threat to public health.

Blood Safety Update

In 2023 the American Red Cross updated blood donation recommendations.

***Questions?** Contact Red Cross Donor and Client Support Center:* **1-866-236-3276**

Ineligible: Persons who have been diagnosed with vCJD, CJD or other prion diseases, or have a blood relative who has been diagnosed with a genetic form of prion disease. Persons who received an injection of growth hormones derived from human pituitary glands before April 1985. Persons who received a human cadaveric dura mater transplant.

Updated Eligibility: Donors who were previously deferred if they spent time in the UK, France, Ireland, or they received a blood transfusion in the UK, France, or Ireland may now be eligible if they meet all other eligibility criteria. Persons who have a blood relative who was diagnosed with sporadic prion disease are now eligible if they meet all other eligibility criteria.

The CJD Foundation

HELPLINE: 1-800-659-1991

help@cjdfoundation.org

cjdfoundation.org

We are available to help you with questions and concerns Monday through Friday, 9 am to 5 pm ET. Messages left on our voicemail after hours concerning patients will be returned evenings and weekends.