

Human Prion Disease in Washington State, 2012–2021

Human prion disease

Prion diseases, also referred to as transmissible spongiform encephalopathies (TSE), are a rare group of progressive neurodegenerative disorders that can occur in humans and animals. Prion diseases can be sporadic, inherited, iatrogenic, or acquired.

Creutzfeldt-Jakob disease (CJD) is the most common human prion disease. It is a rare, fatal disease characterized by rapidly progressing dementia, poor balance, visual changes and/or muscle jerks. Sporadic CJD (sCJD) has no known cause and accounts for about 85% of all CJD cases. Familial CJD (fCJD) results from an inherited mutation and accounts for 10–15% of cases. Other inherited prion diseases include Fatal Familial Insomnia (FFI) and Gertsmann-Straussler-Scheinker syndrome (GSS). In 1996, a new variant CJD (vCJD) recognized in the United Kingdom was associated with eating cattle products from cows affected with bovine spongiform encephalopathy (“mad cow disease”). *To date, no cases of variant CJD are thought to have been acquired in Washington or the United States.*

Iatrogenic transmission of CJD has been linked to the use of contaminated human growth hormone, dura mater and corneal grafts, or neurosurgical equipment. All of the equipment-related cases occurred before the routine implementation of sterilization procedures currently used in health care facilities. No equipment-related cases have been reported since 1976. In the United States, 29 iatrogenic CJD cases have been linked to the use of pituitary human growth hormone (hGH) in patients treated before 1977. The growth hormone now used for treatment poses no threat of infection with CJD.

The only currently available method of confirming the diagnosis of prion diseases is the pathologic examination of brain tissue (autopsy or biopsy). Clinical symptoms in conjunction with some non-confirmatory diagnostic tests (14-3-3 protein in cerebrospinal fluid, MRI, and EEG) are used to make a *pre mortem* clinical diagnosis of probable CJD. In 2018, the Centers for Disease Control and Prevention (CDC) updated the CJD diagnostic criteria to incorporate positive real-time quake-induced Conversion (RT-QuIC). For sporadic, familial, iatrogenic, and variant CJD case definitions please see: <http://www.cdc.gov/prions/cjd/diagnostic-criteria.html> and <http://www.cdc.gov/prions/vcjd/diagnostic-criteria.html>

Human prion disease in Washington State

Beginning in 2004, the Washington State Department of Health (DOH) has been collaborating with CDC and the National Prion Disease Pathology Surveillance Center (NPDPSC) for the purpose of identifying and confirming prion disease in Washington State. Healthcare providers in Washington are required to report suspected human prion disease to the local health jurisdiction for the patient’s county of residence.

During 2012–2021, 143 cases of prion disease were detected in Washington (average 14 cases per year). Five cases (4%) were inherited prion disease (4 fCJD cases and 1 GSS case). One case of hGH-related iatrogenic CJD was reported in 2013. Most cases were sporadic CJD (136 or 95%), including one case of Variably Protease-Sensitive Prionopathy (VPSPr) in 2012. Of the sporadic prion disease cases (sCJD and VPSPr), 83 (58%) were tissue confirmed, and 53 (37%) were clinically diagnosed and did not undergo confirmatory autopsy or biopsy testing. All clinically diagnosed patients had a presentation consistent with sporadic CJD. Fifteen patients were less than 55 years old at the time of death. Of these, 5 were confirmed sCJD, 2 were confirmed fCJD, 1 was confirmed GSS, and 7 were clinically diagnosed sCJD.

Table 1. Characteristics of prion disease cases, Washington State, 2012–2021 (n=143)

Characteristic	No. Cases (%)
Male	82 (57%)
Median age [interquartile range]	68 years [37–90 years]
Median duration of illness [interquartile range]	4 months [1–70 months]
Average incidence	1.91 cases/million population*
Autopsy and/or biopsy performed	90 (63%)

*Worldwide incidence of human prion diseases is approximately 1–2 cases per million population per year.

The following graphs show the number of CJD cases by type and year of death, and by case classification and year of death in Washington State during 2012-2021.

Figure 1. CJD cases by type and year of death

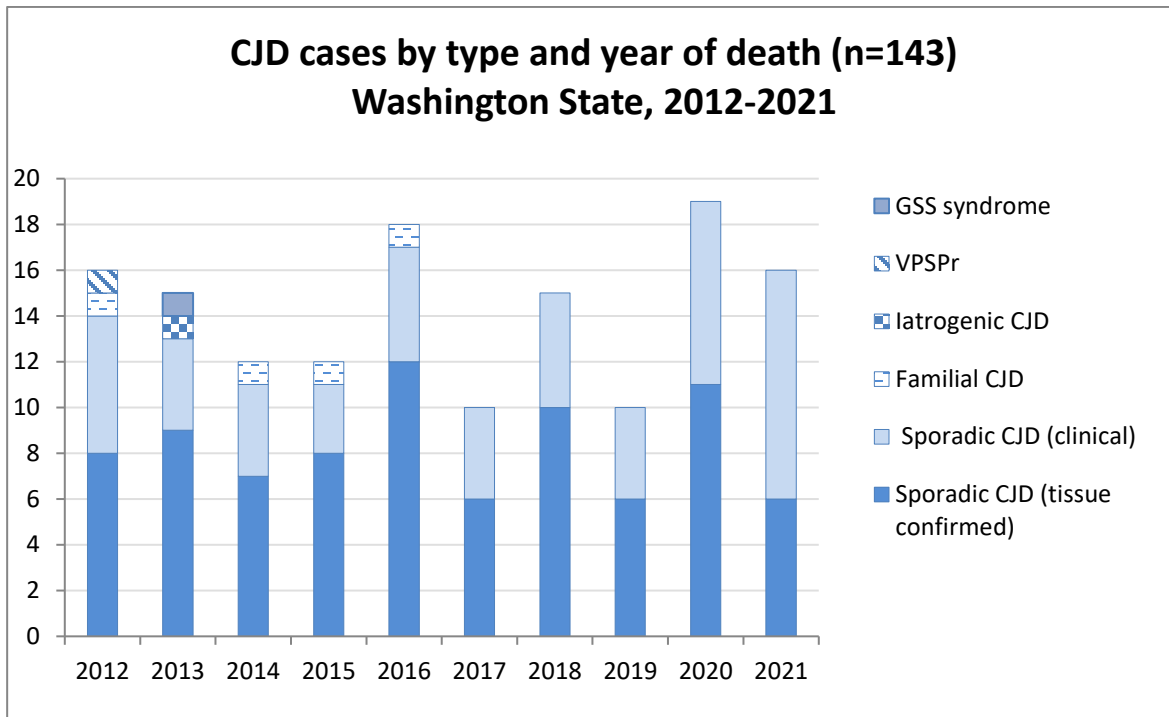
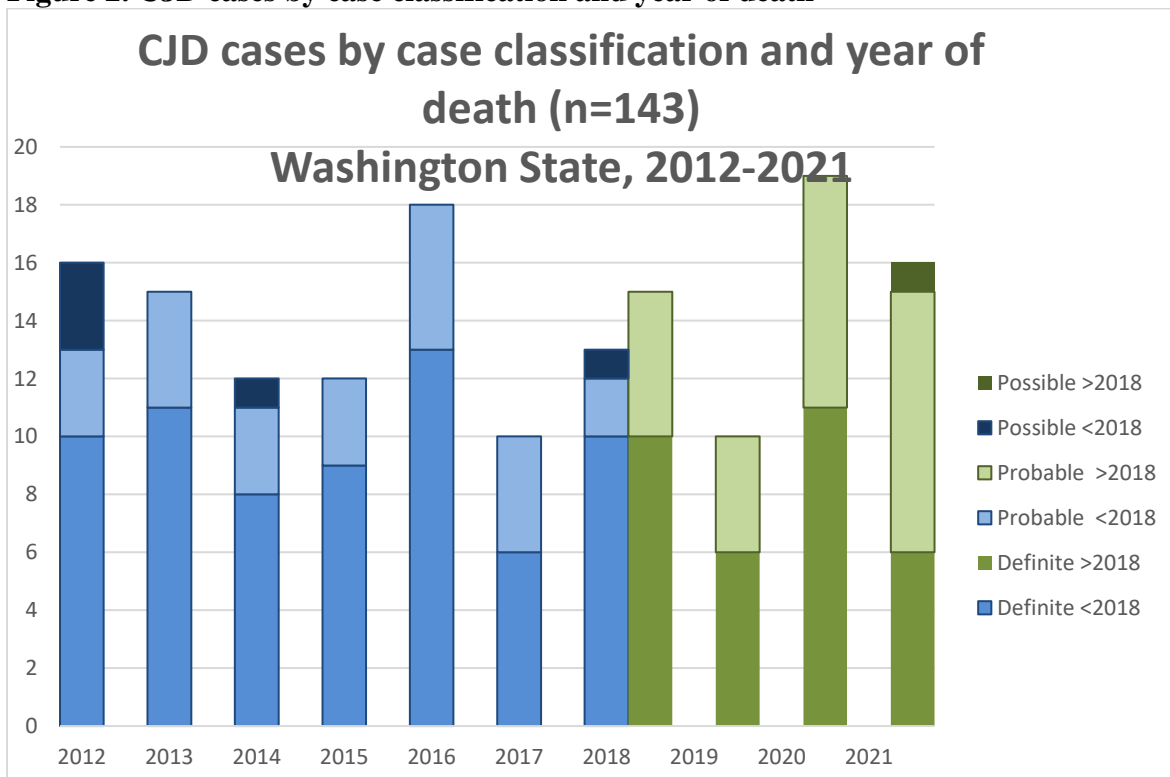


Figure 2. CJD cases by case classification and year of death



* In 2018, CDC updated the CJD diagnostic criteria to incorporate positive real-time quake-induced Conversion (RT-QuIC). Here 2018 data are represented with cases meeting the criteria using the updated 2018 criteria and cases meeting the previous criteria.

Summary: The incidence of human prion disease in Washington State is consistent with reported rates worldwide. During 2011–2020, 61% of sporadic CJD cases, and 63% of all prion disease cases were confirmed by examination of brain tissue. No variant CJD was diagnosed.