

Questions and Answers about Creutzfeldt-Jakob Disease Surveillance and Reporting

Brief Background Information:

Creutzfeldt-Jakob Disease (CJD) is a fatal brain disease that is caused by a novel protein called a prion. The annual incidence of CJD has been reported as 1 case per million. There are no known causes of 80% of the cases of CJD, which are referred to as sporadic CJD (sCJD). A familiar or inherited form of the disease comprised about 15% of the cases and 5% of the cases are iratogenic. Iratogenic cases have been linked to use of human pituitary extracts, dura mater transplants, contaminated EEG electrodes and neuro-surgical instruments. In 1996 a variant of CJD (vCJD) was described in the United Kingdom and linked to the Bovine Spongiform Encephalopathy (Mad Cow Disease), which was epidemic in the UK. There are several differences between sCJD and vCJD. Important differences include the young age of the variant (mean 26 years old) as compared to the sCJD (mean 69 years old). In addition the individuals with the variant form of the disease live longer (median 14 months) than those with the sporadic form (median 4 months).

CJD became a reportable in Florida in June 2003. Prior to 2003 CJD has been monitored through the use of death certificates and pathology reports.

The Florida Department of Health web site has additional detail on CJD and links to many other sites.

Reporting of Creutzfeldt-Jakob Disease (CJD)

The CJD Case report form

The CJD case report form is designed to collect information that is used to meet the CJD case definition, to distinguish between sporadic, familiar, iratogenic and variant forms of the disease.

Hints for completing the case report form and reporting CJD:

SSN: The social security number can be found both in the medical record and on the death certificate.

Outcome: Most of the patients will have died by the time of reporting, please enter the date of death on the case report form in addition to recorded the outcome as died in Merlin.

Imported: Should be entered as unknown. If a person had received human growth hormone, a dura mater transplant, had previous neurosurgery, lived in a county where vCJD is found or had other risk factors for acquiring the disease the case should be investigated in detail and discussed with the lead epidemiologist investigating CJD in the Bureau of Epidemiology.

Date Onset: Frequently the exact date of onset for CJD is unknown. However it is important to determine the data as accurately as possible.

DX Status: See the following case definition.

The following information pertains to the questions on the CJD case report form.

Akinetic mutism –loss of the voluntary ability to speak due to an extrapyramidal disorder

Ataxia – Frequently patients will have problems maintaining their balance and other difficulties with coordination early in the disease process. Ataxia will progress and the patient will lose the ability to walk.

Dementia – The development of dementia in CJD patients is very pronounced over a short period of time (days or weeks) unlike dementia associated with Alzheimer’s Disease.

Dysesthesia and painful sensory symptoms – Frequently patients complain of a new onset of pain or other disagreeable sensations in extremities or other areas not related to injury or stimulation.

Visual signs – refers to changes in vision not due to structural problems typically associated with vision loss

Myoclonus - refers to sudden, involuntary jerking of a muscle or group of muscles

Chorea – irregular, spasmodic, involuntary movements of the limbs or facial muscles, often accompanied by hypotonia

Dystonia- A state of abnormal (either hypo- or hyper-) tonicity in any of the tissues resulting in impairment of voluntary movement

Progressive neuropsychiatric disorder – In the variant form of the disease the patients have a progressive neuropsychiatric disorder lasting at least 6 months. In the sporadic form if neuropsychiatric disorders are present they usually occurs at or around the same time a physical manifestations of the disease.

Early psychiatric symptoms – In variant CJD the first symptoms of the diseases are psychiatric.

EEG – The EEG characteristic of CJD is often described as slow and triphasic in nature. A copy of the report should be attached to the case report form.

MRI scans - The MRI scan is often reported as normal, or with mild atrophy changes. A MRI reporting abnormality bright basal ganglia is frequently found in sporadic CJD. MRI scans are useful in ruling out other etiologies such as tumor or stroke. A copy of the report should be attached to the case report form.

14-3-3 CSF protein analyses – This laboratory test has a high degree of sensitivity and specificity for CJD, but by itself is not diagnostic. A few specialty laboratories perform the test. The National Prion Disease Pathology Surveillance Center at Case Western Reserve University is the preferred laboratory to send CSF for 14-3-3 testing. A copy of the report should be attached to the case report form.

Alternative non-CJD diagnosis – Cancer, stroke, infection and other diagnosis can result in symptoms similar to CJD. Test should be done to rule out a treatable etiology.

Past Medical and Social History – This series of question helps determine if there are risk factors for acquiring CJD and if there are risks for transmitting the disease.

Neuropathology Information – This information is required to confirm the diagnosis. If the patient is alive at the time of reporting effort should be made to arrange for an autopsy prior to death. The National Prion Disease Pathology Surveillance Center can assist in making arrangement for an autopsy and sending specimens to their laboratory.

Death Certificates: Death certificates should be obtained for all patients. These can be obtained at the county health department vital statistics office. If death certificates are incorrect they should be amended. The physician that signed the original death certificate has to sign the form to amend the death certificate. The form to amend a death certificate can be obtained at the local vital statistics office.

Case Definition for: **Creutzfeldt-Jakob Disease (CJD)** reporting code = 04610

Case report form: N/A

A progressive uniformly fatal dementia characterized by: Myoclonus, visual or cerebellar signs, akinetic mutism and pyramidal or extrapyramidal signs,

Laboratory criteria for diagnosis

- Standard neuropathological techniques; and/or immunocytochemically; and/or Western blot confirmed protease-resistant PrP; and/or presence of scrapie-associated fibrils conducted on brain tissue
- 14-3-3 proteins in CSF (test not specific for CJD)
- Periodic sharp and slow wave complexes (PSWC) in EEG (Test suggestive but not specific for CJD)

Case classification

Confirmed: A clinically compatible case diagnosed by standard neuropathological techniques; and/or immunocytochemically; and/or Western blot confirmed protease-resistant PrP; and/or presence of scrapie-associated fibrils.

Probable: Progressive dementia; and at least two out of the following four clinical features:

- Myoclonus
- Visual or cerebellar signs
- Pyramidal/extrapyramidal signs
- Akinetic mutism

AND

- A typical EEG during an illness of any duration; and/or a positive 14-3-3 CSF assay and a clinical duration to death of < 2 years
- Routine investigations should not suggest an alternative diagnosis

Suspect: Progressive dementia; and at least two out of the following four clinical features:

- Myoclonus
- Visual or cerebellar signs
- Pyramidal/extrapyramidal signs
- Akinetic mutism

AND

- No EEG or atypical EEG and duration < 2 years

Comment:

Cases under the age of 55 years old should be evaluated for the variant form of CJD. Brain tissue for diagnosis and CSF for the 14-3-3 protein should be sent to the National Prion disease Pathology Surveillance Center at Case Western Reserve University. Information about the center, shipping and mailing instructions can be found on their web site:

<http://www.cjdsurveillance.com>.