

A brain autopsy is recommended for all physician-diagnosed CJD cases.

Creutzfeldt-Jakob Disease (CJD)

2011 Case Definition (North Carolina)*

Definite (NC Confirmed):

Diagnosed by standard neuropathological techniques; and/or immunocytochemically; and/or Western blot confirmed protease-resistant PrP; and /or presence of scrapie-associated fibrils.

Probable:

Rapidly 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 positive result on at least one of the following laboratory tests:

- a typical EEG (periodic sharp wave complexes) during an illness of any duration; and/or
- a positive 14-3-3 cerebrospinal fluid (CSF) assay in patients with a disease duration of less than 2 years
- Magnetic resonance imaging (MRI) high signal abnormalities in caudate nucleus and/or putamen on diffusion-weighted imaging (DWI) or fluid attenuated inversion recovery (FLAIR)

AND without routine investigations indicating an alternative diagnosis.

Possible (NC 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 the absence of a positive result for any of the three laboratory tests (listed above) that would classify a case as “probable”

AND duration of illness less than two years

AND without routine investigations indicating an alternative diagnosis.

2. Iatrogenic CJD

Progressive cerebellar syndrome in a recipient of human cadaveric-derived pituitary hormone; or sporadic CJD with a recognized exposure risk, e.g., antecedent neurosurgery with dura mater implantation.

3. Familial CJD

Definite or probable CJD plus definite or probable CJD in a first degree relative; and/or Neuropsychiatric disorder plus disease-specific PrP gene mutation.

*Effective January 1, 2011, and adapted from the CDC Diagnostic Criteria for Creutzfeldt-Jakob Disease (CJD), 2010 accessible at:

http://www.cdc.gov/ncidod/dvrd/cjd/diagnostic_criteria.html .