

Merlin disease code: 04610 Creutzfeldt-Jakob Disease (CJD)

[Paper case report form](#) required
No Merlin extended data

Background

CJD is a progressive uniformly fatal dementia characterized by myoclonus, visual or cerebellar signs, akinetic mutism, and pyramidal or extrapyramidal signs.

Clinical criteria for case classification

Confirmatory:

A clinically compatible illness.

Presumptive:

All of the following:

- Progressive dementia,
- **And** a clinical duration to death <2 years,
- **And** at least 2 of the following clinical features:
 - Myoclonus
 - Visual or cerebellar signs
 - Pyramidal or extrapyramidal signs
 - Akinetic mutism,
- **And** no alternative diagnosis suggested during routine investigation.

Laboratory criteria for case classification

Confirmatory:

Diagnosed by one or more of the following:

- Standard neuropathological techniques,
- **Or** immunocytochemical testing,
- **Or** Western blot confirmed protease-resistant prion protein,
- **Or** presence of scrapie-associated fibrils conducted on brain tissue.

Presumptive:

Positive real-time quaking induced conversion (RT-QuIC) in cerebrospinal fluid (CSF) or other tissues.

Supportive:

One or more of the following:

- Positive 14-3-3 CSF assay,
- **Or** typical electroencephalogram (EEG) (periodic sharp wave complexes),
- **Or** high signal in caudate/putamen on magnetic resonance imaging (MRI) brain scan or at least two cortical regions (temporal, parietal, occipital) either on diffusion-weighted imaging (DWI) or fluid attenuated inversion recovery (FLAIR).

Epidemiological criteria for case classification

Not applicable.

Case classification

Confirmed:

A person with confirmatory clinical criteria and confirmatory laboratory criteria.

Probable:

Either of the following:

- A person with confirmatory or presumptive clinical criteria and presumptive laboratory criteria
- **Or** a person with presumptive clinical criteria and supportive laboratory criteria.

Suspect:

A person with presumptive clinical criteria.

Criteria to distinguish a new case from previous reports

Not applicable.

Comments

Cases under the age of 55 years old should be evaluated for the variant form of CJD. Brain tissue for diagnosis and CSF for RT-QuIC and 14-3-3 assays should be sent to the National Prion Disease Pathology Surveillance Center at Case Western Reserve University. Information about the center and shipping instructions can be found on their web site: www.cjdsurveillance.com. Please notify Bureau of Epidemiology to assist with case evaluation and laboratory testing.