Creutzfeldt-Jakob Disease (CJD)

Merlin disease code=04610
Case report form (CRF): Creutzfeldt-Jakob Disease Worksheet
PAPER CRF REQUIRED

Background
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 evidence.

**Probable:**
Either of the following:
- A person with confirmatory or presumptive clinical criteria and presumptive laboratory evidence
- Or a person with presumptive clinical criteria and supportive laboratory evidence.

**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](http://www.cjdsurveillance.com). Please notify Bureau of Epidemiology to assist with case evaluation and laboratory testing.

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