Creutzfeldt-Jakob disease (CJD)

Australian national notifiable diseases case definition

This document contains the surveillance case definitions for Creutzfeldt-Jakob disease (CJD) and closely related prion diseases[i], which are nationally notifiable within Australia, and excluding variant CJD[ii]. State and territory health departments use this definition to decide whether to notify the Australian Centre for Disease Control of a case.

Version	Status	Last reviewed	Implementation date
1.0	Initial CDNA case definition	CDWG November 2009	1 July 2010
2.0	Complete review by CJD IPCG and Surveillance Case Definition Working Group Clarified that case definition applies to fatal familial insomnia and Gerstmann-Straeussler-Scheinker syndrome **Laboratory Definitive Evidence** Updated with greater detail **Probable Case** Added several new evidence streams based around expert assessment of progressive neuropsychiatric disorder compatible with CJD coupled with clinical tests **Clinical Evidence** Updated 1. from “Progressive dementia of less than two years duration” to “Rapidly progressive cognitive impairment” **Laboratory suggestive evidence** Removed	CDNA November 2025	1 January 2026

Reporting

Confirmed and **probable** cases should be notified[iii].

Confirmed case

A confirmed case requires **laboratory definitive evidence**

Laboratory definitive evidence

Brain neuropathological confirmation by immunochemical detection of abnormal prion protein (typically protease-resistant PrP^{Sc}) by western blot or immunocytochemistry.

Probable case

1. progressive neuropsychiatric disorder compatible with CJD as determined by an appropriate expert[iv] AND positive real time-quaking induced conversion (RT-QuIC) in CSF or other tissues ;

OR

2. Progressive neuropsychiatric disorder compatible with CJD and other related prion disease as determined by an appropriate expert[iv] AND definite or probable prion disease in 1st degree relative

OR

3. Progressive neuropsychiatric disorder compatible with CJD and other related prion disease as determined by an appropriate expert[iv] AND recognised pathogenic *PRNP* sequence variation

OR

4. Clinical evidence AND presence of 14-3-3 protein in cerebrospinal fluid (CSF)

OR

5. Clinical evidence AND a typical MRI brain scan (that is, high signal in caudate/putamen or at least two cortical regions (temporal, parietal, occipital) either on DWI or FLAIR)

OR

6. Clinical evidence AND a typical electroencephalogram (EEG) (that is, generalised periodic complexes)

Clinical evidence

1. Rapidly progressive cognitive impairment;

   AND

2. At least 2 of the following clinical features:
   - myoclonus
   - visual or cerebellar signs
   - pyramidal/extrapyramidal signs
   - akinetic mutism.

[i] Phenotypes of fatal familial insomnia (mostly genetic, but also sporadic), and Gerstmann-Straeussler-Scheinker syndrome should also be reported under this definition.
[ii] there is a separate case definition for variant CJD: https://www.health.gov.au/resources/publications/variant-creutzfeldt-jakob-disease-vcjd-surveillance-case-definition?language=en
[iii] This includes sporadic, acquired/accidental and genetic cases
[iv] An appropriate expert such as a neurologist or neuropsychiatrist