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 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 expertiv 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 expertiv 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

At least 2 of the following clinical features:

- myoclonus
- visual or cerebellar signs
- pyramidal/extrapyramidal signs

- akinetic mutism.

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[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