Facts about prion disease (CJD)

CJD is always fatal and unfortunately distinctive because of its typically rapid clinical progression. The duration of illness can vary but approximately 80% of patients die within a year of symptom onset with only a small number of patients surviving for more than two years.

There is no test available to detect CJD prior to the onset of symptoms.

Currently, there is no disease-modifying therapy or cure for any form of prion disease, including CJD. Treatment is symptomatic and supportive.

CJD is not contagious and therefore cannot be transmitted by normal contact. Nursing of a CJD patient or kissing a loved one with CJD does not pose any risk.

A brain-only autopsy can provide a definite diagnosis of CJD and can confirm or rule out if the patient had variant CJD (vCJD). However, it cannot establish if the cause of the patient's illness was sporadic or genetic without testing the DNA of the patient (*PRNP* testing).

Every year in Australia about 50 people die of CJD. Approximately 85–90% suffer with sporadic CJD and 10–15% suffer with a genetic form of prion disease. Acquired forms of prion disease are now extremely rare.

CJD is unique in that it can be genetic as well as transmissible. Ruling out a genetic cause for a patient's illness with CJD can remove ramifications around blood donation and surgery that involves high-infectivity tissue.

ALSO AVAILABLE

- **Information packs for families**
- **Information packs for healthcare professionals**

CJD Support Group Network is a registered charity

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Because prions are remarkably resistant to conventional disinfection and sterilisation practices the National CJD Infection Control Guidelines still recognise risk of transmission for surgical procedures, involving high-infectivity or medium-infectivity tissue, on a patient with an identified risk of CJD. Although there have been no reported cases of transmission via surgical instruments since the 1970s, a theoretical risk cannot be ruled out and special precautions are still recommended for an 'at risk of CJD' patient. However, these precautions should only apply for procedures in which high-infectivity tissue will be exposed (e.g. neurosurgery, spinal cord surgery, ophthalmic surgery, pituitary surgery).

Most forms of genetic CJD are impossible to differentiate from sporadic CJD without *PRNP* testing. Only by testing the DNA of the patient – either during life if there is a family history, or by the next of kin consenting to testing, usually after the autopsy result is reported – can a genetic cause be established or ruled out. The case is then classified as sporadic CJD if the *PRNP* test result is negative for a genetic mutation. Genetic prion disease is often identified from a family history, but in 60% of genetic cases there is no known family history. Families should consider either *PRNP* testing or taking of a blood sample in life for DNA extraction and storage for future *PRNP* testing if/when required.

If a patient's *PRNP* test result is normal, and no mutation is identified, then family members of that patient have the same risk of developing CJD as the general population i.e. background risk. By providing proof of a patient's 'normal' *PRNP* test results, relatives can then donate blood if they fulfil other donation criteria.

This information is provided by the

CJD Support Group Network (CJDSGN)

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CJD SUPPORT GROUP NETWORK

What is CJD?

Creutzfeldt-Jakob disease

What is Creutzfeldt-Jakob disease (CJD)?

CJD is a rare neurodegenerative brain disease in humans. It is the most common human form of a group of diseases that affect humans and animals which are known as transmissible spongiform encephalopathies (TSE) or prion diseases.

In animals the best-known TSEs are bovine spongiform encephalopathy (BSE) in cattle, scrapie in sheep and chronic wasting disease (CWD) in cervids i.e. deer, elk, etc.

BSE and CWD have NEVER been found in Australian livestock.

In humans the three forms of prion diseases are:

- **sporadic CJD (sCJD)**
- **genetic prion disease**

1. familial CJD (fCJD)
2. Gerstmann-Sträussler-Scheinker syndrome (GSS)
3. fatal familial insomnia (FFI)

- **acquired CJD**

1. iatrogenic or medically acquired CJD
2. kuru
3. variant CJD (vCJD)

Sporadic CJD (sCJD) is a rapidly progressive disease that has no known cause but is believed to result from a spontaneous conformational change in the native or normal form of the prion protein. It occurs at random in about 1 to 2 people per million of the population per year and accounts for 85–90% of all cases of prion disease. This equates to a lifetime risk of 1 in 5,000.

Genetic prion disease accounts for approximately 10–15% of cases and occurs due to a mutation in a gene called the prion protein gene (*PRNP*) that encodes the normal form of the prion protein. Mutations in the *PRNP* gene can lead to familial Creutzfeldt-Jakob disease (fCJD), Gerstmann-Sträussler-Scheinker syndrome (GSS) and fatal familial insomnia (FFI). One in 50,000 people carry a pathogenic *PRNP* mutation.

Genetic prion disease is inherited in an autosomal dominant fashion. Each child of a person carrying a mutation for prion disease has a 50% chance of inheriting that mutation. Someone who carries a mutation will most likely develop the illness in their lifetime – if they live long enough. Penetrance, the likelihood of a mutation carrier to become symptomatic, can vary with different *PRNP* mutations, and also between individuals who carry the same mutation or within the same family.

Acquired prion disease is very rare and has three subtypes.

Iatrogenic or medically acquired CJD (iCJD) although very rare, is associated with transmission via medical treatments and surgical procedures. In the mid-1970s it became evident that CJD could be transmitted from one person to another through invasive medical procedures including corneal transplants or contaminated surgical instruments. In the 1980s it was recognised that explanted material, such as extracted human pituitary hormones for fertility and short stature and dura mater grafts accidentally contaminated with prion protein, could also transmit this disease to recipients.

The risk of transmission of CJD from surgical instruments used on patients, who are 'at increased risk of CJD' and undergo procedures involving high infectivity tissue (i.e. surgery involving the central nervous system), albeit low, is still a recognised risk and requires special precautions.
www.cjdsupport.org.au/site/wp-content/uploads/2011/10/ CJD-Infection-Control-Guidelines-Jan-13.pdf

Kuru is a historical prion disease that once affected the Fore people of the Eastern Highlands of Papua New Guinea. The study of kuru was instrumental in establishing that CJD is transmissible. In the 1960s it was realised that kuru was transmitted through ritualistic endocannibalism. This practice was outlawed in the late 1950s resulting in the incidence of kuru gradually declining to its now essential eradication.

Variant CJD (vCJD) was first reported in 1996 in the United Kingdom following the first death of an affected individual in 1994. Globally, 232 cases have been reported up to 2020, mainly in the UK, with the vast majority resulting from the consumption of contaminated meat products from bovine spongiform encephalopathy (BSE) affected cattle. Variant CJD can also be transmitted through blood and blood products. Variant CJD differs clinically from sporadic CJD typically manifesting as a longer duration of illness (median ~14 months) presenting with psychiatric and sensory symptoms. It usually affects a younger age group (median age at death is 28 years).

Deposits of the abnormal, misfolded form of the prion protein are often found in peripheral lympho-reticular tissues such as the spleen, lymph nodes and tonsils, and hence pose additional risks of transmission that do not occur with sporadic CJD.

Variant CJD is often incorrectly referred to as 'mad cow disease'. To date there have been no reported cases of variant CJD in Australia.

Clinical features of CJD

The accumulation of misfolded prion protein causes neuronal dysfunction which results in spongiform changes in the brain. Patients typically present with rapidly progressive dementia and gross motor impairment (such as unsteady gait), culminating in death on average within 4–6 months. Symptoms characteristically include cognitive decline, behavioural changes, impaired balance, lack of coordination (ataxia), muscle jerking (myoclonus), visual disturbances, weakness and spasticity. Patients are usually 50–70 years old but illness onset can be in adolescence or as late as the ninth decade.

Diagnostic tools

Characteristic diagnostic test findings are: generalised period complexes in an electroencephalogram (EEG), T2 hyperintensities in the caudate/putamen and/or multiple cerebral cortical regions in brain magnetic resonance imaging (MRI), elevated 14-3-3 and total tau proteins in cerebrospinal fluid (CSF) and ability to amplify abnormal misfolded prion protein ("seeding activity") using protein amplification techniques such as the real-time quaking induced conversion (RT-QuIC) assay.

A brain-only autopsy is the only way to confirm a definite diagnosis of prion disease.