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## Creutzfeldt-Jakob disease (CJD)

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### Summary

- CJD is extremely rare. It affects at least one person in every million people, within the total population each year.
  - Researchers have found no firm evidence of transmission of classical CJD (sporadic, medically acquired or genetic forms) through exposure to blood and blood products.
  - In almost 90 per cent of cases, it is not understood how or why CJD occurs.
  - The only proven cases of medically acquired CJD have been those which have had direct exposure to highly infective contaminated tissue (for example, CJD brain tissue) and neurosurgical equipment.
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Creutzfeldt-Jakob disease (CJD) is a rapidly progressive disease. It is one of a group of rare diseases that affects humans and animals, known as transmissible spongiform encephalopathies (TSEs) or prion diseases. Creutzfeldt-Jakob disease is characterised by physical deterioration of the brain, which commonly causes dementia and walking difficulties. Death can occur up to two years after the first symptoms; however, the majority of people die within six months. There is no treatment or cure.

### Transmissible spongiform encephalopathies (TSEs) or prion diseases

CJD is the human form of TSE or prion disease. This group of diseases occurs in humans and in animals such as cattle, sheep, elk and deer. CJD was first recognised in humans in the 1920s.

The human TSEs or prion diseases include:

- sporadic CJD, which causes 85 to 90 per cent of cases
- genetic or familial CJD, which causes 10 to 15 per cent of cases
- Kuru
- variant CJD
- medically acquired CJD.

### Sporadic CJD

Sporadic CJD accounts for the greatest number of human deaths from this group of diseases. CJD affects approximately one person per million people each year. So, in a population of 10 million people, there are likely to be at least 10 cases in one year. CJD most often affects people between the ages of 50 to 70 years of age. The cause of sporadic CJD is unknown.

### Genetic CJD

Genetic prion disease is extremely rare and is most often recognised from a family history of the illness in brothers, sisters or parents. It is an inherited disease, passed from a parent to child at conception. The disease is not always passed on; each child born has a 50 per cent chance of contracting the disease.

### Kuru

Kuru is a human prion disease found only in the central highlands of New Guinea. It was caused by the practice of ritualised cannibalism of deceased relatives. The practice has been discontinued and the number of Kuru cases has correspondingly declined over time. Kuru has never been found outside of Papua New Guinea.

### Variant CJD

Variant CJD was first recognised in 1996 in the United Kingdom. It has not been found in Australia. Variant CJD is linked to an epidemic of bovine spongiform encephalopathy (BSE) or 'mad cow disease', as it is known in the media. BSE is a prion disease which occurs in cattle.

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The transmission of the naturally occurring TSE scrapie in sheep to cattle through the food chain is believed to have caused the epidemic of BSE in the United Kingdom in the 1980s. BSE has not occurred in Australian cattle. Concern remains in the UK about the number of people who may be at risk of developing variant CJD.

### **Variant CJD has different symptoms**

Variant CJD is distinguished from the other human prion diseases by distinctly different symptoms. The different symptoms can include the duration of the disease, which can be longer (up to a year) than sporadic CJD. People are also developing variant CJD at a younger age and autopsies reveal distinct cellular changes in the brain not found in other human prion diseases.

### **Medically acquired CJD**

Medically acquired CJD has occurred worldwide as a result of a number of medical treatments. Treatments shown to have transmitted CJD include:

- the use of human pituitary extract hormone for infertility or short stature
- dura mater grafts used in brain surgery to repair damage to the membrane covering the brain
- corneal grafts – three cases have been recorded worldwide
- exposure to contaminated neurosurgical equipment – five cases have been recorded worldwide.

Hormone and grafts treatments previously used tissue derived from human cadavers that were probably contaminated with CJD. Once CJD was identified as a medical hazard, the use of these products was immediately discontinued and replaced by synthetic alternatives. Sterilising procedures were increased for CJD and patients screened for CJD risk before neurosurgery. Stricter guidelines were also established for the use of organ transplants.

### **Australian cases of medically acquired CJD**

There have been nine cases of medically acquired CJD in Australia in total. They include four confirmed CJD deaths following treatment with pituitary extract hormone for infertility. There have been no further CJD-related deaths arising from infertility or short stature hormone treatments since 1991.

Five medically acquired CJD deaths have been caused by dura grafting during brain surgery, where the covering of the brain has been repaired. The last dura-related CJD death occurred in 2000. There have been no other medically acquired or transmitted CJD deaths in Australia.

### **Blood transfusion**

The possibility of transmission of CJD through blood transfusion or the use of blood products is sometimes suggested. There is no evidence to support the argument that blood products may cause sporadic or medically acquired CJD.

Variant CJD has been confirmed to be transmissible through blood transfusion. This has been supported in both the experimental laboratory setting and subsequently by the experience of variant CJD in the United Kingdom.

In response to the recognised transmission of variant CJD by blood transfusion in the United Kingdom, Australian blood-banking guidelines defer donors who may have some risk for CJD. Before donating blood, screening is performed to identify potential donors who may have CJD, those who may have been medically exposed and also some relatives from families where a CJD-related death has occurred. These people are routinely deferred and asked not to donate blood.

These people include:

- the first-degree relatives of those who have died from sporadic CJD
- families with genetic CJD
- recipients of human pituitary hormone extract
- anyone who received neurosurgery on the head, neck or spinal cord between 1972 and 1987 (and who may have had dura grafts).

### **Blood donations in Australia**

In September 2000, a ban was placed on blood donations from people who lived in the United Kingdom for six months or more from 1980 to 1996 or who have received blood transfusions in the UK since 1 January 1980. Similar precautionary measures have been taken in New Zealand, Canada and the USA.

### **Symptoms of CJD**

CJD is difficult to diagnose. Early symptoms can be vague and there are no diagnostic tests to confirm exposure, or diagnose CJD, until symptoms are well advanced. As the disease progresses, extensive investigations are necessary to exclude the possibility of other treatable diseases. A possible diagnosis is made only as the illness progresses. Examination of brain tissue after death is the only way to definitely confirm this disease.

Symptoms may include:

- confusion or disorientation, which rapidly advances to a dementia
- personality changes
- behavioural changes
- weakness, loss of balance and muscle control causing difficulty walking
- muscle spasms
- visual symptoms such as double vision or blindness.

### **Brain tissue changes cause CJD symptoms**

The symptoms of CJD are caused by changes occurring to brain tissue. Normal prion protein is manufactured in the healthy brain. Although the function of normal prion protein is not understood, it is known that prion diseases are caused when the normal prion protein changes its physical structure or shape. This abnormal prion protein causes damage and cell death in the brain. Once present, it is believed the abnormal protein acts as a template, converting other normal prion protein into the abnormal, disease-causing form.

It is not known what causes the initial change to the prion protein structure. It is thought it could be triggered by:

- Changes in the brain's chemical environment
- Exposure to abnormal prion protein – for example, in medical exposures described previously
- Genetic influences that may cause some people to be more vulnerable to spontaneously producing the abnormal prion protein.

### **A viral connection remains unproven**

CJD is sometimes referred to as a slow virus. However, the viral theory has never been confirmed.

### **Why hospitals sometimes screen patients for CJD risks**

The altered prion protein causing CJD is known to be extremely tough. It remains active outside the body and survives accepted and routine levels of hospital sterilisation for other disease-causing agents like viruses and bacteria.

Recommended levels of sterilisation required for CJD exceed routine standards of sterilisation. However, these cannot be performed routinely as the higher levels cause significant damage to medical instruments and equipment.

### **CJD is not transmitted by casual contact**

CJD is not transmitted by casual contact like drinking from the same cup, kissing or close physical contact with an individual suffering from CJD.

### **CJD is now a notifiable disease**

Public health authorities in Australia have made CJD a notifiable disease. This means any newly diagnosed case must be reported to local health departments by their treating doctors.

### **Where to get help**

- Your **GP (doctor)**
- The **CJD Support Group Network** Tel. 1800 052 466

- **Clinical geneticist**

**This page has been produced in consultation with and approved by:**

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