

### Introduction

Alzheimer's disease is one type of a large group of disorders known as "dementias." It is an irreversible disease of the brain in which the progressive degeneration of brain cells causes thinking ability and memory to deteriorate. Alzheimer's disease also affects behaviour, mood and emotions, and the ability to perform daily living activities.

Other forms of dementia resemble Alzheimer's disease in that they also involve a progressive degeneration of brain cells that is currently irreversible. They include the dementia associated with vascular dementia, frontotemporal dementia, Creutzfeldt-Jakob Disease (CJD), Lewy body dementia, Huntington disease, and Parkinson's disease.

Sometimes a person may have different symptoms in the early stages of the disease, such as memory loss, behaviour changes, or difficulties with speech and movement. These symptoms may suggest a form of dementia other than Alzheimer's disease. A person should always seek a thorough medical assessment if any of these symptoms is present.

Regardless of the type of dementia, individuals are encouraged to obtain information and support from the Alzheimer Society.

### What is Creutzfeldt-Jakob disease?

Creutzfeldt-Jakob Disease (CJD) is a rare, fatal brain disease. It is caused by an abnormal form of a substance called the prion protein. In its normal form, prion protein is made by most body cells and doesn't cause disease. In its abnormal form, prion protein is toxic to brain cells and causes disease.

Prion diseases also occur in animals. Examples include:

- Bovine spongiform encephalopathy (BSE) in cattle
- Scrapie in sheep and goats
- Chronic wasting disease (CWD) in deer, elk and moose

The brain damage in people or animals with prion disease can be seen with a microscope. One kind of damage is "spongiform change", in which the brain tissue looks like a sponge with many tiny holes. Many of the brain's nerve cells die.

CJD can be caused in different ways. Most cases (about 90%) occur in older people, without warning or a clear reason. This is called "sporadic" CJD, meaning it happens occasionally or unpredictably. Sporadic CJD likely begins with the forming of abnormal prion protein by one or a few brain cells. The abnormal condition then spreads to the normal prion protein in the rest of the brain.

Most other CJD cases (about 10%) have genetic changes called mutations. These are found in the gene that tells body cells how to make prion protein. Mutations increase the chances that the prion protein will become abnormal, and that prion disease will develop.

A small number of CJD cases has occurred after infection with prions from other humans or from animals. Infection with human prions has occurred by accident through certain medical procedures involving human tissues. This kind of CJD is called “iatrogenic”, meaning it is caused by a medical treatment.

Some cases have resulted from human exposure to BSE, a prion disease of cattle. These patients developed a new form called “variant CJD”.

## How does Creutzfeldt-Jakob disease affect the person?

The symptoms of CJD and their timing can vary greatly from person to person. Usually a mental or neurological problem appears first. However, early symptoms may seem mild, sometimes like depression. A family member is often the first to notice mood swings, social withdrawal or lack of interest.

After it starts, CJD usually progresses rapidly. Eventually the person loses the ability to move, speak or care for themselves, and needs full-time care. Most people with CJD die within 6 months from when the illness begins. Some can live as long as 1 year, but rarely longer.

## Sporadic CJD

Patients may experience any combination of the following symptoms:

- dementia (loss of memory and thinking abilities)
- ataxia (unsteadiness when walking or standing, marked clumsiness)
- myoclonus (sudden jerky movements)
- psychological problems (depression, irritability, changes in behaviour)
- vision problems (including blindness)
- aphasia (loss of ability to speak or understand speech)
- stiffness of arms or legs
- difficulty swallowing

## Genetic prion diseases

More than 50 different rare genetic mutations have been found in the prion protein gene. Individuals carrying different mutations can have different symptoms. So can family members who have the same mutation.

Symptoms can differ enough that some genetic prion diseases have been given special names, such as:

- Genetic CJD
- Gerstmann-Sträussler-Scheinker disease (GSS)
- Fatal familial insomnia (FFI)

In genetic CJD, symptoms are similar to those of sporadic CJD.

GSS usually starts with a clumsiness or unsteadiness when standing or walking, and later progresses to dementia. The disease usually lasts longer than sporadic CJD or genetic CJD, and the person may survive for several years after the illness begins.

In FFI, the main symptom is a severe, progressive and untreatable form of insomnia. FFI leads to reduced control of basic bodily functions, such as blood pressure. Coma and death eventually follow.

## Iatrogenic CJD

Symptoms of iatrogenic CJD are like those of sporadic CJD. Many cases have shown early symptoms of movement disorder, with dementia appearing later.

## Variant CJD

People with variant CJD tend to be much younger than those with other types of CJD, often in their 20s. They also often show early symptoms that are less common in other forms:

- psychological problems such as anxiety, depression, withdrawal
- unsteadiness when walking or standing, marked clumsiness
- persistent pain or other strange sensations

In variant CJD, dementia tends to begin later in the disease. Eventually the person loses the ability to move or speak, and needs constant care. Death usually occurs 1 to 2 years after symptoms begin.

## How is Creutzfeldt-Jakob disease diagnosed?

CJD can be difficult to diagnose, especially in the early stages. There is no completely accurate test to diagnose it in a living person. The only way to confirm whether a person has CJD, or not, is to examine brain tissue after death during an autopsy. However, doctors can do a detailed exam and many tests to help with the diagnosis.

The following steps may be taken:

- **Detailed medical history:** This will help the doctor learn when the person's signs and symptoms started, as CJD usually progresses rapidly.
- **Magnetic resonance imaging (MRI):** An MRI produces a picture of the brain that can show signs of disease. Many people with CJD show specific changes by MRI. MRI can also distinguish between sporadic and variant CJD.
- **Electroencephalogram (EEG):** An EEG measures the brain's electrical activity. Often, but not always, there is a specific EEG pattern that helps to diagnose CJD.
- **Lumbar puncture (LP):** Cerebrospinal fluid (CSF) can be taken from the person's lower back and tested in the lab. The amounts of certain protein markers in the fluid, which indicate brain cell degeneration, are usually higher than normal in CJD. Other tests on the fluid can help determine if the person has a condition that isn't CJD.
- **Blood test:** There is no useful blood test for CJD. But a blood sample is often used to prepare DNA, which can be tested to diagnose genetic prion disease.
- **Brain autopsy:** The best way to diagnose CJD is by looking at the whole brain after death, using a microscope. Brain autopsies are performed only in certain large hospitals in Canada. The Canadian CJD Surveillance System (see contact information at end of this sheet) can help make arrangements for a brain autopsy if CJD is suspected and if the next of kin gives consent.

## What are the risk factors for Creutzfeldt-Jakob Disease?

### Sporadic CJD

Sporadic CJD is found worldwide, mostly in people over 50. Canada, the United States, United Kingdom (UK), Australia, and several European countries carefully monitor sporadic CJD through national surveillance. About 1 or 2 cases typically occur per million people each year.

### Genetic prion diseases

The human prion protein gene (PRNP) contains the DNA instructions to make the normal prion protein. Many different rare genetic variations have been found in this gene. Some variations don't cause disease. People with other variations, called mutations, are much more likely to develop prion disease.

Genetic prion diseases mostly affect older people. In some families genetic prion disease can develop at younger ages. However, some people with a genetic mutation never develop the illness. There is currently no approved medical treatment to prevent prion disease in a person with a genetic CJD mutation.

The effects of some genetic variations in the human prion protein gene -- especially the rarest ones -- aren't well understood. This means it can be hard to predict their long-term health effects.

As with any genetic disease, the risk of developing genetic prion disease can be passed down from parent to child. The chance that a parent with a PRNP mutation will pass it down is 50% for each child. If a person has a brother or sister with a mutation, there is a 50% chance that they have it too and a 50% chance of not having the mutation (see the diagram below).

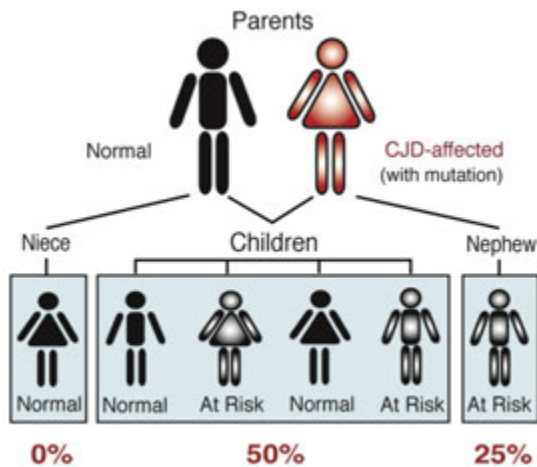


Figure 1: Risk of genetic CJD decreases with more distant relationship to an affected person.

A family at risk for genetic prion disease needs medical support to use this knowledge in personal decisions. Genetic counseling is strongly recommended for anyone who has questions about their genetic risk for prion disease. Genetic counseling services are available in most large medical centres in Canada. If you have questions about genetic testing, you may contact the Canadian CJD Surveillance System at 1-888-489-2999.

## Iatrogenic CJD

Most cases of iatrogenic CJD have been found in the United States, UK, France and Japan, with only four cases to date in Canada. However, because CJD can be silent for many years, some cases are still being found in other countries.

Scientific knowledge and medical technology have greatly improved since iatrogenic CJD was first identified. As a result, risks of passing CJD from one person to another in this way have now been greatly reduced.

As a precaution, it is usually assumed that all forms of human prion disease can be passed to others given the right circumstances. Hospitals use special procedures to ensure that medical and surgical instruments are safe to use on patients. Lab workers take precautions when handling specimens during diagnostic testing procedures.

Funeral services workers also need to follow their provincial regulations when handling the remains of a person suspected to have had CJD.

## Variante CJD

In 1994, a few cases of CJD were found in unexpectedly young people in the UK. Because this disease differed from all known forms of CJD, it was called variant CJD. By 1996, scientists knew the disease looked in some ways like BSE and the UK began very strict food safety measures to prevent contamination of food with BSE.

As of 2016, 231 cases of variant CJD had been identified worldwide, including 2 in Canadians exposed to BSE while living in other countries. The Canadian government put in place safety measures similar to those in the UK in 2003 and it regulates the Canadian beef industry to minimize the risk of human contact with BSE through food. With these safeguards, the risk of human exposure to BSE is now considered extremely low.

Five people in the UK were probably infected with variant CJD after receiving blood transfusions. The blood products were from donors who were healthy at the time of donation but developed the disease later. The Canadian blood system (Canadian Blood Services and Héma Québec) has taken steps to prevent blood donations by people who may have silent CJD and reduce the risk that variant CJD would be spread by blood transfusion.

## Is there treatment?

There is currently no known cure for CJD and no effective way to slow its progression. Supportive nursing care is focused on keeping the person as comfortable as possible.

## For more information:

Visit the Alzheimer Society's website at [www.alzheimer.ca](http://www.alzheimer.ca) or contact your local Alzheimer Society.

More information on CJD is available:

Visit the CJD Surveillance System of Canada website at:

<http://www.phac-aspc.gc.ca/hcai-iamss/cjd-mcj/cjd-eng.php>

To contact a nurse from the Canadian CJD Surveillance System:

Creutzfeldt-Jakob Disease Surveillance System

Public Health Agency of Canada

130 Colonnade Rd, AL: 6502A

Ottawa, ON K1A 0K9

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Telephone: 1-888-489-2999

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## Alzheimer *Society*

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