Alzheimer Society

Creutzfeldt-Jakob disease

Creutzfeldt-Jakob disease (sometimes called CJD) is a rare and fatal brain disease that leads to dementia. As the disease progresses, it causes mood swings, social withdrawal and lack of interest. Eventually the person loses the ability to move, speak or care for themselves, and needs full-time care.

Creutzfeldt-Jakob disease is caused when a substance called prion protein develops an abnormal shape.

Most body cells make prion protein that is a normal shape and that doesn't cause disease. But if prion protein folds into the wrong shape, it is toxic to brain cells and causes disease. We still don't understand why some prion proteins change shape and become toxic to the brain. Researchers are working to try and find answers.

There are different types of prion-related diseases that can happen in humans and animals. Creutzfeldt-Jakob disease is the most common type of prion-related disease in humans. And some examples of prion disease in animals include **Bovine spongiform encephalopathy** in cattle, **scrapie** in sheep and goats and **chronic wasting disease** in deer, elk and moose.

The brain damage in people or animals with a prion disease can be seen with a microscope. One kind of damage is **spongiform change**, when brain tissue looks like a sponge with many tiny holes. In prion diseases, the brain's cells will eventually die.

Creutzfeldt-Jakob disease can be caused in humans in different ways. The majority of people living with this disease have either the **sporadic** or **familial** types. But in rare cases an infection or exposure can cause **Creutzfeldt-Jakob disease**, leading to what is called **iatrogenic** or **variant** types. (See sidebar for more information about these types.)

People living with Creutzfeldt-Jakob disease may experience a wide range of symptoms.

Researchers have identified more than 50 variations of **Creutzfeldt-Jakob disease.** These variations can have different symptoms, and their timing can change greatly from person to person.

Usually, a mental or neurological problem appears first. Early symptoms may seem mild, sometimes like depression. A family member or friend is often the first to notice mood swings, social withdrawal or lack of interest.

There are four main types of Creutzfeldt-Jakob disease.

Sporadic. Approximately 90% of people diagnosed with of Creutzfeldt-Jakob disease have what is considered the sporadic type. Sporadic Creutzfeldt-Jakob disease typically occurs in people around the age of 60 or older, without warning or a clear reason. These cases appear unpredictably and cannot be linked to other cases. Sporadic Creutzfeldt-Jakob disease likely begins when prion protein forms an abnormal shape in one or a few brain cells. The abnormal shape probably then spreads to the normal prion protein in the rest of the brain.

Familial. Most other Creutzfeldt-Jakob disease cases (about 10%) are related to genetic changes called mutations. These mutations are found in the gene that tells body cells how to make prion protein. Mutations increase the chance that prion protein will fold into the wrong shape, becoming abnormal, and causing prion-related disease.

Iatrogenic. A small number of Creutzfeldt-Jakob disease cases have occurred after a person is infected with misfolded or abnormally shaped prion proteins from other humans or animals. Infection with human prions has occurred by accident through certain medical procedures involving human brain tissues.

Variant. This type is very rare. Cases happen when humans are exposed to **bovine spongiform encephalopathy**. This encephalopathy is more commonly known as **mad cow disease**.

After it starts, **Creutzfeldt-Jakob disease** usually progresses rapidly. Eventually the person loses the ability to move, speak or care for themselves, and they need full-time care. Most people with this disease die within six months after their illness began. Some can live as long as one year, and rarely longer.

SYMPTOMS	RISK FACTORS
Sporadic type of Creutzfeldt-Jakob disease	
People may experience any combination of the following symptoms: • Dementia: loss of memory and thinking abilities • Ataxia: unsteadiness when walking or standing • Myoclonus: sudden jerking movements • Psychological problems: depression, irritability and/or changes in behaviour • Vision problems: including blindness, seeing double and/or hallucinations • Aphasia: loss of ability to speak or understand speech • Stiffness of arms or legs • Difficulty swallowing	• Researchers are currently trying to identify possible risk factors for sporadic type Creutzfeldt-Jakob disease.
Familial type of Creutzfeldt-Jakob disease	
Many different and rare genetic variations have been linked to this type of Creutzfeldt-Jakob disease. Symptoms can differ enough that some genetic prion diseases have been given special names, such as: • **Genetic Creutzfeldt-Jakob disease:** This has similar symptoms to the sporadic type of this disease. • **Gerstmann-Sträussler-Scheinker disease:** Symptoms include clumsiness when standing or walking. Later symptoms progress to dementia. Often lasts longer than sporadic or genetic forms of the disease. A person with this disease can survive for several years. • **Fatal familial insomnia:** Key symptom is a severe, progressive and untreatable form of insomnia. Can also lead to loss of basic bodily functions, such as blood pressure control. Coma and death eventually occur.	• People who are over the age of 50 are at greater risk, though these diseases can develop at younger ages. • People who have a mutation in the **human prion protein gene** are much more likely to develop prion diseases. • Prion disease mutations can be passed from parent to child; people who have a parent or sibling with the mutation are at greater risk than those who do not. • Specifically, if a parent has a human prion protein gene mutation, the chance they will pass it down is 50% for each child. If a person has a sibling with a mutation, there is a 50% chance that they have it too. • **Genetic counselling** is strongly recommended for anyone who has questions about their genetic risk for prion disease. Genetic counselling services are available in most large medical centres in Canada. If you have questions about genetic testing for this disease, you can contact nurses at the **Canadian CJD Surveillance System** at **1-888-489-2999**.
Iatrogenic type of Creutzfeldt-Jakob disease	
Similar symptoms to sporadic Creutzfeldt-Jakob disease. Many cases show early symptoms of movement disorder, with dementia appearing later.	• Many risk factors for this type have now been reduced, as scientific knowledge and medical technology have improved since this type of Creutzfeldt-Jakob disease was first identified.

	• To reduce risk, hospitals use special procedures to ensure that medical and surgical instruments are safe to use on patients. Lab workers also take safety precautions when handling specimens during diagnostic testing procedures. • Also to reduce risk, funeral services workers follow their provincial regulations when handling the remains of a person suspected to have had Creutzfeldt-Jakob disease. It is important to discuss a diagnosis of this disease with the funeral director when making funeral arrangements.
Variant type of Creutzfeldt-Jakob disease	
Often younger than those with other types of the disease. People in their 20s can be impacted, for example. Symptoms can include: • Anxiety, depression and withdrawal. • Ataxia or feeling unsteady when walking or standing. • Persistent pain or other strange sensations • Dementia that often begins later with this type. Death usually occurs one to two years after symptoms begin.	• The main risk factor is being exposed to **bovine spongiform encephalopathy** (also known as **mad cow disease**). • Overall risk in Canada is now considered extremely low, as the Canadian government requires the national beef industry to follow certain safety measures to reduce risk. • Receiving a blood transfusion from someone with the disease also increases risk considerably. In the UK, there have been five people who were likely 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. • To reduce the risk around blood transfusion, the Canadian blood donation system (among others) now avoids blood donations by people who may have Creutzfeldt-Jakob disease.

It can be difficult to diagnose Creutzfeldt-Jakob disease, especially in its early stages.

There is no test to accurately diagnose Creutzfeldt-Jakob disease in a living person. The best way to confirm whether a person has had this disease is to examine brain tissue after death via an autopsy.

Doctors can, however, do detailed exams and many tests to help reach a diagnosis during a person's lifetime.

Doctors may take the following steps towards diagnosis:

- **Medical history and neurological exam.** Creutzfeldt-Jakob disease usually progresses rapidly. A detailed history and neurological exam will help doctors learn when signs and symptoms started. It will also help them check for signs and characteristic symptoms of this disease.
- **Magnetic resonance imaging (MRI).** An MRI produces a picture of the brain that can show signs of disease. Many people with Creutzfeldt-Jakob disease and other neurological diseases show specific changes by MRI. An MRI can also sometimes distinguish between sporadic and variant forms of this disease.
- **Electroencephalography (EEG).** An EEG measures the brain's electrical activity. Sometimes, there is a specific EEG pattern called **sharp waves** that can help to diagnose Creutzfeldt-Jakob disease.
- **Lumbar puncture.** If a lumbar puncture finds abnormal prion protein in cerebrospinal fluid, that can be a strong indication of Creutzfeldt-Jakob disease. In Canada, two related tests (EP-QuIC and RT-QuIC) are available to help diagnose the disease.

- **Blood test.** There is no useful blood test for Creutzfeldt-Jakob disease. But a blood sample is often used to prepare DNA that can be tested to diagnose genetic prion disease. Sometimes, people who have no family history of disease can still have a mutation, so genetic testing is still recommended if doctors suspect Creutzfeldt-Jakob disease.

Again, the best way to confirm Creutzfeldt-Jakob disease is actually by looking at the brain after death, using a microscope. Brain autopsies are performed only in specific hospitals in Canada. The **Canadian CJD Surveillance System** can help make arrangements for a brain autopsy if CJD is suspected and if next of kin give consent.

There is currently no known cure or treatments for Creutzfeldt-Jakob disease.

No medical treatments have been approved to prevent Creutzfeldt-Jakob disease, and there is no effective way to slow its progression. Researchers are working on this.

In terms of managing symptoms, supportive nursing care is recommended in middle and later stages to focus on keeping the person as comfortable as possible.

Support is available.

Your local Alzheimer Society is here to support people impacted by dementia in your area—including dementia related to Creutzfeldt-Jakob disease. Visit **alzheimer.ca/Find** or call **1-855-705-4636 (INFO)** for more information and support.

Additional resources.

Canadian CJD Association. canadiancjdassociation.com

CJD Surveillance System of Canada. https://www.canada.ca/en/public-health/services/surveillance/blood-safety-contribution-program/creutzfeldt-jakob-disease.html 1-888-489-2999.

Prion Diseases. canada.ca/en/public-health/services/diseases/prion-diseases.html

Canadian Association of Genetic Counsellors. cagc-accg.ca

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