CONDITION

Creutzfeldt-jakob disease

A rare condition in which abnormal prion proteins cause rapid, severe damage to the brain.

WHAT IT IS
Rare, rapidly progressive brain condition caused by prions

BODY SYSTEM
Nervous system

MAIN SYMPTOMS
Rapid mental decline, poor coordination, muscle jerks, vision changes

WHAT CAUSES IT
Misfolded prion proteins; spontaneous, inherited, or rarely acquired

HOW IT IS DIAGNOSED
Brain tissue testing, usually after death

MAIN TREATMENT
Supportive and palliative care

CAN IT BE CURED?
No - no treatment stops prion damage

IS IT AN EMERGENCY?
Sometimes - sudden confusion, seizure, choking, or unresponsiveness

LONG-TERM OUTLOOK
Rapid progression, usually fatal within months to a year

Overview

Creutzfeldt-Jakob disease, usually shortened to CJD, is a rare condition that causes brain cells to become damaged and die. It is caused by abnormal proteins called prions .

Proteins normally fold into specific shapes so they can work correctly. In CJD, a prion protein folds into an abnormal shape. It can then cause nearby proteins to misfold as well. This process damages brain tissue.

CJD usually gets worse much faster than more common causes of dementia. Problems with thinking, movement, balance, vision, or behaviour may develop over weeks or months. The condition eventually affects basic functions such as swallowing, speaking, and responding to other people.

There are several forms of CJD. Most cases are sporadic , meaning they happen without an

identifiable reason. Some are caused by an inherited gene change. A very small number have been acquired through particular medical exposures or, in variant CJD, past exposure to cattle infected with bovine spongiform encephalopathy.

CJD is not spread through touching, hugging, sharing meals, coughing, sex, or providing ordinary personal care.

Symptoms

Symptoms differ between people, but they usually progress quickly. Early symptoms may include:

As the condition progresses, movement problems often become more noticeable. Myoclonus means brief, sudden muscle jerks that the person cannot control. Muscles may also become stiff, weak, or difficult to coordinate.

Thinking and communication usually decline. A person may become confused, have difficulty recognising people or places, or need help with everyday activities. Hallucinations can occur, meaning seeing or hearing things that are not present.

Later symptoms may include difficulty swallowing, loss of bladder or bowel control, seizures, and reduced awareness of the surroundings. Some people eventually enter a state called akinetic mutism , in which they appear awake but have little movement or speech.

Variant CJD can have a different early pattern. Mood changes, anxiety, withdrawal, pain, or unusual sensations may appear before clear problems with memory and movement.

These symptoms can also be caused by infections, strokes, immune conditions, medication effects, or other treatable problems. Symptoms alone cannot confirm CJD.

- Problems with memory, concentration, planning, or judgment
- Changes in mood, personality, sleep, or behaviour
- Difficulty walking or poor balance
- Blurred vision, double vision, or trouble understanding what is seen
- Slurred speech or difficulty finding words
- Unusual sensations, dizziness, or clumsiness

Causes and risk factors

CJD is caused by abnormal prion proteins building up in the brain. It is not caused by a

bacterium or virus.

The main forms are:

Ordinary contact with someone who has CJD does not place relatives, friends, or care workers at risk. Special handling is needed for certain brain, spinal, and eye tissues because standard sterilisation may not destroy prions. Healthcare and laboratory teams follow specific infection-control procedures for these materials.

Having a relative with sporadic CJD does not by itself mean the condition was inherited.

- Sporadic CJD: The prion change appears to happen spontaneously. There is usually no family history or known exposure.
- Genetic CJD: A change in the `PRNP` gene makes abnormal prion folding more likely. The gene change can be passed through families, although family histories are not always clear.
- Iatrogenic CJD: This is linked to certain medical procedures performed in the past, including treatment with contaminated human-derived hormones or particular tissue grafts. Modern controls have greatly reduced these exposures.
- Variant CJD: This has been linked mainly to eating beef products contaminated with the agent that causes bovine spongiform encephalopathy, sometimes called "mad cow disease."

Diagnosis

Diagnosing CJD during life can be difficult because other conditions can cause rapid changes in thinking and movement. The medical team will first look for potentially treatable causes.

Assessment usually includes a detailed history, neurological examination, medication review, and blood tests. Relatives or close friends may help describe when symptoms began and how quickly they changed.

Tests used in the assessment include:

Results are considered together with the symptoms and their rate of progression. MRI and RT-QuIC can provide strong evidence, but no single routine test answers every case.

Definitive confirmation requires examination and specialised testing of brain tissue. This is most often done after death. A brain biopsy during life is uncommon because it has risks and may not change treatment. Arrangements for specialist testing can be discussed sensitively with the person and family.

- MRI brain scan: This can show characteristic changes in particular areas of the brain.
- Lumbar puncture: A needle is used to collect fluid from around the spinal cord. A test called RT-QuIC looks for evidence of abnormal prion activity. Other fluid proteins may provide supporting evidence.
- EEG: Sensors placed on the scalp record the brain's electrical activity. Some people with CJD develop a characteristic pattern.
- Genetic testing: Testing of the `PRNP` gene may be offered when an inherited form is possible. Genetic counselling is important before and after testing.

Treatment and management

There is currently no treatment that cures CJD or stops prion-related brain damage. Care focuses on comfort, safety, symptom relief, and support for the person and those close to them.

Treatment may help with muscle jerks, seizures, pain, anxiety, low mood, agitation, or sleep problems. Medicines are chosen according to the symptom and the person's overall condition. They need regular review because sedating medicines can worsen alertness, swallowing, or balance.

Physiotherapists and occupational therapists can advise on safer movement, seating, wheelchairs, and equipment at home. Speech and language therapists assess communication and swallowing. Changes in food texture, eating position, or assistance with meals may reduce discomfort and choking risk.

As swallowing becomes harder, the team may discuss assisted feeding. Feeding tubes do not stop the underlying condition, and they may not prevent all aspiration, which is when food or fluid enters the airway. Decisions should reflect the person's wishes, comfort, and goals of care.

Palliative care is appropriate from an early stage. It can be provided alongside neurological care and is not limited to the final days of life. It supports symptom control, emotional needs, advance care planning, and family caregivers. Hospice care may become helpful as the condition advances.

Complications

CJD can affect movement, awareness, and the ability to swallow. Possible complications

include falls, injuries, dehydration, poor nutrition, pressure sores, and loss of communication.

Swallowing problems can allow saliva, food, or drink to enter the lungs. This may cause choking or aspiration pneumonia. Reduced mobility also raises the risk of chest infections and blood clots.

In advanced CJD, the person usually becomes fully dependent on others for care and may become unresponsive.

Prevention

Most cases of sporadic CJD cannot be prevented because they occur without a known exposure or avoidable cause.

Inherited CJD also cannot be prevented through lifestyle changes. If a disease-causing `PRNP` gene change is known in a family, genetic counselling can explain inheritance, testing choices, and family-planning options. Genetic testing is a personal decision and can have emotional and family consequences.

Public health controls reduce acquired forms of CJD. These include monitoring cattle and food supplies, screening donated blood or tissue where appropriate, and using special procedures for instruments exposed to higher-risk tissues.

You cannot prevent CJD by avoiding normal contact with an affected person. Separate dishes, clothing, bathrooms, or living spaces are not needed.

Living with it and what to expect

CJD usually changes quickly over weeks and months rather than over many years. Most people eventually lose the ability to walk, communicate, swallow safely, and manage personal care. The condition is usually fatal within months to a year, although the exact course cannot be predicted for one person. Some forms, including variant and certain inherited prion conditions, may progress differently.

Needs can change faster than services and equipment are normally arranged. Early involvement of neurology, community nursing, therapy, social care, and palliative care can make transitions easier.

While the person can still take part, it may help to discuss where they would prefer to receive care, who should speak for them if they lose decision-making ability, and which treatments would or would not fit their wishes. Written advance care plans can guide relatives and

professionals later.

Familiar voices, music, gentle touch, and a calm environment may remain comforting even when communication becomes limited. Caregivers may need practical help, respite, and bereavement support. Accepting this support is part of caring for both the affected person and the family.

When to get medical help

Arrange a prompt medical assessment for rapidly worsening memory, behaviour, balance, coordination, speech, or vision. These symptoms do not necessarily mean CJD, but they need investigation because some possible causes are treatable.

During planned follow-up, report new falls, increasing muscle jerks, medication side effects, changes in eating or drinking, constipation, pain, sleep problems, or growing care needs. These issues may require changes to support or symptom management.

Seek urgent same-day help for:

Call emergency services for sudden severe confusion, stroke-like symptoms, serious breathing difficulty, prolonged or repeated seizures, major choking, or unresponsiveness. Sudden symptoms should not be assumed to be part of CJD.

- New difficulty swallowing or repeated choking
- Inability to drink enough fluid
- Repeated falls or a significant injury
- Fever, cough, breathing changes, or possible chest infection
- Severe agitation or behaviour that creates an immediate safety risk

Common questions

Is Creutzfeldt-Jakob disease contagious?

Not through everyday contact. It is not spread by touching, hugging, coughing, sharing food, sexual contact, or routine caregiving. Certain tissues and medical instruments require specialist handling, but this does not create a risk during ordinary home care.

Is CJD the same as dementia?

CJD can cause dementia, meaning a decline in thinking that interferes with daily life. However, it is not the same as common dementia conditions such as Alzheimer's disease. CJD usually

progresses much faster and often causes early balance, vision, coordination, or muscle-jerk symptoms.

Can an MRI confirm CJD?

An MRI can show changes that strongly support the diagnosis, but it is interpreted with the symptoms and other tests. Cerebrospinal fluid RT-QuIC testing can add strong evidence. Definitive confirmation usually requires specialised examination of brain tissue.

Does CJD run in families?

Some cases are caused by inherited changes in the `PRNP` gene, but most are not. A person with sporadic CJD may have no affected relatives. When an inherited form is suspected, a genetics service can explain testing and what a result could mean for relatives.

Is there any cure or treatment that slows CJD?

There is currently no proven cure or treatment that stops its progression. Treatment can still ease muscle jerks, seizures, pain, anxiety, sleep problems, and other symptoms. Therapy, nursing, social care, and palliative care can support comfort and safety.

Is CJD related to mad cow disease?

Variant CJD is linked to the agent that causes bovine spongiform encephalopathy in cattle. It is different from classic CJD, which is usually sporadic or inherited. Food-safety measures were introduced to reduce the risk of further exposure.

Sources

- Centers for Disease Control and Prevention - About Classic Creutzfeldt-Jakob Disease
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- World Health Organization - Infection Control Guidelines for Transmissible Spongiform Encephalopathies