

Causes

Creutzfeldt-Jakob disease

Overview (Link: www.nhs.uk/conditions/creutzfeldt-jakob-disease-cjd/)

- Symptoms (Link: www.nhs.uk/conditions/creutzfeldt-jakob-disease-cjd/symptoms/)
- **Causes**
- Diagnosis (Link: www.nhs.uk/conditions/creutzfeldt-jakob-disease-cjd/diagnosis/)
- Treatment (Link: www.nhs.uk/conditions/creutzfeldt-jakob-disease-cjd/treatment/)
- Prevention (Link: www.nhs.uk/conditions/creutzfeldt-jakob-disease-cjd/prevention/)

Creutzfeldt-Jakob disease (CJD) is caused by an abnormal infectious protein in the brain called a prion.

Proteins are molecules made up of amino acids that help the cells in our body function.

They begin as a string of amino acids that then fold themselves into a 3-dimensional shape.

This "protein folding" allows them to perform useful functions within our cells.

Normal (harmless) prion proteins are found in almost all body tissues, but are at the highest levels in brain and nerve cells.

The exact role of normal prion proteins is unknown, but it's thought they may play a role in transporting messages between certain brain cells.

Mistakes sometimes occur during protein folding and the prion protein can't be used by the body.

Normally, these misfolded prion proteins are recycled by the body, but they can build up in the brain if they aren't recycled.

How prions cause CJD

Prions are misfolded prion proteins that build up in the brain and cause other prion proteins to misfold as well.

This causes the brain cells to die, releasing more prions to infect other brain cells.

Eventually, clusters of brain cells are killed and deposits of misfolded prion protein called plaques may appear in the brain.

Prion infections also cause small holes to develop in the brain, so it becomes sponge-like.

The damage to the brain causes the mental and physical impairment associated with CJD, and eventually leads to death.

Prions can survive in nerve tissue, such as the brain or spinal cord, for a very long time, even after death.

Types of CJD

The different types of CJD are all caused by a build-up of prions in the brain. But the reason why this happens is different for each type.

Sporadic CJD

Even though sporadic CJD is very rare, it's the most common type of CJD, accounting for around 8 in every 10 cases.

It's not known what triggers sporadic CJD, but it may be that a normal prion protein spontaneously changes into a prion, or a normal gene spontaneously changes into a faulty gene that produces prions.

Sporadic CJD is more likely to occur in people who have specific versions of the prion protein gene.

At present, nothing else has been identified that increases the risk of developing sporadic CJD.

Variant CJD

There's clear evidence that variant CJD (vCJD) is caused by the same strain of prions that causes bovine spongiform encephalopathy (BSE, or "mad cow" disease).

In 2000, a government inquiry concluded that the prion was spread through cattle that were fed meat-and-bone mix containing traces of infected brains or spinal cords.

The prion then ended up in processed meat products, such as beef burgers, and entered the human food chain.

Strict controls have been in place since 1996 to prevent BSE entering the human food chain, and the use of meat-and-bone mix has been made illegal.

It appears not everyone who's exposed to BSE-infected meat will go on to develop vCJD.

Almost all definite cases of vCJD occurred in people with a specific version (MM) of the prion protein gene, which affects how the body makes a number of amino acids.

It's estimated up to 4 in 10 of the UK population have this version of the gene.

Cases of vCJD peaked in the year 2000, in which there were 28 deaths from this type of CJD. There have been no confirmed deaths from 2017 to 2020.

Some experts believe that the food controls have worked and further cases of vCJD will continue to decline, but this doesn't rule out the possibility that other cases may be identified in the future.

It's also possible for vCJD to be transmitted by blood transfusion (Link: www.nhs.uk/tests-and-treatments/blood-transfusion/), although this is very rare and measures have been put in place to reduce the risk of it happening.

We don't know how many people in the UK population could develop vCJD in the future and how long it'll take for symptoms to appear, if they ever will.

A study published in October 2013 that tested random tissue samples suggested around 1 in 2,000 people in the UK population may be infected with vCJD, but show no symptoms to date.

Familial or inherited CJD

Familial or inherited CJD is a rare form of CJD caused by an inherited mutation (abnormality) in the gene that produces the prion protein.

The altered gene seems to produce misfolded prions that cause CJD. Everyone has 2 copies of the prion protein gene, but the mutated gene is dominant.

This means you only need to inherit 1 mutated gene to develop the condition. So if 1 parent has the mutated gene, there's a 50% chance it will be passed on to their children.

As the symptoms of familial CJD don't usually begin until a person is in their 50s, many people with the condition are unaware that their children are also at risk of inheriting this condition when they decide to start a family.

Iatrogenic CJD

Iatrogenic CJD (iCJD) is where the infection is spread from someone with CJD through medical or surgical treatment.

Most cases of iatrogenic CJD have occurred through the use of human growth hormone to treat children with restricted growth (Link: www.nhs.uk/conditions/restricted-growth/).

Between 1958 and 1985, thousands of children were treated with the hormone, which at the time was extracted from the pituitary glands (a gland at the base of the skull) of human corpses.

A minority of those children developed CJD, as the hormones they received were taken from glands infected with CJD.

Since 1985, all human growth hormone in the UK has been artificially manufactured, so there's now no risk.

But a small number of people exposed before 1985 are still developing iCJD.

A few other cases of iCJD have occurred after people received transplants of infected dura mater (tissue that covers the brain) or came into contact with surgical instruments that were contaminated with CJD.

This happened because prions are tougher than viruses or bacteria, so the normal process of sterilising surgical instruments had no effect.

Once the risk was recognised, the Department of Health tightened the guidelines on organ donation (Link: <https://www.organdonation.nhs.uk/>) and the reuse of surgical equipment. As a result, cases of iCJD are now very rare.

BSE ('mad cow' disease)

Bovine spongiform encephalopathy (BSE), also known as "mad cow" disease, is a relatively new disease that first occurred in the UK during the 1980s.

One theory about why BSE developed is that an older prion disease that affects sheep, called scrapie, may have mutated.

The mutated disease may have then spread to cows that were fed meat-and-bone mix from sheep containing traces of this new mutated prion.

Is CJD contagious?

In theory, CJD can be transmitted from an affected person to others, but only through an injection or consuming infected brain or nervous tissue.

There's no evidence that sporadic CJD is spread through ordinary day-to-day contact with those affected or by airborne droplets, blood or sexual contact.

But in the UK, variant CJD has been transmitted on 5 occasions by blood transfusion.

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