

Types of Prion Disease

There are three different subtypes of prion disease categorized by how the disease is contracted. All differ slightly with regards to typical signs, symptoms and duration of illness. The subtypes are:

- Sporadic
- Genetic
- Acquired

Sporadic

Sporadic Creutzfeldt-Jakob disease (sCJD)

The cause of "classic" or "sporadic" CJD is unknown, which means it occurs in people without any known risk factors or gene mutations. Typical symptoms include imbalance and incoordination, memory loss and impaired thinking, and psychiatric symptoms such as anxiety or depression. Once the symptoms do appear, CJD progresses very quickly and is usually fatal within a few months of symptom onset. sCJD typically affects people in their 60s and is rarely seen in people younger than 40 years old. Sporadic CJD is the most common form.

Sporadic Fatal Insomnia (sFI)

Like sCJD, sFI is caused by a misfolded protein, but also like sCJD, what causes that protein to misfold is unknown. Symptoms include difficulty falling asleep (insomnia), difficulty walking, weight loss and excessive tears in the eye. The symptoms rapidly progress to loss of consciousness and death.

Genetic

Familial Creutzfeldt-Jakob disease (fCJD)

Inherited mutations in the prion protein gene (PRNP) cause the familial form of prion disease. This prion gene provides instructions to your cells regarding how to make the prion protein. In fCJD, the mutations in this gene cause the cells to produce an abnormal form of the prion protein instead of the normal form. In most cases, the patient with fCJD inherits the altered gene from one affected parent. In some people, a new mutation in the gene causes fCJD. Although such people most likely do not have an affected parent, they can pass the genetic change to their children. The symptoms depend on type of mutation, but they often look like classic CJD: balance and coordination problems, memory loss and impaired thinking. Typically these symptoms appear earlier in fCJD than in sCJD. The duration of the illness is usually longer than in the sporadic form. The genetically inherited subtype accounts for about 15% of CJD cases.

Gerstmann-Sträussler-Scheinker syndrome (GSS)

Fatal Familial Insomnia (FFI)

Acquired

CJD acquired by exposure to the abnormal prion protein accounts for less than 1% of known CJD cases. It is important to realize that CJD caused by exposure to the prion is extremely rare. There are three subtypes of this form:

Iatrogenic Creutzfeldt-Jakob disease (iCJD)

"Iatrogenic" literally means "physician-induced". Therefore, an iatrogenic disease is one associated with a medical practitioner or treatment. The signs and symptoms often look like classic CJD. The age at onset depends on the age at exposure and incubation time. The number of new cases of iCJD has fallen dramatically since practices changed to prevent contamination.

Cases of CJD have been linked to treatments using growth hormone prepared from human pituitary glands. Fortunately, a synthetic version of human growth hormone was developed in the 1980s, so growth hormone is now created in the laboratory rather than collected from people.

A few CJD cases have been linked to infected tissue transplants and grafts from donors who turned out to have CJD. There are no known instances where a sporadic or familial form of CJD has been passed to others through blood transfusions. Several cases of the type of CJD associated with eating infected beef have been passed through blood transfusions (see description below). Organ, tissue, and blood donors are now screened for CJD risk factors and not allowed to donate if they could potentially pass on abnormal prions to a recipient.

There have also been a few cases linked to contaminated instruments used in brain surgery. Because typical sterilization procedures do not eradicate abnormal prions, current recommendations are to destroy instruments that have been used on a patient with CJD or suspected CJD.

Variant Creutzfeldt-Jakob disease (vCJD)

This form has been linked to eating beef contaminated with Bovine Spongiform Encephalopathy (BSE or "mad cow disease") in cattle. In the early stages patients often present with personality changes and psychiatric symptoms such as depression or withdrawal. Psychiatric symptoms are often the most prominent feature early in vCJD, but dementia develops later. The motor symptoms of vCJD (stumbling, falls and difficulty walking) also tend to appear earlier in vCJD than in classic CJD. The estimated incubation period is 5 to 40 years, and the duration of illness is typically 12-14 months after signs and symptoms appear. vCJD affects people in their 20s, much earlier in age than people with sporadic CJD. One person with variant CJD has been identified in the United States and one in Canada, however, both lived in the UK during the BSE epidemic and contracted their illness from exposure in the UK.

Kuru

In the 1950s-1960s, kuru reached epidemic proportions in the South Fore tribe of Papua New Guinea. Although researchers do not know how it started, they know it spread when tribal members ritualistically consumed the tissue of affected people during funeral rites. Kuru is characterized by walking problems, shaking of the limbs, slurred speech and mood changes, but little or no dementia. It is usually fatal within 6 to 12 months. Kuru disappeared with the end of cannibalistic practices in New Guinea.