Transmissible Spongiform Encephalopathies

Transmissible spongiform encephalopathies (TSEs) are also known as prion diseases. They are a group of rare degenerative brain disorders. They are characterized by tiny holes that give the brain a "spongy" appearance. These holes can be seen when brain tissue is viewed under a microscope.

Creutzfeldt-Jakob disease (CJD) is the most well-known of the human TSEs. It is a rare type of dementia. It affects about one in every one million people each year. Other human TSEs include:

- Kuru.
- Fatal familial insomnia (FFI).
- Gerstmann-Straussler-Scheinker disease (GSS).

Kuru was identified in people of an isolated tribe in Papua New Guinea. It has now almost disappeared. FFI and GSS are rare hereditary diseases. They are found in just a few families around the world. A new type of CJD, called variant CJD (vCJD), was first described in 1996. It has been found in Great Britain and several other European countries. The initial symptoms of vCJD are different from those of classic CJD. And the disorder typically occurs in younger patients. Research suggests that vCJD may have resulted from human consumption of beef from cattle with a TSE disease. The disease is called bovine spongiform encephalopathy (BSE). It is also known as "mad cow disease." Other TSEs found in animals include:

- Scrapie. It affects sheep and goats.
- Chronic wasting disease. This affects elk and deer.
- Transmissible mink encephalopathy.

In a few rare cases, TSEs have occurred in other mammals such as zoo animals. These cases are probably caused by contaminated feed. CJD and other TSEs also can be transmitted experimentally to mice and other animals in the laboratory.

CAUSES

Research suggests that TSEs are caused by an abnormal version of a protein called a *prion. Prion* is short for *proteinaceous infectious particle*. Prion proteins occur in 2 forms. The normal form is a harmless protein found in the body's cells. But the infectious form causes disease. The harmless and infectious forms of the prion protein are nearly identical. But the infectious form takes on a different folded shape from the normal protein.

Human TSEs can occur three ways:

- Sporadically.
- Hereditary diseases.
- Transmission from infected individuals.

Sporadic TSEs may develop because some of a person's normal prions spontaneously change into the infectious form of the protein. They then alter the prions in other cells in a chain reaction. Inherited cases arise from a change, or mutation, in the prion protein gene that causes the prions to be shaped in an abnormal way. This genetic change may be transmitted to an individual's offspring. Transmission of TSEs from infected individuals is relatively rare. TSEs cannot be transmitted through the air or through touching or most other forms of casual contact. Bu they may be transmitted through contact with infected tissue, body fluids, or contaminated medical instruments. Normal sterilization procedures such as boiling or irradiating materials do not prevent transmission of TSEs.

SYMPTOMS

Symptoms of TSEs vary. But they commonly include:

- Personality changes.
- Psychiatric problems such as depression.
- Lack of coordination.
- Unsteady gait.

Patients also may experience:

- Involuntary jerking movements (*myoclonus*).
- Unusual sensations.
- Insomnia.
- Confusion.
- Memory problems.
- In the later stages of the disease, patients have severe mental impairment. They also lose the ability to move or speak.

TREATMENT

TSEs tend to progress rapidly. They usually result in death over the course of a few months to a few years. There is currently no treatment that can halt progression of any of the TSEs. Treatment is aimed at relieving symptoms and making the patient as comfortable as possible.

RESEARCH BEING DONE

The NINDS conducts and supports research on TSEs. This research is aimed at:

- Determining how abnormal prion proteins lead to disease.
- Finding better tests for diagnosing CJD and other disorders.
- Finding ways to treat TSEs.

Research is continually being done to learn more about this problem.

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