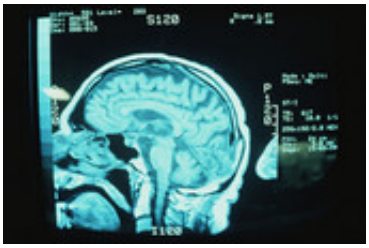
Deadly brain illness discovered in British family

November 13 2013, by Randy Dotinga, Healthday Reporter



Condition destroys tissue much like mad cow disease, but is inherited and not infectious.

(HealthDay)—A British family harbors a deadly inherited illness that destroys the brain in a way that is similar to the dreaded mad cow disease, researchers report.

The newly discovered condition doesn't appear to be contagious, unlike mad cow disease. As a result, it seems to pose no threat to people who aren't part of the family.

But researchers believe members of the family face a 50 percent likelihood that they'll develop the disease in mid-life and die. There is no treatment.

"It has been passed on from generation to generation," said study author Dr. Simon Mead, a neurologist and lead clinician at London's National

Prion Clinic. "But we are developing treatments, and hope to start clinical trials soon."

Researchers believe the condition isn't caused by any of the usual suspects that make people sick—bacteria, viruses, parasites and fungus. Instead, the culprits are "prions"—renegade proteins that cause death by poking holes, Swiss cheese-style, in the brain.

Mad cow disease, spread by the consumption of infected tissue, is the most famous form of prion disease, but there are several others. One, fatal familial insomnia, is inherited and causes increasing sleeplessness that leads to death. Another, sporadic Creutzfeldt-Jakob disease, occurs in people for no apparent reason. Yet another type, called kuru, has been found in cannibals who eat infected human brains.

In the new study, researchers say they've confirmed that a disease caused by prions has infected multiple members of the British family who are descended from a single couple who had three children; it's not clear if the disease was present in even earlier generations. They reached their conclusions after studying six of 11 family members who seem to have been affected.

It appears that physicians misdiagnosed those family members with the disease after they began suffering from chronic diarrhea in their 30s and a series of other symptoms, such as nerve pain. The apparent sufferers developed thinking problems and seizures in their 40s and died, on average, at the age of 57, although one lived to be 70.

The researchers believe that nine people in five generations had the illness and died; another two, from the most recent generation, have the disease and are still alive.

Mead said the disease is unusual because the prions seem to have

damaged the intestines, not just the brain, leading to the diarrhea. It's possible that the prions damaged nerves in the bowels, he said. "Most prion diseases cause damage in the nervous system," he said. "Prions are toxic to brain cells."

Mead said researchers are trying to develop a treatment that will rally the body's immune system against the renegade proteins.

Robert Rohwer, an associate professor of neurology at the University of Maryland at Baltimore School of Medicine who is familiar with prion diseases, said the study is "an excellent piece of work."

Unfortunately, he said, there's no way for members of the family to prevent the spread of the disease other than by simply not having children.

In the bigger picture, he said, scientists are studying the possible connections between prion disease and conditions like Alzheimer's disease, which slowly destroys the brain in a similar fashion. That leads to the prospect that better understanding of one type of disease could lead to more insight into the other.

The study appeared in the Nov. 14 issue of *The New England Journal of Medicine*.

More information: For more about prion diseases, try the U.S. National Library of Medicine.

family.html