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Newresearch shows mad cow disease also caused by genetic mutation

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New findings about the causes of mad cow disease show that sometimes it may be genetic. "We now know it's also in the genes of cattle," said Juergen A. Richt, Regents Distinguished Professor of Diagnostic Medicine and Pathobiology at Kansas State University's College of Veterinary Medicine.

Until several years ago, Richt said, it was thought that the cattle prion disease bovine spongiform encephalopathy -- also called BSE or mad cow disease -- was a foodborne disease. But his team's new findings suggest that mad cow disease also is caused by a genetic mutation within a gene called Prion Protein Gene. Prion proteins are proteins expressed abundantly in the brain and immune cells of mammals.

The research shows, for the first time, that a 10-year-old cow from Alabama with an atypical form of bovine spongiform encephalopathy had the same type of prion protein gene mutation as found in human patients with the genetic form of Creutzfeldt-Jakob disease, also called genetic CJD for short. Besides having a genetic origin, other human forms of prion diseases can be sporadic, as in sporadic CJD, as well as foodborne. That is, they are contracted when people eat products contaminated with mad cow disease. This form of Creutzfeldt-Jakob disease is called variant CJD.

"Our findings that there is a genetic component to BSE are significant because they tell you we can have this disease everywhere in the world, even in so-called BSE-free countries," Richt said.

An article by Richt and colleague Mark Hall of the National Veterinary Services Laboratories in Ames, Iowa, was published online in the journal *PLoS Pathogens*. Richt conducted the research while working at the National Animal Disease Center operated in Ames, Iowa, by the U.S. Department of Agriculture's Agricultural Research Service.

Richt said that prion diseases including mad cow disease are referred to as "slow diseases."

"It's a slow process for infectious prion proteins to develop," he said. "That's why the disease takes a long time -- as long as several years -- to show up."

Richt said mad cow disease caused by genetics is extremely rare. A recent epidemiological study estimated that the mutation affects less than 1 in 2,000 cattle. The study was done in collaboration with the U.S. Department of Agriculture-U.S. Meat Animal Research Center in Clay Center, Neb., which is operated by the Agricultural Research Service.

Richt said the upside of knowing that mad cow disease has a genetic component is that it offers ways of stamping out the disease through selective breeding and culling of genetically affected animals. Therefore, Richt and his colleagues developed high throughput assays to offer the possibility for genetic surveillance of cattle for this rare pathogenic mutation.

"Genetic BSE we can combat," Richt said. "We have submitted a patent for a test system that can assess all bulls and cows before they're bred to see whether they have this mutation."

Source: Kansas State University