

Mutated gene found in dog disease the same in humans, researchers find

April 18 2011



This is Topper, a Tibetan terrier owned by Lynn Steinhaus of Columbia, before he began suffering from NCL. Credit: MU News Bureau

University of Missouri researchers believe both man and animal will benefit from their discovery that the same gene mutation found in Tibetan Terrier dogs can also be found in a fatal human neurological disorder related to Parkinson's disease.

Fabiana Farias, a doctoral candidate in Area Genetics at the University of Missouri, found the mutation as part of her thesis research. Gary Johnson, associate professor of Veterinary Pathobiology; Martin Katz, professor of Veterinary Pathobiology, and Dennis O'Brien, a professor in the Department of Veterinary Medicine and Surgery, along with a host of researchers from MU's College of Veterinary Medicine; College

of Agriculture, Food and Natural Resources (CAFNR) and the Mason Eye Institute, recently published the findings in *Neurobiology of Disease*.

The disease in Tibetan Terriers is called adult-onset neuronal ceroid-lipofuscinosis (NCL). Within the dogs' cells in the brain and eye, material that should be "recycled" builds up and interferes with nerve cell function. Due to this buildup, around the age of five years old, the dog begins to exhibit dementia, impaired visual behavior, loss of coordination, and shows unwarranted aggression.

NCL ultimately took the life of Topper, a Tibetan Terrier owned by Lynn Steinhaus of Columbia. Steinhaus said Topper showed increased shyness around age five, and showed a loss of muscle control later. Topper also suffered seizures before he was euthanized in July of 2009. Topper's DNA was used to further the study.

"This is really hard disease for dog owners to go through," Steinhaus said. "Those seizures are just terrible."

While there are many forms of NCL in humans, the symptoms of NCL are similar in people and [dogs](#), and the disease is ultimately fatal for both. Utilizing the canine genome map and DNA samples from dogs diagnosed with NCL, the researchers were able to pinpoint the specific gene that causes NCL. The mutation they discovered in dogs, however, causes a hereditary form of Parkinson's disease in humans. This suggests that the recycling that goes awry in NCL may also be involved in degenerative diseases like Parkinson's.

Now, DNA from dogs can be tested to identify the presence of the mutated gene, and that test can ensure that Tibetan Terrier breeders do not pass it on to the next generation. The researchers also believe that they may be able to test -potential human therapies on the animal population because they can use the DNA test to identify affected dogs

before they start to show symptoms.

"Looking through samples collected from hundreds of dogs over many years, we got to the point where we're able to say this is a disease caused by the mutation of one gene," Katz said. "Finding that gene was like finding a single house in a very large city – but we had the dog family history and the tools to look through the city in a systematic way to locate address of the mutation responsible for the disease."

The publication is the result of almost 10 years of work, and the researchers believe it couldn't have occurred without the unique combination of animal and human medical science at the University of Missouri.

"Dogs and people suffer from the same diseases, and it's much easier to discover gene issues in dogs because of the unique genetics of pure-bred dogs," O'Brien said. "Because we have a medical school and veterinary school near each other, we can find the [genes](#) in the dog and then find out if they cause a similar disease in people."

Provided by University of Missouri-Columbia

Citation: Mutated gene found in dog disease the same in humans, researchers find (2011, April 18) retrieved 2 May 2024 from <https://phys.org/news/2011-04-mutated-gene-dog-disease-humans.html>

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