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# Researchers find gene responsible for neurodegenerative disease in dogs, possibly in humans

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(PhysOrg.com) -- A North Carolina State University researcher has helped to locate and identify a gene responsible for a fatal neurodegenerative disease that affects American Staffordshire terriers. This same gene may be responsible for a similar rare, fatal disease in humans. Its discovery will lead to improved screening and diagnosis of the disease in dogs and is the first step in working toward a cure for both canines and humans.

Dr. Natasha Olby, associate professor of neurology, was part of a multi-national team of researchers who located the gene responsible for a variant of neuronal ceroid lipofuscinoses (NCL), a family of diseases that result in mental and motor deterioration - and eventually death - in the dogs.

The team's results were published in the Aug. 17 issue of the Proceedings of the National Academy of Sciences.

NCLs, while rare in humans, are most common in children, although an adult-onset form of the disease - known as Kufs' disease - does occur. In this adult disease, neurons within the brain gradually die, causing loss of vision, epilepsy, dementia and loss of coordination.

Olby saw the first case of a canine version of adult-onset NCL in American Staffordshire terriers in 2000. Over subsequent years, she

found that the disease was a widespread and hereditary problem within the breed, affecting one of every 400 registered dogs. The disease kills the neurons in the cerebellum, which controls balance. Over time, the cerebellum shrinks, motor control deteriorates, and the patient dies or is euthanized.

“The disease became so prevalent because it was a recessive disease with a late onset,” says Olby. “Carriers of a single copy of the mutated gene never develop symptoms, and dogs with two copies of the gene might not show symptoms until five or six years of age, so the mutation was able to take hold in the breeding population.”

Through genetic analysis, the research group was able to locate the specific gene - an entirely novel mutation that has not been reported in people. According to Olby, the novel nature of the mutation means that researchers can now test samples from humans with NCL to determine whether this same mutation causes Kufs’ disease in people.

“The canine disease is a good model of the adult human form of the disease,” says Olby. “We hope that this discovery will provide insight into the development of this disease.”

**More information:** “A canine Arylsulfatase G (ARSG) mutation leading to a sulfatase deficiency is associated with neuronal ceroid lipofuscinosis”, Published: Aug. 17, 2010, in *Proceedings of the National Academy of Sciences.*

Provided by North Carolina State University