

Genetic cause of deadly skin condition afflicting bull terriers discovered

March 22 2018



LAD affected puppy in the middle of two nonaffected littermates. A pronounced growth delay and a subtle coat color dilution are visible. Credit: Anina Bauer and colleagues

In a new study published March 22nd, 2018 in *PLOS Genetics*, Anina Bauer of the University of Bern and a large international research team, report the discovery of a mutation that causes lethal acrodermatitis (LAD), a deadly condition that causes skin lesions on the paws and face of affected dogs.

LAD is an inherited disease in [dogs](#) that causes painful, infected wounds on the paws, as well as poor growth, immune problems and death, usually before two years of age. The disease tends to run in families of [bull](#) terriers and miniature bull terriers but is poorly understood and has no known cure. The clinical course of LAD is very similar as in human acrodermatitis enteropathica, which is caused by zinc deficiency. However, in contrast to the deadly dog disease, human acrodermatitis enteropathica can easily be cured by supplementation with zinc. Researchers compared the genomes of LAD affected and healthy dogs to pinpoint the cause to a mutation in the gene that codes for the protein muskelin 1. The mutation alters the splicing of the messenger RNA and thus leads to a lack of functional muskelin 1, which plays diverse roles in cellular shape, adhesion and spreading, and intracellular transport. While the physiological cause of LAD is still unknown, the findings reveal a novel role for muskelin 1 in maintaining a healthy immune system and skin.

The discovery of the mutation that causes LAD will allow veterinarians and breeders to test for the [disease](#), for rapid diagnosis and to prevent the breeding of affected dogs. The study also provides a starting point to investigate the physiological cause of the various clinical manifestations related to LAD, which thus far has eluded researchers. For human patients with unsolved zinc-unresponsive acrodermatitis or immune deficiency problems, [mutations](#) affecting muskelin 1 are one potential cause worth investigating.

Corresponding author Tosso Leeb adds: "LAD has been known for more

than 30 years and the genetic defect is present at an alarmingly high frequency in Bull Terriers and Miniature Bull Terriers. The results of our research will facilitate genetic testing and should allow to immediately end the unintentional breeding of further affected dogs. I am very curious to see when we will finally understand the diverse functions of the enigmatic protein muskelin 1".

More information: Bauer A, Jagannathan V, Högler S, Richter B, McEwan NA, Thomas A, et al. (2018) MKLN1 splicing defect in dogs with lethal acrodermatitis. *PLoS Genet* 14(3): e1007264.
doi.org/10.1371/journal.pgen.1007264

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