Gene Mutation Found at UC Davis

ID of mutation in dogs will also aid humans.

In November 2008 the University of California, Davis, announced that the gene mutation that causes high levels of uric acid in all Dalmatian dogs and bladder stones in some Dalmatians has been identified by a team of researchers at the School of Veterinary Medicine. Veterinary geneticist and lead author of the study Danika Bannasch said, “This defect, which in dogs is peculiar to the entire Dalmatian breed, has been reported for nearly a century and was probably unintentionally introduced as breeders worked to select more distinctive spotting patterns.” The findings of the study were published Nov. 7 in Public Library of Science, a scientific journal.

The identification of the mutation gives Dalmatian breeders the ability to eliminate the trait from the breed. "It is now possible that this trait can be removed from the breed by crossing Dalmatians with the normal offspring of the original Dalmatian-Pointer breeding that occurred in the early 1970s," Bannasch said. On Dec. 1 the Veterinary Genetics Laboratory at UC Davis began offering DNA testing for the mutation in dogs.

What is Uric Acid?
All mammals excrete waste products in their urine: toxins, salts, water and other elements that if allowed to build up can cause harm to the individual. Only humans, great apes and Dalmatian dogs, however, always produce elevated levels of uric acid in their urine and blood; other breeds of dogs do not usually produce uric acid, although researchers discovered the same gene mutation in Bulldogs and Black Russian Terriers. Hyperuricosuria is the medical term that refers to elevated levels of uric acid in the urine. In humans this condition can result in kidney stones, hypertension and gout, a painful inflammation of joints. In Dalmatians hyperuricosuria results in the formation of bladder stones (and sometimes kidney stones) that often must be removed surgically and can be difficult to treat. Although the presence of this trait in Dalmatians was identified in the early 1900s, until now the gene responsible remained unidentified.

Study Findings
UC Davis researchers collected both DNA and urine samples from hundreds of dogs in order to identify the gene responsible for high uric acid levels. Further analysis of dogs that go back to the Pointer/Dalmatian backcross revealed that mutations to the gene SLC2A9 are the cause of elevated uric acid levels in Dals; the same gene was also recently reported to aid in the regulation of uric acid levels in humans.

During the study the same mutations also were also found in samples from some Bulldogs and Black Russian Terriers, breeds that are not known to be closely related to Dalmatians. According to UC Davis researchers, this suggests that the gene mutation must be quite old, even predating formation of the Dalmatian breed. Alternatively, the mutation could have been introduced to those breeds by crosses between breeds, researchers noted.

Because the gene mutation does not always occur in Bulldogs and Black Russian Terriers, breeders can simply use genetic selection to eliminate the undesirable trait from those breeds. Because the mutation occurs in all AKC registered Dalmatians, however, breeders must look outside the breed to correct the problem.

"In recent years, dogs that are about 99 percent Dalmatian and one percent Pointer have been bred, successfully eliminating the elevated uric acid trait," said Bannasch. "The result is a healthy dog that that looks like a Dalmatian, maintains the Dalmatian breed characteristics and is genetically almost identical." Although humans also carry the SLC2A9 gene, scientists have not yet identified the exact mechanism that causes humans and great apes to have elevated uric acid levels, but identification of the responsible mutation in dogs will lead to better understanding of the problem in humans.

Information on the testing program will be available online.
– Editor

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