

New genetic test can help eliminate a form of inherited blindness in dogs

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Dogs with progressive retinal atrophy have normal sight at birth, but by the age of four or five they will be totally blind. There is no treatment. Credit: University of Cambridge

Progressive retinal atrophy (PRA) is a group of inherited diseases that causes progressive degeneration of the light sensitive cells at the back of the eye. Dogs with PRA have normal sight at birth, but by the age of four or five they will be totally blind. There is no treatment.

Now a team led by the University of Cambridge has identified the genetic mutation that causes PRA in English Shepherd Dogs, and developed a DNA test for it. By identifying [dogs](#) carrying the disease before their eyesight starts to fail, this provides a tool to guide breeding decisions so the disease is not passed on to puppies.

Owners usually don't realize their dog has PRA until it is middle-aged, by which time it might have bred and passed on the faulty gene to its puppies. This has made it a difficult disease to control.

The new discovery means that progressive retinal atrophy can now be completely eliminated from the English Shepherd Dog population very quickly. The results are published in the journal *Genes*.

"Once the dog's eyesight starts to fail there's no treatment—it will end up totally blind," said Katherine Stanbury, a researcher in the University of Cambridge's Department of Veterinary Medicine and first author of the report.

She added, "Now we have a DNA test, there's no reason why another English Shepherd Dog ever needs to be born with this form of progressive retinal atrophy—it gives breeders a way of totally

eliminating the disease."



The new DNA test means there is no reason why another English Shepherd Dog ever needs to be born with progressive retinal atrophy, which causes total blindness. Credit: University of Cambridge

The genetic mutation identified by the team is recessive, which means it only causes blindness if the English Shepherd Dog inherits two copies of it. If the dog only has one copy, this makes it a carrier—it will not develop PRA but can pass the mutation on to its puppies. If two carriers are bred together, about one in four of the puppies will be affected with

PRA.

Dog breeds are very inbred, so many individuals are related—giving them a much higher chance of being affected by recessive diseases than humans.

The team began the research after being contacted by a distraught owner of an English Shepherd Dog that had been recently diagnosed with PRA. The dog had been working as a search and rescue dog but had to retire due to visual deterioration that has resulted in total blindness.

The researchers put out a call for DNA samples from other [owners](#) or breeders of this breed, and received samples from six English Shepherds with PRA and 20 without it. This was enough for them to pinpoint the genetic mutation responsible for PRA using whole genome sequencing.

The team offers a commercial canine genetic testing service providing DNA tests to dog breeders to help them avoid breeding dogs that will develop inherited diseases. As part of this, they will now offer a DNA test for Progressive Retinal Atrophy in English Shepherds. Anyone can buy a testing kit, costing just £48, to take a swab from inside their dog's mouth and send it back for testing.



Shola has Progressive Retinal Atrophy (PRA) – an incurable inherited disease that has caused her to go blind. Credit: University of Cambridge



Shola worked for the Edale Mountain Rescue Team before she was unable to carry on due to failing eyesight. Credit: University of Cambridge

"An owner won't necessarily notice their dog has got anything wrong with its eyes until it starts bumping into the furniture. Unlike humans who will speak up if their sight isn't right, dogs just have to get on with things," said Dr. Cathryn Mellersh in the University of Cambridge's Department of Veterinary Medicine, senior author of the report.

She added, "For the price of a decent bag of dog food, people can now have their English Shepherd tested for Progressive Retinal Atrophy prior to breeding. It's about prevention, rather than a cure, and it means a huge amount to the people who breed these dogs. They no longer need to worry about whether the puppies are going to be healthy or are going to develop this horrible disease in a few years' time."

The English Shepherd is a breed of herding dog popular in the United States and is closely related to the Border Collie.

The new discovery is the 33rd genetic mutation causing an inherited disease in dogs that the team has found—23 of which cause eye diseases. They say that the health and well-being of many dogs has been compromised because of how they have been bred by humans.

PRA occurs in many dog breeds, including the English Shepherd Dog. And it is similar to a disease called [retinitis pigmentosa](#) in humans, which also causes blindness. The researchers say that their work with dogs could shed light on the human version of the disease and potentially identify targets for gene therapy in the future.

More information: Exonic SINE insertion in FAM161A is associated with autosomal recessive progressive retinal atrophy in the English Shepherd, *Genes* (2024). [www.repository.cam.ac.uk/items ... 3c-9c4c-e8ed75c561b2](https://www.repository.cam.ac.uk/items/3c-9c4c-e8ed75c561b2)

Provided by University of Cambridge

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