NARRATOR: Since they were first domesticated from wolves, dogs have nuzzled, pawed, and pranced their way into hearts all over the world. People have herded and hunted with them, relied on them for protection, and bred them for function, fashion, and prestige. Some of us even count them among our family members. But dogs provide more than just good company; we learn from them too. Dogs are teaching us about our own health in ways that might surprise you. Biologist Elinor Karlsson studies dogs to discover the genetic basis of traits, including those related to human diseases.

[Elinor Karlsson, Professor, UMass Chan Medical School & Broad Institute]

ELINOR KARLSSON: So I always say that this is my dirty secret, except that it’s not much of a secret because I keep telling people. Not only did I not know anything about dogs when I first got involved in dog genetic research, but I am also a totally unapologetic cat person.

[cat meows]

NARRATOR: So what makes dogs such good subjects for studying genetics, and what can they teach us about human health?

ELINOR: Dogs have a very interesting history as a species. They were domesticated from wolves probably somewhere between 15,000 and 20,000 years ago. Much more recently, we decided to create these things called dog breeds.

[old-fashioned camera snaps]

NARRATOR: Most modern breeds began with British dog breeders in the 1800s.

ELINOR: They decided that they wanted to establish a whole group of dogs that all looked the same as each other. Not only that, they sometimes went for some pretty extreme looks.

NARRATOR: Though the British were far from the first to breed dogs, they set rigid physical standards for traits like size, color, and shape. In less than 200 years, they created or helped shape most of the dog breeds we know today. To get the most desirable versions of each breed’s traits, breeders often selected and bred dogs within the same family.

ELINOR: Within a dog breed, the dogs pretty much all look the same, and the way they’ve done this is by closing off that group of dogs in a genetic sense. They’re not letting any new dogs in. It means that all of the dogs within that breed are genetically quite similar to each other.

NARRATOR: In other words, dogs within a breed have very similar genetic information, or DNA. DNA contains many sections that affect traits, including genes. An organism’s complete set of DNA is their genome. Differences in genomes can be associated with differences in traits, including size and shape,
color, and even the risk of certain diseases. To find differences in the genome associated with traits, Elinor studies dogs within a breed. What makes these dogs ideal for her work?

ELINOR: Even though there’s still going to be differences between every individual dog in a breed, there’s not going to be nearly as many differences between each of those individual dogs than there are between a kind of randomly, you know, just a group of dogs that you kind of picked off of the street. Because we’re looking within a single breed, we’ve eliminated all of this background noise, all of these other differences that we’re not interested in.

NARRATOR: Elinor and her team used a breed called boxers to study one trait in particular.

ELINOR: We knew that we had this really interesting coat color phenotype in the boxers where some of them were completely white and some of them had almost no white fur at all.

NARRATOR: Elinor’s team can obtain DNA from cells in the dogs’ saliva. They analyzed the DNA from two groups of boxers, white boxers and solid-color boxers, to search for differences among their genomes. But since these genomes have billions of nucleotides, they focused on specific positions where nucleotides are known to vary among dogs. These positions are called single nucleotide polymorphisms, or SNPs.

ELINOR: Single means one, nucleotide is basically a base of DNA, and then polymorphism is a change. So some dogs, for example, will have an A and other dogs will have a C. Rather than looking at all 2.4 billion bases in the genome, we could actually focus in on these SNPs where we already knew that dogs tended to be different from one another. But what we were looking for was the place in the DNA where all of the white dogs looked different from all of the solid dogs.

NARRATOR: In other words, they were searching for SNPs associated with coat color.

ELINOR: So we looked at 20,000 positions in 10 white dogs and 20,000 positions in the genome in 10 dogs that didn’t have any white, and we got a place in the genome where we could see that the white dogs and the solid dogs looked extremely different from one another, much more than we’d expect by random chance. And it was sitting right over a gene named MITF.

NARRATOR: To find differences in the genome associated with coat color, they first needed a way to collect dog DNA.

ELINOR: We usually just get a saliva swab. It looks kind of like a giant Q-tip. And we basically ask the owner to put the swab into the cheek of their dog and just kind of move it around a bit and collect a bunch of saliva onto that giant Q-tip. The dogs don’t seem to mind getting swabbed at all, and I have to say that when we’re swabbing the puppies, they actually seem to get a kick out of chewing on the swabs.
NARRATOR: The location of SNPs like this suggested that MITF could be associated with coat color. Additional studies also suggested that the expression of MITF determines how much white color a boxer will have. The genomes of dogs and humans are very similar, and dogs and humans share many diseases. So, Elinor can use her methods in dogs to find parts of the genome associated with human diseases, like certain types of cancer.

ELINOR: Because we know that a lot of the same genes are involved in causing the cancer in people and in dogs, by figuring out what those causes are of the cancers in dog breeds that are at high risk, we can actually translate that directly over to human medicine.

NARRATOR: One of the cancers that Elinor studied is a type of bone cancer.

ELINOR: It’s a really terrible disease. It’s often a pediatric cancer, meaning that it’s happening in children or in teenagers. And it’s been really difficult to treat.

[razor buzzing]

NARRATOR: This cancer affects both humans and dogs. It occurs most commonly in breeds like the greyhound, Rottweiler, and Irish Wolfhound.

ELINOR: And so what we did is we took exactly like we’d done with coat color in boxers. We took a whole bunch of greyhounds that had gotten bone cancer and greyhounds that didn’t have bone cancer. And we did the same thing in the Rottweilers and the Irish Wolfhounds. And within each breed, we compared the ones that had the cancer to the controls, the one that didn’t have the cancer, and basically looked for differences between them.

NARRATOR: Elinor and her team found SNPs associated with the cancer in multiple parts of the genome, including certain genes. Scientists can now investigate whether these genes affect this bone cancer in both dogs and humans. But it’s not just bone cancers that dogs can help us learn about.

ELINOR: There’s many, many different diseases that dogs and people share that we’re hoping we’re actually going to be able to learn a lot more about by studying dogs. And so that includes, as I mentioned cancer, but it also includes heart disease, kidney disease, and psychiatric disorders.

NARRATOR: For Elinor, studying dogs to understand human diseases brings together all the best parts of being a scientist.

ELINOR: The first thing that as a scientist honestly keeps me doing science is just that I love discovering new things. The other thing is really the people. Both working with the graduate students and the staff scientists in my group, and working together as a team, none of us can do science on our own. We’re actually collaborating with all of the pet owners as well. And I think that the best science really should be a collaboration between the scientists and everybody else in our communities as well. With the dog work, we’re getting much closer to that than I have in other science that I’ve done.

[various dogs bark]