The discovery of various disease-associated genetic mutations has greatly changed the way some inherited canine diseases are managed. These mutations are not single basepair alterations but rather involve entire genes or regulatory regions that can be inherited from parent dogs. Through the use of genetic testing developed to identify these mutations, veterinarians can now more accurately predict which puppies are at risk of developing a specific disease and can sometimes even assume that a dog’s carrier status is linked to the presence of a mutation. This knowledge can be used by owners to help them breed their dogs responsibly and by veterinarians to obtain an accurate, definitive diagnosis and in some cases, may offer treatment strategies.

Pet Owner vs Breeder

The accuracy and specificity of an inherited disease diagnosis are particularly important in the world of dog breeding. Many puppies carrying specific mutations, such as carriers of the BLAD, are often bought as pets by unknowing dog owners who may not know the specific underlying molecular mechanisms of a disease (especially when there is also some breed-specific trait linked to the same allele). It is important to realize that not all puppies that develop canine diseases are the direct result of their breeders' carelessness. Breeders around the world are eliminating specific health conditions from their bloodlines through informed genetic testing and responsible breeding practices. However, in some cases, breeders may not be aware of specific mutations or may not make the time to implement the recommendations to prevent the cause of a disease. It is important to consider which mutation or mutations are known to be inherited in the particular breed presented to them. Though some ancient mutations that cause disease may be inherited in a breed, new genetic testing can be used to screen for the presence of many diverse dog breeds, mutations arising after the advent of our modern breeds may only be found in a single breed or even a single bloodline.

Blindness and Progressive Retinal Atrophy

Progressive retinal atrophy (PRA) is a hereditary disease process involving interaction of gene products from dozens of various genes resulting in the conversion of light to an electrical signal which is eventually interpreted by the brain as vision. Disruption of any of these gene products via genetic mutation may result in various inherited diseases of vision. Therefore, finding the specific mutation linked to a disease in a puppy means that, in most cases, the problem in the puppy’s parents can be ruled out. PRA is not always an easy task, but a crucial one for dog breeders, if possible. Though definitely not the breeds with the highest incidence of PRA, there are many dog breeds that have been found to have the disease. Some of these breeds include the Dalmatian, Pembroke Welsh Corgi, and other breeds that are prone to blindness and in blood clothing abnormalities. Progressive retinal atrophy and von Willebrand’s disease are two diseases of dogs with numerous associated mutations resulting in blindness and clothing dysfunction, respectively. In addition, many breeds are now undergoing genetic testing to find new substrates that could be used to breed into new breeds, subgroups based upon the underlying mutation or nuance in clinical signs.

Blindness and Progressive Retinal Atrophy

Progressive retinal atrophy (PRA) is a genetic disease syndrome that affects the retina of the eye, leading to a slow, irreversible loss of vision. It is caused by mutations in several genes, which can result in a range of clinical manifestations. The disease is characterized by progressive degeneration of the photoreceptors, leading to a loss of light-sensing function in the retina. The specific mutations associated with PRA can vary greatly, and genetic testing is essential to identify the specific mutation in a particular dog.

Evolutionary Biology and Dog Breeding

The fact that some dog breeds are more prone to certain diseases than others is not a coincidence. Over the course of thousands of years, breeders have intentionally bred for specific traits, such as color, size, and temperament, that have become associated with certain diseases. For example, certain dog breeds have a higher incidence of hip dysplasia, a condition that can cause lameness and pain. This is because breeders have selected for body structure that is aesthetically pleasing, but not necessarily functional.

Von Willebrand disease

Von Willebrand disease (VWD) is a genetic disorder that affects the body’s ability to control bleeding after injury. It is caused by mutations in the genes that encode components of the von Willebrand factor (vWF) and the von Willebrand factor-cleaving protease (vWF-C3). The disease is characterized by a deficiency or abnormality of one or both of these factors, which play a critical role in the formation of blood clots.

Genetics of Disease

The genetics of disease are complex and can involve multiple genes and environmental factors. Mutations in a single gene can lead to disease, but in many cases, the interaction of multiple genes and environmental factors is required. For example, a person with a mutation in a gene that regulates blood clotting may develop VWD if they are also exposed to certain environmental factors, such as smoking or certain medications.

Other Dog Breeds with Common Diseases

In addition to the breeds mentioned above, many other dog breeds are also at risk of developing specific diseases. For example, the Boxer is prone to developing elbow dysplasia and hip dysplasia, and the Beagle is prone to developing canine ataxia. It is important for breeders to be aware of the specific diseases that are most common in their breed and to take steps to reduce the risk of these diseases in their breeding programs.

Contact Paw Print Genetics

If you have questions about a case involving blindness or blood clotting abnormalities or have any other questions related to inherited canine diseases, please feel free to contact the Friendly Paw Print Genetics team at pawprintgenetics.com or contact us at 1-800-555-5555. We are here to help you with any questions you may have.

Reference: