

Role of genetic testing for diseases of companion animals and genetic counseling in modern veterinary medicine



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Abstract

Several hundred genetic mutations have now been associated with development of clinical diseases in dogs and cats. Many diagnostic testing laboratories now offer genetic tests and samples may be easily submitted. However, interpretation of the results and how it may impact an individual animal or a breeding program can be quite complicated. Regardless, with their veterinary medical knowledge, the veterinarian has a critical role in interpreting genetic results.

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Over 200 genetic mutations (variations, deletions, and insertions) have now been associated with development of a clinical disease in dogs and cats and an additional 100 or so companion animal diseases have been identified without a mutation yet identified. As veterinarians have become better at understanding and preventing nutritional and infectious diseases, disease prevalence has decreased; however, we are faced with several familial disease, particularly, but not limited to, the purebred dog or cat.

Several diagnostic laboratories now offer genetic tests for several canine and feline diseases. Samples may be submitted in a variety of forms including buccal swabs, blood samples in EDTA and semen straws, and samples can be submitted with or without the involvement of a veterinarian. However, the role of a veterinarian is critical in interpretation of genetic results. Although submission and running of genetic tests is not complicated, interpretation of the results and how it may impact an individual animal or a breeding program can be quite complicated and requires the medical knowledge of a veterinarian to help an owner understand the many complicated aspects of genetics, including variations in penetrance and expression. Without careful counseling and discussion with a trained veterinary professional, a pet owner or breeder could lose enthusiasm and trust in genetic testing and we could lose this important tool.

Once genetic testing becomes available for a disease, counseling a pet owner or breeder on how to apply test results is very important. A breed association may be quick to immediately remove animals with a mutation from the breeding pool. Although this approach may appear as a good idea, it can have substantial negative impact on the breed. If the mutant gene exists in 40% of a cat or dog population, a sudden reduction

in 40% of the gene pool could dramatically alter the breed. Additionally, due to complicated genetic phenomena noted in many genetic diseases, not all animals that have the mutation will develop a clinical form of the disease. These concepts need to be carefully weighed against the issues of continuing to produce animals that may carry a mutation and could potentially suffer from the disease. For the breeding animal, recommendations to continue breeding should be based on many factors including size of the breed association, type of disease, risk that the animals produced will develop disease from the mutation, and severity of the disease. Positive attributes of the animal should be considered.

Besides the development of healthier animals through breeding practices, genetics are a key way for veterinarians to become more proactively involved in patient care. Ability to develop a proactive management plan for animals that test positive for a disease mutation is the true benefit of clinical genetics. Although identification of mutations can be used for testing and developing a breeding program, it is unlikely that we can ever completely remove many of these variants from the population. Larger gains will be in the value of testing young animals for these mutations and developing clinical management plans that can be applied for the lifespan of that pet to provide the best possible care. Veterinary medicine needs to move away from being 'reactive-responding' once a disease has been diagnosed, to become as proactive as possible, identify animals at risk of a disease, and implement management plans to prevent or alter outcomes.