

VetGen's DNA Test to Detect Phosphofructokinase Deficiency in English Springer Spaniels

The Disease

Canine Phosphofructokinase (PFK) deficiency is an autosomal recessive genetic disease which prevents the metabolism of glucose into available energy resulting in exercise intolerance and muscle disease in Springer Spaniels. PFK deficiency also destroys red blood cells in affected dogs, leading to anemia. The PFK deficiency gene frequency in Springers is estimated to be 10% of the population. Because it is an autosomal recessive disorder, dogs that are "Carriers" of the disease show no signs of PFK deficiency, yet can pass the gene along and perpetuate the disease through breeding. Ultimately, the result is more affected animals. Although there is a significant frequency of this defect, there are no effective treatments for PFK deficiency. Therefore, breeders have heretofore been unable to combat the disease by using responsible breeding strategies to reduce the incidence of PFK deficiency in future generations of dogs.

The VetGen Genetic Solution

Based on research conducted at the University of Pennsylvania, VetGen has developed a highly reliable, non-invasive DNA test that provides a definitive diagnosis whether a dog is PFK deficiency - "Affected", "Carrier", or "Clear". With this information in hand, Springer and Cocker Spaniel breeders and owners have key insight into their bloodstock and can proceed to make informed decisions about training, showing, and breeding plans for each dog. By following the simple instructions provided in VetGen's DNA Sample Collection Kit, dog owners and breeders collect DNA samples using a soft cheek brush. By gently brushing the inside of the dog's cheek, cells containing DNA are removed. It is this DNA sample that VetGen analyzes to determine the genetic status of each dog. As soon as VetGen receives the completed DNA Sample Collection Kit, the DNA samples are processed and a diagnosis is formed. Within two to four weeks, this diagnosis is provided to the customer in a summary report. Useful for dogs of any age, the DNA sample collection, analysis, and reporting activities can be completed before puppies are placed at 6 to 10 weeks. As a supplement to the summary report, VetGen can assist its customers with genetic counseling services to further help them make informed breeding decisions to eliminate the PFK deficiency gene from their bloodlines while maintaining the integrity of their breed.

PFK Deficiency Genetic Registry

The results of the PFK deficiency test can be formally registered with the Orthopedic Foundation for Animals (OFA). VetGen recommends registering your tested dog with the OFA, regardless of the test findings. When used for breeding, dogs proven "Clear" of the disease gene should afford the breeder a significant advantage in selling puppies. For further information about the OFA's Registry for Phosphofructokinase Deficiency in Springer Spaniels, contact: Orthopedic Foundation for Animals, 2300 E. Nifong Boulevard, Columbia, MO 65201-3856, or telephone (573) 442-0418.