

## Phosphofructokinase Deficiency in the English Springer Spaniel

**Phosphofructokinase (PFK) deficiency** is an **inherited** disease that affects both the field trial and show lines of English Springer Spaniels. M-PFK is an enzyme required for the metabolism of glucose into useable energy. Without the PFK enzyme, some cells, such as muscle cells and red blood cells, cannot produce adequate energy for their needs. Therefore, affected dogs display the following intermittent, clinical signs: weakness, lethargy, exercise intolerance, poor performance, muscle cramps, anemia, jaundice, and dark-colored urine. Dark-colored urine, a hallmark of this disorder, usually appears after strenuous exercise or after excessive barking, panting, and heat exposure.

PFK deficiency is caused by a mutation of the gene that codes for the enzyme M-PFK. Because PFK deficiency is an autosomal recessive trait, a dog must have two copies of the mutated gene (one from each parent) to show clinical signs. A dog with one copy may be healthy but will pass the mutation to its offspring. The mutation has so far been documented in over 100 English Springer Spaniels, especially in field trial lines, but the actual prevalence of the mutation is unknown.

Researchers at the University of Pennsylvania have developed a simple, reliable test to detect the mutation that causes PFK deficiency. This test requires a DNA sample obtained from a cheek swab or blood sample.

### The following three results are possible:

- **Clear** - the dog has two normal PFK genes and does not carry the mutation. The dog will not show clinical signs and will not pass the mutation to its offspring. This dog can be used for breeding.
- **Carrier** - The dog is heterozygous and has one normal and one mutant gene (allele). The dog will not show any clinical signs; therefore, a carrier can be shown and used for field trials without compromising the dog's health. A carrier will pass the mutant gene to approximately half of its offspring, producing further carriers and continuing the presence of the mutant gene in the general population. Carrier animals should only be bred if they otherwise meet the health, temperament, and quality criteria for breeding; with full disclosure to the owner of the mated dog of the carrier status; with confirmation that the mated dog is not itself a carrier, as such a breeding would produce affected offspring; and with agreement between both parties to the mating that all puppies will be tested prior to placement with all puppies identified as carriers placed with a spay/neuter requirement.
- **Affected** - the dog has two copies of the mutant gene (allele). The dog will show intermittent clinical signs of disease and will not perform well in field trials. Further, affected dogs will pass the mutation to 100 percent of its offspring. Affected dogs should never be bred.

By testing and breeding appropriately, PFK deficiency can be rapidly eliminated from the breed. Based on the information this test provides the continued spread of this disease, the costs for diagnosis and management, and animal suffering can all be prevented.