

Phosphofructokinase Deficiency (PFK)

Disease Description

Phosphofructokinase (PFK), a major regulatory enzyme in all cells of the body, catalyzes the metabolism of sugar, and thereby is pivotal in the production of energy to maintain normal cell function. In particular, dogs with this enzyme deficiency have diseased red blood cells and muscle cells. PFK deficiency can present as mild to life-threatening episodic illness.

Particularly in field trial dogs, clinical signs of weakness, exercise intolerance, poor performance to outright refusal to move, and muscle cramps may be observed. Clinical manifestations usually resolve within hours to days. Affected dogs have a relatively normal life expectancy, however, situations that can precipitate such crises should be avoided.

This disorder is inherited as an autosomal recessive trait in English Springer Spaniels. The same disease has also been found in the Cocker Spaniel, the Whippet and mixed breed dogs. PFK deficiency and the associated clinical features are caused by single base pair change (mutation) in the genetic code of the gene for the enzyme. A molecular genetic screening test for PFK deficiency accurately determines whether a dog is affected, a carrier, or a normal dog.

Because of the intermittent and variable clinical signs and the suspected high prevalence of PFK deficiency in the English Springer Spaniel breed, we recommend the testing of all English Springer Spaniels used for field trialing, breeding, or prior to purchase of a puppy and any Springer or mixed breed dog with suggestive clinical signs.

Clinical Signs

A hallmark sign of this disease is intermittent dark urine, with the color of the urine ranging from orange to dark coffee-brown, which commonly develops following strenuous exercise, prolonged barking, and extensive panting. In severe forms, pale gums (anemia) or jaundice (yellow coloration of skin and gums) with fever and poor appetite can be observed. Particularly in field trial dogs, clinical signs of weakness, exercise intolerance, poor performance to outright refusal to move, and muscle cramps may be observed. Clinical manifestations usually resolve within hours to days.

Affected Breeds

English Springer Spaniel
American Cocker Spaniel
Whippets
Mixed Breed Dogs

Required Samples

EDTA Blood, 1-2 mL or 2-3 Buccal Swabs

Cost

Please see PennGen Price List for current DNA Test pricing.

*To submit a sample for PFK Testing please use our **DNA Submission Form***