



DNA Test for Phosphofructokinase Deficiency in English Springer Spaniels.

Phosphofructokinase deficiency is an inherited disorder of English Springer Spaniels characterised by haemolysis of the red blood cells and intolerance to exercise. The genetic defect underlying phosphofructokinase deficiency in English Springer Spaniels has been identified by research groups at the University of Pennsylvania. It is a tiny change (point mutation) in the M-type phosphofructokinase gene. Breeders will be sent results identifying their dog as belonging to one of three categories:

CLEAR: The dog has 2 copies of the normal gene and will neither develop phosphofructokinase deficiency, nor pass a phosphofructokinase deficiency gene to its offspring.

CARRIER: The dog has one copy of the normal gene and one copy of the mutant gene that causes phosphofructokinase deficiency. It will not develop phosphofructokinase deficiency but will, if bred from, pass on the mutant gene to, on average, 50% of its offspring.

AFFECTED: The dog has 2 copies of the mutant gene that causes phosphofructokinase deficiency and will develop the disease.

Inheritance

Phosphofructokinase deficiency is an autosomal recessive disorder. Crossing the three genotypes - clear, carrier or affected - will produce the following litters:

Parents	Litters
clear x clear	all clear
clear x carrier	approximately 50% clear 50% carriers
clear x affected	all carriers
carrier x carrier	approximately 25% clear, 50% carriers 25% affected
carrier x affected	approximately 50% clear 50% affecteds
affected x affected	all affecteds

Tests can be ordered through our Webshop <http://www.ahtdnatesting.co.uk>.