

VETERINARY TECHNICAL DATASHEET

Phosphofructokinase (PFK) Deficiency or Glycogen Storage Disease Type VII



Mutation Found In :Cocker Spaniel, English Cocker Spaniel, English Springer Spaniel, Whippet

Disorder Type

- Blood

Disease Severity

- Moderate

Background

Phosphofructokinase (PFK) is an enzyme that is crucial for production of energy from sugar sources in all cells of the body, especially red blood cells and muscle cells. Lack of this enzyme causes breakdown of the muscle cells (exertional myopathy) and red blood cells (hemolysis) particularly during exercise or excitement resulting in a range of effects including weakness and muscle cramps, discolored urine, anemia, and jaundice. Hereditary PFK deficiency affects mainly spaniel and mixed breed dogs.

Key Signs

- Hemolytic anemia
- Hemolytic crises
- Mild metabolic myopathy
- Pigmenturia
- Cardiac problems (observed in the Whippet only)

Clinical Description

Dogs suffering from PFK deficiency display hemolytic anemia (breakdown of red blood cells) especially with alkaline blood. Barking or panting, rigorous exercising, or high environmental temperature may trigger the clinical signs by increasing the body's pH value. This may lead to a hemolytic crisis causing anemia, fatigue, and fever. Affected dogs can also show pigmenturia and jaundice after an episode. Other milder clinical signs include muscle weakness and reduced exercise tolerance. Cardiac problems have also been observed in Whippets.

Mode of Inheritance

- autosomal recessive

Gene Name

- PFKM

Next Steps

Treatment is supportive care dictated by the severity of the clinical signs during an episode. Owners of affected dogs should be advised on how to manage their dog so that hemolytic episodes can be prevented.

References

Gerber K, Harvey JW, D'Agorne S, Wood J, Giger U. Hemolysis, myopathy, and cardiac disease associated with hereditary phosphofructokinase deficiency in two Whippets. *Vet Clin Pathol* 38(1):46-51, 2009.

Giger U, Harvey JW, Yamaguchi RA, McNulty PK, Chiapella A, Beutler E. Inherited phosphofructokinase deficiency in dogs with hyperventilation-induced hemolysis: increased in vitro and in vivo alkaline fragility of erythrocytes. *Blood* 65(2):345-51, 1985.

Hillström A, Tvedten H, Rowe A, Giger U. Hereditary phosphofructokinase deficiency in wachtelhunds. *J Am Anim Hosp Assoc* 47(2):145-50, 2011.

Inal Gultekin G, Raj K, Lehman S, Hillström A, Giger U. Missense mutation in PFKM associated with muscle-type phosphofructokinase deficiency in the Wachtelhund dog. *Mol Cell Probes* 26(6):243-247, 2012.

Smith BF, Stedman H, Rajpurohit Y, Henthorn PS, Wolfe JH, Patterson DF, Giger U. Molecular basis of canine muscle type phosphofructokinase deficiency. *J BiolChem* 16:271(33):20070-20074, 1996.