

# 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

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